



June 2018

List of rare diseases and synonyms:

Listed in alphabetical order

www.orpha.net

www.orphadata.org

METHODOLOGY

Orphanet provides a comprehensive inventory of rare diseases in Europe, published biannually as a list. Rare diseases registered in Orphanet are defined according to two scopes:

- Every entity is defined by its clinical homogeneity, regardless of its etiology or the number of causing genes identified;
- The rarity is defined according to the European legislation defining a prevalence threshold of not more than 5 affected persons per 10'000 (Regulation (EC) N°141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products, http://ec.europa.eu/health/files/eudralex/vol-1/reg_2000_141/reg_2000_141_en.pdf).

Registered rare diseases have been described in the international scientific literature (peer-reviewed articles) with at least two cases confirming that the clinical signs are not associated fortuitously. However, some diseases are registered although only one case has been reported in order to reproduce the comprehensiveness of a specific classification (notably within the classification of inborn errors of metabolism).

Rare diseases are registered with a preferred name and as many synonyms as necessary. A unique identifier, the ORPHA number, is randomly attributed by the database to each disease. This number is never re-used, so it is stable in time.

ORPHA number of rare diseases registered in the past may be absent from the current inventory. This is due to:

- Obsolescence of entries (e.g. duplicated entities, diseases that are not rare anymore);
- Deprecation of entities when an entity no longer exists per se but has been recognised as being another entity. In this case, information regarding the deprecated entity is moved and the users are redirected to the target entry.

Data collection

As new scientific knowledge arises, the Orphanet rare diseases inventory is updated through the regular addition/update of diseases via two non-exclusive sources: documented sources and/or expert advice.

The scientific knowledge is monitored through:

- A bi-monthly analysis of a defined set of international peer-reviewed scientific journals covering the diversity of medical specialities represented in Orphanet;
- A monthly Medline search algorithm: (nosology[Title] OR classification[Title] OR nomenclature[Title] OR terminology[Title]) AND (rare disease* OR syndrome* OR disorder*);
- Specific Medline queries according to requests from experts, users of the database or needs arising from services newly registered in Orphanet (e.g. diagnostic test, expert centre, patient organisation).

Update of the inventory of rare diseases is assessed monthly by a medical and scientific committee within Orphanet and further validated by consulted experts.

The Orphanet rare disease nomenclature is produced in English and is translated into other languages. A medical validation of the translations is carried out.

Data presentation

Preferred names and synonyms of diseases are listed by alphabetical order with their ORPHA number.

Deprecated entities are listed with the ORPHA number to be used preceded by the sign “→”. A table in annex lists the name of the rare disease and its ORPHA number to be used instead of the deprecated entries.

Obsolete entries are not listed here. In the case of duplicates, the nomenclature of the obsolete entry has been added to the rare disease listed here.

Rare diseases listed in alphabetical order

ORPHA number	Disease name
289157	1-alpha-hydroxylase deficiency
431361	2,4-dienoyl-CoA reductase deficiency
976	2,8-dihydroxyadenine urolithiasis
79154	2-aminoacidic 2-oxoadipic aciduria
391417	2-methyl-3-hydroxybutyric aciduria
391428	2-methyl-3-hydroxybutyric aciduria, classic type
391428	2-methyl-3-hydroxybutyric aciduria, infantile type
391457	2-methyl-3-hydroxybutyric aciduria, neonatal type
391417	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
391428	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency, classic type
391428	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency, infantile type
391457	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency, neonatal type
79095	2-methylacyl-CoA racemase deficiency
79157	2-methylbutyric aciduria
79157	2-methylbutyryl-CoA dehydrogenase deficiency
255182	2-oxoglutarate complex deficiency
869	2A syndrome
2616	3-M syndrome
79301	3-beta-hydroxy-delta-5-C27-steroid oxidoreductase deficiency
20	3-hydroxy-3-methylglutaric aciduria
20	3-hydroxy-3-methylglutaryl-CoA lyase deficiency
35701	3-hydroxy-3-methylglutaryl-CoA synthase deficiency
939	3-hydroxyisobutyric aciduria
134	3-ketothiolase deficiency
1035	3-mercaptopyruvate sulfurtransferase deficiency
6	3-methylcrotonyl-CoA carboxylase deficiency
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
445038	3-methylglutaconic aciduria type 7

ORPHA number	Disease name
505208	3-methylglutaconic aciduria type 8
505216	3-methylglutaconic aciduria type 9
352328	3-methylglutaconic aciduria with deafness-encephalopathy-Leigh-like syndrome
445038	3-methylglutaconic aciduria-cataract-neurologic involvement-neutropenia syndrome
505216	3-methylglutaconic aciduria-epilepsy-spasticity-severe intellectual disability syndrome
67046	3-methylglutaconyl-CoA hydratase deficiency
134	3-oxothiolase deficiency
79351	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form
2671	3-phosphoglycerate dehydrogenase deficiency, neonatal form
79350	3-phosphoserine phosphatase deficiency
869	3A syndrome
7	3C syndrome
2616	3M syndrome
293843	3MC syndrome
→293843	3MC1 syndrome
→293843	3MC2 syndrome
→293843	3MC3 syndrome
67046	3MG-CoA hydratase deficiency
2118	4-HPPD deficiency
2118	4-alpha-hydroxyphenylpyruvate hydroxylase deficiency
22	4-hydroxybutyric aciduria
2118	4-hydroxyphenylpyruvic acid dioxygenase deficiency
869	4A syndrome
88637	4H syndrome
250977	5-amino-4-imidazole carboxamide ribosiduria
217064	5-fluorouracil intoxication
217064	5-fluorouracil poisoning
33572	5-oxoprolinase deficiency
99135	6-phosphogluconate dehydrogenase deficiency
13	6-pyruvoyl-tetrahydropterin synthase deficiency
818	7-dehydrocholesterol reductase deficiency
168588	11-beta-hydroxysteroid dehydrogenase deficiency type 1
320	11-beta-hydroxysteroid dehydrogenase deficiency type 2
752	17-beta-hydroxysteroid dehydrogenase 3 deficiency
752	17-ketoreductase deficiency
752	17-ketosteroidreductase deficiency
99763	18-hydroxylase deficiency
99763	18-oxidase deficiency
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 syndrome
2975	46,XX disorder of sex development-skeletal anomalies syndrome
243	46,XX gonadal dysgenesis
243	46,XX ovarian dysgenesis
444048	46,XX ovarian dysgenesis-short stature syndrome
2138	46,XX ovotesticular DSD
2138	46,XX ovotesticular disorder of sex development
243	46,XX pure gonadal dysgenesis
393	46,XX testicular DSD
393	46,XX testicular disorder of sex development
199310	46,XX/46,XY chimerism
242	46,XY CGD
753	46,XY DSD due to 5-alpha-reductase 2 deficiency
755	46,XY DSD due to LH resistance or LHB deficiency
325448	46,XY DSD due to LHB 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 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
251510	46,XY PGD
242	46,XY complete gonadal dysgenesis

→ This disease is deprecated and has been moved to another (see annex). The indicated Orphanumber should now be used.

ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
96266	46,XY disorder of sex development due to partial LH receptor inactivation	9	48,XXXX syndrome	228415	5q35 microduplication syndrome
96266	46,XY disorder of sex development due to partial LH resistance	96263	48,XXXY syndrome	96125	6p subtelomeric deletion syndrome
96266	46,XY disorder of sex development due to partial luteinizing hormone resistance	10	48,XXYY syndrome	251046	6p22 microdeletion syndrome
752	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	99329	48,XYYY syndrome	96125	6p25 microdeletion syndrome
753	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency	11	49,XXXXX syndrome	75857	6q terminal deletion syndrome
755	46,XY disorder of sex development due to LH resistance or LHB deficiency	96264	49,XXXY syndrome	171829	6q16 deletion syndrome
325448	46,XY disorder of sex development due to LHB deficiency	261534	49,XXXYY syndrome	251056	6q25 microdeletion syndrome
96265	46,XY disorder of sex development due to complete LH receptor inactivation	99330	49,XYYYY syndrome	314034	7p22.1 microduplication syndrome
96265	46,XY disorder of sex development due to complete LH resistance	293948	1p21.3 microdeletion syndrome	96121	7q11.23 microduplication syndrome
96265	46,XY disorder of sex development due to complete luteinizing hormone receptor inactivation	401986	1p31p32 microdeletion syndrome	251061	7q31 microdeletion syndrome
96265	46,XY disorder of sex development due to complete luteinizing hormone resistance	456298	1p35.2 microdeletion syndrome	459074	7q36.3 microduplication syndrome
90796	46,XY disorder of sex development due to isolated 17,20-lyase deficiency	1606	1p36 deletion syndrome	96092	8p inverted duplication/deletion syndrome
755	46,XY disorder of sex development due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency	250989	1q21.1 microdeletion syndrome	168953	8p11 myeloproliferative syndrome
325448	46,XY disorder of sex development due to luteinizing hormone subunit beta deficiency	250994	1q21.1 microduplication syndrome	251066	8p11.2 deletion syndrome
443087	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	250999	1q41-q42 microdeletion syndrome	251076	8p23.1 duplication syndrome
168558	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	238769	1q44 microdeletion syndrome	251071	8p23.1 microdeletion syndrome
168563	46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome	363680	2p13.2 microdeletion syndrome	228399	8q12 microduplication syndrome
325345	46,XY ovotesticular DSD	261349	2p15-p16.1 microdeletion syndrome	2496	8q13 microdeletion syndrome
325345	46,XY ovotesticular disorder of sex development	261349	2p15p16.1 microdeletion syndrome	284160	8q21.11 microdeletion syndrome
251510	46,XY partial gonadal dysgenesis	163693	2p21 deletion syndrome	178303	8q22.1 microdeletion syndrome
251510	46,XY partial testicular dysgenesis	163693	2p21 microdeletion syndrome	508488	8q24.3 deletion syndrome
242	46,XY pure gonadal dysgenesis	369881	2p21 microdeletion syndrome without cystinuria	261112	9p deletion syndrome
3375	47,XXX syndrome	228402	2q23.1 microdeletion syndrome	261112	9p- syndrome
8	47,XYY syndrome	313947	2q23.1 microduplication syndrome	324313	9p13 microdeletion syndrome
		1617	2q24 microdeletion syndrome	96147	9q subtelomeric deletion syndrome
		251014	2q31.1 microdeletion syndrome	96147	9qSTDS
		294026	2q31.1 microduplication syndrome	352665	9q21 microdeletion syndrome
		251019	2q32-q33 microdeletion syndrome	401923	9q31.1q31.3 microdeletion syndrome
		251019	2q32q33 microdeletion syndrome	495818	9q33.3-q34.11 microdeletion syndrome
		251028	2q33.1 microdeletion syndrome	495818	9q33.3q34.11 microdeletion syndrome
		1001	2q37 microdeletion syndrome	284169	10p12p11 microdeletion syndrome
		1620	3p- syndrome	276413	10q22.3q23.3 microdeletion syndrome
		435638	3p25.3 microdeletion syndrome	276422	10q22.3q23.3 microduplication syndrome
		65286	3q subtelomere deletion syndrome	1307	10q24 microduplication syndrome
		65286	3qter deletion	52022	11p11.2 deletion
		1621	3q13 microdeletion syndrome	300305	11p15.4 microduplication syndrome
		96095	3q26 microduplication syndrome	444002	11q22.2-q22.3 deletion syndrome
		356947	3q26-q27microdeletion syndrome	444002	11q22.2q22.3 microdeletion syndrome
		356947	3q26q27 microdeletion syndrome	313884	12p12.1 microdeletion syndrome
		397695	3q27.3 microdeletion syndrome	280325	12p13.33 microdeletion syndrome
		65286	3q29 microdeletion syndrome	94063	12q14 microdeletion syndrome
		251038	3q29 microduplication syndrome	289513	12q15q21.1 microdeletion syndrome
		280	4p- syndrome	412035	13q12.3 microdeletion syndrome
		96072	4p16.3 microduplication syndrome	1590	13q32 deletion
		238750	4q21 microdeletion syndrome		
		502437	4q25 proximal deletion syndrome		
		329802	5p13 microduplication syndrome		
		86841	5q- syndrome		
		228384	5q14.3 microdeletion syndrome		
		436003	5q23 microdeletion syndrome		
		314655	5q31.3 microdeletion syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
261120	14q11.2 microdeletion syndrome		syndrome	916	Aase-Smith syndrome
261229	14q11.2 microduplication syndrome	261279	17q23.1-q23.2 microdeletion syndrome	69663	ABCB4 gene mutation-associated cholelithiasis
261144	14q12 microdeletion syndrome	261279	17q23.1q23.2 microdeletion syndrome	→897	ABCD syndrome
→3157	14q22 microdeletion syndrome	1598	18p- syndrome	2970	Abdominal muscle deficiency syndrome
264200	14q22-q23 microdeletion syndrome	1600	18q deletion syndrome	800	Aberfeld syndrome
264200	14q22q23 microdeletion syndrome	1600	18q- syndrome	85446	ABeta2Mwt amyloidosis
401935	14q24.1q24.3 microdeletion syndrome	254346	19p13.12 microdeletion syndrome	324723	ABeta amyloidosis, Arctic type
488280	14q32 duplication syndrome	357001	19p13.13 microdeletion syndrome	100006	ABeta amyloidosis, Dutch type
314585	15q overgrowth syndrome	447980	19p13.3 microduplication syndrome	324718	ABeta amyloidosis, Flemish type
238446	15q11-q13 duplication syndrome	217346	19q13.11 microdeletion syndrome	324708	ABeta amyloidosis, Iowa type
238446	15q11-q13 microduplication syndrome	313781	20p subtelomeric deletion syndrome	324713	ABeta amyloidosis, Italian type
261183	15q11.2 BP1-BP2 microdeletion syndrome	261295	20p12.3 microdeletion syndrome	324703	ABeta amyloidosis, Piedmont type
261183	15q11.2 microdeletion syndrome	313781	20p13 microdeletion syndrome	324718	ABetaA21G amyloidosis
238446	15q11q13 duplication syndrome	444051	20q11.2 microdeletion syndrome	324718	ABetaA21G-related amyloidosis
238446	15q11q13 microduplication syndrome	363659	20q11.2 microduplication syndrome	324708	ABetaD23N amyloidosis
199318	15q13.3 microdeletion syndrome	261311	20q13.33 microdeletion syndrome	324723	ABetaE22G amyloidosis
261190	15q14 microdeletion syndrome	574	21q deletion syndrome	324713	ABetaE22K amyloidosis
94065	15q24 microdeletion syndrome	574	21q- syndrome	100006	ABetaE22Q amyloidosis
1596	15q26 deletion syndrome	261323	21q22.11-q22.12 microdeletion syndrome	324703	ABetaL34V amyloidosis
363992	15q26.3 microdeletion syndrome	261323	21q22.11q22.12 microdeletion syndrome	324703	ABetaL34V-related amyloidosis
261211	16p11.2-p12.2 microdeletion syndrome	268261	21q22.13-q22.2 microdeletion syndrome	14	Abetalipoproteinemia
261211	16p11.2p12.2 microdeletion syndrome	268261	21q22.13q22.2 microdeletion syndrome	920	Ablepharon macrostomia syndrome
261204	16p11.2p12.2 microduplication syndrome	567	22q11DS	99089	Abnormal number of coronary ostia
485405	16p12.1p12.3 triplication syndrome	567	22q11.2 deletion syndrome	99050	Abnormal origin of right or left pulmonary artery from the aorta
261236	16p13.11 microdeletion syndrome	1727	22q11.2 microduplication syndrome	1164	ABPA
261243	16p13.11 microduplication syndrome	48652	22q13 deletion	97345	ABri amyloidosis
500055	16p13.2 microdeletion syndrome	85445	AA amyloidosis	921	Abruzzo-Erickson syndrome
96078	16p13.3 microduplication syndrome	869	AAA syndrome	69739	ABSD
352629	16q24.1 microdeletion syndrome	35708	AADC deficiency	2310	Absence deformity of leg-cataract syndrome
261250	16q24.3 microdeletion syndrome	91385	AAE	99112	Absence of brachiocephalic vein
819	17p11.2 microdeletion syndrome	100055	AAE 2	1658	Absence of dermatoglyphics-congenital milia syndrome
1713	17p11.2 microduplication syndrome	100055	AAE II	1658	Absence of fingerprints-congenital milia syndrome
477817	17p11.2p12 microduplication syndrome	1414	Aagenaes syndrome	99112	Absence of innominate vein
217385	17p13.3 duplication syndrome	284460	AAOR	101206	Absence of pulmonary valve-Fallot tetralogy-absence of ductus arteriosus syndrome
217385	17p13.3 microduplication syndrome	93560	AApoAI amyloidosis	99048	Absence of pulmonary valve-ventricular septal defect-persistent ductus arteriosus syndrome
97685	17q11 microdeletion syndrome	238269	AApoAll amyloidosis	980	Absence of the pulmonary artery
139474	17q11.2 microduplication syndrome	439232	AApoAIV amyloidosis	99114	Absence of the superior caval vein
261265	17q12 microdeletion syndrome	915	Aarskog syndrome	99114	Absence of the superior vena cava
261272	17q12 microduplication syndrome	1974	Aarskog-like syndrome	2985	Absent eyebrows and eyelashes-intellectual disability syndrome
363958	17q21.31 microdeletion syndrome	3163	Aarskog-Ose-Pande syndrome	85201	Absent patellae-scrotal hypoplasia-renal anomalies-facial dysmorphism-intellectual disability syndrome
217340	17q21.31 microduplication	915	Aarskog-Scott syndrome		
		124	Aase syndrome		
		916	Aase-Smith I syndrome		
		124	Aase-Smith II syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
3016	Absent radius-anogenital anomalies syndrome	93296	Achondrogenesis, Langer-Saldino type	231401	Acquired HbH disease
2951	Absent thumb-short stature-immunodeficiency syndrome	93298	Achondrogenesis, Parenti-Fraccaro type	231401	Acquired hemoglobin H disease
988	Absent tibia-polydactyly syndrome	15	Achondroplasia	158057	Acquired hemophagocytic lymphohistiocytosis associated with malignant disease
3328	Absent tibia-polydactyly-arachnoid cyst syndrome	935	Achondroplasia-SCID syndrome	73274	Acquired hemophilia
225147	ABSN	935	Achondroplasia-severe combined immunodeficiency syndrome	2221	Acquired hypertrichosis lanuginosa
99901	ACAD9 deficiency	935	Achondroplasia-Swiss type agammaglobulinemia syndrome	26348	Acquired hypoprothrombinemia
42	ACADM deficiency	49382	Achromatopsia	454	Acquired ichthyosis
26792	ACADS deficiency	355	Acid beta-glucuronidase deficiency	75564	Acquired idiopathic sideroblastic anemia
945	Acalvaria	333	Acid ceramidase deficiency	37559	Acquired kinky hair syndrome
67043	Acanthamoeba keratitis	424046	Acinar cell carcinoma of pancreas	79086	Acquired lipoatrophic diabetes
79468	Acanthokeratolytic verrucous nevus	40366	Acitretin/etretinate embryopathy	464453	Acquired methemoglobinemia
300504	Acanthoma of the nail matrix	79099	Ackerman dermatitis syndrome	91136	Acquired monoclonal Ig light chain-associated Fanconi syndrome
90301	Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome	2561	Ackerman fused molar rooth syndrome	91136	Acquired monoclonal immunoglobulin light chain-associated Fanconi syndrome
926	Acatalasemia	79099	Ackerman syndrome	589	Acquired myasthenia
2508	ACC-abnormal genitalia syndrome	51890	ACNES	95626	Acquired neurogenic diabetes insipidus
561	Accelerated skeletal maturation-facial dysmorphism-failure to thrive syndrome	43115	Aconitase deficiency	84142	Acquired neuromyotonia
180182	Accessory breasts	252175	Acoustic neurilemoma	91385	Acquired non histamine-induced angioedema
99061	Accessory mitral valve tissue	252175	Acoustic neuroma	79087	Acquired partial lipodystrophy
141096	Accessory nostril	65759	ACPS2	314697	Acquired porencephaly
674	Accessory pancreas	65798	ACPS4	729	Acquired primary erythrocytosis
95462	Accessory tricuspid valve tissue	306431	Acquired adult-onset immunodeficiency	26348	Acquired prothrombin deficiency
1005	ACD-intellectual disability syndrome	90065	Acquired aneurysmal subarachnoid hemorrhage	228247	Acquired pseudoxanthoma elasticum
210122	ACDMPV	91385	Acquired angioedema	49566	Acquired purpura fulminans
502444	ACER3-related early childhood-onset progressive leukodystrophy	100056	Acquired angioedema type 1	228247	Acquired PXE
48818	Aceruloplasminemia	100055	Acquired angioedema type 2	206575	Acquired rippling muscle disease
464458	Acetaminophen poisoning	91385	Acquired angioneurotic edema	485275	Acquired schizencephaly
99736	Acetazolamide-responsive congenital myotonia	100056	Acquired angioneurotic edema type 1	93585	Acquired thrombotic thrombocytopenic purpura
99736	Acetazolamide-responsive myotonia	100055	Acquired angioneurotic edema type 2	93585	Acquired TTP
2008	ACFS	91385	Acquired bradykinine-induced angioedema	99147	Acquired von Willebrand disease
930	Achalasia cardia	91385	Acquired C1 inhibitor deficiency	99147	Acquired von Willebrand syndrome
869	Achalasia-addisonianism-alacrima syndrome	95626	Acquired CDI	263534	Acral deciduous skin
→869	Achalasia-alacrimia syndrome	95626	Acquired central diabetes insipidus	97360	Acral dysostosis with facial and genital abnormalities
929	Achalasia-microcephaly syndrome	454700	Acquired Creutzfeldt-Jakob disease	158673	Acral dystrophic epidermolysis bullosa
294983	Acheiria	228285	Acquired cutis laxa	263534	Acral peeling skin syndrome
931	Acheiropodia	404514	Acquired cystic disease-associated renal cell carcinoma	90396	Acral persistent papular mucinosis
931	Acheiropody	46487	Acquired epidermolysis bullosa	263534	Acral PSS
49382	ACHM	98818	Acquired epileptic aphasia	281127	Acral self-healing collodion baby
932	Achondrogenesis	91136	Acquired Fanconi syndrome secondary to monoclonal gammopathy	281127	Acral SHCB
93299	Achondrogenesis type 1A	79086	Acquired generalized lipodystrophy	978	Acro-dermato-ungual-lacrimal-tooth syndrome
93298	Achondrogenesis type 1B	228247	Acquired Gronblad-Strandberg-Touraine syndrome	958	Acro-renal-mandibular syndrome
93296	Achondrogenesis type 2			959	Acro-renal-ocular syndrome
93299	Achondrogenesis, Houston-Harris type			36	Acrocallosal syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
63446	Acrocapitofemoral dysplasia	1827	Acromelic frontonasal dysplasia		repetitive partial seizures
2008	Acrocardiofacial syndrome	968	Acromesomelic dwarfism	363549	Acute encephalopathy with biphasic seizures and late reduced diffusion
221054	Acrocephalopolydactylous dysplasia	2098	Acromesomelic dysplasia, Grebe type	279888	Acute endophthalmitis
221054	Acrocephalopolydactyly	968	Acromesomelic dysplasia, Hunter-Thompson type	318	Acute erythroid leukemia
65759	Acrocephalopolysyndactyly type 2	40	Acromesomelic dysplasia, Maroteaux type	243367	Acute fatty liver of pregnancy
65798	Acrocephalopolysyndactyly type 4	2500	Acrometageria	3243	Acute febrile neutrophilic dermatosis
87	Acrocephalosyndactyly type 1	969	Acromicric dysplasia	293173	Acute generalized exanthematous pustulosis
794	Acrocephalosyndactyly type 3	955	Acroosteolysis dominant type	99920	Acute graft versus host disease
710	Acrocephalosyndactyly type 5	955	Acroosteolysis with osteoporosis and changes in skull and mandible	90062	Acute hepatic failure
63440	Acrocephaly	363665	Acroosteolysis-keloid-like lesions-premature aging syndrome	98916	Acute idiopathic demyelinating polyneuropathy
949	Acrocraniofacial dysostosis	2980	Acrootoocular syndrome	363549	Acute infantile encephalopathy predominantly affecting the frontal lobes
955	Acrodentoosteodysplasia	85203	Acropectoral syndrome	217371	Acute infantile liver failure due to synthesis defect of mitochondrial DNA-encoded proteins
163931	Acrodermatitis continua of Hallopeau	956	Acropectororenal dysplasia	217371	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins
37	Acrodermatitis enteropathica	957	Acropectorovertebral dysplasia	466794	Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome
37	Acrodermatitis enteropathica, zinc deficiency type	41	Acropigmentation of Dohi	370088	Acute infantile liver failure-multisystemic involvement syndrome
978	Acrodermatounguallacrimaltooth syndrome	1133	Acrorenal defect-ectodermal dysplasia-diabetes syndrome	98916	Acute inflammatory demyelinating polyradiculoneuropathy
950	Acrodysostosis	971	Acrorenal syndrome	98916	Acute inflammatory polyneuropathy
280651	Acrodysostosis with multiple hormone resistance	85203	ACRP syndrome	79276	Acute intermittent porphyria
950	Acrodysplasia	36	ACS	79126	Acute interstitial pneumonia
2956	Acrodysplasia scoliosis	87	ACS1	79126	Acute interstitial pneumonitis
1786	Acrofacial dysostosis, Catania type	794	ACS3	73423	Acute intoxication by Blighia sapida
246	Acrofacial dysostosis, Genee-Wiedmann type	710	ACS5	90062	Acute liver failure
64542	Acrofacial dysostosis, Kennedy-Teebi type	98904	Actin myopathy	178320	Acute lung injury
1787	Acrofacial dysostosis, Palagonia type	254395	Actinic lichen planus	488239	Acute macular neuroretinopathy
1788	Acrofacial dysostosis, Rodríguez type	254395	Actinic LP	518	Acute megakaryoblastic leukemia
952	Acrofacial dysostosis, Weyers type	330061	Actinic prurigo	99887	Acute megakaryoblastic leukemia in Down syndrome
1784	Acrofrontofacionasal dysostosis	330064	Actinic reticuloid	329469	Acute megakaryoblastic leukemia without Down syndrome
2211	Acrofrontofacionasal dysostosis type 2	457095	Actinomycosis	518	Acute megakaryocytic leukemia
2211	Acrofrontofacionasal syndrome type 2	163696	Action myoclonus-renal failure syndrome	514	Acute monoblastic leukemia
2500	Acrogeria	397596	Activated PI3K-delta syndrome	514	Acute monocytic leukemia
2500	Acrogeria, Gottron type	101089	Activation-induced cytidine deaminase deficiency	98917	Acute motor and sensory axonal neuropathy
38	Acrokeratoelastoidosis of Costa	73423	Acute ackee fruit intoxication	98918	Acute motor axonal neuropathy
166113	Acrokeratosis of Bazex	95409	Acute adrenal failure	98917	Acute motor-sensory axonal GBS
166113	Acrokeratosis paraneoplastica	95409	Acute adrenal insufficiency	98917	Acute motor-sensory axonal Guillain-Barré syndrome
166113	Acrokeratosis paraneoplastica of Bazex	95409	Acute adrenocortical insufficiency		
79151	Acrokeratosis verruciformis of Hopf	99870	Acute and disseminated Langerhans cell histiocytosis		
965	Acromegaloid facial appearance syndrome	284460	Acute annular outer retinopathy		
963	Acromegaly	86849	Acute basophilic leukemia		
→2796	Acromegaly-cutis verticis gyrata-corneal leukoma syndrome	225147	Acute bilateral striatal necrosis		
39	Acromelanosis	98837	Acute biphenotypic leukemia		
1827	Acromelic frontonasal dysostosis	2901	Acute brachial plexus neuritis		
		83597	Acute disseminated encephalitis		
		83597	Acute disseminated encephalomyelitis		
		163703	Acute encephalitis with refractory		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
228157	Acute multiple sclerosis, Marburg type	517	Acute myelomonocytic leukemia	100008	ACys amyloidosis
228157	Acute multiple sclerosis, Marburg variant	86843	Acute myelosclerosis	99736	ACZ-responsive congenital myotonia
520	Acute myeloblastic leukemia 3	263524	Acute necrotizing encephalopathy of childhood	99736	ACZ-responsive myotonia
98833	Acute myeloblastic leukemia M1	247546	Acute neonatal citrullinemia type 1	93608	AD dRTA
98834	Acute myeloblastic leukemia M2	247546	Acute neonatal citrullinemia type I	428	AD hypocalcemia
98834	Acute myeloblastic leukemia with maturation	77260	Acute neuronopathic Gaucher disease	314889	AD pRTA
98833	Acute myeloblastic leukemia without maturation	163703	Acute non-herpetic encephalitis with severe refractory status epilepticus	169189	AD-CNM
86843	Acute myelodysplasia with myelofibrosis	35889	Acute opioid poisoning	1810	AD-HED
86843	Acute myelofibrosis	231457	Acute panautonomic GBS	2314	AD-HIES
86845	Acute myeloid leukaemia with myelodysplasia-related features	231457	Acute panautonomic Guillain-Barré syndrome	447753	AD-SPG9A
102379	Acute myeloid leukemia and myelodysplastic syndromes related to alkylating agent	231457	Acute panautonomic neuropathy	447757	AD-SPG9B
164726	Acute myeloid leukemia and myelodysplastic syndromes related to radiation	231457	Acute pandysautonomia	277	ADA deficiency
102381	Acute myeloid leukemia and myelodysplastic syndromes related to topoisomerase type 2 inhibitor	86843	Acute panmyelosis with myelofibrosis	973	Adactyly of hand, unilateral
318	Acute myeloid leukemia M6	90064	Acute peripheral arterial occlusion	216796	Adair-Dighton syndrome
518	Acute myeloid leukemia M7	43119	Acute poisoning by drugs with membrane-stabilizing effect	55881	Adamantinoma
98831	Acute myeloid leukemia with 11q23 abnormalities	520	Acute promyelocytic leukemia	55881	Adamantinoma of long bones
98829	Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)	98918	Acute pure motor GBS	974	Adams-Oliver syndrome
319480	Acute myeloid leukemia with CEBPA somatic mutations	98918	Acute pure motor Guillain-Barré syndrome	97346	ADan amyloidosis
402020	Acute myeloid leukemia with inv3(q21;q26.2) or t(3;3)(q21;q26.2)	231450	Acute pure sensory GBS	88619	ADANE
98832	Acute myeloid leukemia with minimal differentiation	231450	Acute pure sensory Guillain-Barré syndrome	314404	ADCA-DN syndrome
86845	Acute myeloid leukemia with multilineage dysplasia	231450	Acute pure sensory neuropathy	90348	ADCL
402026	Acute myeloid leukemia with NPM1 somatic mutations	454831	Acute radiation sickness	86814	ADCME
402014	Acute myeloid leukemia with t(6;9)(p23;q34)	454831	Acute radiation syndrome	85138	Addison disease
370026	Acute myeloid leukemia with t(8;16)(p11;p13) translocation	140896	Acute respiratory coronavirus infection	95409	Addisonian crisis
102724	Acute myeloid leukemia with t(8;21)(q22;q22) translocation	3099	Acute rheumatic fever	2953	Adducted thumb-clubfoot syndrome
402017	Acute myeloid leukemia with t(9;11)(p22;q23)	90059	Acute sensorineural hearing loss by acute acoustic trauma or sudden deafness or surgery induced acoustic trauma	2952	Adducted thumbs-arthrogryposis syndrome, Christian type
520	Acute myeloid leukemia with t(15;17)(q22;q12);(PML/RARalpha) and variants	231466	Acute sensory ataxic GBS	101046	ADEAF
98835	Acute myeloid leukemia, minimal differentiation, FAB M0	231466	Acute sensory ataxic Guillain-Barré syndrome	83597	ADEM
		231466	Acute sensory ataxic neuropathy	976	Adenine phosphoribosyltransferase deficiency
		139417	Acute transverse myelitis	213504	Adenocarcinoma of ovary
		43117	Acute tricyclic antidepressant poisoning	424016	Adenocarcinoma of the anal canal
		91500	Acute tubulointerstitial nephritis and uveitis syndrome	213772	Adenocarcinoma of the cervix uteri
		98835	Acute undifferentiated leukemia	99976	Adenocarcinoma of the esophagus
		284454	Acute zonal occult outer retinopathy	424991	Adenocarcinoma of the gallbladder and EBT
		137754	ACY1D	424991	Adenocarcinoma of the gallbladder and extrahepatic biliary tract
		141	ACY2 deficiency	424943	Adenocarcinoma of the liver and IBT
		99901	Acyl-CoA dehydrogenase 9 deficiency	424943	Adenocarcinoma of the liver and intrahepatic biliary tract
				363478	Adenocarcinoma of the paratestis
				398053	Adenocarcinoma of the penis
				104075	Adenocarcinoma of the small bowel
				104075	Adenocarcinoma of the small intestine
				494454	Adenocarcinoma of the vulva
				95512	Adenohypophysitis
				213828	Adenoid basal carcinoma of the cervix uteri

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
213823	Adenoid cystic carcinoma of the cervix uteri	88949	ADTKD-MUC1	247585	Adult-onset citrin deficiency
213741	Adenoid cystic carcinoma of the corpus uteri	217330	ADTKD-REN	247573	Adult-onset citrullinemia type 1
93292	Adenoma of the pancreas	88950	ADTKD-UMOD	247585	Adult-onset citrullinemia type 2
26790	Adenomucinosis	70578	Adult acute respiratory distress syndrome	247573	Adult-onset citrullinemia type I
213792	Adenosarcoma of the cervix uteri	70578	Adult ARDS	247585	Adult-onset citrullinemia type II
213600	Adenosarcoma of the corpus uteri	93605	Adult Bartter syndrome	329336	Adult-onset CPEO with mitochondrial myopathy
45	Adenosine monophosphate deaminase deficiency	157846	Adult basal ganglia disease	411641	Adult-onset cystinosis
28	Adenosylcobalamin deficiency	874	Adult cardiac tumor	329478	Adult-onset distal myopathy due to VCP mutation
91127	Adenovirus infection in immunocompromised patients	221	Adult dermatomyositis	199351	Adult-onset dystonia-parkinsonism
46	Adenylosuccinate deficiency	2666	Adult familial nephronophthisis-spastic quadriparegia syndrome	99000	Adult-onset foveomacular dystrophy
46	Adenylosuccinate lyase deficiency	309169	Adult GM2 gangliosidosis 0 variant	99000	Adult-onset foveomacular dystrophy with choroidal neovascularization
482601	Adenylosuccinate synthetase-like 1-related distal myopathy	210159	Adult hepatocellular carcinoma	99000	Adult-onset foveomacular vitelliform dystrophy
137817	Adhesive arachnoiditis	247676	Adult hypophosphatasia	79257	Adult-onset GM1 gangliosidosis
89937	ADHR	2688	Adult idiopathic neutropenia	306431	Adult-onset immunodeficiency with anti-interferon-gamma autoantibodies
454718	Adie syndrome	178487	Adult intestinal botulism	313808	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia
36397	Adiposalgia	178487	Adult intestinal colonization botulism	329314	Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency
36397	Adipose tissue rheumatism	178487	Adult intestinal toxemia botulism	329314	Adult-onset multiple mtDNA deletion syndrome due to DGUOK deficiency
36397	Adiposis dolorosa	206448	Adult Krabbe disease	391490	Adult-onset myasthenia gravis
289290	ADK hypermethioninemia	79262	Adult NCL	171442	Adult-onset nemaline myopathy
99027	ADLD	79262	Adult neuronal ceroid lipofuscinosis	276608	Adult-onset non-insulinoma persistent hyperinsulinemic hypoglycemia
101046	ADLTE	247676	Adult phosphoethanolaminuria	206572	Adult-onset overlap myositis
178464	ADMERF	206583	Adult polyglucosan body disease	35689	Adult-onset PLS
98784	ADNFLE	902	Adult progeria	35689	Adult-onset primary lateral sclerosis
329211	ADNIV	99874	Adult pulmonary Langerhans cell histiocytosis	829	Adult-onset Still disease
404448	ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder	98872	Adult pure red cell aplasia	99000	Adult-onset vitelliform macular dystrophy
1544	Adolescent benign focal crisis	247676	Adult Rathburn disease	3086	ADVIRC
306588	ADOS	773	Adult Refsum disease	682	Adynamia episodica hereditaria
36355	ADP platelet receptor P2Y12 defect	978	ADULT syndrome	1071	AEC syndrome
2924	ADPCLD	86875	Adult T-cell leukemia/lymphoma	281139	AEI
101046	ADPEAF	391490	Adult-onset acquired myasthenia	163703	AERRPS
254892	adPEO	79280	Adult-onset Alpha-N-acetylgalactosaminidase deficiency	363549	AESD
730	ADPKD	391490	Adult-onset autoimmune myasthenia gravis	178345	AEXS
95409	Adrenal crisis	99027	Adult-onset autosomal dominant demyelinating leukodystrophy	37	AEZ
869	Adrenal insufficiency-achalasia-alacrima syndrome	99027	Adult-onset autosomal dominant leukodystrophy	220460	AFAP
1501	Adrenocortical carcinoma	284289	Adult-onset autosomal recessive cerebellar ataxia	313772	AFG3L2-related spastic ataxia-myoclonic epilepsy-neuropathy syndrome
231625	Adrenocortical carcinoma with pure aldosterone hypersecretion	255132	Adult-onset autosomal recessive sideroblastic anemia		
95409	Adrenocortical crisis	420492	Adult-onset cervical dystonia, DYT23 type		
99889	Adrenocorticotropic hormone secretion syndrome	329336	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy		
139399	Adrenomyeloneuropathy				
977	Adrenomyodystrophy				
228169	ADSD				
46	ADSL deficiency				
482601	ADSSL1-related distal myopathy				
34149	ADTKD				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
93562	AFib amyloidosis	250977	AICA-ribosiduria	141242	Alar cleft
243367	AFLP	50	Aicardi syndrome	141242	Alar rim cleft
1827	AFND	51	Aicardi-Goutières syndrome	319671	Alazami syndrome
398147	AFP	101089	AID deficiency	53	Albers-Schönberg osteopetrosis
139507	African iron overload	98916	AIDP	→897	Albinism-black lock-cell migration disorder of the neurocytes of the gut-sensorineural deafness syndrome
101334	African tick typhus	90081	AIDS wasting syndrome	998	Albinism-deafness syndrome
3385	African trypanosomiasis	178333	AIED	→457059	Albright hereditary osteodystrophy
33110	Agammaglobulinemia, non-Bruton type	363549	AIEF	1001	Albright hereditary osteodystrophy type 3
83617	Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	86886	AILT	1001	Albright hereditary osteodystrophy-like syndrome
388	Aganglionic megacolon	103919	AIP	79443	Albright hereditary osteodystrophy-PHP syndrome Ia
35704	AGAT deficiency	280302	AIP type 1	79445	Albright hereditary osteodystrophy-PPHP syndrome
353217	AGC1 deficiency	280315	AIP type 2	98841	ALCL
85448	AGel amyloidosis	75564	AISA	60039	Alcock syndrome
180142	Agenesis and aplasia of uterine body	33355	AK2 deficiency	1915	Alcohol-related birth defects
50	Agenesis of corpus callosum with chorioretinal abnormality	38	AKE	1915	Alcohol-related neurodevelopmental disorder
99114	Agenesis of the superior caval vein	→357225	Akesson syndrome	36899	Alcohol-responsive dystonia
99114	Agenesis of the superior vena cava	79085	AKT2-related familial partial lipodystrophy	43	ALD
99114	Agenesis of the SVC	79085	AKT2-related FPLD	324977	ALDD syndrome
293173	AGEP	79151	AKV of Hopf	35664	ALDH18A1-related De Barsy syndrome
873	Aggressive fibromatosis	85443	AL amyloidosis	99763	Aldosterone synthase deficiency
86873	Aggressive NK-cell leukemia	2232	AI Awadi-Farag-Teebi syndrome	99764	Aldosterone synthase deficiency unrelated to CYP11B2
86873	Aggressive NK-cell lymphoma	2879	AI Awadi-Raas-Rothschild syndrome	99764	Aldosterone synthase deficiency unrelated to the aldosterone synthase gene
98850	Aggressive systemic mastocytosis	→3157	AI Frayh-Facharzt-Haque syndrome	369929	Aldosterone-producing adenoma with seizures and neurological abnormalities
989	Aglossia-adactylia syndrome	2725	AI Gazali-Al Talabani syndrome	369929	Aldosterone-secreting adenoma with seizures and neurological abnormalities
990	Agnathia-holoprosencephaly-situs inversus syndrome	2865	AI Gazali-Aziz-Salem syndrome	85332	Aldred syndrome
824	Agnogenic myeloid metaplasia	2153	AI Gazali-Donnai-Muller syndrome	439224	ALECT2 amyloidosis
100070	Agrammatic variant of PPA	2725	AI Gazali-Lytte syndrome	158799	Aleukemic mast cell leukemia
100070	Agrammatic variant of primary progressive aphasia	2773	AI Gazali-Nair syndrome	58	Alexander disease
442582	AH amyloidosis	→324737	AI-Gazali-Dattani syndrome	363717	Alexander disease type I
2131	AHC	404454	Alacrimia-choreoarthetosis-liver dysfunction syndrome	363722	Alexander disease type II
412069	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	100924	ALAD porphyria	261112	Alfi syndrome
59	AHDS	52	Alagille syndrome	79327	ALG1-CDG
50812	Ahn-Lerman-Sagie syndrome	261600	Alagille syndrome due to 20p12 microdeletion	79326	ALG2-CDG
79443	AHO-PHP syndrome Ia	261619	Alagille syndrome due to a JAG1 point mutation	79321	ALG3-CDG
79445	AHO-PPHP syndrome	261629	Alagille syndrome due to a NOTCH2 point mutation	79320	ALG6-CDG
2134	aHUS	261600	Alagille syndrome due to monosomy 20p12	79325	ALG8-CDG
93581	aHUS with anti-factor H antibodies	52	Alagille-Watson syndrome	79328	ALG9-CDG
93578	aHUS with B factor anomaly	261619	Alagille-Watson syndrome due to a JAG1 point mutation	280071	ALG11-CDG
93575	aHUS with C3 anomaly	261629	Alagille-Watson syndrome due to a NOTCH2 point mutation	79324	ALG12-CDG
357008	aHUS with DGKE deficiency	261600	Alagille-Watson syndrome due to monosomy 20p12		
93579	aHUS with H factor anomaly	178333	Åland Islands eye disease		
93580	aHUS with I factor anomaly	2007	Alar cartilages hypoplasia-coloboma-telecanthus syndrome		
93576	aHUS with MCP/CD46 anomaly				
217023	aHUS with thrombomodulin anomaly				

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324422	ALG13-CDG	734	Alpha dense granule deficiency		macrothrombocytopenia
99995	Algodystrophy	134	Alpha methylacetooacetic aciduria	→182050	Alport syndrome with macrothrombocytopenia
300895	ALK+ ALCL	721	Alpha storage pool deficiency		Alport syndrome-intellectual disability syndrome-midface hypoplasia-elliptocytosis syndrome
300895	ALK+ anaplastic large cell lymphoma	98791	Alpha thalassemia-intellectual disability syndrome, deletion type	86818	ALPS
364043	ALK+ large B-cell lymphoma	98791	Alpha thalassemia-mental retardation syndrome	3261	ALPS due to CTLA4 haploinsufficiency
364043	ALK+ LBCL	365	Alpha-1,4-glucosidase acid deficiency	275517	ALPS with recurrent viral infections
300903	ALK- ALCL	308552	Alpha-1,4-glucosidase acid deficiency, infantile onset	803	ALS
300903	ALK- anaplastic large cell lymphoma	420429	Alpha-1,4-glucosidase acid deficiency, late-onset	357043	ALS4
300903	ALK-negative anaplastic large cell lymphoma	60	Alpha-1-antitrypsin deficiency	86815	ALSG
300895	ALK-positive anaplastic large cell lymphoma	79154	Alpha-aminoacidic aciduria	313808	ALSP
364043	ALK-positive large B-cell lymphoma	399058	Alpha-B crystallin-related late-onset distal myopathy	64	Alström syndrome
502444	Alkaline ceramidase 3 deficiency	324	Alpha-galactosidase A deficiency	99971	ALT
56	Alkaptonuria	100025	Alpha-HCD	2131	Alternating hemiplegia of childhood
59	Allan-Herndon-Dudley syndrome	100025	Alpha-heavy chain disease	210122	Alveolar capillary dysplasia with misalignment of pulmonary veins
1164	Allergic aspergillosis	31	Alpha-ketoglutarate dehydrogenase deficiency	210122	Alveolar capillary dysplasia with misalignment of pulmonary vessels
1164	Allergic bronchopulmonary aspergillosis	349	Alpha-L-fucosidase deficiency	199306	Alveolar cleft lip and palate
869	Allgrove syndrome	579	Alpha-L-iduronidase deficiency	284	Alveolar echinococcosis
69063	Alloimmune neonatal renal disease	61	Alpha-mannosidosis	99756	Alveolar rhabdomyosarcoma
93925	Alobar holoprosencephaly	309288	Alpha-mannosidosis, adult form	163699	Alveolar soft part sarcoma
1006	Alopecia antibody deficiency	309282	Alpha-mannosidosis, infantile form	163699	Alveolar soft tissue sarcoma
700	Alopecia totalis	134	Alpha-methyl-acetoacetyl-CoA thiolase deficiency	→1071	Alveolar synechia-ankyloblepharon-ectodermal dysplasia syndrome
701	Alopecia universalis	79095	Alpha-methyl-acyl-CoA racemase deficiency	306542	ALX1-related frontonasal dysplasia
2316	Alopecia-anosmia-conductive hearing loss-hypogonadism syndrome	3137	Alpha-N-acetylgalactosaminidase deficiency	391474	ALX3-related frontonasal dysplasia
2316	Alopecia-anosmia-deafness-hypogonadism syndrome	79279	Alpha-N-acetylgalactosaminidase deficiency type 1	228390	ALX4-related FNDAG
1005	Alopecia-contractures-dwarfism-intellectual disability syndrome	79280	Alpha-N-acetylgalactosaminidase deficiency type 2	169095	Alymphoid cystic thymic dysgenesis
202	Alopecia-deafness-hypogonadism syndrome	79281	Alpha-N-acetylgalactosaminidase deficiency type 3	93561	ALys amyloidosis
2574	Alopecia-epilepsy-intellectual disability syndrome, Moynahan type	62	Alpha-sarcoglycanopathy	79095	AMACR deficiency
1008	Alopecia-epilepsy-pyorrhea-intellectual disability syndrome	846	Alpha-thalassemia	98918	AMAN
→3464	Alopecia-hypogonadism-extrapyramidal syndrome	163596	Alpha-thalassemia hydrops fetalis	65	Amaurosis congenita of Leber
2850	Alopecia-intellectual disability syndrome	93616	Alpha-thalassemia intermedia	1021	Amaurosis-hypertrichosis syndrome
1014	Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome	163596	Alpha-thalassemia major	→95699	Ambiguous genitalia-disordered steroidogenesis Antley-Bixler-like syndrome
157954	Alopecia-progressive neurological defect-endocrinopathy syndrome	98791	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	1023	Ambras syndrome
202	Alopecia-sensorineural deafness-hypogonadism syndrome	231401	Alpha-thalassemia-myelodysplastic syndrome	294969	Amelia of lower limb
726	Alpers progressive sclerosing poliodystrophy	847	Alpha-thalassemia-X-linked intellectual disability syndrome	294967	Amelia of upper limb
726	Alpers syndrome	63	Alport deafness-nephropathy	314422	Ameloblastic carcinoma
726	Alpers-Huttenlocher syndrome	63	Alport syndrome	314419	Ameloblastoma
734	Alpha delta granule deficiency	→182050	Alport syndrome with leukocyte inclusions and	1946	Amelocerebrohypohidrotic syndrome
				88661	Amelogenesis imperfecta
				100031	Amelogenesis imperfecta type 1
				100033	Amelogenesis imperfecta type 2
				100032	Amelogenesis imperfecta type 3
				100034	Amelogenesis imperfecta type 4
				171836	Amelogenesis imperfecta-gingival hyperplasia syndrome
				1031	Amelogenesis imperfecta-

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	nephrocalcinosis syndrome	86818	AMME syndrome	71	Anderson disease
1028	Amelonychohypohidrotic syndrome	517	AMMoL	324	Anderson-Fabry disease
83595	American mountain fever	488239	AMNR	99916	Androblastoma
3386	American trypanosomiasis	251663	aMOA	329813	Androgenetic/biparental mosaicism
2116	Aminoaciduria, Hartnup type	67	Amoebiasis due to Entamoeba histolytica	157954	ANE syndrome
141	Aminoacylase 2 deficiency	68	Amoebiasis due to free-living amoebae	263524	ANEC
1908	Aminopterin embryopathy syndrome	45	AMP deaminase deficiency	1054	Aneurysm of sinus of Valsalva
221120	Aminopterin syndrome-like sine aminopterin	1035	Ampola syndrome	95484	Aneurysm or dilatation of ascending aorta
1908	Aminopterin/methotrexate embryofetopathy	66529	Ampulla cardiomyopathy	284984	Aneurysm-osteoarthritis syndrome
→33364	Amish brittle hair syndrome	300557	Ampullary carcinoma	480553	Aneurysmal bone cyst
171714	Amish infantile epilepsy syndrome	300557	Ampulloma	353344	Aneurysmal telangiectasia
99742	Amish lethal microcephaly	163696	AMRF	63442	Angel-shaped phalango-epiphyseal dysplasia
98902	Amish nemaline myopathy	98917	AMSAN	72	Angelman syndrome
518	AMKL	366	Amylo-1,6-glucosidase deficiency	411511	Angelman syndrome due to a point mutation
102379	AML and myelodysplastic syndromes related to alkylating agent	49804	Amyloid lichen	411515	Angelman syndrome due to imprinting defect in 15q11-q13
164726	AML and myelodysplastic syndromes related to radiation	319635	Amyloidosis cutis dyschromia	98794	Angelman syndrome due to maternal 15q11q13 deletion
102381	AML and myelodysplastic syndromes related to topoisomerase type 2 inhibitor	319635	Amyloidosis cutis dyschromica	98794	Angelman syndrome due to maternal monosomy 15q11q13
98832	AML M0	85450	Amyloidosis, Ostertag type	98795	Angelman syndrome due to paternal uniparental disomy of chromosome 15
98833	AML M1	367	Amylopectinosis	364039	Angiocentric cutaneous T-cell lymphoma of childhood
98834	AML M2	488586	Amyoplasia congenita	251671	Angiocentric glioma
520	AML M3	803	Amyotrophic lateral sclerosis	86879	Angiocentric T-cell lymphoma
517	AML M4	357043	Amyotrophic lateral sclerosis type 4	79093	Angiodysgenetic necrotizing myelopathy
514	AML M5	94091	Amyotrophic lateral sclerosis, hemiplegic type	98839	Angioendotheliomatosis proliferans systemisata
318	AML M6	90020	Amyotrophic lateral sclerosis-parkinsonism-dementia complex	160	Angiofollicular ganglionic hyperplasia
518	AML M7	90020	Amyotrophic lateral sclerosis-parkinsonism-dementia of Guam syndrome	160	Angiofollicular lymph hyperplasia
98831	AML with 11q23 abnormalities	2615	Amyotrophy-fat tissue anomaly syndrome	86886	Angioimmunoblastic T-cell lymphoma
98829	AML with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)	228113	Anal fistula	324	Angiokeratoma corporis diffusum
319480	AML with CEBPA somatic mutations	31150	Analphalipoproteinemia	95429	Angioma serpiginosum
402020	AML with inv3(q21;q26.2) or t(3;3)(q21;q26.2)	761	Anaphylactoid purpura	2346	Angioosteohypertrophic syndrome
86845	AML with multilineage dysplasia	251589	Anaplastic astrocytoma	75508	Angioosteohypotrophic syndrome
86845	AML with myelodysplasia-related features	251646	Anaplastic ependymoma	263413	Angiosarcoma
402026	AML with NPM1 somatic mutations	251957	Anaplastic ganglioglioma	74	Angiostrongylasis
402014	AML with t(6;9)(p23;q34)	98841	Anaplastic large cell lymphoma	98839	Angiotropic large cell lymphoma
370026	AML with t(8;16)(p11;p13) translocation	251663	Anaplastic oligoastrocytoma	370039	Angora hair nevus
102724	AML with t(8;21)(q22;q22) translocation	251630	Anaplastic oligodendrogloma	76	Anguilluliasis
402017	AML with t(9;11)(p22;q23)	142	Anaplastic thyroid carcinoma	76	Anguillulosis
520	AML with t(15;17)(q22;q12);(PML/RARalpha) and variants	251855	Anaplastic/large cell medulloblastoma	238468	Anhidrotic ectodermal dysplasia
86818	AMME complex	93347	Anauxetic dysplasia	98813	Anhidrotic ectodermal dysplasia with immunodeficiency
		79262	ANCL	69088	Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome
		78	Ancylostomiasis		
		1496	Andermann syndrome		
		37553	Andersen cardiодysrhythmic periodic paralysis		
		367	Andersen disease		
		37553	Andersen syndrome		
		37553	Andersen-Tawil syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
→1071	Anhidrotic ectodermic dysplasia-cleft lip/palate syndrome	2470	Anophthalmia-pulmonary hypoplasia syndrome		antibody-negative pauci-immune glomerulonephritis
1069	Aniridia-absent patella syndrome	1106	Anophthalmia-syndactyly syndrome	2821	Antinolo-Nieto-Borrego syndrome
1065	Aniridia-cerebellar ataxia-intellectual disability syndrome	77298	Anophthalmia/microphthalmia-esophageal atresia syndrome	3006	Antiquitin deficiency
1068	Aniridia-intellectual disability syndrome	1882	ANOTHER syndrome	81	Antisynthetase syndrome
1067	Aniridia-ptosis-intellectual disability-familial obesity syndrome	93976	Anotia	83	Antley-Bixler syndrome
1064	Aniridia-renal agenesis-psychomotor retardation syndrome	2987	Antecubital pterygium syndrome	→95699	Antley-Bixler syndrome type 2
1070	Anisakiasis	93604	Antenatal Bartter syndrome		Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis
86873	ANKCL	294	Antenatal CMV infection	→95699	Antley-Bixler syndrome, POR-related
1072	Ankyloblepharon filiforme adnatum-cleft palate syndrome	294	Antenatal cytomegalovirus infection	1190	AO1
1074	Ankyloblepharon filiforme adnatum-imperforate anus syndrome	70596	Antenatal EBV infection	56304	AO2
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	292	Antenatal enterovirus infection	56305	AO3
2206	Ankylosing vertebral hyperostosis with tylosis	70596	Antenatal Epstein-Barr virus infection	1168	AOA1
1077	Ankylosis of teeth	293	Antenatal herpes simplex virus infection	64753	AOA2
78	Ankylostomiasis	178148	Antenatal multiminicore disease with arthrogryposis multiplex congenita	459033	AOA4
254411	Annular atrophic lichen planus	291	Antenatal varicella virus infection	99000	AOFMD
254411	Annular atrophic LP	98956	Anterior basement membrane dystrophy	1190	AOI
281139	Annular epidermolytic ichthyosis	3344	Anterior bowing of legs with dwarfism	56304	AOII
254424	Annular lichen planus	51890	Anterior cutaneous nerve entrapment syndrome	56305	AOIII
254424	Annular LP	1931	Anterior encephalocele	1457	Aorta coarctation
675	Annular pancreas	98961	Anterior limiting membrane dystrophy type 1	60030	Aortic aneurysm syndrome due to TGF-beta receptors anomalies
229	Annuloaortic ectasia	98960	Anterior limiting membrane dystrophy type 2	1110	Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome
457205	ANOAC	98961	Anterior limiting membrane dystrophy type I	2299	Aortic arch interruption
99797	Anodontia	98960	Anterior limiting membrane dystrophy type II	→91387	Aortic dilatation-joint hypermobility-arterial tortuosity syndrome
101932	Anomaly of the mitral subvalvular apparatus	95512	Anterior pituitary hypopituitarism	95448	Aortic valve atresia
99055	Anomaly of the tricuspid valve chordae	435372	Anterior urethral valve	101043	Aortic valve dysplasia
94150	Anonychia congenita totalis	90079	Anthracycline extravasation	99071	Aorto-left ventricular tunnel
69125	Anonychia with flexural pigmentation	36412	Anti-C1q vasculitis	99070	Aorto-right ventricular tunnel
1094	Anonychia-microcephaly syndrome	375	Anti-GBM syndrome	3400	Aorto-ventricular tunnel
90390	Anonychia-onychodystrophy syndrome	375	Anti-glomerular basement membrane disease	99086	Aortopulmonary coronary arterial course
1487	Anonychia-onychodystrophy with hypoplasia or absence of distal phalanges syndrome	2194	Anti-HLA hyperimmunization	974	AOS
1104	Anophthalmia plus syndrome	206569	Anti-HMG-CoA myopathy	829	AOSD
→2470	Anophthalmia-heart and pulmonary anomalies-intellectual disability syndrome	420789	Anti-IgLON5 disease	280763	AP4 deficiency syndrome
→3157	Anophthalmia-hypothalamo-pituitary insufficiency syndrome	420789	Anti-IgLON5 syndrome	369929	APA with seizures and neurological abnormalities
1101	Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome	81	Anti-Jo1 syndrome	747	aPAP
		275944	Anti-K HDN	206583	APBD
		639	Anti-MAG neuropathy	247806	APC-related AFAP
		454710	Anti-p200 pemphigoid	247806	APC-related attenuated familial adenomatous polyposis
		206569	Anti-SRP myopathy	247806	APC-related attenuated familial polyposis coli
		97564	Antineutrophil cytoplasmic	247806	APC-related attenuated FAP
				397596	APDS
				3453	APECED syndrome
				87	Apert syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
162521	Apertura pyriformis with holoprosencephaly	3143	APS2	466775	ARCMT2X
1112	Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome	227982	APS3	1133	AREDYLD syndrome
1113	Aphalangy-syndactyly-microcephaly syndrome	227990	APS4	101096	Aregenerative anemia
49	Aphallia	3453	APS type 1	→702	Arena syndrome
324540	Aphonia-deafness-retinal dystrophy-bifid halluces-intellectual disability syndrome	3143	APS type 2	75377	Areolar atrophy of the macula
324540	Aphonia-deafness-retinal dystrophy-duplicated halluces-intellectual disability syndrome	227982	APS type 3	319223	Argentine hemorrhagic fever
324540	Aphonia-hearing loss-retinal dystrophy-duplicated halluces-intellectual disability syndrome	227990	APS type 4	319223	Argentinian hemorrhagic fever
66529	Apical ballooning syndrome	101206	APV/ADA, Fallot type	90	Arginase deficiency
324530	APLAID	99048	APV/PDA, non-Fallot type	90	Argininemia
1114	Aplasia cutis congenita	498359	Aquagenic keratoderma	23	Argininosuccinate deficiency
3339	Aplasia cutis congenita-epibulbar dermoids syndrome	498359	Aquagenic palmoplantar keratoderma	247525	Argininosuccinate synthase deficiency
1116	Aplasia cutis congenita-intestinal lymphangiectasia syndrome	498359	Aquagenic syringeal acrokeratoderma	247525	Argininosuccinate synthetase deficiency
370046	Aplasia cutis congenita-nevus sebaceus syndrome	498359	Aquagenic wrinkling of the palms	23	Argininosuccinate lyase deficiency
1117	Aplasia cutis-myopia syndrome	402041	AR dRTA	23	Argininosuccinic acid lyase deficiency
86815	Aplasia of lacrimal and salivary glands	→402041	AR dRTA with deafness	247525	Argininosuccinic acid synthase deficiency
3329	Aplasia of tibia with split-hand/split-foot deformity	→402041	AR dRTA with hearing loss	247525	Argininosuccinic acid synthetase deficiency
2879	Aplasia/hypoplasia of limbs and pelvis	→402041	AR dRTA without deafness	23	Argininosuccinic aciduria
520	APML	93607	AR pRTA	60014	Argyria
70590	Apnea of infancy	90119	AR-CMT2 with acrodystrophy	289176	ARHR
99981	Apnea of prematurity	90118	AR-CMT2, Ouvrier type	79235	Arias syndrome
425	ApoA-I deficiency	98856	AR-CMT2B1	2318	Arima syndrome
294986	Apodia	101101	AR-CMT2B2	950	Arkless-Graham syndrome
93560	Apolipoprotein A-I amyloidosis	228374	AR-CMT2B5	85276	Armfield syndrome
425	Apolipoprotein A-I deficiency	101102	AR-CMT2C	1915	ARND
238269	Apolipoprotein A-II amyloidosis	495274	AR-CMT2T	167635	Arndt-Gottron disease
439232	Apolipoprotein A-IV amyloidosis	169186	AR-CNM	268882	Arnold-Chiari malformation type 1
320	Apparent mineralocorticoid excess	248	AR-HED	1136	Arnold-Chiari malformation type 2
391723	Appendiceal mucinous adenocarcinoma	88616	AR-NSID	268882	Arnold-Chiari malformation type I
100079	Appendiceal NEN	731	AR-PKD	1136	Arnold-Chiari malformation type II
100079	Appendiceal neuroendocrine neoplasm	447760	AR-SPG9B	91	Aromatase deficiency
1201	Apple peel syndrome	1129	Arachnodactyly-abnormal ossification-intellectual disability syndrome	178345	Aromatase excess syndrome
506307	Apple-peel intestinal atresia-ocular anomalies-microcephaly syndrome	1130	Arachnodactyly-intellectual disability-dysmorphism syndrome	35708	Aromatic L-amino acid decarboxylase deficiency
1126	Aprosencephaly cerebellar dysgenesis	2356	Arachnoid cyst	254886	arPEO
976	APRT deficiency	137817	Arachnoiditis	99916	Arrhenoblastoma
3453	APS1	324442	ARAN-NM	1135	Arrhinia-choanal atresia-microphthalmia syndrome
		1915	ARBD	260305	ARSA
		2697	ARC syndrome	98	ARSACS
		88644	ARCA1	314603	ARSAL
		139485	ARCA2	583	ARSB deficiency
		90349	ARCL1	357107	Arterial cervical rib syndrome
		221145	ARCL1C	357107	Arterial costoclavicular syndrome
		357074	ARCL2, classic type	1682	Arterial dissection-lentiginosis syndrome
		357074	ARCL2, Debré type	357107	Arterial hyperabduction syndrome
		357064	ARCL2, progeroid type	357107	Arterial scalenus anticus syndrome
		357058	ARCL2A	357107	Arterial thoracic outlet compression syndrome
		357064	ARCL2B		
		324442	ARCMT2-NM		
		101097	ARCMT2K		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
357107	Arterial thoracic outlet syndrome	2302	Asbestosis	1180	Ataxia-hypogonadism-choroidal dystrophy syndrome
3342	Arterial tortuosity syndrome	1253	Ascher syndrome	370022	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome
357107	Arterial TOS	447997	ASCT1 deficiency	1168	Ataxia-oculomotor apraxia type 1
52	Arteriohepatic dysplasia	1478	ASD	64753	Ataxia-oculomotor apraxia type 2
261619	Arteriohepatic dysplasia due to a JAG1 point mutation	352490	ASD due to AUTS2 deficiency	459033	Ataxia-oculomotor apraxia type 4
261629	Arteriohepatic dysplasia due to a NOTCH2 point mutation	99104	ASD, coronary sinus type	2585	Ataxia-pancytopenia syndrome
261600	Arteriohepatic dysplasia due to monosomy 20p12	99106	ASD, ostium primum type	1184	Ataxia-photosensitivity-short stature syndrome
141174	Arteriovenous malformation of mandible	99103	ASD, ostium secundum type	1178	Ataxia-tapetoretinal degeneration syndrome
141171	Arteriovenous malformation of maxilla	99105	ASD, sinus venosus type	100	Ataxia-telangiectasia
29207	Arthritis urethritis	54251	Aseptic abscesses syndrome	370109	Ataxia-telangiectasia variant
955	Arthrodentoosteodysplasia	97337	Aseptic necrosis of patella	647	Ataxia-telangiectasia, variant 1
994	Arthrogryposis multiplex congenita-pulmonary hypoplasia syndrome	3314	Aseptic necrosis of phalangeal epiphyses	251347	Ataxia-telangiectasia-like disorder
1150	Arthrogryposis multiplex congenita-whistling face syndrome	2380	Aseptic necrosis of the capital femoral epiphysis	352403	Ataxie spinocérébelleuse à début infantile avec retard psychomoteur
3200	Arthrogryposis-ectodermal dysplasia-other anomalies syndrome	97336	Aseptic necrosis of the capital humerus	1183	Ataxo-opso-myoclonus syndrome
1485	Arthrogryposis-hyperkeratosis syndrome, lethal form	97332	Aseptic necrosis of the lunate bone	1190	Atelosteogenesis type 1
1144	Arthrogryposis-like hand anomaly-sensorineural deafness syndrome	2054	Aseptic necrosis of the tarsal bone	56304	Atelosteogenesis type 2
1149	Arthrogryposis-like syndrome	97335	Aseptic necrosis of the tibial tubercle	56305	Atelosteogenesis type 3
1154	Arthrogryposis-oculomotor limitation-electroretinal anomalies syndrome	57194	Aseptic osteitis	1190	Atelosteogenesis type I
2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	54251	Aseptic systemic abscesses	56304	Atelosteogenesis type II
65720	Arthrogryposis-severe scoliosis syndrome	137686	Asherman syndrome	56305	Atelosteogenesis type III
2848	Arthropathy-camptodactyly syndrome	276198	Asidan	69739	Athabascan brainstem dysgenesis syndrome
1187	Arts syndrome	23	ASL deficiency	69739	Athabaskan brainstem dysgenesis syndrome
512	Arylsulfatase A deficiency	391376	Asparagine synthetase deficiency	1192	Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome
309271	Arylsulfatase A deficiency, adult form	141	Aspartoacylase deficiency	95713	Athyreosis
309263	Arylsulfatase A deficiency, juvenile form	93	Aspartylglucosaminidase deficiency	1226	Athyroidal hypothyroidism-spiky hair-cleft palate syndrome
309256	Arylsulfatase A deficiency, late infantile form	93	Aspartylglucosaminuria	250977	ATIC deficiency
583	Arylsulfatase B deficiency	63442	ASPED	1193	Atkin-Flaitz syndrome
276212	Arylsulfatase B deficiency, rapidly progressing	1163	Aspergillosis	99666	Atlantoaxial subluxation
276223	Arylsulfatase B deficiency, slowly progressing	474	Asphyxiating thoracic dystrophy of the newborn	251347	ATLD
81	AS syndrome	163699	ASPS	86875	ATLL
23	ASA deficiency	247525	ASS deficiency	139423	ATM/TM
231466	ASAN	221120	ASSA	231401	ATMDS
583	ASB deficiency	85175	Astley-Kendall dysplasia	163934	Atopic keratoconjunctivitis
2302	Asbestos intoxication	251679	Astroblastoma	357107	ATOS
		647	AT V1	139557	ATP7A-related distal motor neuropathy
		96	Ataxia with isolated vitamin E deficiency	31150	ATP-binding cassette transporter A1 deficiency
		3008	Ataxia with lactic acidosis type 2	98791	ATR syndrome linked to chromosome 16
		3008	Ataxia with lactic acidosis type II	98791	ATR syndrome, deletion type
		94147	Ataxia with pigmentary retinopathy	98791	ATR-16 syndrome
		96	Ataxia with vitamin E deficiency	847	ATR-X syndrome
		1188	Ataxia-deafness-intellectual disability syndrome	1201	Atresia of small intestine
		137639	Ataxia-delayed dentition-hypomyelination syndrome	105	Atresia of urethra
		1227	Ataxia-diabetes-goiter-gonadal insufficiency syndrome		
		1188	Ataxia-hearing loss-intellectual disability syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
1344	Atrial cardiomyopathy with heart block	314721	Atypical dentin dysplasia due to SMOC2 deficiency	261501	Atypical Norrie disease due to Xp11.3 microdeletion
99107	Atrial septal aneurysm	398147	Atypical facial pain	216873	Atypical pantothenate kinase-associated neurodegeneration
1478	Atrial septal defect	309252	Atypical Gaucher disease due to saposin C deficiency	251902	Atypical papilloma of choroid plexus
99104	Atrial septal defect, coronary sinus type	289863	Atypical glycine encephalopathy	79474	Atypical progeroid syndrome
99106	Atrial septal defect, ostium primum type	98961	Atypical granular corneal dystrophy	99750	Atypical progressive supranuclear palsy syndrome
99103	Atrial septal defect, ostium secundum type	238523	Atypical HCS	99750	Atypical PSP syndrome
99105	Atrial septal defect, sinus venosus type	2134	Atypical hemolytic-uremic syndrome	3095	Atypical Rett syndrome
1479	Atrial septal defect-atrioventricular conduction defects syndrome	93581	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	3095	Atypical RTT
1344	Atrial standstill	93578	Atypical hemolytic-uremic syndrome with B factor anomaly	99966	Atypical teratoid rhabdoid tumor
844	Atrial tachyarrhythmia with short PR interval	93575	Atypical hemolytic-uremic syndrome with C3 anomaly	90393	Atypical tuberous myxedema of Jadassohn-Dosseker
86819	Atrichia with papular lesions	357008	Atypical hemolytic-uremic syndrome with DGKE deficiency	79474	Atypical Werner syndrome
392	Atriodigital dysplasia type 1	93579	Atypical hemolytic-uremic syndrome with H factor anomaly	16	Atypical X-linked achromatopsia
1350	Atriodigital dysplasia type 2	93580	Atypical hemolytic-uremic syndrome with I factor anomaly	453504	Au-Kline syndrome
1342	Atriodigital dysplasia type 3	93576	Atypical hemolytic-uremic syndrome with MCP/CD46 anomaly	166415	Audiogenic seizures
168796	Atriodigital dysplasia, Slovenian type	217023	Atypical hemolytic-uremic syndrome with thrombomodulin anomaly	1074	Aughton-Hufnagle syndrome
1352	Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	2134	Atypical HUS	1488	Aural atresia-multiple congenital anomalies-intellectual disability syndrome
86813	Atrophia areata	93581	Atypical HUS with anti-factor H antibodies	→794	Auralcephalosyndactyly
649	Atrophia bulborum hereditaria	93578	Atypical HUS with B factor anomaly	77300	Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome
254449	Atrophic lichen planus	93575	Atypical HUS with C3 anomaly	137888	Auriculocondylar syndrome
254449	Atrophic LP	357008	Atypical HUS with DGKE deficiency	114	Auriculosteodysplasia
79100	Atrophoderma vermiculata	93579	Atypical HUS with H factor anomaly	→794	Aurocephalosyndactyly
99966	ATRT	93580	Atypical HUS with I factor anomaly	1995	Ausems-Wittebol Post-Hennekam syndrome
71289	ATRUS syndrome	93576	Atypical HUS with MCP/CD46 anomaly	352490	Autism spectrum disorder due to AUTS2 deficiency
3342	ATS	217023	Atypical HUS with thrombomodulin anomaly	370943	Autism spectrum disorder-epilepsy-arthrogryposis syndrome
86818	ATS-MR	2134	Atypical HUS	308410	Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency
352723	Attenuated Chédiak-Higashi syndrome	93581	Atypical HUS with anti-factor H antibodies	137911	Autism-facial port-wine stain syndrome
220460	Attenuated familial adenomatous polyposis	93578	Atypical HUS with B factor anomaly	324636	Autoerythrocyte sensitization syndrome
220460	Attenuated familial polyposis coli	93575	Atypical HUS with C3 anomaly	85138	Autoimmune Addison disease
220460	Attenuated FAP	357008	Atypical HUS with DGKE deficiency	85138	Autoimmune adrenalitis
85451	ATTR cardiomyopathy	93579	Atypical HUS with H factor anomaly	420789	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea
85451	ATTRV122I amyloidosis	93580	Atypical HUS with I factor anomaly	391487	Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome
85451	ATTRV122I-related amyloidosis	93576	Atypical HUS with MCP/CD46 anomaly	37042	Autoimmune enteropathy type 1
85447	ATTRV30M amyloidosis	217023	Atypical HUS with thrombomodulin anomaly	103916	Autoimmune enteropathy type 2
85447	ATTRV30M-related amyloidosis	238523	Atypical hypotonia-cystinuria syndrome	103917	Autoimmune enteropathy type 3
330001	ATTRwt amyloidosis	391411	Atypical juvenile parkinsonism	1959	Autoimmune hemolytic anemia and
330001	ATTRwt-related amyloidosis	86797	Atypical lichen myxedematosus		
199627	Atypical autism	99971	Atypical lipoma		
352723	Atypical Chédiak-Higashi syndrome	99971	Atypical lipomatous tumor		
251902	Atypical choroid plexus papilloma	314466	Atypical Meigs syndrome		
98824	Atypical chronic myeloid leukemia	2578	Atypical MRKH syndrome		
1456	Atypical coarctation of aorta	289863	Atypical NKA		
251902	Atypical CPP	289863	Atypical non-ketotic hyperglycinemia		
314466	Atypical Demons-Meigs syndrome	261501	Atypical Norrie disease due to del(X)(p11.3)		
		261501	Atypical Norrie disease due to monosomy Xp11.3		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	autoimmune thrombocytopenia		proteinosis		Marie-Tooth disease type 2 due to NAGLU mutation
90033	Autoimmune hemolytic anemia, warm type	93585	Autoimmune thrombotic thrombocytopenic purpura	435819	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation
444463	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome	3143	Autoimmune thyroid disease and/or type 1 diabetes-Addison disease syndrome	435387	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to VCP mutation
2137	Autoimmune hepatitis	324977	Autoinflammation-lipodystrophy-dermatosis syndrome	401964	Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons
36913	Autoimmune hypoparathyroidism	324530	Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation	99946	Autosomal dominant Charcot-Marie-Tooth disease type 2A1
3453	Autoimmune hypoparathyroidism-chronic candidiasis-Addison disease syndrome	210115	Autoinflammatory disease due to interleukin-1 receptor antagonist deficiency	99947	Autosomal dominant Charcot-Marie-Tooth disease type 2A2
444092	Autoimmune interstitial lung disease-arthritis syndrome	329173	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis	99936	Autosomal dominant Charcot-Marie-Tooth disease type 2B
3261	Autoimmune lymphoproliferative syndrome	33110	Autosomal agammaglobulinemia	99937	Autosomal dominant Charcot-Marie-Tooth disease type 2C
436159	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsufficiency	209335	Autosomal dominant adult-onset proximal SMA	99938	Autosomal dominant Charcot-Marie-Tooth disease type 2D
275517	Autoimmune lymphoproliferative syndrome with recurrent viral infections	209335	Autosomal dominant adult-onset proximal spinal muscular atrophy	99939	Autosomal dominant Charcot-Marie-Tooth disease type 2E
589	Autoimmune myasthenia gravis	88918	Autosomal dominant Alport syndrome	99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F
206569	Autoimmune necrotizing myositis	1810	Autosomal dominant anhidrotic ectodermal dysplasia	99941	Autosomal dominant Charcot-Marie-Tooth disease type 2G
103919	Autoimmune pancreatitis	314399	Autosomal dominant aplasia and myelodysplasia	99942	Autosomal dominant Charcot-Marie-Tooth disease type 2I
280302	Autoimmune pancreatitis type 1	314399	Autosomal dominant aplastic anemia and myelodysplasia	99943	Autosomal dominant Charcot-Marie-Tooth disease type 2J
280315	Autoimmune pancreatitis type 2	1216	Autosomal dominant benign distal spinal muscular atrophy	99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K
747	Autoimmune PAP	314652	Autosomal dominant beta2-microglobulinic amyloidosis	99945	Autosomal dominant Charcot-Marie-Tooth disease type 2L
3453	Autoimmune polyendocrine syndrome type 1	93304	Autosomal dominant brachyolmia	228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M
3143	Autoimmune polyendocrine syndrome type 2	169189	Autosomal dominant centronuclear myopathy	228174	Autosomal dominant Charcot-Marie-Tooth disease type 2N
227982	Autoimmune polyendocrine syndrome type 3	314404	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	284232	Autosomal dominant Charcot-Marie-Tooth disease type 2O
227990	Autoimmune polyendocrine syndrome type 4	487814	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation	329258	Autosomal dominant Charcot-Marie-Tooth disease type 2Q
3453	Autoimmune polyendocrinopathy type 1	488333	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to HARS mutation	397735	Autosomal dominant Charcot-Marie-Tooth disease type 2U
3143	Autoimmune polyendocrinopathy type 2	324611	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation	447964	Autosomal dominant Charcot-Marie-Tooth disease type 2V
227982	Autoimmune polyendocrinopathy type 3	397735	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to MARS mutation	488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W
227990	Autoimmune polyendocrinopathy type 4	466768	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to MORC2 mutation	435387	Autosomal dominant Charcot-Marie-Tooth disease type 2Y
3453	Autoimmune polyglandular syndrome type 1	447964	Autosomal dominant Charcot-	466768	Autosomal dominant Charcot-Marie-Tooth disease type 2Z
3143	Autoimmune polyglandular syndrome type 2			98975	Autosomal dominant CHED
227982	Autoimmune polyglandular syndrome type 3			363447	Autosomal dominant childhood-onset proximal spinal muscular
227990	Autoimmune polyglandular syndrome type 4				
747	Autoimmune pulmonary alveolar				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	atrophy	73229	Autosomal dominant familial hematuria-retinal arteriolar tortuosity-contractures syndrome	324585	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain
363454	Autosomal dominant childhood-onset proximal spinal muscular atrophy with contractures	329466	Autosomal dominant focal dystonia, DYT25 type	90635	Autosomal dominant isolated neurosensory deafness type DFNA
209341	Autosomal dominant childhood-onset proximal spinal muscular atrophy without contractures	402003	Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering	90635	Autosomal dominant isolated neurosensory hearing loss type DFNA
79344	Autosomal dominant chondrodysplasia punctata	2024	Autosomal dominant gingival fibromatosis	90635	Autosomal dominant isolated sensorineural deafness type DFNA
→2526	Autosomal dominant chorioretinopathy-microcephaly syndrome	2024	Autosomal dominant gingival hyperplasia	90635	Autosomal dominant isolated sensorineural hearing loss type DFNA
1455	Autosomal dominant coarctation of aorta	139491	Autosomal dominant hereditary hemochromatosis	93325	Autosomal dominant Kenny-Caffey syndrome
447753	Autosomal dominant complex spastic paraparesis type 9A	401964	Autosomal dominant hereditary motor and sensory neuropathy type 2 with giant axons	2334	Autosomal dominant keratitis
447757	Autosomal dominant complex spastic paraparesis type 9B	2314	Autosomal dominant HIES	293936	Autosomal dominant keratoconus with early-onset anterior polar cataracts
1216	Autosomal dominant congenital benign spinal muscular atrophy	2314	Autosomal dominant hyper-IgE syndrome	503	Autosomal dominant Larsen syndrome
98975	Autosomal dominant congenital hereditary endothelial dystrophy	2314	Autosomal dominant hyperimmunoglobulin E syndrome	411602	Autosomal dominant late-onset Parkinson disease
86814	Autosomal dominant cortical myoclonus and epilepsy	276580	Autosomal dominant hyperinsulinemic hypoglycemia due to Kir6.2 deficiency	67042	Autosomal dominant late-onset retinal degeneration
90348	Autosomal dominant cutis laxa	276575	Autosomal dominant hyperinsulinemic hypoglycemia due to SUR1 deficiency	209335	Autosomal dominant late-onset spinal muscular atrophy, Finkel type
75381	Autosomal dominant cystoid macular edema	276580	Autosomal dominant hyperinsulinism due to Kir6.2 deficiency	101046	Autosomal dominant lateral temporal lobe epilepsy
79499	Autosomal dominant deafness-onychodystrophy syndrome	276575	Autosomal dominant hyperinsulinism due to SUR1 deficiency	313808	Autosomal dominant leukoencephalopathy with neuroaxonal spheroids
2337	Autosomal dominant diffuse palmoplantar keratoderma, Norrbotten type	428	Autosomal dominant hypocalcemia	266	Autosomal dominant limb-girdle muscular dystrophy type 1A
476093	Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome	1810	Autosomal dominant hypohidrotic ectodermal dysplasia	264	Autosomal dominant limb-girdle muscular dystrophy type 1B
139518	Autosomal dominant distal juvenile spinal muscular atrophy type 1	89937	Autosomal dominant hypophosphatemia	265	Autosomal dominant limb-girdle muscular dystrophy type 1C
93608	Autosomal dominant distal renal tubular acidosis	89937	Autosomal dominant hypophosphatemic rickets	34516	Autosomal dominant limb-girdle muscular dystrophy type 1D
98808	Autosomal dominant dopa-responsive dystonia	457193	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	34517	Autosomal dominant limb-girdle muscular dystrophy type 1E
→231568	Autosomal dominant dystrophic epidermolysis bullosa, Cockayne-Touraine type	100043	Autosomal dominant intermediate Charcot-Marie-Tooth disease type A	55595	Autosomal dominant limb-girdle muscular dystrophy type 1F
231568	Autosomal dominant dystrophic epidermolysis bullosa, Pasini and Cockayne-Touraine types	100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	55596	Autosomal dominant limb-girdle muscular dystrophy type 1G
→231568	Autosomal dominant dystrophic epidermolysis bullosa, Pasini type	100045	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C	238755	Autosomal dominant limb-girdle muscular dystrophy type 1H
300576	Autosomal dominant ectodermal dysplasia-cancer predisposition syndrome	100046	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	140957	Autosomal dominant macrothrombocytopenia
98853	Autosomal dominant Emery-Dreifuss muscular dystrophy	93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	34149	Autosomal dominant medullary cystic kidney disease
101046	Autosomal dominant epilepsy with auditory features	352670	Autosomal dominant intermediate Charcot-Marie-Tooth disease type F	88950	Autosomal dominant medullary cystic kidney disease type 2
				319581	Autosomal dominant mendelian susceptibility to mycobacterial

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	diseases due to partial IFNgammaR1 deficiency	306588	Autosomal dominant Opitz syndrome		detachment
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	67036	Autosomal dominant optic atrophy and cataract	3107	Autosomal dominant Robinow syndrome
319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 1 deficiency	→1215	Autosomal dominant optic atrophy and congenital deafness	247511	Autosomal dominant secondary erythrocytosis
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 2 deficiency	250932	Autosomal dominant optic atrophy and peripheral neuropathy	247511	Autosomal dominant secondary polycythemia
457050	Autosomal dominant mitochondrial myopathy with exercise intolerance	1215	Autosomal dominant optic atrophy plus syndrome	98808	Autosomal dominant Segawa syndrome
319581	Autosomal dominant MSMD due to partial IFNgammaR1 deficiency	67036	Autosomal dominant optic atrophy type 3	486	Autosomal dominant severe congenital neutropenia
319589	Autosomal dominant MSMD due to partial IFNgammaR2 deficiency	98673	Autosomal dominant optic atrophy, classic form	140481	Autosomal dominant slowed nerve conduction velocity
319581	Autosomal dominant MSMD due to partial interferon gamma receptor 1 deficiency	98673	Autosomal dominant optic atrophy, Kjer type	251282	Autosomal dominant spastic ataxia type 1
319589	Autosomal dominant MSMD due to partial interferon gamma receptor 2 deficiency	2783	Autosomal dominant osteopetrosis type 1	1182	Autosomal dominant spastic ataxia type 7
65743	Autosomal dominant multiple pterygium syndrome	1798	Autosomal dominant osteosclerosis, Stanescu type	100984	Autosomal dominant spastic paraplegia type 3
99846	Autosomal dominant myoglobinuria	2790	Autosomal dominant osteosclerosis, Worth type	100985	Autosomal dominant spastic paraplegia type 4
440354	Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome	1010	Autosomal dominant palmoplantar hyperkeratosis and congenital alopecia	100988	Autosomal dominant spastic paraplegia type 6
329211	Autosomal dominant neovascular inflammatory vitreoretinopathy	1010	Autosomal dominant palmoplantar keratoderma and congenital alopecia	100989	Autosomal dominant spastic paraplegia type 8
98784	Autosomal dominant nocturnal frontal lobe epilepsy	730	Autosomal dominant polycystic kidney disease	100990	Autosomal dominant spastic paraplegia type 9
178469	Autosomal dominant non-syndromic intellectual disability	88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	100991	Autosomal dominant spastic paraplegia type 10
90635	Autosomal dominant non-syndromic neurosensory deafness type DFNA	2924	Autosomal dominant polycystic liver disease	100993	Autosomal dominant spastic paraplegia type 12
90635	Autosomal dominant non-syndromic neurosensory hearing loss type DFNA	1300	Autosomal dominant popliteal pterygium syndrome	100994	Autosomal dominant spastic paraplegia type 13
90635	Autosomal dominant non-syndromic sensorineural deafness type DFNA	476119	Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome	100998	Autosomal dominant spastic paraplegia type 17
90635	Autosomal dominant non-syndromic sensorineural hearing loss type DFNA	34528	Autosomal dominant primary hypomagnesemia with hypocalciuria	100999	Autosomal dominant spastic paraplegia type 19
93328	Autosomal dominant omodysplasia	2514	Autosomal dominant primary microcephaly	101009	Autosomal dominant spastic paraplegia type 29
306588	Autosomal dominant Opitz BBB/G syndrome	2964	Autosomal dominant prognathism	101011	Autosomal dominant spastic paraplegia type 31
306588	Autosomal dominant Opitz G/BBB syndrome	254892	Autosomal dominant progressive external ophthalmoplegia	320365	Autosomal dominant spastic paraplegia type 36
	→ This disease is deprecated and has been moved to another (see annex). The indicated Orphanumber should now be used.	88659	Autosomal dominant progressive nephropathy with hypertension	171612	Autosomal dominant spastic paraplegia type 37
		314889	Autosomal dominant proximal renal tubular acidosis	171617	Autosomal dominant spastic paraplegia type 38
		171871	Autosomal dominant pseudohypoaldosteronism type 1	320355	Autosomal dominant spastic paraplegia type 41
		209867	Autosomal dominant rhegmatogenous retinal	171863	Autosomal dominant spastic paraplegia type 42
				444099	Autosomal dominant spastic paraplegia type 73
				1797	Autosomal dominant spondylocostal dysostosis
				1797	Autosomal dominant

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	spondylocostal dysplasia	169186	Autosomal recessive centronuclear myopathy		Marie-Tooth type 2 with acrodystryphy
228169	Autosomal dominant striatal neurodegeneration	453521	Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	293603	Autosomal recessive CHED
466806	Autosomal dominant thrombocytopenia with platelet secretion defect	352641	Autosomal recessive cerebellar ataxia due to GBA2 deficiency	508093	Autosomal recessive childhood-onset dystonia, DYT29 type
3357	Autosomal dominant trichodontoonychodyplasia-syndactyly	412057	Autosomal recessive cerebellar ataxia due to STUB1 deficiency	2518	Autosomal recessive chorioretinopathy-microcephaly syndrome
34149	Autosomal dominant tubulointerstitial kidney disease	88644	Autosomal recessive cerebellar ataxia type 1	2518	Autosomal recessive chorioretinopathy-microcephaly-intellectual disability syndrome
3086	Autosomal dominant vitreoretinochoroidopathy	139485	Autosomal recessive cerebellar ataxia type 2	506353	Autosomal recessive complex spastic paraplegia due to Kennedy pathway dysfunction
79278	Autosomal erythropoietic protoporphoria	352641	Autosomal recessive cerebellar ataxia with late-onset spasticity	447760	Autosomal recessive complex spastic paraplegia type 9B
88919	Autosomal recessive Alport syndrome	404499	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency	506353	Autosomal recessive complex SPG due to Kennedy pathway dysfunction
1027	Autosomal recessive amelia	404493	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency	363432	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency
248	Autosomal recessive anhidrotic ectodermal dysplasia	284282	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency	363432	Autosomal recessive congenital cerebellar ataxia due to ionotropic glutamate receptor delta-2 subunit deficiency
1116	Autosomal recessive aplasia cutis	284271	Autosomal recessive cerebellar ataxia-psychomotor retardation syndrome	324262	Autosomal recessive congenital cerebellar ataxia due to metabotropic glutamate receptor 1 deficiency
139485	Autosomal recessive ataxia due to coenzyme Q10 deficiency	363429	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome	324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency
247815	Autosomal recessive ataxia due to PEX10 deficiency	95434	Autosomal recessive cerebellar ataxia-saccadic intrusion syndrome	293603	Autosomal recessive congenital hereditary endothelial dystrophy
139485	Autosomal recessive ataxia due to ubiquinone deficiency	1170	Autosomal recessive cerebelloparenchymal disorder type 3	99951	Autosomal recessive congenital hypomyelinating neuropathy
88644	Autosomal recessive ataxia, Beauce type	363969	Autosomal recessive cerebral atrophy	90349	Autosomal recessive cutis laxa type 1
101101	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2B2	466775	Autosomal recessive Charcot-Marie-Tooth disease type 2 due to SPG11 mutation	221145	Autosomal recessive cutis laxa type 1C
101097	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2K	324442	Autosomal recessive Charcot-Marie-Tooth disease type 2 with neuromyotonia	357074	Autosomal recessive cutis laxa type 2, classic type
495274	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2T	98856	Autosomal recessive Charcot-Marie-Tooth disease type 2B1	357074	Autosomal recessive cutis laxa type 2, Debré type
90119	Autosomal recessive axonal Charcot-Marie-Tooth disease with acrodystryphy	228374	Autosomal recessive Charcot-Marie-Tooth disease type 2B5	357064	Autosomal recessive cutis laxa type 2, progeroid type
98856	Autosomal recessive axonal CMT4C1	466775	Autosomal recessive Charcot-Marie-Tooth disease type 2X	357058	Autosomal recessive cutis laxa type 2A
101102	Autosomal recessive axonal CMT4C2	101097	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	357064	Autosomal recessive cutis laxa type 2B
101101	Autosomal recessive axonal CMT4C3	90118	Autosomal recessive Charcot-Marie-Tooth disease, Ouvrier type	90349	Autosomal recessive cutis laxa with severe systemic involvement
101097	Autosomal recessive axonal CMT4C4	90119	Autosomal recessive Charcot-	90349	Autosomal recessive cutis laxa, pulmonary emphysema type
324442	Autosomal recessive axonal neuropathy with neuromyotonia			79500	Autosomal recessive deafness-
139455	Autosomal recessive bestrophinopathy				
448242	Autosomal recessive brachyolmia				
→3460	Autosomal recessive carpotarsal osteolysis				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	onychodystrophy syndrome		to SUR1 deficiency	268	Autosomal recessive limb-girdle muscular dystrophy type 2B
2776	Autosomal recessive distal osteolysis syndrome	79644	Autosomal recessive hyperinsulinism due to Kir6.2 deficiency	353	Autosomal recessive limb-girdle muscular dystrophy type 2C
402041	Autosomal recessive distal renal tubular acidosis	79643	Autosomal recessive hyperinsulinism due to SUR1 deficiency	62	Autosomal recessive limb-girdle muscular dystrophy type 2D
→402041	Autosomal recessive distal renal tubular acidosis with deafness	248	Autosomal recessive hypohidrotic ectodermal dysplasia	119	Autosomal recessive limb-girdle muscular dystrophy type 2E
→402041	Autosomal recessive distal renal tubular acidosis with hearing loss	289176	Autosomal recessive hypophosphatemic rickets	219	Autosomal recessive limb-girdle muscular dystrophy type 2F
→402041	Autosomal recessive distal renal tubular acidosis without deafness	300547	Autosomal recessive infantile hypercalcemia	34514	Autosomal recessive limb-girdle muscular dystrophy type 2G
→402041	Autosomal recessive distal renal tubular acidosis without hearing loss	93591	Autosomal recessive infantile nephronophthisis	1878	Autosomal recessive limb-girdle muscular dystrophy type 2H
402041	Autosomal recessive distal RTA	93591	Autosomal recessive infantile NPHP	34515	Autosomal recessive limb-girdle muscular dystrophy type 2I
→402041	Autosomal recessive distal RTA with deafness	352530	Autosomal recessive intellectual disability due to TRAPP/C9 deficiency	140922	Autosomal recessive limb-girdle muscular dystrophy type 2J
98920	Autosomal recessive distal spinal muscular atrophy type 1	217055	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	86812	Autosomal recessive limb-girdle muscular dystrophy type 2K
139552	Autosomal recessive distal spinal muscular atrophy type 2	254334	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B	206549	Autosomal recessive limb-girdle muscular dystrophy type 2L
139547	Autosomal recessive distal spinal muscular atrophy type 3	369867	Autosomal recessive intermediate Charcot-Marie-Tooth disease type C	206554	Autosomal recessive limb-girdle muscular dystrophy type 2M
206580	Autosomal recessive distal spinal muscular atrophy type 4	435998	Autosomal recessive intermediate Charcot-Marie-Tooth disease type D	206559	Autosomal recessive limb-girdle muscular dystrophy type 2N
314485	Autosomal recessive distal spinal muscular atrophy type 5	210110	Autosomal recessive intermediate osteopetrosis	206564	Autosomal recessive limb-girdle muscular dystrophy type 2O
101150	Autosomal recessive dopa-responsive dystonia	90636	Autosomal recessive isolated neurosensory deafness type DFNB	280333	Autosomal recessive limb-girdle muscular dystrophy type 2P
79408	Autosomal recessive dystrophic epidermolysis bullosa generalisata gravis	98676	Autosomal recessive isolated optic atrophy	254361	Autosomal recessive limb-girdle muscular dystrophy type 2Q
89842	Autosomal recessive dystrophic epidermolysis bullosa generalisata mitis	90636	Autosomal recessive isolated sensorineural deafness type DFNB	363543	Autosomal recessive limb-girdle muscular dystrophy type 2R
89842	Autosomal recessive dystrophic epidermolysis bullosa, generalized other	93324	Autosomal recessive Kenny-Caffey syndrome	369840	Autosomal recessive limb-girdle muscular dystrophy type 2S
79408	Autosomal recessive dystrophic epidermolysis bullosa, Hallopeau-Siemens type	1842	Autosomal recessive lethal chondrodysplasia, round femoral inferior epiphysis type	363623	Autosomal recessive limb-girdle muscular dystrophy type 2T
98855	Autosomal recessive Emery-Dreifuss muscular dystrophy	33108	Autosomal recessive lethal multiple pterygium syndrome	352479	Autosomal recessive limb-girdle muscular dystrophy type 2U
289586	Autosomal recessive exfoliative ichthyosis	314572	Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome	466801	Autosomal recessive limb-girdle muscular dystrophy type 2W
1974	Autosomal recessive faciodigitogenital syndrome	363543	Autosomal recessive limb-girdle muscular dystrophy due to desmin deficiency	476084	Autosomal recessive limb-girdle muscular dystrophy type 2X
329329	Autosomal recessive frontotemporal pachygyria	352479	Autosomal recessive limb-girdle muscular dystrophy due to ISPD deficiency	424261	Autosomal recessive limb-girdle muscular dystrophy type 2Y
331226	Autosomal recessive hyper-IgE syndrome due to TYK2 deficiency	254361	Autosomal recessive limb-girdle muscular dystrophy due to plectin deficiency	480682	Autosomal recessive limb-girdle muscular dystrophy type 2Z
79644	Autosomal recessive hyperinsulinemic hypoglycemia due to Kir6.2 deficiency	267	Autosomal recessive limb-girdle muscular dystrophy type 2A	476084	Autosomal recessive limb-girdle muscular dystrophy-cardiac arrhythmia syndrome
79643	Autosomal recessive hyperinsulinemic hypoglycemia due			206580	Autosomal recessive lower motor neuron disease with childhood onset
				238505	Autosomal recessive lymphoproliferative disease

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
667	Autosomal recessive malignant osteopetrosis	90636	Autosomal recessive non-syndromic neurosensory deafness type DFNB	247378	Autosomal recessive secondary erythrocytosis, non-Chuvash type
477857	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency	90636	Autosomal recessive non-syndromic sensorineural deafness type DFNB	247378	Autosomal recessive secondary polycythemia not associated with VHL gene
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	98676	Autosomal recessive nonsyndromic optic atrophy	247378	Autosomal recessive secondary polycythemia, non-Chuvash type
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	93329	Autosomal recessive omodysplasia	101150	Autosomal recessive Segawa syndrome
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 1 deficiency	67047	Autosomal recessive optic atrophy plus syndrome	970	Autosomal recessive sensory radicular neuropathy
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 2 deficiency	67047	Autosomal recessive optic atrophy type 3	70594	Autosomal recessive sepiapterin reductase-deficient DRD
175	Autosomal recessive metaphyseal chondrodysplasia	227976	Autosomal recessive optic atrophy, OPA7 type	420702	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency
621	Autosomal recessive methemoglobinemia	178389	Autosomal recessive osteoclast-poor osteopetrosis with hypogammaglobulinemia	420699	Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency
477857	Autosomal recessive MSMD due to complete RORgamma receptor deficiency	178389	Autosomal recessive osteopetrosis type 7	331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency
319569	Autosomal recessive MSMD due to partial IFNgammaR1 deficiency	1366	Autosomal recessive palmoplantar hyperkeratosis and congenital alopecia	423384	Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency
319574	Autosomal recessive MSMD due to partial IFNgammaR2 deficiency	1366	Autosomal recessive palmoplantar keratoderma and congenital alopecia	260305	Autosomal recessive sideroblastic anemia
319569	Autosomal recessive MSMD due to partial interferon gamma receptor 1 deficiency	731	Autosomal recessive polycystic kidney disease	98	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
319574	Autosomal recessive MSMD due to partial interferon gamma receptor 2 deficiency	1234	Autosomal recessive popliteal pterygium syndrome	397946	Autosomal recessive spastic ataxia type 2
93307	Autosomal recessive multiple epiphyseal dysplasia	88628	Autosomal recessive posterior column ataxia and retinitis pigmentosa	314603	Autosomal recessive spastic ataxia type 3
2990	Autosomal recessive multiple pterygium syndrome	477857	Autosomal recessive primary immunodeficiency due to RORC mutation	254343	Autosomal recessive spastic ataxia type 4
424261	Autosomal recessive muscular dystrophy due to LAP1B deficiency	437552	Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity	313772	Autosomal recessive spastic ataxia type 5
424261	Autosomal recessive muscular dystrophy due to Torsin-1A-interacting protein 1 deficiency	437552	Autosomal recessive primary immunodeficiency with defective spontaneous NK cell cytotoxicity	98	Autosomal recessive spastic ataxia type 6
319332	Autosomal recessive myogenic AMC	2512	Autosomal recessive primary microcephaly	314603	Autosomal recessive spastic ataxia with leukoencephalopathy
319332	Autosomal recessive myogenic arthrogryposis multiplex congenita	254886	Autosomal recessive progressive external ophthalmoplegia	254343	Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome
280654	Autosomal recessive nail dysplasia	93607	Autosomal recessive proximal renal tubular acidosis	100986	Autosomal recessive spastic paraplegia type 5A
2990	Autosomal recessive non-lethal multiple pterygium syndrome	171876	Autosomal recessive pseudohypoaldosteronism type 1	2822	Autosomal recessive spastic paraplegia type 11
88616	Autosomal recessive non-syndromic intellectual disability	1507	Autosomal recessive Robinow syndrome	100995	Autosomal recessive spastic paraplegia type 14
		247378	Autosomal recessive secondary erythrocytosis not associated with VHL gene	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

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	paraplegia type 21		paraplegia type 65	95433	Autosomal recessive spinocerebellar ataxia-blindness-hearing loss syndrome
101003	Autosomal recessive spastic paraplegia type 23	401815	Autosomal recessive spastic paraplegia type 66	2311	Autosomal recessive spondylocostal dysostosis
101004	Autosomal recessive spastic paraplegia type 24	401820	Autosomal recessive spastic paraplegia type 67	401979	Autosomal recessive spondylometaphyseal dysplasia, M��garban�� type
101005	Autosomal recessive spastic paraplegia type 25	401825	Autosomal recessive spastic paraplegia type 68	250984	Autosomal recessive Stickler syndrome
101006	Autosomal recessive spastic paraplegia type 26	401830	Autosomal recessive spastic paraplegia type 69	745	Autosomal recessive thrombophilia due to congenital protein C deficiency
101007	Autosomal recessive spastic paraplegia type 27	401835	Autosomal recessive spastic paraplegia type 70	743	Autosomal recessive thrombophilia due to congenital protein S deficiency
101008	Autosomal recessive spastic paraplegia type 28	401840	Autosomal recessive spastic paraplegia type 71	745	Autosomal recessive thrombophilia due to PC deficiency
171622	Autosomal recessive spastic paraplegia type 32	468661	Autosomal recessive spastic paraplegia type 74	280365	Autosomal semi-dominant severe lipodystrophic laminopathy
171629	Autosomal recessive spastic paraplegia type 35	459056	Autosomal recessive spastic paraplegia type 75	300345	Autosomal SLE
139480	Autosomal recessive spastic paraplegia type 39	488594	Autosomal recessive spastic paraplegia type 76	101010	Autosomal spastic paraplegia type 30
320370	Autosomal recessive spastic paraplegia type 43	466722	Autosomal recessive spastic paraplegia type 77	401849	Autosomal spastic paraplegia type 72
320401	Autosomal recessive spastic paraplegia type 44	101005	Autosomal recessive spastic paraplegia-disc herniation syndrome	300345	Autosomal systemic lupus erythematosus
320396	Autosomal recessive spastic paraplegia type 45	98920	Autosomal recessive spinal muscular atrophy with respiratory distress	168629	Autosomal thrombocytopenia with normal platelets
320391	Autosomal recessive spastic paraplegia type 46	1170	Autosomal recessive spinocerebellar ataxia type 2	352490	AUTS2 syndrome
306511	Autosomal recessive spastic paraplegia type 48	95433	Autosomal recessive spinocerebellar ataxia type 3	96	AVED
320385	Autosomal recessive spastic paraplegia type 49	284332	Autosomal recessive spinocerebellar ataxia type 6	98963	Avellino corneal dystrophy
319199	Autosomal recessive spastic paraplegia type 53	284324	Autosomal recessive spinocerebellar ataxia type 7	454836	Avian influenza
320380	Autosomal recessive spastic paraplegia type 54	139485	Autosomal recessive spinocerebellar ataxia type 9	99000	AVMD
320375	Autosomal recessive spastic paraplegia type 55	284289	Autosomal recessive spinocerebellar ataxia type 10	58	AxD
320411	Autosomal recessive spastic paraplegia type 56	284271	Autosomal recessive spinocerebellar ataxia type 11	363717	AxD type I
431329	Autosomal recessive spastic paraplegia type 57	284282	Autosomal recessive spinocerebellar ataxia type 12	363722	AxD type II
397946	Autosomal recessive spastic paraplegia type 58	324262	Autosomal recessive spinocerebellar ataxia type 13	98978	Axenfeld anomaly
401795	Autosomal recessive spastic paraplegia type 59	352403	Autosomal recessive spinocerebellar ataxia type 14	782	Axenfeld syndrome
401800	Autosomal recessive spastic paraplegia type 60	404499	Autosomal recessive spinocerebellar ataxia type 15	782	Axenfeld-Rieger syndrome
401780	Autosomal recessive spastic paraplegia type 61	397709	Autosomal recessive spinocerebellar ataxia type 20	1834	Axial mesodermal dysplasia spectrum
401785	Autosomal recessive spastic paraplegia type 62	466794	Autosomal recessive spinocerebellar ataxia type 21	2777	Axial osteosclerosis
401805	Autosomal recessive spastic paraplegia type 63	95433	Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome	168549	Axial spondylometaphyseal dysplasia
401810	Autosomal recessive spastic paraplegia type 64			401911	AXIN2-related AFAP
320396	Autosomal recessive spastic			401911	AXIN2-related attenuated familial adenomatous polyposis
				401911	AXIN2-related attenuated familial polyposis coli
				401911	AXIN2-related attenuated FAP
				101102	Axonal Charcot-Marie-Tooth disease with pyramidal involvement
				457205	Axonal neuropathy-optic atrophy-cognitive deficit syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
209004	Axonal polyneuropathy associated with IgG/IgM/IgA monoclonal gammopathy	1227	Bangstad syndrome	100976	Bathing suit ichthyosis
1435	Ayazi syndrome	130	Bangungut	1948	Battaglia-Neri syndrome
1272	Aymé-Gripp syndrome	1228	Banki syndrome	79264	Batten disease
284454	AZOOR	109	Bannayan-Riley-Ruvalcaba syndrome	→1071	Baughman syndrome
→399805	Azoospermia due to maturation arrest	139507	Bantu siderosis	166113	Bazex syndrome
→399805	Azoospermia due to meiosis defect	289539	BAP1-related tumor predisposition syndrome	113	Bazex-Dupré-Christol syndrome
3471	Azoospermia-sinopulmonary infections syndrome	1229	Baraitser-Brett-Piesowicz syndrome	65284	BBGD
98757	Azorean disease of the nervous system	2753	Baraitser-Burn syndrome	363444	BBIS
99121	Azygos continuation of the inferior caval vein	1229	Baraitser-Reardon syndrome	110	BBS
99121	Azygos continuation of the inferior vena cava	2995	Baraitser-Winter cerebrofrontofacial syndrome	401777	BBSOAS
99121	Azygos continuation of the IVC	2237	Barakat syndrome	41751	BCD
79332	B4GALT1-CDG	1231	Barber-Say syndrome	1997	BCD syndrome
75496	B4GALT7-CDG	110	Bardet-Biedl syndrome	312	BCIE
99860	B-ALL	34592	Bare lymphocyte syndrome type 1	511	BCKD deficiency
67038	B-cell chronic lymphocytic leukemia	572	Bare lymphocyte syndrome type 2	511	BCKDH deficiency
67038	B-cell chronic lymphoid leukemia	3317	Barnes syndrome	247203	BDC
464336	B-cell expansion with NF-κB and T-cell anergy disease	443084	Baroreflex failure	113	BDCS
86852	B-cell prolymphocytic leukemia	79087	Barraquer-Simons syndrome	115	Beals syndrome
67038	B-CLL	2698	Bart-Pumphrey syndrome	115	Beals-Hecht syndrome
404560	B-K mole syndrome	111	Barth syndrome	1059	Bean syndrome
86852	B-PLL	64692	Bartonellosis due to Bartonella bacilliformis infection	1555	Beare-Stevenson cutis gyrata syndrome
108	Babesiosis	50839	Bartonellosis due to Bartonella henselae infection	363444	Beaulieu-Boycott-Innes syndrome
206994	Bacterial myositis	64694	Bartonellosis due to Bartonella quintana infection	98895	Becker dystrophinopathy
36234	Bacterial toxic-shock syndrome	1234	Bartsocas-Papas syndrome	98895	Becker muscular dystrophy
36234	Bacterial TSS	112	Bartter syndrome	64755	Becker nevus syndrome
69736	BADI	93605	Bartter syndrome type 3	116	Beckwith-Wiedemann syndrome
86814	BAFME	89938	Bartter syndrome type 4	231127	Beckwith-Wiedemann syndrome due to 11p15 microdeletion
2819	Bahemuka-Brown syndrome	263417	Bartter syndrome type 5	96076	Beckwith-Wiedemann syndrome due to 11p15 microduplication
352577	Bainbridge-Ropers syndrome	93605	Bartter syndrome type III	231130	Beckwith-Wiedemann syndrome due to 11p15 translocation/inversion
1658	Baird syndrome	89938	Bartter syndrome type IV	231120	Beckwith-Wiedemann syndrome due to CDKN1C mutation
139471	Bakrania-Ragge syndrome	263417	Bartter syndrome type V	231117	Beckwith-Wiedemann syndrome due to imprinting defect of 11p15
1223	Balantidiasis	93604	Bartter syndrome with hypocalcemia	238613	Beckwith-Wiedemann syndrome due to NSD1 mutation
1223	Balantidiosis	93604	Bartter syndrome, furosemide type	96193	Beckwith-Wiedemann syndrome due to paternal uniparental disomy of chromosome 11
139450	Balikova-Vermeesch syndrome	494451	Basal cell carcinoma of vulva	1945	BECRS
363746	Balint syndrome	377	Basal cell nevus syndrome	1945	BECTS
363746	Balint-Holmes syndrome	268829	Basal encephalocele	2572	Bedouin spastic ataxia syndrome
93395	Ballard syndrome	→1658	Basan syndrome	322	BEEC
1225	Baller-Gerold syndrome	79301	BASD1	1237	Beemer-Ertbruggen syndrome
66529	Ballooning cardiomyopathy	79303	BASD2	275864	Behavioral variant of frontotemporal dementia
228165	Baló concentric sclerosis	79302	BASD3	2705	Behrens-Baumann-Vogel syndrome
634	Bamboo hair syndrome	79095	BASD4	117	Behçet disease
1226	Bamforth syndrome	50810	Basel-Vanagaite-Sirota syndrome	476102	Behçet-like disease due to HA20
1226	Bamforth-Lazarus syndrome	244283	BASM syndrome	476102	Behçet-like disease due to haploinsufficiency of A20
98955	Band-shaped and whorled microcystic dystrophy of the corneal epithelium	14	Bassen-Kornzweig disease		
		1875	Bassoe syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
247203	Bellini carcinoma		syndrome		deficiency
247203	Bellini duct carcinoma	464359	Benign metanephric tumour	65287	Beta-alanine synthase deficiency
1240	Bellini syndrome	168816	Benign multicystic peritoneal mesothelioma	309310	Beta-D-galactosidase deficiency
100978	Benallegue-Lacete syndrome	86909	Benign myoclonic epilepsy of infancy	354	Beta-galactosidase-1 deficiency
1241	Bencze syndrome	86909	Benign myoclonus epilepsy of infancy	584	Beta-glucuronidase deficiency
86814	Benign adult familial myoclonic epilepsy	140927	Benign neonatal-infantile epilepsy	134	Beta-ketothiolase deficiency
86814	Benign adult familial myoclonus epilepsy	209973	Benign nocturnal alternating hemiplegia of childhood	118	Beta-mannosidase deficiency
610	Benign autosomal dominant myopathy	64545	Benign nonfamilial neonatal seizures	118	Beta-mannosidosis
157997	Benign cephalic histiocytosis	25968	Benign occipital epilepsy	1035	Beta-mercaptolactate cysteine disulfiduria
98816	Benign childhood occipital epilepsy, Gastaut type	342	Benign paroxysmal peritonitis	329284	Beta-propeller protein-associated neurodegeneration
98815	Benign childhood occipital epilepsy, Panayiotopoulos type	1179	Benign paroxysmal tonic upgaze of childhood with ataxia	119	Beta-sarcoglycanopathy
2841	Benign chronic familial pemphigus of Hailey-Hailey	71518	Benign paroxysmal torticollis of infancy	848	Beta-thalassemia
251287	Benign concentric annular macular dystrophy	166299	Benign partial epilepsy of infancy with complex partial seizures	231222	Beta-thalassemia intermedia
440233	Benign congenital sixth cranial nerve palsy	166302	Benign partial epilepsy with secondarily generalized seizures in infancy	231214	Beta-thalassemia major
254864	Benign COX deficiency	65682	Benign recurrent intrahepatic cholestasis	→33364	Beta-thalassemia-trichothiodystrophy syndrome
1945	Benign epilepsy of childhood with centrotemporal spikes	99960	Benign recurrent intrahepatic cholestasis type 1	231393	Beta-thalassemia-X-linked thrombocytopenia syndrome
276148	Benign epithelial tumor of salivary glands	99961	Benign recurrent intrahepatic cholestasis type 2	65287	Beta-ureidopropionase deficiency
1429	Benign familial chorea	342	Benign recurrent polyserositis	610	Bethlem myopathy
1945	Benign familial epilepsy of childhood with rolandic spikes	1945	Benign rolandic epilepsy	2114	Beukes familial hip dysplasia
306	Benign familial infantile convulsions	324581	Benign Samaritan congenital myopathy	2114	BFHD
306	Benign familial infantile epilepsy	252164	Benign schwannoma	306	BFIE
306	Benign familial infantile seizures	180237	Benign tumor of fallopian tubes	306	BFIS
163717	Benign familial mesial temporal lobe epilepsy	2198	Bennion-Patterson syndrome	127	BFLS
1949	Benign familial neonatal convulsions	54247	Benson syndrome	140927	BNFIS
1949	Benign familial neonatal epilepsy	464336	BENTA disease	1949	BFNS
1949	Benign familial neonatal seizures	528	Berardinelli-Seip syndrome	293284	BH4-responsive HPA/PKU
140927	Benign familial neonatal-infantile seizures	171839	Berant syndrome	293284	BH4-responsive hyperphenylalaninemia/phenylketonuria
163717	Benign FMTLE	528	Berardinelli-Seip congenital lipodystrophy	1429	BHC
65684	Benign focal amyotrophy	2241	Berdon syndrome	93311	BHMED
1544	Benign focal seizures of adolescence	647	Berlin breakage syndrome	98964	Biber-Haab-Dimmer dystrophy
1429	Benign hereditary chorea	1816	Berlin syndrome	180086	Bicervical bicornuate uterus
254704	Benign hyperferritinemia	274	Bernard-Soulier syndrome	180106	Bicervical bicornuate uterus and blind hemivagina
64545	Benign idiopathic neonatal seizures	178528	Berti lymphoma	180111	Bicervical bicornuate uterus with patent cervix and vagina
166308	Benign infantile focal epilepsy with midline spikes and waves during sleep	133	Berylliosis	2088	Bickel-Fanconi glycogenosis
166305	Benign infantile seizures associated with mild gastroenteritis	797	Besnier-Boeck-Schaumann disease	2182	Bickers-Adams syndrome
238624	Benign intracranial hypertension	321	Bessel-Hagen disease	79138	Bickerstaff brainstem encephalitis
285	Benign joint hypermobility	1243	Best disease	3286	Bidirectional tachycardia induced by catecholamine
		1243	Best macular dystrophy	→33364	BIDS syndrome
		1243	Best vitelliform macular dystrophy	1246	Biemond syndrome
		79332	Beta-1,4-galactosyltransferase	141333	Biemond syndrome type 2
				41751	Bietti crystalline corneoretinal dystrophy
				41751	Bietti crystalline dystrophy
				41751	Bietti crystalline retinopathy
				1986	Bifid femur-monodactylous

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	ectrodactyly syndrome	98836	Bilineal acute leukemia	98885	Bleeding diathesis due to glycoprotein VI deficiency
2695	Bifid nose	415286	Bilirubin encephalopathy	98886	Bleeding diathesis due to integrin alpha2-beta1 deficiency
217266	Bifid nose with or without anorectal and renal anomalies	205	Bilirubin uridinediphosphate glucuronosyltransferase deficiency	220443	Bleeding diathesis due to thromboxane synthesis deficiency
99771	Bifid uvula	79234	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 1		Bleeding disorder due to calcium- and DAG-regulated guanine exchange factor-1 deficiency
99771	Bifidity of the uvula	79235	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 2	420566	Bleeding disorder due to CalDAG-GEFI deficiency
300	Bifunctional enzyme deficiency	205	Bilirubin-UGT deficiency	1997	Blepharo-cheilo-odontic syndrome
637	Bilateral acoustic neurofibromatosis	79234	Bilirubin-UGT deficiency type 1	1253	Blepharochalasis-double lip syndrome
69736	Bilateral acute depigmentation of the iris	79235	Bilirubin-UGT deficiency type 2	1997	Blepharocheilodontic syndrome
319205	Bilateral adrenal hemorrhage	1799	Billard-Toutain-Maheut syndrome	→2353	Blepharofacioskeletal syndrome
325124	Bilateral anorchia	166308	BIMSE	1252	Blepharonasofacial malformation syndrome
2048	Bilateral anterior opercular syndrome	1248	Binder syndrome	2728	Blepharophimosis syndrome, Ohdo type
1229	Bilateral band-like calcification with polymicrogyria	3304	Bindewald-Ulmer-Müller syndrome	126	Blepharophimosis types 1 and 2
208444	Bilateral frontal polymicrogyria	64545	BINS	261572	Blepharophimosis types 1 and 2 due to a point mutation
101070	Bilateral frontoparietal polymicrogyria	65284	Biotin-responsive basal ganglia disease	261579	Blepharophimosis types 1 and 2 due to copy number variations
208447	Bilateral generalized polymicrogyria	65284	Biotin-thiamine-responsive basal ganglia disease	261559	Blepharophimosis-epicanthus inversus-ptosis due to 3q23 rearrangement syndrome
93311	Bilateral hereditary micro-epiphyseal dysplasia	79241	Biotinidase deficiency	261579	Blepharophimosis-epicanthus inversus-ptosis due to a CNV
438117	Bilateral hip and radial head dislocations-short stature-scoliosis-carpal coalitions-pes cavus-facial dysmorphism syndrome	54247	Biparietal Alzheimer disease	261572	Blepharophimosis-epicanthus inversus-ptosis due to a point mutation syndrome
319205	Bilateral massive adrenal hemorrhage	364198	Bipartite talus	261579	Blepharophimosis-epicanthus inversus-ptosis due to copy number variations
97364	Bilateral MCDK	99908	Bird fancier lung	126	Blepharophimosis-epicanthus inversus-ptosis syndrome
140963	Bilateral microtia-deafness-cleft palate syndrome	2617	Bird-headed dwarfism, Montreal type	→2707	Blepharophimosis-intellectual disability syndrome due to UBE3B deficiency
97364	Bilateral multicystic dysplastic kidney	179	Birdshot chorioretinitis	293725	Blepharophimosis-intellectual disability syndrome type V
97364	Bilateral multicystic renal dysplasia	179	Birdshot chorioretinopathy	293707	Blepharophimosis-intellectual disability syndrome, Maat-Kievit-Brunner type
208441	Bilateral parasagittal parieto-occipital polymicrogyria	179	Birdshot retinochoroiditis	293707	Blepharophimosis-intellectual disability syndrome, MKB type
98889	Bilateral perisylvian polymicrogyria	122	Birdshot retinochoroidopathy	2728	Blepharophimosis-intellectual disability syndrome, Ohdo type
268940	Bilateral polymicrogyria	79133	Bitemporal aplasia cutis congenita	3047	Blepharophimosis-intellectual disability syndrome, SBBYS type
1980	Bilateral striopallidodentate calcinosis	2213	Bixler-Christian-Gorlin syndrome	293725	Blepharophimosis-intellectual disability syndrome, Verloes type
1314	Bilateral symmetrical thalamic gliosis	285	BJHS	2057	Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome
276066	Bile acid CoA ligase deficiency and defective amidation	123	Björnstad syndrome		
70567	Bile duct cancer	124	Blackfan-Diamond anemia		
1276	Bilginturan brachydactyly	93930	Bladder exstrophy		
1276	Bilginturan syndrome	322	Bladder exstrophy-epispadias-cloacal extrophy complex		
1247	Bilharziasis	37202	Bladder pain syndrome		
244283	Biliary atresia with splenic malformation syndrome	98922	Blake pouch cyst		
424982	Biliary cystadenocarcinoma	254379	Blaschkoid lichen planus		
386	Biliary hamartoma	254379	Blaschkoid LP		
→2697	Biliary tract malformation-renal failure syndrome	86870	Blastic NK-cell lymphoma		
		86870	Blastic plasmacytoid dendritic cell neoplasm		
		1834	Blastogenesis defect		
		90340	Blau syndrome		
		50945	BLC		
		1229	BLC-PMG		
		73271	Bleeding diathesis due to a collagen receptor defect		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
1968	Blepharophimosis-telecanthus-microstomia syndrome		proptosis-hydrocephalus syndrome	93383	Brachydactyly type B
1259	Blepharoptosis-myopia-ectopia lentis syndrome	2934	Bonneau syndrome	140908	Brachydactyly type B2
93964	Blepharospasm-oromandibular dystonia syndrome	163	Bonneau-Beaumont syndrome	93384	Brachydactyly type C
171844	Blindness-scoliosis-arachnodactyly syndrome	2941	Bonnemann-Meinecke syndrome	93387	Brachydactyly type E
464	Bloch-Siemens syndrome	1261	Bonnemann-Meinecke-Reich syndrome	1276	Brachydactyly type E, with short stature and hypertension
464	Bloch-Sulzberger syndrome	53719	Bonnet-Dechaume-Blanc syndrome	93395	Brachydactyly types B and E combined
50945	Blomstrand chondrodysplasia	1262	Böök syndrome	93388	Brachydactyly, Farabee type
50945	Blomstrand lethal chondrodysplasia	1263	Boomerang dysplasia	2946	Brachydactyly, long thumb type
125	Bloom syndrome	1303	BOOP	93396	Brachydactyly, Mohr-Wriedt type
2768	Blount disease	1933	Booth-Haworth-Dilling syndrome	93397	Brachydactyly, Smorgasbord type
88629	Blue colour blindness	107	BOR syndrome	93394	Brachydactyly, Temtamy type
16	Blue cone monochromacy	206473	Borderline epithelial tumor of ovary	1276	Brachydactyly-arterial hypertension syndrome
16	Blue cone monochromatism	206473	Borderline ovarian epithelial tumor	1275	Brachydactyly-elbow wrist dysplasia syndrome
94086	Blue diaper syndrome	127	Borjeson-Forssman-Lehmann syndrome	1001	Brachydactyly-intellectual disability syndrome
1059	Blue rubber bleb nevus	1264	Bork syndrome	1275	Brachydactyly-joint dysplasia syndrome
98989	Blue-dot cataract	90001	Bornholm eye disease	2946	Brachydactyly-long thumb syndrome
319205	BMAH	36273	Borrmann gastric cancer type 4	1277	Brachydactyly-mesomelia-intellectual disability-heart defects syndrome
1243	BMD	97297	BOS syndrome	1246	Brachydactyly-nystagmus-cerebellar ataxia syndrome
98895	BMD	401777	Bosch-Boonstra-Schaaf optic atrophy syndrome	1278	Brachydactyly-preaxial hallux varus syndrome
293725	BMRS type V	69737	Bosley-Salih-Alorainy syndrome	2956	Brachydactyly-scoliosis-carpal fusion syndrome
293707	BMRS, Maat-Kievit-Brunner type	2250	Bosma arhinia-microphthalmia syndrome	391646	Brachydactyly-short stature-microcephaly syndrome
293707	BMRS, MKB type	2250	Bosma-Henkin-Christiansen syndrome	166035	Brachydactyly-short stature-retinitis pigmentosa syndrome
2728	BMRS, Ohdo type	85128	Bothnia retinal dystrophy	3168	Brachydactyly-symphalangism syndrome
293725	BMRS, Verloes type	128	Bothriocephalosis	93409	Brachydactyly-syndactyly, Zhao type
353253	BMS	1267	Botulism	93394	Brachymesophalangy II and V
217266	BNAR syndrome	1180	Boucher-Neuhäuser syndrome	1292	Brachymorphism-onychodysplasia-dysphalangism syndrome
50945	BOCD	805	Bourneville syndrome	→448242	Brachyolmia type 1, Hobaek type
217008	Bockenheimer syndrome	83313	Boutonneuse fever	→448242	Brachyolmia type 1, Toledo type
1292	BOD syndrome	→912	Bowen syndrome	93302	Brachyolmia type 2
2724	Boder syndrome	1270	Bowen syndrome, Hutterite type	93304	Brachyolmia type 3
48686	Body cavity-based lymphoma	1270	Bowen-Conradi syndrome	448242	Brachyolmia, Hobaek/Toledo type
91135	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	97353	Boxer's dementia	93302	Brachyolmia, Maroteaux type
797	Boeck sarcoid	50814	Boyadjiev-Jabs syndrome	2899	Brachyolmia-amelogenesis imperfecta syndrome
797	Boeck's sarcoid	329284	BPAN	79345	Brachytelephalangic chondrodysplasia punctata
1297	BOFS	70589	BPD	1295	Brachytelephalangy-dysmorphism-Kallmann syndrome
97297	Bohring syndrome	86870	BPDCN		
97297	Bohring-Opitz syndrome	2901	Brachial plexus neuritis		
84081	Boichis disease	199	Brachmann-de Lange syndrome		
401874	BOLA3 deficiency	1519	Brachycephalofrontonasal dysplasia		
319229	Bolivian hemorrhagic fever	1272	Brachycephaly-deafness-cataract-intellectual disability syndrome		
1842	Bone dysplasia, lethal Holmgren type	2619	Brachydactylyous dwarfism, Mseleni type		
85182	Bone dysplasia-medullary fibrosarcoma syndrome	93388	Brachydactyly type A1		
300284	Bone fragility-contractures-arterial rupture-deafness syndrome	93396	Brachydactyly type A2		
2050	Bone fragility-craniosynostosis-	93394	Brachydactyly type A4		
		93389	Brachydactyly type A5		
		93382	Brachydactyly type A6		
		93397	Brachydactyly type A7		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
441	Bradbury-Eggleston syndrome	783	Broad thumb-hallux syndrome	312	Bullous ichthyosis
52047	Braddock syndrome	783	Broad thumbs-halluces syndrome	36237	Bullous impetigo
3323	Braddock-Carey syndrome	412	Broad-betalipoproteinemia	33408	Bullous lichen planus
1538	Braddock-Jones-Superneau syndrome	53347	Brody myopathy	703	Bullous pemphigoid
75374	Bradyopsia	66529	Broken heart syndrome	→193	Buntinx-Lormans-Martin syndrome
178506	Brain calcification, Rajab type	97287	Bronchial NET	98976	Buphthalmia
168598	Brain demyelination due to methionine adenosyltransferase deficiency	97287	Bronchial neuroendocrine tumor	98976	Buphthalmos
352649	Brain dopamine-serotonin vesicular transport disease	→3471	Bronchiectasis-oligospermia syndrome	98976	Buphthalmus
75389	Brain malformation-congenital heart disease-postaxial polydactyly syndrome	1302	Bronchiolitis obliterans organizing pneumonia	543	Burkitt lymphoma
500150	Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	1303	Bronchiolitis obliterans with obstructive pulmonary disease	1200	Burn-McKeown syndrome
467166	Brain stem asymmetry-superior cerebellar and basal ganglia dysplasia syndrome	2357	Bronchogenic cyst	353253	Burning mouth syndrome
209905	Brain-lung-thyroid syndrome	70589	Bronchopulmonary dysplasia	800	Burton skeletal dysplasia
99084	Branch pulmonary artery stenosis	1116	Bronspiegel-Zelnick syndrome	800	Burton syndrome
255182	Branched chain alpha-ketoacid dehydrogenase complex deficiency	99829	Bronze John	352763	Buschke scleredema
511	Branched-chain 2-ketoacid dehydrogenase deficiency	79493	Brooke-Spiegler syndrome	79501	Buschke-Fischer-Brauer syndrome
511	Branched-chain ketoaciduria	97229	Brown-Vialetto-van Laere syndrome	1306	Buschke-Ollendorff syndrome
1296	Branchial dysplasia-intellectual disability-inguinal hernia syndrome	109	BRRS	99001	Butterfly-shaped pattern dystrophy
1297	Branchio-oculo-facial syndrome	2353	BRSS	99001	Butterfly-shaped pigment dystrophy
50815	Branchiogenic deafness syndrome	1304	Brucellosis	1307	Buttiens-Fryns syndrome
52429	Branchioototic syndrome	2771	Bruck syndrome	132	Butyrylcholinesterase deficiency
107	Branchiootorenal syndrome	130	Brugada syndrome	275864	bv-FTD
1299	Branchioskeletalgenital syndrome	3057	Brunner syndrome	1243	BVMD
79133	Brauer syndrome	1305	Brunner-Winter syndrome	116	BWS
2669	Braun-Bayer syndrome	391641	Brunner-Winter syndrome type 1	79306	Byler disease
319239	Brazilian hemorrhagic fever	391646	Brunner-Winter syndrome type 2	459353	C1 inhibitor deficiency
1059	BRBN	528	Brunzell syndrome	280133	C3 deficiency
1945	BRE	→528	Brunzell syndrome	→329931	C3 deposition glomerulonephritis without proliferation
85284	BRESEK syndrome	47	Bruton type agammaglobulinemia	329931	C3 glomerulonephritis
85284	BRESHECK syndrome	528	BSCL	329918	C3 glomerulopathy
65682	BRIC	79304	BSEP deficiency	401901	C9ORF72-related Huntington disease phenocopy
99960	BRIC1	1299	BSG syndrome	401901	C9ORF72-related Huntington disease-like syndrome
99961	BRIC2	100976	BSI	1308	C syndrome
99960	BRIC type 1	1980	BSPDC	231242	C-beta-thalassemia
99961	BRIC type 2	125	BSyn	97297	C-like syndrome
99990	Brill disease	65284	BTBGD	495844	C11ORF73-related autosomal recessive hypomyelinating leukodystrophy
99990	Brill-Zinsser disease	79241	BTD deficiency	495844	C11ORF73-related autosomal recessive hypomyelinating leukoencephalopathy
666	Brittle bone disease	111	BTHS	401948	CA-VA deficiency
90354	Brittle cornea syndrome	47	BTK-deficiency	85293	Cabezas syndrome
→33364	Brittle hair syndrome, Sabinas type	2314	Buckley syndrome	504476	CABV syndrome
→33364	Brittle hair-mental deficiency syndrome	131	Budd-Chiari syndrome	1309	Cacchi-Ricci disease
		36258	Buerger disease	75377	CACD
		2285	Bull-Nixon syndrome	135	CACH syndrome
		312	Bullous congenital ichthyosiform erythroderma	2848	CACP syndrome
		312	Bullous congenital ichthyosiform erythroderma of Brock	159	CACT deficiency
		280785	Bullous DCM		
		280785	Bullous diffuse cutaneous mastocytosis		
		1867	Bullous dystrophy, macular type		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
56425	CAD		clubfoot syndrome		deficiency
448010	CAD-CDG	1321	Camptodactyly-fibrous tissue hyperplasia-skeletal dysplasia syndrome	147	Carbamoyl-phosphate synthetase deficiency
136	CADASIL	1323	Camptodactyly-joint contractures-facial skeletal defects syndrome	147	Carbamoyl-phosphate synthetase I deficiency
369942	CADDS	3447	Camptodactyly-overgrowth-unusual facies syndrome	79328	Carbohydrate deficient glycoprotein syndrome type 1L
2566	CAEBV syndrome	85164	Camptodactyly-tall stature-scoliosis-hearing loss syndrome	79318	Carbohydrate deficient glycoprotein syndrome type 1a
1310	Caffey disease	1325	Camptodactyly-taurinuria syndrome	79319	Carbohydrate deficient glycoprotein syndrome type 1b
436174	CAGSSS	1766	CAMRQ syndrome	79320	Carbohydrate deficient glycoprotein syndrome type 1c
90791	CAH due to 3-beta-hydroxysteroid dehydrogenase deficiency	141194	CAMS1	79321	Carbohydrate deficient glycoprotein syndrome type 1d
90795	CAH due to 11-beta-hydroxylase deficiency	53719	CAMS2	79322	Carbohydrate deficient glycoprotein syndrome type 1e
90793	CAH due to 17-alpha-hydroxylase deficiency	141199	CAMS3	79323	Carbohydrate deficient glycoprotein syndrome type 1f
1375	CAHMR syndrome	3319	CAMT	79324	Carbohydrate deficient glycoprotein syndrome type 1g
435988	CAID syndrome	1328	Camurati-Engelmann disease	79325	Carbohydrate deficient glycoprotein syndrome type 1h
99429	CAIS	3261	Canale-Smith syndrome	79326	Carbohydrate deficient glycoprotein syndrome type 1i
289601	Calcification of joints and arteries	141	Canavan disease	397941	Carbohydrate deficient glycoprotein syndrome type II due to MAN1B1 deficiency
499182	Calcified epithelial carcinoma of Malherbe	289385	Cancer diagnosed during pregnancy	79329	Carbohydrate deficient glycoprotein syndrome type 1Ia
199260	Calcifying aponeurotic fibroma	180242	Cancer of fallopian tubes	79330	Carbohydrate deficient glycoprotein syndrome type 1Ib
499182	Calcifying epitheliocarcinoma	71505	Cancer-associated retinopathy	79332	Carbohydrate deficient glycoprotein syndrome type 1Id
90290	Calcinosis-Raynaud phenomenon-esophageal involvement-sclerodactyly-telangiectasia syndrome	2700	Cancrum oris	79333	Carbohydrate deficient glycoprotein syndrome type 1Ie
280062	Calciphylaxis	325004	CANDLE syndrome	238459	Carbohydrate deficient glycoprotein syndrome type 1If
280065	Calciphylaxis cutis	71279	CANOMAD syndrome	263508	Carbohydrate deficient glycoprotein syndrome type 1Ig
1416	Calcium pyrophosphate dihydrate crystal deposition disease	2233	Cantalamessa-Baldini-Ambrosi syndrome	95428	Carbohydrate deficient glycoprotein syndrome type 1Ih
1408	Calderón-González-Cantu syndrome	1335	Cantrell deformity	263487	Carbohydrate deficient glycoprotein syndrome type 1Ii
228123	California disease	1335	Cantrell syndrome	263501	Carbohydrate deficient glycoprotein syndrome type 1Ij
83483	Californian encephalitis	363705	Cantu craniofaciofrontodigital syndrome	314667	Carbohydrate deficient glycoprotein syndrome type 1Ik
289601	CALJA	504476	CANVAS	468699	Carbohydrate deficient glycoprotein syndrome type 1In
85192	Calvarial doughnut lesions-bone fragility syndrome	171881	Cap disease	468684	Carbohydrate deficient glycoprotein syndrome type 1Io
→1466	CAMAK syndrome	160148	Cap inflammatory polyposis	466703	Carbohydrate deficient glycoprotein syndrome type 1Ip
3003	Camera syndrome	171881	Cap myopathy	86309	Carbohydrate deficient glycoprotein syndrome type 1Ij
2163	Camero-Lituania-Cohen syndrome	160148	Cap polyposis		
→1466	CAMFAK syndrome	85199	CAP syndrome		
79395	Camisa disease	166260	Capdepont teeth		
83472	CAMOS syndrome	75327	CAPE dystrophy		
1318	Campomelia, Cumming type	75327	CAPED		
140	Campomelic dwarfism	188	Capillary hyperpermeability syndrome		
140	Campomelic dysplasia	188	Capillary leak syndrome		
1319	Camptobrachydactyly	79490	Capillary lymphangioma		
295016	Camptodactyly of fingers	79490	Capillary lymphatic malformation		
1327	Camptodactyly syndrome, Guadalajara type 1	137667	Capillary malformation-arteriovenous malformation		
1326	Camptodactyly syndrome, Guadalajara type 2	1171	CAPOS syndrome		
488434	Camptodactyly syndrome, Guadalajara type 3	171839	Capra-DeMarco syndrome		
2848	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	464343	CAPS		
376	Camptodactyly-cleft palate-	71505	CAR syndrome		
		199354	CARASIL		
		147	Carbamoyl-phosphate synthetase 1		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
79327	Carbohydrate deficient glycoprotein syndrome type I κ	70474	Cardiomyopathy with myopathy due to COX deficiency	228305	Carnitine palmitoyl transferase II deficiency, hepatocardiomuscular form
91131	Carbohydrate deficient glycoprotein syndrome type I μ	1345	Cardiomyopathy-cataract-hip spine disease syndrome	228308	Carnitine palmitoyl transferase II deficiency, lethal systemic form
244310	Carbohydrate deficient glycoprotein syndrome type I η	91130	Cardiomyopathy-hypotonia-lactic acidosis syndrome	228302	Carnitine palmitoyl transferase II deficiency, myopathic form
263494	Carbohydrate deficient glycoprotein syndrome type I δ	90022	Cardiomyopathy-renal anomalies syndrome	228308	Carnitine palmitoyl transferase II deficiency, neonatal form
280071	Carbohydrate deficient glycoprotein syndrome type I ρ	111	Cardioskeletal myopathy with neutropenia and abnormal mitochondria	228305	Carnitine palmitoyl transferase II deficiency, severe infantile form
300536	Carbohydrate deficient glycoprotein syndrome type I τ	111	Cardioskeletal myopathy-neutropenia syndrome	157	Carnitine palmitoyltransferase deficiency type 2
329178	Carbohydrate deficient glycoprotein syndrome type I μ	3238	Cardiospondylocarpofacial syndrome	157	Carnitine palmitoyltransferase II deficiency
370924	Carbohydrate deficient glycoprotein syndrome type I χ	2072	Cardiovascular Gaucher disease	158	Carnitine transporter defect
370927	Carbohydrate deficient glycoprotein syndrome type I γ	1358	Carey-Fineman-Ziter syndrome	158	Carnitine uptake deficiency
448010	Carbohydrate deficient glycoprotein syndrome type I ζ	79403	Carmi syndrome	159	Carnitine-acylcarnitine translocase deficiency
306686	Carbon monoxide-induced parkinsonism	→293843	Carnevale syndrome	1361	Carnosinase deficiency
2785	Carbonic anhydrase 2 deficiency	2947	Carnevale-Hernández-del Castillo syndrome	1361	Carnosinemia
213605	Carcinofibroma of the corpus uteri	→293843	Carnevale-Krajewska-Fischetto syndrome	53035	Caroli disease
100093	Carcinoid syndrome	1359	Carney complex	480520	Caroli syndrome
319308	Carcinoma associated with MITF/TFE translocation	319340	Carney complex variant	65759	Carpenter syndrome
418945	Carcinoma of esophagus, salivary gland type	319340	Carney complex-trismus-pseudocamptodactyly syndrome	93973	Carpenter-Waziri syndrome
423781	Carcinoma of stomach, salivary gland type	97286	Carney dyad	2767	Carpotarsal osteochondromatosis
300557	Carcinoma of the ampulla of Vater	1359	Carney syndrome	64692	Carrion disease
213787	Carcinosarcoma of the cervix uteri	139411	Carney triad	175	Cartilage-hair hypoplasia
213610	Carcinosarcoma of the corpus uteri	97286	Carney-Stratakis dyad	→175	Cartilage-hair hypoplasia-like-skeletal dysplasia without hypotrichosis syndrome
369891	Cardiac anomalies-developmental delay-facial dysmorphism syndrome	97286	Carney-Stratakis syndrome	65282	Carvajal syndrome
137628	Cardiac anomalies-heterotaxy syndrome	42	Carnitine deficiency secondary to medium-chain acyl-CoA dehydrogenase deficiency	209908	CAS
168796	Cardiac conduction disease-dilated cardiomyopathy-brachydactyly syndrome	156	Carnitine palmitoyl transferase 1A deficiency	56425	CAS
1686	Cardiac diverticulum	228302	Carnitine palmitoyl transferase deficiency type 2, adult-onset form	94095	Casamassima-Morton-Nance syndrome
875	Cardiac tumor of child	228305	Carnitine palmitoyl transferase deficiency type 2, hepatocardiomuscular form	275517	Caspase 8 deficiency syndrome
2872	Cardiocranial syndrome, Pfeiffer type	228308	Carnitine palmitoyl transferase deficiency type 2, lethal systemic form	1101	Cassia Stocco dos Santos syndrome
37553	Cardiodysrhythmic potassium-sensitive periodic paralysis	228302	Carnitine palmitoyl transferase deficiency type 2, myopathic form	160	Castleman disease
1340	Cardiofaciocutaneous syndrome	228308	Carnitine palmitoyl transferase deficiency type 2, neonatal form	2513	Castro Gago-Pombo-Novó syndrome
97292	Cardiogenic shock	228305	Carnitine palmitoyl transferase deficiency type 2, severe infantile form	195	Cat-eye syndrome
2229	Cardiogenital syndrome	156	Carnitine palmitoyl transferase IA deficiency	50839	Cat-scratch disease
1342	Cardiomelic syndrome type 3	228302	Carnitine palmitoyl transferase II deficiency, adult-onset form	926	Catalase deficiency
500	Cardiomyopathic lentiginosis	1373	Cataract-aberrant oral frenula-growth delay syndrome	1366	Cataract-alopelia-sclerodactyly syndrome
70474	Cardiomyopathy with hypotonia due to cytochrome C oxidase deficiency	1368	Cataract-ataxia-deafness syndrome	314993	Cataract-congenital heart disease-neural tube defect syndrome
		314993	Cataract-deafness-hypogonadism syndrome	1383	Cataract-deafness-hypogonadism syndrome
		162	Cataract-glucoma syndrome	436174	Cataract-growth hormone

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	79283	CblD defect	79327	CDG1K
		79284	CblF defect	79328	CDG1L
		369955	CblJ defects	91131	CDG1M
1375	Cataract-hypertrichosis-intellectual disability syndrome	70567	CCA	244310	CDG1N
1381	Cataract-intellectual disability-anal atresia-urinary defects syndrome	115	CCA syndrome	263494	CDG1O
1387	Cataract-intellectual disability-hypogonadism syndrome	2444	CCAM	280071	CDG1P
→1466	Cataract-microcephaly-arthrogryposis-kyphosis syndrome	280832	CCAM type 1	324737	CDG1Q
→1466	Cataract-microcephaly-failure to thrive-kyphoscoliosis syndrome	280840	CCAM type 2	300536	CDG1R
1377	Cataract-microcornea syndrome	280847	CCAM type 3	324422	CDG1S
2712	Cataract-microphtalmia-radiculomegaly-cardiac septal defect syndrome	468684	CCDC115-CDG	319646	CDG1T
1380	Cataract-nephropathy-encephalopathy syndrome	98972	CCDF	329178	CDG1U
100990	Cataracts-motor neuropathy-short stature-skeletal anomalies syndrome	48431	CCFDN	370921	CDG1W
464343	Catastrophic antiphospholipid syndrome	2008	CCGE syndrome	370924	CDG1X
464343	Catastrophic APS	99827	CCHF	370927	CDG1Y
567	CATCH 22	661	CCHS	448010	CDG1Z
3286	Catecholaminergic polymorphic ventricular tachycardia	289499	CCMCO	79329	CDG2A
800	Catel-Hempel syndrome	319276	CCRCC	79330	CDG2B
1388	Catel-Manzke syndrome	2505	CCSF	99843	CDG2C
228337	Cathepsin D deficiency	457246	CCSK	79332	CDG2D
60015	Catlin marks	280779	CCV	79333	CDG2E
85164	CATSHL syndrome	86870	CD4+/CD56+ hematodermic neoplasm	238459	CDG2F
1123	Caudal appendage-deafness syndrome	437552	CD16 deficiency	263508	CDG2G
1756	Caudal duplication	238505	CD27 deficiency	95428	CDG2H
3027	Caudal dysplasia	98841	CD30 positive anaplastic large cell lymphoma	263487	CDG2I
3027	Caudal regression sequence	293825	CDA due to KLF1 mutation	263501	CDG2J
99994	Causalgia	98869	CDA I	314667	CDG2K
1329	CAVC	98873	CDA II	464443	CDG2L
99068	CAVC-Fallot tetralogy syndrome	98870	CDA III	356961	CDG2M
99066	CAVC-left heart obstruction syndrome	293825	CDA IV	468699	CDG2N
99067	CAVC-ventricle hypoplasia syndrome	98869	CDA type 1	468684	CDG2O
2124	Cavernous hemangiomas of face-supraumbilical midline raphe syndrome	98873	CDA type 2	466703	CDG2P
79489	Cavernous lymphangioma	98870	CDA type 3	79318	CDG syndrome type Ia
79489	Cavernous lymphatic malformation	293825	CDA type 4	79319	CDG syndrome type Ib
165958	Cavitory myiasis	98869	CDA type I	79320	CDG syndrome type Ic
567	Cayler cardiofacial syndrome	98873	CDA type II	79321	CDG syndrome type Id
94122	Cayman ataxia	98870	CDA type III	79322	CDG syndrome type Ie
363972	CBL syndrome	293825	CDA type IV	79323	CDG syndrome type If
79282	CblC defect	85199	CDAGS syndrome	79324	CDG syndrome type Ig
		293825	CDAN4	79325	CDG syndrome type Ih
		247203	CDC	79326	CDG syndrome type IIi
		163681	CDFE syndrome	79329	CDG syndrome type IIa
		163681	CDFES	79330	CDG syndrome type IIb
		79318	CDG1A	99843	CDG syndrome type IIc
		79319	CDG1B	79332	CDG syndrome type IId
		79320	CDG1C	79333	CDG syndrome type IIe
		79321	CDG1D	238459	CDG syndrome type IIff
		79322	CDG1E	263508	CDG syndrome type IIg
		79323	CDG1F	95428	CDG syndrome type IIh
		79324	CDG1G	263487	CDG syndrome type IIIi
		79325	CDG1H	263501	CDG syndrome type IIj
		79326	CDG1I	314667	CDG syndrome type IIk
		86309	CDG1J	464443	CDG syndrome type IIIl
				356961	CDG syndrome type IIIm

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
468699	CDG syndrome type IIn	324422	CDG-Is	637	Central neurofibromatosis
468684	CDG syndrome type Ilo	319646	CDG-It	295004	Central polydactyly
466703	CDG syndrome type IIp	329178	CDG-Iu	759	Central precocious puberty
86309	CDG syndrome type Ij	370921	CDG-Iw	75327	Central retinal pigment epithelial dystrophy
79327	CDG syndrome type Ik	370924	CDG-Ix	411527	Central retinal vein occlusion
79328	CDG syndrome type IL	370927	CDG-Iy	443079	Central serous chorioretinopathy
91131	CDG syndrome type Im	448010	CDG-Iz	90156	Centrifugal lipodystrophy
244310	CDG syndrome type In	2140	CDH	89841	Centripetal dystrophic epidermolysis bullosa
263494	CDG syndrome type Io	1529	CDHS	89841	Centripetal recessive dystrophic epidermolysis bullosa
280071	CDG syndrome type Ip	178029	CDI	89841	Centripetalis recessive dystrophic epidermolysis bullosa
324737	CDG syndrome type Iq	505652	CDKL5 deficiency	319160	Centronuclear myopathy type 4
300536	CDG syndrome type Ir	505652	CDKL5 deficiency disorder	1945	Centrotemporal epilepsy
324422	CDG syndrome type Is	505652	CDKL5 disorder	79277	CEP
319646	CDG syndrome type It	505652	CDKL5-related epileptic encephalopathy	2398	Cephalothoracic lipodystrophy
329178	CDG syndrome type Iu	3194	CDO syndrome	79506	CEPT deficiency
370921	CDG syndrome type Iw	1490	CDPD	276183	Cerebellar ataxia with azoospermia and intellectual disability
370924	CDG syndrome type Ix	35173	CDPX2	504476	Cerebellar ataxia with bilateral vestibulopathy syndrome
370927	CDG syndrome type Iy	35173	CDPXD	504476	Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome
448010	CDG syndrome type Iz	158	CDSP	94122	Cerebellar ataxia, Cayman type
79318	CDG-Ia	468641	CEAS	1171	Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome
79319	CDG-Ib	1459	CEC	1174	Cerebellar ataxia-ectodermal dysplasia syndrome
79320	CDG-Ic	2718	Cecato de Lima-Pinheiro syndrome	1173	Cerebellar ataxia-hypogonadism syndrome
79321	CDG-ID	1515	CED	1766	Cerebellar ataxia-intellectual disability-dysequilibrium syndrome
79322	CDG-Ie	66631	CEDNIK syndrome	83472	Cerebellar ataxia-intellectual disability-optic atrophy-skin abnormalities syndrome
79323	CDG-If	275517	CEDS	97249	Cerebellar atrophy with progressive microcephaly
79324	CDG-Ig	293208	Celiac artery compression syndrome	2246	Cerebellar hypoplasia-tapetoretinal degeneration syndrome
79325	CDG-Ih	1459	Celiac disease-epilepsy-cerebral calcification syndrome	251931	Cerebellar liponeurocytoma
79326	CDG-Ii	93942	Celosomia	94147	Cerebellar syndrome-pigmentary maculopathy syndrome
79329	CDG-IIa	3258	Cenani syndactyly	1454	Cerebellar vermis hypoplasia-oligophrenia-congenital ataxia-coloboma-hepatic fibrosis
79330	CDG-IIb	3258	Cenani-Lenz syndactyly	444072	Cerebellar-facial-dental syndrome
99843	CDG-IIc	75377	Cenani-Lenz syndrome	444072	Cerebellofaciodental syndrome
79332	CDG-IIId	75377	Central areolar choroidal dystrophy	2318	Cerebellooculorenal syndrome
79333	CDG-IIle	75327	Central areolar choroidal sclerosis	475	Cerebelloparenchymal disorder IV
238459	CDG-IIIf	2431	Central areolar pigment epithelial dystrophy	1532	Cerebellotrigeminal-dermal
263508	CDG-IIlg	98972	Central bilateral macrogryria		
95428	CDG-IIlh	98972	Central cloudy corneal dystrophy of François		
263487	CDG-IIli	98972	Central cloudy dystrophy of François		
263501	CDG-IIlj	661	Central congenital hypoventilation syndrome		
314667	CDG-IIlk	597	Central core disease		
464443	CDG-IIIL	178029	Central diabetes insipidus		
356961	CDG-IIIm	→98967	Central discoid corneal dystrophy		
468699	CDG-IIn	99832	Central hypothyroidism due to TRH receptor deficiency		
468684	CDG-Ilo	3240	Central nervous system calcification-deafness-tubular acidosis-anemia syndrome		
466703	CDG-IIp	73256	Central neurocytoma		
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				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	dysplasia syndrome	213828	Cervical adenoid basal carcinoma	101081	Charcot-Marie-Tooth disease type 1A
46724	Cerebral arteriovenous malformation	213823	Cervical adenoid cystic carcinoma	101082	Charcot-Marie-Tooth disease type 1B
136	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy	213792	Cervical adenosarcoma	101083	Charcot-Marie-Tooth disease type 1C
199354	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy	99079	Cervical aortic arch	101084	Charcot-Marie-Tooth disease type 1D
66631	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome	213787	Cervical carcinosarcoma	90658	Charcot-Marie-Tooth disease type 1E
821	Cerebral gigantism	141046	Cervical dermoid cyst	101085	Charcot-Marie-Tooth disease type 1F
→1900	Cerebral gigantism, Nevo type	213837	Cervical germ cell cancer	98856	Charcot-Marie-Tooth disease type 2B1
2081	Cerebral gigantism-jaw cysts syndrome	2218	Cervical hypertrichosis-peripheral neuropathy syndrome	101101	Charcot-Marie-Tooth disease type 2B2
77261	Cerebral juvenile and adult form of Gaucher disease	213807	Cervical leiomyosarcoma	228374	Charcot-Marie-Tooth disease type 2B5
221126	Cerebral proliferative glomeruloid vasculopathy	213837	Cervical malignant germ cell tumor	101102	Charcot-Marie-Tooth disease type 2H
329217	Cerebral sinovenous thrombosis	213787	Cervical malignant Müllerian mixed tumor	300319	Charcot-Marie-Tooth disease type 2P
447788	Cerebral visual impairment	213812	Cervical malignant peripheral neuroectodermal tumor	397968	Charcot-Marie-Tooth disease type 2R
397922	Cerebro-cutaneous syndrome with iron overload	213817	Cervical papillary carcinoma	443073	Charcot-Marie-Tooth disease type 2S
→2995	Cerebro-oculo-facial-lymphatic syndrome	213812	Cervical peripheral neuroectodermal cancer	495274	Charcot-Marie-Tooth disease type 2T
1393	Cerebrocostomandibular syndrome	213802	Cervical rhabdomyosarcoma	64748	Charcot-Marie-Tooth disease type 3
141194	Cerebrofacial arteriovenous metameric syndrome type 1	268392	Cervical spina bifida aperta	99948	Charcot-Marie-Tooth disease type 4A
53719	Cerebrofacial arteriovenous metameric syndrome type 2	268762	Cervical spina bifida cystica	99955	Charcot-Marie-Tooth disease type 4B1
141199	Cerebrofacial arteriovenous metameric syndrome type 3	213767	Cervical squamous cell carcinoma	99956	Charcot-Marie-Tooth disease type 4B2
314679	Cerebrofacioarticular syndrome	141067	Cervicofacial enchondroma	363981	Charcot-Marie-Tooth disease type 4B3
1394	Cerebrofaciothoracic dysplasia	141067	Cervicofacial fibrochondroma	99949	Charcot-Marie-Tooth disease type 4C
912	Cerebrohepatorenal syndrome	3456	Cervicooculoacoustic syndrome	99950	Charcot-Marie-Tooth disease type 4D
2406	Cerebromedullospinal disconnection	268397	Cervicothoracic spina bifida aperta	99951	Charcot-Marie-Tooth disease type 4E
1458	Cerebrooculodentoauriculoskeletal syndrome	268766	Cervicothoracic spina bifida cystica	99952	Charcot-Marie-Tooth disease type 4F
1466	Cerebrooculofacioskeletal syndrome	195	CES	99953	Charcot-Marie-Tooth disease type 4G
66625	Cerebrooculonasal syndrome	231573	CEVD	99954	Charcot-Marie-Tooth disease type 4H
505242	Cerebrorenal syndrome, Perez type	586	CF	139515	Charcot-Marie-Tooth disease type 4J
1396	Cerebrorenodigital syndrome	2032	CFA	391351	Charcot-Marie-Tooth disease type 4K
313838	Cerebroretinal microangiopathy with calcifications and cysts	1340	CFC syndrome	90120	Charcot-Marie-Tooth disease type 6
→247691	Cerebroretinal vasculopathy	1520	CFND	363981	Charcot-Marie-Tooth disease with focally folded myelin
909	Cerebrotendinous xanthomatosis	1520	CFNS		
1980	Cerebrovascular ferrocalcinosis	2020	CFTDM		
169079	Cernunnos deficiency	379	CGD		
169079	Cernunnos XLF	2026	CGHT		
169079	Cernunnos-XLF deficiency	2388	ChAc		
98989	Cerulean cataract	307766	CHAC syndrome		
213772	Cervical adenocarcinoma	307766	CHACS		
		3386	Chagas disease		
		436159	CHAI		
		→1071	CHAND syndrome		
		98979	Chandler syndrome		
		→1071	CHANDS		
		2235	Chang-Davidson-Carlson syndrome		
		88642	Channelopathy-associated CIP		
		88642	Channelopathy-associated congenital insensitivity to pain		
		3282	Chaotic atrial tachycardia		
		319244	Chapare hemorrhagic fever		
		46627	Char syndrome		
		1964	Char-Douglas-Dungan syndrome		
		803	Charcot disease		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
90658	Charcot-Marie-Tooth disease-deafness syndrome	363677	Childhood-onset autosomal recessive myopathy with external ophthalmoplegia		oxosteroid 5-beta-reductase deficiency
90103	Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome	284324	Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia	1414	Cholestasis-lymphedema syndrome
93114	Charcot-Marie-Tooth disease-nephropathy syndrome	497906	Childhood-onset basal ganglia degeneration syndrome	1415	Cholestasis-pigmentary retinopathy-cleft palate syndrome
64751	Charcot-Marie-Tooth disease-pyramidal features syndrome	494541	Childhood-onset benign chorea with striatal involvement	→2697	Cholestatic jaundice-renal tubular insufficiency syndrome
138	CHARGE association	487809	Childhood-onset collagenous gastritis	75234	Cholesterol ester storage disease
138	CHARGE syndrome	508093	Childhood-onset generalized dystonia-optic atrophy syndrome	79506	Cholesterol-ester transfer protein deficiency
921	CHARGE-like syndrome	33402	Childhood-onset HCC	75234	Cholesteryl ester storage disease
1496	Charlevoix disease	33402	Childhood-onset hepatocellular carcinoma	79344	Chondrodysplasia punctata, Sheffield type
1406	Charlie M syndrome	247667	Childhood-onset hypophosphatasia	79346	Chondrodysplasia punctata, tibial-metacarpal type
168577	CHC type 2	500180	Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	79347	Chondrodysplasia punctata, Toriello type
98975	CHED1	171439	Childhood-onset nemaline myopathy	263463	Chondrodysplasia with congenital joint dislocations, CHST3 type
293603	CHED2	247667	Childhood-onset phosphoethanolaminuria	280586	Chondrodysplasia with joint dislocations, gPAPP type
98975	CHEDI	466921	Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome	3144	Chondrodysplasia with snail-like pelvis
167	Chédiak-Higashi disease	247667	Childhood-onset Rathburn disease	50945	Chondrodysplasia, Blomstrand type
167	Chédiak-Higashi syndrome	101000	Childhood-onset spastic paraparesis-distal muscle wasting syndrome	2098	Chondrodysplasia, Grebe type
381	Chédiak-Higashi-like syndrome	401866	Childhood-onset spasticity with hyperglycinemia	166272	Chondrodysplasia-dentinogenesis imperfecta-joint laxity syndrome
167	Chédiak-Higashi-Steinbrink syndrome	401866	Childhood-onset spasticity with variant non-ketotic hyperglycinemia	1422	Chondrodysplasia-disorder of sex development syndrome
293603	CHEDII	3474	CHIME syndrome	1422	Chondrodysplasia-pseudohermaphroditism syndrome
1221	Cheilitis glandularis	2888	Chitayat-Meunier-Hodgkinson syndrome	35173	Chondrodstrophia calcificans congenita
99647	Cheirospolyloenchondromatosis	3218	Chitty-Hall-Baraitser syndrome	289	Chondroectodermal dysplasia
955	Cheney syndrome	757	Chloride shunt syndrome	319195	Chondroectodermal dysplasia with night blindness
812	Cherry-red spot-myoclonus syndrome	86850	Chloroma	404507	Chondromyxoid fibroma
184	Cherubism	180	CHM	55880	Chondrosarcoma
3019	Cherubism-gingival fibromatosis-intellectual disability syndrome	1434	CHM-hypopituitarism syndrome	444077	CHOPS syndrome
→672	CHHS	137914	Choanal atresia	251674	Chordoid glioma
1398	Chiari 4 malformation	137920	Choanal atresia, bilateral	178	Chordoma
1398	Chiari IV malformation	137917	Choanal atresia, unilateral	2388	Chorea-acanthocytosis
268882	Chiari malformation type 1	1200	Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome	2388	Choreoacanthocytosis
1136	Chiari malformation type 2	70567	Cholangiocarcinoma	209905	Choreoathetosis-hypothyroidism-neonatal respiratory distress syndrome
268882	Chiari malformation type I	480501	Choledochal cyst	252015	Choriocarcinoma of the central nervous system
1136	Chiari malformation type II	69663	Cholelithiasis with ABCB4 gene mutation	251899	Choroid plexus carcinoma
324625	Chikungunya	173	Cholera	2807	Choroid plexus papilloma
90280	Chilblain lupus	79303	Cholestasis with delta(4)-3-	1433	Choroidal atrophy-alopecia syndrome
139	CHILD nevus			39044	Choroidal melanoma
139	CHILD syndrome			180	Choroideremia
64280	Childhood absence epilepsy			1435	Choroideremia-deafness-obesity
439175	Childhood AIS				
209908	Childhood apraxia of speech				
439175	Childhood arterial ischemic stroke				
135	Childhood ataxia with diffuse central nervous system hypomyelination				
168782	Childhood disintegrative disorder				
293955	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency				
391497	Childhood myasthenia gravis				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	syndrome	2566	Chronic Epstein-Barr virus infection syndrome	83418	Chronic spinal muscular atrophy
1434	Choroideremia-hypopituitarism syndrome	99921	Chronic graft versus host disease	70591	Chronic thromboembolic pulmonary hypertension
94087	CHP	521	Chronic granulocytic leukemia	97353	Chronic traumatic encephalopathy
440727	CHR-RPE	379	Chronic granulomatous disease	37748	Chronic urticaria with gammopathy
181	Christ-Siemens-Touraine syndrome	396	Chronic hiccup	37748	Chronic urticaria with macroglobulinemia
1436	Christian syndrome	1451	Chronic infantile neurological cutaneous articular syndrome	263463	CHST3-related skeletal dysplasia
85278	Christianson syndrome	83418	Chronic infantile spinal muscular atrophy	93971	Chudley-Lowry syndrome
1808	Christianson-Fourie syndrome	2932	Chronic inflammatory demyelinating polyneuropathy	93971	Chudley-Lowry-Hoar syndrome
98879	Christmas disease	2932	Chronic inflammatory demyelinating polyradiculoneuropathy	314597	Chudley-McCullough syndrome
182	Chromoblastomycosis	294422	Chronic intestinal failure	3068	Chudley-Rozdilsky syndrome
182	Chromomycosis	2978	Chronic intestinal pseudoobstruction	183	Churg-Strauss syndrome
319303	Chromophobe renal cell adenocarcinoma	284448	Chronic lymphocytic inflammation with pontine perivascular enhancement responsive to steroids	238557	Chuvash erythrocytosis
319303	Chromophobe renal cell carcinoma	1334	Chronic mucocutaneous candidiasis	238557	Chuvash polycythemia
3380	Chromosome 18 duplication	99873	Chronic multifocal Langerhans cell histiocytosis	71	Chylomicron retention disease
1445	Chromosome 21 en anneau	521	Chronic myelogenous leukemia	1160	Chylous ascites
330064	Chronic actinic dermatitis	521	Chronic myeloid leukemia	46486	Cicatricial pemphigoid
314928	Chronic adult hydrocephalus	98823	Chronic myelomonocytic leukemia	217390	CID due to DOCK8 deficiency
99871	Chronic and localized Langerhans cell histiocytosis	86830	Chronic myeloproliferative disease, unclassifiable	505227	CID due to GINS1 deficiency
137817	Chronic arachnoiditis	77261	Chronic neuronopathic Gaucher disease	317473	CID due to IKAROS deficiency
71279	Chronic ataxic neuropathy-ophthalmoplegia-IgM paraprotein-cold agglutinins-disialosyl antibodies syndrome	86829	Chronic neutrophilic leukemia	445018	CID due to LRBA deficiency
435988	Chronic atrial and intestinal dysrhythmia syndrome	324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	317476	CID due to MAGT1 deficiency
435988	Chronic atrial dysrhythmia-intestinal motility disorder	439202	Chronic obstetric brachial plexus injury	504530	CID due to Moesin deficiency
325004	Chronic atypical neutrophilic dermatosis-lipodystrophy-elevated temperature syndrome	439202	Chronic obstetric brachial plexus palsy	317428	CID due to ORAI1 deficiency
2137	Chronic autoimmune hepatitis	95426	Chronic pain requiring intraspinal analgesia	231154	CID due to partial RAG1 deficiency
133	Chronic berylliosis	330064	Chronic photosensitivity dermatitis	443811	CID due to PGM3 deficiency
133	Chronic beryllium disease	91359	Chronic pneumonitis of infancy	157949	CID due to RAG 1/2 deficiency
133	Chronic beryllium lung disease	499085	Chronic recurrent isolated optic neuritis	317430	CID due to STIM1 deficiency
56425	Chronic cold agglutinin disease	77297	Chronic recurrent multifocal osteomyelitis-congenital dyserythropoietic anemia-neutrophilic dermatosis syndrome	314689	CID due to STK4 deficiency
79078	Chronic dacryoadenitis and sialadenitis	499085	Chronic relapsing inflammatory optic neuropathy	476113	CID due to TFRC deficiency
103907	Chronic diarrhea due to glucoamylase deficiency	217566	Chronic respiratory distress with surfactant metabolism deficiency	231154	CID with expansion of gamma delta T cells
314373	Chronic diarrhea due to guanylate cyclase 2C overactivity	71279	Chronic sensory ataxic neuropathy with anti-dygalosyl IgM antibodies	436252	CID-MIA/early-onset IBD
397606	Chronic diarrhea with hereditary sensory and autonomic neuropathy	379	Chronic septic granulomatosis	435651	CIDEC-related familial partial lipodystrophy
397606	Chronic diarrhea with HSAN			435651	CIDEC-related FPLD
1670	Chronic diarrhea with villous atrophy			2932	CIDP
2566	Chronic EBV infection syndrome			79394	CIE
279891	Chronic endophthalmitis			294422	CIF
468641	Chronic enteropathy associated with SLCO2A1 gene			1223	Ciliary dysentery
168940	Chronic eosinophilic leukemia			2114	Cilliers-Beighton syndrome
2902	Chronic eosinophilic pneumonia			1451	CINCA syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
157820	CISS	98845	Classic Hodgkin lymphoma, lymphocyte-rich type	508476	Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-hearing loss syndrome
247525	Citrullinemia type 1	98844	Classic Hodgkin lymphoma, mixed cellularity type	199306	Cleft lip-alveolus-palate syndrome
247585	Citrullinemia type 2	98843	Classic Hodgkin lymphoma, nodular sclerosis type	1995	Cleft lip-cone rod dystrophy syndrome
247525	Citrullinemia type I	394	Classic homocystinuria	3429	Cleft lip-limb and heart malformations syndrome
247585	Citrullinemia type II	475	Classic Joubert syndrome	1995	Cleft lip-progressive retinopathy syndrome
251383	CK syndrome	313	Classic lamellar ichthyosis	1995	Cleft lip-retinopathy syndrome
508476	CL/P-cor triatriatum sinister syndrome	98964	Classic lattice corneal dystrophy	199306	Cleft lip/palate
90790	CLAH	268145	Classic maple syrup urine disease	888	Cleft lip/palate with mucous cysts of lower lip
97249	CLAM	158796	Classic mast cell leukemia	2319	Cleft lip/palate-abnormal thumbs-microcephaly syndrome
168984	CLAPO syndrome	251867	Classic medulloblastoma	508476	Cleft lip/palate-cor triatriatum sinister syndrome
188	Clarkson disease	324604	Classic MmD	2003	Cleft lip/palate-deafness-sacral lipoma syndrome
466026	Class I G6PD deficiency	268145	Classic MSUD	3253	Cleft lip/palate-ectodermal dysplasia syndrome
466026	Class I glucose-6-phosphate dehydrogenase deficiency	324604	Classic multiminicore disease	→1896	Cleft lip/palate-ectrodactyly syndrome
90794	Classic 21-OHD CAH	324604	Classic multiminicore myopathy	2328	Cleft lip/palate-facial, eye, heart and intestinal anomalies syndrome
315306	Classic 21-OHD CAH, salt wasting form	2584	Classic mycosis fungoides	2001	Cleft lip/palate-intestinal malrotation-cardiopathy syndrome
315311	Classic 21-OHD CAH, simple virilizing form	329977	Classic neuroendocrine tumor of appendix	3253	Cleft lip/palate-syndactyly-pili torti syndrome
85138	Classic Addison's disease	216866	Classic pantothenate kinase-associated neurodegeneration	95465	Cleft mitral valve
329977	Classic appendiceal neuroendocrine tumor	163898	Classic paraneoplastic limbic encephalitis	141242	Cleft nose
329977	Classic appendix neuroendocrine tumor	163898	Classic paraneoplastic limbic encephalitis, with or without intracellular antigens	2008	Cleft palate-cardiac defect-genital anomalies-ectrodactyly syndrome
93605	Classic Bartter syndrome	93258	Classic Pfeiffer syndrome	921	Cleft palate-coloboma-deafness syndrome
268145	Classic BCKD deficiency	79254	Classic phenylketonuria	2013	Cleft palate-large ears-small head syndrome
268145	Classic branched-chain alpha-ketoacid dehydrogenase deficiency	79254	Classic PKU	2016	Cleft palate-lateral synchia syndrome
268145	Classic branched-chain ketoaciduria	280219	Classic PMD	2167	Cleft palate-Potter sequence-congenital heart anomalies-mesoaxial polydactyly-multiple malformations syndrome
247525	Classic citrullinemia	240071	Classic progressive supranuclear palsy syndrome	2015	Cleft palate-short stature-vertebral anomalies syndrome
247546	Classic citrullinemia type 1	240071	Classic PSP syndrome	2010	Cleft palate-stapes fixation-oligodontia syndrome
247546	Classic citrullinemia type I	773	Classic Refsum disease	99772	Cleft soft palate
325524	Classic CLAH	18	Classic RTA	99772	Cleft velum
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	443192	Classic SPS	99772	Cleft velum palatinum
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	443192	Classic stiff person syndrome	1997	Clefting-ectropion-conical teeth syndrome
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	3467	Classic xanthinuria	1452	Cleidocranial dysostosis
325524	Classic congenital lipid adrenal hyperplasia due to STAR deficiency	2272	Clayton Smith-Donnai syndrome	1452	Cleidocranial dysplasia
93930	Classic extrophy of the bladder	485350	CLCN4-related X-linked intellectual disability syndrome	3472	Cleidocranial dysplasia-
79239	Classic galactosemia	398971	Clear cell adenocarcinoma of the ovary		
98962	Classic GCD	404511	Clear cell papillary renal cell carcinoma		
289857	Classic glycine encephalopathy	319276	Clear cell renal carcinoma		
98962	Classic granular corneal dystrophy	319276	Clear cell renal cell adenocarcinoma		
58017	Classic hairy cell leukemia	457246	Clear cell sarcoma of kidney		
391	Classic Hodgkin disease	97338	Clear cell sarcoma of the tendons and aponeuroses		
391	Classic Hodgkin lymphoma	101023	Cleft hard palate		
98846	Classic Hodgkin lymphoma, lymphocyte-depleted type	141291	Cleft lip and alveolus		
		199306	Cleft lip and palate		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	micrognathia-absent thumbs syndrome	99763	CMO I	99952	CMT4F
1453	Cleidiorhizomelic syndrome	99763	CMO II	99953	CMT4G
284448	CLIPPERS	238459	CMP-sialic acid transporter deficiency	99954	CMT4H
228329	CLN1 disease	86830	CMPD-U	139515	CMT4J
228349	CLN2 disease	71	CMRD	391351	CMT4K
228346	CLN3 disease	590	CMS	101078	CMT4X
228340	CLN4A disease	101081	CMT1A	99014	CMT5X
228343	CLN4B disease	101082	CMT1B	90120	CMT6
228360	CLN5 disease	101083	CMT1C	352675	CMT6X
228363	CLN6 disease	101084	CMT1D	1556	CMTC
228366	CLN7 disease	90658	CMT1E	100043	CMTDIA
228354	CLN8 disease	101085	CMT1F	100044	CMTDIB
1947	CLN8 disease, Northern epilepsy variant	101075	CMT1X	100045	CMTDIC
228357	CLN9 disease	487814	CMT2 due to DGAT2 mutation	100046	CMTDID
228337	CLN10 disease	324611	CMT2 due to KIF5A mutation	93114	CMTDIE
314629	CLN11 disease	435819	CMT2 due to TFG mutation	352670	CMTDIF
314632	CLN12 disease	435387	CMT2 due to VCP mutation	101075	CMTX1
352709	CLN13 disease	401964	CMT2 with giant axons	101076	CMTX2
93929	Cloacal extrophy	99946	CMT2A1	101077	CMTX3
314950	Clonal hypereosinophilic syndrome	99947	CMT2A2	101078	CMTX4
268366	Closed iniencephaly	99936	CMT2B	99014	CMTX5
189	Clouston syndrome	99937	CMT2C	352675	CMTX6
100978	Cloverleaf skull-asphyxiating thoracic dysplasia syndrome	99938	CMT2D	468635	CMUSE
93274	Cloverleaf skull-micromelic bone dysplasia syndrome	99939	CMT2E	137698	CMV disease in patients with impaired cell mediated immunity deemed at risk
93267	Cloverleaf skull-multiple congenital anomalies syndrome	99940	CMT2F	319160	CNM4
140944	CLOVES syndrome	99941	CMT2G	324964	CNO/CRMO
411493	CLP1-related pontocerebellar hypoplasia	101102	CMT2H	306686	CO-induced parkinsonism
3253	CLPED1	99942	CMT2I	1454	COACH syndrome
192	CLS	99943	CMT2J	1456	Coarctation of the abdominal aorta
85136	CLWM	99944	CMT2K	397725	COASY protein-associated neurodegeneration
137667	CM-AVM	99945	CMT2L	190	Coats disease
289504	CMAMMA	228179	CMT2M	313838	Coats plus syndrome
1334	CMC	228174	CMT2N	79282	Cobalamin C defect
258	CMD1A	284232	CMT2O	79283	Cobalamin D defect
98893	CMD1B	300319	CMT2P	79284	Cobalamin F defect
→370953	CMD1C	329258	CMT2Q	369955	Cobalamin J defect
370959	CMD with cerebellar involvement	397968	CMT2R	53721	Cobb syndrome
370968	CMD with intellectual disability	443073	CMT2S	352682	Cobblestone lissencephaly without muscular or eye involvement
329178	CMD with intellectual disability and severe epilepsy	495274	CMT2T	352682	Cobblestone lissencephaly without muscular or ocular involvement
370980	CMD without intellectual disability	397735	CMT2U	1911	Cocaine embryofetopathy
370959	CMD-CRB	447964	CMT2V	90068	Cocaine intoxication
370968	CMD-MR	488333	CMT2W	228123	Coccidioides infection
370980	CMD-no MR	466775	CMT2X	228123	Coccidioidomycosis
371007	CMDH	435387	CMT2Y	502318	Cochlear nerve deficiency
521	CML	466768	CMT2Z	3233	Cochleosaccular degeneration-cataract syndrome
98823	CMML	101077	CMT3X	502305	Cochleovestibular dysplasia
252202	CMMR-D syndrome	99948	CMT4A	191	Cockayne syndrome
		99955	CMT4B1	90321	Cockayne syndrome type 1
		99956	CMT4B2		
		363981	CMT4B3		
		99949	CMT4C		
		99950	CMT4D		
		99951	CMT4E		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
90322	Cockayne syndrome type 2	1471	Coloboma of macula-brachydactyly type B syndrome	79282	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIC
90324	Cockayne syndrome type 3	98947	Coloboma of optic disc	79283	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbID
90321	Cockayne syndrome type I	1475	Coloboma of optic nerve with renal disease	79284	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIF
90322	Cockayne syndrome type II	98947	Coloboma of optic papilla	369955	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIJ
90324	Cockayne syndrome type III	155884	Coloboma of superior eyelid	369962	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIX
1458	CODAS syndrome	3474	Coloboma-congenital heart disease-ichthyosiform dermatosis-intellectual disability-ear anomalies syndrome	35909	Combined deficiency of factor V and factor VIII
192	Coffin-Lowry syndrome	138	Coloboma-heart defects-atresia choanae-retardation of growth and development-genitourinary problems-ear abnormalities syndrome	99732	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase
1465	Coffin-Siris syndrome	468672	Colobomatous macrophtalmia-microcornea syndrome	308386	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type A
1466	COFS syndrome	98938	Colobomatous microphthalmia	308393	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type B
263508	COG1-CDG	363741	Colobomatous microphthalmia-obesity-hypogenitalism-intellectual disability syndrome	308400	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type C
435934	COG2-CDG	424099	Colobomatous microphthalmia-rhizomelic dysplasia syndrome	440727	Combined hamartoma of the retina and retinal pigment epithelium
435934	COG2-related congenital disorder of glycosylation	435930	Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	440727	Combined hamartoma of the retina and RPE
263501	COG4-CDG	→138	Colobomatous-microphthalmia-heart disease-hearing loss syndrome	221078	Combined hyperactive dysfunction syndrome of the cranial nerves
263487	COG5-CDG	1198	Colonic atresia	169082	Combined immunodeficiency due to CD3gamma deficiency
464443	COG6-CGD	100080	Colonic NET	169090	Combined immunodeficiency due to CRAC channel dysfunction
79333	COG7-CDG	16	Color blindness, blue monocone monochromatic type	217390	Combined immunodeficiency due to dedicator of cytokinesis 8 protein deficiency
95428	COG8-CDG	83595	Colorado tick encephalitis	217390	Combined immunodeficiency due to DOCK8 deficiency
98956	Cogan microcystic epithelial dystrophy	83595	Colorado tick fever	505227	Combined immunodeficiency due to GINS1 deficiency
1467	Cogan syndrome	83595	Colorado tick-borne disease	317473	Combined immunodeficiency due to IKAROS deficiency
98980	Cogan-Reese syndrome	733	Colorectal adenomatous polyposis	445018	Combined immunodeficiency due to LRBA deficiency
444077	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	261584	Colorectal adenomatous polyposis due to monosomy 5q22.2	317476	Combined immunodeficiency due to MAGT1 deficiency
193	Cohen syndrome	90793	Combined 17-hydroxylase/17,20-lyase deficiency		
2969	Cohen-Hayden syndrome	734	Combined alpha-delta platelet storage pool deficiency		
79144	COIF	445062	Combined cerebellar and peripheral ataxia-hearing loss-diabetes mellitus syndrome		
79144	COIF syndrome	370114	Combined cervical dystonia		
36383	COL4A1-related brain small vessel disease with hemorrhage	356978	Combined D-2-hydroxyglutaric aciduria and L-2-hydroxyglutaric aciduria		
36383	COL4A1-related familial vascular leukoencephalopathy	356978	Combined D-2-hydroxyglutaric aciduria and L-2-hydroxyglutaric aciduria		
36383	COL4A1-related retinal arteriolar tortuosity-infantile hemiparesis-autosomal dominant leukoencephalopathy syndrome	26	Combined defect in adenosylcobalamin and methylcobalamin synthesis		
31824	Colchicine poisoning				
56425	Cold agglutinin disease				
56425	Cold agglutinin syndrome				
157820	Cold-induced sweating syndrome				
324561	Cole disease				
2050	Cole-Carpenter syndrome				
84087	Collagen type III glomerulopathy				
84087	Collagenofibrotic glomerulopathy				
247203	Collecting duct carcinoma				
2412	Collins-Pope syndrome				
98942	Coloboma of choroid and retina				
98943	Coloboma of eye lens				
98946	Coloboma of eyelid				
155889	Coloboma of inferior eyelid				
98944	Coloboma of iris				
98945	Coloboma of macula				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
397964	Combined immunodeficiency due to MALT1 deficiency		deficiency		deficiencies, genetic forms
504530	Combined immunodeficiency due to Moesin deficiency	289504	Combined malonic and methylmalonic acidemia	139406	Combined prosaposin deficiency
317428	Combined immunodeficiency due to ORAI1 deficiency	289504	Combined malonic and methylmalonic aciduria	300564	Combined pulmonary fibrosis-emphysema syndrome
431149	Combined immunodeficiency due to OX40 deficiency	254920	Combined oxidative phosphorylation defect type 2	166286	Comedo nevus of the palm
231154	Combined immunodeficiency due to partial RAG1 deficiency	254925	Combined oxidative phosphorylation defect type 4	141276	Commissural facial cleft
443811	Combined immunodeficiency due to PGM3 deficiency	137908	Combined oxidative phosphorylation defect type 5	141061	Commissural lip fistula
157949	Combined immunodeficiency due to RAG 1/2 deficiency	254930	Combined oxidative phosphorylation defect type 7	3384	Common aorticopulmonary trunk
317430	Combined immunodeficiency due to STIM1 deficiency	319504	Combined oxidative phosphorylation defect type 8	3384	Common arterial trunk
314689	Combined immunodeficiency due to STK4 deficiency	319509	Combined oxidative phosphorylation defect type 9	1329	Common atrioventricular canal
476113	Combined immunodeficiency due to TFRC deficiency	314637	Combined oxidative phosphorylation defect type 10	→288	Common hereditary elliptocytosis
911	Combined immunodeficiency due to ZAP70 deficiency	324535	Combined oxidative phosphorylation defect type 11	620	Common mesentery
431149	Combined immunodeficiency with childhood-onset Kaposi sarcoma	314051	Combined oxidative phosphorylation defect type 12	1572	Common variable immunodeficiency
231154	Combined immunodeficiency with expansion of gamma delta T cells	319514	Combined oxidative phosphorylation defect type 13	280821	Communicating congenital bronchopulmonary-foregut malformation
221139	Combined immunodeficiency with faciooculoskeletal anomalies	319519	Combined oxidative phosphorylation defect type 14	280133	Complement component 3 deficiency
39041	Combined immunodeficiency with hypereosinophilia	319524	Combined oxidative phosphorylation defect type 15	99429	Complete androgen insensitivity syndrome
431149	Combined immunodeficiency with impaired immunity to HHV-8	352563	Combined oxidative phosphorylation defect type 16	99429	Complete androgen resistance syndrome
431149	Combined immunodeficiency with impaired immunity to human herpes virus 8	369913	Combined oxidative phosphorylation defect type 17	1329	Complete atrioventricular canal
505227	Combined immunodeficiency with intrauterine growth retardation-natural killer cell deficiency-neutropenia	420728	Combined oxidative phosphorylation defect type 20	99066	Complete atrioventricular canal-left heart obstruction syndrome
505227	Combined immunodeficiency with intrauterine growth retardation-NK cell deficiency-neutropenia	420733	Combined oxidative phosphorylation defect type 21	99068	Complete atrioventricular canal-tetralogy of Fallot syndrome
157949	Combined immunodeficiency with skin granulomas	444013	Combined oxidative phosphorylation defect type 23	99067	Complete atrioventricular canal-ventricle hypoplasia syndrome
228423	Combined immunodeficiency with susceptibility to mycobacterial, viral and fungal infections	444458	Combined oxidative phosphorylation defect type 24	1329	Complete atrioventricular septal defect
436252	Combined immunodeficiency-enteropathy spectrum	447954	Combined oxidative phosphorylation defect type 25	98949	Complete cryptophthalmia
169079	Combined immunodeficiency-microcephaly-growth retardation-sensitivity to ionizing radiation syndrome	477684	Combined oxidative phosphorylation defect type 26	289916	Complete deficiency of methylmalonyl-CoA mutase
1979	Combined insulin, insulin-like growth factor 1 (IGF1) and epidermal growth factor (EGF)	477774	Combined oxidative phosphorylation defect type 27	633	Complete growth hormone insensitivity
		466784	Combined oxidative phosphorylation defect type 28	254688	Complete hydatidiform mole
		478029	Combined oxidative phosphorylation defect type 29	79293	Complete LCAT deficiency
		478042	Combined oxidative phosphorylation defect type 30	29	Complete mevalonate kinase deficiency
		309111	Combined pancreatic lipase-colipase deficiency	254688	Complete molar pregnancy
		95494	Combined pituitary hormone	49382	Complete or incomplete color blindness

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	Barnes-Gistelinck type	2879	Congenital absence of ulna and fibula	93322	Congenital aplasia and dysplasia of the tibia with intact fibula
83452	Complex regional pain syndrome	294975	Congenital absence of upper arm and forearm with hand present	2926	Congenital aplasia of the extensor muscles of the fingers and thumb associated with generalized polyneuropathy
99995	Complex regional pain syndrome type 1	247775	Congenital absence of uterus and vagina	353334	Congenital arteriovenous anastomoses of the retina
99994	Complex regional pain syndrome type 2	96269	Congenital absence of vagina	353334	Congenital arteriovenous communication of the retina
306644	Complication after organ transplantation	973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral	1195	Congenital atransferrinemia
268316	Complication in hemodialysis	294988	Congenital absence/hypoplasia of thumb	60041	Congenital atrioventricular block
458758	Composite hemangioendothelioma	324353	Congenital achiasma	162526	Congenital auditory ossicle malformation without external ear abnormality
168966	Composite Hodgkin and non-Hodgkin lymphoma	93583	Congenital ADAMTS-13 deficiency	1216	Congenital benign spinal muscular atrophy with contractures
168966	Composite lymphoma	90791	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	48	Congenital bilateral absence of vas deferens
634	Comèl-Netherton syndrome	90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	48	Congenital bilateral agenesis of vas deferens
228165	Concentric demyelination	90793	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	48	Congenital bilateral aplasia of vas deferens
383	Conductive deafness with stapes fixation	95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	93177	Congenital bilateral megacalycosis
3216	Conductive deafness-malformed external ear syndrome	95699	Congenital adrenal hyperplasia due to cytochrome POR deficiency	79301	Congenital bile acid synthesis defect type 1
3236	Conductive deafness-ptosis-skeletal anomalies syndrome	495875	Congenital agenesis of labia majora or scrotum-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome	79303	Congenital bile acid synthesis defect type 2
3216	Conductive hearing loss-malformed external ear syndrome	495879	Congenital agenesis of the scrotum	79302	Congenital bile acid synthesis defect type 3
1871	Cone dystrophy	33355	Congenital aleukocytosis	79095	Congenital bile acid synthesis defect type 4
209932	Cone dystrophy with supernormal rod electroretinogram	79	Congenital alpha2-antiplasmin deficiency	2292	Congenital bowing of long bones
209932	Cone dystrophy with supernormal rod ERG	210122	Congenital alveolar capillary dysplasia	71278	Congenital brain dysgenesis due to glutamine synthetase deficiency
209932	Cone dystrophy with supernormal rod response	3319	Congenital amegakaryocytic thrombocytopenia	2040	Congenital bronchobiliary fistula
209932	Cone dystrophy with supernormal scotopic electroretinogram	3319	Congenital amegakaryocytic thrombocytopenic purpura	3161	Congenital bronchopulmonary sequestration
1872	Cone rod dystrophy	488586	Congenital amyoplasia	289499	Congenital cataract microcornea with corneal opacity
1873	Cone rod dystrophy-amelogenesis imperfecta syndrome	86816	Congenital analbuminemia	300313	Congenital cataract-deafness-severe developmental delay syndrome
221142	Confetti-like macular atrophy	217399	Congenital analgesia with hyperhidrosis	300313	Congenital cataract-hearing loss-severe developmental delay syndrome
440233	Congenital abducens nerve palsy	453510	Congenital analgesia with severe intellectual disability	1369	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome
294979	Congenital absence of both forearm and hand	95507	Congenital anomaly of hepatic vein	464738	Congenital cataract-microcephaly-nevus flammeus simplex-severe intellectual disability syndrome
294981	Congenital absence of both lower leg and foot	91489	Congenital anterior megalophthalmia	330054	Congenital cataract-progressive muscular hypotonia-deafness-developmental delay syndrome
289465	Congenital absence of fingerprints	95449	Congenital aortic valve insufficiency	330054	Congenital cataract-progressive
294986	Congenital absence of foot	3093	Congenital aortic valve stenosis		
294983	Congenital absence of hand	2037	Congenital aortopulmonary artery fistula		
294973	Congenital absence of humerus	2037	Congenital aortopulmonary septal defect		
86815	Congenital absence of lacrimal puncta and salivary glands	2037	Congenital aortopulmonary window		
217399	Congenital absence of pain with hyperhidrosis				
453510	Congenital absence of pain with severe intellectual disability				
495879	Congenital absence of the scrotum				
294977	Congenital absence of thigh and lower leg with foot present				
93322	Congenital absence of tibia				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	muscular hypotonia-hearing loss-developmental delay syndrome	280847	Congenital cystic adenomatous malformation of the lung type 3		type 1o
48431	Congenital cataracts-facial dysmorphism-neuropathy syndrome	280854	Congenital cystic adenomatous malformation of the lung type 4	280071	Congenital disorder of glycosylation type 1p
661	Congenital central alveolar hypoventilation syndrome	480501	Congenital cystic dilatation of the biliary tract	324737	Congenital disorder of glycosylation type 1q
99803	Congenital central alveolar hypoventilation-Hirschsprung disease syndrome	2444	Congenital cystic disease of the lung	300536	Congenital disorder of glycosylation type 1r
831	Congenital cervical spinal stenosis	280832	Congenital cystic disease of the lung type 1	324422	Congenital disorder of glycosylation type 1s
2345	Congenital cervical vertebral fusion	280840	Congenital cystic disease of the lung type 2	319646	Congenital disorder of glycosylation type 1t
53689	Congenital chloride diarrhea	280847	Congenital cystic disease of the lung type 3	329178	Congenital disorder of glycosylation type 1u
329242	Congenital chronic diarrhea with exudative enteropathy	168612	Congenital deficiency in alpha-fetoprotein	370921	Congenital disorder of glycosylation type 1w
329242	Congenital chronic diarrhea with protein-losing enteropathy	2140	Congenital diaphragmatic hernia	370924	Congenital disorder of glycosylation type 1x
264688	Congenital chylothorax	488635	Congenital disorder of glycosylation due to PIGG deficiency	370927	Congenital disorder of glycosylation type 1y
2505	Congenital circumferential skin folds	3474	Congenital disorder of glycosylation due to PIGL deficiency	448010	Congenital disorder of glycosylation type 1z
91413	Congenital Claude-Bernard-Horner syndrome	83639	Congenital disorder of glycosylation due to PIGM deficiency	397941	Congenital disorder of glycosylation type 2 due to MAN1B1 deficiency
440221	Congenital CNIII lesion	280633	Congenital disorder of glycosylation due to PIGN deficiency	79329	Congenital disorder of glycosylation type 2a
98686	Congenital CNIV palsy	369837	Congenital disorder of glycosylation due to PIGT deficiency	79330	Congenital disorder of glycosylation type 2b
440233	Congenital CNVI palsy	79318	Congenital disorder of glycosylation type 1a	79332	Congenital disorder of glycosylation type 2d
269505	Congenital communicating hydrocephalus	79319	Congenital disorder of glycosylation type 1b	79333	Congenital disorder of glycosylation type 2e
99129	Congenital complete agenesis of pericardium	79320	Congenital disorder of glycosylation type 1c	238459	Congenital disorder of glycosylation type 2f
115	Congenital contractual arachnodactyly	79321	Congenital disorder of glycosylation type 1d	263508	Congenital disorder of glycosylation type 2g
178382	Congenital convex foot	79322	Congenital disorder of glycosylation type 1e	95428	Congenital disorder of glycosylation type 2h
178382	Congenital convex pes valgus	79323	Congenital disorder of glycosylation type 1f	263487	Congenital disorder of glycosylation type 2i
53691	Congenital cornea plana	79324	Congenital disorder of glycosylation type 1g	263501	Congenital disorder of glycosylation type 2j
95491	Congenital coronary aneurysm	79325	Congenital disorder of glycosylation type 1h	314667	Congenital disorder of glycosylation type 2k
95491	Congenital coronary artery aneurysm	79326	Congenital disorder of glycosylation type 1i	464443	Congenital disorder of glycosylation type 2l
2444	Congenital cystic adenomatoid malformation of the lung	86309	Congenital disorder of glycosylation type 1j	356961	Congenital disorder of glycosylation type 2m
280827	Congenital cystic adenomatoid malformation of the lung type 0	79327	Congenital disorder of glycosylation type 1k	468699	Congenital disorder of glycosylation type 2n
280832	Congenital cystic adenomatoid malformation of the lung type 1	79328	Congenital disorder of glycosylation type 1L	468684	Congenital disorder of glycosylation type 2o
280840	Congenital cystic adenomatoid malformation of the lung type 2	91131	Congenital disorder of glycosylation type 1m	466703	Congenital disorder of glycosylation type 2p
280847	Congenital cystic adenomatoid malformation of the lung type 3	244310	Congenital disorder of glycosylation type 1n	79318	Congenital disorder of glycosylation type 1a
280854	Congenital cystic adenomatoid malformation of the lung type 4	263494	Congenital disorder of glycosylation	79319	Congenital disorder of glycosylation
2444	Congenital cystic adenomatous malformation of the lung				
280827	Congenital cystic adenomatous malformation of the lung type 0				
280832	Congenital cystic adenomatous malformation of the lung type 1				
280840	Congenital cystic adenomatous malformation of the lung type 2				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	type Ib		type Ia	231573	Congenital erosive and vesicular dermatosis with reticulated supple scarring
79320	Congenital disorder of glycosylation type Ic	280071	Congenital disorder of glycosylation type Ia	90042	Congenital erythrocytosis due to erythropoietin receptor mutation
79321	Congenital disorder of glycosylation type Id	324737	Congenital disorder of glycosylation type Iq	369992	Congenital erythroderma-hypotrichosis-recurrent infections-multiple food allergies syndrome
79322	Congenital disorder of glycosylation type Ie	300536	Congenital disorder of glycosylation type Ir	79277	Congenital erythropoietic porphyria
79323	Congenital disorder of glycosylation type If	324422	Congenital disorder of glycosylation type Is	91358	Congenital esophageal diverticulum
79324	Congenital disorder of glycosylation type Ig	319646	Congenital disorder of glycosylation type It	91358	Congenital esophageal pouch
79325	Congenital disorder of glycosylation type Ih	329178	Congenital disorder of glycosylation type Iu	215	Congenital essential nyctalopia
79326	Congenital disorder of glycosylation type II	370921	Congenital disorder of glycosylation type Iw	91	Congenital estrogen deficiency
397941	Congenital disorder of glycosylation type II due to MAN1B1 deficiency	370924	Congenital disorder of glycosylation type Ix	280811	Congenital extrapulmonary sequestration
79329	Congenital disorder of glycosylation type Ila	370927	Congenital disorder of glycosylation type ly	99176	Congenital eyelid retraction
79330	Congenital disorder of glycosylation type Ilb	293825	Congenital dyserythropoietic anemia due to KLF1 mutation	570	Congenital facial diplegia
79332	Congenital disorder of glycosylation type Ild	98869	Congenital dyserythropoietic anemia type 1	325	Congenital factor II deficiency
79333	Congenital disorder of glycosylation type Ile	98873	Congenital dyserythropoietic anemia type 2	326	Congenital factor V deficiency
238459	Congenital disorder of glycosylation type IIf	98870	Congenital dyserythropoietic anemia type 3	327	Congenital factor VII deficiency
263508	Congenital disorder of glycosylation type IIg	293825	Congenital dyserythropoietic anemia type 4	328	Congenital factor X deficiency
95428	Congenital disorder of glycosylation type IIh	98869	Congenital dyserythropoietic anemia type I	329	Congenital factor XI deficiency
263487	Congenital disorder of glycosylation type III	98873	Congenital dyserythropoietic anemia type II	330	Congenital factor XII deficiency
263501	Congenital disorder of glycosylation type IIj	98870	Congenital dyserythropoietic anemia type III	331	Congenital factor XIII deficiency
314667	Congenital disorder of glycosylation type IIk	293825	Congenital dyserythropoietic anemia type IV	2020	Congenital fiber-type disproportion myopathy
464443	Congenital disorder of glycosylation type IIII	67044	Congenital dyserythropoietic anemia with thrombocytopenia	335	Congenital fibrinogen deficiency
356961	Congenital disorder of glycosylation type IIIm	70596	Congenital EBV infection	45358	Congenital fibrosis of extraocular muscles
468699	Congenital disorder of glycosylation type IIIn	91491	Congenital ectropion uveae	90045	Congenital folate malabsorption
468684	Congenital disorder of glycosylation type IIlo	295227	Congenital elbow dislocation, bilateral	98686	Congenital fourth cranial nerve palsy
466703	Congenital disorder of glycosylation type IIp	295225	Congenital elbow dislocation, unilateral	2345	Congenital fused cervical segments
86309	Congenital disorder of glycosylation type Ij	103910	Congenital enterocyte heparan sulfate deficiency	476406	Congenital generalized hypercontractile muscle stiffness syndrome
79327	Congenital disorder of glycosylation type Ik	168601	Congenital enterokinase deficiency	2026	Congenital generalized hypertrichosis terminalis
91131	Congenital disorder of glycosylation type Im	168601	Congenital enteropathy due to enteropeptidase deficiency	1023	Congenital generalized hypertrichosis, Ambras type
244310	Congenital disorder of glycosylation type In	292	Congenital enterovirus infection	79495	Congenital generalized hypertrichosis, Macias-Flores type
263494	Congenital disorder of glycosylation	70596	Congenital Epstein-Barr virus infection	295232	Congenital genu flexum
		157826	Congenital epulis	295229	Congenital genu recurvatum
		231573	Congenital erosive and vesicular dermatosis	99095	Congenital Gerbode defect

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	defects		microcephalus-tetraplegia syndrome	295034	Congenital knee dislocation
98975	Congenital hereditary endothelial dystrophy type 1	631	Congenital IGHD	495875	Congenital labioscrotal agenesis-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome
293603	Congenital hereditary endothelial dystrophy type 2	231662	Congenital IGHD type IA	53690	Congenital lactase deficiency
98975	Congenital hereditary endothelial dystrophy type I	231671	Congenital IGHD type IB	70472	Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type
293603	Congenital hereditary endothelial dystrophy type II	231679	Congenital IGHD type II	313	Congenital lamellar ichthyosis
306530	Congenital hereditary facial palsy with variable deafness	231692	Congenital IGHD type III	99872	Congenital Langerhans cell histiocytosis
306530	Congenital hereditary facial palsy with variable hearing loss	306504	Congenital ILNEB syndrome	141124	Congenital laryngeal cyst
306530	Congenital hereditary facial paralysis with variable deafness	217399	Congenital indifference to pain with hyperhidrosis	137932	Congenital laryngeal palsy
306530	Congenital hereditary facial paralysis-variable deafness syndrome	64752	Congenital insensitivity to pain and thermal analgesia	2374	Congenital laryngeal web
306530	Congenital hereditary facial paralysis-variable hearing loss syndrome	217399	Congenital insensitivity to pain with hyperhidrosis	2373	Congenital laryngomalacia
101068	Congenital hereditary stromal dystrophy	391397	Congenital insensitivity to pain with hyperhidrosis and gastrointestinal dysfunction	1954	Congenital lethal erythroderma
293	Congenital herpes simplex virus infection	453510	Congenital insensitivity to pain with preserved temperature sensation	210163	Congenital lethal myopathy, Compton-North type
483	Congenital high-molecular-weight kininogen deficiency	453510	Congenital insensitivity to pain with severe intellectual disability	90790	Congenital lipoid adrenal hyperplasia due to STAR deficiency
91413	Congenital Horner syndrome	453510	Congenital insensitivity to pain with severe non-progressive cognitive delay	140944	Congenital lipomatous overgrowth-vascular malformation-epidermal nevi-skeletal anomaly syndrome
2185	Congenital hydrocephalus	642	Congenital insensitivity to pain-anhidrosis syndrome	140944	Congenital lipomatous overgrowth-vascular malformation-epidermal nevi-spinal anomaly syndrome
2190	Congenital hydronephrosis	306504	Congenital interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome	1928	Congenital lobar emphysema
478	Congenital hypogonadotropic hypogonadism with anosmia	388	Congenital intestinal aganglionosis	1928	Congenital lobar hyperinflation
294973	Congenital hypoplasia of humerus	280802	Congenital intrapulmonary sequestration	93323	Congenital longitudinal deficiency of the fibula
124	Congenital hypoplastic anemia, Blackfan-Diamond type	1229	Congenital intrauterine infection-like syndrome	93321	Congenital longitudinal deficiency of the radius
→672	Congenital hypothalamic hamartoma syndrome	332	Congenital intrinsic factor deficiency	93322	Congenital longitudinal deficiency of the tibia
226313	Congenital hypothyroidism due to maternal intake of antithyroid drugs	199296	Congenital isolated ACTH deficiency	93320	Congenital longitudinal deficiency of the ulna
95715	Congenital hypothyroidism due to transplacental passage of maternal TSH-binding inhibitory antibodies	631	Congenital isolated GH deficiency	2430	Congenital macroglossia
1195	Congenital hypotransferrinemia	231662	Congenital isolated GH deficiency type IA	95430	Congenital major airway collapse
→113	Congenital hypotrichosis-milia syndrome	231671	Congenital isolated GH deficiency type IB	83620	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells
88621	Congenital ichthyosis type 4	231679	Congenital isolated GH deficiency type II	93109	Congenital megacalycosis
352333	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome	231692	Congenital isolated GH deficiency type III	280671	Congenital megaconial myopathy
352333	Congenital ichthyosis-intellectual disability-spastic tetraplegia syndrome	631	Congenital isolated growth hormone deficiency	69063	Congenital membranous nephropathy due to maternal anti-neutral endopeptidase alloimmunization
2271	Congenital ichthyosis-microcephalus-quadriplegia syndrome	231662	Congenital isolated growth hormone deficiency type IA	2665	Congenital mesoblastic nephroma
2271	Congenital ichthyosis-	231671	Congenital isolated growth hormone deficiency type IB	621	Congenital methemoglobinemia
		231679	Congenital isolated growth hormone deficiency type II	391376	Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome
		231692	Congenital isolated growth hormone deficiency type III	566	Congenital microcoria

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2290	Congenital microvillus atrophy		syndrome	2772	Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome
566	Congenital miosis	590	Congenital myasthenic syndrome	465	Congenital PAI-1 deficiency
99057	Congenital mitral stenosis	353327	Congenital myasthenic syndromes with glycosylation defect	2805	Congenital pancreatic agenesis
98905	Congenital multicore myopathy with external ophthalmoplegia	98904	Congenital myopathy with excess of thin filaments	313906	Congenital pancreatic cyst
508512	Congenital multiple café-au-lait macules-increased sister chromatid exchange syndrome	319160	Congenital myopathy with internal nuclei and atypical cores	139414	Congenital panfollicular nevus
258	Congenital muscular dystrophy due to laminin alpha2 deficiency	424107	Congenital myopathy with myasthenic-like onset	264675	Congenital PAP
157973	Congenital muscular dystrophy due to LMNA mutation	199329	Congenital myopathy, Paradas type	99130	Congenital partial agenesis of pericardium
280671	Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect	168572	Congenital myopathy-cleft palate-malignant hyperthermia syndrome	99124	Congenital partial pulmonary venous return anomaly
258	Congenital muscular dystrophy type 1A	289380	Congenital myosclerosis, Löwenthal type	295036	Congenital patella dislocation
98893	Congenital muscular dystrophy type 1B	831	Congenital narrowing of cervical spinal canal	99072	Congenital patent ductus arteriosus aneurysm
→370953	Congenital muscular dystrophy type 1C	162521	Congenital nasal pyriform aperture stenosis with holoprosencephaly	332	Congenital pernicious anemia
→370953	Congenital muscular dystrophy type 1D	168486	Congenital NCL	626	Congenital pigmented nevus
370959	Congenital muscular dystrophy with cerebellar involvement	306504	Congenital NEP syndrome	465	Congenital plasminogen activator inhibitor type 1 deficiency
371007	Congenital muscular dystrophy with hyperlaxity	443988	Congenital nephrosis-cerebral ventriculomegaly syndrome	2907	Congenital poikiloderma with bullae, Weary type
34520	Congenital muscular dystrophy with integrin alpha-7 deficiency	839	Congenital nephrotic syndrome, Finnish type	90042	Congenital polycythemia due to erythropoietin receptor mutation
370968	Congenital muscular dystrophy with intellectual disability	306504	Congenital nephrotic syndrome-interstitial lung disease-epidermolysis bullosa-pulmonary disease syndrome	480531	Congenital portosystemic shunt
329178	Congenital muscular dystrophy with intellectual disability and severe epilepsy	168486	Congenital neuronal ceroid lipofuscinosis	480531	Congenital portosystemic venous fistula
34520	Congenital muscular dystrophy with ITGA7 deficiency	369852	Congenital neutropenia-bone marrow fibrosis-nephromegaly syndrome	124	Congenital PRCA
280671	Congenital muscular dystrophy with mitochondrial structural abnormalities	369852	Congenital neutropenia-myelofibrosis-nephromegaly syndrome	749	Congenital prekallikrein deficiency
370980	Congenital muscular dystrophy without intellectual disability	79394	Congenital non-bullous ichthyosiform erythroderma	83461	Congenital primary aphakia
486815	Congenital muscular dystrophy, Davignon-Chauveau type	269510	Congenital non-communicating hydrocephalus	79452	Congenital primary lymphedema
272	Congenital muscular dystrophy, Fukuyama type	269505	Congenital non-obstructive hydrocephalus	617	Congenital primary megaloureter
75840	Congenital muscular dystrophy, Ullrich type	1216	Congenital nonprogressive spinal muscular atrophy	617	Congenital primary megaureter
370980	Congenital muscular dystrophy-dystroglycanopathy without intellectual disability	208513	Congenital nonprogressive spinocerebellar ataxia	238654	Congenital primary megaureter, nonrefluxing and unobstructed form
1875	Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome	269510	Congenital obstructive hydrocephalus	238646	Congenital primary megaureter, obstructed form
486815	Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity	440221	Congenital oculomotor nerve palsy	238650	Congenital primary megaureter, refluxing form
		79144	Congenital onychodysplasia of the index fingers	327	Congenital proconvertin deficiency
		157713	Congenital or early infantile CACH syndrome	508542	Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome
				66630	Congenital pseudarthrosis of the clavicle
				295020	Congenital pseudarthrosis of the femur
				295022	Congenital pseudarthrosis of the fibula
				157808	Congenital pseudarthrosis of the limbs
				295024	Congenital pseudarthrosis of the radius
				295018	Congenital pseudarthrosis of the tibia

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
295026	Congenital pseudarthrosis of the ulna		limb anomalies	306474	Congenital sucrase-isomaltose malabsorption with starch and lactose intolerance
66630	Congenital pseudoarthrosis of the clavicle	974	Congenital scalp defects with distal limb reduction anomalies	306436	Congenital sucrase-isomaltose malabsorption with starch intolerance
295020	Congenital pseudoarthrosis of the femur	495879	Congenital scrotal absence	306462	Congenital sucrase-isomaltose malabsorption without starch intolerance
295022	Congenital pseudoarthrosis of the fibula	495879	Congenital scrotal agenesis	35122	Congenital sucrose intolerance
157808	Congenital pseudoarthrosis of the limbs	2301	Congenital short bowel syndrome	306446	Congenital sucrose intolerance with minimal starch tolerance
295024	Congenital pseudoarthrosis of the radius	1987	Congenital short femur	306474	Congenital sucrose intolerance with starch and lactose intolerance
295018	Congenital pseudoarthrosis of the tibia	295030	Congenital shoulder dislocation	306436	Congenital sucrose intolerance with starch intolerance
295026	Congenital pseudoarthrosis of the ulna	93400	Congenital sialidosis type 2	306462	Congenital sucrose intolerance without starch intolerance
91411	Congenital ptosis	260305	Congenital sideroblastic anemia	35122	Congenital sucrose intolerance
2444	Congenital pulmonary airway malformation	369861	Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome	306446	Congenital sucrose intolerance with minimal starch tolerance
280827	Congenital pulmonary airway malformation type 0	263435	Congenital smooth muscle hamartoma	306474	Congenital sucrose intolerance with starch and lactose intolerance
280832	Congenital pulmonary airway malformation type 1	103908	Congenital sodium diarrhea	306436	Congenital sucrose intolerance with starch intolerance
280840	Congenital pulmonary airway malformation type 2	94068	Congenital spondyloepiphyseal dysplasia	306462	Congenital sucrose intolerance without starch intolerance
280847	Congenital pulmonary airway malformation type 3	215	Congenital stationary night blindness	306486	Congenital sucrose-isomaltose malabsorption without sucrose intolerance
280854	Congenital pulmonary airway malformation type 4	75382	Congenital stationary night blindness, Oguchi type	98686	Congenital superior oblique palsy
264675	Congenital pulmonary alveolar proteinosis	831	Congenital stenosis of the cervical spine	3465	Congenital suprabulbar paresis
2414	Congenital pulmonary lymphangiectasia	99122	Congenital stenosis of the inferior caval vein	99059	Congenital supravalvular mitral ring
3161	Congenital pulmonary sequestration	99122	Congenital stenosis of the inferior vena cava	98948	Congenital symblepharon
3189	Congenital pulmonary valve stenosis	99122	Congenital stenosis of the IVC	499009	Congenital syphilis
3188	Congenital pulmonary veins atresia or stenosis	3197	Congenital stiff man syndrome	99856	Congenital syringomyelia
185	Congenital pulmonary venolobar syndrome	101068	Congenital stromal corneal dystrophy	2039	Congenital systemic arteriovenous fistula
124	Congenital pure red cell aplasia	328	Congenital Stuart factor deficiency	210576	Congenital temporomandibular joint ankylosis
3269	Congenital radioulnar synostosis	141121	Congenital subglottic stenosis	440221	Congenital third cranial nerve palsy
97598	Congenital renal artery stenosis	35122	Congenital sucrase-isomaltase deficiency	93583	Congenital thrombotic thrombocytopenic purpura
97598	Congenital renovascular hypoplasia	306446	Congenital sucrase-isomaltase deficiency with minimal starch tolerance	99125	Congenital total pulmonary venous return anomaly
281190	Congenital reticular ichthyosiform erythroderma	306474	Congenital sucrase-isomaltase deficiency with starch and lactose intolerance	858	Congenital toxoplasmosis
353334	Congenital retinal arteriovenous anastomoses	306436	Congenital sucrase-isomaltase deficiency with starch intolerance	141127	Congenital tracheal stenosis
353334	Congenital retinal arteriovenous communication	306462	Congenital sucrase-isomaltase deficiency without starch intolerance	3347	Congenital tracheobronchomegaly
91495	Congenital retinal detachment	306486	Congenital sucrase-isomaltase deficiency without sucrose intolerance	95430	Congenital tracheomalacia
190	Congenital retinal telangiectasia	35122	Congenital sucrase-isomaltose malabsorption	95459	Congenital tricuspid stenosis
178382	Congenital rocker-bottom foot	306446	Congenital sucrase-isomaltose malabsorption with minimal starch tolerance	231013	Congenital trigeminal anesthesia
290	Congenital rubella syndrome			210576	Congenital trismus
974	Congenital scalp defects with distal			88629	Congenital tritanopia

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
1864	Congenital valvular dysplasia	1303	Constrictive bronchiolitis		hyperkeratosis-laryngeal dyskeratosis syndrome
291	Congenital varicella syndrome	369942	Contiguous ABCD1 DXS1357E deletion syndrome	3177	Corneal-cerebellar syndrome
2291	Congenital velopharyngeal incompetence	84142	Continuous muscle fiber activity syndrome	199	Cornelia de Lange syndrome
178382	Congenital vertical talus	725	Continuous spikes and waves during sleep	3194	Corneodermatoosseous syndrome
295203	Congenital vertical talus, bilateral	725	Continuous spikes and waves during slow-wave sleep	2041	Coronaro-cardiac fistula
295201	Congenital vertical talus, unilateral	436003	Contractures-developmental delay-Pierre Robin syndrome	2041	Coronary arterial fistulas
137932	Congenital vocal cord paralysis	1484	Contractures-ectodermal dysplasia-cleft lip/palate syndrome	2041	Coronary arterial malformations
216694	Congenitally corrected transposition of the great arteries	314002	Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome	99085	Coronary artery intramyocardial course
216694	Congenitally corrected transposition of the great vessels	1487	Cooks syndrome	99087	Coronary ostial stenosis or atresia
2391	Congenitally short costocoracoid ligament	231214	Cooley anemia	99118	Coronary sinus atresia
860	Congenitally uncorrected transposition of the great arteries	1488	Cooper-Jabs syndrome	99117	Coronary sinus stenosis
216729	Congenitally uncorrected transposition of the great arteries with cardiac malformation	1302	COP	2508	Corpus callosum agenesis-abnormal genitalia syndrome
99042	Congenitally uncorrected transposition of the great arteries with coarctation	444092	COPA syndrome	3338	Corpus callosum agenesis-blepharophimosis-Robin sequence syndrome
860	Congenitally uncorrected transposition of the great vessels	397725	CoPAN	1493	Corpus callosum agenesis-cataract-immunodeficiency syndrome
216729	Congenitally uncorrected transposition of the great vessels with cardiac malformation	2062	Copenhagen syndrome	52055	Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome
99042	Congenitally uncorrected transposition of the great vessels with coarctation	98984	Coppock-like cataract	459074	Corpus callosum agenesis-macrocephaly-hypertelorism syndrome
99827	Congo fever	457185	COQ4-related neonatal encephalomyopathy	1496	Corpus callosum agenesis-neuronopathy syndrome
99827	Congo hemorrhagic fever	99098	Cor triatriatum dexter	1553	Corpus callosum agenesis-polysyndactyly syndrome
97566	Congo red-negative amyloidosis-like glomerulopathy	99098	Cor triatriatum dextrum	→3157	Corpus callosum dysgenesis-hypopituitarism syndrome
97231	Conjunctivitis lignosa	99099	Cor triatriatum sinister	275543	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome
369929	Conn adenoma with seizures and neurological abnormalities	99099	Cor triatriatum sinistrum	2318	CORS
280210	Connatal PMD	98990	Coralliform cataract	1389	Cortical blindness-intellectual disability-polydactyly syndrome
300284	Connective tissue disorder due to LH3 deficiency	180118	Cordiform uterus	300570	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation
300284	Connective tissue disorder due to lysyl hydroxylase-3 deficiency	366	Cori disease	268994	Cortical dysplasia, Taylor type
→2909	Connective tissue dysplasia, Spellacy type	366	Cori-Forbes disease	163681	Cortical dysplasia-focal epilepsy syndrome
420794	Cono-spondylar dysplasia	1051	Corneal anesthesia-deafness-intellectual disability syndrome	3152	Cortical hyperostosis-syndactyly syndrome
140969	Conorenal syndrome	1661	Corneal dystrophy epithelial-short stature syndrome	447788	Cortical visual impairment
567	Conotruncal anomaly face syndrome	98962	Corneal dystrophy Groenouw type I	454887	Corticobasal syndrome
35173	Conradi-Hünermann-Happle syndrome	98969	Corneal dystrophy Groenouw type II	199247	Corticosteroid-binding globulin deficiency
319651	Constitutional megaloblastic anemia with severe neurologic disease	98961	Corneal dystrophy of Bowman layer type 1	54251	Corticosteroid-sensitive aseptic abscess syndrome
252202	Constitutional mismatch repair deficiency syndrome	98960	Corneal dystrophy of Bowman layer type 2	99763	Corticosterone methyloxidase deficiency type I
295000	Constriction rings syndrome	1490	Corneal dystrophy of Bowman layer type I	96253	Corticotroph pituitary adenoma
		98960	Corneal dystrophy of Bowman layer type II	141163	Cosack syndrome
		1490	Corneal dystrophy with progressive deafness		
		1490	Corneal dystrophy-perceptive deafness syndrome		
		352662	Corneal intraepithelial dyskeratosis-palmoplantar		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
67047	Costeff optic atrophy syndrome	156	CPT1A deficiency		ectodermal anomalies-intellectual disability syndrome
67047	Costeff syndrome	157	CPT2	1516	Craniofacial dyssynostosis
3071	Costello syndrome	228302	CPT2, adult-onset form	1529	Craniofacial-deafness-hand syndrome
1507	Costovertebral segmentation defect-mesomelia syndrome	228305	CPT2, hepatocardiomuscular form	293843	Craniofacial-ulnar-renal syndrome
93333	Cousin syndrome	228308	CPT2, lethal systemic form	363705	Craniofaciofrontodigital syndrome
1507	COVESDEM syndrome	228302	CPT2, myopathic form	1520	Craniofrontonasal dysplasia
101078	Cowchock syndrome	228308	CPT2, neonatal form	228390	Craniofrontonasal dysplasia with alopecia and hypogonadism
201	Cowden disease	228305	CPT2, severe infantile form	1519	Craniofrontonasal dysplasia, Teebi type
201	Cowden syndrome	157	CPTII	1521	Craniofrontonasal dysplasia-Poland anomaly syndrome
99932	Cow's milk hypersensitivity	228302	CPTII, adult-onset form	1520	Craniofrontonasal syndrome
70472	COX deficiency, French-Canadian type	228305	CPTII, hepatocardiomuscular form	50814	Craniolenticulosutural dysplasia
781	Coxiellosis	228308	CPTII, lethal systemic form	85184	Craniometadiaphyseal dysplasia, wormian bone type
1508	Coxoauricular syndrome	228302	CPTII, myopathic form	1522	Craniometaphyseal dysplasia
1509	Coxopodopatellar syndrome	228308	CPTII, neonatal form	1524	Craniomicromelic syndrome
254920	COXPD2	228305	CPTII, severe infantile form	54595	Craniopharyngioma
254925	COXPD4	3286	CPVT	63260	Craniorachischisis
137908	COXPD5	35173	CPXD	157832	Craniorhiny
254930	COXPD7	2081	Cramer-Niederdellmann syndrome	1541	Craniosynostosis, Boston type
319504	COXPD8	202	Crandall syndrome	2145	Craniosynostosis, Herrmann-Opitz type
319509	COXPD9	1512	Crane-Heise syndrome	1527	Craniosynostosis, Philadelphia type
314637	COXPD10	97339	Cranial dural arteriovenous fistula	1541	Craniosynostosis, Warman type
324535	COXPD11	97339	Cranial dural arteriovenous malformations	1532	Craniosynostosis-alopecia-brain defect syndrome
314051	COXPD12	268820	Cranial meningocele	85199	Craniosynostosis-anal anomalies-porokeratosis syndrome
319514	COXPD13	98919	Cranial variant of GBS	1530	Craniosynostosis-cataract syndrome
319519	COXPD14	98919	Cranial variant of Guillain-Barré syndrome	2872	Craniosynostosis-congenital heart disease-intellectual disability syndrome
319524	COXPD15	420485	Cranio-cervical dystonia with laryngeal and upper-limb involvement	1538	Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome
352563	COXPD16	2115	Cranio-facio-digitogenital syndrome	284149	Craniosynostosis-dental anomalies
369913	COXPD17	1525	Cranio-osteopathia	→53271	Craniosynostosis-dysmorphism-brachydactyly syndrome
420728	COXPD20	2053	Craniocarpotarsal dysplasia	1533	Craniosynostosis-fibular aplasia syndrome
420733	COXPD21	2053	Craniocarpotarsal dystrophy	171839	Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome
444013	COXPD23	7	Craniocerebellocardiac dysplasia	52054	Craniosynostosis-intracranial calcifications syndrome
444458	COXPD24	1513	Craniodiaphyseal dysplasia	1540	Craniosynostosis-midfacial hypoplasia-foot abnormalities syndrome
447954	COXPD25	1514	Craniodigital-intellectual disability syndrome	1528	Craniotelencephalic dysplasia
477684	COXPD26	1515	Cranoectodermal dysplasia	2095	Craniosynostosis-dysostosis-diaphyseal hyperplasia syndrome
477774	COXPD27	85168	Craniofacial conodysplasia	314555	Craniofacial dysplasia-osteopenia syndrome
466784	COXPD28	1681	Craniofacial duplication	459061	Craniofacial dysplasia-short stature-
478029	COXPD29	1777	Craniofacial dysmorphism-coloboma-corpus callosum agenesis syndrome		
478042	COXPD30	→1394	Craniofacial dysmorphism-skeletal anomalies-intellectual disability syndrome		
2444	CPAM	1798	Craniofacial dysostosis-diaphyseal hyperplasia syndrome		
280827	CPAM type 0	2095	Craniofacial dysostosis-genital, dental, cardiac anomalies syndrome		
280832	CPAM type 1	314555	Craniofacial dysplasia-osteopenia syndrome		
280840	CPAM type 2	459061	Craniofacial dysplasia-short stature-		
280847	CPAM type 3				
280854	CPAM type 4				
475	CPD IV				
300564	CPFE				
91359	CPI				
2016	CPLS syndrome				
2807	CPP				
759	CPP				
147	CPS1 deficiency				
147	CPS1D				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	hypertrichosis-hypoplasia of labia majora syndrome	1548	Cryptorchidism-arachnodactyly-intellectual disability syndrome		muscle or bone atrophy
75373	CRAPB	1549	Cryptosporidiosis	889	Cutaneous hypersensitivity vasculitis
275543	CRASH syndrome	357329	Cryptosporidiosis-chronic cholangitis-liver disease syndrome	178475	Cutaneous infectious botulism
184	CRBM	98967	Crystalline stromal dystrophy	423717	Cutaneous larva migrans
71	CRD	101068	CSCD	889	Cutaneous leukocytoclastic angiitis
52503	Creatine transporter deficiency	443079	CSCR	889	Cutaneous leukocytoclastic vasculitis
99854	Cree leukoencephalopathy	35122	CSID	79455	Cutaneous local mastocytoma
504	Creeping myiasis	306446	CSID with minimal starch tolerance	79490	Cutaneous lymphangioma circumscripum
280569	Crescentic glomerulonephritis	306474	CSID with starch and lactose intolerance	79455	Cutaneous mastocytoma
90290	CREST syndrome	306436	CSID with starch intolerance	90395	Cutaneous mucinosis of infancy
281	Cri du chat syndrome	306462	CSID without starch intolerance	79140	Cutaneous neuroendocrine carcinoma
281190	CRIE	306486	CSID without sucrose intolerance	439729	Cutaneous PAN
205	Crigler-Najjar syndrome	1465	CSS	439729	Cutaneous periarteritis nodosa
79234	Crigler-Najjar syndrome type 1	100008	CST3-related amyloidosis	2881	Cutaneous photosensitivity-lethal colitis syndrome
79235	Crigler-Najjar syndrome type 2	329217	CSV	439729	Cutaneous polyarteritis nodosa
99827	Crimean hemorrhagic fever	725	CSWS	451607	Cutaneous pseudolymphoma
99827	Crimean-Congo hemorrhagic fever	725	CSWSS syndrome	889	Cutaneous small vessel vasculitis
499085	CRION	70591	CTEPH	178475	Cutaneous toxin-mediated botulism
1545	Crisponi syndrome	436159	CTLA-4 haploinsufficiency with autoimmune infiltration disease	464321	Cutaneovisceral angiomatosis-thrombocytopenia syndrome
1461	Criss-cross atrioventricular relationships	247525	CTLN1	1555	Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome
1461	Criss-cross heart	247585	CTLN2	228285	Cutis laxa acquisita
891	Criswick-Schepens syndrome	909	CTX	221145	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies
313838	CRMCC	158	CUD	2962	Cutis laxa-corneal clouding-intellectual disability syndrome
1380	Crome syndrome	420584	Culler-Jones syndrome	171719	Cutis laxa-Marfanoid syndrome
2930	Cronkhite-Canada syndrome	3207	Curatolo-Cilio-Pessagno syndrome	1556	Cutis marmorata telangiectatica congenita
2719	Cross syndrome	98960	Curly fiber corneal dystrophy	→357225	Cutis verticis gyrata-intellectual disability syndrome
2935	Crossed polysyndactyly	307766	Curly hair-acral keratoderma-caries syndrome	→357225	Cutis verticis gyrata-retinitis pigmentosa-neurosensory deafness syndrome
439881	Croupous bronchitis	→1071	Curly hair-ankyloblepharon-nail dysplasia syndrome	→357225	Cutis verticis gyrata-retinitis pigmentosa-neurosensory hearing loss syndrome
207	Crouzon craniofacial dysostosis	1525	Currarino disease	→357225	Cutis verticis gyrata-retinitis pigmentosa-sensorineural deafness syndrome
207	Crouzon disease	1525	Currarino idiopathic osteoarthropathy	→357225	Cutis verticis gyrata-retinitis pigmentosa-sensorineural hearing loss syndrome
93262	Crouzon syndrome-acanthosis nigricans syndrome	1552	Currarino syndrome	→357225	Cutis verticis gyrata-thyroid aplasia-intellectual disability syndrome
93262	Crouzon-dermoskeletal syndrome	1552	Currarino triad	3327	Cutler-Bass-Romshe syndrome
2905	Crow-Fukase syndrome	640	Current pressure-sensitive neuropathy	1572	CVID
290	CRS	952	Curry-Hall syndrome	466670	Cyanide poisoning
→247691	CRV	1553	Curry-Jones syndrome		
411527	CRVO	96253	Cushing disease		
91139	Cryoglobulinemia type 1	99889	Cushing syndrome due to ectopic ACTH secretion		
91138	Cryoglobulinemic vasculitis	189427	Cushing syndrome due to macronodular adrenal hyperplasia		
1546	Cryptococcosis	53721	Cutaneomeningo spinal angiomas		
2032	Cryptogenic fibrosing alveolitis	2451	Cutaneous and mucosal venous malformation		
163708	Cryptogenic late-onset epileptic spasms	280779	Cutaneous collagenous vasculopathy		
468635	Cryptogenic multifocal ulcerous stenosing enteritis	329324	Cutaneous hemangioma with		
1302	Cryptogenic organizing pneumonia				
1547	Cryptomicrotia-brachydactyly syndrome				
1547	Cryptomicrotia-brachydactyly-excess fingertip arch syndrome				
2052	Cryptophthalmos-syndactyly syndrome				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
306692	Cyanide-induced parkinsonism	2437	Czeizel-Losonci syndrome	2806	Dawson's encephalitis
2686	Cyclic neutropenia	90038	D+HUS	1425	DBQD
228379	Cyclosporine-induced folliculodystrophy	356978	D,L-2-HGA	2143	DBS/FOAR syndrome
210	Cyclosporiasis	356978	D,L-2-hydroxyglutaric aciduria	1775	DC
79493	CYLD cutaneous syndrome	79315	D-2-HGA	79456	DCM
171886	Cylindrical spirals myopathy	79315	D-2-hydroxyglutaric aciduria	66634	DCMA syndrome
90795	CYP11B1 deficiency	79315	D-2-hydroxyglutaric aciduria	75381	DCMD
2674	Cyprus facial-neuromusculoskeletal syndrome	93599	D-glycerate dehydrogenase deficiency	1653	DD
212	Cystathionase deficiency	941	D-glycerate kinase deficiency	99789	DD-I
212	Cystathione gamma-lyase deficiency syndrome	941	D-glyceric aciduria	99791	DD-II
394	Cystathionine beta-synthase deficiency	2134	D-HUS	→231568	DDEB, Cockayne-Touraine type
212	Cystathioninuria	93581	D-HUS with anti-factor H antibodies	231568	DDEB, generalized
100008	Cystatin amyloidosis	93578	D-HUS with B factor anomaly	231568	DDEB, Pasini and Cockayne-Touraine types
400	Cystic echinococcosis	93575	D-HUS with C3 anomaly	→231568	DDEB, Pasini type
586	Cystic fibrosis	357008	D-HUS with DGKE deficiency	231568	DDEB-gen
2575	Cystic fibrosis-gastritis-megaloblastic anemia syndrome	93579	D-HUS with H factor anomaly	99970	DDLS
2111	Cystic hamartoma of lung and kidney	93580	D-HUS with I factor anomaly	79499	DDOD syndrome
79486	Cystic hygroma	93576	D-HUS with MCP/CD46 anomaly	52368	DDON syndrome
85136	Cystic leukoencephalopathy without megalencephaly	217023	D-HUS with thrombomodulin anomaly	300536	DDOST-CDG
229	Cystic medial necrosis of aorta	1146	DA1	488647	DDX41-related hematologic malignancy predisposition syndrome
1560	Cysticercosis	1146	DA1A	2962	De Barsy syndrome
213	Cystinosis	329457	DA5D	1130	De Die-Smulders-Vles-Fryns syndrome
214	Cystinuria	1495	Da Silva syndrome	1598	De Grouchy syndrome
93612	Cystinuria type A	251515	DA10	→782	De Hauwere syndrome
93613	Cystinuria type B	458768	Dabska tumor	→782	De Hauwere-Chitty syndrome
214	Cystinuria-lysuria syndrome	141083	Dacryocele	56304	De la Chapelle dysplasia
75381	Cystoid macular dystrophy	1562	Dacryocystitis-osteopoikilosis syndrome	393	De la Chapelle syndrome
472	Cystoisosporiasis	141083	Dacryocystocele	3157	De Morsier syndrome
180261	Cystsarcoma phyllodes of the breast	2186	Daentl-Townsend-Siegel syndrome	→910	De Sanctis-Cacchione syndrome
498228	Cystsarcoma phyllodes of the prostate	1563	Dahlberg syndrome	1570	De Smet-Fabry-Fryns syndrome
70472	Cytochrome C oxidase deficiency, French-Canadian type	1563	Dahlberg-Borer-Newcomer syndrome	33355	De Vaal disease
70472	Cytochrome oxidase deficiency, Saguenay-Lac-Saint-Jean type	2181	Daish-Hardman-Lamont syndrome	71277	De Vivo disease
95702	Cytomegalic congenital adrenal hypoplasia	275523	DALD	3214	Deaf blind hypopigmentation syndrome, Yemenite type
137698	Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	1183	Dancing eye syndrome	90024	Deafness with labyrinthine aplasia, microtia, and microdontia
94087	Cytophagic histiocytic panniculitis	1183	Dancing eye-dancing feet syndrome	2663	Deafness-cataract-skeletal anomalies syndrome
477787	Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder	→42775	Dandy-Walker malformation-facial hemangioma syndrome	3241	Deafness-craniofacial syndrome
137678	Czech dysplasia, metatarsal type	1566	Dandy-Walker malformation-postaxial polydactyly syndrome	52368	Deafness-dystonia-optic neuronopathy syndrome
2736	Czeizel syndrome	2091	Daneman-Davy-Mancer syndrome	3232	Deafness-ear malformation-facial palsy syndrome
2917	Czeizel-Brooser syndrome	34587	Danon disease	3220	Deafness-enamel hypoplasia-nail defects syndrome
		→1426	Dappled diaphyseal dysplasia	254898	Deafness-encephaloneuropathy-obesity-valvulopathy syndrome
		218	Darier disease	3218	Deafness-epiphyseal dysplasia-short stature syndrome
		316	Darier-Gottron disease	3224	Deafness-genital anomalies-metacarpal and metatarsal
		218	Darier-White disease		
		390	Darling disease		
		293978	DAVID syndrome		
		75565	Davies disease		

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	synostosis syndrome	158	Deficiency of plasma-membrane carnitine transporter	444002	Del(11)(q22.2q22.3)
3237	Deafness-Hermann type symphalangism syndrome	141330	Degner syndrome	2308	Del(11)(q23.3)
90646	Deafness-hypogonadism syndrome	679	Degos disease	2308	Del(11)(qter)
94064	Deafness-infertility syndrome	315	Degos genodermatosis "en cocardes"	313884	Del(12)(p12.1)
85321	Deafness-intellectual disability syndrome, Martin-Probst type	3202	Dehydrated hereditary stomatocytosis	280325	Del(12)(p13.33)
3226	Deafness-lymphedema-leukemia syndrome	64748	Dejerine-Sottas syndrome	94063	Del(12)(q14)
2408	Deafness-nephritis-ano-rectal malformation syndrome	2318	Dekaban-Arima syndrome	289513	Del(12)(q15)(q21.1)
3230	Deafness-oligodontia syndrome	1627	Del (5)(q35)	412035	Del(13)(q12.3)
		1627	Del (5)(qter)	1587	Del(13)(q14)
79500	Deafness-onychodystrophy-osteodystrophy-intellectual disability syndrome	401986	Del(1)(p31p32)	96168	Del(13)(q34)
79500	Deafness-onychodystrophy-osteodystrophy-intellectual disability-seizures syndrome	456298	Del(1)(p35.2)	261120	Del(14)(q11.2)
		1606	Del(1)(p36)	261144	Del(14)(q12)
79500	Deafness-onychodystrophy-osteodystrophy-intellectual disability syndrome	250989	Del(1)(q21)	264200	Del(14)(q22q23)
		250999	Del(1)(q41q42)	401935	Del(14)(q24.1q24.3)
→52368	Deafness-opticoacoustic nerve atrophy-dementia syndrome	238769	Del(1)(q44)	261183	Del(15)(q11.2)
123	Deafness-pili torti-hypogonadism syndrome	293948	Del(1)p(21.3)	199318	Del(15)(q13.3)
3219	Deafness-skeletal dysplasia-coarse face with full lips syndrome	363680	Del(2)(p13.2)	261190	Del(15)(q14)
3219	Deafness-skeletal dysplasia-lip granuloma syndrome	261349	Del(2)(p15p16.1)	94065	Del(15)(q24)
3217	Deafness-small bowel diverticulosis-neuropathy syndrome	163693	Del(2)(p21)	261211	Del(16)(p11.2p12.2)
3221	Deafness-thyroid hormone resistance syndrome	369881	Del(2)(p21) without cystinuria	261236	Del(16)(p13.11)
3239	Deafness-vitiligo-achalasia syndrome	228402	Del(2)(q23.1)	500055	Del(16)(p13.2)
→2697	Deal-Barrat-Dillon syndrome	1617	Del(2)(q24)	352629	Del(16)(q24.1)
79411	DEB, bullous dermolysis of the newborn	251014	Del(2)(q31.1)	261250	Del(16)(q24.3)
89843	DEB, pruriginosa	251019	Del(2)(q32)	97685	Del(17)(q11)
79411	DEB-BDN	251019	Del(2)(q32q33)	261265	Del(17)(q12)
89843	DEB-Pr	251028	Del(2)(q33.1)	363958	Del(17)(q21.31)
79410	DEB-Pt	1001	Del(2)(q37)	261279	Del(17)(q23.1q23.2)
431361	DECR deficiency with hyperlysinemia	1621	Del(3)(q13)	254346	Del(19)(p13.12)
99970	Dedifferentiated liposarcoma	356947	Del(3)(q26q27)	357001	Del(19)(p13.13)
397587	Deep dermatophytosis	397695	Del(3)(q27.3)	217346	Del(19)(q13.11)
31150	Defective adenosine triphosphate-binding cassette transporter A1	65286	Del(3)(q29)	261295	Del(20)(p12.3)
75496	Defective biosynthesis of proteodermatan sulfate	435638	Del(3)p(25.3)	313781	Del(20)(p13)
60	Deficiency in Alpa-1-proteinase inhibitor	238750	Del(4)(q21)	444051	Del(20)(q11.2)
293978	Deficiency in anterior pituitary function-variable immunodeficiency syndrome	228384	Del(5)(q14.3)	261311	Del(20)(q13.33)
404546	Deficiency of IL-36R antagonist	314655	Del(5)(q31.3)	261323	Del(21)(q22.11q22.12)
404546	Deficiency of IL-36Ra	251046	Del(6)(p22)	268261	Del(21)(q22.13q22.2)
		171829	Del(6)(q16)	96123	Del(22)
		251056	Del(6)(q25)	261476	Del(X)(p21)
		251061	Del(7)(q31)	1643	Del(X)(p23)
		251066	Del(8)(p11.2)	3034	Delayed membranous cranial ossification
		251071	Del(8)(p23.1)	3038	Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome
		284160	Del(8)(q21.11)	456298	Deletion 1p35.2
		508488	Del(8)(q24.3)	1606	Deletion 1p36
		2496	Del(8)q(13)	1606	Deletion 1pter
		324313	Del(9)(p13)	1001	Deletion 2q37
		352665	Del(9)(q21)	1001	Deletion 2q37-qter
		401923	Del(9)(q31.1q31.3)	281	Deletion 5p
		495818	Del(9)(q33.3q34.11)	1627	Deletion 5q35
		284169	Del(10)(p11.21p12.31)	904	Deletion 7q11.23
		276413	Del(10)(q22.3q23.3)	284160	Deletion 8q21.11
		893	Del(11)(p13)	502	Deletion 8q24.1

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
508488	Deletion 8q24.3	166265	Dentinogenesis imperfecta type 3	35107	Desmosterolosis
495818	Deletion 9q33.3q34.11	49042	Dentinogenesis imperfecta without osteogenesis imperfecta	98852	Desquamative interstitial pneumonia
284169	Deletion 10p11.21p12.31	166260	Dentinogenesis imperfecta, Shields type 2	158014	Destombes-Rosaï-Dorfman disease
276413	Deletion 10q22.3q23.3	166265	Dentinogenesis imperfecta, Shields type 3	163703	Devastating epileptic encephalopathy in school-aged children
893	Deletion 11p13	71267	Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome	313892	Developmental and speech delay due to SOX5 deficiency
94063	Deletion 12q14	77295	Dentoleukoencephalopathy	79157	Developmental delay due to 2-methylbutyryl-CoA dehydrogenase deficiency
289513	Deletion 12q15q21.1	220	Denys-Drash syndrome	289307	Developmental delay due to ALDH6A1 deficiency
1587	Deletion 13q14	3177	Der Kaloustian-Jarudi-Khoury syndrome	289307	Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency
1590	Deletion 13q32	3270	Der Kaloustian-McIntosh-Silver syndrome	289307	Developmental delay due to MMSDH deficiency
1600	Deletion 18q	369950	Der(8)t(8;12)	329195	Developmental delay with ASD and gait instability
96123	Deletion 22	96170	Der(22)t(11;22) syndrome	329195	Developmental delay with autism spectrum disorder and gait instability
1647	Delleman syndrome	36397	Dercum disease	163988	Developmental delay-deafness syndrome, Hildebrand type
1647	Delleman-Oorthuys syndrome	1656	Dermatitis herpetiformis	79134	Developmental delay-epilepsy-neonatal diabetes syndrome
79101	Delta1-pyrroline-5-carboxylate dehydrogenase deficiency	→137834	Dermato-cardio-skeletal syndrome, Borrone type	99989	Developmental delay-epilepsy-neonatal diabetes syndrome, intermediate form
35664	Delta-1-pyrroline 5-carboxylate synthetase deficiency	31112	Dermatofibrosarcoma protuberans	2101	Developmental delay-hypotonia-extremities hypertrophy syndrome
231237	Delta-beta-thalassemia	1659	Dermatoleukodystrophy	459061	Developmental delay-short stature-dysmorphic features-sparse hair syndrome
219	Delta-sarcoglycanopathy	221	Dermatomyositis	79107	Developmental malformations-deafness-dystonia syndrome
168782	Dementia infantilis	1657	Dermatoosteolysis, Kirghizian type	209908	Developmental verbal dyspraxia
97353	Dementia pugilistica	86920	Dermatopathia pigmentosa reticularis	71211	Devic disease
283	Demodicidosis	36426	Dermatostomatitis, Stevens Johnson type	→3464	Devriendt-Legius-Fryns syndrome
283	Demodicosis	79149	Dermochondrocorneal dystrophy	1014	Devriendt-Vandenbergh-Fryns syndrome
314451	Demons-Meigs syndrome	141051	Dermoid cyst of the face	403	Dexamethasone-sensitive hypertension
79134	DEND syndrome	141046	Dermoid cyst of the neck	1666	Dextrocardia
86903	Dendritic cell sarcoma not otherwise specified	1660	Dermodontodysplasia	→244	Dextrocardia-bronchiectasis-sinusitis syndrome
228423	Dendritic cell, monocyte, B and NK lymphoid deficiency	99688	Dermotrichic syndrome	99828	DF
99828	Dengue fever	1916	DES embryofetopathy	383	DFNX2
99828	Dengue virus infection	1916	DES syndrome	31112	DFSP
2109	Dennis-Fairhurst-Moore syndrome	1425	Desbuquois dysplasia	49042	DGI
93571	Dense deposit disease	1425	Desbuquois syndrome	49042	DGI without OI
1652	Dent disease	163703	DESC syndrome	166260	DGI-2
93622	Dent disease type 1	228123	Desert fever	373	DGSX
93623	Dent disease type 2	228123	Desert rheumatism	319651	DHFR deficiency
1652	Dent syndrome	98909	Desmin-related myofibrillar myopathy		
2095	Dental and eye anomalies-patent ductus arteriosus-normal intelligence syndrome	84132	Desmin-related myopathy with Mallory body-like inclusions		
1077	Dental ankylosis	98909	Desminopathy		
101	Dentatorubral pallidoluysian atrophy	873	Desmoid tumor		
101	Dentatorubropallidoluysian atrophy	873	Desmoid type fibromatosis		
1653	Dentin dysplasia	251940	Desmoplastic infantile astrocytoma/ganglioglioma		
314721	Dentin dysplasia type 1 with microdontia and shape anomalies	83469	Desmoplastic small round cell tumor		
99789	Dentin dysplasia type I	251863	Desmoplastic/nodular medulloblastoma		
99791	Dentin dysplasia type II				
99792	Dentin dysplasia-sclerotic bones syndrome				
49042	Dentinogenesis imperfecta				
166260	Dentinogenesis imperfecta type 2				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
139518	dHMN1	628	Diastrophic dwarfism	168811	Diffuse malignant peritoneal mesothelioma
139525	dHMN2	628	Diastrophic dysplasia	2123	Diffuse neonatal hemangiomatosis
139547	dHMN3 and dHMN4	276603	Diazoxide-resistant focal hyperinsulinism due to Kir6.2 deficiency	86918	Diffuse palmoplantar hyperkeratosis-acrocyanosis syndrome
139536	dHMN5	276598	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	369999	Diffuse palmoplantar keratoderma with painful fissures
98920	dHMN6	2195	Dicarboxylic aminoaciduria	2337	Diffuse palmoplantar keratoderma, Bothnian type
139589	dHMN7	284343	DICER1 syndrome	86918	Diffuse palmoplantar keratoderma-acrocyanosis syndrome
357043	dHMN with upper motor neuron signs	180086	Didelphys uterus	171700	Diffuse panbronchiolitis
139552	dHMNJ	3463	DIDMOAD syndrome	71274	Diffuse peritoneal leiomyomatosis
75376	DHRD	370046	Didymosis aplasticosebacea	66627	Diffuse-type GCT
49042	DI	1672	Diencephalic cachexia	66627	Diffuse-type giant cell tumor
1914	di Sala syndrome	1672	Diencephalic syndrome	567	DiGeorge sequence
166260	DI-2	1672	Diencephalic syndrome of childhood	567	DiGeorge syndrome
251940	DIA/DIG	1672	Diencephalic syndrome of emaciation	238	Digestive duplication
3463	Diabetes insipidus-diabetes mellitus-optic atrophy-deafness syndrome	319192	Diencephalic-mesencephalic junction dysplasia	141071	Digestive duplication cyst of the tongue
3464	Diabetes-hypogonadism-deafness-intellectual disability syndrome	1916	Diethylstilbestrol embryofetopathy	1305	Digital anomalies with short palpebral fissures and atresia of esophagus or duodenum
1926	Diabetic embryopathy	1916	Diethylstilbestrol syndrome	391641	Digital anomalies with short palpebral fissures and atresia of esophagus or duodenum type 1
85446	Dialysis-related amyloidosis	146	Differentiated thyroid carcinoma	352487	Digital anomalies-intellectual disability-short stature syndrome
85446	Dialysis-related arthropathy	90060	Diffuse alveolar hemorrhage	2926	Digital extensor muscle aplasia-polyneuropathy
275523	Dianzani autoimmune lymphoproliferative disease	324	Diffuse angiokeratoma	31828	Digitalis poisoning
494444	DIAPH1-related sensorineural deafness-thrombocytopenia syndrome	251595	Diffuse astrocytoma	→79500	Digitorenocerebral syndrome
494444	DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	404437	Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	1146	Digitotalar dysmorphism
66637	Diaphanospondylodysostosis	79456	Diffuse cutaneous maculopapulous mastocytosis	973	Digits 2-5 hypodactyly, unilateral
255182	Diaphorase deficiency	79456	Diffuse cutaneous mastocytosis	973	Digits 2-5 oligodactyly, unilateral
2141	Diaphragmatic defect-limb deficiency-skull defect syndrome	220393	Diffuse cutaneous systemic scleroderma	319651	Dihydrofolate reductase deficiency
2059	Diaphragmatic hernia-abnormal face-distal limb anomalies syndrome	220393	Diffuse cutaneous systemic sclerosis	79244	Dihydrolipoamide acetyltransferase component of pyruvate dehydrogenase complex deficiency
2143	Diaphragmatic hernia-exomphalos-hypertelorism syndrome	2199	Diffuse erythrodermic palmoplantar keratoderma, Voerner type	2394	Dihydrolipoamide dehydrogenase deficiency
2143	Diaphragmatic hernia-hypertelorism-myopia-deafness syndrome	2199	Diffuse erythrodermic palmoplantar keratoderma, Vörner type	255182	Dihydrolipooyl dehydrogenase deficiency
98920	Diaphragmatic spinal muscular atrophy	702	Diffuse familial brain sclerosis	79244	Dihydrolipoylysine-residue acetyltransferase component of pyruvate dehydrogenase complex deficiency
404521	Diaphragmatic spinal muscular atrophy type 2	3165	Diffuse fasciitis with eosinophilia	226	Dihydropteridine reductase deficiency
1802	Diaphyseal dysplasia-anemia syndrome	497188	Diffuse intrinsic pontine glioma	38874	Dihydropyrimidinase deficiency
85182	Diaphyseal medullary stenosis-bone malignancy syndrome	300849	Diffuse large B-cell lymphoma of the central nervous system	1675	Dihydropyrimidine dehydrogenase deficiency
85182	Diaphyseal medullary stenosis-malignant fibrous histiocytoma syndrome	300888	Diffuse large B-cell lymphoma with chronic inflammation	38874	Dihydropyrimidinuria
97282	Diarrheogenic islet cell tumor	252031	Diffuse leptomeningeal melanocytosis	99102	Dilatation of the left atrial appendage
1671	Diastematomyelia	141209	Diffuse lymphangioma		
		141209	Diffuse lymphangiomatosis		
		141209	Diffuse lymphatic malformation		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
99102	Dilatation of the left auricle		osteopoikilosis	1154	Distal arthrogryposis with ophthalmoplegia
99101	Dilatation of the right atrial appendage	397587	Disseminated granulomatous dermatophytosis	254351	Distal del(7)(q11.23)
99101	Dilatation of the right atrial auricle	141209	Disseminated lymphangioma	261222	Distal del(16)(p11.2)
66634	Dilated cardiomyopathy with ataxia	141209	Disseminated lymphangiomatosis	319171	Distal del(17)(p13.1)
2229	Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome	141209	Disseminated lymphatic malformation	261257	Distal del(17)(p13.3)
231111	DILE	228264	Disseminated nevus anelasticus	261330	Distal del(22)(q11.2)
243343	Dimethylglycine dehydrogenase deficiency	71274	Disseminated peritoneal leiomyomatosis	36367	Distal deletion 1q
→3157	Dincsoy-Salih-Patel syndrome	79152	Disseminated superficial actinic porokeratosis	280	Distal deletion 4p
314002	Dinno syndrome	1620	Distal 3p deletion	96145	Distal deletion 4q
1493	Dionisi-Vici-Sabetta-Gambarara syndrome	1627	Distal 5q deletion	96125	Distal deletion 6p
497188	DIPG	254351	Distal 7q11.23 microdeletion syndrome	96126	Distal deletion 7p
227	Diphallia	261102	Distal 7q11.23 microduplication syndrome	1636	Distal deletion 7q36
1679	Diphtheria	1580	Distal 10p deletion	1642	Distal deletion 9p
128	Diphyllobothriasis	1590	Distal 13q deletion	96148	Distal deletion 10q
1681	Diprosopia	1596	Distal 15q deletion syndrome	2308	Distal deletion 11q
1681	Diprosopus	261222	Distal 16p11.2 microdeletion syndrome	280325	Distal deletion 12p
1756	Dipygus	319171	Distal 17p13.1 microdeletion syndrome	96149	Distal deletion 12q
210115	DIRA	261257	Distal 17p13.3 microdeletion syndrome	96168	Distal deletion 13q34
166291	Dirofilariasis	1597	Distal 17q deletion	96150	Distal deletion 14q
94064	DIS	261330	Distal 22q11.2 microdeletion syndrome	96129	Distal deletion 19p
35122	Disaccharide intolerance	261337	Distal 22q11.2 microduplication syndrome	96152	Distal deletion 20q
306446	Disaccharide intolerance with minimal starch tolerance	63273	Distal ABD-filaminopathy	261102	Distal dup(7)(q11.23)
306474	Disaccharide intolerance with starch and lactose intolerance	399096	Distal anoctaminopathy	261337	Distal dup(22)(q11.2)
306436	Disaccharide intolerance with starch intolerance	178400	Distal anterior compartment myopathy	293939	Distal dup(X)(q28)
306462	Disaccharide intolerance without starch intolerance	1146	Distal arthrogryposis type 1	96069	Distal duplication 1p36
306486	Disaccharide intolerance without sucrose intolerance	2053	Distal arthrogryposis type 2A	96070	Distal duplication 2p
90281	Discoid lupus erythematosus	1147	Distal arthrogryposis type 2B	96094	Distal duplication 2q
216694	Discordant ventriculoarterial and atrioventricular connections	376	Distal arthrogryposis type 3	96071	Distal duplication 3p
99052	Discrete fibromuscular subaortic stenosis	65720	Distal arthrogryposis type 4	96072	Distal duplication 4p
99051	Discrete fixed membranous subaortic stenosis	1154	Distal arthrogryposis type 5	96096	Distal duplication 4q
90394	Discrete papular lichen myxedematosus	329457	Distal arthrogryposis type 5 without ophthalmoparesis	96097	Distal duplication 5q
139420	Disease-associated transverse myelitis	329457	Distal arthrogryposis type 5 without ophthalmoplegia	1745	Distal duplication 6p
210272	Disembarkment syndrome	329457	Distal arthrogryposis type 5D	96098	Distal duplication 6q
2412	Dislocation of the hip-dysmorphism syndrome	1144	Distal arthrogryposis type 6	96074	Distal duplication 7p
2983	Disorder of sex development-intellectual disability syndrome	3377	Distal arthrogryposis type 7	96100	Distal duplication 8q
345	Dissecting cellulitis of the scalp	65743	Distal arthrogryposis type 8	96101	Distal duplication 9q
54251	Disseminated aseptic abscesses	115	Distal arthrogryposis type 9	96102	Distal duplication 10q
1306	Disseminated dermatofibrosis with	251515	Distal arthrogryposis type 10	96103	Distal duplication 11q
		376	Distal arthrogryposis type IIA	96105	Distal duplication 13q
		1154	Distal arthrogryposis type IIB	1705	Distal duplication 14q
		65720	Distal arthrogryposis type IID	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
				139518	Distal hereditary motor neuropathy type 1
				139525	Distal hereditary motor neuropathy type 2

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
139547	Distal hereditary motor neuropathy type 3 and type 4	603	Distal myopathy, Welander type		defects-peripheral vascular anomalies syndrome
139536	Distal hereditary motor neuropathy type 5	98911	Distal myotilinopathy	1916	Distilbene embryofetopathy
98920	Distal hereditary motor neuropathy type 6	399103	Distal nebulin myopathy	1685	Distomatosis
139589	Distal hereditary motor neuropathy type 7	2776	Distal osteolysis-short stature-intellectual disability syndrome	1685	Distomiasis
139536	Distal hereditary motor neuropathy type V	18	Distal renal tubular acidosis	404546	DITRA
357043	Distal hereditary motor neuropathy with upper motor neuron signs	→402041	Distal renal tubular acidosis type 1b	99099	Divided left atrium
139552	Distal hereditary motor neuropathy, Jerash type	→402041	Distal renal tubular acidosis type 1c	99098	Divided right atrium
139536	Distal HMN V	93610	Distal renal tubular acidosis with anemia	91131	DK1-CDG
1307	Distal limb deficiencies-micrognathia syndrome	139525	Distal spinal muscular atrophy type 2	3439	DK phocomelia syndrome
36367	Distal monosomy 1q	139547	Distal spinal muscular atrophy type 3	1775	DKC
1620	Distal monosomy 3p	206580	Distal spinal muscular atrophy type 4	300849	DLBCL of the CNS
280	Distal monosomy 4p	139536	Distal spinal muscular atrophy type 5	300888	DLBCL with chronic inflammation
96145	Distal monosomy 4q	139589	Distal spinal muscular atrophy with vocal cord paralysis	2394	DLD deficiency
96125	Distal monosomy 6p	3248	Distal symphalangism	252031	DLM
96126	Distal monosomy 7p	314588	Distal tetrasomy 15q	273	DM1
254351	Distal monosomy 7q11.23	609	Distal titinopathy	98896	DMD
1636	Distal monosomy 7q36	96069	Distal trisomy 1p36	243343	DMG dehydrogenase deficiency
1642	Distal monosomy 9p	96070	Distal trisomy 2p	243343	DMGDH deficiency
1580	Distal monosomy 10p	96094	Distal trisomy 2q	602	DMRV
96148	Distal monosomy 10q	96071	Distal trisomy 3p	99812	DNA ligase IV deficiency
2308	Distal monosomy 11q	96072	Distal trisomy 4p	443950	DNAJB2-related Charcot-Marie-Tooth disease type 2
280325	Distal monosomy 12p	96096	Distal trisomy 4q	443950	DNAJB2-related CMT2
96149	Distal monosomy 12q	96097	Distal trisomy 5q	251946	DNET
1590	Distal monosomy 13q	1745	Distal trisomy 6p	404443	DNMT3A-related overgrowth syndrome
96150	Distal monosomy 14q	96098	Distal trisomy 6q	1215	DOA+
1596	Distal monosomy 15q	96074	Distal trisomy 7p	91500	Dobrin syndrome
261222	Distal monosomy 16p11.2	261102	Distal trisomy 7q11.23	3262	Dobrow syndrome
261257	Distal monosomy 17p13.3	96100	Distal trisomy 8q	447737	DOCK2 deficiency
1597	Distal monosomy 17q	96101	Distal trisomy 9q	217390	DOCK8 immunodeficiency syndrome
96129	Distal monosomy 19p13.3	96102	Distal trisomy 10q	79322	Dol-P-mannosyltransferase deficiency
96152	Distal monosomy 20q	96103	Distal trisomy 11q	91131	Dolichol kinase deficiency
261330	Distal monosomy 22q11.2	96105	Distal trisomy 13q	2616	Dolichospondylic dysplasia
59135	Distal myopathy type 1	1705	Distal trisomy 14q	86309	Dolichyl-phosphate N-acetylgalactosamine phosphotransferase deficiency
399086	Distal myopathy type 3	1707	Distal trisomy 15q	3427	DOLV
178400	Distal myopathy with anterior tibial onset	96078	Distal trisomy 16p	231226	Dominant beta-thalassemia
34521	Distal myopathy with early respiratory muscle involvement	96106	Distal trisomy 16q	75376	Dominant drusen
63273	Distal myopathy with posterior leg and anterior hand involvement	3379	Distal trisomy 17q	158676	Dominant dystrophic epidermolysis bullosa, nails only
602	Distal myopathy with rimmed vacuoles	1716	Distal trisomy 18q	898	Dominant hyaloideoretinal dystrophy of Wagner
600	Distal myopathy with vocal cord weakness	1717	Distal trisomy 19q	244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis
602	Distal myopathy, Nonaka type	96107	Distal trisomy 20q	276580	Dominant KATP hyperinsulinism due to Kir6.2 deficiency
488650	Distal myopathy, Tateyama type	96109	Distal trisomy 22q	75376	Dominant radial drusen
609	Distal myopathy, Udd type	261337	Distal trisomy 22q11.2	90035	Donath-Landsteiner hemolytic anemia
		293939	Distal Xq28 microduplication syndrome	90035	Donath-Landsteiner syndrome
		98920	Distal-HMN type 6		
		→33001	Distichiasis-congenital heart		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2143	Donnai-Barrow syndrome		catecholamines	314485	dSMA5
508	Donohue syndrome	3411	Double uterus and obstructed hemivagina syndrome	139557	DSMAX
79500	DOOR syndrome	3411	Double uterus-hemivagina-renal agenesis syndrome	83469	DSRCT
79500	DOORS syndrome	8	Double Y syndrome	412181	DST-related epidermolysis bullosa simplex
1942	Doose syndrome	95474	Double-orifice mitral valve	99789	DTDP1
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency	79145	Dowling-Degos disease	99791	DTDP2
230	Dopamine beta-hydroxylase deficiency	870	Down syndrome	2639	Du Pan syndrome
98907	Dorfman-Chanarin disease	75376	Doyne honeycomb retinal dystrophy	50817	Duane anomaly-myopathy-scoliosis syndrome
3426	DORV	86309	DPAGT1-CDG	233	Duane retraction syndrome
423712	DORV with atrioventricular septal defect, pulmonary stenosis, heterotaxy	314621	DPG-plus syndrome	233	Duane syndrome
99046	DORV with non-committed subpulmonary VSD	71274	DPL	93293	Duane-radial ray syndrome
423693	DORV with subaortic or doubly committed VSD	79322	DPM1-CDG	261647	Duane-radial ray syndrome due to a point mutation
99043	DORV with subaortic or doubly committed VSD with pulmonary stenosis	329178	DPM2-CDG	261638	Duane-radial ray syndrome due to monosomy 20q13
99045	DORV with subpulmonary VSD	263494	DPM3-CDG	234	Dubin-Johnson syndrome
99043	DORV, Fallot type	231	Dracunculiasis	234	Dubin-Sprinz disease
99045	DORV-TGA	231	Dracunculosis	235	Dubowitz syndrome
869	Double A syndrome	220	Drash syndrome	98896	Duchenne muscular dystrophy
216694	Double discordance	33069	Dravet syndrome	280315	Duct-centric pancreatitis
1464	Double inlet left ventricle	→79500	DRC syndrome	293208	Dunbar syndrome
141091	Double nose	70594	DRD due to SRD	2442	Duncan disease
3427	Double outlet left ventricle	130	Dream disease	2348	Dunnigan syndrome
3426	Double outlet right ventricle	139402	DRESS syndrome	→293864	Duodenal and extrahepatic biliary atresia-hypoplastic pancreas-intestinal malrotation syndrome
423712	Double outlet right ventricle with atrioventricular septal defect, pulmonary stenosis, heterotaxy	101	DRPLA	1203	Duodenal atresia
→423693	Double outlet right ventricle with doubly committed ventricular septal defect	233	DRS	261102	Dup7q11.23D
99046	Double outlet right ventricle with non-committed subpulmonary ventricular septal defect	18	dRTA	250994	Dup(1)(q21.1)
423693	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect	→402041	dRTA type 1b	313947	Dup(2)(q23.1)
99043	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect with pulmonary stenosis	→402041	dRTA type 1c	294026	Dup(2)(q31.1)
→423693	Double outlet right ventricle with subaortic ventricular septal defect	93610	dRTA with anemia	96095	Dup(3)(q26)
99043	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect with pulmonary stenosis	264978	Drug or radiation exposure-related interstitial lung disease	96095	dup(3q) syndrome
99045	Double outlet right ventricle with subpulmonary ventricular septal defect	139402	Drug rash with eosinophilia and systemic symptoms	329802	Dup(5)(p13)
99037	Drug-induced AIHA	139402	Drug reaction eosinophilic systemic syndrome	228415	Dup(5)(q35)
90037	Drug-induced autoimmune hemolytic anemia	90037	Drug-induced AIHA	314034	Dup(7)(p22.1)
90037	Drug-induced localized lipodystrophy	90157	Drug-induced localized lipodystrophy	96121	Dup(7)(q11.23)
231111	Drug-induced lupus erythematosus	231111	Drug-induced lupus erythematosus	459074	Dup(7)(q36.3)
464453	Drug-induced methemoglobinemia	464453	Drug-induced methemoglobinemia	251076	Dup(8)(p23.1p23.1)
251325	Drug-induced vasculitis	251325	Drug-induced vasculitis	228399	Dup(8)(q12)
97368	Drug-related renal tubular dysgenesis	97368	Drug-related renal tubular dysgenesis	276422	Dup(10)(q22.3q23.3)
94086	Drummond syndrome	94086	Drummond syndrome	300305	Dup(11)p(15.4)
33069	DS	33069	DS	261229	Dup(14)(q11.2)
99887	DS-AMKL	99887	DS-AMKL	488280	Dup(14)(q32)
98920	dSMA1	98920	dSMA1	238446	Dup(15)(q11q13)
139525	dSMA2	139525	dSMA2	261204	Dup(16)(p11.2p12.2)
139547	dSMA3	139547	dSMA3	261243	Dup(16)(p13.11)
206580	dSMA4	206580	dSMA4	96078	Dup(16)(p13.3)
3286	Double tachycardia induced by			477817	Dup(17)(p11.2p12)
				217385	Dup(17)(p13.3)
				139474	Dup(17)(q11.2)
				261272	Dup(17)(q12)

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
217340	Dup(17)(q21.31)		mutation	→98808	Dystonia 14
261290	Dup(17p)	296	Dyschondroplasia	210571	Dystonia 16
447980	Dup(19)(p13.13)	1765	Dyschondrosteosis-nephritis syndrome	98811	Dystonia 18
363659	Dup(20)(q11.2)	41	Dyschromatosis symmetrica hereditaria	420492	Dystonia 23
261318	Dup(20p)	241	Dyschromatosis universalis hereditaria	420485	Dystonia 24
1727	Dup(22)(q11)	251946	Dysembryoplastic neuroepithelial tumor	329466	Dystonia 25
284180	Dup(X)(p22)	1766	Dysequilibrium syndrome	508093	Dystonia 29
284180	Dup(X)(p22.13p22.2)	99912	Dysgerminomatous germ cell cancer of the ovary	256	Dystonia musculorum deformans
314389	Dup(X)(q12-q13.3)	3010	Dysharmonic skeletal maturation-muscular fiber disproportion syndrome	412217	Dystonia-aphonia syndrome
261483	Dup(X)(q27.3q28)	1775	Dyskeratosis congenita	199351	Dystonia-parkinsonism, Pisan-Ruiz type
261344	Duplication 1q	3088	Dyskeratosis congenita with bilateral exudative retinopathy	293381	Dystrophia Helsinglandica
1738	Duplication 4p	412	Dyslipidemia type 3	293381	Dystrophia Smolandiensis
1742	Duplication 5p	1779	Dysmorphism-cleft palate-loose skin syndrome	79409	Dystrophic epidermolysis bullosa inversa
264450	Duplication 8p	289553	Dysmorphism-conductive hearing loss-heart defect syndrome	89843	Dystrophic epidermolysis bullosa pruriginosa
1752	Duplication 8q	1780	Dysmorphism-multiple structural anomalies syndrome	256	DYT1
96167	Duplication 8q/deletion 8p	2104	Dysmorphism-pectus carinatum-joint laxity syndrome	99657	DYT2
236	Duplication 9p	2282	Dysmorphism-short stature-deafness-disorder of sex development syndrome	53351	DYT3
1699	Duplication 12p	2282	Dysmorphism-short stature-deafness-pseudohermaphroditism syndrome	98805	DYT4
1715	Duplication 18p	1782	Dysosteoclerosis	98808	DYT5a
1727	Duplication 22q11.2	800	Dysostosis enchondralis metaepiphysaria, Catel-Hempel type	101150	DYT5b
261318	Duplication of 20p	1798	Dysostosis, Stanescu type	98806	DYT6
314621	Duplication of the pituitary gland	99082	Dysphagia lusoria	53583	DYT9
314621	Duplication of the pituitary gland-plus syndrome	1822	Dysplasia epiphysealis hemimelica	71517	DYT12
1738	Duplication of the short arm of chromosome 4	168621	Dysplasia of head of femur, Meyer type	98807	DYT13
1742	Duplication of the short arm of chromosome 5	2204	Dysplastic cortical hyperostosis	→98808	DYT14
236	Duplication of the short arm of chromosome 9	65285	Dysplastic gangliocytoma of the cerebellum	→36899	DYT15
1715	Duplication of the short arm of chromosome 18	325	Dysprothrombinemia	210571	DYT16
237	Duplication of urethra	2476	Dysraphism-cleft lip/palate-limb reduction defects syndrome	98811	DYT18
284180	Duplication Xp22	156731	Dyssegmental dysplasia, Rolland-Desbuquois type	306734	DYT21
3306	Duplication/inversion 15q11	1865	Dyssegmental dysplasia, Silverman-Handmaker type	420492	DYT23
97339	Dural sinus malformation	1804	Dyssegmental dysplasia-glaucoma syndrome	420485	DYT24
1656	Durhing-Brocq disease	85198	Dysspondyloenchondromatosis	329466	DYT25
233	DURS	71517	Dystonia 12	508093	DYT29
→331176	Dursun syndrome			2394	E3-deficient maple syrup urine disease
98984	Dusty cataract			231249	E-beta-thalassemia
3377	Dutch-Kentucky syndrome			2970	Eagle-Barret syndrome
→969	Dwarfism-stiff joint-ocular abnormalities syndrome			40923	Eales disease
→2616	Dwarfism-tall vertebrae syndrome			2554	Ear-patella-short stature syndrome
1566	DWM with postaxial polydactyly			1934	Early infantile epileptic encephalopathy
239	Dyggve-Melchior-Claussen disease			1934	Early infantile epileptic encephalopathy with suppression-bursts
2274	Dykes-Marks-Harper syndrome			1935	Early myoclonic encephalopathy
464306	DYRK1A-related intellectual disability syndrome			1935	Early myoclonic encephalopathy with suppression-bursts
268261	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion			98988	Early-onset anterior polar cataract
464311	DYRK1A-related intellectual disability syndrome due to a point			98988	Early-onset anterior subcapsular cataract
				1020	Early-onset autosomal dominant

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	Alzheimer disease		encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	1880	Ebstein malformation
98815	Early-onset benign childhood occipital epilepsy	496756	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome	313920	EBV-associated gastric carcinoma
98985	Early-onset cataract with Y-shaped suture opacities	352654	Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	289661	EBV-positive DLBCL of the elderly
1177	Early-onset cerebellar ataxia with retained tendon reflexes	→90340	Early-onset sarcoidosis	313920	EBVaGC
84132	Early-onset desmin-related myopathy	96369	Early-onset schizophrenia	494424	ECAA
1667	Early-onset diabetes mellitus with multiple epiphyseal dysplasia	505237	Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome	494424	ECCA
210571	Early-onset dystonia parkinsonism	364055	Early-onset severe retinal dystrophy	50944	Eccrine tumors-ectodermal dysplasia
488635	Early-onset epilepsy-intellectual disability-brain anomalies syndrome	313772	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	284	Echinococcus multilocularis infection
289266	Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation	98985	Early-onset sutural cataract	199332	ECO syndrome
411986	Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	256	Early-onset torsion dystonia	→1896	ECP syndrome
1020	Early-onset familial autosomal dominant Alzheimer disease	1243	Early-onset vitelliform macular dystrophy	99102	Ectasia of the left atrial appendage
494348	Early-onset familial noncirrhotic portal hypertension	98890	Early-onset X-linked optic atrophy	99102	Ectasia of the left auricle
256	Early-onset generalized limb-onset dystonia	98995	Early-onset zonular cataract	99101	Ectasia of the right atrial appendage
256	Early-onset generalized torsion dystonia	199343	EAST syndrome	99101	Ectasia of the right atrial auricle
88660	Early-onset hypertension with exacerbation in pregnancy	391320	East Texas bleeding disorder	35737	Ectasic coloboma
324290	Early-onset Lafora body disease	83594	Eastern equine encephalitis	→1071	Ectodermal dysplasia syndrome, Rapp-Hodgkin type
441452	Early-onset lamellar cataract	83594	Eastern equine encephalomyelitis	69083	Ectodermal dysplasia with natal teeth, Turnpenny type
79242	Early-onset multiple carboxylase deficiency	1973	Eastman-Bixler syndrome	1816	Ectodermal dysplasia, Berlin type
289377	Early-onset myopathy with fatal cardiomyopathy	166418	Eating epilepsy	→1071	Ectodermal dysplasia, Rapp-Hodgkin type
439212	Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome	166418	Eating reflex epilepsy	1818	Ectodermal dysplasia, trichodontoonychial type
91492	Early-onset non-syndromic cataract	166418	Eating seizures	→1658	Ectodermal dysplasia-absent dermatoglyphs syndrome
98991	Early-onset nuclear cataract	86880	EATL	140936	Ectodermal dysplasia-acanthosis nigricans syndrome
2828	Early-onset Parkinson disease	79406	EB progressive	→2036	Ectodermal dysplasia-adrenal cyst syndrome
2379	Early-onset parkinsonism-intellectual disability syndrome	79405	EBJ-I	1806	Ectodermal dysplasia-blindness syndrome
98992	Early-onset partial cataract	98956	EBMD	247827	Ectodermal dysplasia-cutaneous syndactyly syndrome
98993	Early-onset posterior polar cataract	319218	Ebola fever	1897	Ectodermal dysplasia-ectrodactyly-macular dystrophy syndrome
441447	Early-onset posterior subcapsular cataract	319218	Ebola hemorrhagic fever	1812	Ectodermal dysplasia-intellectual disability-central nervous system malformation syndrome
256	Early-onset primary dystonia	319218	Ebola virus disease	1883	Ectodermal dysplasia-sensorineural deafness syndrome
157941	Early-onset prion disease with prominent psychiatric features	89838	EBS, autosomal recessive K14	1883	Ectodermal dysplasia-sensorineural hearing loss syndrome
496641	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	79399	EBS, generalized intermediate	423454	Ectodermal dysplasia-short stature syndrome
500144	Early-onset progressive	79396	EBS, generalized severe	158668	Ectodermal dysplasia-skin fragility syndrome
		412181	EBS-AR BP230	247820	Ectodermal dysplasia-syndactyly syndrome
		412189	EBS-AR exophilin 5	448270	Ectopia cordis
		89838	EBS-AR KRT14	1885	Ectopia lentis syndrome
		79400	EBS-loc	1884	Ectopia lentis-chorioretinal
		257	EBS-MD		
		158681	EBS-migr		
		79397	EBS-MP		
		79401	EBS-O		
		158684	EBS-PA		
		89839	EBSS		
		1880	Ebstein anomaly of the tricuspid valve		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	dystrophy-myopia syndrome	230839	EDS, classic-like type		progressive kyphoscoliosis, myopathy, and hearing loss
99889	Ectopic ACTH secreting tumor	300179	EDS, kyphoscoliotic and hearing loss type	1899	Ehlers-Danlos syndrome, arthrochalasia type
231632	Ectopic aldosterone-producing tumor	1900	EDS, kyphoscoliotic type	1899	Ehlers-Danlos syndrome, arthrochalasis type
99889	Ectopic Cushing syndrome	2953	EDS, musculocontractural type	230851	Ehlers-Danlos syndrome, cardiac valvular type
95496	Ectopic neurohypophysis	1900	EDS, oculoscoliotic type	287	Ehlers-Danlos syndrome, classic type
2440	Ectrodactyly	75496	EDS, progeroid type	230839	Ehlers-Danlos syndrome, classic-like type
→1896	Ectrodactyly-cleft palate syndrome	157965	EDS, spondylocheirodysplastic type	1901	Ehlers-Danlos syndrome, dermatosparaxis type
→1896	Ectrodactyly-ectodermal dysplasia without clefting syndrome	230845	EDS, vascular-like type	75501	Ehlers-Danlos syndrome, fibronectin-deficient
1896	Ectrodactyly-ectodermal dysplasia-cleft lip/palate syndrome	230857	EDS/OI syndrome	75501	Ehlers-Danlos syndrome, fibronectinemic type
1892	Ectrodactyly-polydactyly syndrome	247820	EDSS	285	Ehlers-Danlos syndrome, hypermobile type
1894	Ectrodactyly-spina bifida-cardiopathy syndrome	247820	EDSS1	285	Ehlers-Danlos syndrome, hypermobility type
1997	Ectropion inferior-cleft lip and/or palate syndrome	247827	EDSS2	2953	Ehlers-Danlos syndrome, Kosho type
906	Eczema-thrombocytopenia-immunodeficiency syndrome	178464	Edström Myopathy	300179	Ehlers-Danlos syndrome, kyphoscoliotic and deafness type
98813	EDA-ID	3380	Edwards syndrome	300179	Ehlers-Danlos syndrome, kyphoscoliotic and hearing loss type
247827	EDCS	2668	Edwards-Patton-Dilly syndrome	1900	Ehlers-Danlos syndrome, kyphoscoliotic type
293936	EDICT syndrome	322	EEC	2953	Ehlers-Danlos syndrome, musculocontractural type
1895	Edinburgh malformation syndrome	1896	EEC syndrome	1900	Ehlers-Danlos syndrome, oculoscoliotic type
93308	EDM1	→1896	EEC syndrome without cleft lip/palate	75392	Ehlers-Danlos syndrome, periodontitis type
93307	EDM4	1897	EEM syndrome	75496	Ehlers-Danlos syndrome, progeroid type
93311	EDM5	357131	Effort subclavian vein thrombosis	157965	Ehlers-Danlos syndrome, spondylocheirodysplastic type
261	EDMD	101039	EFMR	286	Ehlers-Danlos syndrome, vascular type
98853	EDMD2	2070	EGE	230845	Ehlers-Danlos syndrome, vascular-like type
98855	EDMD3	183	EGPA	230857	Ehlers-Danlos/osteogenesis imperfecta syndrome
90309	EDS I	319218	EHF	1902	Ehrlichiosis
90318	EDS II	312	EHK	820	Ehrmann-Sneddon syndrome
285	EDS III	230839	Ehlers-Danlos syndrome due to tenascin-X deficiency	312	EI
286	EDS IV	90309	Ehlers-Danlos syndrome type 1	1934	EIEE
198	EDS IX	90318	Ehlers-Danlos syndrome type 2	165991	EIHI
286	EDS type 4	285	Ehlers-Danlos syndrome type 3	79106	Eiken syndrome
75497	EDS V	286	Ehlers-Danlos syndrome type 4	97214	Eisenmenger syndrome
1900	EDS VIA	75497	Ehlers-Danlos syndrome type 5	476096	EKC syndrome
1899	EDS VII	1900	Ehlers-Danlos syndrome type 6A	317	EKV
99875	EDS VIIA	1899	Ehlers-Danlos syndrome type 7	228240	Elastoderma
99876	EDS VIIIB	99875	Ehlers-Danlos syndrome type 7A		
1901	EDS VIIIC	99876	Ehlers-Danlos syndrome type 7B		
75392	EDS VIII	1901	Ehlers-Danlos syndrome type 7C		
82004	EDS with periventricular heterotopia	75392	Ehlers-Danlos syndrome type 8		
300179	EDS with progressive kyphoscoliosis, myopathy, and deafness	198	Ehlers-Danlos syndrome type 9		
300179	EDS with progressive kyphoscoliosis, myopathy, and hearing loss	75501	Ehlers-Danlos syndrome type 10		
75501	EDS X	2295	Ehlers-Danlos syndrome type 11		
2295	EDS XI	286	Ehlers-Danlos syndrome type IV		
230851	EDS, cardiac valvular type	198	Ehlers-Danlos syndrome type IX		
287	EDS, classic type	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		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
228243	Elastofibroma dorsi		calcification-retinal degeneration syndrome	449566	Eosinophilic angiocentric fibrosis
228254	Elastoma	3205	Encephalotrigeminal angiomas	901	Eosinophilic cellulitis
79148	Elastosis perforans serpiginosa	296	Enchondromatosis	402035	Eosinophilic colitis
228236	Elastotic striae	99075	Encircling double aortic arch	75566	Eosinophilic endocarditis
33445	Elejalde disease	83315	Endemic typhus	2070	Eosinophilic enteritis
221054	Elejalde syndrome	199332	Endocrine-cerebro-osteodysplasia syndrome	73247	Eosinophilic esophagitis
289	Ellis Van Creveld syndrome	876	Endodermal sinus tumor	3165	Eosinophilic fasciitis
2516	Ellis-Yale-Winter syndrome	252006	Endodermal sinus tumor of central nervous system	2070	Eosinophilic gastroenteritis
1299	Elsahy-Waters syndrome	252006	Endodermal sinus tumor of CNS	2070	Eosinophilic gastroenterocolitis
1997	Elsching syndrome	98974	Endoepithelial corneal dystrophy	99871	Eosinophilic granuloma
96170	Emanuel syndrome	213741	Endometrial adenoid cystic carcinoma	183	Eosinophilic granulomatosis with polyangiitis
439212	EMARDD	213726	Endometrial capillary carcinoma	482	Eosinophilic lymphogranuloma
1942	EMAS	213716	Endometrial squamous cell carcinoma	364055	EOSRD
3226	Emberger syndrome	213711	Endometrial stromal sarcoma	256	EOTD
180226	Embryonal carcinoma	213746	Endometrial transitional cell carcinoma	251880	Ependymoblastoma
48736	Embryonal carcinoma of the central nervous system	213721	Endometrial undifferentiated carcinoma	251636	Ependymoma
48736	Embryonal carcinoma of the CNS	454723	Endometrioid carcinoma of ovary	99169	Epiblepharon
99757	Embryonal rhabdomyosarcoma	137820	Endometriosis outside pelvis	185	Epibronchial right pulmonary vein syndrome
178315	Embryonal sarcoma of the liver	2022	Endomyocardial fibroelastosis	231742	Epibulbar lipodermoid-preauricular appendage-polythelia syndrome
1664	Embryonary disorganization syndrome	199323	Endophthalmitis	83314	Epidemic typhus
983	Embryonic testicular regression syndrome	209959	Endophthalmitis phacoanaphylactica	35125	Epidermal hamartoma syndrome
139431	EMEA	2790	Endosteal hyperostosis, Worth type	35125	Epidermal nevus syndrome
261	Emery-Dreifuss muscular dystrophy	85186	Endosteal sclerosis-cerebellar hypoplasia syndrome	497737	Epidermal nevus with epidermolytic hyperkeratosis
1927	Emery-Nelson syndrome	293936	Endothelial dystrophy-iris hypoplasia-congenital cataract-stromal thinning syndrome	302	Epidermodysplasia verruciformis
485418	EMILIN-1-related connective tissue disease	137602	Endotheliitis	46487	Epidermolysis bullosa acquisita
1031	Enamel-renal syndrome	1937	Eng-Strom syndrome	79404	Epidermolysis bullosa letalis
83600	Encephalitis lethargica	53540	Enhanced S-cone syndrome	412181	Epidermolysis bullosa simplex due to BP230 deficiency
221126	Encephaloclastic proliferative vasculopathy	60015	Enlarged parietal foramina	412189	Epidermolysis bullosa simplex due to exophilin 5 deficiency
2396	Encephalocranioscutaneous lipomatosis	83620	Enteric anendocrinosis	158668	Epidermolysis bullosa simplex due to plakophilin deficiency
3205	Encephalofacial angiomas	141071	Enteric duplication cyst of the tongue	79400	Epidermolysis bullosa simplex of palms and soles
71277	Encephalopathy due to GLUT1 deficiency	86880	Enteropathy-associated T-cell lymphoma	89839	Epidermolysis bullosa simplex superficialis
79155	Encephalopathy due to hydroxykynureninuria	86880	Enteropathy-type T-cell lymphoma	2325	Epidermolysis bullosa simplex with anodontia/hypodontia
139406	Encephalopathy due to prosaposin deficiency	85438	Enthesitis-related JIA	158681	Epidermolysis bullosa simplex with circinate migratory erythema
833	Encephalopathy due to sulfite oxidase deficiency	85438	Enthesitis-related juvenile idiopathic arthritis	79397	Epidermolysis bullosa simplex with mottled pigmentation
210128	Encephalopathy due to urocanase deficiency	1177	EOCA	257	Epidermolysis bullosa simplex with muscular dystrophy
51	Encephalopathy with basal ganglia calcification	1177	EOCARR	158684	Epidermolysis bullosa simplex with pyloric atresia
51	Encephalopathy with intracranial calcification and chronic lymphocytosis of cerebrospinal fluid	370334	EOE	89838	Epidermolysis bullosa simplex, autosomal recessive K14
319678	Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	73247	EoE	79396	Epidermolysis bullosa simplex, Dowling-Meara type
1261	Encephalopathy-intracerebral	1020	EOFAD	79399	Epidermolysis bullosa simplex,
		289377	EOMFC		
		168829	EOPPC		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	generalized intermediate	399329	Epiphysiolysis of the hip	502499	Erythema multiforme major
79396	Epidermolysis bullosa simplex, generalized severe	399329	Epiphysiolysis of the upper femur	502499	Erythema multiforme majus
79396	Epidermolysis bullosa simplex, herpetiformis	649	Episkopi blindness	231031	Erythema palmarum hereditarium
79399	Epidermolysis bullosa simplex, K��bner type	37612	Episodic ataxia type 1	308473	Erythrocyte epimerase deficiency galactosemia
79399	Epidermolysis bullosa simplex, Koebner type	97	Episodic ataxia type 2	308473	Erythrocyte galactose epimerase deficiency
79401	Epidermolysis bullosa simplex, Ogna type	79135	Episodic ataxia type 3	308473	Erythrocyte GALE deficiency
79400	Epidermolysis bullosa simplex, Weber-Cockayne type	79136	Episodic ataxia type 4	308473	Erythrocyte GALE-D
497737	Epidermolytic epidermal nevus	211067	Episodic ataxia type 5	171690	Erythrocyte lactate transporter defect
312	Epidermolytic hyperkeratosis	209967	Episodic ataxia type 6	308473	Erythrocyte UDP-galactose-4-epimerase deficiency
312	Epidermolytic ichthyosis	209970	Episodic ataxia type 7	308473	Erythrocyte uridine diphosphate galactose-4-epimerase deficiency
497737	Epidermolytic nevus	401953	Episodic ataxia type 8	314	Erythroderma desquamativum
2199	Epidermolytic palmoplantar keratoderma	37612	Episodic ataxia with myokymia	79394	Erythrodermic ichthyosis
2199	Epidermolytic palmoplantar keratoderma of Voerner	401953	Episodic ataxia with slurred speech	247165	Erythroedema polyneuritis
2199	Epidermolytic palmoplantar keratoderma of V��rner	79135	Episodic ataxia-vertigo-tinnitus-myokymia syndrome	315	Erythrokeratoderma "en cocardes"
497737	Epidermolytic verrucous epidermal nevus	53583	Episodic choreoathetosis/spasticity	316	Erythrokeratoderma progressiva symmetrica
141077	Epignathus	29822	Episodic spontaneous hypothermia	317	Erythrokeratoderma variabilis
65683	Epilepsy due to FCD	93928	Epispadias	317	Erythrokeratoderma variabilis, Mendes da Costa type
86911	Epilepsy with myoclonic absences	98956	Epithelial basement membrane dystrophy	1955	Erythrokeratoderma with ataxia
1942	Epilepsy with myoclonic-astatic seizures	293381	Epithelial recurrent erosion dystrophy	476096	Erythrokeratoderma-cardiomyopathy syndrome
1942	Epilepsy with myoclonic-tonic seizures	157791	Epithelioid hemangioendothelioma	50943	Erythrokeratolysis hiemalis
411986	Epilepsy-cortical blindness-intellectual disability-facial dysmorphism syndrome	293202	Epithelioid sarcoma	318	Erythroleukemia
1946	Epilepsy-dementia-amelogenesis imperfecta syndrome	254698	Epithelioid trophoblastic tumor	1956	Erythromelalgia
1948	Epilepsy-microcephaly-skeletal dysplasia syndrome	91414	Epithelioma calcificans of Malherbe	280379	Erythropoietic uroporphyrin associated with myeloid malignancy
1951	Epilepsy-telangiectasia syndrome	501	EPM2	99977	ESCC
725	Epileptic encephalopathy with continuous spike-and-wave during slow sleep	263516	EPM3	2405	Escher-Hirt syndrome
353217	Epileptic encephalopathy with global cerebral demyelination	163696	EPM4	2990	Escobar syndrome
79238	Epimerase deficiency galactosemia	402082	EPM5	2990	Escobar variant multiple pterygium syndrome
1819	Epimetaphyseal skeletal dysplasia	280620	EPM6	99976	Esophageal adenocarcinoma
1825	Epiphyseal dysplasia-hearing loss-dysmorphism syndrome	435438	EPM7	1199	Esophageal atresia
1824	Epiphyseal dysplasia-microcephaly-nystagmus syndrome	424027	EPM8	418945	Esophageal carcinoma, salivary gland type
1952	Epiphyseal stippling syndrome-osteoclastic hyperplasia syndrome	457265	EPM9	100047	Esophageal duplication cyst
496751	Epiphysial-vertebral-ear dysplasia-nose-plus associated findings syndrome	79278	EPP	99977	Esophageal epidermoid carcinoma
		2199	EPPK	506136	Esophageal NEN
		→182050	Epstein syndrome	506136	Esophageal neuroendocrine neoplasm
		313920	Epstein-Barr virus-associated gastric carcinoma	99977	Esophageal squamous cell carcinoma
		289661	Epstein-Barr virus-positive diffuse large B-cell lymphoma of the elderly	91138	Essential cryoglobulinemia
		85438	ERA	2056	Essential fructosuria
		35687	Erdheim-Chester disease	98981	Essential iris atrophy
		293381	ERED	91138	Essential mixed cryoglobulinemia
		999	Ermine phenotype	2843	Essential pentosuria
		160148	Eroded polypoid hyperplasia	3318	Essential thrombocythemia
		→79500	Eronen-Somer-Gustafsson syndrome	3318	Essential thrombocytosis
		222	Erosive pustular dermatosis of the scalp		
		228264	Eruptive collagenoma		
		90000	Erythema elevatum diutinum		
		502499	Erythema exudativum multiforme majus		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
1957	Esthesioneuroblastoma	182127	Extragonadal germinoma	466950	Facial dysmorphism-development delay-behavioral abnormalities syndrome due to WAC point mutation
785	Estrogen resistance syndrome	883	Extragonadal teratoma	352712	Facial dysmorphism-immunodeficiency-livedo-short stature syndrome
3318	ET	280811	Extralobar congenital bronchopulmonary sequestration	2588	Facial dysmorphism-intellectual disability-short stature-hearing loss syndrome
31826	Ethylene glycol poisoning	280811	Extralobar congenital pulmonary sequestration	412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-nontraumatic conjunctive cysts syndrome
51188	Ethylmalonic encephalopathy	2800	Extramammary Paget disease	412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome
983	ETRS	86850	Extramedullary myeloid tumor	1970	Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome
86880	ETTL	100022	Extramedullary soft tissue plasmacytoma	1778	Facial dysmorphism-shawl scrotum-joint laxity syndrome
2892	Euhidrotic ectodermal dysplasia	100002	Extraneural perineurioma	221083	Facial hemispasm
99172	Euryblepharon	52417	Extranodal marginal zone B-cell lymphoma	3020	Facial nerve palsy due to herpes zoster infection
466682	Euthyroid Graves ophthalmopathy	86879	Extranodal nasal NK/T cell lymphoma	3020	Facial nerve palsy due to VZV
466682	Euthyroid Graves orbitopathy	370334	Extraosseous Ewing sarcoma	85162	Facial onset sensory and motor neuronopathy
1959	Evans syndrome	370334	Extraosseous Ewing tumor	3237	Facio-audio-symphalangism
444463	Evans syndrome associated with primary immunodeficiency	137820	Extrapelvic endometriosis	1974	Facio-digito-genital syndrome, Kuwait type
496751	EVEN-plus syndrome	370334	Extraskeletal Ewing sarcoma	1300	Facio-genito-popliteal syndrome
2990	EVMPS	370334	Extraskeletal Ewing tumor	2143	Facio-oculo-acoustico-renal syndrome
251927	EVN	209916	Extraskeletal myxoid chondrosarcoma	2048	Facio-pharyngo-glossal diplegia with automatic-voluntary movement dissociation
319	Ewing sarcoma	1964	Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome	2048	Facio-pharyngo-glosso-masticatory diplegia
99734	Exercise-induced delayed-onset myotonia	251927	Extraventricular neurocytoma	374	Facioauriculovertebral dysplasia
165991	Exercise-induced hyperinsulinemic hypoglycemia	2725	Eye defects-arachnodactyly-cardiopathy syndrome	1973	Faciocardiorectal syndrome
165991	Exercise-induced hyperinsulinism	3172	Eyebrow duplication-syndactyly syndrome	3071	Faciocutaneoskeletal syndrome
466650	Exercise-induced malignant hyperthermia	139431	Eyelid myoclonia with and without absences	915	Faciocutaneoskeletal syndrome
466650	Exertional heat stroke	35909	F5F8D	915	Faciogenital dysplasia
289586	Exfoliative ichthyosis	957	F syndrome	269	Faciocapulohumeral dystrophy
→955	Exner syndrome	95	FA	269	Faciocapulohumeral muscular dystrophy
116	Exomphalos-macroglossia-gigantism syndrome	324	Fabry disease	269	Faciocapulohumeral myopathy
1962	Exostoses-anetodermia-brachydactyly type E syndrome	1969	FACES syndrome	98879	Factor IX deficiency
374	Expanded spectrum hemifacial microsomia	1167	Facial asymmetry-temporal seizures syndrome	220436	Factor V Quebec
322	Exstrophy-epispadias complex	141051	Facial dermoid cyst	98878	Factor VIII deficiency
440724	Extensive peripapillary myelinated nerve fibers	480701	Facial diplegia with paresthesias	300359	FACU
3294	Extensor tendons of finger anomalies	480701	Facial diplegia with paresthesias variant of GBS	306550	FADD-related immunodeficiency
141074	External auditory canal aplasia/hypoplasia	480701	Facial diplegia with paresthesias variant of Guillain-Barré syndrome	994	FADS
3023	External auditory canal atresia-vertical talus-hypertelorism syndrome	→3157	Facial dysmorphism-ambiguous genitalia-hypopituitarism-short limbs syndrome		
141074	External auditory canal stenosis/atresia	1969	Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome		
508533	EXTL3-related neuro-immuno-skeletal dysplasia syndrome	284169	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion		
231632	Extra-adrenal aldosterone-producing tumor				
168829	Extra-ovarian primary peritoneal carcinoma				
494424	Extracranial carotid artery aneurysm				
66662	Extracutaneous mastocytoma				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
882	FAH deficiency		deficiency		microdeletion
329308	FAHN	1416	Familial articular chondrocalcinosis	238578	Familial clubfoot due to 17q23.1q23.2 microduplication
→168569	Faisalabad histiocytosis	334	Familial atrial fibrillation	293150	Familial clubfoot due to PITX1 point mutation
3304	Fallot complex-intellectual disability-growth delay syndrome	615	Familial atrial myxoma	199315	Familial clubfoot with or without associated lower limb anomalies
86814	FAME	436242	Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease	464760	Familial CODA
397685	Familial hyperprolactinemia	300359	Familial atypical cold urticaria	47045	Familial cold autoinflammatory syndrome
86	Familial abdominal aortic aneurysm	404560	Familial atypical mole syndrome	247868	Familial cold autoinflammatory syndrome type 2
88619	Familial acute necrotizing encephalopathy	404560	Familial atypical multiple mole melanoma syndrome	47045	Familial cold urticaria
733	Familial adenomatous polyposis	404560	Familial atypical multiple mole melanoma-pancreatic carcinoma syndrome	300359	Familial cold urticaria with common variable immunodeficiency
261584	Familial adenomatous polyposis due to 5q22.2 microdeletion	86820	Familial avascular necrosis of femoral head	440437	Familial colorectal cancer Type X
261584	Familial adenomatous polyposis due to del(5)(q22.2)	402075	Familial BAV	238722	Familial congenital contralateral synkinesis
261584	Familial adenomatous polyposis due to monosomy 5q22.2	2398	Familial benign cervical lipomatosis	95494	Familial congenital hypopituitarism
404	Familial adrenal adenoma	2841	Familial benign chronic pemphigus	238722	Familial congenital mirror movements
95700	Familial adrenal hypoplasia with absent pituitary LH	1551	Familial benign copper deficiency	451612	Familial congenital nasolacrimal duct obstruction
95700	Familial adrenal hypoplasia with absent pituitary luteinizing hormone	363989	Familial benign flecked retina	91498	Familial congenital palsy of trochlear nerve
95700	Familial adrenal hypoplasia, miniature type	405	Familial benign hypercalcemia	86814	Familial cortical myoclonic tremor and epilepsy
86814	Familial adult myoclonic epilepsy	1551	Familial benign hypocupremia	319189	Familial cortical myoclonus
164736	Familial advanced sleep-phase syndrome	231160	Familial berry aneurysm	1416	Familial CPPD
98880	Familial afibrinogenemia	402075	Familial bicuspid aortic valve	85453	Familial cutaneous amyloidosis
1020	Familial Alzheimer disease	221061	Familial brain cavernous angioma	53296	Familial cutaneous collagenoma
280397	Familial Alzheimer-like prion disease	221061	Familial brain cavernous hemangioma	313846	Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome
319465	Familial AML	227535	Familial breast cancer	211	Familial cylindromatosis
85450	Familial amyloid nephropathy	227535	Familial breast carcinoma	97345	Familial dementia, British type
93560	Familial amyloid nephropathy due to apolipoprotein A-I variant	36382	Familial CAD	97346	Familial dementia, Danish type
238269	Familial amyloid nephropathy due to apolipoprotein A-II variant	2678	Familial café-au-lait spots	313808	Familial dementia, Neumann type
93562	Familial amyloid nephropathy due to fibrinogen A alpha-chain variant	1416	Familial calcium pyrophosphate deposition	1799	Familial developmental dysphasia
93561	Familial amyloid nephropathy due to lysozyme variant	1768	Familial caudal dysgenesis	26106	Familial diffuse cancer of stomach
85447	Familial amyloid polyneuropathy type I	464760	Familial cavitory optic disc anomaly	26106	Familial diffuse gastric cancer
85448	Familial amyloid polyneuropathy type IV	1416	Familial CC	85169	Familial digital arthropathy-brachydactyly
85447	Familial amyloid polyneuropathy, Portuguese-Swedish-Japanese type	169085	Familial CD8 deficiency	300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation
85448	Familial amyloidosis, Finnish type	892	Familial cerebelloretinal angiomas	18	Familial distal primary acidosis
228277	Familial anetoderma	439254	Familial cerebral amyloid angiopathy	85192	Familial doughnut lesions of skull
199279	Familial angilipomatosis	221061	Familial cerebral cavernoma	75376	Familial drusen
91378	Familial angioneurotic edema	221061	Familial cerebral cavernous malformation	1764	Familial dysautonomia
229	Familial aortic dissection	231160	Familial cerebral saccular aneurysm	314381	Familial dysautonomia with contractures
425	Familial apoA-I deficiency	36382	Familial cervical artery dissection	412	Familial dysbetalipoproteinemia
309020	Familial apoC-II deficiency	481662	Familial Chilblain lupus	98881	Familial dysfibrinogenemia
309020	Familial apolipoprotein C-II	1428	Familial chondromalacia patellae	324588	Familial dyskinesia and facial
		444490	Familial chylomicronemia syndrome		
		404560	Familial Clark nevus syndrome		
		293144	Familial clubfoot due to 5q31		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	myokymia		hyperostosis syndrome		epilepsy
404560	Familial dysplastic nevus syndrome	682	Familial hyperPP	352582	Familial infantile myoclonus epilepsy
1885	Familial ectopia lentis	99763	Familial hyperreninemic hypoaldosteronism type 1	225154	Familial infantile striatonigral degeneration
2762	Familial ectopic ossification	99764	Familial hyperreninemic hypoaldosteronism type 2	225154	Familial infantile striatonigral necrosis
85110	Familial encephalopathy with neuroserpin inclusion bodies	424	Familial hyperthyroidism due to mutations in TSH receptor	508410	Familial intestinal malrotation
391384	Familial episodic pain syndrome	427	Familial hypoaldosteronism	2454	Familial intestinal malrotation-facial anomalies syndrome
391392	Familial episodic pain syndrome with predominantly lower limb involvement	425	Familial hypoalphalipoproteinemia	2300	Familial intestinal polyatresia syndrome
391389	Familial episodic pain syndrome with predominantly upper body involvement	405	Familial hypocalciuric hypercalcemia	231160	Familial intracranial saccular aneurysm
90042	Familial erythrocytosis	93372	Familial hypocalciuric hypercalcemia type 1	217656	Familial isolated arrhythmogenic right ventricular cardiomyopathy
225968	Familial essential thrombocythemia	101049	Familial hypocalciuric hypercalcemia type 2	217656	Familial isolated arrhythmogenic right ventricular dysplasia
85195	Familial expansile osteolysis	101050	Familial hypocalciuric hypercalcemia type 3	217656	Familial isolated arrhythmogenic ventricular cardiomyopathy
891	Familial exudative vitreoretinopathy	248408	Familial hypodysfibrinogenemia	293899	Familial isolated arrhythmogenic ventricular cardiomyopathy, biventricular form
98820	Familial focal epilepsy with variable foci	101041	Familial hypofibrinogenemia	293910	Familial isolated arrhythmogenic ventricular cardiomyopathy, classic form
314022	Familial fundic gland polyposis with gastric cancer	440	Familial hypospadias	293888	Familial isolated arrhythmogenic ventricular cardiomyopathy, left dominant form
464756	Familial gastric type 1 neuroendocrine tumor	225154	Familial IBSN	293910	Familial isolated arrhythmogenic ventricular cardiomyopathy, right dominant form
231040	Familial generalized lentiginosis	1677	Familial idiopathic dilatation of the right atrium	217656	Familial isolated arrhythmogenic ventricular dysplasia
99819	Familial gestational hyperthyroidism	656	Familial idiopathic nephrotic syndrome	293899	Familial isolated arrhythmogenic ventricular dysplasia, biventricular form
361	Familial glucocorticoid deficiency	656	Familial idiopathic steroid-resistant nephrotic syndrome	293910	Familial isolated arrhythmogenic ventricular dysplasia, classic form
3000	Familial gonadotropin-independent male-limited sexual precocity	93214	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation	293888	Familial isolated arrhythmogenic ventricular dysplasia, left dominant form
540	Familial hemophagocytic lymphohistiocytosis	93217	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	293910	Familial isolated arrhythmogenic ventricular dysplasia, right dominant form
32960	Familial Hibernian fever	93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis	217656	Familial isolated arrhythmogenic ventricular dysplasia
540	Familial HLH	93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	293899	Familial isolated arrhythmogenic ventricular dysplasia, biventricular form
2604	Familial hollow visceral myopathy	93216	Familial idiopathic steroid-resistant nephrotic syndrome with minimal changes	293910	Familial isolated arrhythmogenic ventricular dysplasia, classic form
403	Familial hyperaldosteronism type I	225154	Familial infantile bilateral striatal necrosis	293888	Familial isolated arrhythmogenic ventricular dysplasia, left dominant form
404	Familial hyperaldosteronism type II	300373	Familial infantile gigantism	293910	Familial isolated arrhythmogenic ventricular dysplasia, right dominant form
251274	Familial hyperaldosteronism type III	448348	Familial infantile gigantism due to a point mutation	217656	Familial isolated ARVC
403	Familial hyperaldosteronism type I	448372	Familial infantile gigantism due to dup(X)q(26)	217656	Familial isolated ARVD
404	Familial hyperaldosteronism type II	448372	Familial infantile gigantism due to Xq26 microduplication	295014	Familial isolated clinodactyly of fingers
251274	Familial hyperaldosteronism type III	300547	Familial infantile hypercalcemia with suppressed intact parathyroid hormone	101351	Familial isolated congenital asplenia
79506	Familial hyperalphalipoproteinemia	352582	Familial infantile myoclonic	154	Familial isolated dilated cardiomyopathy
94086	Familial hypercalcemia-nephrocalcinosis-indicanuria syndrome			99879	Familial isolated hyperparathyroidism
238475	Familial hypercholanemia			2238	Familial isolated
178345	Familial hyperestrogenism				
757	Familial hyperkalemic hypertension				
682	Familial hyperkalemic periodic paralysis				
412	Familial hyperlipoproteinemia type 3				
306661	Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	hypoparathyroidism	495930	Familial monosomy 7 syndrome		dyskinesia
2239	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	99361	Familial MTC	342	Familial paroxysmal polyserositis
189466	Familial isolated hypoparathyroidism due to impaired PTH secretion	276399	Familial multinodular goiter	228140	Familial paroxysmal ventricular fibrillation, non Brugada type
314777	Familial isolated pituitary adenoma	35909	Familial multiple coagulation factor deficiency	98820	Familial partial epilepsy with variable foci
397685	Familial isolated prolactin receptor deficiency	523	Familial multiple cutaneous leiomyomas	79084	Familial partial lipodystrophy type 1
75249	Familial isolated restrictive cardiomyopathy	338	Familial multiple fibrofolliculoma	2348	Familial partial lipodystrophy type 2
411788	Familial isolated trichomegaly	500	Familial multiple lentigines syndrome	79083	Familial partial lipodystrophy type 3
96	Familial isolated vitamin E deficiency	231040	Familial multiple lentigines syndrome without systemic involvement	2348	Familial partial lipodystrophy, Dunnigan type
2295	Familial joint instability syndrome	199276	Familial multiple lipomatosis	79084	Familial partial lipodystrophy, Körberling type
2295	Familial joint laxity	263662	Familial multiple meningioma	466729	Familial patent arterial duct
180176	Familial juvenile gigantomastia	624	Familial multiple nevi flammei	871	Familial PCCD
209886	Familial juvenile gouty nephropathy	624	Familial multiple port-wine stains	93333	Familial pelvis-scapular dysplasia
180176	Familial juvenile hypertrophy of the breast	867	Familial multiple trichoepithelioma	29072	Familial pheochromocytoma-paraganglioma
209886	Familial juvenile hyperuricemic nephropathy type 1	922	Familial nasal acilia	98809	Familial PKD
217330	Familial juvenile hyperuricemic nephropathy type 2	209886	Familial nephropathy with gout	71290	Familial platelet disorder with associated myeloid malignancy
493	Familial keratoacanthoma	424	Familial non-immune hyperthyroidism	71290	Familial platelet syndrome with predisposition to acute myelogenous leukemia
293936	Familial keratoconus with cataract	306658	Familial normophosphatemic tumoral calcinosis	330061	Familial polymorphous light eruption of American Indians
3267	Familial lambdoid synostosis	280403	Familial omphalocele syndrome with facial dysmorphism	733	Familial polyposis coli
79293	Familial LCAT deficiency	154	Familial or idiopathic dilated cardiomyopathy	261584	Familial polyposis coli due to monosomy 5q22.2
523	Familial leiomyomatosis and renal cell cancer	75249	Familial or idiopathic restrictive cardiomyopathy	99810	Familial porencephaly
523	Familial leiomyomatosis cutis et uteri	569	Familial or sporadic hemiplegic migraine	443062	Familial porphyria cutanea tarda
523	Familial leiomyomatosis with renal carcinoma	443236	Familial orthostatic tachycardia due to norepinephrine transporter deficiency	2196	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement
231040	Familial lentigines profusa	435329	Familial ossifying fibroma	31043	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement
871	Familial Lenègre disease	251262	Familial osteochondritis dissecans	34527	Familial primary hypomagnesemia with normocalciuria and normocalcemia
871	Familial Lev disease	2769	Familial osteodysplasia, Anderson type	353220	Familial primary localized cutaneous amyloidosis
871	Familial Lev-Lenègre disease	2801	Familial osteoectasia	65748	Familial primary self-healing squamous epithelioma of the skin, Ferguson-Smith type
309015	Familial lipoprotein lipase deficiency	86820	Familial osteonecrosis of the femoral head	871	Familial progressive cardiac conduction defect
75381	Familial macular edema	79093	Familial osteosclerosis with abnormalities of the nervous system and meninges	871	Familial progressive heart block
3000	Familial male-limited precocious puberty	1333	Familial pancreatic cancer	280628	Familial progressive hyper- and hypopigmentation
401942	Familial median cleft of the upper and lower lips	1333	Familial pancreatic carcinoma	79146	Familial progressive
342	Familial Mediterranean fever	319487	Familial papillary or follicular thyroid carcinoma		
99361	Familial medullary thyroid carcinoma	97290	Familial papillary thyroid carcinoma with renal papillary neoplasia		
35858	Familial megaloblastic anemia	99877	Familial parathyroid adenoma		
618	Familial melanoma	99878	Familial parathyroids hyperplasia		
165805	Familial mesial temporal lobe epilepsy with febrile seizures	97	Familial paroxysmal ataxia		
741	Familial mitral valve prolapse	98809	Familial paroxysmal kinesigenic		
276399	Familial MNG				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	hyperpigmentation		pneumothorax	439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease
488197	Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome	3197	Familial startle disease	439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease
313808	Familial progressive subcortical gliosis	506334	Familial steroid-resistant nephrotic syndrome with adrenal insufficiency	439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease
1767	Familial progressive vestibulocochlear dysfunction	280406	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	439854	Fatal congenital hypertrophic cardiomyopathy due to GSD
1331	Familial prostate cancer	1325	Familial streblodactyly with aminoaciduria	466	Fatal familial insomnia
90044	Familial pseudohyperkalemia	2456	Familial supernumerary nipples	1561	Fatal infantile cardioencephalomyopathy due to cytochrome C oxidase deficiency
→3202	Familial pseudohyperkalemia type 1	370034	Familial syringomyelia	1561	Fatal infantile COX deficiency
2989	Familial pterygium of the conjunctiva	300345	Familial systemic lupus erythematosus	1561	Fatal infantile cytochrome C oxidase deficiency
275777	Familial pulmonary arterial hypertension	91387	Familial TAAD	166073	Fatal infantile encephalopathy with mitochondrial respiratory chain defects
319487	Familial pure nonmedullary thyroid carcinoma	98819	Familial temporal lobe epilepsy	166063	Fatal infantile encephalopathy with olivopontocerebellar hypoplasia
1675	Familial pyrimidinemia	91387	Familial thoracic aortic aneurysm and aortic dissection	→370114	Fatal infantile encephalopathy-pulmonary hypertension syndrome
79147	Familial reactive perforating collagenosis	71493	Familial thrombocythemia	289527	Fatal infantile HCM due to mitochondrial complex I deficiency
46348	Familial rectal pain	71493	Familial thrombocytosis	280553	Fatal infantile hypertonic myofibrillar myopathy
69126	Familial recurrent arthritis	329319	Familial thrombocytosis with transverse limb defect	289527	Fatal infantile hypertrophic cardiomyopathy due to mitochondrial complex I deficiency
2809	Familial recurrent Bell palsy	3324	Familial thrombomodulin anomalies	289527	Fatal infantile hypertrophic cardiomyopathy due to NADH-coenzyme Q reductase deficiency
2809	Familial recurrent peripheral facial palsy	93953	Familial thyroglossal duct cyst	289527	Fatal infantile hypertrophic cardiomyopathy due to NADH-CoQ reductase deficiency
85450	Familial renal amyloidosis	95716	Familial thyroid dyshormonogenesis	17	Fatal infantile lactic acidosis with methylmalonic aciduria
93560	Familial renal amyloidosis due to apolipoprotein A-I variant	53372	Familial trembling of the chin	168566	Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3
238269	Familial renal amyloidosis due to apolipoprotein A-II variant	93583	Familial TTP	168566	Fatal mitochondrial disease due to COXPD3
93561	Familial renal amyloidosis due to lysozyme variant	53715	Familial tumoral calcinosis	391343	Fatal post-viral neurodegenerative disorder
69076	Familial renal glucosuria	289365	Familial vesicoureteral reflux	2492	FATCO syndrome
284247	Familial retinal arterial macroaneurysm	2604	Familial visceral myopathy	816	Fatty acid alcohol oxidoreductase deficiency
231108	Familial rhabdoid tumor	2808	Familial vocal cord dysfunction	329308	Fatty acid hydroxylase-associated neurodegeneration
→168569	Familial Rosaï-Dorfman disease	289365	Familial VUR	2064	Faulk-Epstein-Jones syndrome
168624	Familial scaphocephaly syndrome, McGillivray type	170	Familial woolly hair syndrome	→97229	Fazio-Londe disease
171839	Familial scaphocephaly-radioulnar synostosis syndrome	170	Familial woolly hair syndrome	405	FBH
3135	Familial Scheuermann disease	404560	FAMM-PC syndrome	405	FBHH
3135	Familial Scheuermann juvenile kyphosis	404560	FAMMM syndrome	404451	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome
481986	Familial schizencephaly	84	Fanconi anemia	47045	FCAS
→168569	Familial SHML	84	Fanconi pancytopenia		
51083	Familial short QT syndrome	→2697	Fanconi syndrome-ichthyosis-dysmorphism syndrome		
166282	Familial sick sinus syndrome	2088	Fanconi-Bickel disease		
→168569	Familial sinus histiocytosis with massive lymphadenopathy	733	FAP		
166282	Familial sinus node dysfunction	261584	FAP due to monosomy 5q22.2		
300345	Familial SLE	2792	Fara-Chlupackova syndrome		
3135	Familial spinal osteochondrosis	333	Farber disease		
2903	Familial spontaneous	333	Farber lipogranulomatosis		
		99906	Farmer's lung disease		
		1915	FAS		
		3261	FAS deficiency		
		1915	FASD		
		164736	FASPS		
		166105	FASTKD2-related infantile mitochondrial encephalomyopathy		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
247868	FCAS2	60015	Fenestrae parietales symmetricae	254492	FFA
440437	FCCTX	85110	FENIB	398166	FFDD
98970	FCD	1184	Fenton-Wilkinson-Toselano syndrome	79133	FFDD1
268961	FCD type I	45358	FEOM	398173	FFDD2
268973	FCD type Ia	391384	FEPS	1807	FFDD3
268980	FCD type Ib	65748	Ferguson-Smith disease	398189	FFDD4
268987	FCD type Ic	2180	Ferlini-Ragno-Calzolari syndrome	79133	FFDD type I
268994	FCD type II	157846	Ferritin-related neurodegeneration	398173	FFDD type II
269001	FCD type IIa	397922	Ferro-cerebro-cutaneous syndrome	1807	FFDD type III
269008	FCD type IIb	139491	Ferroportin disease	398189	FFDD type IV
272	FCMD	40366	Fetal acitretin/etretinate syndrome	98820	FFEVF
86814	FCMTE	994	Fetal akinesia deformation sequence	1988	FFS
99654	FCPD	363409	Fetal akinesia-cerebral and retinal hemorrhage syndrome	2019	FFU complex
3071	FCS syndrome	1915	Fetal alcohol spectrum disorders	93932	FG syndrome type 1
47045	FCU	1915	Fetal alcohol syndrome	313855	FGFR2-related bent bone dysplasia
324	FD	1908	Fetal aminopterin syndrome	1305	FGLDS
324588	FDFM	1041	Fetal anasarca	391641	FGLDS1
26106	FDGC	853	Fetal and neonatal alloimmune thrombocytopenia	391646	FGLDS2
412022	FDLAB syndrome	370076	Fetal carbamazepine syndrome	403	FH1
163703	Febrile infection-related epilepsy syndrome	1911	Fetal cocaine syndrome	404	FH2
98974	FECD	294	Fetal cytomegalovirus syndrome	251274	FH3
→182050	Fechtner syndrome	1912	Fetal dihydantoin syndrome	403	FH-I
79292	FED	465824	Fetal encasement syndrome	404	FH-II
247165	Fee disease	97360	Fetal face syndrome	251274	FH-III
98969	Fehr corneal dystrophy	85212	Fetal Gaucher disease	→168569	FHC
1192	Feigenbaum-Bergeron-Richardson syndrome	1912	Fetal hydantoin syndrome	401920	FHCC
1305	Feingold syndrome	1041	Fetal hydrops	405	FHH
391641	Feingold syndrome type 1	1909	Fetal indomethacin syndrome	93372	FHH type 1
391646	Feingold syndrome type 2	1910	Fetal iodine syndrome	101049	FHH type 2
53693	Fellman disease	1055	Fetal left ventricular aneurysm	101050	FHH type 3
47612	Felty syndrome	284362	Fetal lung interstitial tumor	99763	FHHA1
488191	Female infertility due to oocyte meiotic arrest	1917	Fetal methylmercury syndrome	99764	FHHA2
404466	Female infertility due to zona pellucida defect	1918	Fetal minoxidil syndrome	2196	FHHNC with severe ocular involvement
2973	Female pseudohermaphroditism-anorectal anomalies syndrome	295	Fetal parvovirus syndrome	31043	FHHNC without severe ocular involvement
2975	Female pseudohermaphroditism-skeletal anomalies syndrome	290	Fetal rubella syndrome	263479	FHI
101039	Female restricted epilepsy with intellectual disability	3312	Fetal thalidomide syndrome	397618	FHONDA syndrome
1987	Femoral agenesis/hypoplasia	1913	Fetal trimethadione syndrome	1988	FHUFS
399329	Femoral head epiphysiolytic	1906	Fetal valproate syndrome	251601	Fibrillary astrocytoma
1988	Femoral hypoplasia-unusual facies syndrome	1906	Fetal valproic acid syndrome	331	Fibrin-stabilizing factor deficiency
1987	Femoral intercalary meromelia	166068	Fetal-onset olivopontocerebellar hypoplasia	93562	Fibrinogen A alpha-chain amyloidosis
1988	Femoral-facial syndrome	95431	Feto-fetal transfusion syndrome	439881	Fibrinous bronchitis
294977	Femorotibiofibular intercalary transverse meromelia	69063	Fetomaternal alloimmunization with antenatal glomerulopathies	477650	Fibroblastic rheumatism
2019	Femur-fibula-ulna complex	464724	Fever-associated acute infantile liver failure syndrome	99654	Fibrocalculous pancreatic diabetes
2019	Femur-fibula-ulna dysostosis	163703	Fever-induced refractory epileptic encephalopathy in school-aged children	99654	Fibrocalculous pancreatopathy
2019	Femur-fibula-ulna syndrome	891	FEVR	2021	Fibrochondrogenesis

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	carcinoma	293812	Fixed drug eruption		hyperkeratosis
79105	Fibromyxosarcoma	3092	Fixed subaortic stenosis	2200	Focal palmoplantar and gingival keratoderma
84090	Fibronectin glomerulopathy	209886	FJHN type 1	370002	Focal palmoplantar keratoderma with joint keratoses
2030	Fibrosarcoma	217330	FJHN type 2	443804	Focal stiff limb syndrome
63999	Fibrosing mediastinitis	1968	Flat face-microstomia-ear anomaly syndrome	443804	Focal stiff-person syndrome
249	Fibrous dysplasia of bone	79293	FLD	79093	Foix-Alajouanine syndrome
2639	Fibular aplasia-complex brachydactyly syndrome	83315	Flea-borne typhus	2048	Foix-Chavany-Marie syndrome
1118	Fibular aplasia-ectrodactyly syndrome	98970	Fleck corneal dystrophy	79097	Folinic acid-responsive seizures
2492	Fibular aplasia-tibial campomelia-oligosyndactyly syndrome	409	Fleigel disease	113	Follicular atrophoderma and basal cell carcinomas
1757	Fibular dimelia-diplopodia syndrome	284362	FLIT	79459	Follicular atrophoderma-basal cell carcinoma
93323	Fibular hemimelia	2044	Floating-Harbor syndrome	300552	Follicular cholangitis and pancreatitis
2854	Fibular hypoplasia or aplasia-femoral bowing-oligodactyly syndrome	83451	Florid cemento-osseous dysplasia	86902	Follicular dendritic cell sarcoma
93323	Fibular longitudinal meromelia	83451	Florid osseous dysplasia	69745	Follicular dyskeratoma
2256	Fibulo-ulnar hypoplasia-renal anomalies syndrome	2045	FLOTCH syndrome	525	Follicular lichen planus
79306	FIC1 deficiency	199306	FLP	545	Follicular lymphoma
29207	Fiessinger-Leroy disease	99734	Fluctuating myotonia	300552	Follicular pancreatocholangitis
29207	Fiessinger-Leroy-Reiter syndrome	1685	Fluke infection	243	Follicular stimulating hormone-resistant ovaries
2756	Figuera syndrome	2047	Flynn-Aird syndrome	79100	Folliculitis ulerythematosus reticulata
99879	FIHPT	69063	FMAIG	178512	Folliculotropic mycosis fungoides
3255	Filippi syndrome	342	FMF	228371	Foodborne botulism
352712	FILS syndrome	276399	FMNG	3454	Foot contractures-muscle atrophy-oculomotor apraxia syndrome
352582	FIME	3000	FMPP	337	FOP
1272	Fine-Lubinsky syndrome	319487	FNMTC	60015	Foramina parietalia permagna
369979	Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome	137675	Foamy myocardial transformation of infancy	366	Forbes disease
97232	Fingerprint body myopathy	2143	FOAR syndrome	141071	Foregut duplication cyst of the tongue
209335	Finkel disease	308013	Focal acral hyperkeratosis	51208	Formiminoglutamic aciduria
2036	Finlay-Marks syndrome	83451	Focal cemento-osseous dysplasia	51208	Formiminotransferase cyclodeaminase deficiency
839	Finnish congenital nephrosis	2092	Focal dermal hypoplasia	3238	Forney syndrome
609	Finnish tibial muscular dystrophy	352587	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	3238	Forney-Robinson-Pascoe syndrome
399086	Finnish upper limb-onset distal myopathy	352587	Focal epilepsy-intellectual disability-dysarthria-ataxia syndrome	178333	Forsius-Eriksson syndrome
1825	Finucane-Kurtz-Scott syndrome	398166	Focal facial dermal dysplasia	178333	Forsius-Eriksson type ocular albinism
314777	FIPA	79133	Focal facial dermal dysplasia 1, Brauer type	85162	FOSMN syndrome
163703	FIRES	398173	Focal facial dermal dysplasia 2, Brauer-Setleis type	3219	Fountain syndrome
141136	First branchial arch syndrome	1807	Focal facial dermal dysplasia 3, Setleis type	141037	Fourth branchial cleft anomaly
141013	First branchial cleft anomaly	398189	Focal facial dermal dysplasia 4	141037	Fourth branchial cleft cyst
141013	First branchial cleft cyst	79133	Focal facial dermal dysplasia type 1	141037	Fourth branchial cleft fistula
141013	First branchial cleft fistula	79133	Focal facial dermal dysplasia type I	397618	Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome
79292	Fish-eye disease	398173	Focal facial dermal dysplasia type II	2253	Foveal hypoplasia-presenile cataract syndrome
98919	Fisher syndrome	1807	Focal facial dermal dysplasia type III	221126	Fowler syndrome
840	Fistulous vegetative verrucous hydadenoma	398189	Focal facial dermal dysplasia type IV	2795	Fowler-Christmas-Chapple syndrome
2824	Fitzsimmons-McLachlan-Gilbert syndrome	398189	Focal facial preauricular dysplasia		
2820	Fitzsimmons-Walson-Mellor syndrome	221083	Focal myoclonus of face		
		48918	Focal myositis		
		48918	Focal nodular myositis		
		2200	Focal palmoplantar and gingival		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
169095	FOXN1 deficiency	141168	Frontonasal arteriovenous malformation	90062	Fulminant hepatic failure
1799	FOXP2-associated dysphasia	391474	Frontonasal dysplasia type 1	35063	Fulminant viral hepatitis
275777	FPAH	228390	Frontonasal dysplasia type 2	24	Fumarase deficiency
71290	FPD/AML syndrome	306542	Frontonasal dysplasia type 3	24	Fumaryl aciduria
280628	FPHH	228390	Frontonasal dysplasia with alopecia and genital abnormality	882	Fumarylacetoacetate deficiency
353220	FPLCA	228390	Frontonasal dysplasia with alopecia and genital anomaly	882	Fumarylacetoacetate hydrolase deficiency
79084	FPLD1	306542	Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome	622	Functional methionine synthase deficiency
2348	FPLD2	391474	Frontorhinny	308380	Functional methionine synthase deficiency type cblDv1
79083	FPLD3	275872	Frontotemporal dementia with amyotrophic lateral sclerosis	2169	Functional methionine synthase deficiency type cblE
280356	FPLD4	275872	Frontotemporal dementia with motor neuron disease	2170	Functional methionine synthase deficiency type cblG
435651	FPLD5	293848	Frontotemporal dementia, right temporal atrophy variant	91348	Functioning gonadotropic adenoma
435660	FPLD6	2141	Froster-Huch syndrome	91348	Functioning pituitary gonadotropic adenoma
71290	FPS/AML syndrome	2215	Froster-Iskenius-Waterson-Hall syndrome	227796	Fundus albipunctatus
313808	FPSG	2056	Fructokinase deficiency	827	Fundus flavimaculatus
69126	FRA	348	Fructose-1,6-bisphosphatase deficiency	99004	Fundus pulverulentus
908	Fragile X syndrome	2057	Frydman-Cohen-Karmon syndrome	207000	Fungal myositis
93256	Fragile X-associated tremor/ataxia syndrome	2429	Fryns macrocephaly	→60030	Furlong syndrome
284247	FRAM	1104	Fryns microphthalmia syndrome	2579	Furukawa-Takagi-Nakao syndrome
861	Franceschetti-Klein syndrome	2059	Fryns syndrome	591	Furuncular myiasis
2523	Franek-Bocker-Kahlen syndrome	→2995	Fryns-Aftimos syndrome	591	Furunculoid myiasis
137834	Frank-Ter Haar syndrome	2497	Fryns-Hofkens-Fabry syndrome	591	Furunculous myiasis
100026	Franklin disease	2058	Fryns-Smeets-Thiry syndrome	228119	Fusariosis
2108	François dyscephalic syndrome	1305	FS	228119	Fusarium infection
79149	François syndrome	391641	FS1	2287	Fused mandibular incisors
98970	François-Neetens speckled corneal dystrophy	391646	FS2	2498	Fusion of metacarpals 4 and 5
2052	Fraser syndrome	269	FSH dystrophy	35909	FV and FVIII combined deficiency
→2052	Fraser-like syndrome	243	FSH-RO	908	FXS
347	Frasier syndrome	269	FSHD	93256	FXTAS syndrome
908	FraX syndrome	51208	FTCD deficiency	364	G6P deficiency
908	FRAXA syndrome	275872	FTD-ALS	79258	G6P deficiency type 1a
100973	FRAXE intellectual disability	275872	FTD-MND	79259	G6P deficiency type 1b
100974	FRAXF syndrome	247790	FTH1-associated iron overload	79259	G6P translocase deficiency
95	FRDA	247790	FTH1-related iron overload	79259	G6PT deficiency
834	Free sialic acid storage disease	98974	Fuchs endothelial corneal dystrophy	25	GA1
309324	Free sialic acid storage disease, infantile form	263479	Fuchs heterochromic iridocyclitis	2066	GABA transaminase deficiency
2053	Freeman-Sheldon syndrome	349	Fucosidosis	79402	GABEB
1147	Freeman-Sheldon syndrome variant	2854	Fuhrmann syndrome	506358	Gabriele de Vries syndrome
2673	Freire Maia-Pinheiro-Opitz syndrome	2854	Fuhrmann-Rieger-de Sousa syndrome	90041	Gaisböck syndrome
2723	Freire-Maia syndrome	→85199	Fukuda-Miyanomae-Nakata syndrome	487	Galactocerebrosidase deficiency
→264200	Frias syndrome	551	Fukuhara syndrome	79237	Galactokinase deficiency
85335	Fried syndrome	272	Fukuyama congenital muscular dystrophy	79237	Galactokinase deficiency galactosemia
2487	Fried-Goldberg-Mundel syndrome			309297	Galactosamine-6-sulfatase deficiency
1969	Friedman-Goodman syndrome			79238	Galactose epimerase deficiency
95	Friedreich ataxia			79239	Galactose-1-phosphate uridylyltransferase deficiency
96	Friedreich-like ataxia			79239	Galactosemia type 1
99672	Fried's tooth and nail syndrome				
1931	Frontal encephalocele				
254492	Frontal fibrosing alopecia				
1791	Frontofacionasal dysplasia				
1826	Frontometaphyseal dysplasia				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
79237	Galactosemia type 2	913	Gastrinoma	324636	GDS
79238	Galactosemia type 3	2069	Gastrocutaneous syndrome	36387	GEFS+
351	Galactosialidosis	2930	Gastrointestinal polyposis-ectodermal changes syndrome	411777	GEKA
487	Galactosylceramidase deficiency	2930	Gastrointestinal polyposis-skin pigmentation-aloppecia-fingernail changes syndrome	26790	Gelatinous ascites
75496	Galactosyltransferase I deficiency	44890	Gastrointestinal stromal sarcoma	98957	Gelatinous drop-like corneal dystrophy
487	GALC deficiency	44890	Gastrointestinal stromal tumor	2623	Geleophysic dwarfism
79238	GALE deficiency	2368	Gastroschisis	2623	Geleophysic dysplasia
79238	GALE-D	355	Gaucher disease	2073	Gélineau disease
79237	GALK deficiency	77259	Gaucher disease type 1	85448	Gelsolin amyloidosis
79237	GALK-D	77260	Gaucher disease type 2	2074	Gemignani syndrome
100086	Gallbladder neuroendocrine tumor	77261	Gaucher disease type 3	251604	Gemistocytic astrocytoma
2065	Galloway syndrome	2072	Gaucher disease type 3C	2084	GEMSS syndrome
2065	Galloway-Mowat syndrome	77261	Gaucher disease, subacute neuronopathic type	51608	Generalized arterial calcification of infancy
309297	GALNS deficiency	2072	Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome	79402	Generalized atrophic benign epidermolysis bullosa
79239	GALT deficiency	2072	Gaucher-like disease	508529	Generalized basal epidermolysis bullosa simplex with skin atrophy, scarring and hair loss
2325	Gamborg-Nielsen syndrome	308712	GBE deficiency, adult neuromuscular form	168632	Generalized basaloid follicular hamartoma syndrome
3035	Game-Friedman-Paradice syndrome	308684	GBE deficiency, childhood combined hepatic and myopathic form	98806	Generalized cervical and upper-limb-onset dystonia
2066	Gamma-aminobutyric acid transaminase deficiency	308698	GBE deficiency, childhood neuromuscular form	528	Generalized congenital lipodystrophy
212	Gamma-cystathionase deficiency	308670	GBE deficiency, congenital neuromuscular form	228429	Generalized congenital lipodystrophy type 4
33573	Gamma-glutamyl transpeptidase deficiency	308655	GBE deficiency, fatal perinatal neuromuscular form	228429	Generalized congenital lipodystrophy with myopathy
33574	Gamma-glutamylcysteine synthetase deficiency	308638	GBE deficiency, non progressive hepatic form	263543	Generalized deciduous skin
100026	Gamma-HCD	308621	GBE deficiency, progressive hepatic form	263548	Generalized deciduous skin type A
100026	Gamma-heavy chain disease	360	GBM	263553	Generalized deciduous skin type B
22	Gamma-hydroxybutyric aciduria	98916	GBS, acute inflammatory demyelinating polyradiculoneuropathic form	263558	Generalized deciduous skin type C
353	Gamma-sarcoglycanopathy	329984	GCC	231568	Generalized dominant dystrophic epidermolysis bullosa
682	Gamstorp disease	98962	GCD1	79399	Generalized EBS, non-Dowling-Meara type
682	Gamstorp episodic adynamy	98963	GCD2	99647	Generalized enchondromatosis with platyspondyly
382	GAMT deficiency	25	GCDHD	79399	Generalized epidermolysis bullosa simplex, non-Dowling-Meara type
643	GAN	98962	GCDI	36387	Generalized epilepsy with febrile seizures-plus
251937	Gangliocytoma	98963	GCDII	79137	Generalized epilepsy-paroxysmal dyskinesia syndrome
251949	Ganglioglioma	438274	GCGR-related hyperglucagonemia	308487	Generalized epimerase deficiency galactosemia
251877	Ganglioneuroblastoma	528	GCL	157991	Generalized eruptive histiocytoma
251992	Ganglioneuroma	228429	GCL4	157991	Generalized eruptive histiocytosis
2067	GAPO syndrome	2095	GCM syndrome	411777	Generalized eruptive keratoacanthoma
314022	GAPPS	380	GCPS	411777	Generalized eruptive keratoacanthomas of Grzybowski
3469	Garcia-Lurie syndrome	363976	GCT of bone	280774	Generalized essential telangiectasia
79665	Gardner syndrome	98957	GDCD		
324636	Gardner-Diamond syndrome	53697	GDD		
2075	Gardner-Silengo-Wachtel syndrome	366	GDE deficiency		
99000	Gass disease				
314022	Gastric adenocarcinoma and proximal polyposis of the stomach				
423781	Gastric carcinoma, salivary gland type				
141071	Gastric duplication cyst of the tongue				
332	Gastric intrinsic factor deficiency				
36273	Gastric linitis plastica				
100075	Gastric NET				
100075	Gastric neuroendocrine tumor				
418959	Gastric squamous cell carcinoma				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
36236	Generalized exfoliative disease	35686	Geographic helicoid peripapillary choroidopathy	849	Glanzmann thrombasthenia
1041	Generalized fetal edema	79137	GEPD	666	Glass bone disease
308487	Generalized galactose epimerase deficiency	2808	Gerhardt syndrome	→53271	Glass-Chapman-Hockley syndrome
308487	Generalized GALE deficiency	213837	Germ cell cancer of the cervix uteri	213833	Glassy cell carcinoma of the cervix uteri
308487	Generalized GALE-D	213751	Germ cell cancer of the corpus uteri		
33355	Generalized hematopoietic hypoplasia	2077	German syndrome	238763	Glaucoma secondary to spherophakia/ectopia lentis and megalocornea
79402	Generalized junctional epidermolysis bullosa, non-Herlitz type	91352	Germinoma of the central nervous system	2084	Glaucoma-ectopia-microspherophakia-stiff joints-short stature syndrome
329971	Generalized juvenile polyposis/juvenile polyposis coli	2078	Geroderma osteodysplastica	2085	Glaucoma-sleep apnea syndrome
167635	Generalized lichenoid papular eruption	496693	Gershoni-Baruch syndrome	354	GLB1 deficiency
435628	Generalized lipodystrophy-progeroid features-severe intellectual disability syndrome	1117	Gershoni-Baruch-Leibo syndrome	360	Glioblastoma
141209	Generalized lymphatic anomaly	221117	Gerstmann syndrome	360	Glioblastoma multiforme
89842	Generalized mitis RDEB	356	Gerstmann-Straussler-Scheinker syndrome	269197	Glioependymal/ependymal cyst
167635	Generalized papular and sclerodermoid lichen myxedematosus	99926	Gestational choriocarcinoma	251582	Gliomatosis cerebri
263543	Generalized peeling skin syndrome	63275	Gestational pemphigoid	251576	Gliosarcoma
263548	Generalized peeling skin syndrome type A	280774	GET	404476	Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome
263553	Generalized peeling skin syndrome type B	84090	GFND		Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome
263558	Generalized peeling skin syndrome type C	314769	GH and PRL cosecreting pituitary adenoma	488613	Global developmental delay-osteopenia-ectodermal defect syndrome
171876	Generalized pseudohypoaldosteronism type 1	633	GH receptor deficiency		Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome
263543	Generalized PSS	1802	Ghosal hematodiaphyseal dysplasia	73223	Globodontia
247353	Generalized pustular psoriasis	1802	Ghosal syndrome	480898	Globoid cell leukodystrophy
3221	Generalized resistance to thyroid hormone	83450	Ghost teeth		Globangiomytosis
308487	Generalized UDP-galactose-4-epimerase deficiency	314811	Ghrelin receptor deficiency	→69735	Glomerulonephritis-sparse hair-telangiectasis syndrome
308487	Generalized uridine diphosphate galactose-4-epimerase deficiency	180267	Giant adenofibroma of the breast	84090	Glomerulopathy with fibronectin deposits
254704	Genetic hyperferritinemia without iron overload	643	Giant axonal neuropathy	391651	Glomus tumor
99845	Genetic recurrent myoglobinuria	397	Giant cell arteritis	83454	Glomuvenous malformation
226316	Genetic transient congenital hypothyroidism	1190	Giant cell chondrodyplasia	2616	Gloomy face syndrome
2075	Genitopalatocardiac syndrome	251579	Giant cell glioblastoma	141163	Glossopalatine ankylosis
85201	Genitopatellar syndrome	139436	Giant cell histiocytomatosis	221098	Glossopharyngeal neuralgia
2163	Genoa syndrome	363976	Giant cell tumor of bone	404476	GLOW syndrome
85197	Genochondromatosis type 1	626	Giant congenital melanocytic nevus	255132	GLRX5-related sideroblastic anemia
93398	Genochondromatosis type 2	2494	Giant hypertrophic gastritis	97280	Glucagonoma
329813	Genome-wide paternal uniparental disomy mosaicism	626	Giant pigmented hairy nevus	97280	Glucagonoma syndrome
1454	Gentile syndrome	274	Giant platelet syndrome	355	Glucocerebrosidase deficiency
217008	Genuine diffuse phlebectasia	1065	Gillespie syndrome	786	Glucocorticoid resistance
98961	Geographic corneal dystrophy	2025	Gingival fibromatosis-facial dysmorphism syndrome	403	Glucocorticoid-remediable aldosteronism
		3473	Gingival fibromatosis-hepatosplenomegaly-other anomalies syndrome	403	Glucocorticoid-sensitive hypertension
		2026	Gingival fibromatosis-hypertrichosis syndrome	79272	Glucosamine N-acetyl-6-sulfatase
		2027	Gingival fibromatosis-progressive deafness syndrome		
		2709	Gingival hypertrophy-corneal dystrophy		
		44890	GIST		
		97286	GIST-paraganglioma dyad		
		358	Gitelman syndrome		
		3268	Giuffré-Tsukahara syndrome		
		141209	GLA		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	deficiency	57	Glycogen storage disease due to aldolase A deficiency		lactate dehydrogenase H-subunit deficiency
71277	Glucose transporter type 1 deficiency	364	Glycogen storage disease due to G6P deficiency	284426	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency
35710	Glucose-galactose malabsorption	79258	Glycogen storage disease due to G6P deficiency type 1a	34587	Glycogen storage disease due to LAMP-2 deficiency
79330	Glucosidase 1 deficiency	79259	Glycogen storage disease due to G6P deficiency type 1b	79240	Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency
79320	Glucosyltransferase 1 deficiency	364	Glycogen storage disease due to glucose-6-phosphatase deficiency	369	Glycogen storage disease due to liver glycogen phosphorylase deficiency
79325	Glucosyltransferase 2 deficiency	79258	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a	2089	Glycogen storage disease due to liver glycogen synthase deficiency
71277	Glut1-DS	79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1b	264580	Glycogen storage disease due to liver phosphorylase kinase deficiency
71277	Glut-1 deficiency syndrome	2088	Glycogen storage disease due to GLUT2 deficiency	137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency
51208	Glutamate formiminotransferase deficiency	367	Glycogen storage disease due to glycogen branching enzyme deficiency	99849	Glycogen storage disease due to muscle beta-enolase deficiency
2195	Glutamate-aspartate transport defect	308712	Glycogen storage disease due to glycogen branching enzyme deficiency, adult neuromuscular form	368	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
33574	Glutamate-cysteine ligase deficiency	308684	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	371	Glycogen storage disease due to muscle phosphofructokinase deficiency
25	Glutaric acidemia type 1	308698	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood neuromuscular form	715	Glycogen storage disease due to muscle phosphorylase kinase deficiency
26791	Glutaric acidemia type 2	308670	Glycogen storage disease due to glycogen branching enzyme deficiency, congenital neuromuscular form	→319646	Glycogen storage disease due to phosphoglucomutase deficiency
35706	Glutaric acidemia type 3	308655	Glycogen storage disease due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form	713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency
25	Glutaric aciduria type 1	308638	Glycogen storage disease due to glycogen branching enzyme deficiency, non progressive hepatic form	97234	Glycogen storage disease due to phosphoglycerate mutase deficiency
26791	Glutaric aciduria type 2	308621	Glycogen storage disease due to glycogen branching enzyme deficiency, progressive hepatic form	2089	Glycogen storage disease type 0a
35706	Glutaric aciduria type 3	366	Glycogen storage disease due to glycogen debranching enzyme deficiency	137625	Glycogen storage disease type 0b
25	Glutaryl-CoA dehydrogenase deficiency	2089	Glycogen storage disease due to hepatic glycogen synthase deficiency	364	Glycogen storage disease type 1
35706	Glutaryl-CoA oxidase deficiency	2364	Glycogen storage disease due to lactate dehydrogenase deficiency	79258	Glycogen storage disease type 1a
25	Glutaryl-coenzyme A dehydrogenase deficiency	284435	Glycogen storage disease due to	79259	Glycogen storage disease type 1b
32	Glutathione synthetase deficiency			→79259	Glycogen storage disease type 1C
289846	Glutathione synthetase deficiency with 5-oxoprolinuria			→79259	Glycogen storage disease type 1D
289849	Glutathione synthetase deficiency without 5-oxoprolinuria			365	Glycogen storage disease type 2
33573	Glutathionuria			308552	Glycogen storage disease type 2, infantile onset
284414	Glycerol kinase deficiency, adult form			420429	Glycogen storage disease type 2, late-onset
284408	Glycerol kinase deficiency, infantile form			366	Glycogen storage disease type 3
284411	Glycerol kinase deficiency, juvenile form			367	Glycogen storage disease type 4
261476	Glycerol kinase deficiency-contiguous gene syndrome			308712	Glycogen storage disease type 4, adult neuromuscular form
255182	Glycine cleavage system L protein deficiency			308684	Glycogen storage disease type 4,
407	Glycine encephalopathy				
289891	Glycine N-methyltransferase deficiency				
365	Glycogen storage disease due to acid maltase deficiency				
308552	Glycogen storage disease due to acid maltase deficiency, infantile onset				
420429	Glycogen storage disease due to acid maltase deficiency, late-onset				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	childhood combined hepatic and myopathic form	369	Glycogen storage disease type VI	284426	Glycogenesis due to lactate dehydrogenase M-subunit deficiency
308698	Glycogen storage disease type 4, childhood neuromuscular form	371	Glycogen storage disease type VII	34587	Glycogenesis due to LAMP-2 deficiency
308670	Glycogen storage disease type 4, congenital neuromuscular form	2088	Glycogen storage disease type XI	79240	Glycogenesis due to liver and muscle phosphorylase kinase deficiency
308655	Glycogen storage disease type 4, fatal perinatal neuromuscular form	57	Glycogen storage disease type XII	369	Glycogenesis due to liver glycogen phosphorylase deficiency
308638	Glycogen storage disease type 4, non progressive hepatic form	→319646	Glycogen storage disease type XIV	264580	Glycogenesis due to liver phosphorylase kinase deficiency
308621	Glycogen storage disease type 4, progressive hepatic form	263297	Glycogen storage disease type XV	137625	Glycogenesis due to muscle and heart glycogen synthase deficiency
368	Glycogen storage disease type 5	263297	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	99849	Glycogenesis due to muscle beta-enolase deficiency
369	Glycogen storage disease type 6	365	Glycogenesis due to acid maltase deficiency	368	Glycogenesis due to muscle glycogen phosphorylase deficiency
371	Glycogen storage disease type 7	308552	Glycogenesis due to acid maltase deficiency, infantile onset	371	Glycogenesis due to muscle phosphofructokinase deficiency
264580	Glycogen storage disease type 9A	57	Glycogenesis due to aldolase A deficiency	715	Glycogenesis due to muscle phosphorylase kinase deficiency
79240	Glycogen storage disease type 9B	79258	Glycogenesis due to glucose-6-phosphatase deficiency type 1a	→319646	Glycogenesis due to phosphoglucomutase deficiency
264580	Glycogen storage disease type 9C	79259	Glycogenesis due to glucose-6-phosphatase deficiency type 1b	713	Glycogenesis due to phosphoglycerate kinase 1 deficiency
715	Glycogen storage disease type 9D	79258	Glycogenesis due to glucose-6-phosphatase deficiency type 1a	97234	Glycogenesis due to phosphoglycerate mutase deficiency
715	Glycogen storage disease type 9E	79259	Glycogenesis due to glucose-6-phosphatase transport defect type Ib	2089	Glycogenesis type 0a
2088	Glycogen storage disease type 11	2088	Glycogenesis due to GLUT2 deficiency	137625	Glycogenesis type 0b
284426	Glycogen storage disease type 11	367	Glycogenesis due to glycogen branching enzyme deficiency	364	Glycogenesis type 1
57	Glycogen storage disease type 12	308712	Glycogenesis due to glycogen branching enzyme deficiency, adult neuromuscular form	79259	Glycogenesis type 1b
→319646	Glycogen storage disease type 14	308684	Glycogenesis due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	365	Glycogenesis type 2
263297	Glycogen storage disease type 15	308698	Glycogenesis due to glycogen branching enzyme deficiency, childhood neuromuscular form	308552	Glycogenesis type 2, infantile onset
364	Glycogen storage disease type I	308670	Glycogenesis due to glycogen branching enzyme deficiency, congenital neuromuscular form	420429	Glycogenesis type 2, late-onset
79259	Glycogen storage disease type Ib	308655	Glycogenesis due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form	366	Glycogenesis type 3
365	Glycogen storage disease type II	308638	Glycogenesis due to glycogen branching enzyme deficiency, non progressive hepatic form	367	Glycogenesis type 4
308552	Glycogen storage disease type II, infantile onset	308621	Glycogenesis due to glycogen branching enzyme deficiency, progressive hepatic form	308712	Glycogenesis type 4, adult neuromuscular form
420429	Glycogen storage disease type II, late-onset	366	Glycogenesis due to glycogen debranching enzyme deficiency	308684	Glycogenesis type 4, childhood combined hepatic and myopathic form
366	Glycogen storage disease type III	2364	Glycogenesis due to lactate dehydrogenase deficiency	308698	Glycogenesis type 4, childhood neuromuscular form
367	Glycogen storage disease type IV	284435	Glycogenesis due to lactate dehydrogenase H-subunit deficiency	308670	Glycogenesis type 4, congenital neuromuscular form
308712	Glycogen storage disease type IV, adult neuromuscular form			308655	Glycogenesis type 4, fatal perinatal neuromuscular form
308684	Glycogen storage disease type IV, childhood combined hepatic and myopathic form			308638	Glycogenesis type 4, non progressive hepatic form
308698	Glycogen storage disease type IV, childhood neuromuscular form			308621	Glycogenesis type 4, progressive hepatic form
308670	Glycogen storage disease type IV, congenital neuromuscular form			368	Glycogenesis type 5
308655	Glycogen storage disease type IV, fatal perinatal neuromuscular form			369	Glycogenesis type 6
308638	Glycogen storage disease type IV, non progressive hepatic form			371	Glycogenesis type 7
308621	Glycogen storage disease type IV, progressive hepatic form				
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				
368	Glycogen storage disease type V				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
264580	Glycogenosis type 9A	309192	GM2 gangliosidosis, B variant, adult form	376	Gordon syndrome
79240	Glycogenosis type 9B	309178	GM2 gangliosidosis, B variant, infantile form	1173	Gordon-Holmes syndrome
264580	Glycogenosis type 9C	309185	GM2 gangliosidosis, B variant, juvenile form	73	Gorham disease
715	Glycogenosis type 9D	845	GM2 gangliosidosis, B, B1 variant	73	Gorham syndrome
715	Glycogenosis type 9E	309239	GM2 gangliosidosis, B1 variant	73	Gorham-Stout disease
284426	Glycogenosis type 11	101006	GM2 synthase deficiency	377	Gorlin syndrome
57	Glycogenosis type 12	626	GMN	2095	Gorlin-Chaudhry-Moss syndrome
99849	Glycogenosis type 13	2090	GMS syndrome	377	Gorlin-Goltz syndrome
→319646	Glycogenosis type 14	53697	Gnathodiaphyseal dysplasia	66629	GOSHS
263297	Glycogenosis type 15	602	GNE myopathy	280620	GOSR2-related progressive myoclonus ataxia
364	Glycogenosis type I	100075	GNET	2500	Gottron syndrome
79258	Glycogenosis type Ia	79272	GNS deficiency	59135	Gowers disease
79259	Glycogenosis type Ib	329984	Goblet cell adenocarcinoid	900	GPA
365	Glycogenosis type II	329984	Goblet cell carcinoid	280586	gPAPP deficiency
308552	Glycogenosis type II, infantile onset	329984	Goblet cell carcinoma	247353	GPP
420429	Glycogenosis type II, late-onset	329984	Goblet cell tumor	721	GPS
366	Glycogenosis type III	705	Goiter-deafness syndrome	313808	GPSC
367	Glycogenosis type IV	373	Golabi-Rosen syndrome	403	GRA
308712	Glycogenosis type IV, adult neuromuscular form	351	Goldberg syndrome	2763	Gracile bone dysplasia
308684	Glycogenosis type IV, childhood combined hepatic and myopathic form	66629	Goldberg-Shprintzen megacolon syndrome	53693	GRACILE syndrome
308698	Glycogenosis type IV, childhood neuromuscular form	166272	Goldblatt chondrodysplasia	39812	Graft versus host disease
308670	Glycogenosis type IV, congenital neuromuscular form	166272	Goldblatt syndrome	505	Graham Little syndrome
308655	Glycogenosis type IV, fatal perinatal neuromuscular form	3026	Goldblatt-Viljoen syndrome	505	Graham Little-Piccardi-Lassueur syndrome
308638	Glycogenosis type IV, non progressive hepatic form	2261	Goldblatt-Wallis syndrome	2111	Graham-Boyle-Troxell syndrome
308621	Glycogenosis type IV, progressive hepatic form	374	Goldenhar syndrome	52055	Graham-Cox syndrome
264580	Glycogenosis type IXa	53540	Goldmann-Favre syndrome	→247691	Grand-Kaine-Fulling syndrome
79240	Glycogenosis type IXb	3032	Goldston syndrome	79094	Grange occlusive arterial syndrome
264580	Glycogenosis type IXc	1791	Gollop syndrome	79094	Grange syndrome
715	Glycogenosis type IXd	1986	Gollop-Wolfgang complex	2097	Grant syndrome
715	Glycogenosis type IXe	2092	Goltz syndrome	98962	Granular corneal dystrophy type 1
368	Glycogenosis type V	2092	Goltz-Gorlin syndrome	98963	Granular corneal dystrophy type 2
369	Glycogenosis type VI	1532	Gómez-López-Hernández syndrome	98961	Granular corneal dystrophy type 3
371	Glycogenosis type VII	206484	Gonadoblastoma	98962	Granular corneal dystrophy type I
57	Glycogenosis type XII	91348	Gonadotroph adenoma	98963	Granular corneal dystrophy type II
→319646	Glycogenosis type XIV	432	Gonadotropic deficiency	98961	Granular corneal dystrophy type III
263297	Glycogenosis type XV	759	Gonadotropin-dependant precocious puberty	98963	Granular-lattice corneal dystrophy
263297	Glycogenosis with severe cardiomyopathy due to glycogenin deficiency	562	Gonadotropin-independent female-limited sexual precocity	86850	Granulocytic sarcoma
93598	Glycolic aciduria	2090	Goniodygenesis-intellectual disability-short stature syndrome	900	Granulomatosis with polyangiitis
354	GM1 gangliosidosis	1482	Gonococcal conjunctivitis	183	Granulomatous allergic angiitis
79255	GM1 gangliosidosis type 1	3034	Gonzales-del Angel syndrome	64722	Granulomatous mastitis
79256	GM1 gangliosidosis type 2	169105	Good syndrome	33111	Granulomatous slack skin
79257	GM1 gangliosidosis type 3	1321	Goodman camptodactyly	99915	Granulosa cell cancer
796	GM2 gangliosidosis 0 variant	65798	Goodman syndrome	99915	Granulosa cell malignant tumor
309246	GM2 gangliosidosis, AB variant	375	Goodpasture syndrome	35858	Gräsbeck-Imerslund disease
		75389	Goossens-Deviendt syndrome	69665	Gravidic intrahepatic cholestasis
		757	Gordon hyperkalemia-hypertension syndrome	721	Gray platelet syndrome
				293375	Grayson-Wilbrandt corneal dystrophy
				276405	Green jaundice
				99826	Green monkey disease
				1426	Greenberg dysplasia
				380	Greig cephalopolysyndactyly

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	syndrome		late-onset		phosphofructokinase deficiency
495	Greither disease	57	GSD due to aldolase A deficiency	715	GSD due to muscle phosphorylase kinase deficiency
97261	GRF tumor	364	GSD due to G6P deficiency	→319646	GSD due to phosphoglucomutase deficiency
97261	GRFoma	79258	GSD due to G6P deficiency type 1a	713	GSD due to phosphoglycerate kinase 1 deficiency
139474	Grisart-Destrée syndrome	79258	GSD due to G6P deficiency type 1a	97234	GSD due to phosphoglycerate mutase deficiency
381	Griscelli disease	79259	GSD due to G6P deficiency type 1b	2089	GSD type 0a
79476	Griscelli disease type 1	79259	GSD due to G6PT deficiency	137625	GSD type 0b
79477	Griscelli disease type 2	2088	GSD due to GLUT2 deficiency	364	GSD type 1
79478	Griscelli disease type 3	367	GSD due to glycogen branching enzyme deficiency	79259	GSD type 1 non a
381	Griscelli-Pruniéras syndrome	308712	GSD due to glycogen branching enzyme deficiency, adult neuromuscular form	79258	GSD type 1a
79476	Griscelli-Pruniéras syndrome type 1	308684	GSD due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	79259	GSD type 1b
79477	Griscelli-Pruniéras syndrome type 2	308698	GSD due to glycogen branching enzyme deficiency, childhood neuromuscular form	365	GSD type 2
79478	Griscelli-Pruniéras syndrome type 3	308670	GSD due to glycogen branching enzyme deficiency, congenital neuromuscular form	308552	GSD type 2, infantile onset
3217	Groll-Hirschowitz syndrome	308655	GSD due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form	420429	GSD type 2, late-onset
758	Gronblad-Strandberg-Touraine syndrome	308638	GSD due to glycogen branching enzyme deficiency, non progressive hepatic form	366	GSD type 3
314613	Growing teratoma syndrome	308621	GSD due to glycogen branching enzyme deficiency, progressive hepatic form	367	GSD type 4
391348	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	366	GSD due to glycogen debranching enzyme deficiency	308712	GSD type 4, adult neuromuscular form
→264200	Growth deficiency-brachydactyly-dysmorphism syndrome	2089	GSD due to hepatic glycogen synthase deficiency	308684	GSD type 4, childhood combined hepatic and myopathic form
73273	Growth delay due to insulin-like growth factor I resistance	2364	GSD due to lactate dehydrogenase deficiency	308698	GSD type 4, childhood neuromuscular form
73272	Growth delay due to insulin-like growth factor type 1 deficiency	284435	GSD due to lactate dehydrogenase H-subunit deficiency	308670	GSD type 4, congenital neuromuscular form
2067	Growth delay-alopelia-pseudoanodontia-optic atrophy syndrome	284426	GSD due to lactate dehydrogenase M-subunit deficiency	308655	GSD type 4, fatal perinatal neuromuscular form
53693	Growth delay-aminoaciduria-cholestasis-iron overload-lactic acidosis-early death syndrome	34587	GSD due to LAMP-2 deficiency	308638	GSD type 4, non progressive hepatic form
73272	Growth delay-deafness- intellectual disability syndrome	79240	GSD due to liver and muscle phosphorylase kinase deficiency	308621	GSD type 4, progressive hepatic form
3035	Growth delay-hydrocephaly-lung hypoplasia syndrome	369	GSD due to liver glycogen phosphorylase deficiency	368	GSD type 5
314769	Growth hormone and prolactin cosecreting pituitary adenoma	264580	GSD due to liver phosphorylase kinase deficiency	369	GSD type 6
633	Growth hormone receptor deficiency	137625	GSD due to muscle and heart glycogen synthase deficiency	371	GSD type 7
97261	Growth hormone releasing factor tumor	99849	GSD due to muscle beta-enolase deficiency	264580	GSD type 9A
53693	Growth restriction-aminoaciduria-cholestasis-iron overload-lactic acidosis-early death syndrome	368	GSD due to muscle glycogen phosphorylase deficiency	79240	GSD type 9B
391366	Growth retardation-mild developmental delay-chronic hepatitis syndrome	371	GSD due to muscle phosphorylase deficiency	264580	GSD type 9C
2101	Grubben-de Cock-Borghgraef syndrome			715	GSD type 9D
411777	Grzybowski syndrome			715	GSD type 9E
365	GSD due to acid maltase deficiency			97234	GSD type 10
308552	GSD due to acid maltase deficiency, infantile onset			2088	GSD type 11
420429	GSD due to acid maltase deficiency,			284426	GSD type 11

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
264580	GSD type IXa	2957	Guttmacher syndrome	73229	HANAC syndrome
79240	GSD type IXb	39812	GVH	1927	Hand and foot deformity-flat facies syndrome
264580	GSD type IXc	293375	GWCD	2438	Hand-foot-genital syndrome
715	GSD type IXd	99914	Gynandroblastoma	2438	Hand-foot-uterus syndrome
715	GSD type IXe	414	Gyrate atrophy of choroid and retina	99873	Hand-Schüller-Christian disease
368	GSD type V	168569	H syndrome	989	Hanhart syndrome
369	GSD type VI	139441	H-ABC	186	Hanot syndrome
371	GSD type VII	454750	H-type tracheoesophageal fistula	340	Hantavirosis
2088	GSD type XI	2396	Haberland syndrome	340	Hantavirus fever
57	GSD type XII	99803	Haddad syndrome	319247	Hantavirus pulmonary syndrome
263297	GSD type XV	217026	Hadziselimovic syndrome	3294	Hapnes-Boman-Skeie syndrome
263297	GSD with severe cardiomyopathy due to glycogenin deficiency	91378	HAE	2967	Haptocorrin deficiency
79258	GSDIa	100050	HAE 1	1490	Harboyan syndrome
79259	GSDIb	100051	HAE 2	2812	Hard skin syndrome, Parana type
366	GSDIII	100054	HAE 3	899	HARD syndrome
308712	GSDIV, adult neuromuscular form	100050	HAE-I	85182	Hardcastle syndrome
308684	GSDIV, childhood combined hepatic and myopathic form	100051	HAE-II	1415	Hardikar syndrome
308698	GSDIV, childhood neuromuscular form	100054	HAE-III	1177	Harding ataxia
308670	GSDIV, congenital neuromuscular form	966	HAFF	496790	Harel-Yoon syndrome
308655	GSDIV, fatal perinatal neuromuscular form	79263	Hagberg-Santavuori disease	457	Harlequin ichthyosis
308638	GSDIV, non progressive hepatic form	2841	Hailey-Hailey disease	199282	Harlequin syndrome
308621	GSDIV, progressive hepatic form	2342	Haim-Munk syndrome	→216866	HARP syndrome
99849	GSDXIII	1408	Hair defect-photosensitivity-intellectual disability syndrome	2115	Harrod syndrome
→319646	GSDXIV	69084	Hair-nail ectodermal dysplasia	2116	Hartnup disease
2102	GTP cyclohydrolase I deficiency	300878	Hairy cell leukemia variant	2116	Hartnup disorder
98808	GTPCH1-deficient dopa-responsive dystonia	2220	Hairy elbows syndrome	2117	Hartsfield syndrome
98808	GTPCH1-deficient DRD	3387	Hairy throat syndrome	84085	HAS
2102	GTPCH deficiency	955	Hajdu-Cheney syndrome	83601	Hashimoto encephalitis
90020	Guam disease	2157	HAL deficiency	99872	Hashimoto-Pritzker syndrome
319234	Guanarito hemorrhagic fever	2985	Hal-Berg-Rudolph syndrome	2994	Haspeslagh-Fryns-Muelenaere syndrome
382	Guanidinoacetate methyltransferase deficiency	2521	Halal syndrome	3325	HAT
2785	Guibaud-Vainsel syndrome	1809	Halal-Setton-Wang syndrome	2118	Hawkinsinuria
98916	Guillain-Barré syndrome, acute inflammatory demyelinating polyradiculoneuropathic form	185	Halasz syndrome	1071	Hay-Wells syndrome
231	Guinea worm disease	138	Hall-Hittner syndrome	163596	Hb Bart's hydrops fetalis
1661	Guízar Vázquez-Luengas-Muñoz syndrome	2107	Hall-Riggs syndrome	231242	HbC-beta-thalassemia syndrome
2104	Guízar Vázquez-Sánchez-Manzano syndrome	2108	Hallermann-Streiff syndrome	231249	HbE-beta-thalassemia syndrome
1562	Gunal-Seber-Basaran syndrome	2109	Hallermann-Streiff-François syndrome, severe form	93616	HbH disease
79277	Günther disease	2109	Hallermann-Streiff-like syndrome	352657	HBID
1858	Gurrieri-Sammitto-Bellussi syndrome	157850	Hallervorden-Spatz syndrome	330032	HbLepore-beta-thalassemia syndrome
324561	Guttate hypopigmentation and punctate palmoplantar keratoderma	2110	Hallux varus-preaxial polysyndactyly syndrome	251359	HbS-beta-thalassemia syndrome
		3453	HAM syndrome	251365	HbSC disease
		289326	HAM/TSP	251370	HbSD disease
		314555	Hamamy syndrome	251375	HbSE disease
		2926	Hamanishi-Ueba-Tsuiji syndrome	363412	HBSL
		1217	Hamano-Tsukamoto syndrome	88673	HCC
		2869	Hamartomatous intestinal polyposis	86864	HCD
		93946	Hamel cerebro-palato-cardiac syndrome	93556	HCDD
		79126	Hamman-Rich syndrome	85458	HCHWA
				324723	HCHWA, Arctic type
				100006	HCHWA, Dutch type
				324718	HCHWA, Flemish type

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
100008	HCHWA, Icelandic type	244242	HELLP syndrome	714	Hemolytic anemia due to diphosphoglycerate mutase deficiency
324708	HCHWA, Iowa type	1426	HEM dysplasia	99138	Hemolytic anemia due to erythrocyte adenosine deaminase overproduction
324713	HCHWA, Italian type	252054	Hemangioblastoma	712	Hemolytic anemia due to glucophosphate isomerase deficiency
324703	HCHWA, Piedmont type	2330	Hemangioma-thrombocytopenia syndrome	90030	Hemolytic anemia due to glutathione reductase deficiency
100006	HCHWA-D	90053	Hematopoietic stem cell transplantation	248305	Hemolytic anemia due to glyceraldehyde-3-phosphate dehydrogenase deficiency
58017	HCL-C	2128	Hemi 3 syndrome	35120	Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency
300878	HCL-v	86908	Hemiconvulsion-hemiplegia-epilepsy syndrome	766	Hemolytic anemia due to red cell pyruvate kinase deficiency
163690	HCS	2128	Hemicorporal hypertrophy	275944	Hemolytic disease of the newborn with Kell alloimmunization
306741	HD-HA syndrome	443070	Hemicrania continua	90038	Hemolytic-uremic syndrome with diarrhea
26106	HDGC	306741	Hemidystonia-hemiatrophy syndrome	2134	Hemolytic-uremic syndrome without diarrhea
157941	HDL1	1214	Hemifacial atrophy	93581	Hemolytic-uremic syndrome without diarrhea with anti-factor H antibodies
98934	HDL2	141145	Hemifacial hyperplasia	93578	Hemolytic-uremic syndrome without diarrhea with B factor anomaly
157946	HDL3	1241	Hemifacial hyperplasia-strabismus syndrome	93575	Hemolytic-uremic syndrome without diarrhea with C3 anomaly
98759	HDL4	141145	Hemifacial hypertrophy	357008	Hemolytic-uremic syndrome without diarrhea with DGKE deficiency
313808	HDLS	141136	Hemifacial microsomia	93579	Hemolytic-uremic syndrome without diarrhea with H factor anomaly
2237	HDR syndrome	2549	Hemifacial microsomia-radial defects syndrome	93580	Hemolytic-uremic syndrome without diarrhea with I factor anomaly
402823	HDV	141148	Hemifacial myohyperplasia	93576	Hemolytic-uremic syndrome without diarrhea with MCP/CD46 anomaly
288	HE	221083	Hemifacial spasm	217023	Hemolytic-uremic syndrome without diarrhea with thrombomodulin anomaly
67037	Head and neck squamous cell carcinoma	276280	Hemihyperplasia-multiple lipomatosis syndrome	158048	Hemophagocytic syndrome associated with an infection
254898	Hearing loss-encephaloneuropathy-obesity-valvulopathy syndrome	99802	Hemimegalencephaly	98878	Hemophilia A
3225	Hearing loss-familial salivary gland insensitivity to aldosterone syndrome	306669	Hemiparkinsonism-hemiatrophy syndrome	98879	Hemophilia B
1338	Heart defect-tongue hamartoma-polysyndactyly syndrome	99050	Hemitruncus arteriosus	329	Hemophilia C
1354	Heart defects-limb shortening syndrome	139491	Hemochromatosis due to defect in ferroportin	178396	Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation
875	Heart tumor of child	79230	Hemochromatosis type 2	340	Hemorrhagic fever-renal syndrome
392	Heart-hand syndrome type 1	225123	Hemochromatosis type 3		
1350	Heart-hand syndrome type 2	139491	Hemochromatosis type 4		
1342	Heart-hand syndrome type 3	447792	Hemochromatosis type 5		
168796	Heart-hand syndrome, Slovenian type	163596	Hemoglobin Bart's hydrops fetalis		
1342	Heart-hand syndrome, Spanish type	2132	Hemoglobin C disease		
1342	Heart-limb syndrome type 3	231242	Hemoglobin C-beta-thalassemia syndrome		
442582	Heavy chain amyloidosis	90039	Hemoglobin D disease		
93556	Heavy chain deposition disease	2133	Hemoglobin E disease		
86864	Heavy chain disease	231249	Hemoglobin E-beta-thalassemia syndrome		
2119	HEC syndrome	93616	Hemoglobin H disease		
3377	Hecht syndrome	330032	Hemoglobin Lepore-beta-thalassemia syndrome		
3377	Hecht-Beals syndrome	330041	Hemoglobin M disease		
2492	Hecht-Scott syndrome	280615	Hemoglobinopathy Toms River		
238468	HED	244242	Hemolysis, elevated liver enzymes, low platelets in pregnancy		
98813	HED-ID	244242	Hemolysis-elevated liver enzymes-low platelets syndrome		
1882	HEDH syndrome	86817	Hemolytic anemia due to adenylate kinase deficiency		
2787	Heide syndrome				
3220	Heimler syndrome				
99932	Heiner syndrome				
178330	Heinz body anemia				
86813	Helicoid peripapillary chorioretinal degeneration				
168782	Heller syndrome				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
274	Hemorrhagiparous thrombocytic dystrophy	85450	Hereditary amyloid nephropathy	30925	Hereditary central diabetes insipidus
247245	Hemosiderosis of the central nervous system	93560	Hereditary amyloid nephropathy due to apolipoprotein A-I variant	221061	Hereditary cerebral cavernoma
324632	Hendra virus infection	238269	Hereditary amyloid nephropathy due to apolipoprotein A-II variant	221061	Hereditary cerebral cavernous malformation
2136	Hennekam syndrome	93562	Hereditary amyloid nephropathy due to fibrinogen A alpha-chain variant	85458	Hereditary cerebral hemorrhage with amyloidosis
2135	Hennekam-Beemer syndrome	93561	Hereditary amyloid nephropathy due to lysozyme variant	324723	Hereditary cerebral hemorrhage with amyloidosis, Arctic type
761	Henoch-Schönlein purpura	85450	Hereditary amyloidosis with primary renal involvement	100006	Hereditary cerebral hemorrhage with amyloidosis, Dutch type
95159	HEP	85448	Hereditary amyloidosis, Finnish type	324718	Hereditary cerebral hemorrhage with amyloidosis, Flemish type
79269	Heparan sulfamidase deficiency	228277	Hereditary anetoderma	100008	Hereditary cerebral hemorrhage with amyloidosis, Icelandic type
79271	Heparan-alpha-glucosaminide N-acetyltransferase deficiency	91378	Hereditary angioedema	324708	Hereditary cerebral hemorrhage with amyloidosis, Iowa type
3325	Heparin-associated thrombocytopenia	100050	Hereditary angioedema type 1	324713	Hereditary cerebral hemorrhage with amyloidosis, Italian type
3325	Heparin-induced thrombocytopenia type 2	100051	Hereditary angioedema type 2	324703	Hereditary cerebral hemorrhage with amyloidosis, Piedmont type
156	Hepatic carnitine palmitoyl transferase 1 deficiency	100054	Hereditary angioedema type 3	48818	Hereditary ceruloplasmin deficiency
156	Hepatic carnitine palmitoyl transferase I deficiency	91378	Hereditary angioneurotic edema	36382	Hereditary cervical artery dissection
386	Hepatic cystic hamartoma	100050	Hereditary angioneurotic edema type 1	53372	Hereditary chin myoclonus
2031	Hepatic fibrosis-renal cysts-intellectual disability syndrome	100051	Hereditary angioneurotic edema type 2	53372	Hereditary chin-trembling
369	Hepatic glycogen phosphorylase deficiency	100054	Hereditary angioneurotic edema type 3	676	Hereditary chronic pancreatitis
369	Hepatic phosphorylase deficiency	73229	Hereditary angiopathy-nephropathy-aneurysms-muscle cramps syndrome	422526	Hereditary clear cell renal cell adenocarcinoma
100035	Hepatic solitary necrotic nodule	3115	Hereditary areflexic dystasia, Roussy-Lévy type	422526	Hereditary clear cell renal cell carcinoma
890	Hepatic veno-occlusive disease	289601	Hereditary arterial and articular multiple calcification syndrome	293144	Hereditary clubfoot due to 5q31 microdeletion
79124	Hepatic veno-occlusive disease-immunodeficiency syndrome	1416	Hereditary articular chondrocalcinosis	238578	Hereditary clubfoot due to 17q23.1-q23.2 microduplication
90073	Hepatitis B reinfection following liver transplantation	352657	Hereditary benign corneal intraepithelial dyskeratosis	293150	Hereditary clubfoot due to PITX1 point mutation
402823	Hepatitis D virus	352657	Hereditary benign intraepithelial dyskeratosis	98434	Hereditary combined deficiency of factors II, VII, IX and X
402823	Hepatitis delta	91378	Hereditary bradykinine-induced angioedema	98434	Hereditary combined deficiency of vitamin K-dependent clotting factors
449	Hepatoblastoma	221061	Hereditary brain cavernous angioma	238722	Hereditary congenital contralateral synkinesia
54272	Hepatocellular adenoma	221061	Hereditary brain cavernous hemangioma	238722	Hereditary congenital mirror movements
88673	Hepatocellular carcinoma	145	Hereditary breast and ovarian cancer syndrome	972	Hereditary continuous muscle fiber activity
137681	Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	227535	Hereditary breast cancer	79273	Hereditary coproporphyrria
137681	Hepatoencephalopathy due to COXPD1	227535	Hereditary breast carcinoma	60015	Hereditary cranium bifidum
95159	Hepatoerythropoietic porphyria	871	Hereditary bundle branch defect	168577	Hereditary cryohydrocytosis type 2
905	Hepatolenticular degeneration	36382	Hereditary CAD	398088	Hereditary cryohydrocytosis with normal stomatin
64743	Hepatoportal sclerosis	1416	Hereditary calcium pyrophosphate deposition	168577	Hereditary cryohydrocytosis with reduced stomatin
364	Hepatorenal glycogenosis	1416	Hereditary CC	98967	Hereditary crystalline stromal dystrophy of Schnyder
882	Hepatorenal tyrosinemia	30925	Hereditary CDI		
86882	Hepatosplenic T-cell lymphoma				
306539	Hereditary acrokeratotic poikiloderma of Kindler-Weary				
2907	Hereditary acrokeratotic poikiloderma, Weary type				
447964	Hereditary adult-onset painful axonal polyneuropathy				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
100008	Hereditary cystatin C amyloid angiopathy	602	Hereditary inclusion body myopathy type 2	1839	Hereditary mucoepithelial dysplasia
26106	Hereditary diffuse cancer of stomach	79091	Hereditary inclusion body myopathy type 3	171723	Hereditary mucosal leukokeratosis
26106	Hereditary diffuse gastric adenocarcinoma	324381	Hereditary inclusion body myopathy type 4	136	Hereditary multi-infarct dementia
26106	Hereditary diffuse gastric cancer	178464	Hereditary inclusion body myopathy with early respiratory failure	→3460	Hereditary multicentric osteolysis
	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia	79091	Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome	523	Hereditary multiple cutaneous leiomyomas
313808	Hereditary diffuse leukoencephalopathy with spheroids	300373	Hereditary infantile gigantism	83454	Hereditary multiple gliangiomas
288	Hereditary elliptocytosis	397692	Hereditary isolated aplastic anemia	2590	Hereditary myoclonus-progressive distal muscular atrophy syndrome
→247691	Hereditary endotheliopathy-retinopathy-nephropathy-stroke syndrome	332	Hereditary juvenile megaloblastic anemia due to intrinsic factor deficiency	43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency
98873	Hereditary erythroblastic multinuclearity with a positive acidified-serum test (hempas)	2334	Hereditary keratitis	1062	Hereditary neurocutaneous malformation
36899	Hereditary essential myoclonus	493	Hereditary keratoacanthoma	456333	Hereditary neuroendocrine tumor of small bowel
85195	Hereditary expansile polyostotic osteolytic dysplasia	411602	Hereditary late-onset Parkinson disease	456333	Hereditary neuroendocrine tumor of small intestine
157846	Hereditary ferritinopathy	523	Hereditary leiomyomatosis	30925	Hereditary neurogenic diabetes insipidus
221043	Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome	523	Hereditary leiomyomatosis and renal cell cancer	640	Hereditary neuropathy with liability to pressure palsies
90045	Hereditary folate malabsorption	523	Hereditary leiomyomatosis with renal carcinoma	279943	Hereditary neutrophilia
469	Hereditary fructose intolerance	79452	Hereditary lymphedema type I	91378	Hereditary non histamine-induced angioedema
469	Hereditary fructose-1-phosphate aldolase deficiency	90186	Hereditary lymphedema type II	168583	Hereditary North American Indian childhood cirrhosis
469	Hereditary fructosemia	228277	Hereditary macular atrophy	56	Hereditary ochronosis
53372	Hereditary geniospasm	621	Hereditary methemoglobinemia	30	Hereditary orotic aciduria
2024	Hereditary gingival fibromatosis	157794	Hereditary mixed polyposis syndrome	98868	Hereditary ovalocytosis
2024	Hereditary gingival hyperplasia	64748	Hereditary motor and sensory neuropathy type 3	79141	Hereditary painful callosities
774	Hereditary hemorrhagic telangiectasia	773	Hereditary motor and sensory neuropathy type 4	86923	Hereditary palmoplantar hyperkeratosis, Gamborg-Nielsen type
2604	Hereditary hollow visceral myopathy	64751	Hereditary motor and sensory neuropathy type 5	86923	Hereditary palmoplantar keratoderma, Gamborg-Nielsen type
199285	Hereditary hypercarotenemia and vitamin A deficiency	90120	Hereditary motor and sensory neuropathy type 6	47044	Hereditary papillary renal cell carcinoma
238475	Hereditary hypercholanemia	64748	Hereditary motor and sensory neuropathy type III	99878	Hereditary parathyroids hyperplasia
3197	Hereditary hyperekplexia	90119	Hereditary motor and sensory neuropathy with acrodystrophy	476102	Hereditary pediatric Behçet-like disease
3197	Hereditary hyperexplexia	90103	Hereditary motor and sensory neuropathy with deafness, intellectual disability and absent sensory large myelinated fibers	168615	Hereditary persistence of alpha-fetoprotein
163	Hereditary hyperferritinemia with congenital cataracts	99950	Hereditary motor and sensory neuropathy, Lom type	46532	Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome
163	Hereditary hyperferritinemia-cataract syndrome	90117	Hereditary motor and sensory neuropathy, Okinawa type	251380	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome
2801	Hereditary hyperphosphatasia	90117	Hereditary motor and sensory neuropathy, proximal type	29072	Hereditary pheochromocytoma-paraganglioma
157215	Hereditary hypophosphatemic rickets with hypercalciuria	99953	Hereditary motor and sensory neuropathy, Russe Type	300373	Hereditary pituitary hyperplasia
55654	Hereditary hypotrichosis simplex			330061	Hereditary polymorphous light eruption of American Indians
90368	Hereditary hypotrichosis simplex of the scalp			178345	Hereditary prepubertal
217407	Hereditary hypotrichosis with recurrent skin vesicles				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	gynecomastia		neuropathy type I		congenital HRG deficiency
828	Hereditary progressive arthroophthalmopathy	139564	Hereditary sensory and autonomic neuropathy type IB	205	Hereditary unconjugated hyperbilirubinemia
98808	Hereditary progressive dystonia with marked diurnal fluctuation	970	Hereditary sensory and autonomic neuropathy type II	79234	Hereditary unconjugated hyperbilirubinemia type 1
158025	Hereditary progressive mucinous histiocytosis	1764	Hereditary sensory and autonomic neuropathy type III	79235	Hereditary unconjugated hyperbilirubinemia type 2
178464	Hereditary proximal myopathy with early respiratory failure	642	Hereditary sensory and autonomic neuropathy type IV	→247691	Hereditary vascular retinopathy
264675	Hereditary pulmonary alveolar proteinosis	64752	Hereditary sensory and autonomic neuropathy type V	→247691	Hereditary vascular retinopathy-Raynaud phenomenon-migraine syndrome
440427	Hereditary pulmonary alveolar proteinosis with hepatic involvement	314381	Hereditary sensory and autonomic neuropathy type VI	93160	Hereditary vitamin D-resistant rickets
275777	Hereditary pulmonary arterial hypertension	391397	Hereditary sensory and autonomic neuropathy type VII	903	Hereditary von Willebrand disease
→288	Hereditary pyropoikilocytosis	478664	Hereditary sensory and autonomic neuropathy type VIII	98805	Hereditary whispering dysphonia
85450	Hereditary renal amyloidosis	139573	Hereditary sensory and autonomic neuropathy with deafness and global delay	170	Hereditary woolly hair syndrome
93560	Hereditary renal amyloidosis due to apolipoprotein A-I variant	391397	Hereditary sensory and autonomic neuropathy with hyperhidrosis and gastrointestinal dysfunction	170	Hereditary woolly hair syndrome
238269	Hereditary renal amyloidosis due to apolipoprotein A-II variant	139578	Hereditary sensory and autonomic neuropathy with spastic paraparesis	3467	Hereditary xanthinuria
93562	Hereditary renal amyloidosis due to fibrinogen A alpha-chain variant	456318	Hereditary sensory neuropathy-deafness-dementia syndrome	3202	Hereditary xerocytosis
93561	Hereditary renal amyloidosis due to lysozyme variant	456318	Hereditary sensory neuropathy-sensorineural hearing loss-dementia syndrome	773	Heredopathia atactica polyneuritiformis
94088	Hereditary renal hypouricemia	213524	Hereditary site-specific ovarian cancer syndrome	275777	Heritable pulmonary arterial hypertension
788	Hereditary resistance to anti-vitamin K	100996	Hereditary spastic paraparesis type 15	3411	Herlyn-Werner syndrome
357027	Hereditary retinoblastoma	822	Hereditary spherocytosis	79430	Hermansky-Pudlak syndrome
221039	Hereditary sclerosing poikiloderma, Weary type	84093	Hereditary thermosensitive neuropathy	183678	Hermansky-Pudlak syndrome type 2
280598	Hereditary sensorimotor neuropathy with hyperelastic skin	71493	Hereditary thrombocythemia	231531	Hermansky-Pudlak syndrome type 7
36386	Hereditary sensory and autonomic neuropathy type 1	480851	Hereditary thrombocytopenia with early-onset myelofibrosis	231537	Hermansky-Pudlak syndrome type 8
139564	Hereditary sensory and autonomic neuropathy type 1 with cough and gastroesophageal reflux	268322	Hereditary thrombocytopenia with normal platelets	280663	Hermansky-Pudlak syndrome type 9
139564	Hereditary sensory and autonomic neuropathy type 1B	71290	Hereditary thrombocytopenia with normal platelets-hematological cancer predisposition syndrome	183678	Hermansky-Pudlak syndrome with neutropenia
970	Hereditary sensory and autonomic neuropathy type 2	329319	Hereditary thrombocytosis with transverse limb defect	231500	Hermansky-Pudlak syndrome with pulmonary fibrosis
1764	Hereditary sensory and autonomic neuropathy type 3	82	Hereditary thrombophilia due to congenital antithrombin 3 deficiency	231512	Hermansky-Pudlak syndrome without pulmonary fibrosis
642	Hereditary sensory and autonomic neuropathy type 4	82	Hereditary thrombophilia due to congenital antithrombin deficiency	2139	Hernández-Aguirre Negrete syndrome
64752	Hereditary sensory and autonomic neuropathy type 5	217467	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	2786	Hernández-Fragoso syndrome
314381	Hereditary sensory and autonomic neuropathy type 6	217467	Hereditary thrombophilia due to congenital antithrombin 3 deficiency	→247691	HERNS syndrome
391397	Hereditary sensory and autonomic neuropathy type 7			1930	Herpes simplex meningoencephalitis
478664	Hereditary sensory and autonomic neuropathy type 8			1930	Herpes simplex neuroinvasion
36386	Hereditary sensory and autonomic			1930	Herpes simplex virus encephalitis

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
640	Heterozygous microdeletion 17p11.2p12		imperfecta		disability syndrome
3450	Heterozygous OSMED	480541	High grade B-cell lymphoma with MYC and/ or BCL2 and/or BCL6 rearrangement	2153	Hirschsprung disease-nail hypoplasia-dysmorphism syndrome
3450	Heterozygous otospondylomegaepiphseal dysplasia	363396	High myopia-sensorineural deafness syndrome	2150	Hirschsprung disease-type D brachydactyly syndrome
845	Hexosaminidase A deficiency	3181	High scapula	2026	Hirsutism-congenital gingival hyperplasia syndrome
309192	Hexosaminidase A deficiency, adult form	231080	High-grade dysplasia in patients with Barrett esophagus	3283	His bundle tachycardia
309239	Hexosaminidase A deficiency, B1 variant	251646	High-grade ependymoma	2157	HIS deficiency
309178	Hexosaminidase A deficiency, infantile form	213777	High-grade neuroendocrine carcinoma of the cervix uteri	2157	Histidase deficiency
309185	Hexosaminidase A deficiency, juvenile form	213731	High-grade neuroendocrine carcinoma of the corpus uteri	2157	Histidine ammonia-lyase deficiency
309246	Hexosaminidase activator deficiency	213777	High-grade neuroendocrine carcinoma of the uterine cervix	2157	Histidinemia
796	Hexosaminidases A and B deficiency	213731	High-grade neuroendocrine carcinoma of the uterine corpus	2157	Histidinuria
309169	Hexosaminidases A and B deficiency, adult form	101088	HIGM1	2158	Histidinuria-renal tubular defect syndrome
309155	Hexosaminidases A and B deficiency, infantile form	101089	HIGM2	50918	Histiocytic necrotizing lymphadenitis
309162	Hexosaminidases A and B deficiency, juvenile form	101090	HIGM3	86896	Histiocytic sarcoma
1041	HF	101091	HIGM4	137675	Histiocytoid cardiomyopathy
2438	HFGS	101092	HIGM5	390	Histoplasmosis
2744	HGPPS	183663	HIGM with susceptibility to opportunistic infections	3325	HIT
740	HGPS	183666	HIGM without susceptibility to opportunistic infections	→138	Hittner-Hirsch-Kreh syndrome
79271	HGSNAT deficiency	99978	Hilar CCA	443291	HIV-associated cancer
163	HHCS	99978	Hilar cholangiocarcinoma	443291	HIV-related cancer
86908	HHE syndrome	84085	Hinman syndrome	1573	HJMD
415	HHH syndrome	84085	Hinman-Allen syndrome	572	HLA class 2-negative SCID
276280	HHML	1164	Hinson-Pepys disease	572	HLA class 2-negative severe combined immunodeficiency
157215	HHRH	2114	Hip dysplasia, Beukes type	2248	HLHS
774	HHT	3408	Hip dysplasia-enchondromata-ecchondroma syndrome	→444490	HLP type 1
457	HI	411593	Hirata disease	412	HLP type 3
435	HI syndrome	65684	Hirayama disease	70470	HLP type 5
35878	HI/HA syndrome	388	Hirschsprung disease	523	HLRCC
88639	HIBCH deficiency	261537	Hirschsprung disease and intellectual disability due to 2q22 microdeletion	2213	HMC syndrome
602	HIBM2	261552	Hirschsprung disease and intellectual disability due to a ZEB2 point mutation	178464	HMERF
79091	HIBM3	261537	Hirschsprung disease and intellectual disability due to del(2)(q22)	20	HMG-CoA lyase deficiency
324381	HIBM4	261537	Hirschsprung disease and intellectual disability due to monosomy 2q22	35701	HMG-CoA synthase deficiency
178464	HIBM-ERF	2155	Hirschsprung disease-deafness-polydactyly syndrome	157794	HMPS
189	Hidrotic ectodermal dysplasia	2151	Hirschsprung disease-ganglioneuroblastoma syndrome	64748	HMSN 3
1808	Hidrotic ectodermal dysplasia, Christianson-Fourie type	2152	Hirschsprung disease-intellectual	773	HMSN 4
1809	Hidrotic ectodermal dysplasia, Halal type			64751	HMSN 5
343	HIDS			401964	HMSN2 with giant axons
137577	HIE			64748	HMSN III
330012	High altitude pulmonary edema			90119	HMSN with acrodyostrophy
171201	High anorectal malformation			99950	HMSN, Lom type
314029	High bone mass OI			99950	HMSN-Lom
314029	High bone mass osteogenesis			90117	HMSNP

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
1979	Hoepffner-Dreyer-Reimers syndrome		elliptocytosis		gastroesophageal reflux
2349	Hoffman syndrome	98958	Honey-droplet corneal dystrophy	139573	HSAN with deafness and global delay
391665	HoFH	98960	Honeycomb corneal dystrophy	391397	HSAN with hyperhidrosis and gastrointestinal dysfunction
414	HOGA	78	Hookworm infection	139578	HSAN with spastic paraplegia
454718	Holmes-Adie syndrome	307936	HOPP syndrome	2182	HSAS
→994	Holmes-Benacerraf syndrome	2744	Horizontal gaze palsy with progressive scoliosis	388	HSCR
3328	Holmes-Collins syndrome	397	Horton disease	391417	HSD10 deficiency
93970	Holmes-Gang syndrome	392	HOS	85295	HSD10 deficiency, atypical type
2143	Holmes-Schepens syndrome	166412	Hot water reflex epilepsy	391428	HSD10 deficiency, classic type
79242	Holocarboxylase synthetase deficiency	1352	Houlston-Ironton-Temple syndrome	391428	HSD10 deficiency, infantile type
280200	Holoprosencéphalie, minor form	99907	House allergic alveolitis	391457	HSD10 deficiency, neonatal type
2162	Holoprosencephaly	2198	Howell-Evans syndrome	391417	HSD10 disease
2165	Holoprosencephaly-caudal dysgenesis syndrome	3322	Hoyerala-Hreidarsson syndrome	85295	HSD10 disease, atypical type
2163	Holoprosencephaly-craniosynostosis syndrome	306669	HP-HA syndrome	391428	HSD10 disease, classic type
2117	Holoprosencephaly-ectrodactyly-cleft lip/palate syndrome	275777	HPAH	391428	HSD10 disease, infantile type
2570	Holoprosencephaly-fetal akinesia/hypokinesia sequence syndrome	98808	HPD with marked diurnal fluctuation	391457	HSD10 disease, neonatal type
2570	Holoprosencephaly-hypokinesia-congenital contractures syndrome	2162	HPE	1930	HSE
280200	Holoprosencephaly-like	280200	HPE, minor form	30924	HSH
2166	Holoprosencephaly-postaxial polydactyly syndrome	280200	HPE-L	456318	HSN1E
3186	Holoprosencephaly-radial heart renal anomalies syndrome	46532	HPFH-beta-thalassemia syndrome	1930	HSV encephalitis
392	Holt-Oram syndrome	251380	HPFH-sickle cell disease syndrome	1930	HSVE
2167	Holzgreve syndrome	436	HPP	285	HT-EDS
2167	Holzgreve-Wagner-Rehder syndrome	293958	HPPD	289326	HTLV-1-associated myelopathy/tropical spastic paraparesis
30924	HOMG1	47044	HPRCC	482077	HTRA1-related autosomal dominant cerebral angiopathy
34528	HOMG2	79233	HPRT1 partial deficiency	482077	HTRA1-related autosomal dominant cerebral small vessel disease
31043	HOMG3	510	HPRT complete deficiency	228116	Hughes-Stovin syndrome
2168	Homocarnosinase deficiency	510	HPRT deficiency grade IV	438279	Human infection by orthopoxvirus
2168	Homocarnosinosis	79233	HPRT deficiency, grade I	289326	Human T-lymphotropic virus type I-associated myelopathy/tropical spastic paraparesis
394	Homocystinuria due to cystathione beta-synthase deficiency	79233	HPRT partial deficiency	289326	Human T-lymphotropic virus type-1-associated myelopathy/tropical spastic paraparesis
395	Homocystinuria due to methylene tetrahydrofolate reductase deficiency	79233	HPRT-related gout	294973	Humeral agenesis/hypoplasia
622	Homocystinuria without methylmalonic aciduria	79233	HPRT-related hyperuricemia	294973	Humeral intercalary meromelia
56	Homogentisic acid oxidase deficiency	79430	HPS	3265	Humero-radial fusion
163596	Homozygous alpha0-thalassemia	183678	HPS2	295211	Humero-radial fusion, bilateral
391665	Homozygous familial hypercholesterolemia	231531	HPS7	295209	Humero-radial fusion, unilateral
14	Homozygous familial hypobetalipoproteinemia	231537	HPS8	3265	Humero-radial synostosis
→288	Homozygous hereditary	280663	HPS9	295211	Humero-radial synostosis, bilateral
		231500	HPS with pulmonary fibrosis	295209	Humero-radial synostosis, unilateral
		231512	HPS without pulmonary fibrosis	3266	Humero-radio-ulnar fusion
		99880	HPT-JT	295207	Humero-radio-ulnar fusion, bilateral
		2323	HRD syndrome	295205	Humero-radio-ulnar fusion, unilateral
		84085	HS	294975	Humero-radio-ulnar intercalary
		36386	HSAN1		
		139564	HSAN1B		
		456318	HSAN1E		
		970	HSAN2		
		1764	HSAN3		
		642	HSAN4		
		64752	HSAN5		
		314381	HSAN6		
		391397	HSAN7		
		478664	HSAN8		
		139564	HSAN with cough and		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	transverse meromelia	400	Hydatidosis	101088	Hyper-IgM syndrome type 1
3266	Humero-radio-ulnar synostosis	2898	Hyde Forster-McCarthy-Berry syndrome	101089	Hyper-IgM syndrome type 2
295207	Humero-radio-ulnar synostosis, bilateral	2177	Hydranencephaly	101090	Hyper-IgM syndrome type 3
295205	Humero-radio-ulnar synostosis, unilateral	330021	Hydrargyria	101091	Hyper-IgM syndrome type 4
→263463	Humero-spinal dysostosis	330061	Hydroa aestivale	101092	Hyper-IgM syndrome type 5
94056	Humero-ulnar fusion	330058	Hydroa vacciniforme	183663	Hyper-IgM syndrome with susceptibility to opportunistic infections
295215	Humero-ulnar fusion, bilateral	364039	Hydroa vacciniforme-like lymphoma	183666	Hyper-IgM syndrome without susceptibility to opportunistic infections
295213	Humero-ulnar fusion, unilateral	364039	Hydroa-like cutaneous T-cell lymphoma	309147	Hyperalaninemia
94056	Humero-ulnar synostosis	2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	927	Hyperammonemia due to N-acetylglutamate synthase deficiency
295215	Humero-ulnar synostosis, bilateral	899	Hydrocephalus-agyria-retinal dysplasia syndrome	401948	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency
295213	Humero-ulnar synostosis, unilateral	2186	Hydrocephalus-blue sclerae-nephropathy syndrome	168588	Hyperandrogenism due to cortisone reductase deficiency
→263463	Humerospinal dysostosis	916	Hydrocephalus-cleft palate-joint contractures syndrome	90	Hyperargininemia
3383	Humerus trochlea aplasia	2180	Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome	234	Hyperbilirubinemia type 2
580	Hunter syndrome	2119	Hydrocephalus-endocardial fibroelastosis-cataract syndrome	3111	Hyperbilirubinemia, Rotor type
217085	Hunter syndrome type A	2183	Hydrocephalus-obesity-hypogonadism syndrome	276405	Hyperbiliverdinemia
217093	Hunter syndrome type B	1397	Hydrocephaly-cerebellar agenesis syndrome	306661	Hypercalcemic tumoral calcinosis
→35069	Hunter-Carpenter-McDonald syndrome	2184	Hydrocephaly-low insertion umbilicus syndrome	2196	Hypercalciuria-bilateral macular coloboma syndrome
2715	Hunter-Jurenka-Thompson syndrome	2181	Hydrocephaly-tall stature-joint laxity syndrome	209902	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency
97340	Hunter-McAlpine craniosynostosis	221126	Hydrocephaly/hydranencephaly due to cerebral vasculopathy	83639	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency
3365	Hunter-Rudd-Hoffmann syndrome	2189	Hydrocephalus	1032	Hyperdibasic aminoaciduria type 1
1390	Hunter-Thompson-Reed syndrome	2473	Hydrometrocolpos-postaxial polydactyly syndrome	470	Hyperdibasic aminoaciduria type 2
399	Huntington chorea	2704	Hydronephrosis-inverted smile syndrome	3197	Hyperekplexia
399	Huntington disease	1041	Hydrops fetalis	163985	Hyperekplexia-epilepsy syndrome
401901	Huntington disease phenocopy due to C9ORF72 expansions	1426	Hydrops-ectopic calcification-motheaten syndrome	408	Hyperglycerolemia
157941	Huntington disease-like 1	20	Hydroxymethylglutaric aciduria	2410	Hypergonadotropic hypogonadism-cataract syndrome
98934	Huntington disease-like 2	401	Hymenolepisis	243	Hypergonadotropic ovarian dysgenesis
157946	Huntington disease-like 3	309147	Hyper-beta-alaninemia	2157	Hyperhistidinemia
98759	Huntington disease-like 4	343	Hyper-IgD syndrome	742	Hyperimidodipeptiduria
401901	Huntington disease-like syndrome due to C9ORF72 expansions	101090	Hyper-IgM syndrome due to CD40 deficiency	343	Hyperimmunoglobinemia D with recurrent fever
363694	HUPRA syndrome	101088	Hyper-IgM syndrome due to CD40 ligand deficiency	2314	Hyperimmunoglobulin E syndrome type 1
384	Huriez syndrome	101088	Hyper-IgM syndrome due to CD40L deficiency	2314	Hyperimmunoglobulin E-recurrent infection syndrome
93473	Hurler disease	101092	Hyper-IgM syndrome due to UNG deficiency	343	Hyperimmunoglobulinemia D syndrome
93473	Hurler syndrome	101092	Hyper-IgM syndrome due to uracil N-glycosylase	343	Hyperimmunoglobulinemia D with periodic fever
93476	Hurler-Scheie syndrome			79299	Hyperinsulinemic hypoglycemia due to glucokinase deficiency
330061	Hutchinson summer prurigo				
740	Hutchinson-Gilford progeria syndrome				
93160	HVDRR				
364039	HVLL				
→247691	HVR				
53698	Hyaline body myopathy				
498474	Hyaline fibromatosis syndrome				
70587	Hyaline membrane disease				
530	Hyalinosi cutis et mucosae				
508476	Hyaluronidase 2 deficiency				
67041	Hyaluronidase deficiency				
400	Hydatid disease				
99927	Hydatidiform mole				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
324575	Hyperinsulinemic hypoglycemia due to HNF1A deficiency		triacylglycerol lipase deficiency		dehydratase deficiency
263455	Hyperinsulinemic hypoglycemia due to HNF4A deficiency	140905	Hyperlipidemia due to hepatic triglyceride lipase deficiency	226	Hyperphenylalaninemia due to dihydropteridine reductase deficiency
263458	Hyperinsulinemic hypoglycemia due to INSR deficiency	140905	Hyperlipidemia due to HL deficiency	508523	Hyperphenylalaninemia due to DNAJC12 deficiency
263458	Hyperinsulinemic hypoglycemia due to insulin receptor deficiency	140905	Hyperlipidemia due to HTGL deficiency	2102	Hyperphenylalaninemia due to GTP cyclohydrolase deficiency
276603	Hyperinsulinemic hypoglycemia due to Kir6.2 deficiency, diazoxide-resistant focal form	412	Hyperlipidemia type 3	1578	Hyperphenylalaninemia due to pterin-4-alpha-carbinolamine dehydratase deficiency
71212	Hyperinsulinemic hypoglycemia due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	→444490	Hyperlipoproteinemia type 1	238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency
276598	Hyperinsulinemic hypoglycemia due to SUR1 deficiency, diazoxide-resistant focal form	412	Hyperlipoproteinemia type 3	1578	Hyperphenylalaninemia with primapterinuria
276556	Hyperinsulinemic hypoglycemia due to UCP2 deficiency	70470	Hyperlipoproteinemia type 5	2209	Hyperphenylalaninemic embryopathy
79299	Hyperinsulinism due to glucokinase deficiency	2203	Hyperlysinemia	3416	Hyperphosphatasemia tarda
71212	Hyperinsulinism due to glutamodehydrogenase deficiency	2203	Hyperlysinemia type I	247262	Hyperphosphatasia-intellectual disability syndrome
324575	Hyperinsulinism due to HNF1A deficiency	3124	Hyperlysinemia type II	→79189	Hyperpipecolatemia
263455	Hyperinsulinism due to HNF4A deficiency	289891	Hypermethioninemia due to glycine N-methyltransferase deficiency	157798	Hyperplastic polyposis syndrome
263458	Hyperinsulinism due to INSR deficiency	289891	Hypermethioninemia due to GNMT deficiency	682	HyperPP
165991	Hyperinsulinism due to monocarboxylate transporter 1 deficiency	88618	Hypermethioninemia due to S-adenosylhomocysteine hydrolase deficiency	419	Hyperprolinemia type 1
71212	Hyperinsulinism due to SCHAD deficiency	289290	Hypermethioninemia encephalopathy due to adenosine kinase deficiency	79101	Hyperprolinemia type 2
71212	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	289290	Hypermethioninemia encephalopathy due to ADK deficiency	93604	Hyperprostaglandin E syndrome
165991	Hyperinsulinism due to SLC16A1 deficiency	73267	Hypernychthemeral syndrome	889	Hypersensitivity angiitis
276556	Hyperinsulinism due to UCP2 deficiency	414	Hyperornithinemia	1519	Hypertelorism, Teebi type
35878	Hyperinsulinism-hyperammonemia syndrome	414	Hyperornithinemia-gyrate atrophy of choroid and retina syndrome	2211	Hypertelorism-hypospadias-polysyndactyly syndrome
757	Hyperkalemia-hypertension syndrome, Gordon type	415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	2213	Hypertelorism-microtia-facial clefting syndrome
682	Hyperkalemic periodic paralysis	2801	Hyperostosis corticalis deformans juvenilis	2745	Hypertelorism-oesophageal abnormality-hypospadias syndrome
682	Hyperkalemic PP	3416	Hyperostosis corticalis generalisata	293958	Hypertelorism-preauricular sinus-punctual pits-deafness syndrome
409	Hyperkeratosis lenticularis perstans	443098	Hyperostosis cranialis interna	293958	Hypertelorism-preauricular sinus-punctual pits-hearing loss syndrome
1662	Hyperkeratosis-contracture syndrome	77296	Hyperostosis frontalis interna	88660	Hypertension due to gain-of-function mutations in the mineralocorticoid receptor
1336	Hyperkeratosis-hyperpigmentation syndrome	2780	Hyperostosis generalisata with striations	757	Hypertensive hyperkalemia
682	HyperKPP	99880	Hyperparathyroidism-jaw tumor syndrome	423	Hyperthermia of anesthesia
140905	Hyperlipidemia due to hepatic lipase deficiency	295002	Hyperphalangy	2220	Hypertrichosis cubiti
140905	Hyperlipidemia due to hepatic	295140	Hyperphalangy in digits 2-5	2222	Hypertrichosis lanuginosa congenita
		295142	Hyperphalangy, bilateral	2222	Hypertrichosis universalis
		295140	Hyperphalangy, unilateral	2026	Hypertrichosis with or without gingival hyperplasia
		1388	Hyperphalangy-clinodactyly of index finger with Pierre Robin syndrome	966	Hypertrichosis-acromegaloid facial appearance syndrome
		13	Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency	966	Hypertrichosis-acromegaloid facial features syndrome
		238583	Hyperphenylalaninemia due to BH4 deficiency	1231	Hypertrichosis-atrophic skin-ectropion-macrostomia syndrome
		1578	Hyperphenylalaninemia due to		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
966	Hypertrichosis-coarse face syndrome		frontoparietal alopecia syndrome	2237	Hypoparathyroidism-deafness-renal disease syndrome
319182	Hypertrichosis-short stature-facial dysmorphism-developmental delay syndrome	2235	Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome	2323	Hypoparathyroidism-intellectual disability-dysmorphism syndrome
1517	Hypertrichotic osteochondrodysplasia, Cantu type	293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural deafness-dysmorphism syndrome	2323	Hypoparathyroidism-short stature-intellectual disability-seizures syndrome
324525	Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation	293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome	436	Hypophosphatasia
324525	Hypertrophic cardiomyopathy and renal tubular disease due to mtDNA mutation	363523	Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome	314621	Hypophyseal duplication
217601	Hypertrophic cardiomyopathy due to intensive athletic training	238468	Hypohidrotic ectodermal dysplasia	99725	Hypophyseal gigantism
329883	Hypertrophic gastropathy without hypoproteinemia	98813	Hypohidrotic ectodermal dysplasia with immunodeficiency	324561	Hypopigmentation and punctate keratosis of the palms and soles
90282	Hypertrophic or verrucous lupus erythematosus	1882	Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome	42665	Hypopigmentation-deafness syndrome
2224	Hypertryptophanemia	293964	Hypoinsulinemic hypoglycemia and body hemihypertrophy	79477	Hypopigmentation-immunodeficiency with or without neurologic impairment syndrome
363694	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome	681	Hypokalemic periodic paralysis	79476	Hypopigmentation-neurologic impairment syndrome
251523	Hyperzincemia and hypercalprotectinemia	30924	Hypomagnesemia caused by selective magnesium malabsorption	324561	Hypopigmentation-punctate palmoplantar keratoderma syndrome
276429	Hypnic headache	30924	Hypomagnesemia intestinal type 1	91354	Hypopituitarism due to empty sella turcica syndrome
2435	Hypo- and hypermelanotic cutaneous macules-retarded growth-intellectual disability syndrome	1790	Hypomandibular faciocranial dysostosis	→3157	Hypopituitarism-micropenis-cleft lip/palate syndrome
289157	Hypocalcemic vitamin D-dependent rickets	100033	Hypomaturational amelogenesis imperfecta	→3157	Hypopituitarism-microphthalmia syndrome
93160	Hypocalcemic vitamin D-resistant rickets	100034	Hypomaturational-hypoplastic amelogenesis imperfecta with taurodontism	99058	Hypoplasia of the mitral valve annulus
100032	Hypocalcified amelogenesis imperfecta	435	Hypomelanosis of Ito	722	Hypoplasmminogenemia
93297	Hypochondrogenesis	495844	Hypomyelinating leukodystrophy due to hikeshi deficiency	100031	Hypoplastic amelogenesis imperfecta
429	Hypochondroplasia	137639	Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome	2248	Hypoplastic left heart syndrome
36412	Hypocomplementemic urticarial vasculitis	2680	Hypomyelination neuropathy-arthrogryposis syndrome	293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome
2228	Hypodontia-dysplasia of nails syndrome	139441	Hypomyelination with atrophy of basal ganglia and cerebellum	3332	Hypoplastic tibiae-postaxial polydactyly syndrome
2228	Hypodontia-nail dysgenesis syndrome	363412	Hypomyelination with brain stem and spinal cord involvement and leg spasticity	→216866	Hypoprebetalipoproteinemia-acanthocytosis-retinitis pigmentosa-pallidal degeneration syndrome
185	Hypogenetic lung syndrome	447893	Hypomyelination-cerebellar atrophy-hypoplasia of the corpus callosum syndrome	327	Hypoproconvertinemia
989	Hypoglossia-hypodactyly syndrome	85163	Hypomyelination-congenital cataract syndrome	2494	Hypoproteinemic hypertrophic gastropathy
→261483	Hypogonadism-gynecomastia-X-linked intellectual disability syndrome	88637	Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome	325	Hypoprothrombinemia
2233	Hypogonadism-mitral valve prolapse-intellectual disability syndrome	3453	Hypoparathyroidism-Addison disease-mucocutaneous candidiasis syndrome	2250	Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome
141333	Hypogonadism-short stature-coloboma-preaxial polydactyly syndrome			2745	Hypospadias-dysphagia syndrome
2230	Hypogonadotropic hypogonadism-			2745	Hypospadias-hypertelorism syndrome
				→1299	Hypospadias-hypertelorism-coloboma and deafness syndrome
				2261	Hypospadias-intellectual disability, Goldblatt type syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2353	Hypotelorism-cleft palate-hypospadias syndrome	307936	Hypotrichosis-striate palmoplantar hyperkeratosis-acroosteolysis-periodontitis syndrome	79504	Ichthyosis hystrix gravior
443101	Hypothalamic adipsic hypernatraemia syndrome	307936	Hypotrichosis-striate palmoplantar keratoderma-acroosteolysis-periodontitis syndrome	79503	Ichthyosis hystrix of Curth-Macklin
672	Hypothalamic hamartoblastoma syndrome	79233	Hypoxanthine guanine phosphoribosyltransferase 1 partial deficiency	79503	Ichthyosis hystrix, Curth-Macklin type
86906	Hypothalamic hamartomas with gelastic seizures	510	Hypoxanthine guanine phosphoribosyltransferase complete deficiency	281190	Ichthyosis variegata
→3157	Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome	79233	Hypoxanthine guanine phosphoribosyltransferase deficiency, grade I	281190	Ichthyosis with confetti
226307	Hypothyroidism due to deficient transcription factors involved in pituitary development or function	510	Hypoxanthine guanine phosphoribosyltransferase deficiency, grade IV	79504	Ichthyosis, Lambert type
90673	Hypothyroidism due to TSH receptor mutations	79233	Hypoxanthine guanine phosphoribosyltransferase partial deficiency	2269	Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome
1226	Hypothyroidism-cleft palate syndrome	137577	Hypoxic and ischemic brain injury in the newborn	91132	Ichthyosis-follicular atrophoderma-hypotrichosis syndrome
3047	Hypothyroidism-dysmorphism-postaxial polydactyly-intellectual disability syndrome	137577	Hypoxic-ischemic encephalopathy	91132	Ichthyosis-follicular atrophoderma-hypotrichosis-hypohidrosis syndrome
91131	Hypotonia and ichthyosis due to dolichol phosphate deficiency	682	HYPP	2274	Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome
137908	Hypotonia with lactic acidemia and hyperammonemia	63440	Hypsicephaly	91132	Ichthyosis-hypotrichosis syndrome
163690	Hypotonia-cystinuria syndrome	63440	Hypocephaly	59303	Ichthyosis-hypotrichosis-sclerosing cholangitis syndrome
79507	Hypotonia-failure to thrive-microcephaly syndrome	576	I-cell disease	2278	Ichthyosis-intellectual disability-dwarfism-renal impairment syndrome
371364	Hypotonia-speech impairment-severe cognitive delay syndrome	480512	IAD	→1643	Ichthyosis-male hypogonadism syndrome
55654	Hypotrichosis simplex	724	IAEP	2272	Ichthyosis-oral and digital anomalies syndrome
90368	Hypotrichosis simplex of the scalp	158048	IAHS	88621	Ichthyosis-prematurity syndrome
1573	Hypotrichosis with juvenile macular degeneration	293168	IAHSP	363992	Ichthyosis-short stature-brachydactyly-microspherophakia syndrome
1573	Hypotrichosis with juvenile macular dystrophy	254509	Iatrogenic botulism	289347	IDH
444	Hypotrichosis, Marie Unna type	95619	Iatrogenic or traumatic pituitary deficiency	3306	idic(15)
91132	Hypotrichosis-congenital ichthyosis syndrome	363424	IBA57 deficiency	930	Idiopathic achalasia
330029	Hypotrichosis-deafness syndrome	→33364	IBIDS syndrome	930	Idiopathic achalasia of esophagus
2266	Hypotrichosis-intellectual disability, Lopes type	611	IBM	724	Idiopathic acute eosinophilic pneumonia
69735	Hypotrichosis-lymphedema-telangiectasia-membranoproliferative glomerulonephritis syndrome	602	IBM2	139423	Idiopathic acute transverse myelitis
69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	79091	IBM3	480512	Idiopathic adult ductopenia
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar hyperkeratosis syndrome	52430	IBMPFD	422	Idiopathic and/or familial pulmonary arterial hypertension
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar keratoderma syndrome	37202	IC/BPS	280914	Idiopathic anterior uveitis
		37202	IC/PBS	88	Idiopathic aplastic anemia
		31709	ICCA syndrome	206599	Idiopathic asymptomatic hyperCKemia
		64734	ICE syndrome	399307	Idiopathic avascular necrosis
		2268	ICF syndrome	399307	Idiopathic AVN
		455	Ichthyosis bullous of Siemens	1980	Idiopathic basal ganglia calcification
		457	Ichthyosis congenita, Harlequin type	171684	Idiopathic bilateral vestibulopathy
		289586	Ichthyosis exfoliativa	84065	Idiopathic bile acid malabsorption
		457	Ichthyosis fetalis, Harlequin type	88	Idiopathic bone marrow failure
		2273	Ichthyosis follicularis-alopecia-photophobia syndrome	60033	Idiopathic bronchiectasis
		2273	Ichthyosis follicularis-atrichia-photophobia syndrome	1320	Idiopathic camptocormia
				1320	Idiopathic camptocormism
				188	Idiopathic capillary leak syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
163703	Idiopathic catastrophic epileptic encephalopathy	2573	Idiopathic Moyamoya disease		nephrotic syndrome with focal segmental hyalinosis
228000	Idiopathic CD4 lymphocytopenia	2774	Idiopathic multicentric osteolysis with or without nephropathy	93207	Idiopathic steroid-sensitive nephrotic syndrome with minimal change
169615	Idiopathic central precocious puberty	824	Idiopathic myelofibrosis	99858	Idiopathic syringomyelia
2902	Idiopathic chronic eosinophilic pneumonia	45452	Idiopathic neonatal atrial flutter	256	Idiopathic torsion dystonia
95717	Idiopathic congenital hypothyroidism	33577	Idiopathic nodular panniculitis	98806	Idiopathic torsion dystonia of mixed type
209919	Idiopathic copper-associated cirrhosis	51608	Idiopathic obliterative arteriopathy	3347	Idiopathic tracheobronchomegaly
35062	Idiopathic disseminated CMV infection	441	Idiopathic orthostatic hypotension	79153	Idiopathic trachyonychia
35062	Idiopathic disseminated cytomegalovirus infection	280921	Idiopathic panuveitis	209956	Idiopathic uveal effusion syndrome
447881	Idiopathic dropped head syndrome	747	Idiopathic PAP	130	Idiopathic ventricular fibrillation, Brugada type
480512	Idiopathic ductopenia	480524	Idiopathic peliosis hepatitis	228140	Idiopathic ventricular fibrillation, non Brugada type
256	Idiopathic dystonia	480524	Idiopathic peliosis hepatitis	280384	IDMDC
247724	Idiopathic eosinophilia-associated myopathy	444316	Idiopathic phalangeal acroosteolysis	580	Iduronate 2-sulfatase deficiency
247724	Idiopathic eosinophilic myositis	444316	Idiopathic phalangeal acroosteolysis	217085	Iduronate 2-sulfatase deficiency type A
329874	Idiopathic giant cell myocarditis	494428	Idiopathic pleuroparenchymal fibroelastosis	217093	Iduronate 2-sulfatase deficiency type B
64722	Idiopathic granulomatous mastitis	494428	Idiopathic pleuropulmonary fibroelastosis	92050	IED
86908	Idiopathic hemiconvulsion-hemiplegia syndrome	280917	Idiopathic posterior uveitis	91132	IFAH syndrome
2197	Idiopathic hypercalciuria	1320	Idiopathic progressive lumbar kyphosis	2273	IFAP syndrome
3260	Idiopathic hypereosinophilic syndrome	747	Idiopathic pulmonary alveolar proteinosis	332	IFD
33208	Idiopathic hypersomnia	275766	Idiopathic pulmonary arterial hypertension	329903	Ig-mediated membranoproliferative glomerulonephritis
228315	Idiopathic hypersomnia with long sleep time	1676	Idiopathic pulmonary artery dilatation	329903	Ig-mediated MPGN
228318	Idiopathic hypersomnia without long sleep time	2032	Idiopathic pulmonary fibrosis	761	IgA vasculitis
449427	Idiopathic hypertrophic pachymeningitis	99931	Idiopathic pulmonary hemosiderosis	329874	IGCM
1572	Idiopathic immunoglobulin deficiency	35061	Idiopathic recurrent and disabling cutaneous herpes	79099	IGDA
51608	Idiopathic infantile arterial calcification	251307	Idiopathic recurrent pericarditis	73272	IGF-1 deficiency
238624	Idiopathic intracranial hypertension	276174	Idiopathic recurrent stupor	449400	IgG4-related aortitis
85193	Idiopathic juvenile osteoporosis	251307	Idiopathic relapsing pericarditis	79078	IgG4-related dacryoadenitis and sialadenitis
247234	Idiopathic late-onset cerebellar ataxia	40923	Idiopathic retinal perivasculitis	449566	IgG4-related eosinophilic angiocentric fibrosis
314017	Idiopathic linear interstitial keratitis	40923	Idiopathic retinal vasculitis	90003	IgG4-related hepatopathy
33577	Idiopathic lobular panniculitis	209943	Idiopathic retinal vasculitis-aneurysms-neuroretinitis syndrome	449395	IgG4-related kidney disease
90158	Idiopathic localized lipodystrophy	49041	Idiopathic retroperitoneal fibrosis	63999	IgG4-related mediastinitis
353344	Idiopathic macular telangiectasia type 1	458718	Idiopathic SCAD	238593	IgG4-related mesenteritis
353351	Idiopathic macular telangiectasia type 3	35065	Idiopathic severe pneumococcemia	449563	IgG4-related ophthalmic disease
84065	Idiopathic malabsorption due to bile acid synthesis defects	458718	Idiopathic spontaneous coronary artery dissection	449427	IgG4-related pachymeningitis
73	Idiopathic massive osteolysis	69061	Idiopathic steroid-sensitive nephrotic syndrome	280302	IgG4-related pancreatitis
97560	Idiopathic membranous glomerulonephritis	93209	Idiopathic steroid-sensitive nephrotic syndrome with diffuse mesangial proliferation	449400	IgG4-related periaortitis
		93206	Idiopathic steroid-sensitive nephrotic syndrome with focal segmental glomerulosclerosis	49041	IgG4-related retroperitoneal fibrosis
		93206	Idiopathic steroid-sensitive	447764	IgG4-related sclerosing cholangitis
				449432	IgG4-related sialadenitis
				449432	IgG4-related submandibular gland disease
				64744	IgG4-related thyroid disease
				183675	IgG subclass deficiency with IgA

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	subclass deficiency	86886	Immunoblastic lymphadenopathy		and ear anomalies
329235	IGSF1 deficiency syndrome	34592	Immunodeficiency by defective expression of HLA class 1	2759	Imperforate oropharynx-costovertebral anomalies syndrome
364013	IHF	572	Immunodeficiency by defective expression of HLA class 2	71276	Imploding antrum syndrome
86908	IHHS	169147	Immunodeficiency due to a classical component pathway complement deficiency	35069	INAD
371364	IHPRF syndrome	169150	Immunodeficiency due to a late component of complement deficiency	35069	INAD1
91132	IHS	169147	Immunodeficiency due to an early component of complement deficiency	254509	Inadvertent botulism
59303	IHSC	169147	Immunodeficiency due to C1, C4, or C2 component complement deficiency	45453	Incessant infant ventricular tachycardia
238624	IIH	169150	Immunodeficiency due to C5 to C9 component complement deficiency	79263	INCL
85193	IJO	169100	Immunodeficiency due to CD25 deficiency	231226	Inclusion body beta-thalassemia
238569	IL10-related early-onset IBD	331190	Immunodeficiency due to ficolin3 deficiency	199267	Inclusion body fibromatosis
238569	IL10-related early-onset inflammatory bowel disease	70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	602	Inclusion body myopathy type 2
477661	IL21-related infantile IBD	331187	Immunodeficiency due to MASP-2 deficiency	79091	Inclusion body myopathy type 3
477661	IL21-related infantile inflammatory bowel disease	70593	Immunodeficiency due to selective anti-polysaccharide antibody deficiency	52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
100078	Ileal neuroendocrine neoplasm	200421	Immunodeficiency with factor H anomaly	611	Inclusion body myositis
100078	Ileal neuroendocrine tumor	200418	Immunodeficiency with factor I anomaly	254693	Incomplete hydatidiform mole
238621	Ileal pouch anal anastomosis related faecal incontinence	2268	Immunodeficiency-centromeric instability-facial anomalies syndrome	314466	Incomplete Meigs syndrome
1150	Iillum syndrome	647	Immunodeficiency-microcephaly-chromosomal instability syndrome	254693	Incomplete molar pregnancy
79466	ILVEN	935	Immunodeficiency-short limb dwarfism syndrome	157769	Incomplete situs inversus
85173	IMAGe syndrome	761	Immunoglobulin A vasculitis	180079	Incomplete unilateral aplasia of the Müllerian ducts
247718	IMAM	169110	Immunoglobulin heavy chain deficiency	180079	Incomplete unilateral Müllerian aplasia
42062	Iminoglycinuria	329903	Immunoglobulin-mediated membranoproliferative glomerulonephritis	464	Incontinentia pigmenti
284362	Immature interstitial mesenchymal tumor	329903	Immunoglobulin-mediated MPGN	435	Incontinentia pigmenti type 1
398987	Immature teratoma of ovary	100025	Immunoproliferative small intestinal disease	158019	Indeterminate cell histiocytosis
289465	Immigration delay disease	97567	Immunotactoid glomerulonephritis	158019	Indeterminate Dendritic Cell Tumor
→244	Immotile cilia syndrome, Kartagener type	456312	IMNEPD	1388	Index finger anomaly-Pierre Robin syndrome
2901	Immune brachial plexus neuropathy	206569	IMNM	98848	Indolent systemic mastocytosis
169090	Immune dysfunction due to T-cell inactivation due to calcium entry defect	857	Imperforate anus with hand, foot	1909	Indomethacin embryofetopathy
238569	Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome			70587	Infant acute respiratory distress syndrome
37042	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome			70587	Infant ARDS
364013	Immune fetal edema			178478	Infant botulism
364013	Immune fetal hydrops			1943	Infant epilepsy with migrant focal crisis
364013	Immune HF			178478	Infant intestinal botulism
364013	Immune hydrops fetalis			178478	Infant intestinal toxemia botulism
206569	Immune myopathy with myocyte necrosis			178478	Infant intestinal toxin-mediated botulism
1959	Immune pancytopenia			70587	Infant respiratory distress syndrome
3002	Immune thrombocytopenia			178487	Infant-like botulism
3002	Immune thrombocytopenic purpura			247165	Infantile acrodynia
206569	Immune-mediated necrotizing myopathy			99749	Infantile agranulocytosis
206575	Immune-mediated rippling muscle disease			99725	Infantile and juvenile forms of acromegaly
				70590	Infantile apnea
				51608	Infantile arteriosclerosis
				2679	Infantile axonal neuropathy
				89938	Infantile Bartter syndrome with

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	sensorineural deafness		hyperglycinemia	1145	Infantile-onset X-linked spinal muscular atrophy
178478	Infantile botulism	251304	Infantile onset panniculitis with uveitis and systemic granulomatosis	781	Infection due to Coxiella burnetii
314911	Infantile Canavan disease	1186	Infantile onset spinocerebellar ataxia	279922	Infectious anterior uveitis
137675	Infantile cardiomyopathy with histiocytoid change	67047	Infantile optic atrophy with chorea and spastic paraparesis	137593	Infectious epithelial keratitis
217557	Infantile cellular interstitial pneumonitis	85179	Infantile osteopetrosis with neuroaxonal dysplasia	279925	Infectious panuveitis
313850	Infantile cerebellar-retinal degeneration	247651	Infantile phosphoethanolaminuria	279919	Infectious posterior uveitis
402364	Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	247651	Infantile Rathburn disease	289347	Infective dermatitis associated with HTLV-1
77260	Infantile cerebral Gaucher disease	772	Infantile Refsum disease	289347	Infective dermatitis associated with human T-lymphotropic virus type 1
1313	Infantile choriodocerebral calcification syndrome	254864	Infantile reversible cytochrome C oxidase deficiency myopathy	289347	Infective dermatitis associated with human T-lymphotropic virus type I
31709	Infantile convulsions and choreoathetosis	263410	Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome	99123	Inferior caval vein interruption
1310	Infantile cortical hyperostosis	3451	Infantile spasms	155889	Inferior palpebral coloboma
199267	Infantile digital fibromatosis	3173	Infantile spasms-broad thumbs syndrome	99123	Inferior vena cava interruption
87876	Infantile dysmorphic sialidosis	83330	Infantile spinal muscular atrophy	280794	Infiltrative small vesicular DCM
238455	Infantile dystonia-parkinsonism	255241	Infantile subacute necrotizing encephalopathy with leukodystrophy	280794	Infiltrative small vesicular diffuse cutaneous mastocytosis
364063	Infantile epileptic-dyskinetic encephalopathy	255249	Infantile subacute necrotizing encephalopathy with nephrotic syndrome	85445	Inflammatory amyloidosis
300373	Infantile gigantism due to pituitary hyperplasia	2176	Infantile systemic hyalinosis	79466	Inflammatory linear verrucous epidermal nevus
289860	Infantile glycine encephalopathy	2768	Infantile tibia vara	178342	Inflammatory myofibroblastic tumor
79255	Infantile GM1 gangliosidosis	137675	Infantile xanthomatous cardiomyopathy	160148	Inflammatory myoglandular polyps
309155	Infantile GM2 gangliosidosis 0 variant	293168	Infantile-onset ascending hereditary spastic paralysis	247718	Inflammatory myopathy with abundant macrophages
293603	Infantile hereditary endothelial dystrophy	284332	Infantile-onset autosomal recessive nonprogressive cerebellar ataxia	263553	Inflammatory peeling skin syndrome
352563	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	457205	Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome	48918	Inflammatory pseudotumor of skeletal muscle
247651	Infantile hypophosphatasia	494526	Infantile-onset generalized dyskinesia with orofacial involvement	90003	Inflammatory pseudotumor of the liver
371364	Infantile hypotonia-psychomotor retardation-characteristic facies syndrome	391316	Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression	238305	Infundibulo-neurohypophysitis
79076	Infantile juvenile polyposis syndrome	1451	Infantile-onset multisystem inflammatory disease	95513	Infundibulo-panhypophysitis
206436	Infantile Krabbe disease	494526	Infantile-onset orofacial-trunk-limbs dyskinesia	1849	Infundibulopelvic stenosis-multicystic kidney syndrome
1928	Infantile lobar hyperinflation	500062	Infantile-onset periodic fever-panniculitis-dermatosis syndrome	247257	Inhalation anthrax disease
667	Infantile malignant osteopetrosis	352403	Infantile-onset spinocerebellar ataxia-psychomotor delay syndrome	254504	Inhalation botulism
247165	Infantile mercury intoxication	171714	Infantile-onset symptomatic epilepsy syndrome-developmental stagnation-blindness syndrome	247257	Inhalational anthrax
247165	Infantile mercury poisoning			319465	Inhalational botulism
456312	Infantile multisystem neurologic-endocrine-pancreatic disease			319465	Inherited acute myeloid leukemia
2591	Infantile myofibromatosis			319465	Inherited AML
79263	Infantile NCL			319462	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations
93591	Infantile nephronophthisis			282166	Inherited CJD
35069	Infantile neuroaxonal dystrophy			210141	Inherited congenital spastic quadriplegia
79263	Infantile neuronal ceroid lipofuscinoses			210141	Inherited congenital spastic tetraplegia
289860	Infantile NKH			282166	Inherited Creutzfeldt-Jakob disease
289860	Infantile non-ketotic			859	Inherited deficiency of transcobalamin
				100054	Inherited estrogen-associated

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	angioedema		macrocephaly-cerebellar hypotrophy syndrome		abnormalities syndrome
100054	Inherited estrogen-associated angioneurotic edema	329224	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	468678	Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome
100054	Inherited estrogen-dependent angioedema	3454	Intellectual disability-developmental delay-contractures syndrome	457365	Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome
100054	Inherited estrogen-dependent angioneurotic edema	3044	Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome	3068	Intellectual disability-myopathy-short stature-endocrine defect syndrome
71278	Inherited glutamine synthetase deficiency	→280	Intellectual disability-dysmorphism-intrauterine growth retardation syndrome	352530	Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome
71278	Inherited GS deficiency	171851	Intellectual disability-enteropathy-deafness-peripheral neuropathy-ichthyosis-keratoderma syndrome	397973	Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome
289548	Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency	2139	Intellectual disability-epilepsy-bulbous nose syndrome	3082	Intellectual disability-polydactyly-uncombable hair syndrome
225968	Inherited predisposition to essential thrombocythemia	127	Intellectual disability-epilepsy-endocrine disorders syndrome	369837	Intellectual disability-seizures-hypotonia-ophthalmologic-skeletal anomalies syndrome
37	Inherited zinc deficiency	468620	Intellectual disability-epilepsy-extrapyramidal syndrome	369950	Intellectual disability-seizures-macrocephaly-obesity syndrome
63259	Iniencephaly	435638	Intellectual disability-epilepsy-stereotypic hand movement syndrome	391372	Intellectual disability-severe speech delay-mild dysmorphism syndrome
178475	Inoculation botulism	436151	Intellectual disability-expressive aphasia-facial dysmorphism syndrome	3409	Intellectual disability-short stature-hand contractures-genital anomalies syndrome
411593	Insulin autoimmune syndrome	404440	Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	3074	Intellectual disability-short stature-hypertelorism syndrome
2297	Insulin-resistance syndrome type A	370010	Intellectual disability-facial dysmorphism-hand anomalies syndrome	1240	Intellectual disability-short stature-wedge-shaped epiphyses of knees syndrome
2298	Insulin-resistance syndrome type B	363611	Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome	508498	Intellectual disability-skeletal abnormalities-joint laxity-cardiac anomalies syndrome
97279	Insulinoma	369847	Intellectual disability-hyperkinetic movement-truncal ataxia syndrome	3051	Intellectual disability-sparse hair-brachydactyly syndrome
100973	Intellectual disability associated with fragile site FRAXE	1495	Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome	1891	Intellectual disability-spasticity-ectrodactyly syndrome
464311	Intellectual disability syndrome due to a DYRK1A point mutation	314575	Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome	363528	Intellectual disability-strabismus syndrome
166108	Intellectual disability, Birk-Barel type	166108	Intellectual disability-hypotonia-facial dysmorphism syndrome	397941	Intellectual disability-truncal obesity syndrome
3079	Intellectual disability, Buenos-Aires type	356996	Intellectual disability-hypotonia-spasticity-sleep disorder syndrome	75858	Intellectual disability-truncal obesity-retinal dystrophy-micropenis syndrome
→324737	Intellectual disability, Kahrizi type	3451	Intellectual disability-hypsarrhythmia syndrome	1478	Interatrial communication
2557	Intellectual disability, Mietens-Weber type	436151	Intellectual disability-loss of expressive language-facial dysmorphism syndrome	1478	Interauricular communication
3080	Intellectual disability, Wolff type	457279	Intellectual disability-macrocephaly-hypotonia-behavioral	51890	Intercostal nerve syndrome
289483	Intellectual disability-alacrima-achalasia syndrome			86900	Interdigitating cell sarcoma
2466	Intellectual disability-aphasia-shuffling gait-adducted thumbs syndrome			86900	Interdigitating dendritic cell sarcoma
3041	Intellectual disability-balding-patella luxation-acromicria syndrome			210115	Interleukin-1 receptor antagonist deficiency
364577	Intellectual disability-brachydactyly-Pierre Robin syndrome			169100	Interleukin-2 receptor alpha chain
→324737	Intellectual disability-cataract-coloboma-kyphosis syndrome				
3042	Intellectual disability-cataracts-calcified pinnae-myopathy syndrome				
171860	Intellectual disability-cataracts-kyphosis syndrome				
397709	Intellectual disability-coarse face-macrocephaly-cerebellar hypoplasia syndrome				
397709	Intellectual disability-coarse face-				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	deficiency		granulomatosis	329324	Inverse Klippel-Trénaunay syndrome
171208	Intermediate anorectal malformation	314376	Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency	98951	Inverse Marcus-Gunn phenomenon
268162	Intermediate BCKD deficiency	86880	Intestinal T-cell lymphoma	79409	Inverse RDEB
268162	Intermediate branched-chain alpha-ketoacid dehydrogenase deficiency	178481	Intestinal toxemia botulism	79409	Inverse recessive dystrophic epidermolysis bullosa
411634	Intermediate cystinosis	178481	Intestinal toxin-mediated botulism	96092	Inverted 8p duplication/deletion syndrome
99989	Intermediate DEND syndrome	228371	Intoxication botulism	2704	Inverted smile-neurogenic bladder syndrome
86797	Intermediate lichen myxedematosus	46724	Intracranial arteriovenous malformation	1451	IOMID syndrome
268162	Intermediate maple syrup urine disease	252006	Intracranial endodermal sinus tumor	499096	ION
268162	Intermediate MSUD	252006	Intracranial yolk sac tumor	1186	IOSCA
171433	Intermediate nemaline myopathy	137622	Intractable diarrhea-choanal atresia-eye anomalies syndrome	275766	IPAH
210110	Intermediate osteopetrosis	424058	Intraductal papillary mucinous carcinoma of pancreas	747	iPAP
309331	Intermediate severe Salla disease	424982	Intrahepatic bile duct cystadenocarcinoma	238455	IPD
83418	Intermediate spinal muscular atrophy	69665	Intrahepatic cholestasis of pregnancy	37042	IPEX
279914	Intermediate uveitis	280802	Intralobar congenital bronchopulmonary sequestration	1006	Ipp-Gelfand syndrome
268173	Intermittent BCKD deficiency	280802	Intralobar congenital pulmonary sequestration	494428	IPPFE
268173	Intermittent branched-chain alpha-ketoacid dehydrogenase deficiency	99088	Intramural coronary arterial course	88621	IPS
90283	Intermittent cutaneous lupus	100003	Intraneuronal perineurioma	100025	IPSID
329967	Intermittent hydrarthrosis	268139	Intraocular medulloepithelioma	397933	IQSEC2-related syndromic intellectual disability
268173	Intermittent maple syrup urine disease	140436	Intraosseous hemangioma	70592	IRAK4 deficiency
268173	Intermittent MSUD	137686	Intrauterine adhesions	772	IRD
→2686	Intermittent neutropenia	436144	Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	209981	IRIDA syndrome
981	Internal carotid agenesis	85173	Intrauterine growth retardation-metaphyseal dysplasia-adrenal hypoplasia congenita-genital anomalies syndrome	64734	Iridocorneal endothelial syndrome
37202	Interstitial cystitis	137686	Intrauterine synechiae	→782	Iris dysplasia-hypertelorism-deafness syndrome
37202	Interstitial cystitis/bladder pain syndrome	98839	Intravascular large B-cell lymphoma	39044	Iris melanoma
37202	Interstitial cystitis/painful bladder syndrome	98839	Intravascular lymphomatosis	209981	Iron-refractory iron deficiency anemia
79099	Interstitial granulomatous dermatitis with arthritis	332	Intrinsic factor deficiency	43115	Iron-sulfur cluster deficiency myopathy
440427	Interstitial lung and liver disease	324648	iNTS disease	86915	Irons-Bhan syndrome
440402	Interstitial lung disease due to ABCA3 deficiency	457088	Invasive candidiasis-deep dermatophytosis syndrome	86915	Irons-Bianchi syndrome
	Interstitial lung disease due to ATP-binding cassette subfamily A member 3 deficiency	90078	Invasive infections due to vancomycin-resistant enterococci	209943	IRVAN syndrome
440402	Interstitial lung disease due to ATP-binding cassette subfamily A member 3 deficiency	90078	Invasive infections due to VRE	84142	Isaac syndrome
440392	Interstitial lung disease due to SP-C deficiency	99925	Invasive mole	84142	Isaac-Mertens syndrome
440392	Interstitial lung disease due to surfactant protein C deficiency	324648	Invasive non-typhoidal salmonella disease	85200	Ischio-spinal dysostosis
99092	Interventricular septum aneurysm	324648	Invasive non-typhoidal salmonellosis	85200	Ischio-vertebral dysplasia
1201	Intestinal atresia type IIb	3306	Invdup(15)	1509	Ischiopatellar dysplasia
178481	Intestinal botulism	96092	Invdupdel(8p)	43115	ISCU myopathy
178481	Intestinal colonization botulism	79405	Inverse JEB	79144	Iso-Kikuchi syndrome
92050	Intestinal epithelial dysplasia			79159	Isobutyric aciduria
30924	Intestinal hypomagnesemia with secondary hypocalcemia			79159	Isobutyryl-CoA dehydrogenase deficiency
3452	Intestinal lipodystrophy			3309	Isochromosome 5p
3452	Intestinal lipophagia			3310	Isochromosome 9p

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
98797	Isochromosome Yp		adermatoglyphia	99177	Isolated distichiasis
98798	Isochromosome Yq	91416	Isolated congenital alacrima	35093	Isolated dolichocephaly
99731	ISOD	180188	Isolated congenital amastia	1885	Isolated ectopia lentis
3306	Isodicentric 15 chromosome	79143	Isolated congenital anonychia	199647	Isolated encephalocele
263524	Isolated acute necrotizing encephalopathy	88620	Isolated congenital anosmia	221106	Isolated facial myokymia
229717	Isolated agammaglobulinemia	162526	Isolated congenital auditory ossicle malformation	65683	Isolated focal cortical dysplasia
440987	Isolated agenesis of gallbladder	180188	Isolated congenital breast hypoplasia/aplasia	268994	Isolated focal cortical dysplasia type 2
268868	Isolated amyelia	238722	Isolated congenital contralateral synkinesia	268961	Isolated focal cortical dysplasia type I
263524	Isolated ANE	217059	Isolated congenital digital clubbing	268973	Isolated focal cortical dysplasia type Ia
1048	Isolated anencephaly/exencephaly	99171	Isolated congenital ectropion	268980	Isolated focal cortical dysplasia type Ib
140989	Isolated angiitis of the central nervous system	295032	Isolated congenital elbow dislocation	268987	Isolated focal cortical dysplasia type Ic
250923	Isolated aniridia	432	Isolated congenital gonadotropin deficiency	268994	Isolated focal cortical dysplasia type II
91397	Isolated ankyloblepharon filiforme adnatum	485426	Isolated congenital hepatic fibrosis	269001	Isolated focal cortical dysplasia type IIa
79143	Isolated anonychia	141152	Isolated congenital hypoglossia/aglossia	269008	Isolated focal cortical dysplasia type IIb
557	Isolated anorectal malformation	141214	Isolated congenital maxillomandibular fusion	448264	Isolated focal non-epidermolytic palmoplantar keratoderma
3387	Isolated anterior cervical hypertrichosis	91489	Isolated congenital megalocornea	52901	Isolated follicle stimulating hormone deficiency
162516	Isolated apertura pyriformis stenosis	199642	Isolated congenital microcephaly	52901	Isolated FSH deficiency
268936	Isolated arhinencephaly	238722	Isolated congenital mirror movements	468666	Isolated generalized anhidrosis with normal sweat glands
1134	Isolated arrhinia	217059	Isolated congenital nail clubbing	87884	Isolated genetic deafness
1166	Isolated asymmetric crying facies	162516	Isolated congenital nasal pyriform aperture stenosis	408	Isolated glycerol kinase deficiency
206599	Isolated asymptomatic elevation of creatine phosphokinase	79144	Isolated congenital onychodysplasia	231662	Isolated growth hormone deficiency type IA
206599	Isolated asymptomatic hyperCKemia	295032	Isolated congenital radial head dislocation	231671	Isolated growth hormone deficiency type IB
254913	Isolated ATP synthase deficiency	91490	Isolated congenital sclerocornea	231679	Isolated growth hormone deficiency type II
30391	Isolated atresia of bile ducts	141214	Isolated congenital syngnathia	231692	Isolated growth hormone deficiency type III
34528	Isolated autosomal dominant hypomagnesemia	216718	Isolated congenitally uncorrected transposition of the great arteries	2128	Isolated hemihyperplasia
199326	Isolated autosomal dominant hypomagnesemia, Glaudemans type	216718	Isolated congenitally uncorrected transposition of the great vessels	2128	Isolated hemihypertrophy
269221	Isolated bilateral hemispheric cerebellar hypoplasia	1460	Isolated CoQ-cytochrome C reductase deficiency	306527	Isolated hereditary congenital facial paralysis
30391	Isolated biliary atresia	254905	Isolated COX deficiency	229717	Isolated hypogammaglobulinemia
158778	Isolated bone marrow mastocytosis	91396	Isolated cryptophthalmia	183675	Isolated IgG subclass deficiency
35099	Isolated brachycephaly	254905	Isolated cytochrome C oxidase deficiency	2345	Isolated Klippel-Feil syndrome
1398	Isolated cerebellar agenesis	217	Isolated Dandy-Walker malformation	1084	Isolated lissencephaly type 1 without known genetic defects
269203	Isolated cerebellar vermis agenesis	269212	Isolated Dandy-Walker malformation with hydrocephalus	268920	Isolated macrencephaly
199630	Isolated cerebellar vermis hypoplasia	269215	Isolated Dandy-Walker malformation without hydrocephalus	391474	Isolated median cleft face syndrome
485426	Isolated CHF	248340	Isolated delta-SPD	268920	Isolated megalecephaly
199302	Isolated cleft lip	248340	Isolated delta-storage pool disease	238593	Isolated mesenteric lipodystrophy
141242	Isolated cleft of the ala nasi	248340	Isolated dense-SPD	95707	Isolated micropenis
2343	Isolated cloverleaf skull syndrome	248340	Isolated dense-storage pool disease	90641	Isolated mitochondrial
1460	Isolated coenzyme Q-cytochrome C reductase deficiency				
141242	Isolated coloboma of the nose				
2609	Isolated complex I deficiency				
1460	Isolated complex III deficiency				
217059	Isolated congenital acropachy				
289465	Isolated congenital				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	neurosensory deafness		hyperostosis	3236	Jackson-Barr syndrome
2609	Isolated mitochondrial respiratory chain complex I deficiency	3208	Isolated succinate-coenzyme Q reductase deficiency	1540	Jackson-Weiss syndrome
3208	Isolated mitochondrial respiratory chain complex II deficiency	3208	Isolated succinate-CoQ reductase deficiency	2848	Jacobs syndrome
1460	Isolated mitochondrial respiratory chain complex III deficiency	3208	Isolated succinate-ubiquinone reductase deficiency	2308	Jacobsen syndrome
254905	Isolated mitochondrial respiratory chain complex IV deficiency	99731	Isolated sulfite oxidase deficiency	1941	JAE
254913	Isolated mitochondrial respiratory chain complex V deficiency	90674	Isolated thyroid-stimulating hormone deficiency	→636	Jaffe-Campanacci syndrome
90641	Isolated mitochondrial sensorineural deafness	238670	Isolated thyroliberin deficiency	93277	Jaffe-Lichtenstein disease
2609	Isolated NADH-coenzyme Q reductase deficiency	90674	Isolated thyrotropin deficiency	2269	Jagell-Holmgren-Hofer syndrome
2609	Isolated NADH-CoQ reductase deficiency	238670	Isolated thyrotropin-releasing factor deficiency	1873	Jalili syndrome
2609	Isolated NADH-ubiquinone reductase deficiency	238670	Isolated thyrotropin-releasing hormone deficiency	300605	JALS
162516	Isolated nasal pyriform aperture hypoplasia	269206	Isolated total cerebellar vermis agenesis	73423	Jamaican vomiting sickness
447881	Isolated neck extensor myopathy	454750	Isolated tracheoesophageal fistula	73423	Jamaican vomiting syndrome
480556	Isolated neonatal sclerosing cholangitis	103909	Isolated trehalose intolerance	1891	Jancar syndrome
1134	Isolated nose agenesis	238670	Isolated TRF deficiency	2590	Jankovic-Rivera syndrome
137902	Isolated optic nerve hypoplasia/aplasia	238670	Isolated TRH deficiency	168491	Jansky-Bielschowsky disease
499096	Isolated optic neuritis	3366	Isolated trigonocephaly	79139	Japanese encephalitis
166119	Isolated osteopoikilosis	90674	Isolated TSH deficiency	2311	Jarcho-Levin syndrome
63440	Isolated oxycephaly	238670	Isolated TSH-releasing factor deficiency	474	JATD
269209	Isolated partial cerebellar vermis agenesis	1460	Isolated ubiquinone-cytochrome C reductase deficiency	91412	Jaw-winking syndrome
96269	Isolated partial vaginal agenesis	269218	Isolated unilateral hemispheric cerebellar hypoplasia	313795	Jawad syndrome
718	Isolated Pierre Robin sequence	860	Isolated ventriculoarterial discordance	2315	JBS
718	Isolated Pierre Robin syndrome	96	Isolated vitamin E deficiency	397715	JBTS with JATD
35098	Isolated plagiocephaly	472	Isosporiasis	139431	Jeavons syndrome
2924	Isolated polycystic liver disease	2305	Isotretinoin embryopathy	306504	JEB with respiratory and renal involvement
2456	Isolated polythelia	2305	Isotretinoin syndrome	79402	JEB, generalized intermediate
216452	Isolated postlingual genetic deafness	2306	Isotretinoin-like syndrome	79404	JEB, generalized severe
216445	Isolated prelingual genetic deafness	33	Isovaleric acid CoA dehydrogenase deficiency	79404	JEB-H
238670	Isolated prothyroliberin deficiency	33	Isovaleric acidemia	79405	JEB-I
238670	Isolated protirelin deficiency	309324	ISSD	79406	JEB-lo
264691	Isolated pulmonary capillaritis	→33364	Itin syndrome	79402	JEB-nH gen
34528	Isolated renal magnesium wasting	439254	ITM2B amyloidosis	251393	JEB-nH loc
439	Isolated right ventricular hypoplasia	439254	ITM2B-related amyloidosis	79403	JEB-PA
35093	Isolated scaphocephaly	439254	ITM2B-related cerebral amyloid angiopathy	306504	JEB-RR
178311	Isolated SCCH	435	Ito hypomelanosis	1201	Jejunal atresia
440713	Isolated sedoheptulokinase deficiency	3002	ITP	506307	Jejunal atresia-microcephaly-ocular anomalies syndrome
440713	Isolated SHPK deficiency	457375	ITPA-related encephalopathy	100077	Jejunal neuroendocrine neoplasm
457083	Isolated splenogonadal fusion	279914	IU	100077	Jejunal neuroendocrine tumor
2440	Isolated split hand-split foot malformation	99123	IVC interruption	1201	Jejunoileal atresia
178311	Isolated sternocostoclavicular	294415	Ivemark II syndrome	89840	JEN-nH
		97548	Ivemark syndrome	→52368	Jensen syndrome
		2307	IVIC syndrome	90647	Jervell and Lange-Nielsen syndrome
		281190	IWC	33314	Jessner lymphocytic infiltration of the skin
				33314	Jessner-Kanof lymphocytic infiltration of the skin
				3283	JET
				474	Jeune asphyxiating thoracic dystrophy
				474	Jeune syndrome
				248111	JHD
				2929	JIP
				65684	JMADUE

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
307	JME	79404	Junctional epidermolysis bullosa generalisata gravis	85435	Juvenile idiopathic rheumatoid factor-positive polyarthritis
86834	JMML	79402	Junctional epidermolysis bullosa generalisata mitis	2929	Juvenile intestinal polyposis
324999	JMP syndrome	79405	Junctional epidermolysis bullosa inversa	300605	Juvenile Lou Gehrig disease
289596	JNA	306504	Junctional epidermolysis bullosa with respiratory and renal involvement	65684	Juvenile muscular atrophy of distal upper extremity
79264	JNCL	79402	Junctional epidermolysis bullosa, Disentis type	65684	Juvenile muscular atrophy of the distal upper limb
2314	Job syndrome	79402	Junctional epidermolysis bullosa, generalized intermediate	391497	Juvenile myasthenia gravis
2315	Johanson-Blizzard syndrome	79404	Junctional epidermolysis bullosa, generalized severe	86834	Juvenile myelomonocytic leukemia
2316	Johnson neuroectodermal syndrome	79404	Junctional epidermolysis bullosa, Herlitz type	307	Juvenile myoclonic epilepsy
85320	Johnson syndrome	79404	Junctional epidermolysis bullosa, Herlitz-Pearson type	307	Juvenile myoclonus epilepsy
2316	Johnson-McMillin syndrome	89840	Junctional epidermolysis bullosa, non-Herlitz type	289596	Juvenile nasopharyngeal angiofibroma
1112	Johnson-Munson syndrome	79403	Junctional epidermolysis bullosa-pyloric atresia syndrome	79264	Juvenile NCL
1485	Johnston-Aarons-Schelley syndrome	2321	Jung-Wolff-Back-Stahl syndrome	93592	Juvenile nephronophthisis
324999	Joint contractures-muscular atrophy-microcytic anemia-panniculitis-associated lipodystrophy syndrome	319223	Junin hemorrhagic fever	411634	Juvenile nephropathic cystinosis
2295	Joint instability syndrome	989	Jussieu syndrome	79264	Juvenile neuronal ceroid lipofuscinosis
2027	Jones syndrome	1941	Juvenile absence epilepsy	157719	Juvenile or adult CACH syndrome
475	Joubert syndrome	391497	Juvenile acquired myasthenia	85193	Juvenile osteoporosis
475	Joubert syndrome type A	300605	Juvenile amyotrophic lateral sclerosis	329894	Juvenile overlap myositis
1454	Joubert syndrome with congenital hepatic fibrosis	199260	Juvenile aponeurotic fibromatosis	2801	Juvenile Paget disease
1454	Joubert syndrome with hepatic defect	391497	Juvenile autoimmune myasthenia gravis	2801	Juvenile Paget's disease
397715	Joubert syndrome with JATD	314918	Juvenile Canavan disease	247604	Juvenile PLS
397715	Joubert syndrome with Jeune asphyxiating thoracic dystrophy	247794	Juvenile cataract-microcornea-renal glucosuria syndrome	93568	Juvenile PM
220493	Joubert syndrome with ocular defect	300605	Juvenile Charcot disease	93568	Juvenile polymyositis
2318	Joubert syndrome with oculorenal defect	86834	Juvenile chronic myelomonocytic leukemia	79076	Juvenile polyposis of infancy
2754	Joubert syndrome with oral-facial-digital syndrome	411634	Juvenile cystinosis	2929	Juvenile polyposis syndrome
2754	Joubert syndrome with orofaciodigital defect	93672	Juvenile dermatomyositis	247604	Juvenile primary lateral sclerosis
220497	Joubert syndrome with renal defect	93672	Juvenile DM	85436	Juvenile psoriatic arthritis
220493	Joubert syndrome with retinopathy	228254	Juvenile elastoma without osteopoikilosis	85408	Juvenile rheumatoid factor-negative polyarthritis
2318	Joubert syndrome with Senior-Loken syndrome	2929	Juvenile gastrointestinal polyposis	247854	Juvenile rheumatoid factor-negative polyarthritis with anti-nuclear antibodies
475	Joubert-Boltshauser syndrome	98977	Juvenile glaucoma	247861	Juvenile rheumatoid factor-negative polyarthritis without anti-nuclear antibodies
2801	JPG	79256	Juvenile GM1 gangliosidosis	93399	Juvenile sialidosis type 2
247604	JPLS	309162	Juvenile GM2 gangliosidosis 0 variant	83419	Juvenile spinal muscular atrophy
2929	JPS	79230	Juvenile hemochromatosis	585	Juvenile sulfatidosis, Austin type
2318	JS type B	98954	Juvenile hereditary epithelial dystrophy of Meesmann	26137	Juvenile temporal arteritis
1454	JS-H	248111	Juvenile Huntington chorea	158000	Juvenile xanthogranuloma
220493	JS-O	248111	Juvenile Huntington disease	445062	Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome
2318	JS-OR	2028	Juvenile hyaline fibromatosis	79241	Juvenile-onset multiple carboxylase deficiency
220497	JS-R			1243	Juvenile-onset vitelliform macular dystrophy
26137	JTA			99100	Juxtaposition of the atrial appendages
2319	Juberg-Hayward syndrome			99100	Juxtaposition of the atrial auricles
101039	Juberg-Hellman syndrome			1540	JWS
93972	Juberg-Marsidi syndrome				
3283	Junctional ectopic tachycardia				

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2322	Kabuki make-up syndrome		deafness/Hystrix-like ichthyosis-deafness syndrome		syndrome
2322	Kabuki syndrome	447777	Keratocystic odontogenic tumor	499	Kerion celsi
85146	Kaeser syndrome	494	Keratoderma hereditarium mutilans	415286	Kernicterus
254519	Kagami-Ogata syndrome	79395	Keratoderma hereditarium mutilans with ichthyosis	415286	Kernicterus spectrum disorder
29073	Kahler's disease	34217	Keratoderma with woolly hair type I	3351	Kersey syndrome
→324737	Kahrizi syndrome	65282	Keratoderma with woolly hair type II	293807	Ketamine-induced biliary dilatation
2324	Kaler-Garry-Stern syndrome	420686	Keratoderma with woolly hair type IV	438075	Ketoacidosis due to monocarboxylate transporter-1 deficiency
2325	Kallin syndrome	79395	Keratoderma-ichthyosiform dermatosis-elevated beta-glucuronidase syndrome	1399	Ketoaciduria-intellectual disability-ataxia-deafness syndrome
478	Kallmann syndrome	79501	Keratodermia palmoplantaris papulosa, Buschke-Fischer-Brauer type	2056	Ketohexokinase deficiency
2326	Kallmann syndrome-heart disease syndrome	50943	Keratolytic winter erythema	35	Ketotic hyperglycinemia
99179	Kandori fleck retina	495	Keratosis extremitatum hereditaria progrediens	85202	Keutel syndrome
1836	Kantaputra mesomelic dysplasia	218	Keratosis follicularis	2988	Khalifa-Graham syndrome
79280	Kanzaki disease	2340	Keratosis follicularis spinulosa decalvans	98841	Ki-1 positive anaplastic large cell lymphoma
949	Kaplan-Plauchu-Fitch syndrome	2339	Keratosis follicularis-dwarfism-cerebral atrophy syndrome	477	KID syndrome
→3157	Kaplowitz-Bodurtha syndrome	281201	Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome	477	KID/HID syndrome
33276	Kaposi sarcoma	86919	Keratosis palmaris et plantaris-clinodactyly syndrome	97332	Kienbock disease
2122	Kaposiform hemangioendothelioma	678	Keratosis palmoplantar-periodontopathy syndrome	50918	Kikuchi disease
464329	Kaposiform lymphangiomatosis	79141	Keratosis palmoplantaris nummularis	50918	Kikuchi-Fujimoto disease
183675	Kappa-chain deficiency	50942	Keratosis palmoplantaris striata	482	Kimura disease
2328	Kapur-Toriello syndrome	50942	Keratosis palmoplantaris striata et areata	401996	KIN
1381	Karandikar-Maria-Kamble syndrome	495	Keratosis palmoplantaris transgrediens et progrediens	2908	Kindler syndrome
2329	Karsch-Neugebauer syndrome	87503	Keratosis palmoplantaris transgrediens of Siemens	99741	King-Denborough syndrome
→244	Kartagener syndrome	50942	Keratosis palmoplantaris varians of Wachters	565	Kinky hair disease
401996	Karyomegalic interstitial nephritis	34217	Keratosis palmoplantaris with arrhythmogenic cardiomyopathy	565	Kinky hair syndrome
2330	Kasabach-Merritt syndrome	28378	Keratosis palmoplantaris-corneal dystrophy syndrome	1183	Kinsbourne syndrome
1894	Kasznica-Carlson-Coppededge syndrome	50944	Keratosis palmoplantaris-cystic eyelids-hypodontia-hypotrichosis syndrome	100996	Kjellin syndrome
3360	Katsantonis-Papadakou Lagoyanni syndrome	2198	Keratosis palmoplantaris-esophageal carcinoma syndrome	98673	Kjer optic atrophy
2473	Kaufman-Mckusick syndrome	2342	Keratosis palmoplantaris-periodontopathia-onychogryposis	99978	Klatskin tumor
2331	Kawasaki disease			261494	Kleefstra syndrome
2306	Kawashima syndrome			96147	Kleefstra syndrome due to 9q subtelomeric deletion
2533	Kawashima-Tsuji syndrome			96147	Kleefstra syndrome due to 9q34 microdeletion
2332	KBG syndrome			261652	Kleefstra syndrome due to a point mutation
439218	KCNQ2-NEE			96147	Kleefstra syndrome due to del(9)(q34)
439218	KCNQ2-related epileptic encephalopathy			96147	Kleefstra syndrome due to monosomy 9q34
439218	KCNQ2-related neonatal epileptic encephalopathy			896	Klein-Waardenburg syndrome
96169	KdVS			33543	Kleine-Levin syndrome
480	Kearns-Sayre syndrome			2110	Kleiner-Holmes syndrome
199260	Keasby tumor			399081	KLHL9-related early-onset distal myopathy
2662	Keipert syndrome			281201	KLICK syndrome
79233	Kelley-Seegmiller syndrome			447974	Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome
→313795	Kelly-Kirson-Wyatt syndrome			2345	Klippel-Feil malformation
54028	Kelly-Paterson syndrome			2345	Klippel-Feil sequence
481	Kennedy disease			90308	Klippel-Trénaunay syndrome
64542	Kennedy-Teebi syndrome				
2333	Kenny syndrome				
2333	Kenny-Caffey syndrome				
435628	Keppen-Lubinsky syndrome				
477	Keratitis-ichthyosis-				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2346	Klippel-Trénaunay-Weber syndrome	2908	KS	99843	LAD-II
157823	Klüver-Bucy syndrome	293936	KTCNCT	99844	LAD-III
485	Kniest dysplasia	447777	KTOC	2363	LADD syndrome
1571	Knobloch syndrome	306674	Kufor-Rakeb syndrome	1484	Ladda-Zonana-Ramer syndrome
1571	Knobloch-Layer syndrome	79262	Kufs disease	158687	LAEB
2698	Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome	83419	Kugelberg-Welander disease	501	Lafora disease
		→1487	Kumar-Lewick syndrome	1997	Lagophthalmia-cleft lip and palate syndrome
		2505	Kunze-Riehm syndrome	59135	Laing early-onset distal myopathy
		→794	Kurczynski-Casperson syndrome	275761	LAL deficiency
		454745	Kuru	538	LAM
		1149	Kuskokwim disease	306507	LAMB2-related infantile-onset nephrotic syndrome
		767	Küssmaul-Maier disease	1296	Lambert syndrome
		449432	Küttner tumor	43393	Lambert-Eaton myasthenic syndrome
		2798	Kuzniecyk syndrome	313	Lamellar ichthyosis
2349	Kocher-Debré-Semelaigne syndrome	34217	KWWH type I	90024	LAMM syndrome
679	Köhlmeier-Degos disease	65282	KWWH type II	98818	Landau-Kleffner syndrome
679	Köhlmeier-Degos-Delort-Tricot syndrome	420686	KWWH type IV	354	Landing disease
1946	Kohlschutter-Tonz syndrome	319254	Kyasanur forest disease	269	Landouzy-Dejerine myopathy
3197	Kok disease	319254	Kyasanur hemorrhagic fever	231031	Lane disease
99077	Kommerell diverticulum	79155	Kynureninase deficiency	2632	Langer mesomelic dysplasia
3130	Komuragaeri disease	1801	Kyphomelic dysplasia	502	Langer-Giedion syndrome
2764	König disease	496689	Kyphoscoliosis-lateral tongue atrophy-hereditary spastic paraplegia syndrome	86897	Langerhans cell sarcoma
→1215	Konigsmark-Knox-Hussels syndrome	496689	Kyphoscoliosis-lateral tongue atrophy-HSP syndrome	2368	Laparoschisis
96169	Koolen-De Vries syndrome	496686	Kyphosis-lateral tongue atrophy-myofibrillar myopathy syndrome	→1159	Laplane-Fontaine-Lagardere syndrome
363965	Koolen-De Vries syndrome due to a point mutation	275543	L1 syndrome	2363	LARD syndrome
2892	Kopysc-Barczyk-Krol syndrome	275543	L1CAM syndrome	98838	Large cell lymphoma of the mediastinum
254519	KOS	79314	L-2-HGA	626	Large congenital melanocytic nevus
477831	Kosaki overgrowth syndrome	79314	L-2-hydroxyglutaric aciduria	633	Laron syndrome
2839	Kosenow syndrome	35704	L-Arginine:glycine amidinotransferase deficiency	220465	Laron syndrome with immunodeficiency
99749	Kostmann syndrome	157973	L-CMD	220465	Laron-like syndrome
1129	Kosztolanyi syndrome	156	L-CPT1 deficiency	633	Laron-type dwarfism
99741	Koussef-Nichols syndrome	156	L-CPTI deficiency	2370	Larsen-like osseous dysplasia-short stature syndrome
2351	Kousseff syndrome	440731	L-ferritin deficiency	284139	Larsen-like syndrome, B3GAT3 type
629	Kowarski syndrome	93599	L-glyceric aciduria	2808	Laryngeal abductor paralysis
→2462	Kozlowski-Brown-Hardwick syndrome	216694	L-transposition of the great arteries	2375	Laryngeal abductor paralysis-intellectual disability syndrome
3082	Kozlowski-Krajewska syndrome	83483	La Crosse encephalitis	2407	Laryngeal and ocular granulation tissue in children from the Indian subcontinent syndrome
2204	Kozlowski-Tsuruta syndrome	53696	LAAHD	100083	Laryngeal neuroendocrine tumor
487	Krabbe disease	3473	Laband syndrome	2407	Laryngo-onycho-cutaneous syndrome
206436	Krabbe disease, classic form	2363	Lacrimoauriculodentodigital syndrome	2004	Laryngo-tracheo-esophageal cleft
206436	Krabbe disease, early-onset	2363	Lacrimoauriculoradiodental syndrome	280205	Laryngo-tracheo-esophageal cleft type 0
206443	Krabbe disease, late-onset	284426	Lactate dehydrogenase A deficiency	93938	Laryngo-tracheo-esophageal cleft type 1
1345	Krasnow-Qazi syndrome	284435	Lactate dehydrogenase B deficiency	93939	Laryngo-tracheo-esophageal cleft
709	Krause-Kivlin syndrome	2965	Lactotroph adenoma		
709	Krause-van Schooneveld-Kivlin syndrome	2968	LAD		
284149	Kreiborg-Pakistani syndrome	99844	LAD-1 variant		
89838	KRT14-related autosomal recessive EBS	99842	LAD-I		
89838	KRT14-related autosomal recessive epidermolysis bullosa simplex				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	type 2	98964	Lattice corneal dystrophy type 1	54260	Left ventricular noncompaction
93940	Laryngo-tracheo-esophageal cleft type 3	98964	Lattice corneal dystrophy type I	99095	Left ventricular-to-right atrial communication
93941	Laryngo-tracheo-esophageal cleft type 4	99094	Laubry-Pezzi syndrome	1757	Leg duplication-mirror foot syndrome
2004	Laryngo-tracheo-esophageal diastema	2398	Launois-Bensaude lipomatosis	2380	Legg-Calvé-Perthes disease
2372	Laryngocoele	2377	Laurence-Moon syndrome	549	Legionellosis
137935	Laryngotracheal angioma	2378	Laurin-Sandrow syndrome	549	Legionnaires disease
2004	Laryngotracheoesophageal cleft	79086	Lawrence syndrome	137605	Legius syndrome
280205	Laryngotracheoesophageal cleft type 0	79086	Lawrence-Seip syndrome	2789	Lehman syndrome
93938	Laryngotracheoesophageal cleft type 1	137898	LBSL	1647	Leichtman-Wood-Rohn syndrome
93939	Laryngotracheoesophageal cleft type 2	2369	LBWC syndrome	255241	Leigh disease with leukodystrophy
93940	Laryngotracheoesophageal cleft type 3	2004	LC	70474	Leigh disease with myopathy
93941	Laryngotracheoesophageal cleft type 4	99900	LCAD	255249	Leigh disease with nephrotic syndrome
1202	Larynx atresia	650	LCAT deficiency	3008	Leigh necrotizing encephalopathy due to pyruvate carboxylase deficiency
99824	Lassa fever	1486	LCCS1	3008	Leigh syndrome due to PC deficiency
99824	Lassa hemorrhagic fever	137776	LCCS2	3008	Leigh syndrome due to pyruvate carboxylase deficiency
98974	Late hereditary endothelial dystrophy	137783	LCCS3	70474	Leigh syndrome with cardiomyopathy
157716	Late infantile CACH syndrome	363409	LCCS5	255241	Leigh syndrome with leukodystrophy
168491	Late infantile NCL	98964	LCD1	255249	Leigh syndrome with nephrotic syndrome
168491	Late infantile neuronal ceroid lipofuscinosis	93558	LCDD	70472	Leigh syndrome, French-Canadian type
79256	Late-infantile GM1 gangliosidosis	98964	LCDI	70472	Leigh syndrome, Saguenay-Lac-Saint-Jean type
206443	Late-infantile/juvenile Krabbe disease	5	LCHAD deficiency	485421	Leigh-like basal ganglia disease-optic atrophy-peripheral neuropathy syndrome
98816	Late-onset benign childhood occipital epilepsy	5	LCHADD	314	Leiner disease
247573	Late-onset citrullinemia type 1	52416	LCM	71274	Leiomyomatosis peritonealis disseminate
247573	Late-onset citrullinemia type I	626	LCMN	64720	Leiomyosarcoma
399058	Late-onset distal crystallinopathy	363618	LCPS	104076	Leiomyosarcoma of small intestine
98912	Late-onset distal myopathy, Marquesberry-Griggs type	65285	LDD	213807	Leiomyosarcoma of the cervix uteri
228227	Late-onset focal dermal elastosis	2364	LDH deficiency	213625	Leiomyosarcoma of the corpus uteri
163708	Late-onset infantile spasms	284435	LDH-H subunit deficiency	507	Leishmaniasis
199299	Late-onset isolated ACTH deficiency	284426	LDH-M subunit deficiency	140936	Lelis syndrome
79406	Late-onset junctional epidermolysis bullosa	2616	Le Merrer syndrome	137839	Lemierre postanginal sepsis
231556	Late-onset localized junctional epidermolysis bullosa-intellectual disability syndrome	330015	Lead intoxication	137839	Lemierre syndrome
79241	Late-onset multiple carboxylase deficiency	330015	Lead poisoning	497906	Lenk-Ploski syndrome
93589	Late-onset nephronophthisis	3246	Learman syndrome	2382	Lennox-Gastaut syndrome
90186	Late-onset primary lymphedema	65	Leber congenital amaurosis	209959	Lens-induced endophthalmitis
67042	Late-onset retinal degeneration	104	Leber hereditary optic neuropathy	209959	Lens-induced iridocyclitis
2789	Lateral meningocele syndrome	190	Leber miliary aneurysm	209959	Lens-induced uveitis
141136	Laterofacial microsomia	104	Leber optic atrophy	568	Lenz microphthalmia
46059	Lathosterolosis	99718	Leber plus disease	2658	Lenz-Majewski hyperostotic dwarfism
		98955	LECD	500	LEOPARD syndrome
		650	Lecithin-cholesterol acyltransferase deficiency		
		199251	Ledderhose disease		
		71273	Left renal vein entrapment syndrome		
		99111	Left superior caval vein persisting to left-sided atrium		
		99109	Left superior caval vein persisting to the left-sided atrium		
		99111	Left superior vena cava persisting to left-sided atrium		
		99111	Left SVC persisting to left-sided atrium		
		54260	Left ventricular hypertrabeculation		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
330032	Lepore-beta-thalassemia syndrome	435845	Lethal neonatal rigidity-multifocal seizure syndrome		vanishing white matter
508	Leprechaunism	435845	Lethal neonatal spasticity-epileptic encephalopathy syndrome	163684	Leukoencephalopathy-dystonia-motor neuropathy syndrome
548	Leprosy	300313	Lethal neurodegenerative disorder due to copper transport defect	83629	Leukoencephalopathy-metaphyseal chondrodysplasia syndrome
252031	Leptomeningeal melanomatosis	293925	Lethal occipital encephalocele-skeletal dysplasia syndrome	2386	Leukoencephalopathy-palmoplantar keratoderma syndrome
268838	Leptomelolipoma	2736	Lethal omphalocele-cleft palate syndrome	314051	Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome
509	Leptospirosis	216804	Lethal osteogenesis imperfecta	1816	Leukomelanoderma-infantilism-intellectual disability-hypodontia-hypotrichosis syndrome
2900	Leri pleonosteosis	1832	Lethal osteosclerotic bone dysplasia	2387	Leukonychia totalis
240	Léri-Weill dyschondrosteosis	210144	Lethal polymalformative syndrome, Boissel type	210133	Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome
240	Léri-Weill syndrome	1234	Lethal popliteal pterygium syndrome	2045	Leukonychia totalis-trichilemmal cysts-ciliary dystrophy syndrome
510	Lesch-Nyhan syndrome	1423	Lethal recessive chondrodysplasia	79507	Leukotriene C4 synthase deficiency
158687	Lethal acantholytic epidermolysis bullosa	1662	Lethal restrictive dermopathy	2743	Levic-Stefanovic-Nikolic syndrome
314718	Lethal arteriopathy syndrome due to fibulin-4 deficiency	→56304	Lethal short-limb dwarfism, McAlister-Crane type	2388	Levine-Critchley syndrome
53696	Lethal arthrogryposis-anterior horn cell disease syndrome	464366	Lethal skeletal dysplasia-fetal akinesia-contractures-thoracic dysplasia-pulmonary hypoplasia syndrome	216694	Levo-transposition of the great arteries
1187	Lethal ataxia with deafness and optic atrophy	79022	Lethal variant of Simpson-Golabi-Behmel syndrome	95854	Levocardia
1486	Lethal congenital contracture syndrome type 1	99870	Letterer-Siwe disease	95854	Levocardia-situs inversus
137776	Lethal congenital contracture syndrome type 2	58017	Leukemic reticuloendotheliosis	2363	Levy-Hollister syndrome
137783	Lethal congenital contracture syndrome type 3	300878	Leukemic reticuloendotheliosis variant	302	Lewandowsky-Lutz syndrome
363409	Lethal congenital contracture syndrome type 5	2968	Leukocyte adhesion deficiency	→1896	Lewis-Pashayan syndrome
330050	Lethal encephalopathy due to mitochondrial and peroxisomal fission defect	99842	Leukocyte adhesion deficiency type I	48162	Lewis-Sumner syndrome
1972	Lethal faciocardiomyelic dysplasia	99843	Leukocyte adhesion deficiency type II	755	Leydig cell hypoplasia
444069	Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome	99844	Leukocyte adhesion deficiency type III	96265	Leydig cell hypoplasia due to complete LH receptor inactivation
439897	Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome	99844	Leukocyte adhesion deficiency-1 variant	96265	Leydig cell hypoplasia due to complete LH resistance
1046	Lethal hemolytic anemia-genital anomalies syndrome	439224	Leukocyte chemotactic factor-2 amyloidosis	96265	Leydig cell hypoplasia due to complete luteinizing hormone receptor inactivation
480528	Lethal hydranencephaly-diaphragmatic hernia syndrome	502444	Leukodystrophy due to alkaline ceramidase 3 deficiency	96265	Leydig cell hypoplasia due to complete luteinizing hormone resistance
1237	Lethal hydrocephalus-cardiac malformation-dense bones syndrome	77295	Leukodystrophy with oligodontia	325448	Leydig cell hypoplasia due to LHB deficiency
254857	Lethal infantile mitochondrial disease	139444	Leukoencephalopathy with bilateral anterior temporal lobe cysts	325448	Leydig cell hypoplasia due to luteinizing hormone subunit beta deficiency
254857	Lethal infantile mitochondrial myopathy	137898	Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	96266	Leydig cell hypoplasia due to partial LH receptor inactivation
2347	Lethal Kniest-like dysplasia	137898	Leukoencephalopathy with brain stem and spinal cord involvement-lactate elevation syndrome	96266	Leydig cell hypoplasia due to partial LH resistance
2371	Lethal Larsen-like syndrome	363540	Leukoencephalopathy with mild cerebellar ataxia and white matter edema	96266	Leydig cell hypoplasia due to partial luteinizing hormone receptor inactivation
478049	Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome	135	Leukoencephalopathy with	96266	Leydig cell hypoplasia due to partial luteinizing hormone resistance
86879	Lethal midline granuloma				
33108	Lethal multiple pterygium syndrome				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
99824	LF	254395	Lichen planus tropicus	329341	Limbic encephalitis with DPP6 antibodies
844	LGL syndrome	254395	Lichenoid melanodermatitis	329341	Limbic encephalitis with DPPX antibodies
266	LGMD1A	2390	Lichtenstein syndrome	163908	Limbic encephalitis with leucine-rich glioma-inactivated 1 antibodies
264	LGMD1B	448251	Lichtenstein-Knorr syndrome	163908	Limbic encephalitis with LGI1 antibodies
265	LGMD1C	526	Liddle syndrome	217253	Limbic encephalitis with N-methyl-D-aspartate receptor antibodies
34516	LGMD1D	1275	Liebenberg syndrome	163914	Limbic encephalitis with nCMAgs antibodies
34517	LGMD1E	99812	LIG4 syndrome	498700	Limbic encephalitis with neurexin-3 antibodies
55595	LGMD1F	99812	Ligase 4 syndrome	217253	Limbic encephalitis with NMDA receptor antibodies
55596	LGMD1G	93557	Light and heavy chain deposition disease	163914	Limbic encephalitis with novel cell membrane antigen antibodies
238755	LGMD1H	93558	Light chain deposition disease	83467	Limbic encephalitis-neuromyotonia-hyperhidrosis-polyneuropathy syndrome
267	LGMD2A	85443	Light-chain amyloidosis	254857	LIMD
268	LGMD2B	97231	Ligneous conjunctivitis	366	Limit dextrinosis
353	LGMD2C	2369	Limb body wall complex	220402	Limited cutaneous systemic scleroderma
62	LGMD2D	2492	Limb transversal defect-cardiac anomaly syndrome	220402	Limited cutaneous systemic sclerosis
119	LGMD2E	974	Limb, scalp and skull defects	220407	Limited systemic sclerosis
219	LGMD2F	62	Limb-girdle muscular dystrophy due to alpha-sarcoglycan deficiency	254857	LIMM
34514	LGMD2G	119	Limb-girdle muscular dystrophy due to beta-sarcoglycan deficiency	168491	LINCL
1878	LGMD2H	267	Limb-girdle muscular dystrophy due to calpain deficiency	892	Lindau disease
34515	LGMD2I	265	Limb-girdle muscular dystrophy due to caveolin-3 deficiency	3077	Lindsay-Burn syndrome
140922	LGMD2J	219	Limb-girdle muscular dystrophy due to delta-sarcoglycan deficiency	79150	Linear and whorled nevoid hypermelanosis
86812	LGMD2K	268	Limb-girdle muscular dystrophy due to dysferlin deficiency	140933	Linear atrophoderma of Moulin
206549	LGMD2L	34515	Limb-girdle muscular dystrophy due to FKRP deficiency	228236	Linear focal dermal elastosis
206554	LGMD2M	353	Limb-girdle muscular dystrophy due to gamma-sarcoglycan deficiency	228236	Linear focal elastosis
206559	LGMD2N	264	Limb-girdle muscular dystrophy due to lamin A/C deficiency	2611	Linear hamartoma syndrome
206564	LGMD2O	266	Limb-girdle muscular dystrophy due to myotilin deficiency	46488	Linear IgA dermatosis
280333	LGMD2P	445110	Limb-girdle muscular dystrophy due to POMK deficiency	254379	Linear lichen planus
254361	LGMD2Q	34514	Limb-girdle muscular dystrophy due to telethonin deficiency	254379	Linear LP
363543	LGMD2R	1878	Limb-girdle muscular dystrophy due to TRIM32 deficiency	2612	Linear nevus sebaceus syndrome
369840	LGMD2S	257	Limb-girdle muscular dystrophy with epidermolysis bullosa simplex	2611	Linear verrucous nevus syndrome
363623	LGMD2T	52430	Limb-girdle muscular dystrophy with Paget disease of bone	36273	Linitis plastica of the stomach
352479	LGMD2U	86812	Limb-girdle muscular dystrophy-intellectual disability syndrome	888	Lip-pit syndrome
466801	LGMD2W	69085	Limb-mammary syndrome	435660	LIPE-related familial partial lipodystrophy
476084	LGMD2X	171673	Limbal stem cell deficiency	435660	LIPE-related FPLD
424261	LGMD2Y	276402	Limbic encephalitis with caspr2 antibodies	77243	Lipedema
480682	LGMD2Z	329341	Limbic encephalitis with dipeptidyl-peptidase 6 antibodies	255182	Lipoamide dehydrogenase deficiency
445110	LGMD due to POMK deficiency			90160	Lipoatrophia semicircularis
93557	LHCDD			528	Lipoatrophic diabetes
65285	Lhermitte-Duclos disease			90157	Lipoatrophy caused by injected drug
104	LHON				
99718	LHON plus disease				
313	LI				
524	Li-Fraumeni syndrome				
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				

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156156	Lipoatrophy with diabetes, leukomelanodermic papules, liver steatosis, and hypertrophic cardiomyopathy	100016	Lissencephaly with cerebellar hypoplasia type F		disability syndrome
247762	Lipoblastoma	533	Listeria infection	90647	Long QT interval-deafness syndrome
90156	Lipodystrophia centrifugalis abdominalis infantilis	533	Listeriosis	37553	Long QT syndrome type 7
1979	Lipodystrophy due to peptidic growth factors deficiency	820	Livedo racemosa-cerebrovascular accident syndrome	65283	Long QT syndrome type 8
50811	Lipodystrophy-intellectual disability-deafness syndrome	820	Livedo reticularis-cerebrovascular accident syndrome	65283	Long QT syndrome-syndactyly syndrome
3163	Lipodystrophy-Rieger anomaly-diabetes syndrome	79095	Liver disease-retinitis pigmentosa-polyneuropathy-epilepsy syndrome	5	Long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency
401859	Lipoic acid synthetase deficiency	369	Liver glycogen phosphorylase deficiency	180157	Longitudinal vaginal septum
139436	Lipoid dermatopathitis	98818	LKS	52054	Longman-Tolmie syndrome
530	Lipoid proteinosis	363618	LMNA-related cardiocutaneous progeria syndrome	168	Loose anagen syndrome
36397	Lipomatosis dolorosa	157973	LMNA-related congenital muscular dystrophy	411602	LOPD
238593	Lipomatous mesenteritis	33108	LMPS	2832	Lopes-Gorlin syndrome
812	Lipomucopolysaccharidosis	69085	LMS	2266	Lopes-Marques de Faria syndrome
268835	Lipomyelomeningocele	93924	Lobar holoprosencephaly	67042	LORD
329481	Lipoprotein glomerulopathy	666	Lobstein disease	79395	Loricrin keratoderma
69078	Liposarcoma	2440	Lobster-claw deformity	803	Lou Gehrig disease
238593	Liposclerotic mesenteritis	2407	LOC syndrome	100	Louis-Bar syndrome
401862	Lipoyl transferase 1 deficiency	314709	Localized AL amyloidosis	171215	Low anorectal malformation
447795	Lipoyl transferase 2 deficiency	93685	Localized Castleman disease	251633	Low grade ependymoma
98955	Lisch epithelial corneal dystrophy	263534	Localized deciduous skin	69663	Low phospholipid associated cholelithiasis
2400	Lisker-Garcia-Ramos syndrome	79400	Localized epidermolysis bullosa simplex	140949	Low-flow priapism
101003	Lison syndrome	90289	Localized fibrosing scleroderma	213736	Low-grade neuroendocrine tumor of the corpus uteri
531	Lissencephaly due to 17p13.3 deletion	251393	Localized junctional epidermolysis bullosa, non-Herlitz type	213736	Low-grade neuroendocrine tumor of the uterine corpus
95232	Lissencephaly due to LIS1 mutation	90398	Localized lichen myxedematosus with mixed features of different subtypes	1652	Low-molecular-weight proteinuria with hypercalciuria and nephrocalcinosis
171680	Lissencephaly due to TUBA1A mutation	90399	Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms	534	Lowe disease
89844	Lissencephaly syndrome, Norman-Roberts type	178517	Localized pagetoid reticulosis	534	Lowe oculo-cerebro-renal syndrome
2148	Lissencephaly type 1 due to doublecortin gene mutation	263534	Localized PSS	534	Lowe syndrome
352682	Lissencephaly type 2 without muscular or eye involvement	163927	Localized pustular psoriasis	2408	Lowe-Kohn-Cohen syndrome
352682	Lissencephaly type 2 without muscular or ocular involvement	90289	Localized scleroderma	83628	Lower body hemangioma-urogenital anomalies-myelopathy-bony deformities-anorectal and arterial malformations-renal anomalies syndrome
86821	Lissencephaly type 3-familial fetal akinesia sequence syndrome	2406	Locked-in syndrome	363447	Lower extremity-predominant autosomal dominant proximal spinal muscular atrophy
86822	Lissencephaly type 3-metacarpal bone dysplasia syndrome	75566	Loeffler endocarditis	363454	Lower extremity-predominant autosomal dominant proximal spinal muscular atrophy with contractures
100011	Lissencephaly with cerebellar hypoplasia type A	60030	Loeys-Dietz syndrome	209341	Lower extremity-predominant autosomal dominant proximal spinal muscular atrophy without contractures
100012	Lissencephaly with cerebellar hypoplasia type B	2407	LOGIC syndrome	295051	Lower limb hypertrophy
100013	Lissencephaly with cerebellar hypoplasia type C	250831	Logopenic primary progressive aphasia	2487	Lower limb malformation-hypospadias syndrome
100014	Lissencephaly with cerebellar hypoplasia type D	250831	Logopenic progressive aphasia		
100015	Lissencephaly with cerebellar hypoplasia type E	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		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
141064	Lower lip fistula	90283	Lupus erythematosus tumidus	144	Lynch syndrome
276435	Lower motor neuron syndrome with late-adult onset	498251	Luteal-phase-dependent febrile episode	1123	Lynch-Lee-Murday syndrome
844	Lown-Ganong-Levine syndrome	498251	Luteal-phase-dependent periodic fever	3196	Lyngstadaas syndrome
1533	Lowry syndrome	1173	Luteinizing hormone-releasing hormone deficiency with ataxia	98842	LyP
2409	Lowry-MacLean syndrome	302	Lutz-Lewandowsky epidermolytic hyperkeratosis verruciformis	2203	Lysine alpha-ketoglutarate reductase deficiency
1824	Lowry-Wood syndrome	→2697	Lutz-Richner-Landolt syndrome	470	Lysinuric protein intolerance
2003	Lowry-Yong syndrome	54260	LVNC	275761	Lysosomal acid lipase deficiency
254478	LP pemphigoides	79150	LWNH	35121	Lysosomal acid phosphatase deficiency
254463	LP pigmentosa	537	Lyell syndrome	61	Lysosomal alpha-D-mannosidase deficiency
254463	LP pigmentosus	86869	LYG	309288	Lysosomal alpha-D-mannosidase deficiency, adult form
250831	LPA	91546	Lyme borreliosis	309282	Lysosomal alpha-D-mannosidase deficiency, infantile form
69663	LPAC	91546	Lyme disease	34587	Lysosomal glycogen storage disease with normal acid maltase activity
71274	LPD	538	Lymphangiomyomatosis	79284	Lysosomal membrane cobalamin transporter deficiency
329481	LPG	2035	Lymphatic filariasis	93561	Lysozyme amyloidosis
470	LPI	→90186	Lymphedema praecox	90020	Lytic-Bodig disease
309015	LPL deficiency	→90186	Lymphedema tarda	330041	M hemoglobinopathy
163927	LPP	662	Lymphedema with yellow nails	247262	Mabry syndrome
525	LPP	86915	Lymphedema-atrial septal defects-facial changes syndrome	98938	MAC
37553	LQT7	86914	Lymphedema-cerebral arteriovenous anomaly syndrome	36412	Mac Duffie hypocomplementemic urticarial vasculitis
65283	LQT8	33001	Lymphedema-distichiasis syndrome	36412	Mac Duffie syndrome
498481	LRP5-related primary osteoporosis	1563	Lymphedema-hypoparathyroidism syndrome syndrome	2220	MacDermot-Patton-Williams syndrome
314051	LTBL	2136	Lymphedema-lymphangiectasia-intellectual disability syndrome	2083	MacDermot-Winter syndrome
79507	LTC4 synthase deficiency	99141	Lymphedema-posterior choanal atresia syndrome	98757	Machado disease
2004	LTEC	→33001	Lymphedema-ptosis syndrome	98757	Machado-Joseph disease
280205	LTEC0	158793	Lymphadenopathic mastocytosis with eosinophilia	276238	Machado-Joseph disease type 1
93938	LTEC1	86870	Lymphoblastoid variant of NK-cell lymphoma	276241	Machado-Joseph disease type 2
93939	LTEC2	314970	Lymphocytic hypereosinophilic syndrome	276244	Machado-Joseph disease type 3
93940	LTEC3	79128	Lymphocytic interstitial pneumonia	319229	Machupo hemorrhagic fever
93941	LTEC4	314970	Lymphocytic variant HES	79495	Macias Flores-Garcia Cruz-Rivera syndrome
93938	LTEC I	289682	Lymphoepithelial-like carcinoma	1574	Mackay-Shek-Carr syndrome
93939	LTEC II	86886	Lymphogranulomatosis X	468672	MACOM syndrome
93940	LTEC III	314970	Lymphoid HES	357158	Macroblepharon-ectropion-hypertelorism-macrostomia syndrome
93941	LTEC IV	79128	Lymphoid interstitial pneumonia	137893	Macrocephalic sperm head syndrome
53351	Lubag	86869	Lymphomatoid granulomatosis	217335	Macrocephaly-alopelia-cutis laxa-scoliosis syndrome
53351	Lubag syndrome	98842	Lymphomatoid papulosis	60040	Macrocephaly-capillary malformation syndrome
2575	Lubani-Al Saleh-Teebi syndrome	329998	Lymphomatous meningitis	60040	Macrocephaly-cutis marmorata telangiectatica congenita syndrome
2410	Lubinsky syndrome	443159	Lymphoplasmacytic lymphoma without IgM production	397612	Macrocephaly-developmental delay syndrome
→1762	Lubs-Arena syndrome	443159	Lymphoplasmacytic lymphoma without Immunoglobulin M production	210548	Macrocephaly-intellectual
2312	Lucey-Driscoll syndrome	280302	Lymphoplasmacytic sclerosing pancreatitis		
776	Lujan syndrome				
776	Lujan-Frys syndrome				
319213	Lujo hemorrhagic fever				
83628	LUMBAR syndrome				
268388	Lumbosacral spina bifida aperta				
268758	Lumbosacral spina bifida cystica				
97332	Lunatomalacia				
2928	Lundberg syndrome				
1120	Lung agenesis-heart defect-thumb anomalies syndrome				
137631	Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome				
90285	Lupus erythematosus panniculitis				
90285	Lupus erythematosus profundus				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	disability-autism syndrome	26791	MAD deficiency	399808	Male infertility with teratozoospermia due to single gene mutation
466791	Macrocephaly-intellectual disability-left ventricular non compaction syndrome	394532	MAD deficiency, mild type	753	Male pseudohermaphroditism due to 5-alpha-reductase 2 deficiency
457485	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	394529	MAD deficiency, severe neonatal type	752	Male pseudohermaphroditism due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
2563	Macrocephaly-obesity-mental disability-ocular abnormalities syndrome	26791	MADD	755	Male pseudohermaphroditism due to LH resistance or LHB deficiency
2427	Macrocephaly-short stature-paraplegia syndrome	394532	MADD, mild type	755	Male pseudohermaphroditism due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency
2429	Macrocephaly-spastic paraplegia-dysmorphism syndrome	394529	MADD, severe neonatal type	1646	Male sterility due to chromosome Y deletion
79489	Macrocystic lymphangioma	35688	Madelung deformity	3000	Male-limited precocious puberty
79489	Macrocystic lymphatic malformation	295223	Madelung deformity, bilateral	99915	Malignant granulosa cell tumor of the ovary
295044	Macrodactyly of fingers	295221	Madelung deformity, unilateral	289385	Malignancy diagnosed during pregnancy
295241	Macrodactyly of fingers, bilateral	2398	Madelung disease	98839	Malignant angioendotheliomatosis
295239	Macrodactyly of fingers, unilateral	137867	Madras motor neuron disease	679	Malignant atrophic papulosis
295047	Macrodactyly of foot	48162	MADSAM	100093	Malignant carcinoid syndrome
295245	Macrodactyly of foot, bilateral	2583	Madura foot	99912	Malignant dysgerminomatous germ cell tumor of the ovary
295243	Macrodactyly of foot, unilateral	1942	MAE	276145	Malignant epithelial tumor of salivary glands
295044	Macrodactyly of hand	199354	Maeda syndrome	213837	Malignant germ cell tumor of the cervix uteri
295241	Macrodactyly of hand, bilateral	163634	Maffucci syndrome	213751	Malignant germ cell tumor of the corpus uteri
295239	Macrodactyly of hand, unilateral	324972	MAGIC syndrome	206489	Malignant germ cell tumor of the vagina
295047	Macrodactyly of toes	438274	Mahvash disease	423	Malignant hyperthermia of anesthesia
295245	Macrodactyly of toes, bilateral	77297	Majeed syndrome	2215	Malignant hyperthermia-arthrogryposis-torticollis syndrome
295243	Macrodactyly of toes, unilateral	2637	Majewski osteodysplastic primordial dwarfism type II	252050	Malignant melanoma of meninges
2477	Macroencephaly	572	Major histocompatibility complex class II expression deficiency	168999	Malignant melanoma of the mucosa
158061	Macrophage activation syndrome	70470	Major hyperlipidemia	293181	Malignant migrating partial epilepsy of infancy
592	Macrophagic myofasciitis	210272	Mal de débarquement	293181	Malignant migrating partial seizures of infancy
2432	Macrosomia-microphthalmia-cleft palate syndrome	87503	Mal de Meleda	213610	Malignant mixed Müllerian tumor of the corpus uteri
2563	Macrosomia-obesity-macrocephaly-ocular abnormalities syndrome	556	Malakoplakia	213512	Malignant mixed Müllerian tumor of the ovary
141276	Macrostomia	420179	Malan overgrowth syndrome	213787	Malignant Müllerian mixed tumor of the cervix uteri
83619	Macrostomia-preauricular tags-external ophthalmoplegia syndrome	673	Malaria	3148	Malignant neurilemmoma
→182050	Macrothrombocytopenia with leukocyte inclusions	75376	Malattia leventinese	3148	Malignant neurofibroma
220448	Macrothrombocytopenia with mitral valve insufficiency	401973	Male EBP disorder with neurological defects	206538	Malignant non-dysgerminomatous germ cell tumor of ovary
487796	Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome	2234	Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome	99912	Malignant ovarian dysgerminoma
217335	MACS syndrome	171709	Male infertility due to globozoospermia		
137814	Macular amyloidosis	137893	Male infertility due to large-headed multiflagellar polyploid spermatozoa		
91494	Macular coloboma-cleft palate-hallux valgus syndrome	137893	Male infertility due to macrozoospermia		
98969	Macular corneal dystrophy	→399808	Male infertility due to NANOS1 mutation		
79457	Maculopapular cutaneous mastocytosis	171709	Male infertility due to round-headed spermatozoa		
2457	MAD	399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation		
		→399805	Male infertility with normal virilization due to maturation arrest		
		→399805	Male infertility with normal virilization due to meiosis defect		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
3286	Malignant paroxysmal ventricular tachycardia	91412	Mandibulo-palpebral synkinesis-ptosis syndrome		syndrome
252128	Malignant perineurioma	2457	Mandibuloacral dysplasia	2463	Marfanoid habitus-autosomal recessive intellectual disability syndrome
3148	Malignant peripheral nerve sheath tumor	90153	Mandibuloacral dysplasia with type A lipodystrophy	→60030	Marfanoid habitus-craniosynostosis syndrome
252128	Malignant peripheral nerve sheath tumor with perineurial differentiation	90154	Mandibuloacral dysplasia with type B lipodystrophy	314041	Marfanoid habitus-inguinal hernia-advanced bone age syndrome
252212	Malignant peripheral nerve sheath tumor with rhabdomyosarcomatous differentiation	443995	Mandibulofacial dysostosis with alopecia	2464	Marfanoid syndrome, De Silva type
213812	Malignant peripheral neuroectodermal tumor of the cervix uteri	245	Mandibulofacial dysostosis with preaxial limb anomalies	→3253	Margarita island ectodermal dysplasia
213630	Malignant peripheral neuroectodermal tumor of the corpus uteri	861	Mandibulofacial dysostosis without limb anomalies	444	Marie Unna congenital hypotrichosis
168811	Malignant peritoneal mesothelioma	79113	Mandibulofacial dysostosis, Guion-Almeida type	444	Marie Unna hereditary hypotrichosis
499182	Malignant pilomatricoma	1131	Mandibulofacial dysostosis, Toriello type	101104	Marin-Amat syndrome
69077	Malignant rhabdoid tumor	357158	Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome	559	Marinesco-Sjögren syndrome
3148	Malignant schwannoma	79113	Mandibulofacial dysostosis-microcephaly syndrome	2717	Marles syndrome
99916	Malignant Sertoli-Leydig cell tumor of the ovary	306682	Manganese intoxication	2717	Marles-Greenberg-Persaud syndrome
398987	Malignant teratoma of ovary	306682	Manganese poisoning	583	Maroteaux-Lamy disease
99868	Malignant thymoma	306682	Manganism	2767	Maroteaux-Le Merrer-Bensahel syndrome
252212	Malignant triton tumor	2717	Manitoba oculotrichoanal syndrome	950	Maroteaux-Malamut syndrome
180242	Malignant tubal tumor	79327	Mannosyltransferase 1 deficiency	1423	Maroteaux-Stanescu-Cousin syndrome
180242	Malignant tumor of fallopian tubes	79326	Mannosyltransferase 2 deficiency	1040	Maroteaux-Verloes-Stanescu syndrome
943	Malonic aciduria	79321	Mannosyltransferase 6 deficiency	560	Marshall syndrome
943	Malonyl-CoA decarboxylase deficiency	79328	Mannosyltransferase 7-9 deficiency	42642	Marshall syndrome with periodic fever
2229	Malouf syndrome	79324	Mannosyltransferase 8 deficiency	561	Marshall-Smith syndrome
99090	Malposition of the coronary ostium	2459	Mansonelliasis	908	Martin-Bell syndrome
→293843	Malpuech facial clefting syndrome	2459	Mansonellosis	85321	Martin-Probst syndrome
→293843	Malpuech syndrome	52416	Mantle cell lymphoma	→293864	Martínez-Frías syndrome
293843	Malpuech-Michels-Mingarelli-Carnevale syndrome	52416	Mantle zone lymphoma	466718	Martinique crinkled retinal pigment epitheliopathy
293208	MALS	98956	Map-dot-fingerprint dystrophy	1387	Martsolf syndrome
52417	MALT lymphoma	511	Maple syrup urine disease	2466	MASA syndrome
103907	Maltase-glucoamylase deficiency	→2712	Marashi-Gorlin syndrome	→284963	MASS syndrome
52417	MALToma	2785	Marble brain disease	66661	Mast cell sarcoma
180275	Mammary Paget disease	228157	Marburg acute multiple sclerosis	101001	Mast syndrome
50920	Mammary polyadenomatosis	99826	Marburg hemorrhagic fever	2135	Mastocytosis-short stature-hearing loss syndrome
238744	Mammary-digital-nail syndrome	99826	Marburg virus disease	3282	MAT
397941	MAN1B1-CDG	500135	MARCH syndrome	168598	MAT deficiency
244310	Man5GlcNAc2-PP-Dol flippase deficiency	221074	Marchiafava-Bignami disease	168598	MAT I/III deficiency
141174	Mandibular arteriovenous malformation	447	Marchiafava-Michelis disease	254534	Maternal 14q32.2 hypermethylation syndrome
363649	Mandibular hypoplasia-deafness-progeroid syndrome	91412	Marcus-Gunn phenomenon	254528	Maternal 14q32.2 microdeletion syndrome
363649	Mandibular hypoplasia-hearing loss-progeroid syndrome	91412	Marcus-Gunn syndrome	275944	Maternal anti-Kell alloimmunization
246	Mandibulofacial dysostosis with postaxial limb anomalies	2461	Marden-Walker syndrome	254528	Maternal del(14)(q32.2)
		2460	Marden-Walker-like syndrome	2209	Maternal hyperphenylalaninemia
		1120	Mardini-Nyhan syndrome		
		558	Marfan syndrome		
		284963	Marfan syndrome type 1		
		284973	Marfan syndrome type 2		
		2462	Marfanoid craniosynostosis		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2216	Maternal hyperthermia induced birth defects	2470	Matthew-Wood syndrome	88950	MCKD2
254528	Maternal monosomy 14q32.2	552	Maturity-onset diabetes of the young	2473	McKusick-Kaufman syndrome
2209	Maternal phenylketonuria	293603	Maumenee corneal dystrophy	52416	MCL
2209	Maternal PKU	141171	Maxillary arteriovenous malformation	59306	McLeod neuroacanthocytosis syndrome
411712	Maternal riboflavin deficiency	1248	Maxillonasal dysostosis	60040	MCM
251009	Maternal uniparental disomy of chromosome 1	1248	Maxillonasal dysplasia	60040	MCMTC
96179	Maternal uniparental disomy of chromosome 2	→182050	May-Hegglin anomaly	77298	MCOPS3
96180	Maternal uniparental disomy of chromosome 4	→182050	May-Hegglin syndrome	85275	MCOPS4
96181	Maternal uniparental disomy of chromosome 6	→182050	May-Hegglin thrombocytopenia	178364	MCOPS5
96183	Maternal uniparental disomy of chromosome 9	3109	Mayer-Rokitansky-Küster-Hauser syndrome	139471	MCOPS6
97678	Maternal uniparental disomy of chromosome 13	247775	Mayer-Rokitansky-Küster-Hauser syndrome type 1	2556	MCOPS7
96184	Maternal uniparental disomy of chromosome 14	2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2	3434	MCOPS8
96185	Maternal uniparental disomy of chromosome 16	57782	Mazabraud syndrome	2470	MCOPS9
96186	Maternal uniparental disomy of chromosome 20	221074	MBD	77299	MCOPS10
96187	Maternal uniparental disomy of chromosome 21	251858	MBEN	2512	MCPH
96188	Maternal uniparental disomy of chromosome 22	91138	MC	2001	McPherson-Clemens syndrome
261519	Maternal uniparental disomy of chromosome X	71529	MC4R deficiency	2999	McPherson-Hall syndrome
96186	Maternal UPD(20)	93554	MC type II	319287	MCRCC
1349	Maternally-inherited cardiomyopathy and deafness	93555	MC type III	466718	MCRPE
1349	Maternally-inherited cardiomyopathy and hearing loss	254519	MCA due to 14q32.2 maternally expressed gene defect	59	MCT8 deficiency
663	Maternally-inherited chronic progressive external ophthalmoplegia	42	MCAD deficiency	809	MCTD
663	Maternally-inherited CPEO	42	MCADD	523	MCUL
225	Maternally-inherited diabetes and deafness	300496	MCAHS type 2	565	MD
255210	Maternally-inherited infantile subacute necrotizing encephalopathy	369837	MCAHS type 3	273	MD1
255210	Maternally-inherited Leigh disease	→56304	McAlister-Crane syndrome	258	MDC1A
255210	Maternally-inherited Leigh syndrome	60040	MCAP	98893	MDC1B
254851	Maternally-inherited mitochondrial dystonia	368	McArdle disease	→370953	MDC1C
663	Maternally-inherited progressive external ophthalmoplegia	79140	MCC	→370953	MDC1D
320360	Maternally-inherited spastic paraparesis	6	MCC deficiency	210272	MdD
320360	Maternally-inherited SPG	85195	McCabe disease	210272	MDDS
2015	Mathieu-De Broca-Bony syndrome	6	MCCD	1836	MDK
600	MATR3-related distal myopathy	562	McCune-Albright syndrome	238744	MDN syndrome
		93686	MCD	363649	MDP syndrome
		98969	MCD	3097	Meacham syndrome
		1851	MCDK	3097	Meacham-Winn-Culler syndrome
		2471	McDonough syndrome	435438	MEAK
		→357225	McDowall syndrome	370997	MEB disease with bilateral multicystic leucodystrophy
		75327	MCDR1	588	MEB syndrome
		319640	MCDR2	98954	MECD
		1035	MCDU	564	Meckel syndrome
		36412	McDuffie hypocomplementemic urticarial vasculitis	3032	Meckel syndrome type 7
		36412	McDuffie syndrome	564	Meckel-Gruber syndrome
		2953	MCEDS	3032	Meckel-like syndrome type 1
		308425	MCEE deficiency	70588	Meconium aspiration syndrome
		158668	McGrath syndrome	314376	Meconium ileus due to guanylate cyclase 2C deficiency
		34149	MCKD	→1762	MECP2 duplication syndrome
		88949	MCKD1	93308	MED1
				93307	MED4
				93311	MED5
				98838	Med-DLBCL
				3453	MEDAC syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2476	Medeira-Dennis-Donnai syndrome	2478	Megalencephalic leukodystrophy	2485	Melorheostosis
57196	Medial condensing osteitis of the clavicle	2478	Megalencephalic leukoencephalopathy with subcortical cysts	1879	Melorheostosis with osteopoikilosis
293208	Median arcuate ligament syndrome	2477	Megalencephaly	93571	Membranoproliferative glomerulonephritis type 2
2006	Median cleft lip/mandible	60040	Megalencephaly-capillary malformation syndrome	652	MEN1
2006	Median cleft lower facial stage	60040	Megalencephaly-capillary malformation-polymicrogyria syndrome	653	MEN2
141239	Median cleft of the upper lip and maxilla	60040	Megalencephaly-cutis marmorata telangiectatica congenita syndrome	247698	MEN2A
1993	Median cleft of the upper lip-corpus callosum lipoma-cutaneous polyps syndrome	2478	Megalencephaly-cystic leukodystrophy syndrome	247709	MEN2B
2699	Median nodule of the upper lip	83473	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	276152	MEN4
98838	Mediastinal diffuse large-cell lymphoma with sclerosis	457359	Megalencephaly-severe kyphoscoliosis-overgrowth syndrome	401973	MEND syndrome
63999	Mediastinal fibrosis	2479	Megalocornea-intellectual disability syndrome	319552	Mendelian susceptibility to interleukin 12 receptor beta 1 deficiency
370127	Medich giant platelet syndrome	238763	Megalocornea-spherophakia-secondary glaucoma syndrome	99898	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency
370127	Medich macrothrombocytopenia	50815	Mégarbané-Loiselet syndrome	319547	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency
231	Medina worm disease	238637	Megaureter-megacystis syndrome	319558	Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency
231	Medinensis	352328	MEGDEL syndrome	319552	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency
231214	Mediterranean anemia	3038	Mehes syndrome	99898	Mendelian susceptibility to mycobacterial diseases due to complete interferon gamma receptor 1 deficiency
100025	Mediterranean lymphoma	85282	MEHMO syndrome	319547	Mendelian susceptibility to mycobacterial diseases due to complete interferon gamma receptor 2 deficiency
→2882	Mediterranean macrothrombocytopenia	2196	Meier-Blumberg-Imahorn syndrome	319558	Mendelian susceptibility to mycobacterial diseases due to complete interleukin 12B deficiency
83313	Mediterranean spotted fever	2554	Meier-Gorlin syndrome	319563	Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency
42	Medium chain acyl-CoA dehydrogenase deficiency	90186	Meige disease	319600	Mendelian susceptibility to mycobacterial diseases due to partial interferon regulatory factor 8 deficiency
42	Medium chain acyl-coenzyme A dehydrogenase deficiency	93964	Meige dystonia	319600	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency
171851	MEDNIK syndrome	90186	Meige lymphedema	319595	Mendelian susceptibility to mycobacterial diseases due to partial signal transducer and activator of transcription 1 deficiency
88949	Medullary cystic kidney disease type 1	93964	Meige syndrome	319595	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency
29073	Medullary plasmacytoma	→90186	Meige-like disease	2494	Ménétrier disease
1309	Medullary sponge kidney	314451	Meigs syndrome	3216	Mengel-Königsmark syndrome
1332	Medullary thyroid carcinoma	98868	Melanesian elliptocytosis		
616	Medulloblastoma	98868	Melanesian ovalocytosis		
251858	Medulloblastoma with extensive nodularity	252206	Melanoma and neural system tumor syndrome		
251883	Medulloepithelioma of the central nervous system	97338	Melanoma of soft tissue		
98954	Meesmann corneal dystrophy	252206	Melanoma-astrocytoma syndrome		
97252	Mega-cisterna magna	404560	Melanoma-pancreatic cancer syndrome		
66629	Megacolon-microcephaly syndrome	79146	Melanosis diffusa congenita		
280671	Megaconial congenital muscular dystrophy	79146	Melanosis universalis hereditaria		
238637	Megacystis-megaureter syndrome	550	MELAS		
2241	Megacystis-microcolon-intestinal hypoperistalsis syndrome	87503	Meleda disease		
2241	Megacystis-microcolon-intestinal hypoperistalsis-hydronephrosis syndrome	2482	Melhem-Fahl syndrome		
2604	Megaduodenum and/or megacystis	31202	Melioidosis		
402023	Megakaryoblastic acute myeloid leukemia with t(1;22)(p13;q13)	2483	Melkersson-Rosenthal syndrome		
402023	Megakaryoblastic AML with t(1;22)(p13;q13)	2484	Melnick-Needles osteodysplasty		
		2484	Melnick-Needles syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
252046	Meningeal melanocytoma		type	622	Methylcobalamin deficiency
2495	Meningioma	2631	Mesomelic dysplasia, Reardon type	308380	Methylcobalamin deficiency type cbIDv1
→823	Meningocele	85170	Mesomelic dysplasia, Savarirayan type	2169	Methylcobalamin deficiency type cbIE
33475	Meningococcal meningitis	1836	Mesomelic dysplasia, Thai type	2170	Methylcobalamin deficiency type cbIG
565	Menkes disease	171690	Metabolic myopathy due to lactate transporter defect	395	Methylene tetrahydrofolate reductase deficiency
565	Menkes syndrome	2499	Metachondromatosis	308425	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
498251	Menstrual cycle-dependent febrile episode	512	Metachromatic leukodystrophy	308425	Methylmalonic acidemia due to methylmalonyl-CoA racemase deficiency
498251	Menstrual cycle-dependent periodic fever	309271	Metachromatic leukodystrophy, adult form	26	Methylmalonic acidemia with homocystinuria
75858	Mental retardation-truncal obesity-retinal dystrophy-micropenis syndrome	309263	Metachromatic leukodystrophy, juvenile form	79284	Methylmalonic acidemia with homocystinuria type cbIF
508093	MEPAN syndrome	309256	Metachromatic leukodystrophy, late infantile form	79282	Methylmalonic acidemia with homocystinuria, type cbIC
330021	Mercurialism	1240	Metaphyseal acroscyphodysplasia	79283	Methylmalonic acidemia with homocystinuria, type cbID
330021	Mercury intoxication	1040	Metaphyseal anadysplasia	369955	Methylmalonic acidemia with homocystinuria, type cbIJ
330021	Mercury poisoning	33067	Metaphyseal chondrodysplasia, Jansen type	369962	Methylmalonic acidemia with homocystinuria, type cbIX
79140	Merkel cell carcinoma	166038	Metaphyseal chondrodysplasia, Kaitila type	280183	Methylmalonic acidemia, TCb1R type
258	Merosin-negative congenital muscular dystrophy	175	Metaphyseal chondrodysplasia, McKusick type	280183	Methylmalonic acidemia, TCbIR type
551	MERRF	174	Metaphyseal chondrodysplasia, Schmid type	308425	Methylmalonic aciduria due to methylmalonyl-CoA epimerase deficiency
54370	Mesangiocapillary glomerulonephritis	2501	Metaphyseal chondrodysplasia, Spahr type	308425	Methylmalonic aciduria due to methylmalonyl-CoA racemase deficiency
386	Mesenchymal hamartoma of liver	99646	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria	280183	Methylmalonic aciduria due to transcobalamin receptor defect
238593	Mesenteric lipogranuloma	2502	Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome	26	Methylmalonic aciduria with homocystinuria
238593	Mesenteric panniculitis	→175	Metaphyseal dysplasia without hypotrichosis	79282	Methylmalonic aciduria with homocystinuria, type cbIC
99701	Mesial temporal lobe epilepsy with hippocampal sclerosis	85188	Metaphyseal dysplasia, Braun-Tischert type	79283	Methylmalonic aciduria with homocystinuria, type cbID
295004	Mesoaxial polydactyly	3005	Metaphyseal dysplasia, Pyle type	369955	Methylmalonic aciduria with homocystinuria, type cbIJ
157801	Mesoaxial synostotic syndactyly with phalangeal reduction	2504	Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome	369962	Methylmalonic aciduria with homocystinuria, type cbIX
95443	Mesocardia	213531	Metaplastic carcinoma of the breast	280183	Methylmalonic aciduria, TCb1R type
289	Mesodermic dysplasia	2635	Metatropic dwarfism	280183	Methylmalonic aciduria due to transcobalamin receptor defect
2496	Mesomelia-synostoses syndrome	2635	Metatropic dysplasia	26	Methylmalonic aciduria with homocystinuria
2496	Mesomelia-synostoses syndrome, Verloes-David-Pfeiffer type	88639	Methacrylic aciduria	79282	Methylmalonic aciduria with homocystinuria, type cbIC
2632	Mesomelic dwarfism, Langer type	31825	Methanol poisoning	79283	Methylmalonic aciduria with homocystinuria, type cbID
2633	Mesomelic dwarfism, Nievergelt type	1923	Methimazole embryofetopathy	79284	Methylmalonic aciduria with homocystinuria, type cbIF
2634	Mesomelic dwarfism, Reinhardt-Pfeiffer type	1923	Methimazole/carbimazole embryofetopathy	369955	Methylmalonic aciduria with homocystinuria, type cbIJ
2631	Mesomelic dwarfism-cleft palate-camptodactyly syndrome	168598	Methionine adenosyltransferase deficiency	369962	Methylmalonic aciduria with homocystinuria, type cbIX
97360	Mesomelic dwarfism-small genitalia syndrome	86904	Methotrexate-associated lymphoproliferative disorders	27	Methylmalonyl-CoA mutase deficiency
85170	Mesomelic dysplasia with absent fibulas and triangular tibias	1917	Methyl mercury antenatal infection	27	Methylmalonyl-Coenzyme A mutase deficiency
2496	Mesomelic dysplasia with acral synostoses, Verloes-David-Pfeiffer type			502430	Metopic ridging-ptosis-facial
1836	Mesomelic dysplasia, Kantaputra type				
2631	Mesomelic dysplasia, Kozlowski-Reardon type				
2633	Mesomelic dysplasia, Nievergelt				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	dysmorphism syndrome	2643	Microcephalic primordial dwarfism, Toriello type	1305	Microcephaly-digital anomalies-normal intelligence syndrome
29	Mevalonic aciduria	329228	Microcephalic primordial dwarfism, Walsh type	391641	Microcephaly-digital anomalies-normal intelligence syndrome type 1
2710	Meyer-Schwickerath syndrome	436182	Microcephalic primordial dwarfism-insulin resistance syndrome	391646	Microcephaly-digital anomalies-normal intelligence syndrome type 2
443995	MFDA	240760	Microcephaly and chromosomal instability without immunodeficiency	217026	Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type
79113	MFDM syndrome	2512	Microcephaly vera	217026	Microcephaly-faciocardioskeletal syndrome
558	MFS	2513	Microcephaly-albinism-digital anomalies syndrome	2172	Microcephaly-glomerulonephritis-marfanoid habitus syndrome
284963	MFS1	3433	Microcephaly-brachydactyly-kyphoscoliosis syndrome	2065	Microcephaly-hiatus hernia-nephrotic syndrome
284973	MFS2	2523	Microcephaly-brain defect-spasticity-hypernatremia syndrome	2558	Microcephaly-hypergonadotropic hypogonadism-short stature syndrome
67046	MGA1	294016	Microcephaly-capillary malformation syndrome	3132	Microcephaly-hypogammaglobulinemia-abnormal immunity syndrome
111	MGA2	2516	Microcephaly-cardiac defect-lung malsegmentation syndrome	647	Microcephaly-immunodeficiency-lymphoreticuloma syndrome
67047	MGA3	2515	Microcephaly-cardiomyopathy syndrome	→3255	Microcephaly-intellectual disability-phalangeal and neurological anomalies syndrome
67048	MGA4	329332	Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome	457351	Microcephaly-intellectual disability-sensorineural deafness-epilepsy-abnormal muscle tone syndrome
66634	MGA5	329332	Microcephaly-cerebellar hypoplasia-congenital heart conduction defect syndrome	457351	Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome
445038	MGA7	434179	Microcephaly-cerebral malformation-orofaciocdigital syndrome	1305	Microcephaly-intellectual disability-tracheoesophageal fistula syndrome
505208	MGA8	2522	Microcephaly-cervical spine fusion anomalies syndrome	391641	Microcephaly-intellectual disability-tracheoesophageal fistula syndrome type 1
505216	MGA9	2521	Microcephaly-cleft palate-abnormal retinal pigmentation syndrome	391646	Microcephaly-intellectual disability-tracheoesophageal fistula syndrome type 2
79329	MGAT2-CDG	423894	Microcephaly-complex motor and sensory axonal neuropathy syndrome	1229	Microcephaly-intracranial calcification-intellectual disability syndrome
→182050	MHA	488168	Microcephaly-congenital cataract-psoriasisiform dermatitis syndrome	2526	Microcephaly-lymphedema-chorioretinopathy syndrome
443162	MHAC	2508	Microcephaly-corpus callosum agenesis-abnormal genitalia syndrome	2528	Microcephaly-microcornea syndrome, Seemanova type
391417	MHBD deficiency	500159	Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrome	3434	Microcephaly-microphthalmia-ectrodactyly of lower limbs-prognathism syndrome
391428	MHBD deficiency, classic type	457284	Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome	1305	Microcephaly-oculo-digit-esophageal-duodenal syndrome syndrome
391428	MHBD deficiency, infantile type	294016	Microcephaly-cutaneous capillary malformation syndrome	391641	Microcephaly-oculo-digit-esophageal-duodenal syndrome syndrome type 1
391457	MHBD deficiency, neonatal type	2533	Microcephaly-deafness-intellectual disability syndrome	171703	Microcephaly-polymicrogyria-corpus callosum agenesis syndrome
572	MHC class II expression deficiency	→313795	Microcephaly-digital anomalies-intellectual disability syndrome		
99826	MHF				
386	MHL				
79651	mHPA				
294016	MIC-CAP syndrome				
294016	MIC-CM syndrome				
→293843	Michels syndrome				
163937	MICPCH				
2510	Micro syndrome				
2511	Microbrachycephaly-ptosis-cleft lip syndrome				
2512	Microcephalia vera				
85172	Microcephalic osteodysplastic dysplasia, Saul-Wilson type				
2637	Microcephalic osteodysplastic primordial dwarfism type II				
2636	Microcephalic osteodysplastic primordial dwarfism types I and III				
2636	Microcephalic osteodysplastic primordial dwarfism, Taybi-Linder type				
468631	Microcephalic primordial dwarfism due to RTTN deficiency				
329228	Microcephalic primordial dwarfism due to ZNF335 deficiency				
319671	Microcephalic primordial dwarfism, Alazami type				
319675	Microcephalic primordial dwarfism, Dauber type				
2617	Microcephalic primordial dwarfism, Montreal type				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2519	Microcephaly-seizures-intellectual disability-heart disease syndrome	2556	Microphthalmia with linear skin defects syndrome		syndrome
423306	Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome	568	Microphthalmia, Lenz type	293181	Migrating partial epilepsy of infancy
397951	Microcephaly-thin corpus callosum-intellectual disability syndrome	85275	Microphthalmia-ankyloblepharon-intellectual disability syndrome	293181	Migrating partial seizures of infancy
2670	Microcoria-congenital nephrosis syndrome	98938	Microphthalmia-anophthalmia-coloboma syndrome	504	Migratory myiasis
2535	Microcornea-corectopia-macular hypoplasia syndrome	77299	Microphthalmia-brain atrophy syndrome	93926	MIH
2536	Microcornea-glucoma-absent frontal sinuses syndrome	424099	Microphthalmia-coloboma-rhizomelic skeletal dysplasia	93926	MIH type HPE
369970	Microcornea-myopic chorioretinal atrophy-telecanthus syndrome	2556	Microphthalmia-dermal aplasia-sclerocornea syndrome	93926	MIHF
231736	Microcornea-posterior megalolenticonus-persistent fetal vasculature-coloboma syndrome	→2510	Microphthalmia-intellectual disability syndrome	93926	MIHV
263347	Microcornea-rod-cone dystrophy-cataract-posterior staphyloma syndrome	2547	Microphthalmia-microtia-fetal akinesia syndrome	2558	Mikati-Najjar-Sahli syndrome
79490	Microcystic infiltrating lymphatic malformation	2705	Microphthalmia-optic nerve aplasia syndrome	79078	Mikulicz disease
79490	Microcystic lymphangioma	251279	Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	314918	Mild Canavan disease
79490	Microcystic lymphatic malformation	727	Micropolyangiitis	169799	Mild factor IX deficiency
83642	Microcytic anemia with liver iron overload	727	Microscopic polyangiitis	169808	Mild factor VIII deficiency
77301	Microdeletion 9q22.3	727	Microscopic polyarteritis	169808	Mild hemophilia A
567	Microdeletion 22q11.2	2551	Microspherophakia-metaphyseal dysplasia syndrome	169799	Mild hemophilia B
90024	Microdontia-type I microtia-deafness syndrome	2552	Microsporidiosis	79651	Mild HPA
101081	Microduplication 17p12	83463	Microtia	79651	Mild hyperphenylalaninemia
217377	Microduplication Xp11.22-p11.23 syndrome	2306	Microtia-aortic arch syndrome	171439	Mild nemaline myopathy
280200	Microform holoprosencephaly	139450	Microtia-eye coloboma-imperforation of the nasolacrimal duct syndrome	216796	Mild osteogenesis imperfecta
280200	Microform HPE	289522	Microtriplication 11q24.1	247815	Mild peroxisomal disorder due to PEX10 deficiency
2538	Microgastria-limb reduction defect syndrome	2290	Microvillous inclusion disease	79253	Mild phenylketonuria
1388	Micrognathia digital syndrome	2290	Microvillus inclusion disease	411536	Mild phosphoribosylpyrophosphate synthetase superactivity
476126	Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	166430	Micturition-induced seizures	79253	Mild PKU
1083	Microlissencephaly	1456	Mid-aortic dysplastic syndrome	411536	Mild PRPP synthetase superactivity
89844	Microlissencephaly type A	1456	Mid-aortic syndrome	411536	Mild PRPS1 superactivity
50810	Microlissencephaly-micromelia syndrome	228299	Mid-dermal elastolysis	93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis
93329	Micromelic dysplasia-dislocation of radius syndrome	1456	Midaortic syndrome	246	Miller syndrome
139471	Microphthalmia with brain and digit anomalies	2556	MIDAS syndrome	531	Miller-Dieker syndrome
98938	Microphthalmia with colobomatous cyst	225	MIDD	98919	Miller-Fisher syndrome
1104	Microphthalmia with facial clefting	1456	Middle aortic syndrome	94091	Mills syndrome
1106	Microphthalmia with limb anomalies	100084	Middle ear neuroendocrine tumor	79452	Milroy disease
		93926	Middle interhemispheric fusion variant	→79452	Milroy-like disease
		93926	Middle interhemispheric variant of holoprosencephaly	255210	MILS
		141288	Midline cervical cleft	1917	Minamata disease
		95443	Midline heart	457485	MINDS syndrome
		93926	Midline interhemispheric variant of holoprosencephaly	506112	MiNEN of pancreas
		2557	Mietens syndrome	757	Mineralocorticoid resistant hyperkalemia
		2867	Mievis-Verellen-Dumoulin	→293843	Mingarelli syndrome
				352734	Minimal pigment oculocutaneous albinism type 1
				98832	Minimally differentiated acute myeloblastic leukemia
				822	Minkowski-Chauffard disease
				1918	Minoxidil antenatal infection
				494433	MIRAGE syndrome
				94125	MIRAS
				→193	Mirhosseini-Holmes-Walton syndrome
				2378	Mirror hands and feet-nasal

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	defects syndrome		episodes		neurosensory deafness with susceptibility to aminoglycoside exposure
3004	Mirror polydactyly-vertebral segmentation-limbs defects syndrome	1933	Mitochondrial encephalomyopathy-aminoacidopathy syndrome	168609	Mitochondrial non-syndromic neurosensory hearing loss with susceptibility to aminoglycoside exposure
498494	Mirror-image polydactyly	508093	Mitochondrial enoyl CoA reductase protein-associated neurodegeneration syndrome	90641	Mitochondrial non-syndromic sensorineural deafness
319308	MTT family translocation renal cell carcinoma	280288	Mitochondrial HSP60 chaperonopathy	168609	Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure
293822	MITF-related melanoma and renal cell carcinoma predisposition syndrome	314637	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	168609	Mitochondrial non-syndromic sensorineural hearing loss with susceptibility to aminoglycoside exposure
134	Mitochondrial acetoacetyl-coenzyme A thiolase deficiency		Mitochondrial isolated neurosensory deafness with susceptibility to aminoglycoside exposure	447784	Mitochondrial pyruvate carrier deficiency
353217	Mitochondrial aspartate-glutamate carrier 1 deficiency	168609	Mitochondrial isolated neurosensory hearing loss with susceptibility to aminoglycoside exposure	254881	Mitochondrial spinocerebellar ataxia with epilepsy
225	Mitochondrial diabetes	168609	Mitochondrial isolated sensorineural deafness with susceptibility to aminoglycoside exposure	746	Mitochondrial trifunctional protein deficiency
352470	Mitochondrial DNA deletion syndrome with limb-girdle weakness		Mitochondrial isolated sensorineural hearing loss with susceptibility to aminoglycoside exposure	1205	Mitral atresia
352470	Mitochondrial DNA deletion syndrome with progressive myopathy	168609	Mitochondrial membrane protein-associated neurodegeneration	3238	Mitral regurgitation-deafness-skeletal anomalies syndrome
1933	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	289560	Mitochondrial myopathy and sideroblastic anemia	99062	Mitral valve agenesis
255235	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy	2598	Mitochondrial myopathy with reversible complex IV deficiency	→284963	Mitral valve-aorta-skeleton-skin syndrome
369897	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	254864	Mitochondrial myopathy with reversible COX deficiency	295012	Mitten hand
279934	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency	254864	Mitochondrial myopathy with reversible cytochrome C oxidase deficiency	90036	Mixed AIHA
363534	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form	550	Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes	809	Mixed connective tissue disease
254875	Mitochondrial DNA depletion syndrome, myopathic form	502423	Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	91138	Mixed cryoglobulinemia
352447	Mitochondrial DNA maintenance syndrome due to MGME1 deficiency	502423	Mitochondrial myopathy-cerebellar atrophy-pigmentary retinopathy syndrome	93555	Mixed cryoglobulinemia type III
1194	Mitochondrial encephalo-cardio-myopathy due to F1Fo ATPase deficiency	2597	Mitochondrial myopathy-lactic acidosis-deafness syndrome	458792	Mixed cystic lymphangioma
1194	Mitochondrial encephalo-cardio-myopathy due to isolated ATP synthase deficiency	2597	Mitochondrial myopathy-lactic acidosis-hearing loss syndrome	458792	Mixed cystic lymphatic malformation
1194	Mitochondrial encephalo-cardio-myopathy due to isolated mitochondrial respiratory chain complex V deficiency	298	Mitochondrial neurogastrointestinal encephalomyopathy	180234	Mixed germ cell tumor
238329	Mitochondrial encephalomyopathy due to combined oxidative phosphorylation defect 6	90641	Mitochondrial non-syndromic neurosensory deafness	252021	Mixed germ cell tumor of central nervous system
238329	Mitochondrial encephalomyopathy due to COXPD6	168609	Mitochondrial non-syndromic	252021	Mixed germ cell tumor of CNS
550	Mitochondrial encephalomyopathy, lactic acidosis and stroke-like			213610	Mixed Müllerian cancer of corpus uteri

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
98757	MJD		brain injury	250999	Monosomy 1q41-q42
565	MK	178145	Moderate multiminicore disease with hand involvement	250999	Monosomy 1q41q42
423461	ML 3 alpha/beta	169796	Moderately severe factor IX deficiency	238769	Monosomy 1q44
423470	ML 3 gamma	169805	Moderately severe factor VIII deficiency	36367	Monosomy 1qter
423461	ML III alpha/beta	169805	Moderately severe hemophilia A	261349	Monosomy 2p15-p16.1
423470	ML III gamma	169796	Moderately severe hemophilia B	261349	Monosomy 2p15p16.1
2598	MILASA	263335	Moderately-differentiated thymic neuroendocrine carcinoma	163693	Monosomy 2p21
2478	MLC	552	MODY	228402	Monosomy 2q23.1
2526	MLCRD	93111	MODY5	1617	Monosomy 2q24
512	MLD	570	Moebius syndrome	251014	Monosomy 2q31.1
309271	MLD, adult form	2560	Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome	251019	Monosomy 2q32
309263	MLD, juvenile form	3198	Moersch-Woltman syndrome	251019	Monosomy 2q32-q33
309256	MLD, late infantile form	2549	Moeschler-Clarren syndrome	251028	Monosomy 2q33.1
59306	MLS	79330	MOGS-CDG	1001	Monosomy 2q37-qter
2556	MLS syndrome	2751	Mohr syndrome	435638	Monosomy 3p25.3
464321	MLT	2753	Mohr-Majewski syndrome	1620	Monosomy 3pter
369970	MMCAT syndrome	52368	Mohr-Tranebaerg syndrome	1621	Monosomy 3q13
598	MmD	99927	Molar pregnancy	356947	Monosomy 3q26-q27
399096	MMD3	1433	Moloney syndrome	356947	Monosomy 3q26q27
497757	MME-related autosomal dominant Charcot Marie Tooth disease type 2	397973	MOMES syndrome	65286	Monosomy 3q29
497757	MME-related autosomal dominant CMT2	2563	MOMO syndrome	65286	Monosomy 3qter
497757	MME-related autosomal dominant hereditary motor and sensory neuropathy type 2	371428	MONA spectrum	238750	Monosomy 4q21
3434	MMEP syndrome	573	Monilethrix	96145	Monosomy 4qter
592	MMF	573	Moniliform hair syndrome	281	Monosomy 5p
268249	MMF embryopathy	319254	Monkey disease	228384	Monosomy 5q14.3
1923	MMI/CMZ embryofetopathy	319254	Monkey fever	314655	Monosomy 5q31.3
1923	MMI/CMZ embryopathy	3057	Monoamine oxidase A deficiency	1627	Monosomy 5q35
2241	MMIHS	59	Monocarboxylate transporter 8 deficiency	251046	Monosomy 6p22
213512	MMMT of the ovary	228423	Monocyte-B-natural killer-dendritic cell deficiency syndrome	96125	Monosomy 6p25
641	MMN	228423	Monocytopenia and mycobacterial infection syndrome	171829	Monosomy 6q16
641	MMNCB	228423	Monocytopenia with susceptibility to infections	251056	Monosomy 6q25
137867	MMND	99885	Monogenic diabetes of infancy	96126	Monosomy 7pter
293181	MMPEI	228423	MonoMAC	904	Monosomy 7q11.23
293181	MMPSI	65684	Monomelic amyotrophy	251061	Monosomy 7q31
2479	MMR syndrome	86870	Monomorphic NK-cell lymphoma	1636	Monosomy 7qter
1305	MMT	2565	Mononen-Karnes-Senac syndrome	251066	Monosomy 8p11.2
391641	MMT type 1	2901	Mononeuritis multiplex with brachial predilection	251071	Monosomy 8p23.1
391646	MMT type 2	293948	Monosomy 1p21.3	2496	Monosomy 8q13
298	MNGIE	401986	Monosomy 1p31p32	284160	Monosomy 8q21.11
565	MNK	456298	Monosomy 1p35.2	178303	Monosomy 8q22.1
251656	MOA	1606	Monosomy 1p36	502	Monosomy 8q24.1
77299	MOBA syndrome	1606	Monosomy 1pter	508488	Monosomy 8q24.3
570	Möbius syndrome	250989	Monosomy 1q21.1	261112	Monosomy 9p
99732	MOCOD			324313	Monosomy 9p13
308386	MOCOD type A			1642	Monosomy 9pter
308393	MOCOD type B			77301	Monosomy 9q22.3
308400	MOCOD type C			401923	Monosomy 9q31.1q31.3
1305	MODED syndrome			495818	Monosomy 9q33.3-q34.11
391641	MODED syndrome type 1			495818	Monosomy 9q33.3q34.11
90056	Moderate and severe traumatic			284169	Monosomy 10p11.2p12.31
				1580	Monosomy 10pter
				276413	Monosomy 10q22.3q23.3

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
96148	Monosomy 10qter	567	Monosomy 22q11	1711	Mosaic trisomy chromosome 17
893	Monosomy 11p13	48652	Monosomy 22q13	1724	Mosaic trisomy chromosome 20
444002	Monosomy 11q22.2-q22.3	99226	Monosomy X	96068	Mosaic trisomy chromosome 22
444002	Monosomy 11q22.2q22.3	93277	Monostotic fibrous dysplasia	1052	Mosaic variegated aneuploidy syndrome
2308	Monosomy 11qter	158003	Montgomery syndrome	54057	Moschcowitz disease
313884	Monosomy 12p12.1	→969	Moore-Federman syndrome	2717	MOTA syndrome
94063	Monosomy 12q14	2637	MOPD type II	294	Mother-to-child transmission of cytomegalovirus syndrome
289513	Monosomy 12q15q21.1	2636	MOPD types I and III	292	Mother-to-child transmission of enterovirus infection
96149	Monosomy 12qter	141327	Moran-Barroso syndrome	70596	Mother-to-child transmission of Epstein-Barr virus infection
412035	Monosomy 13q12.3	52056	Morava-Mehes syndrome	293	Mother-to-child transmission of herpes simplex virus infection
1587	Monosomy 13q14	77296	Morgagni-Stewart-Morel syndrome	295	Mother-to-child transmission of parvovirus syndrome
1590	Monosomy 13q32	75858	MORM syndrome	290	Mother-to-child transmission of rubella syndrome
96168	Monosomy 13q34	35737	Morning glory syndrome	499009	Mother-to-child transmission of syphilis
261120	Monosomy 14q11.2	582	Morquio disease	858	Mother-to-child transmission of toxoplasmosis
261144	Monosomy 14q12	309297	Morquio disease type A	291	Mother-to-child transmission of varicella syndrome
→3157	Monosomy 14q22	309310	Morquio disease type B	254516	Motor developmental delay due to 14q32.2 paternally expressed gene defect
264200	Monosomy 14q22-q23	2570	Morse-Rawnsley-Sargent syndrome	3347	Mounier-Kühn syndrome
264200	Monosomy 14q22q23	83467	Morvan syndrome	83595	Mountain fever
401935	Monosomy 14q24.1q24.3	83467	Morvan's fibrillary chorea	83595	Mountain tick fever
261183	Monosomy 15q11.2	329813	Mosaic genome-wide paternal uniparental disomy	2572	Mousa-Al Din-Al Nassar syndrome
199318	Monosomy 15q13.3	329813	Mosaic genome-wide paternal UPD	324972	Mouth and genital ulcers with inflamed cartilage
261190	Monosomy 15q14	99228	Mosaic monosomy X	2152	Mowat-Wilson syndrome
94065	Monosomy 15q24	96193	Mosaic paternal uniparental disomy of chromosome 11	261537	Mowat-Wilson syndrome due to 2q22 microdeletion
1596	Monosomy 15q26	1692	Mosaic trisomy 1	261552	Mowat-Wilson syndrome due to a ZEB2 point mutation
261211	Monosomy 16p11.2-p12.2	1723	Mosaic trisomy 2	261537	Mowat-Wilson syndrome due to del(2)q(22)
261211	Monosomy 16p11.2p12.2	100071	Mosaic trisomy 3	261537	Mowat-Wilson syndrome due to monosomy 2q22
261236	Monosomy 16p13.11	96059	Mosaic trisomy 4	280679	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome
500055	Monosomy 16p13.2	96060	Mosaic trisomy 5	2573	Moyamoya disease
352629	Monosomy 16q24.1	1747	Mosaic trisomy 7	401945	Moyamoya disease with early-onset achalasia
261250	Monosomy 16q24.3	96061	Mosaic trisomy 8	280679	Moyamoya disease-short stature-facial dysmorphism-hypergonadotropic hypogonadism
531	Monosomy 17p13.3	99776	Mosaic trisomy 9	2574	Moynahan syndrome
97685	Monosomy 17q11	96063	Mosaic trisomy 10	352734	MP OCA type 1
261265	Monosomy 17q12	1698	Mosaic trisomy 12	727	MPA
363958	Monosomy 17q21.31	1703	Mosaic trisomy 14		
261279	Monosomy 17q23.1-q23.2	1706	Mosaic trisomy 15		
261279	Monosomy 17q23.1q23.2	1708	Mosaic trisomy 16		
1597	Monosomy 17qter	1711	Mosaic trisomy 17		
1598	Monosomy 18p	1724	Mosaic trisomy 20		
1600	Monosomy 18q	96068	Mosaic trisomy 22		
254346	Monosomy 19p13.12	1692	Mosaic trisomy chromosome 1		
357001	Monosomy 19p13.13	1723	Mosaic trisomy chromosome 2		
217346	Monosomy 19q13.11	100071	Mosaic trisomy chromosome 3		
261295	Monosomy 20p12.3	96059	Mosaic trisomy chromosome 4		
313781	Monosomy 20p13	96060	Mosaic trisomy chromosome 5		
444051	Monosomy 20q11	1747	Mosaic trisomy chromosome 7		
261311	Monosomy 20q13.33	96061	Mosaic trisomy chromosome 8		
96152	Monosomy 20qter	99776	Mosaic trisomy chromosome 9		
574	Monosomy 21	96063	Mosaic trisomy chromosome 10		
261323	Monosomy 21q22.11-q22.12	1698	Mosaic trisomy chromosome 12		
261323	Monosomy 21q22.11q22.12	1703	Mosaic trisomy chromosome 14		
268261	Monosomy 21q22.13-q22.2	1706	Mosaic trisomy chromosome 15		
268261	Monosomy 21q22.13q22.2	1708	Mosaic trisomy chromosome 16		
96123	Monosomy 22				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
289560	MPAN	276223	MPSVI, slowly progressing		deficiency
59135	MPD1	584	MPSVII	504530	MSN-related combined immunodeficiency
399086	MPD3	99967	MRCLS	157801	MSSD
79323	MPDU1-CDG	263347	MRCS syndrome	65748	MSSE
293181	MPEI	67045	MRGH	511	MSUD
54370	MPGN	3109	MRKH syndrome	1332	MTC
79319	MPI-CDG	247775	MRKH syndrome type 1	499009	MTCT of syphilis
79253	mpKU	2578	MRKH syndrome type 2	352470	mtDNA deletion syndrome with limb-girdle weakness
3148	MPNST	→457240	MRX35	352470	mtDNA deletion syndrome with progressive myopathy
252212	MPNST with rhabdomyosarcomatous differentiation	85274	MRXS7		mtDNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria
2587	MPO deficiency	85324	MRXS9	255235	mtDNA depletion syndrome, encephalomyopathic form with renal tubulopathy
231736	MPPC syndrome	93952	MRXSH	369897	mtDNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies
83473	MPPH syndrome	2598	MSA	363534	mtDNA depletion syndrome, hepatocerebrorenal form
579	MPS1	102	MSA	254875	mtDNA depletion syndrome, myopathic form
93473	MPS1H	227510	MSA, cerebellar type	352447	mtDNA maintenance syndrome due to MGME1 deficiency
93476	MPS1H/S	98933	MSA, parkinsonian type	395	MTHFR deficiency
93474	MPS1S	227510	MSA-c	99701	MTLE-HS
580	MPS2	98933	MSA-p	252212	MTT
217085	MPS2A	1879	MSBD syndrome	86904	MTX-associated lymphoproliferative disorders
217093	MPS2B	254881	MSCAE	86904	MTX-LPD
581	MPS3	585	MSD	100024	mu-HCD
79269	MPS3A	2619	Mseleni joint disease	100024	Mu-heavy chain disease
79270	MPS3B	480536	MSH3-related AFAP	88949	MUC1-related autosomal dominant medullary cystic kidney disease
79271	MPS3C	480536	MSH3-related attenuated familial adenomatous polyposis	88949	MUC1-related autosomal dominant tubulointerstitial kidney disease
79272	MPS3D	480536	MSH3-related attenuated familial polyposis coli	88949	MUC1-related ADTKD
582	MPS4	480536	MSH3-related attenuated FAP	398961	Mucinous adenocarcinoma of ovary
309297	MPS4A	1309	MSK	391723	Mucinous adenocarcinoma of the appendix
309310	MPS4B	99898	MSMD due to complete IFNgammaR1 deficiency	424053	Mucinous cystadenocarcinoma of the pancreas
583	MPS6	319547	MSMD due to complete IFNgammaR2 deficiency	206470	Mucinous cystadenoma of ovary in childhood
276212	MPS6, rapidly progressing	319558	MSMD due to complete IL12B deficiency	319322	Mucinous tubular and spindle cell renal carcinoma
276223	MPS6, slowly progressing	319552	MSMD due to complete IL12RB1 deficiency	575	Muckle-Wells syndrome
584	MPS7	99898	MSMD due to complete interferon gamma receptor 1 deficiency	2331	Mucocutaneous lymph node syndrome
67041	MPS9	319547	MSMD due to complete interferon gamma receptor 2 deficiency	2451	Mucocutaneous venous malformations
579	MPSI	319552	MSMD due to complete interleukin 12 receptor beta 1 deficiency	423461	Mucolipidosis type 3 alpha/beta
293181	MPSI	319558	MSMD due to complete interleukin 12B deficiency		
93473	MPSIH	319563	MSMD due to complete ISG15 deficiency		
93476	MPSIH/S	319600	MSMD due to partial interferon regulatory factor 8 deficiency		
580	MPSII	319600	MSMD due to partial IRF8 deficiency		
217085	MPSIIA	319595	MSMD due to partial signal transducer and activator of transcription 1 deficiency		
217093	MPSIIB	319595	MSMD due to partial STAT1		
581	MPSIII				
79269	MPSIIIA				
79270	MPSIIIB				
79271	MPSIIIC				
79272	MPSIID				
93474	MPSIS				
582	MPSIV				
309297	MPSIVA				
309310	MPSIVB				
67041	MPSIX				
583	MPSVI				
276212	MPSVI, rapidly progressing				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
423470	Mucolipidosis type 3 gamma	276212	Mucopolysaccharidosis type VI, rapidly progressing		lymphangiomyomatosis with thrombocytopenia
576	Mucolipidosis type II	276223	Mucopolysaccharidosis type VI, slowly progressing	464321	Multifocal lymphangiomyomatosis-thrombocytopenia syndrome
576	Mucolipidosis type II alpha/beta	584	Mucopolysaccharidosis type VII	641	Multifocal motor neuropathy
577	Mucolipidosis type III	505248	Mucopolysaccharidosis-like plus disease	641	Multifocal motor neuropathy with conduction block
423461	Mucolipidosis type III alpha/beta	505248	Mucopolysaccharidosis-like syndrome with congenital heart defects and hematopoietic disorders	2033	Multifocal muscular fibrosis-obstructed vessels syndrome
423470	Mucolipidosis type III gamma	73263	Mucormycosis	99003	Multifocal pattern dystrophy simulating fundus flavimaculatus
578	Mucolipidosis type IV	52417	Mucosa-associated lymphatic tissue lymphoma	99003	Multifocal pattern dystrophy simulating Stargardt disease
579	Mucopolysaccharidosis type 1	52417	Mucosa-associated lymphoid tissue lymphoma	3286	Multifocal ventricular premature beats
93473	Mucopolysaccharidosis type 1H	46486	Mucosal pemphigoid	319287	Multilocular clear cell adenocarcinoma
93476	Mucopolysaccharidosis type 1H/S	585	Mucosulfatidosis	319287	Multilocular clear cell carcinoma
93474	Mucopolysaccharidosis type 1S	46486	Mucosynechial pemphigoid	319287	Multilocular clear cell renal cell adenocarcinoma
580	Mucopolysaccharidosis type 2	46486	Mucous membrane pemphigoid	319287	Multilocular clear cell renal cell carcinoma
217093	Mucopolysaccharidosis type 2, attenuated form	586	Mucoviscidosis	97366	Multilocular cyst of the kidney
217085	Mucopolysaccharidosis type 2, severe form	53271	Muenke syndrome	319287	Multilocular cystic renal cell adenocarcinoma
217085	Mucopolysaccharidosis type 2A	444	MUHH	319287	Multilocular cystic renal cell carcinoma
217093	Mucopolysaccharidosis type 2B	587	Muir-Torre syndrome	319287	Multilocular cystic renal neoplasm of low malignant potential
581	Mucopolysaccharidosis type 3	2576	MULIBREY dwarfism	168816	Multilocular peritoneal inclusion cyst
79269	Mucopolysaccharidosis type 3A	2576	MULIBREY nanism	97366	Multilocular renal cyst
79270	Mucopolysaccharidosis type 3B	247768	Müllerian aplasia and hyperandrogenism	97366	Multiloculated renal cyst
79271	Mucopolysaccharidosis type 3C	1655	Müllerian derivatives-lymphangiectasia-polydactyly syndrome	598	Multiminicore disease
79272	Mucopolysaccharidosis type 3D	2491	Müllerian duct anomalies-limb anomalies syndrome	598	Multiminicore myopathy
582	Mucopolysaccharidosis type 4	2578	Müllerian duct aplasia-renal dysplasia-cervical somite anomalies syndrome	2091	Multinodular goiter-cystic kidney-polydactyly syndrome
309297	Mucopolysaccharidosis type 4A	247768	Müllerian duct failure and hyperandrogenism	500135	Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome
309310	Mucopolysaccharidosis type 4B	2774	Multicentric carpo-tarsal osteolysis with or without nephropathy	26791	Multiple acyl-CoA dehydrogenase deficiency
583	Mucopolysaccharidosis type 6	93686	Multicentric Castleman disease	394532	Multiple acyl-CoA dehydrogenase deficiency, mild type
276212	Mucopolysaccharidosis type 6, rapidly progressing	93686	Multicentric giant lymph node hyperplasia	394529	Multiple acyl-CoA dehydrogenase deficiency, severe neonatal type
276223	Mucopolysaccharidosis type 6, slowly progressing	371428	Multicentric osteolysis-nodulosis-artropathy spectrum	2505	Multiple benign circumferential skin creases on limbs
584	Mucopolysaccharidosis type 7	85196	Multicentric osteolysis-nodulosis-artropathy syndrome	2678	Multiple café-au-lait spots
67041	Mucopolysaccharidosis type 9	139436	Multicentric reticulohistiocytosis	2678	Multiple café-au-lait syndrome
579	Mucopolysaccharidosis type I	1851	Multicystic dysplastic kidney	321	Multiple cartilaginous exostoses
93473	Mucopolysaccharidosis type IH	168816	Multicystic mesothelioma	254519	Multiple congenital anomalies due to 14q32.2 maternally expressed gene defect
93476	Mucopolysaccharidosis type IH/S	1851	Multicystic renal dysplasia		
580	Mucopolysaccharidosis type II	48162	Multifocal acquired demyelinating sensory and motor neuropathy		
217093	Mucopolysaccharidosis type II, attenuated form	3282	Multifocal atrial tachycardia		
217085	Mucopolysaccharidosis type II, severe form	99873	Multifocal eosinophilic granuloma		
217085	Mucopolysaccharidosis type IIA	464321	Multifocal		
217093	Mucopolysaccharidosis type IIB				
581	Mucopolysaccharidosis type III				
79269	Mucopolysaccharidosis type IIIA				
79270	Mucopolysaccharidosis type IIIB				
79271	Mucopolysaccharidosis type IIIC				
79272	Mucopolysaccharidosis type IID				
93474	Mucopolysaccharidosis type IS				
582	Mucopolysaccharidosis type IV				
309297	Mucopolysaccharidosis type IVA				
309310	Mucopolysaccharidosis type IVB				
67041	Mucopolysaccharidosis type IX				
583	Mucopolysaccharidosis type VI				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
280633	Multiple congenital anomalies-hypotonia-seizures syndrome	294049	multiple joint dislocations-short stature-hyperlaxity-craniofacial dysmorphism syndrome	370997	Muscle-eye-brain disease with bilateral multicystic leucodystrophy
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	493	Multiple keratoacanthoma	588	Muscle-eye-brain syndrome
369837	Multiple congenital anomalies-hypotonia-seizures syndrome type 3	65748	Multiple keratoacanthoma, Ferguson-Smith type	2576	Muscle-liver-brain-eye nanism
1486	Multiple contracture syndrome, Finnish type	587	Multiple keratoacanthoma, Muir-Torre type	2579	Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome
137776	Multiple contracture syndrome, Israeli-Bedouin type	79455	Multiple mastocytoma		Muscular dystrophy with progressive weakness, distal contractures and rigid spine
523	Multiple cutaneous and uterine leiomyomas	401869	Multiple mitochondrial dysfunctions syndrome type 1	199340	Muscular dystrophy, Selcen type
3453	Multiple endocrine deficiency-Addison disease-candidiasis syndrome	401874	Multiple mitochondrial dysfunctions syndrome type 2	1877	Muscular dystrophy-white matter spongiosis syndrome
652	Multiple endocrine neoplasia type 1	363424	Multiple mitochondrial dysfunctions syndrome type 3	99849	Muscular enolase deficiency
653	Multiple endocrine neoplasia type 2	457406	Multiple mitochondrial dysfunctions syndrome type 4	324416	Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome
247698	Multiple endocrine neoplasia type 2A	29073	Multiple myeloma	2349	Muscular pseudohypertrophy-hypothyroidism syndrome
247709	Multiple endocrine neoplasia type 2B	→636	Multiple non-ossifying fibromatosis	2953	Musculocontractural Ehlers-Danlos syndrome
247709	Multiple endocrine neoplasia type 3	435329	Multiple ossifying fibroma	3079	Mutchinick syndrome
276152	Multiple endocrine neoplasia type 4	321	Multiple osteochondromas	494	Mutilating keratoderma of Vohwinkel
166002	Multiple epiphyseal dysplasia due to collagen 9 anomaly	324299	Multiple paragangliomas associated with erythrocytosis	494	Mutilating keratoderma plus deafness
93308	Multiple epiphyseal dysplasia type 1	324299	Multiple paragangliomas associated with polycythemia	659	Mutilating palmoplantar hyperkeratosis with periorificial keratotic plaques
93307	Multiple epiphyseal dysplasia type 4	95494	Multiple pituitary hormone deficiencies, genetic forms	659	Mutilating palmoplantar keratoderma with periorificial keratotic plaques
93311	Multiple epiphyseal dysplasia type 5	→1234	Multiple pterygium syndrome, Aslan type	247798	MUTYH-related AFAP
166016	Multiple epiphyseal dysplasia with Robin phenotype	2215	Multiple pterygium-malignant hyperthermia syndrome	247798	MUTYH-related attenuated familial adenomatous polyposis
166024	Multiple epiphyseal dysplasia, Al-Gazali type	3151	Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome	247798	MUTYH-related attenuated familial polyposis coli
166011	Multiple epiphyseal dysplasia, Beighton type	65748	Multiple self-healing squamous epithelioma	247798	MUTYH-related attenuated FAP
166016	Multiple epiphyseal dysplasia, Lowry type	585	Multiple sulfatase deficiency	29	MVA
166032	Multiple epiphyseal dysplasia, with miniepipyses	2398	Multiple symmetric lipomatosis	2290	MVID
166029	Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia	3237	Multiple synostoses syndrome	2582	Myalgia-eosinophilia syndrome associated with tryptophan
166024	Multiple epiphyseal dysplasia-macrocephaly-distinctive facies syndrome	102	Multiple system atrophy	589	Myasthenia gravis
166011	Multiple epiphyseal dysplasia-myopia-deafness syndrome	227510	Multiple system atrophy, cerebellar type	498693	MYBPC1-related autosomal recessive non-lethal AMC syndrome
50920	Multiple fibroadenoma of the breast	98933	Multiple system atrophy, parkinsonian type	498693	MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome
83454	Multiple glomus tumors	102	Multisystem atrophy	2583	Mycetoma
201	Multiple hamartoma syndrome	404463	Multisystemic smooth muscle dysfunction syndrome	314946	Mycobacterium xenopi infection
2300	Multiple intestinal atresia	2959	Mulvihill-Smith syndrome	268249	Mycophenolate mofetil embryopathy
284139	Multiple joint dislocations-short stature-craniofacial dysmorphism-congenital heart defects syndrome	2578	MURCS association	83482	Mycoplasma encephalitis
		83315	Murine typhus	2584	Mycosis fungoides, Alibert-Bazin type
		2028	Murray-Puretic-Drescher syndrome	178512	Mycosis fungoides-associated
		99849	Muscle enolase deficiency		
		171445	Muscle filaminopathy		
		97234	Muscle phosphoglycerate mutase deficiency		
		588	Muscle-eye-brain disease		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	follicular mucinosis		progressive encephalopathies	273	Myotonic dystrophy type 1
183713	MyD88 deficiency	86909	Myoclonic epilepsy of infancy	606	Myotonic dystrophy type 2
59298	Myelinoclastic diffuse sclerosis	86913	Myoclonic status in non-progressive encephalopathies	→52430	Myotonic dystrophy type 3
135	Myelinosis centralis diffusa	1942	Myoclonic-astatic epilepsy	800	Myotonic myopathy, dwarfism, chondrodytrophy, ocular and facial anomalies
2585	Myelocerebellar disorder	1942	Myoclonic-astatic epilepsy in early childhood		
268813	Myelocystocele	435438	Myoclonus epilepsy and ataxia due to potassium channel mutation	508542	MYSM1 deficiency
494433	Myelodysplasia-infection-restriction of growth-adrenal hypoplasia-genital anomalies-enteropathy syndrome	551	Myoclonus epilepsy associated with ragged-red fibres	79105	Myxofibrosarcoma
494433	Myelodysplasia-infection-restriction of growth-adrenal hypoplasia-genital phenotypes-enteropathy syndrome	86913	Myoclonus epilepsy in non-progressive encephalopathies	79105	Myxoid malignant fibrous histiocytoma
86841	Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality	2589	Myoclonus-cerebellar ataxia-deafness syndrome	99967	Myxoid/round cell liposarcoma
824	Myelofibrosis with myeloid metaplasia	36899	Myoclonus-dystonia syndrome	57782	Myxoma with fibrous dysplasia
86850	Myeloid sarcoma	→36899	Myoclonus-dystonia type 15	1359	Myxoma-spotty pigmentation-endocrine overactivity syndrome
168953	Myeloid/lymphoid neoplasm associated with FGFR1 rearrangement	163696	Myoclonus-nephropathy syndrome	251643	Myxopapillary ependymoma
168947	Myeloid/lymphoid neoplasm associated with PDGFRA rearrangement	178464	Myofibrillar myopathy with early respiratory failure	2608	N syndrome
168950	Myeloid/lymphoid neoplasm associated with PDGFRB rearrangement	104077	Myopathic intestinal pseudoobstruction	79270	N-acetyl-alpha-glucosaminidase deficiency
29073	Myelomatosis	2596	Myopathy and diabetes mellitus	583	N-acetylgalactosamine 4-sulfatase deficiency
93969	Myelomeningocele	88635	Myopathy due to calsequestrin and SERCA1 protein overload	309297	N-acetylgalactosamine-6-sulfate sulfatase deficiency
2587	Myeloperoxidase deficiency	97234	Myopathy due to phosphoglycerate mutase deficiency	576	N-acetylglucosamine 1-phosphotransferase deficiency
437572	MYH7-related late-onset scapuloperoneal muscular dystrophy	43115	Myopathy with exercise intolerance, Swedish type	79329	N-acetylglucosaminyltransferase 2 deficiency
437572	MYH7-related late-onset scapuloperoneal syndrome	171889	Myopathy with hexagonally cross-linked tubular arrays	137754	N-acetyl-L-amino acid amidohydrolase deficiency
437572	MYH7-related late-onset SPMD	2598	Myopathy, lactic acidosis and sideroblastic anemia	103908	Na-H exchange deficiency
182050	MYH9-RD	2601	Myopathy-growth delay-intellectual disability-hypospadias syndrome	178303	Nabbius mask-like facial syndrome
182050	MYH9-related disease	1358	Myopathy-Moebius-Robin syndrome	439196	NAE
182050	MYH9-related disorder	289685	Myopericytoma	69087	Naegeli syndrome
182050	MYH9-related syndrome	368	Myophosphorylase deficiency	69087	Naegeli-Franceschetti-Jadassohn syndrome
182050	MYH9-related syndromic thrombocytopenia	178493	Myopic macular degeneration	840	Naevus syringocystadenomatous papilliferus
2588	Myhre syndrome	178493	Myopic maculopathy	245	NAFD
109	Myhre-Riley-Smith syndrome	289380	Myosclerosis	3137	NAGA deficiency
480491	MYO5B deficiency	337	Myositis ossificans progressiva	79279	NAGA deficiency type 1
480491	MYO5B-related progressive familial intrahepatic cholestasis	764	Myositis purulenta tropica	79280	NAGA deficiency type 2
45	Myoadenylate deaminase deficiency	764	Myositis tropicans	79281	NAGA deficiency type 3
1942	Myoclonic atonic epilepsy	306553	Myospherulosis	245	Nager acrofacial dysostosis
36899	Myoclonic dystonia	614	Myotonia congenita	245	Nager syndrome
→36899	Myoclonic dystonia 15	99734	Myotonia fluctuans	927	NAGS deficiency
86913	Myoclonic epilepsy in non-	99735	Myotonia permanens	2211	Naguib-Richieri-Costa syndrome
		3101	Myotonia-intellectual disability-skeletal anomalies syndrome	423454	Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome
		99736	Myotonia-painful contractions syndrome	→1487	Nail dysplasia-camptodactyly-brachydactyly type B syndrome
		800	Myotonic chondrodytrophy	2614	Nail-patella syndrome
				2613	Nail-patella-like renal disease
				158676	Nails-only DDEB
				853	NAIT

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
101	Naito-Oyanagi disease	240760	NBS-like disorder	398097	Neonatal Hughes syndrome
2229	Najjar syndrome	240760	NBSLD	137577	Neonatal hypoxic and ischemic brain injury
1063	Nakagawa angioblastoma	217560	NCHI	59303	Neonatal ichthyosis-sclerosing cholangitis syndrome
2615	Nakajo-Nishimura syndrome	1947	NCL, Northern epilepsy variant	294023	Neonatal inflammatory skin and bowel disease
2822	Nakamura-Osame syndrome	2481	NCM	247598	Neonatal intrahepatic cholestasis caused by citrin deficiency
44	NALD	75327	NCMD	247598	Neonatal intrahepatic cholestasis due to citrin deficiency
206569	NAM	91495	NCRNA disease	238688	Neonatal iodine exposure
→1359	NAME syndrome	443162	NDE1-related microhydranencephaly	398124	Neonatal lupus erythematosus
383	Nance deafness	1398	Near total absence of cerebellum	284979	Neonatal Marfan syndrome
627	Nance-Horan syndrome	399103	Nebulin-related early-onset distal myopathy	69063	Neonatal membranous glomerulopathy with maternal NEP deficiency
251279	Nanophtalmos-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	158011	Necrobiotic xanthogranuloma	69063	Neonatal membranous glomerulopathy with maternal neutral endopeptidase deficiency
35612	Nanophthalmia	439196	Necrolytic acral erythema	284979	Neonatal MFS
85196	NAO syndrome	391673	Necrotizing enterocolitis	79242	Neonatal multiple carboxylase deficiency
247868	NAPS12	440368	Necrotizing soft tissue infection	391504	Neonatal myasthenia gravis
83465	Narcolepsy without cataplexy	217560	NEHI	→42738	Neonatal neutropenia
2073	Narcolepsy-cataplexy syndrome	464366	NEK9-related lethal skeletal dysplasia	289857	Neonatal NKH
644	NARP syndrome	199244	Nelson syndrome	289857	Neonatal non-ketotic hyperglycinemia
141103	Nasal dermoid cyst	100079	NEN of appendix	56304	Neonatal osseous dysplasia type 1
141103	Nasal dermoid sinus cyst	506136	NEN of esophagus	3455	Neonatal progeroid syndrome
141219	Nasal dorsum fistula/cyst	217563	Neonatal acute respiratory distress due to SP-B deficiency	70587	Neonatal respiratory distress syndrome
141118	Nasal encephalocele	217563	Neonatal acute respiratory distress due to surfactant protein B deficiency	3206	Neonatal Schwartz-Jampel syndrome
141115	Nasal ganglioglioma	44	Neonatal adrenoleukodystrophy	398127	Neonatal scleroderma
141112	Nasal glial heterotopia	398109	Neonatal AHA	466784	Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect
141112	Nasal glioma	398109	Neonatal AIHA	417	Neonatal severe primary hyperparathyroidism
86879	Nasal T/natural killer-cell lymphoma	464370	Neonatal alloimmune neutropenia	1451	Neonatal-onset multisystem inflammatory disease
2662	Nasodigitoacoustic syndrome	398097	Neonatal antiphospholipid antibody syndrome	314950	Neoplastic hypereosinophilic syndrome
141083	Nasolacrimal duct cyst	398097	Neonatal antiphospholipid syndrome	94058	Neovascular glaucoma
141083	Nasolacrimal mucocele	398109	Neonatal autoimmune hemolytic anemia	654	Nephroblastoma
2399	Nasopalpebral lipoma-coloboma syndrome	137929	Neonatal brainstem dysfunction	2849	Nephroblastomatosis-fetal ascites-macrosomia-Wilms tumor syndrome
150	Nasopharyngeal carcinoma	314911	Neonatal Canavan disease	223	Nephrogenic diabetes insipidus
141107	Nasopharyngeal teratoma	313906	Neonatal congenital pancreatic cyst	3145	Nephrogenic diabetes insipidus-intracranial calcification syndrome
2770	Nasu-Hakola disease	398117	Neonatal dermatomyositis	137617	Nephrogenic fibrosing dermatopathy
1654	Natal teeth-intestinal pseudoobstruction-patent ductus syndrome	79118	Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome	93606	Nephrogenic syndrome of inappropriate antidiuresis
2663	Nathalie syndrome	398117	Neonatal DM		
168572	Native American myopathy	457185	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome		
69739	Navajo brainstem syndrome	69063	Neonatal glomerulopathy due to Neprilysin alloimmunization		
255229	Navajo neurohepatopathy	69063	Neonatal glomerulopathy due to neprilysin alloimmunization		
255229	Navajo neuropathy	289857	Neonatal glycine encephalopathy		
34217	Naxos disease	446	Neonatal hemochromatosis		
377	NBCCS				
157850	NBIA1				
216873	NBIA1, atypical form				
216866	NBIA1, classic form				
289560	NBIA4				
329284	NBIA5				
397725	NBIA6				
289560	NBIA due to C19orf12 mutation				
647	NBS				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
137617	Nephrogenic systemic fibrosis		mutation	137605	Neurofibromatosis 1-like syndrome
93622	Nephrolithiasis type 1	397725	Neurodegeneration with brain iron accumulation due to COASY mutation	636	Neurofibromatosis type 1
93623	Nephrolithiasis type 2	157850	Neurodegeneration with brain iron accumulation type 1	363700	Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion
655	Nephronophthisis	216873	Neurodegeneration with brain iron accumulation type 1, atypical form	97685	Neurofibromatosis type 1 microdeletion syndrome
3156	Nephronophthisis with retinal dystrophy	216866	Neurodegeneration with brain iron accumulation type 1, classic form	638	Neurofibromatosis type 1-Noonan syndrome
84081	Nephronophthisis-hepatocarcinoma syndrome	289560	Neurodegeneration with brain iron accumulation type 4	637	Neurofibromatosis type 2
411629	Nephropathic infantile cystinosis	329284	Neurodegeneration with brain iron accumulation type 5	93921	Neurofibromatosis type 3
2668	Nephropathy-deafness-hyperparathyroidism syndrome	217382	Neurodegenerative syndrome due to cerebral folate transport deficiency	2678	Neurofibromatosis type 6
2669	Nephrosis-deafness-urinary tract-digital malformations syndrome	453499	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome	638	Neurofibromatosis-Noonan syndrome
2065	Nephrosis-neuronal dysmigration syndrome	352665	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to 9q21 microdeletion	3148	Neurofibrosarcoma
300333	Nephrotic syndrome-deafness-pretribial epidermolysis bullosa syndrome	453504	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to a point mutation	970	Neurogenic acroosteolysis
300333	Nephrotic syndrome-hearing loss-pretribial epidermolysis bullosa syndrome	3474	Neuroectodermal dysplasia, CHIME type	1143	Neurogenic arthrogryposis multiplex congenita
2337	NEPPK	33445	Neuroectodermal melanolysosomal disease	100073	Neurogenic cervical rib syndrome
280576	Nestor-Guillermo progeria syndrome	3474	Neuroectodermal syndrome, Zunich type	100073	Neurogenic costoclavicular syndrome
100082	NET of anal canal	2676	Neuroectodermal-endocrine syndrome	178029	Neurogenic diabetes insipidus
100075	NET of stomach	506098	Neuroendocrine carcinoma of pancreas	644	Neurogenic muscle weakness-ataxia-retinitis pigmentosa syndrome
100080	NET of the colon	217560	Neuroendocrine cell hyperplasia of infancy	98593	Neurogenic palpebral tumor
100081	NET of the rectum	100079	Neuroendocrine neoplasm of appendix	3148	Neurogenic sarcoma
634	Netherton syndrome	506136	Neuroendocrine neoplasm of esophagus	431255	Neurogenic scapuloperoneal amyotrophy, New England type
2671	Neu-Laxova syndrome	100080	Neuroendocrine neoplasm of the colon	85146	Neurogenic scapuloperoneal syndrome, KAESER type
99078	Neuhauser anomaly	100075	Neuroendocrine tumor of stomach	100073	Neurogenic thoracic outlet compression syndrome
2479	Neuhäuser syndrome	100080	Neuroendocrine tumor of the colon	100073	Neurogenic thoracic outlet syndrome
3350	Neuhauser-Daly-Magnelli syndrome	100081	Neuroendocrine tumor of the rectum	100073	Neurogenic TOS
2672	Neuhauser-Eichner-Opitz syndrome	2677	Neuroepithelioma	94093	Neuroleptic malignant syndrome
157826	Neumann tumor	2673	Neurofaciocutaneous syndrome	36397	Neuropilomatosis
2901	Neuralgic amyotrophy	157846	Neuroferritinopathy	163746	Neurologic Waardenburg-Shah syndrome
2901	Neuralgic shoulder amyotrophy	252183	Neurofibroma	137754	Neurological conditions associated with aminoacylase 1 deficiency
351	Neuraminidase deficiency with beta-galactosidase deficiency			206586	Neurolymphomatosis
268865	Neurenteric cyst			71211	Neuromyelitis optica
252164	Neurilemmoma			1947	Neuronal ceroid lipofuscinosis, Northern epilepsy variant
93921	Neurilemmomatosis			99811	Neuronal intestinal pseudoobstruction
252164	Neurilemoma			2289	Neuronal intranuclear inclusion disease
508533	Neuro-immuno-skeletal dysplasia syndrome due to EXTL3 deficiency			639	Neuropathy associated with monoclonal IgM antibodies to myelin-associated glycoprotein
635	Neuroblastoma			139512	Neuropathy with hearing impairment
2481	Neurocutaneous melanocytosis			644	Neuropathy-ataxia-retinitis
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				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	pigmentosa syndrome	289356	NGCO	436166	NLRC4-related autoinflammatory syndrome with macrophage activation syndrome
217622	Neurosensory deafness with dilated cardiomyopathy	404454	NGLY1 deficiency	436166	NLRC4-related autoinflammatory syndrome with MAS
217622	Neurosensory hearing loss with dilated cardiomyopathy	404454	NGLY1-CDDG	436166	NLRC4-related infantile enterocolitis-autoinflammatory syndrome
137596	Neurotrophic keratitis	280576	NGPS	436166	NLRC4-related macrophage activation syndrome
137596	Neurotrophic keratopathy	2770	NHD	436166	NLRC4-related MAS
98907	Neutral lipid storage disease with ichthyosis	169079	NHEJ1 deficiency	247868	NLRP12-associated hereditary periodic fever syndrome
98908	Neutral lipid storage disease with myopathy without ichthyosis	276608	NI-PHH	98907	NLSDI
98908	Neutral lipid storage myopathy	247598	NICCD	98908	NLSDM
→86872	Neutropenia-hyperlymphocytosis with large granular lymphocytes syndrome	141179	NICH	443167	NMC
2690	Neutropenia-monocytopenia-deafness syndrome	3051	Nicolaides-Baraitser syndrome	391504	NMG
183707	Neutrophil immunodeficiency syndrome	77292	Niemann-Pick disease type A	86867	NMZL
169142	Neutrophil-specific granule deficiency	77293	Niemann-Pick disease type B	2615	NNS
575	Neutrophilic urticaria	646	Niemann-Pick disease type C	1884	Noble-Bass-Sherman syndrome
370059	NEVADA syndrome	216986	Niemann-Pick disease type C, adult neurologic onset	31204	Nocardiosis
→1359	Nevi-atrial myxoma-myxoid neurofibromata-ephelides syndrome	216981	Niemann-Pick disease type C, classic form	→98784	Nocturnal paroxysmal dystonia
→1900	Nevo syndrome	216981	Niemann-Pick disease type C, juvenile neurologic onset	86867	Nodal marginal zone B-cell lymphoma
377	Nevoid basal cell carcinoma syndrome	216978	Niemann-Pick disease type C, late infantile neurologic onset	137810	Nodular cutaneous amyloidosis
228264	Nevus anelasticus	216975	Niemann-Pick disease type C, severe early infantile neurologic onset	477742	Nodular fasciitis
64754	Nevus comedonicus syndrome	216972	Niemann-Pick disease type C, severe perinatal form	90393	Nodular lichen myxedematosus
228254	Nevus elasticus	→646	Niemann-Pick disease type D	86893	Nodular lymphocyte predominant Hodgkin lymphoma
370059	Nevus epidermicus verrucosus with angiodysplasia and aneurysms	99022	Niemann-Pick disease type E	2149	Nodular neuronal heterotopia
263432	Nevus fuscocaeeruleus acromiodeltoideus	→646	Niemann-Pick disease, Nova Scotia type	33577	Nodular non-suppurative panniculitis
263425	Nevus fusculocaeeruleus ophthalmomaxillaris	2633	Nievergelt syndrome	48372	Nodular regenerative hyperplasia of the liver
263432	Nevus of Ito	1390	Night blindness-skeletal anomalies-dysmorphism syndrome	158772	Nodular urticaria pigmentosa
263425	Nevus of Ota	98757	Nigro-spino-dentatal degeneration with nuclear ophthalmoplegia	85196	Nodulosis-arthropathy-osteolysis syndrome
2612	Nevus sebaceus of Jadassohn	432	nIHH	2700	Noma
2612	Nevus sebaceus syndrome	2322	Niikawa-Kuroki syndrome	1451	NOMID syndrome
363558	New-onset refractory status epilepticus	647	Nijmegen breakage syndrome	73267	Non-24-hour sleep-wake syndrome
83471	Nezelof syndrome	240760	Nijmegen breakage syndrome-like disorder	231720	Non-acquired combined pituitary hormone deficiency-deafness-rigid cervical spine syndrome
636	NF1	447731	NIK deficiency	231720	Non-acquired combined pituitary hormone deficiency-sensorineuronal hearing loss-spine abnormalities syndrome
97685	NF1 microdeletion syndrome	781	Nine Mile fever	631	Non-acquired isolated growth hormone deficiency
137605	NF1-like syndrome	99825	Nipah encephalitis	97566	Non-amyloid fibrillary glomerulonephritis
637	NF2	99825	Nipah fever	97566	Non-amyloid fibrillary glomerulopathy
93921	NF3	59303	NISCH syndrome	86861	Non-amyloid MIDD
2678	NF6	1422	Nivelon-Nivelon-Mabille syndrome	86861	Non-amyloid monoclonal
69087	NFJ syndrome	263665	NK-cell enteropathy		
638	NFNS	86873	NK-cell large granular lymphocyte leukemia		
91349	NFPA	86873	NK-cell LGL leukemia		
401869	NFU1 deficiency	86879	NK/T-cell lymphoma		
		407	NKA		
		86879	NKTCL		
		86893	NLPHL		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	immunoglobulin deposition disease		neuroendocrine neoplasm of pancreas	436271	Non-progressive predominantly posterior cavitating leukoencephalopathy with peripheral neuropathy
79394	Non-bullous congenital ichthyosiform erythroderma	506075	Non-functioning well-differentiated pancreatic NEN	439202	Non-recovering OBPI
289362	Non-central nervous system-localized embryonal carcinoma	506075	Non-functioning well-differentiated pancreatic neuroendocrine neoplasm	439202	Non-recovering OBPL
77259	Non-cerebral juvenile Gaucher disease	26137	Non-giant cell granulomatous temporal arteritis with eosinophilia	439202	Non-recovering obstetric brachial plexus lesion
48372	Non-cirrhotic nodulation	→79452	Non-hereditary congenital primary lymphedema	101106	Non-secreting chemodectoma
854	Non-cirrhotic portal vein thrombosis	→90186	Non-hereditary late-onset primary lymphedema	94080	Non-secreting paraganglioma
325529	Non-classic congenital lipid adrenal hyperplasia due to STAR deficiency	357034	Non-hereditary retinoblastoma	363494	Non-seminomatous germ cell tumor of testis
289362	Non-CNS-localized embryonal carcinoma	163924	Non-herpetic acute limbic encephalitis	91364	Non-specific idiopathic interstitial pneumonia
216796	Non-deforming osteogenesis imperfecta	329883	Non-hypoproteinemic hypertrophic gastropathy	91364	Non-specific interstitial pneumonia
96136	Non-distal deletion 7p	329918	Non-Ig-mediated membranoproliferative glomerulonephritis	206572	Non-specific myositis
1581	Non-distal deletion 10q	363999	Non-immune fetal edema	90031	Non-spherocytic hemolytic anemia due to hexokinase deficiency
96160	Non-distal deletion 12q	363999	Non-immune fetal hydrops	35099	Non-syndromic bicoronal synostosis
96164	Non-distal deletion 20q	363999	Non-immune HF	30391	Non-syndromic biliary atresia
96112	Non-distal duplication 9q	363999	Non-immune hydrops fetalis	91495	Non-syndromic congenital retinal non-attachment
1695	Non-distal duplication 10q	329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis	49042	Non-syndromic dentinogenesis imperfecta
1702	Non-distal duplication 13q	329918	Non-immunoglobulin-mediated MPGN	49042	Non-syndromic DGI
96136	Non-distal monosomy 7p	263548	Non-inflammatory generalized peeling skin syndrome type A.	87884	Non-syndromic genetic deafness
1581	Non-distal monosomy 10q	263548	Non-inflammatory peeling skin syndrome type A	276234	Non-syndromic male infertility due to asthenozoospermia
96160	Non-distal monosomy 12q	141179	Non-involuting congenital hemangioma	276234	Non-syndromic male infertility due to sperm motility disorder
96164	Non-distal monosomy 20q	407	Non-ketotic hyperglycinemia	3366	Non-syndromic metopic craniosynostosis
3306	Non-distal tetrasomy 15q	98890	Non-Leber type optic atrophy with early-onset	35093	Non-syndromic sagittal synostosis
96112	Non-distal trisomy 9q	411641	Non-nephropathic cystinosis	35098	Non-syndromic unicoronal synostosis
1695	Non-distal trisomy 10q	84085	Non-neurogenic neurogenic bladder	96136	Non-telomeric monosomy 7p
1702	Non-distal trisomy 13q	209989	Non-papillary transitional cell carcinoma of the bladder	1581	Non-telomeric monosomy 10q
329469	Non-DS-AMKL	209989	Non-papillary urothelial carcinoma	96160	Non-telomeric monosomy 12q
206538	Non-dysgerminomatous germ cell cancer of ovary	238583	Non-phenylketonuric hyperphenylalaninemia	96164	Non-telomeric monosomy 20q
363494	Non-dysgerminomatous germ cell tumor of testis	508523	Non-phenylketonuric non-BH4-deficiency hyperphenylalaninemia	3306	Non-telomeric tetrasomy 15q
2337	Non-epidermolytic palmoplantar keratoderma	79651	Non-PKU HPA	96112	Non-telomeric trisomy 9q
→2199	Non-epidermolytic palmoplantar keratoderma	→144	Non-polyposis Turcot syndrome	1695	Non-telomeric trisomy 10q
2972	Non-eruption of teeth-maxillary hypoplasia-genu valgum syndrome	314647	Non-progressive cerebellar ataxia with intellectual disability	1702	Non-telomeric trisomy 13q
100070	Non-fluent variant PPA	1766	Non-progressive cerebellar ataxia-intellectual disability syndrome	411703	Non-tuberculous mycobacterial lung disease
506075	Non-functioning neuroendocrine tumor of pancreas			209919	Non-Wilsonian hepatic copper toxicosis of infancy and childhood
506075	Non-functioning pancreatic NET			602	Nonaka myopathy
506075	Non-functioning pancreatic neuroendocrine tumor			79452	Nonne-Milroy lymphedema
94080	Non-functioning paraganglioma			648	Noonan syndrome
91349	Non-functioning pituitary adenoma			500	Noonan syndrome with multiple lentigines
506075	Non-functioning PNET			363972	Noonan syndrome-like disorder with JMML
506075	Non-functioning well-differentiated NEN of pancreas			363972	Noonan syndrome-like disorder
506075	Non-functioning well-differentiated				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	with juvenile myelomonocytic leukemia	97297	Oberklaid-Danks syndrome		sensorineural deafness
2701	Noonan syndrome-like disorder with loose anagen hair	397615	Obesity due to CEP19 deficiency	1000	Ocular albinism with late-onset sensorineural deafness
230	Noradrenaline deficiency	66628	Obesity due to congenital leptin deficiency	54	Ocular albinism, Nettleship-Falls type
230	Norepinephrine deficiency	179494	Obesity due to leptin receptor gene deficiency	411641	Ocular cystinosis
314928	Normal pressure hydrocephalus	71529	Obesity due to melanocortin 4 receptor deficiency	2788	Ocular form of osteogenesis imperfecta
2254	Norman disease	71526	Obesity due to pro-opiomelanocortin deficiency	1125	Ocular motor apraxia, Cogan type
79255	Norman-Landing disease	71528	Obesity due to prohormone convertase 1 deficiency	99922	Ocular pemphigoid
→682	Normokalemic periodic paralysis	369873	Obesity due to SIM1 deficiency	534	Oculo-cerebro-renal dystrophy
→682	Normokalemic PP	88643	Obesity-colitis-hypothyroidism-cardiac hypertrophy-developmental delay syndrome	534	Oculo-cerebro-renal syndrome
→682	NormoKPP	1303	Obliterative bronchiolitis	1305	Oculo-digit-esophageal-duodenal syndrome
812	Normomorphic sialidosis	64743	Obliterative portal venopathy	391641	Oculo-digit-esophageal-duodenal syndrome type 1
→682	NormoPP	2970	Obrinsky syndrome	→1200	Oculo-oto-facial dysplasia
432	Normosmic congenital hypogonadotropic hypogonadism	3411	Obstructed hemivagina and ipsilateral renal anomaly	2307	Oculo-oto-radial syndrome
432	Normosmic idiopathic hypogonadotropic hypogonadism	352731	OCA1	2714	Oculo-palato-cerebral dwarfism
649	Norrie disease	352734	OCA1-MP	2714	Oculo-palato-cerebral syndrome
649	Norrie-Warburg disease	352737	OCA1-TS	→293843	Oculo-skeletal-abdominal syndrome
363558	NORSE	79431	OCA1A	157962	Oculoauricular syndrome, Schorderet type
75327	North Carolina macular dystrophy	79434	OCA1B	398156	Oculoauriculofrontonasal syndrome
75327	North Carolina macular dystrophy, retinal 1	79432	OCA2	374	Oculoauriculovertebral dysplasia
280620	North Sea progressive myoclonus epilepsy	79433	OCA3	2549	Oculoauriculovertebral spectrum with radial defects
1947	Northern epilepsy	79435	OCA4	374	Oculoauriculovertebral syndrome
79293	Norum disease	370091	OCA5	2705	Oculocerebral dysplasia
178	Notochordal sarcoma	370097	OCA6	2719	Oculocerebral hypopigmentation syndrome, Cross type
2703	Nova syndrome	352745	OCA7	2720	Oculocerebral hypopigmentation syndrome, Preus type
314928	NPH	217017	Occipital atretic cephalocele-unusual facies-large feet syndrome	1647	Oculocerebrocutaneous syndrome
3032	NPHP3-related Meckel-like syndrome	268823	Occipital encephalocele	2707	Oculocerebrofacial syndrome, Kaufman type
480476	NR1H4 deficiency	198	Occipital horn syndrome	534	Oculocerebrorenal dystrophy
634	NS	280640	Occipital malformations of cortical development	534	Oculocerebrorenal syndrome of Lowe
88616	NS-ARID	280640	Occipital MCD	352731	Oculocutaneous albinism type 1
2701	NS/LAH	280640	Occipital pachygyria and polymicrogyria	79431	Oculocutaneous albinism type 1A
417	NSHPT	353351	Occlusive idiopathic juxtapatelloellar retinal telangiectasis	79434	Oculocutaneous albinism type 1B
93606	NSIAD	51608	Occlusive infantile arteriopathy	79432	Oculocutaneous albinism type 2
91364	NSIP	1647	OCCS	79433	Oculocutaneous albinism type 3
440368	NSTI	99889	Occult ectopic ACTH secretion	79435	Oculocutaneous albinism type 4
454840	NTHL1-related AFAP	247834	Occult macular dystrophy	370091	Oculocutaneous albinism type 5
454840	NTHL1-related attenuated familial adenomatous polyposis	84085	Occult neuropathic bladder	370097	Oculocutaneous albinism type 6
454840	NTHL1-related attenuated FAP	2704	Ochoa syndrome	352745	Oculocutaneous albinism type 7
100073	NTOS	247834	OCMD	79434	Oculocutaneous albinism, Amish type
314790	Null pituitary adenoma	534	OCR	28378	Oculocutaneous tyrosinemia
280234	Null syndrome	534	OCRL	2709	Oculodental syndrome, Rutherford type
443167	NUT midline carcinoma	664	OCT deficiency	2710	Oculodentodigital dysplasia
54	OA1	54	Ocular albinism type 1		
398156	OAFNS	352740	Ocular albinism with congenital		
1106	OAS				
374	OAV dysplasia				
374	OAVS				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2710	Oculodentoosseous dysplasia	2919	OFD5		arthritis with anti-nuclear antibodies
3339	Oculoectodermal syndrome	2754	OFD6	247846	Oligoarticular juvenile idiopathic arthritis without anti-nuclear antibodies
2712	Oculofaciocardiodental syndrome	→2750	OFD7	251656	Oligoastrocytoma
1876	Oculogastrointestinal muscular dystrophy	2755	OFD8	75378	Oligcone syndrome
2108	Oculomandibulofacial syndrome	141007	OFD9	75378	Oligcone trichromacy
1794	Oculomaxillofacial dysostosis	2756	OFD10	251627	Oligodendrogioma
1154	Oculomelic amyoplasia	141000	OFD11	99798	Oligodontia
1125	Oculomotor apraxia, Cogan type	141327	OFD12	300576	Oligodontia-cancer predisposition syndrome
2713	Oculoosteocutaneous syndrome	141330	OFD13	2260	Oligomeganephronia
99806	Oculootodental syndrome	434179	OFD14	2260	Oligomeganephronic renal hypoplasia
→293843	Oculopalatoskeletal syndrome	508501	OFD18	137831	Oligophrenin-1 syndrome
98897	Oculopharyngeal distal myopathy	2750	OFDI	2920	Oliver syndrome
270	Oculopharyngeal muscular dystrophy	2750	OFDSI	3363	Oliver-McFarlane syndrome
98897	Oculopharyngodistal myopathy	391655	Off-periods in Parkinson disease not responding to oral treatment	2732	Olivopontocerebellar atrophy-deafness syndrome
2715	Oculorenocerebellar syndrome	424080	OGCT of pancreas	166063	Olivopontocerebellar hypoplasia
2717	Oculotrichoanal syndrome	276432	Ogden syndrome	296	Ollier disease
2718	Oculotrichodysplasia	75382	Oguchi disease	659	Olmsted syndrome
166272	ODCD	75382	Oguchi syndrome	1183	OMA syndrome
2710	ODDD syndrome	1186	Ohaha syndrome	247834	OMD
1305	ODED syndrome	2728	Ohdo syndrome	39041	Omenn syndrome
391641	ODED syndrome type 1	2728	Ohdo-Madokoro-Sonoda syndrome	2741	OMM syndrome
999	O'Doherty syndrome	64739	OHSS	2733	Omodyplasia
2253	O'Donnell-Pappas syndrome	1934	Ohtahara syndrome	660	Omphalocele
2722	Odonto-onycho dysplasia-alopelia syndrome	3411	OHVIRA syndrome	3164	Omphalocele syndrome, Shprintzen-Goldberg type
2721	Odonto-onycho-dermal dysplasia	666	OI	93929	Omphalocele-cloacal exstrophy-imperforate anus-spinal defect syndrome
→2036	Odonto-onycho-hypohidrotic dysplasia-midline scalp defects syndrome	216796	OI type 1	496693	Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome
69082	Odonto-tricho-ungual-digitopalmar syndrome	216804	OI type 2	490	Omphalomesenteric cyst
69082	Odonto-tricho-ungual-digitopalmar syndrome, Mendoza-Valiente type	216812	OI type 3	210115	OMPP
166272	Odontochondrodysplasia	216820	OI type 4	1183	OMS
447777	Odontogenic keratocystoma	216828	OI type 5	319266	Omsk hemorrhagic fever
247685	Odontohypophosphatasia	2729	Okamoto syndrome	3191	Onat syndrome
77295	Odontoleukodystrophy	93293	Okihiro syndrome	2737	Onchocerciasis
2724	Odontomatosis-aortae esophagus stenosis syndrome	261638	Okihiro syndrome due to 20q13 microdeletion	137675	Oncocytic cardiomyopathy
1811	Odontomericonychial dysplasia	261647	Okihiro syndrome due to a point mutation	352540	Oncogenic hypophosphatemic osteomalacia
2723	Odontotrichomelic syndrome	261638	Okihiro syndrome due to del(20)(q13)	352540	Oncogenic osteomalacia
1487	ODP	261638	Okihiro syndrome due to monosomy 20q13	661	Ondine curse
93929	OEIS complex	69088	OL-EDA-ID	661	Ondine syndrome
2676	Oerter-Friedman-Anderson syndrome	→113	Oley syndrome	99803	Ondine-Hirschsprung disease
2792	OFC syndrome	478	Olfacto-genital pathological sequence	99803	Ondine-Hirschsprung syndrome
2712	OFCD syndrome	1957	Olfactory neuroblastoma	→33364	ONMR syndrome
488265	OFD	85410	Oligoarticular JIA	238744	Onycho-digitomammary syndrome
2750	OFD1	247839	Oligoarticular JIA with anti-nuclear antibodies	→33364	Onycho-tricho-dysplasia-neutropenia syndrome
2751	OFD2	247846	Oligoarticular JIA without anti-nuclear antibodies		
2752	OFD3	85410	Oligoarticular juvenile idiopathic arthritis		
2753	OFD4	247839	Oligoarticular juvenile idiopathic		

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300504	Onychocytic matricoma	496790	Optic atrophy-peripheral neuropathy-developmental delay syndrome	2750	Orofaciodigital syndrome type 1
300512	Onychomatricoma	313800	Optic nerve edema-splenomegaly syndrome	2751	Orofaciodigital syndrome type 2
2614	Onychoosteodysplasia	2086	Optic pathway glioma	2752	Orofaciodigital syndrome type 3
2786	OCHS	499107	Optic perineuritis	2753	Orofaciodigital syndrome type 4
99806	OOD	353253	Oral dysesthesia	2919	Orofaciodigital syndrome type 5
2721	OODD	31142	Oral erosive lichen	2754	Orofaciodigital syndrome type 6
98890	OPA2	357154	Oral submucous fibrosis	→2750	Orofaciodigital syndrome type 7
67036	OPA3, autosomal dominant	457252	Oral tongue squamous cell carcinoma	2755	Orofaciodigital syndrome type 8
49042	Opalescent teeth without OI	2750	Oral-facial-digital syndrome type 1	141007	Orofaciodigital syndrome type 9
49042	Opalescent teeth without osteogenesis imperfecta	2751	Oral-facial-digital syndrome type 2	2756	Orofaciodigital syndrome type 10
90650	OPD I syndrome	2752	Oral-facial-digital syndrome type 3	141000	Orofaciodigital syndrome type 11
90652	OPD II syndrome	2753	Oral-facial-digital syndrome type 4	141327	Orofaciodigital syndrome type 12
90650	OPD syndrome 1	2919	Oral-facial-digital syndrome type 5	141330	Orofaciodigital syndrome type 13
90652	OPD syndrome 2	2754	Oral-facial-digital syndrome type 6	434179	Orofaciodigital syndrome type 14
98897	OPDM	→2750	Oral-facial-digital syndrome type 7	508501	Orofaciodigital syndrome type 18
268363	Open iniencephaly	2755	Oral-facial-digital syndrome type 8	2756	Orofaciodigital syndrome with fibular aplasia
137831	OPHN1 syndrome	141007	Oral-facial-digital syndrome type 9	141007	Orofaciodigital syndrome with retinal abnormalities
1106	Ophthalmomacromelic syndrome	2756	Oral-facial-digital syndrome type 10	2755	Orofaciodigital syndrome, Edwards type
2741	Ophthalmomandibulomelic dysplasia	141000	Oral-facial-digital syndrome type 11	141000	Orofaciodigital syndrome, Gabrielli type
1186	Ophthalmoplegia-hypotonia-ataxia-hypoacusis-athetosis syndrome	141327	Oral-facial-digital syndrome type 12	2919	Orofaciodigital syndrome, Thurston type
2743	Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome	141330	Oral-facial-digital syndrome type 13	93958	Oromandibular dystonia
1308	Opitz C trigonocephaly	434179	Oral-facial-digital syndrome type 14	141077	Oropharyngeal teratoma
2745	Opitz G/BBB syndrome	508501	Oral-facial-digital syndrome type 18	30	Oroticaciduria
2745	Opitz syndrome	141007	Oral-facial-digital syndrome with retinal abnormalities	30	Orotidylid decarboxylase deficiency
1308	Opitz trigonocephaly C syndrome	508501	Oral-facial-digital syndrome with short stature and brachymesophalangia	64692	Oroya fever
1308	Opitz trigonocephaly syndrome	2755	Oral-facial-digital syndrome, Edwards type	443236	Orthostatic intolerance due to NET deficiency
97297	Opitz trigonocephaly-like syndrome	141000	Oral-facial-digital syndrome, Gabrielli type	→293843	OSA syndrome
1786	Opitz-Caltabiano syndrome	500062	ORAS	93382	Osebold-Remondini syndrome
2745	Opitz-Frias syndrome	1647	Orbital cyst with cerebral and focal dermal malformations	97335	Osgood-Schlatter disease
93932	Opitz-Kaveggia syndrome	52994	Orbital leiomyoma	2760	OSLAM syndrome
270	OPMD	268139	Orbital medulloepithelioma	729	Osler-Vaquez disease
499107	OPN	2612	Organoid nevus syndrome	1427	OSMED
256	Oppenheim dystonia	166421	Orgasm-induced seizures	357154	OSMF
2788	OPPG	49041	Ormond disease	140436	Osseous venous malformation
2746	Opsismodysplasia	414	Ornithine aminotransferase deficiency	73230	Ossification anomalies-psychomotor developmental delay syndrome
1183	Opsoclonus-myoclonus syndrome	664	Ornithine carbamoyltransferase deficiency	57196	Osteitis condensans of the clavicle
1183	Opsoclonus-myoclonus-ataxia syndrome	415	Ornithine carrier deficiency	58040	Osteoblastoma
363746	Optic ataxia-gaze apraxia-simultanagnosia syndrome	664	Ornithine transcarbamylase deficiency	2764	Osteochondritis dissecans
98673	Optic atrophy type 1	415	Ornithine translocase deficiency	251262	Osteochondritis dissecans and short stature
98890	Optic atrophy type 2	415	ORNT1 deficiency	3314	Osteochondritis of phalangeal epiphyses
1215	Optic atrophy-deafness-polyneuropathy-myopathy syndrome	2319	Orocraniodigital syndrome	2054	Osteochondritis of tarsal/metatarsal bone
401777	Optic atrophy-intellectual disability syndrome	353253	Orodynia	2380	Osteochondritis of the capital femoral epiphysis
→1215	Optic atrophy-ophthalmoplegia-ptosis-deafness-myopathy syndrome			97332	Osteochondritis of the lunate bone

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
97335	Osteochondritis of the tibial tubercle		type 2	64739	Ovarian hyperstimulation syndrome
2653	Osteochondrodysplastic dwarfism-deafness-retinitis pigmentosa syndrome	2785	Osteopetrosis with renal tubular acidosis	398987	Ovarian immature teratoma
2653	Osteochondrodysplastic nanism-deafness-retinitis pigmentosa syndrome	178389	Osteopetrosis-hypogammaglobulinemia syndrome	213512	Ovarian malignant mixed epithelial mesenchymal tumor
800	Osteochondromuscular dystrophy	94063	Osteopoikilosis-short stature-intellectual disability syndrome	213512	Ovarian malignant mixed Müllerian tumor
2768	Osteochondrosis deformans tibiae	2787	Osteoporosis-macrocephaly-blindness-joint hyperlaxity syndrome	99916	Ovarian malignant Sertoli-Leydig cell tumor
97337	Osteochondrosis of patella	2786	Osteoporosis-oculocutaneous hypopigmentation syndrome	398987	Ovarian malignant teratoma
3314	Osteochondrosis of phalangeal epiphyses	2788	Osteoporosis-pseudoglioma syndrome	398961	Ovarian mucinous adenocarcinoma
2380	Osteochondrosis of the capital femoral epiphysis	666	Osteopsathyrosis	99916	Ovarian Sertoli-Leydig cell cancer
97336	Osteochondrosis of the capital humerus	668	Osteosarcoma	206473	Ovarian tumor of low malignant potential
97332	Osteochondrosis of the lunate bone	2760	Osteosarcoma-limb anomalies-erythroid macrocytosis syndrome	99853	Ovarioleukodystrophy
2054	Osteochondrosis of the tarsal bone	2790	Ostéosclérose autosomique dominante type Worth	498488	Overgrowth syndrome with 2q37 translocation
97335	Osteochondrosis of the tibial tubercle	178377	Osteosclerosis-developmental delay-craniosynostosis syndrome	137634	Overgrowth-macrocephaly-facial dysmorphism syndrome
424080	Osteoclastic giant cell tumor of pancreas	75325	Osteosclerosis-ichthyosis-premature ovarian failure syndrome	498485	Overgrowth-metaphyseal undermodeling-spondylar dysplasia syndrome
363976	Osteoclastoma	500548	Osteosclerotic metaphyseal dysplasia	3203	Overhydrated hereditary stomatocytosis
2763	Osteocraniosplenic syndrome	2905	Osteosclerotic myeloma	206572	Overlap myositis
2763	Osteocraniostenosis	1338	Ostravik-Lindemann-Solberg syndrome	326	Owren disease
488265	Osteofibrous dysplasia	99965	O'Sullivan-McLeod syndrome	832	OXCT1 deficiency
666	Osteogenesis imperfecta	664	OTC deficiency	31	Oxoglutaricaciduria
216796	Osteogenesis imperfecta type 1	1308	OTCS	33572	Oxoprolinuria due to oxoprolinase deficiency
216804	Osteogenesis imperfecta type 2	2791	Otodental dysplasia	79302	Oxysterol 7-alpha-hydroxylase deficiency
216812	Osteogenesis imperfecta type 3	2791	Otodental syndrome	36355	P2Y12 defect
216820	Osteogenesis imperfecta type 4	2792	Otofaciocervical syndrome	35664	P5CS deficiency
216828	Osteogenesis imperfecta type 5	141136	Otomandibular dysostosis	35120	P5N deficiency
2771	Osteogenesis imperfecta-congenital joint contractures syndrome	141136	Otomandibular syndrome	98971	PACD
2773	Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome	2793	Otoonychoperoneal syndrome	2796	Pachydermoperiostosis
668	Osteogenic sarcoma	90650	Otopalatodigital syndrome type 1	2798	Pachygyria-intellectual disability-epilepsy syndrome
2645	Osteoglophonic dwarfism	90652	Otopalatodigital syndrome type 2	2309	Pachyonychia congenita
2645	Osteoglosphonic dysplasia	1427	Otospondylomegaepiphyseal dysplasia	1952	Pacman dysplasia
2777	Osteomesopyknosis	457252	OTSCC	140989	PACNS
824	Osteomyelofibrosis	69082	OTUDP syndrome	477749	PADMAL
399293	Osteonecrosis of the jaw	500062	OTULIN deficiency	441	PAF
2780	Osteopathia striata-cranial sclerosis syndrome	500062	OTULIN-related autoinflammatory syndrome	95232	PAFAH1B1-related lissencephaly
2779	Osteopathia striata-pigmentary dermopathy-white forelock syndrome	500062	Otulipenia	180275	Paget disease of the breast
2324	Osteopenia-intellectual disability-sparse hair syndrome	50943	Oudtshoorn disease	180275	Paget disease of the nipple
91133	Osteopenia-myopia-hearing loss-intellectual disability-facial dysmorphism syndrome	1179	Ouvrier-Billson syndrome	357131	Paget-Schrotter disease
53	Osteopetrosis autosomal dominant	213504	Ovarian adenocarcinoma	52430	Pagetoid amyotrophic lateral sclerosis
		213512	Ovarian carcinosarcoma	52430	Pagetoid neuroskeletal syndrome
		398971	Ovarian clear cell adenocarcinoma	178517	Pagetoid reticulosis, Woringer-Kolopp type
		314473	Ovarian fibroma	180275	Paget's disease of the nipple
		314478	Ovarian fibrothecoma	991	PAGOD syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2802	Pagon-Bird-Detter syndrome	1366	Palmoplantar keratoderma and congenital alopecia, Wallis type		tumor
716	PAH deficiency	34217	Palmoplantar keratoderma with arrhythmogenic cardiomyopathy	97278	Pancreatic polypeptidoma
1993	Pai syndrome	→2199	Palmoplantar keratoderma with tonotubular keratin	424073	Pancreatic serous cystadenocarcinoma
37202	Painful bladder syndrome	140966	Palmoplantar keratoderma, Nagashima type	424065	Pancreatic solid pseudopapillary carcinoma
324636	Painful bruising syndrome	86919	Palmoplantar keratoderma-clinodactyly syndrome	424039	Pancreatic squamous cell carcinoma
99736	Painful congenital myotonia	50944	Palmoplantar keratoderma-cystic eyelids-hypodontia-hypotrichosis syndrome	309031	Pancreatic triacylglycerol lipase deficiency
99736	Painful myotonia	2202	Palmoplantar keratoderma-deafness syndrome	309031	Pancreatic triglyceride lipase deficiency
64686	Painful ophthalmoplegia	2198	Palmoplantar keratoderma-esophageal carcinoma syndrome	424080	Pancreatic undifferentiated carcinoma with osteoclast-like giant cells
300501	Painful orbital and systemic neurofibromas-marfanoid habitus syndrome	2202	Palmoplantar keratoderma-hearing loss syndrome	677	Pancreatoblastoma
90797	PAIS	2342	Palmoplantar keratoderma-periodontopathia-onychogryposis syndrome	317473	Pancytopenia due to IKZF1 mutations
477993	Palatal anomalies-multiple diastemata-facial dysmorphism-developmental delay syndrome	384	Palmoplantar keratoderma-sclerodactyly syndrome	401764	Pancytopenia-developmental delay syndrome
477993	Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	2201	Palmoplantar keratoderma-spastic paralysis syndrome	66624	PANDAS
1388	Palatodigital syndrome, Catel-Manzke type	85112	Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome	95513	Panhypophysitis
163921	PALE	→79502	Palmoplantar porokeratosis of Mantoux	90695	Panhypopituitarism
171695	Pallidopyramidal syndrome	163927	Palmoplantar pustulosis	97336	Panner disease
3138	Pallister ulnar-mammary syndrome	767	PAN	90159	Panniculitis-induced localized lipodystrophy
672	Pallister-Hall syndrome	98815	Panayiotopoulos syndrome	157850	Pantothenate kinase-associated neurodegeneration
884	Pallister-Killian syndrome	424046	Pancreatic acinar cell carcinoma	440427	PAP, Reunion island type
2804	Pallister-W syndrome	93292	Pancreatic adenoma	69126	PAPA syndrome
737	Palmar, plantar and disseminated porokeratosis	65288	Pancreatic and cerebellar agenesis	213817	Papillary carcinoma of the cervix uteri
2184	Palmer-Pagon syndrome	97282	Pancreatic cholera	213726	Papillary carcinoma of the corpus uteri
659	Palmoplantar and periorificial keratoderma	309108	Pancreatic colipase deficiency	251962	Papillary glioneuronal tumor
34217	Palmoplantar hyperkeratosis with arrhythmogenic cardiomyopathy	2255	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome	146	Papillary or follicular thyroid carcinoma
140966	Palmoplantar hyperkeratosis, Nagashima type	811	Pancreatic insufficiency and bone marrow dysfunction	319298	Papillary renal cell adenocarcinoma
50944	Palmoplantar hyperkeratosis-cystic eyelids-hypodontia-hypotrichosis syndrome	199337	Pancreatic insufficiency-anemia-hyperostosis syndrome	319298	Papillary renal cell carcinoma
2202	Palmoplantar hyperkeratosis-deafness syndrome	424058	Pancreatic intraductal papillary mucinous carcinoma	251915	Papillary tumor of the pineal region
2198	Palmoplantar hyperkeratosis-esophageal carcinoma syndrome	506112	Pancreatic MiNEN	1475	Papillo-renal syndrome
2202	Palmoplantar hyperkeratosis-hearing loss syndrome	506112	Pancreatic mixed neuroendocrine-nonneuroendocrine neoplasm	2807	Papilloma of choroid plexus
2342	Palmoplantar hyperkeratosis-periodontopathia-onychogryposis syndrome	424053	Pancreatic mucinous cystadenocarcinoma	2750	Papillon-Léage-Psaume syndrome
384	Palmoplantar hyperkeratosis-sclerodactyly syndrome	506098	Pancreatic NEC	678	Papillon-Lefèvre syndrome
2201	Palmoplantar hyperkeratosis-spastic paralysis syndrome	506098	Pancreatic neuroendocrine carcinoma	86819	Papular atrichia
85112	Palmoplantar hyperkeratosis-XX sex reversal-predisposition to squamous cell carcinoma syndrome	424080	Pancreatic osteoclastic giant cell	228264	Papular elastorrhexis
1010	Palmoplantar keratoderma and congenital alopecia, Stevanovic type			313936	Papular epidermal nevi with skyline basal cell layers syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	sarcoma	→98784	Paroxysmal hypnagogic dystonia	261318	Partial trisomy of the short arm of chromosome 20
324299	Paraganglioma-somatostatinoma-polycythemia syndrome	→98784	Paroxysmal hypnogenic dyskinesia	458785	Partially involuting congenital hemangioma
326	Parahemophilia	98809	Paroxysmal kinesigenic choreathetosis	85453	Partington disease
141242	Paramedian nasal cleft	98809	Paroxysmal kinesigenic dyskinesia	94083	Partington syndrome
684	Paramyotonia congenita	31709	Paroxysmal kinesigenic dyskinesia and infantile convulsions	→193	Partington-Anderson syndrome
684	Paramyotonia congenita of Von Eulenburg	→98784	Paroxysmal nocturnal dyskinesia	94083	Partington-Mulley syndrome
2812	Parana hard skin syndrome	447	Paroxysmal nocturnal hemoglobinuria	295	Parvovirus antenatal infection
99889	Paraneoplastic Cushing syndrome	98810	Paroxysmal non-kinesigenic dyskinesia	1394	Pascual-Castroviejo syndrome type 1
1183	Paraneoplastic opsclonus-myoclonus	98810	Paroxysmic non-kinesigenic choreoathetosis	42775	Pascual-Castroviejo syndrome type 2
1183	Paraneoplastic opsclonus-myoclonus-ataxia syndrome	1214	Parry-Romberg syndrome	289478	PASH syndrome
63455	Paraneoplastic pemphigus	574	Partial 21q monosomy	1252	Pashayan syndrome
71505	Paraneoplastic retinopathy	2805	Partial agenesis of the pancreas	1252	Pashayan-Prozansky syndrome
279928	Paraneoplastic uveitis	381	Partial albinism-immunodeficiency syndrome	2278	Passwell-Goodman-Siprkowski syndrome
231445	Paraparetic variant of GBS	90797	Partial androgen insensitivity syndrome	3378	Patau syndrome
231445	Paraparetic variant of Guillain-Barré syndrome	90797	Partial androgen resistance syndrome	→1509	Patella aplasia-coxa vara-tarsal synostosis syndrome
2824	Paraplegia-intellectual disability-hyperkeratosis syndrome	1330	Partial atrioventricular canal	86789	Patella aplasia/hypoplasia
31827	Paraquat poisoning	1330	Partial atrioventricular canal defect	228190	Patent arterial duct-bicuspid aortic valve-hand anomalies syndrome
2646	Parastremmatic dwarfism	1646	Partial chromosome Y deletion	46627	Patent ductus arteriosus with facial dysmorphism and abnormal fifth digits
363478	Paratesticular adenocarcinoma	401959	Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome	228190	Patent ductus arteriosus-bicuspid aortic valve-hand anomalies syndrome
143	Parathyroid carcinoma	98950	Partial cryptophthalmia	431341	Patent urachus
443227	Paratyphoid fever	90076	Partial deep dermal and full thickness burns	254531	Paternal 14q32.2 hypomethylation syndrome
2825	PARC syndrome	79312	Partial deficiency of methylmalonyl-CoA mutase	254525	Paternal 14q32.2 microdeletion syndrome
268826	Parietal encephalocele	261318	Partial duplication of chromosome 20p	261304	Paternal 20q13.2-q13.3 microdeletion syndrome
251290	Parietal foramina with clavicular hypoplasia	261318	Partial duplication of the short arm of chromosome 20	261304	Paternal 20q13.2q13.3 microdeletion syndrome
251290	Parietal foramina with cleidocranial dysplasia	101046	Partial epilepsy with auditory aura	254525	Paternal del(14)(q32.2)
851	Paris-Trousseau thrombocytopenia	101046	Partial epilepsy with auditory features	261304	Paternal del(20)(q13.2q13.3)
306674	PARK9	2704	Partial facial palsy with urinary abnormalities	254525	Paternal monosomy 14q32.2
199351	PARK14	744	Partial gigantism-nevi-hemihypertrophy-macrocephaly syndrome	261304	Paternal monosomy 20q13.2-q13.3
90307	Parkes Weber syndrome	254693	Partial hydatidiform mole	261304	Paternal monosomy 20q13.2q13.3
171695	Parkinsonian-pyramidal syndrome	79292	Partial LCAT deficiency	251004	Paternal uniparental disomy of chromosome 1
314632	Parkinsonism due to ATP13A2 deficiency	343	Partial mevalonate kinase deficiency	96190	Paternal uniparental disomy of chromosome 5
178509	Parkinsonism with alveolar hypoventilation and mental depression	254693	Partial molar pregnancy	96191	Paternal uniparental disomy of chromosome 6
97355	Parkinsonism with dementia of Guadeloupe	2805	Partial pancreatic agenesis	96192	Paternal uniparental disomy of chromosome 7
90020	Parkinsonism-dementia-ALS complex	180129	Partial septate uterus	99324	Paternal uniparental disomy of chromosome 13
90035	Paroxysmal cold hemoglobinuria	157769	Partial situs inversus	96334	Paternal uniparental disomy of
53583	Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	261318	Partial trisomy of chromosome 20p		
98811	Paroxysmal exertion-induced dyskinesia				
46348	Paroxysmal extreme pain disorder				
157835	Paroxysmal hemicrania				
→98784	Paroxysmal hypnagogic dyskinesia				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	chromosome 14	369920	PCH9	99807	PEHO-like syndrome
96194	Paternal uniparental disomy of chromosome 20	97249	PCH with optic atrophy	48686	PEL
96195	Paternal uniparental disomy of chromosome 21	97249	PCH without dyskinesia	702	Pelizaeus-Merzbacher brain sclerosis
261524	Paternal uniparental disomy of chromosome X	411493	PCH10	702	Pelizaeus-Merzbacher disease
96194	Paternal UPD(20)	71528	PCI deficiency	280229	Pelizaeus-Merzbacher disease in female carriers
2976	Patterson pseudoleprechaunism syndrome	454714	PCL	280210	Pelizaeus-Merzbacher disease type II
2976	Patterson syndrome	2924	PCLD	280219	Pelizaeus-Merzbacher disease, classic form
2439	Patterson-Stevenson syndrome	178536	PCMZL	280210	Pelizaeus-Merzbacher disease, connatal form
2439	Patterson-Stevenson-Fontaine syndrome	438134	PCNA-related progressive neurodegenerative photosensitivity syndrome	280234	Pelizaeus-Merzbacher disease, null syndrome
79136	PATX	46135	PCNSL	280224	Pelizaeus-Merzbacher disease, transitional form
93126	Pauci-immune glomerulonephritis	140989	PCNSV	280270	Pelizaeus-Merzbacher-like disease
97563	Pauci-immune glomerulonephritis with ANCA	101330	PCT	280293	Pelizaeus-Merzbacher-like disease due to AIMP1 mutation
97563	Pauci-immune glomerulonephritis with antineutrophil cytoplasmic antibody	163746	PCWH	280282	Pelizaeus-Merzbacher-like disease due to GJC2 mutation
97564	Pauci-immune glomerulonephritis without ANCA	90020	PDALS	280288	Pelizaeus-Merzbacher-like disease due to HSPD1 mutation
97564	Pauci-immune glomerulonephritis without antineutrophil cytoplasmic antibody	293462	PDCD	97352	Pellagra
85410	Pauciarticular chronic arthritis	289157	PDDRI	→220295	Pellagra-like skin rash-neurological manifestations syndrome
247839	Pauciarticular chronic arthritis with anti-nuclear antibodies	439822	PDE4D haploinsufficiency syndrome	137672	Pellucid marginal degeneration
247846	Pauciarticular chronic arthritis without anti-nuclear antibodies	765	PDH	2840	Pelvic dysplasia-arthrogryposis of lower limbs syndrome
1330	PAVC	79246	PDH phosphatase deficiency	2839	Pelvis-shoulder dysplasia
2038	PAVM	79243	PDHAD	93333	Pelviscapular dysplasia
186	PBC	255138	PDHBD	63275	Pemphigoid gestationis
75373	PBCRA	765	PDHC	79480	Pemphigus erythematosus
289666	PBL	2796	PDP	79481	Pemphigus foliaceus
2309	PC	85453	PDR	79479	Pemphigus vegetans
54247	PCA	75496	PDS	704	Pemphigus vulgaris
88628	PCARP	699	Pearson syndrome	994	Pena-Shokeir syndrome type 1
231426	PCB variant of GBS	2835	Pectus excavatum-macrocephaly-dysplastic nails syndrome	1466	Pena-Shokeir syndrome type 2
231426	PCB variant of Guillain-Barré syndrome	98811	PED	705	Pendred syndrome
247198	PCCA	439175	Pediatric AIS	398053	Penile adenocarcinoma
244	PCD	439175	Pediatric arterial ischemic stroke	49	Penile agenesis
178544	PCDLBCL,LT	66624	Pediatric autoimmune disorders associated with Streptococcus infections	398058	Penile squamous cell carcinoma
178540	PCFCL	66624	Pediatric autoimmune neuropsychiatric disorders associated with Streptococcus infections	49	Penis agenesis
90035	PCH	93682	Pediatric Castleman disease	2842	Penoscrotal transposition
2254	PCH1	487809	Pediatric collagenous gastritis	313936	PENS syndrome
2524	PCH2	33402	Pediatric HCC	11	Penta-X
97249	PCH3	33402	Pediatric hepatocellular carcinoma	1335	Pentalogy of Cantrell
166063	PCH4	477738	Pediatric multiple sclerosis	11	Pentasomy X
166068	PCH5	93552	Pediatric systemic lupus erythematosus	2843	Pentosuria
166073	PCH6	263548	Peeling skin syndrome type A	352447	PEO-myopathy-emaciation syndrome
284339	PCH7	263553	Peeling skin syndrome type B	2905	PEP syndrome
324569	PCH8	263558	Peeling skin syndrome type C	2880	PEPCK deficiency
		444138	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	2576	Perheentupa syndrome
		2836	PEHO syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
767	Periarteritis nodosa		neuroectodermal tumor		syndrome
2847	Pericardial and diaphragmatic defect	99084	Peripheral pulmonary stenosis	2869	Peutz-Jeghers syndrome
2576	Pericardial constriction-growth failure syndrome	97927	Peripheral resistance to thyroid hormones	42642	PFAPA syndrome
2848	Pericarditis-arthropathy-camptodactyly syndrome	168816	Peritoneal cystic mesothelioma	1980	PFBC
137577	Perinatal asphyxia	171676	Periventricular leukomalacia	90042	PFCP
137577	Perinatal hypoxia	98892	Periventricular nodular heterotopia	412206	PFE
313855	Perinatal lethal bent bone dysplasia	2849	Perlman syndrome	710	Pfeiffer syndrome
85212	Perinatal lethal Gaucher disease	438266	PERM	93258	Pfeiffer syndrome type 1
247623	Perinatal lethal hypophosphatasia	99885	Permanent neonatal diabetes mellitus	93259	Pfeiffer syndrome type 2
247623	Perinatal lethal phosphoethanolaminuria	65288	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome	93260	Pfeiffer syndrome type 3
247623	Perinatal lethal Rathburn disease	2850	Perniola-Krajewska-Carnevale syndrome	3224	Pfeiffer-Kapferer syndrome
95706	Perineal, scrotal or penoscrotal hypospadias	2971	Peroxisomal acyl-CoA oxidase deficiency	2921	Pfeiffer-Mayer syndrome
65250	Perineural cyst	93598	Peroxisomal alanine-glyoxylate aminotransferase deficiency	2871	Pfeiffer-Palm-Teller syndrome
342	Periodic disease	2855	Perrault syndrome	2872	Pfeiffer-Singer-Zschiesche syndrome
42642	Periodic fever-aphtous stomatitis-pharyngitis-adenopathy syndrome	75374	PERRS	33577	Pfeiffer-Weber-Christian syndrome
	Periodic fever-infantile enterocolitis-autoinflammatory syndrome	178509	Perry syndrome	2019	PFFD
436166		99120	Persistent eustachian valve	172	PFIC
→682	Periodic paralysis type 3	91495	Persistent fetal vasculature syndrome	79306	PFIC1
397750	Periodic paralysis with later-onset distal motor neuropathy	99076	Persistent fifth aortic arch	79304	PFIC2
397755	Periodic paralysis with transient compartment-like syndrome	91495	Persistent hyperplastic primary vitreous	79305	PFIC3
79136	Periodic vestibulocerebellar ataxia	398147	Persistent idiopathic facial pain	480483	PFIC4
139426	Perioral myoclonia with absences	99109	Persistent left superior caval vein connecting to the left-sided atrium	480476	PFIC5
563	Peripartum cardiomyopathy	99109	Persistent left superior vena cava connecting to the left-sided atrium	91495	PFVS
	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	99109	Persistent left SVC connecting to the left-sided atrium	397937	PGBM1
163746		2856	Persistent Müllerian derivatives	319646	PGM1-CDG
1795	Peripheral dysostosis	2856	Persistent Müllerian duct syndrome	443811	PGM3-CDG
252164	Peripheral fibroblastoma	97341	Persistent placoid maculopathy	443811	PGM3-related congenital disorder of glycosylation
2400	Peripheral motor neuropathy-dysautonomia syndrome	300324	Persistent polyclonal B-cell lymphocytosis	251962	PGNT
84142	Peripheral nerve hyperexcitability	300324	Persistent polyclonal B-cell lymphocytosis with binucleated lymphocytes	1214	PHA
213812	Peripheral neuroectodermal cancer of cervix uteri	2380	Perthes disease	757	PHA2
213630	Peripheral neuroectodermal cancer of the corpus uteri	1489	Pertussis	88938	PHA2A
90120	Peripheral neuropathy and optic atrophy	708	Peters anomaly	88939	PHA2B
171848	Peripheral neuropathy, Fiskerstrand type	709	Peters anomaly with short limb dwarfism	88940	PHA2C
397744	Peripheral neuropathy-myopathy-hoarseness-deafness syndrome	101033	Peters anomaly-cataract syndrome	300525	PHA2D
397744	Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome	708	Peters congenital glaucoma	300530	PHA2E
370348	Peripheral PNET	709	Peters plus syndrome	756	PHA type 1
370348	Peripheral primitive	2776	Petit-Fryns syndrome	42775	PHACE syndrome
		2963	Petty syndrome	209959	Phacoallergic endophthalmitis
		2963	Petty-Laxova-Wiedemann	209959	Phacoanaphylactic uveitis
				209959	Phacoantigenic endophthalmitis
				757	PHAI
				209959	Phako-anaphylactic endophthalmitis
				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

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	type 5	498228	Phyllodes tumor of the prostate	251295	Pigmented paravenous retinochoroidal atrophy
79485	Phakomatosis spilorosea	773	Phytanic-CoA hydroxylase deficiency	66627	Pigmented villonodular synovitis
352636	Phalangeal microgeodic syndrome	2882	Phytosterolemia	280633	PIGN-CDG
352636	Phalangeal osteolysis	→33364	PIBIDS syndrome	369837	PIGT-CDG
171848	PHARC syndrome	505	Piccardi-Lassueur-Little syndrome	480506	PIHL
231426	Pharyngeal-cervical-brachial variant of Guillain-Barré syndrome	2885	Piebald trait-neurologic defects syndrome	169	Pili annulati
231426	Pharyngeal-cervical-brachial weakness	2884	Piebaldism	720	Pili bifurcati
231426	Pharyngo-cervico-brachial variant of GBS	→1263	Piepkorn dysplasia	79492	Pili gemini
231426	Pharyngo-cervico-brachial variant of Guillain-Barré syndrome	487825	Pierpont syndrome	79492	Pili multigemini
2876	PHAVER syndrome	1566	Pierquin syndrome	2889	Pili torti
228410	PHD syndrome	2886	Pierre Robin sequence-congenital heart defect-talipes syndrome	2891	Pili torti-developmental delay-neurological abnormalities syndrome
48652	Phelan-McDermid syndrome	2888	Pierre Robin sequence-faciодigital anomaly syndrome	2890	Pili torti-onychodysplasia syndrome
1919	Phenobarbital embryopathy	3450	Pierre Robin sequence-fetal chondrodysplasia syndrome	1410	Pili trianguli et canaliculi
84064	Phenotypic diarrhea	1388	Pierre Robin sequence-hyperphalangy-clinodactyly syndrome	2741	Pillay syndrome
716	Phenylalanine hydroxylase deficiency	3104	Pierre Robin sequence-oligodactyly syndrome	251612	Pilocytic astrocytoma
716	Phenylketonuria	2886	Pierre Robin syndrome-congenital heart defect-talipes syndrome	2892	Pilodental dysplasia-refractive errors syndrome
226	Phenylketonuria type 2	2888	Pierre Robin syndrome-faciодigital anomaly syndrome	91414	Pilomatrixcoma
2209	Phenylketonuric embryopathy	3450	Pierre Robin syndrome-fetal chondrodysplasia syndrome	499182	Pilomatrix carcinoma
1912	Phenytoin embryofetopathy	1388	Pierre Robin syndrome-hyperphalangy-clinodactyly syndrome	228379	Pilomatrix dysplasia
→168569	PHID	2670	Pierson syndrome	91414	Pilomatrixoma
75508	Phlebectatic osteohypoplastic angiodyplasia	398147	PIFP	251615	Pilomyxoid astrocytoma
69084	PHNED	217557	PIG	2894	Pilotto syndrome
2879	Phocomelia, Schinzel type	99908	Pigeon-breeder lung disease	251919	Pineal parenchymal tumor of intermediate differentiation
2878	Phocomelia-ectrodactyly-deafness-sinus arrhythmia syndrome	488635	PIGG-CDG	251909	Pineoblastoma
3439	Phocomelia-thrombocytopenia-encephalocele-urogenital malformations syndrome	3474	PIGL-CDG	251912	Pineocytoma
534	Phosphatidylinositol 4,5-biphosphate 5-phosphatase deficiency	83639	PIGM-CDG	49382	Pingelapse blindness
2880	Phosphoenolpyruvate carboxykinase deficiency	978	Pigment anomaly-ectrodactyly-hypodontia syndrome	3353	Pinheiro-Freire Maia-Miranda syndrome
436	Phosphoethanolaminuria	999	Pigmentary disorder with hearing loss	247165	Pink disease
→319646	Phosphoglucomutase 1 deficiency	64755	Pigmentary hairy epidermal nevus	155838	Pinnae fistula or cyst
319646	Phosphoglucomutase-1 deficiency	435	Pigmentary mosaicism, Ito type	→2510	Pinsky-Di George-Harley syndrome
35069	Phospholipase A2-associated neurodegeneration	313808	Pigmentary orthochromatic leukodystrophy	279904	PIOL
79318	Phosphomannomutase 2 deficiency	→193	Pigmentary retinopathy-intellectual disability syndrome	→79189	Pipecolic acidemia
79319	Phosphomannose isomerase deficiency	447961	Pigmentation defects-palmoplantar keratoderma-skin carcinoma syndrome	2896	Pitt-Hopkins syndrome
3222	Phosphoribosylpyrophosphate synthetase superactivity	→168569	Pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome	221150	Pitt-Hopkins-like syndrome
284417	Phosphoserine aminotransferase deficiency			→280	Pitt-Rogers-Danks syndrome
166409	Photosensitive epilepsy			93395	Pitt-Williams brachydactyly
91495	PHPV			251623	Pituicytoma
30924	PHSH			95613	Pituitary apoplexy
180261	Phyllodes tumor of the breast			300385	Pituitary carcinoma
				96253	Pituitary corticotroph microadenoma
				91354	Pituitary deficiency due to empty sella turcica syndrome
				91350	Pituitary deficiency due to Rathke's cleft cysts
				91351	Pituitary dermoid and epidermoid cysts
				99725	Pituitary gigantism

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2965	Pituitary lactotrophic adenoma	85166	Platyspondylic dysplasia, Torrance type	352596	PMED
165994	Pituitary resistance to thyroid hormone	85166	Platyspondylic dysplasia, Torrance-Luton type	217260	PML
95496	Pituitary stalk interruption syndrome	85166	Platyspondylic lethal skeletal dysplasia, Torrance type	280270	PMLD
91347	Pituitary thyrotrophic adenoma	2899	Platyspondyly-amelogenesis imperfecta syndrome	280282	PMLD1
96253	Pituitary-dependent Cushing syndrome	300359	PLCG2-associated antibody deficiency and immune dysregulation	79318	PMM2-CDG
2897	Pityriasis rubra pilaris	137810	PLCNA	26790	PMP
1078	Piussan-Lenaerts-Mathieu syndrome	99969	Pleomorphic liposarcoma	476394	PMP2-related Charcot-Marie-Tooth disease type 1
2869	PJS	293199	Pleomorphic rhabdomyosarcoma	476394	PMP2-related Charcot-Marie-Tooth neuropathy type 1
157850	PKAN	454821	Pleomorphic salivary gland adenoma	476394	PMP2-related CMT1
216873	PKAN, atypical form	251607	Pleomorphic xanthoastrocytoma	476394	PMP2-related hereditary motor and sensory neuropathy type 1
216866	PKAN, classic form	449266	Pleural empyema	477817	PMP22-RAI1 contiguous gene duplication syndrome
238455	PKDYS	50251	Pleural mesothelioma	500533	PMSE syndrome
716	PKU	99131	Pleuro-pericardial cyst	99885	PNDM
226	PKU type 2	284343	Pleuro-pulmonary blastoma familial tumor susceptibility syndrome	64741	Pneumoblastoma
477787	PLA2G4A-related platelet dysfunction	64742	Pleuropulmonary blastoma	55655	Pneumococcal meningitis
199351	PLA2G6-related dystonia-parkinsonism	284343	Pleuropulmonary blastoma familial tumor susceptibility syndrome	723	Pneumocystosis
439167	Placental insufficiency	99933	Pleuropulmonary blastoma type 1	90066	Pneumonia caused by Pseudomonas aeruginosa infection
99928	Placental site trophoblastic tumor	99934	Pleuropulmonary blastoma type 2	447	PNH
444138	PLACK syndrome	99935	Pleuropulmonary blastoma type 3	760	PNP deficiency
707	Plague	280356	PLIN1-related familial partial lipodystrophy	760	PNPase deficiency
300359	PLAID	280356	PLIN1-related FPLD	79096	PNPO deficiency
79141	Plamoplantar hyperkeratosis nummularis	2770	PLO-SL	79096	PNPO-related neonatal epileptic encephalopathy
79141	Plamoplantar keratoderma nummularis	2770	PLOSSL	246	POADS
35069	PLAN	2375	Plotz syndrome	2905	POEMS syndrome
199251	Plantar fibromatosis	280234	PLP1 null syndrome	2762	POH
251515	Plantar flexion contracture	678	PLS	2908	Poikiloderma of Kindler
487825	Plantar lipomatosis-facial dysmorphism-developmental delay syndrome	35689	PLS	2909	Poikiloderma of Rothmund-Thomson
487825	Plantar lipomatosis-unusual facies-developmental delay syndrome	99969	PLS	221008	Poikiloderma of Rothmund-Thomson type 1
158769	Plaque-form urticaria pigmentosa	85166	PLSD-T	221016	Poikiloderma of Rothmund-Thomson type 2
454714	Plasma cell leukemia	330015	Plumbism	221046	Poikiloderma with neutropenia
29073	Plasma cell myeloma	54028	Plummer-Vinson syndrome	221046	Poikiloderma with neutropenia, Clericuzio type
329	Plasma thromboplastin antecedent deficiency	732	PM	2825	Poikiloderma-alopecia-retrognathism-cleft palate syndrome
289666	Plasmablastic lymphoma	764	PM	221043	POIKTMP syndrome
86855	Plasmacytoma	454706	PMA	279947	POIS
722	Plasminogen deficiency type 1	702	PMD	130	Pokkuri death syndrome
439881	Plastic bronchitis	2856	PMDS	2911	Poland anomaly
721	Platelet alpha-granule deficiency	308	PME type 1	2911	Poland sequence
477787	Platelet dysfunction due to cytosolic phospholipase-A2 alpha deficiency	501	PME type 2	2911	Poland syndrome
		263516	PME type 3	313808	POLD
		402082	PME type 5	2912	Poliomyelitis
		280620	PME type 6	330009	Poliomyelitis in patients with immunodeficiencies deemed at risk
		435438	PME type 7	→33364	Pollitt syndrome
		424027	PME type 8		
		457265	PME type 9		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
11	Poly-X		IgM monoclonal gammopathy with anti-MAG		bladder
767	Polyarteritis nodosa	2905	Polyneuropathy-endocrinopathy-plasma cell dyscrasia syndrome	213777	Poorly differentiated neuroendocrine carcinoma of the cervix uteri
29207	Polyarthritis enterica	2926	Polyneuropathy-hand defect syndrome	213731	Poorly differentiated neuroendocrine carcinoma of the corpus uteri
85435	Polyarthritis with rheumatoid factor	171848	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	213731	Poorly differentiated neuroendocrine carcinoma of the endometrium
85408	Polyarthritis without rheumatoid factor	2928	Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome	213777	Poorly differentiated neuroendocrine cervical carcinoma
247854	Polyarthritis without rheumatoid factor with anti-nuclear antibodies	93276	Polyostotic fibrous dysplasia	263339	Poorly differentiated thymic neuroendocrine carcinoma
247861	Polyarthritis without rheumatoid factor without anti-nuclear antibodies	160148	Polypoid prolapsing folds	506098	Poorly-differentiated NEN of pancreas
450322	Polyclonal hyperviscosity syndrome	2869	Polyps and spots syndrome	506098	Poorly-differentiated neuroendocrine neoplasm of pancreas
2770	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	208981	Polyradiculoneuropathy associated with IgG/IgA/IgM monoclonal gammopathy without known antibodies	506098	Poorly-differentiated pancreatic NEN
2795	Polycystic ovaries-urethral sphincter dysfunction syndrome	141091	Polyrhinia	506098	Poorly-differentiated pancreatic neuroendocrine neoplasm
729	Polycythemia rubra vera	141091	Polyrrhinia	1300	Popliteal web syndrome
729	Polycythemia vera	93338	Polysyndactyly	95699	POR deficiency
93339	Polydactyly of a biphalangeal thumb	93405	Polysyndactyly, Haas type	666	Porak and Durante disease
93336	Polydactyly of a triphalangeal thumb	2934	Polysyndactyly-cardiac malformation syndrome	95699	PORD
93337	Polydactyly of an index finger	228410	Polyvalvular heart disease syndrome	2940	Porencephaly
2919	Polydactyly postaxial with median cleft of upper lip	139426	POMA	2941	Porencephaly-cerebellar hypoplasia-internal malformations syndrome
2754	Polydactyly-cleft lip/palate-psychomotor retardation syndrome	1183	POMA syndrome	306547	Porencephaly-microcephaly-bilateral congenital cataract syndrome
2917	Polydactyly-myopia syndrome	71526	POMC deficiency	370022	Poretti-Boltshauser syndrome
180229	Polyembryoma	365	Pompe disease	735	Porokeratosis of Mibelli
453533	Polyendocrine-polyneuropathy syndrome	308552	Pompe disease, infantile onset	737	Porokeratosis plantaris palmaris et disseminata
93308	Polyepiphyseal dysplasia type 1	420429	Pompe disease, late-onset	166286	Porokeratotic eccrine nevus
93307	Polyepiphyseal dysplasia type 4	99748	Pontiac fever	166286	Porokeratotic eccrine ostial and dermal duct nevus
93311	Polyepiphyseal dysplasia type 5	477749	Pontine autosomal dominant microangiopathy with leukoencephalopathy	101330	Porphyria cutanea tarda
397937	Polyglucosan body myopathy type 1	269229	Pontine tegmental cap dysplasia	443057	Porphyria cutanea tarda type I
456369	Polyglucosan body myopathy type 2	324569	Pontocerebellar hypoplasia due to CHMP1A mutation	443062	Porphyria cutanea tarda type II
500533	Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	2254	Pontocerebellar hypoplasia type 1	100924	Porphyria due to ALA dehydratase deficiency
180182	Polymastia	2524	Pontocerebellar hypoplasia type 2	100924	Porphyria due to ALAD deficiency
447877	Polymerase proofreading-related adenomatous polyposis	97249	Pontocerebellar hypoplasia type 3	100924	Porphyria due to delta-aminolevulinate dehydratase deficiency
300573	Polymicrogyria due to TUBB2B mutation	166063	Pontocerebellar hypoplasia type 4	100924	Porphyria of Doss
250972	Polymicrogyria with optic nerve hypoplasia	166068	Pontocerebellar hypoplasia type 5	79473	Porphyria variegata
64745	Polymorphic eruption of pregnancy	166073	Pontocerebellar hypoplasia type 6	2703	Port-wine nevi-mega cisterna magna-hydrocephalus syndrome
1243	Polymorphic vitelline macular degeneration	284339	Pontocerebellar hypoplasia type 7	70568	Post-transplant lymphoproliferative disease
93569	Polymyalgia rheumatica	324569	Pontocerebellar hypoplasia type 8		
732	Polymyositis	369920	Pontocerebellar hypoplasia type 9		
639	Polyneuropathy associated with	411493	Pontocerebellar hypoplasia type 10		
		284339	Pontocerebellar hypoplasia-46,XY disorder of sex development syndrome		
		284400	Poorly differentiated neuroendocrine carcinoma of the		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
137839	Postanginal sepsis secondary to orophyngeal infection	238606	POT		camptodactyly syndrome
246	Postaxial acrodyssostosis	→682	Potassium-sensitive normokalemic periodic paralysis	739	Prader-Willi syndrome
246	Postaxial acrofacial dysostosis	640	Potato-grubbing palsy	398069	Prader-Willi syndrome due to a point mutation
93334	Postaxial polydactyly type A	1713	Potocki-Lupski syndrome	177910	Prader-Willi syndrome due to imprinting mutation
93335	Postaxial polydactyly type B	52022	Potocki-Shaffer syndrome		Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15
420584	Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome	3316	Potter sequence-cleft lip/palate-cardiopathy syndrome	98754	Prader-Willi syndrome due to paternal 15q11q13 deletion
2916	Postaxial polydactyly-dental and vertebral anomalies syndrome	217067	Pouchitis	98793	Prader-Willi syndrome due to paternal 15q11q13 deletion
2920	Postaxial polydactyly-intellectual disability syndrome	2876	Powell-Chandra-Saal syndrome	177901	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1
93406	Postaxial syndactyly with metacarpal synostosis	2201	Powell-Venencie-Gordon syndrome		Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2
2730	Postaxial tetramelic oligodactyly	314566	PPAOS	177907	Prader-Willi syndrome due to translocation
263352	Postcardiotomy right ventricular failure	447877	PPAP	398073	Prader-Willi-like syndrome
97349	Postencephalitic parkinsonism	79083	PPARG-related familial partial lipodystrophy	398079	Prader-Willi-like syndrome due to a point mutation
98971	Posterior amorphous corneal dystrophy	79083	PPARG-related FPLD	171829	Prader-Willi-like syndrome due to deletion 6q16
98971	Posterior amorphous stromal dystrophy	284343	PPB familial tumor susceptibility syndrome	2956	Prata-Liberai-Goncalves syndrome
88628	Posterior column ataxia-retinitis pigmentosa syndrome	284343	PPBFTDS	293462	Pre-Descemet corneal dystrophy
54247	Posterior cortical atrophy	300324	PPBL	245	Preaxial acrodyssostosis
2064	Posterior fusion of lumbosacral vertebrae-blepharoptosis syndrome	168829	PPC	2957	Preaxial deficiency-postaxial polydactyly-hypospadias syndrome
95706	Posterior hypospadias	98973	PPCD	93339	Preaxial polydactyly type 1
268810	Posterior meningocele	93339	PPD1	93336	Preaxial polydactyly type 2
98973	Posterior polymorphous corneal dystrophy	93336	PPD2	93337	Preaxial polydactyly type 3
98973	Posterior polymorphous dystrophy	93337	PPD3	93338	Preaxial polydactyly type 4
93110	Posterior urethral valve	93338	PPD4	2921	Preaxial polydactyly-clobomata-intellectual disability syndrome
48435	Postinfectious vasculitis	75567	PPFG	1309	Precalicial canicular ectasia
216452	Postlingual non-syndromic genetic deafness	411696	PPI-REE	99860	Precursor B-cell acute lymphoblastic leukemia
477673	Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	411696	PPI-responsive esophageal eosinophilia	99860	Precursor B-cell acute lymphoblastic leukemia/lymphoma
279947	Postorgasmic illness syndrome	411696	PPIRee	99860	Precursor B-cell acute lymphocytic leukemia
563	Postpartum cardiomyopathy	494	PPK mutilans and deafness	99860	Precursor B-cell acute lymphocytic leukemia/lymphoma
443173	Postpartum psychosis	79141	PPK nummularis	99861	Precursor T-cell acute lymphoblastic leukemia
2942	Postpolio sequelae	86923	PPK, Gamborg-Nielsen type	99861	Precursor T-cell acute lymphoblastic leukemia/lymphoma
2942	Postpolio syndrome	140966	PPK, Nagashima type	99861	Precursor T-cell acute lymphocytic leukemia
2942	Postpoliomylitic syndrome	1010	PPK-CA, Stevanovic type	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
2942	Postpoliomylitis sequelae	1366	PPK-CA, Wallis type	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
2942	Postpoliomylitis syndrome	2202	PPK-deafness syndrome	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
98913	Postsynaptic congenital myasthenic syndromes	79501	PPKP1	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
163921	Posttransplant acute limbic encephalitis	79502	PPKP2	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
443236	Postural tachycardia syndrome due to NET deficiency	38	PPKP3	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		308013	PPKP3 without elastoidosis	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		3077	PPM-X	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		189439	PPNAD	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		370348	PPNET	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		97278	PPoma	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		163927	PPP	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		308013	PPPK3 without elastoidosis	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		79502	PPPP	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		251295	PPRCA	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
		398980	PPSPC	488280	Predisposition to adult-onset myeloproliferative neoplasm due to 14q32 duplication
		324977	PRAAS	457088	Predisposition to invasive fungal
		739	Prader-Labhart-Willi syndrome		
		3409	Prader-Willi habitus-osteopenia-		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	disease due to CARD9 deficiency		systemic scleroderma	90026	Primary erythermalgia
275555	Preeclampsia	314684	Primary bone lymphoma	357220	Primary essential cutis verticis gyrata
69665	Pregnancy-related cholestasis	46135	Primary brain lymphoma	412206	Primary failure of tooth eruption
216445	Prelingual non-syndromic genetic deafness	300865	Primary C-ALCL	98957	Primary familial amyloidosis of the cornea
276432	Premature aging appearance-development delay-cardiac arrhythmia syndrome	267	Primary calpainopathy	90042	Primary familial and congenital polycythemia
363665	Premature aging syndrome, Penttinen type	169464	Primary CD59 deficiency	1980	Primary familial brain calcification
→2512	Premature chromosome condensation with microcephaly and intellectual disability	46135	Primary central nervous system lymphoma	90042	Primary familial polycythemia
95486	Premature closure of the arterial duct	140989	Primary central nervous system vasculitis	3337	Primary Fanconi renotubular syndrome
95486	Premature closure of the patent ductus arteriosus	244	Primary ciliary dyskinesia	3337	Primary Fanconi syndrome
2114	Premature degenerative osteoarthropathy of the hip	→244	Primary ciliary dyskinesia, Kartagener type	633	Primary GH insensitivity
247638	Prenatal benign hypophosphatasia	247522	Primary ciliary dyskinesia-retinitis pigmentosa syndrome	633	Primary GH resistance
247638	Prenatal benign phosphoethanolaminuria	46135	Primary CNS lymphoma	633	Primary growth hormone insensitivity
247638	Prenatal benign Rathburn disease	477781	Primary condylar hyperplasia	633	Primary growth hormone resistance
486811	Prenatal-onset spinal muscular atrophy with congenital bone fractures	90042	Primary congenital erythrocytosis	100085	Primary hepatic neuroendocrine carcinoma
90160	Pressure-induced localized lipoatrophy	98976	Primary congenital glaucoma	480506	Primary hepatolithiasis
98914	Presynaptic congenital myasthenic syndromes	91138	Primary cryoglobulinemia	314950	Primary HES
79410	Pretibial DEB	178528	Primary cutaneous aggressive epidermotropic CD8+ T-cell lymphoma	314950	Primary hypereosinophilic syndrome
79410	Pretibial dystrophic epidermolysis bullosa	300865	Primary cutaneous anaplastic large cell lymphoma	2232	Primary hypergonadotropic hypogonadism-partial alopecia syndrome
2958	Prieto-Badia-Mulas syndrome	178522	Primary cutaneous CD4+ small/medium-sized pleomorphic T-cell lymphoma	682	Primary hyperkalemic periodic paralysis
1451	Prieur-Griselli syndrome	178544	Primary cutaneous diffuse large B-cell lymphoma, leg type	416	Primary hyperoxaluria
945	Primary acalvaria	178528	Primary cutaneous epidermotropic cytotoxic CD8+ T-cell lymphoma	93598	Primary hyperoxaluria type 1
930	Primary achalasia	178540	Primary cutaneous follicle center lymphoma	93599	Primary hyperoxaluria type 2
75564	Primary acquired sideroblastic anemia	178533	Primary cutaneous gamma/delta-positive T-cell lymphoma	93600	Primary hyperoxaluria type 3
85138	Primary Addison's disease	178536	Primary cutaneous marginal zone B-cell lymphoma	682	Primary hyperPP
506334	Primary adrenal insufficiency-steroid-resistant nephrotic syndrome due to SGPL1 deficiency	86885	Primary cutaneous peripheral T-cell lymphoma NOS	33208	Primary hypersomnia
874	Primary adult heart tumor	86885	Primary cutaneous peripheral T-cell lymphoma not otherwise specified	1572	Primary hypogammaglobulinemia
85443	Primary amyloidosis	451602	Primary cutaneous plasmacytosis	30924	Primary hypomagnesemia with secondary hypocalcemia
228272	Primary anetoderma	86885	Primary cutaneous unspecified peripheral T-cell lymphoma	75391	Primary immunodeficiency due to MCM4 deficiency
140989	Primary angiitis of the central nervous system	98807	Primary dystonia with mixed phenotype	90023	Primary immunodeficiency syndrome due to p14 deficiency
1572	Primary antibody deficiency	99657	Primary dystonia, DYT2 type	90023	Primary immunodeficiency syndrome with short stature
2285	Primary basilar invagination	98805	Primary dystonia, DYT4 type	447731	Primary immunodeficiency with multifaceted aberrant lymphoid immunity
189427	Primary bilateral macronodular adrenal hyperplasia	98806	Primary dystonia, DYT6 type	75391	Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency
186	Primary biliary cholangitis	98807	Primary dystonia, DYT13 type	431166	Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection
186	Primary biliary cirrhosis	370103	Primary dystonia, DYT17 type	431166	Primary immunodeficiency with post-MMR vaccine viral infection
779	Primary biliary cirrhosis and	306734	Primary dystonia, DYT21 type		
		464440	Primary dystonia, DYT27 type		
		48686	Primary effusion lymphoma		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
73272	Primary insulin-like growth factor deficiency	168829	Primary peritoneal carcinoma	213630	Primitive neuroectodermal tumor of the corpus uteri
90362	Primary intestinal lymphangiectasia	168829	Primary peritoneal serous carcinoma	854	Primitive portal vein thrombosis
480506	Primary intrahepatic lithiasis	398980	Primary peritoneal serous/papillary carcinoma	3033	Primitive renal tubule syndrome
458768	Primary intralymphatic angioendothelioma	189439	Primary pigmented nodular adrenal dysplasia	2636	Primordial microcephalic dwarfism, Crachami type
279904	Primary intraocular lymphoma	189439	Primary pigmented nodular adrenocortical disease	→2637	Primordial short stature-microdontia-opalescent and rootless teeth syndrome
279904	Primary intraocular non-Hodgkin's lymphoma	100021	Primary plasmacytoma of the bone	3042	Primrose syndrome
140436	Primary intraosseous venous malformation	439737	Primary polyarteritis nodosa	397606	Prion protein systemic amyloidosis
137926	Primary laryngeal lymphangioma	314566	Primary progressive apraxia of speech	412066	PRKAR1B-related neurodegenerative dementia with intermediate filaments
35689	Primary lateral sclerosis	75567	Primary progressive freezing gait	2965	PRL-secreting pituitary adenoma
314709	Primary localized amyloidosis	275766	Primary pulmonary arterial hypertension	2965	PRLoma
137810	Primary localized cutaneous nodular amyloidosis	2257	Primary pulmonary hypoplasia	326	Proaccelerin deficiency
319667	Primary lymphoid conjunctival tumor	2420	Primary pulmonary lymphoma	141099	Proboscis lateralis
319667	Primary lymphoma of the conjunctiva	358	Primary renal tubular hypokalemic hypomagnesemia with hypocaliuria	740	Progeria
228272	Primary macular atrophy	412206	Primary retention of teeth	99706	Progeria-associated arthropathy
168811	Primary malignant peritoneal mesothelioma	171	Primary sclerosing cholangitis	2959	Progeria-short stature-pigmented nevi syndrome
98838	Primary mediastinal clear cell lymphoma of B-cell type	289390	Primary Sjögren syndrome	300382	Progeroid and marfanoid aspect-lipodystrophy syndrome
98838	Primary mediastinal large B-cell lymphoma	289390	Primary Sjögren-Gougerot syndrome	435953	Progeroid features-hepatocellular carcinoma predisposition syndrome
238642	Primary megaureter, adult-onset form	99856	Primary syringomyelia	2962	Progeroid syndrome, De Barsy type
252050	Primary melanoma of the central nervous system	98841	Primary systemic ALCL	2963	Progeroid syndrome, Petty type
252050	Primary melanoma of the CNS	314701	Primary systemic amyloidosis	79094	Progressive arterial occlusive disease-hypertension-heart defects-bone fragility-brachysyndactyly syndrome
54370	Primary membranoproliferative glomerulonephritis	268861	Primary tethered cord syndrome	448251	Progressive autosomal recessive ataxia-deafness syndrome
306558	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome	268861	Primary tethered spinal cord syndrome	448251	Progressive autosomal recessive ataxia-sensorineural hearing loss syndrome
391408	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	99867	Primary thymic epithelial neoplasm	75373	Progressive bifocal chorioretinal atrophy
824	Primary myelofibrosis	263310	Primary thymic epithelial neoplasm type A	→97229	Progressive bulbar palsy of childhood
357225	Primary non-essential cutis verticis gyrata	263324	Primary thymic epithelial neoplasm type AB	→97229	Progressive bulbar paralysis of childhood
289356	Primary non-gestational choriocarcinoma of ovary	263317	Primary thymic epithelial neoplasm type B	139447	Progressive cavitating leukoencephalopathy
289356	Primary non-gestational ovarian choriocarcinoma	99867	Primary thymic epithelial tumor	79087	Progressive cephalothoracic lipodystrophy
279897	Primary oculocerebral lymphoma	263310	Primary thymic epithelial tumor type A	247198	Progressive cerebello-cerebral atrophy
279897	Primary oculocerebral non-Hodgkin lymphoma	263324	Primary thymic epithelial tumor type AB	1871	Progressive cone dystrophy
238606	Primary orthostatic tremor	263317	Primary thymic epithelial tumor type B	220393	Progressive cutaneous systemic scleroderma
439737	Primary PAN	98807	Primary torsion dystonia with predominant craniocervical or upper limb onset	220393	Progressive cutaneous systemic sclerosis
99878	Primary parathyroid hyperplasia	231580	Primary unilateral adrenal hyperplasia	3235	Progressive deafness with stapes fixation
875	Primary pediatric heart tumor	140989	Primary vasculitis of the central nervous system		
439737	Primary periarteritis nodosa	213812	Primitive neuroectodermal tumor of the cervix uteri		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
216812	Progressive deforming osteogenesis imperfecta		to CERS1 deficiency		vertebral fusion
1328	Progressive diaphyseal dysplasia	263516	Progressive myoclonic epilepsy due to KCTD7 deficiency	2762	Progressive osseous heteroplasia
495	Progressive diffuse palmoplantar keratoderma	435438	Progressive myoclonic epilepsy due to KV3.1 deficiency	3322	Progressive pancytopenia-immunodeficiency-cerebellar hypoplasia syndrome
495	Progressive diffuse PPK	457265	Progressive myoclonic epilepsy due to LMNB2 deficiency	217396	Progressive polyneuropathy with bilateral striatal necrosis
438266	Progressive encephalomylitis with rigidity and myoclonus	308	Progressive myoclonic epilepsy type 1	1159	Progressive pseudorheumatoid arthropathy of childhood
2836	Progressive encephalopathy with edema, hypersarrhythmia and optic atrophy	501	Progressive myoclonic epilepsy type 2	352718	Progressive retinal dystrophy due to retinol transport defect
431361	Progressive encephalopathy with leukodystrophy due to DECR deficiency	263516	Progressive myoclonic epilepsy type 3	447977	Progressive scapulohumeroperoneal distal myopathy
99852	Progressive encephalopathy with severe infantile anorexia	163696	Progressive myoclonic epilepsy type 4	228012	Progressive sensorineural deafness-hypertrophic cardiomyopathy syndrome
2836	Progressive encephalopathy-optic atrophy syndrome	402082	Progressive myoclonic epilepsy type 5	228012	Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome
1947	Progressive epilepsy-intellectual disability syndrome, Finnish type	280620	Progressive myoclonic epilepsy type 6		Progressive spondyloepimetaphyseal dysplasia-short stature-short fourth metatarsals-intellectual disability syndrome
457212	Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	435438	Progressive myoclonic epilepsy type 7	683	Progressive supranuclear palsy
2744	Progressive external ophthalmoplegia and scoliosis	424027	Progressive myoclonic epilepsy type 8	240112	Progressive supranuclear palsy-apraxia of speech syndrome
352447	Progressive external ophthalmoplegia-myopathy-emaciation syndrome	457265	Progressive myoclonic epilepsy type 9	240103	Progressive supranuclear palsy-corticobasal syndrome
1214	Progressive facial hemiatrophy	352596	Progressive myoclonic epilepsy with dystonia	240085	Progressive supranuclear palsy-parkinsonism syndrome
172	Progressive familial intrahepatic cholestasis	308	Progressive myoclonus epilepsy type 1	240112	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome
79306	Progressive familial intrahepatic cholestasis type 1	501	Progressive myoclonus epilepsy type 2	240094	Progressive supranuclear palsy-pure akinesia with gait freezing syndrome
79304	Progressive familial intrahepatic cholestasis type 2	263516	Progressive myoclonus epilepsy type 3	316	Progressive symmetric erythrokeratoderma
79305	Progressive familial intrahepatic cholestasis type 3	402082	Progressive myoclonus epilepsy type 5	316	Progressive symmetric erythrokeratoderma, Gottron type
480483	Progressive familial intrahepatic cholestasis type 4	280620	Progressive myoclonus epilepsy type 6	2965	Prolactin-secreting pituitary adenoma
480476	Progressive familial intrahepatic cholestasis type 5	435438	Progressive myoclonus epilepsy type 7	2965	Prolactinoma
75327	Progressive foveal dystrophy	424027	Progressive myoclonus epilepsy type 8	742	Prolidase deficiency
1214	Progressive hemifacial atrophy	457265	Progressive myoclonus epilepsy type 9	492	Proliferating trichilemmal cyst
199282	Progressive isolated segmental anhidrosis	352596	Progressive myoclonus epilepsy with dystonia	86872	Proliferation of large granular lymphocytes
73	Progressive massive osteolysis	726	Progressive neuronal degeneration of childhood with liver disease	221126	Proliferative vasculopathy and hydranencephaly/hydrocephaly
477814	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome	228012	Progressive neurosensory deafness-hypertrophic cardiomyopathy syndrome	419	Proline oxidase deficiency
217260	Progressive multifocal leukoencephalitis	228012	Progressive neurosensory hearing loss-hypertrophic cardiomyopathy syndrome	75374	Prolonged electroretinal response suppression
217260	Progressive multifocal leukoencephalopathy	158022	Progressive nodular histiocytosis	300878	Prolymphocytic variant of hairy cell leukemia
454706	Progressive muscular atrophy	100070	Progressive non-fluent aphasia		
424027	Progressive myoclonic epilepsy due	2062	Progressive non-infectious anterior		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
300878	Prolymphocytic variant of HCL		mellitus-cerebellar ataxia syndrome	2976	Pseudoleprechaunism syndrome, Patterson type
2083	Prominent glabella-microcephaly-hypogenitalism syndrome	397606	PrP systemic amyloidosis	26790	Pseudomyxoma peritonei
2966	Properdin deficiency	3222	PRPP synthetase superactivity	251962	Pseudopapillary ganglioglioneurocytoma
35	Propionic acidemia	3222	PRPS1 superactivity	251962	Pseudopapillary neurocytoma with glial differentiation
35	Propionic aciduria	47159	pRTA	2980	Pseudopapilledema-blepharophimosis-hand anomalies syndrome
35	Propionyl-CoA carboxylase deficiency	165994	PRTH	129	Pseudopelade of Brocq
485358	Propylthiouracil embryofetopathy	2970	Prune belly syndrome	2985	Pseudoprogeria syndrome
485358	Propylthiouracil embryopathy	89843	Pruriginous dystrophic epidermolysis bullosa	79445	Pseudopseudohypoparathyroidism
324977	Proteasome disability syndrome	64745	Puritic urticarial papules and plaques of pregnancy	477742	Pseudosarcomatous fasciitis
324977	Proteasome-associated autoinflammatory syndrome	284417	PSAT deficiency	477742	Pseudosarcomatous fibromatosis
213	Protein defect of cystin transport	171	PSC	3103	Pseudothalidomide syndrome
26349	Protein S acquired deficiency	228402	Pseudo-Angelman syndrome	238624	Pseudotumor cerebri
744	Proteus syndrome	99000	Pseudo-Best disease	83316	Pseudotyphus of California
2969	Proteus-like syndrome	314459	Pseudo-Demons-Meigs syndrome	180079	Pseudounicornuate uterus
325	Prothrombin deficiency	577	Pseudo-Hurler polydystrophy	753	Pseudovaginal perineoscrotal hypospadias
411696	Proton-pump inhibitor-responsive esophageal eosinophilia	314459	Pseudo-Meigs syndrome	289157	Pseudovitamin D-deficient rickets
251598	Protoplasmic astrocytoma	439881	Pseudo-membranous bronchitis	758	Pseudoxanthoma elasticum
79473	Protoporphyrinogen oxidase deficiency	263482	Pseudo-Morquio syndrome type 2	228293	Pseudoxanthoma elasticum-like papillary dermal elastolysis
2508	Proud syndrome	2971	Pseudo-NALD	436274	Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa
2508	Proud-Levine-Carpenter syndrome	2971	Pseudo-neonatal adrenoleukodystrophy	91135	Pseudoxanthoma elasticum-like syndrome
52022	Proximal 11p deletion syndrome	1229	Pseudo-TORCH syndrome	228227	Pseudoxanthoma-like late-onset focal dermal elastosis
261197	Proximal 16p11.2 microdeletion syndrome	2166	Pseudo-trisomy 13 syndrome	280794	Pseudoxanthomatous DCM
370079	Proximal 16p11.2 microduplication syndrome	99000	Pseudo-vitelliform macular dystrophy	280794	Pseudoxanthomatous diffuse cutaneous mastocytosis
502437	Proximal del(4)(q25)	52530	Pseudo-von Willebrand disease	95496	PSIS
261197	Proximal del(16)(p11.2)	52530	Pseudo-von Willebrand disease type 2B	85436	Psoriasis-related JIA
370079	Proximal dup(16)(p11.2)	→300	Pseudo-Zellweger syndrome	85436	Psoriasis-related juvenile idiopathic arthritis
502437	Proximal monosomy 4q25	750	Pseudoachondroplasia	683	PSP syndrome
261197	Proximal monosomy 16p11.2	750	Pseudoachondroplastic dysplasia	240112	PSP-AOS
401768	Proximal myopathy with extrapyramidal signs	750	Pseudoachondroplastic spondyloepiphyseal dysplasia	240103	PSP-CBS
606	Proximal myotonic dystrophy	2971	Pseudoadrenoleukodystrophy	240103	PSP-corticobasal syndrome
606	Proximal myotonic myopathy	526	Pseudoaldosteronism	240085	PSP-p
47159	Proximal renal tubular acidosis	221120	Pseudoaminopterin syndrome	240094	PSP-PAGF
93607	Proximal renal tubular acidosis with ocular abnormalities and intellectual disability	85174	Pseudodiastrophic dysplasia	240085	PSP-parkinsonism
70	Proximal spinal muscular atrophy	2983	Pseudohermaphroditism-intellectual disability syndrome	240112	PSP-PNFA
83330	Proximal spinal muscular atrophy type 1	526	Pseudohyperaldosteronism type 1	240094	PSP-pure akinesia with gait freezing
83418	Proximal spinal muscular atrophy type 2	88660	Pseudohyperaldosteronism type 2	263548	PSS type A
83419	Proximal spinal muscular atrophy type 3	756	Pseudohypoaldosteronism type 1	263553	PSS type B
83420	Proximal spinal muscular atrophy type 4	757	Pseudohypoaldosteronism type 2	99928	PSST
3250	Proximal symphalangism	88938	Pseudohypoaldosteronism type 2A	71519	Psychogenic dystonia
370079	Proximal trisomy 16p11.2	88939	Pseudohypoaldosteronism type 2B	71519	Psychogenic movement disorders
3390	Proximal tubulopathy-diabetes	88940	Pseudohypoaldosteronism type 2C	324636	Psychogenic purpura
		300525	Pseudohypoaldosteronism type 2D	505242	Psychomotor regression-
		300530	Pseudohypoaldosteronism type 2E		
		79443	Pseudohypoparathyroidism type 1A		
		94089	Pseudohypoparathyroidism type 1B		
		79444	Pseudohypoparathyroidism type 1C		
		94090	Pseudohypoparathyroidism type 2		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	oculomotor apraxia-movement disorder-nephropathy syndrome	1207	Pulmonary atresia with ventricular septal defect		hypotonia-seizures-encephalopathy syndrome
88618	Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	1208	Pulmonary atresia-intact ventricular septum syndrome	438216	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation
52530	PT-VWD	64741	Pulmonary blastoma	231625	Pure aldosterone-producing adrenocortical carcinoma
329	PTA deficiency	99084	Pulmonary branch stenosis	231625	Pure aldosterone-secreting adrenocortical carcinoma
247698	PTC syndrome	199241	Pulmonary capillary hemangiomatosis	231625	Pure APAC
97290	PTC-RCC	210136	Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome	441	Pure autonomic failure
269229	PTCD	217080	Pulmonary fungal infections in patients deemed at risk	441	Pure dysautonomia
1578	Pterin-4 alpha-carbinolamine dehydratase deficiency	99874	Pulmonary histiocytosis X	319465	Pure familial acute myeloid leukemia
2988	Pterygium colli-intellectual disability-digital anomalies syndrome	991	Pulmonary hypoplasia-agonadism-dextrocardia-diaphragmatic hernia syndrome	319465	Pure familial AML
86789	PTLAH	217557	Pulmonary interstitial glycogenosis	69084	Pure hair and nail ectodermal dysplasia
70568	PTLD	2414	Pulmonary lymphangiomatosis	441	Pure idiopathic dysautonomia
2999	Ptosis-strabismus-ectopic pupils syndrome	60026	Pulmonary nodular lymphoid hyperplasia	475	Pure Joubert syndrome
→293843	Ptosis-strabismus-rectus abdominis diastasis syndrome	411703	Pulmonary non-tuberculous mycobacterial infection	254854	Pure mitochondrial myopathy
238766	Ptosis-syndactyly-learning difficulties syndrome	60026	Pulmonary pseudolymphoma	2028	Puretic syndrome
228396	Ptosis-upper ocular movement limitation-absence of lacrimal punctum syndrome	99048	Pulmonary valve agenesis-intact ventricular septum-persistent ductus arteriosus syndrome	760	Purine nucleoside phosphorylase deficiency
2997	Ptosis-vocal cord paralysis syndrome	101206	Pulmonary valve agenesis-tetralogy of Fallot-absence of ductus arteriosus syndrome	761	Purpura rheumatica
251915	PTPR	31837	Pulmonary venoocclusive disease	2442	Purtilo syndrome
485358	PTU embryofetopathy	→636	Pulmonic stenosis with 'café-au-lait' spots	293173	Pustular drug eruption
485358	PTU embryopathy	85202	Pulmonic stenosis-brachytelephalangism-calcification of cartilages syndrome	163927	Pustulosis palmaris et plantaris
231580	PUAH	98984	Pulverulent cataract	48377	Pustulosis subcornealis
60039	Pudendal algia	97353	Punch-drunk syndrome	93110	PUV
60039	Pudendal nerve entrapment syndrome	79502	Punctate palmoplantar hyperkeratosis type 2	729	PV
60039	Pudendal neuralgia	38	Punctate palmoplantar hyperkeratosis type 3	101206	PVA/ADA, Fallot type
60039	Pudendal neuralgia by pudendal nerve entrapment	308013	Punctate palmoplantar hyperkeratosis type 3 without elastoidosis	99048	PVA/PDA, non-Fallot type
60039	Pudendalgia	79501	Punctate palmoplantar keratoderma type 1	398069	PWS due to a point mutation
443173	Puerperal psychosis	79502	Punctate palmoplantar keratoderma type 2	398073	PWS-like
984	Pulmonary agenesis	38	Punctate palmoplantar keratoderma type 3	398079	PWS-like due to a point mutation
60025	Pulmonary alveolar microlithiasis	308013	Punctate palmoplantar hyperkeratosis type 3 without elastoidosis	251607	PXA
440427	Pulmonary alveolar proteinosis, Reunion island type	79501	Punctate palmoplantar keratoderma type 1	758	PXE
247257	Pulmonary anthrax	79502	Punctate palmoplantar keratoderma type 2	228227	PXE-like late-onset focal dermal elastosis
→331176	Pulmonary arterial hypertension-leukopenia-atrial septal defect syndrome	38	Punctate palmoplantar keratoderma type 3	228293	PXE-like papillary dermal elastolysis
2038	Pulmonary arteriovenous malformation	308013	Punctate palmoplantar keratoderma type 3 without elastoidosis	91135	PXE-like syndrome
99049	Pulmonary artery coming from patent ductus arteriosus	64745	PUPPP	436274	PXE-like syndrome with retinitis pigmentosa
99050	Pulmonary artery coming from the aorta	438213	PURA-related severe neonatal	763	Pycnodysostosis
99083	Pulmonary artery hypoplasia			293633	PYCR1 deficiency
				293633	PYCR1-related De Barys syndrome
				481152	PYCR2-related microcephaly-progressive leukoencephalopathy
				3003	Pyknochondrogenesis
				763	Pyknodysostosis
				64280	Pyknolepsy
				3005	Pyle disease
				48104	Pyoderma gangrenosum
				289478	Pyoderma gangrenosum-acne-suppurative hidradenitis syndrome

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
69126	Pyogenic arthritis-pyoderma gangrenosum-acne syndrome	79246	Pyruvate dehydrogenase phosphatase deficiency		amegakaryocytic thrombocytopenia syndrome
183713	Pyogenic bacterial infections due to MyD88 deficiency	255182	Pyruvate dehydrogenase protein X component deficiency	→193	Radio-ulnar synostosis-retinal pigment abnormalities syndrome
764	Pyomyositis	766	Pyruvate kinase deficiency of erythrocytes	294979	Radio-ulnar terminal transverse meromelia
2561	Pyramidal molar-glucoma-upper abnormal lip syndrome	781	Q fever	420741	Radiosensitivity-immunodeficiency-dysmorphic features-learning difficulties syndrome
63440	Pyrgocephaly	3010	Qazi-Markouizos syndrome	3269	Radioulnar fusion
79096	Pyridoxal phosphate-dependent seizures	602	Quadriceps-sparing myopathy	3270	Radioulnar synostosis-developmental delay-hypotonia syndrome
79096	Pyridoxal phosphate-responsive seizures	781	Quadrilateral fever	3268	Radioulnar synostosis-microcephaly-scoliosis syndrome
79096	Pyridoxamine 5'-oxidase deficiency	9	Quadruple X	100057	RAE
79096	Pyridoxamine 5'-phosphate oxidase deficiency	84142	Quantal squander syndrome	100019	RAEB-1
3006	Pyridoxine-dependent epilepsy	869	Quaternary A syndrome	100020	RAEB-2
32	Pyroglutamicaciduria	220436	Quebec platelet disorder	168960	RAEB-t
293633	Pyrrolidine-5-carboxylate reductase 1 deficiency	781	Query fever	1832	Raine syndrome
3008	Pyruvate carboxylase deficiency	137888	Question mark ear syndrome	50811	Rajab-Spranger syndrome
353308	Pyruvate carboxylase deficiency type A	346	Quinquaud's folliculitis decalvans	178307	RAK
353314	Pyruvate carboxylase deficiency type B	1437	r(1) syndrome	268114	RALD
353320	Pyruvate carboxylase deficiency type C	1447	r(4) syndrome	99843	Rambam-Hasharon syndrome
353320	Pyruvate carboxylase deficiency, benign type	1450	r(8) syndrome	3018	Rambaud-Gallian syndrome
353308	Pyruvate carboxylase deficiency, infantile type	96175	r(11) syndrome	3018	Rambaud-Gallian-Touchard syndrome
353314	Pyruvate carboxylase deficiency, severe neonatal type	1446	r(22) syndrome	3019	Ramon syndrome
79243	Pyruvate decarboxylase deficiency	261529	r(Y)	1051	Ramos-Arroyo syndrome
79244	Pyruvate dehydrogenase complex component E2 deficiency	100057	RAAS-blocker-induced angioedema	3020	Ramsay Hunt syndrome
255182	Pyruvate dehydrogenase complex component E3 deficiency	100057	RAAS-blocker-induced angioneurotic edema	86861	Randall disease
765	Pyruvate dehydrogenase complex deficiency	770	Rabies	3021	RAPADILINO syndrome
79243	Pyruvate dehydrogenase complex E1 component subunit alpha deficiency	769	Rabson-Mendenhall syndrome	293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome
255138	Pyruvate dehydrogenase complex E1 component subunit beta deficiency	240760	RAD50 deficiency	293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation-neural tumors syndrome
765	Pyruvate dehydrogenase deficiency	93321	Radial clubhand	71517	Rapid-onset dystonia-parkinsonism
79243	Pyruvate dehydrogenase E1-alpha deficiency	1121	Radial deficiency-tibial hypoplasia syndrome	141184	Rapidly involuting congenital hemangioma
255138	Pyruvate dehydrogenase E1-beta deficiency	93321	Radial hemimelia	280569	Rapidly progressive glomerulonephritis
79244	Pyruvate dehydrogenase E2 deficiency	2252	Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome	→1071	Rapp-Hodgkin syndrome
2394	Pyruvate dehydrogenase E3 deficiency	93321	Radial longitudinal meromelia	213528	Rare adenocarcinoma of the breast
255182	Pyruvate dehydrogenase E3-binding protein deficiency	93321	Radial ray agenesis	98345	Rare idiopathic male infertility
		2307	Radial ray defects, hearing impairment, external ophthalmoplegia, and thrombocytopenia	98619	Rare isolated myopia
		3026	Radial ray hypoplasia-choanal atresia syndrome	101685	Rare non-syndromic intellectual disability
		90021	Radiation myelitis	101685	Rare NSID
		70475	Radiation proctitis	213574	Rare variants of adenocarcinoma of the corpus uteri
		99789	Radicular dentin dysplasia		
		→2712	Radiculomegaly of canine teeth-congenital cataract		
		3015	Radio-renal syndrome		
		295219	Radio-ulnar fusion, bilateral		
		295217	Radio-ulnar fusion, unilateral		
		295219	Radio-ulnar synostosis, bilateral		
		295217	Radio-ulnar synostosis, unilateral		
		71289	Radio-ulnar synostosis-		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
75564	RARS		syndrome		blasts in transformation
438114	RARS-related autosomal recessive hypomyelinating leukodystrophy	461	Recessive X-linked ichthyosis	100019	Refractory anemia with excess blasts type 1
268114	RAS-associated autoimmune leukoproliferative disease	281090	Recessive X-linked ichthyosis with extracutaneous manifestations	100020	Refractory anemia with excess blasts type 2
1929	Rasmussen subacute encephalitis	96167	Recombinant 8 syndrome	75564	Refractory anemia with ringed sideroblasts
1929	Rasmussen syndrome	96167	Recombinant chromosome 8 syndrome	398063	Refractory CD
3023	Rasmussen-Johnsen-Thomsen syndrome	99990	Recrudescence typhus	398063	Refractory celiac disease
31205	Rat-bite fever	171220	Rectal duplication	398063	Refractory sprue
436	Rathburn disease	100081	Rectal NET	773	Refsum disease
99852	Ravine syndrome	100081	Rectal neuroendocrine tumor	1525	Reginato-Schiapachasse syndrome
2840	Ray-Peterson-Scott syndrome	424002	Rectal squamous cell carcinoma	1433	Regional choroidal atrophy and alopecia
79127	RB-ILD	51890	Rectus abdominis syndrome	83450	Regional odontodysplasia
98961	RBCD	88619	Recurrent acute necrotizing encephalopathy	300865	Regressive atypical histiocytosis
96175	RC11	64740	Recurrent acute pancreatitis	1040	Regressive metaphyseal dysplasia
93111	RCAD syndrome	2672	Recurrent encephalopathy of childhood	448267	Regressive spondylometaphyseal dysplasia
177	RCDP	90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients	2634	Reinhardt-Pfeiffer mesomelic dysplasia
284388	RCVS	293381	Recurrent hereditary corneal erosions	2634	Reinhardt-Pfeiffer syndrome
79408	RDEB generalisata gravis	499103	Recurrent idiopathic neuroretinitis	98961	Reis-Bücklers corneal dystrophy
89842	RDEB generalisata mitis	169142	Recurrent infection due to specific granule deficiency	29207	Reiter disease
89841	RDEB, centripetalis	183675	Recurrent infections associated with rare immunoglobulin isotypes deficiency	29207	Reiter syndrome
89842	RDEB, generalized intermediate	251523	Recurrent infections-inflammatory syndrome due to zinc metabolism disorder syndrome	99991	Relapsing epidemic typhus
79408	RDEB, Hallopeau-Siemens type	69665	Recurrent intrahepatic cholestasis of pregnancy	33577	Relapsing febrile nodular nonsuppurative panniculitis
89842	RDEB, non-Hallopeau-Siemens type	480864	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome	33577	Relapsing febrile nodular panniculitis
89841	RDEB-Ce	169467	Recurrent Neisseria infections due to factor D deficiency	91547	Relapsing fever
89842	RDEB-generalized other	60032	Recurrent respiratory papillomatosis	728	Relapsing polychondritis
79409	RDEB-I	199267	Recurring digital fibrous tumor of childhood	412	Remnant disease
89842	RDEB-O	79433	Red oculocutaneous albinism	217330	REN-associated familial juvenile hyperuricemic nephropathy
79408	RDEB-sev gen	231031	Red palms disease	217330	REN-associated FJHN
85445	Reactive amyloidosis	838	RED-M	217330	REN-associated kidney disease
29207	Reactive arthritis	97239	Reducing body myopathy	217330	REN-related autosomal dominant tubulointerstitial kidney disease
314962	Reactive hypereosinophilic syndrome	523	Reed syndrome	411709	Renal agenesis
166433	Reading seizures	3221	Refetoff syndrome	1848	Renal agenesis, bilateral
857	REAR syndrome	99995	Reflex sympathetic dystrophy	93100	Renal agenesis, unilateral
1188	Reardon-Baraitser syndrome	98826	Refractory anemia	2838	Renal caliceal diverticuli-deafness syndrome
2631	Reardon-Hall-Slaney syndrome	86839	Refractory anemia with excess blasts	319314	Renal cell carcinoma after neuroblastoma
96167	Rec8 syndrome	168960	Refractory anemia with excess	319314	Renal cell carcinoma associated with neuroblastoma
96167	Rec(8) syndrome			1475	Renal coloboma syndrome
1115	Recessive aplasia cutis congenita of limbs			93111	Renal cysts and diabetes syndrome
79409	Recessive dystrophic epidermolysis bullosa inversa			93111	Renal cysts-maturity-onset diabetes of the young syndrome
89842	Recessive dystrophic epidermolysis bullosa, generalized intermediate			93111	Renal dysfunction-early-onset diabetes syndrome
89842	Recessive dystrophic epidermolysis bullosa, non-Hallopeau-Siemens type			93108	Renal dysplasia
89842	Recessive dystrophic epidermolysis bullosa-generalized other				
280384	Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome				
94125	Recessive mitochondrial ataxia				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
93173	Renal dysplasia, bilateral	3242	Renpenning syndrome		intellectual disability due to del(X)(p11.3)
93172	Renal dysplasia, unilateral	494344	RERE-related neurodevelopmental syndrome	85332	Retinitis pigmentosa and intellectual disability due to monosomy Xp11.3
3404	Renal dysplasia-limb defects syndrome	73273	Resistance to IGF-1	85332	Retinitis pigmentosa and intellectual disability due to Xp11.3 microdeletion
→1768	Renal dysplasia-megalocystis-sirenomelia syndrome	424	Resistance to thyroid stimulating hormone	886	Retinitis pigmentosa-deafness syndrome
3404	Renal dysplasia-mesomelia-radiohumeral fusion syndrome	99832	Resistance to thyrotropin-releasing hormone syndrome	494439	Retinitis pigmentosa-deafness-premature aging-short stature-facial dysmorphism syndrome
3156	Renal dysplasia-retinal aplasia syndrome	247257	Respiratory anthrax	494439	Retinitis pigmentosa-hearing loss-premature aging-short stature-facial dysmorphism syndrome
140969	Renal dysplasia-retinal pigmentary dystrophy-cerebellar ataxia-skeletal dysplasia syndrome	247257	Respiratory anthrax disease	140976	Retinitis pigmentosa-hypopituitarism-nephronophthisis-skeletal dysplasia syndrome
654	Renal embryonic tumor	79127	Respiratory bronchiolitis-interstitial lung disease syndrome	3085	Retinitis pigmentosa-intellectual disability-labyrinthine deafness-hypogenitalism syndrome
1652	Renal Fanconi syndrome with nephrocalcinosis and renal stones	1662	Restrictive dermopathy	3085	Retinitis pigmentosa-intellectual disability-deafness-hypogenitalism syndrome
34528	Renal hypomagnesemia type 2	33355	Reticular dysgenesis	3085	Retinitis pigmentosa-intellectual disability-sensorineural hearing loss-hypogenitalism syndrome
31043	Renal hypomagnesemia type 3	99002	Reticular dystrophy of the retinal pigment epithelium	436245	Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome
93101	Renal hypoplasia	100000	Reticular perineurioma	52427	Retinitis punctata albescens
97362	Renal hypoplasia, bilateral	79145	Reticular pigment anomaly of flexures	790	Retinoblastoma
97361	Renal hypoplasia, unilateral	178307	Reticulate acropigmentation of Kitamura	838	Retinocochleocerebral vasculopathy
319319	Renal medullary carcinoma	86900	Reticulum cell sarcoma	3087	Retinohepatoendocrinologic syndrome
71273	Renal nutcracker syndrome	458763	Retiform hemangioendothelioma	2305	Retinoic acid embryopathy
171871	Renal pseudohypoaldosteronism type 1	284247	Retinal arterial macroaneurysm and supravalvular pulmonic stenosis	40366	Retinoid embryopathy
18	Renal tubular acidosis type 1	75326	Retinal arterial tortuosity	2305	Retinoids embryopathy
47159	Renal tubular acidosis type 2	75326	Retinal arteriolar tortuosity	352718	Retinol dystrophy-iris coloboma-comedogenic acne syndrome
2785	Renal tubular acidosis type 3	71213	Retinal capillary malformation	90050	Retinopathy of prematurity
3033	Renal tubular dysgenesis	1574	Retinal degeneration-nanophthalmos-glaucoma syndrome	139455	Retinopathy, Burgess-Black type
97367	Renal tubular dysgenesis due to twin-twin transfusion	1571	Retinal detachment-occipital encephalocele syndrome	3088	Retinopathy-anemia-central nervous system anomalies syndrome
97369	Renal tubular dysgenesis of genetic origin	397758	Retinal dystrophy with inner nuclear layer and ganglion cell anomalies	838	Retinopathy-encephalopathy-deafness associated with microangiopathy
112	Renal tubular normotensive hypokalemic alkalosis with hypercalciuria	397758	Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies	53540	Retinoschisis with early nyctalopia
254902	Renal tubulopathy-encephalopathy-liver failure syndrome	436245	Retinal dystrophy-juvenile cataract-short stature syndrome	269200	Retrocerebellar arachnoid cyst
857	Renal-ear-anal-radial syndrome	75326	Retinal hemorrhage with vascular tortuosity	269200	Retrocerebellar cyst
1092	Renal-genital-middle ear anomalies	3018	Retinal ischemic syndrome-digestive tract small vessel hyalinosis-diffuse cerebral calcifications syndrome	90050	Retrolental fibroplasia
294415	Renal-hepatic-pancreatic dysplasia	319640	Retinal macular dystrophy type 2	778	Rett syndrome
3032	Renal-hepatic-pancreatic dysplasia-Dandy-Walker cysts syndrome	247691	Retinal vasculopathy and cerebral leukoencephalopathy	3095	Rett syndrome variant
774	Rendu-Osler disease	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations		
774	Rendu-Osler-Weber disease	353356	Retinal vasoproliferative tumor		
93975	Renier-Gabreels-Jasper syndrome	791	Retinitis pigmentosa		
100057	Renin-angiotensin-aldosterone system-blocker-induced angioedema	85332	Retinitis pigmentosa and		
100057	Renin-angiotensin-aldosterone system-blocker-induced angioneurotic edema				
294415	Renohepaticpancreatic dysplasia				
3033	Renotubular dysgenesis				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
294049	Reunion Island Larsen syndrome	309803	Rhizomelic chondrodyplasia punctata type 3		to left-sided atrium
99852	Reunion island-anorexia-vomiting which is irrepressible-neurological signs syndrome	468717	Rhizomelic chondrodyplasia punctata type 5	99110	Right SVC connecting to left-sided atrium
284388	Reversible cerebral vasoconstriction syndrome	2831	Rhizomelic dysplasia, Patterson-Lowry type	97244	Rigid spine congenital muscular dystrophy
254864	Reversible infantile cytochrome C oxidase deficiency	93569	Rhizomelic pseudopolyarthrosis	97244	Rigid spine syndrome
254864	Reversible infantile respiratory chain deficiency	1453	Rhizomelic shortness with clavicular defect	1764	Riley-Day syndrome
3088	Revesz syndrome	3098	Rhizomelic syndrome, Urbach type	217335	RIN2 deficiency
3088	Revesz-DeBuse syndrome	59315	Rhombencephalosynapsis	217335	RIN2 syndrome
3096	Reye syndrome	→1071	RHS	96173	Ring 9
199267	Reye tumor	140976	RHYNS syndrome	1437	Ring 1
779	Reynolds syndrome	217055	RI-CMT type A	96171	Ring 2
244310	RFT1-CDG	254334	RI-CMT type B	96172	Ring 3
251975	RGNT	369867	RI-CMT type C	1447	Ring 4
71275	Rh deficiency syndrome	435998	RI-CMT type D	251043	Ring 5
71275	Rh-null syndrome	97229	Riboflavin transporter deficiency	1448	Ring 6
69077	Rhabdoid tumor	440706	Ribose-5-P isomerase deficiency	1449	Ring 7
231108	Rhabdoid tumor predisposition syndrome	141184	RICH	1450	Ring 8
3097	Rhabdomyomatous dysplasia-cardiopathy-genital anomalies syndrome	1399	Richards-Rundle syndrome	1438	Ring 10
780	Rhabdomyosarcoma	240071	Richardson syndrome	96175	Ring 11
213802	Rhabdomyosarcoma of the cervix uteri	2323	Richardson-Kirk syndrome	1439	Ring 12
213615	Rhabdomyosarcoma of the corpus uteri	3101	Richieri Costa-da Silva syndrome	96176	Ring 13
3099	Rheumatic fever	→2995	Richieri Costa-Guion Almeida syndrome	1440	Ring 14
85408	Rheumatoid factor-negative JIA	2511	Richieri Costa-Guion Almeida-Ramos syndrome	96177	Ring 15
247854	Rheumatoid factor-negative JIA with anti-nuclear antibodies	→2353	Richieri Costa-Guion Almeida-Rodini syndrome	96178	Ring 16
247861	Rheumatoid factor-negative JIA without anti-nuclear antibodies	3102	Richieri Costa-Pereira syndrome	1441	Ring 17
85408	Rheumatoid factor-negative juvenile idiopathic arthritis	1784	Richieri-Costa-Colletto syndrome	1442	Ring 18
247854	Rheumatoid factor-negative juvenile idiopathic arthritis with anti-nuclear antibodies	1794	Richieri-Costa-Gorlin syndrome	1443	Ring 19
247861	Rheumatoid factor-negative juvenile idiopathic arthritis without anti-nuclear antibodies	28378	Richner-Hanhart syndrome	1444	Ring 20
85435	Rheumatoid factor-positive polyarticular JIA	606	Ricker disease	1445	Ring 21
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	606	Ricker syndrome	1446	Ring 22
761	Rheumatoid purpura	83312	Rickettsialpox	1437	Ring chromosome 1
177	Rhizomelic chondrodyplasia punctata	420741	RIDDLE syndrome	1437	Ring chromosome 1 syndrome
309789	Rhizomelic chondrodyplasia punctata type 1	64744	Riedel disease	96171	Ring chromosome 2
309796	Rhizomelic chondrodyplasia punctata type 2	64744	Riedel thyroiditis	96171	Ring chromosome 2 syndrome
		91483	Rieger anomaly	96172	Ring chromosome 3
		3163	Rieger anomaly-partial lipodystrophy syndrome	96172	Ring chromosome 3 syndrome
		782	Rieger syndrome	1447	Ring chromosome 4
		319251	Rift valley fever	1447	Ring chromosome 4 syndrome
		99081	Right aortic arch	251043	Ring chromosome 5
		99119	Right inferior caval vein connecting to left-sided atrium	251043	Ring chromosome 5 syndrome
		99119	Right inferior vena cava connecting to left-sided atrium	1448	Ring chromosome 6
		99119	Right IVC connecting to left-sided atrium	1448	Ring chromosome 6 syndrome
		99110	Right superior caval vein connecting to left-sided atrium	1449	Ring chromosome 7
		99110	Right superior vena cava connecting to left-sided atrium	1449	Ring chromosome 7 syndrome
		99110	Right superior vena cava connecting to left-sided atrium	1450	Ring chromosome 8
				96173	Ring chromosome 9
				96173	Ring chromosome 9 syndrome
				1438	Ring chromosome 10
				1438	Ring chromosome 10 syndrome
				96175	Ring chromosome 11
				96175	Ring chromosome 11 syndrome
				1439	Ring chromosome 12

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
1439	Ring chromosome 12 syndrome	293987	ROHHADNET	1672	Russell diencephalic cachexia
96176	Ring chromosome 13	353298	Roifman syndrome	1672	Russell syndrome
96176	Ring chromosome 13 syndrome	221139	Roifman-Chitayat syndrome	1834	Russell-Weaver-Bull syndrome
1440	Ring chromosome 14	→1855	Roifman-Melamed syndrome	2709	Rutherford syndrome
1440	Ring chromosome 14 syndrome	247775	Rokitansky sequence	3121	Ruvalcaba syndrome
96177	Ring chromosome 15	3109	Rokitansky syndrome	247691	RVCL
96177	Ring chromosome 15 syndrome	1945	Rolandic epilepsy	247691	RVCL-S
96178	Ring chromosome 16	163727	Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	293848	rvFTD
96178	Ring chromosome 16 syndrome			461	RXLI
1441	Ring chromosome 17	163721	Rolandic epilepsy-speech dyspraxia syndrome	16	S cone monochromacy
1441	Ring chromosome 17 syndrome	101016	Romano-Ward long QT syndrome	16	S cone monochromatism
1442	Ring chromosome 18	101016	Romano-Ward syndrome	3105	Saal-Greenstein syndrome
1442	Ring chromosome 18 syndrome	1214	Romberg syndrome	319239	Sabia hemorrhagic fever
1443	Ring chromosome 19	3110	Rombo syndrome	3124	Saccharopine dehydrogenase deficiency
1443	Ring chromosome 19 syndrome	90050	ROP	3124	Saccharopururia
1444	Ring chromosome 20	158014	Rosaï-Dorfman disease	286	Sack-Barabas syndrome
1444	Ring chromosome 20 syndrome	158014	Rosaï-Dorfman-Destombes disease	98841	sACL
1445	Ring chromosome 21	1837	Rosenberg-Lohr syndrome	3027	Sacral agenesis syndrome
1445	Ring chromosome 21 syndrome	329	Rosenthal factor deficiency	397927	Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome
1446	Ring chromosome 22	329	Rosenthal syndrome		→83628
1446	Ring chromosome 22 syndrome	251975	Rosette-forming glioneuronal tumor	2351	Sacral meningocele-conotruncal heart defects syndrome
261529	Ring chromosome Y	2909	Rothmund-Thomson syndrome	3027	Sacral regression syndrome
261529	Ring chromosome Y syndrome	221008	Rothmund-Thomson syndrome type 1	→3027	Sacrococcygeal dysgenesis association
91481	Ring dermoid of cornea	221016	Rothmund-Thomson syndrome type 2	494421	Sacrococcygeal teratoma
91481	Ring dermoid syndrome	3111	Rotor syndrome	85165	SADDAN
169	Ringed hair disease	171709	Round-headed sperm syndrome	794	Saethre-Chotzen syndrome
499103	RINR	3115	Roussy-Lévy syndrome	2872	Sagittal craniostenosis with congenital heart disease, mental deficiency and mandibular ankylosis
97238	Rippling muscle disease	1323	Rozin-camptodactyly syndrome	300493	Sagliker syndrome
206575	Rippling muscle disease with myasthenia gravis	1323	Rozin-Hertz-Goodman syndrome	83484	Saint Louis encephalitis
7	Ritscher-Schinzel syndrome	280569	RPGN	2256	Saito-Kuba-Tsuruta syndrome
1803	Rivera-Perez-Salas syndrome	1507	RRS	1409	Salamon syndrome
294049	RLS	818	RSH syndrome	2613	Salcedo syndrome
93307	rMED	293848	RTLA	140969	Saldino-Mainzer syndrome
420741	RNF168 deficiency	231108	RTPS	404499	Salih ataxia
71273	RNS	2909	RTS	289377	Salih myopathy
3103	Roberts syndrome	221008	RTS1	213557	Salivary gland type cancer of the breast
3103	Roberts-SC phocomelia syndrome	221016	RTS2	213557	Salivary gland type carcinoma of the breast
3104	Robin sequence-oligodactyly syndrome	83616	Rubella panencephalitis	309334	Salla disease
97360	Robinow dwarfism	783	Rubinstein-Taybi syndrome	370938	Salt-and-pepper syndrome
97360	Robinow syndrome	353281	Rubinstein-Taybi syndrome due to 16p13.3 microdeletion	112	Salt-losing tubular disorder, Henle's loop type
3105	Robinow-like syndrome	353277	Rubinstein-Taybi syndrome due to CREBBP mutations	112	Salt-wasting tubulopathy, Henle's loop type
97360	Robinow-Silverman-Smith syndrome	353284	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency	2230	Saliti-Salem syndrome
→794	Robinow-Sorauf syndrome	1768	Rudd-Klimek syndrome		
2780	Robinow-Unger syndrome	→798	Rudiger syndrome		
529	Roch-Leri mesosomatous lipomatosis	79433	Rufous oculocutaneous albinism		
83311	Rocky Mountain spotted fever	435953	Ruijs-Aalfs syndrome		
49382	Rod monochromacy				
49382	Rod monochromatism				
49827	Rogers syndrome				
293987	ROHHAD				

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369992	SAM syndrome	98766	SCA5	1170	SCAR2
53721	SAMS 1-31	98758	SCA6	95433	SCAR3
397623	SAMS syndrome	94147	SCA7	95434	SCAR4
228123	San Joaquin valley fever	98760	SCA8	83472	SCAR5
96167	San Luis Valley syndrome	98761	SCA10	284332	SCAR6
796	Sandhoff disease	98767	SCA11	284324	SCAR7
309169	Sandhoff disease, adult form	98762	SCA12	88644	SCAR8
309155	Sandhoff disease, infantile form	98768	SCA13	139485	SCAR9
309162	Sandhoff disease, juvenile form	98763	SCA14	284289	SCAR10
71272	Sandifer syndrome	98769	SCA15/16	284271	SCAR11
70595	SANDO	→98769	SCA16	284282	SCAR12
2378	Sandrow syndrome	98759	SCA17	324262	SCAR13
581	Sanfilippo disease	98771	SCA18	352403	SCAR14
79269	Sanfilippo syndrome type A	98772	SCA19/22	404499	SCAR15
79270	Sanfilippo syndrome type B	101110	SCA20	412057	SCAR16
79271	Sanfilippo syndrome type C	98773	SCA21	453521	SCAR17
79272	Sanfilippo syndrome type D	→98772	SCA22	363432	SCAR18
2323	Sanjad-Sakati syndrome	101108	SCA23	448251	SCAR19
588	Santavuori congenital muscular dystrophy	101111	SCA25	397709	SCAR20
79263	Santavuori disease	101112	SCA26	466794	SCAR21
79263	Santavuori-Haltia disease	98764	SCA27	404493	SCAR23
2155	Santos-Mateus-Leal syndrome	101109	SCA28	3134	SCARF syndrome
98868	SAO	208513	SCA29	90080	Scarring in glaucoma filtration surgical procedures
247234	SAOA	211017	SCA30	95434	SCASI
793	SAPHO syndrome	217012	SCA31	85297	SCAX3
54368	Sarcocystosis	276183	SCA32	85292	SCAX4
797	Sarcoidosis	1955	SCA34	314978	SCAX5
3129	Sarcosine dehydrogenase complex deficiency	276193	SCA35	284400	SCCB
3129	Sarcosinemia	276198	SCA36	98967	SCCD
54368	Sarcosporidiosis	363710	SCA37	370396	SCCO
1878	Sarcotubular myopathy	423296	SCA38	98967	SCD
3130	Satoyoshi syndrome	423275	SCA40	420402	SCD syndrome
330015	Saturnism	458798	SCA41	449280	Scedosporiosis
425120	SAVI	458803	SCA42	399329	SCFE
3047	Say-Barber-Biesecker-Young-Simpson syndrome	497764	SCA43	398069	Schaaf-Yang syndrome
2013	Say-Barber-Hobbs syndrome	95433	SCABD	1383	Schaap-Taylor-Baraitser syndrome
3132	Say-Barber-Miller syndrome	26792	SCAD deficiency	71212	SCHAD deficiency
3133	Say-Field-Coldwell syndrome	26792	SCADD	370039	Schauder syndrome
3369	Say-Meyer syndrome	254881	SCAE	93474	Scheie syndrome
3047	SBBYSS	1003	Scalp defects-postaxial polydactyl syndrome	2353	Schilbach-Rott syndrome
79157	SBCAD deficiency	370052	SCALP syndrome	59298	Schilder disease
481	SBMA	2036	Scalp-ear-nipple syndrome	59298	Schilder's disease
3103	SC phocomelia	64753	SCAN 2	1830	Schimke immuno-osseous dysplasia
3103	SC pseudothalidomide syndrome	94124	SCAN1	1830	Schimke syndrome
98755	SCA1	840	SCAP	2612	Schimmelpenning syndrome
98756	SCA2	168624	Scaphocephaly-macrocephaly-maxillary retrusion-intellectual disability syndrome	3137	Schindler disease
98757	SCA3	2839	Scapuloiliac dysostosis	79279	Schindler disease type 1
276238	SCA3, Joseph type	431255	Scapuloperoneal neuropathy	79280	Schindler disease type 2
276244	SCA3, Machado type	431255	Scapuloperoneal spinal muscular atrophy	79281	Schindler disease type 3
276241	SCA3, Thomas type	64753	SCAR1	3138	Schinzel syndrome
98765	SCA4			798	Schinzel-Giedion syndrome

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799	Schizencephaly	238593	Sclerosing mesenteritis	808	Seckel syndrome
98973	Schlüchting dystrophy	100001	Sclerosing perineurioma	141022	Second branchial cleft anomaly
3143	Schmidt syndrome	3152	Sclerosteosis	141022	Second branchial cleft cyst
2252	Schmitt-Gillenwater-Kelly syndrome	384	Scleroytosis	141022	Second branchial cleft fistula
3144	Schneckenbecken dysplasia	188	SCLS	139420	Secondary acute transverse myelitis
37748	Schnitzler syndrome	1671	SCM type 1	85445	Secondary amyloidosis
98967	Schnyder corneal dystrophy	331176	SCN4	169618	Secondary central precocious puberty
98967	Schnyder crystalline corneal dystrophy	466677	Scorpion envenomation	314962	Secondary HES
98967	Schnyder crystalline dystrophy sine crystals	832	SCOT deficiency	314962	Secondary hypereosinophilic syndrome
3145	Schofer-Beetz-Bohl syndrome	1514	Scott craniodigital syndrome	2615	Secondary hypertrophic osteoperiostosis with pernio
3041	Scholte-Begeer-van Essen syndrome	806	Scott syndrome	140286	Secondary hypoparathyroidism due to impaired parathormon secretion
50944	Schöpf-Schulz-PassARGE syndrome	1514	Scott-Bryant-Graham syndrome	90363	Secondary intestinal lymphangiectasia
93921	Schwannomatosis	1509	Scott-Taor syndrome	399180	Secondary non-traumatic avascular necrosis
800	Schwartz-Jampel syndrome	86813	SCRA	399180	Secondary non-traumatic AVN
800	Schwartz-Jampel syndrome type 1	83317	Scrub typhus	3452	Secondary non-tropical sprue
3206	Schwartz-Jampel syndrome type 2	794	SCS	439746	Secondary PAN
800	Schwartz-Jampel-Aberfeld syndrome	295193	SD1, Castilla type	420259	Secondary PAP
277	SCID due to adenosine deaminase deficiency	295189	SD1, Lueken type	439746	Secondary periarteritis nodosa
275	SCID due to ARTEMIS deficiency	295191	SD1, Montagu type	439746	Secondary polyarteritis nodosa
357237	SCID due to CARD11 deficiency	295187	SD1, Weidenreich type	420259	Secondary pulmonary alveolar proteinosis
331206	SCID due to complete RAG1/2 deficiency	295187	SD1a	99930	Secondary pulmonary hemosiderosis
228003	SCID due to CORO1A deficiency	295189	SD1b	447774	Secondary sclerosing cholangitis
228003	SCID due to coronin-1A deficiency	295191	SD1c	95427	Secondary short bowel syndrome
420573	SCID due to CTPS1 deficiency	295193	SD1d	99857	Secondary syringomyelia
275	SCID due to DCLRE1C deficiency	295197	SD2, Debeer type	364055	SECORD
317425	SCID due to DNA-PKcs deficiency	295199	SD2, Malik type	459051	SED, Stanescu type
397787	SCID due to IKK2 deficiency	295195	SD2, Vordingborg type	163654	SED-BDS
504523	SCID due to LAT deficiency	295195	SD2a	94068	SEDC
280142	SCID due to LCK deficiency	295197	SD2b	567	Sedlackova syndrome
280142	SCID due to lymphocyte-specific protein tyrosine kinase deficiency	295199	SD2c	647	Seemanova syndrome type 2
33355	SCID with leukopenia	93404	SD3	2528	Seemanova-Lesny syndrome
275	SCID, Athabaskan type	93406	SD5	251618	SEGA
275	SCID, Athabaskan type	84064	SD/THE	2759	Seghers syndrome
276	SCIDX1	263463	SDCD, CHST3 type	67039	Segmental odontomaxillary dysplasia
185	Scimitar syndrome	168577	sdCHC	137608	Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome
70573	SCLC	300869	SDRPL	314662	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia
352763	Scleredema	811	SDS	455	SEI
75840	Scleroatonic muscular dystrophy	370052	Sebaceous nevus-central nervous system malformations-aplasia cutis congenital-limbal dermoid-pigmented nevus syndrome	35069	Seitelberger disease
384	Scleroatrophic syndrome	370052	Sebaceous nevus-CNS malformations-aplasia cutis congenital-limbal dermoid-pigmented nevus syndrome	79156	Seizures-intellectual disability due to hydroxylysinuria syndrome
167635	Scleromyxedema	→182050	Sebastian syndrome	466926	Seizures-scoliosis-macrocephaly syndrome
90400	Scleromyxedema without monoclonal gammopathy	841	Sebocystomatosis		
75325	Sclerosing dysplasia of bone-ichthyosis-premature ovarian failure syndrome	168606	Seborrhea-like dermatitis with psoriasiform elements		
63999	Sclerosing mediastinitis	79480	Seborrheic pemphigus		
		98873	SEC23B-CDG		

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199343	Seizures-sensorineural deafness-ataxia-intellectual disability-electrolyte imbalance syndrome	84081	Senior-Boichis syndrome	157798	Serrated polyposis
35858	Selective cobalamin malabsorption with proteinuria	3156	Senior-Loken syndrome	75508	Servelle-Martorell syndrome
183675	Selective IgG subclass deficiency	1515	Sensenbrenner syndrome	199343	SeSAME syndrome
331235	Selective IgM deficiency	217622	Sensorineural deafness with dilated cardiomyopathy	1807	Setleis syndrome
331235	Selective immunoglobulin M deficiency	857	Sensorineural deafness with imperforate anus and hypoplastic thumbs	85165	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome
165994	Selective pituitary resistance to thyroid hormone	217622	Sensorineural hearing loss with dilated cardiomyopathy	438207	Severe autosomal recessive macrothrombocytopenia
99798	Selective tooth agenesis	2663	Sensorineural hearing loss-cataract-skeletal anomalies-cardiomyopathy syndrome	254930	Severe C12ORF65-related combined oxidative phosphorylation defect
281122	Self-healing collodion baby	66633	Sensorineural hearing loss-early graying-essential tremor syndrome	254930	Severe C12ORF65-related COXPD
90397	Self-healing papular mucinosis	97229	Sensorineural hearing loss-pontobulbar palsy syndrome	314911	Severe Canavan disease
65748	Self-healing squamous epithelioma type 1	70595	Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	277	Severe combined immunodeficiency due to adenosine deaminase deficiency
281122	Self-improving collodion baby	477	Senter syndrome	275	Severe combined immunodeficiency due to ARTEMIS deficiency
281122	Self-improving congenital ichthyosis	90118	SEOAN due to MFN2 deficiency	357237	Severe combined immunodeficiency due to CARD11 deficiency
→1768	Selig-Benacerraf-Greene syndrome	228374	SEOAN due to NEFL deficiency	331206	Severe combined immunodeficiency due to complete RAG1/2 deficiency
3232	Sellars-Beighton syndrome	70594	Sepiapterin reductase deficiency	228003	Severe combined immunodeficiency due to CORO1A deficiency
100069	Semantic dementia	90051	Sepsis in premature infants	228003	Severe combined immunodeficiency due to coronin-1A deficiency
100069	Semantic primary progressive aphasia	180154	Septate vagina	420573	Severe combined immunodeficiency due to CTPS1 deficiency
100069	Semantic variant PPA	137839	Septic phlebitis of the internal jugular vein	275	Severe combined immunodeficiency due to DCLRE1C deficiency
93356	SEMD type 2	3157	Septo-optic dysplasia	317425	Severe combined immunodeficiency due to DNA-PKcs deficiency
171866	SEMD, aggrecan type	3157	Septo-optic dysplasia spectrum	397787	Severe combined immunodeficiency due to IKK2 deficiency
168454	SEMD, Geneviève type	280195	Septopreoptic holoprosencephaly	504523	Severe combined immunodeficiency due to LAT deficiency
93351	SEMD, Irapa type	280195	Septopreoptic HPE	280142	Severe combined immunodeficiency due to LCK deficiency
156728	SEMD, MATN3-related	139466	SERKAL syndrome	280142	Severe combined immunodeficiency due to lymphocyte-specific protein tyrosine kinase deficiency
156728	SEMD, matrilin-3 type	43116	Serotonergic syndrome	33355	Severe combined immunodeficiency with leukopenia
93356	SEMD, Missouri type	43116	Serotonin storm	275	Severe combined immunodeficiency
93352	SEMD, Shohat type	43116	Serotonin syndrome		
93359	SEMD-JL	43116	Serotonin toxicity		
93360	SEMD-MD	43116	Serotonin toxidrome		
168454	SEMDG	506090	Serotonin-producing neuroendocrine tumor of pancreas		
93359	SEMDJL1	506090	Serotonin-producing pancreatic NET		
93360	SEMDJL2	506090	Serotonin-producing pancreatic neuroendocrine tumor		
420402	Semicircular canal dehiscence syndrome	506090	Serotonin-producing PNET		
90160	Semicircular lipoptrophy	424073	Serous cystadenocarcinoma of the pancreas		
220386	Semilobar holoprosencephaly	206470	Serous cystadenoma of ovary in childhood		
842	Seminoma of testis	206470	Serous or mucinous cystadenoma of childhood		
842	Seminomatous germ cell tumor of testis	168829	Serous surface papillary carcinoma		
329284	SENDA	→955	Serpentine fibula-polycystic kidneys syndrome		
79480	Senear-Usher syndrome	35686	Serpiginous choroiditis		
397596	Senescent T-cells-lymphadenopathy-immunodeficiency syndrome due to p110delta-activating mutation				
1369	Sengers syndrome				
2183	Sengers-Hamel-Otten syndrome				
330001	Senile systemic amyloidosis				
1292	Senior syndrome				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	immunodeficiency, Athabascan type	466026	Severe hemolytic anemia due to G6PD deficiency	2879	Severe limb deficit
275	Severe combined immunodeficiency, Athabaskan type	169802	Severe hemophilia A	1236	Severe microbrachycephaly-intellectual disability-athetoid cerebral palsy syndrome
209370	Severe congenital encephalopathy due to MECP2 mutation	169793	Severe hemophilia B		Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome
300298	Severe congenital hypochromic anemia with ringed sideroblasts	745	Severe hereditary thrombophilia due to congenital protein C deficiency		Severe myoclonic epilepsy of infancy
300298	Severe congenital hypochromic sideroblastic anemia	743	Severe hereditary thrombophilia due to congenital protein S deficiency		Severe myoclonus epilepsy of infancy
171430	Severe congenital nemaline myopathy	467176	Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome		Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion
99749	Severe congenital neutropenia type 3	98920	Severe infantile axonal neuropathy with respiratory failure type 1		Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency
331176	Severe congenital neutropenia type 4	404521	Severe infantile axonal neuropathy with respiratory failure type 2	209370	Severe neonatal-onset encephalopathy with microcephaly
331176	Severe congenital neutropenia-pulmonary hypertension-superficial venous angiectasia syndrome	280763	Severe intellectual disability and progressive spastic paraparesis	363400	Severe neurodegenerative syndrome due to BSCL2 deficiency
	Severe dermatitis-multiple allergies-metabolic wasting syndrome	420561	Severe intellectual disability-aplasia/hypoplasia of thumb and hallux syndrome	363400	Severe neurodegenerative syndrome with lipodystrophy
→300751	Severe dilated cardiomyopathy due to lamin A/C mutation	466688	Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome	500545	Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract
→300751	Severe dilated cardiomyopathy with or without myopathy	94066	Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia	216812	Severe osteogenesis imperfecta
98896	Severe dystrophinopathy, Duchenne type	438178	Severe intellectual disability-epilepsy-cataract syndrome due to FAR1 deficiency	411543	Severe phosphoribosylpyrophosphate synthetase superactivity
364055	Severe early-childhood-onset retinal dystrophy	438178	Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency	280210	Severe PMD
228374	Severe early-onset axonal neuropathy due to light neurofilament subunit deficiency	438178	Severe intellectual disability-epilepsy-cataract syndrome due to peroxisomal disorder	468726	Severe primary trimethylaminuria
90118	Severe early-onset axonal neuropathy due to MFN2 deficiency	436141	Severe intellectual disability-hypotonia-strabismus-coarse face-planovalgus syndrome	411543	Severe PRPP synthetase superactivity
228374	Severe early-onset axonal neuropathy due to NEFL deficiency	363686	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	411543	Severe PRPS1 superactivity
329249	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency	397933	Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	163703	Severe refractory status epilepticus owing to presumed encephalitis
440427	Severe early-onset pulmonary alveolar proteinosis due to MARS deficiency	404473	Severe intellectual disability-progressive spastic diplegia syndrome	169095	Severe T-cell immunodeficiency-congenital alopecia-nail dystrophy syndrome
169793	Severe factor IX deficiency	391307	Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome	3078	Severe X-linked intellectual disability, Gustavson type
169802	Severe factor VIII deficiency	324307	Severe lateral tibial bowing with short stature	238329	Severe X-linked mitochondrial encephalomyopathy
352577	Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome			363489	Sex cord-stromal tumor of testis
79408	Severe generalized RDEB			139466	Sex reversion-kidneys, adrenal and lung dysgenesis syndrome
79408	Severe generalized recessive dystrophic epidermolysis bullosa			3162	Sézary lymphoma
488627	Severe growth deficiency-strabismus-extensive dermal melanocytosis-intellectual disability syndrome			3162	Sézary syndrome
2109	Severe Hallermann-Streiff-François syndrome			488232	SFMMP
				373	SGBS
				373	SGBS1
				79022	SGBS2
				457083	SGF
				2063	SGFLD syndrome
				35710	SGLT1 deficiency

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
69076	SGLT2 deficiency		hormone secretagogue receptor deficiency		syndrome-cleft mandible-hand anomalies clubfoot syndrome
2462	SGS	632	Short stature due to isolated growth hormone deficiency with X-linked hypogammaglobulinemia	85442	Short stature-pituitary and cerebellar defects-small sella turcica syndrome
798	SGS	314802	Short stature due to partial GHR deficiency	2868	Short stature-valvular heart disease-characteristic facies syndrome
2407	Shabbir syndrome	314802	Short stature due to partial growth hormone receptor deficiency	2865	Short stature-webbed neck-heart disease syndrome
897	Shah-Waardenburg syndrome	140941	Short stature due to primary acid-labile subunit deficiency	2863	Short stature-wormian bones-dextrocardia syndrome
363523	Shaheen syndrome	220465	Short stature due to STAT5b deficiency	3163	SHORT syndrome
29822	Shapiro syndrome	2867	Short stature, Brussels type	2832	Short tarsus-absence of lower eyelashes syndrome
1506	Sharma-Kapoors-Ramji syndrome	435804	Short stature-advanced bone age-early-onset osteoarthritis syndrome	251515	Short tendo calcaneus
809	Sharp syndrome	397623	Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome	357175	Short ulna-dysmorphism-hypotonia-intellectual disability syndrome
281122	SHCB	464288	Short stature-brachydactyly-obesity-global developmental delay syndrome	57145	Short-lasting unilateral neuralgiform headache attacks with conjunctival injection and tearing
91355	Sheehan syndrome	2994	Short stature-craniofacial anomalies-genital hypoplasia syndrome	935	Short-limb skeletal dysplasia with severe combined immunodeficiency
1147	Sheldon-Hall syndrome	2866	Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome	79157	Short/branched-chain acyl-coA dehydrogenase deficiency
3329	SHFLD syndrome	171706	Short stature-delayed bone age due to thyroid hormone metabolism deficiency	2580	Shoulder and girdle defects-familial intellectual disability syndrome
2440	SHFM	488618	Short stature-developmental delay-congenital heart defect syndrome	→392	Shoulder and thorax deformity-congenital heart disease syndrome
3329	SHFM associated with aplasia of long bones	2332	Short stature-facial and skeletal anomalies-intellectual disability-macrodontia syndrome	314795	SHOX-related short stature
90038	Shiga-like toxin-associated HUS	→2995	Short stature-intellectual disability-eye anomalies-cleft lip/palate syndrome	567	Shprintzen syndrome
810	Shigellosis	420794	Short stature-kyphosis-hypoplasia of basal ilia-cone epiphyses-facial dysmorphism syndrome	2462	Shprintzen-Goldberg syndrome
158014	SHML	1937	Short stature-locking fingers syndrome	3165	Shulman syndrome
1008	Shokeir syndrome	423454	Short stature-nail dysplasia-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome	811	Shwachman syndrome
99063	Shone complex	314394	Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome	811	Shwachman-Bodian-Diamond syndrome
251515	Short Achilles tendon	391677	Short stature-optic atrophy-Pelger-Huët anomaly syndrome	811	Shwachman-Diamond syndrome
26792	Short chain acyl-CoA dehydrogenase deficiency	3102	Short stature-Pierre Robin sequence-cleft mandible-hand anomalies clubfoot syndrome	812	Sialidosis type 1
66518	Short fifth metacarpals-insulin resistance syndrome	3102	Short stature-Pierre Robin	87876	Sialidosis type 2
935	Short limb skeletal dysplasia with SCID			3166	Sialuria
93270	Short rib-polydactyly syndrome type 1			3166	Sialuria, French type
93269	Short rib-polydactyly syndrome type 2			98920	SIANRF
93271	Short rib-polydactyly syndrome type 3			→33364	SIBIDS syndrome
93268	Short rib-polydactyly syndrome type 4			611	sIBM
498497	Short rib-polydactyly syndrome type 5			281122	SICI
93268	Short rib-polydactyly syndrome, Beemer-Langer type			232	Sickle cell anemia
93269	Short rib-polydactyly syndrome, Majewski type			232	Sickle cell disease
93270	Short rib-polydactyly syndrome, Saldino-Noonan type			251359	Sickle cell-beta-thalassemia disease syndrome
93271	Short rib-polydactyly syndrome, Verma-Naumoff type			251365	Sickle cell-hemoglobin C disease syndrome
→1263	Short ribs-craniosynostosis-polysyndactyly syndrome			251370	Sickle cell-hemoglobin D disease syndrome
314811	Short stature due to GHSR deficiency			251375	Sickle cell-hemoglobin E disease
629	Short stature due to growth hormone qualitative anomaly				
633	Short stature due to growth hormone resistance				
314811	Short stature due to growth				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	syndrome	439755	Single-organ periarteritis nodosa	399329	Slipped upper femoral epiphysis
210272	Sickness of disembarkment	439755	Single-organ polyarteritis nodosa	88633	SLK
838	SICRET syndrome	85191	Singleton-Merten dysplasia	818	SLOS
168593	SIDDT	85191	Singleton-Merten syndrome	70472	SLSJ-COX deficiency
54028	Sideropenic dysphagia	1260	Sino-auricular heart block	3156	SLSN
3167	Sieglar-Brewer-Carey syndrome	324321	Sinoatrial node dysfunction and deafness	584	Sly disease
→244	Siewert syndrome	158014	Sinus histiocytosis with massive lymphadenopathy	98849	SM-AHN
369861	SIFD syndrome	890	Sinusoidal obstruction syndrome	98849	SM-AHNMD
314786	Silent pituitary adenoma	247698	Sipple syndrome	70	SMA
71276	Silent sinus syndrome	3169	Sirenomelia	83330	SMA1
3168	Sillence syndrome	2882	Sitosterolemia	83418	SMA2
60014	Silver staining	157769	Situs ambiguus	83419	SMA3
100998	Silver syndrome	157769	Situs ambiguus	83420	SMA4
813	Silver-Russell dwarfism	101063	Situs inversus	83330	SMA type 1
813	Silver-Russell syndrome	101063	Situs inversus totalis	83418	SMA type 2
231137	Silver-Russell syndrome due to 7p11.2-p13 microduplication	488437	SIX2-related FND	83419	SMA type 3
231137	Silver-Russell syndrome due to 7p11.2p13 microduplication	488437	SIX2-related frontonasal dysplasia	83420	SMA type 4
231144	Silver-Russell syndrome due to 11p15 microduplication	816	Sjögren-Larsson syndrome	83330	SMA type I
397590	Silver-Russell syndrome due to a point mutation	800	SJS	83418	SMA type II
231140	Silver-Russell syndrome due to an imprinting defect of 11p15	800	SJS1	83419	SMA type III
231137	Silver-Russell syndrome due to dup(7)(p11.2p13)	3206	SJS2	83420	SMA type IV
96182	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7	95455	SJS-TEN	83330	SMA-I
231147	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 11	506784	SJS/TEN overlap syndrome	83418	SMA-II
231137	Silver-Russell syndrome due to trisomy 7p11.2-p13	1426	Skeletal dysplasia, Greenberg type	83419	SMA-III
231137	Silver-Russell syndrome due to trisomy 7p11.2p13	2565	Skeletal dysplasia-brachydactyly syndrome	83420	SMA-IV
1968	Simosa craniofacial syndrome	1858	Skeletal dysplasia-epilepsy-short stature syndrome	486811	SMABF
1968	Simosa-Penchaszadeh-Bustos syndrome	508533	Skeletal dysplasia-T cell immunodeficiency-developmental delay syndrome	209335	SMAFK
91139	Simple cryoglobulinemia	477831	Skeletal overgrowth-craniofacial dysmorphism-hyperelastic skin-white matter lesions syndrome	363447	SMALED
373	Simpson dysmorphia syndrome	293165	Skin fragility-woolly hair-palmoplantar hyperkeratosis syndrome	209341	SMALED1
373	Simpson-Golabi-Behmel syndrome	293165	Skin fragility-woolly hair-palmoplantar keratoderma syndrome	363454	SMALED2
373	Simpson-Golabi-Behmel syndrome type 1	178475	Skin infectious botulism	284400	Small cell bladder cancer
79022	Simpson-Golabi-Behmel syndrome type 2	178475	Skin toxin-mediated botulism	284400	Small cell bladder carcinoma
500163	SIN3A-related intellectual disability syndrome	52503	SLC6A8 deficiency	284400	Small cell carcinoma of the bladder
500166	SIN3A-related intellectual disability syndrome due to a point mutation	238459	SLC35A1-CDG	370396	Small cell carcinoma of the ovary
97337	Sinding-Larsen-Johansson disease	356961	SLC35A2-CDG	284400	Small cell carcinoma of the urinary bladder
50809	Singh-Williams-McAlister syndrome	370943	SLC35A3-CDG	70573	Small cell lung cancer
2286	Single upper central incisor	99843	SLC35C1-CDG	370396	Small cell ovarian carcinoma
439755	Single-organ PAN	3144	SLC35D1-CDG	838	Small infarctions of cochlear, retinal and encephalic tissue
		468699	SLC39A8 deficiency	1201	Small intestinal atresia
		468699	SLC39A8-CDG	67038	Small lymphocytic lymphoma
		93552	SLE, pediatric onset	543	Small non-cleaved cell lymphoma
		3385	Sleeping sickness	1509	Small patella syndrome
		399329	Slipped capital femoral epiphysis	466962	SMARCA4-deficient sarcoma of thorax
				466962	SMARCA4-deficient thoracic sarcoma
				98920	SMARD1
				404521	SMARD2
				481	SMAX1
				1145	SMAX2
				139557	SMAX3
				98959	SMCD

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85167	SMD-CRD	821	Sotos syndrome	100996	Spastic paraplegia-retinal degeneration syndrome
33069	SMEI	420179	Sotos syndrome 2	464282	Spastic paraplegia-severe developmental delay-epilepsy syndrome
93974	Smith-Fineman-Myers syndrome	98868	Southeast Asian ovalocytosis	3011	Spastic quadriplegia-retinitis pigmentosa-intellectual disability syndrome
457485	Smith-Kingsmore syndrome	352403	SPARCA	447997	Spastic quadriplegia-thin corpus callosum-progressive postnatal microcephaly syndrome
818	Smith-Lemli-Opitz syndrome	352403	SPARCA1	210141	Spastic quadriplegic cerebral palsy
819	Smith-Magenis syndrome	79132	Sparse hair-short stature-skin anomalies syndrome	3011	Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome
178355	Smith-McCort dysplasia	279882	Spasmus nutans	447997	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome
2286	SMMC1	1182	Spastic ataxia with congenital miosis	210141	Spastic quadriplegic cerebral palsy
488168	SMO deficiency	2572	Spastic ataxia-corneal dystrophy syndrome	3011	Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome
158775	Smouldering systemic mastocytosis	2572	Spastic ataxia-ocular anomalies syndrome	447997	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome
3198	SMS	99015	Spastic gait type 2	401866	Spasticity-ataxia-gait anomalies syndrome
86854	SMZL	99015	Spastic paraparesis type 2	251282	SPAX1
449285	Snakebite envenomation	100990	Spastic paraparesis-amyopathy-cataracts-gastroesophageal reflux syndrome	397946	SPAX2
820	Sneddon syndrome	2815	Spastic paraparesis-deafness syndrome	314603	SPAX3
48377	Sneddon-Wilkinson disease	101003	Spastic paraparesis-vitiligo-premature graying-characteristic facies syndrome	254343	SPAX4
91496	Snowflake vitreoretinal degeneration	139480	Spastic paraplegia due to neuropathy target esterase mutation	313772	SPAX5
3063	Snyder-Robinson syndrome	139480	Spastic paraplegia due to NTE mutation	98	SPAX6
3157	SOD	431329	Spastic paraplegia due to partial TFG deficiency	1182	SPAX7
67039	SOD	99015	Spastic paraplegia type 2	158	SPCD
306577	Sodium channelopathy-related small fiber neuropathy	99013	Spastic paraplegia type 7	295195	SPD1
99903	Sudoku	100998	Spastic paraplegia-amyotrophy of hands and feet	295197	SPD2
314394	SOFT syndrome	2816	Spastic paraplegia-epilepsy-intellectual disability syndrome	295199	SPD3
100002	Soft tissue perineurioma	2819	Spastic paraplegia-facial-cutaneous lesions syndrome	295197	SPD, Debeer type
2234	Sohval-Soffer syndrome	2818	Spastic paraplegia-glaucoma-intellectual disability syndrome	295199	SPD, Malik type
137608	SOLAMEN syndrome	2822	Spastic paraplegia-intellectual disability-thin corpus callosum syndrome	295195	SPD, Vordingborg type
97230	Solar urticaria	2820	Spastic paraplegia-nephritis-deafness syndrome	352403	Spectrin-associated autosomal recessive cerebellar ataxia
424065	Solid pseudopapillary carcinoma of the pancreas	2821	Spastic paraplegia-neuropathy-poikiloderma syndrome	352403	Spectrin-associated autosomal recessive cerebellar ataxia type 1
424065	Solid pseudopapillary neoplasm of the pancreas	320406	Spastic paraplegia-optic atrophy-neuropathy syndrome	209908	Speech and language disorder with orofacial dyspraxia
83468	Solitary bone cyst	329475	Spastic paraplegia-Paget disease of bone syndrome	209908	Speech-language disorder type 1
2126	Solitary fibrous tumor	2826	Spastic paraplegia-precocious puberty syndrome	→2909	Spellacy-Gibbs-Watts syndrome
79455	Solitary mastocytoma	464282	Spastic paraplegia-psychomotor retardation-seizures syndrome	2816	SPEMR
2286	Solitary median maxillary central incisor syndrome			1855	SPENCD
100035	Solitary necrotic nodule of the liver			→1855	SPENCDI
86855	Solitary plasmacytoma			99865	Spermatocytic seminoma
209964	Solitary rectal ulcer syndrome			306617	SPG1
2612	Solomon syndrome			99015	SPG2
314769	Somatolactotropinoma			100985	SPG4
314769	Somatotammotropinoma			100986	SPG5A
314769	Somatotropinoma			100988	SPG6
97283	Somatostatinoma			99013	SPG7
2564	Sommer-Hines syndrome			100989	SPG8
1064	Sommer-Rathbun-Battles syndrome			100990	SPG9
1529	Sommer-Young-Wee-Frye syndrome			100991	SPG10
1355	Sonoda syndrome				
391677	SOPH syndrome				
59181	Sorsby pseudoinflammatory fundus dystrophy				
1471	Sorsby syndrome				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2822	SPG11	401840	SPG71	98760	Spinocerebellar ataxia type 8
100993	SPG12	401849	SPG72	98761	Spinocerebellar ataxia type 10
100994	SPG13	444099	SPG73	98767	Spinocerebellar ataxia type 11
100995	SPG14	468661	SPG74	98762	Spinocerebellar ataxia type 12
100996	SPG15	459056	SPG75	98768	Spinocerebellar ataxia type 13
100997	SPG16	488594	SPG76	98763	Spinocerebellar ataxia type 14
100998	SPG17	466722	SPG77	98769	Spinocerebellar ataxia type 15/16
209951	SPG18	268129	Spheroid body myopathy	→98769	Spinocerebellar ataxia type 16
100999	SPG19	3449	Spherophakia-brachymorphia syndrome	98759	Spinocerebellar ataxia type 17
101000	SPG20	306553	Spherulocytosis	98771	Spinocerebellar ataxia type 18
101001	SPG21	79264	Spielmeyer-Vogt disease	98772	Spinocerebellar ataxia type 19/22
101003	SPG23	314432	Spigelian hernia-cryptorchidism syndrome	101110	Spinocerebellar ataxia type 20
101004	SPG24	268369	Spina bifida aperta	98773	Spinocerebellar ataxia type 21
101005	SPG25	3176	Spina bifida-hypospadias syndrome	→98772	Spinocerebellar ataxia type 22
101006	SPG26	53721	Spinal arteriovenous metameric syndrome	101108	Spinocerebellar ataxia type 23
101007	SPG27	1217	Spinal atrophy-opthalmoplegia-pyramidal syndrome	101111	Spinocerebellar ataxia type 25
101008	SPG28	90058	Spinal cord injury	101112	Spinocerebellar ataxia type 26
101009	SPG29	1145	Spinal muscular atrophy with arthrogryposis	98764	Spinocerebellar ataxia type 27
101010	SPG30	98920	Spinal muscular atrophy with respiratory distress type 1	101109	Spinocerebellar ataxia type 28
101011	SPG31	404521	Spinal muscular atrophy with respiratory distress type 2	208513	Spinocerebellar ataxia type 29
171622	SPG32	83420	Spinal muscular atrophy, adult form	211017	Spinocerebellar ataxia type 30
171607	SPG34	73245	Spinal muscular atrophy-Dandy-Walker malformation-cataracts syndrome	217012	Spinocerebellar ataxia type 31
171629	SPG35	2590	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	276183	Spinocerebellar ataxia type 32
320365	SPG36	210584	Spindle cell hemangioendothelioma	1955	Spinocerebellar ataxia type 34
171612	SPG37	210584	Spindle cell hemangioma	276193	Spinocerebellar ataxia type 35
171617	SPG38	1955	Spinocerebellar ataxia and erythrokeratoderma	276198	Spinocerebellar ataxia type 36
139480	SPG39	412057	Spinocerebellar ataxia autosomal recessive type 16	363710	Spinocerebellar ataxia type 37
320355	SPG41	453521	Spinocerebellar ataxia autosomal recessive type 17	423296	Spinocerebellar ataxia type 38
171863	SPG42	404493	Spinocerebellar ataxia autosomal recessive type 23	423275	Spinocerebellar ataxia type 40
320370	SPG43	98755	Spinocerebellar ataxia type 1	458798	Spinocerebellar ataxia type 41
320401	SPG44	98756	Spinocerebellar ataxia type 2	458803	Spinocerebellar ataxia type 42
320396	SPG45	98757	Spinocerebellar ataxia type 3	497764	Spinocerebellar ataxia type 43
320391	SPG46	276238	Spinocerebellar ataxia type 3, Joseph type	363710	Spinocerebellar ataxia with altered vertical eye movements
306511	SPG48	276244	Spinocerebellar ataxia type 3, Machado type	94124	Spinocerebellar ataxia with axonal neuropathy type 1
320385	SPG49	98765	Spinocerebellar ataxia type 4	64753	Spinocerebellar ataxia with axonal neuropathy type 2
319199	SPG53	98766	Spinocerebellar ataxia type 5	254881	Spinocerebellar ataxia with epilepsy
320380	SPG54	98758	Spinocerebellar ataxia type 6	276241	Spinocerebellar ataxia, Thomas type
320375	SPG55	94147	Spinocerebellar ataxia type 7	2074	Spinocerebellar ataxia-amyotrophy-deafness syndrome
320411	SPG56			1185	Spinocerebellar ataxia-dysmorphism syndrome
431329	SPG57			3177	Spinocerebellar degeneration-corneal dystrophy syndrome
397946	SPG58			99903	Spirillary rat-bite fever
401795	SPG59			757	Spitzer-Weinstein syndrome
401800	SPG60			300869	Splenic diffuse red pulp lymphoma
401780	SPG61			300869	Splenic diffuse red pulp small B-cell lymphoma
401785	SPG62			86854	Splenic marginal zone lymphoma
401805	SPG63				
401810	SPG64				
320396	SPG65				
401815	SPG66				
401820	SPG67				
401825	SPG68				
401830	SPG69				
401835	SPG70				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
2063	Splenogonadal fusion-limb defects-micrognathia syndrome		type 2		tarda, Kohn type
47612	Splenomegaly-neutropenia-rheumatoid arthritis syndrome	93360	Spondyloepimetaphyseal dysplasia with joint laxity, Hall type	1159	Spondyloepiphyseal dysplasia tarda-progressive arthropathy syndrome
1671	Split cord malformation type 1	93359	Spondyloepimetaphyseal dysplasia with joint laxity	263463	Spondyloepiphyseal dysplasia with congenital joint dyslocations, CHST3 type
2439	Split foot deformity-mandibulofacial dysostosis syndrome	93359	Spondyloepimetaphyseal dysplasia with joint laxity type 1	→93284	Spondyloepiphyseal dysplasia, Byers type
2440	Split hand foot malformation	93360	Spondyloepimetaphyseal dysplasia with joint laxity type 2	163654	Spondyloepiphyseal dysplasia, Cantu type
2437	Split hand with obstructive uropathy, spina bifida and diaphragmatic defects	93359	Spondyloepimetaphyseal dysplasia with joint laxity, Brighton type	93283	Spondyloepiphyseal dysplasia, Kimberley type
71271	Split hand-split foot-deafness syndrome	93360	Spondyloepimetaphyseal dysplasia with joint laxity, leptodactyl type	163668	Spondyloepiphyseal dysplasia, MacDermot type
2437	Split hand-urinary anomalies-spina bifida syndrome	93360	Spondyloepimetaphyseal dysplasia with multiple dislocations	263482	Spondyloepiphyseal dysplasia, Maroteaux type
3329	Split hand/foot malformation with long bone deficiency	93360	Spondyloepimetaphyseal dysplasia with multiple dislocations, Hall type	163649	Spondyloepiphyseal dysplasia, Nishimura type
958	Split hand/split foot-mandibular hypoplasia syndrome	171866	Spondyloepimetaphyseal dysplasia, aggrecan type	→263463	Spondyloepiphyseal dysplasia, Omani type
2329	Split hand/split foot-nystagmus syndrome	93347	Spondyloepimetaphyseal dysplasia, anauxetic type	163662	Spondyloepiphyseal dysplasia, Reardon type
1756	Split notochord syndrome	168448	Spondyloepimetaphyseal dysplasia, Bieganski type	459051	Spondyloepiphyseal dysplasia, Stanescu type
488232	Split-foot malformation-mesoaxial polydactyly syndrome	168454	Spondyloepimetaphyseal dysplasia, Geneviève type	163654	Spondyloepiphyseal dysplasia-brachydactyly-speech disorder syndrome
488232	Split-foot malformation-mesoaxial polydactyly-nail abnormalities-sensorineural hearing loss syndrome	99642	Spondyloepimetaphyseal dysplasia, Handigodu type	163649	Spondyloepiphyseal dysplasia-craniosynostosis-cleft palate-cataract-intellectual disability syndrome
3329	Split-hand/foot malformation associated with aplasia of long bones	93351	Spondyloepimetaphyseal dysplasia, Irapa type	163668	Spondyloepiphyseal dysplasia-myopia-sensorineural deafness syndrome
320406	SPOAN	370015	Spondyloepimetaphyseal dysplasia, Isidor type	1830	Spondyloepiphyseal dysplasia-nephrotic syndrome
93357	SPONASTRIME dysplasia	156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type	→93284	Spondyloepiphyseal dysplasia-punctate corneal dystrophy syndrome
93357	Spondylar and nasal changes with striations of the metaphyses (SPONASTRIME) dysplasia	93347	Spondyloepimetaphyseal dysplasia, Menger type	353298	Spondyloepiphyseal dysplasia-retinal dystrophy-immunodeficiency syndrome
1190	Spondylo-humero-femoral dysplasia	93356	Spondyloepimetaphyseal dysplasia, Missouri type	→1855	Spondylometaphyseal dysplasia with combined immunodeficiency
228387	Spondylo-megaepiphyseal-metaphyseal dysplasia	93282	Spondyloepimetaphyseal dysplasia, Pakistani type	1855	Spondylometaphyseal dysplasia with enchondromatous changes
85194	Spondylo-ocular syndrome	93282	Spondyloepimetaphyseal dysplasia, PAPSS2 type	93316	Spondylometaphyseal dysplasia with severe genu valgum
3180	Spondylocamptodactyly syndrome	93352	Spondyloepimetaphyseal dysplasia, Shohat type	168555	Spondylometaphyseal dysplasia, A4 type
3275	Spondylocarpotarsal synostosis	93357	Spondyloepimetaphyseal dysplasia, Sponastrime type	93316	Spondylometaphyseal dysplasia, Algerian type
94095	Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome	168451	Spondyloepimetaphyseal dysplasia-abnormal dentition syndrome	93315	Spondylometaphyseal dysplasia, 'corner fracture' type
329252	Spondylocostal dysostosis-hypospadias-intellectual disability syndrome	168443	Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome	370019	Spondylometaphyseal dysplasia,
1855	Spondyloenchondrodysplasia	93358	Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome		
→1855	Spondyloenchondrodysplasia with immune dysregulation	94068	Spondyloepiphyseal dysplasia congenita		
1855	Spondyloenchondromatosis	93284	Spondyloepiphyseal dysplasia tarda		
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type	163665	Spondyloepiphyseal dysplasia		
93356	Spondyloepimetaphyseal dysplasia				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	Czarny-Ratajczak type		necrosis		nasal cavity and paranasal sinuses
168544	Spondylometaphyseal dysplasia, Golden type	225147	Sporadic infantile striatonigral degeneration	500464	Squamous cell carcinoma of the nasal cavity and sinuses
93314	Spondylometaphyseal dysplasia, Kozlowski type	225147	Sporadic infantile striatonigral necrosis	150	Squamous cell carcinoma of the nasopharynx
93316	Spondylometaphyseal dysplasia, Schmidt type	227510	Sporadic olivopontocerebellar atrophy type 1	502363	Squamous cell carcinoma of the oral cavity
93317	Spondylometaphyseal dysplasia, Sedaghatian type	227510	Sporadic OPCA type 1	457252	Squamous cell carcinoma of the oral tongue
93315	Spondylometaphyseal dysplasia, Sutcliffe type	276624	Sporadic pheochromocytoma	500478	Squamous cell carcinoma of the oropharynx
168552	Spondylometaphyseal dysplasia-bowed forearms-facial dysmorphism syndrome	276621	Sporadic pheochromocytoma/secretting paraganglioma	424039	Squamous cell carcinoma of the pancreas
85167	Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	443057	Sporadic porphyria cutanea tarda	398058	Squamous cell carcinoma of the penis
1856	Spondyloperipheral dysplasia-short ulna syndrome	276627	Sporadic secreting paraganglioma	424002	Squamous cell carcinoma of the rectum
141	Spongy degeneration of the brain	826	Sporotrichosis	423968	Squamous cell carcinoma of the small bowel
54260	Spongy myocardium	464282	SPPRS syndrome	423968	Squamous cell carcinoma of the small intestine
443180	Spontaneous cerebrospinal fluid leak	70594	SPR deficiency	418959	Squamous cell carcinoma of the stomach
443180	Spontaneous intracranial hypotension	94068	Spranger-Wiedemann disease	494448	Squamous cell carcinoma of the vulva
29822	Spontaneous periodic hypothermia	3181	Sprengel deformity	324737	SRD5A3-CDG
247234	Sporadic adult-onset ataxia of unknown etiology	70476	Spring catarrh	83601	SREAT
204	Sporadic CJD	234	Sprinz-Nelson syndrome	330001	SSA
204	Sporadic Creutzfeldt-Jakob disease	3198	SPS	22	SSADH deficiency
1665	Sporadic fetal brain disruption sequence	1509	SPS	466926	SSM syndrome
306776	Sporadic hyperekplexia	431255	SPSMA	2806	SSPE
225147	Sporadic IBSN	86884	SPTCL	50944	SSPS
84271	Sporadic idiopathic nephrosis	51083	SQTS	370927	SSR4-CDG
84271	Sporadic idiopathic steroid-resistant nephrotic syndrome	424996	Squamous cell carcinoma of gallbladder and extrahepatic biliary tract	2323	SSS
97555	Sporadic idiopathic steroid-resistant nephrotic syndrome with collapsing glomerulopathy	424996	Squamous cell carcinoma of gallblader and EBT	36236	SSSS
93222	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation	67037	Squamous cell carcinoma of head and neck	83484	St. Louis encephalitis
93220	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	424975	Squamous cell carcinoma of liver and IBT	502434	STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome
93218	Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis	424975	Squamous cell carcinoma of liver and intrahepatic biliary tract	2454	Stalker-Chitayat syndrome
93218	Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	500481	Squamous cell carcinoma of salivary glands	1798	Stanescu osteosclerosis
93221	Sporadic idiopathic steroid-resistant nephrotic syndrome with minimal changes	424019	Squamous cell carcinoma of the anal canal	3235	Stapedo-vestibular ankylosis
611	Sporadic inclusion body myositis	213767	Squamous cell carcinoma of the cervix uteri	140917	Stapes ankylosis with broad thumbs and toes
225147	Sporadic infantile bilateral striatal	423994	Squamous cell carcinoma of the colon	36238	Staphylococcal necrotizing pneumonia
		213716	Squamous cell carcinoma of the corpus uteri	36236	Staphylococcal scalded skin syndrome
		99977	Squamous cell carcinoma of the esophagus	36235	Staphylococcal scarlet fever
		494547	Squamous cell carcinoma of the hypopharynx	99919	Staphylococcal toxic-shock syndrome
		494550	Squamous cell carcinoma of the larynx	140952	STAR syndrome
		502366	Squamous cell carcinoma of the lip	827	Stargardt 1
		500464	Squamous cell carcinoma of the	827	Stargardt disease

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
85146	Stark-Kaeser syndrome	166100	Stickler syndrome, non-ocular type	166277	Suarez-Stickler syndrome
166427	Startle epilepsy	3197	Stiff baby syndrome	101029	Sub-cortical nodular heterotopia
391311	STAT1 deficiency	443804	Stiff leg syndrome	79093	Subacute angiohypertrophic myelomalacia
2314	STAT3 deficiency	3198	Stiff man syndrome	79093	Subacute ascending necrotizing myelitis
438159	STAT3-related early-onset multisystem autoimmune disease	3198	Stiff person syndrome and related disorders	163525	Subacute cutaneous lupus erythematosus
329284	Static encephalopathy of childhood with neurodegeneration in adulthood	2833	Stiff skin syndrome	2806	Subacute inclusion body encephalitis
841	Steatocystoma multiplex	85414	Still disease	206594	Subacute inflammatory demyelinating polyneuropathy
3184	Steatocystoma multiplex-natal teeth syndrome	233	Stilling-Turk-Duane syndrome	206594	Subacute inflammatory demyelinating polyradiculoneuropathy
438117	Steel syndrome	3199	Stimmler syndrome	98824	Subacute myeloid leukemia
240071	Steele-Richardson-Olszewski disease	425120	STING-associated vasculopathy with onset in infancy	79093	Subacute necrotizing myelitis
565	Steely hair disease	2972	Stoelinga-de Koomen-Davis syndrome	2806	Subacute sclerosing leukoencephalitis
565	Steely hair syndrome	3200	Stoll-Alembik-Finck syndrome	2806	Subacute sclerosing panencephalitis
273	Steinert disease	3074	Stoll-Géraudel-Chauvin syndrome	356	Subacute spongiform encephalopathy, Gerstmann-Straussler type
273	Steinert myotonic dystrophy	3201	Stoll-Kieny-Dott syndrome	99113	Subaortic course of brachiocephalic vein
3186	Steinfeld syndrome	2878	Stoll-Lévy-Francfort syndrome	99113	Subaortic course of innominate vein
168953	Stem cell leukemia/lymphoma	168577	Stomatin-deficient cryohydrocytosis	3191	Subaortic stenosis-short stature syndrome
99087	Stenosis or atresia of the coronary ostium	98868	Stomatocytic elliptocytosis	48377	Subcorneal pustular dermatitis
210115	Sterile multifocal osteomyelitis with periostitis and pustulosis	353253	Stomatodynia	48377	Subcorneal pustular dermatosis
3194	Stern-Lubinsky-Durrie syndrome	353253	Stomatopyrosis	99796	Subcortical band heterotopia
→42775	Sternal malformation-vascular dysplasia syndrome	337	Stone man syndrome	313808	Subcortical gliosis of Neumann
753	Steroid 5-alpha-reductase deficiency	3204	Stormorken syndrome	99796	Subcortical laminar heterotopia
3196	Steroid dehydrogenase deficiency-dental anomalies syndrome	3204	Stormorken-Sjaastad-Langslet syndrome	86884	Subcutaneous panniculitic T-cell lymphoma
461	Steroid sulfatase deficiency	99064	Straddling and/or overriding mitral valve	86884	Subcutaneous panniculitis-like T-cell lymphoma
83601	Steroid-responsive encephalopathy associated with autoimmune thyroiditis	95461	Straddling or overriding tricuspid valve	306553	Subcutaneous spherulocystic disease
93207	Steroid-sensitive MCNS	1277	Stratton-Garcia-Young syndrome	251618	Subependymal giant cell astrocytoma
→69061	Steroid-sensitive nephrotic syndrome without renal biopsy	2863	Stratton-Parker syndrome	101030	Subependymal nodular heterotopia
909	Sterol 27-hydroxylase deficiency	99905	Streptobacillary rat-bite fever	251639	Subependymoma
46059	Sterol C5-desaturase deficiency	99918	Streptococcal toxic-shock syndrome	98957	Subepithelial amyloidosis of the cornea
488168	Sterol-C4-methyl oxidase deficiency	99918	Streptococcal TSS	98959	Subepithelial mucinous corneal dystrophy
36426	Stevens-Johnson syndrome	66529	Stress cardiomyopathy	155878	Submucosal cleft palate
506784	Stevens-Johnson syndrome/toxic epidermal necrolysis overlap syndrome	90041	Stress erythrocytosis	3190	Subpulmonary stenosis
95455	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	90041	Stress polycythemia	1606	Subtelomeric 1p36 deletion
506784	Stevens-Johnson/TEN overlap syndrome	50942	Striate palmoplantar keratoderma	96168	Subtelomeric deletion 13q34
506784	Stevens-Johnson/toxic epidermal necrolysis overlap syndrome	137599	Stromal keratitis	1398	Subtotal absence of cerebellum
828	Stickler syndrome	213711	Stromal sarcoma of the corpus uteri	180129	Subtotal septate uterus
90653	Stickler syndrome type 1	506307	Stromme syndrome	→2609	Succinic aciduria
90654	Stickler syndrome type 2	76	Strongyloidiasis		
166100	Stickler syndrome type 3	100984	Strümpell disease		
		370921	STT3A-CDG		
		370924	STT3B-CDG		
		328	Stuart-Prower factor deficiency		
		3205	Sturge-Weber syndrome		
		3205	Sturge-Weber-Dimitri syndrome		
		3205	Sturge-Weber-Krabbe angiomas		
		3206	Sturge-Weber-Krabbe syndrome		
		3206	Stüve-Wiedemann dysplasia		
		3206	Stüve-Wiedemann syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
22	Succinic semialdehyde dehydrogenase deficiency	141096	Supernumerary nostril	465508	Symptomatic form of hemochromatosis type 1
832	Succinyl-CoA acetoacetate transferase deficiency	295002	Supernumerary phalanges	177926	Symptomatic form of hemophilia A in female carriers
832	Succinyl-CoA:3-ketoacid CoA transferase deficiency	295142	Supernumerary phalanges, bilateral	177929	Symptomatic form of hemophilia B in female carriers
832	Succinyl-CoA:3-oxoacid CoA transferase deficiency	295140	Supernumerary phalanges, unilateral	465508	Symptomatic form of HFE-related hereditary hemochromatosis
702	Sudanophilic leukodystrophy, Paelizeus-Merzbacher type	295002	Supernumerary phalanx	206546	Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers
168593	Sudden infant death-dysgenesis of the testes syndrome	295142	Supernumerary phalanx, bilateral	357332	Synactyly-camptodactyly and clinodactyly of fifth fingers-bifid halluces syndrome
130	Sudden unexplained nocturnal death syndrome	295140	Supernumerary phalanx, unilateral	98915	Synaptic congenital myasthenic syndromes
399329	SUFE	1461	Superoinferior ventricles	93404	Syndactyly of fingers 4 and 5
498602	Sugarman brachydactyly	764	Suppurative myositis	93402	Syndactyly type 1
2752	Sugarman syndrome	466695	Supratip dysplasia	295193	Syndactyly type 1, Castilla type
498602	Sugarman-Hager-Kulik syndrome	3193	Supravalvular aortic stenosis	295189	Syndactyly type 1, Lueken type
3412	Sujansky-Leonard syndrome	3192	Supravalvular pulmonary stenosis	295191	Syndactyly type 1, Montagu type
99732	Sulfite oxidase deficiency due to molybdenum cofactor deficiency	391351	SURF1-related Charcot-Marie-Tooth disease type 4	295187	Syndactyly type 1, Weidenreich type
308386	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A	391351	SURF1-related CMT4	295187	Syndactyly type 1a
308393	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type B	391351	SURF1-related severe demyelinating Charcot-Marie-Tooth disease	295189	Syndactyly type 1b
308400	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type C	838	Susac syndrome	295191	Syndactyly type 1c
99731	Sulfocysteinuria	331226	Susceptibility to infection due to TYK2 deficiency	295193	Syndactyly type 1d
65682	Summerskill-Walshe-Tygstrup syndrome	447740	Susceptibility to localized juvenile periodontitis	93403	Syndactyly type 2
254395	Summertime actinic lichenoid eruption	169085	Susceptibility to respiratory infections associated with CD8alpha chain mutation	93404	Syndactyly type 3
3210	Summitt syndrome	391311	Susceptibility to viral and mycobacterial infections	93405	Syndactyly type 4
57145	SUNCT syndrome	3193	SVAS	93406	Syndactyly type 5
130	SUNDS	86813	Sveinsson chorioretinal atrophy	295012	Syndactyly type 6
455	Superficial epidermolytic ichthyosis	3243	Sweet syndrome	3258	Syndactyly type 7
98961	Superficial granular corneal dystrophy	247165	Swift disease	2498	Syndactyly type 8
247245	Superficial hemosiderosis of the central nervous system	247165	Swift-Feer disease	157801	Syndactyly type 9
247245	Superficial hemosiderosis of the CNS	3205	SWS	157801	Syndactyly, Malik-Percin type
79490	Superficial lymphangioma	242	Swyer syndrome	295012	Syndactyly, mitten type
79490	Superficial lymphatic malformation	90038	Sxt-HUS	357332	Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome
247245	Superficial siderosis	306731	Sydenham chorea	3253	Syndactyly-ectodermal dysplasia-cleft/lip palate
247245	Superficial siderosis of the central nervous system	1570	Sybrachydactyly of hands and feet	3259	Syndactyly-polydactyly-ear lobe syndrome
247245	Superficial siderosis of the CNS	60015	Symmetric parietal foramina	85203	Syndactyly-preaxial polydactyly-sternal deformity syndrome
247245	Superficial siderosis of the central nervous system	1314	Symmetrical thalamic calcifications	140952	Syndactyly-telecanthus-anogenital and renal malformations syndrome
247245	Superficial siderosis of the CNS	79098	Sympathetic ophthalmia	→1159	Syndesmodyplastic dwarfism
247245	Superficial siderosis of the CNS	79098	Sympathetic uveitis	2143	Syndrome of ocular and facial anomalies, telecanthus and deafness
247245	Superficial siderosis of the CNS	3246	Symphalangism with multiple anomalies of hands and feet	1447	Syndrome r(4)
247245	Superficial siderosis of the CNS	3250	Symphalangism, Cushing type	52	Syndromic bile duct paucity
247245	Superficial siderosis of the CNS	3237	Symphalangism-brachydactyly syndrome	261619	Syndromic bile duct paucity due to
88633	Superior limbic keratoconjunctivitis	465508	Symptomatic form of classic hemochromatosis		
155884	Superior palpebral coloboma	276630	Symptomatic form of Coffin-Lowry syndrome in female carriers		
180182	Supernumerary breasts	449291	Symptomatic form of fragile X syndrome in female carrier		
96170	Supernumerary der(22) syndrome				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	a JAG1 point mutation	295197	Synpolydactyly type 2		immunodeficiency due to CD45 deficiency
261629	Syndromic bile duct paucity due to a NOTCH2 point mutation	295199	Synpolydactyly type 3	276	T-B+ severe combined immunodeficiency due to gamma chain deficiency
261600	Syndromic bile duct paucity due to monosomy 20p12	295197	Synpolydactyly, Debeer type	169154	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency
84064	Syndromic diarrhea	295199	Synpolydactyly, Malik type	35078	T-B+ severe combined immunodeficiency due to JAK3 deficiency
84064	Syndromic diarrhea/Tricho-hepatointeric syndrome	295195	Synpolydactyly, Vordingborg type	276	T-B+ severe combined immunodeficiency, X-linked
77298	Syndromic microphthalmia type 3	3275	Synspondylism	86871	T-cell chronic lymphocytic leukemia
85275	Syndromic microphthalmia type 4	93926	Syntelencephaly	324294	T-cell immunodeficiency due to RHOH deficiency
178364	Syndromic microphthalmia type 5	840	Syringadenoma papilliferum	324294	T-cell immunodeficiency with epidermodysplasia verruciformis
139471	Syndromic microphthalmia type 6	840	Syringocystadenoma papilliferum	86872	T-cell large granular lymphocyte leukemia
2556	Syndromic microphthalmia type 7	314701	Systemic AL amyloidosis	86872	T-cell LGL leukemia
3434	Syndromic microphthalmia type 8	188	Systemic capillary leak syndrome	86886	T-cell lymphoma, AILD type
2470	Syndromic microphthalmia type 9	→528	Systemic cystic angiomas-Seip syndrome	86871	T-cell prolymphocytic leukemia
77299	Syndromic microphthalmia type 10	364033	Systemic EBV+ T-cell LPD of childhood	300857	T-cell/histiocyte rich large B cell lymphoma
178364	Syndromic microphthalmia/anophthalmia due to OTX2 mutation	364033	Systemic EBV-positive T-cell lymphoproliferative disease of childhood	86872	T-LGL
228426	Syndromic multisystem autoimmune disease due to Itch deficiency	364033	Systemic Epstein-Barr virus-positive T-cell lymphoproliferative disease of childhood	86871	T-PLL
98606	Syndromic orbital border hypoplasia	401996	Systemic karyomegaly	1350	Tabatznik syndrome
281090	Syndromic recessive X-linked ichthyosis	98849	Systemic mastocytosis with an associated clonal hematologic non-mast cell lineage disease	3384	TAC
281090	Syndromic RXLI	98849	Systemic mastocytosis with associated hematologic neoplasm	447896	TACH syndrome
457223	Syndromic sensorineural deafness due to combined oxidative phosphorylation defect	90069	Systemic monochloroacetate poisoning	457077	TAFRO syndrome
457223	Syndromic sensorineural deafness due to COXPD	439762	Systemic PAN	567	Takao syndrome
457223	Syndromic sensorineural hearing loss due to COXPD	439762	Systemic periarteritis nodosa	2905	Takatsuki syndrome
85274	Syndromic X-linked intellectual disability 7	439762	Systemic polyarteritis nodosa	3287	Takayasu arteritis
85279	Syndromic X-linked intellectual disability due to JARID1C mutation	85414	Systemic polyarthritis	487796	Takenouchi-Kosaki syndrome
85295	Syndromic X-linked intellectual disability type 10	158	Systemic primary carnitine deficiency	66529	Tako-Tsubo cardiomyopathy
85286	Syndromic X-linked intellectual disability type 11	90291	Systemic scleroderma	66529	Tako-Tsubo syndrome
319332	SYNE1-related AMC	90291	Systemic sclerosis	66529	Takotsubo cardiomyopathy
319332	SYNE1-related arthrogryposis multiplex congenita	220407	Systemic sclerosis sine scleroderma	66529	Takotsubo syndrome
3263	Syngnathia-cleft palate syndrome	85414	Systemic-onset JIA	101028	TALDO deficiency
3262	Syngnathia-multiple anomalies syndrome	85414	Systemic-onset juvenile idiopathic arthritis	2886	Talipes equinovarus-atrial septal defect-Robin sequence-persistence of the left superior vena cava syndrome
35098	Synostotic plagiocephaly	134	T2 deficiency	217335	Tall forehead-sparse hair-skin hyperextensibility-scoliosis syndrome
3273	Synovial sarcoma	99861	T-ALL	404443	Tall stature-intellectual disability-facial dysmorphism syndrome
3273	Synovialosarcoma	169160	T-B+ SCID due to CD3delta/CD3epsilon/CD3zeta	500095	Tall stature-intellectual disability-renal anomalies syndrome
793	Synovitis-acne-pustulosis-hyperostosis-osteitis syndrome	169157	T-B+ SCID due to CD45 deficiency	329191	Tall stature-scoliosis-macrodactyly of the great toes syndrome
93403	Synpolydactyly	276	T-B+ SCID due to gamma chain deficiency	329191	Tall stature-scoliosis-macrodactyly of the halluces syndrome
295195	Synpolydactyly type 1	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		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
50809	Talo-patello-scaphoid osteolysis		monoclonal gammopathy-perinephric-fluid collections-intrapulmonary shunting syndrome	352737	Temperature-sensitive oculocutaneous albinism type 1
31150	Tangier disease	3293	Telecanthus-hypertelorism-strabismus-pes cavus syndrome	284227	TEMPI syndrome
180	Tapetochoroidal dystrophy	2885	Telfer-Sugar-Jaeger syndrome	96184	Temple syndrome
98839	Tappeiner-Pfleger disease	488642	TELO2-related intellectual disability-neurodevelopmental disorder	420561	Temple-Baraitser syndrome
3320	TAR syndrome	1596	Telomeric 15q deletion syndrome	397	Temporal arteritis
65250	Tarlov cyst	36367	Telomeric deletion 1q	363417	Temtamy preaxial brachydactyly syndrome
2886	TARP syndrome	280	Telomeric deletion 4p	1777	Temtamy syndrome
99170	Tarsal kink syndrome	96145	Telomeric deletion 4q	1777	Temtamy-Shalash syndrome
1412	Tarsal-carpal coalition syndrome	1627	Telomeric deletion 5q	66627	Tenosynovial giant cell tumor
371	Tarui disease	96126	Telomeric deletion 7p	137834	Ter Haar syndrome
404443	Tatton-Brown-Rahman overgrowth syndrome	1636	Telomeric deletion 7q36	252018	Teratoma of the central nervous system
163654	Tattoo dysplasia	1642	Telomeric deletion 9p	141107	Teratoma of the nasopharynx
2731	Taurodontia-absent teeth-sparse hair syndrome	1580	Telomeric deletion 10p	363483	Teratoma of the testis
3289	Taurodontism	96148	Telomeric deletion 10q	169150	Terminal complement pathway deficiency
99045	Taussig-Bing syndrome	2308	Telomeric deletion 11q	88630	Terminal osseous dysplasia-pigmentary defects syndrome
→33364	Tay syndrome	96149	Telomeric deletion 12q	141242	Tessier number 1 cleft
845	Tay-Sachs disease	96150	Telomeric deletion 14q	141258	Tessier number 4 facial cleft
309239	Tay-Sachs disease, B1 variant	531	Telomeric deletion 17p	141261	Tessier number 5 facial cleft
309192	Tay-Sachs disease, B variant, adult form	1597	Telomeric deletion 17q	141265	Tessier number 6 facial cleft
309178	Tay-Sachs disease, B variant, infantile form	96129	Telomeric deletion 19p	325124	Testicular agenesis
309185	Tay-Sachs disease, B variant, juvenile form	96152	Telomeric deletion 20q	363494	Testicular non seminomatous germ cell tumor
90650	Taybi syndrome	1590	Telomeric deletion 13q	363494	Testicular non-dysgerminomatous germ cell tumor
2636	Taybi-Linder syndrome	96069	Telomeric duplication 1p36	983	Testicular regression syndrome
98960	TBCD	96070	Telomeric duplication 2p	842	Testicular seminoma
488632	TBCK-related intellectual disability syndrome	96094	Telomeric duplication 2q	842	Testicular seminomatous germ cell tumor
297	TBE	96071	Telomeric duplication 3p	363489	Testicular sex cord-stromal tumor
499004	TBM	96072	Telomeric duplication 4p	363483	Testicular teratoma
857	TBS	96096	Telomeric duplication 4q	3000	Testotoxicosis
2967	TCI deficiency	96097	Telomeric duplication 5q	3299	Tetanus
103918	TCP	1745	Telomeric duplication 6p	9	Tetra X
397959	TCR-alpha-beta+ T-cell deficiency	96098	Telomeric duplication 6q	294971	Tetra-amelia
397959	TCR-alpha-beta-positive T-cell deficiency	96074	Telomeric duplication 7p	3301	Tetraamelia-multiple malformations syndrome
2655	TD	96100	Telomeric duplication 8q	199310	Tetragametic chimerism
1860	TD1	96101	Telomeric duplication 9q	293284	Tetrahydrobiopterin-responsive HPA/PKU
93274	TD2	96102	Telomeric duplication 10q	293284	Tetrahydrobiopterin-responsive hyperphenylalaninemia/phenylketonuria
3352	TDO syndrome	96103	Telomeric duplication 11q	3303	Tetralogy of Fallot
1519	Teebi hypertelorism syndrome	96105	Telomeric duplication 13q	2564	Tetramelic monodactyly
1519	Teebi syndrome	1705	Telomeric duplication 14q	3305	Tetraploidy
2432	Teebi-Al Saleh-Hassoon syndrome	1707	Telomeric duplication 15q	3309	Tetrasomy 5p
1094	Teebi-Kaurah syndrome	96078	Telomeric duplication 16p	3310	Tetrasomy 9p
1974	Teebi-Naguib-Alawadi syndrome	96106	Telomeric duplication 16q	289522	Tetrasomy 11q24.1
3291	Teebi-Shaltout syndrome	3379	Telomeric duplication 17q	884	Tetrasomy 12p
3292	Tel Hashomer camptodactyly syndrome	1716	Telomeric duplication 18q	314588	Tetrasomy 15(q25-qter)
90389	Telangiectasia macularis eruptiva perstans	1717	Telomeric duplication 19q		
284227	Telangiectasia-erythrocytosis-	96107	Telomeric duplication 20q		
		96109	Telomeric duplication 22q		
		1762	Telomeric duplication Xq		
		1620	Telomeric monosomy 3p		
		75565	TEMF		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
314588	Tetrasomy 15q26		deafness syndrome	436169	Thrombomodulin-related coagulopathy
485405	Tetrasomy 16p12.1-p12.3	98960	Thiel-Behnke corneal dystrophy	54057	Thrombotic thrombocytopenic purpura
485405	Tetrasomy 16p12.1p12.3	3314	Thiemann disease, familial form	2251	Thumb deformity-alopecia-pigmentation anomaly syndrome
3307	Tetrasomy 18p	3235	Thies-Reis syndrome	294988	Thumb hypodactyly
96055	Tetrasomy 21	1506	Thin ribs-tubular bones-dysmorphism syndrome	294988	Thumb oligodactyly
9	Tetrasomy X	166424	Thinking seizures	1078	Thumb stiffness-brachydactyly-intellectual disability syndrome
140917	Teunissen-Cremers syndrome	→300	Thiolase deficiency	2919	Thurston syndrome
746	TFP deficiency	141030	Third branchial cleft anomaly	83471	Thymic aplasia
746	TFPD	141030	Third branchial cleft cyst	99868	Thymic carcinoma
225123	TFR2-related hemochromatosis	141030	Third branchial cleft fistula	99869	Thymic neuroendocrine carcinoma
476113	TFRC-related combined immunodeficiency	363444	THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	97289	Thymic neuroendocrine tumor
216729	TGA with cardiac malformation	3316	Thomas syndrome	3326	Thymic-renal-anal-lung dysplasia
99042	TGA with coarctation	2547	Thomas-Jewett-Raines syndrome	99867	Thymoma
66627	TGCT	2031	Thompson-Baritser syndrome	263310	Thymoma type A
3329	TH-SHFM	614	Thomsen and Becker disease	263324	Thymoma type AB
1780	Thakker-Donnai syndrome	2866	Thong-Douglas-Ferrante syndrome	263317	Thymoma type B
3312	Thalidomide embryopathy	1861	Thoracic dysplasia-hydrocephalus syndrome	169105	Thymoma-immunodeficiency syndrome
2655	Thanatophoric dwarfism	97330	Thoracic outlet compression syndrome	3327	Thyrocerebrorenal syndrome
1860	Thanatophoric dwarfism type 1	97330	Thoracic outlet syndrome	95716	Thyroid dyshormonogenesis
93274	Thanatophoric dwarfism type 2	1759	Thoraco-abdominal enteric duplication	95712	Thyroid ectopia
93274	Thanatophoric dwarfism-cloverleaf skull syndrome	1335	Thoraco-abdominal syndrome	95719	Thyroid hemiogenesis
2655	Thanatophoric dysplasia	3317	Thoracolaryngopelvic dysplasia	95720	Thyroid hypoplasia
1860	Thanatophoric dysplasia type 1	1803	Thoracolimb dysplasia, Rivera type	97285	Thyroid lymphoma
93274	Thanatophoric dysplasia type 2	268384	Thoracolumbosacral spina bifida aperta	91347	Thyroid stimulating hormone-secreting pituitary adenoma
→175	Thanatophoric dysplasia, Glasgow variant	268752	Thoracolumbosacral spina bifida cystica	2091	Thyroid-renal-digital anomalies
500095	Thauvin-Robinet-Faivre syndrome	1803	Thoracomelic dysplasia	79102	Thyrotoxic hypokalemic periodic paralysis
436169	THBD-related bleeding disorder	→2199	Hhost-Unna palmoplantar keratoderma	79102	Thyrotoxic periodic paralysis
436169	THBD-related coagulopathy	300857	THRLBCL	91347	Thyrotroph adenoma
99917	Theca (steroid-producing) cell cancer, not further specified	36258	Thromboangiitis obliterans	2768	Tibia vara Blount
99917	Theca steroid-producing cell malignant tumor of ovary, not further specified	329319	Thrombocythemia with distal limb defects	3329	Tibial aplasia-ectrodactyly syndrome
88633	Theodore's superior limbic keratoconjunctivitis	3204	Thrombocytopathy-asplenia-miosis syndrome	93322	Tibial hemimelia
88633	Theodore's syndrome	67044	Thrombocytopenia with congenital dyserythropoietic anemia	3329	Tibial hemimelia with split hand/foot malformation
268184	Thiamine-responsive BCKD deficiency	3320	Thrombocytopenia-absent radius syndrome	3329	Tibial hemimelia-ectrodactyly syndrome
268184	Thiamine-responsive branched-chain alpha-ketoacid dehydrogenase deficiency	457077	Thrombocytopenia-anasarca-fever-renal insufficiency-organomegaly syndrome	988	Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome
199348	Thiamine-responsive encephalopathy	3323	Thrombocytopenia-Robin sequence syndrome	93322	Tibial longitudinal meromelia
268184	Thiamine-responsive maple syrup urine disease	3002	Thrombocytopenic purpura, autoimmune	609	Tibial muscular dystrophy
49827	Thiamine-responsive megaloblastic anemia syndrome	436169	Thrombomodulin-related bleeding disorder	295028	Tibio-fibular fusion
49827	Thiamine-responsive megaloblastic anemia with diabetes mellitus and sensorineural deafness			295028	Tibio-fibular synostosis
268184	Thiamine-responsive MSUD			294981	Tibiofibular terminal transverse meromelia
2405	Thickened earlobes-conductive			297	Tick-borne encephalitis

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
91500	TINU syndrome	858	Toxoplasma embryopathy	280615	Transient neonatal cyanosis and anemia due to Toms River Hemoglobin
352540	TIO	93164	TPHA	99886	Transient neonatal diabetes mellitus
480483	TJP2 deficit	444463	TPP1I deficiency	329942	Transient neonatal glutaric aciduria type 2
488618	TKT deficiency	444463	TPP1I-related immunodeficiency, autoimmunity, and neurodevelopmental delay with impaired glycolysis and lysosomal expansion disease	329942	Transient neonatal glutaric aciduria type 2
420561	TMBTS		2950	329942	Transient neonatal MAD deficiency
→1394	TMCO1 defect syndrome	412022	Traboulsi syndrome	329942	Transient neonatal MADD
420611	TMD	3346	Tracheal agenesis	329942	Transient neonatal multiple acyl-CoA dehydrogenase deficiency
609	TMD	3347	Tracheobronchomegaly	391504	Transient neonatal myasthenia gravis
314667	TMEM165-CDG	3348	Tracheobronchopathia osteochondroplastica	93164	Transient pseudohypoaldosteronism
466703	TMEM199-CDG	3348	Tracheopathia osteoplastica	498359	Transient reactive papulotranslucent acrokeratoderma
1194	TMEM70-related mitochondrial encephalo-cardio-myopathy	3052	Tranebaerg-Svegaard syndrome	3402	Transient tyrosinemia of the neonate
99886	TNDM	101028	Transaldolase deficiency	3402	Transient tyrosinemia of the newborn
32960	TNF receptor 1-associated periodic syndrome	859	Transcobalamin deficiency	213746	Transitional cell carcinoma of the corpus uteri
64686	Tolosa-Hunt syndrome	2967	Transcobalamin I deficiency	280224	Transitional PMD
1920	Toluene embryopathy	859	Transcobalamin II deficiency	488618	Transketolase deficiency
640	Tomaculous neuropathy	2967	Transcobalamin-1 deficiency	319308	Translocation renal cell carcinoma
→314632	Tomé-Brunet-Fardeau syndrome	199247	Transcortin deficiency	85451	Transthyretin amyloid cardiopathy
454718	Tonic pupil-tendon areflexia syndrome	495	Transgrediens et progrediens palmoplantar keratoderma	85447	Transthyretin amyloid neuropathy
1547	Tonoki-Ohura-Niikawa syndrome	495	Transgrediens et progrediens PPK	85447	Transthyretin amyloid polyneuropathy
2228	Tooth and nail syndrome	87503	Transgrediens palmoplantar keratoderma of Siemens	85451	Transthyretin-related familial amyloid cardiomyopathy
3460	Torg-Winchester syndrome	420611	Transient abnormal myelopoiesis	→221061	Transverse limb deficiency-hemangioma syndrome
1827	Toriello syndrome	98871	Transient acquired pure red cell aplasia	180160	Transverse vaginal septum
3338	Toriello-Carey syndrome	79411	Transient bullous dermolysis of the newborn	32960	TRAPS syndrome
79347	Toriello-Higgins-Miller syndrome	98871	Transient erythroblastopenia of childhood	399175	Traumatic avascular necrosis
3339	Toriello-Lacassie-Droste syndrome	2312	Transient familial neonatal hyperbilirubinemia	399175	Traumatic AVN
51084	Torsade-de-pointes syndrome with short coupling interval	289877	Transient hyperammonemia of the newborn	165955	Traumatic myiasis
3341	Torticollis-keloids-cryptorchidism-renal dysplasia syndrome	169139	Transient hypogammaglobulinemia of infancy	861	Treacher-Collins syndrome
75326	Tortuosity of retinal arteries	300293	Transient infantile hypertriglyceridemia and fatty liver	→1215	Treft-Sanborn-Carey syndrome
97330	TOS	300293	Transient infantile hypertriglyceridemia and hepatosteatosis	103909	Trehalase deficiency
2701	Tosti syndrome	66529	Transient left ventricular apical ballooning syndrome	447896	Tremor-ataxia-central hypomyelination syndrome
294971	Total amelia	420611	Transient myeloproliferative disease	3350	Tremor-nystagmus-duodenal ulcer syndrome
49382	Total color blindness	420611	Transient myeloproliferative syndrome	64694	Trench fever
98994	Total early-onset cataract	391504	Transient neonatal acquired myasthenia	1822	Trevor disease
180126	Total septate uterus	391504	Transient neonatal autoimmune myasthenia gravis	99832	TRH resistance syndrome
268377	Total spina bifida aperta			2970	Triad syndrome
268748	Total spina bifida cystica			444463	TRIANGLE disease
2796	Touraine-Solente-Gole syndrome			85170	Triangular tibia-fibular aplasia syndrome
857	Townes syndrome				
857	Townes-Brocks syndrome				
537	Toxic epidermal necrolysis				
95455	Toxic epidermolyticus				
279894	Toxic maculopathy due to antimalarial drugs				
227972	Toxic oil syndrome				
293173	Toxic pustuloderma				
230800	Toxin-mediated infectious botulism				
230800	Toxin-mediated infective botulism				
3343	Toxocariasis				
858	Toxoplasma embryofetopathy				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
863	Trichinellosis		cardiomyovasculopathy	96074	Trisomy 7pter
863	Trichinosis	1308	Trigonocephaly C syndrome	96121	Trisomy 7q11.23
3352	Tricho-dento-osseous syndrome	3368	Trigonocephaly-bifid nose-acral anomalies syndrome	96061	Trisomy 8 mosaicism
84064	Tricho-hepato-enteric syndrome	3365	Trigonocephaly-broad thumbs syndrome	264450	Trisomy 8p
1264	Tricho-retino-dento-digital syndrome	3369	Trigonocephaly-short stature-developmental delay syndrome	251076	Trisomy 8p23.1
3351	Trichodental syndrome	401764	Trilineage bone marrow failure-developmental delay syndrome	1752	Trisomy 8q
3360	Trichodermal syndrome-intellectual disability syndrome	3374	Triopia	228399	Trisomy 8q12
3353	Trichodermodysplasia-dental alterations syndrome	868	Triose phosphate-isomerase deficiency	96100	Trisomy 8qter
228379	Trichodysplasia spinulosa	485405	Trip(16)(p12.1p12.3)	99776	Trisomy 9 mosaicism
1809	Trichodysplasia-abnormal dermatoglyphics-intellectual disability syndrome	444463	Tripeptidyl-peptidase II deficiency	236	Trisomy 9p
79129	Trichodysplasia-amelogenesis imperfecta syndrome	2950	Triphalangeal thumb-polysyndactyly syndrome	96101	Trisomy 9qter
3361	Trichodysplasia-xeroderma syndrome	2947	Triphalangeal thumbs-brachiectrodactyly syndrome	96063	Trisomy 10 mosaicism
864	Trichofolliculoma	3133	Triphalangeal thumbs-dislocation of patella syndrome	171929	Trisomy 10p
84064	Trichohepatoenteric syndrome	869	Triple A syndrome	276422	Trisomy 10q22.3q23.3
499182	Trichomatrical carcinoma	415	Triple H syndrome	96102	Trisomy 10qter
3363	Trichomegaly-retina pigmentary degeneration-dwarfism syndrome	3375	Triple X syndrome	300305	Trisomy 11p15.4
3355	Trichoodontoonychial dysplasia	3375	Triple-X syndrome	96103	Trisomy 11qter
3355	Trichoodontoonychial dysplasia with bone deficiency in frontoparietal region	3376	Triploidy	1698	Trisomy 12 mosaicism
565	Trichopoliodystrophy	3377	Trismus-pseudocamptodactyly syndrome	1699	Trisomy 12p
77258	Trichorhinophalangeal syndrome type 1 and 3	1692	Trisomy 1 mosaicism	3378	Trisomy 13
502	Trichorhinophalangeal syndrome type 2	96069	Trisomy 1pter	96105	Trisomy 13qter
→33364	Trichorrhexis nodosa syndrome	261344	Trisomy 1q	1703	Trisomy 14 mosaicism
33364	Trichothiodystrophy	250994	Trisomy 1q21.1	261229	Trisomy 14q11.2
→33364	Trichothiodystrophy type B	1723	Trisomy 2 mosaicism	488280	Trisomy 14q32
→33364	Trichothiodystrophy type C	96070	Trisomy 2pter	1705	Trisomy 14qter
→33364	Trichothiodystrophy type D	313947	Trisomy 2q23.1	1706	Trisomy 15 mosaicism
→33364	Trichothiodystrophy type E	294026	Trisomy 2q31.1	238446	Trisomy 15q11-q13
→33364	Trichothiodystrophy type F	96094	Trisomy 2qter	238446	Trisomy 15q11q13
→33364	Trichothiodystrophy type G	100071	Trisomy 3 mosaicism	1707	Trisomy 15qter
→33364	Trichothiodystrophy with congenital ichthyosis	96071	Trisomy 3pter	1708	Trisomy 16 mosaicism
→33364	Trichothiodystrophy-neurocutaneous syndrome syndrome	96095	Trisomy 3q26	261204	Trisomy 16p11.2p12.2
→33364	Trichothiodystrophy-osteosclerosis syndrome	251038	Trisomy 3q29	261243	Trisomy 16p13.11
→33364	Trichothiodystrophy-sun sensitivity syndrome	96059	Trisomy 4 mosaicism	96078	Trisomy 16pter
1209	Tricuspid atresia	1738	Trisomy 4p	96106	Trisomy 16qter
95457	Tricuspid valve agenesis	96072	Trisomy 4pter	1711	Trisomy 17 mosaicism
95458	Tricuspid valve prolapse	96096	Trisomy 4qter	261290	Trisomy 17p
221091	Trigeminal neuralgia	96060	Trisomy 5 mosaicism	1713	Trisomy 17p11.2
98908	Triglyceride deposit	1742	Trisomy 5p	477817	Trisomy 17p11.2-p12
		329802	Trisomy 5p13	477817	Trisomy 17p11.2p12
		228415	Trisomy 5q35	217385	Trisomy 17p13.3
		96097	Trisomy 5qter	139474	Trisomy 17q11.2
		1745	Trisomy 6pter	261272	Trisomy 17q12
		96098	Trisomy 6qter	217340	Trisomy 17q21.31
		1747	Trisomy 7 mosaicism	3379	Trisomy 17qter
		314034	Trisomy 7p22.1	3380	Trisomy 18
				1715	Trisomy 18p
				1716	Trisomy 18qter
				1717	Trisomy 19qter
				1724	Trisomy 20 mosaicism
				261318	Trisomy 20p
				96107	Trisomy 20qter
				870	Trisomy 21
				96068	Trisomy 22 mosaicism

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1727	Trisomy 22q11.2	805	Tuberous sclerosis	79431	Tyrosinase-negative oculocutaneous albinism
96109	Trisomy 22qter	805	Tuberous sclerosis complex	101150	Tyrosine hydroxylase deficiency
1738	Trisomy of the short arm of chromosome 4	88924	Tuberous sclerosis/polycystic kidney disease contiguous gene syndrome	101150	Tyrosine hydroxylase-deficient dopa-responsive dystonia
1742	Trisomy of the short arm of chromosome 5	2593	Tubular aggregate myopathy	69723	Tyrosinemia due to 4-hydroxyphenylpyruvate dioxygenase deficiency
236	Trisomy of the short arm of chromosome 9	100048	Tubular duplication of the esophagus	69723	Tyrosinemia due to 4-hydroxyphenylpyruvic acid oxidase deficiency
1715	Trisomy of the short arm of chromosome 18	73224	Tubular renal disease-cardiomyopathy syndrome	69723	Tyrosinemia due to HPD deficiency
3375	Trisomy X	467166	Tubulinopathy-associated dysgyria	28378	Tyrosinemia due to TAT deficiency
217377	Trisomy Xp11.22-p11.23	319325	Tubulocystic renal cell carcinoma	28378	Tyrosinemia due to tyrosine aminotransferase deficiency
261483	Trisomy Xq27.3-q28	91500	Tubulointerstitial nephritis and uveitis syndrome	882	Tyrosinemia type 1
261483	Trisomy Xq27.3q28	2997	Tucker syndrome	28378	Tyrosinemia type 2
1762	Trisomy Xq28	→2036	Tuffli-Laxova syndrome	69723	Tyrosinemia type 3
88629	Tritan colour blindness	1063	Tufted angioma	882	Tyrosinemia type I
88629	Tritanopia	3392	Tularemia	28378	Tyrosinemia type II
49827	TRMA	640	Tulip-bulb digger's palsy	69723	Tyrosinemia type III
1349	tRNA-LYS-related cardiomyopathy-hearing loss syndrome	32960	Tumor necrosis factor receptor 1 associated periodic syndrome	75840	UCMD
103918	Tropical calcific chronic pancreatitis	289539	Tumor susceptibility linked to germline BAP1 mutations	90002	UCTD
75565	Tropical endomyocardial fibrosis	352540	Tumor-induced osteomalacia	609	Udd myopathy
99654	Tropical pancreatic diabetes	879	Tungiasis	79238	UDP-galactose-4-epimerase deficiency
103918	Tropical pancreatitis	3225	Tungland-Bellman syndrome	178315	UES
764	Tropical pyomyositis	99053	Tunnel subaortic stenosis	205	UGT deficiency
289326	Tropical spastic paraparesis	211	Turban tumor syndrome	79234	UGT deficiency type 1
101000	Troyer syndrome	99818	Turcot syndrome with polyposis	79235	UGT deficiency type 2
983	TRS	881	Turner syndrome	3403	Uhl anomaly
313906	True congenital pancreatic cyst	99413	Turner syndrome due to structural X chromosome anomalies	2032	UIP
2138	True hermaphroditism	2614	Turner-Kieser syndrome	3404	Ulbright-Hodes syndrome
2512	True microcephaly	63440	Turricephaly	308	ULD
180074	True unicornuate uterus	95431	Twin to twin transfusion syndrome	3406	Ulerythema ophryogenes
3357	Trueb-Burg-Bottani syndrome	1461	Twisted atrioventricular connections	320	Ulick syndrome
3384	Truncus arteriosus	2889	Twisted hair	75840	Ullrich disease
228379	TS	2198	Tylosis-oesophageal carcinoma syndrome	2497	Ulna hypoplasia
352737	TS OCA type 1	477781	Type 1 condylar hyperplasia	2249	Ulna hypoplasia-intellectual disability syndrome
3173	Tsao-Ellingson syndrome	3255	Type 1 syndactyly-microcephaly-intellectual disability syndrome	1837	Ulna metaphyseal dysplasia syndrome
66627	TSGCT	→79259	Type 1C glycogenosis	93320	Ulnar clubhand
91347	TSH-oma	→79259	Type 1D glycogenosis	93320	Ulnar hemimelia
91347	TSH-secreting pituitary adenoma	93554	Type II mixed cryoglobulinemia	1122	Ulnar hypoplasia-lobster-claw deformity of feet syndrome
289326	TSP	99745	Typhoid	1122	Ulnar hypoplasia-split foot syndrome
3268	Tsukahara syndrome	99745	Typhoid fever	93320	Ulnar longitudinal meromelia
3387	Tsukahara-Kajii syndrome	99745	Typhoidal salmonellosis	3138	Ulnar-mammary syndrome
83317	Tsutsugamushi disease	90038	Typical hemolytic-uremic syndrome	52056	Ulnar/fibula ray defect-brachydactyly syndrome
83317	Tsutsugamushi fever	90038	Typical HUS	3405	Umbilical cord ulceration-intestinal atresia syndrome
54057	TTT	171436	Typical nemaline myopathy	209886	UMOD-associated familial juvenile
85447	TTR amyloid neuropathy	158766	Typical urticaria pigmentosa		
85451	TTR-related amyloid cardiomyopathy	1895	Typus Edinburgensis		
85451	TTR-related cardiac amyloidosis				
180242	Tubal cancer				
499004	Tubercular meningitis				
3389	Tuberculosis				
499004	Tuberculous meningitis				

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	hyperuricemic nephropathy	180079	Unicornuate uterus with rudimentary horn	2489	Upper limb defect-eye and ear abnormalities syndrome
209886	UMOD-associated FJHN	180074	Unicornuate uterus without rudimentary horn	295049	Upper limb hypertrophy
88950	UMOD-related ADTKD	93176	Unilateral congenital megacalycosis	2497	Upper limb mesomelic dysplasia
88950	UMOD-related autosomal dominant tubulointerstitial kidney disease	268947	Unilateral focal polymicrogyria	268740	Upper thoracic spina bifida aperta
35120	UMPH1 deficiency	101071	Unilateral hemispheric polymicrogyria	268770	Upper thoracic spina bifida cystica
3138	UMS	97363	Unilateral MCDK	2023	UPS
104078	Unclassified intestinal pseudoobstruction	99802	Unilateral megencephaly	93583	Upshaw-Schulman syndrome
98825	Unclassified mixed myelodysplastic/myeloproliferative syndrome	97363	Unilateral multicystic dysplastic kidney	488	Urachal cyst
98827	Unclassified myelodysplastic syndrome	268943	Unilateral polymicrogyria	431347	Urachal diverticulum
98825	Unclassified myelodysplastic/myeloproliferative disease	295012	Unilateral syndactyly of digits 2-5	431344	Urachal sinus
251328	Unclassified vasculitis	1464	Univentricular heart	530	Urbach-Wiethe disease
1410	Uncombable hair syndrome	99069	Univentricular heart with single atrio-ventricular valve	221145	Urban-Rifkin-Davis syndrome
1264	Uncombable hair-retinal pigmentary dystrophy-dental anomalies-brachydactyly syndrome	79146	Universal melanosis	3409	Urban-Rogers-Meyer syndrome
103920	Undetermined colitis	620	Universal mesentery	1839	Urban-Schosser-Spohn syndrome
442835	Undetermined early-onset epileptic encephalopathy	99104	Unroofed coronary sinus	94059	Uremic pruritus
442835	Undetermined EOEE	91140	Unspecified JIA	105	Urethral atresia
418951	Undifferentiated carcinoma of esophagus	91140	Unspecified juvenile idiopathic arthritis	35120	Uridine 5'-monophosphate hydrolase deficiency
424970	Undifferentiated carcinoma of liver and IBT	99139	Unstable hemoglobin disease	79238	Uridine diphosphate galactose-4-epimerase deficiency
424970	Undifferentiated carcinoma of liver and intrahepatic biliary tract	308	Unverricht-Lundborg disease	30	Uridine monophosphate synthetase deficiency
424080	Undifferentiated carcinoma of pancreas with osteoclast-like giant cells	251009	UPD(1)mat	210128	Urocanic aciduria
423786	Undifferentiated carcinoma of stomach	251004	UPD(1)pat	2704	Urofacial syndrome
213721	Undifferentiated carcinoma of the corpus uteri	96179	UPD(2)mat	98606	Urrets-Zavalia syndrome
90002	Undifferentiated connective tissue syndrome	96180	UPD(4)mat	79457	Urticaria pigmentosa
178315	Undifferentiated embryonal sarcoma of the liver	96190	UPD(5)pat	886	USH
418951	Undifferentiated esophageal carcinoma	96181	UPD(6)mat	231169	USH1
423786	Undifferentiated gastric carcinoma	96191	UPD(6)pat	231178	USH2
86830	Undifferentiated myeloproliferative disease	96182	UPD(7)mat	231183	USH3
2023	Undifferentiated pleomorphic sarcoma	96192	UPD(7)pat	886	Usher syndrome
178315	Undifferentiated sarcoma of the liver	96183	UPD(9)mat	231169	Usher syndrome type 1
251332	Unexplained long-lasting fever/inflammatory syndrome	231147	UPD(11)mat	231178	Usher syndrome type 2
83468	Unicameral bone cyst	96193	UPD(11)pat	231183	Usher syndrome type 3
		97678	UPD(13)mat	481665	USP18 deficiency
		99324	UPD(13)pat	2032	Usual interstitial pneumonia
		96184	UPD(14)mat	213610	Uterine carcinosarcoma
		96334	UPD(14)pat	180145	Uterine cervical aplasia and agenesis
		98754	UPD(15)mat	180139	Uterine hypoplasia
		98795	UPD(15)pat	439167	Uteroplacental vascular insufficiency
		96185	UPD(16)mat	180118	Uterus arcuatus
		96186	UPD(20)mat	180118	Uterus cordiformis
		96194	UPD(20)pat	180129	Uterus subseptus
		96187	UPD(21)mat	178338	UV-sensitive syndrome
		96195	UPD(21)pat	1473	Uveal coloboma-cleft lip and palate-intellectual disability
		96188	UPD(22)mat	39044	Uveal melanoma
		261519	UPD(X)mat	3437	Uveomenigitic syndrome
		261524	UPD(X)pat	99771	Uvular cleft
		3408	Upington disease	370109	v-AT
				887	VACTERL association

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
3412	VACTERL with hydrocephalus	228379	VATS	252175	Vestibular schwannoma
887	VACTERL/VATER association	898	VCAN-related vitreoretinopathy	892	VHL
25980	Vacuolar myopathy	600	VCPDM	493348	Vibratory angioedema
2478	Vacuolating megalencephalic leukoencephalopathy with subcortical cysts	289157	VDDI	493342	Vibratory urticaria
65681	Vaginal atresia	93160	VDDR II	1493	Vici syndrome
180247	Vaginal carcinoma	289157	VDDR-I	505395	VIDD
206489	Vaginal germ cell cancer	2460	VDEGS	3433	Viljoen-Kallis-Voges syndrome
206489	Vaginal germ cell malignant tumor	93160	VDRR II	3434	Viljoen-Smart syndrome
180247	Vaginal malignant epithelial tumor	1053	Vein of Galen aneurysm	97282	VIP-secreting tumor
158048	VAHS	1053	Vein of Galen arteriovenous malformations	97282	VIPoma
88639	Valine metabolic defect	3424	Velo-facial-skeletal syndrome	206991	Viral myositis
228123	Valley fever	567	Velocardiofacial syndrome	180176	Virginal breast hypertrophy
99054	Valvular pulmonary stenosis	29207	Venereal arthritis	99916	Virilizing ovarian tumor
1548	Van Benthem-Driessen-Hanveld syndrome	319234	Venezuelan hemorrhagic fever	158048	Virus-associated hemophagocytic syndrome
2806	Van Bogaert disease	357131	Venous cervical rib syndrome	228379	Virus-associated trichodysplasia spinulosa
2806	Van Bogaert encephalitis	357131	Venous costoclavicular syndrome	280068	Visceral calciphylaxis
3416	Van Buchem disease	357131	Venous hyperabduction syndrome	1876	Visceral myopathy-familial external ophthalmoplegia syndrome
1122	Van den Berghe-Dequecker syndrome	83454	Venous malformations with glomus cells	73246	Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome
3417	Van den Bosch syndrome	357131	Venous scalenus anticus syndrome	353344	Visible and exudative idiopathic juxtafoveolar retinal telangiectasis
2460	Van den Ende-Gupta syndrome	357131	Venous thoracic outlet compression syndrome	420556	Visual snow syndrome
216796	Van der Hoeve syndrome	357131	Venous thoracic outlet syndrome	3006	Vitamin B6-dependent seizures
2478	Van der Knaap syndrome	357131	Venous TOS	28	Vitamin B12-responsive methylmalonic acidemia
888	Van der Woude syndrome	505395	Ventilator-induced diaphragmatic dysfunction	79310	Vitamin B12-responsive methylmalonic acidemia type cbIA
314679	Van Maldergem syndrome	3201	Ventricular extrasystoles with syncopal episodes-periodically-Robin sequence syndrome	79311	Vitamin B12-responsive methylmalonic acidemia type cbIB
3419	Van Regemorter-Pierquin-Vamos syndrome	216694	Ventricular inversion	308442	Vitamin B12-responsive methylmalonic acidemia, type cbIDv2
73	Vanishing bone disease	99094	Ventricular septal defect with aortic insufficiency	28	Vitamin B12-responsive methylmalonic aciduria
983	Vanishing testes syndrome	216694	Ventriculoarterial and atrioventricular discordance	79310	Vitamin B12-responsive methylmalonic aciduria type cbIA
983	Vanishing testis syndrome	860	Ventriculoarterial discordance with atrioventricular concordance	79311	Vitamin B12-responsive methylmalonic aciduria, type cbIB
729	Vaquez disease	443988	Ventriculomegaly-cystic kidney disease	308442	Vitamin B12-responsive methylmalonic aciduria, type cbIDv2
2754	Váradí syndrome	508488	Verheij syndrome	27	Vitamin B12-unresponsive methylmalonic acidemia
2754	Váradí-Papp syndrome	2899	Verloes-Bourguignon syndrome	79312	Vitamin B12-unresponsive methylmalonic acidemia type mut-
454742	Variably protease-sensitive prionopathy	2496	Verloes-David syndrome	289916	Vitamin B12-unresponsive methylmalonic acidemia type mut0
314652	Variant ABeta2M amyloidosis	50817	Verloes-Deprez syndrome	27	Vitamin B12-unresponsive methylmalonic aciduria
79253	Variant phenylketonuria	2983	Verloes-Gillerot-Fryns syndrome	79312	Vitamin B12-unresponsive methylmalonic aciduria type mut-
79253	Variant PKU	2551	Verloes-Van Maldergem-de Marneffe syndrome	289916	Vitamin B12-unresponsive methylmalonic aciduria type mut0
79473	Variegate porphyria	3429	Verlooze Vanhorick-Brubakk syndrome	27	Vitamin B12-unresponsive methylmalonic aciduria
404553	Vasculitis due to ADA2 deficiency	70476	Vernal keratoconjunctivitis	79312	Vitamin B12-unresponsive methylmalonic aciduria type mut-
404553	Vasculitis due to DADA2	97282	Verner-Morrison syndrome	289916	Vitamin B12-unresponsive methylmalonic aciduria type mut0
353356	Vasoproliferative tumor of the ocular fundus	464318	Verrucous hemangioma	27	Vitamin B12-unresponsive methylmalonic aciduria
353356	Vasoproliferative tumor of the retina	79467	Verrucous nevus	79312	Vitamin B12-unresponsive methylmalonic aciduria type mut-
→261483	Vasquez-Hurst-Sotos syndrome	26793	Very long chain acyl-CoA dehydrogenase deficiency	289916	Vitamin B12-unresponsive
85128	Västerbotten dystrophy	431347	Vesicourachal diverticulum		
887	VATER association				
52047	Vater-like syndrome with pulmonary hypertension, abnormal ears and growth deficiency				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
	methylmalonic aciduria type mut0	99094	VSD with aortic insufficiency	1914	Warfarin embryofetopathy
289157	Vitamin D dependent rickets type I	357131	VTOS	1914	Warfarin embryopathy
289157	Vitamin D-dependency type I	494454	Vulvar adenocarcinoma	96061	Warkany syndrome
93160	Vitamin D-dependent rickets type II	494451	Vulvar basal cell carcinoma	90033	Warm AIHA
93160	Vitamin D-resistant rickets type II	137583	Vulvar intraepithelial neoplasia	1541	Warman-Mulliken-Hayward syndrome
1914	Vitamin K antagonist embryofetopathy	137583	Vulvar intraepithelial tumor	280558	Warsaw breakage syndrome
1914	Vitamin K antagonist embryopathy	494448	Vulvar squamous cell carcinoma	51636	Warts-hypogammaglobulinemia-infections-myelokathexis syndrome
1243	Vitelliform macular dystrophy type 2	83453	Vulvovaginal gingival syndrome	51636	Warts-infections-leukopenia-myelokatexis syndrome
179	Vitiliginous choroiditis	206492	Vulvovaginal rhabdomyosarcoma	69745	Warty dyskeratoma
898	Vitreoretinal degeneration, Wagner type	53696	Vuopala disease	906	WAS
26793	VLCAD deficiency	888	VWS	1046	Water-West syndrome
26793	VLCADD	2804	W syndrome	100067	Waterhouse-Friderichsen syndrome
386	VMC	2180	Waaler-Aarskog syndrome	97282	Watery diarrhea-hypokalemia-achlorhydria syndrome
443988	VMCKD	1106	Waardenburg anophthalmia syndrome	→636	Watson syndrome
2451	VMCM	3440	Waardenburg syndrome	33577	WCD
83454	VMGLOM	894	Waardenburg syndrome type 1	284395	WDFA
600	Vocal cord and pharyngeal distal myopathy	895	Waardenburg syndrome type 2	97282	WDHA syndrome
79124	VODI syndrome	352740	Waardenburg syndrome type 2 with ocular albinism	99971	WDLS
3437	Vogt-Koyanagi-Harada disease	896	Waardenburg syndrome type 3	603	WDM
494	Vohwinkel syndrome	897	Waardenburg syndrome type 4	3447	Weaver syndrome
79395	Vohwinkel syndrome with ichthyosis	894	Waardenburg syndrome type I	→3447	Weaver-like syndrome
2427	Volcke-Soekarman syndrome	895	Waardenburg syndrome type II	3448	Weaver-Williams syndrome
35737	Volubilis syndrome	896	Waardenburg syndrome type III	33577	Weber-Christian disease
83600	Von Economo encephalitis	896	Waardenburg syndrome with limb anomalies	33577	Weber-Christian panniculitis
364	Von Gierke disease	897	Waardenburg-Hirschsprung syndrome	1521	Webster-Deming syndrome
892	Von Hippel-Lindau disease	98960	Waardenburg-Jonker corneal dystrophy	900	Wegener granulomatosis
892	Von Hippel-Lindau syndrome	897	Waardenburg-Shah syndrome	228254	Weidman juvenile elastoma
238557	Von Hippel-Lindau-dependent polycythemia	280558	WABS	3449	Weill-Marchesani syndrome
386	Von Meyenburg complexes disease	466943	WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	3344	Weismann-Netter syndrome
636	Von Recklinghausen disease	247709	Wagenmann-Froboese syndrome	3344	Weismann-Netter-Stuhl syndrome
363700	Von Recklinghausen disease due to NF1 mutation or intragenic deletion	898	Wagner disease	3450	Weissenbacher-Zweymuller syndrome
3439	Von Voss-Cherstvoy syndrome	898	Wagner syndrome	284395	Well-differentiated fetal adenocarcinoma of the lung
903	Von Willebrand disease	893	WAGR syndrome	99971	Well-differentiated liposarcoma
166078	Von Willebrand disease type 1	90033	wAHA	213736	Well-differentiated neuroendocrine neoplasm of the endometrium
166081	Von Willebrand disease type 2	357332	Wahab syndrome	213736	Well-differentiated neuroendocrine tumor of the corpus uteri
166084	Von Willebrand disease type 2A	90033	wAIHA	213736	Well-differentiated neuroendocrine tumor of the endometrium
166087	Von Willebrand disease type 2B	2379	Waisman syndrome	263331	Well-differentiated thymic neuroendocrine carcinoma
166090	Von Willebrand disease type 2M	33226	Waldenström macroglobulinemia	146	Well-differentiated thyroid carcinoma
166093	Von Willebrand disease type 2N	90362	Waldmann disease	1373	Wellesley-Carman-French syndrome
166096	Von Willebrand disease type 3	1068	Walker-Dyson syndrome	901	Wells syndrome
466934	VPS11-related autosomal recessive hypomyelinating leukodystrophy	899	Walker-Warburg syndrome	2815	Wells-Jankovic syndrome
466934	VPS11-related autosomal recessive hypomyelinating leukoencephalopathy	1453	Wallis-Zieff-Goldblatt syndrome	83330	Werdnig-Hoffmann disease
369852	VPS45 deficiency	2510	WARBM	652	Wermer syndrome
353356	VPTR	2510	Warburg micro syndrome		
		3214	Warburg-Thomsen syndrome		
		1052	Warburton-Anyane-Yeboa syndrome		

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
3332	Werner mesomelic syndrome	94087	Winkelmann cytophagic panniculitis	2834	Wrinkly skin syndrome
902	Werner syndrome	2515	Winship-Viljoen-Leary syndrome	1667	WRS
1979	Werner-like syndrome due to combined growth factor deficiency	906	Wiskott-Aldrich syndrome	902	WS
3451	West syndrome	829	Wissler-Fanconi syndrome	894	WS1
83476	West-Nile encephalitis	2228	Witkop syndrome	895	WS2
83476	West-Nile fever	101068	Witschel dystrophy	896	WS3
2435	Westerhof-Beemer-Cormane syndrome	→280	Wittwer syndrome	897	WS4
83593	Western equine encephalitis	3237	WL syndrome	163746	WS4 plus
83593	Western equine encephalomyelitis	3344	WNS	2834	WSS
681	Westphall disease	247768	WNT4 deficiency	3466	WT limb-blood syndrome
952	Weyers acro dental dysostosis	1667	Wolcott-Rallison syndrome	3459	WTS
952	Weyers acrofacial dysostosis	280	Wolf-Hirschhorn syndrome	3411	Wunderlich syndrome
→2750	Whelan syndrome	3080	Wolff-Zimmermann syndrome	899	WWS
51636	WHIM syndrome	3463	Wolfram syndrome	53719	Wyburn-Mason syndrome
3452	Whipple disease	411590	Wolfram-like syndrome	96201	X small rings
2053	Whistling face syndrome	75233	Wolman disease	43	X-ALD
228290	White fibrous papulosis of the neck	3464	Woodhouse-Sakati syndrome	300373	X-LAG (X-linked acrogigantism)
2475	White forelock with malformations	2571	Woods-Black-Norbury syndrome	448348	X-LAG (X-linked acrogigantism) due to a point mutation
3207	White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome	→3255	Woods-Crouchman-Huson syndrome	448372	X-LAG (X-linked acrogigantism) due to dup(X)q(26)
370131	White platelet syndrome	170	Woolly hair	2182	X-linked aqueductal stenosis
171723	White sponge nevus	79414	Woolly hair nevus	448348	X-linked acrogigantism due to a point mutation
171723	White sponge nevus of Cannon	1409	Woolly hair-hypotrichosis-everted lower lip-outstanding ears syndrome	448372	X-linked acrogigantism due to Xq26 microduplication
1489	Whooping cough	420686	Woolly hair-palmoplantar hyperkeratosis syndrome	43	X-linked adrenoleukodystrophy
2779	Whyte-Murphy syndrome	65282	Woolly hair-palmoplantar hyperkeratosis-dilated cardiomyopathy syndrome	47	X-linked agammaglobulinemia
3454	Wieacker-Wolff syndrome	420686	Woolly hair-palmoplantar keratoderma syndrome	43	X-linked ALD
116	Wiedemann-Beckwith syndrome	65282	Woolly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome	88917	X-linked Alport syndrome
3455	Wiedemann-Rautenstrauch syndrome	170	Wooly hair	85278	X-linked Angelman-like syndrome
319182	Wiedemann-Steiner syndrome	79414	Wooly hair nevus	181	X-linked anhidrotic ectodermal dysplasia
85446	Wild type ABeta2-microglobulinic amyloidosis	1409	Wooly hair-hypotrichosis-everted lower lip-outstanding ears syndrome	85297	X-linked ataxia-deafness syndrome
85446	Wild type ABeta2M amyloidosis	65282	Wooly hair-palmoplantar hyperkeratosis-dilated cardiomyopathy syndrome	85292	X-linked ataxia-dementia syndrome
330001	Wild type ATTR amyloidosis	65282	Wooly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome	139583	X-linked auditory neuropathy with peripheral sensory neuropathy type 1
330001	Wild type ATTR-related amyloidosis	170	Wooly hair	1131	X-linked branchial arch syndrome
3456	Wildervanck syndrome	79414	Wooly hair nevus	481	X-linked BSMA
739	Willi-Prader syndrome	1409	Wooly hair-hypotrichosis-everted lower lip-outstanding ears syndrome	481	X-linked bulbospinal amyotrophy
904	Williams syndrome	65282	Wooly hair-palmoplantar hyperkeratosis-dilated cardiomyopathy syndrome	481	X-linked bulbospinal muscular atrophy
904	Williams-Beuren syndrome	65282	Wooly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome	391327	X-linked calvarial hyperostosis
411501	Williams-Campbell syndrome	166277	Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia	111	X-linked cardioskeletal myopathy and neutropenia
51636	WILM	3465	Worster-Drought syndrome	329235	X-linked central congenital hypothyroidism with late-onset macroorchidism
654	Wilms tumor	2790	Worth syndrome	329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement
220	Wilms tumor and pseudohermaphroditism	178475	Wound botulism	596	X-linked centronuclear myopathy
893	Wilms tumor-aniridia-genitourinary anomalies-intellectual disability syndrome	165955	Wound myiasis	139396	X-linked cerebral adrenoleukodystrophy
905	Wilson disease	2834	Wrinkled skin syndrome		
3459	Wilson-Turner syndrome				
3460	Winchester syndrome				
169095	Winged helix deficiency				

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
163961	X-linked cerebral-cerebellar-coloboma syndrome		atrophy type 3	89936	X-linked hypophosphatemia
101075	X-linked Charcot-Marie-Tooth disease type 1	35173	X-linked dominant chondrodyplasia punctata	89936	X-linked hypophosphatemic rickets
101076	X-linked Charcot-Marie-Tooth disease type 2	163966	X-linked dominant chondrodyplasia, Chassaing-Lacombe type	461	X-linked ichthyosis
101077	X-linked Charcot-Marie-Tooth disease type 3	163966	X-linked dominant chondrodyplasia-hydrocephaly-microphthalmia syndrome	231692	X-linked IGHD
101078	X-linked Charcot-Marie-Tooth disease type 4	443197	X-linked dominant erythropoietic protoporphyria	317476	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia
99014	X-linked Charcot-Marie-Tooth disease type 5	93951	X-linked dominant intellectual disability-epilepsy syndrome	2571	X-linked immunoneurologic disorder
352675	X-linked Charcot-Marie-Tooth disease type 6	443197	X-linked dominant protoporphyria	16	X-linked incomplete achromatopsia
35173	X-linked chondrodyplasia punctata type 2	139557	X-linked dSMA3	364028	X-linked intellectual disability due to GRIA3 anomalies
324601	X-linked cleft palate and ankyloglossia	363727	X-linked dyserythropoetic anemia with abnormal platelets and neutropenia	3242	X-linked intellectual disability due to PQBP1 mutations
431140	X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome	373	X-linked dysplasia gigantism syndrome	67045	X-linked intellectual disability with isolated growth hormone deficiency
431140	X-linked colobomatous microphthalmia-microcephaly-short stature-psychomotor retardation syndrome	53351	X-linked dystonia-parkinsonism	776	X-linked intellectual disability with marfanoid habitus
1497	X-linked complicated corpus callosum dysgenesis	75497	X-linked Ehlers-Danlos syndrome	85273	X-linked intellectual disability, Abidi type
306617	X-linked complicated spastic paraplegia type 1	98863	X-linked Emery-Dreifuss muscular dystrophy	85274	X-linked intellectual disability, Ahmad type
90001	X-linked cone dysfunction syndrome with myopia	293621	X-linked endothelial corneal dystrophy	85276	X-linked intellectual disability, Armfield type
95702	X-linked congenital adrenal hypoplasia	85294	X-linked epilepsy-learning disabilities-behavior disorders syndrome	1193	X-linked intellectual disability, Atkin type
67044	X-linked congenital dyserythropoietic anemia with thrombocytopenia	443197	X-linked erythropoietic protoporphyria	3056	X-linked intellectual disability, Brooks type
79495	X-linked congenital generalized hypertrichosis	500188	X-linked external auditory canal atresia-dilated internal auditory canal-facial dysmorphism syndrome	85293	X-linked intellectual disability, Cabezas type
565	X-linked copper deficiency	480880	X-linked facial dysmorphism-short stature-choanal atresia-intellectual disability syndrome limited to females	85277	X-linked intellectual disability, Cantagrel type
1661	X-linked corneal dermoid	480880	X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	163971	X-linked intellectual disability, Cilliers type
52503	X-linked creatine transporter deficiency	→994	X-linked fetal akinesia syndrome	→93950	X-linked intellectual disability, Fichera type
85453	X-linked cutaneous amyloidosis	139583	X-linked hereditary sensory and autonomic neuropathy with deafness	93947	X-linked intellectual disability, Golabi-Ito-Hall type
198	X-linked cutis laxa	139583	X-linked HSAN with deafness	→457240	X-linked intellectual disability, Gu type
383	X-linked deafness type 2	2182	X-linked HSAS	93952	X-linked intellectual disability, Hedera type
85321	X-linked deafness-intellectual disability syndrome	2182	X-linked hydrocephalus	163961	X-linked intellectual disability, Kroes type
139557	X-linked dHMN3	2182	X-linked hydrocephalus with stenosis of aqueduct of Sylvius	→1762	X-linked intellectual disability, Lubs type
1018	X-linked diffuse leiomyomatosis-Alport syndrome	1397	X-linked hydrocephalus-cerebellar agenesis-intellectual disability syndrome	85283	X-linked intellectual disability, Miles-Carpenter type
1145	X-linked distal arthrogryposis multiplex congenita	101088	X-linked hyper-IgM syndrome	163937	X-linked intellectual disability, Najm type
139557	X-linked distal hereditary motor neuropathy type 3	181	X-linked hypohidrotic ectodermal dysplasia	163956	X-linked intellectual disability, Nascimento type
139557	X-linked distal spinal muscular			85322	X-linked intellectual disability, Pai type
				93945	X-linked intellectual disability, Porteous type

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ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
→776	X-linked intellectual disability, Raymond type	85278	X-linked intellectual disability-craniofacial dysmorphism-epilepsy-ophthalmoplegia-cerebellar atrophy syndrome	163937	X-linked intellectual disability-microcephaly-pontocerebellar hypoplasia syndrome
3242	X-linked intellectual disability, Renpenning type	163979	X-linked intellectual disability-craniofacioskeletal syndrome	163971	X-linked intellectual disability-microcephaly-testicular failure syndrome
85285	X-linked intellectual disability, Schimke type	85280	X-linked intellectual disability-cubitus valgus-dysmorphism syndrome	→3057	X-linked intellectual disability-monoamine oxidase A metabolism anomaly syndrome
85323	X-linked intellectual disability, Seemanova type	1568	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	163956	X-linked intellectual disability-nail dystrophy-seizures syndrome
85286	X-linked intellectual disability, Shashi type	2958	X-linked intellectual disability-dysmorphism-cerebral atrophy syndrome	2898	X-linked intellectual disability-plagiocephaly syndrome
85324	X-linked intellectual disability, Shrimpton type	94083	X-linked intellectual disability-dystonia-dysarthria syndrome	85318	X-linked intellectual disability-precocious puberty-obesity syndrome
85287	X-linked intellectual disability, Siderius type	85319	X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome	3077	X-linked intellectual disability-psychosis-macroorchidism syndrome
3063	X-linked intellectual disability, Snyder type	85282	X-linked intellectual disability-epileptic seizures-hypogenitalism-microcephaly-obesity syndrome	85332	X-linked intellectual disability-retinitis pigmentosa syndrome
85278	X-linked intellectual disability, South African type	480907	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	3052	X-linked intellectual disability-seizures-psoriasis syndrome
85325	X-linked intellectual disability, Stevenson type	3459	X-linked intellectual disability-gynecomastia-obesity syndrome	457240	X-linked intellectual disability-short stature-overweight syndrome
85288	X-linked intellectual disability, Stocco Dos Santos type	85317	X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome	→702	X-linked intellectual disability-spastic paraparesia with iron deposits syndrome
85326	X-linked intellectual disability, Stoll type	3055	X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome	163982	X-linked intellectual disability-spastic quadriplegia syndrome
93950	X-linked intellectual disability, Sutherland-Haan type	59	X-linked intellectual disability-hypotonia syndrome	231692	X-linked isolated growth hormone deficiency
85328	X-linked intellectual disability, Turner type	85329	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	90625	X-linked isolated neurosensory deafness type DFN
163976	X-linked intellectual disability, Van Esch type	457260	X-linked intellectual disability-hypotonia-movement disorder syndrome	90625	X-linked isolated neurosensory hearing loss type DFN
→85293	X-linked intellectual disability, Vitale type	→1762	X-linked intellectual disability-hypotonia-recurrent Infections syndrome	90625	X-linked isolated sensorineural deafness type DFN
85290	X-linked intellectual disability, Wilson type	423479	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	792	X-linked juvenile retinoschisis
→280	X-linked intellectual disability, Wittwer type	85320	X-linked intellectual disability-macrocephaly-macroorchidism syndrome	482606	X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome
→59	X-linked intellectual disability, Zorick type	251383	X-linked intellectual disability-microcephaly-cortical malformation-thin habitus syndrome	79447	X-linked lethal multiple pterygium syndrome
85327	X-linked intellectual disability-acromegaly-hyperactivity syndrome			2148	X-linked lissencephaly type 1
85338	X-linked intellectual disability-ataxia-apraxia syndrome			452	X-linked lissencephaly with abnormal genitalia
324410	X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome			452	X-linked lissencephaly with ambiguous genitalia
137831	X-linked intellectual disability-cerebellar hypoplasia syndrome			452	X-linked lissencephaly-corpus callosum agenesis-genital anomalies syndrome
459070	X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome			2442	X-linked lymphoproliferative disease
85295	X-linked intellectual disability-choreoathetosis-abnormal behavior syndrome				
85330	X-linked intellectual disability-corpus callosum agenesis-spastic quadripareis syndrome				

→ This disease is deprecated and has been moved to another (see annex). The indicated Orphanumber should now be used.

ORPHA number	Disease name	ORPHA number	Disease name	ORPHA number	Disease name
1131	X-linked mandibulofacial dysostosis	90625	X-linked non-syndromic neurosensory hearing loss type DFN		5
1131	X-linked mandibulofacial dysostosis with limb anomalies	90625	X-linked non-syndromic sensorineural deafness type DFN	431272	X-linked SPMD
59306	X-linked McLeod syndrome	90625	X-linked non-syndromic sensorineural hearing loss type DFN	93349	X-linked spondyloepimetaphyseal dysplasia
319605	X-linked mendelian susceptibility to mycobacterial diseases	293707	X-linked Ohdo syndrome	168544	X-linked spondylometaphyseal dysplasia
319623	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency	306597	X-linked Opitz BBB/G syndrome	383	X-linked stapes gusher syndrome
319612	X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency	306597	X-linked Opitz G/BBB syndrome	852	X-linked thrombocytopenia with normal platelets
319612	X-linked mendelian susceptibility to mycobacterial diseases due to NEMO deficiency	306597	X-linked Opitz syndrome	3467	Xanthic urolithiasis
435938	X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	391330	X-linked osteoporosis with fractures	93602	Xanthine dehydrogenase and xanthine aldehyde oxidase dual deficiency
383	X-linked mixed conductive and neurosensory deafness	363654	X-linked parkinsonism-spasticity syndrome	93601	Xanthine dehydrogenase deficiency
383	X-linked mixed conductive and neurosensory hearing loss	1175	X-linked progressive cerebellar ataxia	93601	Xanthine oxidase deficiency
383	X-linked mixed conductive and sensorineural deafness	1652	X-linked recessive hypercalciuric hypophosphatemic rickets	93601	Xanthine oxidoreductase deficiency
383	X-linked mixed conductive and sensorineural hearing loss	83648	X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome	3467	Xanthine stone disease
383	X-linked mixed deafness with perilymphatic gusher	1652	X-linked recessive nephrolithiasis	93601	Xanthinuria type I
504530	X-linked Moesin-associated immunodeficiency	54	X-linked recessive ocular albinism	93602	Xanthinuria type II
319605	X-linked MSMD	85453	X-linked reticulate pigmentary disorder	158003	Xanthoma disseminatum
319623	X-linked MSMD due to CYBB deficiency	1852	X-linked retinal dysplasia	79433	Xanthous oculocutaneous albinism
319612	X-linked MSMD due to IKBKG deficiency	792	X-linked retinoschisis	79155	Xanthurenic aciduria
319612	X-linked MSMD due to NEMO deficiency	431272	X-linked scapuloperoneal muscular dystrophy	67044	XDAT
25980	X-linked myopathy with excessive autophagy	431272	X-linked scapuloperoneal syndrome	93602	XDH and AOX dual deficiency
178461	X-linked myopathy with postural muscle atrophy	86788	X-linked severe congenital neutropenia	93601	XDH deficiency
596	X-linked myotubular myopathy	75563	X-linked sideroblastic anemia	53351	XDP
456328	X-linked myotubular myopathy-abnormal genitalia syndrome	2802	X-linked sideroblastic anemia and spinocerebellar ataxia	293621	XECD
85334	X-linked neurodegenerative syndrome, Bertini type	2802	X-linked sideroblastic anemia with ataxia	910	Xeroderma pigmentosum
85336	X-linked neurodegenerative syndrome, Hamel type	1436	X-linked skeletal dysplasia-intellectual disability syndrome	90342	Xeroderma pigmentosum variant
314978	X-linked non progressive cerebellar ataxia	99015	X-linked spastic paraplegia type 2	→910	Xeroderma pigmentosum with neurologic manifestation
777	X-linked non-specific intellectual disability	100997	X-linked spastic paraplegia type 16	220295	Xeroderma pigmentosum-Cockayne syndrome complex
777	X-linked non-syndromic intellectual disability	171607	X-linked spastic paraplegia type 34	75496	XGPT deficiency
90625	X-linked non-syndromic neurosensory deafness type DFN	3175	X-linked spasticity-intellectual disability-epilepsy syndrome	181	XHED
		481	X-linked spinal and bulbar muscular atrophy	101088	XHIGM
		1145	X-linked spinal muscular atrophy type 2	412069	Xia-Gibbs syndrome
		404521	X-linked spinal muscular atrophy with respiratory distress	3469	XK aprosencephaly syndrome
		85297	X-linked spinocerebellar ataxia type 3	3469	XK syndrome
		85292	X-linked spinocerebellar ataxia type 4	452	XLAG (X-linked lissencephaly with abnormal genitalia) syndrome
		314978	X-linked spinocerebellar ataxia type	596	XLCNM
				443197	XLDPP
				264580	XLG
				89936	XLH
				461	XLI
				596	XLMTM
				54	XLOA
				306597	XLOS
				2442	XLP
				443197	XLP
				85453	XLPDR
				443197	XLPP

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ORPHA number	Disease name
792	XLRS
75563	XLSA
2802	XLSA-A
231393	XLTT
25980	XMEA
317476	XMEN
178461	XMPMA
93601	XO deficiency
93601	XOR deficiency
910	XP
220295	XP/CS complex
261476	Xp21 contiguous gene deletion syndrome
261476	Xp21 microdeletion syndrome
284180	Xp22.13p22.2 duplication syndrome
1643	Xp22.3 microdeletion syndrome
363654	XPDS
90342	XPV
314389	Xq12-q13.3 duplication syndrome
1018	Xq22.3 microdeletion syndrome
261483	Xq27.3-q28 microduplication syndrome
261483	Xq27.3q28 duplication syndrome
456328	Xq28 contiguous gene deletion syndrome
243	XX female gonadal dysgenesis
2855	XX gonadal dysgenesis-deafness syndrome
393	XX, male syndrome
243	XX-GD
3375	XXX syndrome
983	XY gonadal agenesis syndrome
168558	XY sex reversal-adrenal failure
1770	XY type gonadal dysgenesis-associated anomalies syndrome
2843	Xylitol dehydrogenase deficiency
75496	Xylosylprotein 4-beta-galactosyltransferase deficiency
370930	XYLT1-CDG
8	XYY syndrome
8	Y disomy
2616	Yakut short stature syndrome
99829	Yellow fever
99829	Yellow Jack
662	Yellow nail syndrome
79434	Yellow oculocutaneous albinism
3214	Yemenite deaf-blind hypopigmentation syndrome
707	Yersiniosis
99829	YF
662	YNS
876	Yolk sac tumor
252006	Yolk sac tumor of central nervous system

ORPHA number	Disease name
	system
252006	Yolk sac tumor of CNS
2828	YOPD
2255	Yorifuji-Okuno syndrome
3240	Yoshimura-Takeshita syndrome
488642	You-Hoover-Fong syndrome
314485	Young adult-onset dHMN
314485	Young adult-onset distal hereditary motor neuropathy
3471	Young syndrome
3055	Young-Hughes syndrome
2828	Young-onset Parkinson disease
477817	Yuan-Harel-Lupski syndrome
3472	Yunis-Varon syndrome
506358	YY1 haploinsufficiency syndrome
319213	Zambian hemorrhagic fever
98912	ZASP-related myofibrillar myopathy
97240	Zebra body myopathy
217017	Zechi-Ceide syndrome
912	Zellweger syndrome
369942	Zellweger-like contiguous gene deletion syndrome
50812	Zellweger-like syndrome without peroxisomal anomalies
911	Zeta-associated-protein 70 deficiency
448237	Zika virus disease
448237	Zika virus infection
3301	Zimmer phocomelia
3473	Zimmermann-Laband syndrome
439196	Zinc-responsive necrolytic acral erythema
1775	Zinsser-Engman-Cole syndrome
3253	Zlotogora-Ogur syndrome
3253	Zlotogora-Zilberman-Tenenbaum syndrome
913	Zollinger-Ellison syndrome
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

→ This disease is deprecated and has been moved to another (see annex). The indicated Orphanumber should now be used.

List of diseases to be used instead of deprecated entities

→ Use these ORPHA number		instead of the deprecated entities	
ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities
59	Allan-Herndon-Dudley syndrome	85337	X-linked intellectual disability, Zorick type
113	Bazex-Dupré-Christol syndrome	79458	Oley syndrome
113	Bazex-Dupré-Christol syndrome	79458	Congenital hypotrichosis-milia syndrome
138	CHARGE syndrome	1474	Colobomatous-microphthalmia-heart disease-hearing loss syndrome
138	CHARGE syndrome	1474	Hittner-Hirsch-Kreh syndrome
144	Lynch syndrome	99817	Non-polyposis Turcot syndrome
175	Cartilage-hair hypoplasia	1838	Metaphyseal dysplasia without hypotrichosis
175	Cartilage-hair hypoplasia	1838	Cartilage-hair hypoplasia-like-skeletal dysplasia without hypotrichosis syndrome
175	Cartilage-hair hypoplasia	93275	Thanatophoric dysplasia, Glasgow variant
193	Cohen syndrome	3084	Mirhosseini-Holmes-Walton syndrome
193	Cohen syndrome	3084	Pigmentary retinopathy-intellectual disability syndrome
193	Cohen syndrome	2829	Partington-Anderson syndrome
193	Cohen syndrome	3271	Radio-ulnar synostosis-retinal pigment abnormalities syndrome
193	Cohen syndrome	3271	Buntinx-Lormans-Martin syndrome
244	Primary ciliary dyskinesia	98861	Primary ciliary dyskinesia, Kartagener type
244	Primary ciliary dyskinesia	98861	Dextrocardia-bronchiectasis-sinusitis syndrome
244	Primary ciliary dyskinesia	98861	Immotile cilia syndrome, Kartagener type
244	Primary ciliary dyskinesia	98861	Kartagener syndrome
244	Primary ciliary dyskinesia	98861	Siewert syndrome
280	Wolf-Hirschhorn syndrome	85291	X-linked intellectual disability, Wittwer type
280	Wolf-Hirschhorn syndrome	85291	Wittwer syndrome
280	Wolf-Hirschhorn syndrome	98788	Pitt-Rogers-Danks syndrome
280	Wolf-Hirschhorn syndrome	98788	Intellectual disability-dysmorphism-intrauterine growth retardation syndrome
288	Hereditary elliptocytosis	98867	Hereditary pyropoikilocytosis
288	Hereditary elliptocytosis	98864	Common hereditary elliptocytosis
288	Hereditary elliptocytosis	98865	Homozygous hereditary elliptocytosis

→ Use these ORPHA number		instead of the deprecated entities	
ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities
300	Bifunctional enzyme deficiency	2981	Pseudo-Zellweger syndrome
300	Bifunctional enzyme deficiency	2981	Thiolase deficiency
392	Holt-Oram syndrome	1940	Shoulder and thorax deformity-congenital heart disease syndrome
528	Berardinelli-Seip congenital lipodystrophy	1060	Systemic cystic angiomas-Seip syndrome
528	Berardinelli-Seip congenital lipodystrophy	1060	Brunzell syndrome
636	Neurofibromatosis type 1	3444	Watson syndrome
636	Neurofibromatosis type 1	3444	Pulmonic stenosis with 'café-au-lait' spots
636	Neurofibromatosis type 1	2029	Multiple non-ossifying fibromatoses
636	Neurofibromatosis type 1	2029	Jaffe-Campanacci syndrome
646	Niemann-Pick disease type C	79289	Niemann-Pick disease type D
646	Niemann-Pick disease type C	79289	Niemann-Pick disease, Nova Scotia type
672	Pallister-Hall syndrome	2113	Congenital hypothalamic hamartoma syndrome
672	Pallister-Hall syndrome	2113	CHHS
682	Hyperkalemic periodic paralysis	680	Normokalemic periodic paralysis
682	Hyperkalemic periodic paralysis	680	NormoKPP
682	Hyperkalemic periodic paralysis	680	NormoPP
682	Hyperkalemic periodic paralysis	680	Normokalemic PP
682	Hyperkalemic periodic paralysis	680	Periodic paralysis type 3
682	Hyperkalemic periodic paralysis	680	Potassium-sensitive normokalemic periodic paralysis
702	Pelizaeus-Merzbacher disease	85333	X-linked intellectual disability-spastic paraparesis with iron deposits syndrome
702	Pelizaeus-Merzbacher disease	85333	Arena syndrome
776	X-linked intellectual disability with marfanoid habitus	163953	X-linked intellectual disability, Raymond type
782	Axenfeld-Rieger syndrome	1831	De Hauwere syndrome
782	Axenfeld-Rieger syndrome	1831	De Hauwere-Chitty syndrome
782	Axenfeld-Rieger syndrome	1831	Iris dysplasia-hypertelorism-deafness syndrome
794	Saethre-Chotzen syndrome	1219	Aurocephalosyndactyly
794	Saethre-Chotzen syndrome	1219	Auralcephalosyndactyly
794	Saethre-Chotzen syndrome	1219	Kurczynski-Casperton syndrome
794	Saethre-Chotzen syndrome	3106	Robinow-Sorauf syndrome
798	Schinzel-Giedion syndrome	3118	Rudiger syndrome
823	Isolated spina bifida	93968	Meningocele
869	Triple A syndrome	99777	Achalasia-alacrimia syndrome
897	Waardenburg-Shah syndrome	918	ABCD syndrome

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ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities	ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities
897	Waardenburg-Shah syndrome	918	Albinism-black lock-cell migration disorder of the neurocytes of the gut-sensorineural deafness syndrome	1200	Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome	77302	Oculo-oto-facial dysplasia
910	Xeroderma pigmentosum	1569	De Sanctis-Cacchione syndrome	1215	Autosomal dominant optic atrophy plus syndrome	3349	Treft-Sanborn-Carey syndrome
910	Xeroderma pigmentosum	1569	Xeroderma pigmentosum with neurologic manifestation	1215	Autosomal dominant optic atrophy plus syndrome	3349	Optic atrophy-ophthalmoplegia-ptosis-deafness-myopathy syndrome
912	Zellweger syndrome	1271	Bowen syndrome	1215	Autosomal dominant optic atrophy plus syndrome	3212	Autosomal dominant optic atrophy and congenital deafness
955	Acroosteolysis dominant type	2853	Serpentine fibula-polycystic kidneys syndrome	1215	Autosomal dominant optic atrophy plus syndrome	3212	Konigsmark-Knox-Hussels syndrome
955	Acroosteolysis dominant type	2853	Exner syndrome	1234	Bartsocas-Papas syndrome	79446	Multiple pterygium syndrome, Aslan type
969	Acromicric dysplasia	2569	Moore-Federman syndrome	1263	Boomerang dysplasia	156723	Piepkorn dysplasia
969	Acromicric dysplasia	2569	Dwarfism-stiff joint-ocular abnormalities syndrome	1263	Boomerang dysplasia	156723	Short ribs-craniosynostosis-polysyndactyly syndrome
994	Fetal akinesia deformation sequence	995	X-linked fetal akinesia syndrome	1299	Branchioskeletogenital syndrome	157788	Hypospadias-hypertelorism-coloboma and deafness syndrome
994	Fetal akinesia deformation sequence	995	Holmes-Benacerraf syndrome	1359	Carney complex	623	NAME syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	1401	CHAND syndrome	1359	Carney complex	623	Nevi-atrial myxoma-myxoid neurofibromata-ephelides syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	1401	Baughman syndrome	1394	Cerebrofaciothoracic dysplasia	228407	Craniofacial dysmorphismskeletal anomalies-intellectual disability syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	1401	CHANDS	1394	Cerebrofaciothoracic dysplasia	228407	TMCO1 defect syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	1401	Curly hair-ankyloblepharon-nail dysplasia syndrome	1426	Greenberg dysplasia	99645	Dappled diaphyseal dysplasia
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	3022	Rapp-Hodgkin syndrome	1466	COFS syndrome	1317	CAMFAK syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	3022	Anhidrotic ectodermic dysplasia-cleft lip/palate syndrome	1466	COFS syndrome	1317	CAMAK syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	3022	Ectodermal dysplasia syndrome, Rapp-Hodgkin type	1466	COFS syndrome	1317	Cataract-microcephaly-arthrogryposis-kyphosis syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	3022	Ectodermal dysplasia, Rapp-Hodgkin type	1466	COFS syndrome	1317	Cataract-microcephaly-failure to thrive-kyphoscoliosis syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	3022	RHS	1487	Cooks syndrome	2355	Kumar-Levick syndrome
1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	99694	Alveolar synchia-ankylodlepharon-ectodermal dysplasia syndrome	1487	Cooks syndrome	2355	Nail dysplasia-camptodactyly-brachydactyly type B syndrome
1159	Progressive pseudorheumatoid arthropathy of childhood	2654	Syndesmodysplastic dwarfism	1509	Coxopodopatellar syndrome	3112	Patella aplasia-coxa var-tarsal synostosis syndrome
1159	Progressive pseudorheumatoid arthropathy of childhood	2654	Laplane-Fontaine-Lagardere syndrome	1643	Xp22.3 microdeletion syndrome	431	Ichthyosis-male hypogonadism syndrome
				1658	Absence of fingerprints-congenital milia syndrome	1235	Ectodermal dysplasia-absent dermatoglyphs syndrome
				1658	Absence of fingerprints-congenital milia syndrome	1235	Basan syndrome
				1762	Trisomy Xq28	85281	MECP2 duplication syndrome
				1762	Trisomy Xq28	85281	Lubs-Arena syndrome
				1762	Trisomy Xq28	85281	X-linked intellectual disability, Lubs type

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ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities	ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities
1762	Trisomy Xq28	85281	X-linked intellectual disability-hypotonia-recurrent Infections syndrome	2512	Autosomal recessive primary microcephaly	52183	Premature chromosome condensation with microcephaly and intellectual disability
1768	Familial caudal dysgenesis	1850	Renal dysplasia-megalocystis-sirenomelia syndrome	2526	Microcephaly-lymphedema-chorioretinopathy syndrome	1432	Autosomal dominant chorioretinopathy-microcephaly syndrome
1768	Familial caudal dysgenesis	1850	Selig-Benacerraf-Greene syndrome	2609	Isolated complex I deficiency	936	Succinic acidemia
1855	Spondyloenchondrodysplasia	50816	Spondylometaphyseal dysplasia with combined immunodeficiency	2616	3M syndrome	2661	Dwarfism-tall vertebrae syndrome
1855	Spondyloenchondrodysplasia	50816	Roifman-Melamed syndrome	2637	Microcephalic osteodysplastic primordial dwarfism type II	46658	Primordial short stature-microdontia-opalescent and rootless teeth syndrome
1855	Spondyloenchondrodysplasia	50816	SPENCDI	2686	Cyclic neutropenia	2689	Intermittent neutropenia
1855	Spondyloenchondrodysplasia	50816	Spondyloenchondrodysplasia with immune dysregulation	2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	1981	Fanconi syndrome-ichthyosis-dysmorphism syndrome
1896	EEC syndrome	1888	Ectrodactyly-ectodermal dysplasia without clefting syndrome	2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	1981	Deal-Barrat-Dillon syndrome
1896	EEC syndrome	1888	EEC syndrome without cleft lip/palate	2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	3438	Biliary tract malformation-renal failure syndrome
1896	EEC syndrome	1889	Ectrodactyly-cleft palate syndrome	2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	3438	Cholestatic jaundice-renal tubular insufficiency syndrome
1896	EEC syndrome	1889	ECP syndrome	2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	3438	Lutz-Richner-Landolt syndrome
1896	EEC syndrome	2389	Lewis-Pashayan syndrome	2707	Oculocerebrofacial syndrome, Kaufman type	329255	Blepharophimosis-intellectual disability syndrome due to UBE3B deficiency
1896	EEC syndrome	2389	Cleft lip/palate-ectrodactyly syndrome	2712	Oculofaciocardiodental syndrome	3013	Radiculomegaly of canine teeth- congenital cataract
1900	Ehlers-Danlos syndrome, kyphoscoliotic type	2691	Nevo syndrome	2712	Oculofaciocardiodental syndrome	3013	Marashi-Gorlin syndrome
1900	Ehlers-Danlos syndrome, kyphoscoliotic type	2691	Cerebral gigantism, Nevo type	2750	Orofaciodigital syndrome type 1	90649	Orofaciodigital syndrome type 7
2036	Scalp-ear-nipple syndrome	3391	Odonto-onycho-hypohidrotic dysplasia-midline scalp defects syndrome	2750	Orofaciodigital syndrome type 1	90649	OFD7
2036	Scalp-ear-nipple syndrome	3391	Ectodermal dysplasia-adrenal cyst syndrome	2750	Orofaciodigital syndrome type 1	90649	Oral-facial-digital syndrome type 7
2036	Scalp-ear-nipple syndrome	3391	Tuffli-Laxova syndrome	2750	Orofaciodigital syndrome type 1	90649	Whelan syndrome
2052	Fraser syndrome	2051	Fraser-like syndrome	2796	Pachydermoperiostosis	964	Acromegaly-cutis verticis gyrata-corneal leukoma syndrome
2199	Epidermolytic palmoplantar keratoderma	496	Thost-Unna palmoplantar keratoderma	2882	Sitosterolemia	101022	Mediterranean macrothrombocytopenia
2199	Epidermolytic palmoplantar keratoderma	496	Non-epidermolytic palmoplantar keratoderma	2909	Rothmund-Thomson syndrome	3333	Connective tissue dysplasia, Spellacy type
2199	Epidermolytic palmoplantar keratoderma	89833	Palmoplantar keratoderma with tonotubular keratin	2909	Rothmund-Thomson syndrome	3333	Spellacy-Gibbs-Watts syndrome
2353	Schilbach-Rott syndrome	1251	Blepharofaciokkeletal syndrome	2995	Baraitser-Winter cerebrofrontofacial syndrome	2649	Short stature-intellectual disability-eye anomalies-cleft lip/palate syndrome
2353	Schilbach-Rott syndrome	1251	Richieri Costa-Guion Almeida-Rodini syndrome				
2462	Shprintzen-Goldberg syndrome	2352	Kozlowski-Brown-Hardwick syndrome				
2470	Matthew-Wood syndrome	91129	Anophthalmia-heart and pulmonary anomalies-intellectual disability syndrome				
2510	Micro syndrome	2895	Pinsky-Di George-Harley syndrome				
2510	Micro syndrome	2895	Microphthalmia-intellectual disability syndrome				

→ Use these ORPHA number		instead of the deprecated entities		→ Use these ORPHA number		instead of the deprecated entities	
ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities	ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities
2995	Baraits-Winter cerebrofrontofacial syndrome	2649	Richieri Costa-Guion Almeida syndrome	3464	Woodhouse-Sakati syndrome	1011	Devriendt-Legius-Fryns syndrome
2995	Baraits-Winter cerebrofrontofacial syndrome	94084	Cerebro-oculo-facial-lymphatic syndrome	3471	Young syndrome	1301	Bronchiectasis-oligospermia syndrome
2995	Baraits-Winter cerebrofrontofacial syndrome	94084	Fryns-Aftimos syndrome	33001	Lymphedema-distichiasis syndrome	1683	Distichiasis-congenital heart defects-peripheral vascular anomalies syndrome
3027	Caudal regression sequence	1773	Sacrococcygeal dysgenesis association	33001	Lymphedema-distichiasis syndrome	2419	Lymphedema-ptosis syndrome
3057	Monoamine oxidase A deficiency	3065	X-linked intellectual disability-monoamine oxidase A metabolism anomaly syndrome	33364	Trichothiodystrophy	1245	BIDS syndrome
3157	Septo-optic dysplasia spectrum	1102	Anophthalmia-hypothalamo-pituitary insufficiency syndrome	33364	Trichothiodystrophy	1245	Amish brittle hair syndrome
3157	Septo-optic dysplasia spectrum	1102	14q22 microdeletion syndrome	33364	Trichothiodystrophy	1245	Trichothiodystrophy type D
3157	Septo-optic dysplasia spectrum	1102	Al Frayh-Facharzt-Haque syndrome	33364	Trichothiodystrophy	670	PIBIDS syndrome
3157	Septo-optic dysplasia spectrum	1102	Monosomy 14q22	33364	Trichothiodystrophy	670	Trichothiodystrophy type F
3157	Septo-optic dysplasia spectrum	1678	Dincsoy-Salih-Patel syndrome	33364	Trichothiodystrophy	670	Trichothiodystrophy-sun sensitivity syndrome
3157	Septo-optic dysplasia spectrum	1678	Facial dysmorphism-ambiguous genitalia-hypopituitarism-short limbs syndrome	33364	Trichothiodystrophy	453	IBIDS syndrome
3157	Septo-optic dysplasia spectrum	2243	Hypopituitarism-micropenis-cleft lip/palate syndrome	33364	Trichothiodystrophy	453	Tay syndrome
3157	Septo-optic dysplasia spectrum	2244	Hypopituitarism-microphthalmia syndrome	33364	Trichothiodystrophy	453	Trichothiodystrophy type E
3157	Septo-optic dysplasia spectrum	2244	Kaplowitz-Bodurtha syndrome	33364	Trichothiodystrophy	453	Trichothiodystrophy with congenital ichthyosis
3157	Septo-optic dysplasia spectrum	370006	Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome	33364	Trichothiodystrophy	2739	Oncho-tricho-dysplasia-neutropenia syndrome
3157	Septo-optic dysplasia spectrum	93943	Corpus callosum dysgenesis-hypopituitarism syndrome	33364	Trichothiodystrophy	2739	Itin syndrome
3202	Dehydrated hereditary stomatocytosis	100039	Familial pseudohyperkalemia type 1	33364	Trichothiodystrophy	2739	ONMR syndrome
3253	Zlotogora-Ogur syndrome	90338	Margarita island ectodermal dysplasia	33364	Trichothiodystrophy	2739	Trichothiodystrophy type G
3255	Filippi syndrome	137658	Microcephaly-intellectual disability-phalangeal and neurological anomalies syndrome	33364	Trichothiodystrophy	3123	Brittle hair syndrome, Sabinas type
3255	Filippi syndrome	137658	Woods-Crouchman-Huson syndrome	33364	Trichothiodystrophy	3123	Brittle hair-mental deficiency syndrome
3447	Weaver syndrome	3446	Weaver-like syndrome	33364	Trichothiodystrophy	3123	Trichothiodystrophy type B
3460	Torg-Winchester syndrome	2775	Autosomal recessive carpotarsal osteolysis	33364	Trichothiodystrophy	231256	Beta-thalassemia-trichothiodystrophy syndrome
3460	Torg-Winchester syndrome	2775	Hereditary multicentric osteolysis	33364	Trichothiodystrophy	75790	Pollitt syndrome
3464	Woodhouse-Sakati syndrome	1011	Alopecia-hypogonadism-extrapyramidal syndrome	33364	Trichothiodystrophy	75790	Trichorrhexis nodosa syndrome
				33364	Trichothiodystrophy	75790	Trichothiodystrophy type C
				33364	Trichothiodystrophy	75790	Trichothiodystrophy-neurocutaneous syndrome syndrome
				33364	Trichothiodystrophy	75789	SIBIDS syndrome
				33364	Trichothiodystrophy	75789	Trichothiodystrophy-osteosclerosis syndrome
				35069	Infantile neuroaxonal dystrophy	2174	Hunter-Carpenter-McDonald syndrome
				36899	Myoclonus-dystonia syndrome	210566	Myoclonic dystonia 15
				36899	Myoclonus-dystonia syndrome	210566	DYT15
				36899	Myoclonus-dystonia syndrome	210566	Myoclonus-dystonia type 15
				42738	Severe congenital neutropenia	37629	Neonatal neutropenia
				42775	PHACE syndrome	1564	Dandy-Walker malformation-facial hemangioma syndrome

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42775	PHACE syndrome	3195	Sternal malformation-vascular dysplasia syndrome	85199	Craniosynostosis-anal anomalies-porokeratosis syndrome	2060	Fukuda-Miyanomae-Nakata syndrome
52368	Mohr-Tranebaerg syndrome	3213	Deafness-opticoacoustic nerve atrophy-dementia syndrome	85293	X-linked intellectual disability, Cabezas type	85289	X-linked intellectual disability, Vitale type
52368	Mohr-Tranebaerg syndrome	3213	Jensen syndrome	86872	T-cell large granular lymphocyte leukemia	2687	Neutropenia-hyperlymphocytosis with large granular lymphocytes syndrome
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	54238	Myotonic dystrophy type 3	90186	Meige disease	77242	Lymphedema tarda
53271	Muenke syndrome	1535	Craniosynostosis-dysmorphism-brachydactyly syndrome	90186	Meige disease	77241	Lymphedema praecox
53271	Muenke syndrome	1535	Glass-Chapman-Hockley syndrome	90186	Meige disease	90185	Non-hereditary late-onset primary lymphedema
56304	Atelosteogenesis type II	2640	Lethal short-limb dwarfism, McAlister-Crane type	90186	Meige disease	90185	Meige-like disease
56304	Atelosteogenesis type II	2640	McAlister-Crane syndrome	90340	Blau syndrome	90341	Early-onset sarcoidosis
60030	Loeys-Dietz syndrome	97295	Furlong syndrome	91387	Familial thoracic aortic aneurysm and aortic dissection	88636	Aortic dilatation-joint hypermobility-arterial tortuosity syndrome
60030	Loeys-Dietz syndrome	97295	Marfanoid habitus-craniosynostosis syndrome	93284	Spondyloepiphyseal dysplasia tarda	163673	Spondyloepiphyseal dysplasia, Byers type
69061	Idiopathic steroid-sensitive nephrotic syndrome	97552	Steroid-sensitive nephrotic syndrome without renal biopsy	93284	Spondyloepiphyseal dysplasia tarda	163673	Spondyloepiphyseal dysplasia-punctate corneal dystrophy syndrome
69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	2087	Glomerulonephritis-sparse hair-telangiectasis syndrome	93950	X-linked intellectual disability, Sutherland-Haan type	93944	X-linked intellectual disability, Fichera type
79189	Peroxisome biogenesis disorder	34	Pipecolic acidemia	95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis
79189	Peroxisome biogenesis disorder	34	Hyperpipecolatemia	95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Ambiguous genitalia-disordered steroidogenesis Antley-Bixler-like syndrome
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	79261	Glycogen storage disease type 1D	95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Antley-Bixler syndrome type 2
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	79261	Type 1D glycogenosis	95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Antley-Bixler syndrome, POR-related
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	79260	Glycogen storage disease type 1C	97229	Riboflavin transporter deficiency	56965	Progressive bulbar paralysis of childhood
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	79260	Type 1C glycogenosis	97229	Riboflavin transporter deficiency	56965	Fazio-Londe disease
79452	Milroy disease	79450	Non-hereditary congenital primary lymphedema	97229	Riboflavin transporter deficiency	56965	Progressive bulbar palsy of childhood
79452	Milroy disease	79450	Milroy-like disease	98769	Spinocerebellar ataxia type 15/16	98770	Spinocerebellar ataxia type 16
79500	DOORS syndrome	1674	Digitorenocerebral syndrome	98769	Spinocerebellar ataxia type 15/16	98770	SCA16
79500	DOORS syndrome	1674	DRC syndrome	98772	Spinocerebellar ataxia type 19/22	101107	Spinocerebellar ataxia type 22
79500	DOORS syndrome	1674	Eronen-Somer-Gustafsson syndrome	98772	Spinocerebellar ataxia type 19/22	101107	SCA22
79502	Punctate palmoplantar keratoderma type 2	736	Palmoplantar porokeratosis of Mantoux	98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal hypnogenic dyskinesia
83628	LUMBAR syndrome	2125	Sacral hemangiomas-multiple congenital abnormalities syndrome				

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ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities	ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Nocturnal paroxysmal dystonia	221061	Familial cerebral cavernous malformation	2486	Transverse limb deficiency-hemangioma syndrome
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal hypnagogic dyskinesia	231568	Generalized dominant dystrophic epidermolysis bullosa	216989	Autosomal dominant dystrophic epidermolysis bullosa, Pasini type
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal hypnagogic dystonia	231568	Generalized dominant dystrophic epidermolysis bullosa	216989	DDEB, Pasini type
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal nocturnal dyskinesia	231568	Generalized dominant dystrophic epidermolysis bullosa	79407	Autosomal dominant dystrophic epidermolysis bullosa, Cockayne-Touraine type
98808	Autosomal dominant dopa-responsive dystonia	101151	Dystonia 14	231568	Generalized dominant dystrophic epidermolysis bullosa	79407	DDEB, Cockayne-Touraine type
98808	Autosomal dominant dopa-responsive dystonia	101151	DYT14	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	3421	Cerebroretinal vasculopathy
98967	Schnyder corneal dystrophy	98968	Central discoid corneal dystrophy	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	3421	CRV
137834	Frank-Ter Haar syndrome	1266	Dermato-cardio-skeletal syndrome, Borrone type	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	3421	Grand-Kainé-Fulling syndrome
168569	H syndrome	254723	Pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	63261	HERNS syndrome
168569	H syndrome	254723	PHID	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	63261	Hereditary endotheliopathy-retinopathy-nephropathy-stroke syndrome
168569	H syndrome	254712	Familial sinus histiocytosis with massive lymphadenopathy	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	71291	Hereditary vascular retinopathy
168569	H syndrome	254712	Familial Rosaï-Dorfman disease	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	71291	HVR
168569	H syndrome	254712	Familial SHML	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	71291	Hereditary vascular retinopathy-Raynaud phenomenon-migraine syndrome
168569	H syndrome	254707	Faisalabad histiocytosis	261483	Xq27.3q28 duplication syndrome	3423	Vasquez-Hurst-Sotos syndrome
168569	H syndrome	254707	FHC	261483	Xq27.3q28 duplication syndrome	3423	Hypogonadism-gynecomastia-X-linked intellectual disability syndrome
182050	MYH9-related disease	850	May-Hegglin thrombocytopenia	263463	CHST3-related skeletal dysplasia	1792	Humerospinal dysostosis
182050	MYH9-related disease	850	MHA	263463	CHST3-related skeletal dysplasia	93280	Spondyloepiphyseal dysplasia, Omani type
182050	MYH9-related disease	850	May-Hegglin anomaly	263463	CHST3-related skeletal dysplasia	93280	Humero-spinal dysostosis
182050	MYH9-related disease	850	May-Hegglin syndrome				
182050	MYH9-related disease	1984	Fechtner syndrome				
182050	MYH9-related disease	1984	Alport syndrome with leukocyte inclusions and macrothrombocytopenia				
182050	MYH9-related disease	1019	Epstein syndrome				
182050	MYH9-related disease	1019	Alport syndrome with macrothrombocytopenia				
182050	MYH9-related disease	807	Sebastian syndrome				
182050	MYH9-related disease	807	Macrothrombocytopenia with leukocyte inclusions				
216866	Classic pantothenate kinase-associated neurodegeneration	157855	HARP syndrome				
216866	Classic pantothenate kinase-associated neurodegeneration	157855	Hypoprebetalipoproteinemia-acanthocytosis-retinitis pigmentosa-pallidal degeneration syndrome				
220295	Xeroderma pigmentosum-Cockayne syndrome complex	2837	Pellagra-like skin rash-neurological manifestations syndrome				

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264200	14q22q23 microdeletion syndrome	2055	Growth deficiency-brachydactyly-dysmorphism syndrome	319646	PGM1-CDG	711	Glycogen storage disease type XIV
264200	14q22q23 microdeletion syndrome	2055	Frias syndrome	319646	PGM1-CDG	711	Glycogenosis due to phosphoglucomutase deficiency
284963	Marfan syndrome type 1	99715	MASS syndrome	319646	PGM1-CDG	711	Glycogenosis type 14
284963	Marfan syndrome type 1	99715	Mitral valve-aorta-skeleton-skin syndrome	319646	PGM1-CDG	711	Glycogenosis type XIV
293843	3MC syndrome	2453	Malpuech syndrome	319646	PGM1-CDG	711	Phosphoglucomutase 1 deficiency
293843	3MC syndrome	2453	3MC3 syndrome	324737	SRD5A3-CDG	168972	Kahrizi syndrome
293843	3MC syndrome	2453	Malpuech facial clefting syndrome	324737	SRD5A3-CDG	168972	Intellectual disability, Kahrizi type
293843	3MC syndrome	2506	Michels syndrome	324737	SRD5A3-CDG	168972	Intellectual disability-cataract-coloboma-kyphosis syndrome
293843	3MC syndrome	2506	3MC1 syndrome	324737	SRD5A3-CDG	139477	Al-Gazali-Dattani syndrome
293843	3MC syndrome	2506	Oculopalatoskeletal syndrome	329931	C3 glomerulonephritis	93559	C3 deposition glomerulonephritis without proliferation
293843	3MC syndrome	2998	Carnevale syndrome	331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	178503	Dursun syndrome
293843	3MC syndrome	2998	3MC2 syndrome	331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	178503	Pulmonary arterial hypertension-leukopenia-atrial septal defect syndrome
293843	3MC syndrome	2998	Carnevale-Krajewska-Fischetto syndrome	357225	Primary non-essential cutis verticis gyrata	1557	Cutis verticis gyrata-intellectual disability syndrome
293843	3MC syndrome	2998	Mingarelli syndrome	357225	Primary non-essential cutis verticis gyrata	1557	McDowall syndrome
293843	3MC syndrome	2998	OSA syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata-retinitis pigmentosa-sensorineural deafness syndrome
293843	3MC syndrome	2998	Oculo-skeletal-abdominal syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata-retinitis pigmentosa-neurosensory deafness syndrome
293843	3MC syndrome	2998	Ptosis-strabismus-rectus abdominis diastasis syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata-retinitis pigmentosa-neurosensory hearing loss syndrome
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	137862	Martínez-Frías syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata-retinitis pigmentosa-sensorineural deafness syndrome
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	137862	Duodenal and extrahepatic biliary atresia-hypoplastic pancreas-intestinal malrotation syndrome	370114	Combined cervical dystonia	293838	Fatal infantile encephalopathy-pulmonary hypertension syndrome
300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	83618	Severe dilated cardiomyopathy due to lamin A/C mutation	370953	Congenital muscular dystrophy due to dystroglycanopathy	52428	Congenital muscular dystrophy type 1C
300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	83618	Severe dilated cardiomyopathy with or without myopathy	370953	Congenital muscular dystrophy due to dystroglycanopathy	52428	CMD1C
313795	Jawad syndrome	137653	Microcephaly-digital anomalies-intellectual disability syndrome	370953	Congenital muscular dystrophy due to dystroglycanopathy	52428	MDC1C
313795	Jawad syndrome	137653	Kelly-Kirson-Wyatt syndrome				
314632	Parkinsonism due to ATP13A2 deficiency	3336	Tomé-Brunet-Farreau syndrome				
319646	PGM1-CDG	711	Glycogen storage disease due to phosphoglucomutase deficiency				
319646	PGM1-CDG	711	GSD due to phosphoglucomutase deficiency				
319646	PGM1-CDG	711	GSD type 14				
319646	PGM1-CDG	711	GSDXIV				
319646	PGM1-CDG	711	Glycogen storage disease type 14				

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370953	Congenital muscular dystrophy due to dystroglycanopathy	98894	Congenital muscular dystrophy type 1D	423693	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect	99047	Double outlet right ventricle with doubly committed ventricular septal defect
370953	Congenital muscular dystrophy due to dystroglycanopathy	98894	MDC1D	444490	Familial chylomicronemia syndrome	411	Hyperlipoproteinemia type 1
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Male infertility with normal virilization due to meiosis defect	444490	Familial chylomicronemia syndrome	411	HLP type 1
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Azoospermia due to maturation arrest	448242	Autosomal recessive brachyolmia	93301	Brachyolmia type 1, Hobaek type
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Azoospermia due to meiosis defect	448242	Autosomal recessive brachyolmia	93303	Brachyolmia type 1, Toledo type
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Male infertility with normal virilization due to maturation arrest	457059	Pseudohypoparathyroidism with Albright hereditary osteodystrophy	665	Albright hereditary osteodystrophy
399808	Male infertility with teratozoospermia due to single gene mutation	352613	Male infertility due to NANOS1 mutation	457240	X-linked intellectual disability-short stature-overweight syndrome	3059	X-linked intellectual disability, Gu type
402041	Autosomal recessive distal renal tubular acidosis	93609	Autosomal recessive distal renal tubular acidosis without deafness	457240	X-linked intellectual disability-short stature-overweight syndrome	3059	MRX35
402041	Autosomal recessive distal renal tubular acidosis	93609	AR dRTA without deafness				
402041	Autosomal recessive distal renal tubular acidosis	93609	AR dRTA without hearing loss				
402041	Autosomal recessive distal renal tubular acidosis	93609	Autosomal recessive distal renal tubular acidosis without hearing loss				
402041	Autosomal recessive distal renal tubular acidosis	93609	Distal renal tubular acidosis type 1c				
402041	Autosomal recessive distal renal tubular acidosis	93609	dRTA type 1c				
402041	Autosomal recessive distal renal tubular acidosis	93611	Autosomal recessive distal renal tubular acidosis with deafness				
402041	Autosomal recessive distal renal tubular acidosis	93611	AR dRTA with deafness				
402041	Autosomal recessive distal renal tubular acidosis	93611	AR dRTA with hearing loss				
402041	Autosomal recessive distal renal tubular acidosis	93611	Autosomal recessive distal RTA with deafness				
402041	Autosomal recessive distal renal tubular acidosis	93611	Autosomal recessive distal renal tubular acidosis with hearing loss				
402041	Autosomal recessive distal renal tubular acidosis	93611	Distal renal tubular acidosis type 1b				
402041	Autosomal recessive distal renal tubular acidosis	93611	dRTA type 1b				
423693	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect	99044	Double outlet right ventricle with subaortic ventricular septal defect				

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