



March 2016

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.

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
2671	3-Phosphoglycerate dehydrogenase deficiency, neonatal form
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
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	ORPHA number	Disease name
352328	3-methylglutaconic aciduria with deafness - encephalopathy - Leigh-like syndrome	881	45,X/46,XX syndrome
445038	3-methylglutaconic aciduria-cataract-neurologic involvement-neutropenia syndrome	1772	45,X/46,XY MGD
67046	3-methylglutaconyl-CoA hydratase deficiency	1772	45,X/46,XY mixed gonadal dysgenesis
134	3-oxothiolase deficiency	1772	45,X0/46,XY MGD
79351	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form	243	46,XX complete gonadal dysgenesis
79350	3-phosphoserine phosphatase deficiency	2973	46,XX disorder of sex development - anorectal anomalies
869	3A syndrome	2975	46,XX disorder of sex development - skeletal anomalies
7	3C syndrome	243	46,XX gonadal dysgenesis
2616	3M syndrome	243	46,XX ovarian dysgenesis
293843	3MC syndrome	444048	46,XX ovarian dysgenesis-short stature syndrome
→29384	3MC1 syndrome	2138	46,XX ovotesticular DSD
3	3MC2 syndrome	2138	46,XX ovotesticular disorder of sex development
→29384	3MC3 syndrome	243	46,XX pure gonadal dysgenesis
67046	3MG-CoA hydratase deficiency	393	46,XX testicular DSD
2118	4-HPPD deficiency	393	46,XX testicular disorder of sex development
2118	4-alpha-hydroxyphenylpyruvate hydroxylase deficiency	199310	46,XX/46,XY chimerism
22	4-hydroxybutyric aciduria	242	46,XY CGD
2118	4-hydroxyphenylpyruvic acid dioxygenase deficiency	753	46,XY DSD due to 5-alpha-reductase 2 deficiency
869	4A syndrome	755	46,XY DSD due to LH resistance or LHB deficiency
88637	4H syndrome	325448	46,XY DSD due to LHB deficiency
250977	5-amino-4-imidazole carboxamide ribosiduria	96265	46,XY DSD due to complete LH receptor inactivation
217064	5-fluorouracil intoxication	96265	46,XY DSD due to complete LH resistance
217064	5-fluorouracil poisoning	96265	46,XY DSD due to complete luteinizing hormone receptor inactivation
240839	5-fluorouracil toxicity	96265	46,XY DSD due to complete luteinizing hormone resistance
33572	5-oxoprolinase deficiency	755	46,XY DSD due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency
99135	6-phosphogluconate dehydrogenase deficiency	325448	46,XY DSD due to luteinizing hormone subunit beta deficiency
13	6-pyruvoyl-tetrahydropterin synthase deficiency	96266	46,XY DSD due to partial LH receptor inactivation
818	7-dehydrocholesterol reductase deficiency	96266	46,XY DSD due to partial LH resistance
168588	11-beta-hydroxysteroid dehydrogenase deficiency type 1	96266	46,XY DSD due to partial luteinizing hormone resistance
320	11-beta-hydroxysteroid dehydrogenase deficiency type 2	251510	46,XY PGD
752	17-beta-hydroxysteroid dehydrogenase 3 deficiency	242	46,XY complete gonadal dysgenesis
752	17-ketoreductase deficiency	96266	46,XY disorder of sex development due to partial LH receptor inactivation
99763	18-hydroxylase deficiency	96266	46,XY disorder of sex development due to partial LH resistance
99763	18-oxidase deficiency		
881	45,X 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
96266	46,XY disorder of sex development due to partial luteinizing hormone resistance	99330	49,XYYYY syndrome	96121	7q11.23 microduplication syndrome
		293948	1p21.3 microdeletion syndrome	251061	7q31 microdeletion syndrome
		401986	1p31p32 microdeletion syndrome	96092	8p inverted duplication/deletion syndrome
		1606	1p36 deletion syndrome	168953	8p11 myeloproliferative syndrome
168558	46,XY disorder of sex development - adrenal insufficiency due to CYP11A1 deficiency	250989	1q21.1 microdeletion syndrome	251066	8p11.2 deletion syndrome
		250994	1q21.1 microduplication syndrome	251071	8p23.1 microdeletion syndrome
		250999	1q41-q42 microdeletion syndrome	251076	8p23.1 microduplication syndrome
		250999	1q41q42 microdeletion syndrome	228399	8q12 microduplication syndrome
752	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	238769	1q44 microdeletion syndrome	2496	8q13 microdeletion syndrome
753	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency	363680	2p13.2 microdeletion syndrome	284160	8q21.11 microdeletion syndrome
755	46,XY disorder of sex development due to LH resistance or LHB deficiency	261349	2p15-p16.1 microdeletion syndrome	178303	8q22.1 microdeletion syndrome
		261349	2p15p16.1 microdeletion syndrome	261112	9p deletion syndrome
325448	46,XY disorder of sex development due to LHB deficiency	163693	2p21 deletion syndrome	261112	9p- syndrome
		163693	2p21 microdeletion syndrome	324313	9p13 microdeletion syndrome
96265	46,XY disorder of sex development due to complete LH receptor inactivation	369881	2p21 microdeletion syndrome without cystinuria	96147	9q subtelomeric deletion syndrome
		228402	2q23.1 microdeletion syndrome	96147	9qSTDs
		313947	2q23.1 microduplication syndrome	352665	9q21 microdeletion syndrome
		1617	2q24 microdeletion syndrome	401923	9q31.1q31.3 microdeletion syndrome
96265	46,XY disorder of sex development due to complete luteinizing hormone receptor inactivation	251014	2q31.1 microdeletion syndrome	284169	10p11.21p12.31 microdeletion syndrome
		294026	2q31.1 microduplication syndrome	284169	10p12p11 microdeletion syndrome
96265	46,XY disorder of sex development due to complete luteinizing hormone resistance	251019	2q32-q33 microdeletion syndrome	276413	10q22.3q23.3 microdeletion syndrome
		251019	2q32q33 microdeletion syndrome	276422	10q22.3q23.3 microduplication syndrome
		251028	2q33.1 microdeletion syndrome	1307	10q24 microduplication syndrome
90796	46,XY disorder of sex development due to isolated 17,20-lyase deficiency	1001	2q37 microdeletion syndrome	52022	11p11.2 deletion
		1620	3p- syndrome	300305	11p15.4 microduplication syndrome
		435638	3p25.3 microdeletion syndrome	444002	11q22.2-q22.3 deletion syndrome
755	46,XY disorder of sex development due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency	65286	3q subtelomere deletion syndrome	444002	11q22.2q22.3 microdeletion syndrome
		65286	3qter deletion	313884	12p12.1 microdeletion syndrome
		1621	3q13 microdeletion syndrome	280325	12p13.33 microdeletion syndrome
325448	46,XY disorder of sex development due to luteinizing hormone subunit beta deficiency	96095	3q26 microduplication syndrome	94063	12q14 microdeletion syndrome
		356947	3q26-q27microdeletion syndrome	289513	12q15q21.1 microdeletion syndrome
		356947	3q26q27 microdeletion syndrome	412035	13q12.3 microdeletion syndrome
		397695	3q27.3 microdeletion syndrome	1590	13q32 deletion
443087	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	65286	3q29 microdeletion syndrome	261120	14q11.2 microdeletion syndrome
		251038	3q29 microduplication	261229	14q11.2 microduplication syndrome
168563	46,XY gonadal dysgenesis - motor and sensory neuropathy	280	4p- syndrome	261144	14q12 microdeletion syndrome
325345	46,XY ovotesticular DSD	96072	4p16.3 microduplication syndrome	→3157	14q22 microdeletion syndrome
325345	46,XY ovotesticular disorder of sex development	238750	4q21 microdeletion syndrome	264200	14q22-q23 microdeletion syndrome
251510	46,XY partial gonadal dysgenesis	329802	5p13 microduplication syndrome	264200	14q22q23 microdeletion syndrome
251510	46,XY partial testicular dysgenesis	86841	5q- syndrome	401935	14q24.1q24.3 microdeletion syndrome
242	46,XY pure gonadal dysgenesis	228384	5q14.3 microdeletion syndrome	314585	15q overgrowth syndrome
3375	47,XXX syndrome	436003	5q23 microdeletion syndrome	238446	15q11-q13 duplication syndrome
8	47,XYY syndrome	314655	5q31.3 microdeletion syndrome	238446	15q11-q13 microduplication syndrome
9	48,XXXX syndrome	228415	5q35 microduplication syndrome	261183	15q11.2 microdeletion syndrome
96263	48,XXXY syndrome	96125	6p subtelomeric deletion syndrome	238446	15q11q13 duplication syndrome
10	48,XXYY syndrome	251046	6p22 microdeletion syndrome	238446	15q11q13 microduplication syndrome
99329	48,XYYY syndrome	96125	6p25 microdeletion syndrome	261183	15q11.2 microdeletion syndrome
11	49,XXXXX syndrome	75857	6q terminal deletion syndrome	238446	15q11q13 duplication syndrome
96264	49,XXXXY syndrome	171829	6q16 deletion syndrome	238446	15q11q13 microduplication syndrome
261534	49,XXXXY syndrome	251056	6q25 microdeletion syndrome	238446	15q11q13 microduplication syndrome
		314034	7p22.1 microduplication 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
199318	15q13.3 microdeletion syndrome	567	22q11.2 deletion syndrome	99112	Absence of brachiocephalic vein
261190	15q14 microdeletion syndrome	1727	22q11.2 microduplication syndrome	1658	Absence of dermatoglyphics - congenital milia
94065	15q24 microdeletion syndrome	48652	22q13 deletion	1658	Absence of fingerprints - congenital milia
1596	15q26 deletion syndrome	85445	AA amyloidosis	99112	Absence of innominate vein
363992	15q26.3 microdeletion syndrome	869	AAA syndrome	101206	Absence of pulmonary valve - Fallot's tetralogy - absence of ductus arteriosus
261211	16p11.2-p12.2 microdeletion syndrome	35708	AADC deficiency		Absence of pulmonary valve - ventricular septal defect - persistent ductus arteriosus
261211	16p11.2p12.2 microdeletion syndrome	91385	AAE	99048	Absence of the pulmonary artery
		100055	AAE 2	99048	Absence of the superior caval vein
		100055	AAE II	99048	Absence of the superior vena cava
261204	16p11.2p12.2 microduplication syndrome	1414	Aagenaes syndrome	99114	Absence of the SVC
261236	16p13.11 microdeletion syndrome	284460	AAOR	85201	Absent patellae - scrotal hypoplasia - renal anomalies - facial dysmorphism - intellectual disability
261243	16p13.11 microduplication syndrome	93560	AApoAI amyloidosis	3016	Absent radius-anogenital anomalies syndrome
96078	16p13.3 microduplication syndrome	238269	AApoII amyloidosis	2951	Absent thumb-short stature-immunodeficiency syndrome
352629	16q24.1 microdeletion syndrome	439232	AApoIV amyloidosis	988	Absent tibia - polydactyly
261250	16q24.3 microdeletion syndrome	295105	Aapodia, unilateral	3328	Absent tibia - polydactyly - arachnoid cyst
819	17p11.2 microdeletion syndrome	915	Aarskog syndrome	99901	ACAD9 deficiency
1713	17p11.2 microduplication syndrome	1974	Aarskog-like syndrome	42	ACADM deficiency
217385	17p13.3 duplication syndrome	3163	Aarskog-Ose-Pande syndrome	26792	ACADS deficiency
217385	17p13.3 microduplication syndrome	915	Aarskog-Scott syndrome	945	Acalvaria
97685	17q11 microdeletion syndrome	124	Aase syndrome	67043	Acanthamoeba keratitis
139474	17q11.2 microduplication syndrome	916	Aase-Smith I syndrome	79468	Acanthokeratolytic verrucous nevus
261265	17q12 microdeletion syndrome	124	Aase-Smith II syndrome	300504	Acanthoma of the nail matrix
261272	17q12 microduplication syndrome	916	Aase-Smith syndrome	90301	Acanthosis nigricans - insulin resistance - muscle cramps - acral enlargement
363958	17q21.31 microdeletion syndrome	240841	Abacavir toxicity	926	Acatasemia
217340	17q21.31 microduplication syndrome	69663	ABCB4 gene mutation-associated cholelithiasis	561	Accelerated skeletal maturation - peculiar facies - failure to thrive
261279	17q23.1-q23.2 microdeletion syndrome	→897	ABCD syndrome	180182	Accessory breasts
261279	17q23.1q23.2 microdeletion syndrome	2970	Abdominal muscle deficiency syndrome	99061	Accessory mitral valve tissue
		800	Aberfeld syndrome	141096	Accessory nostril
1598	18p- syndrome	85446	ABeta2Mwt amyloidosis	674	Accessory pancreas
1600	18q- syndrome	324723	ABeta amyloidosis, Arctic type	95462	Accessory tricuspid valve tissue
254346	19p13.12 microdeletion syndrome	100006	ABeta amyloidosis, Dutch type	210122	ACDMPV
357001	19p13.13 microdeletion syndrome	324708	ABeta amyloidosis, Iowa type	48818	Aceruloplasminemia
447980	19p13.3 microduplication syndrome	324713	ABeta amyloidosis, Italian type	99736	Acetazolamide-responsive congenital myotonia
217346	19q13.11 microdeletion syndrome	324718	ABetaA21G amyloidosis	99736	Acetazolamide-responsive myotonia
313781	20p subtelomeric deletion syndrome	324718	ABetaA21G-related amyloidosis	2008	ACFS
261295	20p12.3 microdeletion syndrome	324708	ABetaD23N amyloidosis	929	Achalasia - microcephaly
313781	20p13 microdeletion syndrome	324723	ABetaE22G amyloidosis	930	Achalasia cardia
444051	20q11.2 microdeletion syndrome	324713	ABetaE22K amyloidosis	869	Achalasia-addisonianism-alacrima syndrome
363659	20q11.2 microduplication syndrome	100006	ABetaE22Q amyloidosis		
261311	20q13.33 microdeletion syndrome	324703	ABetaL34V amyloidosis		
574	21q deletion syndrome	324703	ABetaL34V-related amyloidosis		
574	21q- syndrome	14	Abetalipoproteinemia		
261323	21q22.11-q22.12 microdeletion syndrome	920	Ablepharon macrostomia syndrome		
261323	21q22.11q22.12 microdeletion syndrome	99089	Abnormal number of coronary ostia		
		99050	Abnormal origin of right or left pulmonary artery from the aorta		
268261	21q22.13-q22.2 microdeletion syndrome	1164	ABPA		
268261	21q22.13q22.2 microdeletion syndrome	97345	ABri amyloidosis		
		921	Abruzzo-Erickson syndrome		
567	22q11DS	69739	ABSD		
		2310	Absence deformity of leg - cataract		

→ 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
→869	Achalasia-alacrimia syndrome	91365	Acquired ciliary dyskinesia	1784	Acro-fronto-facio-nasal dysostosis
294983	Acheiria	228285	Acquired cutis laxa	2211	Acro-fronto-facio-nasal dysostosis type 2
295103	Acheiria, bilateral	46487	Acquired epidermolysis bullosa	2211	Acro-fronto-facio-nasal syndrome type 2
295101	Acheiria, unilateral	98818	Acquired epileptic aphasia	2980	Acro-oto-ocular syndrome
931	Acheiropodia	79086	Acquired generalized lipodystrophy	85203	Acro-pectoral syndrome
931	Acheiropody	228247	Acquired Gronblad-Strandberg-Touraine syndrome	956	Acro-pectoror-renal dysplasia
49382	ACHM	231401	Acquired HbH disease	958	Acro-renal-mandibular syndrome
932	Achondrogenesis	231401	Acquired hemoglobin H disease	959	Acro-renal-ocular syndrome
93299	Achondrogenesis type 1A	158057	Acquired hemophagocytic lymphohistiocytosis associated with a malignant disease	36	Acrocallosal syndrome
93298	Achondrogenesis type 1B	73274	Acquired hemophilia	63446	Acrocapitofemoral dysplasia
93296	Achondrogenesis type 2	2221	Acquired hypertrichosis lanuginosa	221054	Acrocephalopolydactylous dysplasia
93299	Achondrogenesis, Houston-Harris type	26348	Acquired hypoprothrombinemia	221054	Acrocephalopolydactyly
93296	Achondrogenesis, Langer-Saldino type	454	Acquired ichthyosis	65759	Acrocephalopolysyndactyly type 2
93298	Achondrogenesis, Parenti-Fraccaro type	75564	Acquired idiopathic sideroblastic anemia	3128	Acrocephalopolysyndactyly type 3
15	Achondroplasia	404514	Acquired kidney disease-associated renal cell carcinoma	65798	Acrocephalopolysyndactyly type 4
935	Achondroplasia-SCID syndrome	37559	Acquired kinky hair syndrome	87	Acrocephalosyndactyly type 1
935	Achondroplasia-severe combined immunodeficiency syndrome	79086	Acquired lipoatrophic diabetes	794	Acrocephalosyndactyly type 3
935	Achondroplasia-Swiss type agammaglobulinemia syndrome	589	Acquired myasthenia	710	Acrocephalosyndactyly type 5
49382	Achromatopsia	95626	Acquired neurogenic diabetes insipidus	63440	Acrocephaly
355	Acid beta-glucuronidase deficiency	84142	Acquired neuromyotonia	949	Acrocraniofacial dysostosis
333	Acid ceramidase deficiency	91385	Acquired non histamine-induced angioedema	955	Acrodentosteodysplasia
35121	Acid phosphatase deficiency	314697	Acquired porencephaly	163931	Acrodermatitis continua suppurativa of Hallopeau
424046	Acinar cell carcinoma of pancreas	729	Acquired primary erythrocytosis	37	Acrodermatitis enteropathica
40366	Acitretin/etretinate embryopathy	26348	Acquired prothrombin deficiency	950	Acrodysostosis
79099	Ackerman dermatitis syndrome	228247	Acquired pseudoxanthoma elasticum	280651	Acrodysostosis with multiple hormone resistance
2561	Ackerman syndrome	49566	Acquired purpura fulminans	950	Acrodysplasia
43115	Aconitase deficiency	228247	Acquired PXE	2956	Acrodysplasia scoliosis
252175	Acoustic neurilemoma	206575	Acquired rippling muscle disease	1786	Acrofacial dysostosis, Catania type
252175	Acoustic neurinoma	93585	Acquired thrombotic thrombocytopenic purpura	246	Acrofacial dysostosis, Genee-Wiedmann type
252175	Acoustic neuroma	93585	Acquired TTP	64542	Acrofacial dysostosis, Kennedy-Teebi type
65759	ACPS2	99147	Acquired von Willebrand disease	1787	Acrofacial dysostosis, Palagonia type
65798	ACPS4	99147	Acquired von Willebrand syndrome	1788	Acrofacial dysostosis, Rodríguez type
3128	ACPS III	263534	Acral deciduous skin	952	Acrofacial dysostosis, Weyers type
3128	ACPS with leg hypoplasia	97360	Acral dysostosis with facial and genital abnormalities	2500	Acrogeria
306431	Acquired adult-onset immunodeficiency	158673	Acral dystrophic epidermolysis bullosa	2500	Acrogeria, Gottron type
90065	Acquired aneurysmal subarachnoid hemorrhage	263534	Acral peeling skin syndrome	38	Acrokeratoelastoidosis of Costa
91385	Acquired angioedema	90396	Acral persistent papular mucinosis	166113	Acrokeratosis of Bazex
100056	Acquired angioedema type 1	263534	Acral PSS	166113	Acrokeratosis paraneoplastica
100055	Acquired angioedema type 2	281127	Acral self-healing collodion baby	79151	Acrokeratosis verruciformis of Hopf
91385	Acquired angioneurotic edema	281127	Acral SHCB	965	Acromegaloid facial appearance syndrome
100056	Acquired angioneurotic edema type 1	945	Acrania	963	Acromegaly
100055	Acquired angioneurotic edema type 2	2008	Acro-cardio-facial syndrome	→2796	Acromegaly-cutis verticis gyrata-corneal leukoma syndrome
91385	Acquired bradykinine-induced angioedema	978	Acro-dermato-ungual-lacrimal-tooth syndrome	39	Acromelanosis
91385	Acquired C1 inhibitor deficiency			1827	Acromelic frontonasal dysplasia
95626	Acquired CDI			968	Acromesomelic dwarfism
95626	Acquired central diabetes insipidus				

→ 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
2098	Acromesomelic dysplasia, Grebe type	243367	Acute fatty liver of pregnancy	86843	Acute myelofibrosis
968	Acromesomelic dysplasia, Hunter-Thomson type	3243	Acute febrile neutrophilic dermatosis	102379	Acute myeloid leukemia and myelodysplastic syndromes related to alkylating agent
40	Acromesomelic dysplasia, Maroteaux type	293173	Acute generalized exanthematous pustulosis	164726	Acute myeloid leukemia and myelodysplastic syndromes related to radiation
2500	Acrometageria	99920	Acute graft versus host disease	102381	Acute myeloid leukemia and myelodysplastic syndromes related to topoisomerase type 2 inhibitor
969	Acromicric dysplasia	90062	Acute hepatic failure	318	Acute myeloid leukemia M6
955	Acroosteolysis dominant type	98916	Acute idiopathic demyelinating polyneuropathy	518	Acute myeloid leukemia M7
955	Acroosteolysis with osteoporosis and changes in skull and mandible	363549	Acute infantile encephalopathy predominantly affecting the frontal lobes	98831	Acute myeloid leukemia with 11q23 abnormalities
363665	Acroosteolysis-keloid-like lesions-premature aging syndrome	217371	Acute infantile liver failure due to synthesis defect of mitochondrial DNA-encoded proteins	98829	Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)
957	Acropectorovertebral dysplasia	217371	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	319480	Acute myeloid leukemia with CEBPA somatic mutations
41	Acropigmentation of Dohi	370088	Acute infantile liver failure-multisystemic involvement syndrome	402020	Acute myeloid leukemia with inv3(p21;q26.2) or t(3;3)(p21;q26.2)
1133	Acrorenal defect - ectodermal dysplasia - diabetes	98916	Acute inflammatory demyelinating polyradiculoneuropathy	86845	Acute myeloid leukemia with multilineage dysplasia
971	Acrorenal syndrome	98916	Acute inflammatory polyneuropathy	402026	Acute myeloid leukemia with NPM1 somatic mutations
85203	ACRP syndrome	79276	Acute intermittent porphyria	402014	Acute myeloid leukemia with t(6;9)(p23;q34)
36	ACS	79126	Acute interstitial pneumonia	370026	Acute myeloid leukemia with t(8;16)(p11;p13) translocation
87	ACS1	79126	Acute interstitial pneumonitis	102724	Acute myeloid leukemia with t(8;21)(q22;q22) translocation
794	ACS3	73423	Acute intoxication by Blighia sapida	402017	Acute myeloid leukemia with t(9;11)(p22;q23)
710	ACS5	90062	Acute liver failure	520	Acute myeloid leukemia with t(15;17)(q22;q12);(PML/RARalpha) and variants
98904	Actin myopathy	178320	Acute lung injury	517	Acute myelomonocytic leukemia
254395	Actinic lichen planus	518	Acute megakaryoblastic leukemia	86843	Acute myelosclerosis
254395	Actinic LP	99887	Acute megakaryoblastic leukemia in Down syndrome	263524	Acute necrotizing encephalopathy of childhood
330061	Actinic prurigo	329469	Acute megakaryoblastic leukemia without Down syndrome	247546	Acute neonatal citrullinemia type 1
330064	Actinic reticuloid	518	Acute megakaryocytic leukemia	247546	Acute neonatal citrullinemia type I
163696	Action myoclonus-renal failure syndrome	514	Acute monoblastic leukemia	77260	Acute neuronopathic Gaucher disease
397596	Activated PIK3-delta syndrome	514	Acute monocytic leukemia	163703	Acute non-herpetic encephalitis with severe refractory status epilepticus
101089	Activation-induced cytidine deaminase deficiency	98918	Acute motor axonal neuropathy	35889	Acute opioid poisoning
73423	Acute ackee fruit intoxication	98917	Acute motor-sensory axonal GBS	231457	Acute panautonomic GBS
95409	Acute adrenal failure	98917	Acute motor-sensory axonal Guillain-Barré syndrome	231457	Acute panautonomic Guillain-Barré syndrome
95409	Acute adrenal insufficiency	98917	Acute motor-sensory axonal neuropathy	231457	Acute panautonomic neuropathy
95409	Acute adrenocortical insufficiency	228157	Acute multiple sclerosis, Marburg type	231457	Acute pandysautonomia
73423	Acute akee fruit intoxication	228157	Acute multiple sclerosis, Marburg variant	86843	Acute panmyelosis with myelofibrosis
99870	Acute and disseminated Langerhans cell histiocytosis	98833	Acute myeloblastic leukemia M1	90064	Acute peripheral arterial occlusion
284460	Acute annular outer retinopathy	98834	Acute myeloblastic leukemia M2	43119	Acute poisoning by drugs with membrane-stabilizing effect
86849	Acute basophilic leukemia	520	Acute myeloblastic leukemia type 3		
69736	Acute bilateral depigmentation of the iris	98834	Acute myeloblastic leukemia with maturation		
98837	Acute biphenotypic leukemia	98833	Acute myeloblastic leukemia without maturation		
2901	Acute brachial plexus neuritis	86843	Acute myelodysplasia with myelofibrosis		
83597	Acute disseminated encephalitis				
83597	Acute disseminated encephalomyelitis				
163703	Acute encephalitis with refractory repetitive partial seizures				
363549	Acute encephalopathy with biphasic seizures and late reduced diffusion				
279888	Acute endophthalmitis				
318	Acute erythroid leukemia				

→ 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
520	Acute promyelocytic leukemia	2952	Adducted thumbs-arthrogryposis syndrome, Christian type	404448	ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder
98918	Acute pure motor GBS	2953	Adducted thumbs-arthrogryposis syndrome, Dundar type	1544	Adolescent benign focal crisis
98918	Acute pure motor Guillain-Barré syndrome	101046	ADEAF	306588	ADOS
231450	Acute pure sensory GBS	83597	ADEM	36355	ADP platelet receptor P2Y12 defect
231450	Acute pure sensory Guillain-Barré syndrome	976	Adenine phosphoribosyltransferase deficiency	2924	ADPCLD
231450	Acute pure sensory neuropathy	424016	Adenocarcinoma of anal canal	101046	ADPEAF
3099	Acute rheumatic fever	99976	Adenocarcinoma of esophagus	254892	adPEO
90059	Acute sensorineural hearing loss by acute acoustic trauma or sudden deafness or surgery induced acoustic trauma	424991	Adenocarcinoma of gallbladder and EBT	95409	Adrenal crisis
231466	Acute sensory ataxic GBS	424991	Adenocarcinoma of gallbladder and extrahepatic biliary tract	463	Adrenal incidentaloma
231466	Acute sensory ataxic Guillain-Barré syndrome	424943	Adenocarcinoma of liver and IBT	869	Adrenal insufficiency-achalasia-alacrima syndrome
231466	Acute sensory ataxic neuropathy	424943	Adenocarcinoma of liver and intrahepatic biliary tract	1501	Adrenocortical carcinoma
139417	Acute transverse myelitis	213504	Adenocarcinoma of ovary	231625	Adrenocortical carcinoma with pure aldosterone hypersecretion
43117	Acute tricyclic antidepressant poisoning	363478	Adenocarcinoma of paratestis	95409	Adrenocortical crisis
91500	Acute tubulointerstitial nephritis and uveitis syndrome	398053	Adenocarcinoma of penis	99889	Adrenocorticotrophic hormone secretion syndrome
98835	Acute undifferentiated leukemia	104075	Adenocarcinoma of small bowel	139399	Adrenomyeloneuropathy
284454	Acute zonal occult outer retinopathy	104075	Adenocarcinoma of small intestine	977	Adrenomyodystrophy
137754	ACY1D	213772	Adenocarcinoma of the cervix uteri	228169	ADSD
141	ACY2 deficiency	95512	Adenohypophysitis	46	ADSL deficiency
99901	Acyl-CoA dehydrogenase 9 deficiency	213828	Adenoid basal carcinoma of the cervix uteri	70578	Adult acute respiratory distress syndrome
100008	ACys amyloidosis	213823	Adenoid cystic carcinoma of the cervix uteri	70578	Adult ARDS
99736	ACZ-responsive congenital myotonia	213741	Adenoid cystic carcinoma of the corpus uteri	93605	Adult Bartter syndrome
99736	ACZ-responsive myotonia	93292	Adenoma of pancreas	157846	Adult basal ganglia disease
93608	AD dRTA	26790	Adenomucinosis	874	Adult cardiac tumor
428	AD hypocalcemia	213792	Adenosarcoma of the cervix uteri	2666	Adult familial nephronophthisis - spastic quadriplegia
314889	AD pRTA	213600	Adenosarcoma of the corpus uteri	309169	Adult GM2 gangliosidosis 0 variant
169189	AD-CNM	45	Adenosine monophosphate deaminase deficiency	210159	Adult HCC
1810	AD-HED	28	Adenosylcobalamin deficiency	874	Adult heart tumor
2314	AD-HIES	91127	Adenovirus infection in immunocompromised patients	210159	Adult hepatocellular carcinoma
277	ADA deficiency	46	Adenylosuccinase deficiency	247676	Adult hypophosphatasia
435623	Adactyly of foot	46	Adenylosuccinate lyase deficiency	2688	Adult idiopathic neutropenia
295118	Adactyly of foot, bilateral	137817	Adhesive arachnoiditis	178487	Adult intestinal botulism
295116	Adactyly of foot, unilateral	89937	ADHR	178487	Adult intestinal colonization botulism
295114	Adactyly of hand, bilateral	36397	Adiposalgia	178487	Adult intestinal toxin-mediated botulism
973	Adactyly of hand, unilateral	36397	Adipose tissue rheumatism	206448	Adult Krabbe disease
216796	Adair-Dighton syndrome	36397	Adiposis dolorosa	79262	Adult NCL
55881	Adamantinoma	289290	ADK hypermethioninemia	79262	Adult neuronal ceroid lipofuscinosis
974	Adams-Oliver syndrome	99027	ADLD	247676	Adult phosphoethanolaminuria
97346	ADan amyloidosis	101046	ADLTE	206583	Adult polyglucosan body disease
88619	ADANE	178464	ADMERF	902	Adult progeria
314404	ADCA-DN	98784	ADNFLE	99874	Adult pulmonary Langerhans cell histiocytosis
90348	ADCL	329211	ADNIV	98872	Adult pure red cell aplasia
86814	ADCME			247676	Adult Rathburn disease
85138	Addison disease			773	Adult Refsum disease
95409	Addisonian crisis				
2953	Adducted thumb-clubfoot 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
978	ADULT syndrome	247585	Adult-onset type 2 citrullinemia	59	AHDS
86875	Adult T-cell leukemia/lymphoma	247585	Adult-onset type II citrullinemia	50812	Ahn-Lerman-Sagie syndrome
391490	Adult-onset acquired myasthenia	99000	Adult-onset vitelliform macular dystrophy	79443	AHO - PHP Ia
79280	Adult-onset Alpha-N-acetylgalactosaminidase deficiency	3086	ADVIRC	79445	AHO - PPHP
391490	Adult-onset autoimmune myasthenia gravis	682	Adynamia episodica hereditaria	2134	aHUS
99027	Adult-onset autosomal dominant demyelinating leukodystrophy	37	AE	93581	aHUS with anti-factor H antibodies
99027	Adult-onset autosomal dominant leukodystrophy	1071	AEC syndrome	93578	aHUS with B factor anomaly
284289	Adult-onset autosomal recessive cerebellar ataxia	281139	AEI	93575	aHUS with C3 anomaly
255132	Adult-onset autosomal recessive sideroblastic anemia	163703	AERRPS	357008	aHUS with DGKE deficiency
420492	Adult-onset cervical dystonia, DYT23 type	363549	AESD	93579	aHUS with H factor anomaly
329336	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy	178345	AEXS	93580	aHUS with I factor anomaly
247585	Adult-onset citrin deficiency	37	AEZ	93576	aHUS with MCP/CD46 anomaly
247573	Adult-onset citrullinemia type 1	220460	AFAP	217023	aHUS with thrombomodulin anomaly
247573	Adult-onset citrullinemia type I	313772	AFG3L2-related spastic ataxia-neuropathy syndrome	250977	AICA-ribosiduria
329336	Adult-onset CPEO with mitochondrial myopathy	93562	AFib amyloidosis	50	Aicardi syndrome
411641	Adult-onset cystinosis	243367	AFLP	51	Aicardi-Goutières syndrome
329478	Adult-onset distal myopathy due to VCP mutation	398147	AFP	101089	AID deficiency
199351	Adult-onset dystonia-parkinsonism	139507	African iron overload	98916	AIDP
99000	Adult-onset foveomacular dystrophy	101334	African tick typhus	90081	AIDS wasting syndrome
99000	Adult-onset foveomacular dystrophy with choroidal neovascularization	3385	African trypanosomiasis	178333	AIED
99000	Adult-onset foveomacular vitelliform dystrophy	33110	Agammaglobulinemia, non-Bruton type	363549	AIEF
79257	Adult-onset GM1 gangliosidosis	83617	Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	86886	AILT
306431	Adult-onset immunodeficiency with anti-interferon-gamma autoantibodies	388	Aganglionic megacolon	103919	AIP
313808	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	35704	AGAT deficiency	280302	AIP type 1
329314	Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency	85448	AGel amyloidosis	280315	AIP type 2
329314	Adult-onset multiple mtDNA deletion syndrome due to DGUOK deficiency	180142	Agenesis and aplasia of uterine body	75564	AISA
391490	Adult-onset myasthenia gravis	52055	Agenesis of the corpus callosum-intellectual disability-coloboma-micrognathia syndrome	33355	AK2 deficiency
171442	Adult-onset nemaline myopathy	99114	Agenesis of the superior caval vein	38	AKE
276608	Adult-onset non-insulinoma persistent hyperinsulinemic hypoglycemia	99114	Agenesis of the superior vena cava	→35722	Akesson syndrome
35689	Adult-onset PLS	99114	Agenesis of the SVC	5	
35689	Adult-onset primary lateral sclerosis	293173	AGEP	79085	AKT2-related familial partial lipodystrophy
209335	Adult-onset proximal spinal muscular atrophy, autosomal dominant	873	Aggressive fibromatosis	79085	AKT2-related FPLD
829	Adult-onset Still disease	86873	Aggressive NK-cell leukemia	85443	AL amyloidosis
		86873	Aggressive NK-cell lymphoma	2232	Al Awadi-Farag-Teebi syndrome
		98850	Aggressive systemic mastocytosis	2879	Al Awadi-Raas-Rothschild syndrome
		989	Aglossia-adactylia syndrome	→3157	Al Frayh-Facharzt-Haque syndrome
		990	Agnathia-holoprosencephaly-situs inversus syndrome	2725	Al Gazali-Al Talabani syndrome
		824	Agnogenic myeloid metaplasia	2865	Al Gazali-Aziz-Salem syndrome
		100070	Agrammatic variant of PPA	2153	Al Gazali-Donnai-Muller syndrome
		100070	Agrammatic variant of primary progressive aphasia	2725	Al Gazali-Lytte syndrome
		442582	AH amyloidosis	2773	Al Gazali-Nair syndrome
		2131	AHC	→32473	Al-Gazali-Dattani syndrome
		412069	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	7	

→ 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
261629	Alagille syndrome due to a NOTCH2 point mutation	363717	Alexander disease type I	157954	Alopecia-progressive neurological defect-endocrinopathy syndrome
261600	Alagille syndrome due to del(20)(p12)	363722	Alexander disease type II	726	Alpers progressive sclerosing poliodystrophy
261600	Alagille syndrome due to monosomy 20p12	261112	Alfi syndrome	726	Alpers syndrome
52	Alagille-Watson syndrome	79327	ALG1-CDG	726	Alpers-Huttenlocher syndrome
261619	Alagille-Watson syndrome due to a JAG1 point mutation	79326	ALG2-CDG	734	Alpha delta granule deficiency
261629	Alagille-Watson syndrome due to a NOTCH2 point mutation	79321	ALG3-CDG	734	Alpha dense granule deficiency
261600	Alagille-Watson syndrome due to monosomy 20p12	79320	ALG6-CDG	134	Alpha methylacetoacetic aciduria
178333	Åland Islands eye disease	79325	ALG8-CDG	721	Alpha storage pool deficiency
2007	Alar cartilages hypoplasia-coloboma-telecanthus syndrome	79328	ALG9-CDG	98791	Alpha thalassemia-intellectual disability syndrome, deletion type
53	Albers-Schönberg osteopetrosis	280071	ALG11-CDG	98791	Alpha thalassemia-retardation syndrome
→897	Albinism-black lock-cell migration disorder of the neurocytes of the gut-sensorineural deafness syndrome	79324	ALG12-CDG	365	Alpha-1,4-glucosidase acid deficiency
998	Albinism-deafness syndrome	324422	ALG13-CDG	308552	Alpha-1,4-glucosidase acid deficiency, infantile onset
665	Albright hereditary osteodystrophy	99995	Algodystrophy	420429	Alpha-1,4-glucosidase acid deficiency, late onset
79443	Albright hereditary osteodystrophy - PHP Ia	300895	ALK+ ALCL	93594	Alpha-1-antichymotrypsin deficiency
79445	Albright hereditary osteodystrophy - PPHP	364043	ALK+ large B-cell lymphoma	60	Alpha-1-antitrypsin deficiency
1001	Albright hereditary osteodystrophy type 3	364043	ALK+ LBCL	79154	Alpha-aminoacidic aciduria
1001	Albright hereditary osteodystrophy-like syndrome	300903	ALK- ALCL	399058	Alpha-B crystallin-related late-onset distal myopathy
98841	ALCL	300903	ALK- anaplastic large cell lymphoma	324	Alpha-galactosidase A deficiency
60039	Alcock syndrome	300903	ALK-negative anaplastic large cell lymphoma	100025	Alpha-HCD
1915	Alcohol-related birth defects	300895	ALK-positive anaplastic large cell lymphoma	100025	Alpha-heavy chain disease
1915	Alcohol-related neurodevelopmental disorder	364043	ALK-positive large B-cell lymphoma	31	Alpha-ketoglutarate dehydrogenase deficiency
36899	Alcohol-responsive dystonia	56	Alkaptonuria	349	Alpha-L-fucosidase deficiency
43	ALD	59	Allan-Herndon-Dudley syndrome	579	Alpha-L-iduronidase deficiency
324977	ALDD syndrome	1164	Allergic aspergillosis	61	Alpha-mannosidosis
35664	ALDH18A1-related De Barysy syndrome	1164	Allergic bronchopulmonary aspergillosis	309288	Alpha-mannosidosis, adult form
99763	Aldosterone synthase deficiency	93925	Alobar holoprosencephaly	309282	Alpha-mannosidosis, infantile form
99764	Aldosterone synthase deficiency unrelated to CYP11B2	1006	Alopecia antibody deficiency	134	Alpha-methyl-acetoacetyl-CoA thiolase deficiency
99764	Aldosterone synthase deficiency unrelated to the aldosterone synthase gene	700	Alopecia totalis	79095	Alpha-methyl-acyl-CoA racemase deficiency
369929	Aldosterone-producing adenoma with seizures and neurological abnormalities	701	Alopecia universalis	3137	Alpha-N-acetylgalactosaminidase deficiency
369929	Aldosterone-secreting adenoma with seizures and neurological abnormalities	2316	Alopecia-anosmia-deafness-hypogonadism syndrome	79279	Alpha-N-acetylgalactosaminidase deficiency type 1
85332	Aldred syndrome	1005	Alopecia-contractions-dwarfism-intellectual disability syndrome	79280	Alpha-N-acetylgalactosaminidase deficiency type 2
439224	ALECT2 amyloidosis	202	Alopecia-deafness-hypogonadism syndrome	79281	Alpha-N-acetylgalactosaminidase deficiency type 3
158799	Aleukemic mast cell leukemia	2574	Alopecia-epilepsy-oligophrenia syndrome, Moynahan type	62	Alpha-sarcoglycanopathy
58	Alexander disease	1008	Alopecia-epilepsy-pyorrhea-intellectual disability syndrome	846	Alpha-thalassemia
		→3464	Alopecia-hypogonadism-extrapyramidal disorder syndrome	163596	Alpha-thalassemia hydrops fetalis
		2850	Alopecia-intellectual disability syndrome	93616	Alpha-thalassemia intermedia
		1014	Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome	163596	Alpha-thalassemia major
				98791	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16

→ 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
231401	Alpha-thalassemia-myelodysplastic syndrome	294967	Amelia of upper limb	86845	AML with multilineage dysplasia
847	Alpha-thalassemia-X-linked intellectual disability syndrome	295055	Amelia of upper limb, bilateral	402026	AML with NPM1 somatic mutations
63	Alport deafness-nephropathy	295053	Amelia of upper limb, unilateral	402014	AML with t(6;9)(p23;q34)
→18205 0	Alport syndrome with leukocyte inclusions and macrothrombocytopenia	1946	Amelo-cerebro-hypohidrotic syndrome	370026	AML with t(8;16)(p11;p13) translocation
→18205 0	Alport syndrome with macrothrombocytopenia	1028	Amelo-onycho-hypohidrotic syndrome	102724	AML with t(8;21)(q22;q22) translocation
86818	Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	314422	Ameloblastic carcinoma	402017	AML with t(9;11)(p22;q23)
3261	ALPS	314419	Ameloblastoma	520	AML with t(15;17)(q22;q12);(PML/RARalpha) and variants
436159	ALPS due to CTLA4 haploinsufficiency	88661	Amelogenesis imperfecta	86818	AMME complex
268114	ALPS type 4	100031	Amelogenesis imperfecta type 1	86818	AMME syndrome
436159	ALPS type 5	100033	Amelogenesis imperfecta type 2	251663	aMOA
268114	ALPS type IV	100032	Amelogenesis imperfecta type 3	67	Amoebiasis due to Entamoeba histolytica
436159	ALPS type V	100034	Amelogenesis imperfecta type 4	68	Amoebiasis due to free-living amoebae
275517	ALPS with recurrent viral infections	171836	Amelogenesis imperfecta-gingival hyperplasia syndrome	45	AMP deaminase deficiency
803	ALS	1031	Amelogenesis imperfecta-nephrocalcinosis syndrome	1035	Ampola syndrome
357043	ALS4	83595	American mountain fever	66529	Ampulla cardiomyopathy
86815	ALSG	3386	American trypanosomiasis	300557	Ampullary carcinoma
313808	ALSP	2116	Aminoaciduria, Hartnup type	300557	Ampulloma
64	Alström syndrome	141	Aminoacylase 2 deficiency	98917	AMSAN
99971	ALT	1908	Aminopterin embryopathy syndrome	366	Amylo-1,6-glucosidase deficiency
2131	Alternating hemiplegia in childhood	221120	Aminopterin syndrome-like sine aminopterin	49804	Amyloid lichen
2131	Alternating hemiplegia of childhood	1908	Aminopterin/methotrexate embryofetopathy	319635	Amyloidosis cutis dyschromia
210122	Alveolar capillary dysplasia with misalignment of pulmonary veins	→33364	Amish brittle hair syndrome	319635	Amyloidosis cutis dyschromica
210122	Alveolar capillary dysplasia with misalignment of pulmonary vessels	171714	Amish infantile epilepsy syndrome	85450	Amyloidosis, Oster tag type
284	Alveolar echinococcosis	99742	Amish lethal microcephaly	367	Amylopectinosis
99756	Alveolar rhabdomyosarcoma	98902	Amish nemaline myopathy	803	Amyotrophic lateral sclerosis
163699	Alveolar soft-part sarcoma	518	AMKL	357043	Amyotrophic lateral sclerosis type 4
163699	Alveolar soft-tissue sarcoma	102379	AML and myelodysplastic syndromes related to alkylating agent	94091	Amyotrophic lateral sclerosis, hemiplegic type
→1071	Alveolar synechia-ankyloblepharon-ectodermal dysplasia syndrome	164726	AML and myelodysplastic syndromes related to radiation	90020	Amyotrophic lateral sclerosis-parkinsonism-dementia complex
3354	Alves-dos Santos-Castelo syndrome	102381	AML and myelodysplastic syndromes related to topoisomerase type 2 inhibitor	90020	Amyotrophic lateral sclerosis-parkinsonism-dementia of Guam syndrome
306542	ALX1-related frontonasal dysplasia	98832	AML M0	2615	Amyotrophy-fat tissue anomaly syndrome
391474	ALX3-related frontonasal dysplasia	98833	AML M1	228113	Anal fistula
228390	ALX4-related FNDAG	98834	AML M2	31150	Analphalipoproteinemia
169095	Alymphoid cystic thymic dysgenesis	520	AML M3	761	Anaphylactoid purpura
93561	ALys amyloidosis	517	AML M4	251589	Anaplastic astrocytoma
79095	AMACR deficiency	514	AML M5	251646	Anaplastic ependymoma
98918	AMAN	318	AML M6	251957	Anaplastic ganglioglioma
65	Amaurosis congenita of Leber	518	AML M7	98841	Anaplastic large cell lymphoma
1021	Amaurosis-hypertrichosis syndrome	98831	AML with 11q23 abnormalities	251663	Anaplastic oligoastrocytoma
1023	Ambras syndrome	98829	AML with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)	251630	Anaplastic oligodendrogloma
294969	Amelia of lower limb	319480	AML with CEBPA somatic mutations	142	Anaplastic thyroid carcinoma
295059	Amelia of lower limb, bilateral	402020	AML with inv3(p21;q26.2) or t(3;3)(p21;q26.2)	251855	Anaplastic/large cell medulloblastoma
295057	Amelia of lower limb, unilateral			93347	Anauxetic dysplasia

→ 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
79262	ANCL	98813	Anhidrotic ectodermal dysplasia with immunodeficiency	1104	Anophthalmia plus syndrome
78	Ancylostomiasis	69088	Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome	1106	Anophthalmia-syndactyly syndrome
1496	Andermann syndrome	→1071	Anhidrotic ectodermic dysplasia-cleft lip/palate syndrome	77298	Anophthalmia/microphthalmia - esophageal atresia
37553	Andersen cardiодysrhythmic periodic paralysis	1067	Aniridia - ptosis - intellectual disability - familial obesity	1882	ANOTHER syndrome
367	Andersen disease	1064	Aniridia - renal agenesis - psychomotor retardation	93976	Anotia
37553	Andersen syndrome	1069	Aniridia-absent patella syndrome	2987	Antecubital pterygium syndrome
37553	Andersen-Tawil syndrome	1065	Aniridia-cerebellar ataxia-intellectual disability syndrome	93604	Antenatal Bartter syndrome
71	Anderson disease	1068	Aniridia-intellectual disability syndrome	294	Antenatal CMV infection
324	Anderson-Fabry disease	1070	Anisakiasis	294	Antenatal cytomegalovirus infection
99916	Androblastoma	86873	ANKCL	292	Antenatal enterovirus infection
329813	Androgenetic/biparental mosaicism	1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	70596	Antenatal Epstein-Barr virus infection
157954	ANE syndrome	1074	Ankyloblepharon filiforme - imperforate anus	293	Antenatal herpes virus infection
263524	ANEC	1072	Ankyloblepharon filiforme adnatum - cleft palate	178148	Antenatal multiminicore disease with arthrogryposis multiplex congenita
1044	Anemia due to adenosine triphosphatase deficiency	2206	Ankylosing vertebral hyperostosis with tylosis	291	Antenatal varicella virus infection
1054	Aneurysm of sinus of Valsalva	1077	Ankylosis of teeth	1931	Anterior encephalocele
95484	Aneurysm or dilatation of ascending aorta	78	Ankylostomiasis	98961	Anterior limiting membrane dystrophy type I
284984	Aneurysm-osteoarthritis syndrome	254411	Annular atrophic lichen planus	98960	Anterior limiting membrane dystrophy type II
353344	Aneurysmal telangiectasia	254411	Annular atrophic LP	95512	Anterior pituitary hypophysitis
63442	Angel-shaped phalango-epiphyseal dysplasia	281139	Annular epidermolytic ichthyosis	435372	Anterior urethral valve
72	Angelman syndrome	254424	Annular lichen planus	90079	Anthracycline extravasations
411511	Angelman syndrome due to a point mutation	254424	Annular LP	36412	Anti-C1q vasculitis
411515	Angelman syndrome due to imprinting defect in 15q11-q13	675	Annular pancreas	375	Anti-GBM syndrome
98794	Angelman syndrome due to maternal 15q11q13 deletion	229	Annuloaortic ectasia	375	Anti-glomerular basement membrane disease
98794	Angelman syndrome due to maternal monosomy 15q11q13	99797	Anodontia	2194	Anti-HLA hyperimmunization
98795	Angelman syndrome due to paternal uniparental disomy of chromosome 15	101932	Anomaly of the mitral subvalvular apparatus	206569	Anti-HMG-CoA myopathy
251671	Angiocentric glioma	99055	Anomaly of the tricuspid valve chordae	81	Anti-Jo1 syndrome
86879	Angiocentric T-cell lymphoma	1094	Anonychia - microcephaly	639	Anti-MAG neuropathy
79093	Angiodysgenetic necrotizing myelopathy	90390	Anonychia - onychodystrophy	206569	Anti-SRP myopathy
98839	Angioendotheliomatosis proliferans systematisata	1487	Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges	413667	Antidepressant or antipsychotic toxicity or dose selection
160	Angiofollicular ganglionic hyperplasia	94150	Anonychia congenita totalis	2821	Antinolo-Nieto-Borrego syndrome
160	Angiofollicular lymph hyperplasia	69125	Anonychia with flexural pigmentation	3006	Antiquitin deficiency
86886	Angioimmunoblastic T-cell lymphoma	→2470	Anophthalmia - heart and pulmonary anomalies - intellectual disability	81	Antisynthetase syndrome
324	Angiokeratoma corporis diffusum	→3157	Anophthalmia - hypothalamo-pituitary insufficiency	83	Antley-Bixler syndrome
95429	Angioma serpiginosum	1101	Anophthalmia - megalocornea - cardiopathy - skeletal anomalies	→95699	Antley-Bixler syndrome type 2
2346	Angioosteohypertrophic syndrome	2470	Anophthalmia - pulmonary hypoplasia	→95699	Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis
75508	Angioosteohypotrophic syndrome			→95699	Antley-Bixler syndrome, POR-related
263413	Angiosarcoma			→95699	Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis
74	Angiostrongyliasis			1190	AO1
98839	Angiotropic large cell lymphoma			56305	AO3
370039	Angora hair nevus			1168	AOA1
76	Anguilluliasis			64753	AOA2
76	Anguillulosis			99000	AOFMD
238468	Anhidrotic ectodermal dysplasia			1190	AOI

→ 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
70590	AOI	370046	Aplasia cutis congenita-nevus sebaceus syndrome	1129	Arachnodactyly - abnormal ossification - intellectual disability
56305	AOIII	86815	Aplasia of lacrimal and salivary glands	1130	Arachnodactyly - intellectual disability - dysmorphism
1457	Aorta coarctation	3329	Aplasia of tibia with split-hand/split-foot deformity	2356	Arachnoid cyst
60030	Aortic aneurysm syndrome due to TGF-beta receptors anomalies	2879	Aplasia/hypoplasia of limbs and pelvis	137817	Arachnoiditis
1110	Aortic arch anomaly - peculiar facies - intellectual disability	70590	Apnea of infancy	324442	ARAN-NM
2299	Aortic arch interruption	99981	Apnea of prematurity	1915	ARBD
99079	Aortic arch syndrome	425	ApoA-I deficiency	2697	ARC syndrome
→91387	Aortic dilatation - joint hypermobility - arterial tortuosity	294986	Apodia	88644	ARCA1
95448	Aortic valve atresia	295107	Apodia, bilateral	139485	ARCA2
101043	Aortic valve dysplasia	93560	Apolipoprotein A-I amyloidosis	90349	ARCL1
99071	Aorto-left ventricular tunnel	425	Apolipoprotein A-I deficiency	357074	ARCL2, classic type
99070	Aorto-right ventricular tunnel	238269	Apolipoprotein A-II amyloidosis	357074	ARCL2, Debré type
3400	Aorto-ventricular tunnel	439232	Apolipoprotein A-IV amyloidosis	357064	ARCL2, progeroid type
99086	Aortopulmonary coronary arterial course	320	Apparent mineralocorticoid excess	357058	ARCL2A
974	AOS	100079	Appendiceal endocrine tumor	357064	ARCL2B
284984	AOS	391723	Appendiceal mucinous adenocarcinoma	324442	ARCMT2-NM
829	AOSD	1201	Apple peel syndrome	101097	ARCMT2K
280763	AP4 deficiency syndrome	1126	Aprosencephaly cerebellar dysgenesis	1133	AREDYLD syndrome
369929	APA with seizures and neurological abnormalities	976	APRT deficiency	101096	Aregenerative anemia
747	aPAP	3453	APS1	→702	Arena syndrome
206583	APBD	3143	APS2	75377	Areolar atrophy of the macula
247806	APC-related AFAP	227982	APS3	319223	Argentine hemorrhagic fever
247806	APC-related attenuated familial adenomatous polyposis	227990	APS4	319223	Argentinian hemorrhagic fever
247806	APC-related attenuated familial polyposis coli	3453	APS type 1	90	Arginase deficiency
247806	APC-related attenuated FAP	3143	APS type 2	90	Argininemia
397596	APDS	227982	APS type 3	23	Argininosuccinate deficiency
3453	APECED syndrome	227990	APS type 4	247525	Argininosuccinate synthase deficiency
87	Apert syndrome	101206	APV/ADA, Fallot type	247525	Argininosuccinate synthetase deficiency
162521	Apertura pyriformis with holoprosencephaly	99048	APV/PDA, non-Fallot type	23	Argininosuccinatelyase deficiency
1112	Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis	402041	AR dRTA	23	Argininosuccinic acid lyase deficiency
1113	Aphalangy - syndactyly - microcephaly	→40204	AR dRTA with deafness	247525	Argininosuccinic acid synthase deficiency
49	Aphallia	1	AR dRTA with hearing loss	247525	Argininosuccinic acid synthetase deficiency
324540	Aphonia - deafness - retinal dystrophy - bifid halluces - intellectual disability	→40204	AR dRTA without deafness	23	Argininosuccinic aciduria
324540	Aphonia - deafness - retinal dystrophy - duplicated halluces - intellectual disability	1	AR dRTA without hearing loss	60014	Argyria
66529	Apical ballooning syndrome	93607	AR pRTA	97342	Argyrophilic grain disease
324530	APLAID	90119	AR-CMT2 with acrodystryphy	289176	ARHR
1117	Aplasia cutis - myopia	90118	AR-CMT2, Ouvrier type	79235	Arias syndrome
1114	Aplasia cutis congenita	98856	AR-CMT2B1	2318	Arima syndrome
3339	Aplasia cutis congenita - epibulbar dermoids	101101	AR-CMT2B2	950	Arkless-Graham syndrome
1116	Aplasia cutis congenita - intestinal lymphangiectasia	101102	AR-CMT2C	85276	Armfield syndrome
		443950	AR-CMT2T	1915	ARND
		169186	AR-CNMs	167635	Arndt-Gottron disease
		248	AR-HED	268882	Arnold-Chiari malformation type 1
		88616	AR-NSID	1136	Arnold-Chiari malformation type 2
				268882	Arnold-Chiari malformation type I
				1136	Arnold-Chiari malformation type II
				91	Aromatase deficiency
				178345	Aromatase excess syndrome

→ 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
35708	Aromatic L-amino acid decarboxylase deficiency	309263	Arylsulfatase A deficiency, juvenile form	137639	Ataxia - delayed dentition - hypomyelination
254886	arPEO	309256	Arylsulfatase A deficiency, late infantile form	1227	Ataxia - diabetes - goiter - gonadal insufficiency
99916	Arrhenoblastoma	583	Arylsulfatase B deficiency	1180	Ataxia - hypogonadism - choroidal dystrophy
1134	Arrhinia	276212	Arylsulfatase B deficiency, rapidly progressing	1168	Ataxia - oculomotor apraxia type 1
1135	Arrhinia - choanal atresia - microphthalmia	276223	Arylsulfatase B deficiency, slowly progressing	64753	Ataxia - oculomotor apraxia type 2
260305	ARSA	81	AS syndrome	2585	Ataxia - pancytopenia
98	ARSACS	23	ASA deficiency	1184	Ataxia - photosensitivity - short stature
314603	ARSL	231466	ASAN	1178	Ataxia - tapetoretinal degeneration
583	ARSB deficiency	583	ASB deficiency	96	Ataxia with isolated vitamin E deficiency
357107	Arterial cervical rib syndrome	2302	Asbestos intoxication	3008	Ataxia with lactic acidosis type 2
357107	Arterial costoclavicular syndrome	2302	Asbestosis	3008	Ataxia with lactic acidosis type II
1682	Arterial dissection - lentiginosis	1253	Ascher syndrome	94147	Ataxia with pigmentary retinopathy
357107	Arterial hyperabduction syndrome	447997	ASCT1 deficiency	96	Ataxia with vitamin E deficiency
357107	Arterial scalenus anticus syndrome	1478	ASD	1188	Ataxia-deafness-intellectual disability syndrome
357107	Arterial thoracic outlet compression syndrome	352490	ASD due to AUTS2 deficiency	370022	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome
357107	Arterial thoracic outlet syndrome	99104	ASD, coronary sinus type	100	Ataxia-telangiectasia
3342	Arterial tortuosity syndrome	99106	ASD, ostium primum type	370109	Ataxia-telangiectasia variant
357107	Arterial TOS	99103	ASD, ostium secundum type	647	Ataxia-telangiectasia, variant 1
52	Arteriohepatic dysplasia	99105	ASD, sinus venosus type	251347	Ataxia-telangiectasia-like disorder
261619	Arteriohepatic dysplasia due to a JAG1 point mutation	54251	Aseptic abscesses syndrome	1183	Ataxo-opso-myoclonus syndrome
261629	Arteriohepatic dysplasia due to a NOTCH2 point mutation	97337	Aseptic necrosis of patella	2953	ATCS
261600	Arteriohepatic dysplasia due to monosomy 20p12	3314	Aseptic necrosis of phalangeal epiphyses	3469	Atelencephaly
29207	Arthritis urethritica	2380	Aseptic necrosis of the capital femoral epiphysis	1190	Atelosteogenesis type 1
955	Arthrodentosteodysplasia	97336	Aseptic necrosis of the capital humerus	56304	Atelosteogenesis type 2
3200	Arthrogryposis - ectodermal dysplasia - other anomalies	97332	Aseptic necrosis of the lunate bone	56305	Atelosteogenesis type 3
1485	Arthrogryposis - hyperkeratosis, lethal form	2054	Aseptic necrosis of the tarsal bone	1190	Atelosteogenesis type I
2697	Arthrogryposis - renal dysfunction - cholestasis	97335	Aseptic necrosis of the tibial tubercle	56304	Atelosteogenesis type II
65720	Arthrogryposis - severe scoliosis	57194	Aseptic osteitis	56305	Atelosteogenesis type III
1155	Arthrogryposis due to muscular dystrophy	54251	Aseptic systemic abscesses	69739	Athabaskan brainstem dysgenesis syndrome
994	Arthrogryposis multiplex congenita - pulmonary hypoplasia	137686	Asherman syndrome	69739	Athabaskan brainstem dysgenesis syndrome
1150	Arthrogryposis multiplex congenita - whistling face	276198	Asidan	1192	Atherosclerosis - deafness - diabetes - epilepsy - nephropathy
1154	Arthrogryposis with oculomotor limitation and electroretinal anomalies	23	ASL deficiency	95713	Athyreosis
1144	Arthrogryposis-like hand anomaly - sensorineural deafness	391376	Asparagine synthetase deficiency	1226	Athyroidal hypothyroidism-spiky hair-cleft palate syndrome
1149	Arthrogryposis-like syndrome	141	Aspartoacylase deficiency	250977	ATIC deficiency
2848	Arthropathy-camptodactyly syndrome	93	Aspartylglucosaminidase deficiency	1193	Atkin-Flaitz syndrome
1187	Arts syndrome	93	Aspartylglucosaminuria	99666	Atlantoaxial subluxation
512	Arylsulfatase A deficiency	63442	ASPED	251347	ATLD
309271	Arylsulfatase A deficiency, adult form	1163	Aspergillosis	86875	ATLL
		474	Asphyxiating thoracic dystrophy of the newborn	139423	ATM/TM
		163699	ASPS	231401	ATMDS
		247525	ASS deficiency	163934	Atopic keratoconjunctivitis
		221120	ASSA	357107	ATOS
		85175	Astley-Kendall dysplasia		
		251679	Astroblastoma		
		647	AT V1		

→ 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
31150	ATP-binding cassette transporter A1 deficiency	85447	ATTRV30M-related amyloidosis	289863	Atypical NKA
98791	ATR syndrome linked to chromosome 16	330001	ATTRwt amyloidosis	289863	Atypical non-ketotic hyperglycinemia
98791	ATR syndrome, deletion type	330001	ATTRwt-related amyloidosis	261501	Atypical Norrie disease due to del(X)(p11.3)
98791	ATR-16 syndrome	95487	Atypical arterial duct	261501	Atypical Norrie disease due to monosomy Xp11.3
847	ATR-X syndrome	199627	Atypical autism	261501	Atypical Norrie disease due to Xp11.3 microdeletion
30391	Atresia of bile ducts	352723	Atypical Chédiak-Higashi syndrome	216873	Atypical pantothenate kinase-associated neurodegeneration
1201	Atresia of small intestine	98824	Atypical chronic myeloid leukemia	251902	Atypical papilloma of choroid plexus
105	Atresia of urethra	1456	Atypical coarctation of aorta	95487	Atypical patent ductus arteriosus
1344	Atrial cardiomyopathy with heart block	314466	Atypical Demons-Meigs syndrome	79474	Atypical progeroid syndrome
99107	Atrial septal aneurysm	314721	Atypical dentin dysplasia due to SMOC2 deficiency	99750	Atypical progressive supranuclear palsy
1478	Atrial septal defect	398147	Atypical facial pain	99750	Atypical PSP
1479	Atrial septal defect - atrioventricular conduction defects	309252	Atypical Gaucher disease due to saposin C deficiency	3095	Atypical Rett syndrome
99104	Atrial septal defect, coronary sinus type	289863	Atypical glycine encephalopathy	3095	Atypical RTT
99106	Atrial septal defect, ostium primum type	98961	Atypical granular corneal dystrophy	99966	Atypical teratoid rhabdoid tumor
99103	Atrial septal defect, ostium secundum type	238523	Atypical HCS	90393	Atypical tuberous myxedema of Jadassohn-Dosseker
99105	Atrial septal defect, sinus venosus type	2134	Atypical hemolytic-uremic syndrome	79474	Atypical Werner syndrome
1344	Atrial standstill	93581	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	16	Atypical X-linked achromatopsia
844	Atrial tachyarrhythmia with short PR interval	93578	Atypical hemolytic-uremic syndrome with B factor anomaly	166415	Audiogenic seizures
86819	Atrichia with papular lesions	93575	Atypical hemolytic-uremic syndrome with C3 anomaly	1074	Aughton-Hufnagle syndrome
392	Atriodigital dysplasia type 1	357008	Atypical hemolytic-uremic syndrome with DGKE deficiency	1488	Aural atresia - multiple congenital anomalies - intellectual disability
1350	Atriodigital dysplasia type 2	93579	Atypical hemolytic-uremic syndrome with H factor anomaly	→794	Auralcephalosyndactyly
1342	Atriodigital dysplasia type 3	93580	Atypical hemolytic-uremic syndrome with I factor anomaly	77300	Auricular abnormalities - cleft lip with or without cleft palate - ocular abnormalities
168796	Atriodigital dysplasia, Slovenian type	93576	Atypical hemolytic-uremic syndrome with MCP/CD46 anomaly	137888	Auriculocondylar syndrome
1352	Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	217023	Atypical hemolytic-uremic syndrome with thrombomodulin anomaly	71270	Auriculocular anomalies - cleft lip
86813	Atrophia areata	2134	Atypical HUS	114	Auriculosteodysplasia
649	Atrophia bulborum hereditaria	93581	Atypical HUS with anti-factor H antibodies	→794	Aurocephalosyndactyly
254449	Atrophic lichen planus	93578	Atypical HUS with B factor anomaly	1995	Ausems-Wittebol Post-Hennekam syndrome
254449	Atrophic LP	93575	Atypical HUS with C3 anomaly	137911	Autism - facial port-wine stain
79100	Atrophoderma vermiculata	357008	Atypical HUS with DGKE deficiency	352490	Autism spectrum disorder due to AUTS2 deficiency
99966	ATRT	93579	Atypical HUS with H factor anomaly	370943	Autism spectrum disorder-epilepsy-arthrogryposis syndrome
71289	ATRUS syndrome	93580	Atypical HUS with I factor anomaly	308410	Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency
3342	ATS	93576	Atypical HUS with MCP/CD46 anomaly	324636	Autoerythrocyte sensitization syndrome
86818	ATS-MR	217023	Atypical HUS with thrombomodulin anomaly	85138	Autoimmune Addison's disease
352723	Attenuated Chédiak-Higashi syndrome	238523	Atypical hypotonia - cystinuria syndrome	85138	Autoimmune adrenitis
220460	Attenuated familial adenomatous polyposis	391411	Atypical juvenile parkinsonism	420789	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea
220460	Attenuated familial polyposis coli	86797	Atypical lichen myxedematosus	391487	Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome
220460	Attenuated FAP	99971	Atypical lipoma		
85451	ATTR cardiomyopathy	99971	Atypical lipomatous tumor		
85451	ATTRV122I amyloidosis	314466	Atypical Meigs syndrome		
85451	ATTRV122I-related amyloidosis	2578	Atypical MRKH syndrome		
85447	ATTRV30M amyloidosis				

→ 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
37042	Autoimmune enteropathy type 1	227982	Autoimmune polyendocrinopathy type 3	397735	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to MARS mutation
103916	Autoimmune enteropathy type 2	227990	Autoimmune polyendocrinopathy type 4	447964	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to NAGLU mutation
103917	Autoimmune enteropathy type 3	3453	Autoimmune polyglandular syndrome type 1	435819	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation
1959	Autoimmune hemolytic anemia and autoimmune thrombocytopenia	3143	Autoimmune polyglandular syndrome type 2	435387	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to VCP mutation
90033	Autoimmune hemolytic anemia, warm type	227982	Autoimmune polyglandular syndrome type 3	401964	Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons
444463	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome	227990	Autoimmune polyglandular syndrome type 4	99946	Autosomal dominant Charcot-Marie-Tooth disease type 2A1
2137	Autoimmune hepatitis	747	Autoimmune pulmonary alveolar proteinosis	99947	Autosomal dominant Charcot-Marie-Tooth disease type 2A2
36913	Autoimmune hypoparathyroidism	93585	Autoimmune thrombotic thrombocytopenic purpura	99936	Autosomal dominant Charcot-Marie-Tooth disease type 2B
3453	Autoimmune hypoparathyroidism - chronic candidiasis - Addison's disease	3143	Autoimmune thyroid disease and/or type 1 diabetes - Addison disease	99937	Autosomal dominant Charcot-Marie-Tooth disease type 2C
444092	Autoimmune interstitial lung disease-arthritis syndrome	592	Autoimmune/inflammatory syndrome induced by adjuvant with persisting aluminic granuloma	99938	Autosomal dominant Charcot-Marie-Tooth disease type 2D
3261	Autoimmune lymphoproliferative syndrome	324977	Autoinflammation-lipodystrophy-dermatosis syndrome	99939	Autosomal dominant Charcot-Marie-Tooth disease type 2E
436159	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsufficiency	324530	Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation	99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F
268114	Autoimmune lymphoproliferative syndrome type 4	210115	Autoinflammatory disease due to interleukin-1 receptor antagonist deficiency	99941	Autosomal dominant Charcot-Marie-Tooth disease type 2G
436159	Autoimmune lymphoproliferative syndrome type 5	329173	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis	99942	Autosomal dominant Charcot-Marie-Tooth disease type 2I
268114	Autoimmune lymphoproliferative syndrome type IV	33110	Autosomal agammaglobulinemia	99943	Autosomal dominant Charcot-Marie-Tooth disease type 2J
436159	Autoimmune lymphoproliferative syndrome type V	88918	Autosomal dominant Alport syndrome	99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K
275517	Autoimmune lymphoproliferative syndrome with recurrent viral infections	1810	Autosomal dominant anhidrotic ectodermal dysplasia	99945	Autosomal dominant Charcot-Marie-Tooth disease type 2L
589	Autoimmune myasthenia gravis	314399	Autosomal dominant aplasia and myelodysplasia	228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M
206569	Autoimmune necrotizing myopathy	314399	Autosomal dominant aplastic anemia and myelodysplasia	228174	Autosomal dominant Charcot-Marie-Tooth disease type 2N
206569	Autoimmune necrotizing myositis	1216	Autosomal dominant benign distal spinal muscular atrophy	284232	Autosomal dominant Charcot-Marie-Tooth disease type 2O
103919	Autoimmune pancreatitis	314652	Autosomal dominant beta2-microglobulinic amyloidosis	329258	Autosomal dominant Charcot-Marie-Tooth disease type 2Q
280302	Autoimmune pancreatitis type 1	93304	Autosomal dominant brachyolmia	397735	Autosomal dominant Charcot-Marie-Tooth disease type 2U
280315	Autoimmune pancreatitis type 2	169189	Autosomal dominant centronuclear myopathy	447964	Autosomal dominant Charcot-Marie-Tooth disease type 2V
747	Autoimmune PAP	314404	Autosomal dominant cerebellar ataxia, deafness and narcolepsy	98975	Autosomal dominant CHED
3453	Autoimmune polyendocrine syndrome type 1	314404	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	363447	Autosomal dominant childhood-onset proximal spinal muscular atrophy
3143	Autoimmune polyendocrine syndrome type 2	324611	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation	363454	Autosomal dominant childhood-onset proximal spinal muscular atrophy with contractures
227982	Autoimmune polyendocrine syndrome type 3				
227990	Autoimmune polyendocrine syndrome type 4				
3453	Autoimmune polyendocrinopathy - candidiasis - ectodermal dystrophy syndrome				
3453	Autoimmune polyendocrinopathy type 1				
3143	Autoimmune polyendocrinopathy type 2				

→ 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
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 sensorineural hearing loss type DFNA
79344	Autosomal dominant chondrodysplasia punctata	2024	Autosomal dominant gingival fibromatosis	93325	Autosomal dominant Kenny-Caffey syndrome
→2526	Autosomal dominant chorioretinopathy - microcephaly	2024	Autosomal dominant gingival hyperplasia	2334	Autosomal dominant keratitis
1455	Autosomal dominant coarctation of aorta	139491	Autosomal dominant hereditary hemochromatosis	293936	Autosomal dominant keratoconus with early-onset anterior polar cataracts
447753	Autosomal dominant complex spastic paraplegia type 9A	401964	Autosomal dominant hereditary motor and sensory neuropathy type 2 with giant axons	503	Autosomal dominant Larsen syndrome
447757	Autosomal dominant complex spastic paraplegia type 9B	2314	Autosomal dominant HIES	411602	Autosomal dominant late-onset Parkinson disease
1216	Autosomal dominant congenital benign spinal muscular atrophy	2314	Autosomal dominant hyper-IgE syndrome	67042	Autosomal dominant late-onset retinal degeneration
98975	Autosomal dominant congenital hereditary endothelial dystrophy	2314	Autosomal dominant hyperimmunoglobulin E syndrome	101046	Autosomal dominant lateral temporal lobe epilepsy
86814	Autosomal dominant cortical myoclonus and epilepsy	276580	Autosomal dominant hyperinsulinemic hypoglycemia due to Kir6.2 deficiency	313808	Autosomal dominant leukoencephalopathy with neuroaxonal spheroids
90348	Autosomal dominant cutis laxa	276575	Autosomal dominant hyperinsulinemic hypoglycemia due to SUR1 deficiency	266	Autosomal dominant limb-girdle muscular dystrophy type 1A
75381	Autosomal dominant cystoid macular edema	276580	Autosomal dominant hyperinsulinism due to Kir6.2 deficiency	264	Autosomal dominant limb-girdle muscular dystrophy type 1B
79499	Autosomal dominant deafness-onychodystrophy syndrome	276575	Autosomal dominant hyperinsulinism due to SUR1 deficiency	265	Autosomal dominant limb-girdle muscular dystrophy type 1C
2337	Autosomal dominant diffuse palmoplantar keratoderma, Norrbotten type	428	Autosomal dominant hypocalcemia	34516	Autosomal dominant limb-girdle muscular dystrophy type 1D
139518	Autosomal dominant distal juvenile spinal muscular atrophy type 1	1810	Autosomal dominant hypohidrotic ectodermal dysplasia	34517	Autosomal dominant limb-girdle muscular dystrophy type 1E
93608	Autosomal dominant distal renal tubular acidosis	89937	Autosomal dominant hypophosphatemia	55595	Autosomal dominant limb-girdle muscular dystrophy type 1F
98808	Autosomal dominant dopa-responsive dystonia	89937	Autosomal dominant hypophosphatemic rickets	55596	Autosomal dominant limb-girdle muscular dystrophy type 1G
→23156 8	Autosomal dominant dystrophic epidermolysis bullosa, Cockayne-Touraine type	100043	Autosomal dominant intermediate Charcot-Marie-Tooth disease type A	238755	Autosomal dominant limb-girdle muscular dystrophy type 1H
231568	Autosomal dominant dystrophic epidermolysis bullosa, Pasini and Cockayne-Touraine types	100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	140957	Autosomal dominant macrothrombocytopenia
→23156 8	Autosomal dominant dystrophic epidermolysis bullosa, Pasini type	100045	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C	88950	Autosomal dominant medullary cystic kidney disease with hyperuricemia
300576	Autosomal dominant ectodermal dysplasia-cancer predisposition syndrome syndrome	100046	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	34149	Autosomal dominant medullary cystic kidney disease with or without hyperuricemia
98853	Autosomal dominant Emery-Dreifuss muscular dystrophy	93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	88949	Autosomal dominant medullary cystic kidney disease without hyperuricemia
101046	Autosomal dominant epilepsy with auditory features	352670	Autosomal dominant intermediate Charcot-Marie-Tooth disease type F	319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency
73229	Autosomal dominant familial hematuria - retinal arteriolar tortuosity - contractures	324585	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency
100988	Autosomal dominant familial spastic paraplegia type 3	90635	Autosomal dominant isolated neurosensory deafness type DFNA	319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 1 deficiency
329466	Autosomal dominant focal dystonia, DYT25	90635	Autosomal dominant isolated neurosensory hearing loss type DFNA		
		90635	Autosomal dominant isolated sensorineural deafness type DFNA		

→ 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
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 2 deficiency	67036	Autosomal dominant optic atrophy type 3	100988	Autosomal dominant spastic paraplegia type 6
330041	Autosomal dominant methemoglobinemia	98673	Autosomal dominant optic atrophy, classic form	100989	Autosomal dominant spastic paraplegia type 8
2514	Autosomal dominant microcephaly	98673	Autosomal dominant optic atrophy, Kjer type	100990	Autosomal dominant spastic paraplegia type 9
319581	Autosomal dominant MSMD due to partial IFNgammaR1 deficiency	2783	Autosomal dominant osteopetrosis type 1	100991	Autosomal dominant spastic paraplegia type 10
319589	Autosomal dominant MSMD due to partial IFNgammaR2 deficiency	1798	Autosomal dominant osteosclerosis, Stanescu type	100993	Autosomal dominant spastic paraplegia type 12
319581	Autosomal dominant MSMD due to partial interferon gamma receptor 1 deficiency	2790	Autosomal dominant osteosclerosis, Worth type	100994	Autosomal dominant spastic paraplegia type 13
319589	Autosomal dominant MSMD due to partial interferon gamma receptor 2 deficiency	1010	Autosomal dominant palmoplantar hyperkeratosis and congenital alopecia	100998	Autosomal dominant spastic paraplegia type 17
65743	Autosomal dominant multiple pterygium syndrome	1010	Autosomal dominant palmoplantar keratoderma and congenital alopecia	100999	Autosomal dominant spastic paraplegia type 19
99846	Autosomal dominant myoglobinuria	88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	101009	Autosomal dominant spastic paraplegia type 29
440354	Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome	2924	Autosomal dominant polycystic liver disease	101011	Autosomal dominant spastic paraplegia type 31
79153	Autosomal dominant nail dysplasia	1300	Autosomal dominant popliteal pterygium syndrome	320365	Autosomal dominant spastic paraplegia type 36
329211	Autosomal dominant neovascular inflammatory vitreoretinopathy	34528	Autosomal dominant primary hypomagnesemia with hypocalciuria	171612	Autosomal dominant spastic paraplegia type 37
98784	Autosomal dominant nocturnal frontal lobe epilepsy	2964	Autosomal dominant prognathism	171617	Autosomal dominant spastic paraplegia type 38
178469	Autosomal dominant non-syndromic intellectual disability	254892	Autosomal dominant progressive external ophthalmoplegia	320355	Autosomal dominant spastic paraplegia type 41
90635	Autosomal dominant non-syndromic neurosensory deafness type DFNA	88659	Autosomal dominant progressive nephropathy with hypertension	171863	Autosomal dominant spastic paraplegia type 42
90635	Autosomal dominant non-syndromic neurosensory hearing loss type DFNA	314889	Autosomal dominant proximal renal tubular acidosis	444099	Autosomal dominant spastic paraplegia type 73
90635	Autosomal dominant non-syndromic sensorineural deafness type DFNA	171871	Autosomal dominant pseudohypoaldosteronism type 1	1797	Autosomal dominant spondylocostal dysostosis
90635	Autosomal dominant non-syndromic sensorineural hearing loss type DFNA	209867	Autosomal dominant rhegmatogenous retinal detachment	1797	Autosomal dominant spondylocostal dysplasia
90635	Autosomal dominant non-syndromic sensorineural hearing loss type DFNA	3107	Autosomal dominant Robinow syndrome	228169	Autosomal dominant striatal neurodegeneration
93328	Autosomal dominant omodysplasia	247511	Autosomal dominant secondary erythrocytosis	98757	Autosomal dominant striatonigral degeneration
306588	Autosomal dominant Opitz BBB/G syndrome	247511	Autosomal dominant secondary polycythemia	3357	Autosomal dominant trichodontoonychodyplasia-syndactyly
306588	Autosomal dominant Opitz G/BBB syndrome	98808	Autosomal dominant Segawa syndrome	3086	Autosomal dominant vitreoretinochoroidopathy
306588	Autosomal dominant Opitz syndrome	486	Autosomal dominant severe congenital neutropenia	79278	Autosomal erythropoietic protoporphyria
67036	Autosomal dominant optic atrophy and cataract	140481	Autosomal dominant slowed nerve conduction velocity	88919	Autosomal recessive Alport syndrome
→1215	Autosomal dominant optic atrophy and congenital deafness	251282	Autosomal dominant spastic ataxia type 1	1027	Autosomal recessive amelia
255117	Autosomal dominant optic atrophy and late-onset deafness	100984	Autosomal dominant spastic paraplegia type 3	248	Autosomal recessive anhidrotic ectodermal dysplasia
250932	Autosomal dominant optic atrophy and peripheral neuropathy	100985	Autosomal dominant spastic paraplegia type 4	1116	Autosomal recessive aplasia cutis
1215	Autosomal dominant optic atrophy plus syndrome			139485	Autosomal recessive ataxia due to coenzyme Q10 deficiency
				247815	Autosomal recessive ataxia due to PEX10 deficiency

→ 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
139485	Autosomal recessive ataxia due to ubiquinone deficiency	363429	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome	90349	Autosomal recessive cutis laxa, pulmonary emphysema type
88644	Autosomal recessive ataxia, Beauce type	1170	Autosomal recessive cerebelloparenchymal disorder type 3	79500	Autosomal recessive deafness-onychodystrophy syndrome
101101	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2B2	363969	Autosomal recessive cerebral atrophy	2776	Autosomal recessive distal osteolysis syndrome
101097	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2K	324442	Autosomal recessive Charcot-Marie-Tooth disease type 2 with neuromyotonia	402041	Autosomal recessive distal renal tubular acidosis
443950	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2T	98856	Autosomal recessive Charcot-Marie-Tooth disease type 2B1	→40204	Autosomal recessive distal renal tubular acidosis with deafness
90119	Autosomal recessive axonal Charcot-Marie-Tooth disease with acrodystrophy	101097	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	→40204	Autosomal recessive distal renal tubular acidosis with hearing loss
98856	Autosomal recessive axonal CMT4C1	90118	Autosomal recessive Charcot-Marie-Tooth disease, Ouvrier type	→40204	Autosomal recessive distal renal tubular acidosis without deafness
101102	Autosomal recessive axonal CMT4C2	90119	Autosomal recessive Charcot-Marie-Tooth type 2 with acrodystrophy	→40204	Autosomal recessive distal renal tubular acidosis without hearing loss
101101	Autosomal recessive axonal CMT4C3	293603	Autosomal recessive CHED	402041	Autosomal recessive distal RTA
101097	Autosomal recessive axonal CMT4C4	217046	Autosomal recessive childhood-onset cortical cataract	→40204	Autosomal recessive distal RTA with deafness
324442	Autosomal recessive axonal neuropathy with neuromyotonia	2518	Autosomal recessive chorioretinopathy-microcephaly syndrome	98920	Autosomal recessive distal spinal muscular atrophy type 1
139455	Autosomal recessive bestrophinopathy	447760	Autosomal recessive complex spastic paraparesis type 9B	139552	Autosomal recessive distal spinal muscular atrophy type 2
448242	Autosomal recessive brachyolmia, Hobaek/Toledo type	363432	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency	139547	Autosomal recessive distal spinal muscular atrophy type 3
→3460	Autosomal recessive carpotarsal osteolysis	363432	Autosomal recessive congenital cerebellar ataxia due to ionotropic glutamate receptor delta-2 subunit deficiency	206580	Autosomal recessive distal spinal muscular atrophy type 4
169186	Autosomal recessive centronuclear myopathy	324262	Autosomal recessive congenital cerebellar ataxia due to metabotropic glutamate receptor 1 deficiency	314485	Autosomal recessive distal spinal muscular atrophy type 5
95433	Autosomal recessive cerebellar ataxia - blindness - deafness	324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency	101150	Autosomal recessive dopa-responsive dystonia
352403	Autosomal recessive cerebellar ataxia - cognitive defect	293603	Autosomal recessive congenital hereditary endothelial dystrophy	79408	Autosomal recessive dystrophic epidermolysis bullosa generalisata gravis
284271	Autosomal recessive cerebellar ataxia - psychomotor retardation	99951	Autosomal recessive congenital hypomyelinating neuropathy	89842	Autosomal recessive dystrophic epidermolysis bullosa generalisata mitis
95434	Autosomal recessive cerebellar ataxia - saccadic intrusion	90349	Autosomal recessive cutis laxa type 1	89842	Autosomal recessive dystrophic epidermolysis bullosa, generalized other
352641	Autosomal recessive cerebellar ataxia due to GBA2 deficiency	357074	Autosomal recessive cutis laxa type 2, classic type	79408	Autosomal recessive dystrophic epidermolysis bullosa, Hallopeau-Siemens type
412057	Autosomal recessive cerebellar ataxia due to STUB1 deficiency	357074	Autosomal recessive cutis laxa type 2, Debré type	238569	Autosomal recessive early-onset IBD
88644	Autosomal recessive cerebellar ataxia type 1	357064	Autosomal recessive cutis laxa type 2, progeroid type	238569	Autosomal recessive early-onset inflammatory bowel disease
139485	Autosomal recessive cerebellar ataxia type 2	357058	Autosomal recessive cutis laxa type 2A	98855	Autosomal recessive Emery-Dreifuss muscular dystrophy
352641	Autosomal recessive cerebellar ataxia with late-onset spasticity	357064	Autosomal recessive cutis laxa type 2B	289586	Autosomal recessive exfoliative ichthyosis
404499	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to KIAA0226 deficiency	90349	Autosomal recessive cutis laxa with severe systemic involvement	1974	Autosomal recessive facio-digito-genital syndrome
404493	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency			329329	Autosomal recessive frontotemporal pachygyria
284282	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency			331226	Autosomal recessive hyper-IgE syndrome due to TYK2 deficiency

→ 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
79644	Autosomal recessive hyperinsulinemic hypoglycemia due to Kir6.2 deficiency	267	Autosomal recessive limb-girdle muscular dystrophy type 2A	319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 1 deficiency
79643	Autosomal recessive hyperinsulinemic hypoglycemia due to SUR1 deficiency	268	Autosomal recessive limb-girdle muscular dystrophy type 2B	319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 2 deficiency
79644	Autosomal recessive hyperinsulinism due to Kir6.2 deficiency	353	Autosomal recessive limb-girdle muscular dystrophy type 2C	175	Autosomal recessive metaphyseal chondrodysplasia
79643	Autosomal recessive hyperinsulinism due to SUR1 deficiency	62	Autosomal recessive limb-girdle muscular dystrophy type 2D	319569	Autosomal recessive MSMD due to partial IFNgammaR1 deficiency
248	Autosomal recessive hypohidrotic ectodermal dysplasia	119	Autosomal recessive limb-girdle muscular dystrophy type 2E	319574	Autosomal recessive MSMD due to partial IFNgammaR2 deficiency
289176	Autosomal recessive hypophosphatemic rickets	219	Autosomal recessive limb-girdle muscular dystrophy type 2F	319569	Autosomal recessive MSMD due to partial interferon gamma receptor 1 deficiency
300547	Autosomal recessive infantile hypercalcemia	34514	Autosomal recessive limb-girdle muscular dystrophy type 2G	319574	Autosomal recessive MSMD due to partial interferon gamma receptor 2 deficiency
93591	Autosomal recessive infantile nephronophthisis	1878	Autosomal recessive limb-girdle muscular dystrophy type 2H	319569	Autosomal recessive multiple epiphyseal dysplasia
93591	Autosomal recessive infantile NPHP	34515	Autosomal recessive limb-girdle muscular dystrophy type 2I	2990	Autosomal recessive multiple pterygium syndrome
352530	Autosomal recessive intellectual disability due to TRAPPC9 deficiency	140922	Autosomal recessive limb-girdle muscular dystrophy type 2J	424261	Autosomal recessive muscular dystrophy due to LAP1B deficiency
217055	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	86812	Autosomal recessive limb-girdle muscular dystrophy type 2K	424261	Autosomal recessive muscular dystrophy due to Torsin-1A-interacting protein 1 deficiency
254334	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B	206549	Autosomal recessive limb-girdle muscular dystrophy type 2L	319332	Autosomal recessive myogenic AMC
369867	Autosomal recessive intermediate Charcot-Marie-Tooth disease type C	206554	Autosomal recessive limb-girdle muscular dystrophy type 2M	319332	Autosomal recessive myogenic arthrogryposis multiplex congenita
435998	Autosomal recessive intermediate Charcot-Marie-Tooth disease type D	206559	Autosomal recessive limb-girdle muscular dystrophy type 2N	280654	Autosomal recessive nail dysplasia
210110	Autosomal recessive intermediate osteopetrosis	206564	Autosomal recessive limb-girdle muscular dystrophy type 2O	2990	Autosomal recessive non-lethal multiple pterygium syndrome
90636	Autosomal recessive isolated neurosensory deafness type DFNB	280333	Autosomal recessive limb-girdle muscular dystrophy type 2P	88616	Autosomal recessive non-syndromic intellectual disability
98676	Autosomal recessive isolated optic atrophy	254361	Autosomal recessive limb-girdle muscular dystrophy type 2Q	90636	Autosomal recessive non-syndromic neurosensory deafness type DFNB
90636	Autosomal recessive isolated sensorineural deafness type DFNB	363543	Autosomal recessive limb-girdle muscular dystrophy type 2R	90636	Autosomal recessive non-syndromic sensorineural deafness type DFNB
93324	Autosomal recessive Kenny-Caffey syndrome	369840	Autosomal recessive limb-girdle muscular dystrophy type 2S	93329	Autosomal recessive omodyplasia
263463	Autosomal recessive Larsen syndrome	363623	Autosomal recessive limb-girdle muscular dystrophy type 2T	67047	Autosomal recessive optic atrophy plus syndrome
1842	Autosomal recessive lethal chondrodysplasia, round femoral inferior epiphysis type	352479	Autosomal recessive limb-girdle muscular dystrophy type 2U	67047	Autosomal recessive optic atrophy type 3
33108	Autosomal recessive lethal multiple pterygium syndrome	206580	Autosomal recessive lower motor neuron disease with childhood onset	99012	Autosomal recessive optic atrophy, OPA6 type
314572	Autosomal recessive leukoencephalopathy with ischemic stroke-retinitis pigmentosa syndrome	238505	Autosomal recessive lymphoproliferative disease	227976	Autosomal recessive optic atrophy, OPA7 type
363543	Autosomal recessive limb-girdle muscular dystrophy due to desmin deficiency	667	Autosomal recessive malignant osteopetrosis	441344	Autosomal recessive optic atrophy, OPA8 type
352479	Autosomal recessive limb-girdle muscular dystrophy due to ISPD deficiency	319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	178389	Autosomal recessive osteoclast-poor osteopetrosis with hypogammaglobulinemia
254361	Autosomal recessive limb-girdle muscular dystrophy due to plectin deficiency	319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency		

→ 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
178389	Autosomal recessive osteopetrosis type 7	423384	Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency	320401	Autosomal recessive spastic paraplegia type 44
1366	Autosomal recessive palmoplantar hyperkeratosis and congenital alopecia	260305	Autosomal recessive sideroblastic anemia	320396	Autosomal recessive spastic paraplegia type 45
1366	Autosomal recessive palmoplantar keratoderma and congenital alopecia	300345	Autosomal recessive SLE	320391	Autosomal recessive spastic paraplegia type 46
731	Autosomal recessive polycystic kidney disease	254343	Autosomal recessive spastic ataxia - optic atrophy - dysarthria	306511	Autosomal recessive spastic paraplegia type 48
1234	Autosomal recessive popliteal pterygium syndrome	98	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	320385	Autosomal recessive spastic paraplegia type 49
88628	Autosomal recessive posterior column ataxia and retinitis pigmentosa	314603	Autosomal recessive spastic ataxia type 3	319199	Autosomal recessive spastic paraplegia type 53
437552	Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity	254343	Autosomal recessive spastic ataxia type 4	320380	Autosomal recessive spastic paraplegia type 54
437552	Autosomal recessive primary immunodeficiency with defective spontaneous NK cell cytotoxicity	313772	Autosomal recessive spastic ataxia type 5	320375	Autosomal recessive spastic paraplegia type 55
2512	Autosomal recessive primary microcephaly	314603	Autosomal recessive spastic ataxia with leukoencephalopathy	320411	Autosomal recessive spastic paraplegia type 56
254886	Autosomal recessive progressive external ophthalmoplegia	101005	Autosomal recessive spastic paraplegia - disc herniation	431329	Autosomal recessive spastic paraplegia type 57
93607	Autosomal recessive proximal renal tubular acidosis	100986	Autosomal recessive spastic paraplegia type 5A	397946	Autosomal recessive spastic paraplegia type 58
171876	Autosomal recessive pseudohypoaldosteronism type 1	2822	Autosomal recessive spastic paraplegia type 11	401795	Autosomal recessive spastic paraplegia type 59
1507	Autosomal recessive Robinow syndrome	100995	Autosomal recessive spastic paraplegia type 14	401800	Autosomal recessive spastic paraplegia type 60
247378	Autosomal recessive secondary erythrocytosis not associated with VHL gene	100996	Autosomal recessive spastic paraplegia type 15	401780	Autosomal recessive spastic paraplegia type 61
247378	Autosomal recessive secondary erythrocytosis, non-Chuvash type	209951	Autosomal recessive spastic paraplegia type 18	401785	Autosomal recessive spastic paraplegia type 62
247378	Autosomal recessive secondary polycythemia not associated with VHL gene	101000	Autosomal recessive spastic paraplegia type 20	401805	Autosomal recessive spastic paraplegia type 63
247378	Autosomal recessive secondary polycythemia, non-Chuvash type	101001	Autosomal recessive spastic paraplegia type 21	401810	Autosomal recessive spastic paraplegia type 64
101150	Autosomal recessive Segawa syndrome	101003	Autosomal recessive spastic paraplegia type 23	320396	Autosomal recessive spastic paraplegia type 65
970	Autosomal recessive sensory radicular neuropathy	101004	Autosomal recessive spastic paraplegia type 24	401815	Autosomal recessive spastic paraplegia type 66
70594	Autosomal recessive sepiapterin reductase-deficient DRD	101005	Autosomal recessive spastic paraplegia type 25	401820	Autosomal recessive spastic paraplegia type 67
420702	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency	101006	Autosomal recessive spastic paraplegia type 26	401825	Autosomal recessive spastic paraplegia type 68
420699	Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency	101007	Autosomal recessive spastic paraplegia type 27	401830	Autosomal recessive spastic paraplegia type 69
331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	101008	Autosomal recessive spastic paraplegia type 28	401835	Autosomal recessive spastic paraplegia type 70
		101010	Autosomal recessive spastic paraplegia type 30	401840	Autosomal recessive spastic paraplegia type 71
		171622	Autosomal recessive spastic paraplegia type 32	98920	Autosomal recessive spinal muscular atrophy with respiratory distress
		171629	Autosomal recessive spastic paraplegia type 35	284332	Autosomal recessive spinocerebellar ataxia type 6
		139480	Autosomal recessive spastic paraplegia type 39	284324	Autosomal recessive spinocerebellar ataxia type 7
		320370	Autosomal recessive spastic paraplegia type 43	139485	Autosomal recessive spinocerebellar ataxia type 9

→ 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
284289	Autosomal recessive spinocerebellar ataxia type 10	284454	AZOOR	1229	Baraitser-Brett-Piesowicz syndrome
284271	Autosomal recessive spinocerebellar ataxia type 11	3471	Azoospermia - sinopulmonary infections	2753	Baraitser-Burn syndrome
284282	Autosomal recessive spinocerebellar ataxia type 12	→399805	Azoospermia due to maturation arrest	1229	Baraitser-Reardon syndrome
324262	Autosomal recessive spinocerebellar ataxia type 13	→399805	Azoospermia due to meiosis defect	2995	Baraitser-Winter syndrome
352403	Autosomal recessive spinocerebellar ataxia type 14	98757	Azorean disease of the nervous system	2237	Barakat syndrome
404499	Autosomal recessive spinocerebellar ataxia type 15	99121	Azygos continuation of the inferior caval vein	1231	Barber-Say syndrome
397709	Autosomal recessive spinocerebellar ataxia type 20	99121	Azygos continuation of the inferior vena cava	110	Bardet-Biedl syndrome
2311	Autosomal recessive spondylocostal dysostosis	79332	B4GALT1-CDG	34592	Bare lymphocyte syndrome type 1
401979	Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type	75496	B4GALT7-CDG	572	Bare lymphocyte syndrome type 2
250984	Autosomal recessive Stickler syndrome	99860	B-ALL	3317	Barnes syndrome
300345	Autosomal recessive systemic lupus erythematosus	67038	B-cell chronic lymphocytic leukemia	443084	Baroreflex failure
280365	Autosomal semi-dominant severe lipodystrophic laminopathy	67038	B-cell chronic lymphoid leukemia	79087	Barraquer-Simons syndrome
401849	Autosomal spastic paraplegia type 72	86852	B-cell prolymphocytic leukemia	2698	Bart-Pumphrey syndrome
168629	Autosomal thrombocytopenia with normal platelets	67038	B-CLL	111	Barth syndrome
352490	AUTS2 syndrome	404560	B-K mole syndrome	64692	Bartonellosis due to Bartonella bacilliformis infection
96	AVED	86852	B-PLL	50839	Bartonellosis due to Bartonella henselae infection
98963	Avellino corneal dystrophy	108	Babesiosis	64694	Bartonellosis due to Bartonella quintana infection
99000	AVMD	206994	Bacterial myositis	1234	Bartsocas-Papas syndrome
58	AxD	36234	Bacterial toxic-shock syndrome	112	Bartter syndrome
363717	AxD type I	36234	Bacterial TSS	93605	Bartter syndrome type 3
363722	AxD type II	86814	BAFME	89938	Bartter syndrome type 4
98978	Axenfeld anomaly	2819	Bahemuka-Brown syndrome	263417	Bartter syndrome type 5
782	Axenfeld syndrome	352577	Bainbridge-Ropers syndrome	93605	Bartter syndrome type III
782	Axenfeld-Rieger syndrome	1658	Baird syndrome	89938	Bartter syndrome type IV
1834	Axial mesodermal dysplasia spectrum	139471	Bakrania-Ragge syndrome	263417	Bartter syndrome type V
2777	Axial osteosclerosis	1223	Balantidiasis	263417	Bartter syndrome with hypocalcemia
168549	Axial spondylometaphyseal dysplasia	1223	Balantidiosis	93604	Bartter syndrome, furosemide type
401911	AXIN2-related AFAP	139450	Balikova-Vermeesch syndrome	93604	Bartter syndrome, furosemide-amiloride type
401911	AXIN2-related attenuated familial adenomatous polyposis	363746	Balint syndrome	377	Basal cell nevus syndrome
401911	AXIN2-related attenuated familial polyposis coli	363746	Balint-Holmes syndrome	268829	Basal encephalocele
401911	AXIN2-related attenuated FAP	93395	Ballard syndrome	→1658	Basan syndrome
101102	Axonal Charcot-Marie-Tooth disease with pyramidal involvement	1225	Baller-Gerold syndrome	50810	Basel-Vanagaite-Sirota syndrome
209004	Axonal polyneuropathy associated with IgG/IgM/IgA monoclonal gammopathy	66529	Ballooning cardiomyopathy	244283	BASM syndrome
1435	Ayazi syndrome	228165	Baló concentric sclerosis	14	Bassen-Kornzweig disease
413687	Azathioprine or 6-mercaptopurine toxicity or dose selection	634	Bamboo hair syndrome	1875	Bassoe syndrome
		1226	Bamforth syndrome	100976	Bathing suit ichthyosis
		1226	Bamforth-Lazarus syndrome	1948	Battaglia-Neri syndrome
		98955	Band-shaped and whorled microcystic dystrophy of the corneal epithelium	79264	Batten disease
		1227	Bangstad syndrome	→1071	Baughman syndrome
		130	Bangungut	166113	Bazex syndrome
		1228	Banki syndrome	113	Bazex-Dupré-Christol syndrome
		109	Bannayan-Riley-Ruvalcaba syndrome	65284	BBGD
		139507	Bantu siderosis	110	BBS
		289539	BAP1-related tumor predisposition syndrome	41751	BCD
				1997	BCD syndrome
				312	BCIE
				511	BCKD deficiency
				511	BCKDH deficiency
				1236	Bd 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
247203	BDC	2841	Benign chronic familial pemphigus of Hailey-Hailey	65682	Benign recurrent intrahepatic cholestasis
113	BDCS	251287	Benign concentric annular macular dystrophy	99960	Benign recurrent intrahepatic cholestasis type 1
115	Beals syndrome	440233	Benign congenital sixth cranial nerve palsy	99961	Benign recurrent intrahepatic cholestasis type 2
115	Beals-Hecht syndrome	254864	Benign COX deficiency	342	Benign recurrent polyserositis
1059	Bean syndrome	180284	Benign ductal tumor of breast	1945	Benign rolandic epilepsy
1555	Beare-Stevenson cutis gyrata syndrome	1945	Benign epilepsy of childhood with centrotemporal spikes	324581	Benign Samaritan congenital myopathy
98895	Becker dystrophinopathy	276148	Benign epithelial tumor of salivary glands	252164	Benign schwannoma
98895	Becker muscular dystrophy	71269	Benign exophthalmos syndrome	180237	Benign tumor of fallopian tubes
64755	Becker nevus syndrome	1429	Benign familial chorea	2198	Bennion-Patterson syndrome
116	Beckwith-Wiedemann syndrome	1945	Benign familial epilepsy of childhood with rolandic spikes	54247	Benson syndrome
231127	Beckwith-Wiedemann syndrome due to 11p15 microdeletion	306	Benign familial infantile convulsions	528	Berardinelli-Seip syndrome
96076	Beckwith-Wiedemann syndrome due to 11p15 microduplication	306	Benign familial infantile epilepsy	171839	Berant syndrome
231130	Beckwith-Wiedemann syndrome due to 11p15 translocation/inversion	306	Benign familial infantile seizures	528	Berardinelli-Seip congenital lipodystrophy
231120	Beckwith-Wiedemann syndrome due to CDKN1C mutation	163717	Benign familial mesial temporal lobe epilepsy	2241	Berdon syndrome
231117	Beckwith-Wiedemann syndrome due to imprinting defect of 11p15	1949	Benign familial neonatal convulsions	647	Berlin breakage syndrome
238613	Beckwith-Wiedemann syndrome due to NSD1 mutation	1949	Benign familial neonatal epilepsy	274	Bernard-Soulier syndrome
96193	Beckwith-Wiedemann syndrome due to paternal uniparental disomy of chromosome 11	1949	Benign familial neonatal seizures	178528	Berti lymphoma
1945	BECRS	140927	Benign familial neonatal-infantile seizures	133	Berylliosis
1945	BECTS	209973	Benign familial nocturnal alternating hemiplegia in childhood	71269	BES
2572	Bedouin spastic ataxia syndrome	209973	Benign familial nocturnal alternating hemiplegia of childhood	797	Besnier-Boeck-Schaumann disease
322	BEEC	65684	Benign focal amyotrophy	321	Bessel-Hagen disease
1237	Beemer-Ertbruggen syndrome	1544	Benign focal seizures of adolescence	1243	Best disease
275864	Behavioral variant of frontotemporal dementia	64545	Benign idiopathic neonatal seizures	1243	Best macular dystrophy
1239	Behr syndrome	166308	Benign infantile focal epilepsy with midline spikes and wave during sleep	1243	Best vitelliform macular dystrophy
2705	Behrens-Baumann-Vogel syndrome	166305	Benign infantile seizures associated to mild gastroenteritis	324718	Beta amyloidosis, Flemish type
117	Behçet disease	238624	Benign intracranial hypertension	324703	Beta amyloidosis, Piedmont type
2810	Bell palsy	285	Benign joint hypermobility syndrome	79332	Beta-1,4-galactosyltransferase deficiency
247203	Bellini carcinoma	168816	Benign multicystic peritoneal mesothelioma	65287	Beta-alanine synthase deficiency
247203	Bellini duct carcinoma	86909	Benign myoclonic epilepsy of infancy	309310	Beta-D-galactosidase deficiency
1240	Bellini syndrome	86909	Benign myoclonus epilepsy of infancy	354	Beta-galactosidase-1 deficiency
1492	Ben Ari-Shuper-Mimouni syndrome	140927	Benign neonatal-infantile epilepsy	584	Beta-glucuronidase deficiency
100978	Benallegue-Lacete syndrome	25968	Benign occipital epilepsy	134	Beta-ketothiolase deficiency
1241	Bencze syndrome	342	Benign paroxysmal peritonitis	118	Beta-mannosidase deficiency
86814	Benign adult familial myoclonic epilepsy	1179	Benign paroxysmal tonic upgaze of childhood with ataxia	118	Beta-mannosidosis
86814	Benign adult familial myoclonus epilepsy	71518	Benign paroxysmal torticollis of infancy	329284	Beta-propeller protein-associated neurodegeneration
610	Benign autosomal dominant myopathy	166299	Benign partial epilepsy of infancy with complex partial seizures	119	Beta-sarcoglycanopathy
157997	Benign cephalic histiocytosis	166302	Benign partial epilepsy with secondarily generalized seizures in infancy	848	Beta-thalassemia
98816	Benign childhood occipital epilepsy, Gastaut type			→33364	Beta-thalassemia - trichothiodystrophy
98815	Benign childhood occipital epilepsy, Panayiotopoulos type			231393	Beta-thalassemia - X-linked thrombocytopenia

→ 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
2114	BFHD	101070	Bilateral frontoparietal polymicrogyria	54247	Biparietal Alzheimer disease
306	BFIE	208447	Bilateral generalized polymicrogyria	364198	Bipartite talus
306	BFIS	438117	Bilateral hip and radial head dislocations-short stature-scoliosis-carpal coalitions-pes cavus-facial dysmorphism syndrome	99908	Bird fancier lung
127	BFLS			2617	Bird headed-dwarfism, Montreal type
140927	BNFIS	319205	Bilateral massive adrenal hemorrhage	179	Birdshot chorioretinitis
1949	BFNS	97364	Bilateral MCDK	179	Birdshot chorioretinopathy
293284	BH4-responsive HPA/PKU	140963	Bilateral microtia - deafness - cleft palate	179	Birdshot retinochoroiditis
293284	BH4-responsive hyperphenylalaninemia/phenylketonuria	97364	Bilateral multicystic dysplastic kidney	179	Birdshot retinochoroidopathy
98964	Biber-Haab-Dimmer dystrophy	97364	Bilateral multicystic renal dysplasia	122	Birt-Hogg-Dubé syndrome
180086	Bicervical bicornuate uterus	208441	Bilateral parasagittal parieto-occipital polymicrogyria	79133	Bitemporal aplasia cutis congenita
180106	Bicervical bicornuate uterus and blind hemivagina	98889	Bilateral perisylvian polymicrogyria	2213	Bixler-Christian-Gorlin syndrome
180106	Bicervical bicornuate uterus one-eyed hemi-vagina	268940	Bilateral polymicrogyria	285	BJHS
180111	Bicervical bicornuate uterus with patent cervix and vagina	295150	Bilateral PPD2	123	Björnstad syndrome
2088	Bickel-Fanconi glycogenosis	1980	Bilateral striopallidodentate calcinosis	124	Blackfan-Diamond anemia
2182	Bickers-Adams syndrome	276066	Bile acid CoA ligase deficiency and defective amidation	93930	Bladder exstrophy
79138	Bickerstaff brainstem encephalitis	70567	Bile duct cancer	322	Bladder exstrophy-epispadias-cloacal extrophy complex
3286	Bidirectional tachycardia induced by catecholamine	1276	Bilginturan brachydactyly	37202	Bladder pain syndrome
→33364	BIDS syndrome	1276	Bilginturan syndrome	98922	Blake pouch cyst
1246	Biemond syndrome	1247	Bilharziasis	254379	Blaschkoid lichen planus
141333	Biemond syndrome type 2	30391	Biliary atresia	254379	Blaschkoid LP
41751	Bietti crystalline corneoretinal dystrophy	244283	Biliary atresia with splenic malformation syndrome	86870	Blastic NK-cell lymphoma
41751	Bietti crystalline dystrophy	424982	Biliary cystadenocarcinoma	86870	Blastic plasmacytoid dendritic cell neoplasm
41751	Bietti crystalline retinopathy	386	Biliary hamartoma	1834	Blastogenesis defect
1986	Bifid femur - monodactylous ectrodactyly	→2697	Biliary tract malformation - renal failure	90340	Blau syndrome
295006	Bifid great toes	98836	Bilineal acute leukemia	50945	BLC
295177	Bifid great toes, bilateral	415286	Bilirubin encephalopathy	1229	BLC-PMG
295175	Bifid great toes, unilateral	205	Bilirubin uridinediphosphate glucuronosyltransferase deficiency	73271	Bleeding diathesis due to a collagen receptor defect
295006	Bifid halluces	79234	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 1	98885	Bleeding diathesis due to glycoprotein VI deficiency
295177	Bifid halluces, bilateral	79235	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 2	98886	Bleeding diathesis due to integrin alpha2-beta1 deficiency
295175	Bifid halluces, unilateral	205	Bilirubin-UGT deficiency	220443	Bleeding diathesis due to thromboxane synthesis deficiency
295006	Bifid hallux	79234	Bilirubin-UGT deficiency type 1	420566	Bleeding disorder due to calcium- and DAG-regulated guanine exchange factor-1 deficiency
295177	Bifid hallux, bilateral	79235	Bilirubin-UGT deficiency type 2	420566	Bleeding disorder due to CalDAG-GEFI deficiency
295175	Bifid hallux, unilateral	1799	Billard-Toutain-Maheut syndrome	1997	Blepharo-cheilo-odontic syndrome
2695	Bifid nose	1248	Binder syndrome	1253	Blepharochalasis-double lip syndrome
217266	Bifid nose with or without anorectal and renal anomalies	3304	Bindewald-Ulmer-Müller syndrome	1997	Blepharocheilodontic syndrome
99771	Bifid uvula	1249	Binswanger disease	→2353	Blepharofacioskeletal syndrome
99771	Bifidity of the uvula	65284	Biotin-responsive basal ganglia disease	1252	Blepharonasofacial malformation syndrome
300	Bifunctional enzyme deficiency	65284	Biotin-thiamine-responsive basal ganglia disease	126	Blepharophimosis - epicanthus inversus - ptosis
319205	Bilateral adrenal hemorrhage	79241	Biotinidase deficiency	261559	Blepharophimosis - epicanthus inversus - ptosis due to 3q23 rearrangement
325124	Bilateral anorchia				
2048	Bilateral anterior opercular syndrome				
1229	Bilateral band-like calcification with polymicrogyria				
208444	Bilateral frontal polymicrogyria				

→ 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
261572	Blepharophimosis - epicanthus inversus - ptosis due to a point mutation	98989	Blue-dot cataract	69737	Bosley-Salih-Alorainy syndrome
261579	Blepharophimosis - epicanthus inversus - ptosis due to polyA expansion	319205	BMAH	2250	Bosma-Henkin-Christiansen syndrome
2057	Blepharophimosis - ptosis - esotropia - syndactyly - short stature	1243	BMD	85128	Bothnia retinal dystrophy
1256	Blepharophimosis - radioulnar synostosis	98895	BMD	128	Bothriocephalosis
1968	Blepharophimosis - telecanthus - microstomia	293725	BMRS type V	1267	Botulism
2728	Blepharophimosis syndrome, Ohdo type	293707	BMRS, Maat-Kievit-Brunner type	1180	Boucher-Neuhäuser syndrome
126	Blepharophimosis types 1 and 2	293707	BMRS, MKB type	805	Bourneville syndrome
261572	Blepharophimosis types 1 and 2 due to a point mutation	2728	BMRS, Ohdo type	83313	Boutonneuse fever
261579	Blepharophimosis types 1 and 2 due to polyA expansion	293725	BMRS, Verloes type	3331	Bowed tibiae - radial anomalies - osteopenia - fractures
329255	Blepharophimosis-intellectual disability syndrome due to UBE3B deficiency	217266	BNAR syndrome	→912	Bowen syndrome
293725	Blepharophimosis-intellectual disability syndrome type V	50945	BOCD	1270	Bowen syndrome, Hutterite type
293707	Blepharophimosis-intellectual disability syndrome, Maat-Kievit-Brunner type	217008	Bockenheimer syndrome	1270	Bowen-Conradi syndrome
293707	Blepharophimosis-intellectual disability syndrome, MKB type	1292	BOD syndrome	97353	Boxer's dementia
2728	Blepharophimosis-intellectual disability syndrome, Ohdo type	2724	Boder syndrome	50814	Bojadjiev-Jabs syndrome
3047	Blepharophimosis-intellectual disability syndrome, SBBYS type	48686	Body cavity-based lymphoma	2680	Boylan-Dew syndrome
293725	Blepharophimosis-intellectual disability syndrome, Verloes type	91135	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	329284	BPAN
1258	Blepharoptosis - cleft palate - ectrodactyly - dental anomalies	797	Boeck sarcoid	70589	BPD
1259	Blepharoptosis - myopia - ectopia lentis	797	Boeck's sarcoid	86870	BPDCN
93964	Blepharospasm - oromandibular dystonia	1297	BOFS	97342	Braak disease
171844	Blindness-scoliosis-arachnodactyly syndrome	97297	Bohring syndrome	2901	Brachial plexus neuritis
464	Bloch-Siemens syndrome	97297	Bohring-Opitz syndrome	199	Brachmann-de Lange syndrome
464	Bloch-Sulzberger syndrome	84081	Boichis disease	1519	Brachycephalofrontonasal dysplasia
50945	Blomstrand chondrodysplasia	401874	BOLA3 deficiency	1272	Brachycephaly - deafness - cataract - intellectual disability
50945	Blomstrand lethal chondrodysplasia	319229	Bolivian hemorrhagic fever	2619	Brachydactylyous dwarfism, Mseleni type
50945	Blomstrand osteochondrodysplasia	85182	Bone dysplasia - medullary fibrosarcoma	1275	Brachydactyly - elbow wrist dysplasia
125	Bloom syndrome	1844	Bone dysplasia, Azouz type	1275	Brachydactyly - joint dysplasia
2768	Blount disease	1842	Bone dysplasia, lethal Holmgren type	2946	Brachydactyly - long thumb
88629	Blue colour blindness	2050	Bone fragility - craniostenosis - proptosis - hydrocephalus	1277	Brachydactyly - mesomelia - intellectual disability - heart defects
16	Blue cone monochromacy	300284	Bone fragility-contractures-arterial rupture-deafness syndrome	1246	Brachydactyly - nystagmus - cerebellar ataxia
16	Blue cone monochromatism	2934	Bonneau syndrome	1278	Brachydactyly - preaxial hallux varus
94086	Blue diaper syndrome	163	Bonneau-Beaumont syndrome	2956	Brachydactyly - scoliosis - carpal fusion
1059	Blue rubber bleb nevus	2941	Bonnemann-Meinecke syndrome	294996	Brachydactyly of fingers
		1261	Bonnemann-Meinecke-Reich syndrome	295130	Brachydactyly of fingers, bilateral
		53719	Bonnet-Dechaume-Blanc syndrome	295128	Brachydactyly of fingers, unilateral
		1262	Böök syndrome	294998	Brachydactyly of toes
		1263	Boomerang dysplasia	295134	Brachydactyly of toes, bilateral
		1303	BOOP	295132	Brachydactyly of toes, unilateral
		1933	Booth-Haworth-Dilling syndrome	93388	Brachydactyly type A1
		107	BOR syndrome	93396	Brachydactyly type A2
		206473	Borderline epithelial tumor of ovary	93394	Brachydactyly type A4
		206473	Borderline ovarian epithelial tumor	93389	Brachydactyly type A5
		127	Borjeson-Forssman-Lehmann syndrome	93382	Brachydactyly type A6
		1264	Bork syndrome	93397	Brachydactyly type A7
		90001	Bornholm eye disease	93383	Brachydactyly type B
		36273	Borrmann gastric cancer type 4	140908	Brachydactyly type B2
		97297	BOS syndrome	93384	Brachydactyly type C
				93387	Brachydactyly type E

→ 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
1276	Brachydactyly type E, with short stature and hypertension	511	Branched-chain ketoaciduria	47	Bruton type agammaglobulinemia
93395	Brachydactyly types B and E combined	1296	Branchial dysplasia - intellectual disability - inguinal hernia	528	BSCL
93388	Brachydactyly, Farabee type	1297	Branchio-oculo-facial syndrome	79304	BSEP deficiency
2946	Brachydactyly, long thumb type	52429	Branchio-otic syndrome	1299	BSG syndrome
93396	Brachydactyly, Mohr-Wriedt type	1299	Branchio-skeleto-genital syndrome	100976	BSI
93397	Brachydactyly, Smorgasbord type	50815	Branchiogenic deafness syndrome	46489	BSLE
93394	Brachydactyly, Temtamy type	107	Branchiootorenal syndrome	1980	BSPDC
1276	Brachydactyly-arterial hypertension syndrome	79133	Brauer syndrome	125	BSyn
1001	Brachydactyly-intellectual disability	2669	Braun-Bayer syndrome	65284	BTBGD
391646	Brachydactyly-short stature-microcephaly syndrome	319239	Brazilian hemorrhagic fever	79241	BTD deficiency
3168	Brachydactyly-symphalangism syndrome	1945	BRE	111	BTHS
93409	Brachydactyly-syndactyly, Zhao type	85284	BRESEK syndrome	47	BTK-deficiency
93394	Brachymesophalangy II and V	85284	BRESHECK syndrome	2314	Buckley syndrome
1292	Brachymorphism - onychodysplasia - dysphalangism	65682	BRIC	131	Budd-Chiari syndrome
→44824 2	Brachyolmia type 1, Hobaek type	99960	BRIC1	36258	Buerger disease
→44824 2	Brachyolmia type 1, Toledo type	99961	BRIC2	2285	Bull-Nixon syndrome
93302	Brachyolmia type 2	99960	BRIC type 1	312	Bullous congenital ichthyosiform erythroderma
93304	Brachyolmia type 3	99961	BRIC type 2	312	Bullous congenital ichthyosiform erythroderma of Brock
93302	Brachyolmia, Maroteaux type	99990	Brill disease	280785	Bullous DCM
448242	Brachyolmia, recessive type	99990	Brill-Zinsser disease	280785	Bullous diffuse cutaneous mastocytosis
2899	Brachyolmia-amelogenesis imperfecta syndrome	666	Brittle bone disease	1867	Bullous dystrophy, macular type
79345	Brachytelephalangic chondrodysplasia punctata	90354	Brittle cornea syndrome	312	Bullous ichthyosis
1295	Brachytelephalangy - dysmorphism - Kallmann syndrome	→33364	Brittle hair - mental deficiency	36237	Bullous impetigo
441	Bradbury-Eggleston syndrome	→33364	Brittle hair syndrome, Sabinas type	33408	Bullous lichen planus
52047	Braddock syndrome	783	Broad thumb-hallux syndrome	703	Bullous pemphigoid
3323	Braddock-Carey syndrome	783	Broad thumbs-halluces syndrome	46489	Bullous systemic lupus erythematosus
1538	Braddock-Jones-Superneau syndrome	412	Broad-beta lipoproteinemia	→193	Buntinx-Lormans-Martin syndrome
75374	Bradyopsia	53347	Brody myopathy	98976	Buphthalmia
178506	Brain calcification, Rajab type	97287	Bronchial carcinoid tumor	98976	Buphthalmos
168598	Brain demyelination due to methionine adenosyltransferase deficiency	97287	Bronchial endocrine tumor	543	Burkitt lymphoma
352649	Brain dopamine-serotonin vesicular transport disease	97287	Bronchial neuroendocrine tumor	1200	Burn-McKeown syndrome
75389	Brain malformation - congenital heart disease - postaxial polydactyly	→3471	Bronchiectasis - oligospermia	800	Burton disease
36383	Brain small vessel disease with hemorrhage	1302	Bronchiolitis obliterans organizing pneumonia	800	Burton skeletal dysplasia
209905	Brain-lung-thyroid syndrome	1303	Bronchiolitis obliterans with obstructive pulmonary disease	800	Burton syndrome
255182	Branched chain alpha-ketoacid dehydrogenase complex deficiency	2357	Bronchogenic cyst	352763	Buschke scleredema
511	Branched-chain 2-ketoacid dehydrogenase deficiency	70589	Bronchopulmonary dysplasia	79501	Buschke-Fischer-Brauer syndrome
		1116	Bronspiegel-Zelnick syndrome	1306	Buschke-Ollendorff syndrome
		99829	Bronze John	99001	Butterfly-shaped pigment dystrophy
		79493	Brooke-Spiegler syndrome	1307	Buttiens-Fryns syndrome
		97229	Brown-Vialetto-van Laere syndrome	132	Butyrylcholinesterase deficiency
		109	BRRS	275864	bv-FTD
		2353	BRSS	1243	BVMD
		1304	Brucellosis	116	BWS
		2771	Bruck syndrome	79306	Byler disease
		130	Brugada syndrome	280133	C3 deficiency
		1305	Brunner-Winter syndrome	→32993 1	C3 deposition glomerulonephritis without proliferation
		391641	Brunner-Winter syndrome type 1	329931	C3 glomerulonephritis
		391646	Brunner-Winter syndrome type 2	329918	C3 glomerulopathy
		528	Brunzell syndrome		
		→528	Brunzell 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
401901	C9ORF72-related Huntington disease phenocopy	1320	Campnocormia	1171	CAPOS syndrome
401901	C9ORF72-related Huntington disease-like syndrome	1320	Campnocormism	171839	Capra-DeMarco syndrome
1308	C syndrome	376	Camptodactyly - cleft palate- clubfoot	71505	CAR syndrome
231242	C-beta-thalassemia	1321	Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia	199354	CARASIL
97297	C-like syndrome	1323	Camptodactyly - joint contractures - facial skeletal defects	147	Carbamoyl-phosphate synthetase 1 deficiency
401948	CA-VA deficiency	3447	Camptodactyly - overgrowth - unusual facies	147	Carbamoyl-phosphate synthetase 2 deficiency
85293	Cabezas syndrome	85164	Camptodactyly - tall stature - scoliosis - hearing loss	147	Carbamoyl-phosphate synthetase 1 deficiency
1309	Cacchi-Ricci disease	1325	Camptodactyly - taurinuria	1923	Carbamazole embryofetopathy
75377	CACD	295016	Camptodactyly of fingers	79318	Carbohydrate deficient glycoprotein syndrome type Ia
135	CACH syndrome	1327	Camptodactyly syndrome, Guadalajara type 1	79319	Carbohydrate deficient glycoprotein syndrome type Ib
2848	CACP syndrome	1326	Camptodactyly syndrome, Guadalajara type 2	79320	Carbohydrate deficient glycoprotein syndrome type Ic
159	CACT deficiency	2848	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	79321	Carbohydrate deficient glycoprotein syndrome type Id
56425	CAD	1766	CAMRQ syndrome	79322	Carbohydrate deficient glycoprotein syndrome type Ie
448010	CAD-CDG	141194	CAMS1	79323	Carbohydrate deficient glycoprotein syndrome type If
136	CADASIL	53719	CAMS2	79324	Carbohydrate deficient glycoprotein syndrome type Ig
369942	CADDs	141199	CAMS3	79325	Carbohydrate deficient glycoprotein syndrome type Ih
1578	CADH deficiency	3319	CAMT	79326	Carbohydrate deficient glycoprotein syndrome type Ii
1310	Caffey disease	1328	Camurati-Engelmann disease	79329	Carbohydrate deficient glycoprotein syndrome type IIa
436174	CAGSSS	3261	Canale-Smith syndrome	79330	Carbohydrate deficient glycoprotein syndrome type IIb
90791	CAH due to 3-beta-hydroxysteroid dehydrogenase deficiency	141	Canavan disease	79332	Carbohydrate deficient glycoprotein syndrome type IIId
90795	CAH due to 11-beta-hydroxylase deficiency	289385	Cancer diagnosed during pregnancy	79333	Carbohydrate deficient glycoprotein syndrome type IIe
90793	CAH due to 17-alpha-hydroxylase deficiency	180242	Cancer of fallopian tubes	238459	Carbohydrate deficient glycoprotein syndrome type IIIf
1375	CAHMR syndrome	71505	Cancer-associated retinopathy	263508	Carbohydrate deficient glycoprotein syndrome type IIg
435988	CAID syndrome	2700	Cancrum oris	95428	Carbohydrate deficient glycoprotein syndrome type IIh
99429	CAIS	325004	CANDLE syndrome	263487	Carbohydrate deficient glycoprotein syndrome type III
199260	Calcified aponeurotic fibroma	71279	CANOMAD syndrome	263501	Carbohydrate deficient glycoprotein syndrome type IIj
90290	Calcinosis - Raynaud phenomenon - esophageal involvement - sclerodactyly - telangiectasia	2233	Cantalamessa-Baldini-Ambrosi syndrome	86309	Carbohydrate deficient glycoprotein syndrome type Ij
280062	Calciphylaxis	1335	Cantrell deformity	79327	Carbohydrate deficient glycoprotein syndrome type Ik
280065	Calciphylaxis cutis	1335	Cantrell syndrome	79328	Carbohydrate deficient glycoprotein syndrome type Il
1416	Calcium pyrophosphate dihydrate crystal deposition disease	363705	Cantu craniofaciofrontodigital syndrome	91131	Carbohydrate deficient glycoprotein syndrome type Im
1408	Calderón-González-Cantu syndrome	171881	Cap disease		
228123	California disease	160148	Cap inflammatory polypsis		
83483	Californian encephalitis	171881	Cap myopathy		
85192	Calvarial doughnut lesions - bone fragility	160148	Cap polyposis		
→1466	CAMAK syndrome	85199	CAP syndrome		
3003	Camera syndrome	166260	Capdepont teeth		
2163	Camero-Lituania-Cohen syndrome	75327	CAPE dystrophy		
→1466	CAMFAK syndrome	75327	CAPED		
79395	Camisa disease	188	Capillary hyperpermeability syndrome		
83472	CAMOS syndrome	188	Capillary leak syndrome		
1318	Campomelia, Cumming type	79490	Capillary lymphangioma		
140	Campomelic dwarfism	79490	Capillary lymphatic malformation		
140	Campomelic dysplasia	137667	Capillary malformation - arteriovenous malformation		
1319	Camptobrachydactyly				

→ 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
244310	Carbohydrate deficient glycoprotein syndrome type Ia	111	Cardioskeletal myopathy with neutropenia and abnormal mitochondria	228308	Carnitine palmitoyl transferase II deficiency, neonatal form
263494	Carbohydrate deficient glycoprotein syndrome type Ia	111	Cardioskeletal myopathy-neutropenia syndrome	228305	Carnitine palmitoyl transferase II deficiency, severe infantile form
280071	Carbohydrate deficient glycoprotein syndrome type Ia	3238	Cardiospondylocarpofacial syndrome	157	Carnitine palmitoyltransferase deficiency type 2
300536	Carbohydrate deficient glycoprotein syndrome type Ia	2072	Cardiovascular Gaucher disease	157	Carnitine palmitoyltransferase II deficiency
329178	Carbohydrate deficient glycoprotein syndrome type Ia	1358	Carey-Fineman-Ziter syndrome	158	Carnitine transporter defect
370924	Carbohydrate deficient glycoprotein syndrome type Ia	79403	Carmi syndrome	158	Carnitine uptake deficiency
370927	Carbohydrate deficient glycoprotein syndrome type Ia	→29384 3	Carnevale syndrome	159	Carnitine-acylcarnitine translocase deficiency
448010	Carbohydrate deficient glycoprotein syndrome type IZ	2947	Carnevale-Hernández-del Castillo syndrome	1361	Carnosinase deficiency
306686	Carbon monoxide-induced parkinsonism	→29384 3	Carnevale-Krajewska-Fischetto syndrome	1361	Carnosinemia
2785	Carbonic anhydrase 2 deficiency	1359	Carney complex	53035	Caroli disease
213605	Carcinofibroma of the corpus uteri	319340	Carney complex variant	65759	Carpenter syndrome
100093	Carcinoid tumor and carcinoid syndrome	319340	Carney complex-trismus-pseudocamptodactyly syndrome	93973	Carpenter-Waziri syndrome
97289	Carcinoid tumor of thymus	97286	Carney dyad	2767	Carpotarsal osteochondromatosis
319308	Carcinoma associated with MITF/TFE translocation	1359	Carney syndrome	64692	Carrion disease
418945	Carcinoma of esophagus, salivary gland type	139411	Carney triad	175	Cartilage-hair hypoplasia
423781	Carcinoma of stomach, salivary gland type	97286	Carney-Stratakis dyad	→175	Cartilage-hair hypoplasia-like - skeletal dysplasia without hypotrichosis
300557	Carcinoma of the ampulla of Vater	97286	Carney-Stratakis syndrome	65282	Carvajal syndrome
137628	Cardiac anomalies - heterotaxy	42	Carnitine deficiency secondary to medium-chain acyl-CoA dehydrogenase deficiency	209908	CAS
369891	Cardiac anomalies-developmental delay-facial dysmorphism syndrome	156	Carnitine palmitoyl transferase 1A deficiency	56425	CAS
168796	Cardiac conduction disease - dilated cardiomyopathy - brachydactyly	228302	Carnitine palmitoyl transferase deficiency type 2, adult-onset form	94095	Casamassima-Morton-Nance syndrome
1686	Cardiac diverticulum	228305	Carnitine palmitoyl transferase deficiency type 2, hepatocardiomuscular form	275517	Caspase 8 deficiency syndrome
208600	Cardiac papillary fibroelastoma	228308	Carnitine palmitoyl transferase deficiency type 2, lethal systemic form	275517	Caspase eight deficiency state
875	Cardiac tumor of the child	228302	Carnitine palmitoyl transferase deficiency type 2, myopathic form	1101	Cassia Stocco dos Santos syndrome
2872	Cardiocranial syndrome, Pfeiffer type	228308	Carnitine palmitoyl transferase deficiency type 2, neonatal form	160	Castleman disease
37553	Cardiodysrhythmic potassium-sensitive periodic paralysis	228305	Carnitine palmitoyl transferase deficiency type 2, severe infantile form	2513	Castro Gago-Pombo-Novó syndrome
1340	Cardiofaciocutaneous syndrome	156	Carnitine palmitoyl transferase IA deficiency	195	Cat-eye syndrome
97292	Cardiogenic shock	228302	Carnitine palmitoyl transferase II deficiency, adult-onset form	50839	Cat-scratch disease
2229	Cardiogenital syndrome	228305	Carnitine palmitoyl transferase II deficiency, hepatocardiomuscular form	926	Catalase deficiency
1342	Cardiomelic syndrome type 3	228308	Carnitine palmitoyl transferase II deficiency, lethal systemic form	1373	Cataract - aberrant oral frenula - growth delay
500	Cardiomyopathic lentiginosis	156	Carnitine palmitoyl transferase IA deficiency	1366	Cataract - alopecia - sclerodactyly
1345	Cardiomyopathy - cataract - hip spine disease	228302	Carnitine palmitoyl transferase II deficiency, myopathic form	1368	Cataract - ataxia - deafness
91130	Cardiomyopathy - hypotonia - lactic acidosis	228305	Carnitine palmitoyl transferase II deficiency, severe infantile form	1383	Cataract - deafness - hypogonadism
90022	Cardiomyopathy - renal anomalies	156	Carnitine palmitoyl transferase IA deficiency	1375	Cataract - hypertrichosis - intellectual disability
70474	Cardiomyopathy with hypotonia due to cytochrome C oxidase deficiency	228302	Carnitine palmitoyl transferase II deficiency, adult-onset form	1381	Cataract - intellectual disability - anal atresia - urinary defects
70474	Cardiomyopathy with myopathy due to COX deficiency	228305	Carnitine palmitoyl transferase II deficiency, hepatocardiomuscular form	1387	Cataract - intellectual disability - hypogonadism
		228308	Carnitine palmitoyl transferase II deficiency, lethal systemic form	→1466	Cataract - microcephaly - arthrogryposis - kyphosis
		228302	Carnitine palmitoyl transferase II deficiency, myopathic form	→1466	Cataract - microcephaly - failure to thrive - kyphoscoliosis
				2712	Cataract - microphthalmia - radiculomegaly - septal heart defect

→ 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
1380	Cataract - nephropathy - encephalopathy	2008	CCGE syndrome	448010	CDG1Z
98984	Cataract, Coppock-like	99827	CCHF	79329	CDG2A
314993	Cataract-congenital heart disease-neural tube defect syndrome	661	CCHS	79330	CDG2B
162	Cataract-glaucoma syndrome	289499	CCMCO	99843	CDG2C
436174	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	319276	CCRCC	79332	CDG2D
		2505	CCSF	79333	CDG2E
		280779	CCV	238459	CDG2F
		86870	CD4+/CD56+ hematodermic neoplasm	263508	CDG2G
		437552	CD16 deficiency	95428	CDG2H
		238505	CD27 deficiency	263487	CDG2I
		98841	CD30 positive anaplastic large cell lymphoma	263501	CDG2J
1377	Cataract-microcornea syndrome	293825	CDA due to KLF1 mutation	314667	CDG2K
100990	Cataracts motor neuropathy - short stature - skeletal anomalies	98869	CDA I	356961	CDG2M
567	CATCH 22	98873	CDA II	79318	CDG syndrome type Ia
3286	Catecholaminergic polymorphic ventricular tachycardia	98870	CDA III	79319	CDG syndrome type Ib
		293825	CDA IV	79320	CDG syndrome type Ic
		98869	CDA type 1	79321	CDG syndrome type Id
		98873	CDA type 2	79322	CDG syndrome type Ie
		98870	CDA type 3	79323	CDG syndrome type If
		293825	CDA type 4	79324	CDG syndrome type Ig
		98869	CDA type I	79325	CDG syndrome type Ih
1123	Caudal appendage - deafness	98873	CDA type II	79326	CDG syndrome type II
1756	Caudal duplication	98870	CDA type III	79329	CDG syndrome type IIa
3027	Caudal dysplasia	293825	CDA type IV	79330	CDG syndrome type IIb
3027	Caudal regression sequence	98869	CDA type 1	99843	CDG syndrome type IIc
99994	Causalgia	98873	CDA type 2	79332	CDG syndrome type IId
1329	CAVC	98870	CDA type 3	79333	CDG syndrome type IIe
99068	CAVC - Fallot tetralogy	293825	CDA type 4	238459	CDG syndrome type IIIf
99066	CAVC - left heart obstruction	98869	CDA type I	263508	CDG syndrome type IIg
99067	CAVC - ventricle hypoplasia	98873	CDA type II	95428	CDG syndrome type IIh
99066	CAVC type A	98870	CDA type III	263487	CDG syndrome type III
99067	CAVC type B	293825	CDA type IV	263501	CDG syndrome type IIj
99068	CAVC type C	85199	CDAGS syndrome	314667	CDG syndrome type IIk
2124	Cavernous hemangiomas of face - supraumbilical midline raphe	293825	CDAN4	356961	CDG syndrome type IIIm
79489	Cavernous lymphangioma	247203	CDC	86309	CDG syndrome type Ij
79489	Cavernous lymphatic malformation	79318	CDG1A	79327	CDG syndrome type Ik
165958	Cavitory myiasis	79319	CDG1B	79328	CDG syndrome type IL
567	Cayler cardiofacial syndrome	79320	CDG1C	91131	CDG syndrome type Im
94122	Cayman ataxia	79321	CDG1D	244310	CDG syndrome type In
363972	CBL syndrome	79322	CDG1E	263494	CDG syndrome type Io
79282	CblC defect	79323	CDG1F	280071	CDG syndrome type Ip
79283	CblD defect	79324	CDG1G	300536	CDG syndrome type Ir
79284	CblF defect	79325	CDG1H	324422	CDG syndrome type Is
369955	CblJ defects	79326	CDG1I	319646	CDG syndrome type It
70567	CCA	86309	CDG1J	329178	CDG syndrome type Iu
115	CCA syndrome	79327	CDG1K	370921	CDG syndrome type Iw
2444	CCAM	79328	CDG1L	370924	CDG syndrome type Ix
280832	CCAM type 1	91131	CDG1M	370927	CDG syndrome type ly
280840	CCAM type 2	244310	CDG1N	448010	CDG syndrome type Iz
280847	CCAM type 3	263494	CDG1O	79318	CDG-ia
98972	CCDF	280071	CDG1P	79319	CDG-Ib
48431	CCFDN	324737	CDG1Q	79320	CDG-Ic
		300536	CDG1R		
		324422	CDG1S		
		319646	CDG1T		
		329178	CDG1U		
		370921	CDG1W		
		370924	CDG1X		
		370927	CDG1Y		

→ 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
79321	CDG-I α	3258	Cenani-Lenz syndrome	83472	Cerebellar ataxia - intellectual disability - optic atrophy - skin abnormalities
79322	CDG-I ϵ	75377	Central areolar choroidal dystrophy	276183	Cerebellar ataxia with azoospermia and intellectual disability
79323	CDG-I δ	75377	Central areolar choroidal sclerosis	94122	Cerebellar ataxia, Cayman type
79324	CDG-I γ	75327	Central areolar pigment epithelial dystrophy	97249	Cerebellar atrophy with progressive microcephaly
79325	CDG-I η	2431	Central bilateral macrogryria	2246	Cerebellar hypoplasia - tapetoretinal degeneration
79326	CDG-I ι	98972	Central cloudy corneal dystrophy of Francois	251931	Cerebellar liponeurocytoma
79329	CDG-II α	98972	Central cloudy dystrophy of Francois	251858	Cerebellar neuroblastoma
79330	CDG-II β	661	Central congenital hypoventilation syndrome	94147	Cerebellar syndrome - pigmentary maculopathy
99843	CDG-II ζ	597	Central core disease	1454	Cerebellar vermis hypoplasia-oligophrenia-congenital ataxia-coloboma-hepatic fibrosis
79332	CDG-II δ	178029	Central diabetes insipidus	444072	Cerebellar-facial-dental syndrome
79333	CDG-II ϵ	→98967	Central discoid corneal dystrophy	2318	Cerebello-oculo-renal syndrome
238459	CDG-II η	99832	Central hypothyroidism due to TRH receptor deficiency	444072	Cerebellofaciodental syndrome
263508	CDG-II γ	3240	Central nervous system calcification - deafness - tubular acidosis - anemia	475	Cerebelloparenchymal disorder IV
95428	CDG-II ι	73256	Central neurocytoma	1532	Cerebellotrigeminal - dermal dysplasia
263487	CDG-III	295004	Central polydactyly of fingers	1397	Cerebellum agenesis - hydrocephaly
263501	CDG-II \jmath	295173	Central polydactyly of fingers, bilateral	46724	Cerebral arteriovenous fistula
314667	CDG-III κ	295171	Central polydactyly of fingers, unilateral	46724	Cerebral arteriovenous malformation
356961	CDG-III μ	295010	Central polydactyly of foot	46724	Cerebral arteriovenous shunt
86309	CDG-I δ	295004	Central polydactyly of hand	136	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
79327	CDG-I κ	295010	Central polydactyly of toes	199354	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
79328	CDG-IL	295185	Central polydactyly of toes, bilateral	666631	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome
91131	CDG-Im	295183	Central polydactyly of toes, unilateral	821	Cerebral gigantism
244310	CDG-In	759	Central precocious puberty	2081	Cerebral gigantism - jaw cysts
263494	CDG-Lo	75327	Central retinal pigment epithelial dystrophy	→1900	Cerebral gigantism, Nevo type
280071	CDG-Ip	411527	Central retinal vein occlusion	77261	Cerebral juvenile and adult form of Gaucher disease
324737	CDG-Iq	443079	Central serous chorioretinopathy	221126	Cerebral proliferative glomeruloid vasculopathy
300536	CDG-Ir	90156	Centrifugal lipodystrophy	329217	Cerebral sinovenous thrombosis
324422	CDG-Is	89841	Centripetal dystrophic epidermolysis bullosa	447788	Cerebral visual impairment
319646	CDG-It	89841	Centripetal recessive dystrophic epidermolysis bullosa	1393	Cerebro-costo-mandibular syndrome
329178	CDG-Iu	89841	Centripetalis recessive dystrophic epidermolysis bullosa	397922	Cerebro-cutaneous syndrome with iron overload
370921	CDG-Iw	319160	Centronuclear myopathy type 4	314679	Cerebro-facio-articular syndrome
370924	CDG-Ix	1945	Centrotemporal epilepsy	1394	Cerebro-facio-thoracic dysplasia
370927	CDG-Iy	79277	CEP	1458	Cerebro-oculo-dento-auriculo-skeletal syndrome
448010	CDG-IZ	2398	Cephalothoracic lipodystrophy	→2995	Cerebro-oculo-facial-lymphatic syndrome
2140	CDH	79506	CEPT deficiency	66625	Cerebro-oculo-nasal syndrome
1529	CDHS	1171	Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss		
178029	CDI	1174	Cerebellar ataxia - ectodermal dysplasia		
1490	CDPD	1173	Cerebellar ataxia - hypogonadism		
35173	CDPX2	1766	Cerebellar ataxia - intellectual disability - dysequilibrium syndrome		
35173	CDPXD				
158	CDSP				
1459	CEC				
2718	Cecato de Lima-Pinheiro syndrome				
1515	CED				
666631	CEDNIK syndrome				
275517	CEDS				
1459	Celiac disease, epilepsy and cerebral calcification syndrome				
293208	Celiac trunk compression syndrome				
93942	Celosomia				
3258	Cenani syndactyly				
3258	Cenani-Lenz syndactyly				

→ 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
1396	Cerebro-reno-digital syndrome	2020	CFTDM	99953	Charcot-Marie-Tooth disease type 4G
141194	Cerebrofacial arteriovenous metameric syndrome type 1	379	CGD	99954	Charcot-Marie-Tooth disease type 4H
53719	Cerebrofacial arteriovenous metameric syndrome type 2	2026	CGHT	139515	Charcot-Marie-Tooth disease type 4J
141199	Cerebrofacial arteriovenous metameric syndrome type 3	2388	ChAc	391351	Charcot-Marie-Tooth disease type 4K
2995	Cerebrofrontofacial syndrome type 3	307766	CHAC syndrome	90120	Charcot-Marie-Tooth disease type 6
912	Cerebrohepatorenal syndrome	307766	CHACS	363981	Charcot-Marie-Tooth disease with focally folded myelin
2406	Cerebromedullospinal disconnection	3386	Chagas disease	138	CHARGE association
1466	Cerebrooculofacioskeletal syndrome	436159	CHAI	138	CHARGE syndrome
313838	Cerebroretinal microangiopathy with calcifications and cysts	→1071	CHAND syndrome	921	CHARGE-like syndrome
3421	Cerebroretinal vasculopathy	98979	Chandler syndrome	1496	Charlevoix disease
909	Cerebrotendinous xanthomatosis	→1071	CHANDS	1406	Charlie M syndrome
1980	Cerebrovascular ferrocalcinosis	2235	Chang-Davidson-Carlson syndrome	168577	CHC type 2
169079	Cernunnos deficiency	88642	Channelopathy-associated congenital insensitivity to pain	98975	CHED1
169079	Cernunnos XLFD	3282	Chaotic atrial tachycardia	293603	CHED2
169079	Cernunnos-XLF deficiency	319244	Chapare hemorrhagic fever	98975	CHEDI
98989	Cerulean cataract	46627	Char syndrome	167	Chédiak-Higashi disease
213772	Cervical adenocarcinoma	1964	Char-Douglas-Dungan syndrome	167	Chédiak-Higashi syndrome
213828	Cervical adenoid basal carcinoma	803	Charcot disease	381	Chédiak-Higashi-like syndrome
213823	Cervical adenoid cystic carcinoma	90658	Charcot-Marie-Tooth disease - deafness	167	Chédiak-Higashi-Steinbrink syndrome
213792	Cervical adenosarcoma	90103	Charcot-Marie-Tooth disease - deafness - intellectual disability	293603	CHEDII
99079	Cervical aortic arch	93114	Charcot-Marie-Tooth disease - nephropathy	1221	Cheilitis glandularis
141046	Cervical dermoid cyst	64751	Charcot-Marie-Tooth disease - pyramidal features	99647	Cheirospolyochochondromatosis
213837	Cervical germ cell cancer	101081	Charcot-Marie-Tooth disease type 1A	955	Cheney syndrome
2218	Cervical hypertrichosis - peripheral neuropathy	101082	Charcot-Marie-Tooth disease type 1B	812	Cherry-red spot-myoclonus syndrome
213807	Cervical leiomyosarcoma	101083	Charcot-Marie-Tooth disease type 1C	184	Cherubism
213837	Cervical malignant germ cell tumor	101084	Charcot-Marie-Tooth disease type 1D	3019	Cherubism - gingival fibromatosis - intellectual disability
213787	Cervical malignant Müllerian mixed tumor	90658	Charcot-Marie-Tooth disease type 1E	→672	CHHS
213812	Cervical malignant peripheral neuroectodermal tumor	101085	Charcot-Marie-Tooth disease type 1F	268882	Chiari malformation type 1
213817	Cervical papillary carcinoma	98856	Charcot-Marie-Tooth disease type 2B1	1136	Chiari malformation type 2
213812	Cervical peripheral neuroectodermal cancer	101101	Charcot-Marie-Tooth disease type 2B2	268882	Chiari malformation type I
213802	Cervical rhabdomyosarcoma	101102	Charcot-Marie-Tooth disease type 2H	1136	Chiari malformation type II
268392	Cervical spina bifida aperta	300319	Charcot-Marie-Tooth disease type 2P	33402	Childhood-onset HCC
268762	Cervical spina bifida cystica	397968	Charcot-Marie-Tooth disease type 2R	324625	Chikungunya
213767	Cervical squamous cell carcinoma	443073	Charcot-Marie-Tooth disease type 2S	90280	Chilblain lupus
3456	Cervico-oculo-acoustic syndrome	443950	Charcot-Marie-Tooth disease type 2T	139	CHILD nevus
141067	Cervicofacial enchondroma	64748	Charcot-Marie-Tooth disease type 3	139	CHILD syndrome
141067	Cervicofacial fibrochondroma	99948	Charcot-Marie-Tooth disease type 4A	64280	Childhood absence epilepsy
137923	Cervicofacial lymphatic malformation	99955	Charcot-Marie-Tooth disease type 4B1	439175	Childhood AIS
268397	Cervicothoracic spina bifida aperta	99956	Charcot-Marie-Tooth disease type 4B2	209908	Childhood apraxia of speech
268766	Cervicothoracic spina bifida cystica	363981	Charcot-Marie-Tooth disease type 4B3	439175	Childhood arterial ischemic stroke
586	CF	99949	Charcot-Marie-Tooth disease type 4C	135	Childhood ataxia with diffuse central nervous system hypomyelination
2032	CFA	99950	Charcot-Marie-Tooth disease type 4D	168782	Childhood disintegrative disorder
1340	CFC syndrome	99951	Charcot-Marie-Tooth disease type 4E	293955	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency
1520	CFND	99952	Charcot-Marie-Tooth disease type 4F	391497	Childhood myasthenia gravis
1520	CFNS			363677	Childhood-onset autosomal recessive myopathy with external ophthalmoplegia

→ 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
284324	Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia	263463	Chondrodysplasia with congenital joint dislocations, CHST3 type	137817	Chronic arachnoiditis
33402	Childhood-onset hepatocellular carcinoma	280586	Chondrodysplasia with joint dislocations, gPAPP type	71279	Chronic ataxic neuropathy - ophthalmoplegia - IgM paraprotein - cold agglutinins - disialosyl antibodies
247667	Childhood-onset hypophosphatasia	3144	Chondrodysplasia with snail-like pelvis	435988	Chronic atrial and intestinal dysrhythmia syndrome
171439	Childhood-onset nemaline myopathy	50945	Chondrodysplasia, Blomstrand type	435988	Chronic atrial dysrhythmia-intestinal motility disorder
247667	Childhood-onset phosphoethanolaminuria	2098	Chondrodysplasia, Grebe type	325004	Chronic atypical neutrophilic dermatosis-lipodystrophy-elevated temperature syndrome
247667	Childhood-onset Rathburn disease	35173	Chondrodstrophia calcificans congenita	2137	Chronic autoimmune hepatitis
101000	Childhood-onset spastic paraparesis - distal muscle wasting	289	Chondroectodermal dysplasia	133	Chronic berylliosis
3474	CHIME syndrome	319195	Chondroectodermal dysplasia with night blindness	133	Chronic beryllium disease
2888	Chitayat-Meunier-Hodgkinson syndrome	404507	Chondromyxoid fibroma	133	Chronic beryllium lung disease
3218	Chitty-Hall-Baraitser syndrome	55880	Chondrosarcoma	56425	Chronic cold agglutinin disease
3331	Chitty-Hall-Webb syndrome	444077	CHOPS syndrome	79078	Chronic dacryoadenitis and sialadenitis
757	Chloride shunt syndrome	251674	Chordoid glioma	103907	Chronic diarrhea due to glucoamylase deficiency
86850	Chloroma	178	Chordoma	314373	Chronic diarrhea due to guanylate cyclase 2C overactivity
180	CHM	2388	Chorea-acanthocytosis	397606	Chronic diarrhea with hereditary sensory and autonomic neuropathy
137914	Choanal atresia	2388	Choreoacanthocytosis	397606	Chronic diarrhea with HSAN
137920	Choanal atresia, bilateral	209905	Choreoathetosis-hypothyroidism-neonatal respiratory distress syndrome	1670	Chronic diarrhea with villous atrophy
137917	Choanal atresia, unilateral	252015	Choriocarcinoma of the central nervous system	279891	Chronic endophthalmitis
1200	Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome	251899	Choroid plexus carcinoma	168940	Chronic eosinophilic leukemia
70567	Cholangiocarcinoma	1433	Choroidal atrophy - alopecia	2902	Chronic eosinophilic pneumonia
69663	Cholelithiasis with ABCB4 gene mutation	39044	Choroidal melanoma	99921	Chronic graft versus host disease
173	Cholera	180	Choroideremia	521	Chronic granulocytic leukemia
79303	Cholestasis, with delta(4)-3-oxosteroid 5-beta-reductase deficiency	1435	Choroideremia - deafness - obesity	379	Chronic granulomatous disease
1414	Cholestasis-lymphedema syndrome	1434	Choroideremia - hypopituitarism	396	Chronic hiccough
1415	Cholestasis-pigmentary retinopathy-cleft palate syndrome	94087	CHP	396	Chronic hiccup
102069	Cholestatic hepatic amyloidosis	181	Christ-Siemens-Touraine syndrome	1451	Chronic infantile neurological cutaneous articular syndrome
→2697	Cholestatic jaundice - renal tubular insufficiency	1436	Christian syndrome	83418	Chronic infantile spinal muscular atrophy
75234	Cholesterol ester storage disease	85278	Christianson syndrome	2932	Chronic inflammatory demyelinating polyneuropathy
79506	Cholesterol-ester transfer protein deficiency	1808	Christianson-Fourie syndrome	2932	Chronic inflammatory demyelinating polyradiculoneuropathy
75234	Cholesteryl ester storage disease	98879	Christmas disease	294422	Chronic intestinal failure
166272	Chondrodysplasia - dentinogenesis imperfecta - joint laxity	1201	Christmas tree syndrome	2978	Chronic intestinal pseudoobstruction
1422	Chondrodysplasia - disorder of sex development	182	Chromoblastomycosis	284448	Chronic lymphocytic inflammation with pontine perivascular enhancement responsive to steroids
1422	Chondrodysplasia - pseudohermaphroditism	182	Chromomycosis	1334	Chronic mucocutaneous candidiasis
79344	Chondrodysplasia punctata, Sheffield type	319303	Chromophobe renal cell adenocarcinoma	521	Chronic myelogenous leukemia
79346	Chondrodysplasia punctata, tibial-metacarpal type	319303	Chromophobe renal cell carcinoma	521	Chronic myeloid leukemia
79347	Chondrodysplasia punctata, Toriello type	1450	Chromosome 8-derived supernumerary ring /marker	98823	Chronic myelomonocytic leukemia
		3380	Chromosome 18 duplication	77261	Chronic neuronopathic Gaucher disease
		195	Chromosome 22 inversion/duplication	86829	Chronic neutrophilic leukemia
		330064	Chronic actinic dermatitis		
		314928	Chronic adult hydrocephalus		
		99871	Chronic and localized Langerhans cell histiocytosis		
		99873	Chronic and multifocal Langerhans cell histiocytosis		

→ 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
439202	Chronic obstetric brachial plexus injury	435651	CIDEC-related familial partial lipodystrophy	325524	Classic congenital lipoïd adrenal hyperplasia due to STAR deficiency
439202	Chronic obstetric brachial plexus palsy	435651	CIDEC-related FPLD	329977	Classic endocrine tumor of appendix
95426	Chronic pain requiring intraspinal analgesia	2932	CIDP	93930	Classic exstrophy of the bladder
330064	Chronic photosensitivity dermatitis	79394	CIE	79239	Classic galactosemia
91359	Chronic pneumonitis of infancy	294422	CIF	98962	Classic GCD
324964	Chronic recurrent multifocal osteomyelitis	1223	Ciliary dysentery	289857	Classic glycine encephalopathy
	Chronic recurrent multifocal osteomyelitis - congenital dyserythropoietic anemia - neutrophilic dermatosis	2114	Cilliers-Brighton syndrome	98962	Classic granular corneal dystrophy
217566	Chronic respiratory distress with surfactant metabolism deficiency	1451	CINCA syndrome	391	Classic Hodgkin disease
71279	Chronic sensory ataxic neuropathy with anti-dylosyl IgM antibodies	2978	CIPO	391	Classic Hodgkin lymphoma
379	Chronic septic granulomatosis	217410	Circumscribed lymphangioma	98846	Classic Hodgkin lymphoma, lymphocyte-depleted type
83418	Chronic spinal muscular atrophy	217410	Circumscribed lymphatic malformation	98845	Classic Hodgkin lymphoma, lymphocyte-rich type
70591	Chronic thromboembolic pulmonary hypertension	69744	Circumscribed palmoplantar hypokeratosis	98844	Classic Hodgkin lymphoma, mixed cellularity type
97353	Chronic traumatic encephalopathy	309854	Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome	98843	Classic Hodgkin lymphoma, nodular sclerosis type
37748	Chronic urticaria with gammopathy	57777	Cirrhotic cardiomyopathy	394	Classic homocystinuria
37748	Chronic urticaria with macroglobulinemia	240863	Cisplatin toxicity	475	Classic Joubert syndrome
263463	CHST3-related skeletal dysplasia	157820	CISS	313	Classic lamellar ichthyosis
2953	CHST14-related EDS	247525	Citrullinemia type 1	98964	Classic lattice corneal dystrophy
2953	CHST14-related Ehlers-Danlos syndrome	247585	Citrullinemia type 2	268145	Classic maple syrup urine disease
93971	Chudley-Lowry syndrome	247525	Citrullinemia type I	158796	Classic mast cell leukemia
93971	Chudley-Lowry-Hoar syndrome	247585	Citrullinemia type II	251867	Classic medulloblastoma
314597	Chudley-McCullough syndrome	251383	CK syndrome	324604	Classic MmD
3068	Chudley-Rozdilsky syndrome	90790	CLAH	268145	Classic MSUD
183	Churg-Strauss syndrome	97249	CLAM	324604	Classic multiminicore disease
238557	Chuvash erythrocytosis	168984	CLAPO syndrome	324604	Classic multiminicore myopathy
238557	Chuvash polycythemia	188	Clarkson disease	2584	Classic mycosis fungoides
71	Chylomicron retention disease	90794	Classic 21-OHD CAH	216866	Classic pantothenate kinase-associated neurodegeneration
1160	Chylous ascites	315306	Classic 21-OHD CAH, salt wasting form	163898	Classic paraneoplastic limbic encephalitis
46486	Cicatricial pemphigoid	315311	Classic 21-OHD CAH, simple virilizing form	163898	Classic paraneoplastic limbic encephalitis, with or without intracellular antigens
217390	CID due to DOCK8 deficiency	85138	Classic Addison's disease	93258	Classic Pfeiffer syndrome
317473	CID due to ikaros deficiency	329977	Classic appendiceal endocrine tumor	79254	Classic phenylketonuria
445018	CID due to LRBA deficiency	329977	Classic appendix endocrine tumor	79254	Classic PKU
317476	CID due to MAGT1 deficiency	93605	Classic Bartter syndrome	280219	Classic PMD
317428	CID due to ORAI1 deficiency	268145	Classic BCKD deficiency	240071	Classic progressive supranuclear palsy
443811	CID due to PGM3 deficiency	268145	Classic branched-chain 2-ketoacid dehydrogenase deficiency	240071	Classic PSP
157949	CID due to RAG 1/2 deficiency	268145	Classic branched-chain ketoaciduria	773	Classic Refsum disease
317430	CID due to STIM1 deficiency	247525	Classic citrullinemia	18	Classic RTA
314689	CID due to STK4 deficiency	247546	Classic citrullinemia type 1	443192	Classic SPS
231154	CID T+ B+ due to partial RAG1 deficiency	247546	Classic citrullinemia type I	443192	Classic stiff person syndrome
231154	CID with expansion of gamma delta T cells	325524	Classic CLAH	3467	Classic xanthinuria
436252	CID-MIA/early-onset IBD	90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	2272	Clayton Smith-Donnai syndrome
		315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	319276	Clear cell adenocarcinoma
		315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	398971	Clear cell adenocarcinoma of ovary
				404511	Clear cell papillary renal cell carcinoma
				319276	Clear cell renal carcinoma

→ 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
319276	Clear cell renal cell adenocarcinoma	228366	CLN7 disease	101084	CMT1D
319276	Clear cell renal cell carcinoma	228354	CLN8 disease	90658	CMT1E
97338	Clear cell sarcoma of soft tissue	1947	CLN8 disease, Northern epilepsy variant	101085	CMT1F
97338	Clear cell sarcoma of the tendons and aponeuroses	228357	CLN9 disease	101075	CMT1X
3429	Cleft - limb-heart malformation syndrome	228337	CLN10 disease	324611	CMT2 due to KIF5A mutation
101023	Cleft hard palate	314629	CLN11 disease	435819	CMT2 due to TFG mutation
1995	Cleft lip - retinopathy	314632	CLN12 disease	435387	CMT2 due to VCP mutation
2319	Cleft lip/palate - abnormal thumbs - microcephaly	352709	CLN13 disease	401964	CMT2 with giant axons
2003	Cleft lip/palate - deafness - sacral lipoma	93929	Cloacal exstrophy	99946	CMT2A1
→1896	Cleft lip/palate - ectrodactyly	314950	Clonal hypereosinophilic syndrome	99947	CMT2A2
2328	Cleft lip/palate - facial, eye, heart and intestinal anomalies	221083	Clonic hemifacial spasm	99936	CMT2B
2001	Cleft lip/palate - intestinal malrotation - cardiopathy	268366	Closed iniencephaly	99937	CMT2C
888	Cleft lip/palate with mucous cysts of lower lip	189	Clouston syndrome	99938	CMT2D
3253	Cleft lip/palate-ectodermal dysplasia syndrome	140944	CLOVE syndrome	99939	CMT2E
3253	Cleft lip/palate-syndactyly-pili torti syndrome	100978	Cloverleaf skull - asphyxiating thoracic dysplasia	99940	CMT2F
95465	Cleft mitral valve	93274	Cloverleaf skull - micromelic bone dysplasia	99941	CMT2G
141242	Cleft nose	93267	Cloverleaf skull - multiple congenital anomalies	99942	CMT2I
2014	Cleft palate	411493	CLP1-related pontocerebellar hypoplasia	99943	CMT2J
2008	Cleft palate - cardiac defect - genital anomalies - ectrodactyly	3253	CLPED1	99944	CMT2K
2013	Cleft palate - large ears - small head	192	CLS	99945	CMT2L
2015	Cleft palate - short stature - vertebral anomalies	85136	CLWM	228179	CMT2M
2010	Cleft palate - stapes fixation - oligodontia	137667	CM-AVM	228174	CMT2N
921	Cleft palate-coloboma-deafness syndrome	289504	CMAMMA	284232	CMT2O
2016	Cleft palate-lateral synchia syndrome	1334	CMC	300319	CMT2P
99772	Cleft velum	258	CMD1A	329258	CMT2Q
99772	Cleft velum palatinum	98893	CMD1B	397968	CMT2R
1997	Clefting - ectropion - conical teeth	→37095	CMD1C	443073	CMT2S
1452	Cleidocranial dysostosis	3		443950	CMT2T
1452	Cleidocranial dysplasia	370959	CMD with cerebellar involvement	397735	CMT2U
3472	Cleidocranial dysplasia - micrognathia - absent thumbs	370968	CMD with intellectual disability	447964	CMT2V
1453	Cleidorhizomelic syndrome	329178	CMD with intellectual disability and severe epilepsy	101076	CMT2X
284448	CLIPPERS	370980	CMD without intellectual disability	101077	CMT3X
228329	CLN1 disease	370959	CMD-CRB	99948	CMT4A
228349	CLN2 disease	370968	CMD-MR	99955	CMT4B1
228346	CLN3 disease	370980	CMD-no MR	99956	CMT4B2
228340	CLN4A disease	371007	CMDH	363981	CMT4B3
228343	CLN4B disease	521	CML	99949	CMT4C
228360	CLN5 disease	252202	CMMR-D syndrome	99950	CMT4D
228363	CLN6 disease	99763	CMO I	99951	CMT4E
		99763	CMO II	99952	CMT4F
		238459	CMP-sialic acid transporter deficiency	99953	CMT4G
		86830	CMPD-U	99954	CMT4H
		71	CMRD	139515	CMT4J
		590	CMS	391351	CMT4K
		101081	CMT1A	101078	CMT4X
		101082	CMT1B	99014	CMT5X
		101083	CMT1C	90120	CMT6
				352675	CMT6X
				1556	CMTC
				100043	CMTDIA
				100044	CMTDIB
				100045	CMTDIC

→ 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
100046	CMTDID	79333	COG7-CDG	16	Color blindness, blue moncone monochromatic type
93114	CMTDIE	95428	COG8-CDG	83595	Colorado tick encephalitis
352670	CMTDIF	1467	Cogan syndrome	83595	Colorado tick fever
101075	CMTX1	98980	Cogan-Reese syndrome	83595	Colorado tick-borne disease
101076	CMTX2	444077	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	733	Colorectal adenomatous polyposis
101077	CMTX3			261584	Colorectal adenomatous polyposis due to monosomy 5q22.2
101078	CMTX4			90793	Combined 17-hydroxylase/17,20-lyase deficiency
99014	CMTX5			445062	Combined cerebellar and peripheral ataxia-hearing loss-diabetes mellitus syndrome
352675	CMTX6			370114	Combined cervical dystonia
137698	CMV disease in patients with impaired cell mediated immunity deemed at risk	193	Cohen syndrome	356978	Combined D-2-hydroxyglutaric aciduria and L-2-hydroxyglutaric aciduria
319160	CNM4	2969	Cohen-Hayden syndrome	356978	Combined D-2-hydroxyglutaric aciduria and L-2-hydroxyglutaric aciduria
306686	CO-induced parkinsonism	79144	COIF	26	Combined defect in adenosylcobalamin and methylcobalamin synthesis
1454	COACH syndrome	79144	COIF syndrome	79282	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIC
1456	Coarctation of the abdominal aorta	31824	Colchicine poisoning	79283	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbID
397725	COASY protein-associated neurodegeneration	56425	Cold agglutinin disease	79284	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIF
190	Coats disease	56425	Cold agglutinin syndrome	369955	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIJ
313838	Coats plus syndrome	157820	Cold-induced sweating syndrome	369962	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbIX
79282	Cobalamin C defect	324561	Cole disease	35909	Combined deficiency of factor V and factor VIII
79283	Cobalamin D defect	2050	Cole-Carpenter syndrome	99732	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase
79284	Cobalamin F defect	84087	Collagen type III glomerulopathy	308386	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type A
369955	Cobalamin J defect	36205	Collagenous colitis	308393	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type B
53721	Cobb syndrome	247203	Collecting duct carcinoma	308400	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type C
352682	Cobblestone lissencephaly without muscular or eye involvement	2412	Collins-Pope syndrome	440727	Combined hamartoma of the retina and retinal pigment epithelium
352682	Cobblestone lissencephaly without muscular or ocular involvement	168	Coloboma - hair abnormality	440727	Combined hamartoma of the retina and RPE
1911	Cocaine embryofetopathy	98942	Coloboma of choroid and retina		
90068	Cocaine poisoning	98943	Coloboma of eye lens		
228123	Coccidioides infection	98946	Coloboma of eyelid		
228123	Coccidioidomycosis	155889	Coloboma of inferior eyelid		
3233	Cochleosaccular degeneration - cataract	98944	Coloboma of iris		
191	Cockayne syndrome	98945	Coloboma of macula		
90321	Cockayne syndrome type 1	1471	Coloboma of macula - brachydactyly type B		
90322	Cockayne syndrome type 2	1475	Coloboma of optic nerve with renal disease		
90324	Cockayne syndrome type 3	98947	Coloboma of optic papilla		
90321	Cockayne syndrome type I	155884	Coloboma of superior eyelid		
90322	Cockayne syndrome type II	3474	Coloboma-congenital heart disease-ichthyosisform dermatosis-intellectual disability-ear anomalies syndrome		
90324	Cockayne syndrome type III	138	Coloboma-heart defects-atresia choanae-retardation of growth and development-genitourinary problems-ear abnormalities syndrome		
1458	CODAS syndrome	→138	Colobomatous - microphthalmia - heart disease - hearing loss		
240867	Codeine toxicity	98938	Colobomatous microphthalmia		
192	Coffin-Lowry syndrome	363741	Colobomatous microphthalmia-obesity-hypogenitalism-intellectual disability syndrome		
1465	Coffin-Siris syndrome	424099	Colobomatous microphthalmia-rhizomelic dysplasia syndrome		
1466	COFS syndrome	435930	Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome		
263508	COG1-CDG	1198	Colonc atresia		
435934	COG2-related congenital disorder of glycosylation				
263501	COG4-CDG				
263487	COG5-CDG				

→ 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
221078	Combined hyperactive dysfunction syndrome of the cranial nerves	1979	Combined insulin, insulin-like growth factor 1 (IGF1) and epidermal growth factor (EGF) deficiency	3384	Common arterial trunk
169079	Combined immunodeficiency - microcephaly - growth retardation - sensitivity to ionizing radiation	289504	Combined malonic and methylmalonic aciduria	1329	Common atrioventricular canal
169082	Combined immunodeficiency due to CD3gamma deficiency	289504	Combined malonic and methylmalonic aciduria	→288	Common hereditary elliptocytosis
169090	Combined immunodeficiency due to CRAC channel dysfunction	254920	Combined oxidative phosphorylation defect type 2	620	Common mesentery
217390	Combined immunodeficiency due to dedicator of cytokinesis 8 protein deficiency	254925	Combined oxidative phosphorylation defect type 4	1572	Common variable immunodeficiency
217390	Combined immunodeficiency due to DOCK8 deficiency	137908	Combined oxidative phosphorylation defect type 5	280821	Communicating congenital bronchopulmonary-foregut malformation
317473	Combined immunodeficiency due to ikaros deficiency	254930	Combined oxidative phosphorylation defect type 7	280133	Complement component 3 deficiency
445018	Combined immunodeficiency due to LRBA deficiency	319504	Combined oxidative phosphorylation defect type 8	99429	Complete androgen insensitivity syndrome
317476	Combined immunodeficiency due to MAGT1 deficiency	319509	Combined oxidative phosphorylation defect type 9	99429	Complete androgen resistance syndrome
397964	Combined immunodeficiency due to MALT1 deficiency	314637	Combined oxidative phosphorylation defect type 10	1329	Complete atrioventricular canal
317428	Combined immunodeficiency due to ORAI1 deficiency	324535	Combined oxidative phosphorylation defect type 11	99068	Complete atrioventricular canal - Fallot tetralogy
431149	Combined immunodeficiency due to OX40 deficiency	319514	Combined oxidative phosphorylation defect type 13	99066	Complete atrioventricular canal - left heart obstruction
443811	Combined immunodeficiency due to PGM3 deficiency	319519	Combined oxidative phosphorylation defect type 14	99067	Complete atrioventricular canal - ventricle hypoplasia
157949	Combined immunodeficiency due to RAG 1/2 deficiency	319524	Combined oxidative phosphorylation defect type 15	99066	Complete atrioventricular canal type A
317430	Combined immunodeficiency due to STIM1 deficiency	352563	Combined oxidative phosphorylation defect type 16	99067	Complete atrioventricular canal type B
314689	Combined immunodeficiency due to STK4 deficiency	369913	Combined oxidative phosphorylation defect type 17	99068	Complete atrioventricular canal type C
911	Combined immunodeficiency due to ZAP70 deficiency	314051	Combined oxidative phosphorylation deficiency type 12	1329	Complete atrioventricular septal defect
231154	Combined immunodeficiency T+ B+ due to partial RAG1 deficiency	420728	Combined oxidative phosphorylation deficiency type 20	98949	Complete cryptophthalmia
431149	Combined immunodeficiency with childhood-onset Kaposi sarcoma	420733	Combined oxidative phosphorylation deficiency type 21	289916	Complete deficiency of methylmalonyl-CoA mutase
221139	Combined immunodeficiency with facio-oculo-skeletal anomalies	444013	Combined oxidative phosphorylation deficiency type 23	633	Complete growth hormone insensitivity
39041	Combined immunodeficiency with hypereosinophilia	444458	Combined oxidative phosphorylation deficiency type 24	254688	Complete hydatidiform mole
431149	Combined immunodeficiency with impaired immunity to HHV-8	447954	Combined oxidative phosphorylation deficiency type 25	79293	Complete LCAT deficiency
		309111	Combined pancreatic lipase-colipase deficiency	29	Complete mevalonate kinase deficiency
		95494	Combined pituitary hormone deficiencies, genetic forms	254688	Complete molar pregnancy
157949	Combined immunodeficiency with skin granulomas	139406	Combined prosaposin deficiency	49382	Complete or incomplete color blindness
228423	Combined immunodeficiency with susceptibility to mycobacterial, viral and fungal infections	300564	Combined pulmonary fibrosis-emphysema syndrome	101063	Complete situs inversus
436252	Combined immunodeficiency-enteropathy spectrum	166286	Comedo nevus of the palm	101063	Complete situs inversus viscerum
		141276	Commissural facial cleft	180074	Complete unilateral aplasia of the Müllerian ducts
		141061	Commissural lip fistula	180074	Complete unilateral Müllerian aplasia
		3384	Common aortico-pulmonary trunk	83452	Complex regional pain syndrome
				99995	Complex regional pain syndrome type 1
				99994	Complex regional pain syndrome type 2
				306644	Complication after organ transplantation
				268316	Complication in hemodialysis
				168966	Composite Hodgkin and non-Hodgkin lymphoma

→ 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
168966	Composite lymphoma	295118	Congenital absence of toes, bilateral	95449	Congenital aortic valve insufficiency
634	Comèl-Netherton syndrome	295116	Congenital absence of toes, unilateral	3093	Congenital aortic valve stenosis
228165	Concentric demyelination	2879	Congenital absence of ulna and fibula	2037	Congenital aortopulmonary septal defect
3216	Conductive deafness - malformed external ear	294975	Congenital absence of upper arm and forearm with hand present	2037	Congenital aortopulmonary window
3236	Conductive deafness - ptosis - skeletal anomalies	295087	Congenital absence of upper arm and forearm with hand present, bilateral	93322	Congenital aplasia and dysplasia of the tibia with intact fibula
383	Conductive deafness with stapes fixation	295085	Congenital absence of upper arm and forearm with hand present, unilateral	353334	Congenital arteriovenous anastomoses of the retina
1871	Cone dystrophy	247775	Congenital absence of uterus and vagina	353334	Congenital arteriovenous communication of the retina
209932	Cone dystrophy with supernormal rod electroretinogram	96269	Congenital absence of vagina	1195	Congenital atranferrinemia
209932	Cone dystrophy with supernormal rod ERG	294990	Congenital absence/hypoplasia of fingers excluding thumb	60041	Congenital atrioventricular block
209932	Cone dystrophy with supernormal rod response	295114	Congenital absence/hypoplasia of fingers excluding thumb, bilateral	162526	Congenital auditory ossicle malformation without external ear abnormality
209932	Cone dystrophy with supernormal scotopic electroretinogram	973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral	1216	Congenital benign spinal muscular atrophy with contractures
1872	Cone rod dystrophy	294988	Congenital absence/hypoplasia of thumb	48	Congenital bilateral absence of vas deferens
1873	Cone rod dystrophy - amelogenesis imperfecta	295112	Congenital absence/hypoplasia of thumb, bilateral	48	Congenital bilateral agenesis of vas deferens
221142	Confetti-like macular atrophy	295110	Congenital absence/hypoplasia of thumb, unilateral	48	Congenital bilateral aplasia of vas deferens
440233	Congenital abducens nerve palsy	324353	Congenital achiasma	93177	Congenital bilateral megacalycosis
294979	Congenital absence of both forearm and hand	93583	Congenital ADAMTS-13 deficiency	79301	Congenital bile acid synthesis defect type 1
295095	Congenital absence of both forearm and hand, bilateral	90791	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	79303	Congenital bile acid synthesis defect type 2
295093	Congenital absence of both forearm and hand, unilateral	90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	79302	Congenital bile acid synthesis defect type 3
294981	Congenital absence of both lower leg and foot	90793	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	79095	Congenital bile acid synthesis defect type 4
295099	Congenital absence of both lower leg and foot, bilateral	95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	300337	Congenital blindness due to retinal non-attachment
295097	Congenital absence of both lower leg and foot, unilateral	95699	Congenital adrenal hyperplasia due to cytochrome POR deficiency	2292	Congenital bowing of long bones
289465	Congenital absence of fingerprints	95701	Congenital adrenal hypoplasia of maternal cause	71278	Congenital brain dysgenesis due to glutamine synthetase deficiency
294986	Congenital absence of foot	33355	Congenital aleukocytosis	2040	Congenital bronchobiliary fistula
295107	Congenital absence of foot, bilateral	79	Congenital alpha2-antiplasmin deficiency	3161	Congenital bronchopulmonary sequestration
295105	Congenital absence of foot, unilateral	210122	Congenital alveolar capillary dysplasia	1369	Congenital cataract - hypertrophic cardiomyopathy - mitochondrial myopathy
294983	Congenital absence of hand	3319	Congenital amegakaryocytic thrombocytopenia	1376	Congenital cataract - ichthyosis
295103	Congenital absence of hand, bilateral	3319	Congenital amegakaryocytic thrombocytopenic purpura	330054	Congenital cataract - progressive muscular hypotonia - deafness - developmental delay
295101	Congenital absence of hand, unilateral	86816	Congenital analbuminemia	330054	Congenital cataract - progressive muscular hypotonia - hearing loss - developmental delay
86815	Congenital absence of lacrimal puncta and salivary glands	217399	Congenital analgesia with hyperhidrosis	289499	Congenital cataract microcornea with corneal opacity
217399	Congenital absence of pain with hyperhidrosis	95507	Congenital anomaly of hepatic vein	300313	Congenital cataract-deafness-severe developmental delay syndrome
294977	Congenital absence of thigh and lower leg with foot present	91489	Congenital anterior megalophthalmia		
295091	Congenital absence of thigh and lower leg with foot present, bilateral	2037	Congenital aortopulmonary artery fistula		
295089	Congenital absence of thigh and lower leg with foot present, unilateral				
93322	Congenital absence of tibia				
435623	Congenital absence of toes				

→ 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
300313	Congenital cataract-hearing loss-severe developmental delay syndrome	2444	Congenital cystic disease of the lung	79330	Congenital disorder of glycosylation type 2b
48431	Congenital cataracts - facial dysmorphism - neuropathy	280832	Congenital cystic disease of the lung type 1	79332	Congenital disorder of glycosylation type 2d
99803	Congenital central alveolar hypoventilation - Hirschsprung disease	280840	Congenital cystic disease of the lung type 2	79333	Congenital disorder of glycosylation type 2e
661	Congenital central alveolar hypoventilation syndrome	280847	Congenital cystic disease of the lung type 3	238459	Congenital disorder of glycosylation type 2f
2345	Congenital cervical vertebral fusion	168612	Congenital deficiency in alpha-fetoprotein	95428	Congenital disorder of glycosylation type 2h
53689	Congenital chloride diarrhea	2140	Congenital diaphragmatic hernia	356961	Congenital disorder of glycosylation type 2m
329242	Congenital chronic diarrhea with exudative enteropathy	3474	Congenital disorder of glycosylation due to PIGL deficiency	79318	Congenital disorder of glycosylation type 1a
329242	Congenital chronic diarrhea with protein-losing enteropathy	79318	Congenital disorder of glycosylation type 1a	79319	Congenital disorder of glycosylation type 1b
264688	Congenital chylothorax	79319	Congenital disorder of glycosylation type 1b	79320	Congenital disorder of glycosylation type 1c
2505	Congenital circumferential skin folds	79320	Congenital disorder of glycosylation type 1c	79321	Congenital disorder of glycosylation type 1d
91413	Congenital Claude-Bernard-Horner syndrome	79321	Congenital disorder of glycosylation type 1d	79322	Congenital disorder of glycosylation type 1e
440221	Congenital CNIII lesion	79322	Congenital disorder of glycosylation type 1e	79323	Congenital disorder of glycosylation type 1f
98686	Congenital CNIV palsy	79323	Congenital disorder of glycosylation type 1f	79324	Congenital disorder of glycosylation type 1g
440233	Congenital CNVI palsy	79324	Congenital disorder of glycosylation type 1g	79325	Congenital disorder of glycosylation type 1h
269505	Congenital communicating hydrocephalus	79325	Congenital disorder of glycosylation type 1h	79326	Congenital disorder of glycosylation type 1i
99129	Congenital complete agenesis of pericardium	79326	Congenital disorder of glycosylation type 1i	79329	Congenital disorder of glycosylation type IIa
115	Congenital contractual arachnodactyly	86309	Congenital disorder of glycosylation type 1j	79330	Congenital disorder of glycosylation type IIb
178382	Congenital convex foot	79327	Congenital disorder of glycosylation type 1k	79332	Congenital disorder of glycosylation type IId
178382	Congenital convex pes valgus	79328	Congenital disorder of glycosylation type 1L	79333	Congenital disorder of glycosylation type IIe
53691	Congenital cornea plana	91131	Congenital disorder of glycosylation type 1m	238459	Congenital disorder of glycosylation type IIff
95491	Congenital coronary artery aneurysm	244310	Congenital disorder of glycosylation type 1n	263508	Congenital disorder of glycosylation type IIg
2444	Congenital cystic adenomatoid malformation of the lung	280071	Congenital disorder of glycosylation type 1p	95428	Congenital disorder of glycosylation type IIh
280827	Congenital cystic adenomatoid malformation of the lung type 0	324737	Congenital disorder of glycosylation type 1q	263487	Congenital disorder of glycosylation type IIIi
280832	Congenital cystic adenomatoid malformation of the lung type 1	300536	Congenital disorder of glycosylation type 1r	263501	Congenital disorder of glycosylation type IIj
280840	Congenital cystic adenomatoid malformation of the lung type 2	324422	Congenital disorder of glycosylation type 1s	356961	Congenital disorder of glycosylation type IIIm
280847	Congenital cystic adenomatoid malformation of the lung type 3	370924	Congenital disorder of glycosylation type 1x	86309	Congenital disorder of glycosylation type Ij
280854	Congenital cystic adenomatoid malformation of the lung type 4	370927	Congenital disorder of glycosylation type 1y	79327	Congenital disorder of glycosylation type Ik
2444	Congenital cystic adenomatous malformation of the lung	448010	Congenital disorder of glycosylation type 1Z	79328	Congenital disorder of glycosylation type IL
280827	Congenital cystic adenomatous malformation of the lung type 0	79329	Congenital disorder of glycosylation type 2a		
280832	Congenital cystic adenomatous malformation of the lung type 1				
280840	Congenital cystic adenomatous malformation of the lung type 2				
280847	Congenital cystic adenomatous malformation of the lung type 3				
280854	Congenital cystic adenomatous malformation of the lung type 4				

→ 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
91131	Congenital disorder of glycosylation type Im	157826	Congenital epulis	98975	Congenital hereditary endothelial dystrophy type I
244310	Congenital disorder of glycosylation type In	231573	Congenital erosive and vesicular dermatosis	293603	Congenital hereditary endothelial dystrophy type II
263494	Congenital disorder of glycosylation type Io	90042	Congenital erythrocytosis due to erythropoietin receptor mutation	306530	Congenital hereditary facial palsy with variable deafness
280071	Congenital disorder of glycosylation type Ip	369992	Congenital erythroderma-hypotrichosis-recurrent infections-multiple food allergies syndrome	306530	Congenital hereditary facial palsy with variable hearing loss
324737	Congenital disorder of glycosylation type Iq	79277	Congenital erythropoietic porphyria	306530	Congenital hereditary facial paralysis with variable deafness
300536	Congenital disorder of glycosylation type Ir	91358	Congenital esophageal diverticulum	306530	Congenital hereditary facial paralysis with variable hearing loss
324422	Congenital disorder of glycosylation type Is	215	Congenital essential nyctalopia	101068	Congenital hereditary stromal dystrophy
329178	Congenital disorder of glycosylation type Iu	91	Congenital estrogen deficiency	293	Congenital herpes virus infection
370921	Congenital disorder of glycosylation type Iw	280811	Congenital extrapulmonary sequestration	483	Congenital high-molecular-weight kininogen deficiency
370924	Congenital disorder of glycosylation type Ix	99176	Congenital eyelid retraction	91413	Congenital Horner syndrome
370927	Congenital disorder of glycosylation type Iy	570	Congenital facial diplegia	2185	Congenital hydrocephalus
293825	Congenital dyserythropoietic anemia due to KLF1 mutation	325	Congenital factor II deficiency	2190	Congenital hydronephrosis
98869	Congenital dyserythropoietic anemia type 1	326	Congenital factor V deficiency	478	Congenital hypogonadotropic hypogonadism with anosmia
98873	Congenital dyserythropoietic anemia type 2	327	Congenital factor VII deficiency	124	Congenital hypoplastic anemia, Blackfan-Diamond type
98870	Congenital dyserythropoietic anemia type 3	328	Congenital factor X deficiency	→672	Congenital hypothalamic hamartoma syndrome
293825	Congenital dyserythropoietic anemia type 4	329	Congenital factor XI deficiency	226313	Congenital hypothyroidism due to maternal intake of antithyroid drugs
98869	Congenital dyserythropoietic anemia type I	330	Congenital factor XII deficiency	95715	Congenital hypothyroidism due to transplacental passage of maternal TSH-binding inhibitory antibodies
98873	Congenital dyserythropoietic anemia type II	331	Congenital factor XIII deficiency	1195	Congenital hypotransferrinemia
98870	Congenital dyserythropoietic anemia type III	92050	Congenital familial intractable diarrhea with epithelial or epithelium abnormalities	79458	Congenital hypotrichosis - milia
293825	Congenital dyserythropoietic anemia type IV	2020	Congenital fiber-type disproportion myopathy	2271	Congenital ichthyosis - microcephalus - quadriplegia
67044	Congenital dyserythropoietic anemia with thrombocytopenia	335	Congenital fibrinogen deficiency	2271	Congenital ichthyosis - microcephalus - tetraplegia
91491	Congenital ectropion uveae	45358	Congenital fibrosis of extraocular muscles	88621	Congenital ichthyosis type 4
295032	Congenital elbow dislocation	90045	Congenital folate malabsorption	352333	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome
295227	Congenital elbow dislocation, bilateral	98686	Congenital fourth cranial nerve palsy	352333	Congenital ichthyosis-intellectual disability-spastic tetraplegia syndrome
295225	Congenital elbow dislocation, unilateral	2345	Congenital fused cervical segments	631	Congenital IGHD
103910	Congenital enterocyte heparan sulfate deficiency	2026	Congenital generalized hypertrichosis terminalis	231662	Congenital IGHD type IA
168601	Congenital enterokinase deficiency	1023	Congenital generalized hypertrichosis, Ambras type	231671	Congenital IGHD type IB
168601	Congenital enteropathy due to enteropeptidase deficiency	79495	Congenital generalized hypertrichosis, Macias-Flores type	231679	Congenital IGHD type II
292	Congenital enterovirus infection	295232	Congenital genu flexum	231692	Congenital IGHD type III
70596	Congenital Epstein-Barr virus infection	295229	Congenital genu recurvatum	217399	Congenital indifference to pain with hyperhidrosis
		157826	Congenital gingival cell tumor	64752	Congenital insensitivity to pain and thermal analgesia
		98976	Congenital glaucoma	217399	Congenital insensitivity to pain with hyperhidrosis
		157826	Congenital granular cell tumor		
		330	Congenital Hageman factor deficiency		
		60041	Congenital heart block		
		139	Congenital hemidysplasia with ichthyosiform nevus and limbs defects		
		238691	Congenital hepatic hemangioma		
		98975	Congenital hereditary endothelial dystrophy type 1		
		293603	Congenital hereditary endothelial dystrophy type 2		

→ 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
391397	Congenital insensitivity to pain with hyperhidrosis and gastrointestinal dysfunction	768	Congenital long QT syndrome	370968	Congenital muscular dystrophy with intellectual disability
388	Congenital intestinal aganglionosis	93323	Congenital longitudinal deficiency of the fibula	329178	Congenital muscular dystrophy with intellectual disability and severe epilepsy
280802	Congenital intrapulmonary sequestration	93321	Congenital longitudinal deficiency of the radius	34520	Congenital muscular dystrophy with ITGA7 deficiency
1229	Congenital intrauterine infection-like syndrome	93322	Congenital longitudinal deficiency of the tibia	280671	Congenital muscular dystrophy with mitochondrial structural abnormalities
332	Congenital intrinsic factor deficiency	93320	Congenital longitudinal deficiency of the ulna	370980	Congenital muscular dystrophy without intellectual disability
199296	Congenital isolated ACTH deficiency	2430	Congenital macroglossia	272	Congenital muscular dystrophy, Fukuyama type
631	Congenital isolated GH deficiency	95430	Congenital major airway collapse	75840	Congenital muscular dystrophy, Ullrich type
231662	Congenital isolated GH deficiency type IA	83620	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	590	Congenital myasthenic syndrome
231671	Congenital isolated GH deficiency type IB	141214	Congenital maxillomandibular fusion	353327	Congenital myasthenic syndromes with glycosylation defect
231679	Congenital isolated GH deficiency type II	93109	Congenital megacalycosis	168572	Congenital myopathy - cleft palate - malignant hyperthermia
231692	Congenital isolated GH deficiency type III	280671	Congenital megaconial myopathy	98904	Congenital myopathy with excess of thin filaments
631	Congenital isolated growth hormone deficiency	69063	Congenital membranous nephropathy due to maternal anti-neutral endopeptidase alloimmunization	319160	Congenital myopathy with internal nuclei and atypical cores
231662	Congenital isolated growth hormone deficiency type IA	2665	Congenital mesoblastic nephroma	424107	Congenital myopathy with myasthenic-like onset
231671	Congenital isolated growth hormone deficiency type IB	391376	Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome	199329	Congenital myopathy, Paradas type
231679	Congenital isolated growth hormone deficiency type II	566	Congenital microcoria	289380	Congenital myosclerosis, Löwenthal type
231692	Congenital isolated growth hormone deficiency type III	199293	Congenital microgastria	831	Congenital narrowing of cervical spinal canal
209893	Congenital isolated TBG deficiency	2290	Congenital microvillus atrophy	162521	Congenital nasal pyriform aperture stenosis with holoprosencephaly
209893	Congenital isolated thyroxine-binding globulin deficiency	2290	Congenital microvillus atrophy	451612	Congenital nasolacrimal duct obstruction
295034	Congenital knee dislocation	566	Congenital miosis	168486	Congenital NCL
53690	Congenital lactase deficiency	99057	Congenital mitral stenosis	443988	Congenital nephrosis-cerebral ventriculomegaly syndrome
70472	Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type	98905	Congenital multicore myopathy with external ophthalmoplegia	839	Congenital nephrotic syndrome, Finnish type
313	Congenital lamellar ichthyosis	1875	Congenital muscular dystrophy - infantile cataract - hypogonadism	306504	Congenital nephrotic syndrome-interstitial lung disease-epidermolysis bullosa syndrome
99872	Congenital Langerhans cell histiocytosis	258	Congenital muscular dystrophy due to laminin alpha2 deficiency	168486	Congenital neuronal ceroid lipofuscinosis
141124	Congenital laryngeal cyst	157973	Congenital muscular dystrophy due to LMNA mutation	369852	Congenital neutropenia-bone marrow fibrosis-nephromegaly syndrome
137932	Congenital laryngeal palsy	280671	Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect	369852	Congenital neutropenia-myelofibrosis-nephromegaly syndrome
2374	Congenital laryngeal web	258	Congenital muscular dystrophy type 1A	79394	Congenital non-bullous ichthyosiform erythroderma
2373	Congenital laryngomalacia	98893	Congenital muscular dystrophy type 1B	269510	Congenital non-communicating hydrocephalus
1954	Congenital lethal erythroderma	→37095	Congenital muscular dystrophy type 3 1C	269505	Congenital non-obstructive hydrocephalus
210163	Congenital lethal myopathy, Compton-North type	→37095	Congenital muscular dystrophy type 3 1D		
93937	Congenital limb amputation	370959	Congenital muscular dystrophy with cerebellar involvement		
90790	Congenital lipid adrenal hyperplasia due to STAR deficiency	371007	Congenital muscular dystrophy with hyperlaxity		
140944	Congenital lipomatous overgrowth - vascular malformation - epidermal nevi	34520	Congenital muscular dystrophy with integrin alpha-7 deficiency		
238691	Congenital liver hemangioma				
1928	Congenital lobar emphysema				
1928	Congenital lobar hyperinflation				

→ 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
1216	Congenital nonprogressive spinal muscular atrophy	157808	Congenital pseudoarthrosis of the limbs	369861	Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome
208513	Congenital nonprogressive spinocerebellar ataxia	295024	Congenital pseudoarthrosis of the radius	263435	Congenital smooth muscle hamartoma
269510	Congenital obstructive hydrocephalus	295018	Congenital pseudoarthrosis of the tibia	103908	Congenital sodium diarrhea
440221	Congenital oculomotor nerve palsy	295026	Congenital pseudoarthrosis of the ulna	94068	Congenital spondyloepiphyseal dysplasia
79144	Congenital onychodysplasia	91411	Congenital ptosis	215	Congenital stationary night blindness
79144	Congenital onychodysplasia of the index fingers	2444	Congenital pulmonary airway malformation	75382	Congenital stationary night blindness, Oguchi type
157713	Congenital or early infantile CACH syndrome	280827	Congenital pulmonary airway malformation type 0	99122	Congenital stenosis of the inferior caval vein
2772	Congenital osteogenesis imperfecta - microcephaly - cataracts	280832	Congenital pulmonary airway malformation type 1	99122	Congenital stenosis of the inferior vena cava
465	Congenital PAI-1 deficiency	280840	Congenital pulmonary airway malformation type 2	99122	Congenital stenosis of the IVC
2805	Congenital pancreatic agenesis	280847	Congenital pulmonary airway malformation type 3	3197	Congenital stiff man syndrome
313906	Congenital pancreatic cyst	280854	Congenital pulmonary airway malformation type 4	101068	Congenital stromal corneal dystrophy
139414	Congenital panfollicular nevus	264675	Congenital pulmonary alveolar proteinosis	328	Congenital Stuart factor deficiency
264675	Congenital PAP	2414	Congenital pulmonary lymphangiectasia	141121	Congenital subglottic stenosis
99130	Congenital partial agenesis of pericardium	3161	Congenital pulmonary sequestration	35122	Congenital sucrase-isomaltase deficiency
99124	Congenital partial pulmonary venous return anomaly	3189	Congenital pulmonary valve stenosis	306446	Congenital sucrase-isomaltase deficiency with minimal starch tolerance
295036	Congenital patella dislocation	3188	Congenital pulmonary veins atresia or stenosis	306474	Congenital sucrase-isomaltase deficiency with starch and lactose intolerance
295237	Congenital patella dislocation, bilateral	185	Congenital pulmonary venolobar syndrome	306436	Congenital sucrase-isomaltase deficiency with starch intolerance
295234	Congenital patella dislocation, unilateral	124	Congenital pure red cell aplasia	306462	Congenital sucrase-isomaltase deficiency without starch intolerance
99072	Congenital patent ductus arteriosus aneurysm	295032	Congenital radial head dislocation	306486	Congenital sucrase-isomaltase deficiency without sucrose intolerance
332	Congenital pernicious anemia	97598	Congenital renal artery stenosis	35122	Congenital sucrase-isomaltose malabsorption
626	Congenital pigmented nevus	97598	Congenital renovascular hypoplasia	306446	Congenital sucrase-isomaltose malabsorption with minimal starch tolerance
465	Congenital plasminogen activator inhibitor type 1 deficiency	281190	Congenital reticular ichthyosiform erythroderma	306474	Congenital sucrase-isomaltose malabsorption with starch and lactose intolerance
2907	Congenital poikiloderma with bullae, Weary type	353334	Congenital retinal arteriovenous anastomoses	306436	Congenital sucrase-isomaltose malabsorption with starch intolerance
90042	Congenital polycythemia due to erythropoietin receptor mutation	353334	Congenital retinal arteriovenous communication	306446	Congenital sucrase-isomaltose malabsorption without starch intolerance
124	Congenital PRCA	300337	Congenital retinal detachment	35122	Congenital sucrase-isomaltose intolerance
749	Congenital prekallikrein deficiency	190	Congenital retinal telangiectasia	306446	Congenital sucrase-isomaltose intolerance with minimal starch tolerance
83461	Congenital primary aphakia	178382	Congenital rocker-bottom foot	306474	Congenital sucrase-isomaltose malabsorption with starch and lactose intolerance
79452	Congenital primary lymphedema	290	Congenital rubella syndrome	306436	Congenital sucrase-isomaltose malabsorption with starch intolerance
617	Congenital primary megaloureter	974	Congenital scalp defects with distal limb anomalies	306462	Congenital sucrase-isomaltose malabsorption without starch intolerance
617	Congenital primary megaureter	974	Congenital scalp defects with distal limb reduction anomalies	306446	Congenital sucrase-isomaltose malabsorption with starch and lactose intolerance
238654	Congenital primary megaureter, nonrefluxing and unobstructed form	2301	Congenital short bowel syndrome	306474	Congenital sucrase-isomaltose malabsorption with starch and lactose intolerance
238646	Congenital primary megaureter, obstructed form	1987	Congenital short femur	306436	Congenital sucrase-isomaltose malabsorption with starch intolerance
238650	Congenital primary megaureter, refluxing form	295030	Congenital shoulder dislocation	35122	Congenital sucrose intolerance
327	Congenital proconvertin deficiency	93400	Congenital sialidosis type 2	306446	Congenital sucrose intolerance with minimal starch tolerance
66630	Congenital pseudoarthrosis of clavicle	260305	Congenital sideroblastic anemia	306474	Congenital sucrose intolerance with starch and lactose intolerance
295020	Congenital pseudoarthrosis of the femur			306436	Congenital sucrose intolerance with starch intolerance
295022	Congenital pseudoarthrosis of the fibula			306462	Congenital sucrose intolerance without starch intolerance

→ 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
306486	Congenital sucrose-isomaltose malabsorption without sucrose intolerance	860	Congenitally uncorrected transposition of the great vessels	99099	Cor triatriatum sinistrum
3465	Congenital suprabulbar paresis	216729	Congenitally uncorrected transposition of the great vessels with cardiac malformation	98990	Coralliform cataract
99059	Congenital supravalvular mitral ring	99042	Congenitally uncorrected transposition of the great vessels with coarctation	180118	Cordiform uterus
98948	Congenital symblepharon	99827	Congo fever	366	Cori disease
141214	Congenital syngnathia	99827	Congo hemorrhagic fever	366	Cori-Forbes disease
99856	Congenital syringomyelia	97231	Conjunctivitis lignosa	1051	Corneal anesthesia - deafness - intellectual disability
210576	Congenital temporomandibular joint ankylosis	369929	Conn adenoma with seizures and neurological abnormalities	1490	Corneal dystrophy - perceptive deafness
440221	Congenital third cranial nerve palsy	280210	Connatal PMD	1661	Corneal dystrophy epithelial - short stature
93583	Congenital thrombotic thrombocytopenic purpura	300284	Connective tissue disorder due to LH3 deficiency	98962	Corneal dystrophy Groenouw type I
99125	Congenital total pulmonary venous return anomaly	300284	Connective tissue disorder due to lysyl hydroxylase-3 deficiency	98969	Corneal dystrophy Groenouw type II
858	Congenital toxoplasmosis	→2909	Connective tissue dysplasia, Spellacy type	98961	Corneal dystrophy of Bowman layer type I
141127	Congenital tracheal stenosis	420794	Cono-spondylar dysplasia	98960	Corneal dystrophy of Bowman layer type II
3347	Congenital tracheobronchomegaly	140969	Conorenal syndrome	1490	Corneal dystrophy with progressive deafness
95430	Congenital tracheomalacia	567	Conotruncal anomaly face syndrome	352662	Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome
95459	Congenital tricuspid stenosis	35173	Conradi-Hünermann-Happle syndrome	3177	Corneal-cerebellar syndrome
231013	Congenital trigeminal anesthesia	319651	Constitutional megaloblastic anemia with severe neurologic disease	199	Cornelia de Lange syndrome
210576	Congenital trismus	252202	Constitutional mismatch repair deficiency syndrome	96095	Cornelia de Lange-like syndrome
88629	Congenital tritanopia	295000	Constriction rings syndrome	3194	Corneo-dermato-osseous syndrome
98686	Congenital trochlear nerve palsy	1303	Constrictive bronchiolitis	2041	Coronaro-cardiac fistula
93583	Congenital TTP	369942	Contiguous ABCD1 DXS1357E deletion syndrome	2041	Coronary arterial fistulas
141099	Congenital tubular nose	84142	Continuous muscle fiber activity syndrome	2041	Coronary arterial malformations
99060	Congenital unguarded mitral orifice	725	Continuous spikes and waves during sleep	99085	Coronary artery intramyocardial course
95457	Congenital unguarded tricuspid orifice	725	Continuous spikes and waves during slow-wave sleep	99118	Coronary sinus atresia
1166	Congenital unilateral hypoplasia of depressor anguli oris	1484	Contractures - ectodermal dysplasia - cleft lip/palate	99117	Coronary sinus stenosis
2258	Congenital unilateral pulmonary hypoplasia	436003	Contractures-developmental delay-Pierre Robin syndrome	3338	Corpus callosum agenesis - blepharophimosis - Robin sequence
1864	Congenital valvular dysplasia	314002	Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome	1492	Corpus callosum agenesis - double urinary collecting system
2291	Congenital velopharyngeal incompetence	1487	Cooks syndrome	1496	Corpus callosum agenesis - neuropathy
178382	Congenital vertical talus	231214	Cooley anemia	1553	Corpus callosum agenesis - polysyndactyly
295203	Congenital vertical talus, bilateral	1488	Cooper-Jabs syndrome	50	Corpus callosum agenesis of with chorioretinal abnormality
295201	Congenital vertical talus, unilateral	444092	COPA defect	→3157	Corpus callosum dysgenesis - hypopituitarism
137932	Congenital vocal cord paralysis	397725	CoPAN	275543	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome
216694	Congenitally corrected transposition of the great arteries	2062	Copenhagen syndrome	2318	CORS
216694	Congenitally corrected transposition of the great vessels	99098	Cor triatriatum dexter	1389	Cortical blindness - intellectual disability - polydactyly
2391	Congenitally short costocoracoid ligament	99098	Cor triatriatum dextrum	300570	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation
860	Congenitally uncorrected transposition of the great arteries	99099	Cor triatriatum sinister		
216729	Congenitally uncorrected transposition of the great arteries with cardiac malformation				
99042	Congenitally uncorrected transposition of the great arteries with coarctation				

→ 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
163681	Cortical dysplasia - focal epilepsy syndrome	280847	CPAM type 3	→1394	Craniofacial dysmorphism-skeletal anomalies-intellectual disability syndrome
65683	Cortical dysplasia, Taylor type	280854	CPAM type 4	1798	Craniofacial dysostosis - diaphyseal hyperplasia
3152	Cortical hyperostosis - syndactyly	475	CPD IV	2095	Craniofacial dysostosis - genital, dental, cardiac anomalies
447788	Cortical visual impairment	300564	CPFE	314555	Craniofacial dysplasia-osteopenia syndrome
278	Corticobasal degeneration	91359	CPI	1516	Craniofacial dyssynostosis
199247	Corticosteroid-binding globulin deficiency	2016	CPLS syndrome	1529	Craniofacial-deafness-hand syndrome
54251	Corticosteroid-sensitive aseptic abscess syndrome	759	CPP	293843	Craniofacial-ulnar-renal syndrome
99763	Corticosterone methyloxidase deficiency type I	147	CPS1 deficiency	363705	Craniofaciofrontodigital syndrome
96253	Corticotroph pituitary adenoma	147	CPS1D	1520	Craniofrontonasal dysplasia
141163	Cosack syndrome	156	CPT1A deficiency	1521	Craniofrontonasal dysplasia - Poland anomaly
67047	Costeff optic atrophy syndrome	157	CPT2	228390	Craniofrontonasal dysplasia with alopecia and hypogonadism
67047	Costeff syndrome	228302	CPT2, adult-onset form	1519	Craniofrontonasal dysplasia, Teebi type
3071	Costello syndrome	228305	CPT2, hepatocardiomuscular form	1520	Craniofrontonasal syndrome
1507	Costovertebral segmentation defect - mesomelia	228308	CPT2, lethal systemic form	50814	Craniolenticulosutural dysplasia
1914	Coumarin embryopathy	228302	CPT2, myopathic form	85184	Craniometadiaphyseal dysplasia, wormian bone type
93333	Cousin syndrome	228308	CPT2, neonatal form	1522	Craniometaphyseal dysplasia
1507	COVESDEM syndrome	228305	CPT2, severe infantile form	1524	Craniomicromelic syndrome
101078	Cowchock syndrome	157	CPTII	54595	Craniopharyngioma
201	Cowden disease	228302	CPTII, adult-onset form	63260	Craniorachischisis
201	Cowden syndrome	228305	CPTII, hepatocardiomuscular form	157832	Craniorhiny
99932	Cow's milk hypersensitivity	228308	CPTII, lethal systemic form	1532	Craniosynostosis - alopecia - brain defect
70472	COX deficiency, French-Canadian type	228305	CPTII, myopathic form	85199	Craniosynostosis - anal anomalies - porokeratosis
781	Coxiellosis	3286	CPVT	1530	Craniosynostosis - cataract
1508	Coxoauricular syndrome	35173	CPXD	2872	Craniosynostosis - congenital heart disease - intellectual disability
1509	Coxopodopatellar syndrome	2081	Cramer-Niederdellmann syndrome	1538	Craniosynostosis - Dandy-Walker malformation - hydrocephalus
254920	COXPD2	202	Crandall syndrome	1535	Craniosynostosis - dysmorphism - brachydactyly
254925	COXPD4	1512	Crane-Heise syndrome	1533	Craniosynostosis - fibular aplasia
137908	COXPD5	97339	Cranial dural arteriovenous fistula	171839	Craniosynostosis - hydrocephalus - Arnold-Chiari malformation type I - radioulnar synostosis
254930	COXPD7	97339	Cranial dural arteriovenous malformations	52054	Craniosynostosis - intracranial calcifications
319504	COXPD8	268820	Cranial meningocele	1540	Craniosynostosis - midfacial hypoplasia - foot abnormalities
319509	COXPD9	98919	Cranial variant of GBS	284149	Craniosynostosis and dental anomalies
314637	COXPD10	98919	Cranial variant of Guillain-Barré syndrome	1541	Craniosynostosis, Boston type
324535	COXPD11	420485	Cranio-cervical dystonia with laryngeal and upper-limb involvement	2145	Craniosynostosis, Herrmann-Optiz type
314051	COXPD12	2115	Cranio-facio-digitogenital syndrome	1527	Craniosynostosis, Philadelphia type
319514	COXPD13	1525	Cranio-osteoarthropathy	1541	Craniosynostosis, Warman type
319519	COXPD14	2053	Craniocarpotarsal dysplasia		
319524	COXPD15	2053	Craniocarpotarsal dystrophy		
352563	COXPD16	7	Craniocerebellocardiac dysplasia		
369913	COXPD17	1513	Craniodiaphyseal dysplasia		
420728	COXPD20	1514	Craniodigital syndrome - intellectual disability		
420733	COXPD21	1515	Cranioectodermal dysplasia		
444013	COXPD23	2099	Craniofacial and osseous defects - intellectual disability		
444458	COXPD24	85168	Craniofacial conodysplasia		
447954	COXPD25	1777	Craniofacial dysmorphism - coloboma - corpus callosum agenesis		
2444	CPAM				
280827	CPAM type 0				
280832	CPAM type 1				
280840	CPAM type 2				

→ 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
1528	Craniotelencephalic dysplasia	2052	Cryptophthalmos-syndactyly syndrome	329324	Cutaneous hemangioma with muscle or bone atrophy
2095	Cranofacial dysostosis-hypertrichosis-hypoplasia of labia majora syndrome	1548	Cryptorchidism - arachnodactyly - intellectual disability	889	Cutaneous hypersensitivity vasculitis
75373	CRAPB	1549	Cryptosporidiosis	178475	Cutaneous infectious botulism
275543	CRASH syndrome	357329	Cryptosporidiosis - chronic cholangitis - liver disease	423717	Cutaneous larva migrans
184	CRBM	98967	Crystalline stromal dystrophy	889	Cutaneous leukocytoclastic angitis
71	CRD	101068	CSCD	889	Cutaneous leukocytoclastic vasculitis
52503	Creatine transporter deficiency	443079	CSCR	79455	Cutaneous local mastocytoma
99854	Cree leukoencephalopathy	35122	CSID	79490	Cutaneous lymphangioma circumscriptum
504	Creeping myiasis	306446	CSID with minimal starch tolerance	79455	Cutaneous mastocytoma
280569	Crescentic glomerulonephritis	306474	CSID with starch and lactose intolerance	90395	Cutaneous mucinosis of infancy
90290	CREST syndrome	306436	CSID with starch intolerance	79140	Cutaneous neuroendocrine carcinoma
204	Creutzfeldt-Jakob disease	306462	CSID without starch intolerance	439729	Cutaneous PAN
281	Cri du chat syndrome	306486	CSID without sucrose intolerance	439729	Cutaneous periarteritis nodosa
281190	CRIE	1465	CSS	2881	Cutaneous photosensitivity - lethal colitis
205	Crigler-Najjar syndrome	100008	CST3-related amyloidosis	439729	Cutaneous polyarteritis nodosa
79234	Crigler-Najjar syndrome type 1	329217	CSVT	451607	Cutaneous pseudolymphoma
79235	Crigler-Najjar syndrome type 2	725	CSWS	889	Cutaneous small vessel vasculitis
99827	Crimean hemorrhagic fever	725	CSWSS syndrome	178475	Cutaneous toxin-mediated botulism
99827	Crimean-Congo hemorrhagic fever	70591	CTEPH	1555	Cutis gyrata - acanthosis nigricans - craniostenosis
1545	Crisponi syndrome	436159	CTLA-4 haploinsufficiency with autoimmune infiltration disease	2962	Cutis laxa - corneal clouding - intellectual disability
1461	Criss-cross atrioventricular relationships	247525	CTLN1	228285	Cutis laxa acquisita
1461	Criss-cross heart	247585	CTLN2	221145	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies
891	Criswick-Schepens syndrome	909	CTX	171719	Cutis laxa-Marfanoid syndrome
313838	CRMCC	158	CUD	1556	Cutis marmorata telangiectatica congenita
324964	CRMO	→3157	Culler-Jones syndrome	→35722	Cutis verticis gyrata - intellectual disability
1380	Crome syndrome	413693	Curariform drugs toxicity	5	→35722 Cutis verticis gyrata - retinitis pigmentosa - neurosensory deafness
2930	Cronkhite-Canada syndrome	3207	Curatolo-Cilio-Pessagno syndrome	→35722	Cutis verticis gyrata - retinitis pigmentosa - neurosensory hearing loss
2719	Cross syndrome	98960	Curly fiber corneal dystrophy	→35722	Cutis verticis gyrata - retinitis pigmentosa - sensorineural deafness
2935	Crossed polydactyly	→1071	Curly hair - ankyloblepharon - nail dysplasia syndrome	5	→35722 Cutis verticis gyrata - retinitis pigmentosa - sensorineural hearing loss
2935	Crossed polysyndactyly	307766	Curly hair-acral keratoderma-caries syndrome	→35722	Cutis verticis gyrata - thyroid aplasia - intellectual disability
439881	Croupous bronchitis	1525	Currarino disease	3327	Cutler-Bass-Romshe syndrome
207	Crouzon craniofacial dysostosis	1525	Currarino idiopathic osteoarthropathy	1572	CVID
207	Crouzon disease	1552	Currarino syndrome	306692	Cyanide-induced parkinsonism
93262	Crouzon syndrome - acanthosis nigricans	1552	Currarino triad	2686	Cyclic neutropenia
93262	Crouzon-dermoskeletal syndrome	640	Current pressure-sensitive neuropathy	228379	Cyclosporine-induced folliculodystrophy
2905	Crow-Fukase syndrome	952	Curry-Hall syndrome	210	Cyclosporiasis
290	CRS	1553	Curry-Jones syndrome	79493	CYLD cutaneous syndrome
3421	CRV	96253	Cushing disease	171886	Cylindrical spirals myopathy
411527	CRVO	99889	Cushing syndrome due to ectopic ACTH secretion		
91139	Cryoglobulinemia type 1	189427	Cushing syndrome due to macronodular adrenal hyperplasia		
91138	Cryoglobulinemic vasculitis	53721	Cutaneomeningo spinal angiomatosis		
1546	Cryptococcosis	2451	Cutaneous and mucosal venous malformation		
2032	Cryptogenic fibrosing alveolitis	280779	Cutaneous collagenous vasculopathy		
163708	Cryptogenic late-onset epileptic spasms				
1302	Cryptogenic organizing pneumonia				
1547	Cryptomicrotia - brachydactyly - excess fingertip arch				
1547	Cryptomicrotia-brachydactyly 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
90795	CYP11B1 deficiency	941	D-glyceric aciduria	→23156 8	DDEB, Pasini type
2674	Cyprus facial-neuromusculoskeletal syndrome	2134	D-HUS	231568	DDEB-gen
212	Cystathionase deficiency	93581	D-HUS with anti-factor H antibodies	99970	DDLS
212	Cystathione gamma - lyase deficiency	93578	D-HUS with B factor anomaly	79499	DDOD syndrome
394	Cystathionine beta-synthase deficiency	93575	D-HUS with C3 anomaly	52368	DDON syndrome
212	Cystathioninuria	357008	D-HUS with DGKE deficiency	300536	DDOST-CDG
100008	Cystatin amyloidosis	93579	D-HUS with H factor anomaly	2962	De Barsy syndrome
400	Cystic echinococcosis	93580	D-HUS with I factor anomaly	1130	De Die-Smulders-Vles-Fryns syndrome
586	Cystic fibrosis	93576	D-HUS with MCP/CD46 anomaly	1598	De Grouchy syndrome
2575	Cystic fibrosis - gastritis - megaloblastic anemia	217023	D-HUS with thrombomodulin anomaly	→782	De Hauwere syndrome
2111	Cystic hamartoma of lung and kidney	1146	DA1	→782	De Hauwere-Chitty syndrome
79486	Cystic hygroma	1146	DA1A	56304	De la Chapelle dysplasia
85136	Cystic leukoencephalopathy without megalencephaly	329457	DA5D	393	De la Chapelle syndrome
229	Cystic medial necrosis of aorta	1495	Da Silva syndrome	3157	De Morsier syndrome
1560	Cysticercosis	251515	DA10	244275	De novo thrombotic microangiopathy after kidney transplantation
213	Cystinosis	1562	Dacryocystitis - osteopoikilosis	→910	De Sanctis-Cacchione syndrome
214	Cystinuria	141083	Dacryocystocele	1570	De Smet-Fabry-Fryns syndrome
214	Cystinuria - lysinuria	2186	Daentl-Townsend-Siegel syndrome	33355	De Vaal disease
93612	Cystinuria type A	1563	Dahlberg syndrome	71277	De Vivo disease
93613	Cystinuria type B	1563	Dahlberg-Borer-Newcomer syndrome	3214	Deaf blind hypopigmentation syndrome, Yemenite type
75381	Cystoid macular dystrophy	2181	Daish-Hardman-Lamont syndrome	3217	Deafness - small bowel diverticulosis - neuropathy
180261	Cystosarcoma phyllode	275523	DALD	2663	Deafness - cataracts - skeletal anomalies
180261	Cystosarcoma phylloide	1183	Dancing eye syndrome	52368	Deafness - dystonia - optic neuropathy syndrome
70472	Cytochrome C oxidase deficiency, French-Canadian type	1183	Dancing eye-dancing feet syndrome	3232	Deafness - ear malformation - facial palsy
70472	Cytochrome oxidase deficiency, Saguenay-Lac-Saint-Jean type	1564	Dandy-Walker malformation - facial hemangioma	3220	Deafness - enamel hypoplasia - nail defects
95702	Cytomegalic congenital adrenal hypoplasia	1566	Dandy-Walker malformation - postaxial polydactyly	254898	Deafness - encephaloneuropathy - obesity - valvulopathy
137698	Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	2091	Daneman-Davy-Mancer syndrome	3218	Deafness - epiphyseal dysplasia - short stature
94087	Cytophagic histiocytic panniculitis	34587	Danon disease	3224	Deafness - genital anomalies - metacarpal and metatarsal synostosis
137678	Czech dysplasia, metatarsal type	99645	Dappled diaphyseal dysplasia	90646	Deafness - hypogonadism
2736	Czeizel syndrome	218	Darier disease	85321	Deafness - intellectual disability, Martin-Probst type
2917	Czeizel-Brooser syndrome	316	Darier-Gottron disease	3226	Deafness - lymphedema - leukemia
2437	Czeizel-Losonci syndrome	218	Darier-White disease	2408	Deafness - nephritis - ano-rectal malformation
2953	D4ST1-deficient EDS	390	Darling disease	3230	Deafness - oligodontia
2953	D4ST1-deficient Ehlers-Danlos syndrome	293978	DAVID syndrome	→52368	Deafness - opticoacoustic nerve atrophy - dementia
90038	D+HUS	75565	Davies disease	123	Deafness - pili torti - hypogonadism
356978	D,L-2-HGA	2806	Dawson's encephalitis	3219	Deafness - skeletal dysplasia - coarse face with full lips
356978	D,L-2-hydroxyglutaric acidemia	2143	DBS/FOAR syndrome	3219	Deafness - skeletal dysplasia - lip granuloma
356978	D,L-2-hydroxyglutaric aciduria	1775	DC		
79315	D-2-HGA	79456	DCM		
79315	D-2-hydroxyglutaric acidemia	66634	DCMA syndrome		
79315	D-2-hydroxyglutaric aciduria	75381	DCMD		
93599	D-glycerate dehydrogenase deficiency	1653	DD		
941	D-glycerate kinase deficiency	99789	DD-I		
941	D-glyceric aciduria	99791	DD-II		
		→23156 8	DDEB, Cockayne-Touraine type		
		231568	DDEB, generalized		
		231568	DDEB, Pasini and Cockayne-Touraine types		

→ 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
3237	Deafness - symphalangism syndrome, Hermann type	250989	Del(1)(q21)	261190	Del(15)(q14)
		250999	Del(1)(q41q42)	94065	Del(15)(q24)
3221	Deafness - thyroid hormone resistance	238769	Del(1)(q44)	261211	Del(16)(p11.2p12.2)
3239	Deafness - vitiligo - achalasia	293948	Del(1)p(21.3)	261236	Del(16)(p13.11)
90024	Deafness with labyrinthine aplasia, microtia, and microdontia	363680	Del(2)(p13.2)	352629	Del(16)(q24.1)
		261349	Del(2)(p15p16.1)	261250	Del(16)(q24.3)
3241	Deafness-craniofacial syndrome	163693	Del(2)(p21)	97685	Del(17)(q11)
94064	Deafness-infertility syndrome	369881	Del(2)(p21) without cystinuria	261265	Del(17)(q12)
3231	Deafness-onychodystrophy syndrome	228402	Del(2)(q23.1)	363958	Del(17)(q21.31)
		1617	Del(2)(q24)	261279	Del(17)(q23.1q23.2)
79500	Deafness-onychodystrophy-osteodystrophy-intellectual disability syndrome	251014	Del(2)(q31.1)	254346	Del(19)(p13.12)
		251019	Del(2)(q32)	357001	Del(19)(p13.13)
		251019	Del(2)(q32q33)	217346	Del(19)(q13.11)
79500	Deafness-onychoosteodystrophy-intellectual disability syndrome	251028	Del(2)(q33.1)	261295	Del(20)(p12.3)
→2697	Deal-Barrat-Dillon syndrome	1001	Del(2)(q37)	313781	Del(20)(p13)
158673	DEB, acral	1621	Del(3)(q13)	444051	Del(20)(q11.2)
79411	DEB, bullous dermolysis of the newborn	356947	Del(3)(q26q27)	261311	Del(20)(q13.33)
		397695	Del(3)(q27.3)	261323	Del(21)(q22.11q22.12)
89843	DEB, pruriginosa	65286	Del(3)(q29)	268261	Del(21)(q22.13q22.2)
158673	DEB-ac	435638	Del(3)p(25.3)	96123	Del(22)
79411	DEB-BDN	238750	Del(4)(q21)	261476	Del(X)(p21)
158676	DEB-na	228384	Del(5)(q14.3)	1643	Del(X)(p23)
89843	DEB-Pr	314655	Del(5)(q31.3)	3034	Delayed membranous cranial ossification
79410	DEB-Pt	251046	Del(6)(p22)	3038	Delayed speech - facial asymmetry - strabismus - ear lobe creases
431361	DECР deficiency with hyperlysinemia	171829	Del(6)(q16)	1606	Deletion 1p36
99970	Dedifferentiated liposarcoma	251056	Del(6)(q25)	1606	Deletion 1pter
397587	Deep dermatophytosis	251061	Del(7)(q31)	1001	Deletion 2q37
31150	Defective adenosine triphosphate-binding cassette transporter A1	251066	Del(8)(p11.2)	1001	Deletion 2q37-qter
		251071	Del(8)(p23.1)	281	Deletion 5p
75496	Defective biosynthesis of proteodermatan sulfate	284160	Del(8)(q21.11)	1627	Deletion 5q35
		2496	Del(8)q(13)	904	Deletion 7q11.23
60	Deficiency in Alpa-1-proteinase inhibitor	324313	Del(9)(p13)	284160	Deletion 8q21.11
		352665	Del(9)(q21)	502	Deletion 8q24.1
293978	Deficiency in anterior pituitary function-variable immunodeficiency syndrome	401923	Del(9)(q31.1q31.3)	284169	Deletion 10p11.21p12.31
		284169	Del(10)(p11.21p12.31)	276413	Deletion 10q22.3q23.3
169150	Deficiency of complement of terminal pathway	276413	Del(10)(q22.3q23.3)	893	Deletion 11p13
404546	Deficiency of IL-36R antagonist	444002	Del(11)(q22.2q22.3)	94063	Deletion 12q14
404546	Deficiency of IL-36Ra	2308	Del(11)(q23.3)	289513	Deletion 12q15q21.1
		2308	Del(11)(qter)	1587	Deletion 13q14
158	Deficiency of plasma-membrane carnitine transporter	313884	Del(12)(p12.1)	1590	Deletion 13q32
		280325	Del(12)(p13.33)	1600	Deletion 18q
679	Degos disease	94063	Del(12)(q14)	96123	Deletion 22
315	Degos genodermatoses "en cocardes"	289513	Del(12)(q15)(q21.1)	1647	Delleman syndrome
1578	Dehydratase deficiency	412035	Del(13)(q12.3)	1647	Delleman-Oorthuys syndrome
3202	Dehydrated hereditary stomatocytosis	1587	Del(13)(q14)	79101	Delta1-pyrroline-5-carboxylate dehydrogenase deficiency
		96168	Del(13)(q34)	35664	Delta-1-pyrroline 5-carboxylate synthetase deficiency
64748	Dejerine-Sottas syndrome	261120	Del(14)(q11.2)	231237	Delta-beta-thalassemia
2318	Dekaban-Arima syndrome	261144	Del(14)(q12)	219	Delta-sarcoglycanopathy
1627	Del (5)(q35)	264200	Del(14)(q22q23)	168782	Dementia Infantilis
1627	Del (5)(qter)	401935	Del(14)(q24.1q24.3)		
401986	Del(1)(p31p32)	261183	Del(15)(q11.2)		
1606	Del(1)(p36)	199318	Del(15)(q13.3)		

→ 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
97353	Dementia pugilistica	1659	Dermatoleukodystrophy	329195	Developmental delay with autism spectrum disorder and gait instability
283	Demodicidosis	221	Dermatomyositis	79134	Developmental delay-epilepsy-neonatal diabetes syndrome
283	Demodicosis	1657	Dermatoosteolysis, Kirghizian type	99989	Developmental delay-epilepsy-neonatal diabetes syndrome, intermediate form
314451	Demons-Meigs syndrome	86920	Dermopathia pigmentosa reticularis	363444	Developmental delay-microcephaly-facial dysmorphism syndrome, Hutterite type
79134	DEND syndrome	36426	Dermatostomatitis, Stevens Johnson type	79107	Developmental malformations - deafness - dystonia
86903	Dendritic cell sarcoma not otherwise specified	1660	Dermo-odonto dysplasia	209908	Developmental verbal dyspraxia
99828	Dengue fever	79149	Dermochondrocorneal dystrophy	71211	Devic disease
99828	Dengue virus infection	141051	Dermoid cyst of the face	→3464	Devriendt-Legius-Fryns syndrome
2109	Dennis-Fairhurst-Moore syndrome	141046	Dermoid cyst of the neck	1014	Devriendt-Vandenberge-Fryns syndrome
93571	Dense deposit disease	99688	Dermotrichic syndrome	403	Dexamethasone-sensitive hypertension
1652	Dent disease	1916	DES embryofetopathy	1666	Dextrocardia
93622	Dent disease type 1	1916	DES syndrome	→244	Dextrocardia - bronchiectasis - sinusitis
93623	Dent disease type 2	1425	Desbuquois dysplasia	99828	DF
1652	Dent syndrome	1425	Desbuquois syndrome	383	DFNX2
2095	Dental and eye anomalies-patent ductus arteriosus-normal intelligence syndrome	163703	DESC syndrome	31112	DFSP
1077	Dental ankylosis	228123	Desert fever	49042	DGI
101	Dentatorubral pallidoluysian atrophy	228123	Desert rheumatism	49042	DGI without OI
101	Dentatorubropallidoluysian atrophy	98909	Desmin-related myofibrillar myopathy	166260	DGI-2
1653	Dentin dysplasia	84132	Desmin-related myopathy with Mallory body-like inclusions	373	DGSX
99792	Dentin dysplasia - sclerotic bones	98909	Desminopathy	319651	DHFR deficiency
314721	Dentin dysplasia type 1 with microdontia and shape anomalies	873	Desmoid tumor	139518	dHMN1
99789	Dentin dysplasia type I	873	Desmoid type fibromatosis	139525	dHMN2
99791	Dentin dysplasia type II	251940	Desmoplastic infantile astrocytoma/ganglioglioma	139547	dHMN3 and dHMN4
49042	Dentinogenesis imperfecta	83469	Desmoplastic small round cell tumor	139536	dHMN5
71267	Dentinogenesis imperfecta - short stature - hearing loss - intellectual disability	251863	Desmoplastic/nodular medulloblastoma	100998	dHMN5B
166260	Dentinogenesis imperfecta type 2	35107	Desmosterolosis	98920	dHMN6
166265	Dentinogenesis imperfecta type 3	98852	Desquamative interstitial pneumonia	139589	dHMN7
49042	Dentinogenesis imperfecta without osteogenesis imperfecta	158014	Destombes-Rosai-Dorfman disease	357043	dHMN with upper motor neuron signs
166260	Dentinogenesis imperfecta, Shields type 2	163703	Devastating epileptic encephalopathy in school-aged children	139552	dHMNJ
166265	Dentinogenesis imperfecta, Shields type 3	313892	Developmental and speech delay due to SOX5 deficiency	75376	DHRD
77295	Dentoleukoencephalopathy	163988	Developmental delay - deafness, Hildebrand type	49042	DI
228423	Dendritic cell, monocyte, B and NK lymphoid deficiency	2101	Developmental delay - hypotonia - extremities hypertrophy	166260	DI-2
220	Denys-Drash syndrome	79157	Developmental delay due to 2-methylbutyryl-CoA dehydrogenase deficiency	251940	DIA/DIG
3177	Der Kaloustian-Jarudi-Khoury syndrome	289307	Developmental delay due to ALDH6A1 deficiency	3464	Diabetes - hypogonadism - deafness - intellectual disability
3270	Der Kaloustian-McIntosh-Silver syndrome	289307	Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	3463	Diabetes insipidus - diabetes mellitus - optic atrophy - deafness
369950	Der(8)t(8;12)	289307	Developmental delay due to MMSDH deficiency	1926	Diabetic embryopathy
96170	Der(22)t(11;22) syndrome	329195	Developmental delay with ASD and gait instability	85446	Dialysis-related amyloidosis
36397	Dercum's disease	329195	Developmental delay with ASD and gait instability	85446	Dialysis-related arthropathy
1656	Dermatitis herpetiformis			275523	Dianzani autoimmune lymphoproliferative disease
1266	Dermato-cardio-skeletal syndrome, Borromeo type			66637	Diaphanospodylodystostosis
31112	Dermatofibrosarcoma protuberans			255182	Diaphorase deficiency

→ 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
2140	Diaphragmatic agenesis	220393	Diffuse cutaneous systemic sclerosis	2394	Dihydrolipoamide dehydrogenase deficiency
2141	Diaphragmatic defect - limb deficiency - skull defect	2199	Diffuse erythrodermic palmoplantar keratoderma, Voerner type	255182	Dihydrolipoyl dehydrogenase deficiency
2059	Diaphragmatic hernia - abnormal face - distal limb anomalies	2199	Diffuse erythrodermic palmoplantar keratoderma, Vörner type	79244	Dihydrolipoyllysine-residue acetyltransferase component of pyruvate dehydrogenase complex deficiency
2143	Diaphragmatic hernia-exomphalos-hypertelorism syndrome	702	Diffuse familial brain sclerosis	226	Dihydropteridine reductase deficiency
2143	Diaphragmatic hernia-hypertelorism-myopia-deafness syndrome	3165	Diffuse fasciitis with eosinophilia	38874	Dihydropyrimidinase deficiency
98920	Diaphragmatic spinal muscular atrophy	300849	Diffuse large B-cell lymphoma of the central nervous system	1675	Dihydropyrimidine dehydrogenase deficiency
404521	Diaphragmatic spinal muscular atrophy type 2	300888	Diffuse large B-cell lymphoma with chronic inflammation	38874	Dihydropyrimidinuria
1802	Diaphyseal dysplasia - anemia	252031	Diffuse leptomeningeal melanocytosis	99102	Dilatation of the left appendage
85182	Diaphyseal medullary stenosis - bone malignancy	141209	Diffuse lymphangioma	99102	Dilatation of the left auricle
85182	Diaphyseal medullary stenosis - malignant fibrous histiocytoma	141209	Diffuse lymphangiomatosis	99101	Dilatation of the right atrial appendage
103909	Diarrhea-vomiting due to trehalase deficiency	141209	Diffuse lymphatic malformation	99101	Dilatation of the right atrial auricle
97282	Diarrheogenic islet cell tumor	168811	Diffuse malignant peritoneal mesothelioma	2229	Dilated cardiomyopathy - hypergonadotropic hypogonadism
1671	Diastematomyelia	2123	Diffuse neonatal hemangiomatosis	66634	Dilated cardiomyopathy with ataxia
628	Diastrophic dwarfism	86918	Diffuse palmoplantar hyperkeratosis-acrocyanosis syndrome	231111	DILE
628	Diastrophic dysplasia	369999	Diffuse palmoplantar keratoderma with painful fissures	243343	Dimethylglycine dehydrogenase deficiency
276603	Diazoxide-resistant focal hyperinsulinism due to Kir6.2 deficiency	86918	Diffuse palmoplantar keratoderma-acrocyanosis syndrome	→3157	Dincsoy-Salih-Patel syndrome
276598	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	171700	Diffuse panbronchiolitis	314002	Dinno syndrome
2195	Dicarboxylic aminoaciduria	71274	Diffuse peritoneal leiomyomatosis	1493	Dionisi-Vici-Sabett-Gambarara syndrome
284343	DICER1 syndrome	66627	Diffuse-type GCT	227	Diphallia
180086	Didelphys uterus	66627	Diffuse-type giant cell tumor	1679	Diphtheria
3463	DIDMOAD syndrome	567	DiGeorge sequence	128	Diphyllolobothriasis
370046	Didymosis aplasticosebacea	567	DiGeorge syndrome	1681	Diprosopia
1672	Diencephalic cachexia	238	Digestive duplication	1756	Dipygus
1672	Diencephalic syndrome	141071	Digestive duplication cyst of the tongue	210115	DIRA
1672	Diencephalic syndrome of childhood	352487	Digital anomalies - intellectual disability - short stature	166291	Dirofilariasis
1672	Diencephalic syndrome of emaciation	1305	Digital anomalies with short palpebral fissures and atresia of esophagus or duodenum	94064	DIS
319192	Diencephalic-mesencephalic junction dysplasia	391641	Digital anomalies with short palpebral fissures and atresia of esophagus or duodenum type 1	35122	Disaccharide intolerance
1916	Diethylstilbestrol embryofetopathy	31828	Digitalis poisoning	306446	Disaccharide intolerance with minimal starch tolerance
1916	Diethylstilbestrol syndrome	→79500	Digitorenocerebral syndrome	306474	Disaccharide intolerance with starch and lactose intolerance
146	Differentiated thyroid carcinoma	1146	Digitotalar dysmorphism	306436	Disaccharide intolerance with starch intolerance
90060	Diffuse alveolar hemorrhage	294990	Digits 2-5 hypodactyly	306462	Disaccharide intolerance without starch intolerance
324	Diffuse angiokeratoma	295114	Digits 2-5 hypodactyly, bilateral	306486	Disaccharide intolerance without sucrose intolerance
251595	Diffuse astrocytoma	973	Digits 2-5 hypodactyly, unilateral	90281	Discoid lupus erythematosus
404437	Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	294990	Digits 2-5 oligodactyly	216694	Discordant ventriculoarterial and atrioventricular connections
79456	Diffuse cutaneous maculopapulous mastocytosis	295114	Digits 2-5 oligodactyly, bilateral	99052	Discrete fibromuscular subaortic stenosis
79456	Diffuse cutaneous mastocytosis	973	Digits 2-5 oligodactyly, unilateral	99051	Discrete fixed membranous subaortic stenosis
220393	Diffuse cutaneous systemic scleroderma	319651	Dihydrofolate reductase deficiency		
		79244	Dihydrolipoamide acetyltransferase component of pyruvate dehydrogenase complex deficiency		

→ 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
90394	Discrete papular lichen myxedematosus	329457	Distal arthrogryposis type 5 without ophthalmoparesis	1705	Distal duplication 14q
139420	Disease-associated transverse myelitis	329457	Distal arthrogryposis type 5 without ophthalmoplegia	1707	Distal duplication 15q
210272	Disembarkment syndrome	329457	Distal arthrogryposis type 5D	96078	Distal duplication 16p
2412	Dislocation of the hip - dysmorphism	1144	Distal arthrogryposis type 6	96106	Distal duplication 16q
8	Disomy Y	3377	Distal arthrogryposis type 7	3379	Distal duplication 17q
2983	Disorder of sex development - intellectual disability	65743	Distal arthrogryposis type 8	1716	Distal duplication 18q
345	Dissecting cellulitis of the scalp	115	Distal arthrogryposis type 9	1717	Distal duplication 19q
54251	Disseminated aseptic abscesses	251515	Distal arthrogryposis type 10	96107	Distal duplication 20q
1306	Disseminated dermatofibrosis with osteopoikilosis	376	Distal arthrogryposis type IIA	96109	Distal duplication 22q
397587	Disseminated granulomatous dermatophytosis	1154	Distal arthrogryposis type IIB	1762	Distal duplication Xq
141209	Disseminated lymphangioma	65720	Distal arthrogryposis type IID	139518	Distal hereditary motor neuropathy type 1
141209	Disseminated lymphangiomatosis	1154	Distal arthrogryposis with ophthalmoplegia	139525	Distal hereditary motor neuropathy type 2
141209	Disseminated lymphatic malformation	254351	Distal del(7)(q11.23)	139547	Distal hereditary motor neuropathy type 3 and type 4
228264	Disseminated nevus anelasticus	261222	Distal del(16)(p11.2)	139536	Distal hereditary motor neuropathy type 5
71274	Disseminated peritoneal leiomyomatosis	319171	Distal del(17)(p13.1)	100998	Distal hereditary motor neuropathy type 5B
79152	Disseminated superficial actinic porokeratosis	261257	Distal del(17)(p13.3)	98920	Distal hereditary motor neuropathy type 6
1620	Distal 3p deletion	261330	Distal del(22)(q11.2)	139589	Distal hereditary motor neuropathy type 7
1627	Distal 5q deletion	36367	Distal deletion 1q	357043	Distal hereditary motor neuropathy with upper motor neuron signs
254351	Distal 7q11.23 microdeletion syndrome	280	Distal deletion 4p	139552	Distal hereditary motor neuropathy, Jerash type
261102	Distal 7q11.23 microduplication syndrome	96145	Distal deletion 4q	1307	Distal limb deficiencies - micrognathia syndrome
1580	Distal 10p deletion	96125	Distal deletion 6p	36367	Distal monosomy 1q
1590	Distal 13q deletion	96126	Distal deletion 7p	1620	Distal monosomy 3p
1596	Distal 15q deletion syndrome	1636	Distal deletion 7q36	280	Distal monosomy 4p
261222	Distal 16p11.2 microdeletion syndrome	1642	Distal deletion 9p	96145	Distal monosomy 4q
319171	Distal 17p13.1 microdeletion syndrome	96148	Distal deletion 10q	96125	Distal monosomy 6p
261257	Distal 17p13.3 microdeletion syndrome	2308	Distal deletion 11q	96126	Distal monosomy 7p
1597	Distal 17q deletion	280325	Distal deletion 12p	254351	Distal monosomy 7q11.23
261330	Distal 22q11.2 microdeletion syndrome	96149	Distal deletion 12q	1636	Distal monosomy 7q36
261337	Distal 22q11.2 microduplication syndrome	96168	Distal deletion 13q34	1642	Distal monosomy 9p
63273	Distal ABD-filaminopathy	96150	Distal deletion 14q	1580	Distal monosomy 10p
399096	Distal anoctaminopathy	96129	Distal deletion 19p	96148	Distal monosomy 10q
178400	Distal anterior compartment myopathy	96152	Distal deletion 20q	2308	Distal monosomy 11q
1146	Distal arthrogryposis type 1	261102	Distal dup(7)(q11.23)	280325	Distal monosomy 12p
2053	Distal arthrogryposis type 2A	261337	Distal dup(22)(q11.2)	96149	Distal monosomy 12q
1147	Distal arthrogryposis type 2B	293939	Distal dup(X)(q)(28)	1590	Distal monosomy 13q
376	Distal arthrogryposis type 3	96069	Distal duplication 1p36	96150	Distal monosomy 14q
65720	Distal arthrogryposis type 4	96070	Distal duplication 2p	1596	Distal monosomy 15q
1154	Distal arthrogryposis type 5	96094	Distal duplication 2q	261222	Distal monosomy 16p11.2
		96071	Distal duplication 3p	261257	Distal monosomy 17p13.3
		96072	Distal duplication 4p	1597	Distal monosomy 17q
		96096	Distal duplication 4q	96129	Distal monosomy 19p13.3
		96097	Distal duplication 5q	96152	Distal monosomy 20q
		1745	Distal duplication 6p	261330	Distal monosomy 22q11.2
		96098	Distal duplication 6q	59135	Distal myopathy type 1
		96074	Distal duplication 7p		
		96100	Distal duplication 8q		
		96101	Distal duplication 9q		
		96102	Distal duplication 10q		
		96103	Distal duplication 11q		
		96105	Distal duplication 13q		

→ 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
399086	Distal myopathy type 3	3379	Distal trisomy 17q	276580	Dominant KATP hyperinsulinism due to Kir6.2 deficiency
178400	Distal myopathy with anterior tibial onset	1716	Distal trisomy 18q	75376	Dominant radial drusen
34521	Distal myopathy with early respiratory muscle involvement	1717	Distal trisomy 19q	90035	Donath-Landsteiner hemolytic anemia
63273	Distal myopathy with posterior leg and anterior hand involvement	96107	Distal trisomy 20q	90035	Donath-Landsteiner syndrome
602	Distal myopathy with rimmed vacuoles	96109	Distal trisomy 22q	2143	Donnai-Barrow syndrome
600	Distal myopathy with vocal cord weakness	261337	Distal trisomy 22q11.2	508	Donohue syndrome
602	Distal myopathy, Nonaka type	293939	Distal trisomy Xq28	79500	DOOR syndrome
609	Distal myopathy, Udd type	293939	Distal Xq28 microduplication syndrome	79500	DOORS syndrome
603	Distal myopathy, Welander type	98920	Distal-HMN type 6	1942	Doose syndrome
98911	Distal myotilinopathy	→33001	Distichiasis - congenital heart defects - peripheral vascular anomalies	70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency
2776	Distal osteolysis - short stature - intellectual disability	1916	Distilbene embryofetopathy	230	Dopamine beta-hydroxylase deficiency
18	Distal renal tubular acidosis	1685	Distomatosis	98907	Dorfman-Chanarin disease
→40204 1	Distal renal tubular acidosis type 1b	1685	Distomiasis	3426	DORV
→40204 1	Distal renal tubular acidosis type 1c	404546	DITRA	423712	DORV with atrioventricular septal defect, pulmonary stenosis, heterotaxy
93610	Distal renal tubular acidosis with anemia	99099	Divided left atrium	99046	DORV with non-committed subpulmonary VSD
139525	Distal spinal muscular atrophy type 2	99098	Divided right atrium	423693	DORV with subaortic or doubly committed VSD
139547	Distal spinal muscular atrophy type 3	91131	DK1-CDG	99043	DORV with subaortic or doubly committed VSD with pulmonary stenosis
206580	Distal spinal muscular atrophy type 4	3439	DK phocomelia syndrome	99045	DORV with subpulmonary VSD
139536	Distal spinal muscular atrophy type 5	1775	DKC	99043	DORV, Fallot type
139589	Distal spinal muscular atrophy with vocal cord paralysis	300849	DLBCL of the CNS	99045	DORV-TGA
3248	Distal symphalangism	300888	DLBCL with chronic inflammation	869	Double A syndrome
314588	Distal tetrasomy 15q	2394	DLD deficiency	216694	Double discordance
609	Distal titinopathy	252031	DLM	1464	Double inlet left ventricle
96069	Distal trisomy 1p36	221	DM	141091	Double nose
96070	Distal trisomy 2p	273	DM1	3427	Double outlet left ventricle
96094	Distal trisomy 2q	98896	DMD	3426	Double outlet right ventricle
96071	Distal trisomy 3p	243343	DMG dehydrogenase deficiency	423712	Double outlet right ventricle with atrioventricular septal defect, pulmonary stenosis, heterotaxy
96072	Distal trisomy 4p	243343	DMGDH deficiency	→42369 3	Double outlet right ventricle with doubly committed ventricular septal defect
96096	Distal trisomy 4q	602	DMRV	99046	Double outlet right ventricle with non-committed subpulmonary ventricular septal defect
96097	Distal trisomy 5q	99812	DNA ligase IV deficiency	423693	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect
1745	Distal trisomy 6p	251946	DNET	99043	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect with pulmonary stenosis
96098	Distal trisomy 6q	404443	DNMT3A-related overgrowth syndrome	→42369 3	Double outlet right ventricle with subaortic ventricular septal defect
96074	Distal trisomy 7p	251975	DNT of the cerebellum	99046	Double outlet right ventricle with subpulmonary ventricular septal defect
261102	Distal trisomy 7q11.23	1215	DOA+	423693	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect
96100	Distal trisomy 8q	447737	DOCK2 deficiency	99043	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect with pulmonary stenosis
96101	Distal trisomy 9q	217390	DOCK8 immunodeficiency syndrome	→42369 3	Double outlet right ventricle with subaortic ventricular septal defect
96102	Distal trisomy 10q	79322	Dol-P-mannosyltransferase deficiency	99046	Double outlet right ventricle with subpulmonary ventricular septal defect
96103	Distal trisomy 11q	91131	Dolichol kinase deficiency	423693	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect
96105	Distal trisomy 13q	2616	Dolichospondylic dysplasia	99043	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect with pulmonary stenosis
1705	Distal trisomy 14q	86309	Dolichyl-phosphate N-acetylgalactosamine phosphotransferase deficiency	→42369 3	Double outlet right ventricle with subaortic ventricular septal defect
1707	Distal trisomy 15q	3427	DOLV	99046	Double outlet right ventricle with subpulmonary ventricular septal defect
96078	Distal trisomy 16p	231226	Dominant beta-thalassemia	423693	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect
96106	Distal trisomy 16q	75376	Dominant drusen	99043	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect with pulmonary stenosis
		898	Dominant hyaloideoretinal dystrophy of Wagner	→42369 3	Double outlet right ventricle with subaortic ventricular septal defect
		244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis		

→ 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
99045	Double outlet right ventricle with subpulmonary ventricular septal defect	139525	dSMA2	261318	Dup(20p)
99045	Double outlet right ventricle with transposition of the great arteries	139547	dSMA3	1727	Dup(22)(q11)
99043	Double outlet right ventricle, Fallot type	206580	dSMA4	284180	Dup(X)(p22)
3286	Double tachycardia induced by catecholamines	314485	dSMA5	284180	Dup(X)(p22.13p22.2)
3411	Double uterus - hemivagina - renal agenesis	83469	DSRCT	314389	Dup(X)(q12-q13.3)
3411	Double uterus and obstructed hemivagina syndrome	412181	DST-related epidermolysis bullosa simplex	261483	Dup(X)(q27.3q28)
8	Double Y	99789	DTDP1	261344	Duplication 1q
95474	Double-orifice mitral valve	99791	DTDP2	1738	Duplication 4p
79145	Dowling-Degos disease	2639	Du Pan syndrome	1742	Duplication 5p
75376	Doyne honeycomb retinal dystrophy	50817	Duane anomaly - myopathy - scoliosis	264450	Duplication 8p
86309	DPAGT1-CDG	233	Duane retraction syndrome	1752	Duplication 8q
314621	DPG-plus syndrome	233	Duane syndrome	96167	Duplication 8q/deletion 8p
71274	DPL	93293	Duane-radial ray syndrome	236	Duplication 9p
79322	DPM1-CDG	261647	Duane-radial ray syndrome due to a point mutation	1699	Duplication 12p
329178	DPM2-CDG	261638	Duane-radial ray syndrome due to monosomy 20q13	1715	Duplication 18p
263494	DPM3-CDG	234	Dubin-Johnson syndrome	1727	Duplication 22q11.2
231	Dracunculiasis	234	Dubin-Sprinz disease	261318	Duplication of 20p
231	Dracunculosis	235	Dubowitz syndrome	314621	Duplication of the pituitary gland
220	Drash syndrome	98896	Duchenne muscular dystrophy	314621	Duplication of the pituitary gland-plus syndrome
33069	Dravet syndrome	280315	Duct-centric pancreatitis	1738	Duplication of the short arm of chromosome 4
→79500	DRC syndrome	2442	Duncan disease	1742	Duplication of the short arm of chromosome 5
70594	DRD due to SRD	2348	Dunnigan syndrome	236	Duplication of the short arm of chromosome 9
130	Dream disease	→29386	Duodenal and extrahepatic biliary atresia - hypoplastic pancreas - intestinal malrotation	1715	Duplication of the short arm of chromosome 18
139402	DRESS syndrome	1203	Duodenal atresia	237	Duplication of urethra
101	DRPLA	250994	Dup(1)(q21.1)	284180	Duplication Xp22
233	DRS	313947	Dup(2)(q23.1)	3306	Duplication/inversion 15q11
18	dRTA	294026	Dup(2)(q31.1)	97339	Dural sinus malformation
→40204	dRTA type 1b	96095	Dup(3)(q26)	1656	Durhing-Brocq disease
1		329802	Dup(5)(p13)	233	DURS
→40204	dRTA type 1c	228415	Dup(5)(q35)	→33117	Dursun syndrome
93610	dRTA with anemia	314034	Dup(7)(p22.1)	6	
139402	Drug rash with eosinophilia and systemic symptoms	96121	Dup(7)(q11.23)	98984	Dusty cataract
139402	Drug reaction eosinophilic systemic syndrome	251076	Dup(8)(p23.1)	3377	Dutch-Kentucky syndrome
90037	Drug-induced AIHA	228399	Dup(8)(q12)	→969	Dwarfism - stiff joint - ocular abnormalities
90037	Drug-induced autoimmune hemolytic anemia	276422	Dup(10)(q22.3q23.3)	→2616	Dwarfism - tall vertebrae
90157	Drug-induced localized lipodystrophy	300305	Dup(11)p(15.4)	2650	Dwarfism-intellectual disability-eye abnormality syndrome
231111	Drug-induced lupus erythematosus	261229	Dup(14)(q11.2)	1566	DWM with postaxial polydactyly
251325	Drug-induced vasculitis	238446	Dup(15)(q11q13)	239	Dyggve-Melchior-Claussen disease
97368	Drug-related renal tubular dysgenesis	261204	Dup(16)(p11.2p12.2)	2274	Dykes-Markes-Harper syndrome
94086	Drummond syndrome	261243	Dup(16)(p13.11)	296	Dyschondroplasia
33069	DS	96078	Dup(16)(p13.3)	1765	Dyschondrosteosis - nephritis
99887	DS-AMKL	217385	Dup(17)(p13.3)	41	Dyschromatosis symmetrica hereditaria
98920	dSMA1	139474	Dup(17)(q11.2)	241	Dyschromatosis universalis
		261272	Dup(17)(q12)	251946	Dysembryoplastic neuroepithelial tumor
		217340	Dup(17)(q21.31)		
		261290	Dup(17p)		
		447980	Dup(19)(p13.13)		
		363659	Dup(20)(q11.2)		

→ 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
251975	Dysembryoplastic neuroepithelial tumor of cerebellum	293381	Dystrophia Smoldaniensis	1667	Early-onset diabetes mellitus with multiple epiphyseal dysplasia
1766	Dysequilibrium syndrome	79409	Dystrophic epidermolysis bullosa inversa	210571	Early-onset dystonia parkinsonism
99912	Dysgerminomatous germ cell cancer of ovary	89843	Dystrophic epidermolysis bullosa pruriginosa	289266	Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation
3010	Dysharmonic skeletal maturation - muscular fiber disproportion	158676	Dystrophic epidermolysis bullosa, nails only	411986	Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome
1775	Dyskeratosis congenita	256	DYT1	1020	Early-onset familial autosomal dominant Alzheimer disease
3088	Dyskeratosis congenita with bilateral exudative retinopathy	99657	DYT2	256	Early-onset generalized limb-onset dystonia
412	Dyslipidemia type 3	53351	DYT3	256	Early-onset generalized torsion dystonia
1779	Dysmorphism - cleft palate - loose skin	98805	DYT4	88660	Early-onset hypertension with exacerbation in pregnancy
289553	Dysmorphism - conductive hearing loss - heart defect	98808	DYT5a	324290	Early-onset Lafora body disease
1780	Dysmorphism - multiple structural anomalies	101150	DYT5b	441452	Early-onset lamellar cataract
2104	Dysmorphism - pectus carinatum - joint laxity	98806	DYT6	79242	Early-onset multiple carboxylase deficiency
2282	Dysmorphism - short stature - deafness - disorder of sex development	53583	DYT9	289377	Early-onset myopathy with fatal cardiomyopathy
2282	Dysmorphism - short stature - deafness - pseudohermaphroditism	36899	DYT11	439212	Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome
1782	Dysosteosclerosis	71517	DYT12	91492	Early-onset non-syndromic cataract
800	Dysostosis enchondralis metaeipiphysaria, Catel-Hempel type	98807	DYT13	98991	Early-onset nuclear cataract
1798	Dysostosis, Stanescu type	→98808	DYT14	2828	Early-onset Parkinson disease
99082	Dysphagia lusoria	→36899	DYT15	2379	Early-onset parkinsonism - intellectual disability
1822	Dysplasia epiphysealis hemimelica	210571	DYT16	98992	Early-onset partial cataract
168621	Dysplasia of head of femur, Meyer type	98811	DYT18	98993	Early-onset posterior polar cataract
2204	Dysplastic cortical hyperostosis	306734	DYT21	441447	Early-onset posterior subcapsular cataract
65285	Dysplastic gangliocytoma of the cerebellum	420492	DYT23	256	Early-onset primary dystonia
325	Dysprothrombinemia	420485	DYT24	157941	Early-onset prion disease with prominent psychiatric features
2476	Dysraphism - cleft lip/palate - limb reduction defects	2394	E3-deficient maple syrup urine disease	352654	Early-onset progressive neurodegeneration - blindness - ataxia - spasticity
1804	Dyssegmental dysplasia - glaucoma	231249	E-beta-thalassemia	→90340	Early-onset sarcoidosis
156731	Dyssegmental dysplasia, Rolland-Desbuquois type	2970	Eagle-Barret syndrome	96369	Early-onset schizophrenia
1865	Dyssegmental dysplasia, Silverman-Handmaker type	40923	Eales disease	364055	Early-onset severe retinal dystrophy
85198	Dyspondyloenchondromatosis	2554	Ear-patella-short stature syndrome	313772	Early-onset spastic ataxia-neuropathy syndrome
71517	Dystonia 12	1934	Early infantile epileptic encephalopathy	98985	Early-onset sutural cataract
→98808	Dystonia 14	1934	encephalopathy with suppression-bursts	256	Early-onset torsion dystonia
210571	Dystonia 16	369894	Early infantile epileptic encephalopathy without suppression burst	1243	Early-onset vitelliform macular dystrophy
98811	Dystonia 18	1935	Early myoclonic encephalopathy	98890	Early-onset X-linked isolated optic atrophy
420492	Dystonia 23	1935	Early myoclonic encephalopathy with suppression-bursts	98995	Early-onset zonular cataract
420485	Dystonia 24	98988	Early-onset anterior polar cataract	199343	EAST syndrome
256	Dystonia musculorum deformans	98988	Early-onset anterior subcapsular cataract		
412217	Dystonia-aphonia syndrome	1020	Early-onset autosomal dominant Alzheimer disease		
199351	Dystonia-parkinsonism, Paisan-Ruiz type	98815	Early-onset benign childhood occipital epilepsy		
293381	Dystrophia Helsinglandica	98985	Early-onset cataract with Y-shaped suture opacities		
		1177	Early-onset cerebellar ataxia with retained tendon reflexes		
		84132	Early-onset desmin-related myopathy		

→ 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
391320	East Texas bleeding disorder	→1071	Ectodermal dysplasia syndrome, Rapp-Hodgkin type	1900	EDS VIA
83594	Eastern equine encephalitis	69083	Ectodermal dysplasia with natal teeth, Turnpenny type	1899	EDS VII
83594	Eastern equine encephalomyelitis	1809	Ectodermal dysplasia with skin anomalies and intellectual disability	99875	EDS VIIA
1973	Eastman-Bixler syndrome	1816	Ectodermal dysplasia, Berlin type	99876	EDS VIIB
166418	Eating seizures	→1071	Ectodermal dysplasia, Rapp-Hodgkin type	1901	EDS VIIC
86880	EATL	1818	Ectodermal dysplasia, trichodontoonychial type	75392	EDS VIII
79406	EB progressive	1883	Ectodermal dysplasia-sensorineural deafness syndrome	82004	EDS with periventricular heterotopia
79405	EBJ-I	423454	Ectodermal dysplasia-short stature syndrome	300179	EDS with progressive kyphoscoliosis, myopathy, and deafness
319218	Ebola fever	448270	Ectopia Cordis	300179	EDS with progressive kyphoscoliosis, myopathy, and hearing loss
319218	Ebola hemorrhagic fever	1884	Ectopia lenti - chorioretinal dystrophy - myopia	75501	EDS X
319218	Ebola virus disease	1885	Ectopia lenti syndrome	2295	EDS XI
412181	EBS-AR BP230	99889	Ectopic ACTH secreting tumor	2953	EDS, arthrogryposic type
412189	EBS-AR exophilin 5	231632	Ectopic aldosterone-producing tumor	230851	EDS, cardiac valvular type
89838	EBS-AR KRT14	99889	Ectopic Cushing syndrome	287	EDS, classic type
79400	EBS-loc	95496	Ectopic neurohypophysis	230839	EDS, classic-like type
257	EBS-MD	2440	Ectrodactyly	2953	EDS, Kosho type
158681	EBS-migr	→1896	Ectrodactyly - cleft palate	300179	EDS, kyphoscoliotic and hearing loss type
79397	EBS-MP	1896	Ectrodactyly - ectodermal dysplasia - cleft lip/palate	1900	EDS, kyphoscoliotic type
79401	EBS-O	→1896	Ectrodactyly - ectodermal dysplasia without clefting	2953	EDS, musculocontractural type
158684	EBS-PA	1892	Ectrodactyly - polydactyly	1900	EDS, oculoscoliotic type
89839	EBSS	1894	Ectrodactyly - spina bifida - cardiopathy	75496	EDS, progeroid type
1880	Ebstein anomaly of the tricuspid valve	1997	Ectropion inferior - cleft lip and or palate	157965	EDS, spondylocheirodysplastic type
1880	Ebstein malformation	906	Eczema-thrombocytopenia-immunodeficiency syndrome	230845	EDS, vascular-like type
313920	EBV-associated gastric carcinoma	98813	EDA-ID	230857	EDS/OI syndrome
289661	EBV-positive DLBCL of the elderly	247827	EDCS	247820	EDSS
313920	EBVaGC	293936	EDICT syndrome	247820	EDSS1
50944	Eccrine tumors-ectodermal dysplasia	1895	Edinburgh malformation syndrome	247827	EDSS2
199332	ECO syndrome	93308	EDM1	178464	Edström Myopathy
→1896	ECP syndrome	93307	EDM4	3380	Edwards syndrome
99102	Ectasia of the left appendage	93311	EDM5	2668	Edwards-Patton-Dilly syndrome
99102	Ectasia of the left auricle	261	EDMD	322	EEC
99101	Ectasia of the right atrial appendage	98863	EDMD1	1896	EEC syndrome
99101	Ectasia of the right atrial auricle	98853	EDMD2	→1896	EEC syndrome without cleft lip/palate
35737	Ectasic coloboma	98855	EDMD3	1897	EEM syndrome
→1658	Ectodermal dysplasia - absent dermatoglyphs	90309	EDS I	240869	Efavirenz toxicity
140936	Ectodermal dysplasia - acanthosis nigricans	90318	EDS II	357131	Effort subclavian vein thrombosis
→2036	Ectodermal dysplasia - adrenal cyst	285	EDS III	101039	EFMR
1806	Ectodermal dysplasia - blindness	286	EDS IV	2070	EGE
3354	Ectodermal dysplasia - cataracts - kyphoscoliosis	198	EDS IX	183	EGPA
247827	Ectodermal dysplasia - cutaneous syndactyly syndrome	286	EDS type 4	319218	EHF
1897	Ectodermal dysplasia - ectrodactyly - macular dystrophy	75497	EDS V	312	EHK
1812	Ectodermal dysplasia - intellectual disability - central nervous system malformation			230839	Ehlers-Danlos syndrome due to tenascin-X deficiency
158668	Ectodermal dysplasia - skin fragility syndrome			90309	Ehlers-Danlos syndrome type 1
247820	Ectodermal dysplasia - syndactyly syndrome			90318	Ehlers-Danlos syndrome type 2
				285	Ehlers-Danlos syndrome type 3
				286	Ehlers-Danlos syndrome type 4
				75497	Ehlers-Danlos syndrome type 5
				1900	Ehlers-Danlos syndrome type 6A
				1899	Ehlers-Danlos syndrome type 7

→ 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
99875	Ehlers-Danlos syndrome type 7A	75496	Ehlers-Danlos syndrome, progeroid type	221126	Encephaloclastic proliferative vasculopathy
99876	Ehlers-Danlos syndrome type 7B	157965	Ehlers-Danlos syndrome, spondylocheirodysplastic type	2396	Encephalocraniocutaneous lipomatosis
1901	Ehlers-Danlos syndrome type 7C	286	Ehlers-Danlos syndrome, vascular type	3205	Encephalofacial angiomas
75392	Ehlers-Danlos syndrome type 8	230845	Ehlers-Danlos syndrome, vascular-like type	319678	Encephalopathy - hypertrophic cardiomyopathy - renal tubular disease
198	Ehlers-Danlos syndrome type 9	230857	Ehlers-Danlos/osteogenesis imperfecta syndrome	1261	Encephalopathy - intracerebral calcification - retinal degeneration
75501	Ehlers-Danlos syndrome type 10	1902	Ehrlachiosis	1035	Encephalopathy due to beta-mercaptoprolactate-cysteine disulfiduria
2295	Ehlers-Danlos syndrome type 11	820	Ehrmann-Sneddon syndrome	71277	Encephalopathy due to GLUT1 deficiency
286	Ehlers-Danlos syndrome type IV	312	EI	79155	Encephalopathy due to hydroxylkynureninuria
198	Ehlers-Danlos syndrome type IX	1934	EIEE	139406	Encephalopathy due to prosaposin deficiency
82004	Ehlers-Danlos syndrome with periventricular heterotopia	165991	EIHI	833	Encephalopathy due to sulfite oxidase deficiency
75501	Ehlers-Danlos syndrome with platelet dysfunction from fibronectin abnormality	79106	Eiken syndrome	210128	Encephalopathy due to urocanase deficiency
300179	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and deafness	97214	Eisenmenger syndrome	51	Encephalopathy with basal ganglia calcification
300179	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss	317	EKV	51	Encephalopathy with intracranial calcification and chronic lymphocytosis of cerebrospinal fluid
1899	Ehlers-Danlos syndrome, arthrochalasia type	228240	Elastoderma	3205	Encephalotrigeminal angiomas
1899	Ehlers-Danlos syndrome, arthrochalasic type	228243	Elastofibroma dorsi	296	Enchondromatosis
2953	Ehlers-Danlos syndrome, arthrogryposis type	228254	Elastoma	99075	Encircling double aortic arch
230851	Ehlers-Danlos syndrome, cardiac valvular type	79148	Elastosis perforans serpiginosa	100082	Endocrine tumor of anal canal
287	Ehlers-Danlos syndrome, classic type	228236	Elastotic striae	100080	Endocrine tumor of colon
230839	Ehlers-Danlos syndrome, classic-like type	26791	Electron transfer flavoprotein deficiency	100081	Endocrine tumor of rectum
1901	Ehlers-Danlos syndrome, dermatosparaxis type	26791	Electron transfer flavoprotein ubiquinone oxidoreductase deficiency	100079	Endocrine tumor of the appendix
75501	Ehlers-Danlos syndrome, fibronectin-deficient	33445	Elejalde disease	199332	Endocrine-cerebro-osteodysplasia syndrome
75501	Ehlers-Danlos syndrome, fibronectinemic type	221054	Elejalde syndrome	876	Endodermal sinus tumor
285	Ehlers-Danlos syndrome, hypermobile type	289	Ellis Van Creveld syndrome	252006	Endodermal sinus tumor of central nervous system
285	Ehlers-Danlos syndrome, hypermobility type	2516	Ellis-Yale-Winter syndrome	252006	Endodermal sinus tumor of CNS
2953	Ehlers-Danlos syndrome, Kosho type	1997	Elsching syndrome	98974	Endoepithelial corneal dystrophy
300179	Ehlers-Danlos syndrome, kyphoscoliotic and deafness type	96170	Emanuel syndrome	213741	Endometrial adenoid cystic carcinoma
300179	Ehlers-Danlos syndrome, kyphoscoliotic and hearing loss type	439212	EMARDD	213726	Endometrial capillary carcinoma
1900	Ehlers-Danlos syndrome, kyphoscoliotic type	1942	EMAS	213716	Endometrial squamous cell carcinoma
2953	Ehlers-Danlos syndrome, musculocontractural type	3226	Emberger syndrome	213711	Endometrial stromal sarcoma
1900	Ehlers-Danlos syndrome, oculoscoliotic type	1914	Embryofetopathy due to oral anticoagulant therapy	213746	Endometrial transitional cell carcinoma
75392	Ehlers-Danlos syndrome, periodontitis type	180226	Embryonal carcinoma	213721	Endometrial undifferentiated carcinoma
		48736	Embryonal carcinoma of the central nervous system	2022	Endomyocardial fibroelastosis
		48736	Embryonal carcinoma of the CNS	199323	Endophthalmitis
		99757	Embryonal rhabdomyosarcoma	209959	Endophthalmitis phacoanaphylactica
		178315	Embryonal sarcoma of the liver	2790	Endosteal hyperostosis, Worth type
		1664	Embryonal disorganization syndrome	85186	Endosteal sclerosis - cerebellar hypoplasia
		983	Embryonic testicular regression syndrome		
		139431	EMEA		
		98863	Emerinopathy		
		261	Emery-Dreifuss muscular dystrophy		
		1927	Emery-Nelson syndrome		
		83600	Encephalitis lethargica		

→ 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
293936	Endothelial dystrophy-iris hypoplasia-congenital cataract-stromal thinning syndrome	89839	Epidermolysis bullosa simplex superficialis	1825	Epiphyseal dysplasia - hearing loss - dysmorphism
137602	Endotheliitis	2325	Epidermolysis bullosa simplex with anodontia/hypodontia	1824	Epiphyseal dysplasia - microcephaly - nystagmus
1937	Eng-Strom syndrome	158681	Epidermolysis bullosa simplex with circinate migratory erythema	1952	Epiphyseal stippling syndrome - osteoclastic hyperplasia
53540	Enhanced S-cone syndrome	79397	Epidermolysis bullosa simplex with mottled pigmentation	399329	Epiphysiolysis of the hip
60015	Enlarged parietal foramina	257	Epidermolysis bullosa simplex with muscular dystrophy	399329	Epiphysiolysis of the upper femur
83620	Enteric anendocrinosis	158684	Epidermolysis bullosa simplex with pyloric atresia	649	Episkopi blindness
141071	Enteric duplication cyst of the tongue	79396	Epidermolysis bullosa simplex, Dowling-Meara type	79135	Episodic ataxia - vertigo - tinnitus - myokymia
86880	Enteropathy-associated T-cell lymphoma	79396	Epidermolysis bullosa simplex, herpetiformis	37612	Episodic ataxia type 1
86880	Enteropathy-type T-cell lymphoma	79399	Epidermolysis bullosa simplex, Köbner type	97	Episodic ataxia type 2
85438	Enthesitis-related arthritis	79399	Epidermolysis bullosa simplex, Koebner type	79135	Episodic ataxia type 3
1177	EOCA	79401	Epidermolysis bullosa simplex, Ogna type	79136	Episodic ataxia type 4
1177	EOCARR	79400	Epidermolysis bullosa simplex, Weber-Cockayne type	211067	Episodic ataxia type 5
370334	EOE	312	Epidermolytic hyperkeratosis	209967	Episodic ataxia type 6
73247	EoE	312	Epidermolytic ichthyosis	209970	Episodic ataxia type 7
1020	EOFAD	2199	Epidermolytic palmoplantar keratoderma	401953	Episodic ataxia type 8
168829	EOPPC	2199	Epidermolytic palmoplantar keratoderma of Voerner	37612	Episodic ataxia with myokymia
449566	Eosinophilic angiocentric fibrosis	2199	Epidermolytic palmoplantar keratoderma of Vörner	401953	Episodic ataxia with slurred speech
901	Eosinophilic cellulitis	141077	Epignathus	53583	Episodic choreoathetosis/spasticity
402035	Eosinophilic colitis	1946	Epilepsy - dementia - amelogenesis imperfecta	29822	Episodic spontaneous hypothermia
75566	Eosinophilic endocarditis	1948	Epilepsy - microcephaly - skeletal dysplasia	93928	Epispadias
2070	Eosinophilic enteritis	65683	Epilepsy due to FCD	293381	Epithelial recurrent erosion dystrophy
73247	Eosinophilic esophagitis	1951	Epilepsy telangiectasia	103912	Epithelio-exfoliative colitis - deafness
3165	Eosinophilic fasciitis	86911	Epilepsy with myoclonic absences	157791	Epithelioid hemangioendothelioma
2070	Eosinophilic gastroenteritis	1942	Epilepsy with myoclonic-astatic seizures	293202	Epithelioid sarcoma
2070	Eosinophilic gastroenterocolitis	1942	Epilepsy with myoclonic-ataxic seizures	254698	Epithelioid trophoblastic tumor
99871	Eosinophilic granuloma	411986	Epilepsy-cortical blindness-intellectual disability-facial dysmorphism syndrome	91414	Epithelioma calcificans of Malherbe
183	Eosinophilic granulomatosis with polyangiitis	725	Epileptic encephalopathy with continuous spike-and-wave during slow sleep	501	EPM2
482	Eosinophilic lymphogranuloma	353217	Epileptic encephalopathy with global cerebral demyelination	263516	EPM3
364055	EOSRD	79238	Epimerase deficiency galactosemia	163696	EPM4
256	EOTD	1819	Epimetaphyseal skeletal dysplasia	402082	EPMS
251880	Ependymoblastoma			280620	EPM6
251636	Ependymoma			435438	EPM7
99169	Epiblepharon			424027	EPM8
185	Epibronchial right pulmonary artery syndrome			79278	EPP
83314	Epidemic typhus			2199	EPPK
35125	Epidermal hamartoma syndrome			→182050	Epstein syndrome
35125	Epidermal nevus syndrome			313920	Epstein-Barr virus-associated gastric carcinoma
302	Epidermodyplasia verruciformis			289661	Epstein-Barr virus-positive diffuse large B-cell lymphoma of the elderly
46487	Epidermolysis bullosa acquisita			85438	ERA
79404	Epidermolysis bullosa letalis			229	Erdheim disease
412181	Epidermolysis bullosa simplex due to BP230 deficiency			35687	Erdheim-Chester disease
412189	Epidermolysis bullosa simplex due to exophilin 5 deficiency			293381	ERED
158668	Epidermolysis bullosa simplex due to plakophilin deficiency			999	Ermine phenotype
79400	Epidermolysis bullosa simplex of palms and soles			160148	Eroded polypoid hyperplasia
				→79500	Eronen-Somer-Gustafsson 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
222	Erosive pustular dermatosis of the scalp	3318	Essential thrombocytosis	86879	Extranodal nasal NK/T cell lymphoma
228264	Eruptive collagenoma	1957	Esthesioneuroblastoma	370334	Extraosseous Ewing sarcoma
90000	Erythema elevatum diutinum	785	Estrogen resistance syndrome	370334	Extraosseous Ewing tumor
231031	Erythema palmaris hereditarium	3318	ET	370334	Extraskeletal Ewing sarcoma
729	Erythremia	31826	Ethylene glycol poisoning	209916	Extraskeletal myxoid chondrosarcoma
308473	Erythrocyte epimerase deficiency galactosemia	51188	Ethylmalonic encephalopathy	1964	Extrasystoles - short stature - hyperpigmentation - microcephaly
308473	Erythrocyte galactose epimerase deficiency	983	ETRS	251927	Extraventricular neurocytoma
308473	Erythrocyte GALE deficiency	86880	ETTL	2725	Eye defects - arachnodactyly - cardiopathy
308473	Erythrocyte GALE-D	2892	Euhidrotic ectodermal dysplasia	3172	Eyebrow duplication - syndactyly
171690	Erythrocyte lactate transporter defect	99172	Euryblepharon	2985	Eyebrows and eyelashes absence - intellectual disability
308473	Erythrocyte UDP-galactose-4-epimerase deficiency	1959	Evans syndrome	139431	Eyelid myoclonia with and without absences
308473	Erythrocyte uridine diphosphate galactose-4-epimerase deficiency	444463	Evans syndrome associated with primary immunodeficiency	35909	F5F8D
314	Erythroderma desquamativum	2990	EVMPS	957	F syndrome
79394	Erythrodermic ichthyosis	319	Ewing sarcoma	95	FA
247165	Erythroedema polyneuritis	99734	Exercise-induced delayed-onset myotonia	324	Fabry disease
315	Erythrokeratoderma "en cocardes"	165991	Exercise-induced hyperinsulinemic hypoglycemia	1969	FACES syndrome
316	Erythrokeratoderma progressiva symmetrica	165991	Exercise-induced hyperinsulinism	1167	Facial asymmetry - temporal seizures
317	Erythrokeratoderma variabilis	289586	Exfoliative ichthyosis	141051	Facial dermoid cyst
171851	Erythrokeratoderma variabilis 3	→955	Exner syndrome	→3157	Facial dysmorphism - ambiguous genitalia - hypopituitarism - short limbs
171851	Erythrokeratoderma variabilis, Kamouraska type	116	Exomphalos - macroglossia - gigantism	352712	Facial dysmorphism - immunodeficiency - livedo - short stature
317	Erythrokeratoderma variabilis, Mendes da Costa type	1962	Exostoses - anetodermia - brachydactyly type E	2588	Facial dysmorphism - intellectual disability - short stature - hearing loss
1955	Erythrokeratoderma with ataxia	374	Expanded spectrum of hemifacial microsomia	1970	Facial dysmorphism - macrocephaly - myopia - Dandy-Walker malformation
50943	Erythrokeratolysis hiemalis	322	Extrophy-epispadias complex	1778	Facial dysmorphism - shawl scrotum - joint laxity
318	Erythroleukemia	321	EXT1/EXT2-CDG	412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-nontraumatic conjunctive cysts syndrome
1956	Erythromelalgia	440724	Extensive peripapillary myelinated nerve fibers	412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome
280379	Erythropoietic uroporphyrinia associated with myeloid malignancy	3294	Extensor tendons of finger anomalies	221083	Facial hemispasm
99977	ESCC	141074	External auditory canal aplasia/hypoplasia	3020	Facial nerve palsy due to herpes zoster infection
2405	Escher-Hirt syndrome	141074	External auditory canal stenosis/atresia	3020	Facial nerve palsy due to VZV
2990	Escobar syndrome	231632	Extra-adrenal aldosterone-producing tumor	3020	Facial nerve paralysis due to VZV
2990	Escobar variant multiple pterygium syndrome	168829	Extra-ovarian primary peritoneal carcinoma	85162	Facial onset sensory and motor neuropathy
99976	Esophageal adenocarcinoma	66662	Extracutaneous mastocytoma	3237	Facio-audio-symphalangism
1199	Esophageal atresia	182127	Extragonadal germinoma	1974	Facio-digitogenital syndrome, Kuwait type
418945	Esophageal carcinoma, salivary gland type	280811	Extralobar congenital bronchopulmonary sequestration	1300	Facio-genito-popliteal syndrome
100047	Esophageal duplication cyst	280811	Extralobar congenital pulmonary sequestration	2143	Facio-oculo-acoustico-renal syndrome
99977	Esophageal epidermoid carcinoma	2800	Extramammary Paget disease		
99977	Esophageal squamous cell carcinoma	86850	Extramedullary myeloid tumor		
91138	Essential cryoglobulinemia	100022	Extramedullary soft tissue plasmacytoma		
2056	Essential fructosuria	100002	Extraneural perineurioma		
98981	Essential iris atrophy	52417	Extranodal marginal zone B-cell lymphoma		
91138	Essential mixed cryoglobulinemia				
2843	Essential pentosuria				
98682	Essential strabismus				
3318	Essential thrombocythemia				

→ 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
2048	Facio-pharyngo-glosso-masticatory diplegia	238269	Familial amyloid nephropathy due to apolipoprotein A-II variant	221061	Familial cerebral cavernoma
374	Facioauriculovertebral dysplasia	93562	Familial amyloid nephropathy due to fibrinogen A alpha-chain variant	221061	Familial cerebral cavernous malformation
1973	Faciocardiorenal syndrome	93561	Familial amyloid nephropathy due to lysozyme variant	231160	Familial cerebral saccular aneurysm
3071	Faciocutaneoskeletal syndrome	85447	Familial amyloid polyneuropathy type I (Portuguese-Swedish-Japanese Type)	36382	Familial cervical artery dissection
915	Faciodigitogenital syndrome	85448	Familial amyloid polyneuropathy type IV	1428	Familial chondromalacia patellae
915	Faciogenital dysplasia	85448	Familial amyloidosis, Finnish type	444490	Familial chylomicronemia syndrome
269	Facioscapulohumeral dystrophy	228277	Familial anetoderma	404560	Familial Clark nevus syndrome
269	Facioscapulohumeral muscular dystrophy	199279	Familial angioliomatosis	293144	Familial clubfoot due to 5q31 microdeletion
269	Facioscapulohumeral myopathy	91378	Familial angioneurotic edema	238578	Familial clubfoot due to 17q23.1q23.2 microduplication
98879	Factor IX deficiency	229	Familial aortic dissection	293150	Familial clubfoot due to PITX1 point mutation
220436	Factor V Quebec	425	Familial apoA-I deficiency	199315	Familial clubfoot with or without associated lower limb anomalies
98878	Factor VIII deficiency	309020	Familial apoC-II deficiency	47045	Familial cold autoinflammatory syndrome
300359	FACU	309020	Familial apolipoprotein C-II deficiency	247868	Familial cold autoinflammatory syndrome type 2
306550	FADD-related immunodeficiency	1416	Familial articular chondrocalcinosis	47045	Familial cold urticaria
994	FADS	334	Familial atrial fibrillation	300359	Familial cold urticaria with common variable immunodeficiency
882	FAH deficiency	615	Familial atrial myxoma	440437	Familial colorectal cancer Type X
329308	FAHN	436242	Familial atrial tachyarrhythmia-infranodal cardiac conduction disease	238722	Familial congenital contralateral synkinesia
→168569	Faisalabad histiocytosis	300359	Familial atypical cold urticaria	95494	Familial congenital hypopituitarism
3304	Fallot complex - intellectual disability - growth delay	404560	Familial atypical mole syndrome	238722	Familial congenital mirror movements
86814	FAME	404560	Familial atypical multiple mole melanoma syndrome	91498	Familial congenital palsy of trochlear nerve
397685	Familial hyperprolactinemia	404560	Familial atypical multiple mole melanoma-pancreatic carcinoma syndrome	86814	Familial cortical myoclonic tremor and epilepsy
86	Familial abdominal aortic aneurysm	86820	Familial avascular necrosis of femoral head	319189	Familial cortical myoclonus
637	Familial acoustic neurinoma	2398	Familial benign cervical lipomatosis	1416	Familial CPPD
637	Familial acoustic neuroma	2841	Familial benign chronic pemphigus	85453	Familial cutaneous amyloidosis
88619	Familial acute necrotizing encephalopathy	1551	Familial benign copper deficiency	53296	Familial cutaneous collagenoma
733	Familial adenomatous polyposis	363989	Familial benign flecked retina	313846	Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome
261584	Familial adenomatous polyposis due to 5q22.2 microdeletion	405	Familial benign hypercalcemia	211	Familial cylindromatosis
261584	Familial adenomatous polyposis due to del(5)(q22.2)	405	Familial benign hypocalcemic hypercalcemia	97345	Familial dementia, British type
261584	Familial adenomatous polyposis due to monosomy 5q22.2	231160	Familial berry aneurysm	97346	Familial dementia, Danish type
404	Familial adrenal adenoma	402075	Familial bicuspid aortic valve	313808	Familial dementia, Neumann type
95700	Familial adrenal hypoplasia with absent pituitary LH	221061	Familial brain cavernous angioma	1799	Familial developmental dysphasia
95700	Familial adrenal hypoplasia with absent pituitary luteinizing hormone	221061	Familial brain cavernous hemangioma	26106	Familial diffuse cancer of stomach
95700	Familial adrenal hypoplasia, miniature type	227535	Familial breast cancer	26106	Familial diffuse gastric cancer
86814	Familial adult myoclonic epilepsy	227535	Familial breast carcinoma	85169	Familial digital arthropathy-brachydactyly
164736	Familial advanced sleep-phase syndrome	36382	Familial CAD	300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation
98880	Familial afibrinogenemia	2678	Familial café-au-lait spots	18	Familial distal primary acidosis
1020	Familial Alzheimer disease	1416	Familial calcium pyrophosphate deposition	85192	Familial doughnut lesions of skull
280397	Familial Alzheimer-like prion disease	91415	Familial capillary hemangioma	75376	Familial drusen
319465	Familial AML	1768	Familial caudal dysgenesis		
85450	Familial amyloid nephropathy	1416	Familial CC		
93560	Familial amyloid nephropathy due to apolipoprotein A-I variant	169085	Familial CD8 deficiency		
		892	Familial cerebelloretinal angiomas		

→ 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
79142	Familial Dupuytren contracture	412	Familial hyperlipoproteinemia type 3	300547	Familial infantile hypercalcemia with suppressed intact parathyroid hormone
1764	Familial dysautonomia	306661	Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome	352582	Familial infantile myoclonic epilepsy
314381	Familial dysautonomia with contractures	682	Familial hyperPP	352582	Familial infantile myoclonus epilepsy
412	Familial dysbetaipoproteinemia	99763	Familial hyperreninemic hypoaldosteronism type 1	225154	Familial infantile striatonigral degeneration
98881	Familial dysfibrinogenemia	99764	Familial hyperreninemic hypoaldosteronism type 2	225154	Familial infantile striatonigral necrosis
324588	Familial dyskinesia and facial myokymia	424	Familial hyperthyroidism due to mutations in TSH receptor	2454	Familial intestinal malrotation - facial anomalies
404560	Familial dysplastic nevus syndrome	413	Familial hypertriglyceridemia	2300	Familial intestinal polyatresia syndrome
1885	Familial ectopia lentis	427	Familial hypoaldosteronism	231160	Familial intracranial saccular aneurysm
2762	Familial ectopic ossification	425	Familial hypoalphalipoproteinemia	217656	Familial isolated arrhythmogenic right ventricular cardiomyopathy
85110	Familial encephalopathy with neuroserpin inclusion bodies	405	Familial hypocalciuric hypercalcemia	217656	Familial isolated arrhythmogenic right ventricular dysplasia
101039	Familial epilepsy and mental retardation limited to females	93372	Familial hypocalciuric hypercalcemia type 1	217656	Familial isolated arrhythmogenic ventricular cardiomyopathy
391384	Familial episodic pain syndrome	101049	Familial hypocalciuric hypercalcemia type 2	293899	Familial isolated arrhythmogenic ventricular cardiomyopathy, biventricular form
391392	Familial episodic pain syndrome with predominantly lower limb involvement	101050	Familial hypocalciuric hypercalcemia type 3	293910	Familial isolated arrhythmogenic ventricular cardiomyopathy, classic form
391389	Familial episodic pain syndrome with predominantly upper body involvement	248408	Familial hypodysfibrinogenemia	293888	Familial isolated arrhythmogenic ventricular cardiomyopathy, left dominant form
90042	Familial erythrocytosis	101041	Familial hypofibrinogenemia	293910	Familial isolated arrhythmogenic ventricular cardiomyopathy, right dominant form
225968	Familial essential thrombocythemia	440	Familial hypospadias	217656	Familial isolated arrhythmogenic ventricular dysplasia
85195	Familial expansile osteolysis	225154	Familial IBSN	293899	Familial isolated arrhythmogenic ventricular dysplasia, biventricular form
891	Familial exudative vitreoretinopathy	1677	Familial idiopathic dilatation of the right atrium	293888	Familial isolated arrhythmogenic ventricular cardiomyopathy, left dominant form
98820	Familial focal epilepsy with variable foci	656	Familial idiopathic nephrotic syndrome	293910	Familial isolated arrhythmogenic ventricular cardiomyopathy, right dominant form
314022	Familial fundic gland polyposis with gastric cancer	656	Familial idiopathic steroid-resistant nephrotic syndrome	217656	Familial isolated arrhythmogenic ventricular dysplasia
231040	Familial generalized lentiginosis	93214	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation	293899	Familial isolated arrhythmogenic ventricular dysplasia, biventricular form
99819	Familial gestational hyperthyroidism	93217	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	293910	Familial isolated arrhythmogenic ventricular dysplasia, classic form
361	Familial glucocorticoid deficiency	93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis	293888	Familial isolated arrhythmogenic ventricular dysplasia, left dominant form
3000	Familial gonadotropin-independent male-limited sexual precocity	93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	293910	Familial isolated arrhythmogenic ventricular dysplasia, right dominant form
540	Familial hemophagocytic lymphohistiocytosis	93216	Familial idiopathic steroid-resistant nephrotic syndrome with minimal changes	217656	Familial isolated ARVC
32960	Familial Hibernian fever	225154	Familial infantile bilateral striatal necrosis	217656	Familial isolated ARVD
540	Familial HLH	300373	Familial infantile gigantism	295014	Familial isolated clinodactyly of fingers
2604	Familial hollow visceral myopathy	448348	Familial infantile gigantism due to a point mutation	101351	Familial isolated congenital asplenia
403	Familial hyperaldosteronism type 1	448372	Familial infantile gigantism due to dup(X)q(26)	154	Familial isolated dilated cardiomyopathy
404	Familial hyperaldosteronism type 2	448372	Familial infantile gigantism due to Xq26 microduplication	99879	Familial isolated hyperparathyroidism
251274	Familial hyperaldosteronism type 3			2238	Familial isolated hypoparathyroidism
403	Familial hyperaldosteronism type I			2239	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland
404	Familial hyperaldosteronism type II				
251274	Familial hyperaldosteronism type III				
79506	Familial hyperalphalipoproteinemia				
94086	Familial hypercalcemia - nephrocalcinosis - indicanuria				
238475	Familial hypercholanemia				
178345	Familial hyperestrogenism				
757	Familial hyperkalemic hypertension				
682	Familial hyperkalemic periodic paralysis				

→ 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
189466	Familial isolated hypoparathyroidism due to impaired PTH secretion	500	Familial multiple lentigines syndrome	79083	Familial partial lipodystrophy type 3
314777	Familial isolated pituitary adenoma	231040	Familial multiple lentigines syndrome without systemic involvement	2348	Familial partial lipodystrophy, Dunnigan type
397685	Familial isolated prolactin receptor deficiency	199276	Familial multiple lipomatosis	79084	Familial partial lipodystrophy, K��berling type
75249	Familial isolated restrictive cardiomyopathy	263662	Familial multiple meningioma	871	Familial PCCD
411788	Familial isolated trichomegaly	624	Familial multiple nevi flammei	93333	Familial pelvis-scapular dysplasia
96	Familial isolated vitamin E deficiency	624	Familial multiple port-wine stains	29072	Familial pheochromocytoma-paraganglioma
2295	Familial joint instability syndrome	867	Familial multiple trichoepithelioma	98809	Familial PKD
2295	Familial joint laxity	922	Familial nasal acilia	71290	Familial platelet disorder with associated myeloid malignancy
180176	Familial juvenile gigantomastia	209886	Familial nephropathy with gout	71290	Familial platelet syndrome
209886	Familial juvenile gouty nephropathy	424	Familial non-immune hyperthyroidism	71290	Familial platelet syndrome with predisposition to acute myelogenous leukemia
180176	Familial juvenile hypertrophy of the breast	306658	Familial normophosphatemic tumoral calcinosis	330061	Familial polymorphous light eruption of American Indians
209886	Familial juvenile hyperuricemic nephropathy type 1	88632	Familial ocular anterior segment mesenchymal dysgenesis	733	Familial polyposis coli
217330	Familial juvenile hyperuricemic nephropathy type 2	280403	Familial omphalocele syndrome with facial dysmorphism	261584	Familial polyposis coli due to monosomy 5q22.2
493	Familial keratoacanthoma	154	Familial or idiopathic dilated cardiomyopathy	99810	Familial porencephaly
293936	Familial keratoconus with cataract	75249	Familial or idiopathic restrictive cardiomyopathy	443062	Familial porphyria cutanea tarda
3267	Familial lambdoid synostosis	569	Familial or sporadic hemiplegic migraine	2196	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement
79293	Familial LCAT deficiency	443236	Familial orthostatic tachycardia due to norepinephrine transporter deficiency	31043	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement
523	Familial leiomyomatosis and renal cell cancer	435329	Familial ossifying fibroma	34527	Familial primary hypomagnesemia with normocalciuria and normocalcemia
523	Familial leiomyomatosis cutis et uteri	251262	Familial osteochondritis dissecans	353220	Familial primary localized cutaneous amyloidosis
523	Familial leiomyomatosis with renal carcinoma	2769	Familial osteodysplasia, Anderson type	2257	Familial primary pulmonary hypoplasia
231040	Familial lentigines profusa	2801	Familial osteoectasia	65748	Familial primary self-healing squamous epithelioma of the skin, Ferguson-Smith type
871	Familial Len��re disease	86820	Familial osteonecrosis of the femoral head	871	Familial progressive cardiac conduction defect
871	Familial Lev disease	79093	Familial osteosclerosis with abnormalities of the nervous system and meninges	871	Familial progressive heart block
871	Familial Lev-Len��re disease	1333	Familial pancreatic cancer	280628	Familial progressive hyper- and hypopigmentation
309015	Familial lipoprotein lipase deficiency	319487	Familial papillary or follicular thyroid carcinoma	79146	Familial progressive hyperpigmentation
768	Familial long QT syndrome	97290	Familial papillary thyroid carcinoma with renal papillary neoplasia	313808	Familial progressive subcortical gliosis
75381	Familial macular edema	99877	Familial parathyroid adenoma	1767	Familial progressive vestibulocochlear dysfunction
3000	Familial male-limited precocious puberty	99878	Familial parathyroids hyperplasia	1331	Familial prostate cancer
401942	Familial median cleft of the upper and lower lips	97	Familial paroxysmal ataxia	90044	Familial pseudohyperkalemia
342	Familial Mediterranean fever	98809	Familial paroxysmal kinesigenic dyskinesia	→3202	Familial pseudohyperkalemia type 1
99361	Familial medullary thyroid carcinoma	342	Familial paroxysmal polyserositis		
35858	Familial megaloblastic anemia	228140	Familial paroxysmal ventricular fibrillation, not Brugada type		
618	Familial melanoma	98820	Familial partial epilepsy with variable foci		
165805	Familial mesial temporal lobe epilepsy with febrile seizures	79084	Familial partial lipodystrophy type 1		
741	Familial mitral valve prolapse	2348	Familial partial lipodystrophy type 2		
276399	Familial MNG				
99361	Familial MTC				
276399	Familial multinodular goiter				
35909	Familial multiple coagulation factor deficiency				
523	Familial multiple cutaneous leiomyomas				
338	Familial multiple fibrofolliculoma				

→ 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
275777	Familial pulmonary arterial hypertension	71493	Familial thrombocythemia	166073	Fatal infantile encephalopathy with mitochondrial respiratory chain defects
319487	Familial pure nonmedullary thyroid carcinoma	71493	Familial thrombocytosis	166063	Fatal infantile encephalopathy with olivopontocerebellar hypoplasia
1675	Familial pyrimidinemia	329319	Familial thrombocytosis with transverse limb defect	→37011	Fatal infantile encephalopathy-pulmonary hypertension syndrome
79147	Familial reactive perforating collagenosis	3324	Familial thrombomodulin anomalies	289527	Fatal infantile HCM due to mitochondrial complex I deficiency
46348	Familial rectal pain	93953	Familial thyroglossal duct cyst	280553	Fatal infantile hypertonic myofibrillar myopathy
69126	Familial recurrent arthritis	95716	Familial thyroid dyshormonogenesis	289527	Fatal infantile hypertrophic cardiomyopathy due to mitochondrial complex I deficiency
2809	Familial recurrent peripheral facial palsy	53372	Familial trembling of the chin	289527	Fatal infantile hypertrophic cardiomyopathy due to NADH-coenzyme Q reductase deficiency
85450	Familial renal amyloidosis	93583	Familial TTP	289527	Fatal infantile hypertrophic cardiomyopathy due to NADH-CoQ reductase deficiency
93560	Familial renal amyloidosis due to apolipoprotein A-I variant	53715	Familial tumoral calcinosis	17	Fatal infantile lactic acidosis with methylmalonic aciduria
238269	Familial renal amyloidosis due to apolipoprotein A-II variant	36383	Familial vascular leukoencephalopathy	168566	Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3
93561	Familial renal amyloidosis due to lysozyme variant	289365	Familial vesicoureteral reflux	289573	Fatal multiple mitochondrial dysfunction syndrome
69076	Familial renal glucosuria	637	Familial vestibular schwannoma	401869	Fatal multiple mitochondrial dysfunction syndrome type 1
284247	Familial retinal arterial macroaneurysm	2604	Familial visceral myopathy	401874	Fatal multiple mitochondrial dysfunction syndrome type 2
231108	Familial rhabdoid tumor	2808	Familial vocal cord dysfunction	363424	Fatal multiple mitochondrial dysfunction syndrome type 3
→168569	Familial Rosaï-Dorfman disease	289365	Familial VUR	391343	Fatal post-viral neurodegenerative disorder
171839	Familial scaphocephaly - radioulnar synostosis	170	Familial woolly hair syndrome	816	Fatty acid alcohol oxidoreductase deficiency
168624	Familial scaphocephaly syndrome, McGillivray type	170	Familial wooly hair syndrome	329308	Fatty acid hydroxylase-associated neurodegeneration
3135	Familial Scheuermann disease	404560	FAMM-PC syndrome	2064	Faulk-Epstein-Jones syndrome
3135	Familial Scheuermann juvenile kyphosis	404560	FAMMM syndrome	→97229	Fazio-Londe disease
→168569	Familial SHML	84	Fanconi anemia	405	FBH
51083	Familial short QT syndrome	84	Fanconi pancytopenia	405	FBHH
166282	Familial sick sinus syndrome	→2697	Fanconi syndrome - ichthyosis - dysmorphism	404451	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome
→168569	Familial sinus histiocytosis with massive lymphadenopathy	2088	Fanconi-Bickel disease	47045	FCAS
166282	Familial sinus node dysfunction	733	FAP	47045	FCAS1
300345	Familial SLE	261584	FAP due to monosomy 5q22.2	247868	FCAS2
3135	Familial spinal osteochondrosis	2792	Fara-Chlupackova syndrome	440437	FCCTX
2903	Familial spontaneous pneumothorax	333	Farber disease	98970	FCD
3197	Familial startle disease	333	Farber lipogranulomatosis	268961	FCD type I
280406	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	99906	Farmer's lung disease	268973	FCD type Ia
1325	Familial strobloactyly with amino-aciduria	1915	FAS	268980	FCD type Ib
2456	Familial supernumerary nipples	3261	FAS deficiency	268987	FCD type Ic
370034	Familial syringomyelia	1915	FASD		
300345	Familial systemic lupus erythematosus	164736	FASPS		
91387	Familial TAAD	166105	FASTKD2-related infantile mitochondrial encephalomyopathy		
98819	Familial temporal epilepsy	439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease		
91387	Familial thoracic aortic aneurysm and aortic dissection	439854	Fatal congenital hypertrophic cardiomyopathy due to glycogenosis		
		466	Fatal familial insomnia		
		1561	Fatal infantile cardioencephalomyopathy due to cytochrome C oxidase deficiency		
		1561	Fatal infantile COX deficiency		
		1561	Fatal infantile cytochrome C oxidase deficiency		

→ 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
268994	FCD type II	295091	Femorotibiofibular intercalary transverse meromelia, bilateral	69063	Fetomaternal alloimmunization with antenatal glomerulopathies
269001	FCD type IIa	295089	Femorotibiofibular intercalary transverse meromelia, unilateral	163703	Fever-induced refractory epileptic encephalopathy in school-aged children
269008	FCD type IIb	2019	Femur-fibula-ulna complex	891	FEVR
272	FCMD	2019	Femur-fibula-ulna dysostosis	254492	FFA
86814	FCMTE	2019	Femur-fibula-ulna syndrome	398166	FFDD
99654	FCPD	60015	Fenestrae parietales symmetricae	79133	FFDD1
3071	FCS syndrome	85110	FENIB	398173	FFDD2
47045	FCU	1184	Fenton-Wilkinson-Toselano syndrome	1807	FFDD3
324	FD	45358	FEOM	398189	FFDD4
324588	FDFM	391384	FEPS	79133	FFDD type I
26106	FDGC	65748	Ferguson-Smith disease	398173	FFDD type II
412022	FDLAB syndrome	2180	Ferlini-Ragno-Calzolari syndrome	1807	FFDD type III
163703	Febrile infection-related epilepsy syndrome	157846	Ferritin-related neurodegeneration	398189	FFDD type IV
98974	FECD	397922	Ferro-cerebro-cutaneous syndrome	98820	FFEVF
→18205 0	Fechtner syndrome	139491	Ferroportin disease	1988	FFS
79292	FED	40366	Fetal acitretin/etretinate syndrome	2019	FFU complex
247165	Feer disease	994	Fetal akinesia deformation sequence	313855	FGFR2-related bent bone dysplasia
98969	Fehr corneal dystrophy	363409	Fetal akinesia-cerebral and retinal hemorrhage syndrome	1305	FGLDS
1192	Feigenbaum-Bergeron-Richardson syndrome	1915	Fetal alcohol spectrum disorders	391641	FGLDS1
1305	Feingold syndrome	1915	Fetal alcohol syndrome	391646	FGLDS2
391641	Feingold syndrome type 1	1908	Fetal aminopterin syndrome	403	FH1
391646	Feingold syndrome type 2	1041	Fetal anasarca	404	FH2
53693	Fellman disease	853	Fetal and neonatal alloimmune thrombocytopenia	251274	FH3
47612	Felty syndrome	370076	Fetal carbamazepine syndrome	403	FH-I
404466	Female infertility due to zona pellucida defect	1911	Fetal cocaine syndrome	404	FH-II
2973	Female pseudohermaphroditism - anorectal anomalies	294	Fetal cytomegalovirus syndrome	251274	FH-III
2975	Female pseudohermaphroditism - skeletal anomalies	1912	Fetal dihydantoin syndrome	→16856 9	FHC
101039	Female restricted epilepsy with intellectual disability	97360	Fetal face syndrome	401920	FHCC
1987	Femoral agenesis/hypoplasia	85212	Fetal Gaucher disease	405	FHH
295067	Femoral agenesis/hypoplasia, bilateral	1912	Fetal hydantoin syndrome	93372	FHH type 1
295065	Femoral agenesis/hypoplasia, unilateral	1041	Fetal hydrops	101049	FHH type 2
399329	Femoral head epiphysiolysis	1909	Fetal indomethacin syndrome	101050	FHH type 3
1988	Femoral hypoplasia - unusual facies syndrome	1910	Fetal iodine syndrome	99763	FHHA1
1987	Femoral intercalary meromelia	1055	Fetal left ventricular aneurysm	99764	FHHA2
295067	Femoral intercalary meromelia, bilateral	284362	Fetal lung interstitial tumor	2196	FHHNC with severe ocular involvement
295065	Femoral intercalary meromelia, unilateral	1917	Fetal methylmercury syndrome	31043	FHHNC without severe ocular involvement
1863	Femoral trochlear groove insufficiency	1918	Fetal minoxidil syndrome	263479	FHI
1988	Femoral-facial syndrome	295	Fetal parvovirus syndrome	397618	FHONDA syndrome
294977	Femorotibiofibular intercalary transverse meromelia	290	Fetal rubella syndrome	1988	FHUFS
1863	Femoral trochlear groove insufficiency	3312	Fetal thalidomide syndrome	251601	Fibrillary astrocytoma
1988	Femoral-facial syndrome	1913	Fetal trimethadione syndrome	331	Fibrin-stabilizing factor deficiency
294977	Femorotibiofibular intercalary transverse meromelia	1906	Fetal valproate syndrome	93562	Fibrinogen A alpha-chain amyloidosis
1863	Femoral trochlear groove insufficiency	1906	Fetal valproic acid syndrome	439881	Fibrinous bronchitis
1988	Femoral-facial syndrome	291	Fetal varicella syndrome	99654	Fibrocalculus pancreatic diabetes
294977	Femorotibiofibular intercalary transverse meromelia	1914	Fetal warfarin syndrome	99654	Fibrocalculus pancreatopathy
1863	Femoral trochlear groove insufficiency	166068	Fetal-onset olivopontocerebellar hypoplasia	2021	Fibrochondrogenesis
1988	Femoral-facial syndrome	95431	Feto-fetal transfusion syndrome	337	Fibrodysplasia ossificans progressiva

→ 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
122	Fibrofolliculomas with trichodiscomas and acrochordons	840	Fistulous vegetative verrucous hydadenoma	221083	Focal myoclonus of face
401920	Fibrolamellar hepatocarcinoma	2823	Fitzsimmons-Guilbert syndrome	48918	Focal myositis
401920	Fibrolamellar hepatocellular carcinoma	2824	Fitzsimmons-McLachlan-Gilbert syndrome	48918	Focal nodular myositis
79105	Fibromyxosarcoma	2820	Fitzsimmons-Walson-Mellor syndrome	448264	Focal non-epidermolytic palmoplantar keratoderma
84090	Fibronectin glomerulopathy	293812	Fixed pigmented erythema	2200	Focal palmoplantar and gingival hyperkeratosis
2030	Fibrosarcoma	3092	Fixed subaortic stenosis	2200	Focal palmoplantar and gingival keratoderma
63999	Fibrosing mediastinitis	209886	FJHN type 1	370002	Focal palmoplantar keratoderma with joint keratoses
249	Fibrous dysplasia of bone	217330	FJHN type 2	443804	Focal stiff limb syndrome
2639	Fibular aplasia - complex brachydactyly	1968	Flat face - microstomia - ear anomaly	443804	Focal stiff-person syndrome
1118	Fibular aplasia - ectrodactyly	79293	FLD	79093	Foix-Alajouanine syndrome
1757	Fibular dimelia - diplopodia	98970	Fleck corneal dystrophy	2048	Foix-Chavany-Marie syndrome
93323	Fibular hemimelia	409	Flegel disease	79097	Folinic acid-responsive seizures
295083	Fibular hemimelia, bilateral	284362	FLIT	113	Follicular atrophoderma and basal cell carcinomas
295081	Fibular hemimelia, unilateral	2044	Floating-Harbor syndrome	79459	Follicular atrophoderma-basal cell carcinoma
2854	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly	83451	Florid cemento-osseous dysplasia	300552	Follicular cholangitis and pancreatitis
93323	Fibular longitudinal meromelia	83451	Florid osseous dysplasia	86902	Follicular dendritic cell sarcoma
295083	Fibular longitudinal meromelia, bilateral	2045	FLOTCH syndrome	69745	Follicular dyskeratoma
295081	Fibular longitudinal meromelia, unilateral	240871	Flucloxacilline toxicity	2112	Follicular hamartoma - alopecia - cystic fibrosis
2256	Fibulo-ulnar hypoplasia - renal anomalies	99734	Fluctuating myotonia	525	Follicular lichen planus
79306	FIC1 deficiency	1685	Fluke infection	545	Follicular lymphoma
29207	Fiessinger-Leroy disease	2047	Flynn-Aird syndrome	300552	Follicular panreatocholangitis
29207	Fiessinger-Leroy-Reiter syndrome	69063	FMAIG	243	Follicular stimulating hormone-resistant ovaries
2756	Figuera syndrome	342	FMF	79100	Folliculitis ulerythematosa reticulata
99879	FIHPT	276399	FMNG	178512	Folliculotropic mycosis fungoides
3255	Filippi syndrome	3000	FMPP	228371	Foodborne botulism
352712	FILS syndrome	319487	FNMTC	3454	Foot contractures-muscle atrophy-oculomotor apraxia syndrome
352582	FIME	137675	Foamy myocardial transformation of infancy	337	FOP
1272	Fine-Lubinsky syndrome	2143	FOAR syndrome	60015	Foramina parietalia permagna
369979	Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome	308013	Focal acral hyperkeratosis	366	Forbes disease
97232	Fingerprint body myopathy	83451	Focal cemento-osseous dysplasia	141071	Foregut duplication cyst of the tongue
209335	Finkel disease	2092	Focal dermal hypoplasia	51208	Formiminoglutamic aciduria
2036	Finlay-Marks syndrome	352587	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	51208	Formiminotransferase cyclodeaminase deficiency
839	Finnish congenital nephrosis	352587	Focal epilepsy-intellectual disability-dysarthria-ataxia syndrome	3238	Forney syndrome
609	Finnish tibial muscular dystrophy	398166	Focal facial dermal dysplasia	3238	Forney-Robinson-Pascoe syndrome
399086	Finnish upper limb-onset distal myopathy	79133	Focal facial dermal dysplasia 1, Brauer type	178333	Forsius-Eriksson syndrome
1825	Finucane-Kurtz-Scott syndrome	398173	Focal facial dermal dysplasia 2, Brauer-Setleis type	178333	Forsius-Eriksson type ocular albinism
314777	FIPA	1807	Focal facial dermal dysplasia 3, Setleis type	85162	FOSMN syndrome
163703	FIREs	398189	Focal facial dermal dysplasia 4	3219	Fountain syndrome
141136	First branchial arch syndrome	79133	Focal facial dermal dysplasia type 1	141037	Fourth branchial cleft anomaly
141013	First branchial cleft anomaly	79133	Focal facial dermal dysplasia type I	141037	Fourth branchial cleft cyst
141013	First branchial cleft cyst	398173	Focal facial dermal dysplasia type II	141037	Fourth branchial cleft fistula
141013	First branchial cleft fistula	1807	Focal facial dermal dysplasia type III	2253	Foveal hypoplasia - presenile cataract
79292	Fish-eye disease	398189	Focal facial dermal dysplasia type IV		
98919	Fisher syndrome	398189	Focal facial preauricular dysplasia		

→ 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
397618	Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome	1931	Frontal encephalocele	24	Fumarase deficiency
221126	Fowler syndrome	254492	Frontal fibrosing alopecia	24	Fumarylaciduria
2795	Fowler-Christmas-Chapple syndrome	1791	Frontofacinal nasal dysplasia	882	Fumarylacetate deficiency
1799	FOXP2-associated dysphasia	1826	Frontometaphyseal dysplasia	882	Fumarylacetate hydrolase deficiency
275777	FPAH	141168	Frontonasal arteriovenous malformation	622	Functional methionine synthase deficiency
71290	FPD/AML syndrome	228390	Frontonasal dysplasia with alopecia and genital anomaly	308380	Functional methionine synthase deficiency type cblDv1
280628	FPHH	228390	Frontonasal dysplasia with alopecia and genital anomaly	2169	Functional methionine synthase deficiency type cblE
353220	FPLCA		Frontonasal dysplasia-severe	2170	Functional methionine synthase deficiency type cblG
79084	FPLD1	306542	microphthalmia-severe facial clefting syndrome	91348	Functioning gonadotropic adenoma
2348	FPLD2	391474	Frontorhiny	91348	Functioning pituitary gonadotropic adenoma
79083	FPLD3	275872	Frontotemporal dementia with amyotrophic lateral sclerosis	227796	Fundus albipunctatus
280356	FPLD4	275872	Frontotemporal dementia with motor neuron disease	827	Fundus flavimaculatus
435660	FPLD6	293848	Frontotemporal dementia, right temporal atrophy variant	99004	Fundus pulverulentus
71290	FPS/AML syndrome	2141	Froster-Huch syndrome	207000	Fungal myositis
313808	FPSG	2215	Froster-Iskenius-Waterson syndrome	→60030	Furlong syndrome
69126	FRA	2056	Fructokinase deficiency	2579	Furukawa-Takagi-Nakao syndrome
908	Fragile X syndrome	348	Fructose-1,6-bisphosphatase deficiency	591	Furuncular myiasis
93256	Fragile X-associated tremor/ataxia syndrome	2057	Frydman-Cohen-Karmon syndrome	591	Furunculoid myiasis
284247	FRAM	2429	Fryns macrocephaly	591	Furunculous myiasis
861	Franceschetti-Klein syndrome	1104	Fryns microphthalmia syndrome	228119	Fusariosis
2523	Franeck-Bocker-Kahlen syndrome	2059	Fryns syndrome	228119	Fusarium infection
137834	Frank-Ter Haar syndrome	→2995	Fryns-Aftimos syndrome	2287	Fused mandibular incisors
100026	Franklin disease	2497	Fryns-Hofkens-Fabry syndrome	2498	Fusion of metacarpals 4 and 5
2108	François dyscephalic syndrome	2058	Fryns-Smeets-Thiry syndrome	35909	FV and FVIII combined deficiency
79149	François syndrome	1305	FS	908	FXS
98970	François-Neetens speckled corneal dystrophy	391641	FS1	93256	FXTAS syndrome
2052	Fraser syndrome	391646	FS2	364	G6P deficiency
→2052	Fraser-like syndrome	269	FSH dystrophy	79258	G6P deficiency type a
347	Frasier syndrome	243	FSH-RO	79259	G6P deficiency type b
908	FraX syndrome	269	FSHD	79259	G6P translocase deficiency
908	FRAXA syndrome	51208	FTCD deficiency	79259	G6PT deficiency
100973	FRAXE intellectual disability	275872	FTD-ALS	25	GA1
100974	FRAXF syndrome	275872	FTD-MND	2066	GABA transaminase deficiency
95	FRDA	247790	FTH1-associated iron overload	79402	GABEB
834	Free sialic acid storage disease	247790	FTH1-related iron overload	90041	Gaisböck syndrome
309324	Free sialic acid storage disease, infantile form	98974	Fuchs endothelial corneal dystrophy	487	Galactocerebrosidase deficiency
2053	Freeman-Sheldon syndrome	263479	Fuchs heterochromic iridocyclitis	79237	Galactokinase deficiency
1147	Freeman-Sheldon syndrome variant	349	Fucosidosis	79237	Galactokinase deficiency galactosemia
2673	Freire Maia-Pinheiro-Opitz syndrome	2854	Fuhrmann syndrome	309297	Galactosamine-6-sulfatase deficiency
2723	Freire-Maia syndrome	2854	Fuhrmann-Rieger-de Sousa syndrome	79238	Galactose epimerase deficiency
→26420 0	Frias syndrome	2060	Fukuda-Miyanomae-Nakata syndrome	79239	Galactose-1-phosphate uridyltransferase deficiency
85335	Fried syndrome	551	Fukuhara syndrome	79239	Galactosemia type 1
2487	Fried-Goldberg-Mundel syndrome	272	Fukuyama congenital muscular dystrophy	79237	Galactosemia type 2
1969	Friedman-Goodman syndrome	35063	Fulminant viral hepatitis	79238	Galactosemia type 3
95	Friedreich ataxia			351	Galactosialidosis
96	Friedreich-like ataxia				
99672	Fried's tooth and nail 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
487	Galactosylceramidase deficiency	44890	Gastrointestinal stromal sarcoma	2623	Geleophysic dwarfism
75496	Galactosyltransferase I deficiency	44890	Gastrointestinal stromal tumor	2623	Geleophysic dysplasia
487	GALC deficiency	2368	Gastroschisis	2073	Gélineau disease
79238	GALE deficiency	228423	GATA2 deficiency	85448	Gelsolin amyloidosis
79238	GALE-D	355	Gaucher disease	2074	Gemignani syndrome
79237	GALK deficiency	2072	Gaucher disease - ophthalmoplegia - cardiovascular calcification	251604	Gemistocytic astrocytoma
79237	GALK-D	77259	Gaucher disease type 1	2084	GEMSS syndrome
100086	Gallbladder endocrine tumor	77260	Gaucher disease type 2	51608	Generalized arterial calcification of infancy
2065	Galloway syndrome	77261	Gaucher disease type 3	79402	Generalized atrophic benign epidermolysis bullosa
2065	Galloway-Mowat syndrome	2072	Gaucher disease type 3C	168632	Generalized basaloid follicular hamartoma syndrome
309297	GALNS deficiency	77261	Gaucher disease, subacute neuronopathic type	98806	Generalized cervical and upper-limb-onset dystonia
79239	GALT deficiency	2072	Gaucher-like disease	528	Generalized congenital lipodystrophy
2325	Gamborg-Nielsen syndrome	308712	GBE deficiency, adult neuromuscular form	228429	Generalized congenital lipodystrophy type 4
3035	Game-Friedman-Paradice syndrome	308684	GBE deficiency, childhood combined hepatic and myopathic form	228429	Generalized congenital lipodystrophy with myopathy
2066	Gamma-aminobutyric acid transaminase deficiency	308698	GBE deficiency, childhood neuromuscular form	263543	Generalized deciduous skin
212	Gamma-cystathione deficiency	308670	GBE deficiency, congenital neuromuscular form	263548	Generalized deciduous skin type A
33573	Gamma-glutamyl transpeptidase deficiency	308655	GBE deficiency, fatal perinatal neuromuscular form	263553	Generalized deciduous skin type B
33574	Gamma-glutamylcysteine synthetase deficiency	308638	GBE deficiency, non progressive hepatic form	263558	Generalized deciduous skin type C
100026	Gamma-HCD	308621	GBE deficiency, progressive hepatic form	231568	Generalized dominant dystrophic epidermolysis bullosa
100026	Gamma-heavy chain disease	360	GBM	79399	Generalized EBS, non-Dowling-Meara type
353	Gamma-sarcoglycanopathy	98916	GBS, acute inflammatory demyelinating polyradiculoneuropathic form	79399	Generalized epidermolysis bullosa simplex, non-Dowling-Meara type
682	Gamstorp disease	329984	GCC	79137	Generalized epilepsy - paroxysmal dyskinesia
682	Gamstorp episodic adynamy	98962	GCD1	36387	Generalized epilepsy with febrile seizures-plus
382	GAMT deficiency	98963	GCD2	308487	Generalized epimerase deficiency galactosemia
251937	Gangliocytoma	25	GCDHD	157991	Generalized eruptive histiocytoma
251949	Ganglioglioma	98962	GCDI	157991	Generalized eruptive histiocytosis
251877	Ganglioneuroblastoma	98963	GCDII	411777	Generalized eruptive keratoacanthoma
251992	Ganglioneuroma	438274	GCGR-related hyperglucagonemia	411777	Generalized eruptive keratoacanthomas of Grzybowski
2067	GAPO syndrome	528	GCL	280774	Generalized essential telangiectasia
314022	GAPPS	228429	GCL4	36236	Generalized exfoliative disease
3469	Garcia-Lurie syndrome	2095	GCM syndrome	1041	Generalized fetal edema
79665	Gardner syndrome	380	GCPS	308487	Generalized galactose epimerase deficiency
324636	Gardner-Diamond syndrome	79330	GCS1-CDG	308487	Generalized GALE deficiency
2075	Gardner-Silengo-Wachtel syndrome	363976	GCT of bone	308487	Generalized GALE-D
99000	Gass disease	98957	GDCD	33355	Generalized hematopoietic hypoplasia
314022	Gastric adenocarcinoma and proximal polyposis of the stomach	53697	GDD	79402	Generalized junctional epidermolysis bullosa, non-Herlitz type
423781	Gastric carcinoma, salivary gland type	366	GDE deficiency	329971	Generalized juvenile polyposis/juvenile polyposis coli
141071	Gastric duplication cyst of the tongue	324636	GDS		
100075	Gastric endocrine tumor	36387	GEFS+		
332	Gastric intrinsic factor deficiency	411777	GEKA		
36273	Gastric linitis plastica	26790	Gelatinous ascites		
418959	Gastric squamous cell carcinoma	98957	Gelatinous drop-like corneal dystrophy		
913	Gastrinoma				
2069	Gastrocutaneous syndrome				
2930	Gastrointestinal polyposis - ectodermal changes				
2930	Gastrointestinal polyposis - skin pigmentation - alopecia - fingernail changes				

→ 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
167635	Generalized lichenoid papular eruption	356	Gerstmann-Straussler-Scheinker syndrome	360	Glioblastoma
435628	Generalized lipodystrophy-progeroid features-severe intellectual disability syndrome	99926	Gestational choriocarcinoma	360	Glioblastoma multiforme
141209	Generalized lymphatic anomaly	63275	Gestational pemphigoid	269197	Glioependymal/ependymal cyst
89842	Generalized mitis RDEB	280774	GET	251582	Gliomatosis cerebri
167635	Generalized papular and sclerodermoid lichen myxedematosus	84090	GFND	251576	Gliosarcoma
263543	Generalized peeling skin syndrome	314769	GH and PRL cosecreting pituitary adenoma	73223	Global developmental delay - osteopenia - ectodermal defect
263548	Generalized peeling skin syndrome type A	633	GH receptor deficiency	404476	Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome
263553	Generalized peeling skin syndrome type B	1802	Ghosal hematodiaphyseal dysplasia	2791	Globodontia
263558	Generalized peeling skin syndrome type C	1802	Ghosal syndrome	487	Globoid cell leukodystrophy
171876	Generalized pseudohypoaldosteronism type 1	83450	Ghost teeth	83454	Glomangiomatosis
263543	Generalized PSS	314811	Ghrelin receptor deficiency	→69735	Glomerulonephritis-sparse hair-telangiectasis syndrome
247353	Generalized pustular psoriasis	180267	Giant adenofibroma of the breast	84090	Glomerulopathy with fibronectin deposits
3221	Generalized resistance to thyroid hormone	643	Giant axonal neuropathy	391651	Glomus tumor
308487	Generalized UDP-galactose-4-epimerase deficiency	397	Giant cell arteritis	83454	Glomuvenous malformation
308487	Generalized uridine diphosphate galactose-4-epimerase deficiency	1190	Giant cell chondrodysplasia	2616	Gloomy face syndrome
254704	Genetic hyperferritinemia without iron overload	251579	Giant cell glioblastoma	141163	Glossopalatine ankylosis
99845	Genetic recurrent myoglobinuria	139436	Giant cell histiocytomatosis	221098	Glossopharyngeal neuralgia
226316	Genetic transient congenital hypothyroidism	363976	Giant cell tumor of bone	221098	Glossovasopharyngeal neuralgia
2075	Genito-palato-cardiac syndrome	626	Giant congenital melanocytic nevus	404476	GLOW syndrome
85201	Genitopatellar syndrome	2494	Giant hypertrophic gastritis	255132	GLRX5-related sideroblastic anemia
2163	Genoa syndrome	626	Giant pigmented hairy nevus	97280	Glucagonoma
85197	Genochondromatosis type 1	274	Giant platelet syndrome	97280	Glucagonoma syndrome
93398	Genochondromatosis type 2	1065	Gillespie syndrome	355	Glucocerebrosidase deficiency
329813	Genome-wide paternal uniparental disomy mosaicism	2025	Gingival fibromatosis - facial dysmorphism	786	Glucocorticoid resistance
1454	Gentile syndrome	3473	Gingival fibromatosis - hepatosplenomegaly - other anomalies	403	Glucocorticoid-remediable aldosteronism
217008	Genuine diffuse phlebectasia	2027	Gingival fibromatosis - progressive deafness	403	Glucocorticoid-sensitive hypertension
98961	Geographic corneal dystrophy	2026	Gingival fibromatosis-hypertrichosis syndrome	79272	Glucosamine N-acetyl-6-sulfatase deficiency
35686	Geographic helicoid peripapillary choroidopathy	2709	Gingival hypertrophy-corneal dystrophy	71277	Glucose transporter type 1 deficiency
79137	GEPD	44890	GIST	35710	Glucose-galactose malabsorption
99095	Gerbode defect	97286	GIST-paraganglioma dyad	79330	Glucosidase 1 deficiency
2808	Gerhardt syndrome	358	Gitelman syndrome	79320	Glucosyltransferase 1 deficiency
213837	Germ cell cancer of cervix uteri	3268	Giuffré-Tsukahara syndrome	79325	Glucosyltransferase 2 deficiency
213751	Germ cell cancer of corpus uteri	141209	GLA	71277	Glut1-DS
2077	German syndrome	849	Glanzmann thrombasthenia	71277	Glut-1 deficiency Syndrome
91352	Germinoma of the central nervous system	666	Glass bone disease	51208	Glutamate formiminotransferase deficiency
2078	Geroderma osteodysplastica	1535	Glass-Chapman-Hockley syndrome	2195	Glutamate-aspartate transport defect
1117	Gershoni-Baruch-Leibo syndrome	213833	Glassy cell carcinoma of the cervix uteri	33574	Glutamate-cysteine ligase deficiency
221117	Gerstmann syndrome	2084	Glaucoma - ectopia - microspherophakia - stiff joints - short stature	25	Glutaric acidemia type 1
		2085	Glaucoma - sleep apnea	26791	Glutaric acidemia type 2
		238763	Glaucoma secondary to spherophakia/ectopia lentis and megalocornea	35706	Glutaric acidemia type 3
		354	GLB1 deficiency	25	Glutaric aciduria type 1
				26791	Glutaric aciduria type 2
				35706	Glutaric aciduria type 3
				25	Glutaryl-CoA dehydrogenase deficiency

→ 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
35706	Glutaryl-CoA oxidase deficiency	308698	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood neuromuscular form	715	Glycogen storage disease due to muscle phosphorylase kinase deficiency
25	Glutaryl-coenzyme A dehydrogenase deficiency	308670	Glycogen storage disease due to glycogen branching enzyme deficiency, congenital neuromuscular form	→31964 6	Glycogen storage disease due to phosphoglucomutase deficiency
32	Glutathione synthetase deficiency	308655	Glycogen storage disease due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form	713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency
289846	Glutathione synthetase deficiency with 5-oxoprolinuria	308638	Glycogen storage disease due to glycogen branching enzyme deficiency, non progressive hepatic form	97234	Glycogen storage disease due to phosphoglycerate mutase deficiency
289849	Glutathione synthetase deficiency without 5-oxoprolinuria	308621	Glycogen storage disease due to glycogen branching enzyme deficiency, progressive hepatic form	2089	Glycogen storage disease type 0a
33573	Glutathionuria	366	Glycogen storage disease due to glycogen debranching enzyme deficiency	137625	Glycogen storage disease type 0b
284414	Glycerol kinase deficiency, adult form	263297	Glycogen storage disease due to glycogenin deficiency	364	Glycogen storage disease type 1
284408	Glycerol kinase deficiency, infantile form	2089	Glycogen storage disease due to hepatic glycogen synthase deficiency	79258	Glycogen storage disease type 1a
284411	Glycerol kinase deficiency, juvenile form	2364	Glycogen storage disease due to lactate dehydrogenase deficiency	79259	Glycogen storage disease type 1b
261476	Glycerol kinase deficiency-contiguous gene syndrome	284435	Glycogen storage disease due to lactate dehydrogenase H-subunit deficiency	→79259	Glycogen storage disease type 1C
255182	Glycine cleavage system L protein deficiency	284426	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency	→79259	Glycogen storage disease type 1D
407	Glycine encephalopathy	34587	Glycogen storage disease due to LAMP-2 deficiency	365	Glycogen storage disease type 2
289891	Glycine N-methyltransferase deficiency	79240	Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency	308552	Glycogen storage disease type 2, infantile onset
365	Glycogen storage disease due to acid maltase deficiency	369	Glycogen storage disease due to liver glycogen phosphorylase deficiency	420429	Glycogen storage disease type 2, late onset
308552	Glycogen storage disease due to acid maltase deficiency, infantile onset	2089	Glycogen storage disease due to liver glycogen synthase deficiency	366	Glycogen storage disease type 3
420429	Glycogen storage disease due to acid maltase deficiency, late-onset	2364	Glycogen storage disease due to lactate dehydrogenase deficiency	367	Glycogen storage disease type 4
57	Glycogen storage disease due to aldolase A deficiency	284435	Glycogen storage disease due to lactate dehydrogenase H-subunit deficiency	308712	Glycogen storage disease type 4, adult neuromuscular form
364	Glycogen storage disease due to G6P deficiency	284426	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency	308684	Glycogen storage disease type 4, childhood combined hepatic and myopathic form
79258	Glycogen storage disease due to G6P deficiency type a	34587	Glycogen storage disease due to LAMP-2 deficiency	308698	Glycogen storage disease type 4, childhood neuromuscular form
79259	Glycogen storage disease due to G6P deficiency type b	79240	Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency	308670	Glycogen storage disease type 4, congenital neuromuscular form
364	Glycogen storage disease due to glucose-6-phosphatase deficiency	369	Glycogen storage disease due to liver glycogen phosphorylase deficiency	308655	Glycogen storage disease type 4, fatal perinatal neuromuscular form
79258	Glycogen storage disease due to glucose-6-phosphatase deficiency type a	2089	Glycogen storage disease due to liver glycogen synthase deficiency	308638	Glycogen storage disease type 4, non progressive hepatic form
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	264580	Glycogen storage disease due to liver phosphorylase kinase deficiency	308621	Glycogen storage disease type 4, progressive hepatic form
2088	Glycogen storage disease due to GLUT2 deficiency	137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency	368	Glycogen storage disease type 5
367	Glycogen storage disease due to glycogen branching enzyme deficiency	99849	Glycogen storage disease due to muscle beta-enolase deficiency	369	Glycogen storage disease type 6B
308712	Glycogen storage disease due to glycogen branching enzyme deficiency, adult neuromuscular form	368	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	371	Glycogen storage disease type 7
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	264580	Glycogen storage disease type 9A
				79240	Glycogen storage disease type 9B
				264580	Glycogen storage disease type 9C
				715	Glycogen storage disease type 9D
				715	Glycogen storage disease type 9E
				284426	Glycogen storage disease type 11
				57	Glycogen storage disease type 12
				→31964 6	Glycogen storage disease type 14
				263297	Glycogen storage disease type 15
				264580	Glycogen storage disease type IXa
				79240	Glycogen storage disease type IXb
				264580	Glycogen storage disease type IXc

→ 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
715	Glycogen storage disease type IXd	264580	Glycogenosis due to liver phosphorylase kinase deficiency	79258	Glycogenosis type Ia
715	Glycogen storage disease type IXe	137625	Glycogenosis due to muscle and heart glycogen synthase deficiency	79259	Glycogenosis type Ib
263297	Glycogen storage disease type XV	99849	Glycogenosis due to muscle beta-enolase deficiency	264580	Glycogenosis type IXa
365	Glycogenosis due to acid maltase deficiency	368	Glycogenosis due to muscle glycogen phosphorylase deficiency	79240	Glycogenosis type IXb
308552	Glycogenosis due to acid maltase deficiency, infantile onset	371	Glycogenosis due to muscle phosphofructokinase deficiency	264580	Glycogenosis type IXc
57	Glycogenosis due to aldolase A deficiency	715	Glycogenosis due to muscle phosphorylase kinase deficiency	715	Glycogenosis type IXd
79258	Glycogenosis due to glucose-6-phosphatase deficiency type a	→31964	Glycogenosis due to phosphoglucomutase deficiency	715	Glycogenosis type IXe
79259	Glycogenosis due to glucose-6-phosphatase deficiency type b	713	Glycogenosis due to phosphoglycerate kinase 1 deficiency	263297	Glycogenosis type XV
79259	Glycogenosis due to glucose-6-phosphatase transport defect	97234	Glycogenosis due to phosphoglycerate mutase deficiency	93598	Glycolic aciduria
2088	Glycogenosis due to GLUT2 deficiency	2089	Glycogenosis type 0a	354	GM1 gangliosidosis
367	Glycogenosis due to glycogen branching enzyme deficiency	137625	Glycogenosis type 0b	79255	GM1 gangliosidosis type 1
308712	Glycogenosis due to glycogen branching enzyme deficiency, adult neuromuscular form	364	Glycogenosis type 1	79256	GM1 gangliosidosis type 2
308684	Glycogenosis due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	365	Glycogenosis type 2	79257	GM1 gangliosidosis type 3
308698	Glycogenosis due to glycogen branching enzyme deficiency, childhood neuromuscular form	308552	Glycogenosis type 2, infantile onset	796	GM2 gangliosidosis 0 variant
308670	Glycogenosis due to glycogen branching enzyme deficiency, congenital neuromuscular form	420429	Glycogenosis type 2, late onset	309246	GM2 gangliosidosis, AB variant
308655	Glycogenosis due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form	366	Glycogenosis type 3	309192	GM2 gangliosidosis, B variant, adult form
308638	Glycogenosis due to glycogen branching enzyme deficiency, non progressive hepatic form	367	Glycogenosis type 4	309178	GM2 gangliosidosis, B variant, infantile form
308621	Glycogenosis due to glycogen branching enzyme deficiency, progressive hepatic form	308712	Glycogenosis type 4, adult neuromuscular form	309185	GM2 gangliosidosis, B variant, juvenile form
366	Glycogenosis due to glycogen debranching enzyme deficiency	308684	Glycogenosis type 4, childhood combined hepatic and myopathic form	845	GM2 gangliosidosis, B, B1 variant
263297	Glycogenosis due to glycogenin deficiency	308698	Glycogenosis type 4, childhood neuromuscular form	309239	GM2 gangliosidosis, B1 variant
2364	Glycogenosis due to lactate dehydrogenase deficiency	308670	Glycogenosis type 4, congenital neuromuscular form	101006	GM2 synthase deficiency
284435	Glycogenosis due to lactate dehydrogenase H-subunit deficiency	308655	Glycogenosis type 4, fatal perinatal neuromuscular form	626	GMN
284426	Glycogenosis due to lactate dehydrogenase M-subunit deficiency	308638	Glycogenosis type 4, non progressive hepatic form	2090	GMS syndrome
34587	Glycogenosis due to LAMP-2 deficiency	308621	Glycogenosis type 4, progressive hepatic form	53697	Gnathodiaphyseal dysplasia
79240	Glycogenosis due to liver and muscle phosphorylase kinase deficiency	368	Glycogenosis type 5	602	GNE myopathy
369	Glycogenosis due to liver glycogen phosphorylase deficiency	369	Glycogenosis type 6B	79272	GNS deficiency
		371	Glycogenosis type 7	329984	Goblet cell adenocarcinoid
		264580	Glycogenosis type 9A	329984	Goblet cell carcinoid
		79240	Glycogenosis type 9B	329984	Goblet cell carcinoma
		264580	Glycogenosis type 9C	329984	Goblet cell tumor
		715	Glycogenosis type 9D	705	Goiter - deafness
		715	Glycogenosis type 9E	373	Golabi-Rosen syndrome
		284426	Glycogenosis type 11	351	Goldberg syndrome
		57	Glycogenosis type 12	66629	Goldberg-Shprintzen megacolon syndrome
		99849	Glycogenosis type 13	166272	Goldblatt chondrodyplasia
		→31964	Glycogenosis type 14	166272	Goldblatt syndrome
		6		3026	Goldblatt-Viljoen syndrome
		263297	Glycogenosis type 15	2261	Goldblatt-Wallis syndrome
				374	Goldenhar syndrome
				53540	Goldmann-Favre syndrome
				3032	Goldston syndrome
				1791	Gollop syndrome
				1986	Gollop-Wolfgang complex
				2092	Goltz syndrome
				2092	Goltz-Gorlin syndrome
				1532	Gómez-López-Hernández syndrome
				1770	Gonadal dysgenesis, XY type - associated anomalies
				432	Gonadotropic deficiency

→ 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
759	Gonadotropin-dependant precocious puberty	900	Granulomatosis with polyangiitis	314769	Growth hormone and prolactin cosecreting pituitary adenoma
562	Gonadotropin-independent female-limited sexual precocity	183	Granulomatous allergic angiitis	633	Growth hormone receptor deficiency
2090	Goniodysgenesis - intellectual disability - short stature	64722	Granulomatous mastitis	97261	Growth hormone releasing factor tumor
1482	Gonococcal conjunctivitis	33111	Granulomatous slack skin	53693	Growth restriction - aminoaciduria - cholestasis - iron overload - lactic acidosis - early death
3034	Gonzales-del Angel syndrome	99915	Granulosa cell cancer	391366	Growth retardation-mild developmental delay-chronic hepatitis syndrome
169105	Good syndrome	35858	Gräsbeck-Imerslund disease	2101	Grubben-de Cock-Borghgraef syndrome
1321	Goodman camptodactyly	69665	Gravidic intrahepatic cholestasis	411777	Grzybowski syndrome
65798	Goodman syndrome	721	Gray platelet syndrome	365	GSD due to acid maltase deficiency
375	Goodpasture syndrome	293375	Grayson-Wilbrandt corneal dystrophy	308552	GSD due to acid maltase deficiency, infantile onset
75389	Goossens-Deviriendt syndrome	276405	Green jaundice	420429	GSD due to acid maltase deficiency, late onset
757	Gordon hyperkalemia-hypertension syndrome	99826	Green monkey disease	57	GSD due to aldolase A deficiency
376	Gordon syndrome	1426	Greenberg dysplasia	364	GSD due to G6P deficiency
1173	Gordon-Holmes syndrome	380	Greig cephalopolysyndactyly syndrome	79258	GSD due to G6P deficiency type a
73	Gorham disease	495	Greither disease	79259	GSD due to G6P deficiency type b
73	Gorham syndrome	97261	GRF tumor	79259	GSD due to G6PT deficiency
377	Gorham-Stout disease	97261	GRFoma	2088	GSD due to GLUT2 deficiency
2095	Gorlin-Chaudhry-Moss syndrome	139474	Grisart-Destrée syndrome	367	GSD due to glycogen branching enzyme deficiency
377	Gorlin-Goltz syndrome	381	Griselli disease	308712	GSD due to glycogen branching enzyme deficiency, adult neuromuscular form
66629	GOSHS	79476	Griselli disease type 1	308684	GSD due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form
280620	GOSR2-related progressive myoclonus ataxia	79477	Griselli disease type 2	308698	GSD due to glycogen branching enzyme deficiency, childhood neuromuscular form
2500	Gottron syndrome	79478	Griselli disease type 3	308670	GSD due to glycogen branching enzyme deficiency, congenital neuromuscular form
59135	Gowers disease	2099	Grix-Blankenship-Peterson syndrome	308655	GSD due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form
900	GPA	3217	Groll-Hirschowitz syndrome	308638	GSD due to glycogen branching enzyme deficiency, non progressive hepatic form
280586	gPAPP deficiency	758	Gronblad-Strandberg-Touraine syndrome	308621	GSD due to glycogen branching enzyme deficiency, progressive hepatic form
247353	GPP	314613	Growing teratoma syndrome	366	GSD due to glycogen debranching enzyme deficiency
721	GPS	391348	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	263297	GSD due to glycogenin deficiency
313808	GPSC	→26420	Growth deficiency - brachydactyly - dysmorphism	2089	GSD due to hepatic glycogen synthase deficiency
403	GRA	2067	Growth delay - alopecia - pseudoanodontia - optic atrophy	2364	GSD due to lactate dehydrogenase deficiency
2763	Gracile bone dysplasia	53693	Growth delay - aminoaciduria - cholestasis - iron overload - lactic acidosis - early death		
53693	GRACILE syndrome	73272	Growth delay - deafness- intellectual disability		
39812	Graft versus host disease	3035	Growth delay - hydrocephaly - lung hypoplasia		
505	Graham Little syndrome	79113	Growth delay - intellectual disability - mandibulofacial dysostosis - microcephaly - cleft palate		
505	Graham Little-Piccardi-Lassueur syndrome	73273	Growth delay due to insulin-like growth factor I resistance		
2111	Graham-Boyle-Troxell syndrome	73272	Growth delay due to insulin-like growth factor type 1 deficiency		
52055	Graham-Cox syndrome				
3421	Grand-Kaine-Fulling syndrome				
79094	Grange occlusive arterial syndrome				
79094	Grange syndrome				
2097	Grant syndrome				
98962	Granular corneal dystrophy type 1				
98963	Granular corneal dystrophy type 2				
98962	Granular corneal dystrophy type I				
98963	Granular corneal dystrophy type II				
98961	Granular corneal dystrophy type III				
98963	Granular-lattice corneal dystrophy				
86850	Granulocytic sarcoma				

→ 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
284435	GSD due to lactate dehydrogenase H-subunit deficiency	79240	GSD type 9B	324561	Guttate hypopigmentation and punctate palmoplantar keratoderma
284426	GSD due to lactate dehydrogenase M-subunit deficiency	264580	GSD type 9C	2957	Guttmacher syndrome
34587	GSD due to LAMP-2 deficiency	715	GSD type 9D	39812	GVH
79240	GSD due to liver and muscle phosphorylase kinase deficiency	715	GSD type 9E	293375	GWCD
369	GSD due to liver glycogen phosphorylase deficiency	97234	GSD type 10	99914	Gynandroblastoma
264580	GSD due to liver phosphorylase kinase deficiency	284426	GSD type 11	414	Gyrate atrophy of choroid and retina
137625	GSD due to muscle and heart glycogen synthase deficiency	57	GSD type 12	168569	H syndrome
99849	GSD due to muscle beta-enolase deficiency	→31964 6	GSD type 14	139441	H-ABC
368	GSD due to muscle glycogen phosphorylase deficiency	263297	GSD type 15	2396	Haberland syndrome
371	GSD due to muscle phosphofructokinase deficiency	264580	GSD type IXa	99803	Haddad syndrome
715	GSD due to muscle phosphorylase kinase deficiency	79240	GSD type IXb	71212	HADH deficiency
→31964 6	GSD due to phosphoglucomutase deficiency	264580	GSD type IXc	217026	Hadziselimovic syndrome
713	GSD due to phosphoglycerate kinase 1 deficiency	715	GSD type IXd	91378	HAE
97234	GSD due to phosphoglycerate mutase deficiency	715	GSD type IXe	100051	HAE 2
2089	GSD type 0a	263297	GSD type XV	100054	HAE 3
137625	GSD type 0b	79258	GSDIa	100050	HAE-I
364	GSD type 1	79259	GSDIb	100051	HAE-II
79259	GSD type 1 non a	366	GSDIII	100054	HAE-III
79258	GSD type 1a	308712	GSDIV, adult neuromuscular form	966	HAFF
79259	GSD type 1b	308684	GSDIV, childhood combined hepatic and myopathic form	79263	Hagberg-Santavuori disease
365	GSD type 2	308698	GSDIV, childhood neuromuscular form	2841	Hailey-Hailey disease
308552	GSD type 2, infantile onset	308670	GSDIV, congenital neuromuscular form	2342	Haim-Munk syndrome
420429	GSD type 2, late onset	308655	GSDIV, fatal perinatal neuromuscular form	1408	Hair defect - photosensitivity - intellectual disability
366	GSD type 3	308638	GSDIV, non progressive hepatic form	69084	Hair-nail ectodermal dysplasia
367	GSD type 4	308621	GSDIV, progressive hepatic form	58017	Hairy cell leukemia
308712	GSD type 4, adult neuromuscular form	99849	GSDXIII	300878	Hairy cell leukemia variant
308684	GSD type 4, childhood combined hepatic and myopathic form	→31964 6	GSDXIV	2220	Hairy elbows
308698	GSD type 4, childhood neuromuscular form	2102	GTP cyclohydrolase I deficiency	3387	Hairy throat syndrome
308670	GSD type 4, congenital neuromuscular form	98808	GTPCH1-deficient dopa-responsive dystonia	955	Hajdu-Cheney syndrome
308655	GSD type 4, fatal perinatal neuromuscular form	98808	GTPCH1-deficient DRD	2157	HAL deficiency
308638	GSD type 4, non progressive hepatic form	2102	GTPCH deficiency	2985	Hal-Berg-Rudolph syndrome
308621	GSD type 4, progressive hepatic form	90020	Guam disease	2521	Halal syndrome
368	GSD type 5	319234	Guanarito hemorrhagic fever	1809	Halal-Setton-Wang syndrome
369	GSD type 6B	382	Guanidinoacetate methyltransferase deficiency	185	Halasz syndrome
371	GSD type 7	2785	Guibaud-Vainsel syndrome	138	Hall-Hittner syndrome
264580	GSD type 9A	98916	Guillain-Barré syndrome, acute inflammatory demyelinating polyradiculoneuropathic form	2107	Hall-Riggs syndrome
		231	Guinea worm disease	2108	Hallermann-Streiff syndrome
		1661	Guízar Vázquez-Luengas-Muñoz syndrome	2109	Hallermann-Streiff-François syndrome, severe form
		2104	Guízar Vázquez-Sánchez-Manzano syndrome	2109	Hallermann-Streiff-like syndrome
		1562	Gunal-Seber-Basaran syndrome	157850	Hallervorden-Spatz syndrome
		79277	Günther disease	2110	Hallux varus - preaxial polysyndactyly
		1858	Gurrieri-Sammitto-Bellussi syndrome	3453	HAM syndrome
				289326	HAM/TSP
				314555	Hamamy syndrome
				2926	Hamanishi-Ueba-Tsuiji syndrome
				1217	Hamano-Tsukamoto syndrome
				2869	Hamartomatous intestinal polyposis
				93946	Hamel cerebro-palato-cardiac syndrome
				79126	Hamman-Rich 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
73229	HANAC syndrome	324708	HCHWA, Iowa type	90053	Hematopoietic stem cell transplantation
1927	Hand and foot deformity - flat facies	324713	HCHWA, Italian type	2128	Hemi 3 syndrome
2438	Hand-foot-genital syndrome	324703	HCHWA, Piedmont type	86908	Hemiconvulsion-hemiplegia-epilepsy syndrome
2438	Hand-foot-uterus syndrome	100006	HCHWA-D	2128	Hemicorporal hypertrophy
99873	Hand-Schüller-Christian disease	58017	HCL	443070	Hemicrania continua
989	Hanhart syndrome	300878	HCL-v	306741	Hemidystonia-hemiatrophy syndrome
186	Hanot syndrome	163690	HCS	1214	Hemifacial atrophy
340	Hantavirosis	306741	HD-HA syndrome	1241	Hemifacial hyperplasia - strabismus
340	Hantavirus fever	26106	HDGC	141145	Hemifacial hypertrophy
319247	Hantavirus pulmonary syndrome	157941	HDL1	141136	Hemifacial microsomia
3294	Hapnes-Boman-Skeie syndrome	98934	HDL2	2549	Hemifacial microsomia - radial defects
1490	Harboyan syndrome	157946	HDL3	141148	Hemifacial myohyperplasia
899	HARD syndrome	98759	HDL4	276280	Hemihyperplasia-multiple lipomatosis syndrome
2812	Hard-skin syndrome, Parana type	313808	HDLS	99802	Hemimegalencephaly
85182	Hardcastle syndrome	2237	HDR syndrome	306669	Hemiparkinsonism-hemiatrophy syndrome
1415	Hardikar syndrome	288	HE	99050	Hemitruncus arteriosus
1177	Harding ataxia	67037	Head and neck squamous cell carcinoma	139491	Hemochromatosis due to defect in ferroportin
457	Harlequin ichthyosis	254898	Hearing loss - encephaloneuropathy - obesity - valvulopathy	79230	Hemochromatosis type 2
199282	Harlequin syndrome	3225	Hearing loss - familial salivary gland insensitivity to aldosterone	225123	Hemochromatosis type 3
→21686 6	HARP syndrome	1355	Heart defect - round face - congenital developmental delay	139491	Hemochromatosis type 4
2115	Harrod syndrome	1338	Heart defect-tongue hamartoma-polysyndactyly syndrome	447792	Hemochromatosis type 5
2116	Hartnup disease	1354	Heart defects - limb shortening	163596	Hemoglobin Bart's hydrops fetalis
2116	Hartnup disorder	875	Heart tumor of the child	231242	Hemoglobin C - beta-thalassemia
2117	Hartsfield-Bixler-Demyer syndrome	392	Heart-hand syndrome type 1	2132	Hemoglobin C disease
84085	HAS	1350	Heart-hand syndrome type 2	90039	Hemoglobin D disease
83601	Hashimoto encephalitis	1342	Heart-hand syndrome type 3	231249	Hemoglobin E - beta-thalassemia
99872	Hashimoto-Pritzker syndrome	168796	Heart-hand syndrome, Slovenian type	2133	Hemoglobin E disease
2994	Haspeslagh-Fryns-Muelenaere syndrome	1342	Heart-hand syndrome, Spanish type	93616	Hemoglobin H disease
3325	HAT	1342	Heart-limb syndrome type 3	330032	Hemoglobin Lepore - beta-thalassemia
2118	Hawkinsinuria	442582	Heavy chain amyloidosis	330041	Hemoglobin M disease
1071	Hay-Wells syndrome	93556	Heavy chain deposition disease	280615	Hemoglobinopathy Toms River
163596	Hb Bart's hydrops fetalis	86864	Heavy chain disease	86817	Hemolytic anemia due to adenylate kinase deficiency
231242	HbC - beta-thalassemia	2119	HEC syndrome	714	Hemolytic anemia due to diphosphoglycerate mutase deficiency
231249	HbE - beta-thalassemia	3377	Hecht syndrome	99138	Hemolytic anemia due to erythrocyte adenosine deaminase overproduction
93616	HbH disease	3377	Hecht-Beals syndrome	712	Hemolytic anemia due to glucophosphate isomerase deficiency
352657	HBID	2492	Hecht-Scott syndrome	90030	Hemolytic anemia due to glutathione reductase deficiency
330032	HbLepore - beta-thalassemia	238468	HED	248305	Hemolytic anemia due to glyceraldehyde-3-phosphate dehydrogenase deficiency
251359	HbS - beta-thalassemia	98813	HED-ID	35120	Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency
251365	HbSC disease	2787	Heide syndrome	766	Hemolytic anemia due to red cell pyruvate kinase deficiency
251370	HbSD disease	3220	Heimler syndrome		
251375	HbSE disease	99932	Heiner syndrome		
363412	HBSL	178330	Heinz body anemia		
88673	HCC	86813	Helicoid peripapillary chorioretinal degeneration		
86864	HCD	168782	Heller syndrome		
93556	HCDD	252054	Hemangioblastoma		
85458	HCHWA	2330	Hemangioma-thrombocytopenia syndrome		
324723	HCHWA, Arctic type				
100006	HCHWA, Dutch type				
324718	HCHWA, Flemish type				
100008	HCHWA, Icelandic type				

→ 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
275944	Hemolytic disease of the newborn with Kell alloimmunization	369	Hepatic glycogen phosphorylase deficiency	3115	Hereditary areflexic dystasia, Roussy-Lévy type
90038	Hemolytic-uremic syndrome with diarrhea	369	Hepatic phosphorylase deficiency	289601	Hereditary arterial and articular multiple calcification syndrome
2134	Hemolytic-uremic syndrome without diarrhea	890	Hepatic veno-occlusive disease	1416	Hereditary articular chondrocalcinosis
93581	Hemolytic-uremic syndrome without diarrhea with anti-factor H antibodies	79124	Hepatic veno-occlusive disease - immunodeficiency	1429	Hereditary benign chorea
93578	Hemolytic-uremic syndrome without diarrhea with B factor anomaly	90073	Hepatitis B reinfection following liver transplantation	352657	Hereditary benign corneal intraepithelial dyskeratosis
93575	Hemolytic-uremic syndrome without diarrhea with C3 anomaly	402823	Hepatitis delta	352657	Hereditary benign intraepithelial dyskeratosis
357008	Hemolytic-uremic syndrome without diarrhea with DGKE deficiency	449	Hepatoblastoma	91378	Hereditary bradykinin-induced angioedema
93579	Hemolytic-uremic syndrome without diarrhea with H factor anomaly	54272	Hepatocellular adenoma	221061	Hereditary brain cavernous angioma
93580	Hemolytic-uremic syndrome without diarrhea with I factor anomaly	88673	Hepatocellular carcinoma	221061	Hereditary brain cavernous hemangioma
93576	Hemolytic-uremic syndrome without diarrhea with MCP/CD46 anomaly	137681	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1	145	Hereditary breast and ovarian cancer syndrome
217023	Hemolytic-uremic syndrome without diarrhea with thrombomodulin anomaly	137681	Hepatoencephalopathy due to COXPD1	227535	Hereditary breast cancer
158048	Hemophagocytic syndrome associated with an infection	95159	Hepatoerythropoietic porphyria	227535	Hereditary breast carcinoma
98878	Hemophilia A	905	Hepatolenticular degeneration	871	Hereditary bundle branch defect
98879	Hemophilia B	64743	Hepatoportal sclerosis	36382	Hereditary CAD
329	Hemophilia C	364	Hepatorenal glycogenosis	1416	Hereditary calcium pyrophosphate deposition
178396	Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation	882	Hepatorenal tyrosinemia	1416	Hereditary CC
340	Hemorrhagic fever - renal syndrome	86882	Hepatosplenic T-cell lymphoma	30925	Hereditary CDI
274	Hemorrhagiparous thrombocytic dystrophy	306539	Hereditary acrokeratotic poikiloderma of Kindler-Weary	30925	Hereditary central diabetes insipidus
324632	Hendra virus infection	2907	Hereditary acrokeratotic poikiloderma, Weary type	221061	Hereditary cerebral cavernoma
2136	Hennekam syndrome	447964	Hereditary adult-onset painful axonal polyneuropathy	221061	Hereditary cerebral cavernous malformation
2135	Hennekam-Beemer syndrome	85450	Hereditary amyloid nephropathy	85458	Hereditary cerebral hemorrhage with amyloidosis
761	Henoch-Schönlein purpura	93560	Hereditary amyloid nephropathy due to apolipoprotein A-I variant	324723	Hereditary cerebral hemorrhage with amyloidosis, Arctic type
95159	HEP	238269	Hereditary amyloid nephropathy due to apolipoprotein A-II variant	100006	Hereditary cerebral hemorrhage with amyloidosis, Dutch type
79269	Heparan sulfamidase deficiency	93562	Hereditary amyloid nephropathy due to fibrinogen A alpha-chain variant	324718	Hereditary cerebral hemorrhage with amyloidosis, Flemish type
79271	Heparan-alpha-glucosaminide N-acetyltransferase deficiency	93561	Hereditary amyloid nephropathy due to lysozyme variant	100008	Hereditary cerebral hemorrhage with amyloidosis, Icelandic type
3325	Heparin-associated thrombocytopenia	85450	Hereditary amyloidosis with primary renal involvement	324708	Hereditary cerebral hemorrhage with amyloidosis, Iowa type
3325	Heparin-induced thrombocytopenia	85448	Hereditary amyloidosis, Finnish type	324713	Hereditary cerebral hemorrhage with amyloidosis, Italian type
3325	Heparin-induced thrombocytopenia type 2	228277	Hereditary anetoderma	324703	Hereditary cerebral hemorrhage with amyloidosis, Piedmont type
102069	Hepatic amyloidosis with intrahepatic cholestasis	91378	Hereditary angioneurotic edema	48818	Hereditary ceruloplasmin deficiency
156	Hepatic carnitine palmitoyl transferase 1 deficiency	100050	Hereditary angioneurotic edema type 1	36382	Hereditary cervical artery dissection
156	Hepatic carnitine palmitoyl transferase I deficiency	100051	Hereditary angioneurotic edema type 2	53372	Hereditary chin myoclonus
386	Hepatic cystic hamartoma	100054	Hereditary angioneurotic edema type 3	53372	Hereditary chin-trembling
2031	Hepatic fibrosis - renal cysts - intellectual disability	91378	Hereditary angioneurotic edema	676	Hereditary chronic pancreatitis
		100050	Hereditary angioneurotic edema type 1	422526	Hereditary clear cell renal cell adenocarcinoma
		100051	Hereditary angioneurotic edema type 2	422526	Hereditary clear cell renal cell carcinoma
		100054	Hereditary angioneurotic edema type 3		
		73229	Hereditary angiopathy-nephropathy-aneurysms-muscle cramps 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
293144	Hereditary clubfoot due to 5q31 microdeletion	774	Hereditary hemorrhagic telangiectasia	64751	Hereditary motor and sensory neuropathy type 5
238578	Hereditary clubfoot due to 17q23.1-q23.2 microduplication	2604	Hereditary hollow visceral myopathy	90120	Hereditary motor and sensory neuropathy type 6
293150	Hereditary clubfoot due to PITX1 point mutation	199285	Hereditary hypercarotenemia and vitamin A deficiency	90119	Hereditary motor and sensory neuropathy with acrodystrophy
98434	Hereditary combined deficiency of factors II, VII, IX and X	238475	Hereditary hypercholanemia	90103	Hereditary motor and sensory neuropathy with deafness, intellectual disability and absent sensory large myelinated fibers
98434	Hereditary combined deficiency of vitamin K-dependent clotting factors	3197	Hereditary hyperekplexia		
238722	Hereditary congenital contralateral synkinesia	3197	Hereditary hyperexplexia		
238722	Hereditary congenital mirror movements	163	Hereditary hyperferritinemia with congenital cataracts	99950	Hereditary motor and sensory neuropathy, Lom type
972	Hereditary continuous muscle fiber activity	163	Hereditary hyperferritinemia-cataract syndrome	90117	Hereditary motor and sensory neuropathy, Okinawa type
79273	Hereditary coproporphyria	2801	Hereditary hyperphosphatasia	90117	Hereditary motor and sensory neuropathy, proximal type
60015	Hereditary cranium bifidum	157215	Hereditary hypophosphatemic rickets with hypercalciuria	99953	Hereditary motor and sensory neuropathy, Russe Type
168577	Hereditary cryohydrocytosis type 2	55654	Hereditary hypotrichosis simplex	1839	Hereditary mucoepithelial dysplasia
398088	Hereditary cryohydrocytosis with normal stomatin	90368	Hereditary hypotrichosis simplex of the scalp	171723	Hereditary mucosal leukokeratosis
168577	Hereditary cryohydrocytosis with reduced stomatin	217407	Hereditary hypotrichosis with recurrent skin vesicles	136	Hereditary multi-infarct dementia
98967	Hereditary crystalline stromal dystrophy of Schnyder	79091	Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia	→3460	Hereditary multicentric osteolysis
100008	Hereditary cystatin C amyloid angiopathy	602	Hereditary inclusion body myopathy type 2	523	Hereditary multiple cutaneous leiomyomas
26106	Hereditary diffuse cancer of stomach	79091	Hereditary inclusion body myopathy type 3	83454	Hereditary multiple glomangiomas
26106	Hereditary diffuse gastric adenocarcinoma	324381	Hereditary inclusion body myopathy type 4	2590	Hereditary myoclonus - progressive distal muscular atrophy
26106	Hereditary diffuse gastric cancer	178464	Hereditary inclusion body myopathy with early respiratory failure	43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency
313808	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia	300373	Hereditary infantile gigantism	1062	Hereditary neurocutaneous angioma
313808	Hereditary diffuse leukoencephalopathy with spheroids	397692	Hereditary isolated aplastic anemia	30925	Hereditary neurogenic diabetes insipidus
288	Hereditary elliptocytosis	332	Hereditary juvenile megaloblastic anemia due to intrinsic factor deficiency	640	Hereditary neuropathy with liability to pressure palsies
63261	Hereditary endotheliopathy - retinopathy - nephropathy - stroke	2334	Hereditary keratitis	279943	Hereditary neutrophilia
98873	Hereditary erythroblastic multinuclearity with a positive acidified-serum test (hempas)	493	Hereditary keratoacanthoma	91378	Hereditary non histamine-induced angioedema
36899	Hereditary essential myoclonus	411602	Hereditary late-onset Parkinson disease	168583	Hereditary North American Indian childhood cirrhosis
85195	Hereditary expansile polyostotic osteolytic dysplasia	523	Hereditary leiomyomatosis	56	Hereditary ochronosis
157846	Hereditary ferritinopathy	523	Hereditary leiomyomatosis and renal cell cancer	30	Hereditary orotic aciduria
90045	Hereditary folate malabsorption	79452	Hereditary lymphedema type I	98868	Hereditary ovalocytosis
469	Hereditary fructose intolerance	90186	Hereditary lymphedema type II	79141	Hereditary painful callosities
469	Hereditary fructose-1-phosphate aldolase deficiency	228277	Hereditary macular atrophy	86923	Hereditary palmoplantar hyperkeratosis, Gamborg-Nielsen type
469	Hereditary fructosemia	621	Hereditary methemoglobinemia	86923	Hereditary palmoplantar keratoderma, Gamborg-Nielsen type
53372	Hereditary geniospasm	330041	Hereditary methemoglobinemia due to hemoglobin mutation	47044	Hereditary papillary renal cell carcinoma
2024	Hereditary gingival fibromatosis	157794	Hereditary mixed polyposis syndrome	99878	Hereditary parathyroids hyperplasia
2024	Hereditary gingival hyperplasia	64748	Hereditary motor and sensory neuropathy type 3	168615	Hereditary persistence of alpha-fetoprotein
		773	Hereditary motor and sensory neuropathy type 4	46532	Hereditary persistence of fetal hemoglobin - beta-thalassemia

→ 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
251380	Hereditary persistence of fetal hemoglobin - sickle cell disease	64752	Hereditary sensory and autonomic neuropathy type 5	3202	Hereditary xerocytosis
29072	Hereditary pheochromocytoma-paraganglioma	314381	Hereditary sensory and autonomic neuropathy type 6	773	Heredopathia atactica polyneuritiformis
300373	Hereditary pituitary hyperplasia	391397	Hereditary sensory and autonomic neuropathy type 7	275777	Heritable pulmonary arterial hypertension
330061	Hereditary polymorphous light eruption of American Indians	139573	Hereditary sensory and autonomic neuropathy with deafness and global delay	3411	Herlyn-Werner syndrome
178345	Hereditary prepubertal gynecomastia	391397	Hereditary sensory and autonomic neuropathy with hyperhidrosis and gastrointestinal dysfunction	79430	Hermansky-Pudlak syndrome
828	Hereditary progressive arthroophthalmopathy	139578	Hereditary sensory and autonomic neuropathy with spastic paraparesis	183678	Hermansky-Pudlak syndrome type 2
98808	Hereditary progressive dystonia with marked diurnal fluctuation	213524	Hereditary site-specific ovarian cancer syndrome	231531	Hermansky-Pudlak syndrome type 7
158025	Hereditary progressive mucinous histiocytosis	100996	Hereditary spastic paraparesis type 15	231537	Hermansky-Pudlak syndrome type 8
178464	Hereditary proximal myopathy with early respiratory failure	822	Hereditary spherocytosis	280663	Hermansky-Pudlak syndrome type 9
264675	Hereditary pulmonary alveolar proteinosis	84093	Hereditary thermosensitive neuropathy	183678	Hermansky-Pudlak syndrome with neutropenia
440427	Hereditary pulmonary alveolar proteinosis with hepatic involvement	71493	Hereditary thrombocythemia	231500	Hermansky-Pudlak syndrome with pulmonary fibrosis
275777	Hereditary pulmonary arterial hypertension	268322	Hereditary thrombocytopenia with normal platelets	231512	Hermansky-Pudlak syndrome without pulmonary fibrosis
→288	Hereditary pyropoikilocytosis	329319	Hereditary thrombocytosis with transverse limb defect	2139	Hernández-Aguirre Negrete syndrome
85450	Hereditary renal amyloidosis	82	Hereditary thrombophilia due to congenital antithrombin 3 deficiency	2786	Hernández-Fragoso syndrome
93560	Hereditary renal amyloidosis due to apolipoprotein A-I variant	82	Hereditary thrombophilia due to congenital antithrombin deficiency	63261	HERNS syndrome
238269	Hereditary renal amyloidosis due to apolipoprotein A-II variant	217467	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	1930	Herpes simplex encephalitis
93562	Hereditary renal amyloidosis due to fibrinogen A alpha-chain variant	217467	Hereditary thrombophilia due to congenital HRG deficiency	1930	Herpes simplex meningo-encephalitis
93561	Hereditary renal amyloidosis due to lysozyme variant	745	Hereditary thrombophilia due to congenital protein C deficiency	1930	Herpetic encephalitis
94088	Hereditary renal hypouricemia	743	Hereditary thrombophilia due to congenital protein S deficiency	208524	Herpetiform pemphigus
788	Hereditary resistance to anti-vitamin K	745	Hereditary thrombophilia due to PC deficiency	369	Hers disease
357027	Hereditary retinoblastoma	205	Hereditary unconjugated hyperbilirubinemia	1486	Herva disease
221043	Hereditary sclerosing poikiloderma with tendon and pulmonary involvement	79234	Hereditary unconjugated hyperbilirubinemia type 1	314970	HES-L
221039	Hereditary sclerosing poikiloderma, Weary type	79235	Hereditary unconjugated hyperbilirubinemia type 2	314950	HES-M
280598	Hereditary sensorimotor neuropathy with hyperelastic skin	71291	Hereditary vascular retinopathy	314950	HES-N
36386	Hereditary sensory and autonomic neuropathy type 1	71291	Hereditary vascular retinopathy - Raynaud phenomenon - migraine	314962	HES-R
139564	Hereditary sensory and autonomic neuropathy type 1 with cough and gastroesophageal reflux	93160	Hereditary vitamin D-resistant rickets	640	Heterozygous microdeletion 17p11.2p12
139564	Hereditary sensory and autonomic neuropathy type 1B	903	Hereditary von Willebrand disease	3450	Heterozygous OSMED
970	Hereditary sensory and autonomic neuropathy type 2	98805	Hereditary whispering dysphonia	3450	Heterozygous otospondylomegaepiphyseal dysplasia
1764	Hereditary sensory and autonomic neuropathy type 3	170	Hereditary woolly hair syndrome	845	Hexosaminidase A deficiency
642	Hereditary sensory and autonomic neuropathy type 4	170	Hereditary wooly hair syndrome	309192	Hexosaminidase A deficiency, adult form
		3467	Hereditary xanthinuria	309239	Hexosaminidase A deficiency, B1 variant
				309178	Hexosaminidase A deficiency, infantile form
				309185	Hexosaminidase A deficiency, juvenile form
				309246	Hexosaminidase activator deficiency
				796	Hexosaminidases A and B deficiency
				309169	Hexosaminidases A and B deficiency, adult form
				309155	Hexosaminidases A and B deficiency, infantile form
				309162	Hexosaminidases A and B deficiency, juvenile form
				1041	HF

→ 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
2438	HFGS	411593	Hirata disease	64751	HMSN 5
2744	HGPPS	65684	Hirayama disease	401964	HMSN2 with giant axons
740	HGPS	388	Hirschsprung disease	90119	HMSN with acrodystryphy
79271	HGSNAT deficiency	2155	Hirschsprung disease - deafness - polydactyly	99950	HMSN, Lom type
163	HHCS	2151	Hirschsprung disease - ganglioneuroblastoma	99950	HMSN-Lom
86908	HHE syndrome	2152	Hirschsprung disease - intellectual disability	90117	HMSNP
415	HHH syndrome	2150	Hirschsprung disease - type D brachydactyly	99953	HMSNR
276280	HHML	261537	Hirschsprung disease and intellectual disability due to 2q22 microdeletion	69084	HNED
157215	HRRH	261552	Hirschsprung disease and intellectual disability due to a ZEB2 point mutation	93111	HNF1B-MODY
774	HHT	261537	Hirschsprung disease and intellectual disability due to del(2)(q22)	640	HNPP
457	HI	261537	Hirschsprung disease and intellectual disability due to monosomy 2q22	67037	HNSCC
435	HI syndrome	2153	Hirschsprung disease-nail hypoplasia-dysmorphism	1979	Hoepffner-Dreyer-Reimers syndrome
35878	HI/HA syndrome	2026	Hirsutism-congenital gingival hyperplasia syndrome	2349	Hoffman syndrome
88639	HIBCH deficiency	2156	Hirsutism-skeletal dysplasia-intellectual disability syndrome	391665	HoFH
602	HIBM2	3283	His bundle tachycardia	414	HOGA
79091	HIBM3	2157	HIS deficiency	329173	HOIL1 deficiency
324381	HIBM4	2157	Histidase deficiency	→994	Holmes-Benacerraf syndrome
178464	HIBM-ERF	2157	Histidine ammonia-lyase deficiency	3328	Holmes-Collins syndrome
189	Hidrotic ectodermal dysplasia	2157	Histidinemia	93970	Holmes-Gang syndrome
1808	Hidrotic ectodermal dysplasia, Christianson-Fourie type	2157	Histidinuria	2143	Holmes-Schepens syndrome
1809	Hidrotic ectodermal dysplasia, Halal type	2158	Histidinuria - renal tubular defect	79242	Holocarboxylase synthetase deficiency
343	HIDS	50918	Histiocytic necrotizing lymphadenitis	2162	Holoprosencephaly
137577	HIE	86896	Histiocytic sarcoma	2165	Holoprosencephaly - caudal dysgenesis
330012	High altitude pulmonary edema	137675	Histiocytoid cardiomyopathy	2163	Holoprosencephaly - craniostostosis
171201	High anorectal malformation	390	Histoplasmosis	2117	Holoprosencephaly - ectrodactyly - cleft lip palate
314029	High bone mass OI	3325	HIT	2166	Holoprosencephaly - postaxial polydactyly
314029	High bone mass osteogenesis imperfecta	→138	Hittner-Hirsch-Kreh syndrome	3186	Holoprosencephaly - radial heart renal anomalies
363396	High myopia-sensorineural deafness syndrome	443291	HIV-associated cancer	2570	Holoprosencephaly-fetal akinesia/hypokinesia sequence syndrome
3181	High scapula	443291	HIV-related cancer	2570	Holoprosencephaly-hypokinesia-congenital contractures syndrome
231080	High-grade dysplasia in patients with Barrett esophagus	1573	HJMD	392	Holt-Oram syndrome
251646	High-grade ependymoma	572	HLA class 2-negative severe combined immunodeficiency	2167	Holzgreve-Wagner-Rehder syndrome
101088	HIGM1	2248	HLHS	30924	HOMG1
101089	HIGM2	412	HLP type 3	34528	HOMG2
101090	HIGM3	523	HLRCC	31043	HOMG3
101091	HIGM4	2213	HMC syndrome	2168	Homocarnosinase deficiency
101092	HIGM5	178464	HMERF	2168	Homocarnosinosis
183663	HIGM with susceptibility to opportunistic infections	20	HMG-CoA lyase deficiency	394	Homocystinuria due to cystathionine beta-synthase deficiency
183666	HIGM without susceptibility to opportunistic infections	35701	HMG-CoA synthase deficiency	395	Homocystinuria due to methylene tetrahydrofolate reductase deficiency
99978	Hilar CCA	157794	HMPS	622	Homocystinuria without methylmalonic aciduria
99978	Hilar cholangiocarcinoma	64748	HMSN 3	56	Homogentisic acid oxidase deficiency
84085	Hinman syndrome	773	HMSN 4	163596	Homozygous alpha0-thalassemia
84085	Hinman-Allen syndrome			391665	Homozygous familial hypercholesterolemia
1164	Hinson-Pepys disease				
3408	Hip dysplasia - enchondromata - ecchondroma				
2114	Hip dysplasia, Beukes type				

→ 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
14	Homozygous familial hypobetalipoproteinemia	391397	HSAN with hyperhidrosis and gastrointestinal dysfunction	3266	Humero-radio-ulnar synostosis
→288	Homozygous hereditary elliptocytosis	139578	HSAN with spastic paraplegia	295207	Humero-radio-ulnar synostosis, bilateral
98960	Honeycomb corneal dystrophy	2182	HSAS	295205	Humero-radio-ulnar synostosis, unilateral
78	Hookworm infection	388	HSCR	→26346 3	Humero-spinal dysostosis
307936	HOPP syndrome	391417	HSD10 deficiency	94056	Humero-ulnar fusion
2744	Horizontal gaze palsy with progressive scoliosis	85295	HSD10 deficiency, atypical type	295215	Humero-ulnar fusion, bilateral
397	Horton disease	391428	HSD10 deficiency, classic type	295213	Humero-ulnar fusion, unilateral
392	HOS	391428	HSD10 deficiency, infantile type	94056	Humero-ulnar synostosis
166412	Hot water reflex epilepsy	391457	HSD10 deficiency, neonatal type	295215	Humero-ulnar synostosis, bilateral
1352	Houlston-Ironton-Temple syndrome	391417	HSD10 disease	295213	Humero-ulnar synostosis, unilateral
99907	House allergic alveolitis	85295	HSD10 disease, atypical type	→26346 3	Humerospinal dysostosis
2198	Howell-Evans syndrome	391428	HSD10 disease, classic type	3383	Humerus trochlea aplasia
3322	Hoyeraa-Hreidarsson syndrome	391428	HSD10 disease, infantile type	580	Hunter syndrome
306669	HP-HA syndrome	391457	HSD10 disease, neonatal type	217085	Hunter syndrome type A
275777	HPAH	30924	HSH	217093	Hunter syndrome type B
98808	HPD with marked diurnal fluctuation	1930	HSV encephalitis	→35069	Hunter-Carpenter-McDonald syndrome
2162	HPE	285	HT-EDS	2715	Hunter-Jurenka-Thompson syndrome
46532	HPFH - beta-thalassemia	289326	HTLV-1-associated myelopathy/tropical spastic paraparesis	97340	Hunter-McAlpine craniostenosis
251380	HPFH - sickle cell disease	228116	Hughes-Stovin syndrome	3365	Hunter-Rudd-Hoffmann syndrome
247262	HPMR	438279	Human infection by orthopoxvirus	1390	Hunter-Thompson-Reed syndrome
436	HPP	289326	Human T-lymphotropic virus type I-associated myelopathy/tropical spastic paraparesis	399	Huntington chorea
293958	HPPD	289326	Human T-lymphotropic virus type-1-associated myelopathy/tropical spastic paraparesis	399	Huntington disease
47044	HPRCC	294973	Humeral agenesis/hypoplasia	401901	Huntington disease phenocopy due to C9ORF72 expansions
79233	HPRT1 partial deficiency	295063	Humeral agenesis/hypoplasia, bilateral	157941	Huntington disease-like 1
510	HPRT complete deficiency	295061	Humeral agenesis/hypoplasia, unilateral	98934	Huntington disease-like 2
510	HPRT deficiency grade IV	294973	Humeral intercalary meromelia	157946	Huntington disease-like 3
79233	HPRT deficiency, grade I	295063	Humeral intercalary meromelia, bilateral	98759	Huntington disease-like 4
79233	HPRT partial deficiency	295061	Humeral intercalary meromelia, unilateral	401901	Huntington disease-like syndrome due to C9ORF72 expansions
79233	HPRT-related gout	3265	Humero-radial fusion	363694	HUPRA syndrome
79233	HPRT-related hyperuricemia	295211	Humero-radial fusion, bilateral	384	Huriez syndrome
79430	HPS	295209	Humero-radial fusion, unilateral	93473	Hurler disease
183678	HPS2	3265	Humero-radial synostosis	93473	Hurler syndrome
231531	HPS7	295211	Humero-radial synostosis, bilateral	93476	Hurler-Scheie syndrome
231537	HPS8	295209	Humero-radial synostosis, unilateral	330061	Hutchinson summer prurigo
280663	HPS9	3266	Humero-radio-ulnar fusion	740	Hutchinson-Gilford progeria syndrome
231500	HPS with pulmonary fibrosis	295207	Humero-radio-ulnar fusion, bilateral	93160	HVDRR
231512	HPS without pulmonary fibrosis	295205	Humero-radio-ulnar fusion, unilateral	71291	HVR
99880	HPT-JT	294975	Humero-radio-ulnar intercalary transverse meromelia	53698	Hyaline body myopathy
2323	HRD syndrome	295087	Humero-radio-ulnar intercalary transverse meromelia, bilateral	70587	Hyaline membrane disease
84085	HS	295085	Humero-radio-ulnar intercalary transverse meromelia, unilateral	530	Hyalinosis cutis et mucosae
139564	HSAN1B			67041	Hyaluronidase deficiency
970	HSAN2			400	Hydatid disease
1764	HSAN3			99927	Hydatidiform mole
642	HSAN4			400	Hydatidosis
64752	HSAN5				
314381	HSAN6				
391397	HSAN7				
139564	HSAN with cough and gastroesophageal reflux				
139573	HSAN with deafness and global delay				

→ 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
2898	Hyde Forster-McCarthy-Berry syndrome	101091	Hyper-IgM syndrome type 4	263455	Hyperinsulinemic hypoglycemia due to HNF4A deficiency
2177	Hydranencephaly	101092	Hyper-IgM syndrome type 5	263458	Hyperinsulinemic hypoglycemia due to INSR deficiency
330021	Hydargyria	183663	Hyper-IgM syndrome with susceptibility to opportunistic infections	263458	Hyperinsulinemic hypoglycemia due to insulin receptor deficiency
330061	Hydroa aestivale	183666	Hyper-IgM syndrome without susceptibility to opportunistic infections	276603	Hyperinsulinemic hypoglycemia due to Kir6.2 deficiency, diazoxide-resistant focal form
330058	Hydroa vacciniforme	309147	Hyperalaninemia	71212	Hyperinsulinemic hypoglycemia due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency
364039	Hydroa vacciniforme-like lymphoma	927	Hyperammonemia due to N-acetylglutamate synthase deficiency	276598	Hyperinsulinemic hypoglycemia due to SUR1 deficiency, diazoxide-resistant focal form
899	Hydrocephalus - agyria - retinal dysplasia	401948	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	276556	Hyperinsulinemic hypoglycemia due to UCP2 deficiency
2186	Hydrocephalus - blue sclerae - nephropathy	168588	Hyperandrogenism due to cortisone reductase deficiency	79299	Hyperinsulinism due to glucokinase deficiency
1237	Hydrocephalus - cardiac malformation - dense bones	90	Hyperargininemia	71212	Hyperinsulinism due to glutamodehydrogenase deficiency
916	Hydrocephalus - cleft palate - joint contractures	234	Hyperbilirubinemia type 2	71212	Hyperinsulinism due to HADH deficiency
2180	Hydrocephalus - costovertebral dysplasia - Sprengel anomaly	3111	Hyperbilirubinemia, Rotor type	324575	Hyperinsulinism due to HNF1A deficiency
2119	Hydrocephalus - endocardial fibroelastosis - cataract	276405	Hyperbiliverdinemia	263455	Hyperinsulinism due to HNF4A deficiency
2183	Hydrocephalus - obesity - hypogonadism	306661	Hypercalcemic tumoral calcinosis	263458	Hyperinsulinism due to INSR deficiency
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	2196	Hypercalciuria - bilateral macular coloboma	165991	Hyperinsulinism due to monocarboxylate transporter 1 deficiency
899	Hydrocephalus-agyria-retinal dysplasia syndrome	209902	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	71212	Hyperinsulinism due to SCHAD deficiency
2184	Hydrocephaly - low insertion umbilicus	83639	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency	71212	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency
2181	Hydrocephaly - tall stature - joint laxity	1032	Hyperdibasic aminoaciduria type 1	165991	Hyperinsulinism due to SLC16A1 deficiency
221126	Hydrocephaly/hydranencephaly due to cerebral vasculopathy	470	Hyperdibasic aminoaciduria type 2	276556	Hyperinsulinism due to UCP2 deficiency
2189	Hydrolethalus	3197	Hyperekplexia	35878	Hyperinsulinism-hyperammonemia syndrome
2473	Hydrometrocolpos - postaxial polydactyly	163985	Hyperekplexia - epilepsy	682	Hyperkalemic periodic paralysis
2704	Hydronephrosis - inverted smile	408	Hyperglycerolemia	682	Hyperkalemic PP
1426	Hydrops - ectopic calcification - moth-eaten	2410	Hypergonadotropic hypogonadism - cataract syndrome	757	Hyperkaliemia - hypertension, Gordon type
1041	Hydrops fetalis	243	Hypergonadotropic ovarian dysgenesis	409	Hyperkeratosis lenticularis perstans
20	Hydroxymethylglutaric aciduria	2157	Hyperhistidinemia	1662	Hyperkeratosis-contracture syndrome
401	Hymenolepiasis	742	Hyperimidodipeptiduria	1336	Hyperkeratosis-hyperpigmentation syndrome
309147	Hyper-beta-alaninemia	343	Hyperimmunoglobulinemia D with recurrent fever	682	HyperKPP
343	Hyper-IgD syndrome	2314	Hyperimmunoglobulin E syndrome type 1	140905	Hyperlipidemia due to hepatic triglyceride lipase deficiency
101090	Hyper-IgM syndrome due to CD40 deficiency	2314	Hyperimmunoglobulin E-recurrent infection syndrome	412	Hyperlipidemia type 3
101088	Hyper-IgM syndrome due to CD40 ligand deficiency	343	Hyperimmunoglobulinemia D syndrome	411	Hyperlipoproteinemia type 1
101088	Hyper-IgM syndrome due to CD40L deficiency	343	Hyperimmunoglobulinemia D with periodic fever		
101092	Hyper-IgM syndrome due to UNG deficiency	79299	Hyperinsulinemic hypoglycemia due to glucokinase deficiency		
101092	Hyper-IgM syndrome due to uracil N-glycosylase	324575	Hyperinsulinemic hypoglycemia due to HNF1A deficiency		
101088	Hyper-IgM syndrome type 1				
101089	Hyper-IgM syndrome type 2				
101090	Hyper-IgM syndrome type 3				

→ 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
412	Hyperlipoproteinemia type 3	238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	329883	Hypertrophic gastropathy without hypoproteinemia
413	Hyperlipoproteinemia type 4	1578	Hyperphenylalaninemia with primapterinuria	64748	Hypertrophic neuropathy of infancy
70470	Hyperlipoproteinemia type 5	2209	Hyperphenylalaninemic embryopathy	90282	Hypertrophic or verrucous lupus erythematosus
2203	Hyperlysineuria	247262	Hyperphosphatasia-intellectual disability syndrome	2224	Hypertryptophanemia
2203	Hyperlysineuria type I	→79189	Hyperpipecolatemia	217330	Hyperuricemia - anemia - renal failure
3124	Hyperlysineuria type II	157798	Hyperplastic polyposis syndrome	363694	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome
289891	Hypermethioninemia due to glycine N-methyltransferase deficiency	682	HyperPP	251523	Hyperzincemia and hypercalprotectinemia
289891	Hypermethioninemia due to GNMT deficiency	419	Hyperprolinemia type 1	276429	Hypnic headache
	Hypermethioninemia due to S-adenosylhomocysteine hydrolase deficiency	79101	Hyperprolinemia type 2	2435	Hypo- and hypermelanotic cutaneous macules - retarded growth - intellectual disability
88618		93604	Hyperprostaglandin E syndrome	289157	Hypocalcemic vitamin D-dependent rickets
289290	Hypermethioninemia encephalopathy due to adenosine kinase deficiency	889	Hypersensitivity angiitis	93160	Hypocalcemic vitamin D-resistant rickets
289290	Hypermethioninemia encephalopathy due to ADK deficiency	2211	Hypertelorism - hypospadias - polysyndactyly syndrome	100032	Hypocalcified amelogenesis imperfecta
73267	Hypernychthemeral syndrome	1519	Hypertelorism, Teebi type	93297	Hypochondrogenesis
414	Hyperornithinemia	2213	Hypertelorism-microtia-facial clefting syndrome	429	Hypochondroplasia
414	Hyperornithinemia - gyrate atrophy of choroid and retina	2745	Hypertelorism-oesophageal abnormality-hypospadias syndrome	36412	Hypocomplementemic urticarial vasculitis
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	293958	Hypertelorism-preauricular sinus-punctual pits-deafness syndrome	2228	Hypodontia - dysplasia of nails
2801	Hyperostosis corticalis deformans juvenilis	293958	Hypertelorism-preauricular sinus-punctual pits-hearing loss syndrome	2228	Hypodontia - nail dysgenesis
3416	Hyperostosis corticalis generalisata	88660	Hypertension due to gain-of-function mutations in the mineralocorticoid receptor	185	Hypogenetic lung syndrome
443098	Hyperostosis cranialis interna	757	Hypertensive hyperkalemia	989	Hypoglossia-hypodactyly syndrome
77296	Hyperostosis frontalis interna	423	Hyperthermia of anesthesia	→26148	Hypogonadism - gynecomastia - X-linked intellectual disability
2780	Hyperostosis generalisata with striations	1231	Hypertrichosis - atrophic skin - ectropion - macrostomia	2233	Hypogonadism - mitral valve prolapse - intellectual disability
99880	Hyperparathyroidism-jaw tumor syndrome	2220	Hypertrichosis cubiti - short stature	141333	Hypogonadism-short stature-coloboma-preaxial polydactyly syndrome
295002	Hyperphalangy	2222	Hypertrichosis lanuginosa congenita	2230	Hypogonadotropic hypogonadism - frontoparietal alopecia
295140	Hyperphalangy in digits 2-5	2222	Hypertrichosis universalis	2235	Hypogonadotropic hypogonadism - retinitis pigmentosa
295142	Hyperphalangy, bilateral	2026	Hypertrichosis with or without gingival hyperplasia	293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural deafness-dysmorphism syndrome
295140	Hyperphalangy, unilateral	966	Hypertrichosis-acromegaloid facial appearance syndrome	293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome
1388	Hyperphalangy-clinodactyly of index finger with Pierre Robin syndrome	966	Hypertrichosis-acromegaloid facial features syndrome	363523	Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome
238583	Hyperphenylalaninemia	966	Hypertrichosis-coarse face syndrome	238468	Hypohidrotic ectodermal dysplasia
13	Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency	319182	Hypertrichosis-short stature-facial dysmorphism-developmental delay syndrome	1882	Hypohidrotic ectodermal dysplasia - hypothyroidism - ciliary dyskinesia
238583	Hyperphenylalaninemia due to BH4 deficiency	1517	Hypertrichotic osteochondrodysplasia, Cantu type	98813	Hypohidrotic ectodermal dysplasia with immunodeficiency
1578	Hyperphenylalaninemia due to dehydratase deficiency	324525	Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation		
226	Hyperphenylalaninemia due to dihydropteridine reductase deficiency	324525	Hypertrophic cardiomyopathy and renal tubular disease due to mtDNA mutation		
2102	Hyperphenylalaninemia due to GTP cyclohydrolase deficiency	217601	Hypertrophic cardiomyopathy due to intensive athletic training		
1578	Hyperphenylalaninemia due to pterin-4-alpha-carbinolamine dehydratase deficiency				

→ 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
293964	Hypoinsulinemic hypoglycemia and body hemihypertrophy	91354	Hypopituitarism due to empty sella turcica syndrome	363424	Hypotonia-cerebral atrophy-hyperglycinemia syndrome
681	Hypokalemic periodic paralysis	1863	Hypoplasia of the femoral trochlea	163690	Hypotonia-cystinuria syndrome
30924	Hypomagnesemia caused by selective magnesium malabsorption	99058	Hypoplasia of the mitral valve annulus	371364	Hypotonia-speech impairment-severe cognitive delay syndrome
30924	Hypomagnesemia intestinal type 1	722	Hypoplasminogenemia	55654	Hypotrichosis simplex
1790	Hypomandibular faciocranial dysostosis	100031	Hypoplastic amelogenesis imperfecta	90368	Hypotrichosis simplex of the scalp
100033	Hypomaturation amelogenesis imperfecta	2248	Hypoplastic left heart syndrome	1573	Hypotrichosis with juvenile macular degeneration
100034	Hypomaturation-hypoplastic amelogenesis imperfecta with taurodontism	293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	1573	Hypotrichosis with juvenile macular dystrophy
435	Hypomelanosis of Ito	3332	Hypoplastic tibiae - postaxial polydactyly	444	Hypotrichosis, Marie Unna type
85163	Hypomyelination - congenital cataract	→21686	Hypoprebetalipoproteinemia - acanthocytosis - retinitis pigmentosa - pallidal degeneration	91132	Hypotrichosis-congenital ichthyosis syndrome
2680	Hypomyelination neuropathy - arthrogryposis	327	Hypoproconvertinemia	330029	Hypotrichosis-deafness syndrome
139441	Hypomyelination with atrophy of basal ganglia and cerebellum	2494	Hypoproteinemic hypertrophic gastropathy	2266	Hypotrichosis-intellectual disability, Lopes type
363412	Hypomyelination with brain stem and spinal cord involvement and leg spasticity	325	Hypoprothrombinemia	69735	Hypotrichosis-lymphedema-telangiectasia-membranoproliferative glomerulonephritis syndrome
447893	Hypomyelination-cerebellar atrophy-hypoplasia of the corpus callosum syndrome	2250	Hyposmia - nasal and ocular hypoplasia - hypogonadotropic hypogonadism	69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome
88637	Hypomyelination-hypogonadotropic-hypogonadism-hypodontia syndrome	157788	Hypospadias - hypertelorism - coloboma and deafness	307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar hyperkeratosis syndrome
3453	Hypoparathyroidism - Addison's disease - mucocutaneous candidiasis	2261	Hypospadias - intellectual disability, Goldblatt type	307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar keratoderma syndrome
2237	Hypoparathyroidism - deafness - renal disease	2745	Hypospadias-dysphagia syndrome	307936	Hypotrichosis-striate palmoplantar hyperkeratosis-acroosteolysis-periodontitis syndrome
2323	Hypoparathyroidism-intellectual disability-dysmorphism syndrome	2745	Hypospadias-hypertelorism syndrome	307936	Hypotrichosis-striate palmoplantar keratoderma-acroosteolysis-periodontitis syndrome
2323	Hypoparathyroidism-short stature-intellectual disability-seizures syndrome	2353	Hypotelorism - cleft palate - hypospadias	672	Hypothalamic hamartoblastoma syndrome
436	Hypophosphatasia	443101	Hypothalamic adipic hypernatraemia syndrome	86906	Hypothalamic hamartomas with gelastic seizures
314621	Hypophyseal duplication	→3157	Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome	1226	Hypothyroidism - cleft palate
99725	Hypophyseal gigantism	3047	Hypothyroidism - dysmorphism - postaxial polydactyly - intellectual disability	226307	Hypothyroidism due to deficient transcription factors involved in pituitary development or function
79477	Hypopigmentation - immunodeficiency with or without neurologic impairment	90673	Hypothyroidism due to TSH receptor mutations	79507	Hypotonia - failure to thrive - microcephaly
79476	Hypopigmentation - neurologic impairment	91131	Hypotonia and ichthyosis due to dolichol phosphate deficiency	137507	Hypotonia with lactic acidemia and hyperammonemia
324561	Hypopigmentation and punctate keratosis of the palms and soles	→3157	Hypotuitarism - micropenis - cleft lip/palate	137577	I-cell disease
42665	Hypopigmentation-deafness syndrome	→3157	Hypotuitarism - microphthalmia	137577	Hypoxic and ischemic brain injury in the newborn
324561	Hypopigmentation-punctate palmoplantar keratoderma syndrome	→3157	Hypotuitarism - postaxial polydactyly	682	Hypoxic-ischemic encephalopathy
				63440	HYPP
				63440	Hypsicephaly
				576	Hypocephaly

→ 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
724	IAEP	724	Idiopathic acute eosinophilic pneumonia	84065	Idiopathic malabsorption due to bile acid synthesis defects
158048	IAHS	139423	Idiopathic acute transverse myelitis	73	Idiopathic massive osteolysis
293168	IAHSP	422	Idiopathic and/or familial pulmonary arterial hypertension	97560	Idiopathic membranous glomerulonephritis
254509	Iatrogenic botulism	280914	Idiopathic anterior uveitis	2774	Idiopathic multicentric osteolysis with or without nephropathy
95619	Iatrogenic or traumatic pituitary deficiency	88	Idiopathic aplastic anemia	824	Idiopathic myelofibrosis
363424	IBA57 deficiency	399307	Idiopathic avascular necrosis	45452	Idiopathic neonatal atrial flutter
→33364	IBIDS syndrome	399307	Idiopathic AVN	33577	Idiopathic nodular panniculitis
611	IBM	1980	Idiopathic basal ganglia calcification	51608	Idiopathic obliterative arteriopathy
602	IBM2	171684	Idiopathic bilateral vestibulopathy	441	Idiopathic orthostatic hypotension
79091	IBM3	84065	Idiopathic bile acid malabsorption	280921	Idiopathic panuveitis
52430	IBMPFD	88	Idiopathic bone marrow failure	747	Idiopathic PAP
1576	IBSN	60033	Idiopathic bronchiectasis	444316	Idiopathic phalangeal acro-osteolysis
31709	ICCA syndrome	188	Idiopathic capillary leak syndrome	444316	Idiopathic phalangeal acroosteolysis
64734	ICE syndrome	163703	Idiopathic catastrophic epileptic encephalopathy	280917	Idiopathic posterior uveitis
2268	ICF syndrome	228000	Idiopathic CD4 lymphocytopenia	747	Idiopathic pulmonary alveolar proteinosis
2269	Ichthyosis - alopecia - eclabion - ectropion - intellectual disability	169615	Idiopathic central precocious puberty	275766	Idiopathic pulmonary arterial hypertension
2274	Ichthyosis - hepatosplenomegaly - cerebellar degeneration	2902	Idiopathic chronic eosinophilic pneumonia	1676	Idiopathic pulmonary artery dilatation
59303	Ichthyosis - hypotrichosis - sclerosing cholangitis	95717	Idiopathic congenital hypothyroidism	2032	Idiopathic pulmonary fibrosis
2278	Ichthyosis - intellectual disability - dwarfism - renal impairment	209919	Idiopathic copper-associated cirrhosis	99931	Idiopathic pulmonary hemosiderosis
→1643	Ichthyosis - male hypogonadism	447881	Idiopathic dropped head syndrome	35061	Idiopathic recurrent and disabling cutaneous herpes
2272	Ichthyosis - oral and digital anomalies	256	Idiopathic dystonia	251307	Idiopathic recurrent pericarditis
455	Ichthyosis bullosa of Siemens	247724	Idiopathic eosinophilic myositis	276174	Idiopathic recurrent stupor
457	Ichthyosis congenita, harlequin type	2810	Idiopathic facial palsy	251307	Idiopathic relapsing pericarditis
289586	Ichthyosis exfoliativa	329874	Idiopathic giant cell myocarditis	40923	Idiopathic retinal perivasculitis
457	Ichthyosis fetalis, Harlequin type	64722	Idiopathic granulomatous mastitis	40923	Idiopathic retinal vasculitis
2273	Ichthyosis follicularis - alopecia - photophobia	86908	Idiopathic hemiconvulsion-hemiplegia syndrome	209943	Idiopathic retinal vasculitis-aneurysms-neuroretinitis syndrome
2273	Ichthyosis follicularis - atrichia - photophobia	2197	Idiopathic hypercalciuria	35065	Idiopathic severe pneumococcemia
79504	Ichthyosis hystrix gravior	3260	Idiopathic hypereosinophilic syndrome	69061	Idiopathic steroid-sensitive nephrotic syndrome
79503	Ichthyosis hystrix of Curth-Macklin	33208	Idiopathic hypersomnia	93209	Idiopathic steroid-sensitive nephrotic syndrome with diffuse mesangial proliferation
79503	Ichthyosis hystrix, Curth-Macklin type	228315	Idiopathic hypersomnia with long sleep time	93206	Idiopathic steroid-sensitive nephrotic syndrome with focal segmental glomerulosclerosis
88621	Ichthyosis prematurity syndrome	228318	Idiopathic hypersomnia without long sleep time	93206	Idiopathic steroid-sensitive nephrotic syndrome with focal segmental hyalinosis
281190	Ichthyosis variegata	449427	Idiopathic hypertrophic pachymeningitis	93207	Idiopathic steroid-sensitive nephrotic syndrome with minimal change
281190	Ichthyosis with confetti	1572	Idiopathic immunoglobulin deficiency	99858	Idiopathic syringomyelia
79504	Ichthyosis, Lambert type	51608	Idiopathic infantile arterial calcification	256	Idiopathic torsion dystonia
2267	Ichthyosis-cheek-eyebrow syndrome	238624	Idiopathic intracranial hypertension	98806	Idiopathic torsion dystonia of mixed type
91132	Ichthyosis-follicular atrophoderma-hypotrichosis syndrome	85193	Idiopathic juvenile osteoporosis	3347	Idiopathic tracheobronchomegaly
91132	Ichthyosis-follicular atrophoderma-hypotrichosis-hypohidrosis syndrome	247234	Idiopathic late-onset cerebellar ataxia	209956	Idiopathic uveal effusion syndrome
91132	Ichthyosis-hypotrichosis syndrome	314017	Idiopathic linear interstitial keratitis		
363992	Ichthyosis-short stature-brachydactyly-microspherophakia syndrome	33577	Idiopathic lobular panniculitis		
289347	IDH	90158	Idiopathic localized lipodystrophy		
3306	idic(15)	353344	Idiopathic macular telangiectasia type 1		
930	Idiopathic achalasia	353351	Idiopathic macular telangiectasia type 3		
930	Idiopathic achalasia of esophagus				

→ 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
130	Idiopathic ventricular fibrillation, Brugada type	247718	IMAM	200421	Immunodeficiency with factor H anomaly
228140	Idiopathic ventricular fibrillation, not Brugada type	42062	Iminoglycinuria	200418	Immunodeficiency with factor I anomaly
280384	IDMDC	284362	Immature interstitial mesenchymal tumor	935	Immunodeficiency-short limb dwarfism syndrome
580	Iduronate 2-sulfatase deficiency	398987	Immature teratoma of ovary	761	Immunoglobulin A vasculitis
217085	Iduronate 2-sulfatase deficiency type A	289465	Immigration delay disease	169110	Immunoglobulin heavy chain deficiency
217093	Iduronate 2-sulfatase deficiency type B	→244	Immotile cilia syndrome, Kartagener type		Immunoglobulin-mediated membranoproliferative glomerulonephritis
92050	IED	2901	Immune brachial plexus neuropathy	329903	Immunoglobulin-mediated MPGN
91132	IFAH syndrome	169090	Immune dysfunction due to T-cell inactivation due to calcium entry defect	442582	Immunoglobulinic amyloidosis
2273	IFAP syndrome	37042	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	85443	Immunoglobulinic amyloidosis
332	IFD	364013	Immune fetal edema	100025	Immunoproliferative small intestinal disease
329903	Ig-mediated membranoproliferative glomerulonephritis	364013	Immune fetal hydrops	97567	Immunotactoid glomerulopathy
329903	Ig-mediated MPGN	364013	Immune HF	857	Imperforate anus with hand, foot and ear anomalies
761	IgA vasculitis	364013	Immune hydrops fetalis	2759	Imperforate oropharynx - costo vertebral anomalies
329874	IGCM	1959	Immune pancytopenia	71276	Imploding antrum syndrome
79099	IGDA	3002	Immune thrombocytopenia	35069	INAD
73272	IGF-1 deficiency	3002	Immune thrombocytopenic purpura	35069	INAD1
449400	IgG4-related aortitis	206569	Immune-mediated necrotizing myopathy	254509	Inadvertent botulism
79078	IgG4-related dacryoadenitis and sialadenitis	206575	Immune-mediated rippling muscle disease	45453	Incessant infant ventricular tachycardia
449566	IgG4-related eosinophilic angiocentric fibrosis	86886	Immunoblastic lymphadenopathy	79263	INCL
90003	IgG4-related hepatopathy	2268	Immunodeficiency - centromeric instability - facial anomalies	231226	Inclusion body beta-thalassemia
449395	IgG4-related kidney disease	647	Immunodeficiency - microcephaly - chromosomal instability	199267	Inclusion body fibromatosis
63999	IgG4-related mediastinitis	34592	Immunodeficiency by defective expression of HLA class 1	602	Inclusion body myopathy type 2
238593	IgG4-related mesenteritis	572	Immunodeficiency by defective expression of HLA class 2	79091	Inclusion body myopathy type 3
449563	IgG4-related ophthalmic disease	169147	Immunodeficiency due to a C1, C4, or C2 component complement deficiency	52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
449427	IgG4-related pachymeningitis	169150	Immunodeficiency due to a C5 to C9 component complement deficiency	611	Inclusion body myositis
280302	IgG4-related pancreatitis	169150	Immunodeficiency due to a late component of complement deficiency	254693	Incomplete hydatidiform mole
449400	IgG4-related periaortitis	169147	Immunodeficiency due to an early component of complement deficiency	254693	Incomplete molar pregnancy
49041	IgG4-related retroperitoneal fibrosis	169100	Immunodeficiency due to CD25 deficiency	157769	Incomplete situs inversus
447764	IgG4-related sclerosing cholangitis	331190	Immunodeficiency due to ficolin3 deficiency	180079	Incomplete unilateral aplasia of the Müllerian ducts
449432	IgG4-related submandibular gland disease	70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	180079	Incomplete unilateral Müllerian aplasia
64744	IgG4-related thyroid disease	331187	Immunodeficiency due to MASP-2 deficiency	464	Incontinentia pigmenti
183675	IgG subclass deficiency with IgA subclass deficiency	70593	Immunodeficiency due to selective anti-polysaccharide antibody deficiency	435	Incontinentia pigmenti type 1
329235	IGSF1 deficiency syndrome			158019	Indeterminate cell histiocytosis
364013	IHF			1388	Index finger anomaly - Pierre Robin syndrome
86908	IHHS			98848	Indolent systemic mastocytosis
91132	IHS			1909	Indomethacin embryofetopathy
59303	IHSC			70587	Infant acute respiratory distress syndrome
238624	IIH			70587	Infant ARDS
85193	IJO				
100078	Ileal endocrine tumor				
238621	Ileal pouch anal anastomosis related faecal incontinence				
1150	Illum syndrome				
79466	ILVEN				
85173	IMAge 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
178478	Infant botulism	1928	Infantile lobar hyperinflation	171714	Infantile-onset symptomatic epilepsy syndrome - developmental stagnation - blindness
1943	Infant epilepsy with migrant focal crisis	667	Infantile malignant osteopetrosis	781	Infection due to <i>Coxiella burnetii</i>
178478	Infant intestinal botulism	247165	Infantile mercury intoxication	279922	Infectious anterior uveitis
178478	Infant intestinal toxemia botulism	247165	Infantile mercury poisoning	137593	Infectious epithelial keratitis
178478	Infant intestinal toxin-mediated botulism	2591	Infantile myofibromatosis	279925	Infectious panuveitis
70587	Infant respiratory distress syndrome	79263	Infantile NCL	279919	Infectious posterior uveitis
178487	Infant-like botulism	93591	Infantile nephronophthisis	289347	Infective dermatitis associated with HTLV-1
247165	Infantile acrodynia	35069	Infantile neuroaxonal dystrophy	289347	Infective dermatitis associated with human T-lymphotropic virus type 1
99749	Infantile agranulocytosis	79263	Infantile neuronal ceroid lipofuscinosis	289347	Infective dermatitis associated with human T-lymphotropic virus type I
99725	Infantile and juvenile forms of acromegaly	289860	Infantile NKH	99123	Inferior caval vein interruption
70590	Infantile apnea	289860	Infantile non-ketotic hyperglycinemia	155889	Inferior palpebral coloboma
51608	Infantile arteriosclerosis	251304	Infantile onset panniculitis with uveitis and systemic granulomatosis	99123	Inferior vena cava interruption
2679	Infantile axonal neuropathy	1186	Infantile onset spinocerebellar ataxia	280794	Infiltrative small vesicular DCM
89938	Infantile Bartter syndrome with sensorineural deafness	67047	Infantile optic atrophy with chorea and spastic paraparesis	280794	Infiltrative small vesicular diffuse cutaneous mastocytosis
1576	Infantile bilateral striatal necrosis	85179	Infantile osteopetrosis with neuroaxonal dysplasia	85445	Inflammatory amyloidosis
178478	Infantile botulism	247651	Infantile phoshethanolaminuria	79466	Inflammatory linear verrucous epidermal nevus
314911	Infantile Canavan disease	247651	Infantile Rathburn disease	178342	Inflammatory myofibroblastic tumor
137675	Infantile cardiomyopathy with histiocytoid change	772	Infantile Refsum disease	160148	Inflammatory myoglandular polyps
217557	Infantile cellular interstitial pneumonitis	254864	Infantile reversible cytochrome C oxidase deficiency myopathy	247718	Inflammatory myopathy with abundant macrophages
313850	Infantile cerebellar-retinal degeneration	263410	Infantile spasms - psychomotor retardation - progressive brain atrophy - basal ganglia disease	263553	Inflammatory peeling skin syndrome
402364	Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	3451	Infantile spasms	48918	Inflammatory pseudotumor of skeletal muscle
77260	Infantile cerebral Gaucher disease	3173	Infantile spasms - broad thumbs	90003	Inflammatory pseudotumor of the liver
1313	Infantile choroidocerebral calcification syndrome	83330	Infantile spinal muscular atrophy	238305	Infundibulo-neurohypophysitis
31709	Infantile convulsions and choreoathetosis	1576	Infantile striatonigral degeneration	95513	Infundibulo-panhypophysitis
1310	Infantile cortical hyperostosis	1576	Infantile striatonigral necrosis	1849	Infundibulopelvic stenosis - multicystic kidney
199267	Infantile digital fibromatosis	1575	Infantile striothalamic degeneration	247257	Inhalation anthrax disease
87876	Infantile dysmorphic sialidosis	255241	Infantile subacute necrotizing encephalopathy with leukodystrophy	254504	Inhalation botulism
238455	Infantile dystonia-parkinsonism	255249	Infantile subacute necrotizing encephalopathy with nephrotic syndrome	247257	Inhalational anthrax
364063	Infantile epileptic-dyskinetic encephalopathy	3311	Infantile symmetrical thalamic degeneration	254504	Inhalational botulism
300373	Infantile gigantism due to pituitary hyperplasia	2176	Infantile systemic hyalinosis	319465	Inherited acute myeloid leukemia
289860	Infantile glycine encephalopathy	1577	Infantile thalamic degeneration	319465	Inherited AML
79255	Infantile GM1 gangliosidosis	2768	Infantile tibia vara	319462	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations
309155	Infantile GM2 gangliosidosis 0 variant	137675	Infantile xanthomatous cardiomyopathy	282166	Inherited CJD
293603	Infantile hereditary endothelial dystrophy	293168	Infantile-onset ascending hereditary spastic paralysis	210141	Inherited congenital spastic quadriplegia
352563	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	284332	Infantile-onset autosomal recessive nonprogressive cerebellar ataxia	210141	Inherited congenital spastic tetraparesis
247651	Infantile hypophosphatasia	391316	Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression	282166	Inherited Creutzfeldt-Jakob disease
79076	Infantile juvenile polyposis syndrome	1451	Infantile-onset multisystem inflammatory disease	859	Inherited deficiency of transcobalamin
206436	Infantile Krabbe disease				

→ 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
100054	Inherited estrogen-associated angioedema	356996	Intellectual disability - hypotonia - spasticity - sleep disorder	370010	Intellectual disability-facial dysmorphism-hand anomalies syndrome
100054	Inherited estrogen-associated angioneurotic edema	3451	Intellectual disability - hypsarrhythmia	363611	Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome
100054	Inherited estrogen-dependent angioedema	3067	Intellectual disability - microcephaly - phalangeal - facial abnormalities	369847	Intellectual disability-hyperkinetic movement-truncal ataxia syndrome
100054	Inherited estrogen-dependent angioneurotic edema	3068	Intellectual disability - myopathy - short stature - endocrine defect	314575	Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome
71278	Inherited glutamine synthetase deficiency	352530	Intellectual disability - obesity - brain malformations - facial dysmorphism	436151	Intellectual disability-loss of expressive language-facial dysmorphism syndrome
71278	Inherited GS deficiency	3082	Intellectual disability - polydactyly - uncombable hair	397973	Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome
289548	Inherited isolated adrenal insufficiency due to CYP11A1 deficiency	3409	Intellectual disability - short stature - hand contractures - genital anomalies	369837	Intellectual disability-seizures-hypotonia-opthalmologic-skeletal anomalies syndrome
225968	Inherited predisposition to essential thrombocythemia	3074	Intellectual disability - short stature - hypertelorism	369950	Intellectual disability-seizures-macrocephaly-obesity syndrome
37	Inherited zinc deficiency	1240	Intellectual disability - short stature - wedge shaped epiphyses of knees	391372	Intellectual disability-severe speech delay-mild dysmorphism syndrome
63259	Iniencephaly	3051	Intellectual disability - sparse hair - brachydactyly	363528	Intellectual disability-strabismus syndrome
178475	Inoculation botulism	1891	Intellectual disability - spasticity - ectrodactyly	397941	Intellectual disability-truncal obesity syndrome
642	Insensitivity to pain - anhidrosis	75858	Intellectual disability - truncal obesity - retinal dystrophy - micropenis	1478	Interatrial communication
411593	Insulin autoimmune syndrome	100973	Intellectual disability associated with fragile site FRAXE	1478	Interauricular communication
2297	Insulin-resistance syndrome type A	166108	Intellectual disability, Birk-Barel type	86900	Interdigitating cell sarcoma
2298	Insulin-resistance syndrome type B	3079	Intellectual disability, Buenos-Aires type	86900	Interdigitating dendritic cell sarcoma
97279	Insulinoma	→32473 7	Intellectual disability, Kahrizi type	210115	Interleukin-1 receptor antagonist deficiency
127	Intellectual deficiency - epilepsy - endocrine disorders	2557	Intellectual disability, Mietens-Weber type	169100	Interleukin-2 receptor alpha chain deficiency
289483	Intellectual disability - alacrima - achalasia	3080	Intellectual disability, Wolff type	171208	Intermediate anorectal malformation
1236	Intellectual disability - athetosis - microphthalmia	2466	Intellectual disability-aphasia-shuffling gait-adducted thumbs syndrome	268162	Intermediate BCKD deficiency
3041	Intellectual disability - balding - patella luxation - acromicria	364577	Intellectual disability-brachydactyly-Pierre Robin syndrome	268162	Intermediate branched-chain 2-ketoacid dehydrogenase deficiency
→32473 7	Intellectual disability - cataract - coloboma - kyphosis	397709	Intellectual disability-coarse face-macrocephaly-cerebellar hypoplasia syndrome	411634	Intermediate cystinosis
3042	Intellectual disability - cataracts - calcified pinnae - myopathy	397709	Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	99989	Intermediate DEND syndrome
171860	Intellectual disability - cataracts - kyphosis	3454	Intellectual disability-developmental delay-contractures syndrome	86797	Intermediate lichen myxedematosus
329224	Intellectual disability - craniofacial dysmorphism - cryptorchidism	435638	Intellectual disability-epilepsy-stereotypic hand movement syndrome	268162	Intermediate maple syrup urine disease
3044	Intellectual disability - dysmorphism - hypogonadism - diabetes mellitus	436151	Intellectual disability-expressive aphasia-facial dysmorphism syndrome	268162	Intermediate MSUD
→280	Intellectual disability - dysmorphism - intrauterine growth retardation	404440	Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	171433	Intermediate nemaline myopathy
171851	Intellectual disability - enteropathy - deafness - peripheral neuropathy - ichthyosis - keratodermia			210110	Intermediate osteopetrosis
2139	Intellectual disability - epilepsy - bulbous nose			309331	Intermediate severe Salla disease
1495	Intellectual disability - hypoplastic corpus callosum - preauricular tag			83418	Intermediate spinal muscular atrophy
166108	Intellectual disability - hypotonia - facial dysmorphism			268173	Intermittent BCKD deficiency
3050	Intellectual disability - hypotonia - skin hyperpigmentation			268173	Intermittent branched-chain 2-ketoacid dehydrogenase deficiency
				329967	Intermittent hydrarthrosis

→ 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
268173	Intermittent maple syrup urine disease	436144	Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	85200	Ischio-vertebral syndrome
268173	Intermittent MSUD	85173	Intrauterine growth retardation - metaphyseal dysplasia - adrenal hypoplasia congenita - genital anomalies	1509	Ischiopatellar dysplasia
→2686	Intermittent neutropenia	137686	Intrauterine synechiae	43115	ISCU myopathy
981	Internal carotid agenesis	98839	Intravascular large B-cell lymphoma	79144	Iso-Kikuchi syndrome
37202	Interstitial cystitis	98839	Intravascular lymphomatosis	79159	Isobutyric aciduria
79099	Interstitial granulomatous dermatitis with arthritis	332	Intrinsic factor deficiency	79159	Isobutyryl-CoA dehydrogenase deficiency
440427	Interstitial lung and liver disease	3306	Inv dup(15)	3309	Isochromosome 5p
440402	Interstitial lung disease due to ABCA3 deficiency	90078	Invasive infections due to vancomycin-resistant enterococci	3310	Isochromosome 9p
440402	Interstitial lung disease due to ATP-binding cassette subfamily A member 3 deficiency	90078	Invasive infections due to VRE	884	Isochromosome 12p mosaicism
440392	Interstitial lung disease due to SP-C deficiency	99925	Invasive mole	884	Isochromosome 12p syndrome
440392	Interstitial lung disease due to surfactant protein C deficiency	324648	Invasive non-typhoidal salmonellosis	3307	Isochromosome 18p
99092	Interventricular septum aneurysm	96092	Invdupdel(8p)	96055	Isochromosome 21
1201	Intestinal atresia type IIIB	79405	Inverse JEB	98797	Isochromosome Yp
178481	Intestinal botulism	329324	Inverse Klippel-Trénaunay syndrome	98798	Isochromosome Yq
178481	Intestinal colonization botulism	98951	Inverse Marcus-Gunn phenomenon	99731	ISOD
92050	Intestinal epithelial dysplasia	79409	Inverse RDEB	3306	Isodicentric 15 chromosome
30924	Intestinal hypomagnesemia with secondary hypocalcemia	79409	Inverse recessive dystrophic epidermolysis bullosa	263524	Isolated acute necrotizing encephalopathy
3452	Intestinal lipodystrophy	96092	Inverted 8p duplication/deletion syndrome	229717	Isolated agammaglobulinemia
3452	Intestinal lipophagic granulomatosis	2704	Inverted smile - neurogenic bladder	440987	Isolated agenesis of gallbladder
314376	Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency	1451	IOMID syndrome	268868	Isolated amyelia
86880	Intestinal T-cell lymphoma	1186	IOSCA	263524	Isolated ANE
178481	Intestinal toxemia botulism	275766	IPAH	1048	Isolated anencephaly/exencephaly
178481	Intestinal toxin-mediated botulism	747	iPAP	140989	Isolated angiitis of the central nervous system
228371	Intoxication botulism	238455	IPD	250923	Isolated aniridia
46724	Intracranial arteriovenous malformation	37042	IPEX	91397	Isolated ankyloblepharon filiforme adnatum
252006	Intracranial endodermal sinus tumor	88621	IPS	79143	Isolated anonychia
91352	Intracranial germinoma	100025	IPSID	3387	Isolated anterior cervical hypertrichosis
252006	Intracranial yolk sac tumor	70592	IRAK4 deficiency	162516	Isolated apertura pyriformis stenosis
137622	Intractable diarrhea - choanal atresia - eye anomalies	772	IRD	268936	Isolated arhinencephaly
424058	Intraductal papillary mucinous carcinoma of pancreas	209981	IRIDA syndrome	1166	Isolated asymmetric crying facies
424982	Intrahepatic bile duct cystadenocarcinoma	64734	Iridocorneal endothelial syndrome	206599	Isolated asymptomatic elevation of creatine phosphokinase
69665	Intrahepatic cholestasis of pregnancy	240885	Irinotecan toxicity	254913	Isolated ATP synthase deficiency
280802	Intralobar congenital bronchopulmonary sequestration	2995	Iris coloboma-ptosis-intellectual disability syndrome	34528	Isolated autosomal dominant hypomagnesemia
280802	Intralobar congenital pulmonary sequestration	→782	Iris dysplasia - hypertelorism - deafness	199326	Isolated autosomal dominant hypomagnesemia, Glaudemans type
99088	Intramural coronary arterial course	39044	Iris melanoma	269221	Isolated bilateral hemispheric cerebellar hypoplasia
100003	Intraneuronal perineurioma	209981	Iron-refractory iron deficiency anemia	158778	Isolated bone marrow mastocytosis
268139	Intraocular medulloepithelioma	43115	Iron-sulfur cluster deficiency myopathy	35099	Isolated brachycephaly
140436	Intraosseous hemangioma	86915	Irons-Bianchi syndrome	1398	Isolated cerebellar hypoplasia/agenesis
137686	Intrauterine adhesions	209943	IRVAN syndrome	269203	Isolated cerebellar vermis agenesis
		84142	Isaac syndrome	199630	Isolated cerebellar vermis hypoplasia
		84142	Isaac-Mertens syndrome	2343	Isolated cloverleaf skull syndrome
		85200	Ischio-spinal dysostosis	1460	Isolated coenzyme Q-cytochrome C reductase deficiency
		85200	Ischio-vertebral dysplasia	217059	Isolated congenital acropachy

→ 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
289465	Isolated congenital adermatoglyphia	268987	Isolated focal cortical dysplasia type Ic	137902	Isolated optic nerve hypoplasia
91416	Isolated congenital alacrima	268994	Isolated focal cortical dysplasia type II	166119	Isolated osteopoikilosis
180188	Isolated congenital amastia	269001	Isolated focal cortical dysplasia type IIa	63440	Isolated oxycephaly
79143	Isolated congenital anonychia	269008	Isolated focal cortical dysplasia type IIb	269209	Isolated partial cerebellar vermis agenesis
88620	Isolated congenital anosmia	52901	Isolated follicle stimulating hormone deficiency	96269	Isolated partial vaginal agenesis
162526	Isolated congenital auditory ossicle malformation	52901	Isolated FSH deficiency	718	Isolated Pierre Robin sequence
180188	Isolated congenital breast hypoplasia/aplasia	408	Isolated glycerol kinase deficiency	718	Isolated Pierre Robin syndrome
238722	Isolated congenital contralateral synkinesia	231662	Isolated growth hormone deficiency type IA	35098	Isolated plagioccephaly
217059	Isolated congenital digital clubbing	231671	Isolated growth hormone deficiency type IB	2924	Isolated polycystic liver disease
99171	Isolated congenital ectropion	231679	Isolated growth hormone deficiency type II	2456	Isolated polythelia
432	Isolated congenital gonadotropin deficiency	231692	Isolated growth hormone deficiency type III	216452	Isolated postlingual genetic deafness
141152	Isolated congenital hypoglossia/aglossia	2128	Isolated hemihyperplasia	216445	Isolated prelingual genetic deafness
91489	Isolated congenital megalocornea	2128	Isolated hemihypertrophy	238670	Isolated prothyroliberin deficiency
238722	Isolated congenital mirror movements	306527	Isolated hereditary congenital facial paralysis	238670	Isolated protirelin deficiency
217059	Isolated congenital nail clubbing	229717	Isolated hypogammaglobulinemia	264691	Isolated pulmonary capillaritis
162516	Isolated congenital nasal pyriform aperture stenosis	183675	Isolated IgG subclass deficiency	34528	Isolated renal magnesium wasting
91490	Isolated congenital sclerocornea	2345	Isolated Klippel-Feil syndrome	35093	Isolated scaphocephaly
216718	Isolated congenitally uncorrected transposition of the great arteries	1084	Isolated lissencephaly type 1 without known genetic defects	440713	Isolated sedoheptulokinase deficiency
216718	Isolated congenitally uncorrected transposition of the great vessels	268920	Isolated macrencephaly	440713	Isolated SHPK deficiency
1460	Isolated CoQ-cytochrome C reductase deficiency	391474	Isolated median cleft syndrome	178311	Isolated sternocostoclavicular hyperostosis
254905	Isolated COX deficiency	268920	Isolated megalecephaly	3208	Isolated succinate-coenzyme Q reductase deficiency
91396	Isolated cryptophthalmia	238593	Isolated mesenteric lipodystrophy	3208	Isolated succinate-CoQ reductase deficiency
254905	Isolated cytochrome C oxidase deficiency	95707	Isolated micropenis	99731	Isolated sulfite oxidase deficiency
217	Isolated Dandy-Walker malformation	90641	Isolated mitochondrial neurosensory deafness	90674	Isolated thyroid-stimulating hormone deficiency
269212	Isolated Dandy-Walker malformation with hydrocephalus	2609	Isolated mitochondrial respiratory chain complex I deficiency	238670	Isolated thyroliberin deficiency
269215	Isolated Dandy-Walker malformation without hydrocephalus	3208	Isolated mitochondrial respiratory chain complex II deficiency	238670	Isolated thyrotropin-releasing factor deficiency
248340	Isolated delta-SPD	1460	Isolated mitochondrial respiratory chain complex III deficiency	238670	Isolated thyrotropin-releasing hormone deficiency
248340	Isolated delta-storage pool disease	254905	Isolated mitochondrial respiratory chain complex IV deficiency	269206	Isolated total cerebellar vermis agenesis
248340	Isolated dense-SPD	254913	Isolated mitochondrial respiratory chain complex V deficiency	103909	Isolated trehalose intolerance
248340	Isolated dense-storage pool disease	90641	Isolated mitochondrial sensorineural deafness	238670	Isolated TRF deficiency
99177	Isolated distichiasis	2609	Isolated NADH-coenzyme Q reductase deficiency	238670	Isolated TRH deficiency
35093	Isolated dolichocephaly	2609	Isolated NADH-CoQ reductase deficiency	3366	Isolated trigonocephaly
1885	Isolated ectopia lentis	2609	Isolated NADH-ubiquinone reductase deficiency	90674	Isolated TSH deficiency
199647	Isolated encephalocele	162516	Isolated nasal pyriform aperture hypoplasia	238670	Isolated TSH-releasing factor deficiency
221106	Isolated facial myokymia	447881	Isolated neck extensor myopathy	1460	Isolated ubiquinone-cytochrome C reductase deficiency
65683	Isolated focal cortical dysplasia			269218	Isolated unilateral hemispheric cerebellar hypoplasia
268961	Isolated focal cortical dysplasia type I			860	Isolated ventriculoarterial discordance
268973	Isolated focal cortical dysplasia type Ia			96	Isolated vitamin E deficiency
268980	Isolated focal cortical dysplasia type Ib			240887	Isoniazid toxicity
				472	Isosporiasis

→ 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
2305	Isotretinoin embryopathy	90647	Jervell and Lange-Nielsen syndrome	1454	JS-H
2305	Isotretinoin syndrome	33314	Jessner-Kanof lymphocytic infiltration of the skin	220493	JS-O
2306	Isotretinoin-like syndrome	33314	Jessner's benign lymphocytic infiltration of the skin	2318	JS-OR
33	Isovaleric acid CoA dehydrogenase deficiency	33314	Jessner's lymphocytic infiltration of the skin	220497	JS-R
33	Isovaleric acidemia	3283	JET	2319	Juberg-Hayward syndrome
309324	ISSD	474	Jeune asphyxiating thoracic dystrophy	93972	Juberg-Marsidi syndrome
→33364	Itin syndrome	474	Jeune syndrome	3283	Junctional ectopic tachycardia
439254	ITM2B amyloidosis	248111	JHD	79403	Junctional epidermolysis bullosa - pyloric atresia
439254	ITM2B-related amyloidosis	2929	JIP	79404	Junctional epidermolysis bullosa generalisata gravis
439254	ITM2B-related cerebral amyloid angiopathy	65684	JMADUE	79402	Junctional epidermolysis bullosa generalisata mitis
435	Ito hypomelanosis	307	JME	79405	Junctional epidermolysis bullosa inversa
3002	ITP	324999	JMP syndrome	79402	Junctional epidermolysis bullosa, Disentis type
99123	IVC interruption	289596	JNA	79404	Junctional epidermolysis bullosa, Herlitz type
294415	Ivemark II syndrome	79264	JNCL	79404	Junctional epidermolysis bullosa, Herlitz-Pearson type
97548	Ivemark syndrome	2314	Job syndrome	89840	Junctional epidermolysis bullosa, non-Herlitz type
2307	IVIC syndrome	2315	Johanson-Blizzard syndrome	2321	Jung-Wolff-Back-Stahl syndrome
281190	IWC	2316	Johnson neuroectodermal syndrome	319223	Junin hemorrhagic fever
3236	Jackson-Barr syndrome	85320	Johnson syndrome	989	Jussieu syndrome
1540	Jackson-Weiss syndrome	2316	Johnson-McMillin syndrome	1941	Juvenile absence epilepsy
2848	Jacobs syndrome	1112	Johnson-Munson syndrome	391497	Juvenile acquired myasthenia
2308	Jacobsen syndrome	1485	Johnston-Aarons-Schelley syndrome	300605	Juvenile amyotrophic lateral sclerosis
1941	JAE	324999	Joint contractures-muscular atrophy-microcytic anemia-panniculitis-associated lipodystrophy syndrome	199260	Juvenile aponeurotic fibromatosis
2029	Jaffe-Campanacci syndrome	2295	Joint instability syndrome	391497	Juvenile autoimmune myasthenia gravis
93277	Jaffe-Lichtenstein disease	2027	Jones syndrome	314918	Juvenile Canavan disease
2269	Jagell-Holmgren-Hofer syndrome	1256	Jorgenson-Lenz syndrome	247794	Juvenile cataract - microcornea - renal glucosuria
1873	Jalili syndrome	475	Joubert syndrome	300605	Juvenile Charcot disease
300605	JALS	475	Joubert syndrome type A	86834	Juvenile chronic myelomonocytic leukemia
73423	Jamaican vomiting sickness	1454	Joubert syndrome with congenital hepatic fibrosis	411634	Juvenile cystinosis
73423	Jamaican vomiting syndrome	1454	Joubert syndrome with hepatic defect	93672	Juvenile dermatomyositis
1891	Jancar syndrome	397715	Joubert syndrome with JATD	93672	Juvenile DM
2590	Jankovic-Rivera syndrome	397715	Joubert syndrome with Jeune asphyxiating thoracic dystrophy	228254	Juvenile elastoma without osteopoikilosis
168491	Jansky-Bielschowsky disease	220493	Joubert syndrome with ocular defect	2929	Juvenile gastrointestinal polyposis
79139	Japanese encephalitis	2318	Joubert syndrome with oculorenal defect	98977	Juvenile glaucoma
2311	Jarcho-Levin syndrome	2754	Joubert syndrome with oral-facial-digital syndrome	79256	Juvenile GM1 gangliosidosis
474	JATD	2754	Joubert syndrome with orofaciодigital defect	309162	Juvenile GM2 gangliosidosis 0 variant
91412	Jaw-winking syndrome	220497	Joubert syndrome with renal defect	79230	Juvenile hemochromatosis
313795	Jawad syndrome	220493	Joubert syndrome with retinopathy	98954	Juvenile hereditary epithelial dystrophy of Meesmann
397715	JBTS with JATD	2318	Joubert syndrome with Senior-Loken syndrome	248111	Juvenile Huntington chorea
139431	Jeavons syndrome	475	Joubert-Boltshauser syndrome	248111	Juvenile Huntington disease
79404	JEB-H	2801	JPG	2028	Juvenile hyaline fibromatosis
79405	JEB-I	247604	JPLS	2929	Juvenile intestinal polyposis
79406	JEB-lo	2929	JPS		
79402	JEB-nH gen	2318	JS type B		
251393	JEB-nH loc				
79403	JEB-PA				
1201	Jejunal atresia				
100077	Jejunal endocrine tumor				
1201	Jeunoileal atresia				
89840	JEN-nH				
→52368	Jensen 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
300605	Juvenile Lou Gehrig disease	2322	Kabuki make-up syndrome	79395	Keratoderma - ichthyosiform dermatosis - elevated beta-glucuronidase
65684	Juvenile muscular atrophy of distal upper extremity	2322	Kabuki syndrome	494	Keratoderma hereditarium mutilans
65684	Juvenile muscular atrophy of the distal upper limb	85146	Kaeser syndrome	79395	Keratoderma hereditarium mutilans with ichthyosis
391497	Juvenile myasthenia gravis	29073	Kahler's disease	34217	Keratoderma with woolly hair type I
86834	Juvenile myelomonocytic leukemia	→32473 7	Kahrizi syndrome	65282	Keratoderma with woolly hair type II
307	Juvenile myoclonic epilepsy	2324	Kaler-Garrity-Stern syndrome	420686	Keratoderma with woolly hair type IV
307	Juvenile myoclonus epilepsy	2325	Kallin syndrome	79501	Keratoderma palmoplantaris papulosa, Buschke-Fischer-Brauer type
289596	Juvenile nasopharyngeal angiofibroma	478	Kallmann syndrome	50943	Keratolytic winter erythema
79264	Juvenile NCL	2326	Kallmann syndrome - heart disease	495	Keratosis extremitatum hereditaria progrediens
93592	Juvenile nephronophthisis	99179	Kandori fleck retina	218	Keratosis follicularis
411634	Juvenile nephropathic cystinosis	1836	Kantaputra mesomelic dysplasia	2339	Keratosis follicularis - dwarfism - cerebral atrophy
79264	Juvenile neuronal ceroid lipofuscinoses	79280	Kanzaki disease	2340	Keratosis follicularis spinulosa decalvans
157719	Juvenile or adult CACH syndrome	949	Kaplan-Plauchu-Fitch syndrome	281201	Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome
85193	Juvenile osteoporosis	→3157	Kaplowitz-Bodurtha syndrome	86919	Keratosis palmaris et plantaris - clinodactyly
329894	Juvenile overlap myositis	33276	Kaposi sarcoma	678	Keratosis palmoplantar - periodontopathy
2801	Juvenile Paget disease	2122	Kaposiform hemangioendothelioma	28378	Keratosis palmoplantaris - corneal dystrophy
2801	Juvenile Paget's disease	91136	Kappa light chain-associated Fanconi syndrome	50944	Keratosis palmoplantaris - cystic eyelids - hypodontia - hypotrichosis
247604	Juvenile PLS	183675	Kappa-chain deficiency	2342	Keratosis palmoplantaris - periodontopathy - onychogryposis
93568	Juvenile PM	2328	Kapur-Toriello syndrome	79141	Keratosis palmoplantaris nummularis
93568	Juvenile polymyositis	1381	Karandikar-Maria-Kamble syndrome	50942	Keratosis palmoplantaris striata
79076	Juvenile polyposis of infancy	2329	Karsch-Neugebauer syndrome	50942	Keratosis palmoplantaris striata et areata
2929	Juvenile polyposis syndrome	→244	Kartagener syndrome	495	Keratosis palmoplantaris transgrediens et progrediens
247604	Juvenile primary lateral sclerosis	401996	Karyomegalic interstitial nephritis	87503	Keratosis palmoplantaris transgrediens of Siemens
85436	Juvenile psoriatic arthritis	2330	Kasabach-Merritt syndrome	50942	Keratosis palmoplantaris varians of Wachters
85408	Juvenile rheumatoid factor-negative polyarthritis	1894	Kasznica-Carlson-Coppedge syndrome	34217	Keratosis palmoplantaris with arrhythmogenic cardiomyopathy
247854	Juvenile rheumatoid factor-negative polyarthritis with anti-nuclear antibodies	3360	Katsantoni-Papadakou Lagoyanni syndrome	2198	Keratosis palmoplantaris-esophageal carcinoma syndrome
247861	Juvenile rheumatoid factor-negative polyarthritis without anti-nuclear antibodies	2473	Kaufman-Mckusick syndrome	499	Kerion celsi
85435	Juvenile rheumatoid factor-positive polyarthritis	2331	Kawasaki disease	415286	Kernicterus
93399	Juvenile sialidosis type 2	2306	Kawashima syndrome	3351	Kersey syndrome
83419	Juvenile spinal muscular atrophy	2533	Kawashima-Tsuji syndrome	293807	Ketamine-induced biliary dilatation
85438	Juvenile spondylarthropathy	2332	KBG syndrome	438075	Ketoacidosis due to monocarboxylate transporter-1 deficiency
585	Juvenile sulfatidosis, Austin type	439218	KCNQ2-NEE	1399	Ketoaciduria - intellectual disability - ataxia - deafness
26137	Juvenile temporal arteritis	439218	KCNQ2-related epileptic encephalopathy	2056	Ketohexokinase deficiency
158000	Juvenile xanthogranuloma	96169	KdVS		
445062	Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	480	Kearns-Sayre syndrome		
79241	Juvenile-onset multiple carboxylase deficiency	199260	Keasby tumor		
1243	Juvenile-onset vitelliform macular dystrophy	2662	Keipert syndrome		
99100	Juxtaposition of the atrial appendages	79233	Kelley-Seegmiller syndrome		
99100	Juxtaposition of the atrial auricles	137653	Kelly-Kirson-Wyatt syndrome		
1540	JWS	54028	Kelly-Paterson syndrome		
		481	Kennedy disease		
		64542	Kennedy-Teebi syndrome		
		2333	Kenny syndrome		
		2333	Kenny-Caffey syndrome		
		435628	Keppen-Lubinsky syndrome		
		477	Keratitis - ichthyosis - deafness/Hystrix-like ichthyosis - deafness		
		447777	Keratocystic odontogenic tumor		

→ 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
35	Ketotic hyperglycinemia	679	Köhlmeier-Degos-Delort-Tricort syndrome	79314	L-2-hydroxyglutaric aciduria
85202	Keutel syndrome	1946	Kohlschutter-Tonz syndrome	35704	L-Arginine:glycine amidinotransferase deficiency
2988	Khalifa-Graham syndrome	3197	Kok disease	157973	L-CMD
98841	Ki-1 positive anaplastic large cell lymphoma	99077	Kommerell diverticulum	156	L-CPT1 deficiency
477	KID syndrome	2764	König disease	156	L-CPTI deficiency
477	KID/HID syndrome	→1215	Konigsmark-Knox-Hussels syndrome	440731	L-ferritin deficiency
97332	Kienbock disease	96169	Koolen-De Vries syndrome	93599	L-glyceric aciduria
50918	Kikuchi disease	363965	Koolen-De Vries syndrome due to a point mutation	216694	L-transposition of the great arteries
50918	Kikuchi-Fujimoto disease	2892	Kopysc-Barczyk-Krol syndrome	83483	La Crosse encephalitis
482	Kimura disease	2839	Kosenow syndrome	53696	LAAHD
401996	KIN	99749	Kostmann syndrome	3473	Laband syndrome
2908	Kindler syndrome	1129	Kosztolanyi syndrome	2363	Lacrimoauriculodentodigital syndrome
99741	King-Denborough syndrome	99741	Koussef-Nichols syndrome	2363	Lacrimoauriculoradiodental syndrome
565	Kinky hair disease	2351	Kousseff syndrome	284426	Lactate dehydrogenase A deficiency
565	Kinky hair syndrome	629	Kowarski syndrome	284435	Lactate dehydrogenase B deficiency
1183	Kinsbourne syndrome	2352	Kozlowski-Brown-Hardwick syndrome	2965	Lactotroph adenoma
100996	Kjellin syndrome	3082	Kozlowski-Krajewska syndrome	2968	LAD
98673	Kjer optic atrophy	2204	Kozlowski-Tsuruta syndrome	99844	LAD-1 variant
99978	Klatskin tumor	487	Krabbe disease	99842	LAD-I
261494	Kleefstra syndrome	206436	Krabbe disease, classic form	99843	LAD-II
96147	Kleefstra syndrome due to 9q subtelomeric deletion	206436	Krabbe disease, early-onset	99844	LAD-III
96147	Kleefstra syndrome due to 9q34 microdeletion	206443	Krabbe disease, late-onset	2363	LADD syndrome
261652	Kleefstra syndrome due to a point mutation	1345	Krasnow-Qazi syndrome	1484	Ladda-Zonana-Ramer syndrome
96147	Kleefstra syndrome due to del(9)(q34)	709	Krause-Kivlin syndrome	158687	LAEB
96147	Kleefstra syndrome due to monosomy 9q34	709	Krause-van Schooneveld-Kivlin syndrome	501	Lafora disease
896	Klein-Waardenburg syndrome	284149	Kreiborg-Pakistani syndrome	1997	Lagophthalmia - cleft lip and palate
33543	Kleine-Levin syndrome	89838	KRT14-related epidermolysis bullosa simplex	59135	Laing early-onset distal myopathy
2110	Kleiner-Holmes syndrome	2908	KS	275761	LAL deficiency
399081	KLHL9-related childhood-onset distal myopathy	293936	KTCNCT	538	LAM
281201	KLICK syndrome	306674	Kufor-Rakeb syndrome	306507	LAMB2-related infantile-onset nephrotic syndrome
447974	Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome	79262	Kufs disease	1296	Lambert syndrome
2345	Klippel-Feil malformation	83419	Kugelberg-Welander disease	43393	Lambert-Eaton myasthenic syndrome
2345	Klippel-Feil sequence	→1487	Kumar-Levick syndrome	313	Lamellar ichthyosis
90308	Klippel-Trénaunay syndrome	2505	Kunze-Riehm syndrome	137871	Laminopathy type Decaudain-Vigouroux
2346	Klippel-Trénaunay-Weber syndrome	→794	Kurczynski-Casperson syndrome	137871	Laminopathy with severe metabolic syndrome and myopathy
157823	Klüver-Bucy syndrome	1149	Kuskokwim disease	90024	LAMM syndrome
485	Kniest dysplasia	767	Küssmaul-Maier disease	98818	Landau-Kleffner syndrome
1571	Knobloch syndrome	2798	Kuzniecyk syndrome	354	Landing disease
1571	Knobloch-Layer syndrome	34217	KWWH type I	269	Landouzy-Dejerine myopathy
2698	Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome	65282	KWWH type II	231031	Lane disease
2698	Knuckle pads-leukonychia-sensorineural deafness-palmoplantar keratoderma syndrome	420686	KWWH type IV	2632	Langer mesomelic dysplasia
2349	Kocher-Debré-Semelaigne syndrome	319254	Kyasianur forest disease	502	Langer-Giedion syndrome
679	Köhlmeier-Degos disease	319254	Kyasianur hemorrhagic fever	86897	Langerhans cell sarcoma
		79155	Kynureninase deficiency	2368	Laparoschisis
		1801	Kyphomelic dysplasia	→1159	Laplane-Fontaine-Lagardere syndrome
		275543	L1 syndrome	2363	LARD syndrome
		275543	L1CAM syndrome		
		79314	L-2-HGA		
		79314	L-2-hydroxyglutaric acidemia		

→ 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
98838	Large cell lymphoma of the mediastinum	199299	Late-onset isolated ACTH deficiency	650	Lecithin-cholesterol acyltransferase deficiency
626	Large congenital melanocytic nevus	79406	Late-onset junctional epidermolysis bullosa	199251	Ledderhose disease
633	Laron syndrome	231556	Late-onset localized junctional epidermolysis bullosa - intellectual disability	71273	Left renal vein entrapment syndrome
220465	Laron syndrome with immunodeficiency	79241	Late-onset multiple carboxylase deficiency	99111	Left superior caval vein persisting to left-sided atrium
220465	Laron-like syndrome	93589	Late-onset nephronophthisis	99111	Left superior vena cava persisting to left-sided atrium
633	Laron-type dwarfism	90186	Late-onset primary lymphedema	99111	Left SVC persisting to left-sided atrium
2370	Larsen-like osseous dysplasia - short stature	67042	Late-onset retinal degeneration	54260	Left ventricular hypertrabeculation
284139	Larsen-like syndrome, B3GAT3 type	2789	Lateral meningocele syndrome	54260	Left ventricular noncompaction
2808	Laryngeal abductor paralysis	141136	Laterofacial microsomia	99095	Left ventricular-to-right atrial communication
2375	Laryngeal abductor paralysis - intellectual disability	46059	Lathosterolosis	1757	Leg duplication - mirror foot
2407	Laryngeal and ocular granulation tissue in children from the Indian subcontinent syndrome	98964	Lattice corneal dystrophy type 1	2380	Legg-Calvé-Perthes disease
100083	Laryngeal endocrine tumor	98964	Lattice corneal dystrophy type I	549	Legionellosis
2407	Laryngo-onycho-cutaneous syndrome	99094	Laubry-Pezzi syndrome	549	Legionnaires disease
2004	Laryngo-tracheo-esophageal cleft	2398	Launois-Bensaude lipomatosis	137605	Legius syndrome
2005	Laryngo-tracheo-esophageal cleft - pulmonary hypoplasia	2377	Laurence-Moon syndrome	2789	Lehman syndrome
280205	Laryngo-tracheo-esophageal cleft type 0	2378	Laurin-Sandrow syndrome	1647	Leichtman-Wood-Rohn syndrome
93938	Laryngo-tracheo-esophageal cleft type 1	79086	Lawrence syndrome	255241	Leigh disease with leukodystrophy
93939	Laryngo-tracheo-esophageal cleft type 2	79086	Lawrence-Seip syndrome	70474	Leigh disease with myopathy
93940	Laryngo-tracheo-esophageal cleft type 3	137898	LBSL	255249	Leigh disease with nephrotic syndrome
93941	Laryngo-tracheo-esophageal cleft type 4	2369	LBWC syndrome	3008	Leigh necrotizing encephalopathy due to pyruvate carboxylase deficiency
2004	Laryngo-tracheo-esophageal diastema	2004	LC	3008	Leigh syndrome due to PC deficiency
2372	Laryngocoele	99900	LCAD	3008	Leigh syndrome due to pyruvate carboxylase deficiency
137935	Laryngotracheal angioma	650	LCAT deficiency	70474	Leigh syndrome with cardiomyopathy
1202	Larynx atresia	1486	LCCS1	255241	Leigh syndrome with leukodystrophy
99824	Lassa fever	137776	LCCS2	255249	Leigh syndrome with nephrotic syndrome
99824	Lassa hemorrhagic fever	137783	LCCS3	70472	Leigh syndrome, French-Canadian type
98974	Late hereditary endothelial dystrophy	98964	LCD1	70472	Leigh syndrome, Saguénay-Lac-Saint-Jean type
157716	Late infantile CACH syndrome	93558	LCDD	314	Leiner disease
168491	Late infantile NCL	98964	LCDI	71274	Leiomyomatosis peritonealis disseminate
168491	Late infantile neuronal ceroid lipofuscinosi	5	LCHAD deficiency	64720	Leiomyosarcoma
98816	Late onset benign childhood occipital epilepsy	52416	LCM	104076	Leiomyosarcoma of small intestine
79256	Late-infantile GM1 gangliosidosis	626	LCMN	213807	Leiomyosarcoma of the cervix uteri
206443	Late-infantile/juvenile Krabbe disease	363618	LCPS	213625	Leiomyosarcoma of the corpus uteri
247573	Late-onset citrullinemia type 1	65285	LDD	507	Leishmaniasis
247573	Late-onset citrullinemia type I	2364	LDH deficiency	140936	Lelis syndrome
399058	Late-onset distal crystallinopathy	284435	LDH-H subunit deficiency	137839	Lemierre postanginal sepsis
98912	Late-onset distal myopathy, Markesberry-Griggs type	284426	LDH-M subunit deficiency	137839	Lemierre syndrome
228227	Late-onset focal dermal elastosis	2616	Le Merrer syndrome	2382	Lennox-Gastaut syndrome
163708	Late-onset infantile spasms	330015	Lead intoxication	209959	Lens-induced endophthalmitis
		330015	Lead poisoning	209959	Lens-induced iridocyclitis
		3246	Learman syndrome	209959	Lens-induced uveitis
		65	Leber congenital amaurosis	568	Lenz microphthalmia
		104	Leber hereditary optic neuropathy		
		190	Leber miliary aneurysm		
		104	Leber optic atrophy		
		99718	Leber plus disease		
		98955	LECD		

→ 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
2658	Lenz-Majewski hyperostotic dwarfism	2736	Lethal omphalocele-cleft palate syndrome	210133	Leukonychia totalis - acanthosis-nigricans-like lesions - abnormal hair
500	LEOPARD syndrome	216804	Lethal osteogenesis imperfecta	2045	Leukonychia totalis - trichilemmal cysts - ciliary dystrophy
330032	Lepore - beta-thalassemia	1832	Lethal osteosclerotic bone dysplasia	79507	Leukotriene C4 synthase deficiency
508	Leprechaunism	210144	Lethal polymalformative syndrome, Boissel type	2743	Levic-Stefanovic-Nikolic syndrome
548	Leprosy	1234	Lethal popliteal pterygium syndrome	2388	Levine-Critchley syndrome
252031	Leptomeningeal melanomatosis	1423	Lethal recessive chondrodysplasia	216694	Levo-transposition of the great arteries
268838	Leptomelyolipoma	1662	Lethal restrictive dermopathy	95854	Levocardia
509	Leptospirosis	→56304	Lethal short-limb dwarfism, McAlister-Crane type	95854	Levocardia-situs inversus
2900	Leri pleonosteosis	79022	Lethal variant of Simpson-Golabi-Behmel syndrome	2363	Levy-Hollister syndrome
240	Léri-Weill dyschondrosteosis	99870	Letterer-Siwe disease	302	Lewandowsky-Lutz syndrome
240	Léri-Weill syndrome	58017	Leukemic reticuloendotheliosis	→1896	Lewis-Pashayan syndrome
510	Lesch-Nyhan syndrome	300878	Leukemic reticuloendotheliosis variant	48162	Lewis-Sumner syndrome
158687	Lethal acantholytic epidermolysis bullosa	2968	Leukocyte adhesion deficiency	755	Leydig cell hypoplasia
314718	Lethal arteriopathy syndrome due to fibulin-4 deficiency	99842	Leukocyte adhesion deficiency type I	96265	Leydig cell hypoplasia due to complete LH receptor inactivation
53696	Lethal arthrogryposis - anterior horn cell disease	99843	Leukocyte adhesion deficiency type II	96265	Leydig cell hypoplasia due to complete LH resistance
1187	Lethal ataxia with deafness and optic atrophy	99844	Leukocyte adhesion deficiency type III	96265	Leydig cell hypoplasia due to complete luteinizing hormone receptor inactivation
1420	Lethal chondrodysplasia, Moerman type	99844	Leukocyte adhesion deficiency-1 variant	96265	Leydig cell hypoplasia due to complete luteinizing hormone resistance
1421	Lethal chondrodysplasia, Seller type	439224	Leukocyte chemotactic factor-2 amyloidosis	96265	Leydig cell hypoplasia due to complete luteinizing hormone resistance
1486	Lethal congenital contracture syndrome type 1	77295	Leukodystrophy with oligodontia	325448	Leydig cell hypoplasia due to LHB deficiency
137776	Lethal congenital contracture syndrome type 2	163684	Leukoencephalopathy - dystonia - motor neuropathy	325448	Leydig cell hypoplasia due to luteinizing hormone subunit beta deficiency
137783	Lethal congenital contracture syndrome type 3	83629	Leukoencephalopathy - metaphyseal chondrodysplasia	96266	Leydig cell hypoplasia due to partial LH receptor inactivation
330050	Lethal encephalopathy due to mitochondrial and peroxisomal fission defect	139444	Leukoencephalopathy with bilateral anterior temporal lobe cysts	96266	Leydig cell hypoplasia due to partial LH resistance
1972	Lethal faciocardiomelic dysplasia	137898	Leukoencephalopathy with brain stem and spinal cord involvement - high lactate	96266	Leydig cell hypoplasia due to partial luteinizing hormone receptor inactivation
444069	Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome	137898	Leukoencephalopathy with brain stem and spinal cord involvement - lactate elevation	96266	Leydig cell hypoplasia due to partial luteinizing hormone resistance
439897	Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome	363540	Leukoencephalopathy with mild cerebellar ataxia and white matter edema	99824	LF
1046	Lethal hemolytic anemia - genital anomalies	135	Leukoencephalopathy with vanishing white matter	266	LGMD1A
254857	Lethal infantile mitochondrial disease	137639	Leukoencephalopathy-ataxia-hypodontia-hypomyelination syndrome	264	LGMD1B
254857	Lethal infantile mitochondrial myopathy	2386	Leukoencephalopathy-palmoplantar keratoderma syndrome	265	LGMD1C
2347	Lethal Kniest-like dysplasia	314051	Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome	34516	LGMD1D
2371	Lethal Larsen-like syndrome	1816	Leukomelanoderma - intellectual disability - hypotrichosis	34517	LGMD1E
86879	Lethal midline granuloma	2387	Leukonychia totalis	55595	LGMD1F
33108	Lethal multiple pterygium syndrome			55596	LGMD1G
435845	Lethal neonatal rigidity-multifocal seizure syndrome			238755	LGMD1H
435845	Lethal neonatal spasticity-epileptic encephalopathy syndrome			267	LGMD2A
300313	Lethal neurodegenerative disorder due to copper transport defect			268	LGMD2B
293925	Lethal occipital encephalocele-skeletal dysplasia syndrome			353	LGMD2C
				62	LGMD2D
				119	LGMD2E

→ 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
219	LGMD2F	62	Limb-girdle muscular dystrophy due to alpha-sarcoglycan deficiency	220402	Limited cutaneous systemic scleroderma
34514	LGMD2G	119	Limb-girdle muscular dystrophy due to beta-sarcoglycan deficiency	220402	Limited cutaneous systemic sclerosis
1878	LGMD2H	267	Limb-girdle muscular dystrophy due to calpain deficiency	220407	Limited systemic sclerosis
34515	LGMD2I	265	Limb-girdle muscular dystrophy due to caveolin-3 deficiency	168491	LINCL
140922	LGMD2J	219	Limb-girdle muscular dystrophy due to delta-sarcoglycan deficiency	892	Lindau disease
86812	LGMD2K	268	Limb-girdle muscular dystrophy due to dyserlin deficiency	3077	Lindsay-Burn syndrome
206549	LGMD2L	34515	Limb-girdle muscular dystrophy due to FKRP deficiency	79150	Linear and whorled nevoid hypermelanosis
206554	LGMD2M	353	Limb-girdle muscular dystrophy due to gamma-sarcoglycan deficiency	140933	Linear atrophoderma of Moulin
206559	LGMD2N	264	Limb-girdle muscular dystrophy due to lamin A/C deficiency	228236	Linear focal dermal elastosis
206564	LGMD2O	266	Limb-girdle muscular dystrophy due to myotilin deficiency	2611	Linear hamartoma syndrome
280333	LGMD2P	445110	Limb-girdle muscular dystrophy due to POMK deficiency	46488	Linear IgA dermatosis
254361	LGMD2Q	34514	Limb-girdle muscular dystrophy due to telethonin deficiency	254379	Linear lichen planus
363543	LGMD2R	1878	Limb-girdle muscular dystrophy due to TRIM32 deficiency	254379	Linear LP
369840	LGMD2S	257	Limb-girdle muscular dystrophy with epidermolysis bullosa simplex	2612	Linear nevus sebaceus syndrome
363623	LGMD2T	52430	Limb-girdle muscular dystrophy with Paget disease of bone	2611	Linear verrucous nevus syndrome
352479	LGMD2U	69085	Limb-mammary syndrome	36273	Linitis plastica of the stomach
445110	LGMD due to POMK deficiency	171673	Limbal stem cell deficiency	888	Lip-pit syndrome
93557	LHCDD	83467	Limbic encephalitis - neuromyotonia - hyperhidrosis - polyneuropathy	435660	LIPE-related familial partial lipodystrophy
65285	Lhermitte-Duclos disease	276402	Limbic encephalitis with caspr2 antibodies	435660	LIPE-related FPLD
104	LHON	329341	Limbic encephalitis with dipeptidyl-peptidase 6 antibodies	77243	Lipedema
313	LI	329341	Limbic encephalitis with DPP6 antibodies	255182	Lipoamide dehydrogenase deficiency
524	Li-Fraumeni syndrome	329341	Limbic encephalitis with DPPX antibodies	528	Lipoatrophic diabetes
49804	Lichen amyloidosis	163908	Limbic encephalitis with leucine-rich glioma-inactivated 1 antibodies	156156	Lipoatrophy with diabetes, leukomelanodermic papules, liver steatosis, and hypertrophic cardiomyopathy
49804	Lichen amyloidosis	163908	Limbic encephalitis with LGI1 antibodies	247762	Lipoblastoma
525	Lichen follicularis	217253	Limbic encephalitis with N-methyl-D-aspartate receptor antibodies	90156	Lipodystrophia centrifugalis abdominalis infantilis
525	Lichen planopilaris	163914	Limbic encephalitis with nCMAgs antibodies	50811	Lipodystrophy - intellectual disability - deafness
254395	Lichen planus actinus	217253	Limbic encephalitis with NMDA receptor antibodies	3163	Lipodystrophy - Rieger anomaly - diabetes
525	Lichen planus follicularis	163914	Limbic encephalitis with novel cell membrane antigen antibodies	1979	Lipodystrophy due to peptidic growth factors deficiency
254478	Lichen planus pemphigoides	254857	LIMD	401859	Lipoic acid synthetase deficiency
254463	Lichen planus pigmentosa	366	Limit dextrinosis	139436	Lipoid dermatopathitis
254463	Lichen planus pigmentosus			530	Lipoid proteinosis
254463	Lichen planus pigmentosus inversus			36397	Lipomatosis dolorosa
254395	Lichen planus subtropicus			238593	Lipomatous mesenteritis
254395	Lichen planus tropicus			812	Lipomucopolysaccharidosis
254395	Lichenoid melanodermatitis			268835	Lipomyelomeningocele
2390	Lichstenstein syndrome			329481	Lipoprotein glomerulopathy
448251	Lichtenstein-Knorr syndrome			69078	Liposarcoma
526	Liddle syndrome			238593	Liposclerotic mesenteritis
1275	Liebenberg syndrome			401862	Lipoyle transferase 1 deficiency
99812	LIG4 syndrome			447795	Lipoyle transferase 2 deficiency
99812	Ligase 4 syndrome			98955	Lisch epithelial corneal dystrophy
93557	Light and heavy chain deposition disease			2400	Lisker-Garcia-Ramos syndrome
93558	Light chain deposition disease			101003	Lison syndrome
85443	Light-chain amyloidosis				
97231	Ligneous conjunctivitis				
2369	Limb body wall complex				
2492	Limb transversal defect - cardiac anomaly				
974	Limb, scalp and skull defects				
86812	Limb-girdle muscular dystrophy - intellectual disability				

→ 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
531	Lissencephaly due to 17p13.3 deletion	314709	Localized immunoglobulinic amyloidosis	363447	Lower extremity-predominant autosomal dominant proximal spinal muscular atrophy
95232	Lissencephaly due to LIS1 mutation	251393	Localized junctional epidermolysis bullosa, non-Herlitz type	363454	Lower extremity-predominant autosomal dominant proximal spinal muscular atrophy with contractures
171680	Lissencephaly due to TUBA1A mutation	90398	Localized lichen myxedematosus with mixed features of different subtypes	209341	Lower extremity-predominant autosomal dominant proximal spinal muscular atrophy without contractures
89844	Lissencephaly syndrome, Norman-Roberts type	90399	Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms	2487	Lower limb deficiency - hypospadias
2148	Lissencephaly type 1 due to doublecortin gene mutation	178517	Localized pagetoid reticulosis	295051	Lower limb hypertrophy
352682	Lissencephaly type 2 without muscular or eye involvement	263534	Localized PSS	141064	Lower lip fistula
352682	Lissencephaly type 2 without muscular or ocular involvement	163927	Localized pustular psoriasis	276435	Lower motor neuron syndrome with late-adult onset
86821	Lissencephaly type 3 - familial fetal akinesia sequence	90289	Localized scleroderma	844	Lown-Ganong-Levine syndrome
86822	Lissencephaly type 3 - metacarpal bone dysplasia	2406	Locked-in syndrome	1533	Lowry syndrome
100011	Lissencephaly with cerebellar hypoplasia type A	75566	Loeffler endocarditis	2409	Lowry-MacLean syndrome
100012	Lissencephaly with cerebellar hypoplasia type B	60030	Loeys-Dietz syndrome	1824	Lowry-Wood syndrome
100013	Lissencephaly with cerebellar hypoplasia type C	2407	LOGIC syndrome	2003	Lowry-Yong syndrome
100014	Lissencephaly with cerebellar hypoplasia type D	250831	Logopenic primary progressive aphasia	254478	LP pemphigoides
100015	Lissencephaly with cerebellar hypoplasia type E	250831	Logopenic progressive aphasia	254463	LP pigmentosa
100016	Lissencephaly with cerebellar hypoplasia type F	2404	Loiasis	254463	LP pigmentosus
533	Listeriosis	5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	250831	LPA
1680	Little syndrome	99900	Long chain acyl-CoA dehydrogenase deficiency	71274	LPD
820	Livedo racemosa and cerebrovascular accidents	3363	Long eyelashes - intellectual disability	329481	LPG
820	Livedo reticularis and cerebrovascular accidents	90647	Long QT interval - deafness	470	LPI
79095	Liver disease - retinitis pigmentosa - polyneuropathy - epilepsy	65283	Long QT syndrome - syndactyly	309015	LPL deficiency
369	Liver glycogen phosphorylase deficiency	37553	Long QT syndrome type 7	163927	LPP
98818	LKS	65283	Long QT syndrome type 8	525	LPP
363618	LMNA-related cardiocutaneous progeria syndrome	180157	Longitudinal vaginal septum	37553	LQT7
157973	LMNA-related congenital muscular dystrophy	52054	Longman-Tolmie syndrome	65283	LQT8
33108	LMPS	168	Loose anagen syndrome	314051	LTBL
69085	LMS	411602	LOPD	79507	LTC4 synthase deficiency
93924	Lobar holoprosencephaly	2832	Lopes-Gorlin syndrome	2004	LTEC
666	Lobstein disease	2266	Lopes-Marques de Faria syndrome	280205	LTEC0
2440	Lobster-claw deformity	67042	LORD	93938	LTEC1
2407	LOC syndrome	79395	Loricrin keratoderma	93939	LTEC2
314709	Localized AL amyloidosis	803	Lou Gehrig disease	93940	LTEC3
93685	Localized Castleman disease	100	Louis-Bar syndrome	93941	LTEC4
263534	Localized deciduous skin	171215	Low anorectal malformation	93938	LTEC I
79400	Localized epidermolysis bullosa simplex	251633	Low grade ependymoma	93939	LTEC II
90289	Localized fibrosing scleroderma	69663	Low phospholipid associated cholelithiasis	93940	LTEC III
		140949	Low-flow priapism	93941	LTEC IV
		1652	Low-molecular-weight proteinuria with hypercalciuria and nephrocalcinosis	53351	Lubag
		534	Lowe disease	53351	Lubag syndrome
		534	Lowe oculo-cerebro-renal syndrome	2575	Lubani-Al Saleh-Teebi syndrome
		534	Lowe syndrome	2410	Lubinsky syndrome
		2408	Lowe-Kohn-Cohen syndrome	→1762	Lubs-Arena syndrome
				2312	Lucey-Driscoll syndrome
				776	Lujan syndrome
				776	Lujan-Fryns syndrome
				319213	Lujo hemorrhagic fever

→ 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
268388	Lumbosacral spina bifida aperta	329998	Lymphomatous meningitis	2427	Macrocephaly - short stature - paraplegia
268758	Lumbosacral spina bifida cystica	67038	Lymphoplasmacytic leukemia	2429	Macrocephaly - spastic paraplegia - dysmorphism
97332	Lunatomalacia	443159	Lymphoplasmacytic lymphoma without IgM production	217335	Macrocephaly-alopecia-cutis laxa-scoliosis syndrome
2928	Lundberg syndrome	443159	Lymphoplasmacytic lymphoma without Immunoglobulin M production	210548	Macrocephaly-autism syndrome
1120	Lung agenesis - heart defect - thumb anomalies	280302	Lymphoplasmacytic sclerosing pancreatitis	60040	Macrocephaly-capillary malformation syndrome
137631	Lung fibrosis - immunodeficiency - 46,XX gonadal dysgenesis	67038	Lymphoplasmacytoid immunocytoma	397612	Macrocephaly-developmental delay syndrome
90285	Lupus erythematosus panniculitis	144	Lynch syndrome	2563	Macrocephaly-obesity-mental disability-ocular abnormalities syndrome
90285	Lupus erythematosus profundus	1123	Lynch-Lee-Murday syndrome	79489	Macrocystic lymphangioma
90283	Lupus erythematosus tumidus	3196	Lyngstadaas syndrome	79489	Macrocystic lymphatic malformation
1173	Luteinizing hormone-releasing hormone deficiency with ataxia	98842	LyP	295044	Macrodactyly of fingers
302	Lutz-Lewandowsky epidermolympiasis verruciformis	2203	Lysine alpha-ketoglutarate reductase deficiency	295241	Macrodactyly of fingers, bilateral
→2697	Lutz-Richner-Landolt syndrome	470	Lysinuric protein intolerance	295239	Macrodactyly of fingers, unilateral
54260	LVNC	275761	Lysosomal acid lipase deficiency	295047	Macrodactyly of foot
537	Lyell syndrome	61	Lysosomal alpha-D-mannosidase deficiency	295245	Macrodactyly of foot, bilateral
86869	LYG	309288	Lysosomal alpha-D-mannosidase deficiency, adult form	295243	Macrodactyly of foot, unilateral
91546	Lyme borreliosis	309282	Lysosomal alpha-D-mannosidase deficiency, infantile form	295044	Macrodactyly of hand
91546	Lyme disease	34587	Lysosomal glycogen storage disease with normal acid maltase activity	295241	Macrodactyly of hand, bilateral
538	Lymphangioleiomyomatosis	79284	Lysosomal membrane cobalamin transporter deficiency	295239	Macrodactyly of hand, unilateral
2035	Lymphatic filariasis	93561	Lysozyme amyloidosis	295047	Macrodactyly of toes
86915	Lymphedema - atrial septal defects - facial changes	90020	Lytic-Bodig disease	295245	Macrodactyly of toes, bilateral
86914	Lymphedema - cerebral arteriovenous anomaly	330041	M hemoglobinopathy	295243	Macrodactyly of toes, unilateral
86917	Lymphedema - cleft palate	247262	Mabry syndrome	158061	Macrophage activation syndrome
33001	Lymphedema - distichiasis	98938	MAC	592	Macrophagic myofasciitis
1563	Lymphedema - hypoparathyroidism syndrome	2083	Mac Dermot-Winter syndrome	2432	Macrosomia - microphthalmia - cleft palate
2136	Lymphedema - lymphangiectasia - intellectual disability	36412	Mac Duffie hypocomplementemic urticarial vasculitis	2563	Macrosomia-obesity-macrocephaly-ocular abnormalities syndrome
→33001	Lymphedema - ptosis	36412	Mac Duffie syndrome	141276	Macrostomia
→28982 5	Lymphedema praecox	2220	MacDermot-Patton-Williams syndrome	83619	Macrostomia - preauricular tags - external ophthalmoplegia
→28982 5	Lymphedema tarda	98757	Machado disease	→18205 0	Macrothrombocytopenia with leukocyte inclusions
662	Lymphedema with yellow nails	98757	Machado-Joseph disease	220448	Macrothrombocytopenia with mitral valve insufficiency
158793	Lymphadenopathic mastocytosis with eosinophilia	276238	Machado-Joseph disease type 1	217335	MACS syndrome
86870	Lymphoblastoid variant of NK-cell lymphoma	276241	Machado-Joseph disease type 2	137814	Macular amyloidosis
65279	Lymphocytic colitis	276244	Machado-Joseph disease type 3	91494	Macular coloboma-cleft palate-hallux valgus syndrome
314970	Lymphocytic hypereosinophilic syndrome	319229	Machupo hemorrhagic fever	98969	Macular corneal dystrophy
79128	Lymphocytic interstitial pneumonia	79495	Macias Flores-Garcia Cruz-Rivera syndrome	79457	Maculopapular cutaneous mastocytosis
314970	Lymphocytic variant HES	1574	Mackay-Shek-Carr syndrome	90287	Maculopapular lupus rash
289682	Lymphoepithelial-like carcinoma	2477	Macrencephaly	2457	MAD
86886	Lymphogranulomatosis X	357158	Macroblepharon - ectropion - hypertelorism - macrostomia syndrome	26791	MAD deficiency
314970	Lymphoid HES	60040	Macrocephaly - cutis marmorata telangiectatica congenita	26791	MADD
79128	Lymphoid interstitial pneumonia			35688	Madelung deformity
86869	Lymphomatoid granulomatosis			295223	Madelung deformity, bilateral
98842	Lymphomatoid papulosis				

→ 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
295221	Madelung deformity, unilateral	1646	Male sterility due to chromosome Y deletion	398987	Malignant teratoma of ovary
2398	Madelung disease	3000	Male-limited precocious puberty	99868	Malignant thymoma
137867	Madras motor neuron disease	99915	Malignant granulosa cell tumor of ovary	252212	Malignant triton tumor
48162	MADSAM	289385	Malignancy diagnosed during pregnancy	180242	Malignant tubal tumor
2583	Madura foot	98839	Malignant angioendotheliomatosis	180242	Malignant tumor of fallopian tubes
1942	MAE	679	Malignant atrophic papulosis	943	Malonic aciduria
171709	Mae infertility due to round-headed spermatozoa	99912	Malignant dysgerminomatous germ cell tumor of ovary	943	Malonyl-CoA decarboxylase deficiency
199354	Maeda syndrome	276145	Malignant epithelial tumor of salivary glands	2229	Malouf syndrome
163634	Maffucci syndrome	213837	Malignant germ cell tumor of cervix uteri	99090	Malposition of the coronary ostium
324972	MAGIC syndrome	213751	Malignant germ cell tumor of corpus uteri	→29384 3	Malpuech facial clefting syndrome
438274	Mahvash disease	423	Malignant hyperpyrexia	→29384 3	Malpuech syndrome
77297	Majeed syndrome	423	Malignant hyperthermia	52417	MALT lymphoma
2637	Majewski osteodysplastic primordial dwarfism type II	2215	Malignant hyperthermia - arthrogryposis - torticollis	103907	Maltase-glucoamylase deficiency
70470	Major hyperlipidemia	168999	Malignant melanoma of the mucosa	52417	MALToma
210272	Mal de débarquement	293181	Malignant migrating partial epilepsy of infancy	50920	Mammary polyadenomatosis
87503	Mal de Meleda	293181	Malignant migrating partial seizures of infancy	238744	Mammary-digital-nail syndrome
556	Malakoplakia	213512	Malignant mixed epithelial mesenchymal tumor of ovary	397941	MAN1B1-CDG
420179	Malan overgrowth syndrome	213610	Malignant mixed Müllerian tumor of corpus uteri	244310	Man5GlcNAc2-PP-Dol flippase deficiency
673	Malaria	213787	Malignant Müllerian mixed tumor of cervix uteri	141174	Mandibular arteriovenous malformation
75376	Malattia leventinese	3148	Malignant neurilemmoma	363649	Mandibular hypoplasia-deafness-progeroid syndrome
401973	Male EBP disorder with neurological defects	3148	Malignant neurofibroma	246	Mandibulofacial dysostosis with postaxial limb anomalies
2234	Male hypergonadotropic hypogonadism - intellectual disability - skeletal anomalies	206538	Malignant non-dysgerminomatous germ cell tumor of ovary	91412	Mandibulo-palpebral synkinesis - ptosis
171709	Male infertility due to globozoospermia	99912	Malignant ovarian dysgerminoma	2457	Mandibuloacral dysplasia
137893	Male infertility due to large-headed multiflagellar polyploid spermatozoa	3286	Malignant paroxysmal ventricular tachycardia	90153	Mandibuloacral dysplasia with type A lipodystrophy
→39980	Male infertility due to NANOS1 mutation	252128	Malignant perineurioma	90154	Mandibuloacral dysplasia with type B lipodystrophy
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	3148	Malignant peripheral nerve sheath tumor	357158	Mandibulofacial dysostosis - macroblepharon - macrostomia
→39980	Male infertility with normal virilization due to maturation arrest	252212	Malignant peripheral nerve sheath tumor with rhabdomyosarcomatous differentiation	443995	Mandibulofacial dysostosis with alopecia
→39980	Male infertility with normal virilization due to meiosis defect	213812	Malignant peripheral neuroectodermal tumor of cervix uteri	245	Mandibulofacial dysostosis with preaxial limb anomalies
399808	Male infertility with teratozoospermia due to single gene mutation	213630	Malignant peripheral neuroectodermal tumor of corpus uteri	861	Mandibulofacial dysostosis without limb anomalies
753	Male pseudohermaphroditism due to 5-alpha-reductase 2 deficiency	168811	Malignant peritoneal mesothelioma	79113	Mandibulofacial dysostosis, Guion-Almeida type
752	Male pseudohermaphroditism due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	69077	Malignant rhabdoid tumor	1131	Mandibulofacial dysostosis, Toriello type
755	Male pseudohermaphroditism due to LH resistance or LHB deficiency	3148	Malignant schwannoma	79113	Mandibulofacial dysostosis-microcephaly syndrome
755	Male pseudohermaphroditism due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency	99916	Malignant Sertoli-Leydig cell tumor of ovary	306682	Manganese intoxication
				306682	Manganese poisoning
				306682	Manganism
				2717	Manitoba oculotrichoanal syndrome
				79327	Mannosyltransferase 1 deficiency

→ 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
79326	Mannosyltransferase 2 deficiency	→29386 4	Martínez-Frías syndrome	663	Maternally-inherited chronic progressive external ophthalmoplegia
79321	Mannosyltransferase 6 deficiency	1387	Martsolf syndrome	663	Maternally-inherited CPEO
79328	Mannosyltransferase 7-9 deficiency	2466	MASA syndrome	225	Maternally-inherited diabetes and deafness
79324	Mannosyltransferase 8 deficiency	→28496 3	MASS syndrome	255210	Maternally-inherited infantile subacute necrotizing encephalopathy
2459	Mansonelliasis	66661	Mast cell sarcoma	255210	Maternally-inherited Leigh disease
2459	Mansonellosis	101001	Mast syndrome	255210	Maternally-inherited Leigh syndrome
52416	Mantle cell lymphoma	2135	Mastocytosis - short stature - hearing loss	254851	Maternally-inherited mitochondrial dystonia
52416	Mantle zone lymphoma	3282	MAT	663	Maternally-inherited progressive external ophthalmoplegia
511	Maple syrup urine disease	168598	MAT deficiency	320360	Maternally-inherited spastic paraparesia
→2712	Marashi-Gorlin syndrome	168598	MAT I/III deficiency	320360	Maternally-inherited SPG
2785	Marble brain disease	254534	Maternal 14q32.2 hypermethylation syndrome	2015	Mathieu-De Broca-Bony syndrome
228157	Marburg acute multiple sclerosis	254528	Maternal 14q32.2 microdeletion syndrome	2470	Matthew-Wood syndrome
99826	Marburg hemorrhagic fever	275944	Maternal anti-Kell alloimmunization	552	Maturity-onset diabetes of the young
99826	Marburg virus disease	254528	Maternal del(14)(q32.2)	293603	Maumenee corneal dystrophy
221074	Marchiafava-Bignami disease	2209	Maternal hyperphenylalaninemia	141171	Maxillary arteriovenous malformation
447	Marchiafava-Micheli disease	2216	Maternal hyperthermia induced birth defects	1248	Maxillonasal dysostosis
91412	Marcus-Gunn phenomenon	254528	Maternal monosomy 14q32.2	1248	Maxillonasal dysplasia
91412	Marcus-Gunn syndrome	2209	Maternal phenylketonuria	→18205 0	May-Hegglin anomaly
2461	Marden-Walker syndrome	2209	Maternal PKU	→18205 0	May-Hegglin syndrome
2460	Marden-Walker-like syndrome	411712	Maternal riboflavin deficiency	→18205 0	May-Hegglin thrombocytopenia
1120	Mardini-Nyhan syndrome	251009	Maternal uniparental disomy of chromosome 1	3109	Mayer-Rokitansky-Küster-Hauser syndrome
558	Marfan syndrome	96179	Maternal uniparental disomy of chromosome 2	247775	Mayer-Rokitansky-Küster-Hauser syndrome type 1
284963	Marfan syndrome type 1	96180	Maternal uniparental disomy of chromosome 4	2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2
284973	Marfan syndrome type 2	96181	Maternal uniparental disomy of chromosome 6	57782	Mazabraud syndrome
2462	Marfanoid craniosynostosis syndrome	96183	Maternal uniparental disomy of chromosome 9	91138	MC
→60030	Marfanoid habitus - craniosynostosis syndrome	97678	Maternal uniparental disomy of chromosome 13	71529	MC4R deficiency
314041	Marfanoid habitus - inguinal hernia - advanced bone age	96184	Maternal uniparental disomy of chromosome 14	93554	MC type II
2463	Marfanoid habitus - intellectual disability, autosomal recessive	96185	Maternal uniparental disomy of chromosome 16	93555	MC type III
2464	Marfanoid syndrome, De Silva type	96186	Maternal uniparental disomy of chromosome 20	254519	MCA due to 14q32.2 maternally expressed gene defect
→3253	Margarita island ectodermal dysplasia	96187	Maternal uniparental disomy of chromosome 21	42	MCAD deficiency
444	Marie Unna congenital hypotrichosis	96188	Maternal uniparental disomy of chromosome 22	42	MCADD
444	Marie Unna hereditary hypotrichosis	261519	Maternal uniparental disomy of chromosome X	300496	MCAHS type 2
101104	Marin-Amat syndrome	96186	Maternal UPD20	→56304	McAlister-Crane syndrome
559	Marinesco-Sjögren syndrome	1349	Maternally-inherited cardiomyopathy and deafness	60040	MCAP
2717	Marles syndrome	1349	Maternally-inherited cardiomyopathy and hearing loss	368	McArdle disease
2717	Marles-Greenberg-Persaud syndrome			79140	MCC
583	Maroteaux-Lamy disease			6	MCC deficiency
2767	Maroteaux-Le Merrer-Bensahel syndrome			85195	McCabe's disease
950	Maroteaux-Malamut syndrome			6	MCCD
1423	Maroteaux-Stanescu-Cousin syndrome			562	McCune-Albright syndrome
1040	Maroteaux-Verloes-Stanescu syndrome			93686	MCD
560	Marshall syndrome				
42642	Marshall syndrome with periodic fever				
561	Marshall-Smith syndrome				
908	Martin-Bell syndrome				
85321	Martin-Probst 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
98969	MCD	564	Meckel syndrome	2241	Megacystis-microcolon-intestinal hypoperistalsis-hydronephrosis syndrome
1851	MCDK	3032	Meckel syndrome type 7	2604	Megaduodenum and/or megacystis
2471	McDonough syndrome	564	Meckel-Gruber syndrome	402023	Megakaryoblastic acute myeloid leukemia with t(1;22)(p13;q13)
→35722 5	McDowall syndrome	3032	Meckel-like syndrome type 1	402023	Megakaryoblastic AML with t(1;22)(p13;q13)
75327	MCDR1	70588	Meconium aspiration syndrome	2478	Megalencephalic leukodystrophy
319640	MCDR2	314376	Meconium ileus due to guanylate cyclase 2C deficiency	2478	Megalencephalic leukoencephalopathy with subcortical cysts
36412	McDuffie hypocomplementemic urticarial vasculitis	93308	MED1	2477	Megalencephaly
36412	McDuffie syndrome	93307	MED4	60040	Megalencephaly - cutis marmorata telangiectatica congenita
308425	MCEE deficiency	93311	MED5	2478	Megalencephaly - cystic leukodystrophy
158668	McGrath syndrome	98838	Med-DLBCL	83473	Megalencephaly - polymicrogyria - postaxial polydactyly - hydrocephalus
2473	McKusick-Kaufman syndrome	3453	MEDAC syndrome	60040	Megalencephaly-capillary malformation syndrome
52416	MCL	2476	Medeira-Dennis-Donnai syndrome	60040	Megalencephaly-capillary malformation-polymicrogyria syndrome
59306	McLeod neuroacanthocytosis syndrome	57196	Medial condensing osteitis of the clavicle	238763	Megalocornea - spherophakia - secondary glaucoma
60040	MCM	2006	Median cleft lip/mandibule	2479	Megalocornea-intellectual disability syndrome
60040	MCMTC	2006	Median cleft lower facial stage	50815	Mégarbané-Loiselet syndrome
77298	MCOPS3	1993	Median cleft of the upper lip - corpus callosum lipoma - cutaneous polyps	238637	Megaureter-megacystis syndrome
85275	MCOPS4	141239	Median cleft of the upper lip and maxilla	352328	MEGDEL syndrome
178364	MCOPS5	2699	Median nodule of the upper lip	3038	Mehes syndrome
139471	MCOPS6	98838	Mediastinal diffuse large-cell lymphoma with sclerosis	85282	MEHMO syndrome
2556	MCOPS7	63999	Mediastinal fibrosis	2196	Meier-Blumberg-Imahorn syndrome
3434	MCOPS8	370127	Medich giant platelet syndrome	2554	Meier-Gorlin syndrome
2470	MCOPS9	370127	Medich macrothrombocytopenia	90186	Meige disease
77299	MCOPS10	231	Medina worm disease	93964	Meige dystonia
2512	MCPH	231	Medinensis	90186	Meige lymphedema
2001	McPherson-Clemens syndrome	231214	Mediterranean anemia	93964	Meige syndrome
2999	McPherson-Hall syndrome	100025	Mediterranean lymphoma	→90186	Meige-like disease
228418	MCSZ	101022	Mediterranean macrothrombocytopenia	314451	Meigs syndrome
59	MCT8 deficiency	42	Medium chain acyl-CoA dehydrogenase deficiency	98868	Melanesian elliptocytosis
809	MCTD	171851	MEDNIK syndrome	98868	Melanesian ovalocytosis
523	MCUL	3050	Medrano-Roldan syndrome	252206	Melanoma and neural system tumor syndrome
565	MD	29073	Medullary plasmacytoma	97338	Melanoma of soft parts
273	MD1	1309	Medullary sponge kidney	97338	Melanoma of soft tissue
258	MDC1A	1332	Medullary thyroid carcinoma	252206	Melanoma-astrocytoma syndrome
98893	MDC1B	616	Medulloblastoma	404560	Melanoma-pancreatic cancer syndrome
→37095 3	MDC1C	251858	Medulloblastoma with extensive nodularity	79146	Melanosis diffusa congenita
→37095 3	MDC1D	251883	Medulloepithelioma	79146	Melanosis universalis hereditaria
210272	MdD	98954	Meesmann corneal dystrophy	550	MELAS
210272	MDDS	97252	Mega-cisterna magna	87503	Meleda disease
1836	MDK	66629	Megacolon - microcephaly		
238744	MDN syndrome	280671	Megaonial congenital muscular dystrophy		
363649	MDP syndrome	238637	Megacystis-megaureter syndrome		
3097	Meacham syndrome	2241	Megacystis-microcolon-intestinal hypoperistalsis syndrome		
3097	Meacham-Winn-Culler syndrome				
435438	MEAK				
370997	MEB disease with bilateral multicystic leucodystrophy				
588	MEB syndrome				
98954	MECD				

→ 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
2482	Melhem-Fahl syndrome	319595	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency	2496	Mesomelic dysplasia with acral synostoses, Verloes-David-Pfeiffer type
31202	Melioidosis	2494	Menetrier disease	1836	Mesomelic dysplasia, Kantaputra type
2483	Melkersson-Rosenthal syndrome	3216	Mengel-Konigsmark syndrome	85170	Mesomelic dysplasia, Savarirayan type
2484	Melnick-Needles osteodysplasty	252046	Meningeal melanocytoma	1836	Mesomelic dysplasia, Thai type
2484	Melnick-Needles syndrome	2495	Meningioma	171690	Metabolic myopathy due to lactate transporter defect
2485	Melorheostosis	→823	Meningocele	2499	Metachondromatosis
1879	Melorheostosis with osteopoikilosis	33475	Meningococcal meningitis	512	Metachromatic leukodystrophy
93571	Membranoproliferative glomerulonephritis type 2	565	Menkes disease	309271	Metachromatic leukodystrophy, adult form
652	MEN1	565	Menkes syndrome	309263	Metachromatic leukodystrophy, juvenile form
653	MEN2	75858	Mental retardation - truncal obesity - retinal dystrophy - micropenis	309256	Metachromatic leukodystrophy, late infantile form
247698	MEN2A	330021	Mercurialism	1240	Metaphyseal acroscyphodysplasia
247709	MEN2B	330021	Mercury intoxication	1040	Metaphyseal anadysplasia
276152	MEN4	330021	Mercury poisoning	166035	Metaphyseal chondrodysplasia - retinitis pigmentosa
401973	MEND syndrome	79140	Merkel cell carcinoma	33067	Metaphyseal chondrodysplasia, Jansen type
319552	Mendelian susceptibility to interleukin 12 receptor beta 1 deficiency	258	Merosin-negative congenital muscular dystrophy	166038	Metaphyseal chondrodysplasia, Kaitila type
99898	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency	551	MERRF	175	Metaphyseal chondrodysplasia, McKusick type
319547	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency	54370	Mesangiocapillary glomerulonephritis	174	Metaphyseal chondrodysplasia, Schmid type
319558	Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency	386	Mesenchymal hamartoma of liver	2501	Metaphyseal chondrodysplasia, Spahr type
319552	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	238593	Mesenteric lipogranuloma	99646	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria
99898	Mendelian susceptibility to mycobacterial diseases due to complete interferon gamma receptor 1 deficiency	238593	Mesenteric panniculitis	2502	Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome
319547	Mendelian susceptibility to mycobacterial diseases due to complete interferon gamma receptor 2 deficiency	99701	Mesial temporal lobe epilepsy with hippocampal sclerosis	2504	Metaphyseal dysplasia - maxillary hypoplasia - brachydacty
319558	Mendelian susceptibility to mycobacterial diseases due to complete interleukin 12B deficiency	295004	Mesoaxial polydactyly of fingers	→175	Metaphyseal dysplasia without hypotrichosis
319563	Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency	295173	Mesoaxial polydactyly of fingers, bilateral	85188	Metaphyseal dysplasia, Braun-Tischert type
319600	Mendelian susceptibility to mycobacterial diseases due to partial interferon regulatory factor 8 deficiency	295171	Mesoaxial polydactyly of fingers, unilateral	3005	Metaphyseal dysplasia, Pyle type
319600	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency	295010	Mesoaxial polydactyly of toes	213531	Metaplastic carcinoma of the breast
319595	Mendelian susceptibility to mycobacterial diseases due to partial signal transducer and activator of transcription 1 deficiency	295185	Mesoaxial polydactyly of toes, bilateral	2635	Metatropic dwarfism
		295183	Mesoaxial polydactyly of toes, unilateral	2635	Metatropic dysplasia
		157801	Mesoaxial synostotic syndactyly with phalangeal reduction	88639	Methacrylic aciduria
		95443	Mesocardia	31825	Methanol poisoning
		289	Mesodermic dysplasia	1923	Methimazole embryofetopathy
		2496	Mesomelia-synostoses syndrome	168598	Methionine adenosyltransferase deficiency
		2496	Mesomelia-synostoses syndrome, Verloes-David-Pfeiffer type	413690	Methotrexate toxicity or dose selection
		2631	Mesomelic dwarfism - cleft palate - camptodactyly	86904	Methotrexate-associated lymphoproliferative disorders
		2632	Mesomelic dwarfism, Langer type		
		2633	Mesomelic dwarfism, Nievergelt type		
		2634	Mesomelic dwarfism, Reinhardt-Pfeiffer type		
		97360	Mesomelic dwarfism-small genitalia syndrome		
		85170	Mesomelic dysplasia with absent fibulas and triangular tibias		

→ 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
1917	Methyl mercury antenatal infection	67046	MGA1	2516	Microcephaly - cardiac defect - lung malsegmentation
622	Methylcobalamin deficiency	111	MGA2	2515	Microcephaly - cardiomyopathy
308380	Methylcobalamin deficiency type cblDv1	67047	MGA3	2522	Microcephaly - cervical spine fusion anomalies
2169	Methylcobalamin deficiency type cblE	67048	MGA4	2521	Microcephaly - cleft palate
2170	Methylcobalamin deficiency type cblG	66634	MGA5	2533	Microcephaly - deafness - intellectual disability
395	Methylene tetrahydrofolate reductase deficiency	445038	MGA7	137653	Microcephaly - digital anomalies - intellectual disability
		→18205 0	MHA	217026	Microcephaly - facio-cardio-skeletal syndrome, Hadziselimovic type
308425	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	443162	MHAC	2172	Microcephaly - glomerulonephritis - marfanoid habitus
308425	Methylmalonic acidemia due to methylmalonyl-CoA racemase deficiency	391417	MHBD deficiency	2065	Microcephaly - hiatus hernia - nephrotic syndrome
26	Methylmalonic acidemia with homocystinuria	391428	MHBD deficiency, classic type	2558	Microcephaly - hypergonadotropic hypogonadism - short stature
79284	Methylmalonic acidemia with homocystinuria type cblF	391428	MHBD deficiency, infantile type	3132	Microcephaly - hypogammaglobulinemia - abnormal immunity
79282	Methylmalonic acidemia with homocystinuria, type cblC	391457	MHBD deficiency, neonatal type	647	Microcephaly - immunodeficiency - lymphoreticuloma
79283	Methylmalonic acidemia with homocystinuria, type cblD	99826	MHF	137658	Microcephaly - intellectual disability - phalangeal and neurological anomalies
369955	Methylmalonic acidemia with homocystinuria, type cblJ	386	MHL	1305	Microcephaly - intellectual disability - tracheoesophageal fistula
369962	Methylmalonic acidemia with homocystinuria, type cblX	79651	mHPA	391641	Microcephaly - intellectual disability - tracheoesophageal fistula type 1
280183	Methylmalonic acidemia, TCb1R type	294016	MIC-CAP syndrome	1229	Microcephaly - intracranial calcification - intellectual disability
280183	Methylmalonic acidemia, TCb1R type	294016	MIC-CM syndrome	2526	Microcephaly - lymphedema - chorioretinopathy
308425	Methylmalonic aciduria due to methylmalonyl-CoA epimerase deficiency	2505	Michelin tire baby syndrome	3434	Microcephaly - microphthalmia - ectrodactyly of lower limbs - prognathism
308425	Methylmalonic aciduria due to methylmalonyl-CoA racemase deficiency	→29384 3	Michels syndrome	1305	Microcephaly - oculo-digito-esophageal-duodenal syndrome
280183	Methylmalonic aciduria due to transcobalamin receptor defect	163937	MICPCH	391641	Microcephaly - oculo-digito-esophageal-duodenal syndrome type 1
26	Methylmalonic aciduria with homocystinuria	2510	Micro syndrome	171703	Microcephaly - polymicrogyria - corpus callosum agenesis
79282	Methylmalonic aciduria with homocystinuria, type cblC	2511	Microbrachycephaly - ptosis - cleft lip	2519	Microcephaly - seizures - intellectual disability - heart disease
79283	Methylmalonic aciduria with homocystinuria, type cblD	2512	Microcephalia vera	240760	Microcephaly and chromosomal instability without immunodeficiency
79284	Methylmalonic aciduria with homocystinuria, type cblF	85172	Microcephalic osteodysplastic dysplasia, Saul-Wilson type	2512	Microcephaly vera
369955	Methylmalonic aciduria with homocystinuria, type cblJ	2637	Microcephalic osteodysplastic primordial dwarfism type II	294016	Microcephaly-capillary malformation syndrome
369962	Methylmalonic aciduria with homocystinuria, type cblX	2636	Microcephalic osteodysplastic primordial dwarfism types I and III	329332	Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome
280183	Methylmalonic aciduria with homocystinuria, type cblI	2636	Microcephalic osteodysplastic primordial dwarfism, Taybi-Linder type	329332	Microcephaly-cerebellar hypoplasia-congenital heart conduction defect syndrome
26	Methylmalonic aciduria with homocystinuria	329228	Microcephalic primordial dwarfism due to ZNF335 deficiency		
79282	Methylmalonic aciduria with homocystinuria, type cblC	319671	Microcephalic primordial dwarfism, Alazami type		
79283	Methylmalonic aciduria with homocystinuria, type cblD	319675	Microcephalic primordial dwarfism, Dauber type		
79284	Methylmalonic aciduria with homocystinuria, type cblF	2643	Microcephalic primordial dwarfism, Toriello type		
369955	Methylmalonic aciduria with homocystinuria, type cblJ	329228	Microcephalic primordial dwarfism, Walsh type		
369962	Methylmalonic aciduria with homocystinuria, type cblX	436182	Microcephalic primordial dwarfism-insulin resistance syndrome		
29	Mevalonic aciduria	2513	Microcephaly - albinism - digital anomalies		
2710	Meyer-Schwickerath syndrome	3433	Microcephaly - brachydactyly - kyphoscoliosis		
443995	MFDA	2523	Microcephaly - brain defect - spasticity - hypernatremia		
79113	MFDM syndrome				
558	MFS				
284963	MFS1				
284973	MFS2				

→ 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
434179	Microcephaly-cerebral malformation-orofaciogigital syndrome	280200	Microform HPE	100084	Middle ear endocrine tumor
423894	Microcephaly-complex motor and sensory axonal neuropathy syndrome	2538	Microgastria - limb reduction defect	93926	Middle interhemispheric fusion variant
2508	Microcephaly-corpus callosum agenesis-abnormal genitalia syndrome	1388	Micrognathia digital syndrome	93926	Middle interhemispheric variant of holoprosencephaly
294016	Microcephaly-cutaneous capillary malformation syndrome	50810	Microlissencephaly - micromelia	141288	Midline cervical cleft
1305	Microcephaly-digital anomalies-normal intelligence syndrome	89844	Microlissencephaly type A	95443	Midline heart
391641	Microcephaly-digital anomalies-normal intelligence syndrome type 1	2641	Micromelic dwarfism, Fryns type	93926	Midline interhemispheric variant of holoprosencephaly
391646	Microcephaly-digital anomalies-normal intelligence syndrome type 2	93329	Micromelic dysplasia - dislocation of radius	2557	Mietens syndrome
217026	Microcephaly-faciocardioskeletal syndrome	85275	Microphthalmia - ankyloblepharon - intellectual disability	2867	Mievis - Verellen-Dumoulin syndrome
391646	Microcephaly-intellectual disability-tracheoesophageal fistula syndrome type 2	98938	Microphthalmia - anophthalmia - coloboma	293181	Migrating partial epilepsy of infancy
2528	Microcephaly-microcornea syndrome, Seemanova type	77299	Microphthalmia - brain atrophy	293181	Migrating partial seizures of infancy
228418	Microcephaly-seizures-developmental delay syndrome	2556	Microphthalmia - dermal aplasia - sclerocornea	504	Migratory myiasis
423306	Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome	→2510	Microphthalmia - mental deficiency	93926	MIH
397951	Microcephaly-thin corpus callosum-intellectual disability syndrome	2547	Microphthalmia - microtia - fetal akinesia	93926	MIH type HPE
2670	Microcoria - congenital nephrosis	2705	Microphthalmia - optic nerve aplasia	93926	MIHF
2535	Microcornea - corectopia - macular hypoplasia	251279	Microphthalmia - retinitis pigmentosa - foveoschisis - optic disc drusen	93926	MIHV
2536	Microcornea - glaucoma - absent frontal sinuses	139471	Microphthalmia with brain and digit anomalies	2558	Mikati-Najjar-Sahli syndrome
231736	Microcornea - posterior megalolenticonus - persistent fetal vasculature - coloboma	98938	Microphthalmia with colobomatous cyst	79078	Mikulicz disease
263347	Microcornea - rod-cone dystrophy - cataract - posterior staphyoma	1104	Microphthalmia with facial clefting	314918	Mild Canavan disease
369970	Microcornea-myopic chorioretinal atrophy-telecanthus syndrome	1106	Microphthalmia with limb anomalies	169799	Mild factor IX deficiency
98956	Microcystic corneal dystrophy	2556	Microphthalmia with linear skin defects syndrome	169808	Mild factor VIII deficiency
79490	Microcystic infiltrating lymphatic malformation	568	Microphthalmia, Lenz type	169808	Mild hemophilia A
79490	Microcystic lymphangioma	424099	Microphthalmia-coloboma-rhizomelic skeletal dysplasia	169799	Mild hemophilia B
79490	Microcystic lymphatic malformation	727	Micropolyangiitis	79651	Mild HPA
83642	Microcytic anemia with liver iron overload	58220	Microscopic colitis	79651	Mild hyperphenylalaninemia
77301	Microdeletion 9q22.3	727	Microscopic polyangiitis	171439	Mild nemaline myopathy
567	Microdeletion 22q11.2	2551	Microscopic polyarteritis	216796	Mild osteogenesis imperfecta
90024	Microdontia - type I microtia - deafness	2552	Microspherophakia - metaphyseal dysplasia	247815	Mild peroxisomal disorder due to PEX10 deficiency
101081	Microduplication 17p12	83463	Microsporidiosis	79253	Mild phenylketonuria
217377	Microduplication Xp11.22-p11.23 syndrome	2306	Microtia	411536	Mild phosphoribosylpyrophosphate synthetase superactivity
280200	Microform holoprosencephaly	139450	Microtia-aortic arch syndrome	79253	Mild PKU
		289522	Microtia-eye coloboma-imperforation of the nasolacrimal duct syndrome	411536	Mild PRPP synthetase superactivity
		2290	Microtripllication 11q24.1	411536	Mild PRPS1 superactivity
		2290	Microvillous inclusion disease	93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis
		166430	Microvillus inclusion disease	246	Miller syndrome
		1456	Micturition-induced seizures	531	Miller-Dieker syndrome
		1456	Mid-aortic dysplastic syndrome	98919	Miller-Fisher syndrome
		1456	Mid-aortic syndrome	94091	Mills syndrome
		228299	Mid-dermal elastolysis	79452	Milroy disease
		1456	Midaortic syndrome	→79452	Milroy-like disease
		2556	MIDAS syndrome	255210	MILS
		225	MIDD	1917	Minamata disease
		1456	Middle aortic syndrome	757	Mineralocorticoid resistant hyperkalemia
		→29384 3		→29384 3	Mingarelli 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
352734	Minimal pigment oculocutaneous albinism type 1	1194	Mitochondrial encephalo-cardio-myopathy due to isolated mitochondrial respiratory chain complex V deficiency	168609	Mitochondrial non-syndromic neurosensory hearing loss with susceptibility to aminoglycoside exposure
98832	Minimally differentiated acute myeloblastic leukemia	1933	Mitochondrial encephalomyopathy - aminoacidopathy	90641	Mitochondrial non-syndromic sensorineural deafness
822	Minkowski-Chauffard disease	238329	Mitochondrial encephalomyopathy due to combined oxidative phosphorylation deficiency 6	168609	Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure
1918	Minoxidil antenatal infection	238329	Mitochondrial encephalomyopathy due to COXP6	168609	Mitochondrial non-syndromic sensorineural hearing loss with susceptibility to aminoglycoside exposure
94125	MIRAS	550	Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes	447784	Mitochondrial pyruvate carrier deficiency
→193	Mirhosseini-Holmes-Walton syndrome	280288	Mitochondrial HSP60 chaperonopathy	254881	Mitochondrial spinocerebellar ataxia with epilepsy
295010	Mirror foot	314637	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	746	Mitochondrial trifunctional protein deficiency
295185	Mirror foot, bilateral	168609	Mitochondrial isolated neurosensory deafness with susceptibility to aminoglycoside exposure	1205	Mitral atresia
295183	Mirror foot, unilateral	168609	Mitochondrial isolated neurosensory hearing loss with susceptibility to aminoglycoside exposure	3238	Mitral regurgitation - deafness - skeletal anomalies
295004	Mirror hand	168609	Mitochondrial isolated sensorineural deafness with susceptibility to aminoglycoside exposure	99062	Mitral valve agenesis
295173	Mirror hand, bilateral	168609	Mitochondrial isolated sensorineural hearing loss with susceptibility to aminoglycoside exposure	→28496	Mitral valve-aorta-skeleton-skin 3 syndrome
295171	Mirror hand, unilateral	168609	Mitochondrial isolated sensorineural hearing loss with susceptibility to aminoglycoside exposure	295012	Mitten hand
2378	Mirror hands and feets - nasal defects	289560	Mitochondrial membrane protein-associated neurodegeneration	90036	Mixed AIHA
3004	Mirror polydactyly - vertebral segmentation - limbs defects	2597	Mitochondrial myopathy - lactic acidosis - deafness	809	Mixed connective tissue disease
293822	MITF-related melanoma and renal cell carcinoma predisposition syndrome	2597	Mitochondrial myopathy - lactic acidosis - hearing loss	91138	Mixed cryoglobulinemia
134	Mitochondrial acetoacetyl-coenzyme A thiolase deficiency	2598	Mitochondrial myopathy and sideroblastic anemia	93555	Mixed cryoglobulinemia type III
353217	Mitochondrial aspartate-glutamate carrier 1 deficiency	254864	Mitochondrial myopathy with reversible complex IV deficiency	180234	Mixed germ cell tumor
225	Mitochondrial diabetes	254864	Mitochondrial myopathy with reversible COX deficiency	252021	Mixed germ cell tumor of central nervous system
352470	Mitochondrial DNA deletion syndrome with limb-girdle weakness	254864	Mitochondrial myopathy with reversible cytochrome C oxidase deficiency	252021	Mixed germ cell tumor of CNS
352470	Mitochondrial DNA deletion syndrome with progressive myopathy	550	Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes	213610	Mixed Müllerian cancer of corpus uteri
1933	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	298	Mitochondrial neurogastrointestinal encephalomyopathy	251656	Mixed oligoastrocytoma
255235	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy	90641	Mitochondrial non-syndromic neurosensory deafness	2785	Mixed renal tubular acidosis
369897	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	168609	Mitochondrial non-syndromic neurosensory deafness with susceptibility to aminoglycoside exposure	2785	Mixed RTA
279934	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency			1879	Mixed sclerosing bone dystrophy
363534	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form			324364	Mixed sclerosing bone dystrophy with extra-skeletal manifestations
254875	Mitochondrial DNA depletion syndrome, myopathic form			90036	Mixed-type autoimmune hemolytic anemia
352447	Mitochondrial DNA maintenance syndrome due to MGME1 deficiency			399096	Miyoshi muscular dystrophy type 3
1194	Mitochondrial encephalo-cardio-myopathy due to F1Fo ATPase deficiency			45448	Miyoshi myopathy
1194	Mitochondrial encephalo-cardio-myopathy due to isolated ATP synthase deficiency			98757	MJD
				565	MK
				423461	ML 3 alpha/beta
				423470	ML 3 gamma
				423461	ML III alpha/beta
				423470	ML III gamma
				2598	MLASA
				2478	MLC

→ 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
2526	MLCRD	1420	Moerman-Vandenberghen-Fryns syndrome	251019	Monosomy 2q32q33
512	MLD	3198	Moersch-Woltman syndrome	251028	Monosomy 2q33.1
309271	MLD, adult form	2549	Moeschler-Clarren syndrome	1001	Monosomy 2q37-qter
309263	MLD, juvenile form	2751	Mohr syndrome	435638	Monosomy 3p25.3
309256	MLD, late infantile form	52368	Mohr-Tranebjærg syndrome	1620	Monosomy 3pter
59306	MLS	99927	Molar pregnancy	1621	Monosomy 3q13
2556	MLS syndrome	2650	Mollica-Pavone-Antener syndrome	356947	Monosomy 3q26-q27
369970	MMCAT syndrome	1433	Moloney syndrome	356947	Monosomy 3q26q27
598	MmD	397973	MOMES syndrome	65286	Monosomy 3q29
399096	MMD3	2563	MOMO syndrome	65286	Monosomy 3qter
3434	MMEP syndrome	371428	MONA spectrum	238750	Monosomy 4q21
592	MMF	573	Monilethrix	96145	Monosomy 4qter
268249	MMF embryopathy	573	Moniliform hair syndrome	281	Monosomy 5p
2241	MMIHS	319254	Monkey disease	228384	Monosomy 5q14.3
641	MMN	319254	Monkey fever	314655	Monosomy 5q31.3
641	MMNCB	3057	Monoamine oxidase A deficiency	1627	Monosomy 5q35
137867	MMND	59	Monocarboxylate transporter 8 deficiency	251046	Monosomy 6p22
293181	MMPEI	91136	Monoclonal Ig light chain-associated Fanconi syndrome	96125	Monosomy 6p25
293181	MMPSI	91136	Monoclonal kappa Ig light chain-associated Fanconi syndrome	171829	Monosomy 6q16
2479	MMR syndrome	228423	Monocyte - B - natural killer - dendritic cell deficiency	251056	Monosomy 6q25
1305	MMT	228423	Monocytopenia and mycobacterial infection syndrome	96126	Monosomy 7pter
391641	MMT type 1	228423	Monocytopenia with susceptibility to infections	904	Monosomy 7q11.23
391646	MMT type 2	99885	Monogenic diabetes of infancy	251061	Monosomy 7q31
298	MNGIE	228423	MonoMAC	1636	Monosomy 7qter
565	MNK	65684	Monomelic amyotrophy	251066	Monosomy 8p11.2
251656	MOA	86870	Monomorphic NK-cell lymphoma	251071	Monosomy 8p23.1
77299	MOBA syndrome	2565	Mononen-Karnes-Senac syndrome	2496	Monosomy 8q13
570	Möbius syndrome	2901	Mononeuritis multiplex with brachial predilection	284160	Monosomy 8q21.11
2560	Möbius syndrome - axonal neuropathy - hypogonadotropic hypogonadism	293948	Monosomy 1p21.3	178303	Monosomy 8q22.1
99732	MOCOD	401986	Monosomy 1p31p32	502	Monosomy 8q24.1
308386	MOCOD type A	1606	Monosomy 1p36	261112	Monosomy 9p
308393	MOCOD type B	1606	Monosomy 1pter	324313	Monosomy 9p13
308400	MOCOD type C	250989	Monosomy 1q21.1	1642	Monosomy 9pter
1305	MODED syndrome	250999	Monosomy 1q41-q42	77301	Monosomy 9q22.3
391641	MODED syndrome type 1	250999	Monosomy 1q41q42	401923	Monosomy 9q31.1q31.3
90056	Moderate and severe traumatic brain injury	238769	Monosomy 1q44	284169	Monosomy 10p11.21p12.31
178145	Moderate multiminicore disease with hand involvement	36367	Monosomy 1qter	1580	Monosomy 10pter
169796	Moderately severe factor IX deficiency	261349	Monosomy 2p15-p16.1	276413	Monosomy 10q22.3q23.3
169805	Moderately severe factor VIII deficiency	261349	Monosomy 2p15p16.1	96148	Monosomy 10qter
169805	Moderately severe hemophilia A	163693	Monosomy 2p21	893	Monosomy 11p13
169796	Moderately severe hemophilia B	228402	Monosomy 2q23.1	444002	Monosomy 11q22.2-q22.3
263335	Moderately-differentiated thymic neuroendocrine carcinoma	1617	Monosomy 2q24	444002	Monosomy 11q22.2q22.3
552	MODY	251014	Monosomy 2q31.1	2308	Monosomy 11qter
93111	MODY5	251019	Monosomy 2q32	313884	Monosomy 12p12.1
570	Moebius syndrome	251019	Monosomy 2q32-q33	94063	Monosomy 12q14
				289513	Monosomy 12q15q21.1
				96149	Monosomy 12qter
				412035	Monosomy 13q12.3
				1587	Monosomy 13q14
				1590	Monosomy 13q32
				96168	Monosomy 13q34
				261120	Monosomy 14q11.2

→ 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
261144	Monosomy 14q12	309297	Morquio disease type A	401945	Moyamoya disease with early-onset achalasia
→3157	Monosomy 14q22	309310	Morquio disease type B	280679	Moyamoya disease-short stature-facial dysmorphism-hypergonadotropic hypogonadism
264200	Monosomy 14q22-q23	2570	Morse-Rawnsley-Sargent syndrome	2574	Moynahan syndrome
264200	Monosomy 14q22q23	83467	Morvan syndrome	352734	MP OCA type 1
401935	Monosomy 14q24.1q24.3	83467	Morvan's fibrillary chorea	727	MPA
261183	Monosomy 15q11.2	329813	Mosaic genome-wide paternal uniparental disomy	289560	MPAN
199318	Monosomy 15q13.3	329813	Mosaic genome-wide paternal UPD	59135	MPD1
261190	Monosomy 15q14	99228	Mosaic monosomy X	399086	MPD3
94065	Monosomy 15q24	96193	Mosaic paternal uniparental disomy of chromosome 11	79323	MPDU1-CDG
1596	Monosomy 15q26	1692	Mosaic trisomy 1	293181	MPEI
261211	Monosomy 16p11.2-p12.2	1723	Mosaic trisomy 2	54370	MPGN
261211	Monosomy 16p11.2p12.2	100071	Mosaic trisomy 3	79319	MPI-CDG
261236	Monosomy 16p13.11	96059	Mosaic trisomy 4	79253	mPKU
352629	Monosomy 16q24.1	96060	Mosaic trisomy 5	3148	MPNST
261250	Monosomy 16q24.3	1747	Mosaic trisomy 7	252212	MPNST with rhabdomyosarcomatous differentiation
531	Monosomy 17p13.3	96061	Mosaic trisomy 8	2587	MPO deficiency
97685	Monosomy 17q11	99776	Mosaic trisomy 9	231736	MPPC syndrome
261265	Monosomy 17q12	96063	Mosaic trisomy 10	83473	MPPH syndrome
363958	Monosomy 17q21.31	1698	Mosaic trisomy 12	579	MPS1
261279	Monosomy 17q23.1-q23.2	1703	Mosaic trisomy 14	93473	MPS1H
261279	Monosomy 17q23.1q23.2	1706	Mosaic trisomy 15	93476	MPS1H/S
1597	Monosomy 17qter	1708	Mosaic trisomy 16	93474	MPS1S
1598	Monosomy 18p	1711	Mosaic trisomy 17	580	MPS2
1600	Monosomy 18q	1724	Mosaic trisomy 20	217085	MPS2A
254346	Monosomy 19p13.12	96068	Mosaic trisomy 22	217093	MPS2B
357001	Monosomy 19p13.13	1052	Mosaic variegated aneuploidy syndrome	581	MPS3
217346	Monosomy 19q13.11	54057	Moschcowitz disease	79269	MPS3A
261295	Monosomy 20p12.3	2717	MOTA syndrome	79270	MPS3B
313781	Monosomy 20p13	254516	Motor developmental delay due to 14q32.2 paternally expressed gene defect	79271	MPS3C
444051	Monosomy 20q11	3347	Mounier-Kühn syndrome	79272	MPS3D
261311	Monosomy 20q13.33	83595	Mountain fever	582	MPS4
96152	Monosomy 20qter	83595	Mountain tick fever	309297	MPS4A
574	Monosomy 21	2572	Mousa-Al Din-Al Nassar syndrome	309310	MPS4B
261323	Monosomy 21q22.11-q22.12	324972	Mouth and genital ulcers with inflamed cartilage	583	MPS6
261323	Monosomy 21q22.11q22.12	2152	Mowat-Wilson syndrome	276212	MPS6, rapidly progressing
268261	Monosomy 21q22.13-q22.2	261537	Mowat-Wilson syndrome due to 2q22 microdeletion	276223	MPS6, slowly progressing
268261	Monosomy 21q22.13q22.2	261552	Mowat-Wilson syndrome due to a ZEB2 point mutation	584	MPS7
96123	Monosomy 22	261537	Mowat-Wilson syndrome due to del(2)q(22)	67041	MPS9
567	Monosomy 22q11	261537	Mowat-Wilson syndrome due to monosomy 2q22	579	MPSI
48652	Monosomy 22q13	280679	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	293181	MPSI
99226	Monosomy X	2573	Moyamoya disease	93473	MPSIH
261476	Monosomy Xp21			93476	MPSIH/S
93277	Monostotic fibrous dysplasia			580	MPSII
158003	Montgomery syndrome			217085	MPSIIA
→969	Moore-Federman syndrome			217093	MPSIIB
2637	MOPD type II			581	MPSIII
2636	MOPD types I and III			79269	MPSIIIA
52056	Morava-Mehes syndrome			79270	MPSIIIB
77296	Morgagni-Stewart-Morel syndrome			79271	MPSIIIC
75858	MORM syndrome				
35737	Morning glory syndrome				
582	Morquio disease				

→ 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
79272	MPSIID	319595	MSMD due to partial signal transducer and activator of transcription 1 deficiency	93474	Mucopolysaccharidosis type 1S
93474	MPSIS	319595	MSMD due to partial STAT1 deficiency	580	Mucopolysaccharidosis type 2
582	MPSIV	157801	MSSD	217093	Mucopolysaccharidosis type 2, attenuated form
309297	MPSIVA	65748	MSSE	217085	Mucopolysaccharidosis type 2, severe form
309310	MPSIVB	511	MSUD	217085	Mucopolysaccharidosis type 2A
67041	MPSIX	2505	MTBS	217093	Mucopolysaccharidosis type 2B
583	MPSVI	1332	MTC	581	Mucopolysaccharidosis type 3
276212	MPSVI, rapidly progressing	352470	mtDNA deletion syndrome with limb-girdle weakness	79269	Mucopolysaccharidosis type 3A
276223	MPSVI, slowly progressing	352470	mtDNA deletion syndrome with progressive myopathy	79270	Mucopolysaccharidosis type 3B
584	MPSVII	1933	mtDNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	79271	Mucopolysaccharidosis type 3C
99967	MRCLS	255235	mtDNA depletion syndrome, encephalomyopathic form with renal tubulopathy	79272	Mucopolysaccharidosis type 3D
263347	MRCS syndrome	369897	mtDNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	582	Mucopolysaccharidosis type 4
67045	MRGH	363534	mtDNA depletion syndrome, hepatocerebrorenal form	309297	Mucopolysaccharidosis type 4A
3109	MRKH syndrome	254875	mtDNA depletion syndrome, myopathic form	309310	Mucopolysaccharidosis type 4B
247775	MRKH syndrome type 1	352447	mtDNA maintenance syndrome due to MGME1 deficiency	583	Mucopolysaccharidosis type 6
2578	MRKH syndrome type 2	395	MTHFR deficiency	276212	Mucopolysaccharidosis type 6, rapidly progressing
3059	MRX35	252212	MTT	276223	Mucopolysaccharidosis type 6, slowly progressing
85274	MRXS7	100024	mu-HCD	584	Mucopolysaccharidosis type 7
85324	MRXS9	100024	Mu-heavy chain disease	67041	Mucopolysaccharidosis type 9
93952	MRXSH	398961	Mucinous adenocarcinoma of ovary	579	Mucopolysaccharidosis type I
2598	MSA	391723	Mucinous adenocarcinoma of the appendix	93473	Mucopolysaccharidosis type IH
102	MSA	424053	Mucinous cystadenocarcinoma of pancreas	93476	Mucopolysaccharidosis type IH/S
227510	MSA, cerebellar type	319322	Mucinous tubular and spindle cell carcinoma	580	Mucopolysaccharidosis type II
98933	MSA, parkinsonian type	575	Muckle-Wells syndrome	217093	Mucopolysaccharidosis type II, attenuated form
227510	MSA-c	2331	Mucocutaneous lymph node syndrome	217085	Mucopolysaccharidosis type II, severe form
98933	MSA-p	2451	Mucocutaneous venous malformations	217085	Mucopolysaccharidosis type IIA
1879	MSBD syndrome	423461	Mucolipidosis type 3 alpha/beta	217093	Mucopolysaccharidosis type IIB
254881	MSCAE	423470	Mucolipidosis type 3 gamma	581	Mucopolysaccharidosis type III
585	MSD	576	Mucolipidosis type II	79269	Mucopolysaccharidosis type IIIA
2619	Mseleni joint disease	577	Mucolipidosis type III	79270	Mucopolysaccharidosis type IIIB
1309	MSK	423461	Mucolipidosis type III alpha/beta	79271	Mucopolysaccharidosis type IIIC
99898	MSMD due to complete IFNgammaR1 deficiency	423470	Mucolipidosis type III gamma	79272	Mucopolysaccharidosis type IIID
319547	MSMD due to complete IFNgammaR2 deficiency	576	Mucolipidosis type II	93474	Mucopolysaccharidosis type IS
319558	MSMD due to complete IL12B deficiency	577	Mucolipidosis type III	582	Mucopolysaccharidosis type IV
319552	MSMD due to complete IL12RB1 deficiency	423461	Mucolipidosis type III alpha/beta	309297	Mucopolysaccharidosis type IVA
99898	MSMD due to complete interferon gamma receptor 1 deficiency	423470	Mucolipidosis type III gamma	309310	Mucopolysaccharidosis type IVB
319547	MSMD due to complete interferon gamma receptor 2 deficiency	576	Mucolipidosis type II	67041	Mucopolysaccharidosis type IX
319552	MSMD due to complete interleukin 12 receptor beta 1 deficiency	577	Mucolipidosis type III	583	Mucopolysaccharidosis type VI
319558	MSMD due to complete interleukin 12B deficiency	423461	Mucolipidosis type III alpha/beta	276212	Mucopolysaccharidosis type VI, rapidly progressing
319563	MSMD due to complete ISG15 deficiency	423470	Mucolipidosis type III gamma	276223	Mucopolysaccharidosis type VI, slowly progressing
319600	MSMD due to partial interferon regulatory factor 8 deficiency	578	Mucolipidosis type IV	584	Mucopolysaccharidosis type VII
319600	MSMD due to partial IRF8 deficiency	579	Mucopolysaccharidosis type 1	73263	Mucormycosis
		93473	Mucopolysaccharidosis type 1H	52417	Mucosa-associated lymphatic tissue lymphoma
		93476	Mucopolysaccharidosis type 1H/S	52417	Mucosa-associated lymphoid tissue lymphoma

→ 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
46486	Mucosal pemphigoid	319287	Multilocular cystic renal cell adenocarcinoma	166024	Multiple epiphyseal dysplasia, Al-Gazali type
585	Mucosulfatidosis	319287	Multilocular cystic renal cell carcinoma	166011	Multiple epiphyseal dysplasia, Beighton type
46486	Mucosynechial pemphigoid	168816	Multilocular peritoneal inclusion cyst	166016	Multiple epiphyseal dysplasia, Lowry type
46486	Mucus membrane pemphigoid	97366	Multilocular renal cyst	166032	Multiple epiphyseal dysplasia, with miniepipyses
586	Mucoviscidosis	97366	Multiloculated renal cyst	166029	Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia
53271	Muenke syndrome	598	Multiminicore disease	50920	Multiple fibroadenoma of the breast
444	MUHH	598	Multiminicore myopathy	83454	Multiple glomus tumors
587	Muir-Torre syndrome	2091	Multinodular goiter - cystic kidney - polydactyly	201	Multiple hamartoma syndrome
2576	MULIBREY dwarfism	26791	Multiple acyl-CoA dehydrogenase deficiency	2300	Multiple intestinal atresia
2576	MULIBREY nanism	394532	Multiple acyl-CoA dehydrogenation deficiency, mild type	284139	Multiple joint dislocations - short stature - craniofacial dysmorphism - congenital heart defects
247768	Müllerian aplasia and hyperandrogenism	394529	Multiple acyl-CoA dehydrogenation deficiency, severe neonatal type	294049	multiple joint dislocations-short stature-hyperlaxity-craniofacial dysmorphism syndrome
1655	Müllerian derivatives - lymphangiectasia - polydactyly	2505	Multiple benign circumferential skin creases on limbs	493	Multiple keratoacanthoma
2491	Müllerian duct anomalies - limb anomalies	2678	Multiple café-au-lait spots	65748	Multiple keratoacanthoma, Ferguson-Smith type
2578	Müllerian duct aplasia-renal dysplasia-cervical somite anomalies syndrome	2678	Multiple café-au-lait syndrome	587	Multiple keratoacanthoma, Muir-Torre type
247768	Müllerian duct failure and hyperandrogenism	321	Multiple cartilaginous exostoses	79455	Multiple mastocytoma
2774	Multicentric carpo-tarsal osteolysis with or without nephropathy	280633	Multiple congenital anomalies - hypotonia - seizures syndrome	29073	Multiple myeloma
93686	Multicentric Castleman disease	254519	Multiple congenital anomalies due to 14q32.2 maternally expressed gene defect	2029	Multiple non-ossifying fibromatosis
93686	Multicentric giant lymph node hyperplasia	300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	435329	Multiple ossifying fibroma
85196	Multicentric osteolysis - nodulosis - arthropathy	1486	Multiple contracture syndrome, Finnish type	321	Multiple osteochondromas
371428	Multicentric osteolysis-nodulosis-arthropathy spectrum	137776	Multiple contracture syndrome, Israeli-Bedouin type	324299	Multiple paragangliomas associated with erythrocytosis
139436	Multicentric reticulohistiocytosis	523	Multiple cutaneous and uterine leiomyomas	324299	Multiple paragangliomas associated with polycythemia
1851	Multicystic dysplastic kidney	3453	Multiple endocrine deficiency - Addison's disease - candidiasis	95494	Multiple pituitary hormone deficiencies, genetic forms
168816	Multicystic mesothelioma	652	Multiple endocrine neoplasia type 1	→1234	Multiple pterygium syndrome, Aslan type
1851	Multicystic renal dysplasia	653	Multiple endocrine neoplasia type 2	3151	Multiple sclerosis - ichthyosis - factor VIII deficiency
48162	Multifocal acquired demyelinating sensory and motor neuropathy	247698	Multiple endocrine neoplasia type 2A	65748	Multiple self-healing squamous epithelioma
3282	Multifocal atrial tachycardia	247709	Multiple endocrine neoplasia type 2B	585	Multiple sulfatase deficiency
99873	Multifocal eosinophilic granuloma	247709	Multiple endocrine neoplasia type 3	2398	Multiple symmetric lipomatosis
641	Multifocal motor neuropathy	276152	Multiple endocrine neoplasia type 4	3237	Multiple synostoses syndrome
641	Multifocal motor neuropathy with conduction block	166024	Multiple epiphyseal dysplasia - macrocephaly - distinctive facies	102	Multiple system atrophy
2033	Multifocal muscular fibrosis - obstructed vessels	166011	Multiple epiphyseal dysplasia - myopia - deafness	227510	Multiple system atrophy, cerebellar type
99003	Multifocal pattern dystrophy simulating fundus flavimaculatus	166002	Multiple epiphyseal dysplasia due to collagen 9 anomaly	98933	Multiple system atrophy, parkinsonian type
3286	Multifocal ventricular premature beats	93308	Multiple epiphyseal dysplasia type 1	99096	Multiple ventricular septal defects
319287	Multilocular clear cell adenocarcinoma	93307	Multiple epiphyseal dysplasia type 4	102	Multisystem atrophy
319287	Multilocular clear cell carcinoma	93311	Multiple epiphyseal dysplasia type 5	404463	Multisystemic smooth muscle dysfunction syndrome
319287	Multilocular clear cell renal cell adenocarcinoma	166016	Multiple epiphyseal dysplasia with Robin phenotype	2959	Mulvihill-Smith syndrome
97366	Multilocular cyst of the kidney				

→ 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
2578	MURCS association	2585	Myelocerebellar disorder	163696	Myoclonus-nephropathy syndrome
83315	Murine typhus	268813	Myelocystocele	178464	Myofibrillar myopathy with early respiratory failure
2028	Murray-Puretic-Drescher syndrome	86841	Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality	104077	Myopathic intestinal pseudoobstruction
99849	Muscle enolase deficiency	824	Myelofibrosis with myeloid metaplasia	2601	Myopathy - growth delay - intellectual disability - hypospadias
171445	Muscle filaminopathy	168953	Myeloid neoplasm associated with FGFR1 rearrangement	1358	Myopathy - Moebius - Robin syndrome
97234	Muscle phosphoglycerate mutase deficiency	168947	Myeloid neoplasm associated with PDGFRA rearrangement	2596	Myopathy and diabetes mellitus
588	Muscle-eye-brain disease	168950	Myeloid neoplasm associated with PDGFRB rearrangement	88635	Myopathy due to calsequestrin and SERCA1 protein overload
370997	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	86850	Myeloid sarcoma	97234	Myopathy due to phosphoglycerate mutase deficiency
588	Muscle-eye-brain syndrome	91136	Myeloma-associated Fanconi syndrome	43115	Myopathy with exercise intolerance, Swedish type
2576	Muscle-liver-brain-eye nanism	29073	Myelomatosis	171889	Myopathy with hexagonally cross-linked tubular arrays
2579	Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus	93969	Myelomeningocele	2598	Myopathy, lactic acidosis and sideroblastic anemia
1877	Muscular dystrophy - white matter spongiosis	2587	Myeloperoxidase deficiency	289685	Myopericytoma
424261	Muscular dystrophy with progressive weakness, distal contractures and rigid spine	824	Myelosclerosis with myeloid metaplasia	368	Myophosphorylase deficiency
199340	Muscular dystrophy, Selcen type	437572	MYH7-related late-onset scapuloperoneal muscular dystrophy	178493	Myopic macular degeneration
99849	Muscular enolase deficiency	437572	MYH7-related late-onset scapuloperoneal syndrome	178493	Myopic maculopathy
324416	Muscular hypertrophy - hepatomegaly - polyhydramnios	437572	MYH7-related late-onset SPMD	289380	Myosclerosis
2349	Muscular pseudohypertrophy - hypothyroidism	182050	MYH9-RD	337	Myositis ossificans progressiva
3079	Mutchnick syndrome	182050	MYH9-related disease	764	Myositis purulenta tropica
494	Mutilating keratoderma of Vohwinkel	182050	MYH9-related disorder	764	Myositis tropicans
494	Mutilating keratoderma plus deafness	182050	MYH9-related syndrome	306553	Myospherulosis
	Mutilating palmoplantar hyperkeratosis with periorificial keratotic plaques	182050	MYH9-related syndromic thrombocytopenia	275534	Myostatin-related muscle hypertrophy
659	Mutilating palmoplantar keratoderma with periorificial keratotic plaques	2588	Myhre syndrome	3101	Myotonia - intellectual disability - skeletal anomalies
247798	MUTYH-related AFAP	109	Myhre-Riley-Smith syndrome	99736	Myotonia - painful contractions
247798	MUTYH-related attenuated familial adenomatous polyposis	45	Myoadenylate deaminase deficiency	614	Myotonia congenita
247798	MUTYH-related attenuated familial polyposis coli	1942	Myoclonic atonic epilepsy	99734	Myotonia fluctuans
247798	MUTYH-related attenuated FAP	36899	Myoclonic dystonia	99735	Myotonia permanens
29	MVA	→36899	Myoclonic dystonia 15	800	Myotonic chondrodystrophy
2290	MVID	86913	Myoclonic epilepsy in non-progressive encephalopathies	273	Myotonic dystrophy type 1
2582	Myalgia-eosinophilia syndrome associated with tryptophan	86909	Myoclonic epilepsy of infancy	606	Myotonic dystrophy type 2
589	Myasthenia gravis	1942	Myoclonic-astasic epilepsy	→52430	Myotonic dystrophy type 3
2583	Mycetoma	1942	Myoclonic-astatic epilepsy in early childhood	800	Myotonic myopathy, dwarfism, chondrodystrophy, ocular and facial anomalies
314946	Mycobacterium xenopi infection	435438	Myoclonus epilepsy and ataxia due to potassium channel mutation	596	Myotubular myopathy
268249	Mycophenolate mofetil embryopathy	551	Myoclonus epilepsy associated with ragged-red fibres	79105	Myxofibrosarcoma
83482	Mycoplasma encephalitis	86913	Myoclonus epilepsy in non-progressive encephalopathies	79105	Myxoid malignant fibrous histiocytoma
2584	Mycosis fungoides, Alibert-Bazin type	2589	Myoclonus-cerebellar ataxia-deafness syndrome	99967	Myxoid/round cell liposarcoma
178512	Mycosis fungoides-associated follicular mucinosis	36899	Myoclonus-dystonia syndrome	1359	Myxoma - spotty pigmentation - endocrine overactivity
183713	MyD88 deficiency	→36899	Myoclonus-dystonia type 15	57782	Myxoma with fibrous dysplasia
59298	Myelinoclastic diffuse sclerosis			251643	Myxopapillary ependymoma
135	Myelinosis centralis diffusa			2608	N 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
79270	N-acetyl-alpha-glucosaminidase deficiency	35612	Nanophthalmia	217560	NEHI
583	N-acetylgalactosamine 4-sulfatase deficiency	85196	NAO syndrome	199244	Nelson syndrome
309297	N-acetylgalactosamine-6-sulfate sulfatase deficiency	247868	NAPS12	217563	Neonatal acute respiratory distress due to SP-B deficiency
576	N-acetylglucosamine 1-phosphotransferase deficiency	83465	Narcolepsy without cataplexy	217563	Neonatal acute respiratory distress due to surfactant protein B deficiency
79329	N-acetylglucosaminyltransferase 2 deficiency	2073	Narcolepsy-cataplexy	44	Neonatal adrenoleukodystrophy
137754	N-acyl-L-amino acid amidohydrolase deficiency	644	NARP syndrome	398109	Neonatal AHA
103908	Na-H exchange deficiency	141103	Nasal dermoid cyst	398109	Neonatal AIHA
178303	Nablus mask-like facial syndrome	141103	Nasal dermoid sinus cyst	398097	Neonatal antiphospholipid antibody syndrome
139373	NADH-cytochrome b5reductase deficiency type 1	141219	Nasal dorsum fistula/cyst	398097	Neonatal antiphospholipid syndrome
139380	NADH-cytochrome b5reductase deficiency type 2	141118	Nasal encephalocele	398109	Neonatal autoimmune hemolytic anemia
139373	NADH-diaphorase deficiency type 1	141115	Nasal ganglioglioma	137929	Neonatal brainstem dysfunction
139380	NADH-diaphorase deficiency type 2	141112	Nasal glial heterotopia	314911	Neonatal Canavan disease
439196	NAE	141112	Nasal glioma	313906	Neonatal congenital pancreatic cyst
69087	Naegeli syndrome	86879	Nasal T/natural killer-cell lymphoma	398117	Neonatal dermatomyositis
69087	Naegeli-Franceschetti-Jadassohn syndrome	2662	Nasodigitoacoustic syndrome	79118	Neonatal diabetes - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys
840	Naevus syringocystadenomatous papilliferus	141083	Nasolacrimal duct cyst	398117	Neonatal DM
245	NAFD	141083	Nasolacrimal mucocele	289857	Neonatal glycine encephalopathy
3137	NAGA deficiency	2399	Nasopalpebral lipoma - coloboma - telecanthus	446	Neonatal hemochromatosis
79279	NAGA deficiency type 1	150	Nasopharyngeal carcinoma	398097	Neonatal Hughes syndrome
79280	NAGA deficiency type 2	141107	Nasopharyngeal teratoma	137577	Neonatal hypoxic and ischemic brain injury
79281	NAGA deficiency type 3	2770	Nasu-Hakola disease	294023	Neonatal inflammatory skin and bowel disease
245	Nager acrofacial dysostosis	1654	Natal teeth - intestinal pseudoobstruction - patent ductus	247598	Neonatal intrahepatic cholestasis caused by citrin deficiency
245	Nager syndrome	2663	Nathalie syndrome	247598	Neonatal intrahepatic cholestasis due to citrin deficiency
927	NAGS deficiency	168572	Native American myopathy	238688	Neonatal iodine exposure
2211	Naguib-Richieri-Costa syndrome	69739	Navajo brainstem syndrome	398124	Neonatal lupus erythematosus
423454	Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome	255229	Navajo neurohepatopathy	284979	Neonatal Marfan syndrome
→1487	Nail dysplasia - camptodactyly - brachydactyly type B	255229	Navajo neuropathy	69063	Neonatal membranous glomerulopathy with maternal NEP deficiency
2614	Nail-patella syndrome	34217	Naxos disease	69063	Neonatal membranous glomerulopathy with maternal neutral endopeptidase deficiency
2613	Nail-patella-like renal disease	377	NBCCS	284979	Neonatal MFS
158676	Nails-only DEB	157850	NBIA1	79242	Neonatal multiple carboxylase deficiency
853	NAIT	216873	NBIA1, atypical form	391504	Neonatal myasthenia gravis
101	Naito-Oyanagi disease	216866	NBIA1, classic form	→42738	Neonatal neutropenia
2229	Najjar syndrome	289560	NBIA4	289857	Neonatal NKH
1063	Nakagawa angioblastoma	329284	NBIA5	289857	Neonatal non-ketotic hyperglycinemia
2615	Nakajo-Nishimura syndrome	397725	NBIA6	56304	Neonatal osseous dysplasia type 1
2822	Nakamura-Osame syndrome	289560	NBIA due to C19orf12 mutation	3455	Neonatal progeroid syndrome
44	NALD	647	NBS	70587	Neonatal respiratory distress syndrome
206569	NAM	240760	NBS-like disorder		
→1359	NAME syndrome	240760	NBSLD		
383	Nance deafness	217560	NCHI		
627	Nance-Horan syndrome	1947	NCL, Northern epilepsy variant		
		2481	NCM		
		75327	NCMD		
		300337	NCRNA disease		
		443162	NDE1-related microhydranencephaly		
		399103	Nebulin-related early-onset distal myopathy		
		158011	Necrobiotic xanthogranuloma		
		439196	Necrolytic acral erythema		
		391673	Necrotizing enterocolitis		
		440368	Necrotizing soft tissue infection		

→ 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
398127	Neonatal scleroderma	635	Neuroblastoma	100073	Neurogenic cervical rib syndrome
417	Neonatal severe primary hyperparathyroidism	2481	Neurocutaneous melanocytosis	100073	Neurogenic costoclavicular syndrome
1451	Neonatal-onset multisystem inflammatory disease	2481	Neurocutaneous melanosis	178029	Neurogenic diabetes insipidus
314950	Neoplastic hypereosinophilic syndrome	35664	Neurocutaneous syndrome, Bicknell type	644	Neurogenic muscle weakness - ataxia - retinitis pigmentosa
94058	Neovascular glaucoma	88639	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	3148	Neurogenic sarcoma
654	Nephroblastoma	289560	Neurodegeneration with brain iron accumulation due to C19orf12 mutation	431255	Neurogenic scapuloperoneal amyotrophy, New England type
2849	Nephroblastomatosis - fetal ascites - macrosomia - Wilms tumor	397725	Neurodegeneration with brain iron accumulation due to COASY mutation	85146	Neurogenic scapuloperoneal syndrome, Kaeser type
223	Nephrogenic diabetes insipidus	157850	Neurodegeneration with brain iron accumulation type 1	100073	Neurogenic thoracic outlet compression syndrome
3145	Nephrogenic diabetes insipidus - intracranial calcification	216873	Neurodegeneration with brain iron accumulation type 1, atypical form	100073	Neurogenic thoracic outlet syndrome
137617	Nephrogenic fibrosing dermopathy	216866	Neurodegeneration with brain iron accumulation type 1, classic form	100073	Neurogenic TOS
93606	Nephrogenic syndrome of inappropriate antidiuresis	289560	Neurodegeneration with brain iron accumulation type 4	94093	Neuroleptic malignant syndrome
137617	Nephrogenic systemic fibrosis	329284	Neurodegeneration with brain iron accumulation type 5	36397	Neurolipomatosis
93622	Nephrolithiasis type 1	217382	Neurodegenerative syndrome due to cerebral folate transport deficiency	163746	Neurologic Waardenburg-Shah syndrome
93623	Nephrolithiasis type 2	3474	Neuroectodermal dysplasia, CHIME type	137754	Neurological conditions associated with aminoacylase 1 deficiency
655	Nephronophthisis	33445	Neuroectodermal melanolysosomal disease	206586	Neurolymphomatosis
3156	Nephronophthisis with retinal dystrophy	3474	Neuroectodermal syndrome, Zunich type	71211	Neuromyelitis optica
84081	Nephronophthisis-hepatic fibrosis syndrome	2676	Neuroectodermal-endocrine syndrome	1947	Neuronal ceroid lipofuscinosis, Northern epilepsy variant
411629	Nephropathic infantile cystinosis	217560	Neuroendocrine cell hyperplasia of infancy	99811	Neuronal intestinal pseudoobstruction
2668	Nephropathy-deafness-hyperparathyroidism syndrome	2677	Neuroepithelioma	2289	Neuronal intranuclear inclusion disease
2669	Nephrosis - deafness - urinary tract - digital malformations	2673	Neurofaciodigitorenal syndrome	644	Neuropathy - ataxia - retinitis pigmentosa
2065	Nephrosis - neuronal dysmigration syndrome	157846	Neuroferritinopathy	639	Neuropathy associated with monoclonal IgM antibodies to myelin-associated glycoprotein
300333	Nephrotic syndrome-deafness-pretilbial epidermolysis bullosa syndrome	252183	Neurofibroma	139512	Neuropathy with hearing impairment
300333	Nephrotic syndrome-hearing loss-pretilbial epidermolysis bullosa syndrome	137605	Neurofibromatosis 1-like syndrome	217622	Neurosensory deafness with dilated cardiomyopathy
2337	NEPPK	636	Neurofibromatosis type 1	217622	Neurosensory hearing loss with dilated cardiomyopathy
280576	Nestor-Guillermo progeria syndrome	363700	Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion	137596	Neutrophic keratitis
634	Netherton syndrome	97685	Neurofibromatosis type 1 microdeletion syndrome	137596	Neutrophic keratopathy
2671	Neu-Laxova syndrome	638	Neurofibromatosis type 1-Noonan syndrome	98907	Neutral lipid storage disease with ichthyosis
99078	Neuhauser anomaly	637	Neurofibromatosis type 2	98908	Neutral lipid storage disease with myopathy without ichthyosis
2479	Neuhäuser syndrome	93921	Neurofibromatosis type 3	98908	Neutral lipid storage myopathy
3350	Neuhauser-Daly-Magnelli syndrome	2678	Neurofibromatosis type 6	→86872	Neutropenia - hyperlymphocytosis with large granular lymphocytes
2672	Neuhauser-Eichner-Opitz syndrome	638	Neurofibromatosis-Noonan syndrome	2690	Neutropenia - moncytopenia - deafness
2901	Neuralgic amyotrophy	3148	Neurofibrosarcoma	183707	Neutrophil immunodeficiency syndrome
2901	Neuralgic shoulder amyotrophy	970	Neurogenic acroosteolysis	169142	Neutrophil-specific granule deficiency
351	Neuraminidase deficiency with beta-galactosidase deficiency	1143	Neurogenic arthrogryposis multiplex congenita	575	Neutrophilic urticaria
268865	Neurenteric cyst			370059	NEVADA syndrome
252164	Neurilemmoma				
93921	Neurilemmomatosis				
252164	Neurilemoma				

→ 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
→1359	Nevi - atrial myxoma - myxoid neurofibromata - ephelides	216972	Niemann-Pick disease type C, severe perinatal form	2700	Noma
→1900	Nevo syndrome	→646	Niemann-Pick disease type D	1451	NOMID syndrome
377	Nevoid basal cell carcinoma syndrome	99022	Niemann-Pick disease type E	73267	Non-24-hour sleep-wake syndrome
228264	Nevus anelasticus	→646	Niemann-Pick disease, Nova Scotia type	231720	Non-acquired combined pituitary hormone deficiency with spine abnormalities
64754	Nevus comedonicus syndrome	2633	Nievergelt syndrome	631	Non-acquired isolated growth hormone deficiency
228254	Nevus elasticus	1390	Night blindness - skeletal anomalies - dysmorphism	97566	Non-amyloid fibrillary glomerulopathy
370059	Nevus epidermicus verrucosus with angiodyplasia and aneurysms	98757	Nigro-spino-dental degeneration with nuclear ophthalmoplegia	86861	Non-amyloid MIDD
263432	Nevus fuscocaeuleus acromiodeltoideus	432	nIHH	86861	Non-amyloid monoclonal immunoglobulin deposition disease
263425	Nevus fusculoceruleus ophthalmomaxillaris	2322	Niikawa-Kuroki syndrome	79394	Non-bullous congenital ichthyosiform erythroderma
263432	Nevus of Ito	647	Nijmegen breakage syndrome	289362	Non-central nervous system-localized embryonal carcinoma
263425	Nevus of Ota	240760	Nijmegen breakage syndrome-like disorder	77259	Non-cerebral juvenile Gaucher disease
2612	Nevus sebaceus of Jadassohn	447731	NIK deficiency	48372	Non-cirrhotic nodulation
2612	Nevus sebaceus syndrome	781	Nine Mile fever	325529	Non-classic congenital lipoid adrenal hyperplasia due to STAR deficiency
363558	New-onset refractory status epilepticus	99825	Nipah encephalitis	289362	Non-CNS-localized embryonal carcinoma
83471	Nezelof syndrome	99825	Nipah fever	216796	Non-deforming osteogenesis imperfecta
636	NF1	59303	NISCH syndrome	96136	Non-distal deletion 7p
97685	NF1 microdeletion syndrome	1422	Nivelon-Nivelon-Mabille syndrome	1581	Non-distal deletion 10q
137605	NF1-like syndrome	263665	NK-cell enteropathy	96160	Non-distal deletion 12q
637	NF2	86873	NK-cell large granular lymphocyte leukemia	96164	Non-distal deletion 20q
93921	NF3	86873	NK-cell LGL leukemia	96112	Non-distal duplication 9q
2678	NF6	86879	NK/T-cell lymphoma	1695	Non-distal duplication 10q
69087	NFJ syndrome	407	NKA	1702	Non-distal duplication 13q
638	NFNS	86879	NKTCL	96136	Non-distal monosomy 7p
91349	NFPA	86893	NLPHL	1581	Non-distal monosomy 10q
401869	NFU1 deficiency	247868	NLRP12-associated hereditary periodic fever syndrome	96160	Non-distal monosomy 12q
289356	NGCO	98907	NLSDI	96164	Non-distal monosomy 20q
404454	NGLY1 deficiency	98908	NLSDM	3306	Non-distal tetrasomy 15q
404454	NGLY1-CDDG	443167	NMC	96112	Non-distal trisomy 9q
280576	NGPS	391504	NMG	1695	Non-distal trisomy 10q
2770	NHD	86867	NMZL	1702	Non-distal trisomy 13q
169079	NHEJ1 deficiency	2615	NNS	329469	Non-DS-AMKL
276608	NI-PHH	1884	Noble-Bass-Sherman syndrome	206538	Non-dysgerminomatous germ cell cancer of ovary
247598	NICCD	31204	Nocardiosis	363494	Non-dysgerminomatous germ cell tumor of testis
141179	NICH	→98784	Nocturnal paroxysmal dystonia	2337	Non-epidermolytic palmoplantar keratoderma
3051	Nicolaides-Baraitser syndrome	86867	Nodal marginal zone B-cell lymphoma	→2199	Non-epidermolytic palmoplantar keratoderma
77292	Niemann-Pick disease type A	137810	Nodular cutaneous amyloidosis	2972	Non-eruption of teeth - maxillary hypoplasia - genu valgum
77293	Niemann-Pick disease type B	90393	Nodular lichen myxedematosus	100070	Non-fluent variant PPA
646	Niemann-Pick disease type C	86893	Nodular lymphocyte predominant Hodgkin lymphoma	91349	Non-functioning pituitary adenoma
216986	Niemann-Pick disease type C, adult neurologic onset	2149	Nodular neuronal heterotopia		
216981	Niemann-Pick disease type C, classic form	33577	Nodular non-suppurative panniculitis		
216981	Niemann-Pick disease type C, juvenile neurologic onset	48372	Nodular regenerative hyperplasia of the liver		
216978	Niemann-Pick disease type C, late infantile neurologic onset	158772	Nodular urticaria pigmentosa		
216975	Niemann-Pick disease type C, severe early infantile neurologic onset	85196	Nodulosis-arthropathy-osteolysis 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
26137	Non-giant cell granulomatous temporal arteritis with eosinophilia	94080	Non-secreting paraganglioma	812	Normomorphic sialidosis
→79452	Non-hereditary congenital primary lymphedema	363494	Non-seminomatous germ cell tumor of testis	→682	NormoPP
→90186	Non-hereditary late-onset primary lymphedema	91364	Non-specific idiopathic interstitial pneumonia	432	Normosmic congenital hypogonadotropic hypogonadism
357034	Non-hereditary retinoblastoma	91364	Non-specific interstitial pneumonia	432	Normosmic idiopathic hypogonadotropic hypogonadism
163924	Non-herpetic acute limbic encephalitis	90031	Non-spherocytic hemolytic anemia due to hexokinase deficiency	649	Norrie disease
329883	Non-hypoproteinemic hypertrophic gastropathy	35099	Non-syndromic bicornal synostosis	649	Norrie-Warburg disease
329918	Non-Ig-mediated membranoproliferative glomerulonephritis	30391	Non-syndromic biliary atresia	363558	NORSE
329918	Non-Ig-mediated MPGN	300337	Non-syndromic congenital retinal non-attachment	75327	North Carolina macular dystrophy
363999	Non-immune fetal edema	49042	Non-syndromic dentinogenesis imperfecta	75327	North Carolina macular dystrophy, retinal 1
363999	Non-immune fetal hydrops	49042	Non-syndromic DGI	280620	North Sea progressive myoclonus epilepsy
363999	Non-immune HF	276234	Non-syndromic male infertility due to asthenozoospermia	1947	Northern epilepsy
363999	Non-immune hydrops fetalis	276234	Non-syndromic male infertility due to sperm motility disorder	79293	Norum disease
329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis	3366	Non-syndromic metopic craniosynostosis	1134	Nose agenesis
329918	Non-immunoglobulin-mediated MPGN	35093	Non-syndromic sagittal synostosis	77304	Not NOTCH3-related small vessel disease of the brain
263548	Non-inflammatory generalized peeling skin syndrome type A.	35098	Non-syndromic unicoronal synostosis	178	Notochordal sarcoma
263548	Non-inflammatory peeling skin syndrome type A	96136	Non-telomeric monosomy 7p	2703	Nova syndrome
141179	Non-involuting congenital hemangioma	1581	Non-telomeric monosomy 10q	2005	Novak syndrome
407	Non-ketotic hyperglycinemia	96160	Non-telomeric monosomy 12q	314928	NPH
98890	Non-Leber type optic atrophy with early-onset	96164	Non-telomeric monosomy 20q	3032	NPHP3-related Meckel-like syndrome
411641	Non-nephropathic cystinosis	3306	Non-telomeric tetrasomy 15q	634	NS
84085	Non-neurogenic neurogenic bladder	96112	Non-telomeric trisomy 9q	88616	NS-ARID
209989	Non-papillary transitional cell carcinoma of the bladder	1695	Non-telomeric trisomy 10q	2701	NS/LAH
209989	Non-papillary urothelial carcinoma	1702	Non-telomeric trisomy 13q	417	NSHPT
238583	Non-phenylketonuric hyperphenylalaninemia	411703	Non-tuberculous mycobacterial lung disease	93606	NSIAD
79651	Non-PKU HPA	209919	Non-Wilsonian hepatic copper toxicosis of infancy and childhood	91364	NSIP
99817	Non-polyposis Turcot syndrome	602	Nonaka myopathy	100073	NTOS
1766	Non-progressive cerebellar ataxia - intellectual disability	79452	Nonne-Milroy lymphedema	314790	Null pituitary adenoma
314647	Non-progressive cerebellar ataxia with intellectual disability	648	Noonan syndrome	280234	Null syndrome
436271	Non-progressive predominantly posterior cavitating leukoencephalopathy with peripheral neuropathy	500	Noonan syndrome with multiple lentigines	443167	NUT midline carcinoma
439202	Non-recovering OBPI	363972	Noonan syndrome-like disorder with JMML	54	OA1
439202	Non-recovering OBPL	363972	Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	398156	OAFNS
439202	Non-recovering obstetric brachial plexus lesion	2701	Noonan syndrome-like disorder with loose anagen hair	1106	OAS
101106	Non-secreting chemodectoma	230	Noradrenaline deficiency	374	OAV dysplasia
		230	Norepinephrine deficiency	374	OAVS
		314928	Normal pressure hydrocephalus	97297	Oberklaid-Danks syndrome
		2254	Norman disease	88643	Obesity - colitis - hypothyroidism - cardiac hypertrophy - developmental delay
		79255	Norman-Landing disease	397615	Obesity due to CEP19 deficiency
		→682	Normokalemic periodic paralysis	66628	Obesity due to congenital leptin deficiency
		→682	Normokalemic PP	179494	Obesity due to leptin receptor gene deficiency
		→682	NormoKPP	217031	Obesity due to MC3R deficiency
				71529	Obesity due to melanocortin 4 receptor deficiency
				71526	Obesity due to pro-opiomelanocortin deficiency

→ 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
71528	Obesity due to prohormone convertase 1 deficiency	534	Oculo-cerebro-renal syndrome	→29384 3	Oculopalatoskeletal syndrome
369873	Obesity due to SIM1 deficiency	1305	Oculo-digito-esophageal-duodenal syndrome	98897	Oculopharyngeal distal myopathy
1303	Obliterative bronchiolitis	391641	Oculo-digito-esophageal-duodenal syndrome type 1	270	Oculopharyngeal muscular dystrophy
64743	Obliterative portal venopathy	→1200	Oculo-oto-facial dysplasia	98897	Oculopharyngodistal myopathy
2970	Obrinsky syndrome	2307	Oculo-oto-radial syndrome	2715	Oculorenocerebellar syndrome
3411	Obstructed hemivagina and ipsilateral renal anomaly	2714	Oculo-palato-cerebral dwarfism	2717	Oculotrichoanal syndrome
352731	OCA1	2714	Oculo-palato-cerebral syndrome	2718	Oculotrichodysplasia
352734	OCA1-MP	→29384 3	Oculo-skeletal-abdominal syndrome	166272	ODCD
352737	OCA1-TS	2716	Oculo-skeletal-renal syndrome	2710	ODDD syndrome
79431	OCA1A	157962	Oculoauricular syndrome, Schorderet type	1305	ODED syndrome
79434	OCA1B	398156	Oculoauriculofrontonasal syndrome	391641	ODED syndrome type 1
79432	OCA2	374	Oculoauriculovertebral dysplasia	999	O'Doherty syndrome
79433	OCA3	2549	Oculoauriculovertebral spectrum with radial defects	2253	O'Donnell-Pappas syndrome
79435	OCA4	374	Oculoauriculovertebral syndrome	2722	Odonto-onycho dysplasia - alopecia
370091	OCA5	2705	Oculocerebral dysplasia	2721	Odonto-onycho-dermal dysplasia
370097	OCA6	2719	Oculocerebral hypopigmentation syndrome, Cross type	→2036	Odonto-onycho-hypohidrotic dysplasia - midline scalp defects
352745	OCA7	2720	Oculocerebral hypopigmentation syndrome, Preus type	69082	Odonto-tricho-ungual-digitopalmar syndrome
217017	Occipital atretic cephalocele - unusual facies - large feet	1647	Oculocerebrocutaneous syndrome	69082	Odonto-tricho-ungual-digitopalmar syndrome, Mendoza-Valiente type
268823	Occipital encephalocele	2707	Oculocerebrofacial syndrome, Kaufman type	166272	Odontochondrodyplasia
198	Occipital horn syndrome	534	Oculocerebrorenal dystrophy	447777	Odontogenic keratocystoma
280640	Occipital malformations of cortical development	534	Oculocerebrorenal syndrome of Lowe	247685	Odontohypophosphatasia
280640	Occipital MCD	352731	Oculocutaneous albinism type 1	77295	Odontoleukodystrophy
280640	Occipital pachygyria and polymicrogyria	79431	Oculocutaneous albinism type 1A	2724	Odontomatosis - aortae esophagus stenosis
353351	Occlusive idiopathic juxtafoveolar retinal telangiectasis	79434	Oculocutaneous albinism type 1B	1811	Odontomicrognathia dysplasia
51608	Occlusive infantile arteriopathy	79432	Oculocutaneous albinism type 2	2723	Odontotrichomelic syndrome
1647	OCCS	79433	Oculocutaneous albinism type 3	1487	ODP
99889	Occult ectopic ACTH secretion	79435	Oculocutaneous albinism type 4	93929	OEIS complex
247834	Occult macular dystrophy	370091	Oculocutaneous albinism type 5	2676	Oerter-Friedman-Anderson syndrome
84085	Occult neuropathic bladder	370097	Oculocutaneous albinism type 6	2792	OFC syndrome
2704	Ochoa syndrome	352745	Oculocutaneous albinism type 7	2712	OFCD syndrome
247834	OCMD	79434	Oculocutaneous albinism, Amish type	2750	OFD1
534	OCR	28378	Oculocutaneous tyrosinemia	2751	OFD2
534	OCRL	2709	Oculodental syndrome, Rutherford type	2752	OFD3
664	OCT deficiency	2710	Oculodentodigital dysplasia	2753	OFD4
54	Ocular albinism type 1	2710	Oculodentoosseous dysplasia	2919	OFD5
352740	Ocular albinism with congenital sensorineural deafness	3339	Oculoectodermal syndrome	2754	OFD6
1000	Ocular albinism with late-onset sensorineural deafness	2712	Oculofaciocardiodental syndrome	→2750	OFD7
54	Ocular albinism, Nettleship-Falls type	1876	Oculogastrointestinal muscular dystrophy	2755	OFD8
195	Ocular coloboma - imperforate anus	1794	Oculomaxillofacial dysostosis	141007	OFD9
411641	Ocular cystinosis	1154	Oculomelic amyoplasia	2756	OFD10
2788	Ocular form of osteogenesis imperfecta	1125	Oculomotor apraxia, Cogan type	141000	OFD11
1125	Ocular motor apraxia, Cogan type	2713	Oculoosteocutaneous syndrome	141327	OFD12
99922	Ocular pemphigoid	99806	Oculootodental syndrome	141330	OFD13
534	Oculo-cerebro-renal dystrophy			434179	OFD14
				2750	OFDI
				2750	OFDSI
				391655	Off-periods in Parkinson disease not responding to oral treatment

→ 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
424080	OGCT of pancreas	39041	Omenn syndrome	1308	Opitz C trigonocephaly
276432	Ogden syndrome	2741	OMM syndrome	2745	Opitz G/BBB syndrome
75382	Oguchi disease	2733	Omodyplasia	2745	Opitz syndrome
75382	Oguchi syndrome	660	Omphalocele	1308	Opitz trigonocephaly C syndrome
1186	Ohaha syndrome	93929	Omphalocele - cloacal extrophy - imperforate anus - spinal defect	1308	Opitz trigonocephaly syndrome
2728	Ohdo syndrome	3164	Omphalocele syndrome, Shprintzen-Goldberg type	97297	Opitz trigonocephaly-like syndrome
2728	Ohdo-Madokoro-Sonoda syndrome	490	Omphalomesenteric cyst	1786	Opitz-Caltabiano syndrome
64739	OHSS	210115	OMPP	2745	Opitz-Frias syndrome
1934	Ohtahara syndrome	1183	OMS	270	OPMD
3411	OHVIRA syndrome	319266	Omsk hemorrhagic fever	256	Oppenheim dystonia
666	OI	3191	Onat syndrome	2788	OPPG
216796	OI type 1	2737	Onchocerciasis	2746	Opsismodysplasia
216804	OI type 2	137675	Oncocytic cardiomyopathy	1183	Opsoclonus-myoclonus syndrome
216812	OI type 3	352540	Oncogenic hypophosphatemic osteomalacia	1183	Opsoclonus-myoclonus-ataxia syndrome
216820	OI type 4	352540	Oncogenic osteomalacia	363746	Optic ataxia-gaze apraxia-simultanagnosia syndrome
216828	OI type 5	661	Ondine curse	1215	Optic atrophy - deafness-polyneuropathy - myopathy
2729	Okamoto syndrome	661	Ondine syndrome	→1215	Optic atrophy - ophthalmoplegia - ptosis - deafness - myopathy
93293	Okihiro syndrome	99803	Ondine-Hirschsprung disease	98673	Optic atrophy type 1
261638	Okihiro syndrome due to 20q13 microdeletion	99803	Ondine-Hirschsprung syndrome	98890	Optic atrophy type 2
261647	Okihiro syndrome due to a point mutation	→33364	ONMR syndrome	401777	Optic atrophy-intellectual disability syndrome
261638	Okihiro syndrome due to del(20)(q13)	238744	Onycho-digitomammary syndrome	313800	Optic nerve edema-splenomegaly syndrome
261638	Okihiro syndrome due to monosomy 20q13	→33364	Onycho-tricho-dysplasia - neutropenia	2086	Optic pathway glioma
69088	OL-EDA-ID	300504	Onychocytic matricoma	413681	Oral antidiabetic drugs toxicity or dose selection
79458	Oley syndrome	79153	Onychodystrophy totalis	31142	Oral erosive lichen
478	Olfacto-genital pathological sequence	300512	Onychomatricoma	357154	Oral submucous fibrosis
1957	Olfactory neuroblastoma	2614	Onychoosteodysplasia	2750	Oral-facial-digital syndrome type 1
85410	Oligoarticular juvenile arthritis	2786	OOCHS	2751	Oral-facial-digital syndrome type 2
247839	Oligoarticular juvenile arthritis with anti-nuclear antibodies	99806	OOD	2752	Oral-facial-digital syndrome type 3
247846	Oligoarticular juvenile arthritis without anti-nuclear antibodies	2721	OODD	2753	Oral-facial-digital syndrome type 4
251656	Oligoastrocytoma	98890	OPA2	2919	Oral-facial-digital syndrome type 5
75378	Oligocone syndrome	67036	OPA3, autosomal dominant	2754	Oral-facial-digital syndrome type 6
75378	Oligocone trichromacy	49042	Opalescent teeth without OI	→2750	Oral-facial-digital syndrome type 7
251627	Oligodendrogloma	49042	Opalescent teeth without osteogenesis imperfecta	2755	Oral-facial-digital syndrome type 8
99798	Oligodontia	90650	OPD I syndrome	141007	Oral-facial-digital syndrome type 9
300576	Oligodontia - cancer predisposition syndrome	90652	OPD II syndrome	2756	Oral-facial-digital syndrome type 10
2260	Oligomeganephronia	90650	OPD syndrome 1	141000	Oral-facial-digital syndrome type 11
2260	Oligomeganephronic renal hypoplasia	90652	OPD syndrome 2	141327	Oral-facial-digital syndrome type 12
137831	Oligophrenin-1 syndrome	98897	OPDM	141330	Oral-facial-digital syndrome type 13
2920	Oliver syndrome	268363	Open iniencephaly	434179	Oral-facial-digital syndrome type 14
3363	Oliver-McFarlane syndrome	137831	OPHN1 syndrome	141007	Oral-facial-digital syndrome with retinal abnormalities
2732	Olivopontocerebellar atrophy - deafness	1106	Ophthalmacromelic syndrome	2755	Oral-facial-digital syndrome, Edwards type
166063	Olivopontocerebellar hypoplasia	2741	Ophthalmomandibulomelic dysplasia	141000	Oral-facial-digital syndrome, Gabrielli type
296	Ollier disease	1186	Ophthalmoplegia - hypotonia - ataxia - hypoacusis - athetosis	1647	Orbital cyst with cerebral and focal dermal malformations
659	Olmsted syndrome	2743	Ophthalmoplegia - intellectual disability - lingua scrotalis		
1183	OMA syndrome	2742	Ophthalmoplegia - myalgia - tubular aggregates		
247834	OMD				

→ 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
52994	Orbital leiomyoma	140436	Osseous vascular malformation	2324	Osteopenia - intellectual disability - sparse hair
268139	Orbital medulloepithelioma	73230	Ossification anomalies - psychomotor development delay	91133	Osteopenia - myopia - hearing loss - intellectual disability - facial dysmorphism
2612	Organoid nevus syndrome	58040	Osteoblastoma	178389	Osteopetrosis - hypogammaglobulinemia
166421	Orgasm-induced seizures	2764	Osteochondritis dissecans	53	Osteopetrosis autosomal dominant type 2
49041	Ormond disease	251262	Osteochondritis dissecans and short stature	2785	Osteopetrosis with renal tubular acidosis
414	Ornithine aminotransferase deficiency	3314	Osteochondritis of phalangeal epiphyses	94063	Osteopoikilosis - short stature - intellectual disability
664	Ornithine carbamoyltransferase deficiency	2054	Osteochondritis of tarsal/metatarsal bone	2787	Osteoporosis - macrocephaly - blindness - joint hyperlaxity
415	Ornithine carrier deficiency	2380	Osteochondritis of the capital femoral epiphysis	2786	Osteoporosis - oculocutaneous hypopigmentation syndrome
664	Ornithine transcarbamylase deficiency	97332	Osteochondritis of the lunate bone	2788	Osteoporosis - pseudoglioma
415	ORNT1 deficiency	97335	Osteochondritis of the tibial tubercle	666	Osteopsathyrosis
2319	Orocraniodigital syndrome	2653	Osteochondrodysplastic dwarfism - deafness - retinitis pigmentosa	668	Osteosarcoma
2750	Orofaciodigital syndrome type 1	2653	Osteochondrodysplastic nanism - deafness - retinitis pigmentosa	2760	Osteosarcoma - limb anomalies - erythroid macrocytosis
2751	Orofaciodigital syndrome type 2	800	Osteochondromuscular dystrophy	75325	Osteosclerosis - ichthyosis - premature ovarian failure
2752	Orofaciodigital syndrome type 3	2768	Osteochondrosis deformans tibiae	178377	Osteosclerosis-developmental delay-craniosynostosis syndrome
2753	Orofaciodigital syndrome type 4	97337	Osteochondrosis of patella	2905	Osteosclerotic myeloma
2919	Orofaciodigital syndrome type 5	3314	Osteochondrosis of phalangeal epiphyses	1338	Ostravik-Lindemann-Solberg syndrome
2754	Orofaciodigital syndrome type 6	2380	Osteochondrosis of the capital femoral epiphysis	99965	O'Sullivan-McLeod syndrome
→2750	Orofaciodigital syndrome type 7	97336	Osteochondrosis of the capital humerus	664	OTC deficiency
2755	Orofaciodigital syndrome type 8	97332	Osteochondrosis of the lunate bone	1308	OTCS
141007	Orofaciodigital syndrome type 9	2054	Osteochondrosis of the tarsal bone	2791	Otodontal dysplasia
2756	Orofaciodigital syndrome type 10	97335	Osteochondrosis of the tibial tubercle	2791	Otodontal syndrome
141000	Orofaciodigital syndrome type 11	424080	Osteoclastic giant cell tumor of pancreas	2792	Otofaciocervical syndrome
141327	Orofaciodigital syndrome type 12	2763	Osteocraniosplenic syndrome	141136	Otomandibular dysostosis
141330	Orofaciodigital syndrome type 13	2763	Osteocraniostenosis	141136	Otomandibular syndrome
434179	Orofaciodigital syndrome type 14	666	Osteogenesis imperfecta	2793	Otoonychoperoneal syndrome
2756	Orofaciodigital syndrome with fibular aplasia	2771	Osteogenesis imperfecta - congenital joint contractures	669	Otopalatodigital syndrome
141007	Orofaciodigital syndrome with retinal abnormalities	2773	Osteogenesis imperfecta - retinopathy - seizures - intellectual disability	90650	Otopalatodigital syndrome type 1
2755	Orofaciodigital syndrome, Edwards type	216796	Osteogenesis imperfecta type 1	90652	Otopalatodigital syndrome type 2
141000	Orofaciodigital syndrome, Gabrielli type	216804	Osteogenesis imperfecta type 2	1427	Otospondylomegaphyseal dysplasia
2919	Orofaciodigital syndrome, Thurston type	216812	Osteogenesis imperfecta type 3	69082	OTUDP syndrome
93958	Oromandibular dystonia	216820	Osteogenesis imperfecta type 4	50943	Oudtshoorn disease
141077	Oropharyngeal teratoma	216828	Osteogenesis imperfecta type 5	1179	Ouvrier-Billson syndrome
30	Oroticaciduria	668	Osteogenic sarcoma	213504	Ovarian adenocarcinoma
30	Orotidylid decarboxylase deficiency	2645	Osteoglophonic dwarfism	213512	Ovarian carcinosarcoma
64692	Oroya fever	2777	Osteomesopyknosis	398971	Ovarian clear cell adenocarcinoma
443236	Orthostatic intolerance due to NET deficiency	399293	Osteonecrosis of the jaw	314473	Ovarian fibroma
→29384	OSA syndrome	2780	Osteopathia striata - cranial sclerosis	314478	Ovarian fibrothecoma
3		2779	Osteopathia striata - pigmentary dermopathy - white forelock	206484	Ovarian gonadoblastoma
93382	Osebold-Remondini syndrome			64739	Ovarian hyperstimulation syndrome
97335	Osgood-Schlatter disease			398987	Ovarian immature teratoma
2760	OSLAM syndrome				
729	Osler-Vaquez disease				
1427	OSMED				
357154	OSMF				

→ 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
99916	Ovarian malignant Sertoli-Leydig cell tumor	1388	Palatodigital syndrome, Catel-Manzke type	2202	Palmoplantar keratoderma-hearing loss syndrome
398987	Ovarian malignant teratoma	171695	Pallidopyramidal syndrome	384	Palmoplantar keratoderma-sclerodactyly syndrome
398961	Ovarian mucinous adenocarcinoma	3138	Pallister ulnar-mammary syndrome	2201	Palmoplantar keratoderma-spastic paralysis syndrome
99916	Ovarian Sertoli-Leydig cell cancer	672	Pallister-Hall syndrome	→79502	Palmoplantar porokeratosis of Mantoux
206473	Ovarian tumor of low malignant potential	884	Pallister-Killian syndrome	163927	Palmoplantar pustulosis
99853	Ovarioleukodystrophy	2804	Pallister-W syndrome	767	PAN
137634	Overgrowth - macrocephaly - facial dysmorphism	737	Palmar, plantar and disseminated porokeratosis	98815	Panayiotopoulos syndrome
3203	Overhydrated hereditary stomatocytosis	2184	Palmer-Pagon syndrome	424046	Pancreatic acinar cell carcinoma
326	Owren disease	659	Palmoplantar and periorificial keratoderma	93292	Pancreatic adenoma
832	OXCT1 deficiency	50944	Palmoplantar hyperkeratosis - cystic eyelids - hypodontia - hypotrichosis	65288	Pancreatic and cerebellar agenesis
31	Oxoglutaricaciduria	2342	Palmoplantar hyperkeratosis - periodontopathia - onychogryposis	97282	Pancreatic cholera
33572	Oxoprolinuria due to oxoprolinase deficiency	85112	Palmoplantar hyperkeratosis - XX sex reversal - predisposition to squamous cell carcinoma	309108	Pancreatic colipase deficiency
79302	Oxysterol 7-alpha-hydroxylase deficiency	34217	Palmoplantar hyperkeratosis with arrhythmogenic cardiomyopathy	2255	Pancreatic hypoplasia - diabetes - congenital heart disease
36355	P2Y12 defect	140966	Palmoplantar hyperkeratosis, Nagashima type	199337	Pancreatic insufficiency - anemia - hyperostosis
35664	P5CS deficiency	2202	Palmoplantar hyperkeratosis-deafness syndrome	811	Pancreatic insufficiency and bone marrow dysfunction
35120	P5N deficiency	2198	Palmoplantar hyperkeratosis-esophageal carcinoma syndrome	424058	Pancreatic intraductal papillary mucinous carcinoma
98971	PACD	2202	Palmoplantar hyperkeratosis-hearing loss syndrome	424053	Pancreatic mucinous cystadenocarcinoma
2796	Pachydermoperiostosis	384	Palmoplantar hyperkeratosis-sclerodactyly syndrome	424080	Pancreatic osteoclastic giant cell tumor
→2995	Pachygyria - epilepsy - intellectual disability - dysmorphism	2201	Palmoplantar hyperkeratosis-spastic paralysis syndrome	97278	Pancreatic polypeptidoma
2798	Pachygyria - intellectual disability - epilepsy	86919	Palmoplantar keratoderma - clinodactyly	424073	Pancreatic serous cystadenocarcinoma
2309	Pachyonychia congenita	50944	Palmoplantar keratoderma - cystic eyelids - hypodontia - hypotrichosis	424065	Pancreatic solid pseudopapillary carcinoma
1952	Pacman dysplasia	2342	Palmoplantar keratoderma - periodontopathia - onychogryposis	424039	Pancreatic squamous cell carcinoma
140989	PACNS	85112	Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma	309031	Pancytopenia due to IKZF1 mutations
706	PAD	1010	Palmoplantar keratoderma and congenital alopecia, Stevanovic type	401764	Pancytopenia-developmental delay syndrome
441	PAF	1366	Palmoplantar keratoderma and congenital alopecia, Wallis type	66624	PANDAS
95232	PAFAH1B1-related lissencephaly	34217	Palmoplantar keratoderma with arrhythmogenic cardiomyopathy	95513	Panhypophysitis
180275	Paget disease of the nipple	→2199	Palmoplantar keratoderma with tonotubular keratin	90695	Panhypopituitarism
357131	Paget-Schrotter disease	140966	Palmoplantar keratoderma, Nagashima type	97336	Panner disease
52430	Pagetoid amyotrophic lateral sclerosis	2202	Palmoplantar keratoderma-deafness syndrome	90159	Panniculitis and localized lipodystrophy
52430	Pagetoid neuroskeletal syndrome	2198	Palmoplantar keratoderma-esophageal carcinoma syndrome	157850	Pantothenate kinase-associated neurodegeneration
178517	Pagetoid reticulosclerosis, Woringer-Kolopp type			440427	PAP, Reunion island type
180275	Paget's disease of the nipple			69126	PAPA syndrome
991	PAGOD syndrome				
716	PAH deficiency				
1993	Pai syndrome				
37202	Painful bladder syndrome				
324636	Painful bruising syndrome				
99736	Painful congenital myotonia				
99736	Painful myotonia				
64686	Painful ophthalmoplegia				
300501	Painful orbital and systemic neurofibromas-marfanoid habitus syndrome				
90797	PAIS				

→ 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
213817	Papillary carcinoma of the cervix uteri	251290	Parietal foramina with cleidocranial dysostosis	90076	Partial deep dermal and full thickness burns
213726	Papillary carcinoma of the corpus uteri	251290	Parietal foramina with cleidocranial dysplasia	79312	Partial deficiency of methylmalonyl-CoA mutase
208600	Papillary fibroelastoma of the heart	851	Paris-Trousseau thrombocytopenia	261318	Partial duplication of chromosome 20p
251962	Papillary glioneuronal tumor	306674	PARK9	261318	Partial duplication of the short arm of chromosome 20
146	Papillary or follicular thyroid carcinoma	199351	PARK14	101046	Partial epilepsy with auditory aura
319298	Papillary renal cell adenocarcinoma	90307	Parkes Weber syndrome	101046	Partial epilepsy with auditory features
319298	Papillary renal cell carcinoma	171695	Parkinsonian-pyramidal syndrome	2704	Partial facial palsy with urinary abnormalities
251915	Papillary tumour of the pineal region	314632	Parkinsonism due to ATP13A2 deficiency	744	Partial gigantism - nevi - hemihypertrophy - macrocephaly
1475	Papillo-renal syndrome	178509	Parkinsonism with alveolar hypoventilation and mental depression	254693	Partial hydatidiform mole
2807	Papilloma of choroid plexus	97355	Parkinsonism with dementia of Guadeloupe	79292	Partial LCAT deficiency
2750	Papillon-Léage-Psaume syndrome	90020	Parkinsonism-dementia-ALS complex	343	Partial mevalonate kinase deficiency
678	Papillon-Lefèvre syndrome	90035	Paroxysmal cold hemoglobinuria	254693	Partial molar pregnancy
86819	Papular atrichia	53583	Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	2805	Partial pancreatic agenesis
228264	Papular elastorrhexis	98811	Paroxysmal exertion-induced dyskinesia	157769	Partial situs inversus
313936	Papular epidermal nevi with skyline basal cell layers syndrome	46348	Paroxysmal extreme pain disorder	261318	Partial trisomy of chromosome 20p
90395	Papular mucinosis of infancy	157835	Paroxysmal hemicrania	261318	Partial trisomy of the short arm of chromosome 20
158008	Papular xanthoma	→98784	Paroxysmal hypnagogic dyskinesia	85453	Partington disease
679	Papulosis atrophicana maligna	→98784	Paroxysmal hypnagogic dystonia	94083	Partington syndrome
99056	Parachute tricuspid valve	→98784	Paroxysmal hypnogenic dyskinesia	→193	Partington-Anderson syndrome
73260	Paracoccidioidomycosis	98809	Paroxysmal kinesigenic choreathetosis	94083	Partington-Mulley syndrome
324299	Paraganglioma - somatostatinoma - polycythemia	98809	Paroxysmal kinesigenic dyskinesia	295	Parvovirus antenatal infection
97286	Paraganglioma and gastric stromal sarcoma	31709	Paroxysmal kinesigenic dyskinesia and infantile convulsions	1394	Pascual-Castroviejo syndrome type 1
326	Parahemophilia	→98784	Paroxysmal nocturnal dyskinesia	42775	Pascual-Castroviejo syndrome type 2
141242	Paramedian nasal cleft	447	Paroxysmal nocturnal hemoglobinuria	289478	PASH syndrome
684	Paramyotonia congenita	98810	Paroxysmal non-kinesigenic dyskinesia	1252	Pashayan syndrome
684	Paramyotonia congenita of Von Eulenburg	98810	Paroxystic non-kinesigenic choreoathetosis	1252	Pashayan-Prozansky syndrome
2812	Parana hard-skin syndrome	1214	Parry-Romberg syndrome	2278	Passwell-Goodman-Sipkowski syndrome
99889	Paraneoplastic Cushing syndrome	574	Partial 21q monosomy	3378	Patau syndrome
1183	Paraneoplastic opsoclonus-myoclonus	79087	Partial acquired lipodystrophy	→1509	Patella aplasia - coxa vara - tarsal synostosis
1183	Paraneoplastic opsoclonus-myoclonus-ataxia syndrome	2805	Partial agenesis of the pancreas	86789	Patella aplasia/hypoplasia
63455	Paraneoplastic pemphigus	381	Partial albinism - immunodeficiency	295041	Patella aplasia/hypoplasia, bilateral
71505	Paraneoplastic retinopathy	90797	Partial androgen insensitivity syndrome	295038	Patella aplasia/hypoplasia, unilateral
231445	Paraparetic variant of GBS	90797	Partial androgen resistance syndrome	706	Patent arterial duct
231445	Paraparetic variant of Guillain-Barré syndrome	1330	Partial atrioventricular canal	228190	Patent arterial duct - bicuspid aortic valve - hand anomalies
2823	Paraplegia - brachydactyly - cone-shaped epiphysis	1330	Partial atrioventricular canal defect	706	Patent ductus arteriosus
2824	Paraplegia - intellectual disability - hyperkeratosis	1646	Partial chromosome Y deletion	228190	Patent ductus arteriosus - bicuspid aortic valve - hand anomalies
31827	Paraquat poisoning	401959	Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome		Patent ductus arteriosus with facial dysmorphism and abnormal fifth digits
2646	Parastremmatic dwarfism	98950	Partial cryptophthalmia	46627	Patent foramen ovale
363478	Paratesticular adenocarcinoma			99108	Patent urachus
143	Parathyroid carcinoma			431341	
443227	Paratyphoid fever				
2825	PARC syndrome				
268826	Parietal encephalocele				

→ 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
254531	Paternal 14q32.2 hypomethylation syndrome	1578	PCBD deficiency	33402	Pediatric HCC
254525	Paternal 14q32.2 microdeletion syndrome	247198	PCCA	33402	Pediatric hepatocellular carcinoma
261304	Paternal 20q13.2-q13.3 microdeletion syndrome	244	PCD	93552	Pediatric systemic lupus erythematosus
261304	Paternal 20q13.2q13.3 microdeletion syndrome	178544	PCDLBCL,LT	263548	Peeling skin syndrome type A
254525	Paternal del(14)(q32.2)	178540	PCFCL	263553	Peeling skin syndrome type B
261304	Paternal del(20)(q13.2q13.3)	90035	PCH	444138	Peeling skin-leukonuchia-acral punctate keratoses-cheilitis-knuckle pads syndrome
254525	Paternal monosomy 14q32.2	2254	PCH1	2836	PEHO syndrome
261304	Paternal monosomy 20q13.2-q13.3	2524	PCH2	99807	PEHO-like syndrome
261304	Paternal monosomy 20q13.2q13.3	97249	PCH3	48686	PEL
251004	Paternal uniparental disomy of chromosome 1	166063	PCH4	702	Pelizaeus-Merzbacher brain sclerosis
96190	Paternal uniparental disomy of chromosome 5	166068	PCH5	702	Pelizaeus-Merzbacher disease
96191	Paternal uniparental disomy of chromosome 6	166073	PCH6	280229	Pelizaeus-Merzbacher disease in female carriers
96192	Paternal uniparental disomy of chromosome 7	284339	PCH7	280210	Pelizaeus-Merzbacher disease type II
99324	Paternal uniparental disomy of chromosome 13	324569	PCH8	280219	Pelizaeus-Merzbacher disease, classic form
96334	Paternal uniparental disomy of chromosome 14	369920	PCH9	280210	Pelizaeus-Merzbacher disease, connatal form
96194	Paternal uniparental disomy of chromosome 20	97249	PCH with optic atrophy	280234	Pelizaeus-Merzbacher disease, null syndrome
96195	Paternal uniparental disomy of chromosome 21	97249	PCH without dyskinesia	280224	Pelizaeus-Merzbacher disease, transitional form
261524	Paternal uniparental disomy of chromosome X	411493	PCH10	280270	Pelizaeus-Merzbacher-like disease
96194	Paternal UPD20	71528	PCI deficiency	280293	Pelizaeus-Merzbacher-like disease due to AIMP1 mutation
2439	Patterson-Stevenson syndrome	2924	PCLD	280282	Pelizaeus-Merzbacher-like disease due to GJC2 mutation
2439	Patterson-Stevenson-Fontaine syndrome	178536	PCMZL	280288	Pelizaeus-Merzbacher-like disease due to HSPD1 mutation
79136	PATX	438134	PCNA-related progressive neurodegenerative photosensitivity syndrome	97352	Pellagra
93126	Pauci-immune glomerulonephritis	46135	PCNSL	2837	Pellagra-like skin rash - neurological manifestations
97563	Pauci-immune glomerulonephritis with ANCA	140989	PCNSV	137672	Pellucid marginal degeneration
97564	Pauci-immune glomerulonephritis without ANCA	101330	PCT	2840	Pelvic dysplasia - arthrogryposis of lower limbs
85410	Pauciarticular chronic arthritis	163746	PCWH	83628	PELVIS syndrome
247839	Pauciarticular chronic arthritis with anti-nuclear antibodies	90020	PDALS	2839	Pelvis-shoulder dysplasia
247846	Pauciarticular chronic arthritis without anti-nuclear antibodies	293462	PDCD	93333	Pelviscapular dysplasia
1330	PAVC	289157	PDDRI	63275	Pemphigoid gestationis
75373	PBCRA	439822	PDE4D haploinsufficiency syndrome	79480	Pemphigus erythematosus
289666	PBL	765	PDH	79481	Pemphigus foliaceus
2309	PC	79246	PDH phosphatase deficiency	79479	Pemphigus vegetans
54247	PCA	79243	PDHAD	704	Pemphigus vulgaris
88628	PCARP	255138	PDHBD	994	Pena-Shokeir syndrome type 1
231426	PCB variant of GBS	765	PDHC	1466	Pena-Shokeir syndrome type 2
231426	PCB variant of Guillain-Barré syndrome	2796	PDP	705	Pendred syndrome
		85453	PDR	398053	Penile adenocarcinoma
		75496	PDS	49	Penile agenesis
		699	Pearson syndrome	398058	Penile squamous cell carcinoma
		2835	Pectus excavatum - macrocephaly - dysplastic nails	49	Penis agenesis
		98811	PED	2842	Penoscrotal transposition
		439175	Pediatric AIS		
		439175	Pediatric arterial ischemic stroke		
		66624	Pediatric autoimmune disorders associated with Streptococcus infections		
		66624	Pediatric autoimmune neuropsychiatric disorders associated with Streptococcus infections		
		93682	Pediatric Castleman disease		

→ 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
313936	PENS syndrome	213812	Peripheral neuroectodermal cancer of cervix uteri	2380	Perthes disease
11	Penta-X	213630	Peripheral neuroectodermal cancer of corpus uteri	1489	Pertussis
1335	Pentalogy of Cantrell	90120	Peripheral neuropathy and optic atrophy	708	Peters anomaly
11	Pentasomy X	171848	Peripheral neuropathy, Fiskerstrand type	101033	Peters anomaly - cataract
2843	Pentosuria	397744	Peripheral neuropathy-myopathy-hoarseness-deafness syndrome	709	Peters anomaly with short limb dwarfism
352447	PEO - myopathy - emaciation	397744	Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome	708	Peters congenital glaucoma
2905	PEP syndrome	370348	Peripheral PNET	709	Peters plus syndrome
79316	PEPCK1 deficiency	370348	Peripheral primitive neuroectodermal tumor	2776	Petit-Fryns syndrome
2880	PEPCK deficiency	97927	Peripheral resistance to thyroid hormones	2963	Petty syndrome
2576	Perheentupa syndrome	168816	Peritoneal cystic mesothelioma	2963	Petty-Laxova-Wiedemann syndrome
767	Periarteritis nodosa	171676	Periventricular leukomalacia	2869	Peutz-Jeghers syndrome
2847	Pericardial and diaphragmatic defect	98892	Periventricular nodular heterotopia	42642	PFAPA syndrome
2576	Pericardial constriction - growth failure	2849	Perlman syndrome	90042	PFCP
2848	Pericarditis - arthropathy - camptodactyly	438266	PERM	412206	PFE
137577	Perinatal asphyxia	99885	Permanent neonatal diabetes mellitus	710	Pfeiffer syndrome
137577	Perinatal hypoxia	65288	Permanent neonatal diabetes mellitus - pancreatic and cerebellar agenesis	93258	Pfeiffer syndrome type 1
313855	Perinatal lethal bent bone dysplasia	2850	Perniola-Krajewska-Carnevale syndrome	93259	Pfeiffer syndrome type 2
85212	Perinatal lethal Gaucher disease	2971	Peroxisomal acyl-CoA oxidase deficiency	93260	Pfeiffer syndrome type 3
247623	Perinatal lethal hypophosphatasia	93598	Peroxisomal alanine-glyoxylate aminotransferase deficiency	3224	Pfeiffer-Kapferer syndrome
247623	Perinatal lethal phosphoethanolaminuria	2855	Perrault syndrome	2921	Pfeiffer-Mayer syndrome
247623	Perinatal lethal Rathburn disease	75374	PERRS	2871	Pfeiffer-Palm-Teller syndrome
83628	Perineal hemangioma - external genitalia malformations - lipomyelomeningocele - vesicorenal abnormalities - imperforate anus	178509	Perry syndrome	2872	Pfeiffer-Singer-Zschiesche syndrome
95706	Perineal, scrotal or penoscrotal hypospadias	99120	Persistent eustachian valve	33577	Pfeiffer-Weber-Christian syndrome
65250	Perineural cyst	91495	Persistent fetal vasculature syndrome	2019	PFFD
342	Periodic disease	99076	Persistent fifth aortic arch	172	PFIC
42642	Periodic fever-aphtous stomatitis-pharyngitis-adenopathy syndrome	91495	Persistent hyperplastic primary vitreous	79306	PFIC1
436166	Periodic fever-infantile enterocolitis-autoinflammatory syndrome	398147	Persistent idiopathic facial pain	79304	PFIC2
→682	Periodic paralysis type 3	99109	Persistent left superior caval vein connecting to the left-sided atrium	79305	PFIC3
397750	Periodic paralysis with later-onset distal motor neuropathy	99109	Persistent left superior vena cava connecting to the left-sided atrium	91495	PFVS
397755	Periodic paralysis with transient compartment-like syndrome	99109	Persistent left SVC connecting to the left-sided atrium	319646	PGM1-CDG
79136	Periodic vestibulocerebellar ataxia	2856	Persistent Müllerian derivatives	443811	PGM3-CDG
139426	Perioral myoclonia with absences	2856	Persistent Müllerian duct syndrome	443811	PGM3-related congenital disorder of glycosylation
563	Peripartum cardiomyopathy	706	Persistent patency of the arterial duct	251962	PGNT
163746	Peripheral demyelinating neuropathy - central dysmyelinating leukodystrophy - Waardenburg syndrome - Hirschsprung disease	97341	Persistent placoid maculopathy	1214	PHA
1795	Peripheral dysostosis	300324	Persistent polyclonal B-cell lymphocytosis	757	PHA2
252164	Peripheral fibroblastoma	300324	Persistent polyclonal B-cell lymphocytosis with binucleated lymphocytes	88938	PHA2A
2400	Peripheral motor neuropathy - dysautonomia			88939	PHA2B
84142	Peripheral nerve hyperexcitability			88940	PHA2C

→ 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
2874	Phakomatosis pigmentokeratotica	79318	Phosphomannomutase 2 deficiency	447961	Pigmentation defects-palmoplantar keratoderma-skin carcinoma syndrome
2875	Phakomatosis pigmentovascularis	79319	Phosphomannose isomerase deficiency	→16856 9	Pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome
79483	Phakomatosis pigmentovascularis type 2	3222	Phosphoribosylpyrophosphate synthetase superactivity	251295	Pigmented paravenous retinochoroidal atrophy
79485	Phakomatosis pigmentovascularis type 3	284417	Phosphoserine aminotransferase deficiency	66627	Pigmented villonodular synovitis
79484	Phakomatosis pigmentovascularis type 5	166409	Photosensitive epilepsy	169	Pili annulati
79485	Phakomatosis spilorosea	91495	PHPV	720	Pili bifurcati
352636	Phalangeal microgeodic syndrome	30924	PHSH	79492	Pili gemini
352636	Phalangeal osteolysis	180261	Phyllode tumor	79492	Pili multigemini
171848	PHARC syndrome	180261	Phylloide tumor	2889	Pili torti
231426	Pharyngeal-cervical-brachial variant of Guillain-Barré syndrome	773	Phytanic-CoA hydroxylase deficiency	2891	Pili torti - developmental delay - neurological abnormalities
231426	Pharyngeal-cervical-brachial weakness	2882	Phytosterolemia	2890	Pili torti - onychodysplasia
231426	Pharyngo-cervico-brachial variant of GBS	→33364	PIBIDS syndrome	1410	Pili trianguli et canaliculi
231426	Pharyngo-cervico-brachial variant of Guillain-Barré syndrome	505	Piccardi-Lassueur-Little syndrome	2741	Pillay syndrome
2876	PHAVER syndrome	2885	Piebald trait - neurologic defects	251612	Pilocytic astrocytoma
228410	PHD syndrome	2884	Piebaldism	2892	Pilodental dysplasia - refractive errors
48652	Phelan-McDermid syndrome	→1263	Piepkorn dysplasia	91414	Pilomatricoma
1919	Phenobarbital embryopathy	1566	Pierquin syndrome	228379	Pilomatrix dysplasia
84064	Phenotypic diarrhea	2886	Pierre Robin sequence - congenital heart defect - talipes	91414	Pilomatrixoma
716	Phenylalanine hydroxylase deficiency	2888	Pierre Robin sequence - faciodigital anomaly	251615	Pilomyxoid astrocytoma
716	Phenylketonuria	3450	Pierre Robin sequence - fetal chondrodysplasia	2894	Pilotto syndrome
226	Phenylketonuria type 2	1388	Pierre Robin sequence - hyperphalangy - clinodactyly	251919	Pineal parenchymal tumor of intermediate differentiation
2209	Phenylketonuric embryopathy	3104	Pierre Robin sequence - oligodactyly	251909	Pineoblastoma
1912	Phenytoin embryofetopathy	2886	Pierre Robin syndrome - congenital heart defect - talipes	251912	Pineocytoma
414750	Phenytoin or carbamazepine toxicity	2888	Pierre Robin syndrome - faciodigital anomaly	49382	Pingelapse blindness
→16856 9	PHID	3450	Pierre Robin syndrome - fetal chondrodysplasia	3353	Pinheiro-Freire Maia-Miranda syndrome
75508	Phlebectatic osteohypoplastic angiodyplasia	1388	Pierre Robin syndrome - hyperphalangy - clinodactyly	247165	Pink disease
69084	PHINED	2670	Pierson syndrome	155838	Pinnae fistula or cyst
294975	Phocomelia	398147	PIFP	→2510	Pinsky-Di George-Harley syndrome
2878	Phocomelia - ectrodactyly - deafness - sinus arrhythmia	217557	PIG	279904	PIOL
3439	Phocomelia - thrombocytopenia - encephalocele - urogenital malformations	99908	Pigeon-breeder lung disease	→79189	Pipecolic acidemia
2879	Phocomelia, Schinzel type	3474	PIGL-CDG	2896	Pitt-Hopkins syndrome
534	Phosphatidylinositol 4,5-biphosphate 5-phosphatase deficiency	83639	PIGM-CDG	221150	Pitt-Hopkins-like syndrome
79316	Phosphoenolpyruvate carboxykinase 1 deficiency	978	Pigment anomaly - ectrodactyly - hypodontia	→280	Pitt-Rogers-Danks syndrome
2880	Phosphoenolpyruvate carboxykinase deficiency	999	Pigmentary disorder with hearing loss	93395	Pitt-Williams brachydactyly
436	Phosphoethanolaminuria	64755	Pigmentary hairy epidermal nevus	251623	Pituicytoma
→31964 6	Phosphoglucomutase 1 deficiency	435	Pigmentary mosaicism, Ito type	95613	Pituitary apoplexy
35069	Phospholipase A2-associated neurodegeneration	313808	Pigmentary orthochromatic leukodystrophy	300385	Pituitary carcinoma
		→193	Pigmentary retinopathy - intellectual disability	96253	Pituitary corticotroph micro-adenoma
				91354	Pituitary deficiency due to empty sella turcica syndrome
				91350	Pituitary deficiency due to Rathke's pouch cysts
				91351	Pituitary dermoid and epidermoid cysts
				99725	Pituitary gigantism
				2965	Pituitary lactotrophic adenoma

→ 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
95496	Pituitary stalk interruption syndrome	284343	Pleuro-pulmonary blastoma family tumor susceptibility syndrome	2908	Poikiloderma of Kindler
91347	Pituitary thyrotrophic adenoma	64742	Pleuropulmonary blastoma	2909	Poikiloderma of Rothmund-Thomson
96253	Pituitary-dependent Cushing syndrome	284343	Pleuropulmonary blastoma family tumor susceptibility syndrome	221008	Poikiloderma of Rothmund-Thomson type 1
2897	Pityriasis rubra pilaris	99933	Pleuropulmonary blastoma type 1	221016	Poikiloderma of Rothmund-Thomson type 2
1078	Piussan-Lenaerts-Mathieu syndrome	99934	Pleuropulmonary blastoma type 2	221046	Poikiloderma with neutropenia
2869	PJS	99935	Pleuropulmonary blastoma type 3	221046	Poikiloderma with neutropenia, Clericuzio type
157850	PKAN	280356	PLIN1-related familial partial lipodystrophy	279947	POIS
216873	PKAN, atypical form	280356	PLIN1-related FPLD	130	Pokkuri death syndrome
216866	PKAN, classic form	2770	PLO-SL	2911	Poland anomaly
238455	PKDYS	2770	PLOSL	2911	Poland sequence
716	PKU	2375	Plott syndrome	2911	Poland syndrome
226	PKU type 2	280234	PLP1 null syndrome	313808	POLD
199351	PLA2G6-related dystonia-parkinsonism	678	PLS	2912	Poliomyelitis
439167	Placental insufficiency	35689	PLS	330009	Poliomyelitis in patients with immunodeficiencies deemed at risk
99928	Placental site trophoblastic tumor	99969	PLS	→33364	Pollitt syndrome
444138	PLACK syndrome	85166	PLSD-T	11	Poly-X
707	Plague	330015	Plumbism	29207	Polyarteritis enterica
300359	PLAID	54028	Plummer-Vinson syndrome	767	Polyarteritis nodosa
79141	Plamoplantar hyperkeratosis nummularis	732	PM	85435	Polyarthritis with rheumatoid factor
79141	Plamoplantar keratoderma nummularis	764	PM	85408	Polyarthritis without rheumatoid factor
35069	PLAN	702	PMD	247854	Polyarthritis without rheumatoid factor with anti-nuclear antibodies
199251	Plantar fibromatosis	2856	PMDS	247861	Polyarthritis without rheumatoid factor without anti-nuclear antibodies
251515	Plantar flexion contracture	308	PME type 1	450322	Polyclonal hyperviscosity syndrome
158769	Plaque-form urticaria pigmentosa	501	PME type 2	2770	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
29073	Plasma cell myeloma	263516	PME type 3	2795	Polycystic ovaries - urethral sphincter dysfunction
329	Plasma thromboplastin antecedent deficiency	402082	PME type 5	729	Polycythemia rubra vera
289666	Plasmablastic lymphoma	280620	PME type 6	729	Polycythemia vera
86855	Plasmacytoma	435438	PME type 7	2754	Polydactyly - cleft lip/palate - psychomotor retardation
722	Plasminogen deficiency type 1	424027	PME type 8	93339	Polydactyly of a biphalangeal thumb
439881	Plastic bronchitis	352596	PMED	295146	Polydactyly of a biphalangeal thumb, bilateral
721	Platelet alpha-granule deficiency	280270	PMLD	295144	Polydactyly of a biphalangeal thumb, unilateral
79434	Platinum oculocutaneous albinism	280282	PMLD1	93336	Polydactyly of a triphalangeal thumb
85166	Platyspondylic dysplasia, Torrance type	79318	PMM2-CDG	295150	Polydactyly of a triphalangeal thumb, bilateral
85166	Platyspondylic dysplasia, Torrance-Luton type	26790	PMP	295148	Polydactyly of a triphalangeal thumb, unilateral
85166	Platyspondylic lethal skeletal dysplasia, Torrance type	99885	PNDM	93337	Polydactyly of an index finger
2899	Platyspondyly - amelogenesis imperfecta	64741	Pneumoblastoma	295154	Polydactyly of an index finger, bilateral
300359	PLCG2-associated antibody deficiency and immune dysregulation	55655	Pneumococcal meningitis	295152	Polydactyly of an index finger, unilateral
137810	PLCNA	723	Pneumocystosis		
99969	Pleomorphic liposarcoma	90066	Pneumonia caused by Pseudomonas aeruginosa infection		
293199	Pleomorphic rhabdomyosarcoma	447	PNH		
251607	Pleomorphic xanthoastrocytoma	760	PNP deficiency		
449266	Pleural empyema	760	PNPase deficiency		
50251	Pleural mesothelioma	79096	PNPO deficiency		
99131	Pleuro-pericardial cyst	79096	PNPO-related neonatal epileptic encephalopathy		
		246	POADS		
		2905	POEMS syndrome		
		2825	Poikiloderma - alopecia - retrognathism - cleft palate		

→ 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
2919	Polydactyly postaxial with median cleft of upper lip	99748	Pontiac fever	2703	Port-wine nevi - mega cisterna magna - hydrocephalus
2917	Polydactyly-myopia syndrome	269229	Pontine tegmental cap dysplasia	854	Portal vein thrombosis
180229	Polyembryoma	284339	Pontocerebellar hypoplasia - 46,XY disorder of sex development	137839	Postanginal sepsis secondary to oropharyngeal infection
93308	Polyepiphyseal dysplasia type 1	324569	Pontocerebellar hypoplasia due to CHMP1A mutation	246	Postaxial acrodysostosis
93307	Polyepiphyseal dysplasia type 4	2254	Pontocerebellar hypoplasia type 1	246	Postaxial acrofacial dysostosis
93311	Polyepiphyseal dysplasia type 5	2524	Pontocerebellar hypoplasia type 2	2916	Postaxial polydactyly - dental and vertebral anomalies
397937	Polyglucosan body myopathy	97249	Pontocerebellar hypoplasia type 3	2920	Postaxial polydactyly - intellectual disability
180182	Polymastia	166063	Pontocerebellar hypoplasia type 4	295008	Postaxial polydactyly of foot
447877	Polymerase proofreading-associated adenomatous polyposis	166068	Pontocerebellar hypoplasia type 5	295008	Postaxial polydactyly of toes
2925	Polymicrogyria - turricephaly - hypogenitalism	166073	Pontocerebellar hypoplasia type 6	295181	Postaxial polydactyly of toes, bilateral
300573	Polymicrogyria due to TUBB2B mutation	284339	Pontocerebellar hypoplasia type 7	295179	Postaxial polydactyly of toes, unilateral
250972	Polymicrogyria with optic nerve hypoplasia	324569	Pontocerebellar hypoplasia type 8	93334	Postaxial polydactyly type A
64745	Polymorphic eruption of pregnancy	369920	Pontocerebellar hypoplasia type 9	295165	Postaxial polydactyly type A, bilateral
1243	Polymorphic vitelline macular degeneration	411493	Pontocerebellar hypoplasia type 10	295163	Postaxial polydactyly type A, unilateral
93569	Polymyalgia rheumatica	213777	Poorly differentiated endocrine carcinoma of the cervix uteri	93335	Postaxial polydactyly type B
732	Polymyositis	213731	Poorly differentiated endocrine carcinoma of the corpus uteri	295169	Postaxial polydactyly type B, bilateral
2905	Polyneuropathy - endocrinopathy - plasma cell dyscrasia	213777	Poorly differentiated endocrine cervical carcinoma	295167	Postaxial polydactyly type B, unilateral
2926	Polyneuropathy - hand defect	284400	Poorly differentiated neuroendocrine carcinoma of the bladder	420584	Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome
171848	Polyneuropathy - hearing loss - ataxia - retinitis pigmentosa - cataract	263339	Poorly differentiated thymic neuroendocrine carcinoma	93406	Postaxial syndactyly with metacarpal synostosis
2928	Polyneuropathy - intellectual disability - acromicria - premature menopause	1300	Popliteal web syndrome	2730	Postaxial tetramelic oligodactyly
639	Polyneuropathy associated with IgM monoclonal gammopathy with anti-MAG	95699	POR deficiency	263352	Postcardiotomy right ventricular failure
93276	Polyostotic fibrous dysplasia	666	Porak and Durante disease	97349	Postencephalitic parkinsonism
160148	Polypoid prolapsing folds	95699	PORD	98971	Posterior amorphous corneal dystrophy
2869	Polyps and spots syndrome	2940	Porencephaly	98971	Posterior amorphous stromal dystrophy
208981	Polyradiculoneuropathy associated with IgG/IgA/IgM monoclonal gammopathy without known antibodies	2941	Porencephaly - cerebellar hypoplasia - internal malformations	88628	Posterior column ataxia - retinitis pigmentosa
141091	Polyrhinia	306547	Porencephaly-microcephaly-bilateral congenital cataract syndrome	54247	Posterior cortical atrophy
141091	Polyrrhinia	735	Porokeratosis of Mibelli	2064	Posterior fusion of lumbosacral vertebrae - blepharoptosis
93338	Polysyndactyly	737	Porokeratosis plantaris palmaris et disseminata	95706	Posterior hypospadias
2934	Polysyndactyly - cardiac malformation	166286	Porokeratotic eccrine nevus	268810	Posterior meningocele
295161	Polysyndactyly, bilateral	166286	Porokeratotic eccrine ostial and dermal duct nevus	98973	Posterior polymorphous corneal dystrophy
93405	Polysyndactyly, Haas type	101330	Porphyria cutanea tarda	98973	Posterior polymorphous dystrophy
295159	Polysyndactyly, unilateral	443057	Porphyria cutanea tarda type I	93110	Posterior urethral valve
228410	Polyvalvular heart disease syndrome	443062	Porphyria cutanea tarda type II	48435	Postinfectious vasculitis
139426	POMA	100924	Porphyria due to ALA dehydratase deficiency	216452	Postlingual non-syndromic genetic deafness
1183	POMA syndrome	100924	Porphyria due to ALAD deficiency	279947	Postorgasmic illness syndrome
71526	POMC deficiency		Porphyria due to delta-aminolevulinate dehydratase deficiency	563	Postpartum cardiomyopathy
365	Pompe disease	100924	Porphyria of Doss	443173	Postpartum psychosis
308552	Pompe disease, infantile onset	79473	Porphyria variegata		
420429	Pompe disease, late onset				

→ 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
2942	Postpolio sequelae	308013	PPKP3 without elastoidosis	295154	Preaxial polydactyly type 3, bilateral
2942	Postpolio syndrome	3077	PPM-X	295152	Preaxial polydactyly type 3, unilateral
2942	Postpoliomyelic syndrome	189439	PPNAD	93338	Preaxial polydactyly type 4
2942	Postpoliomyelitis sequelae	370348	PPNET	295161	Preaxial polydactyly type 4, bilateral
2942	Postpoliomyelitis syndrome	97278	PPoma	295159	Preaxial polydactyly type 4, unilateral
98913	Postsynaptic congenital myasthenic syndromes	163927	PPP	1309	Precalicial canalicular ectasia
		308013	PPPK3 without elastoidosis	99860	Precursor B-cell acute lymphoblastic leukemia
163921	Posttransplant acute limbic encephalitis	79502	PPPP	99860	Precursor B-cell acute lymphoblastic leukemia/lymphoma
70568	Posttransplant lymphoproliferative disease	251295	PPRCA	99860	Precursor B-cell acute lymphocytic leukemia
443236	Postural tachycardia syndrome due to NET deficiency	398980	PPSPC	99860	Precursor B-cell acute lymphocytic leukemia/lymphoma
238606	POT	324977	PRAAS	99860	Precursor B-cell acute lymphocytic leukemia/lymphoma
→682	Potassium-sensitive normokalemic periodic paralysis	739	Prader-Labhart-Willi syndrome	99861	Precursor T-cell acute lymphoblastic leukemia
640	Potato-grubbing palsy	3409	Prader-Willi habitus - osteopenia - camptodactyly	99861	Precursor T-cell acute lymphoblastic leukemia/lymphoma
1713	Potocki-Lupski syndrome	739	Prader-Willi syndrome	99861	Precursor T-cell acute lymphocytic leukemia
52022	Potocki-Shaffer syndrome	177910	Prader-Willi syndrome due to imprinting mutation	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
3316	Potter sequence - cleft lip/palate - cardiopathy	98754	Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15	99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
217067	Pouchitis	98793	Prader-Willi syndrome due to paternal 15q11q13 deletion	275555	Preeclampsia
2876	Powell-Chandra-Saal syndrome	177901	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1	69665	Pregnancy-related cholestasis
2201	Powell-Venecie-Gordon syndrome	177904	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2	216445	Prelingual non-syndromic genetic deafness
314566	PPAO	398069	Prader-Willi syndrome due to point mutation	276432	Premature aging appearance-developmental delay-cardiac arrhythmia syndrome
447877	PPAP	177907	Prader-Willi syndrome due to translocation	363665	Premature aging syndrome, Pentinen type
79083	PPARG-related familial partial lipodystrophy	398073	Prader-Willi-like syndrome	52183	Premature chromosome condensation with microcephaly and intellectual disability
79083	PPARG-related FPLD	171829	Prader-Willi-like syndrome due to deletion 6q16	95486	Premature closure of the arterial duct
284343	PPB family tumor susceptibility syndrome	398079	Prader-Willi-like syndrome due to point mutation	95486	Premature closure of the patent ductus arteriosus
284343	PPBFTDS	245	Preaxial acrodysostosis	2114	Premature degenerative osteoarthropathy of the hip
300324	PPBL	2957	Preaxial deficiency - postaxial polydactyly - hypospadias	247638	Prenatal benign hypophosphatasia
168829	PPC	2921	Preaxial polydactyly - colobomata - intellectual disability	247638	Prenatal benign phosphoethanolaminuria
98973	PPCD	295006	Preaxial polydactyly of foot	247638	Prenatal benign Rathburn disease
93339	PPD1	295006	Preaxial polydactyly of toes	90160	Pressure-induced localized lipoatrophy
93336	PPD2	295177	Preaxial polydactyly of toes, bilateral	98914	Presynaptic congenital myasthenic syndromes
93337	PPD3	295175	Preaxial polydactyly of toes, unilateral	79410	Pretibial DEB
93338	PPD4	93339	Preaxial polydactyly type 1	79410	Pretibial dystrophic epidermolysis bullosa
411696	PPI-REE	295146	Preaxial polydactyly type 1, bilateral	2958	Prieto-Badia-Mulas syndrome
411696	PPI-responsive esophageal eosinophilia	295144	Preaxial polydactyly type 1, unilateral	1451	Prieur-Griscelli syndrome
411696	PPIRee	93336	Preaxial polydactyly type 2	930	Primary achalasia
494	PPK mutilans and deafness	295150	Preaxial polydactyly type 2, bilateral	75564	Primary acquired sideroblastic anemia
79141	PPK nummularis	295148	Preaxial polydactyly type 2, unilateral		
86923	PPK, Gamburg-Nielsen type	93337	Preaxial polydactyly type 3		
140966	PPK, Nagashima type				
1010	PPK-CA, Stevanovic type				
1366	PPK-CA, Wallis type				
2202	PPK-deafness syndrome				
79501	PPKP1				
79502	PPKP2				
38	PPKP3				

→ 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
85138	Primary Addison's disease	98807	Primary dystonia, DYT13 type	73272	Primary insulin-like growth factor deficiency
85443	Primary amyloidosis	370103	Primary dystonia, DYT17 type	90362	Primary intestinal lymphangiectasia
228272	Primary anetoderma	306734	Primary dystonia, DYT21 type	279904	Primary intraocular lymphoma
140989	Primary angiitis of the central nervous system	48686	Primary effusion lymphoma	279904	Primary intraocular non-Hodgkin's lymphoma
		90026	Primary erythermalgia	140436	Primary intraosseous vascular malformation
1572	Primary antibody deficiency	357220	Primary essential cutis verticis gyrata	137926	Primary laryngeal lymphangioma
2285	Primary basilar invagination	412206	Primary failure of tooth eruption	35689	Primary lateral sclerosis
189427	Primary bilateral macronodular adrenal hyperplasia	98957	Primary familial amyloidosis of the cornea	314709	Primary localized amyloidosis
186	Primary biliary cirrhosis	90042	Primary familial and congenital polycythemia	137810	Primary localized cutaneous nodular amyloidosis
779	Primary biliary cirrhosis and systemic scleroderma	90042	Primary familial polycythemia	319667	Primary lymphoid conjunctival tumor
314684	Primary bone lymphoma	3337	Primary Fanconi renotubular syndrome	319667	Primary lymphoma of the conjunctiva
46135	Primary brain lymphoma	3337	Primary Fanconi syndrome	228272	Primary macular atrophy
300865	Primary C-ALCL	633	Primary GH insensitivity	168811	Primary malignant peritoneal mesothelioma
267	Primary calpainopathy	633	Primary GH resistance	98838	Primary mediastinal clear cell lymphoma of B-cell type
169464	Primary CD59 deficiency	633	Primary growth hormone insensitivity	98838	Primary mediastinal large B-cell lymphoma
46135	Primary central nervous system lymphoma	633	Primary growth hormone resistance	238642	Primary megaureter, adult-onset form
140989	Primary central nervous system vasculitis	100085	Primary hepatic carcinoid tumor	252050	Primary melanoma of the central nervous system
244	Primary ciliary dyskinesia	100085	Primary hepatic neuroendocrine carcinoma	54370	Primary membranoproliferative glomerulonephritis
247522	Primary ciliary dyskinesia - retinitis pigmentosa	314950	Primary HES	306558	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome
→244	Primary ciliary dyskinesia, Kartagener type	314950	Primary hypereosinophilic syndrome	391408	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome
46135	Primary CNS lymphoma	2232	Primary hypergonadotropic hypogonadism - partial alopecia	824	Primary myelofibrosis
90042	Primary congenital erythrocytosis	682	Primary hyperkalemic periodic paralysis	357225	Primary non-essential cutis verticis gyrata
98976	Primary congenital glaucoma	416	Primary hyperoxaluria	289356	Primary non-gestational choriocarcinoma of ovary
91138	Primary cryoglobulinemia	93598	Primary hyperoxaluria type 1	289356	Primary non-gestational ovarian choriocarcinoma
178528	Primary cutaneous aggressive epidermotropic CD8+ T-cell lymphoma	93599	Primary hyperoxaluria type 2	279897	Primary oculocerebral lymphoma
300865	Primary cutaneous anaplastic large cell lymphoma	93600	Primary hyperoxaluria type 3	279897	Primary oculocerebral non-Hodgkin lymphoma
178522	Primary cutaneous CD4+ small/medium-sized pleomorphic T-cell lymphoma	682	Primary hyperPP	238606	Primary orthostatic tremor
		33208	Primary hypersomnia	439737	Primary PAN
178544	Primary cutaneous diffuse large B-cell lymphoma, leg type	1572	Primary hypogammaglobulinemia	99878	Primary parathyroid hyperplasia
178528	Primary cutaneous epidermotropic cytotoxic CD8+ T-cell lymphoma	30924	Primary hypomagnesemia with secondary hypocalcemia	875	Primary pediatric cardiac tumor
178540	Primary cutaneous follicle center lymphoma	75391	Primary immunodeficiency due to MCM4 deficiency	439737	Primary periarthritis nodosa
178533	Primary cutaneous gamma/delta-positive T-cell lymphoma	431166	Primary immunodeficiency due to STAT2 deficiency	168829	Primary peritoneal carcinoma
178536	Primary cutaneous marginal zone B-cell lymphoma	90023	Primary immunodeficiency syndrome due to p14 deficiency	168829	Primary peritoneal serous carcinoma
451602	Primary cutaneous plasmacytosis	90023	Primary immunodeficiency syndrome with short stature	398980	Primary peritoneal serous/papillary carcinoma
86885	Primary cutaneous unspecified peripheral T-cell lymphoma	431166	Primary immunodeficiency with disseminated vaccine-strain measles	189439	Primary pigmented nodular adrenocortical disease
98807	Primary dystonia with mixed phenotype	447731	Primary immunodeficiency with multifaceted aberrant lymphoid immunity		
99657	Primary dystonia, DYT2 type	75391	Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency		
98805	Primary dystonia, DYT4 type				
98806	Primary dystonia, DYT6 type				

→ 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
100021	Primary plasmacytoma of the bone	300382	Progeroid and marfanoid aspect-lipodystrophy syndrome	352447	Progressive external ophthalmoplegia - myopathy - emaciation
439737	Primary polyarteritis nodosa	435953	Progeroid features-hepatocellular carcinoma predisposition syndrome	2744	Progressive external ophthalmoplegia and scoliosis
314566	Primary progressive apraxia of speech	2962	Progeroid syndrome, De Barys type	1214	Progressive facial hemiatrophy
75567	Primary progressive freezing gait	2963	Progeroid syndrome, Petty type	172	Progressive familial intrahepatic cholestasis
275766	Primary pulmonary arterial hypertension	79094	Progressive arterial occlusive disease - hypertension - heart defects - bone fragility - brachysyndactyly	79306	Progressive familial intrahepatic cholestasis type 1
2420	Primary pulmonary lymphoma	448251	Progressive autosomal recessive ataxia-deafness syndrome	79304	Progressive familial intrahepatic cholestasis type 2
358	Primary renal tubular hypokalemic hypomagnesemia with hypocalciuria	448251	Progressive autosomal recessive ataxia-sensorineural hearing loss syndrome	79305	Progressive familial intrahepatic cholestasis type 3
412206	Primary retention of teeth	75373	Progressive bifocal chorioretinal atrophy	75327	Progressive foveal dystrophy
171	Primary sclerosing cholangitis	→97229	Progressive bulbar palsy of childhood	1214	Progressive hemifacial atrophy
99856	Primary syringomyelia	→97229	Progressive bulbar paralysis of childhood	199282	Progressive isolated segmental anhidrosis
98841	Primary systemic ALCL	139447	Progressive cavitating leukoencephalopathy	73	Progressive massive osteolysis
314701	Primary systemic amyloidosis	79087	Progressive cephalothoracic lipodystrophy	217260	Progressive multifocal leukoencephalitis
268861	Primary tethered chord syndrome	247198	Progressive cerebello-cerebral atrophy	217260	Progressive multifocal leukoencephalopathy
268861	Primary tethered spinal cord syndrome	1871	Progressive cone dystrophy	424027	Progressive myoclonic epilepsy due to CERS1 deficiency
99867	Primary thymic epithelial neoplasm	220393	Progressive cutaneous systemic scleroderma	263516	Progressive myoclonic epilepsy due to KCTD7 deficiency
263310	Primary thymic epithelial neoplasm type A	220393	Progressive cutaneous systemic sclerosis	435438	Progressive myoclonic epilepsy due to KV3.1 deficiency
263324	Primary thymic epithelial neoplasm type AB	3235	Progressive deafness with stapes fixation	308	Progressive myoclonic epilepsy type 1
263317	Primary thymic epithelial neoplasm type B	216812	Progressive deforming osteogenesis imperfecta	501	Progressive myoclonic epilepsy type 2
99867	Primary thymic epithelial tumor	217396	Progressive demyelinating neuropathy with bilateral striatal necrosis	263516	Progressive myoclonic epilepsy type 3
263310	Primary thymic epithelial tumor type A	1328	Progressive diaphyseal dysplasia	163696	Progressive myoclonic epilepsy type 4
263324	Primary thymic epithelial tumor type AB	495	Progressive diffuse palmoplantar keratoderma	402082	Progressive myoclonic epilepsy type 5
263317	Primary thymic epithelial tumor type B	495	Progressive diffuse PPK	280620	Progressive myoclonic epilepsy type 6
98807	Primary torsion dystonia with predominant craniocervical or upper limb onset	438266	Progressive encephalomyelitis with rigidity and myoclonus	435438	Progressive myoclonic epilepsy type 7
231580	Primary unilateral adrenal hyperplasia	2836	Progressive encephalopathy - optic atrophy	424027	Progressive myoclonic epilepsy type 8
140989	Primary vasculitis of the central nervous system	2836	Progressive encephalopathy with edema, hypsarrhythmia and optic atrophy	352596	Progressive myoclonic epilepsy with dystonia
3033	Primitive renal tubule syndrome	431361	Progressive encephalopathy with leukodystrophy due to DECR deficiency	308	Progressive myoclonus epilepsy type 1
2636	Primordial microcephalic dwarfism, Crachami type	99852	Progressive encephalopathy with severe infantile anorexia	501	Progressive myoclonus epilepsy type 2
→2637	Primordial short stature - microdontia - opalescent and rootless teeth	1947	Progressive epilepsy - intellectual disability, Finnish type	263516	Progressive myoclonus epilepsy type 3
3042	Primrose syndrome			402082	Progressive myoclonus epilepsy type 5
412066	PRKAR1B-related neurodegenerative dementia with intermediate filaments			280620	Progressive myoclonus epilepsy type 6
2965	PRL-secreting pituitary adenoma			435438	Progressive myoclonus epilepsy type 7
2965	PRLOma			424027	Progressive myoclonus epilepsy type 8
326	Proaccelerin deficiency			352596	Progressive myoclonus epilepsy with dystonia
141099	Proboscis lateralis				
740	Progeria				
2959	Progeria - short stature - pigmented nevi				
99706	Progeria-associated arthropathy				

→ 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
726	Progressive neuronal degeneration of childhood with liver disease	2966	Properdin deficiency	89843	Pruriginous dystrophic epidermolysis bullosa
228012	Progressive neurosensory deafness - hypertrophic cardiomyopathy	35	Propionic acidemia	64745	Pruritic urticarial papules and plaques of pregnancy
228012	Progressive neurosensory hearing loss - hypertrophic cardiomyopathy	35	Propionic aciduria	284417	PSAT deficiency
158022	Progressive nodular histiocytosis	324977	Proteasome disability syndrome	171	PSC
100070	Progressive non-fluent aphasia	324977	Proteasome-associated autoinflammatory syndrome	228402	Pseudo-Angelman syndrome
2062	Progressive non-infectious anterior vertebral fusion	213	Protein defect of cystin transport	99000	Pseudo-Best disease
2762	Progressive osseous heteroplasia	2967	Protein R deficiency	314459	Pseudo-Demons-Meigs syndrome
3322	Progressive pancytopenia - immunodeficiency - cerebellar hypoplasia	26349	Protein S acquired deficiency	577	Pseudo-Hurler polydystrophy
1159	Progressive pseudorheumatoid arthropathy of childhood	744	Proteus syndrome	314459	Pseudo-Meigs syndrome
352718	Progressive retinal dystrophy due to retinol transport defect	2969	Proteus-like syndrome	439881	Pseudo-membranous bronchitis
447977	Progressive scapulohumeral peroneal distal myopathy	325	Prothrombin deficiency	263482	Pseudo-Morquio syndrome type 2
228012	Progressive sensorineural deafness - hypertrophic cardiomyopathy	411696	Proton-pump inhibitor-responsive esophageal eosinophilia	2971	Pseudo-NALD
228012	Progressive sensorineural hearing loss - hypertrophic cardiomyopathy	251598	Protoplasmic astrocytoma	2971	Pseudo-neonatal adrenoleukodystrophy
683	Progressive supranuclear palsy	79473	Protoporphyrinogen oxidase deficiency	1229	Pseudo-TORCH syndrome
240112	Progressive supranuclear palsy - apraxia of speech	2508	Proud-Levine-Carpenter syndrome	2166	Pseudo-trisomy 13 syndrome
240103	Progressive supranuclear palsy - corticobasal syndrome	52022	Proximal 11p deletion syndrome	99000	Pseudo-vitelliform macular dystrophy
240085	Progressive supranuclear palsy - parkinsonism	261197	Proximal 16p11.2 microdeletion syndrome	52530	Pseudo-von Willebrand disease
240112	Progressive supranuclear palsy - progressive non fluent aphasia	370079	Proximal 16p11.2 microduplication syndrome	52530	Pseudo-von Willebrand disease type 2B
240094	Progressive supranuclear palsy - pure akinesia with gait freezing	261197	Proximal del(16)(p11.2)	→300	Pseudo-Zellweger syndrome
316	Progressive symmetric erythrokeratoderma	370079	Proximal dup(16)(p11.2)	750	Pseudoachondroplasia
316	Progressive symmetric erythrokeratoderma, Gottron type	2019	Proximal focal femoral deficiency	750	Pseudoachondroplastic dysplasia
2965	Prolactin-secreting pituitary adenoma	261197	Proximal monosomy 16p11.2	750	Pseudoachondroplastic spondyloepiphyseal dysplasia
2965	Prolactinoma	401768	Proximal myopathy with extrapyramidal signs	2971	Pseudoadrenoleukodystrophy
742	Polidase deficiency	606	Proximal myotonic dystrophy	526	Pseudoaldosteronism
492	Proliferating trichilemmal cyst	606	Proximal myotonic myopathy	221120	Pseudoaminopterin syndrome
86872	Proliferation of large granular lymphocytes	47159	Proximal renal tubular acidosis	85174	Pseudodiastrophic dysplasia
221126	Proliferative vasculopathy and hydranencephaly/hydrocephaly	93607	Proximal renal tubular acidosis with ocular abnormalities and intellectual disability	2983	Pseudohermaphroditism - intellectual disability
419	Proline oxidase deficiency	70	Proximal spinal muscular atrophy	526	Pseudohyperaldosteronism type 1
75374	Prolonged electroretinal response suppression	83330	Proximal spinal muscular atrophy type 1	88660	Pseudohyperaldosteronism type 2
300878	Polymphocytic variant of hairy cell leukemia	83418	Proximal spinal muscular atrophy type 2	756	Pseudohypoaldosteronism type 1
300878	Polymphocytic variant of HCL	83419	Proximal spinal muscular atrophy type 3	757	Pseudohypoaldosteronism type 2
2083	Prominent glabella - microcephaly - hypogenitalism	83420	Proximal spinal muscular atrophy type 4	88938	Pseudohypoaldosteronism type 2A
		370079	Proximal symphalangism	88939	Pseudohypoaldosteronism type 2B
		3390	Proximal trisomy 16p11.2	88940	Pseudohypoaldosteronism type 2C
		3390	Proximal tubulopathy - diabetes mellitus - cerebellar ataxia	300525	Pseudohypoaldosteronism type 2D
		3222	PRPP synthetase superactivity	300530	Pseudohypoaldosteronism type 2E
		3222	PRPS1 superactivity	79443	Pseudohypoparathyroidism type 1A
		47159	pRTA	94089	Pseudohypoparathyroidism type 1B
		2970	Prune belly syndrome	79444	Pseudohypoparathyroidism type 1C
				94090	Pseudohypoparathyroidism type 2
				2976	Pseudoleprechaunism syndrome, Patterson type
				26790	Pseudomyxoma peritonei
				251962	Pseudopapillary ganglioglioneurocytoma
				251962	Pseudopapillary neurocytoma with glial differentiation

→ 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
2980	Pseudopapilledema - blepharophimosis - hand anomalies	2999	Ptosis - strabismus - ectopic pupils	101206	Pulmonary valve agenesis - Fallot's tetralogy - absence of ductus arteriosus
129	Pseudopelade of Brocq	→29384 3	Ptosis - strabismus - rectus abdominis diastasis	99048	Pulmonary valve agenesis - ventricular septal defect - persistent ductus arteriosus
2985	Pseudoprogeria syndrome	238766	Ptosis - syndactyly - learning difficulties	31837	Pulmonary venoocclusive disease
79445	Pseudopseudohypoparathyroidism	228396	Ptosis - upper ocular movement limitation - absence of lacrimal punctum	85202	Pulmonic stenosis - brachytelephalangism - calcification of cartilages
3103	Pseudothalidomide syndrome	2997	Ptosis - vocal cord paralysis	→636	Pulmonic stenosis with 'café-au-lait' spots
2518	Pseudotoxoplasmosis syndrome	231580	PUAH	98984	Pulverulent cataract
238624	Pseudotumor cerebri	60039	Pudendal algia	97353	Punch-drunk syndrome
83316	Pseudotyphus of California	60039	Pudendal nerve entrapment syndrome	79502	Punctate palmoplantar hyperkeratosis type 2
180079	Pseudounicornuate uterus	60039	Pudendal neuralgia	38	Punctate palmoplantar hyperkeratosis type 3
753	Pseudovaginal perineoscrotal hypospadias	60039	Pudendal neuralgia by pudendal nerve entrapment	308013	Punctate palmoplantar hyperkeratosis type 3 without elastoidosis
289157	Pseudovitamin D-deficient rickets	60039	Pudendalgia	79501	Punctate palmoplantar keratoderma type 1
758	Pseudoxanthoma elasticum	443173	Puerperal psychosis	79502	Punctate palmoplantar keratoderma type 2
228293	Pseudoxanthoma elasticum-like papillary dermal elastocytosis	2038	Pulmonary arteriovenous aneurysm	38	Punctate palmoplantar keratoderma type 3
436274	Pseudoxanthoma elasticum-like skin manifestations with retinis pigmentosa	984	Pulmonary agenesis	308013	Punctate palmoplantar keratoderma type 3 without elastoidosis
91135	Pseudoxanthoma elasticum-like syndrome	60025	Pulmonary alveolar microlithiasis		
228227	Pseudoxanthoma-like late-onset focal dermal elastosis	440427	Pulmonary alveolar proteinosis, Reunion island type		
280794	Pseudoxanthomatous DCM	247257	Pulmonary anthrax		
280794	Pseudoxanthomatous diffuse cutaneous mastocytosis	→33117 6	Pulmonary arterial hypertension - leukopenia - atrial septal defect		
95496	PSIS	2038	Pulmonary arteriovenous fistula		
683	PSP	99049	Pulmonary artery coming from patent ductus arteriosus		
240112	PSP-AOS	99050	Pulmonary artery coming from the aorta	438213	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome
240103	PSP-CBS	99083	Pulmonary artery hypoplasia	438216	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation
240085	PSP-corticobasal syndrome	1208	Pulmonary atresia - intact ventricular septum	231625	Pure aldosterone-producing adrenocortical carcinoma
240094	PSP-p	1207	Pulmonary atresia with ventricular septal defect	231625	Pure aldosterone-secreting adrenocortical carcinoma
240094	PSP-PAGF	64741	Pulmonary blastoma	231625	Pure APAC
240085	PSP-parkinsonism	99084	Pulmonary branch stenosis	441	Pure autonomic failure
240112	PSP-PNFA	199241	Pulmonary capillary hemangiomatosis	441	Pure dysautonomia
240094	PSP-pure akinesia with gait freezing	210136	Pulmonary fibrosis - hepatic hyperplasia - bone marrow hypoplasia	319465	Pure familial acute myeloid leukemia
263548	PSS type A	217080	Pulmonary fungal infections in patients deemed at risk	319465	Pure familial AML
263553	PSS type B	99874	Pulmonary histiocytosis X	69084	Pure hair and nail ectodermal dysplasia
99928	PSST	991	Pulmonary hypoplasia - agonadism - dextrocardia - diaphragmatic hernia syndrome	441	Pure idiopathic dysautonomia
324636	Psychogenic purpura	217557	Pulmonary interstitial glycogenosis	475	Pure Joubert syndrome
88618	Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	2414	Pulmonary lymphangiomatosis	254854	Pure mitochondrial myopathy
52530	PT-VWD	60026	Pulmonary nodular lymphoid hyperplasia	2028	Puretic syndrome
329	PTA deficiency	411703	Pulmonary non-tuberculous mycobacterial infection	760	Purine nucleoside phosphorylase deficiency
247698	PTC syndrome	60026	Pulmonary pseudolymphoma	761	Purpura rheumatica
97290	PTC-RCC			2442	Purtilo syndrome
269229	PTCD			293173	Pustular drug eruption
2988	Pterygium colli - intellectual disability - digital anomalies				
2989	Pterygium of the conjunctiva, familial form				
86789	PTLAH				
70568	PTLD				

→ 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
163927	Pustulosis palmaris et plantaris	353320	Pyruvate carboxylase deficiency, benign type	93321	Radial hemimelia
48377	Pustulosis subcornealis	353308	Pyruvate carboxylase deficiency, infantile form	295071	Radial hemimelia, bilateral
93110	PUV	353314	Pyruvate carboxylase deficiency, severe neonatal type	295069	Radial hemimelia, unilateral
729	PV	79243	Pyruvate decarboxylase deficiency	2252	Radial hypoplasia - triphalangeal thumbs - hypospadias - maxillary diastema
101206	PVA/ADA, Fallot type	79244	Pyruvate dehydrogenase complex component E2 deficiency	93321	Radial longitudinal meromelia
99048	PVA/PDA, non-Fallot type	255182	Pyruvate dehydrogenase complex component E3 deficiency	295071	Radial longitudinal meromelia, bilateral
398069	PWS due to point mutation	765	Pyruvate dehydrogenase complex deficiency	295069	Radial longitudinal meromelia, unilateral
398073	PWS-like	79243	Pyruvate dehydrogenase complex E1 component subunit alpha deficiency	93321	Radial ray agenesis
398079	PWS-like due to point mutation	255138	Pyruvate dehydrogenase complex E1 component subunit beta deficiency	2307	Radial ray defects, hearing impairment, external ophthalmoplegia, and thrombocytopenia
251607	PXA	765	Pyruvate dehydrogenase E1-alpha deficiency	3026	Radial ray hypoplasia - choanal atresia
758	PXE	79243	Pyruvate dehydrogenase E1-beta deficiency	90021	Radiation myelitis
228227	PXE-like late-onset focal dermal elastosis	255138	Pyruvate dehydrogenase E2 deficiency	70475	Radiation proctitis
228293	PXE-like papillary dermal elastocytosis	79244	Pyruvate dehydrogenase E3 deficiency	99789	Radicular dentin dysplasia
91135	PXE-like syndrome	2394	Pyruvate dehydrogenase E3-binding protein deficiency	→2712	Radiculomegaly of canine teeth-congenital cataract
436274	PXE-like syndrome with retinis pigmentosa	255182	Pyruvate dehydrogenase phosphatase deficiency	3015	Radio-renal syndrome
763	Pycnodynostosis	79246	Pyruvate dehydrogenase protein X component deficiency	3269	Radio-ulnar fusion
293633	PYCR1 deficiency	766	Pyruvate kinase deficiency of erythrocytes	295219	Radio-ulnar fusion, bilateral
293633	PYCR1-related De Barsy syndrome	781	Q fever	295217	Radio-ulnar fusion, unilateral
3003	Pyknoachondrogenesis	3010	Qazi-Markouizos syndrome	3269	Radio-ulnar synostosis
763	Pyknodynostosis	602	Quadriceps-sparing myopathy	71289	Radio-ulnar synostosis - amegakaryocytic thrombocytopenia
64280	Pyknolepsy	781	Quadrilateral fever	3270	Radio-ulnar synostosis - intellectual disability - hypotonia
3005	Pyle disease	9	Quadruple X	→193	Radio-ulnar synostosis - retinal pigment abnormalities
48104	Pyoderma gangrenosum	84142	Quantal squander syndrome	295219	Radio-ulnar synostosis, bilateral
289478	Pyoderma gangrenosum - acne - suppurative hidradenitis	869	Quaternary A syndrome	295217	Radio-ulnar synostosis, unilateral
69126	Pyogenic arthritis - pyoderma gangrenosum - acne	220436	Quebec platelet disorder	294979	Radio-ulnar terminal transverse meromelia
183713	Pyogenic bacterial infections due to MyD88 deficiency	781	Query fever	295095	Radio-ulnar terminal transverse meromelia, bilateral
764	Pyomyositis	137888	Question mark ear syndrome	295093	Radio-ulnar terminal transverse meromelia, unilateral
2561	Pyramidal molar - glaucoma - upper abnormal lip	346	Quinquaud's folliculitis decalvans	420741	Radiosensitivity-immunodeficiency-dysmorphic features-learning difficulties syndrome
63440	Pyrgocephaly	261529	r(Y)	100057	RAE
79096	Pyridoxal phosphate-dependent seizures	100057	RAAS-blocker-induced angioedema	100019	RAEB-1
79096	Pyridoxal phosphate-responsive seizures	100057	RAAS-blocker-induced angioneurotic edema	100020	RAEB-2
79096	Pyridoxamine 5'-oxidase deficiency	770	Rabies	168960	RAEB-t
79096	Pyridoxamine 5'-phosphate oxidase deficiency	769	Rabson-Mendenhall syndrome	1832	Raine syndrome
3006	Pyridoxine-dependent epilepsy	240760	RAD50 deficiency	50811	Rajab-Spranger syndrome
32	Pyroglutamicaciduria	93321	Radial clubhand	268114	RALD
293633	Pyrroline-5-carboxylate reductase 1 deficiency	1121	Radial deficiency - tibial hypoplasia	240905	Raltegravir toxicity
3008	Pyruvate carboxylase deficiency	353308	Pyruvate carboxylase deficiency type A	99843	Rambam-Hasharon syndrome
353308	Pyruvate carboxylase deficiency type A	353314	Pyruvate carboxylase deficiency type B		
353314	Pyruvate carboxylase deficiency type B	353320	Pyruvate carboxylase deficiency type C		

→ 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
3018	Rambaud-Gallian syndrome	89842	RDEB, non-Hallopeau-Siemens type	169142	Recurrent infection due to specific granule deficiency
3018	Rambaud-Gallian-Touchard syndrome	89841	RDEB-Ce	251523	Recurrent infections - inflammatory syndrome due to zinc metabolism disorder
3019	Ramon syndrome	89842	RDEB-generalized other	183675	Recurrent infections associated with rare immunoglobulin isotypes deficiency
1051	Ramos-Arroyo syndrome	79409	RDEB-I	69665	Recurrent intrahepatic cholestasis of pregnancy
3020	Ramsay Hunt syndrome	89842	RDEB-O	169467	Recurrent Neisseria infections due to factor D deficiency
86861	Randall disease	79408	RDEB-sev gen	60032	Recurrent respiratory papillomatosis
3021	RAPADILINO syndrome	85445	Reactive amyloidosis	199267	Recurring digital fibrous tumor of childhood
293987	Rapid-onset childhood obesity - hypothalamic dysfunction - hypoventilation - autonomic dysregulation syndrome	29207	Reactive arthritis	79433	Red oculocutaneous albinism
		314962	Reactive hypereosinophilic syndrome	231031	Red palms disease
		166433	Reading seizures	838	RED-M
		857	REAR syndrome	97239	Reducing body myopathy
		1188	Reardon-Baraitser syndrome	523	Reed syndrome
		2631	Reardon-Hall-Slaney syndrome	3221	Refetoff syndrome
		96167	Rec8 syndrome	99995	Reflex sympathetic dystrophy
		96167	Rec(8) syndrome	98826	Refractory anemia
293987	Rapid-onset childhood obesity - hypothalamic dysfunction - hypoventilation-autonomic dysregulation - neural tumors	1115	Recessive aplasia cutis congenita of limbs	86839	Refractory anemia with excess blasts
		139373	Recessive congenital methemoglobinemia type 1	168960	Refractory anemia with excess blasts in transformation
		139380	Recessive congenital methemoglobinemia type 2	100019	Refractory anemia with excess blasts type 1
		79409	Recessive dystrophic epidermolysis bullosa inversa	100020	Refractory anemia with excess blasts type 2
		89842	Recessive dystrophic epidermolysis bullosa, non-Hallopeau-Siemens type	75564	Refractory anemia with ringed sideroblasts
		89842	Recessive dystrophic epidermolysis bullosa-generalized other	398063	Refractory CD
		139373	Recessive hereditary methemoglobinemia type 1	398063	Refractory celiac disease
		139380	Recessive hereditary methemoglobinemia type 2	398063	Refractory sprue
280384	Rare variants of adenocarcinoma of the corpus uteri	280384	Recessive intellectual disability - motor dysfunction - multiple joint contractures	773	Refsum disease
		94125	Recessive mitochondrial ataxia syndrome	1525	Reginato-Schiapachasse syndrome
		461	Recessive X-linked ichthyosis	1433	Regional choroidal atrophy and alopecia
		96167	Recombinant 8 syndrome	83450	Regional odontodysplasia
		96167	Recombinant chromosome 8 syndrome	300865	Regressive atypical histiocytosis
		99990	Recrudescence typhus	1040	Regressive metaphyseal dysplasia
		171220	Rectal duplication	448267	Regressive spondylometaphyseal dysplasia
		100081	Rectal endocrine tumor	2634	Reinhardt-Pfeiffer mesomelic dysplasia
284388	RAS-associated autoimmune leukoproliferative disease	424002	Rectal squamous cell carcinoma	2634	Reinhardt-Pfeiffer syndrome
		88619	Recurrent acute necrotizing encephalopathy	98961	Reis-Bücklers corneal dystrophy
		64740	Recurrent acute pancreatitis	29207	Reiter syndrome
		2672	Recurrent encephalopathy of childhood	99991	Relapsing epidemic typhus
		90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients	33577	Relapsing febrile nodular nonsuppurative panniculitis
		293381	Recurrent hereditary corneal erosions	33577	Relapsing febrile nodular panniculitis
				91547	Relapsing fever

→ 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
728	Relapsing polychondritis	112	Renal tubular normotensive hypokalemic alkalosis with hypercalcioria	71213	Retinal cavernous hemangioma
412	Remnant disease	254902	Renal tubulopathy - encephalopathy - liver failure	1574	Retinal degeneration - nanophthalmos - glaucoma
217330	REN-associated familial juvenile hyperuricemic nephropathy	857	Renal-ear-anal-radial syndrome	1571	Retinal detachment - occipital encephalocele
217330	REN-associated FJHN	1092	Renal-genital-middle ear anomalies	397758	Retinal dystrophy with inner nuclear layer and ganglion cell anomalies
217330	REN-associated kidney disease	294415	Renal-hepatic-pancreatic dysplasia	397758	Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies
411709	Renal agenesis	3032	Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts	436245	Retinal dystrophy-juvenile cataract-short stature syndrome
1848	Renal agenesis, bilateral	774	Rendu-Osler disease	75326	Retinal hemorrhage with vascular tortuosity
93100	Renal agenesis, unilateral	774	Rendu-Osler-Weber disease	3018	Retinal ischemic syndrome - digestive tract small vessel hyalinosis - diffuse cerebral calcifications
2838	Renal caliceal diverticuli - deafness	93975	Renier-Gabreels-Jasper syndrome	319640	Retinal macular dystrophy type 2
319314	Renal cell carcinoma after neuroblastoma	100057	Renin-angiotensin-aldosterone system-blocker-induced angioedema	353356	Retinal vasoproliferative tumor
319314	Renal cell carcinoma associated with neuroblastoma	100057	Renin-angiotensin-aldosterone system-blocker-induced angioneurotic edema	791	Retinitis pigmentosa
1475	Renal coloboma syndrome	294415	Renohepatopancreatic dysplasia	886	Retinitis pigmentosa - deafness
93111	Renal cysts - maturity-onset diabetes of the young	3033	Renotubular dysgenesis	140976	Retinitis pigmentosa - hypopituitarism - nephronophthisis - skeletal dysplasia
93111	Renal cysts and diabetes syndrome	3242	Renpenning syndrome	3085	Retinitis pigmentosa - intellectual disability - deafness - hypogenitalism
93111	Renal dysfunction - early-onset diabetes	364195	Resistance to bleomycine in the treatment of testicular cancer	85332	Retinitis pigmentosa and intellectual disability due to del(X)(p11.3)
93108	Renal dysplasia	240935	Resistance to clopidogrel	85332	Retinitis pigmentosa and intellectual disability due to monosomy Xp11.3
3404	Renal dysplasia - limb defects	73273	Resistance to IGF-1	436245	Retinitis pigmentosa-juvenile
3404	Renal dysplasia - mesomelia - radiohumeral fusion	240947	Resistance to tamoxifene	52427	Retinitis punctata albescens
3156	Renal dysplasia - retinal aplasia	424	Resistance to thyroid stimulating hormone	790	Retinoblastoma
140969	Renal dysplasia - retinal pigmentary dystrophy - cerebellar ataxia - skeletal dysplasia	99832	Resistance to thyrotropin-releasing hormone syndrome	838	Retinocochleocerebral vasculopathy
93173	Renal dysplasia, bilateral	413684	Resistance to vitamin K antagonists	3087	Retinohepatoendocrinologic syndrome
93172	Renal dysplasia, unilateral	247257	Respiratory anthrax	2305	Retinoic acid embryopathy
→1768	Renal dysplasia-megalocystis-sirenomelia syndrome	247257	Respiratory anthrax disease	40366	Retinoid embryopathy
654	Renal embryonic tumor	79127	Respiratory bronchiolitis - interstitial lung disease	2305	Retinoids embryopathy
1652	Renal Fanconi syndrome with nephrocalcinosis and renal stones	284102	Response to antiviral treatment in hepatitis C	352718	Retinol dystrophy-iris coloboma-comedogenic acne syndrome
69076	Renal glucosuria	284102	Response to PEG/IFN-ribavirin in HCV	90050	Retinopathy of prematurity
34528	Renal hypomagnesemia type 2	1662	Restrictive dermopathy	139455	Retinopathy, Burgess-Black type
31043	Renal hypomagnesemia type 3	33355	Reticular dysgenesis	3088	Retinopathy-anemia-central nervous system anomalies syndrome
93101	Renal hypoplasia	99002	Reticular dystrophy of the retinal pigment epithelium	838	Retinopathy-encephalopathy-deafness associated with microangiopathy
97362	Renal hypoplasia, bilateral	100000	Reticular perineurioma	53540	Retinoschisis with early nyctalopia
97361	Renal hypoplasia, unilateral	79145	Reticular pigment anomaly of flexures	269200	Retrocerebellar cyst
319319	Renal medullary carcinoma	178307	Reticulate acropigmentation of Kitamura	90050	Retrolental fibroplasia
71273	Renal nutcracker syndrome	86900	Reticulum cell sarcoma		
171871	Renal pseudohypoaldosteronism type 1	284247	Retinal arterial macroaneurysm and supravalvular pulmonic stenosis		
18	Renal tubular acidosis type 1	75326	Retinal arterial tortuosity		
47159	Renal tubular acidosis type 2	75326	Retinal arteriolar tortuosity		
2785	Renal tubular acidosis type 3	36383	Retinal arteriolar tortuosity - infantile hemiparesis - autosomal dominant leukoencephalopathy		
3033	Renal tubular dysgenesis				
97367	Renal tubular dysgenesis due to twin-twin transfusion				
97369	Renal tubular dysgenesis of genetic origin				

→ 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
49041	Retroperitoneal fibrosis	435998	RI-CMT type D	251043	Ring chromosome 5
778	Rett syndrome	97229	Riboflavin transporter deficiency	1448	Ring chromosome 6
3095	Rett syndrome variant	440706	Ribose-5-P isomerase deficiency	1449	Ring chromosome 7
99852	Reunion island - anorexia - vomiting which is irrepressible - neurological signs	141184	RICH	1450	Ring chromosome 8
		1399	Richards-Rundle syndrome	96173	Ring chromosome 9
294049	Reunion Island Larsen syndrome	240071	Richardson syndrome	1438	Ring chromosome 10
284388	Reversible cerebral vasoconstriction syndrome	2323	Richardson-Kirk syndrome	96175	Ring chromosome 11
254864	Reversible infantile cytochrome C oxidase deficiency	3101	Richieri Costa-da Silva syndrome	1439	Ring chromosome 12
254864	Reversible infantile respiratory chain deficiency	2649	Richieri Costa-Guion Almeida syndrome	96176	Ring chromosome 13
3088	Revesz syndrome	2511	Richieri Costa-Guion Almeida-Ramos syndrome	1440	Ring chromosome 14
3088	Revesz-DeBuse syndrome	→2353	Richieri Costa-Guion Almeida-Rodini syndrome	96177	Ring chromosome 15
3096	Reye syndrome	3102	Richieri Costa-Pereira syndrome	96178	Ring chromosome 16
199267	Reye tumor	1784	Richieri-Costa-Colletto syndrome	1441	Ring chromosome 17
779	Reynolds syndrome	1794	Richieri-Costa-Gorlin syndrome	1442	Ring chromosome 18
244310	RFT1-CDG	28378	Richner-Hanhart syndrome	1443	Ring chromosome 19
251975	RGNT	606	Ricker disease	1444	Ring chromosome 20
71275	Rh deficiency syndrome	606	Ricker syndrome	1445	Ring chromosome 21
71275	Rh-null syndrome	83312	Rickettsialpox	1446	Ring chromosome 22
69077	Rhabdoid tumor	420741	RIDDLE syndrome	261529	Ring chromosome Y
231108	Rhabdoid tumor predisposition syndrome	64744	Riedel disease	91481	Ring dermoid of cornea
3097	Rhabdomyomatous dysplasia - cardiopathy - genital anomalies	64744	Riedel thyroiditis	91481	Ring dermoid syndrome
780	Rhabdomyosarcoma	91483	Rieger anomaly	169	Ringed hair disease
213802	Rhabdomyosarcoma of the cervix uteri	3163	Rieger anomaly - partial lipodystrophy	97238	Rippling muscle disease
213615	Rhabdomyosarcoma of the corpus uteri	782	Rieger syndrome	206575	Rippling muscle disease with myasthenia gravis
3099	Rheumatic fever	319251	Rift valley fever	7	Ritscher-Schinzel syndrome
761	Rheumatoid purpura	99081	Right aortic arch	1803	Rivera-Perez-Salas syndrome
177	Rhizomelic chondrodysplasia punctata	99119	Right inferior caval vein connecting to left-sided atrium	294049	RLS
309789	Rhizomelic chondrodysplasia punctata type 1	99119	Right inferior vena cava connecting to left-sided atrium	93307	rMED
309796	Rhizomelic chondrodysplasia punctata type 2	99119	Right IVC connecting to left-sided atrium	137634	RNF135-related overgrowth syndrome
309803	Rhizomelic chondrodysplasia punctata type 3	99110	Right superior caval vein connecting to left-sided atrium	420741	RNF168 deficiency
2831	Rhizomelic dysplasia, Patterson-Lowry type	99110	Right superior vena cava connecting to left-sided atrium	71273	RNS
93569	Rhizomelic pseudopolyarthrosis	99110	Right SVC connecting to left-sided atrium	3103	Roberts syndrome
1453	Rhizomelic shortness with clavicular defect	293848	Right temporal lobar atrophy	3103	Roberts-SC phocomelia syndrome
3098	Rhizomelic syndrome, Urbach type	439	Right ventricular hypoplasia	3104	Robin sequence - oligodactyly
59315	Rhombencephalosynapsis	97244	Rigid spine congenital muscular dystrophy	97360	Robinow dwarfism
→1071	RHS	97244	Rigid spine syndrome	97360	Robinow syndrome
140976	RHYNS syndrome	1764	Riley-Day syndrome	3105	Robinow-like syndrome
217055	RI-CMT type A	217335	RIN2 deficiency	97360	Robinow-Silverman-Smith syndrome
254334	RI-CMT type B	217335	RIN2 syndrome	→794	Robinow-Sorauf syndrome
369867	RI-CMT type C	1437	Ring chromosome 1	2780	Robinow-Unger syndrome
		96171	Ring chromosome 2	529	Roch-Leri mesosomatous lipomatosis
		96172	Ring chromosome 3	83311	Rocky Mountain spotted fever
		1447	Ring chromosome 4	49382	Rod monochromacy

→ 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
→1855	Roifman-Melamed syndrome	3121	Ruvalcaba syndrome	309155	Sandhoff disease, infantile form
247775	Rokitansky sequence	293848	rvFTD	309162	Sandhoff disease, juvenile form
3109	Rokitansky syndrome	461	RXLI	71272	Sandifer syndrome
1945	Rolandic epilepsy	16	S cone monochromacy	70595	SANDO
163727	Rolandic epilepsy - paroxysmal exercise-induced dystonia - writer's cramp	16	S cone monochromatism	2378	Sandrow syndrome
163721	Rolandic epilepsy - speech dyspraxia	3105	Saal-Greenstein syndrome	581	Sanfilippo disease
101016	Romano-Ward long QT syndrome	319239	Sabia hemorrhagic fever	79269	Sanfilippo syndrome type A
101016	Romano-Ward syndrome	3124	Saccharopine dehydrogenase deficiency	79270	Sanfilippo syndrome type B
1214	Romberg syndrome	3124	Saccharopinuria	79271	Sanfilippo syndrome type C
3110	Rombo syndrome	286	Sack-Barabas syndrome	79272	Sanfilippo syndrome type D
1088	Rommen-Mueller-Sybert syndrome	98841	sACL	2323	Sanjad-Sakati syndrome
90050	ROP	3027	Sacral agenesis syndrome	588	Santavuori congenital muscular dystrophy
158014	Rosaï-Dorfman disease	397927	Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome	79263	Santavuori disease
158014	Rosaï-Dorfman-Destombes disease	→83628	Sacral hemangiomas - multiple congenital abnormalities	79263	Santavuori-Haltia disease
1837	Rosenberg-Lohr syndrome	2351	Sacral meningocele - conotruncal heart defects	2155	Santos-Mateus-Leal syndrome
329	Rosenthal factor deficiency	3027	Sacral regression syndrome	98868	SAO
329	Rosenthal syndrome	1773	Sacrococcygeal dysgenesis association	247234	SAOA
251975	Rosette-forming glioneuronal tumor of fourth ventricule	85165	SADDAN	793	SAPHO syndrome
90339	Rosselli-Gulienetti syndrome	794	Saethre-Chotzen syndrome	54368	Sarcocystosis
2909	Rothmund-Thomson syndrome	2872	Sagittal craniostenosis with congenital heart disease, mental deficiency and mandibular ankylosis	797	Sarcoidosis
221008	Rothmund-Thomson syndrome type 1	300493	Sagliker syndrome	3129	Sarcosine dehydrogenase complex deficiency
221016	Rothmund-Thomson syndrome type 2	83484	Saint Louis encephalitis	3129	Sarcosinemia
3111	Rotor syndrome	2256	Saito-Kuba-Tsuruta syndrome	54368	Sarcosporidiosis
3115	Roussy-Lévy syndrome	3128	Sakati syndrome	1878	Sarcotubular myopathy
1323	Rozin-camptodactyly syndrome	3128	Sakati-Nyhan syndrome	140896	SARS
1323	Rozin-Hertz-Goodman syndrome	3128	Sakati-Nyhan-Tisdale syndrome	140896	SARS-associated coronavirus
280569	RPGN	1409	Salomon syndrome	140896	SARS-CoV
1507	RRS	2613	Salcedo syndrome	3130	Satoyoshi syndrome
818	RSH syndrome	140969	Saldino-Mainzer syndrome	330015	Saturnism
293848	RTLA	213557	Salivary gland type cancer of the breast	425120	SAVI
231108	RTPS	213557	Salivary gland type carcinoma of the breast	3047	Say-Barber-Biesecker-Young-Simpson syndrome
2909	RTS	309334	Salla disease	2013	Say-Barber-Hobbs syndrome
221008	RTS1	370938	Salt-and-pepper syndrome	3132	Say-Barber-Miller syndrome
221016	RTS2	112	Salt-losing tubular disorder, Henle's loop type	3133	Say-Field-Coldwell syndrome
83616	Rubella panencephalitis	112	Salt-wasting tubulopathy, Henle's loop type	3369	Say-Meyer syndrome
783	Rubinstein-Taybi syndrome	2230	Salti-Salem syndrome	3047	SBBYSS
353281	Rubinstein-Taybi syndrome due to 16p13.3 microdeletion	369992	SAM syndrome	79157	SBCAD deficiency
353277	Rubinstein-Taybi syndrome due to CREBBP mutations	53721	SAMS 1-31	481	SBMA
353284	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency	397623	SAMS syndrome	3103	SC phocomelia
1768	Rudd-Klimek syndrome	228123	San Joaquin valley fever	3103	SC pseudothalidomide syndrome
→798	Rudiger syndrome	96167	San Luis Valley syndrome	98755	SCA1
79433	Rufous oculocutaneous albinism	796	Sandhoff disease	98756	SCA2
435953	Ruijs-Aalfs syndrome	309169	Sandhoff disease, adult form	98757	SCA3
1672	Russell diencephalic cachexia			276238	SCA3, Joseph type
1672	Russell syndrome			276244	SCA3, Machado type
1834	Russell-Weaver-Bull syndrome			276241	SCA3, Thomas type
2709	Rutherford syndrome			98765	SCA4
				98766	SCA5
				98758	SCA6
				94147	SCA7

→ 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
98760	SCA8	139485	SCAR9	3041	Scholte-Begeer-van Essen syndrome
98761	SCA10	284289	SCAR10	50944	Schöpf-Schulz-Passarge syndrome
98767	SCA11	284271	SCAR11	93921	Schwannomatosis
98762	SCA12	284282	SCAR12	800	Schwartz-Jampel syndrome
98768	SCA13	324262	SCAR13	800	Schwartz-Jampel syndrome type 1
98763	SCA14	352403	SCAR14	800	Schwartz-Jampel-Aberfeld syndrome
98769	SCA15/16	404499	SCAR15	277	SCID due to adenosine deaminase deficiency
→98769	SCA16	412057	SCAR16	275	SCID due to artemis deficiency
98759	SCA17	363432	SCAR18	357237	SCID due to CARD11 deficiency
98771	SCA18	448251	SCAR19	331206	SCID due to complete RAG1/2 deficiency
98772	SCA19/22	397709	SCAR20	228003	SCID due to CORO1A deficiency
101110	SCA20	3134	SCARF syndrome	228003	SCID due to coronin-1A deficiency
98773	SCA21	90080	Scarring in glaucoma filtration surgical procedures	420573	SCID due to CTPS1 deficiency
→98772	SCA22	95434	SCASI	275	SCID due to DCLRE1C deficiency
101108	SCA23	85297	SCAX3	317425	SCID due to DNA-PKcs deficiency
101111	SCA25	85292	SCAX4	397787	SCID due to IKK2 deficiency
101112	SCA26	284400	SCCB	280142	SCID due to LCK deficiency
98764	SCA27	98967	SCCD	280142	SCID due to lymphocyte-specific protein tyrosine kinase deficiency
101109	SCA28	370396	SCCO	33355	SCID with leukopenia
208513	SCA29	91365	SCD	275	SCID, Athabascan type
211017	SCA30	98967	SCD	275	SCID, Athabaskan type
217012	SCA31	449280	Scedosporiosis	276	SCIDX1
276183	SCA32	1383	Schaap-Taylor-Baraitser syndrome	185	Scimitar syndrome
1955	SCA34	71212	SCHAD deficiency	70573	SCLC
276193	SCA35	370039	Schauder syndrome	352763	Scleredema
276198	SCA36	93474	Scheie syndrome	75840	Scleroatonic muscular dystrophy
363710	SCA37	2353	Schilbach-Rott syndrome	384	Scleroatrophic syndrome
423296	SCA38	59298	Schilder disease	167635	Scleromyxedema
423275	SCA40	59298	Schilder's disease	90400	Scleromyxedema without monoclonal gammopathy
26792	SCAD deficiency	1830	Schimke immuno-osseous dysplasia	75325	Sclerosing dysplasia of bone - ichthyosis - premature ovarian failure
26792	SCADD	1830	Schimke syndrome	63999	Sclerosing mediastinitis
254881	SCAE	2612	Schimmelpenning syndrome	238593	Sclerosing mesenteritis
1003	Scalp defects - postaxial polydactyly	3137	Schindler disease	100001	Sclerosing perineurioma
370052	SCALP syndrome	79279	Schindler disease type 1	3152	Sclerosteosis
2036	Scalp-ear-nipple syndrome	79280	Schindler disease type 2	384	Sclerotylosis
64753	SCAN 2	79281	Schindler disease type 3	188	SCLS
94124	SCAN1	3138	Schinzel syndrome	331176	SCN4
840	SCAP	798	Schinzel-Giedion syndrome	439746	Ssecondary PAN
168624	Scaphocephaly - macrocephaly - maxillary retrusion - intellectual disability	63862	Schisis association	832	SCOT deficiency
2839	Scapuloiliac dysostosis	1247	Schistosomiasis	1514	Scott craniodigital syndrome
431255	Scapuloperoneal neuronopathy	799	Schizencephaly	806	Scott syndrome
431255	Scapuloperoneal spinal muscular atrophy	98973	Schlüchting dystrophy	1514	Scott-Bryant-Graham syndrome
64753	SCAR1	3143	Schmidt syndrome	1509	Scott-Taor syndrome
1170	SCAR2	2252	Schmitt-Gillenwater-Kelly syndrome	86813	SCRA
95433	SCAR3	3144	Schneckenbecken dysplasia	83317	Scrub typhus
95434	SCAR4	37748	Schnitzler syndrome	794	SCS
83472	SCAR5	98967	Schnyder corneal dystrophy	295193	SD1, Castilla type
284332	SCAR6	98967	Schnyder crystalline corneal dystrophy sine crystals	295189	SD1, Lueken type
284324	SCAR7	3145	Schofer-Beetz-Bohl syndrome		
88644	SCAR8				

→ 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
295191	SD1, Montagu type	3452	Secondary non-tropical sprue	93356	SEMD type 2
295187	SD1, Weidenreich type	420259	Secondary PAP	171866	SEMD, aggrecan type
295187	SD1a	439746	Secondary periarteritis nodosa	93351	SEMD, Irapa type
295189	SD1b	439746	Secondary polyarteritis nodosa	156728	SEMD, MATN3-related
295191	SD1c	420259	Secondary pulmonary alveolar proteinosis	156728	SEMD, matrilin-3 type
295193	SD1d	99930	Secondary pulmonary hemosiderosis	93356	SEMD, Missouri type
295197	SD2, Debeer type	447774	Secondary sclerosing cholangitis	93352	SEMD, Shohat type
295199	SD2, Malik type	95427	Secondary short bowel syndrome	93359	SEMD-JL
295195	SD2, Vordingborg type	99857	Secondary syringomyelia	93360	SEMD-MD
295195	SD2a	364055	SECORD	93359	SEMDJL1
295197	SD2b	163654	SED-BDS	93360	SEMDJL2
295199	SD2c	94068	SEDC	420402	Semicircular canal dehiscence syndrome
93404	SD3	567	Sedlackova syndrome	220386	Semilobar holoprosencephaly
93406	SD5	647	Seemanova syndrome type 2	842	Seminoma of testis
84064	SD/THE	2528	Seemanova-Lesny syndrome	842	Seminomatous germ cell tumor of testis
263463	SDCD, CHST3 type	251618	SEGA	329284	SENDA
168577	sdCHC	2759	Seghers syndrome	79480	Senear-Usher syndrome
300869	SDRPL	67039	Segmental odontomaxillary dysplasia	397596	Senescent T-cells-lymphadenopathy-immunodeficiency syndrome due to p110delta-activating mutation
811	SDS	137608	Segmental outgrowth - lipomatosis - arteriovenous malformation - epidermal nevus	1369	Sengers syndrome
373	SDYS	314662	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	2183	Sengers-Hamel-Otten syndrome
158029	Sea-blue histiocytosis	455	SEI	330001	Senile systemic amyloidosis
1778	Seaver-Cassidy syndrome	35069	Seitelberger disease	1292	Senior syndrome
370052	Sebaceous nevus-central nervous system malformations-aplasia cutis congenital-limbal dermoid-pigmented nevus syndrome	79156	Seizures - intellectual disability due to hydroxyllysinuria	84081	Senior-Boichis syndrome
370052	Sebaceous nevus-CNS malformations-aplasia cutis congenital-limbal dermoid-pigmented nevus syndrome	199343	Seizures - sensorineural deafness - ataxia - intellectual disability - electrolyte imbalance	3156	Senior-Loken syndrome
→18205 0	Sebastian syndrome	357194	Selection of therapeutic option in colorectal cancer	1515	Sensenbrenner syndrome
841	Sebocystomatosis	357191	Selection of therapeutic option in non-small cell lung carcinoma	217622	Sensorineural deafness with dilated cardiomyopathy
168606	Seborrhea-like dermatitis with psoriasisiform elements	35858	Selective cobalamin malabsorption with proteinuria	857	Sensorineural deafness with imperforate anus and hypoplastic thumbs
79480	Seborrheic pemphigus	183675	Selective IgG subclass deficiency	66633	Sensorineural hearing loss - early graying - essential tremor
98873	SEC23B-CDG	331235	Selective IgM deficiency	97229	Sensorineural hearing loss - pontobulbar palsy
808	Seckel syndrome	331235	Selective immunoglobulin M deficiency	217622	Sensorineural hearing loss with dilated cardiomyopathy
141022	Second branchial cleft anomaly	165994	Selective pituitary resistance to thyroid hormone	70595	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis
141022	Second branchial cleft cyst	99798	Selective tooth agenesis	477	Senter syndrome
141022	Second branchial cleft fistula	281122	Self-healing collodion baby	90118	SEOAN due to MFN2 deficiency
139420	Secondary acute transverse myelitis	90397	Self-healing papular mucinosis	70594	Sepiapterin reductase deficiency
85445	Secondary amyloidosis	65748	Self-healing squamous epithelioma type 1	90051	Sepsis in premature infants
169618	Secondary central precocious puberty	→1768	Selig-Benacerraf-Greene syndrome	180154	Septate vagina
91365	Secondary ciliary dyskinesia	3232	Sellars-Beighton syndrome	137839	Septic phlebitis of the internal jugular vein
314962	Secondary HES	100069	Semantic dementia	3157	Septo-optic dysplasia
314962	Secondary hypereosinophilic syndrome	100069	Semantic primary progressive aphasia	3157	Septo-optic dysplasia spectrum
2615	Secondary hypertrophic osteoperiostosis with pernio	100069	Semantic variant PPA	280195	Septopreoptic holoprosencephaly
90363	Secondary intestinal lymphangiectasia			280195	Septopreoptic HPE
399180	Secondary non-traumatic avascular necrosis			139466	SERKAL syndrome
399180	Secondary non-traumatic AVN				

→ 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
43116	Serotonergic syndrome	275	Severe combined immunodeficiency, Athabaskan type	420561	Severe intellectual disability-aplasia/hypoplasia of thumb and hallux syndrome
43116	Serotonin storm	209370	Severe congenital encephalopathy due to MECP2 mutation	94066	Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia
43116	Serotonin syndrome	300298	Severe congenital hypochromic anemia with ringed sideroblasts	438178	Severe intellectual disability-epilepsy-cataract syndrome due to FAR1 deficiency
43116	Serotonin toxicity	300298	Severe congenital hypochromic sideroblastic anemia	438178	Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency
43116	Serotonin toxidrome	171430	Severe congenital nemaline myopathy	438178	Severe intellectual disability-epilepsy-cataract syndrome due to peroxisomal disorder
424073	Serous cystadenocarcinoma of pancreas	331176	Severe congenital neutropenia - pulmonary hypertension - superficial venous angiectasis	436141	Severe intellectual disability-hypotonia-strabismus-coarse face-planovalgus syndrome
206470	Serous or mucinous cystadenoma of childhood	99749	Severe congenital neutropenia type 3	363686	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome
168829	Serous surface papillary carcinoma	331176	Severe congenital neutropenia type 4	397933	Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome
→955	Serpentine fibula - polycystic kidneys	369992	Severe dermatitis-multiple allergies-metabolic wasting syndrome	404473	Severe intellectual disability-progressive spastic diplegia syndrome
35686	Serpiginous choroiditis	→30075	Severe dilated cardiomyopathy due to lamin A/C mutation	391307	Severe intellectual disability-short stature-behavioral troubles-facial dysmorphism syndrome
157798	Serrated polyposis	→30075	Severe dilated cardiomyopathy with or without myopathy	324307	Severe lateral tibial bowing with short stature
2901	Serum neuritis	98896	Severe dystrophinopathy, Duchenne type	2879	Severe limb deficit
75508	Servelle-Martorell syndrome	364055	Severe early-childhood-onset retinal dystrophy	369939	Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome
199343	SeSAME syndrome	228374	Severe early-onset axonal neuropathy due to light neurofilament subunit deficiency	33069	Severe myoclonic epilepsy of infancy
1807	Setleis syndrome	90118	Severe early-onset axonal neuropathy due to MFN2 deficiency	33069	Severe myoclonus epilepsy of infancy
85165	Severe achondroplasia - developmental delay - acanthosis nigricans	228374	Severe early-onset axonal neuropathy due to NEFL deficiency	314655	Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion
140896	Severe acute respiratory syndrome	329249	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency	397593	Severe neonatal lactic acidosis due to NFS1-1SD11 complex deficiency
438207	Severe autosomal recessive macrothrombocytopenia	440427	Severe early-onset pulmonary alveolar proteinosis due to MARS deficiency	209370	Severe neonatal-onset encephalopathy with microcephaly
314911	Severe Canavan disease	169793	Severe factor IX deficiency	363400	Severe neurodegenerative syndrome due to BSCL2 deficiency
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	169802	Severe factor VIII deficiency	363400	Severe neurodegenerative syndrome with lipodystrophy
275	Severe combined immunodeficiency due to artemis deficiency	352577	Severe feeding difficulties - failure to thrive - microcephaly due to ASXL3 deficiency	216812	Severe osteogenesis imperfecta
357237	Severe combined immunodeficiency due to CARD11 deficiency	79408	Severe generalized RDEB	411543	Severe phosphoribosylpyrophosphate synthetase superactivity
331206	Severe combined immunodeficiency due to complete RAG1/2 deficiency	79408	Severe generalized recessive dystrophic epidermolysis bullosa	280210	Severe PMD
228003	Severe combined immunodeficiency due to CORO1A deficiency	2109	Severe Hallermann-Streiff-François syndrome	411543	Severe PRPP synthetase superactivity
228003	Severe combined immunodeficiency due to coronin-1A deficiency	169802	Severe hemophilia A	411543	Severe PRPS1 superactivity
420573	Severe combined immunodeficiency due to CTPS1 deficiency	169793	Severe hemophilia B		
275	Severe combined immunodeficiency due to DCLRE1C deficiency	98920	Severe infantile axonal neuropathy with respiratory failure type 1		
317425	Severe combined immunodeficiency due to DNA-PKcs deficiency	404521	Severe infantile axonal neuropathy with respiratory failure type 2		
397787	Severe combined immunodeficiency due to IKK2 deficiency	280763	Severe intellectual disability and progressive spastic paraparesis		
280142	Severe combined immunodeficiency due to LCK deficiency				
280142	Severe combined immunodeficiency due to lymphocyte-specific protein tyrosine kinase deficiency				
33355	Severe combined immunodeficiency with leukopenia				
275	Severe combined immunodeficiency, Athabascan type				

→ 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
163703	Severe refractory status epilepticus owing to presumed encephalitis	93268	Short rib-polydactyly syndrome type 4	435804	Short stature-advanced bone age-early-onset osteoarthritis syndrome
169095	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy	93268	Short rib-polydactyly syndrome, Beemer-Langer type	397623	Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome
3078	Severe X-linked intellectual disability, Gustavson type	93269	Short rib-polydactyly syndrome, Majewski type		Short stature-delayed bone age due to thyroid hormone metabolism deficiency
238329	Severe X-linked mitochondrial encephalomyopathy	93270	Short rib-polydactyly syndrome, Saldino-Noonan type	1088	Short stature-heart defect-craniofacial anomalies syndrome
363489	Sex cord-stromal tumor of testis	93271	Short rib-polydactyly syndrome, Verma-Naumoff type	420794	Short stature-kyphosis-hypoplasia of basal ilia-cone epiphyses-facial dysmorphism syndrome
139466	Sex reversion - kidneys, adrenal and lung dysgenesis	→1263	Short ribs - craniostostosis - polysyndactyly	423454	Short stature-nail dysplasia-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome
3162	Sézary lymphoma	2994	Short stature - craniofacial anomalies - genital hypoplasia	314394	Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome
3162	Sézary syndrome	2866	Short stature - deafness - neutrophil dysfunction - dysmorphism	391677	Short stature-optic atrophy-Pelger-Hüet anomaly syndrome
373	SGBS	2332	Short stature - facial and skeletal anomalies - intellectual disability - macrodontia	3163	SHORT syndrome
373	SGBS1	2649	Short stature - intellectual disability - eye anomalies - cleft lip/palate	2832	Short tarsus - absence of lower eyelashes
79022	SGBS2	1937	Short stature - locking fingers	251515	Short tendo calcaneus
35710	SGLT1 deficiency	3102	Short stature - Pierre Robin sequence - cleft mandible - hand anomalies clubfoot	294998	Short toes
69076	SGLT2 deficiency	3102	Short stature - Pierre Robin syndrome - cleft mandible - hand anomalies clubfoot	295134	Short toes, bilateral
2462	SGS	85442	Short stature - pituitary and cerebellar defects - small sella turcica	295132	Short toes, unilateral
798	SGS	2868	Short stature - valvular heart disease - characteristic facies	357175	Short ulna - dysmorphism - hypotonia - intellectual disability
2407	Shabbir syndrome	2865	Short stature - webbed neck - heart disease	57145	Short-lasting unilateral neuralgiform headache attacks with conjunctival injection and tearing
897	Shah-Waardenburg syndrome	2863	Short stature - wormian bones - dextrocardia	935	Short-limb skeletal dysplasia with severe combined immunodeficiency
29822	Shapiro syndrome	314811	Short stature due to GHSR deficiency	79157	Short/branched-chain acyl-coA dehydrogenase deficiency
1506	Sharma-Kapoor-Ramji syndrome	629	Short stature due to growth hormone qualitative anomaly	2580	Shoulder and girdle defects - familial intellectual disability
809	Sharp syndrome	633	Short stature due to growth hormone resistance	1940	Shoulder and thorax deformity - congenital heart disease
281122	SHCB	314811	Short stature due to growth hormone secretagogue receptor deficiency	314795	SHOX-related short stature
91355	Sheehan syndrome	632	Short stature due to isolated growth hormone deficiency with X-linked hypogammaglobulinemia	567	Shprintzen syndrome
1147	Sheldon-Hall syndrome	314802	Short stature due to partial GHR deficiency	2462	Shprintzen-Goldberg syndrome
3329	SHFLD syndrome	314802	Short stature due to partial growth hormone receptor deficiency	3165	Shulman syndrome
2440	SHFM	140941	Short stature due to primary acid-labile subunit deficiency	811	Shwachman syndrome
3329	SHFM associated with aplasia of long bones	220465	Short stature due to STAT5b deficiency	811	Shwachman-Bodian-Diamond syndrome
90038	Shiga-like toxin-associated HUS	2867	Short stature, Brussels type	811	Shwachman-Diamond syndrome
810	Shigellosis			812	Sialidosis type 1
158014	SHML			87876	Sialidosis type 2
1008	Shokeir syndrome			3166	Sialuria
99063	Shone complex			3166	Sialuria, French type
251515	Short Achilles tendon			98920	SIANRF
26792	Short chain acyl-CoA dehydrogenase deficiency			→33364	SIBIDS syndrome
66518	Short fifth metacarpals - insulin resistance			611	sIBM
294996	Short fingers				
295130	Short fingers, bilateral				
295128	Short fingers, unilateral				
935	Short limb skeletal dysplasia with SCID				
93270	Short rib-polydactyly syndrome type 1				
93269	Short rib-polydactyly syndrome type 2				
93271	Short rib-polydactyly syndrome type 3				

→ 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
251359	Sickle cell - beta-thalassemia disease	50809	Singh-Williams-McAlister syndrome	584	Sly disease
251365	Sickle cell - hemoglobin C disease	2286	Single upper central incisor	70	SMA
251370	Sickle cell - hemoglobin D disease	99097	Single ventricular septal defect	83330	SMA1
251375	Sickle cell - hemoglobin E disease	439755	Single-organ PAN	83418	SMA2
232	Sickle cell anemia	439755	Single-organ periarteritis nodosa	83419	SMA3
232	Sickle cell disease	439755	Single-organ polyarteritis nodosa	83420	SMA4
210272	Sickness of disembarkment	85191	Singleton-Merten dysplasia	83330	SMA type 1
838	SICRET syndrome	85191	Singleton-Merten syndrome	83418	SMA type 2
168593	SIDDT	1260	Sino-auricular heart block	83419	SMA type 3
54028	Sideropenic dysphagia	324321	Sinoatrial node dysfunction and deafness	83420	SMA type 4
2267	Sidransky-Feinstein-Goodman syndrome	158014	Sinus histiocytosis with massive lymphadenopathy	83330	SMA type I
3167	Sieglar-Brewer-Carey syndrome	890	Sinusoidal obstruction syndrome	83418	SMA type II
→244	Siewert syndrome	247698	Sipple syndrome	83419	SMA type III
369861	SIFD syndrome	3169	Sirenomelia	83420	SMA type IV
314786	Silent pituitary adenoma	2882	Sitosterolemia	83330	SMA-I
71276	Silent sinus syndrome	157769	Situs ambiguous	83418	SMA-II
3168	Sillence syndrome	157769	Situs ambiguus	83419	SMA-III
60014	Silver staining	101063	Situs inversus	83420	SMA-IV
100998	Silver syndrome	101063	Situs inversus totalis	363447	SMALED
813	Silver-Russell dwarfism	816	Sjögren-Larsson syndrome	209341	SMALED1
813	Silver-Russell syndrome	800	SJS	363454	SMALED2
231137	Silver-Russell syndrome due to 7p11.2-p13 microduplication	800	SJS1	284400	Small cell bladder cancer
231137	Silver-Russell syndrome due to 7p11.2p13 microduplication	95455	SJS-TEN	284400	Small cell bladder carcinoma
231144	Silver-Russell syndrome due to 11p15 microduplication	2565	Skeletal dysplasia - brachydactyly	284400	Small cell carcinoma of the bladder
397590	Silver-Russell syndrome due to a point mutation	1858	Skeletal dysplasia - epilepsy - short stature	370396	Small cell carcinoma of the ovary
231140	Silver-Russell syndrome due to an imprinting defect of 11p15	1436	Skeletal dysplasia - intellectual disability	284400	Small cell carcinoma of the urinary bladder
231137	Silver-Russell syndrome due to dup(7)(p11.2p13)	166277	Skeletal dysplasia with wormian bone - multiple fractures - dentin abnormality	70573	Small cell lung cancer
96182	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7	1426	Skeletal dysplasia, Greenberg type	370396	Small cell ovarian carcinoma
231147	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 11	293165	Skin fragility-woolly hair- palmoplantar hyperkeratosis syndrome	838	Small infarctions of cochlear, retinal and encephalic tissue
231137	Silver-Russell syndrome due to trisomy 7p11.2-p13	293165	Skin fragility-woolly hair- palmoplantar keratoderma syndrome	1201	Small intestinal atresia
231137	Silver-Russell syndrome due to trisomy 7p11.2p13	178475	Skin infectious botulism	67038	Small lymphocytic lymphoma
1968	Simosa-Penchaszadeh-Bustos syndrome	178475	Skin toxin-mediated botulism	543	Small non-cleaved cell lymphoma
91139	Simple cryoglobulinemia	52503	SLC6A8 deficiency	1509	Small patella syndrome
373	Simpson dysmorphia syndrome	238459	SLC35A1-CDG	98920	SMARD1
373	Simpson-Golabi-Behmel syndrome	356961	SLC35A2-CDG	404521	SMARD2
373	Simpson-Golabi-Behmel syndrome type 1	370943	SLC35A3-CDG	481	SMAX1-related spinobulbar muscular atrophy
79022	Simpson-Golabi-Behmel syndrome type 2	99843	SLC35C1-CDG	1145	SMAX2
97337	Sinding-Larsen-Johansson disease	3144	SLC35D1-CDG	1145	SMAX2-related spinal muscular atrophy
		93552	SLE, pediatric onset	139557	SMAX3-related distal spinal muscular atrophy
		3385	Sleeping sickness	98959	SMCD
		88633	SLK	85167	SMD-CRD
		818	SLOS	33069	SMEI
		70472	SLSJ-COX deficiency	93974	Smith-Fineman-Myers syndrome
		3156	SLSN	818	Smith-Lemli-Opitz syndrome
				819	Smith-Magenis syndrome
				178355	Smith-McCort dysplasia
				2286	SMMC1
				158775	Smouldering systemic mastocytosis

→ 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
3198	SMS	2815	Spastic paraparesis - deafness	295195	SPD1
86854	SMZL	101003	Spastic paraparesis - vitiligo - premature graying - characteristic facies	295197	SPD2
449285	Snakebite envenomation			295199	SPD3
820	Sneddon syndrome	99015	Spastic paraparesis type 2	295197	SPD, Debeer type
48377	Sneddon-Wilkinson disease	2816	Spastic paraplegia - epilepsy - intellectual disability	295199	SPD, Malik type
91496	Snowflake vitreoretinal degeneration	2819	Spastic paraplegia - facial-cutaneous lesions	295195	SPD, Vordingborg type
3063	Snyder-Robinson syndrome	2818	Spastic paraplegia - glaucoma - intellectual disability	352403	Spectrin-associated autosomal recessive cerebellar ataxia
3157	SOD	2822	Spastic paraplegia - intellectual disability - thin corpus callosum	352403	Spectrin-associated autosomal recessive cerebellar ataxia type 1
67039	SOD	2820	Spastic paraplegia - nephritis - deafness	209908	Speech and language disorder with orofacial dyspraxia
306577	Sodium channelopathy-related small fiber neuropathy	2821	Spastic paraplegia - neuropathy - poikiloderma	209908	Speech-language disorder type 1
99903	Sudoku	329475	Spastic paraplegia - Paget disease of bone	→2909	Spellacy-Gibbs-Watts syndrome
99772	Soft cleft palate	2826		1855	SPENCD
314394	SOFT syndrome	100996	Spastic paraplegia - retinal degeneration	→1855	SPENCDI
100002	Soft tissue perineurioma	139480	Spastic paraplegia due to neuropathy target esterase mutation	2816	SPERM
2234	Sohval-Soffer syndrome	139480		99865	Spermatocytic seminoma
137608	SOLAMEN syndrome	431329	Spastic paraplegia due to partial TGF deficiency	306617	SPG1
97230	Solar urticaria	99015		99015	SPG2
424065	Solid pseudopapillary carcinoma of pancreas	99013	Spastic paraplegia type 2	100985	SPG4
83468	Solitary bone cyst	100998		100986	SPG5A
2126	Solitary fibrous tumor	320406	Spastic paraplegia-optic atrophy-neuropathy syndrome	100988	SPG6
79455	Solitary mastocytoma	3011		99013	SPG7
2286	Solitary median maxillary central incisor syndrome	447997	Spastic quadriplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	100989	SPG8
100035	Solitary necrotic tumor of the liver	210141		100990	SPG9
86855	Solitary plasmacytoma	3011	Spastic tetraplegia - retinitis pigmentosa - intellectual disability	100991	SPG10
209964	Solitary rectal ulcer syndrome	447997		2822	SPG11
2612	Solomon syndrome	3175	Spasticity - intellectual disability - X-linked epilepsy	100993	SPG12
314769	Somatotammotropinoma	401866		100994	SPG13
97283	Somatostatinoma	251282	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	100995	SPG14
2564	Sommer-Hines syndrome	314603		100996	SPG15
1064	Sommer-Rathbun-Battles syndrome	254343	Spasticity-ataxia-gait anomalies syndrome	100997	SPG16
1529	Sommer-Young-Wee-Frye syndrome	313772		100998	SPG17
1355	Sonoda syndrome	158	Spasticity-ataxia-gait anomalies syndrome	209951	SPG18
391677	SOPH syndrome	171622		100999	SPG19
1471	Sorsby syndrome	171607	Spastic quadriplegic cerebral palsy	101000	SPG20
59181	Sorsby's fundus dystrophy	101001		101001	SPG21
821	Sotos syndrome	101003	Spastic quadriplegia - retinitis pigmentosa - intellectual disability	101003	SPG23
420179	Sotos syndrome 2	101004		101004	SPG24
98868	Southeast Asian ovalocytosis	101005	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	101005	SPG25
352403	SPARCA	101006		101006	SPG26
352403	SPARCA1	101007	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	101007	SPG27
79132	Sparse hair - short stature - skin anomalies	101008		101008	SPG28
279882	Spasmus nutans	101009	Spasticity - intellectual disability - X-linked epilepsy	101009	SPG29
2572	Spastic ataxia - corneal dystrophy	101010		101010	SPG30
2572	Spastic ataxia - ocular anomalies	101011	Spasticity-ataxia-gait anomalies syndrome	101011	SPG31
1182	Spastic ataxia with congenital miosis	171622		171622	SPG32
1680	Spastic diplegia, infantile type	171607	SPAX1	171607	SPG34
99015	Spastic gait type 2	171629		171629	SPG35
100990	Spastic paraparesis - amyopathy - cataracts - gastroesophageal reflux	320365	SPAX3	320365	SPG36
		171612		171612	SPG37
		171617	SPAX4	171617	SPG38
		313772			
		158			

→ 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
139480	SPG39	210584	Spindle cell hemangioma	64753	Spinocerebellar ataxia with axonal neuropathy type 2
320355	SPG41	2074	Spinocerebellar ataxia - amyotrophy - deafness	254881	Spinocerebellar ataxia with epilepsy
171863	SPG42	1185	Spinocerebellar ataxia - dysmorphism	276241	Spinocerebellar ataxia, Thomas type
320370	SPG43	1955	Spinocerebellar ataxia and erythrokeratoderma	448251	Spinocerebellar autosomal recessive ataxia 19
320401	SPG44	412057	Spinocerebellar ataxia autosomal recessive type 16	3177	Spinocerebellar degeneration - corneal dystrophy
320396	SPG45	98755	Spinocerebellar ataxia type 1	99903	Spirillary rat-bite fever
320391	SPG46	94124	Spinocerebellar ataxia type 1 with axonal neuropathy	757	Spitzer-Weinstein syndrome
306511	SPG48	98756	Spinocerebellar ataxia type 2	300869	Splenic diffuse red pulp B-cell lymphoma
320385	SPG49	98757	Spinocerebellar ataxia type 3	300869	Splenic diffuse red pulp lymphoma
319199	SPG53	276238	Spinocerebellar ataxia type 3, Joseph type	86854	Splenic marginal zone lymphoma
320380	SPG54	276244	Spinocerebellar ataxia type 3, Machado type	2063	Splenogonadal fusion - limb defects - micrognathia
320375	SPG55	98765	Spinocerebellar ataxia type 4	47612	Splenomegaly-neutropenia-rheumatoid arthritis syndrome
320411	SPG56	98766	Spinocerebellar ataxia type 5	294994	Split foot
431329	SPG57	98758	Spinocerebellar ataxia type 6	2439	Split foot deformity - mandibulofacial dysostosis
397946	SPG58	94147	Spinocerebellar ataxia type 7	295126	Split foot, bilateral
401795	SPG59	98760	Spinocerebellar ataxia type 8	295124	Split foot, unilateral
401800	SPG60	98761	Spinocerebellar ataxia type 10	294992	Split hand
401780	SPG61	98767	Spinocerebellar ataxia type 11	71271	Split hand - split foot - deafness
401785	SPG62	98762	Spinocerebellar ataxia type 12	2437	Split hand - urinary anomalies - spina bifida
401805	SPG63	98768	Spinocerebellar ataxia type 13	2440	Split hand foot malformation
401810	SPG64	98763	Spinocerebellar ataxia type 14	2437	Split hand with obstructive uropathy, spina bifida and diaphragmatic defects
320396	SPG65	98769	Spinocerebellar ataxia type 15/16	295122	Split hand, bilateral
401815	SPG66	→98769	Spinocerebellar ataxia type 16	295120	Split hand, unilateral
401820	SPG67	98759	Spinocerebellar ataxia type 17	2440	Split hand-split foot malformation
401825	SPG68	98771	Spinocerebellar ataxia type 18	3329	Split hand/foot malformation with long bone deficiency
401830	SPG69	98772	Spinocerebellar ataxia type 19/22	958	Split hand/split foot - mandibular hypoplasia
401835	SPG70	101110	Spinocerebellar ataxia type 20	2329	Split hand/split foot - nystagmus
401840	SPG71	98773	Spinocerebellar ataxia type 21	1756	Split notochord syndrome
401849	SPG72	→98772	Spinocerebellar ataxia type 22	3329	Split-hand/foot malformation associated with aplasia of long bones
444099	SPG73	101108	Spinocerebellar ataxia type 23	320406	SPOAN
268129	Spheroid body myopathy	101111	Spinocerebellar ataxia type 25	93357	SPONASTRIME dysplasia
3449	Spherophakia - brachymorphia	101112	Spinocerebellar ataxia type 26	1190	Spondylo-humero-femoral dysplasia
306553	Spherulocytosis	98764	Spinocerebellar ataxia type 27	228387	Spondylo-megaepiphyseal-metaphyseal dysplasia
79264	Spielmeyer-Vogt disease	101109	Spinocerebellar ataxia type 28	85194	Spondylo-ocular syndrome
314432	Spigelian hernia-cryptorchidism syndrome	208513	Spinocerebellar ataxia type 29	3180	Spondylocamptodactyly syndrome
3176	Spina bifida - hypospadias	211017	Spinocerebellar ataxia type 30	3275	Spondylocarpotarsal synostosis
268369	Spina bifida aperta	217012	Spinocerebellar ataxia type 31	94095	Spondylocostal dysostosis - anal and genitourinary malformations
53721	Spinal arteriovenous metameric syndrome	276183	Spinocerebellar ataxia type 32	329252	Spondylocostal dysostosis - hypospadias - intellectual disability
1217	Spinal atrophy - ophthalmoplegia - pyramidal syndrome	1955	Spinocerebellar ataxia type 34	1855	Spondyloenchondrodyplasia
90058	Spinal cord injury	276193	Spinocerebellar ataxia type 35		
73245	Spinal muscular atrophy - Dandy-Walker malformation - cataracts	276198	Spinocerebellar ataxia type 36		
1145	Spinal muscular atrophy with arthrogryposis	363710	Spinocerebellar ataxia type 37		
98920	Spinal muscular atrophy with respiratory distress type 1	423296	Spinocerebellar ataxia type 38		
404521	Spinal muscular atrophy with respiratory distress type 2	423275	Spinocerebellar ataxia type 40		
83420	Spinal muscular atrophy, adult form	363710	Spinocerebellar ataxia with altered vertical eye movements		
210584	Spindle cell hemangiobendothelioma				

→ 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
→1855	Spondyloenchondrodyplasia with immune dysregulation	163654	Spondyloepiphyseal dysplasia - brachydactyly - speech disorder	370019	Spondylometaphyseal dysplasia, Czarny-Ratajczak type
1855	Spondyloenchondromatosis	163649	Spondyloepiphyseal dysplasia - craniostenosis - cleft palate - cataract - intellectual disability	168544	Spondylometaphyseal dysplasia, Golden type
168451	Spondyloepimetaphyseal dysplasia - abnormal dentition	163668	Spondyloepiphyseal dysplasia - myopia - sensorineural deafness	93314	Spondylometaphyseal dysplasia, Kozlowski type
168443	Spondyloepimetaphyseal dysplasia - hypotrichosis	1830	Spondyloepiphyseal dysplasia - nephrotic syndrome	93316	Spondylometaphyseal dysplasia, Schmidt type
93358	Spondyloepimetaphyseal dysplasia - short limb - abnormal calcification	→93284	Spondyloepiphyseal dysplasia - punctate corneal dystrophy	93317	Spondylometaphyseal dysplasia, Sedaghatian type
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type	353298	Spondyloepiphyseal dysplasia - retinal dystrophy - immunodeficiency	93315	Spondylometaphyseal dysplasia, Sutcliffe type
93356	Spondyloepimetaphyseal dysplasia type 2	94068	Spondyloepiphyseal dysplasia congenita	1856	Spondyloperipheral dysplasia - short ulna
93360	Spondyloepimetaphyseal dysplasia with joint laxity, Hall type	93284	Spondyloepiphyseal dysplasia tarda	141	Spongy degeneration of the brain
93359	Spondyloepimetaphyseal dysplasia with joint laxity	1159	Spondyloepiphyseal dysplasia tarda - progressive arthropathy	54260	Spongy myocardium
93359	Spondyloepimetaphyseal dysplasia with joint laxity type 1	163665	Spondyloepiphyseal dysplasia tarda, Kohn type	443180	Spontaneous cerebrospinal fluid leak
93360	Spondyloepimetaphyseal dysplasia with joint laxity type 2	263463	Spondyloepiphyseal dysplasia with congenital joint dyslocations, CHST3 type	443180	Spontaneous intracranial hypotension
93360	Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type	→93284	Spondyloepiphyseal dysplasia, Byers type	29822	Spontaneous periodic hypothermia
93360	Spondyloepimetaphyseal dysplasia with multiple dislocations	163654	Spondyloepiphyseal dysplasia, Cantu type	247234	Sporadic adult-onset ataxia of unknown etiology
93360	Spondyloepimetaphyseal dysplasia with multiple dislocations, Hall type	93283	Spondyloepiphyseal dysplasia, Kimberley type	1665	Sporadic fetal brain disruption sequence
171866	Spondyloepimetaphyseal dysplasia, aggrecan type	163668	Spondyloepiphyseal dysplasia, MacDermot type	306776	Sporadic hyperekplexia
93347	Spondyloepimetaphyseal dysplasia, anauxetic type	263482	Spondyloepiphyseal dysplasia, Maroteaux type	225147	Sporadic IBSN
168448	Spondyloepimetaphyseal dysplasia, Bieganski type	163649	Spondyloepiphyseal dysplasia, Nishimura type	84271	Sporadic idiopathic nephrosis
168454	Spondyloepimetaphyseal dysplasia, Geneviève type	→26346	Spondyloepiphyseal dysplasia, Omani type	84271	Sporadic idiopathic steroid-resistant nephrotic syndrome
99642	Spondyloepimetaphyseal dysplasia, Handigodu type	163662	Spondyloepiphyseal dysplasia, Reardon type	97555	Sporadic idiopathic steroid-resistant nephrotic syndrome with collapsing glomerulopathy
93351	Spondyloepimetaphyseal dysplasia, Irapa type	168552	Spondylometaphyseal dysplasia - bowed forearms - facial dysmorphism	93222	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation
370015	Spondyloepimetaphyseal dysplasia, Isidor type	85167	Spondylometaphyseal dysplasia - cone-rod dystrophy	93220	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis
156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type	→1855	Spondylometaphyseal dysplasia with combined immunodeficiency	93218	Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis
93347	Spondyloepimetaphyseal dysplasia, Menger type	1855	Spondylometaphyseal dysplasia with enchondromatous changes	93218	Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis
93356	Spondyloepimetaphyseal dysplasia, Missouri type	93316	Spondylometaphyseal dysplasia with severe genu valgum	93221	Sporadic idiopathic steroid-resistant nephrotic syndrome with minimal changes
93282	Spondyloepimetaphyseal dysplasia, Pakistani type	168555	Spondylometaphyseal dysplasia, A4 type	611	Sporadic inclusion body myositis
93282	Spondyloepimetaphyseal dysplasia, PAPSS2 type	93316	Spondylometaphyseal dysplasia, Algerian type	225147	Sporadic infantile bilateral striatal necrosis
93352	Spondyloepimetaphyseal dysplasia, Shohat type	93315	Spondylometaphyseal dysplasia, 'corner fracture' type	225147	Sporadic infantile striatonigral degeneration
93357	Spondyloepimetaphyseal dysplasia, Sponastrime type			227510	Sporadic olivopontocerebellar atrophy type 1

→ 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
227510	Sporadic OPCA type 1	83484	St. Louis encephalitis	828	Stickler syndrome
276624	Sporadic pheochromocytoma	2454	Stalker-Chitayat syndrome	90653	Stickler syndrome type 1
276621	Sporadic pheochromocytoma/secretting paraganglioma	1798	Stanescu osteosclerosis	90654	Stickler syndrome type 2
		3235	Stapedo-vestibular ankylosis	166100	Stickler syndrome type 3
443057	Sporadic porphyria cutanea tarda	140917	Stapes ankylosis with broad thumbs and toes	166100	Stickler syndrome, non-ocular type
276627	Sporadic secreting paraganglioma	36238	Staphylococcal necrotizing pneumonia	3197	Stiff baby syndrome
826	Sporotrichosis	36236	Staphylococcal scalded skin syndrome	443804	Stiff leg syndrome
70594	SPR deficiency	36235	Staphylococcal scarlet fever	3198	Stiff man syndrome
94068	Spranger-Wiedemann disease	99919	Staphylococcal toxic-shock syndrome	3198	Stiff person syndrome and related disorders
3181	Sprengel deformity	99919	Staphylococcal TSS	2833	Stiff skin syndrome
70476	Spring catarrh	140952	STAR syndrome	85414	Still disease
234	Sprinz-Nelson syndrome	827	Stargardt 1	233	Stilling-Turk-Duane syndrome
3198	SPS	827	Stargardt disease	3199	Stimmler syndrome
1509	SPS	85146	Stark-Kaeser syndrome	425120	STING-associated vasculopathy with onset in infancy
431255	SPSMA	166427	Startle epilepsy	2972	Stoelinga-de Koomen-Davis syndrome
86884	SPTCL	391311	STAT1 deficiency	3200	Stoll-Alembik-Finck syndrome
51083	SQTS	2314	STAT3 deficiency	3074	Stoll-Géraudel-Chauvin syndrome
424019	Squamous cell carcinoma of anal canal	438159	STAT3-related early-onset multisystem autoimmune disease	3201	Stoll-Kieny-Dott syndrome
423994	Squamous cell carcinoma of colon	329284	Static encephalopathy of childhood with neurdegeneration in adulthood	2878	Stoll-Lévy-Francfort syndrome
99977	Squamous cell carcinoma of esophagus	413696	Statin toxicity	168577	Stomatin-deficient cryohydrocytosis
424996	Squamous cell carcinoma of gallbladder and extrahepatic biliary tract	841	Steatocystoma multiplex	98868	Stomatocytic elliptocytosis
424996	Squamous cell carcinoma of gallblader and EBT	3184	Steatocystoma multiplex - natal teeth	337	Stone man syndrome
67037	Squamous cell carcinoma of head and neck	438117	Steel syndrome	3204	Stormorken-Sjaastad-Langslet syndrome
424975	Squamous cell carcinoma of liver and IBT	240071	Steele-Richardson-Olszewski disease	99064	Straddling and/or overriding mitral valve
424975	Squamous cell carcinoma of liver and intrahepatic biliary tract	565	Steely hair disease	95461	Straddling or overriding tricuspid valve
424039	Squamous cell carcinoma of pancreas	565	Steely hair syndrome	1277	Stratton-Garcia-Young syndrome
398058	Squamous cell carcinoma of penis	273	Steinert disease	2863	Stratton-Parker syndrome
424002	Squamous cell carcinoma of rectum	273	Steinert myotonic dystrophy	99905	Streptobacillary rat-bite fever
423968	Squamous cell carcinoma of small bowel	3186	Steinfeld syndrome	99918	Streptococcal toxic-shock syndrome
423968	Squamous cell carcinoma of small intestine	168953	Stem cell leukemia/lymphoma	99918	Streptococcal TSS
418959	Squamous cell carcinoma of stomach	99087	Stenosis or atrophy of the coronary ostium	66529	Stress cardiomyopathy
213767	Squamous cell carcinoma of the cervix uteri	210115	Sterile multifocal osteomyelitis with periostitis and pustulosis	90041	Stress erythrocytosis
213716	Squamous cell carcinoma of the corpus uteri	3194	Stern-Lubinsky-Durrie syndrome	90041	Stress polycythemia
324737	SRD5A3-CDG	3195	Sternal malformation - vascular dysplasia	50942	Striate palmoplantar keratoderma
83601	SREAT	753	Steroid 5-alpha-reductase deficiency	137599	Stromal keratitis
330001	SSA	3196	Steroid dehydrogenase deficiency - dental anomalies	213711	Stromal sarcoma of the corpus uteri
2806	SSPE	461	Steroid sulfatase deficiency	76	Strongyloidiasis
50944	SSPS	83601	Steroid-responsive encephalopathy associated with autoimmune thyroiditis	100984	Strümpell disease
370927	SSR4-CDG	93207	Steroid-sensitive MCNS	370921	STT3A-CDG
2323	SSS	→69061	Steroid-sensitive nephrotic syndrome without renal biopsy	370924	STT3B-CDG
36236	SSSS	909	Sterol 27-hydroxylase deficiency	328	Stuart-Prower factor deficiency
		46059	Sterol C5-desaturase deficiency	3205	Sturge-Weber syndrome
		36426	Stevens-Johnson syndrome	3205	Sturge-Weber-Dimitri syndrome
				3205	Sturge-Weber-Krabbe angiomas
				3205	Sturge-Weber-Krabbe syndrome
				3206	Stüve-Wiedemann dysplasia
				3206	Stüve-Wiedemann syndrome
				166277	Suarez-Stickler 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
101029	Sub-cortical nodular heterotopia	702	Sudanophilic leukodystrophy, Paelizeus-Merzbacher type	284113	Susceptibility to adverse reaction due to mercaptopurine
79093	Subacute angiohypertrophic myelomalacia	168593	Sudden infant death - dysgenesis of the testes	2566	Susceptibility to chronic infection by Epstein-Barr virus
79093	Subacute ascending necrotizing myelitis	130	Sudden unexplained nocturnal death syndrome	331226	Susceptibility to infection due to TYK2 deficiency
163525	Subacute cutaneous lupus erythematosus	2752	Sugarman syndrome	449306	Susceptibility to infection in immunocompromised patient
2806	Subacute inclusion body encephalitis	3412	Sujansky-Leonard syndrome	447740	Susceptibility to localized juvenile periodontitis
206594	Subacute inflammatory demyelinating polyneuropathy	99732	Sulfite oxidase deficiency due to molybdenum cofactor deficiency	169085	Susceptibility to respiratory infections associated with CD8alpha chain mutation
206594	Subacute inflammatory demyelinating polyradiculoneuropathy	308386	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A	391311	Susceptibility to viral and mycobacterial infections
98824	Subacute myeloid leukemia	308393	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type B	319269	Susceptibility/resistance to HIV infection
79093	Subacute necrotizing myelitis	308400	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type C	3193	SVAS
2806	Subacute sclerosing leukoencephalitis	99731	Sulfocysteinuria	86813	Sveinsson chorioretinal atrophy
2806	Subacute sclerosing panencephalitis	65682	Summerskill-Walshe-Tygstrup syndrome	3243	Sweet syndrome
356	Subacute spongiform encephalopathy, Gerstmann-Straussler type	254395	Summertime actinic lichenoid eruption	247165	Swift disease
99113	Subaortic course of brachiocephalic vein	3210	Summitt syndrome	247165	Swift-Feir disease
99113	Subaortic course of innominate vein	57145	SUNCT syndrome	3205	SWS
3191	Subaortic stenosis - short stature	130	SUNDS	242	Swyer syndrome
48377	Subcorneal pustular dermatitis	455	Superficial epidermolytic ichthyosis	90038	Sxt-HUS
48377	Subcorneal pustular dermatosis	98961	Superficial granular corneal dystrophy	306731	Sydenham chorea
99796	Subcortical band heterotopia	79490	Superficial lymphangioma	295138	Symbrachydactyly of hand and foot, bilateral
313808	Subcortical gliosis of Neumann	79490	Superficial lymphatic malformation	295136	Symbrachydactyly of hand and foot, unilateral
99796	Subcortical laminar heterotopia	247245	Superficial siderosis	1570	Symbrachydactyly of hands and feet
86884	Subcutaneous panniculitic T-cell lymphoma	88633	Superior limbic keratoconjunctivitis	60015	Symmetric parietal foramina
86884	Subcutaneous panniculitis-like T-cell lymphoma	155884	Superior palpebral coloboma	1314	Symmetrical thalamic calcifications
251618	Subependymal giant cell astrocytoma	180182	Supernumerary breasts	79098	Sympathetic ophthalmia
101030	Subependymal nodular heterotopia	96170	Supernumerary der(22) syndrome	79098	Sympathetic uveitis
251639	Subependymoma	141096	Supernumerary nostril	635	Sympathoblastoma
98957	Subepithelial amyloidosis of the cornea	295002	Supernumerary phalanges	3237	Symphalangism - brachydactyly
98959	Subepithelial mucinous corneal dystrophy	295142	Supernumerary phalanges, bilateral	3246	Symphalangism with multiple anomalies of hands and feet
155878	Submucosal cleft palate	295140	Supernumerary phalanges, unilateral	3250	Symphalangism, Cushing type
3190	Subpulmonary stenosis	295002	Supernumerary phalanx	276630	Symptomatic form of Coffin-Lowry syndrome in female carriers
1606	Subtelomeric 1p36 deletion	295142	Supernumerary phalanx, bilateral	449291	Symptomatic form of fragile X syndrome in female carrier
96168	Subtelomeric deletion 13q34	295140	Supernumerary phalanx, unilateral	177926	Symptomatic form of hemophilia A in female carriers
180129	Subtotal septate uterus	1450	Supernumerary ring/marker 8	177929	Symptomatic form of hemophilia B in female carriers
→2609	Succinic acidemia	1461	Superoinferior ventricles	206546	Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers
22	Succinic semialdehyde dehydrogenase deficiency	764	Suppurative myositis	357332	Synactyly - camptodactyly and clinodactyly of fifth fingers - bifid halluces
		3193	Supravalvular aortic stenosis		
		3192	Supravalvular pulmonary stenosis		
		391351	SURF1-related Charcot-Marie-Tooth disease type 4		
		391351	SURF1-related CMT4		
		391351	SURF1-related severe demyelinating Charcot-Marie-Tooth disease		
		838	Susac 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
98915	Synaptic congenital myasthenic syndromes	77299	Syndromic microphthalmia type 10	314701	Systemic immunoglobulinic amyloidosis
357332	Syndactyly - camptodactyly and clinodactyly of fifth fingers - bifid toes	178364	Syndromic microphthalmia/anophthalmia due to OTX2 mutation	401996	Systemic karyomegaly
85203	Syndactyly - preaxial polydactyly - sternal deformity	228426	Syndromic multisystem autoimmune disease due to Itch deficiency	98849	Systemic mastocytosis with an associated clonal hematologic non-mast cell lineage disease
140952	Syndactyly - telecanthus - anogenital and renal malformations	98606	Syndromic orbital border hypoplasia	90069	Systemic monochloroacetate poisoning
93404	Syndactyly of fingers 4 and 5	281090	Syndromic recessive X-linked ichthyosis	439762	Systemic PAN
93402	Syndactyly type 1	281090	Syndromic RXLI	439762	Systemic periarteritis nodosa
3255	Syndactyly type 1 - microcephaly - intellectual disability	281090	Syndromic X-linked ichthyosis	439762	Systemic polyarteritis nodosa
295193	Syndactyly type 1, Castilla type	85274	Syndromic X-linked intellectual disability 7	85414	Systemic polyarthritides
295189	Syndactyly type 1, Lueken type	85279	Syndromic X-linked intellectual disability due to JARID1C mutation	158	Systemic primary carnitine deficiency
295191	Syndactyly type 1, Montagu type	85295	Syndromic X-linked intellectual disability type 10	90291	Systemic scleroderma
295187	Syndactyly type 1, Weidenreich type	85286	Syndromic X-linked intellectual disability type 11	90291	Systemic sclerosis
295187	Syndactyly type 1a	319332	SYNE1-related AMC	220407	Systemic sclerosis sine scleroderma
295189	Syndactyly type 1b	319332	SYNE1-related arthrogryposis multiplex congenita	85414	Systemic-onset JIA
295191	Syndactyly type 1c	3263	Syngnathia - cleft palate	85414	Systemic-onset juvenile idiopathic arthritis
295193	Syndactyly type 1d	3262	Syngnathia multiple anomalies	134	T2 deficiency
93403	Syndactyly type 2	3268	Synostosis - microcephaly - scoliosis	99861	T-ALL
93404	Syndactyly type 3	35098	Synostotic plagiocephaly	169160	T-B+ SCID due to CD3delta/CD3epsilon/CD3zeta
93405	Syndactyly type 4	3273	Synovial sarcoma	169157	T-B+ SCID due to CD45 deficiency
93406	Syndactyly type 5	3273	Synovialosarcoma	276	T-B+ SCID due to gamma chain deficiency
295012	Syndactyly type 6	793	Synovitis-acne-pustulosis-hyperostosis-osteitis syndrome	169154	T-B+ SCID due to IL-7Ralpha deficiency
3258	Syndactyly type 7	93403	Synpolydactyly	35078	T-B+ SCID due to JAK3 deficiency
2498	Syndactyly type 8	295195	Synpolydactyly type 1	169160	T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta
157801	Syndactyly type 9	295197	Synpolydactyly type 2	169157	T-B+ severe combined immunodeficiency due to CD45 deficiency
157801	Syndactyly, Malik-Percin type	295199	Synpolydactyly type 3	276	T-B+ severe combined immunodeficiency due to gamma chain deficiency
295012	Syndactyly, mitten type	295197	Synpolydactyly, Debeer type	169154	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency
3253	Syndactyly-ectodermal dysplasia-cleft/lip palate	295199	Synpolydactyly, Malik type	35078	T-B+ severe combined immunodeficiency due to JAK3 deficiency
3259	Syndactyly-polydactyly-ear lobe syndrome	295195	Synpolydactyly, Vordingborg type	169160	T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta
→1159	Syndesmodyplastic dwarfism	3275	Synspondylism	169157	T-B+ severe combined immunodeficiency due to CD45 deficiency
2143	Syndrome of ocular and facial anomalies, telecanthus and deafness	93926	Syntelencephaly	276	T-B+ severe combined immunodeficiency due to gamma chain deficiency
52	Syndromic bile duct paucity	840	Syringadenoma papilliferum	169154	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency
261619	Syndromic bile duct paucity due to a JAG1 point mutation	840	Syringocystadenoma papilliferum	35078	T-B+ severe combined immunodeficiency due to JAK3 deficiency
261629	Syndromic bile duct paucity due to a NOTCH2 point mutation	314701	Systemic AL amyloidosis	276	T-B+ severe combined immunodeficiency, X-linked
261600	Syndromic bile duct paucity due to monosomy 20p12	2039	Systemic arteriovenous fistula	86871	T-cell chronic lymphocytic leukemia
84064	Syndromic diarrhea	188	Systemic capillary leak syndrome	324294	T-cell immunodeficiency due to RHOH deficiency
84064	Syndromic diarrhea/Tricho-hepatointestinal syndrome	→528	Systemic cystic angiomas - Seip syndrome	324294	T-cell immunodeficiency with epidermodysplasia verruciformis
77298	Syndromic microphthalmia type 3	364033	Systemic EBV+ T-cell LPD of childhood	86872	T-cell large granular lymphocyte leukemia
85275	Syndromic microphthalmia type 4	364033	Systemic EBV-positive T-cell lymphoproliferative disease of childhood	86872	T-cell LGL leukemia
178364	Syndromic microphthalmia type 5	364033	Systemic Epstein-Barr virus-positive T-cell lymphoproliferative disease of childhood	86886	T-cell lymphoma, AILD type
139471	Syndromic microphthalmia type 6				
2556	Syndromic microphthalmia type 7				
3434	Syndromic microphthalmia type 8				
2470	Syndromic microphthalmia type 9				

→ 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
86871	T-cell prolymphocytic leukemia	2636	Taybi-Linder syndrome	1745	Telomeric duplication 6p
300857	T-cell/histiocyte rich large B cell lymphoma	98960	TBCD	96098	Telomeric duplication 6q
		857	TBS	96074	Telomeric duplication 7p
86872	T-LGL	103918	TCP	96100	Telomeric duplication 8q
86871	T-PLL	397959	TCR-alpha-beta+ T-cell deficiency	96101	Telomeric duplication 9q
1350	Tabatznik syndrome	397959	TCR-alpha-beta-positive T-cell deficiency	96102	Telomeric duplication 10q
3384	TAC	2655	TD	96103	Telomeric duplication 11q
447896	TACH	1860	TD1	96105	Telomeric duplication 13q
241043	Tacrolimus dose selection	93274	TD2	1705	Telomeric duplication 14q
567	Takao syndrome	3352	TDO syndrome	1707	Telomeric duplication 15q
2905	Takatsuki syndrome	1519	Teebi hypertelorism syndrome	96078	Telomeric duplication 16p
3287	Takayasu arteritis	1519	Teebi syndrome	96106	Telomeric duplication 16q
66529	Tako-Tsubo cardiomyopathy	2432	Teebi-Al Saleh-Hassoon syndrome	3379	Telomeric duplication 17q
66529	Tako-tsubo syndrome	1094	Teebi-Kaurah syndrome	1716	Telomeric duplication 18q
66529	Takotsubo cardiomyopathy	1974	Teebi-Naguib-Alawadi syndrome	1717	Telomeric duplication 19q
66529	Takotsubo syndrome	3291	Teebi-Shaltout syndrome	96107	Telomeric duplication 20q
101028	TALDO	3292	Tel Hashomer camptodactyly syndrome	96109	Telomeric duplication 22q
2886	Talipes equinovarus - atrial septal defect - Robin sequence - Persistence of the left superior vena cava	284227	Telangiectasia - erythrocytosis - monoclonal gammopathy - perinephric-fluid collections - intrapulmonary shunting	1762	Telomeric duplication Xq
217335	Tall forehead-sparse hair-skin hyperextensibility-scoliosis syndrome	90389	Telangiectasia macularis eruptiva perstans	1620	Telomeric monosomy 3p
329191	Tall stature - scoliosis - macrodactyly of the great toes	3293	Telecanthus - hypertelorism - strabismus - pes cavus	75565	TEMF
329191	Tall stature - scoliosis - macrodactyly of the halluces	2885	Telfer-Sugar-Jaeger syndrome	352737	Temperature-sensitive oculocutaneous albinism type 1
404443	Tall stature-intellectual disability-facial dysmorphism syndrome	1596	Telomeric 15q deletion syndrome	284227	TEMPI syndrome
50809	Talo-patello-scaphoid osteolysis	36367	Telomeric deletion 1q	420561	Temple-Baraitser syndrome
31150	Tangier disease	280	Telomeric deletion 4p	397	Temporal arteritis
180	Tapetochoroidal dystrophy	96145	Telomeric deletion 4q	363417	Temtamy preaxial brachydactyly syndrome
98839	Tappeiner-Pfleger disease	1627	Telomeric deletion 5q	1777	Temtamy syndrome
3320	TAR syndrome	96126	Telomeric deletion 7p	1777	Temtamy-Shalash syndrome
65250	Tarlov cyst	1636	Telomeric deletion 7q36	66627	Tenosynovial giant cell tumor
2886	TARP syndrome	1642	Telomeric deletion 9p	137834	Ter Haar syndrome
99170	Tarsal kink syndrome	1580	Telomeric deletion 10p	883	Teratoma
1412	Tarsal-carpal coalition syndrome	96148	Telomeric deletion 10q	252018	Teratoma of the central nervous system
371	Tarui disease	2308	Telomeric deletion 11q	141107	Teratoma of the nasopharynx
163654	Tattoo dysplasia	96149	Telomeric deletion 12q	363483	Teratoma of the testis
2731	Taurodontia - absent teeth - sparse hair	96150	Telomeric deletion 14q	88630	Terminal osseous dysplasia - pigmentary defects
3289	Taurodontism	531	Telomeric deletion 17p	93937	Terminal transverse defects of arm
99045	Taussig-Bing syndrome	1597	Telomeric deletion 17q	141242	Tessier number 1 cleft
→33364	Tay syndrome	96129	Telomeric deletion 19p	141258	Tessier number 4 facial cleft
845	Tay-Sachs disease	96152	Telomeric deletion 20q	141261	Tessier number 5 facial cleft
309239	Tay-Sachs disease, B1 variant	1590	Telomeric deletion13q	141265	Tessier number 6 facial cleft
309192	Tay-Sachs disease, B variant, adult form	96069	Telomeric duplication 1p36	325124	Testicular agenesis
309178	Tay-Sachs disease, B variant, infantile form	96070	Telomeric duplication 2p	363494	Testicular non seminomatous germ cell tumor
309185	Tay-Sachs disease, B variant, juvenile form	96094	Telomeric duplication 2q	363494	Testicular non-dysgerminomatous germ cell tumor
669	Taybi syndrome	96071	Telomeric duplication 3p	983	Testicular regression syndrome
90650	Taybi syndrome	96072	Telomeric duplication 4p	842	Testicular seminoma
		96096	Telomeric duplication 4q	842	Testicular seminomatous germ cell tumor
		96097	Telomeric duplication 5q	363489	Testicular sex cord-stromal tumor

→ 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
363483	Testicular teratoma	88633	Theodore's syndrome	67044	Thrombocytopenia with congenital dyserythropoietic anemia
3000	Testotoxicosis	268184	Thiamine-responsive BCKD deficiency	3002	Thrombocytopenic purpura, autoimmune
3299	Tetanus	268184	Thiamine-responsive branched-chain 2-ketoacid dehydrogenase deficiency	436169	Thrombomodulin-related bleeding disorder
9	Tetra X	199348	Thiamine-responsive encephalopathy	436169	Thrombomodulin-related coagulopathy
294971	Tetra-amelia	268184	Thiamine-responsive maple syrup urine disease	54057	Thrombotic thrombocytopenic purpura
3301	Tetraamelia - multiple malformations	49827	Thiamine-responsive megaloblastic anemia syndrome	2251	Thumb deformity - alopecia - pigmentation anomaly
199310	Tetragametic chimerism	49827	Thiamine-responsive megaloblastic anemia with diabetes mellitus and sensorineural deafness	294988	Thumb hypodactyly
293284	Tetrahydrobiopterin-responsive HPA/PKU	268184	Thiamine-responsive MSUD	295112	Thumb hypodactyly, bilateral
293284	Tetrahydrobiopterin-responsive hyperphenylalaninemia/phenylketonuria	2405	Thickened earlobes - conductive deafness	295110	Thumb hypodactyly, unilateral
3303	Tetralogy of Fallot	98960	Thiel-Behnke corneal dystrophy	294988	Thumb oligodactyly
2564	Tetramelic monodactyly	3314	Thiemann disease, familial form	295112	Thumb oligodactyly, bilateral
3305	Tetraploidy	3235	Thies-Reis syndrome	295110	Thumb oligodactyly, unilateral
3309	Tetrasomy 5p	1506	Thin ribs - tubular bones - dysmorphism	1078	Thumb stiffness - brachydactyly - intellectual disability
3310	Tetrasomy 9p	166424	Thinking seizures	2919	Thurston syndrome
289522	Tetrasomy 11q24.1	→300	Thiolase deficiency	83471	Thymic aplasia
884	Tetrasomy 12p	3315	Thiopurine S-methyltransferase deficiency	99868	Thymic carcinoma
314588	Tetrasomy 15(q25-qter)	141030	Third branchial cleft anomaly	97289	Thymic endocrine tumor
314588	Tetrasomy 15q26	141030	Third branchial cleft cyst	99869	Thymic neuroendocrine carcinoma
3307	Tetrasomy 18p	141030	Third branchial cleft fistula	97289	Thymic neuroendocrine tumor
96055	Tetrasomy 21	3316	Thomas syndrome	3326	Thymic-renal-anal-lung dysplasia
9	Tetrasomy X	2547	Thomas-Jewett-Raines syndrome	99867	Thymoma
140917	Teunissen-Cremers syndrome	2031	Thompson-Baraitser syndrome	263310	Thymoma type A
746	TFP deficiency	614	Thomsen and Becker disease	263324	Thymoma type AB
746	TFPD	2866	Thong-Douglas-Ferrante syndrome	263317	Thymoma type B
225123	TFR2-related hemochromatosis	1861	Thoracic dysplasia-hydrocephalus syndrome	169105	Thymoma-immunodeficiency syndrome
216729	TGA with cardiac malformation	97330	Thoracic outlet compression syndrome	3327	Thycerebrorenal syndrome
99042	TGA with coarctation	97330	Thoracic outlet syndrome	95716	Thyroid dyshormonogenesis
66627	TGCT	1759	Thoraco-abdominal enteric duplication	95712	Thyroid ectopia
3329	TH-SHFM	1335	Thoraco-abdominal syndrome	95719	Thyroid hemiogenesis
1780	Thakker-Donnai syndrome	3317	Thoracolaryngopelvic dysplasia	95720	Thyroid hypoplasia
3312	Thalidomide embryopathy	268384	Thoracolumbosacral spina bifida aperta	97285	Thyroid lymphoma
2655	Thanatophoric dwarfism	268752	Thoracolumbosacral spina bifida cystica	91347	Thyroid stimulating hormone-secreting pituitary adenoma
93274	Thanatophoric dwarfism - cloverleaf skull	1803	Thoracomelic dysplasia	2091	Thyroid-renal-digital anomalies
1860	Thanatophoric dwarfism type 1	→2199	Thost-Unna palmoplantar keratoderma	79102	Thyrotoxic hypokalemic periodic paralysis
93274	Thanatophoric dwarfism type 2	300857	THRLBCL	79102	Thyrotoxic periodic paralysis
2655	Thanatophoric dysplasia	36258	Thromboangiitis obliterans	91347	Thyrotroph adenoma
1860	Thanatophoric dysplasia type 1	3204	Thrombocytopathy - asplenia - miosis	2768	Tibia vara Blount
93274	Thanatophoric dysplasia type 2	3320	Thrombocytopenia - absent radius	3329	Tibial aplasia - ectrodactyly
→175	Thanatophoric dysplasia, Glasgow variant	3323	Thrombocytopenia - Robin sequence	93322	Tibial hemimelia
436169	THBD-related bleeding disorder			3329	Tibial hemimelia with split hand/foot malformation
436169	THBD-related coagulopathy			295079	Tibial hemimelia, bilateral
99917	Theca (steroid-producing) cell cancer, not further specified			295077	Tibial hemimelia, unilateral
99917	Theca steroid-producing cell malignant tumor of ovary, not further specified				
88633	Theodore's superior limbic keratoconjunctivitis				

→ 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
3329	Tibial hemimelia-ectrodactyly syndrome	268377	Total spina bifida aperta	289877	Transient hyperammonemia of the newborn
93322	Tibial longitudinal meromelia	268748	Total spina bifida cystica	169139	Transient hypogammaglobulinemia of infancy
295079	Tibial longitudinal meromelia, bilateral	2796	Touraine-Solente-Gole syndrome	300293	Transient infantile hypertriglyceridemia and fatty liver
295077	Tibial longitudinal meromelia, unilateral	857	Townes syndrome	300293	Transient infantile hypertriglyceridemia and hepatosteatosis
609	Tibial muscular dystrophy	857	Townes-Brocks syndrome	66529	Transient left ventricular apical ballooning syndrome
295028	Tibio-fibular fusion	95455	Toxic epidermal necrolysis	420611	Transient myeloproliferative disease
295028	Tibio-fibular synostosis	95455	Toxic epidermolysis	420611	Transient myeloproliferative syndrome
294981	Tibiofibular terminal transverse meromelia	279894	Toxic maculopathy due to antimalarial drugs	391504	Transient neonatal acquired myasthenia
295099	Tibiofibular terminal transverse meromelia, bilateral	227972	Toxic oil syndrome	391504	Transient neonatal autoimmune myasthenia gravis
295097	Tibiofibular terminal transverse meromelia, unilateral	293173	Toxic pustuloderma	280615	Transient neonatal cyanosis and anemia due to Toms River Hemoglobin
297	Tick-borne encephalitis	284121	Toxicity or absent response to clozapine	99886	Transient neonatal diabetes mellitus
42665	Tietz syndrome	230800	Toxin-mediated infectious botulism	329942	Transient neonatal glutaric aciduria type 2
1662	Tight skin contracture syndrome	230800	Toxin-mediated infective botulism	329942	Transient neonatal glutaric aciduria type 2
65283	Timothy syndrome	3343	Toxocariasis	329942	Transient neonatal MAD deficiency
91500	TINU syndrome	858	Toxoplasma embryofetopathy	329942	Transient neonatal MADD
352540	TIO	858	Toxoplasma embryopathy	329942	Transient neonatal multiple acyl-CoA dehydrogenase deficiency
→1394	TMCO1 defect syndrome	93164	TPHA	391504	Transient neonatal myasthenia gravis
420611	TMD	444463	TPP11 deficiency	93164	Transient pseudohypoaldosteronism
609	TMD	444463	TPP11-related immunodeficiency, autoimmunity, and neurodevelopmental delay with impaired glycolysis and lysosomal expansion disease	3402	Transient tyrosinemia of the neonate
314667	TMEM165-CDG	2950	TPT-PS syndrome	3402	Transient tyrosinemia of the newborn
1194	TMEM70-related mitochondrial encephalo-cardio-myopathy	412022	Traboulsi syndrome	213746	Transitional cell carcinoma of the corpus uteri
99886	TNDM	3346	Tracheal agenesis	280224	Transitional PMD
32960	TNF receptor 1-associated periodic syndrome	2042	Tracheo-esophageal fistula - hypospadias	319308	Translocation carcinoma
64686	Tolosa-Hunt syndrome	3347	Tracheobronchomegaly	319308	Translocation renal cell carcinoma
1920	Toluene embryopathy	3348	Tracheobronchopathia osteochondroplastica	85451	Transthyretin amyloid cardiopathy
640	Tomaculous neuropathy	3348	Tracheopathia osteoplastica	85447	Transthyretin amyloid neuropathy
→31463 2	Tomé-Brunet-Fardeau syndrome	3052	Tranebjærg-Svejgaard syndrome	85447	Transthyretin amyloid polyneuropathy
1547	Tonoki-Ohura-Niikawa syndrome	101028	Transaldolase deficiency	85451	Transthyretin-related familial amyloid cardiomyopathy
2228	Tooth and nail syndrome	859	Transcobalamin deficiency	2486	Transverse limb deficiency - hemangioma
3460	Torg-Winchester syndrome	859	Transcobalamin II deficiency	180160	Transverse vaginal septum
1827	Toriello syndrome	199247	Transcortin deficiency	32960	TRAPS syndrome
3338	Toriello-Carey syndrome	495	Transgrediens et progrediens palmoplantar keratoderma	399175	Traumatic avascular necrosis
79347	Toriello-Higgins-Miller syndrome	495	Transgrediens et progrediens PPK	399175	Traumatic AVN
3339	Toriello-Lacassie-Droste syndrome	87503	Transgrediens palmoplantar keratoderma of Siemens	861	Treacher-Collins syndrome
51084	Torsade-de-pointes syndrome with short coupling interval	420611	Transient abnormal myelopoiesis	→1215	Treft-Sanborn-Carey syndrome
3341	Torticollis - keloids - cryptorchidism - renal dysplasia	98871	Transient acquired pure red cell aplasia	3350	Tremor - nystagmus - duodenal ulcer
75326	Tortuosity of retinal arteries	79411	Transient bullous dermolytic of the newborn	447896	Tremor-ataxia-central hypomyelination syndrome
97330	TOS	98871	Transient erythroblastopenia of childhood		
2701	Tosti syndrome	2312	Transient familial neonatal hyperbilirubinemia		
294971	Total amelia				
49382	Total color blindness				
98994	Total early-onset cataract				
180126	Total septate uterus				

→ 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
64694	Trench fever	3368	Trigonocephaly - bifid nose - acral anomalies	171929	Trisomy 10p
1822	Trevor disease	3365	Trigonocephaly - broad thumbs	276422	Trisomy 10q22.3q23.3
99832	TRH resistance syndrome	3369	Trigonocephaly - short stature - developmental delay	96102	Trisomy 10qter
2970	Triad syndrome	1308	Trigonocephaly C syndrome	300305	Trisomy 11p15.4
444463	TRIANGLE disease	401764	Trilineage bone marrow failure- developmental delay syndrome	96103	Trisomy 11qter
85170	Triangular tibia - fibular aplasia	3374	Triopia	1699	Trisomy 12p
863	Trichinellosis	868	Triose phosphate-isomerase deficiency	3378	Trisomy 13
863	Trichinosis	444463	Tripeptidyl-peptidase II deficiency	96105	Trisomy 13qter
3352	Tricho-dento-osseous syndrome	2950	Triphalangeal thumb - polysyndactyly syndrome	261229	Trisomy 14q11.2
84064	Tricho-hepato-enteric syndrome	2947	Triphalangeal thumbs - brachyectrodactyly	1705	Trisomy 14qter
3354	Tricho-oculo-dermo-vertebral syndrome	3133	Triphalangeal thumbs - dislocation of patella	238446	Trisomy 15q11-q13
1264	Tricho-retino-dento-digital syndrome	869	Triple A syndrome	238446	Trisomy 15q11q13
3351	Trichodental syndrome	415	Triple H syndrome	1707	Trisomy 15qter
3360	Trichodermal syndrome - intellectual disability	3375	Triple X syndrome	261204	Trisomy 16p11.2p12.2
3353	Trichodermodysplasia - dental alterations	3375	Triple-X syndrome	261243	Trisomy 16p13.11
79129	Trichodysplasia - amelogenesis imperfecta	3376	Triploidy	96078	Trisomy 16pter
3361	Trichodysplasia - xeroderma	3377	Trismus - pseudocampodactyly	96106	Trisomy 16qter
228379	Trichodysplasia spinulosa	96069	Trisomy 1pter	261290	Trisomy 17p
864	Trichofolliculoma	261344	Trisomy 1q	1713	Trisomy 17p11.2
3363	Trichomegaly - retina pigmentary degeneration - dwarfism	250994	Trisomy 1q21.1	217385	Trisomy 17p13.3
3355	Trichoodontoonychial dysplasia	96070	Trisomy 2pter	139474	Trisomy 17q11.2
3355	Trichoodontoonychial dysplasia with bone deficiency in frontoparietal region	313947	Trisomy 2q23.1	261272	Trisomy 17q12
565	Trichopoliodystrophy	294026	Trisomy 2q31.1	217340	Trisomy 17q21.31
77258	Trichorhinophalangeal syndrome type 1 and 3	96094	Trisomy 2qter	3379	Trisomy 17qter
502	Trichorhinophalangeal syndrome type 2	96071	Trisomy 3pter	3380	Trisomy 18
→33364	Trichorrhexis nodosa syndrome	96095	Trisomy 3q26	1715	Trisomy 18p
33364	Trichothiodystrophy	251038	Trisomy 3q29	1716	Trisomy 18qter
→33364	Trichothiodystrophy - neurocutaneous syndrome	1738	Trisomy 4p	1717	Trisomy 19qter
→33364	Trichothiodystrophy - osteosclerosis	96072	Trisomy 4pter	261318	Trisomy 20p
→33364	Trichothiodystrophy - sun sensitivity	96096	Trisomy 4qter	96107	Trisomy 20qter
→33364	Trichothiodystrophy type B	1742	Trisomy 5p	1727	Trisomy 22q11.2
→33364	Trichothiodystrophy type C	329802	Trisomy 5p13	96109	Trisomy 22qter
→33364	Trichothiodystrophy type D	228415	Trisomy 5q35	1738	Trisomy of the short arm of chromosome 4
→33364	Trichothiodystrophy type E	96097	Trisomy 5qter	1742	Trisomy of the short arm of chromosome 5
→33364	Trichothiodystrophy type F	1745	Trisomy 6pter	236	Trisomy of the short arm of chromosome 9
→33364	Trichothiodystrophy type G	96098	Trisomy 6qter	1715	Trisomy of the short arm of chromosome 18
→33364	Trichothiodystrophy with congenital ichthyosis	314034	Trisomy 7p22.1	3375	Trisomy X
1209	Tricuspid atresia	96074	Trisomy 7pter	217377	Trisomy Xp11.22-p11.23
95457	Tricuspid valve agenesis	96121	Trisomy 7q11.23	261483	Trisomy Xq27.3-q28
95458	Tricuspid valve prolapse	264450	Trisomy 8p	261483	Trisomy Xq27.3q28
221091	Trigeminal neuralgia	251076	Trisomy 8p23.1	1762	Trisomy Xq28
98908	Triglyceride deposit cardiomyovasculopathy	1752	Trisomy 8q	88629	Tritan colour blindness
		228399	Trisomy 8q12	88629	Tritanopia
		96100	Trisomy 8qter	49827	TRMA
		236	Trisomy 9p	1349	tRNA-LYS-related cardiomyopathy - hearing loss
		96101	Trisomy 9qter	1863	Trochlear dysplasia
				103918	Tropical calcific chronic pancreatitis
				75565	Tropical endomyocardial fibrosis

→ 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
99654	Tropical pancreatic diabetes	99053	Tunnel subaortic stenosis	79235	UGT deficiency type 2
103918	Tropical pancreatitis	211	Turban tumor syndrome	3403	Uhl anomaly
764	Tropical pyomyositis	99818	Turcot syndrome with polyposis	2032	UIP
289326	Tropical spastic paraparesis	881	Turner syndrome	3404	Ulbright-Hodes syndrome
101000	Troyer syndrome	99413	Turner syndrome due to structural X chromosome anomalies	308	ULD
983	TRS	2614	Turner-Kieser syndrome	3406	Ulerythema ophryogenes
313906	True congenital pancreatic cyst	63440	Turricephaly	320	Ulick syndrome
2138	True hermaphroditism	79153	Twenty-nail dystrophy	75840	Ullrich disease
2512	True microcephaly	95431	Twin to twin transfusion syndrome	2497	Ulna hypoplasia
180074	True unicornuate uterus	1461	Twisted atrioventricular connections	2249	Ulna hypoplasia - intellectual disability
3357	Trueb-Burg-Bottani syndrome	2889	Twisted hair	1837	Ulna metaphyseal dysplasia syndrome
3384	Truncus arteriosus	2198	Tylosis - oesophageal carcinoma	93320	Ulnar clubhand
228379	TS	→79259	Type 1C glycogenosis	93320	Ulnar hemimelia
352737	TS OCA type 1	→79259	Type 1D glycogenosis	295073	Ulnar hemimelia, bilateral
3173	Tsao-Ellingson syndrome	93554	Type II mixed cryoglobulinemia	295075	Ulnar hemimelia, unilateral
66627	TSGCT	99745	Typhoid	1122	Ulnar hypoplasia - lobster-claw deformity of feet
91347	TSH-oma	99745	Typhoid fever	1122	Ulnar hypoplasia - split foot
91347	TSH-secreting pituitary adenoma	99038	Typical hemolytic-uremic syndrome	93320	Ulnar longitudinal meromelia
289326	TSP	90038	Typical HUS	295073	Ulnar longitudinal meromelia, bilateral
3268	Tsukahara syndrome	171436	Typical nemaline myopathy	295075	Ulnar longitudinal meromelia, unilateral
3387	Tsukahara-Kajii syndrome	158766	Typical urticaria pigmentosa	3138	Ulnar-mammary syndrome
83317	Tsutsugamushi disease	1895	Typus Edinburgensis	52056	Ulnar/fibula ray defect - brachydactyly
83317	Tsutsugamushi fever	79431	Tyrosinase-negative oculocutaneous albinism	3405	Umbilical cord ulceration - intestinal atresia
54057	TTP	101150	Tyrosine hydroxylase deficiency	209886	UMOD-associated familial juvenile hyperuricemic nephropathy
85447	TTR amyloid neuropathy	101150	Tyrosine hydroxylase-deficient dopamine-responsive dystonia	209886	UMOD-associated FJHN
85451	TTR-related amyloid cardiomyopathy	69723	Tyrosinemia due to 4-hydroxyphenylpyruvate dioxygenase deficiency	35120	UMPH1 deficiency
85451	TTR-related cardiac amyloidosis	69723	Tyrosinemia due to 4-hydroxyphenylpyruvic acid oxidase deficiency	3138	UMS
180242	Tubal cancer	69723	Tyrosinemia due to HPD deficiency	86830	Unclassified chronic myeloproliferative disease
3389	Tuberculosis	28378	Tyrosinemia due to TAT deficiency	104078	Unclassified intestinal pseudoobstruction
805	Tuberous sclerosis	28378	Tyrosinemia due to tyrosine aminotransferase deficiency	98825	Unclassified mixed myelodysplastic/myeloproliferative syndrome
805	Tuberous sclerosis complex	882	Tyrosinemia type 1	98827	Unclassified myelodysplastic syndrome
88924	Tuberous sclerosis/polycystic kidney disease contiguous gene syndrome	28378	Tyrosinemia type 2	98825	Unclassified myelodysplastic/myeloproliferative disease
2593	Tubular aggregate myopathy	69723	Tyrosinemia type 3	251316	Unclassified overlapping connective tissue disease
100048	Tubular duplication of the esophagus	882	Tyrosinemia type I	1264	Uncombable hair - retinal pigmentary dystrophy - dental anomalies - brachydactyly
73224	Tubular renal disease - cardiomyopathy	28378	Tyrosinemia type II	1410	Uncombable hair syndrome
319325	Tubulocystic carcinoma	69723	Tyrosinemia type III	442835	Undetermined early-onset epileptic encephalopathy
91500	Tubulointerstitial nephritis and uveitis syndrome	75840	UCMD		
2997	Tucker syndrome	90002	UCTD		
→2036	Tuffli-Laxova syndrome	609	Udd myopathy		
1063	Tufted angioma	79238	UDP-galactose-4-epimerase deficiency		
92050	Tufting enteropathy	178315	UES		
3392	Tularemia	205	UGT deficiency		
640	Tulip-bulb digger's palsy	79234	UGT deficiency type 1		
32960	Tumor necrosis factor receptor 1 associated periodic syndrome				
289539	Tumor susceptibility linked to germline BAP1 mutations				
352540	Tumor-induced osteomalacia				
879	Tungiasis				
3225	Tungland-Bellman 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
442835	Undetermined EOEE	96179	UPD(2)mat	79457	Urticaria pigmentosa
418951	Undifferentiated carcinoma of esophagus	96180	UPD(4)mat	886	USH
424970	Undifferentiated carcinoma of liver and IBT	96190	UPD(5)pat	231169	USH1
424970	Undifferentiated carcinoma of liver and intrahepatic biliary tract	96181	UPD(6)mat	231178	USH2
424970	Undifferentiated carcinoma of liver and intrahepatic biliary tract	96191	UPD(6)pat	231183	USH3
424080	Undifferentiated carcinoma of pancreas with osteoclast-like giant cells	96182	UPD(7)mat	886	Usher syndrome
424080	Undifferentiated carcinoma of pancreas with osteoclast-like giant cells	96192	UPD(7)pat	231169	Usher syndrome type 1
423786	Undifferentiated carcinoma of stomach	96183	UPD(9)mat	231178	Usher syndrome type 2
213721	Undifferentiated carcinoma of the corpus uteri	231147	UPD(11)mat	231183	Usher syndrome type 3
90002	Undifferentiated connective tissue syndrome	96193	UPD(11)pat	2032	Usual interstitial pneumonia
178315	Undifferentiated embryonal sarcoma of the liver	97678	UPD(13)mat	180145	Uterine cervical aplasia and agenesis
418951	Undifferentiated esophageal carcinoma	99324	UPD(13)pat	180139	Uterine hypoplasia
423786	Undifferentiated gastric carcinoma	96184	UPD(14)mat	439167	Uteroplacental vascular insufficiency
86830	Undifferentiated myeloproliferative disease	96334	UPD(14)pat	180118	Uterus arcuatus
2023	Undifferentiated pleomorphic sarcoma	98754	UPD(15)mat	180118	Uterus cordiformis
178315	Undifferentiated sarcoma of the liver	98795	UPD(15)pat	178338	UV-sensitive syndrome
319658	Unexplained intellectual disability	96185	UPD(16)mat	1473	Uveal coloboma-cleft lip and palate-intellectual disability
83468	Unicameral bone cyst	96186	UPD(20)mat	39044	Uveal melanoma
180079	Unicornuate uterus with rudimentary horn	96194	UPD(20)pat	3437	Uveomenigitic syndrome
180074	Unicornuate uterus without rudimentary horn	96187	UPD(21)mat	99771	Uvular cleft
93176	Unilateral congenital megacalycosis	96195	UPD(21)pat	370109	v-AT
268947	Unilateral focal polymicrogyria	96188	UPD(22)mat	887	VACTERL association
101071	Unilateral hemispheric polymicrogyria	261519	UPD(X)mat	3412	VACTERL with hydrocephalus
97363	Unilateral MCDK	261524	UPD(X)pat	887	VACTERL/VATER association
99802	Unilateral megalencephaly	3408	Upington disease	25980	Vacuolar myopathy
97363	Unilateral multicystic dysplastic kidney	2489	Upper limb defect - eye and ear abnormalities	2478	Vacuolating megalencephalic leukoencephalopathy with subcortical cysts
97363	Unilateral multicystic renal dysplasia	295049	Upper limb hypertrophy	65681	Vaginal atresia
268943	Unilateral polymicrogyria	2497	Upper limb mesomelic dysplasia	180247	Vaginal carcinoma
295148	Unilateral PPD2	268740	Upper thoracic spina bifida aperta	206489	Vaginal germ cell cancer
295012	Unilateral syndactyly of digits 2-5	268770	Upper thoracic spina bifida cystica	206489	Vaginal germ cell malignant tumor
1464	Univentricular heart	2023	UPS	180247	Vaginal malignant epithelial tumor
99069	Univentricular heart with single atrio-ventricular valve	93583	Upshaw-Schulman syndrome	158048	VAHS
79146	Universal melanosis	488	Urachal cyst	88639	Valine metabolic defect
620	Universal mesentery	431347	Urachal diverticulum	228123	Valley fever
84096	Unknown leukodystrophy	431344	Urachal sinus	99054	Valvular pulmonary stenosis
99104	Unroofed coronary sinus	530	Urbach-Wiethe disease	1548	Van Benthem-Driessen-Hanveld syndrome
99139	Unstable hemoglobin disease	221145	Urban-Rifkin-Davis syndrome	2806	Van Bogaert disease
308	Unverricht-Lundborg disease	3409	Urban-Rogers-Meyer syndrome	2806	Van Bogaert encephalitis
251009	UPD(1)mat	1839	Urban-Schosser-Spohn syndrome	3416	Van Buchem disease
251004	UPD(1)pat	94059	Uremic pruritus	1122	Van den Berghe-Dequecker syndrome
		105	Urethral atresia	3417	Van den Bosch syndrome
		35120	Uridine 5'-monophosphate hydrolase deficiency	2460	Van den Ende-Gupta syndrome
		79238	Uridine diphosphate galactose-4-epimerase deficiency	216796	Van der Hoeve syndrome
		30	Uridine monophosphate synthetase deficiency	2478	Van der Knaap syndrome
		210128	Urocanic aciduria	888	Van der Woude syndrome
		2704	Urofacial syndrome	314679	Van Maldergem syndrome
		83628	Urorectal septum malformation sequence	3419	Van Regemorter-Pierquin-Vamos syndrome
		98606	Urrets-Zavalia syndrome	73	Vanishing bone disease

→ 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
983	Vanishing testes syndrome	443988	Ventriculomegaly-cystic kidney disease	308442	Vitamin B12-responsive methylmalonic aciduria, type cblDv2
983	Vanishing testis syndrome	2899	Verloes-Bourguignon syndrome	27	Vitamin B12-unresponsive methylmalonic acidemia
729	Vaquez disease	2496	Verloes-David syndrome	79312	Vitamin B12-unresponsive methylmalonic acidemia type mut-
2754	Váradi syndrome	50817	Verloes-Deprez syndrome	289916	Vitamin B12-unresponsive methylmalonic acidemia type mut0
2754	Váradi-Papp syndrome	2983	Verloes-Gillerot-Fryns syndrome	27	Vitamin B12-unresponsive methylmalonic aciduria
314652	Variant ABeta2M amyloidosis	2551	Verloes-Van Maldergem-de Marneffe syndrome	79312	Vitamin B12-unresponsive methylmalonic aciduria type mut-
79253	Variant phenylketonuria	3429	Verloo Van Horick-Brubakk syndrome	289916	Vitamin B12-unresponsive methylmalonic aciduria type mut0
79253	Variant PKU	70476	Vernal keratoconjunctivitis	27	Vitamin B12-unresponsive methylmalonic aciduria
79473	Variegate porphyria	97282	Verner-Morrison syndrome	79312	Vitamin B12-unresponsive methylmalonic aciduria type mut-
404553	Vasculitis due to ADA2 deficiency	79467	Verrucous nevus	289916	Vitamin B12-unresponsive methylmalonic aciduria type mut0
353356	Vasoproliferative tumor of ocular fundus	26793	Very long chain acyl-CoA dehydrogenase deficiency	289157	Vitamin D dependent rickets type I
353356	Vasoproliferative tumor of retina	431347	Vesicourachal diverticulum	289157	Vitamin D-dependency type I
→26148 3	Vasquez-Hurst-Sotos syndrome	252175	Vestibular schwannoma	93160	Vitamin D-dependent rickets type II
85128	Västerbotten dystrophy	892	VHL	93160	Vitamin D-resistant rickets type II
887	VATER association	1493	Vici syndrome	1914	Vitamin K antagonists embryofetopathy
52047	Vater-like syndrome with pulmonary hypertension, abnormal ears and growth deficiency	3433	Viljoen-Kallis-Voges syndrome	413674	Vitamin K antagonists toxicity or dose selection
228379	VATS	3434	Viljoen-Smart syndrome	1243	Vitelliform macular dystrophy type 2
898	VCAN-related vitreoretinopathy	97282	VIP-secreting tumor	179	Vitiliginous choroiditis
289157	VDDI	97282	VIPoma	247871	Vitiligo-associated autoimmune disease
93160	VDDR II	206991	Viral myositis	898	Vitreoretinal degeneration, Wagner type
289157	VDDR-I	180176	Virginal breast hypertrophy	26793	VLCAD deficiency
2460	VDEGS	99916	Virilizing ovarian tumor	26793	VLCADD
93160	VDRR II	158048	Virus-associated hemophagocytis syndrome	386	VMC
1053	Vein of Galen aneurysm	228379	Virus-associated trichodysplasia spinulosa	443988	VMCKD
1053	Vein of Galen arteriovenous malformations	280068	Visceral calciphylaxis	2451	VMCM
3424	Velo-facial-skeletal syndrome	1876	Visceral myopathy - familial external ophthalmoplegia	83454	VMGLOM
567	Velocardiofacial syndrome	73246	Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay	79124	VODI syndrome
29207	Venereal arthritis	353344	Visible and exudative idiopathic juxtafoveolar retinal telangiectasis	3437	Vogt-Koyanagi-Harada disease
319234	Venezuelan hemorrhagic fever	420556	Visual snow syndrome	494	Vohwinkel syndrome
357131	Venous cervical rib syndrome	3006	Vitamin B6-dependent seizures	79395	Vohwinkel syndrome - ichthyosis
357131	Venous costoclavicular syndrome	28	Vitamin B12-responsive methylmalonic acidemia	2427	Volcke-Soekarman syndrome
357131	Venous hyperabduction syndrome	79310	Vitamin B12-responsive methylmalonic acidemia type cbIA	35737	Volubilis syndrome
83454	Venous malformations with glomus cells	79311	Vitamin B12-responsive methylmalonic acidemia type cbIB	83600	Von Economo encephalitis
357131	Venous scalenus anticus syndrome	308442	Vitamin B12-responsive methylmalonic acidemia, type cblDv2	364	Von Gierke disease
357131	Venous thoracic outlet compression syndrome	28	Vitamin B12-responsive methylmalonic aciduria	98941	Von Hippel anomaly
357131	Venous thoracic outlet syndrome	79310	Vitamin B12-responsive methylmalonic aciduria type cbIA	892	Von Hippel-Lindau disease
357131	Venous TOS	79311	Vitamin B12-responsive methylmalonic aciduria type cbIB	892	Von Hippel-Lindau syndrome
3201	Ventricular extrasystoles with syncopal episodes - perodactyly - Robin sequence	308442	Vitamin B12-responsive methylmalonic acidemia, type cblDv2	238557	Von Hippel-Lindau-dependent polycythemia
216694	Ventricular inversion	28	Vitamin B12-responsive methylmalonic aciduria	386	Von Meyenburg complexes disease
99094	Ventricular septal defect with aortic insufficiency	79310	Vitamin B12-responsive methylmalonic aciduria type cbIA	636	Von Recklinghausen disease
216694	Ventriculoarterial and atrioventricular discordance	79311	Vitamin B12-responsive methylmalonic aciduria, type cbIB	363700	Von Recklinghausen disease due to NF1 mutation or intragenic deletion
860	Ventriculoarterial discordance with atrioventricular concordance			3439	Von Voss-Cherstvoy syndrome
				903	Von Willebrand disease
				166078	Von Willebrand disease type 1

→ 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
166081	Von Willebrand disease type 2	96061	Warkany syndrome	1979	Werner-like syndrome due to combined growth factor deficiency
166084	Von Willebrand disease type 2A	90033	Warm AIHA	3451	West syndrome
166087	Von Willebrand disease type 2B	1541	Warman-Mulliken-Hayward syndrome	83476	West-Nile encephalitis
166090	Von Willebrand disease type 2M	280558	Warsaw breakage syndrome	83476	West-Nile fever
166093	Von Willebrand disease type 2N	51636	Warts-hypogammaglobulinemia-infections-myelokathesis syndrome	2435	Westerhof-Beemer-Cormane syndrome
240921	Voriconazole toxicity	51636	Warts-infections-leukopenia-myelokathesis syndrome	83593	Western equine encephalitis
369852	VPS45 deficiency	69745	Warty dyskeratoma	83593	Western equine encephalomyelitis
353356	VPTR	906	WAS	681	Westphall disease
99094	VSD with aortic insufficiency	1046	Water-West syndrome	952	Weyers acrodental dysostosis
357131	VTOS	100067	Waterhouse-Friderichsen syndrome	952	Weyers acrofacial dysostosis
137583	Vulvar intraepithelial neoplasia	97282	Watery diarrhea - hypokalemia - achlorhydria	→2750	Whelan syndrome
137583	Vulvar intraepithelial tumor	→636	Watson syndrome	51636	WHIM syndrome
83453	Vulvovaginal gingival syndrome	33577	WCD	3452	Whipple disease
206492	Vulvovaginal rhabdomyosarcoma	284395	WDFA	2053	Whistling face syndrome
53696	Vuopala disease	97282	WDHA syndrome	228290	White fibrous papulosis of the neck
888	VWS	99971	WDLS	2475	White forelock with malformations
2804	W syndrome	603	WDM	3207	White matter hypoplasia - corpus callosum agenesis - intellectual disability
2180	Waaler-Aarskog syndrome	3447	Weaver syndrome		370131
1106	Waardenburg anophthalmia syndrome	→3447	Weaver-like syndrome	171723	White sponge nevus
3440	Waardenburg syndrome	3448	Weaver-Williams syndrome	171723	White sponge nevus of Cannon
894	Waardenburg syndrome type 1	33577	Weber-Christian disease	1489	Whooping cough
895	Waardenburg syndrome type 2	33577	Weber-Christian panniculitis	2779	Whyte-Murphy syndrome
352740	Waardenburg syndrome type 2 with ocular albinism	1521	Webster-Deming syndrome	3454	Wieacker-Wolff syndrome
896	Waardenburg syndrome type 3	900	Wegener granulomatosis	116	Wiedemann-Beckwith syndrome
897	Waardenburg syndrome type 4	228254	Weidman juvenile elastoma	2156	Wiedemann-Oldigs-Oppermann syndrome
896	Waardenburg syndrome with limb anomalies	3449	Weill-Marchesani syndrome	3455	Wiedemann-Rautenstrauch syndrome
897	Waardenburg-Hirschsprung syndrome	3344	Weismann-Netter syndrome	319182	Wiedemann-Steiner syndrome
98960	Waardenburg-Jonker corneal dystrophy	3450	Weissenbacher- Zweymuller syndrome	85446	Wild type ABeta2-microglobulinic amyloidosis
897	Waardenburg-Shah syndrome	213736	Well-differentiated endocrine neoplasm of corpus uteri	85446	Wild type ABeta2M amyloidosis
280558	WABS	213736	Well-differentiated endocrine neoplasm of endometrium	330001	Wild type ATTR amyloidosis
247709	Wagenmann-Froboese syndrome	213736	Well-differentiated endocrine tumor of corpus uteri	330001	Wild type ATTR-related amyloidosis
898	Wagner disease	213736	Well-differentiated endocrine tumor of endometrium	3456	Wildervanck syndrome
898	Wagner syndrome	284395	Well-differentiated fetal adenocarcinoma of the lung	739	Willi-Prader syndrome
893	WAGR syndrome	99971	Well-differentiated liposarcoma	904	Williams syndrome
90033	wAHA	263331	Well-differentiated thymic neuroendocrine carcinoma	904	Williams-Beuren syndrome
357332	Wahab syndrome	146	Well-differentiated thyroid carcinoma	411501	Williams-Campbell syndrome
90033	wAIHA	1373	Wellesley-Carman-French syndrome	51636	WILM
2379	Waisman syndrome	901	Wells syndrome	654	Wilms tumor
33226	Waldenström macroglobulinemia	2815	Wells-Jankovic syndrome	893	Wilms tumor - aniridia - genitourinary anomalies - intellectual disability
90362	Waldmann disease	83330	Werdnig-Hoffmann disease	220	Wilms tumor and pseudohermaphroditism
1068	Walker-Dyson syndrome	652	Wermer syndrome	905	Wilson disease
899	Walker-Warburg syndrome	3332	Werner mesomelic syndrome	3459	Wilson-Turner syndrome
1453	Wallis-Zieff-Goldblatt syndrome	902	Werner syndrome	3460	Winchester syndrome
2078	Walt Disney dwarfism			169095	Winged helix deficiency
2510	WARBM			2901	Winged scapula
2510	Warburg micro syndrome				
3214	Warburg-Thomsen syndrome				
1052	Warburton-Anyane-Yeboa 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
94087	Winkelmann cytophagic panniculitis	2834	WSS	101078	X-linked Charcot-Marie-Tooth disease type 4
2515	Winship-Viljoen-Leary syndrome	3466	WT limb-blood syndrome	99014	X-linked Charcot-Marie-Tooth disease type 5
906	Wiskott-Aldrich syndrome	3459	WTS	352675	X-linked Charcot-Marie-Tooth disease type 6
829	Wissler-Fanconi syndrome	3411	Wunderlich syndrome	35173	X-linked chondrodysplasia punctata type 2
2228	Witkop syndrome	899	WWS	324601	X-linked cleft palate and ankyloglossia
101068	Witschel dystrophy	53719	Wyburn-Mason syndrome	431140	X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome
85291	Wittwer syndrome	96201	X small rings		X-linked complicated corpus callosum dysgenesis
3237	WL syndrome	43	X-ALD	1497	X-linked complicated spastic paraplegia type 1
247768	WNT4 deficiency	300373	X-LAG (X-linked acrogigantism)	306617	X-linked cone dysfunction syndrome with myopia
1667	Wolcott-Rallison syndrome	448348	X-LAG (X-linked acrogigantism) due to a point mutation	90001	X-linked congenital adrenal hypoplasia
280	Wolf-Hirschhorn syndrome	448372	X-LAG (X-linked acrogigantism) due to dup(X)q(26)	95702	X-linked congenital dyserythropoietic anemia with thrombocytopenia
3080	Wolff-Zimmermann syndrome	2182	X-linked aqueductal stenosis	67044	X-linked congenital generalized hypertrichosis
3463	Wolfram syndrome	448348	X-linked acrogigantism due to a point mutation	1661	X-linked corneal dermoid
411590	Wolfram-like syndrome	448372	X-linked acrogigantism due to Xq26 microduplication	52503	X-linked creatine transporter deficiency
75233	Wolman disease	43	X-linked adrenoleukodystrophy	85453	X-linked cutaneous amyloidosis
3464	Woodhouse-Sakati syndrome	47	X-linked agammaglobulinemia	198	X-linked deafness - intellectual disability syndrome
2571	Woods-Black-Norbury syndrome	43	X-linked ALD	383	X-linked deafness type 2
137658	Woods-Crouchman-Huson syndrome	88917	X-linked Alport syndrome	139557	X-linked dHMN type 3
170	Woolly hair	85278	X-linked Angelman-like syndrome	1018	X-linked diffuse leiomyomatosis - Alport syndrome
1409	Woolly hair - hypotrichosis - everted lower lip - outstanding ears	181	X-linked anhidrotic ectodermal dysplasia	1145	X-linked distal arthrogryposis multiplex congenita
79414	Woolly hair nevus	85297	X-linked ataxia-deafness syndrome	139557	X-linked distal hereditary motor neuropathy type 3
420686	Woolly hair-palmoplantar hyperkeratosis syndrome	85292	X-linked ataxia-dementia syndrome	139557	X-linked distal spinal muscular atrophy type 3
65282	Woolly hair-palmoplantar hyperkeratosis-dilated cardiomyopathy syndrome	139583	X-linked auditory neuropathy with peripheral sensory neuropathy type 1	163966	X-linked dominant chondrodysplasia - hydrocephaly - microphthalmia
420686	Woolly hair-palmoplantar keratoderma syndrome	1131	X-linked branchial arch syndrome	35173	X-linked dominant chondrodysplasia punctata
65282	Woolly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome	481	X-linked BSMA	163966	X-linked dominant chondrodysplasia, Chassaing-Lacombe type
170	Wooly hair	481	X-linked bulbospinal amyotrophy	443197	X-linked dominant erythropoietic protoporphyria
1409	Wooly hair - hypotrichosis - everted lower lip - outstanding ears	481	X-linked bulbospinal muscular atrophy	93951	X-linked dominant intellectual disability - epilepsy
65282	Wooly hair - palmoplantar keratoderma - dilated cardiomyopathy	391327	X-linked calvarial hyperostosis	443197	X-linked dominant protoporphyria
79414	Wooly hair nevus	111	X-linked cardioskeletal myopathy and neutropenia	139557	X-linked dSMA type 3
65282	Wooly hair-palmoplantar hyperkeratosis-dilated cardiomyopathy syndrome	329235	X-linked central congenital hypothyroidism with late-onset macroorchidism		
3465	Worster-Drought syndrome	329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement		
2790	Worth syndrome	596	X-linked centronuclear myopathy		
178475	Wound botulism	163961	X-linked cerebral - cerebellar - coloboma syndrome		
165955	Wound myiasis	139396	X-linked cerebral adrenoleukodystrophy		
2834	Wrinkled skin syndrome	101075	X-linked Charcot-Marie-Tooth disease type 1		
2834	Wrinkly skin syndrome	101076	X-linked Charcot-Marie-Tooth disease type 2		
1667	WRS	101077	X-linked Charcot-Marie-Tooth disease type 3		
902	WS				
894	WS1				
895	WS2				
896	WS3				
897	WS4				
163746	WS4 plus				

→ 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
363727	X-linked dyserythropoetic anemia with abnormal platelets and neutropenia	85280	X-linked intellectual disability - cubitus valgus - dysmorphism	67045	X-linked intellectual disability with isolated growth hormone deficiency
373	X-linked dysplasia gigantism syndrome	2958	X-linked intellectual disability - dysmorphism - cerebral atrophy	776	X-linked intellectual disability with marfanoid habitus
53351	X-linked dystonia-parkinsonism	94083	X-linked intellectual disability - dystonia - dysarthria	85273	X-linked intellectual disability, Abidi type
75497	X-linked Ehlers-Danlos syndrome	85319	X-linked intellectual disability - epilepsy - progressive joint contractures - dysmorphism	85274	X-linked intellectual disability, Ahmad type
98863	X-linked Emery-Dreifuss muscular dystrophy	85282	X-linked intellectual disability - epileptic seizures - hypogenitalism - microcephaly - obesity	85276	X-linked intellectual disability, Armfield type
293621	X-linked endothelial corneal dystrophy	85317	X-linked intellectual disability - hypogammaglobulinemia - progressive neurological deterioration	1193	X-linked intellectual disability, Atkin type
85294	X-linked epilepsy - learning disabilities - behavior disorders	85331	X-linked intellectual disability - hypogonadism - ichthyosis - obesity - short stature	3056	X-linked intellectual disability, Brooks type
443197	X-linked erythropoietic protoporphyrinia	59	X-linked intellectual disability - hypotonia	85293	X-linked intellectual disability, Cabezas type
→994	X-linked fetal akinesia syndrome	85329	X-linked intellectual disability - hypotonia - facial dysmorphism - aggressive behavior	85277	X-linked intellectual disability, Cantagrel type
139583	X-linked hereditary sensory and autonomic neuropathy with deafness	→1762	X-linked intellectual disability - hypotonia - recurrent Infections	163971	X-linked intellectual disability, Cilliers type
139583	X-linked HSAN with deafness	85320	X-linked intellectual disability - macrocephaly - macroorchidism	→93950	X-linked intellectual disability, Fichera type
2182	X-linked HSAS	251383	X-linked intellectual disability - microcephaly - cortical malformation - thin habitus	93947	X-linked intellectual disability, Golabi-Ito-Hall type
2182	X-linked hydrocephalus	163937	X-linked intellectual disability - microcephaly - pontocerebellar hypoplasia	3059	X-linked intellectual disability, Gu type
2182	X-linked hydrocephalus with stenosis of aqueduct of Sylvius	163971	X-linked intellectual disability - microcephaly - testicular failure	93952	X-linked intellectual disability, Hedera type
101088	X-linked hyper-IgM syndrome	→3057	X-linked intellectual disability - monoamine oxidase A metabolism anomaly	163961	X-linked intellectual disability, Kroes type
181	X-linked hypohidrotic ectodermal dysplasia	163956	X-linked intellectual disability - nail dystrophy - seizures	→1762	X-linked intellectual disability, Lubs type
89936	X-linked hypophosphatemia	2898	X-linked intellectual disability - plagioccephaly	775	X-linked intellectual disability, Martinez type
89936	X-linked hypophosphatemic rickets	85318	X-linked intellectual disability - precocious puberty - obesity	85283	X-linked intellectual disability, Miles-Carpenter type
461	X-linked ichthyosis	3077	X-linked intellectual disability - psychosis - macroorchidism	163937	X-linked intellectual disability, Najm type
231692	X-linked IGHD	3052	X-linked intellectual disability - seizures - psoriasis	163956	X-linked intellectual disability, Nascimento type
317476	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia	3055	X-linked intellectual disability - short stature - obesity	85322	X-linked intellectual disability, Pai type
2571	X-linked immunoneurologic disorder	163982	X-linked intellectual disability - spastic quadripareisis	93945	X-linked intellectual disability, Porteous type
16	X-linked incomplete achromatopsia	364028	X-linked intellectual disability due to GRIA3 anomalies	→776	X-linked intellectual disability, Raymond type
1145	X-linked infantile spinal muscular atrophy	3242	X-linked intellectual disability due to PQBP1 mutations	3061	X-linked intellectual disability, Raynaud type
85327	X-linked intellectual disability - acromegaly - hyperactivity			3242	X-linked intellectual disability, Renpenning type
85338	X-linked intellectual disability - ataxia - apraxia			85285	X-linked intellectual disability, Schimke type
324410	X-linked intellectual disability - cardiomegaly - congestive heart failure			3062	X-linked intellectual disability, Schutz type
137831	X-linked intellectual disability - cerebellar hypoplasia			85323	X-linked intellectual disability, Seemanova type
85330	X-linked intellectual disability - corpus callosum agenesis - spastic quadripareisis				
85278	X-linked intellectual disability - craniofacial dysmorphism - epilepsy - ophthalmoplegia - cerebellar atrophy				
163979	X-linked intellectual disability - craniofacioskeletal 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
85286	X-linked intellectual disability, Shashi type	90625	X-linked isolated sensorineural hearing loss type DFN	85334	X-linked neurodegenerative syndrome, Bertini type
85324	X-linked intellectual disability, Shrimpton type	792	X-linked juvenile retinoschisis	85336	X-linked neurodegenerative syndrome, Hamel type
85287	X-linked intellectual disability, Siderius type	79447	X-linked lethal multiple pterygium syndrome	314978	X-linked non progressive cerebellar ataxia
3063	X-linked intellectual disability, Snyder type	452	X-linked lissencephaly - agenesis of the corpus callosum - genital anomalies	777	X-linked non-specific intellectual disability
85278	X-linked intellectual disability, South African type	2148	X-linked lissencephaly type 1	777	X-linked non-syndromic intellectual disability
85325	X-linked intellectual disability, Stevenson type	452	X-linked lissencephaly with abnormal genitalia	90625	X-linked non-syndromic neurosensory deafness type DFN
85288	X-linked intellectual disability, Stocco Dos Santos type	452	X-linked lissencephaly with ambiguous genitalia	90625	X-linked non-syndromic neurosensory hearing loss type DFN
85326	X-linked intellectual disability, Stoll type	2442	X-linked lymphoproliferative disease	90625	X-linked non-syndromic sensorineural deafness type DFN
93950	X-linked intellectual disability, Sutherland-Haan type	1131	X-linked mandibulofacial dysostosis	90625	X-linked non-syndromic sensorineural hearing loss type DFN
85328	X-linked intellectual disability, Turner type	1131	X-linked mandibulofacial dysostosis with limb anomalies	293707	X-linked Ohdo syndrome
163976	X-linked intellectual disability, Van Esch type	59306	X-linked McLeod syndrome	306597	X-linked Opitz BBB/G syndrome
85289	X-linked intellectual disability, Vitale type	319605	X-linked mendelian susceptibility to mycobacterial diseases	306597	X-linked Opitz G/BBB syndrome
85290	X-linked intellectual disability, Wilson type	319623	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency	306597	X-linked Opitz syndrome
3064	X-linked intellectual disability, Wittner type	319612	X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency	391330	X-linked osteoporosis with fractures
85291	X-linked intellectual disability, Wittwer type	319612	X-linked mendelian susceptibility to mycobacterial diseases due to NEMO deficiency	363654	X-linked parkinsonism-spasticity syndrome
85337	X-linked intellectual disability, Zorick type	776	X-linked mental retardation with marfanoid habitus	1175	X-linked progressive cerebellar ataxia
85295	X-linked intellectual disability-choreoathetosis-abnormal behavior syndrome	435938	X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	1652	X-linked recessive hypercalciuric hypophosphatemic rickets
1568	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	383	X-linked mixed conductive and neurosensory deafness	83648	X-linked recessive intellectual disability - macrocephaly - ciliary dysfunction
3459	X-linked intellectual disability-gynecomastia-obesity syndrome	383	X-linked mixed conductive and neurosensory hearing loss	1652	X-linked recessive nephrolithiasis
423479	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	383	X-linked mixed conductive and sensorineural deafness	54	X-linked recessive ocular albinism
85332	X-linked intellectual disability-retinitis pigmentosa syndrome	383	X-linked mixed conductive and sensorineural hearing loss	85453	X-linked reticulate pigmentary disorder with systemic manifestations
→702	X-linked intellectual disability-spastic paraplegia with iron deposits syndrome	383	X-linked mixed deafness with perilymphatic gusher	1852	X-linked retinal dysplasia
231692	X-linked isolated growth hormone deficiency	319605	X-linked MSMD	792	X-linked retinoschisis
90625	X-linked isolated neurosensory deafness type DFN	319623	X-linked MSMD due to CYBB deficiency	431272	X-linked scapuloperoneal muscular dystrophy
90625	X-linked isolated neurosensory hearing loss type DFN	319612	X-linked MSMD due to IKBKG deficiency	431272	X-linked scapuloperoneal syndrome
90625	X-linked isolated sensorineural deafness type DFN	319612	X-linked MSMD due to NEMO deficiency	86788	X-linked severe congenital neutropenia
		25980	X-linked myopathy with excessive autophagy	75563	X-linked sideroblastic anemia
		178461	X-linked myopathy with postural muscle atrophy	2802	X-linked sideroblastic anemia and ataxia
				2802	X-linked sideroblastic anemia with ataxia
				99015	X-linked spastic paraplegia type 2
				100997	X-linked spastic paraplegia type 16
				171607	X-linked spastic paraplegia type 34
				481	X-linked spinal and bulbar muscular atrophy
				1145	X-linked spinal muscular atrophy type 2

→ 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
404521	X-linked spinal muscular atrophy with respiratory distress	412069	Xia-Gibbs syndrome	370930	XYLT1-CDG
85297	X-linked spinocerebellar ataxia type 3	3469	XK aprosencephaly	2616	Yakut short stature syndrome
85292	X-linked spinocerebellar ataxia type 4	452	XLAG (X-linked lissencephaly with abnormal genitalia) syndrome	99829	Yellow fever
431272	X-linked SPMD	596	XLCNM	99829	Yellow Jack
93349	X-linked spondyloepimetaphyseal dysplasia	443197	XLDPP	662	Yellow nail syndrome
168544	X-linked spondylometaphyseal dysplasia	264580	XLG	79434	Yellow oculocutaneous albinism
383	X-linked stapes gusher syndrome	89936	XLH	3214	Yemenite deaf-blind hypopigmentation syndrome
852	X-linked thrombocytopenia with normal platelets	461	XLI	707	Yersiniosis
3467	Xanthic urolithiasis	776	XLMR with marfanoid habitus	99829	YF
93602	Xanthine dehydrogenase and xanthine aldehyde oxidase dual deficiency	596	XLMTM	662	YNS
93601	Xanthine dehydrogenase deficiency	54	XLOA	876	Yolk sac tumor
93601	Xanthine oxidase deficiency	306597	XLOS	252006	Yolk sac tumor of central nervous system
93601	Xanthine oxidoreductase deficiency	2442	XLP	252006	Yolk sac tumor of CNS
3467	Xanthine stone disease	443197	XLP	2828	YOPD
93601	Xanthinuria type I	85453	XLPDR	2255	Yorifuji-Okuno syndrome
93602	Xanthinuria type II	443197	XLPP	3240	Yoshimura-Takeshita syndrome
158003	Xanthoma disseminatum	792	XLRS	314485	Young adult-onset dHMN
2882	Xanthomatosis with sisterolemia	75563	XLSA	314485	Young adult-onset distal hereditary motor neuropathy
79433	Xanthous oculocutaneous albinism	2802	XLSA-A	3471	Young syndrome
79155	Xanthurenic aciduria	231393	XLTT	3055	Young-Hughes syndrome
67044	XDAT	25980	XMEA	2828	Young-onset Parkinson disease
93602	XDH and AOX dual deficiency	317476	XMEN	3472	Yunis-Varon syndrome
93601	XDH deficiency	93601	XO deficiency	319213	Zambian hemorrhagic fever
53351	XDP	93601	XOR deficiency	98912	ZASP-related myofibrillar myopathy
293621	XECD	910	XP	97240	Zebra body myopathy
910	Xeroderma pigmentosum	220295	XP/CS complex	217017	Zechi-Ceide syndrome
276249	Xeroderma pigmentosum complementation group A	261476	Xp21 microdeletion syndrome	912	Zellweger syndrome
276252	Xeroderma pigmentosum complementation group B	284180	Xp22.13p22.2 duplication syndrome	369942	Zellweger-like contiguous gene deletion syndrome
276255	Xeroderma pigmentosum complementation group C	1643	Xp22.3 microdeletion syndrome	50812	Zellweger-like syndrome without peroxisomal anomalies
276258	Xeroderma pigmentosum complementation group D	276249	XPA	911	Zeta-associated-protein 70 deficiency
276261	Xeroderma pigmentosum complementation group E	276252	XPB	448237	Zika virus disease
276264	Xeroderma pigmentosum complementation group F	276255	XPC	448237	Zika virus infection
276267	Xeroderma pigmentosum complementation group G	276258	XPD	3301	Zimmer phocomelia
90342	Xeroderma pigmentosum variant	363654	XPDS	3473	Zimmermann-Laband syndrome
→910	Xeroderma pigmentosum with neurologic manifestation	276261	XPE	439196	Zinc-responsive necrolytic acral erythema
220295	Xeroderma pigmentosum-Cockayne syndrome complex	276264	XPF	1775	Zinsser-Engman-Cole syndrome
75496	XGPT deficiency	276267	XPG	3253	Zlotogora-Ogur syndrome
181	XHED	90342	XPV	3253	Zlotogora-Zilberman-Tenenbaum syndrome
101088	XHIGM	314389	Xq12-q13.3 duplication syndrome	913	Zollinger-Ellison syndrome
		1018	Xq22.3 microdeletion syndrome	2835	Zori-Stalker-Williams syndrome
		261483	Xq27.3-q28 microduplication syndrome	912	ZS
		261483	Xq27.3q28 duplication syndrome	3474	Zunich-Kaye syndrome
		243	XX female gonadal dysgenesis	295187	Zygodactyl type 1
		2855	XX gonadal dysgenesis - deafness	295189	Zygodactyl type 2
		393	XX, male syndrome	295191	Zygodactyl type 3
		243	XX-GD	295193	Zygodactyl type 4
		3375	XXX syndrome		
		168558	XY sex reversal - adrenal failure		
		2843	Xylitol dehydrogenase deficiency		
		75496	Xylosylprotein 4-beta-galactosyltransferase deficiency		

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

ORPHA number	Disease name
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
138	CHARGE syndrome	1474	Colobomatous - microphthalmia - heart disease - hearing loss
138	CHARGE syndrome	1474	Hittner-Hirsch-Kreh syndrome
175	Cartilage-hair hypoplasia	1838	Metaphyseal dysplasia without hypotrichosis
175	Cartilage-hair hypoplasia	1838	Cartilage-hair hypoplasia-like skeletal dysplasia without hypotrichosis
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
193	Cohen syndrome	2829	Partington-Anderson syndrome
193	Cohen syndrome	3271	Radio-ulnar synostosis - retinal pigment abnormalities
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
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	98788	Pitt-Rogers-Danks syndrome
280	Wolf-Hirschhorn syndrome	98788	Intellectual disability - dysmorphism - intrauterine growth retardation
288	Hereditary elliptocytosis	98867	Hereditary pyropoikilocytosis
288	Hereditary elliptocytosis	98864	Common hereditary elliptocytosis
288	Hereditary elliptocytosis	98865	Homozygous hereditary elliptocytosis
300	Bifunctional enzyme deficiency	2981	Pseudo-Zellweger syndrome
300	Bifunctional enzyme deficiency	2981	Thiolase deficiency
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
646	Niemann-Pick disease type C	79289	Niemann-Pick disease type D

→ Use these ORPHA number		instead of the deprecated entities	
ORPHA number	Disease to be used	Deprecated ORPHA number	Deprecated entities
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
794	Saethre-Chotzen syndrome	1219	Aurocephalosyndactyly
794	Saethre-Chotzen syndrome	1219	Auralcephalosyndactyly
794	Saethre-Chotzen syndrome	1219	Kurczynski-Casperson 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
897	Waardenburg-Shah syndrome	918	Albinism-black lock-cell migration disorder of the neurocytes of the gut-sensorineural deafness syndrome
910	Xeroderma pigmentosum	1569	De Sanctis-Cacchione syndrome
910	Xeroderma pigmentosum	1569	Xeroderma pigmentosum with neurologic manifestation
912	Zellweger syndrome	1271	Bowen syndrome
955	Acroosteolysis dominant type	2853	Serpentine fibula - polycystic kidneys
955	Acroosteolysis dominant type	2853	Exner 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
969	Acromicric dysplasia	2569	Moore-Federman syndrome Dwarfism - stiff joint - ocular abnormalities	1263	Boomerang dysplasia	156723	Short ribs - craniosynostosis - polysyndactyly
969	Acromicric dysplasia	2569	X-linked fetal akinesia syndrome	1359	Carney complex	623	NAME syndrome
994	Fetal aknesia deformation sequence	995	Holmes-Benacerraf syndrome	1359	Carney complex	623	Nevi - atrial myxoma - myxoid neurofibromata - ephelides
994	Fetal aknesia deformation sequence	995	CHAND syndrome	1394	Cerebro-facio-thoracic dysplasia	228407	Craniofacial dysmorphism-skeletal anomalies-intellectual disability syndrome
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	1401	Baughman syndrome	1394	Cerebro-facio-thoracic dysplasia	228407	TMCO1 defect syndrome
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	1401	CHANDS	1466	COFS syndrome	1317	CAMFAK syndrome
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	1401	Curly hair - ankyloblepharon - nail dysplasia syndrome	1466	COFS syndrome	1317	CAMAK syndrome
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	1401	Rapp-Hodgkin syndrome	1466	COFS syndrome	1317	Cataract - microcephaly - arthrogryposis - kyphosis
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	3022	Anhidrotic ectodermic dysplasia-cleft lip/palate syndrome	1466	COFS syndrome	1317	Cataract - microcephaly - failure to thrive - kyphoscoliosis
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	3022	Ectodermal dysplasia syndrome, Rapp-Hodgkin type	1487	Cooks syndrome	2355	Kumar-Levick syndrome
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	3022	Ectodermal dysplasia, Rapp-Hodgkin type	1487	Cooks syndrome	2355	Nail dysplasia - camptodactyly - brachydactyly type B
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	3022	RHS	1509	Coxopodopatellar syndrome	3112	Patella aplasia - coxa vara - tarsal synostosis
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate	99694	Alveolar synechia- ankyloblepharon-ectodermal dysplasia syndrome	1643	Xp22.3 microdeletion syndrome	431	Ichthyosis - male hypogonadism
1159	Progressive pseudorheumatoid arthropathy of childhood	2654	Syndesmodyplastic dwarfism	1658	Absence of fingerprints - congenital milia	1235	Ectodermal dysplasia - absent dermatoglyphs
1159	Progressive pseudorheumatoid arthropathy of childhood	2654	Laplane-Fontaine-Lagardere syndrome	1658	Absence of fingerprints - congenital milia	1235	Basan syndrome
1200	Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome	77302	Oculo-oto-facial dysplasia	1762	Trisomy Xq28	85281	X-linked intellectual disability, Lubs type
1215	Autosomal dominant optic atrophy plus syndrome	3349	Treft-Sanborn-Carey syndrome	1762	Trisomy Xq28	85281	Lubs-Arena syndrome
1215	Autosomal dominant optic atrophy plus syndrome	3349	Optic atrophy - ophthalmoplegia - ptosis - deafness - myopathy	1762	Trisomy Xq28	85281	X-linked intellectual disability - hypotonia - recurrent Infections
1215	Autosomal dominant optic atrophy plus syndrome	3212	Autosomal dominant optic atrophy and congenital deafness	1768	Familial caudal dysgenesis	1850	Renal dysplasia-megalocystis-sirenomelia syndrome
1215	Autosomal dominant optic atrophy plus syndrome	3212	Konigsmark-Knox-Hussell syndrome	1768	Familial caudal dysgenesis	1850	Selig-Benacerraf-Greene syndrome
1234	Bartsocas-Papas syndrome	79446	Multiple pterygium syndrome, Aslan type	1855	Spondyloenchondrodysplasia	50816	Spondylometaphyseal dysplasia with combined immunodeficiency
1263	Boomerang dysplasia	156723	Piepkorn dysplasia	1855	Spondyloenchondrodysplasia	50816	Roifman-Melamed syndrome
				1855	Spondyloenchondrodysplasia	50816	SPENCDI
				1855	Spondyloenchondrodysplasia	50816	Spondyloenchondrodysplasia with immune dysregulation
				1896	EEC syndrome	1888	Ectrodactyly - ectodermal dysplasia without clefting
				1896	EEC syndrome	1888	EEC syndrome without cleft lip/palate
				1896	EEC syndrome	1889	Ectrodactyly - cleft palate
				1896	EEC syndrome	1889	ECP syndrome
				1896	EEC syndrome	2389	Lewis-Pashayan syndrome
				1896	EEC syndrome	2389	Cleft lip/palate - ectrodactyly

→ 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
1900	Ehlers-Danlos syndrome, kyphoscoliotic type	2691	Nevo syndrome	2750	Orofaciodigital syndrome type 1	90649	Whelan syndrome
1900	Ehlers-Danlos syndrome, kyphoscoliotic type	2691	Cerebral gigantism, Nevo type	2796	Pachydermoperiostosis	964	Acromegaly-cutis verticis gyrata-corneal leukoma syndrome
2036	Scalp-ear-nipple syndrome	3391	Odonto-onycho-hypohidrotic dysplasia - midline scalp defects	2909	Rothmund-Thomson syndrome	3333	Connective tissue dysplasia, Spellacy type
2036	Scalp-ear-nipple syndrome	3391	Ectodermal dysplasia - adrenal cyst	2909	Rothmund-Thomson syndrome	3333	Spellacy-Gibbs-Watts syndrome
2036	Scalp-ear-nipple syndrome	3391	Tuffli-Laxova syndrome	2995	Baraits-Winter syndrome	94084	Pachygyria - epilepsy - intellectual disability - dysmorphism
2052	Fraser syndrome	2051	Fraser-like syndrome	2995	Baraits-Winter syndrome	94084	Cerebro-oculo-facial-lymphatic syndrome
2199	Epidermolytic palmoplantar keratoderma	496	Thost-Unna palmoplantar keratoderma	2995	Baraits-Winter syndrome	94084	Fryns-Aftimos syndrome
2199	Epidermolytic palmoplantar keratoderma	496	Non-epidermolytic palmoplantar keratoderma	3057	Monoamine oxidase A deficiency	3065	X-linked intellectual disability - monoamine oxidase A metabolism anomaly
2199	Epidermolytic palmoplantar keratoderma	89833	Palmoplantar keratoderma with tonotubular keratin	3157	Septo-optic dysplasia spectrum	1102	Anophthalmia - hypothalamo-pituitary insufficiency
2353	Schilbach-Rott syndrome	1251	Blepharofacioskeletal syndrome	3157	Septo-optic dysplasia spectrum	1102	14q22 microdeletion syndrome
2353	Schilbach-Rott syndrome	1251	Richieri Costa-Guion Almeida-Rodini syndrome	3157	Septo-optic dysplasia spectrum	1102	Al Frayh-Facharzt-Haque syndrome
2470	Matthew-Wood syndrome	91129	Anophthalmia - heart and pulmonary anomalies - intellectual disability	3157	Septo-optic dysplasia spectrum	1102	Monosomy 14q22
2510	Micro syndrome	2895	Pinsky-Di George-Harley syndrome	3157	Septo-optic dysplasia spectrum	1678	Dincsoy-Salih-Patel syndrome
2510	Micro syndrome	2895	Microphthalmia - mental deficiency	3157	Septo-optic dysplasia spectrum	1678	Facial dysmorphism - ambiguous genitalia - hypopituitarism - short limbs
2526	Microcephaly - lymphedema - chorioretinopathy	1432	Autosomal dominant chorioretinopathy - microcephaly	3157	Septo-optic dysplasia spectrum	2245	Hypopituitarism - postaxial polydactyly
2609	Isolated NADH-CoQ reductase deficiency	936	Succinic acidemia	3157	Septo-optic dysplasia spectrum	2245	Culler-Jones syndrome
2616	3M syndrome	2661	Dwarfism - tall vertebrae	3157	Septo-optic dysplasia spectrum	2243	Hypopituitarism - micropenis - cleft lip/palate
2637	Microcephalic osteodysplastic primordial dwarfism type II	46658	Primordial short stature - microdontia - opalescent and rootless teeth	3157	Septo-optic dysplasia spectrum	2244	Hypopituitarism - microphthalmia
2686	Cyclic neutropenia	2689	Intermittent neutropenia	3157	Septo-optic dysplasia spectrum	2244	Kaplowitz-Bodurtha syndrome
2697	Arthrogryposis - renal dysfunction - cholestasis	1981	Fanconi syndrome - ichthyosis - dysmorphism	3157	Septo-optic dysplasia spectrum	370006	Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome
2697	Arthrogryposis - renal dysfunction - cholestasis	1981	Deal-Barrat-Dillon syndrome	3157	Septo-optic dysplasia spectrum	93943	Corpus callosum dysgenesis - hypopituitarism
2697	Arthrogryposis - renal dysfunction - cholestasis	3438	Biliary tract malformation - renal failure	3202	Dehydrated hereditary stomatocytosis	100039	Familial pseudohyperkalemia type 1
2697	Arthrogryposis - renal dysfunction - cholestasis	3438	Cholestatic jaundice - renal tubular insufficiency	3253	Zlotogora-Ogur syndrome	90338	Margarita island ectodermal dysplasia
2697	Arthrogryposis - renal dysfunction - cholestasis	3438	Lutz-Richner-Landolt syndrome	3447	Weaver syndrome	3446	Weaver-like syndrome
2712	Oculoaciocardiodental syndrome	3013	Radiculomegaly of canine teeth- congenital cataract	3460	Torg-Winchester syndrome	2775	Autosomal recessive carpotarsal osteolysis
2712	Oculoaciocardiodental syndrome	3013	Marashi-Gorlin syndrome	3460	Torg-Winchester syndrome	2775	Hereditary multicentric osteolysis
2750	Orofaciodigital syndrome type 1	90649	Orofaciodigital syndrome type 7				
2750	Orofaciodigital syndrome type 1	90649	OFD7				
2750	Orofaciodigital syndrome type 1	90649	Oral-facial-digital syndrome type 7				

→ 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
3464	Woodhouse-Sakati syndrome	1011	Alopecia-hypogonadism-extrapyramidal disorder syndrome	52368	Mohr-Tranebjaerg syndrome	3213	Jensen syndrome
3464	Woodhouse-Sakati syndrome	1011	Devriendt-Legius-Fryns syndrome	52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	54238	Myotonic dystrophy type 3
3471	Young syndrome	1301	Bronchiectasis - oligospermia	56304	Atelosteogenesis type II	2640	Lethal short-limb dwarfism, McAlister-Crane type
33001	Lymphedema - distichiasis	1683	Distichiasis - congenital heart defects - peripheral vascular anomalies	56304	Atelosteogenesis type II	2640	McAlister-Crane syndrome
33001	Lymphedema - distichiasis	2419	Lymphedema - ptosis	60030	Loeys-Dietz syndrome	97295	Furlong syndrome
33364	Trichothiodystrophy	1245	BIDS syndrome	60030	Loeys-Dietz syndrome	97295	Marfanoid habitus - craniosynostosis syndrome
33364	Trichothiodystrophy	1245	Amish brittle hair syndrome	69061	Idiopathic steroid-sensitive nephrotic syndrome	97552	Steroid-sensitive nephrotic syndrome without renal biopsy
33364	Trichothiodystrophy	1245	Trichothiodystrophy type D	69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	2087	Glomerulonephritis-sparse hair-telangiectasia syndrome
33364	Trichothiodystrophy	670	PIBDS syndrome	79189	Peroxisome biogenesis disorder-Zellweger syndrome spectrum	34	Pipecolic acidemia
33364	Trichothiodystrophy	670	Trichothiodystrophy - sun sensitivity	79189	Peroxisome biogenesis disorder-Zellweger syndrome spectrum	34	Hyperpipecolatemia
33364	Trichothiodystrophy	670	Trichothiodystrophy type F	79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	79261	Glycogen storage disease type 1D
33364	Trichothiodystrophy	453	IBIDS syndrome	79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	79261	Type 1D glycogenosis
33364	Trichothiodystrophy	453	Tay syndrome	79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	79260	Glycogen storage disease type 1C
33364	Trichothiodystrophy	453	Trichothiodystrophy type E	79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	79260	Type 1C glycogenosis
33364	Trichothiodystrophy	453	Trichothiodystrophy with congenital ichthyosis	79452	Milroy disease	79450	Non-hereditary congenital primary lymphedema
33364	Trichothiodystrophy	2739	Oncyo-tricho-dysplasia - neutropenia	79452	Milroy disease	79450	Milroy-like disease
33364	Trichothiodystrophy	2739	Itin syndrome	79500	DOORS syndrome	1674	Digitorenocerebral syndrome
33364	Trichothiodystrophy	2739	ONMR syndrome	79500	DOORS syndrome	1674	DRC syndrome
33364	Trichothiodystrophy	2739	Trichothiodystrophy type G	79500	DOORS syndrome	1674	Eronen-Somer-Gustafsson syndrome
33364	Trichothiodystrophy	3123	Brittle hair syndrome, Sabinas type	79502	Punctate palmoplantar keratoderma type 2	736	Palmoplantar porokeratosis of Mantoux
33364	Trichothiodystrophy	3123	Brittle hair - mental deficiency	83628	PELVIS syndrome	2125	Sacral hemangiomas - multiple congenital abnormalities
33364	Trichothiodystrophy	3123	Trichothiodystrophy type B	86872	T-cell large granular lymphocyte leukemia	2687	Neutropenia - hyperlymphocytosis with large granular lymphocytes
33364	Trichothiodystrophy	231256	Beta-thalassemia - trichothiodystrophy	90186	Meige disease	90185	Non-hereditary late-onset primary lymphedema
33364	Trichothiodystrophy	75790	Pollitt syndrome	90186	Meige disease	90185	Meige-like disease
33364	Trichothiodystrophy	75790	Trichorrhexis nodosa syndrome				
33364	Trichothiodystrophy	75790	Trichothiodystrophy - neurocutaneous syndrome				
33364	Trichothiodystrophy	75790	Trichothiodystrophy type C				
33364	Trichothiodystrophy	75789	SIBIDS syndrome				
33364	Trichothiodystrophy	75789	Trichothiodystrophy - osteosclerosis				
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				
52368	Mohr-Tranebjaerg syndrome	3213	Deafness - opticoacoustic nerve atrophy - dementia				

→ 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
90340	Blau syndrome	90341	Early-onset sarcoidosis	98808	Autosomal dominant dopa-responsive dystonia	101151	DYT14
91387	Familial thoracic aortic aneurysm and aortic dissection	88636	Aortic dilatation - joint hypermobility - arterial tortuosity	98967	Schnyder corneal dystrophy	98968	Central discoid corneal dystrophy
93284	Spondyloepiphyseal dysplasia tarda	163673	Spondyloepiphyseal dysplasia, Byers type	168569	H syndrome	254723	Pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome
93284	Spondyloepiphyseal dysplasia tarda	163673	Spondyloepiphyseal dysplasia - punctate corneal dystrophy	168569	H syndrome	254723	PHID
93950	X-linked intellectual disability, Sutherland-Haan type	93944	X-linked intellectual disability, Fichera type	168569	H syndrome	254712	Familial sinus histiocytosis with massive lymphadenopathy
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis	168569	H syndrome	254712	Familial Rosaï-Dorfman disease
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Antley-Bixler syndrome type 2	168569	H syndrome	254712	Familial SHML
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Antley-Bixler syndrome, POR-related	168569	H syndrome	254707	Faisalabad histiocytosis
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	63269	Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis	168569	H syndrome	254707	FHC
97229	Riboflavin transporter deficiency	56965	Progressive bulbar paralysis of childhood	182050	MYH9-related disease	850	May-Hegglin thrombocytopenia
97229	Riboflavin transporter deficiency	56965	Fazio-Londe disease	182050	MYH9-related disease	850	MHA
97229	Riboflavin transporter deficiency	56965	Progressive bulbar palsy of childhood	182050	MYH9-related disease	850	May-Hegglin anomaly
98769	Spinocerebellar ataxia type 15/16	98770	Spinocerebellar ataxia type 16	182050	MYH9-related disease	850	May-Hegglin syndrome
98769	Spinocerebellar ataxia type 15/16	98770	SCA16	182050	MYH9-related disease	1984	Fechtner syndrome
98772	Spinocerebellar ataxia type 19/22	101107	Spinocerebellar ataxia type 22	182050	MYH9-related disease	1984	Alport syndrome with leukocyte inclusions and macrothrombocytopenia
98772	Spinocerebellar ataxia type 19/22	101107	SCA22	182050	MYH9-related disease	1019	Epstein syndrome
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal hypnogenic dyskinesia	182050	MYH9-related disease	1019	Alport syndrome with macrothrombocytopenia
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Nocturnal paroxysmal dystonia	182050	MYH9-related disease	807	Sebastian syndrome
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal hypnagogic dyskinesia	216866	Classic pantothenate kinase-associated neurodegeneration	157855	Macrothrombocytopenia with leukocyte inclusions
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal hypnagogic dyskinesia	216866	Classic pantothenate kinase-associated neurodegeneration	157855	HARP syndrome
98784	Autosomal dominant nocturnal frontal lobe epilepsy	98812	Paroxysmal hypnagogic dyskinesia	231568	Generalized dominant dystrophic epidermolysis bullosa	216989	Hypoprebetalipoproteinemia - acanthocytosis - retinitis pigmentosa - pallidal degeneration
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 nocturnal dyskinesia	231568	Generalized dominant dystrophic epidermolysis bullosa	79407	DDEB, Pasini type
98808	Autosomal dominant dopa-responsive dystonia	101151	Dystonia 14	231568	Generalized dominant dystrophic epidermolysis bullosa	79407	Autosomal dominant dystrophic epidermolysis bullosa, Cockayne-Touraine type
				261483	Xq27.3q28 duplication syndrome	3423	DDEB, Cockayne-Touraine type
				261483	Xq27.3q28 duplication syndrome	3423	Vasquez-Hurst-Sotos syndrome
				263463	CHST3-related skeletal	1792	Humerospinal dysostosis

→ 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
	dysplasia			319646	PGM1-CDG	711	GSD type 14
263463	CHST3-related skeletal dysplasia	93280	Spondyloepiphyseal dysplasia, Omani type	319646	PGM1-CDG	711	GSDXIV
263463	CHST3-related skeletal dysplasia	93280	Humero-spinal dysostosis	319646	PGM1-CDG	711	Glycogen storage disease type 14
264200	14q22q23 microdeletion syndrome	2055	Growth deficiency - brachydactyly - dysmorphism	319646	PGM1-CDG	711	Glycogenosis due to phosphoglucomutase deficiency
264200	14q22q23 microdeletion syndrome	2055	Frias syndrome	319646	PGM1-CDG	711	Glycogenosis type 14
284963	Marfan syndrome type 1	99715	MASS syndrome	319646	PGM1-CDG	711	Phosphoglucomutase 1 deficiency
284963	Marfan syndrome type 1	99715	Mitral valve-aorta-skeleton-skin syndrome	324737	SRD5A3-CDG	168972	Kahrizi syndrome
289825	Late-onset primary lymphedema	77242	Lymphedema tarda	324737	SRD5A3-CDG	168972	Intellectual disability - cataract - coloboma - kyphosis
289825	Late-onset primary lymphedema	77241	Lymphedema praecox	324737	SRD5A3-CDG	168972	Intellectual disability, Kahrizi type
293843	3MC syndrome	2453	Malpuech syndrome	324737	SRD5A3-CDG	139477	Al-Gazali-Dattani syndrome
293843	3MC syndrome	2453	3MC3 syndrome	329931	C3 glomerulonephritis	93559	C3 deposition glomerulonephritis without proliferation
293843	3MC syndrome	2453	Malpuech facial clefting syndrome	331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	178503	Dursun syndrome
293843	3MC syndrome	2506	Michels syndrome	331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	178503	Pulmonary arterial hypertension - leukopenia - atrial septal defect
293843	3MC syndrome	2506	3MC1 syndrome	357225	Primary non-essential cutis verticis gyrata	1557	Cutis verticis gyrata - intellectual disability
293843	3MC syndrome	2506	Oculopalatoskeletal syndrome	357225	Primary non-essential cutis verticis gyrata	1557	McDowall syndrome
293843	3MC syndrome	2998	Carnevale syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata - retinitis pigmentosa - sensorineural deafness
293843	3MC syndrome	2998	3MC2 syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata - retinitis pigmentosa - neurosensory deafness
293843	3MC syndrome	2998	Carnevale-Krajewska-Fischetto syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata - retinitis pigmentosa - neurosensory hearing loss
293843	3MC syndrome	2998	Mingarelli syndrome	357225	Primary non-essential cutis verticis gyrata	217315	Cutis verticis gyrata - retinitis pigmentosa - sensorineural hearing loss
293843	3MC syndrome	2998	OSA syndrome	357225	Primary non-essential cutis verticis gyrata	79482	Cutis verticis gyrata - thyroid aplasia - intellectual disability
293843	3MC syndrome	2998	Oculo-skeletal-abdominal syndrome	357225	Primary non-essential cutis verticis gyrata	79482	Akesson syndrome
293843	3MC syndrome	2998	Ptosis - strabismus - rectus abdominis diastasis	370114	Combined cervical dystonia	293838	Fatal infantile encephalopathy-pulmonary hypertension syndrome
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	137862	Martínez-Frías syndrome	370953	Congenital muscular dystrophy due to dystroglycanopathy	52428	Congenital muscular dystrophy type 1C
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	137862	Duodenal and extrahepatic biliary atresia - hypoplastic pancreas - intestinal malrotation	370953	Congenital muscular dystrophy due to dystroglycanopathy	52428	CMD1C
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	MDC1C
300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	83618	Severe dilated cardiomyopathy with or without myopathy				
314632	Parkinsonism due to ATP13A2 deficiency	3336	Tomé-Brunet-Fardeau syndrome				
319646	PGM1-CDG	711	Glycogen storage disease due to phosphoglucomutase deficiency				
319646	PGM1-CDG	711	GSD due to phosphoglucomutase deficiency				

→ 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
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	448242	Brachyolmia, recessive type	93301	Brachyolmia type 1, Hobaek type
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Male infertility with normal virilization due to meiosis defect	448242	Brachyolmia, recessive type	93303	Brachyolmia type 1, Toledo type
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Azoospermia due to maturation arrest				
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Azoospermia due to meiosis defect				
399805	Male infertility with azoospermia or oligozoospermia due to single gene mutation	217034	Male infertility with normal virilization due to maturation arrest				
399808	Male infertility with teratozoospermia due to single gene mutation	352613	Male infertility due to NANOS1 mutation				
402041	Autosomal recessive distal renal tubular acidosis	93609	Autosomal recessive distal renal tubular acidosis without deafness				
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				

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

Editor-in-chief :Ana Rath – Editor: David Kelly

Technical support: Samuel Demarest, Valérie Lanneau - Photography: Alliance Maladies Rares / Karine Lhémond

The correct form when quoting this document is :

« List of rare diseases and synonyms listed in alphabetical order », Orphanet Report Series, Rare Diseases collection,
March 2016,

[This Orphanet Report Series is part of the joint action 677024 RD-ACTION which has received funding from the European Union's Health Programme \(2014-2020\).](http://www.orpha.net/orphacom/cahiers/docs/GB>List_of_rare_diseases_in_alpha.pdf</p></div><div data-bbox=)

The content of this Orphanet Report Series represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.