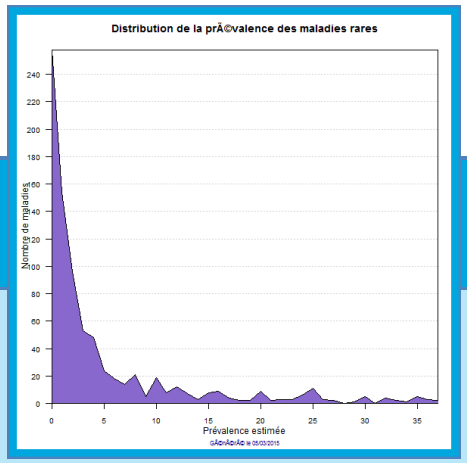

Orphanet Report Series
Rare Diseases collection

Number 2 | October 2024

Prevalence and incidence of rare diseases: Bibliographic data

Diseases listed by decreasing prevalence, incidence or number of published cases

RÉPUBLIQUE FRANÇAISE
Liberté
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Inserm
La science pour la santé
From science to health

MINISTÈRE DE LA SANTÉ ET DE LA PRÉVENTION
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Methodology

Orphanet carries out a systematic survey of literature in order to estimate the prevalence and incidence of rare diseases. This study aims to collect new data regarding point prevalence, birth prevalence and incidence, and to update already published data according to new scientific studies or other available data.

This data is presented in the following reports published biannually:

- Prevalence, incidence or number of published cases listed by diseases (in alphabetical order) ;
- Diseases listed by decreasing prevalence, incidence or number of published cases ;

Data collection

A number of different sources are used :

- Registries (RARECARE, EUROCAT, etc) ;
- National/international health institutes and agencies (Institut National de Veille Sanitaire (French Institute of Health Surveillance); American Center of Disease Control and Prevention, American National Cancer Institute, European Medicines Agency, World Health Organization etc) ;
- Medline is consulted using the following search algorithm : «Disease names» AND Epidemiology[MeSH:NoExp] OR Incidence[Title/abstract] OR Prevalence[Title/ abstract] OR Epidemiology[Title/abstract];
- Medical texts, grey literature and reports from experts ;
- Orphanet collaborating experts

Data characteristics

The data published in this document are worldwide estimations, or European estimations if a worldwide estimation is not available.

The published data is raw collected data or extrapolations of raw data at worldwide or European level when no genetic founder effect is suspected as a cause of a disease.

If a range of national data is available, the average is calculated to estimate the worldwide or European prevalence or incidence. When a range of data sources is available, the most recent data source that meets a certain number of quality criteria is favoured (registries, meta-analyses, population-based studies, large cohort studies).

For congenital diseases, the prevalence is estimated, so that:
Prevalence = birth prevalence x (patient life expectancy/general population life expectancy).

When only incidence data is documented, the prevalence is estimated when possible, so that:
Prevalence = incidence x disease mean duration.

When neither prevalence nor incidence data is available, which is the case for very rare diseases, the number of cases or families documented in the medical literature is provided.

Without specification, published figures are worldwide. An asterisk * indicates European data.
BP indicates birth prevalence
**This data is currently bing reevaluated.

Limitations of the study

The prevalence and incidence data presented in this report are only estimations and cannot be considered to be absolutely correct.
The average values presented in this report do not take into account the heterogeneous nature of the methodologies employed by the studies considered in the literature survey.

The validity and exactitude of raw data sources is taken for granted and have not been verified. Thus, confusion between terms such as incidence and prevalence and/or birth prevalence is possible due to the interchangeable use of these terms in certain sources.

It is possible that prevalence is overestimated in some cases as epidemiological studies are generally based on hospital data in regions with higher prevalence.

Data presentation

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Please note that this is just a selection of Orphanet's rare disease epidemiological data. Currently 4363 rare diseases are annotated with prevalence or incidence information in the Orphanet database. To access the complete data sets visit Orphadata (www.orphadata.com).

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List of diseases by decreasing prevalence

ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
870	Down syndrome**	Disorder	95.0 BP
870	Down syndrome**	Disorder	57.0 *
870	Down syndrome**	Disorder	101.0 BP*
199306	Cleft lip/palate	Disorder	80.0 BP
853	Fetal and neonatal alloimmune thrombocytopenia	Disorder	39.6307
2965	Prolactinoma	Disorder	50.7 *
93100	Renal agenesis, unilateral	Subtype of disorder	50.0 BP
90066	Pneumonia caused by Pseudomonas aeruginosa infection	Disorder	50.0 *
8	47,XYY syndrome	Disorder	50.0 BP*
63259	Iniencephaly	Disorder	50.0 BP*
48	Congenital bilateral absence of vas deferens	Disorder	50.0 *
289390	Primary Sjögren syndrome	Disorder	48.99 *
67038	B-cell chronic lymphocytic leukemia	Disorder	48.0 *
2185	Congenital hydrocephalus	Disorder	46.5 BP*
391673	Necrotizing enterocolitis	Disorder	45.0
275555	Preeclampsia	Disorder	45.0 *
137686	Asherman syndrome	Disorder	44.0 *
536	Systemic lupus erythematosus	Disorder	43.7
93108	Renal dysplasia	Disorder	43.5 BP*
3375	Trisomy X	Disorder	42.5 *
363999	Non-immune hydrops fetalis	Subtype of disorder	42.0 BP
97292	Cardiogenic shock	Disorder	40.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
90059	Sudden sensorineural hearing loss	Disorder	40.0 *
402823	Hepatitis delta	Disorder	40.0 *
294	Fetal cytomegalovirus syndrome	Disorder	40.0 *
101016	Romano-Ward syndrome	Disorder	40.0 *
853	Fetal and neonatal alloimmune thrombocytopenia	Disorder	66.6667 BP
730	Autosomal dominant polycystic kidney disease	Disorder	39.6 *
90056	Moderate and severe traumatic brain injury	Disorder	37.8 *
567	22q11.2 deletion syndrome	Disorder	9.6 BP*
545	Follicular lymphoma	Disorder	37.0 *
340	Hemorrhagic fever-renal syndrome	Disorder	37.0 *
209989	Non-papillary transitional cell carcinoma of the bladder	Disorder	37.0 *
231080	High-grade dysplasia in patients with Barrett esophagus	Disorder	36.0 *
1457	Aorta coarctation	Disorder	35.6 BP*
94059	Uremic pruritus	Disorder	35.0 *
70475	Radiation proctitis	Disorder	35.0 *
2764	Osteochondritis dissecans	Disorder	35.0 *
1048	Isolated anencephaly/exencephaly	Disorder	35.0 BP*
3303	Tetralogy of Fallot	Disorder	34.0 BP
636	Neurofibromatosis type 1	Disorder	21.3 *
858	Congenital toxoplasmosis	Disorder	33.0 BP*
439167	Placental insufficiency	Disorder	33.0
908	Fragile X syndrome	Disorder	32.5
908	Fragile X syndrome	Disorder	2.4 BP*
90058	Spinal cord injury	Disorder	32.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
90051	Sepsis in premature infants	Disorder	32.0 *
70476	Vernal keratoconjunctivitis	Disorder	32.0 *
791	Retinitis pigmentosa	Disorder	30.0 *
729	Polycythemia vera	Disorder	30.0 *
563	Peripartum cardiomyopathy	Disorder	30.0 BP
33208	Idiopathic hypersomnia	Disorder	30.0 *
314701	Primary systemic amyloidosis	Subtype of disorder	30.0 *
2140	Congenital diaphragmatic hernia	Disorder	30.0 BP
2073	Narcolepsy type 1	Disorder	30.0 *
1330	Partial atrioventricular septal defect	Disorder	20.0 BP*
577	Mucolipidosis type III	Disorder	29.55 *
3303	Tetralogy of Fallot	Disorder	29.3 BP*
411527	Central retinal vein occlusion	Disorder	28.0 *
582	Mucopolysaccharidosis type 4	Disorder	0.45 BP*
582	Mucopolysaccharidosis type 4	Disorder	0.07 BP
1656	Dermatitis herpetiformis	Disorder	27.0 *
791	Retinitis pigmentosa	Disorder	26.7
70568	Post-transplant lymphoproliferative disease	Disorder	26.2 *
137698	Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	Disorder	25.5 *
95719	Thyroid hemiagenesis	Disorder	25.0
93402	Syndactyly type 1	Disorder	25.0 BP*
703	Bullous pemphigoid	Disorder	25.0 *
701	Alopecia universalis	Disorder	25.0 *
3002	Immune thrombocytopenia	Disorder	25.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
186	Primary biliary cholangitis	Disorder	21.05
140286	Secondary hypoparathyroidism due to impaired parathormon secretion	Disorder	24.75 *
94058	Neovascular glaucoma	Disorder	24.4 *
1199	Esophageal atresia	Disorder	24.3 BP*
860	Congenitally uncorrected transposition of the great arteries	Disorder	24.25 BP*
2137	Autoimmune hepatitis	Disorder	23.5
1851	Multicystic dysplastic kidney	Disorder	23.26 BP
97363	Unilateral multicystic dysplastic kidney	Subtype of disorder	23.2 BP
90080	Scarring in glaucoma filtration surgical procedures	Disorder	22.0 *
217080	Pulmonary fungal infections in patients deemed at risk	Disorder	22.0 *
217067	Pouchitis	Disorder	22.0 *
636	Neurofibromatosis type 1	Disorder	33.3 BP
2140	Congenital diaphragmatic hernia	Disorder	21.2 BP*
186	Primary biliary cholangitis	Disorder	25.0 *
1646	Chromosome Y microdeletion	Disorder	20.8
85410	Oligoarticular juvenile idiopathic arthritis	Disorder	20.5 *
908	Fragile X syndrome	Disorder	20.0 *
90081	AIDS wasting syndrome	Disorder	20.0 *
90062	Acute liver failure	Disorder	20.0 *
797	Sarcoidosis	Disorder	20.0 *
70	Proximal spinal muscular atrophy	Disorder	20.0 BP*
66627	Tenosynovial giant cell tumor	Disorder	20.0 *
60	Alpha-1-antitrypsin deficiency	Disorder	20.0 *
589	Myasthenia gravis	Disorder	20.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
558	Marfan syndrome	Disorder	15.0
35122	Congenital sucrase-isomaltase deficiency	Disorder	20.0 *
261197	Proximal 16p11.2 microdeletion syndrome	Disorder	20.0 *
171673	Limbal stem cell deficiency	Disorder	20.0 *
1646	Chromosome Y microdeletion	Disorder	20.0 *
137583	Vulvar intraepithelial neoplasia	Disorder	20.0 *
1330	Partial atrioventricular septal defect	Disorder	30.0 *
1329	Complete atrioventricular septal defect	Disorder	20.0 BP*
130	Brugada syndrome	Disorder	20.0 *
586	Cystic fibrosis	Disorder	19.3912 BP*
95706	Non-syndromic posterior hypospadias	Disorder	19.25 BP*
30391	Isolated biliary atresia	Disorder	2.9 BP*
228113	Anal fistula	Disorder	18.3 *
91127	Adenovirus infection in immunocompromised patients	Disorder	18.0 *
704	Pemphigus vulgaris	Disorder	18.0 *
2248	Hypoplastic left heart syndrome	Disorder	18.0 BP
154	Familial isolated dilated cardiomyopathy	Disorder	17.5 *
2368	Gastroschisis	Disorder	16.9 BP*
3380	Trisomy 18	Disorder	16.7 BP
461	Recessive X-linked ichthyosis	Disorder	16.6 *
2032	Idiopathic pulmonary fibrosis	Disorder	16.125
90064	Acute peripheral arterial occlusion	Disorder	16.0 *
774	Hereditary hemorrhagic telangiectasia	Disorder	16.0 *
54370	Primary membranoproliferative glomerulonephritis	Disorder	16.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
36258	Buerger disease	Disorder	16.0
83463	Microtia	Disorder	13.0 BP*
90291	Systemic sclerosis	Disorder	15.4 *
2248	Hypoplastic left heart syndrome	Disorder	15.1 BP*
558	Marfan syndrome	Disorder	20.0 *
388	Hirschsprung disease	Disorder	13.2 BP*
388	Hirschsprung disease	Disorder	15.0
388	Hirschsprung disease	Disorder	13.2 *
309297	Mucopolysaccharidosis type 4A	Subtype of disorder	15.0 *
2828	Young-onset Parkinson disease	Disorder	15.0 *
2382	Lennox-Gastaut syndrome	Disorder	15.0 *
221061	Familial cerebral cavernous malformation	Disorder	15.0
163934	Atopic keratoconjunctivitis	Disorder	15.0 *
97363	Unilateral multicystic dysplastic kidney	Subtype of disorder	14.8 BP*
166260	Dentinogenesis imperfecta type 2	Subtype of disorder	14.6 *
49042	Dentinogenesis imperfecta	Disorder	14.5 *
95712	Thyroid ectopia	Disorder	14.3 *
683	Progressive supranuclear palsy	Disorder	5.26
238624	Idiopathic intracranial hypertension	Disorder	14.0 *
214	Cystinuria	Disorder	14.0
2162	Holoprosencephaly	Disorder	13.4 BP*
3193	Supravalvular aortic stenosis	Disorder	4.0 BP*
388	Hirschsprung disease	Disorder	15.0 BP
83463	Microtia	Disorder	15.5 BP

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
827	Stargardt disease	Disorder	13.0 *
70589	Bronchopulmonary dysplasia	Disorder	13.0 *
449266	Pleural empyema	Disorder	13.0 *
44890	Gastrointestinal stromal tumor	Disorder	13.0 *
423461	Mucolipidosis type III alpha/beta	Subtype of disorder	13.0
3376	Triploidy	Disorder	12.6 BP*
85138	Addison disease	Disorder	12.5 *
285	Hypermobile Ehlers-Danlos syndrome	Disorder	12.5 *
273	Steinert myotonic dystrophy	Disorder	5.0 *
828	Stickler syndrome	Disorder	1.0 BP*
86870	Blastic plasmacytoid dendritic cell neoplasm	Disorder	12.0 *
70573	Small cell lung cancer	Disorder	12.0 *
42	Medium chain acyl-CoA dehydrogenase deficiency	Disorder	12.0 BP*
399	Huntington disease	Disorder	2.7
29073	Multiple myeloma	Disorder	11.9 *
660	Omphalocele	Disorder	11.7 BP*
716	Phenylketonuria	Disorder	11.4 BP*
716	Phenylketonuria	Disorder	4.1366
716	Phenylketonuria	Disorder	11.5079 *
98878	Hemophilia A	Disorder	4.85
98878	Hemophilia A	Disorder	8.0 *
586	Cystic fibrosis	Disorder	11.1319 *
635	Neuroblastoma	Disorder	11.0 *
3109	Mayer-Rokitansky-Küster-Hauser syndrome	Disorder	11.0 BP

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
700	Alopecia totalis	Disorder	10.5 *
3380	Trisomy 18	Disorder	10.4 BP*
3366	Non-syndromic metopic craniosynostosis	Disorder	6.7 BP*
903	Von Willebrand disease	Disorder	10.0
90076	Partial deep dermal and full thickness burns	Disorder	10.0 *
90065	Acquired aneurysmal subarachnoid hemorrhage	Disorder	10.0 *
778	Rett syndrome	Disorder	5.0 BP*
654	Nephroblastoma	Disorder	10.0 BP*
569	Familial or sporadic hemiplegic migraine	Disorder	10.0 *
51	Aicardi-Goutières syndrome	Disorder	10.0 *
412	Dysbetalipoproteinemia	Disorder	7.8 *
36258	Buerger disease	Disorder	10.0 *
3286	Catecholaminergic polymorphic ventricular tachycardia	Disorder	10.0 *
3157	Septo-optic dysplasia spectrum	Disorder	10.0 BP*
31112	Dermatofibrosarcoma protuberans	Disorder	10.0 *
233	Duane retraction syndrome	Disorder	10.0 *
232	Sickle cell anemia	Disorder	10.0 *
1114	Aplasia cutis congenita	Disorder	10.0 BP
98896	Duchenne muscular dystrophy	Disorder	9.9 BP
567	22q11.2 deletion syndrome	Disorder	37.5 BP
79665	Gardner syndrome	Subtype of disorder	9.1 BP
99125	Congenital total pulmonary venous return anomaly	Disorder	9.0 BP
99125	Congenital total pulmonary venous return anomaly	Disorder	9.0
900	Granulomatosis with polyangiitis	Disorder	9.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
306644	Complication after organ transplantation	Disorder	9.0 *
138	CHARGE syndrome	Disorder	6.5 BP
1203	Duodenal atresia	Disorder	9.0 BP*
1203	Duodenal atresia	Disorder	9.0 *
1201	Small bowel atresia	Disorder	9.0 BP*
137914	Choanal atresia	Disorder	8.6 BP*
99981	Apnea of prematurity	Disorder	8.5 *
2444	Congenital pulmonary airway malformation	Disorder	8.2 BP*
171	Primary sclerosing cholangitis	Disorder	7.84 *
666	Osteogenesis imperfecta	Disorder	8.06
98878	Hemophilia A	Disorder	11.25 BP
95702	X-linked adrenal hypoplasia congenita	Disorder	8.0
95702	X-linked adrenal hypoplasia congenita	Disorder	8.0 BP
930	Idiopathic achalasia	Disorder	8.0
85408	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	Disorder	8.0 *
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Disorder	1.0 BP*
171	Primary sclerosing cholangitis	Disorder	8.1
412	Dysbetalipoproteinemia	Disorder	10.0
589	Myasthenia gravis	Disorder	7.77
247234	Sporadic adult-onset ataxia of unknown etiology	Disorder	7.6 *
221	Dermatomyositis	Disorder	7.5312
72	Angelman syndrome	Disorder	7.5
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	Subtype of disorder	7.5 BP*
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	Subtype of disorder	7.5 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
2004	Laryngotracheoesophageal cleft	Disorder	7.5 BP*
1464	Univentricular heart	Disorder	7.5 BP
821	Sotos syndrome	Disorder	7.1 BP
732	Polymyositis	Disorder	7.1 *
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Disorder	7.0 *
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Disorder	7.0 BP
90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients	Disorder	7.0 *
705	Pendred syndrome	Disorder	7.0 *
261236	16p13.11 microdeletion syndrome	Disorder	7.0 BP
2059	Fryns syndrome	Disorder	7.0 BP*
42	Medium chain acyl-CoA dehydrogenase deficiency	Disorder	6.85
57145	SUNCT syndrome	Disorder	6.7 *
3366	Non-syndromic metopic craniosynostosis	Disorder	10.2979 *
238468	Hypohidrotic ectodermal dysplasia	Disorder	6.7 *
42062	Iminoglycinuria	Disorder	6.67 BP*
42062	Iminoglycinuria	Disorder	6.68 *
324	Fabry disease	Disorder	6.66 BP
50839	Cat-scratch disease	Disorder	6.6 *
138	CHARGE syndrome	Disorder	9.0 *
716	Phenylketonuria	Disorder	6.4 BP
79254	Classic phenylketonuria	Subtype of disorder	6.0
79254	Classic phenylketonuria	Subtype of disorder	6.0 BP
79254	Classic phenylketonuria	Subtype of disorder	6.34 *
887	VACTERL/VATER association	Disorder	6.25 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
905	Wilson disease	Disorder	2.25 BP
905	Wilson disease	Disorder	2.02
79254	Classic phenylketonuria	Subtype of disorder	6.34 BP*
790	Retinoblastoma	Disorder	6.0 BP
733	Familial adenomatous polyposis	Disorder	6.0 *
609	Tibial muscular dystrophy	Disorder	6.0 *
521	Chronic myeloid leukemia	Disorder	6.0 *
46724	Cerebral arteriovenous malformation	Disorder	6.0 *
411703	Pulmonary non-tuberculous mycobacterial infection	Disorder	6.0 *
3451	Infantile epileptic spasms syndrome	Disorder	6.0 *
3451	Infantile epileptic spasms syndrome	Disorder	3.7 BP
252164	Benign schwannoma	Disorder	6.0 *
635	Neuroblastoma	Disorder	5.8 BP*
85438	Enthesitis-related juvenile idiopathic arthritis	Disorder	5.7 *
1209	Tricuspid atresia	Disorder	5.5625 BP*
85443	AL amyloidosis	Disorder	5.127
93372	Familial hypocalciuric hypercalcemia type 1	Subtype of disorder	5.5
881	Turner syndrome	Disorder	5.5 BP*
2440	Isolated split hand-split foot malformation	Disorder	5.4 BP*
819	Smith-Magenis syndrome	Disorder	4.0
683	Progressive supranuclear palsy	Disorder	14.0 *
803	Amyotrophic lateral sclerosis	Disorder	5.2 *
85443	AL amyloidosis	Disorder	5.5311 *
98838	Primary mediastinal large B-cell lymphoma	Disorder	5.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
79271	Sanfilippo syndrome type C	Subtype of disorder	5.0 *
792	X-linked retinoschisis	Disorder	5.0
778	Rett syndrome	Disorder	10.0 *
766	Hemolytic anemia due to red cell pyruvate kinase deficiency	Disorder	5.0 *
718	Isolated Pierre Robin syndrome	Disorder	5.0 BP*
53	Albers-Schönberg osteopetrosis	Disorder	1.0
469	Hereditary fructose intolerance	Disorder	5.0 *
39812	Graft versus host disease	Disorder	5.0 *
287	Classical Ehlers-Danlos syndrome	Disorder	5.0
273	Steinert myotonic dystrophy	Disorder	12.5
244	Primary ciliary dyskinesia	Disorder	5.0 BP*
214	Cystinuria	Disorder	5.0 *
1332	Medullary thyroid carcinoma	Disorder	5.0 *
1198	Colonic atresia	Disorder	5.0 BP
88629	Tritanopia	Disorder	4.8 *
3384	Common arterial trunk	Disorder	4.3 BP
15	Achondroplasia	Disorder	4.73 BP
60041	Congenital heart block	Disorder	4.54 BP
85446	Wild type ABeta2M amyloidosis	Disorder	4.5 *
792	X-linked retinoschisis	Disorder	4.5 *
269	Facioscapulohumeral dystrophy	Disorder	4.5 *
60015	Enlarged parietal foramina	Disorder	4.3 *
3384	Common arterial trunk	Disorder	4.8 BP*
1143	Neurogenic arthrogryposis multiplex congenita	Disorder	4.3 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
104	Leber hereditary optic neuropathy	Disorder	4.3
727	Microscopic polyangiitis	Disorder	4.2843
137599	Herpes simplex virus stromal keratitis	Disorder	4.2091
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	Disorder	4.2 *
2116	Hartnup disease	Disorder	4.2
137596	Neurotrophic keratopathy	Disorder	4.2 *
391655	Off-periods in Parkinson disease not responding to oral treatment	Disorder	4.15 *
93110	Posterior urethral valve	Disorder	2.0 *
2300	Multiple intestinal atresia	Disorder	4.05 BP
96253	Cushing disease	Disorder	4.0 *
95716	Familial thyroid dyshormonogenesis	Disorder	4.0 *
884	Tetrasomy 12p	Disorder	4.0 BP*
819	Smith-Magenis syndrome	Disorder	5.35 *
79140	Cutaneous neuroendocrine carcinoma	Disorder	4.0 *
564	Meckel syndrome	Disorder	4.0 BP
52417	MALT lymphoma	Disorder	4.0 *
3193	Supravalvular aortic stenosis	Disorder	13.3 *
1928	Congenital lobar emphysema	Disorder	4.0 BP
178029	Arginine vasopressin deficiency	Disorder	4.0 *
101330	Porphyria cutanea tarda	Disorder	4.0 *
803	Amyotrophic lateral sclerosis	Disorder	3.85
79126	Acute interstitial pneumonia	Disorder	3.8 *
481	Kennedy disease	Disorder	3.8 *
478	Kallmann syndrome	Subtype of disorder	3.75 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
818	Smith-Lemli-Opitz syndrome	Disorder	3.7 BP*
60015	Enlarged parietal foramina	Disorder	3.7
3465	Worster-Drought syndrome	Disorder	3.7 *
3451	Infantile epileptic spasms syndrome	Disorder	3.5 BP*
3378	Trisomy 13	Disorder	3.7 BP*
2932	Chronic inflammatory demyelinating polyneuropathy	Disorder	3.7 *
102	Multiple system atrophy	Disorder	3.7 *
15	Achondroplasia	Disorder	3.62 BP*
95720	Thyroid hypoplasia	Disorder	3.5
95713	Athyreosis	Disorder	3.5 *
81	Antisynthetase syndrome	Disorder	3.5
640	Hereditary neuropathy with liability to pressure palsies	Disorder	3.5 *
52416	Mantle cell lymphoma	Disorder	3.5 *
3205	Sturge-Weber syndrome	Disorder	3.5 BP*
2655	Thanatophoric dysplasia	Disorder	3.5 BP*
1880	Ebstein malformation of the tricuspid valve	Disorder	1.25 *
116	Beckwith-Wiedemann syndrome	Disorder	3.5 BP*
102	Multiple system atrophy	Disorder	3.5
218	Darier disease	Disorder	3.4 *
53271	Muenke syndrome	Disorder	3.33 BP
3306	Inverted duplicated chromosome 15 syndrome	Disorder	3.33 BP
750	Pseudoachondroplasia	Disorder	3.3
652	Multiple endocrine neoplasia type 1	Disorder	3.3 *
33069	Dravet syndrome	Disorder	3.3 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
926	Acatalasemia	Disorder	3.2 *
158	Systemic primary carnitine deficiency	Disorder	3.2 BP*
100075	Neuroendocrine tumor of stomach	Disorder	3.2 *
767	Polyarteritis nodosa	Disorder	3.16 *
98916	Acute inflammatory demyelinating polyradiculoneuropathy	Disorder	3.1 *
85414	Systemic-onset juvenile idiopathic arthritis	Disorder	3.1
739	Prader-Willi syndrome	Disorder	3.1 BP*
50251	Pleural mesothelioma	Disorder	3.1 *
2322	Kabuki syndrome	Disorder	3.1 *
93930	Bladder exstrophy	Subtype of disorder	3.05 BP
429	Hypochondroplasia	Disorder	3.0303 BP
429	Hypochondroplasia	Disorder	3.0303
98879	Hemophilia B	Disorder	3.0 *
86875	Adult T-cell leukemia/lymphoma	Disorder	3.0 *
824	Primary myelofibrosis	Disorder	3.0 *
794	Saethre-Chotzen syndrome	Disorder	3.0 BP*
673	Malaria	Disorder	3.0 *
565782	Methotrexate toxicity	Disorder	3.0 *
365	Glycogen storage disease due to acid maltase deficiency	Disorder	0.8 BP*
36234	Bacterial toxic-shock syndrome	Disorder	3.0
321	Multiple osteochondromas	Disorder	3.0 *
238621	Ileal pouch anal anastomosis related faecal incontinence	Disorder	3.0 *
216694	Congenitally corrected transposition of the great arteries	Disorder	3.0 BP
136	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	Disorder	3.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
653	Multiple endocrine neoplasia type 2	Disorder	2.9 *
30391	Isolated biliary atresia	Disorder	18.5 BP
98896	Duchenne muscular dystrophy	Disorder	2.8
506	Leigh syndrome	Disorder	2.8 BP*
169802	Severe hemophilia A	Subtype of disorder	2.8 *
626	Large/giant congenital melanocytic nevus	Disorder	2.75 *
49382	Achromatopsia	Disorder	2.7
399	Huntington disease	Disorder	12.0 *
747	Autoimmune pulmonary alveolar proteinosis	Disorder	2.66
6	3-methylcrotonyl-CoA carboxylase deficiency	Disorder	2.65 BP*
564	Meckel syndrome	Disorder	2.6 BP*
79432	Oculocutaneous albinism type 2	Disorder	2.55
93321	Isolated radial hemimelia	Disorder	2.5 BP
758	Pseudoxanthoma elasticum	Disorder	2.5 *
65	Leber congenital amaurosis	Disorder	2.5 BP
65	Leber congenital amaurosis	Disorder	2.5
393	46,XX testicular difference of sex development	Disorder	2.5
358	Gitelman syndrome	Disorder	2.5 *
352731	Oculocutaneous albinism type 1	Disorder	2.5
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	Subtype of disorder	2.5 *
2337	Diffuse palmoplantar keratoderma, Bothnian type	Disorder	2.5 *
2138	46,XX ovotesticular difference of sex development	Disorder	2.5 BP
1872	Cone rod dystrophy	Disorder	2.5 *
1600	Monosomy 18q	Disorder	2.5 BP

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
107	BOR syndrome	Disorder	2.5
100070	Progressive non-fluent aphasia	Disorder	2.5 *
70588	Meconium aspiration syndrome	Disorder	2.44 *
98933	Multiple system atrophy, parkinsonian type	Subtype of disorder	2.4 *
93928	Isolated epispadias	Subtype of disorder	2.4 BP*
247525	Citrullinemia type I	Disorder	2.4 *
330015	Lead poisoning	Disorder	2.3 *
104	Leber hereditary optic neuropathy	Disorder	2.3 *
905	Wilson disease	Disorder	6.0 *
98976	Congenital glaucoma	Disorder	2.2 BP*
98895	Becker muscular dystrophy	Disorder	2.0 *
98895	Becker muscular dystrophy	Disorder	1.53
454750	Isolated tracheoesophageal fistula	Disorder	2.2 BP
454750	Isolated tracheoesophageal fistula	Disorder	2.2
2869	Peutz-Jeghers syndrome	Disorder	2.2 BP
137605	Legius syndrome	Disorder	2.2 BP
89936	X-linked hypophosphatemia	Disorder	1.66 *
70567	Cholangiocarcinoma	Disorder	2.1
217	Isolated Dandy-Walker malformation	Disorder	1.0 BP*
71211	Neuromyelitis optica spectrum disorder	Disorder	2.071
280921	Idiopathic panuveitis	Disorder	2.0194 *
98895	Becker muscular dystrophy	Disorder	2.2 BP*
98841	Anaplastic large cell lymphoma	Disorder	2.0 *
98673	Autosomal dominant optic atrophy, classic form	Disorder	2.0

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
95	Friedreich ataxia	Disorder	2.0 *
93110	Posterior urethral valve	Disorder	4.125 BP*
90073	Hepatitis B reinfection following liver transplantation	Disorder	2.0 *
861	Treacher-Collins syndrome	Disorder	2.0 BP*
83418	Proximal spinal muscular atrophy type 2	Subtype of disorder	2.0 BP*
54595	Craniopharyngioma	Disorder	2.0 *
506	Leigh syndrome	Disorder	2.0 *
480	Kearns-Sayre syndrome	Disorder	2.0 *
447	Paroxysmal nocturnal hemoglobinuria	Disorder	2.0 *
377	Gorlin syndrome	Disorder	2.0 *
3392	Tularemia	Disorder	2.0 *
3346	Tracheal agenesis	Disorder	2.0 BP*
3129	Sarcosinemia	Disorder	2.0 BP
280	Wolf-Hirschhorn syndrome	Disorder	2.0 BP*
275761	Lysosomal acid lipase deficiency	Disorder	2.0 *
26790	Pseudomyxoma peritonei	Disorder	2.0 *
2345	Isolated Klippel-Feil syndrome	Disorder	2.0 *
217064	5-fluorouracil poisoning	Disorder	2.0 *
2017	Sternal cleft	Disorder	2.0 BP*
185	Scimitar syndrome	Disorder	2.0 BP*
180	Choroideremia	Disorder	2.0 *
1699	Trisomy 12p	Disorder	2.0 BP
168782	Childhood disintegrative disorder	Disorder	2.0 *
166119	Isolated osteopoikilosis	Disorder	2.0

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
1598	Monosomy 18p	Disorder	2.0 BP*
150	Nasopharyngeal carcinoma	Disorder	2.0 *
10	48,XXYY syndrome	Disorder	1.9 BP*
140	Campomelic dysplasia	Disorder	0.0003
363958	17q21.31 microdeletion syndrome	Subtype of disorder	1.82 *
675	Annular pancreas	Disorder	1.8 BP*
664	Ornithine transcarbamylase deficiency	Disorder	1.0 *
420429	Glycogen storage disease due to acid maltase deficiency, late-onset	Subtype of disorder	1.75 BP
330001	Wild type ATTR amyloidosis	Disorder	1.72
251076	8p23.1 duplication syndrome	Disorder	1.72
637	Full NF2-related schwannomatosis	Disorder	1.7 *
623615	Autoimmune limbic encephalitis	Disorder	1.7 *
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	Subtype of disorder	1.7 BP
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	Subtype of disorder	1.7
2152	Mowat-Wilson syndrome	Disorder	1.7 BP*
1848	Renal agenesis, bilateral	Subtype of disorder	1.7 BP*
141077	Epignathus	Subtype of disorder	0.0017
475	Joubert syndrome	Disorder	1.6666 BP
98879	Hemophilia B	Disorder	1.665 BP
89936	X-linked hypophosphatemia	Disorder	2.14
899	Walker-Warburg syndrome	Disorder	1.65 BP*
394	Homocystinuria due to cystathionine beta-synthase deficiency	Disorder	0.3 BP
79241	Biotinidase deficiency	Disorder	1.6 BP
79241	Biotinidase deficiency	Disorder	1.6 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
1915	Fetal alcohol syndrome	Disorder	1.6 BP*
183	Eosinophilic granulomatosis with polyangiitis	Disorder	1.56 *
633228	Isolated proximal femoral focal deficiency	Disorder	1.55
633228	Isolated proximal femoral focal deficiency	Disorder	1.55 BP
98757	Spinocerebellar ataxia type 3	Disorder	1.5
98756	Spinocerebellar ataxia type 2	Disorder	1.5
98755	Spinocerebellar ataxia type 1	Disorder	1.5
641	Multifocal motor neuropathy	Disorder	1.5
45453	Incessant infant ventricular tachycardia	Disorder	1.5 BP*
45452	Idiopathic neonatal atrial flutter	Disorder	1.5 BP*
389	Langerhans cell histiocytosis	Disorder	1.5 *
35689	Primary lateral sclerosis	Disorder	1.5 *
2911	Poland syndrome	Disorder	1.5 BP*
213	Cystinosis	Disorder	0.75 BP
213	Cystinosis	Disorder	1.5 *
2019	Femur-fibula-ulna complex	Disorder	1.5 BP*
192	Coffin-Lowry syndrome	Disorder	1.5
192	Coffin-Lowry syndrome	Disorder	1.5 *
183	Eosinophilic granulomatosis with polyangiitis	Disorder	1.5
131	Budd-Chiari syndrome	Disorder	1.5 *
512	Metachromatic leukodystrophy	Disorder	1.47 BP*
79269	Sanfilippo syndrome type A	Subtype of disorder	0.32 *
474	Jeune syndrome	Disorder	1.4 BP*
195	Cat-eye syndrome	Disorder	1.35 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
3287	Takayasu arteritis	Disorder	1.34 *
79434	Oculocutaneous albinism type 1B	Subtype of disorder	1.3
79431	Oculocutaneous albinism type 1A	Subtype of disorder	1.3
79408	Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	Disorder	1.3 BP*
72	Angelman syndrome	Disorder	1.3 BP*
499009	Congenital syphilis	Disorder	1.3 *
499009	Congenital syphilis	Disorder	1.3 BP*
355	Gaucher disease	Disorder	1.0 *
281090	Syndromic recessive X-linked ichthyosis	Disorder	1.3 *
2481	Neurocutaneous melanocytosis	Disorder	1.25 *
1880	Ebstein malformation of the tricuspid valve	Disorder	3.5 BP*
199	Cornelia de Lange syndrome	Disorder	1.24 BP*
628	Diastrophic dysplasia	Disorder	1.2 *
464	Incontinentia pigmenti	Disorder	1.2 BP*
2750	Orofaciodigital syndrome type 1	Disorder	1.2 BP*
263432	Nevus of Ito	Disorder	1.17 *
1896	EEC syndrome	Disorder	1.11 BP*
93323	Isolated fibular hemimelia	Disorder	1.1033 BP
93323	Isolated fibular hemimelia	Disorder	1.1033
377	Gorlin syndrome	Disorder	1.1
289	Ellis Van Creveld syndrome	Disorder	0.4 BP*
275766	Idiopathic pulmonary arterial hypertension	Subtype of disorder	1.1 *
131	Budd-Chiari syndrome	Disorder	1.1
1906	Fetal valproate spectrum disorder	Disorder	1.02 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
99789	Dentin dysplasia type I	Subtype of disorder	1.0 *
98863	X-linked Emery-Dreifuss muscular dystrophy	Subtype of disorder	1.0 BP
98863	X-linked Emery-Dreifuss muscular dystrophy	Subtype of disorder	1.0
96263	48,XXXY syndrome	Disorder	1.0 BP*
95715	Congenital hypothyroidism due to transplacental passage of TSH-binding inhibitory antibodies	Disorder	1.0 *
94068	Spondyloepiphyseal dysplasia congenita	Disorder	1.0 BP*
90068	Cocaine intoxication	Disorder	1.0 *
90060	Diffuse alveolar hemorrhage	Disorder	1.0 *
87503	Mal de Meleda	Disorder	1.0
86867	Nodal marginal zone B-cell lymphoma	Disorder	1.0 *
828	Stickler syndrome	Disorder	12.2 BP
79435	Oculocutaneous albinism type 4	Disorder	1.0
79258	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia	Subtype of disorder	1.0 BP*
79086	Acquired generalized lipodystrophy	Disorder	1.0 *
77259	Gaucher disease type 1	Subtype of disorder	1.0 *
746	Mitochondrial trifunctional protein deficiency	Disorder	1.0 *
710	Pfeiffer syndrome	Disorder	1.0 BP*
681	Hypokalemic periodic paralysis	Disorder	1.0 *
67043	Amoebic keratitis	Disorder	1.0 *
664	Ornithine transcarbamylase deficiency	Disorder	1.77 BP
647	Nijmegen breakage syndrome	Disorder	1.0 BP
646	Niemann-Pick disease type C	Disorder	1.0 *
616	Medulloblastoma	Disorder	1.0 *
614	Thomsen and Becker disease	Disorder	1.0

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
606	Proximal myotonic myopathy	Disorder	1.0 *
602	GNE myopathy	Disorder	1.0
579	Mucopolysaccharidosis type 1	Disorder	1.0 BP*
579	Mucopolysaccharidosis type 1	Disorder	0.82 BP
531	Miller-Dieker syndrome	Disorder	1.0 BP*
53	Albers-Schönberg osteopetrosis	Disorder	5.0 *
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Disorder	8.0 *
487	Krabbe disease	Disorder	1.0 *
487	Krabbe disease	Disorder	1.0 BP*
487	Krabbe disease	Disorder	0.7 BP
43393	Lambert-Eaton myasthenic syndrome	Disorder	1.0 *
422	Idiopathic/heritable pulmonary arterial hypertension	Disorder	1.0 *
396	Chronic hiccup	Disorder	1.0 *
364	Glycogen storage disease due to glucose-6-phosphatase deficiency	Disorder	1.0 BP
360	Glioblastoma	Disorder	1.0
355	Gaucher disease	Disorder	1.3 BP
3449	Weill-Marchesani syndrome	Disorder	1.0
3403	Uhl anomaly	Disorder	1.0 BP
331206	Severe combined immunodeficiency due to complete RAG1/2 deficiency	Disorder	1.0 *
33	Isovaleric acidemia	Disorder	1.0 *
296	Ollier disease	Disorder	1.0 *
2924	Isolated polycystic liver disease	Disorder	1.0 *
286	Vascular Ehlers-Danlos syndrome	Disorder	1.0
267	Calpain-3-related limb-girdle muscular dystrophy R1	Disorder	1.0 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
264580	Glycogen storage disease due to liver phosphorylase kinase deficiency	Disorder	1.0 BP*
2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2	Subtype of disorder	1.0 BP*
25	Glutaryl-CoA dehydrogenase deficiency	Disorder	1.0 BP
23	Argininosuccinic aciduria	Disorder	1.0 *
217	Isolated Dandy-Walker malformation	Disorder	2.1 *
2134	Atypical hemolytic uremic syndrome	Disorder	1.0 *
205	Crigler-Najjar syndrome	Disorder	0.1 BP*
1900	Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency	Subtype of disorder	1.0 BP
189	Hidrotic ectodermal dysplasia	Disorder	1.0 *
180242	Malignant tumor of fallopian tubes	Disorder	1.0 *
16	Blue cone monochromatism	Disorder	1.0 BP
16	Blue cone monochromatism	Disorder	1.0
157	Carnitine palmitoyltransferase II deficiency	Disorder	1.0 *
141	Canavan disease	Disorder	1.0 BP
3169	Sirenomelia	Disorder	0.98 BP
3169	Sirenomelia	Disorder	0.71 BP*
444490	Familial chylomicronemia syndrome	Disorder	0.97 *
79408	Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	Disorder	0.963 *
623626	Paraneoplastic cerebellar degeneration	Disorder	0.9553 *
79278	Autosomal erythropoietic protoporphyria	Disorder	0.92 *
882	Tyrosinemia type 1	Disorder	0.9 BP
48162	Lewis-Sumner syndrome	Subtype of disorder	0.9 *
207	Crouzon syndrome	Disorder	0.9 BP*
581	Mucopolysaccharidosis type 3	Disorder	0.87 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
99429	Complete androgen insensitivity syndrome	Disorder	0.83
579	Mucopolysaccharidosis type 1	Disorder	0.5 *
52	Alagille syndrome	Disorder	0.8 BP*
365	Glycogen storage disease due to acid maltase deficiency	Disorder	3.0 *
169793	Severe hemophilia B	Subtype of disorder	0.8 *
1461	Criss-cross heart	Disorder	0.8 BP*
3312	Thalidomide embryopathy	Disorder	0.77
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	Disorder	0.75 BP*
93929	Cloacal exstrophy	Subtype of disorder	0.75 BP*
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	Disorder	0.75 BP*
667	Autosomal recessive malignant osteopetrosis	Disorder	0.75 BP*
354	GM1 gangliosidosis	Disorder	0.75 BP*
213	Cystinosis	Disorder	0.5 BP*
181	X-linked hypohidrotic ectodermal dysplasia	Subtype of disorder	0.75 BP*
1501	Adrenocortical carcinoma	Disorder	0.75 *
3169	Sirenomelia	Disorder	0.01
93473	Hurler syndrome	Subtype of disorder	0.5 *
813	Silver-Russell syndrome	Disorder	0.7 BP*
783	Rubinstein-Taybi syndrome	Disorder	0.7 BP*
726	Alpers-Huttenlocher syndrome	Disorder	0.7 BP*
580	Mucopolysaccharidosis type 2	Disorder	0.7 BP*
580	Mucopolysaccharidosis type 2	Disorder	0.68 BP
392	Holt-Oram syndrome	Disorder	0.7 BP*
177	Rhizomelic chondrodysplasia punctata	Disorder	0.7 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
528	Congenital generalized lipodystrophy	Disorder	0.5 *
580	Mucopolysaccharidosis type 2	Disorder	0.2 *
796	Sandhoff disease	Disorder	0.67 BP*
511	Maple syrup urine disease	Disorder	0.67 BP
448270	Ectopia cordis	Disorder	0.67 BP
3282	Multifocal atrial tachycardia	Disorder	0.67 BP
2591	Infantile myofibromatosis	Disorder	0.67 BP*
1335	Pentalogy of Cantrell	Disorder	0.55 BP*
124	Diamond-Blackfan anemia	Disorder	0.67 BP*
90053	Complications after hematopoietic stem cell transplantation	Disorder	0.65 *
84	Fanconi anemia	Disorder	0.3 *
3463	Wolfram syndrome	Disorder	0.13
294975	Isolated absence of upper arm and forearm with hand present	Disorder	0.62 BP
994	Fetal akinesia deformation sequence	Disorder	0.6 BP*
98809	Paroxysmal kinesigenic dyskinesia	Disorder	0.6
79098	Sympathetic ophthalmia	Disorder	0.6 *
550	MELAS	Disorder	0.6 *
248111	Juvenile Huntington disease	Disorder	0.6 *
240103	Progressive supranuclear palsy-corticobasal syndrome	Subtype of disorder	0.6 *
2345	Isolated Klippel-Feil syndrome	Disorder	0.6 BP*
169799	Mild hemophilia B	Subtype of disorder	0.6 *
169796	Moderate hemophilia B	Subtype of disorder	0.6 *
54	X-linked recessive ocular albinism	Disorder	0.58 BP*
96264	49,XXXXY syndrome	Disorder	0.55 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
562	McCune-Albright syndrome	Disorder	0.55 *
1335	Pentalogy of Cantrell	Disorder	0.67 BP
93929	Cloacal exstrophy	Subtype of disorder	0.54 BP
79276	Acute intermittent porphyria	Disorder	0.54 *
71277	Classic glucose transporter type 1 deficiency syndrome	Disorder	0.538
93473	Hurler syndrome	Subtype of disorder	0.7 BP*
92050	Congenital tufting enteropathy	Disorder	0.5 BP*
915	Aarskog-Scott syndrome	Disorder	0.5 BP*
902	Werner syndrome	Disorder	0.5 *
86854	Splenic marginal zone lymphoma	Disorder	0.5 *
821	Sotos syndrome	Disorder	0.5 BP*
811	Shwachman-Diamond syndrome	Disorder	0.5 BP
79242	Holocarboxylase synthetase deficiency	Disorder	0.5 BP*
782	Axenfeld-Rieger syndrome	Disorder	0.5 *
682	Hyperkalemic periodic paralysis	Disorder	0.5 *
64742	Pleuropulmonary blastoma	Disorder	0.5 BP*
634	Netherton syndrome	Disorder	0.5 BP*
634	Netherton syndrome	Disorder	0.5 *
611	Inclusion body myositis	Disorder	0.5 *
528	Congenital generalized lipodystrophy	Disorder	0.6812
379	Chronic granulomatous disease	Disorder	0.46 BP
35909	Combined deficiency of factor V and factor VIII	Disorder	0.5 *
35858	Imerslund-Gräsbeck syndrome	Disorder	0.5 *
3427	Double outlet left ventricle	Disorder	0.5 BP

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3320	Thrombocytopenia-absent radius syndrome	Disorder	0.5 BP*
122	Birt-Hogg-Dubé syndrome	Disorder	0.5 *
1215	Autosomal dominant optic atrophy plus syndrome	Disorder	0.5 *
110	Bardet-Biedl syndrome	Disorder	0.5 BP*
100	Ataxia-telangiectasia	Disorder	0.49 *
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	Disorder	0.47 *
379	Chronic granulomatous disease	Disorder	0.5 BP*
582	Mucopolysaccharidosis type 4	Disorder	27.6 *
676	Hereditary chronic pancreatitis	Disorder	0.43 *
88	Idiopathic aplastic anemia	Disorder	0.4 *
77293	Chronic visceral acid sphingomyelinase deficiency	Disorder	0.4 BP*
503	Larsen syndrome	Disorder	0.4 BP*
3008	Pyruvate carboxylase deficiency	Disorder	0.4 BP*
289	Ellis Van Creveld syndrome	Disorder	1.1 BP
2869	Peutz-Jeghers syndrome	Disorder	0.4 *
256	Early-onset generalized limb-onset dystonia	Disorder	0.4 *
2315	Johanson-Blizzard syndrome	Disorder	0.4 BP*
217085	Mucopolysaccharidosis type 2, severe form	Subtype of disorder	0.4 BP*
1452	Cleidocranial dysplasia	Disorder	0.1
99885	Isolated permanent neonatal diabetes mellitus	Disorder	0.38 BP*
3440	Waardenburg syndrome	Disorder	0.37 BP*
43393	Lambert-Eaton myasthenic syndrome	Disorder	0.35
290	Congenital rubella syndrome	Disorder	0.35 BP*
179	Birdshot chorioretinopathy	Disorder	0.35

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
576	Mucolipidosis type II	Disorder	0.34 BP*
510	Lesch-Nyhan syndrome	Disorder	0.34 BP*
96	Ataxia with vitamin E deficiency	Disorder	0.33 *
565	Menkes disease	Disorder	0.33 BP*
327	Congenital factor VII deficiency	Disorder	0.33 *
79473	Variegate porphyria	Disorder	0.32 *
79269	Sanfilippo syndrome type A	Subtype of disorder	1.4 BP
391665	Homozygous familial hypercholesterolemia	Disorder	0.3194
845	Tay-Sachs disease	Disorder	0.31 BP*
99886	Transient neonatal diabetes mellitus	Disorder	0.3 BP*
90647	Jervell and Lange-Nielsen syndrome	Disorder	0.3
84	Fanconi anemia	Disorder	0.62 BP*
79394	Congenital ichthyosiform erythroderma	Disorder	0.3 *
633	Laron syndrome	Disorder	0.3 *
628	Diastrophic dysplasia	Disorder	0.3 BP*
590	Congenital myasthenic syndrome	Disorder	0.3 *
581	Mucopolysaccharidosis type 3	Disorder	0.3 *
394	Homocystinuria due to cystathionine beta-synthase deficiency	Disorder	1.65 *
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	Disorder	0.3
3004	Mirror polydactyly-vertebral segmentation-limbs defects syndrome	Disorder	0.3 *
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	Disorder	0.3 BP*
261	Emery-Dreifuss muscular dystrophy	Disorder	0.3 *
258	Laminin subunit alpha 2-related congenital muscular dystrophy	Disorder	0.3 *
2299	Aortic arch interruption	Disorder	0.3 BP*

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
229717	Isolated agammaglobulinemia	Disorder	0.3
219	Delta-sarcoglycan-related limb-girdle muscular dystrophy R6	Disorder	0.3 *
182050	MYH9-related disease	Disorder	0.3 *
845	Tay-Sachs disease	Disorder	0.28 BP
811	Shwachman-Diamond syndrome	Disorder	0.28
93571	Dense deposit disease	Subtype of disorder	0.25
77292	Infantile neurovisceral acid sphingomyelinase deficiency	Disorder	0.25 BP*
702	Pelizaeus-Merzbacher disease	Disorder	0.25 *
678	Papillon-Lefèvre syndrome	Disorder	0.25
538	Lymphangioleiomyomatosis	Disorder	0.15
35173	X-linked dominant chondrodysplasia punctata	Disorder	0.25 BP*
910	Xeroderma pigmentosum	Disorder	0.23 BP*
271861	Hereditary ATTR amyloidosis	Disorder	0.2222
47	X-linked agammaglobulinemia	Subtype of disorder	0.1 *
111	Barth syndrome	Disorder	0.22 *
98813	Hypohidrotic ectodermal dysplasia with immunodeficiency	Disorder	0.2 BP*
893	WAGR syndrome	Disorder	0.2 BP
808	Seckel syndrome	Disorder	0.2 BP*
79270	Sanfilippo syndrome type B	Subtype of disorder	0.2 *
596	X-linked centronuclear myopathy	Disorder	0.2 *
585	Multiple sulfatase deficiency	Disorder	0.2
534	Oculocerebrorenal syndrome of Lowe	Disorder	0.2
534	Oculocerebrorenal syndrome of Lowe	Disorder	0.2 *
375	Anti-glomerular basement membrane disease	Disorder	0.2 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
353	Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5	Disorder	0.2 *
35	Propionic acidemia	Disorder	0.2 *
3006	Pyridoxine-dependent epilepsy	Disorder	0.2 BP*
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	Disorder	0.2 *
2635	Metatropic dysplasia	Disorder	0.2 BP*
2614	Nail-patella syndrome	Disorder	0.2 BP*
238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	Disorder	0.2
235	Dubowitz syndrome	Disorder	0.2 BP*
209916	Extraskeletal myxoid chondrosarcoma	Disorder	0.2 *
2052	Fraser syndrome	Disorder	0.2 BP*
191	Cockayne syndrome	Disorder	0.2 BP*
178478	Infant botulism	Subtype of disorder	0.2 BP*
1361	Carnosinase deficiency	Disorder	0.2 BP
166096	Von Willebrand disease type 3	Subtype of disorder	0.1865
52427	Retinitis punctata albescens	Disorder	0.125
79404	Severe generalized junctional epidermolysis bullosa	Disorder	0.17 BP
407	Glycine encephalopathy	Disorder	0.17 *
280219	Pelizaeus-Merzbacher disease, classic form	Subtype of disorder	0.17 *
1456	Middle aortic syndrome	Disorder	0.17 BP*
745	Severe hereditary thrombophilia due to congenital protein C deficiency	Disorder	0.16 BP
722	Hypoplasminogenemia	Disorder	0.16 *
583	Mucopolysaccharidosis type 6	Disorder	0.16 BP*
583	Mucopolysaccharidosis type 6	Disorder	0.16 *
79430	Hermansky-Pudlak syndrome	Disorder	0.15

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
538	Lymphangioleiomyomatosis	Disorder	0.25 *
335	Congenital fibrinogen deficiency	Disorder	0.15 *
223	Arginine vasopressin resistance	Disorder	0.15 *
169154	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	Disorder	0.15 BP
157850	Pantothenate kinase-associated neurodegeneration	Disorder	0.15 *
118	Beta-mannosidosis	Disorder	0.14 BP*
763	Pycnodysostosis	Disorder	0.13
3463	Wolfram syndrome	Disorder	0.62 *
52427	Retinitis punctata albescens	Disorder	0.175 *
33364	Trichothiodystrophy	Disorder	0.12 BP*
623789	Body integrity dysphoria	Disorder	0.11
1308	C syndrome	Disorder	0.11 *
620102	Non-syndromic unicoronal craniosynostosis	Disorder	0.1049
99842	Leukocyte adhesion deficiency type I	Subtype of disorder	0.1 *
98810	Paroxysmal non-kinesigenic dyskinesia	Disorder	0.1
93322	Isolated tibial hemimelia	Disorder	0.1 BP*
93322	Isolated tibial hemimelia	Disorder	0.1 *
93262	Crouzon syndrome-acanthosis nigricans syndrome	Disorder	0.1 BP
90793	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	Disorder	0.1 *
906	Wiskott-Aldrich syndrome	Disorder	0.1 *
86834	Juvenile myelomonocytic leukemia	Disorder	0.1 *
773	Refsum disease	Disorder	0.1 *
61	Alpha-mannosidosis	Disorder	0.1 *
512	Metachromatic leukodystrophy	Disorder	0.1 *

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507	Leishmaniasis	Disorder	0.1 *
47	X-linked agammaglobulinemia	Subtype of disorder	0.22
367	Glycogen storage disease due to glycogen branching enzyme deficiency	Disorder	0.1 BP
3329	Tibial aplasia-ectrodactyly syndrome	Disorder	0.1 *
32960	Tumor necrosis factor receptor 1 associated periodic syndrome	Disorder	0.1 *
329	Congenital factor XI deficiency	Disorder	0.1 *
326	Congenital factor V deficiency	Disorder	0.1 *
31824	Colchicine poisoning	Disorder	0.1 *
298	Mitochondrial neurogastrointestinal encephalomyopathy	Disorder	0.1 *
289560	Mitochondrial membrane protein-associated neurodegeneration	Disorder	0.1
2686	Cyclic neutropenia	Disorder	0.1 *
209335	Autosomal dominant adult-onset proximal spinal muscular atrophy	Disorder	0.1 *
205	Crigler-Najjar syndrome	Disorder	1.0 *
1959	Evans syndrome	Disorder	0.1 *
1775	Dyskeratosis congenita	Disorder	0.1 *
1452	Cleidocranial dysplasia	Disorder	0.4 BP*
142	Anaplastic thyroid carcinoma	Disorder	0.1 *
119	Beta-sarcoglycan-related limb-girdle muscular dystrophy R4	Disorder	0.1 *
48818	Aceruloplasminemia	Disorder	0.09
2485	Melorheostosis	Disorder	0.09 *
204	Sporadic Creutzfeldt-Jakob disease	Disorder	0.088
275777	Heritable pulmonary arterial hypertension	Subtype of disorder	0.08 *
189427	Cushing syndrome due to bilateral macronodular adrenocortical disease	Disorder	0.08 *
337	Fibrodysplasia ossificans progressiva	Disorder	0.05

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726	Alpers-Huttenlocher syndrome	Disorder	0.07 *
217563	Neonatal acute respiratory distress due to SP-B deficiency	Disorder	0.067 BP
77261	Gaucher disease type 3	Subtype of disorder	0.05 *
633124	Invasive scopulariopsis infection	Disorder	0.05 *
337	Fibrodysplasia ossificans progressiva	Disorder	0.078 *
331	Congenital factor XIII deficiency	Disorder	0.05 *
325	Congenital factor II deficiency	Disorder	0.05 *
2788	Osteoporosis-pseudoglioma syndrome	Disorder	0.05 *
620113	Non-syndromic unilambdoid craniosynostosis	Disorder	0.0442
99718	Leber plus disease	Disorder	0.04 *
69087	Naegeli-Franceschetti-Jadassohn syndrome	Disorder	0.035 *
34520	Congenital muscular dystrophy with integrin alpha-7 deficiency	Disorder	0.03 *
280224	Pelizaeus-Merzbacher disease, transitional form	Subtype of disorder	0.03 *
280210	Pelizaeus-Merzbacher disease, connatal form	Subtype of disorder	0.03 *
93976	Anotia	Disorder	0.028 BP*
740	Hutchinson-Gilford progeria syndrome	Disorder	0.025 BP
227	Diphallia	Disorder	0.02 BP
620139	Non-syndromic unifrontosphenoidal craniosynostosis	Disorder	0.0136
77260	Gaucher disease type 2	Subtype of disorder	0.01 *
584	Mucopolysaccharidosis type 7	Disorder	0.01 *
3169	Sirenomelia	Disorder	0.009 *
90308	Klippel-Trénaunay syndrome	Disorder	0.007 *
740	Hutchinson-Gilford progeria syndrome	Disorder	0.005
141077	Epignathus	Subtype of disorder	1.68 BP

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated prevalence (/100,000)
140	Campomelic dysplasia	Disorder	1.875 BP

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List of diseases by decreasing incidence

ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated incidence (/100,000)
99828	Dengue fever	Disorder	714.0
91546	Lyme disease	Disorder	177.5 *
673	Malaria	Disorder	73.0
558	Marfan syndrome	Disorder	25.0 *
507	Leishmaniasis	Disorder	25.0
178320	Acute lung injury	Disorder	25.0 *
91546	Lyme disease	Disorder	21.9
813	Silver-Russell syndrome	Disorder	15.5 *
461	Recessive X-linked ichthyosis	Disorder	15.0 *
268316	Complication in hemodialysis	Disorder	13.0 *
641350	Immunotherapy induced hypophysitis	Disorder	12.8074 *
1546	Cryptococcosis	Disorder	11.0 *
848	Beta-thalassemia	Disorder	1.0
2209	Maternal phenylketonuria	Disorder	10.0 *
137839	Lemierre syndrome	Disorder	10.0 *
3467	Hereditary xanthinuria	Disorder	9.05 *
1489	Whooping cough	Disorder	8.9 *
1941	Juvenile absence epilepsy	Disorder	7.5 *
289390	Primary Sjögren syndrome	Disorder	6.92
3002	Immune thrombocytopenia	Disorder	6.75 *
29073	Multiple myeloma	Disorder	6.0
213504	Adenocarcinoma of ovary	Disorder	5.97 *
2032	Idiopathic pulmonary fibrosis	Disorder	5.55
146	Differentiated thyroid carcinoma	Disorder	5.25
99977	Squamous cell carcinoma of the esophagus	Disorder	3.357 *
536	Systemic lupus erythematosus	Disorder	5.14
3099	Rheumatic fever	Disorder	5.0 *
139417	Acute transverse myelitis	Disorder	4.72
494550	Squamous cell carcinoma of the larynx	Disorder	4.61 *
89936	X-linked hypophosphatemia	Disorder	4.5
213767	Squamous cell carcinoma of the cervix uteri	Disorder	4.28 *
70591	Chronic thromboembolic pulmonary hypertension	Disorder	4.2 *
70567	Cholangiocarcinoma	Disorder	4.2
70567	Cholangiocarcinoma	Disorder	4.0 *
585867	Acute myeloid leukemia with t(9;22)(q34.1;q11.2)	Disorder	4.0
2929	Juvenile polyposis syndrome	Disorder	3.85 *
548	Leprosy	Disorder	3.7
213528	Rare adenocarcinoma of the breast	Disorder	3.55 *
502363	Squamous cell carcinoma of the oral cavity	Disorder	3.51 *
99977	Squamous cell carcinoma of the esophagus	Disorder	5.2
99976	Adenocarcinoma of the esophagus	Disorder	3.264 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated incidence (/100,000)
210159	Adult hepatocellular carcinoma	Disorder	3.22 *
500478	Squamous cell carcinoma of the oropharynx	Disorder	3.12 *
99745	Typhoid	Disorder	3.0 *
99429	Complete androgen insensitivity syndrome	Disorder	3.0 *
360	Glioblastoma	Disorder	3.0
186	Primary biliary cholangitis	Disorder	2.57 *
154	Familial isolated dilated cardiomyopathy	Disorder	2.91 *
454821	Pleomorphic salivary gland adenoma	Subtype of disorder	2.725
95716	Familial thyroid dyshormonogenesis	Disorder	2.67
424991	Adenocarcinoma of the gallbladder and extrahepatic biliary tract	Disorder	2.62 *
70	Proximal spinal muscular atrophy	Disorder	2.6 *
186	Primary biliary cholangitis	Disorder	3.0
91349	Non-functioning pituitary adenoma	Disorder	1.05
360	Glioblastoma	Disorder	2.52 *
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	Disorder	2.5
2038	Pulmonary arteriovenous malformation	Disorder	2.5
391	Classic Hodgkin lymphoma	Disorder	2.463 *
29073	Multiple myeloma	Disorder	2.4 *
803	Amyotrophic lateral sclerosis	Disorder	2.2 *
707	Plague	Disorder	2.2 *
545	Follicular lymphoma	Disorder	2.192 *
79239	Classic galactosemia	Disorder	2.1 *
166119	Isolated osteopoikilosis	Disorder	2.0
146	Differentiated thyroid carcinoma	Disorder	2.0 *
319276	Clear cell renal carcinoma	Disorder	1.99 *
729	Polycythemia vera	Disorder	1.9 *
50251	Pleural mesothelioma	Disorder	1.9 *
102	Multiple system atrophy	Disorder	1.8
854	Non-malignant and non-cirrhotic portal vein thrombosis	Disorder	1.72 *
842	Testicular seminomatous germ cell tumor	Disorder	1.71 *
589	Myasthenia gravis	Disorder	1.7 *
355	Gaucher disease	Disorder	1.7 *
810	Shigellosis	Disorder	1.68 *
583861	Isolated mesenteric vein thrombosis	Disorder	1.6 *
618	Familial melanoma	Disorder	1.5 *
598216	Upper tract urothelial carcinoma	Disorder	1.5
35	Propionic acidemia	Disorder	1.5
26106	Hereditary diffuse gastric cancer	Disorder	1.5 *
549	Legionnaires disease	Disorder	1.4 *
803	Amyotrophic lateral sclerosis	Disorder	1.35
250923	Isolated aniridia	Disorder	1.31 *
98843	Classic Hodgkin lymphoma, nodular sclerosis type	Subtype of disorder	1.28 *
494547	Squamous cell carcinoma of the hypopharynx	Disorder	1.27 *
635	Neuroblastoma	Disorder	1.26
521	Chronic myeloid leukemia	Disorder	1.25 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated incidence (/100,000)
363494	Non-seminomatous germ cell tumor of testis	Disorder	1.21 *
673	Malaria	Disorder	1.2 *
313920	Epstein-Barr virus-associated gastric carcinoma	Disorder	1.2
2137	Autoimmune hepatitis	Disorder	1.2
85443	AL amyloidosis	Disorder	1.044
91349	Non-functioning pituitary adenoma	Disorder	2.55 *
85443	AL amyloidosis	Disorder	1.1177 *
502366	Squamous cell carcinoma of the lip	Disorder	1.02
213772	Adenocarcinoma of the cervix uteri	Disorder	1.01 *
848	Beta-thalassemia	Disorder	10.0 *
824	Primary myelofibrosis	Disorder	1.0 *
727	Microscopic polyangiitis	Disorder	1.0 *
69078	Liposarcoma	Disorder	1.0 *
54595	Craniopharyngioma	Disorder	1.0
44890	Gastrointestinal stromal tumor	Disorder	1.0 *
44890	Gastrointestinal stromal tumor	Disorder	1.0
400	Cystic echinococcosis	Disorder	1.0 *
3148	Malignant peripheral nerve sheath tumor	Disorder	1.0
209964	Solitary rectal ulcer syndrome	Disorder	1.0 *
157798	Serrated polyposis syndrome	Disorder	1.0
577	Mucolipidosis type III	Disorder	0.985 *
221	Dermatomyositis	Disorder	0.9704
97560	Primary membranous glomerulonephritis	Disorder	0.8103
2023	Undifferentiated pleomorphic sarcoma	Disorder	0.9 *
900	Granulomatosis with polyangiitis	Disorder	0.85 *
398961	Mucinous adenocarcinoma of ovary	Disorder	0.85 *
97560	Primary membranous glomerulonephritis	Disorder	0.9194 *
454723	Endometrioid carcinoma of ovary	Disorder	0.81 *
424019	Squamous cell carcinoma of the anal canal	Disorder	0.81 *
33226	Waldenström macroglobulinemia	Disorder	0.81 *
930	Idiopathic achalasia	Disorder	0.77
171	Primary sclerosing cholangitis	Disorder	0.77 *
2137	Autoimmune hepatitis	Disorder	0.75 *
48104	Pyoderma gangrenosum	Disorder	0.74
340	Hemorrhagic fever-renal syndrome	Disorder	0.74 *
276145	Malignant epithelial tumor of salivary glands	Disorder	0.73 *
99976	Adenocarcinoma of the esophagus	Disorder	0.7
100070	Progressive non-fluent aphasia	Disorder	0.7 *
98823	Chronic myelomonocytic leukemia	Disorder	0.68
289596	Juvenile nasopharyngeal angiofibroma	Disorder	0.6666
79139	Japanese encephalitis	Disorder	0.65 *
683	Progressive supranuclear palsy	Disorder	0.65
171	Primary sclerosing cholangitis	Disorder	0.65
85414	Systemic-onset juvenile idiopathic arthritis	Disorder	0.6
83597	Acute disseminated encephalomyelitis	Disorder	0.6 *
101330	Porphyria cutanea tarda	Disorder	0.6 *

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780	Rhabdomyosarcoma	Disorder	0.59 *
104075	Adenocarcinoma of the small intestine	Disorder	0.588 *
732	Polymyositis	Disorder	0.585 *
398058	Squamous cell carcinoma of the penis	Disorder	0.57 *
86830	Chronic myeloproliferative disease, unclassifiable	Disorder	0.53 *
589	Myasthenia gravis	Disorder	0.53
99971	Well-differentiated liposarcoma	Subtype of disorder	0.51 *
180275	Paget disease of the nipple	Disorder	0.51 *
280921	Idiopathic panuveitis	Disorder	0.5051 *
99828	Dengue fever	Disorder	0.5 *
980	Absence of the pulmonary artery	Disorder	0.5 *
39044	Uveal melanoma	Disorder	0.5 *
2584	Classic mycosis fungoides	Disorder	0.5 *
191	Cockayne syndrome	Disorder	0.5 *
3318	Essential thrombocythemia	Disorder	0.48 *
963	Acromegaly	Disorder	0.47
533	Listeriosis	Disorder	0.43 *
98844	Classic Hodgkin lymphoma, mixed cellularity type	Subtype of disorder	0.42 *
424943	Adenocarcinoma of the liver and intrahepatic biliary tract	Disorder	0.412 *
86872	T-cell large granular lymphocyte leukemia	Disorder	0.4 *
820	Sneddon syndrome	Disorder	0.4 *
570422	Galactose mutarotase deficiency	Disorder	0.4
83484	St. Louis encephalitis	Disorder	0.38 *
399	Huntington disease	Disorder	0.38
36426	Stevens-Johnson syndrome	Subtype of disorder	0.36 *
150	Nasopharyngeal carcinoma	Disorder	0.36 *
728	Relapsing polychondritis	Disorder	0.35
54057	Thrombotic thrombocytopenic purpura	Disorder	0.35
500464	Squamous cell carcinoma of the nasal cavity and paranasal sinuses	Disorder	0.35
49041	IgG4-related retroperitoneal fibrosis	Subtype of disorder	0.35 *
33276	Kaposi sarcoma	Disorder	0.34 *
533	Listeriosis	Disorder	0.337
398971	Clear cell adenocarcinoma of the ovary	Disorder	0.32 *
1070	Anisakiasis	Disorder	0.32
873	Desmoid tumor	Disorder	0.3 *
52417	MALT lymphoma	Disorder	0.3 *
293173	Acute generalized exanthematous pustulosis	Disorder	0.3
29072	Hereditary pheochromocytoma-paraganglioma	Disorder	0.3
1930	Herpes simplex virus encephalitis	Disorder	0.3
178478	Infant botulism	Subtype of disorder	0.3 *
93672	Juvenile dermatomyositis	Disorder	0.295
58017	Classic hairy cell leukemia	Disorder	0.29 *
99970	Dedifferentiated liposarcoma	Subtype of disorder	0.27 *
79140	Cutaneous neuroendocrine carcinoma	Disorder	0.27
83330	Proximal spinal muscular atrophy type 1	Subtype of disorder	0.26 *
168999	Malignant melanoma of the mucosa	Disorder	0.26 *

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567548	Idiopathic steroid-resistant nephrotic syndrome	Disorder	0.2582
424016	Adenocarcinoma of the anal canal	Disorder	0.253 *
97279	Insulinoma	Disorder	0.25
623615	Autoimmune limbic encephalitis	Disorder	0.25 *
329977	Classic neuroendocrine tumor of appendix	Subtype of disorder	0.25
251627	Oligodendroglioma	Disorder	0.25 *
139423	Idiopathic acute transverse myelitis	Subtype of disorder	0.25 *
55880	Chondrosarcoma	Disorder	0.24 *
668	Osteosarcoma	Disorder	0.23 *
623626	Paraneoplastic cerebellar degeneration	Disorder	0.2225 *
1332	Medullary thyroid carcinoma	Disorder	0.22 *
423786	Undifferentiated carcinoma of stomach	Disorder	0.211 *
96253	Cushing disease	Disorder	0.2 *
3392	Tularemia	Disorder	0.2 *
100085	Primary hepatic neuroendocrine carcinoma	Disorder	0.2
95455	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	Disorder	0.19
71211	Neuromyelitis optica spectrum disorder	Disorder	0.1877
183	Eosinophilic granulomatosis with polyangiitis	Disorder	0.18 *
543	Burkitt lymphoma	Disorder	0.17 *
517	Acute myelomonocytic leukemia	Disorder	0.17 *
142	Anaplastic thyroid carcinoma	Disorder	0.17 *
781	Q fever	Disorder	0.16 *
284	Alveolar echinococcosis	Disorder	0.16 *
251636	Ependymoma	Disorder	0.16 *
599480	Acquired hemophilia A	Disorder	0.1505 *
913	Zollinger-Ellison syndrome	Disorder	0.15 *
86839	Myelodysplastic neoplasm with increased blasts	Disorder	0.15 *
33402	Pediatric hepatocellular carcinoma	Disorder	0.15 *
329918	C3 glomerulopathy	Subtype of disorder	0.15 *
363976	Giant cell tumor of bone	Disorder	0.1404
99867	Thymoma	Disorder	0.14 *
654	Nephroblastoma	Disorder	0.14 *
319298	Papillary renal cell carcinoma	Disorder	0.14 *
79140	Cutaneous neuroendocrine carcinoma	Disorder	0.13 *
514	Acute monoblastic/monocytic leukemia	Disorder	0.13 *
319	Skeletal Ewing sarcoma	Disorder	0.13 *
913	Zollinger-Ellison syndrome	Disorder	0.125
99915	Malignant granulosa cell tumor of the ovary	Disorder	0.12 *
86893	Nodular lymphocyte predominant Hodgkin lymphoma	Disorder	0.12
509	Leptospirosis	Disorder	0.12 *
324625	Chikungunya	Disorder	0.12 *
213716	Squamous cell carcinoma of the corpus uteri	Disorder	0.12 *
213512	Malignant mixed Müllerian tumor of the ovary	Disorder	0.12 *
2086	Optic pathway glioma	Disorder	0.12
204	Sporadic Creutzfeldt-Jakob disease	Disorder	0.118
418959	Squamous cell carcinoma of the stomach	Disorder	0.115 *

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424002	Squamous cell carcinoma of the rectum	Disorder	0.113 *
616	Medulloblastoma	Disorder	0.11 *
520	Acute promyelocytic leukemia	Disorder	0.11 *
99967	Myxoid/round cell liposarcoma	Subtype of disorder	0.1 *
98919	Miller Fisher syndrome	Disorder	0.1 *
98845	Classic Hodgkin lymphoma, lymphocyte-rich type	Subtype of disorder	0.1 *
53035	Caroli disease	Disorder	0.1
37553	Andersen-Tawil syndrome	Disorder	0.1 *
26790	Pseudomyxoma peritonei	Disorder	0.1
2382	Lennox-Gastaut syndrome	Disorder	0.1 *
2314	Autosomal dominant hyper-IgE syndrome due to STAT3 deficiency	Disorder	0.1 *
228371	Foodborne botulism	Subtype of disorder	0.1 *
1822	Dysplasia epiphysealis hemimelica	Disorder	0.1
178475	Wound botulism	Subtype of disorder	0.1 *
131	Budd-Chiari syndrome	Disorder	0.1
112	Bartter syndrome	Disorder	0.1 *
86893	Nodular lymphocyte predominant Hodgkin lymphoma	Disorder	0.095 *
75564	Acquired idiopathic sideroblastic anemia	Disorder	0.09 *
251630	Anaplastic oligodendroglioma	Disorder	0.09 *
1304	Brucellosis	Disorder	0.09 *
3287	Takayasu arteritis	Disorder	0.084 *
375	Anti-glomerular basement membrane disease	Disorder	0.08 *
398987	Malignant teratoma of ovary	Disorder	0.07 *
79277	Congenital erythropoietic porphyria	Disorder	0.065 *
86843	Acute panmyelosis with myelofibrosis	Disorder	0.06 *
863	Trichinellosis	Disorder	0.06 *
213531	Metaplastic carcinoma of the breast	Disorder	0.06 *
99969	Pleomorphic liposarcoma	Subtype of disorder	0.05 *
86852	B-cell prolymphocytic leukemia	Disorder	0.05 *
790	Retinoblastoma	Disorder	0.05 *
213557	Salivary gland type cancer of the breast	Disorder	0.05 *
418951	Undifferentiated carcinoma of esophagus	Disorder	0.044 *
99931	Idiopathic pulmonary hemosiderosis	Disorder	0.0425 *
99912	Ovarian dysgerminoma	Disorder	0.04 *
98846	Classic Hodgkin lymphoma, lymphocyte-depleted type	Subtype of disorder	0.04 *
454714	Plasma cell leukemia	Disorder	0.04 *
331	Congenital factor XIII deficiency	Disorder	0.04 *
300385	Pituitary carcinoma	Disorder	0.04 *
248111	Juvenile Huntington disease	Disorder	0.04 *
168960	Refractory anemia with excess blasts in transformation	Disorder	0.04 *
357034	Non-hereditary retinoblastoma	Subtype of disorder	0.038 *
83476	West-Nile encephalitis	Disorder	0.036 *
2573	Moyamoya disease	Disorder	0.035 *
424996	Squamous cell carcinoma of gallbladder and extrahepatic biliary tract	Disorder	0.032 *
99865	Spermatocytic seminoma	Disorder	0.03 *

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ORPHAcode	Disease or Subtype of disease	Classification Level	Estimated incidence (/100,000)
46487	Epidermolysis bullosa acquisita	Disorder	0.03 *
33355	Reticular dysgenesis	Disorder	0.03 *
290	Congenital rubella syndrome	Disorder	0.03 *
251576	Gliosarcoma	Subtype of disorder	0.03 *
1501	Adrenocortical carcinoma	Disorder	0.03 *
424046	Acinar cell carcinoma of pancreas	Disorder	0.029 *
423994	Squamous cell carcinoma of the colon	Disorder	0.026 *
401920	Fibrolamellar hepatocellular carcinoma	Disorder	0.025 *
329984	Goblet cell carcinoma	Subtype of disorder	0.025
3299	Tetanus	Disorder	0.024 *
424039	Squamous cell carcinoma of pancreas	Disorder	0.023 *
1267	Botulism	Disorder	0.022 *
99928	Placental site trophoblastic tumor	Disorder	0.02 *
98834	Acute myeloblastic leukemia with maturation	Disorder	0.02 *
86850	Myeloid sarcoma	Disorder	0.02 *
518	Acute megakaryoblastic leukemia	Disorder	0.02 *
449	Hepatoblastoma	Disorder	0.02 *
363489	Sex cord-stromal tumor of testis	Disorder	0.02 *
251909	Pineoblastoma	Disorder	0.02 *
251679	Astroblastoma	Disorder	0.02 *
251579	Giant cell glioblastoma	Subtype of disorder	0.02 *
1957	Esthesioneuroblastoma	Disorder	0.02 *
143	Parathyroid carcinoma	Disorder	0.02 *
1183	Opsoclonus-myoclonus syndrome	Disorder	0.02 *
424970	Undifferentiated carcinoma of liver and intrahepatic biliary tract	Disorder	0.015 *
31837	Pulmonary venoocclusive disease	Disorder	0.015 *
538	Lymphangioleiomyomatosis	Disorder	0.0135
79276	Acute intermittent porphyria	Disorder	0.013 *
79278	Autosomal erythropoietic protoporphyria	Disorder	0.012 *
424058	Intraductal papillary mucinous carcinoma of pancreas	Disorder	0.011 *
98833	Acute myeloblastic leukemia without maturation	Disorder	0.01 *
98832	Acute myeloid leukemia with minimal differentiation	Disorder	0.01 *
55881	Adamantinoma	Disorder	0.01 *
424053	Mucinous cystadenocarcinoma of the pancreas	Disorder	0.01 *
319303	Chromophobe renal cell carcinoma	Disorder	0.01 *
251899	Choroid plexus carcinoma	Disorder	0.01 *
251863	Desmoplastic/nodular medulloblastoma	Subtype of disorder	0.01 *
251607	Pleomorphic xanthoastrocytoma	Disorder	0.01 *
251598	Protoplasmic astrocytoma	Subtype of disorder	0.01 *
251582	Gliomatosis cerebri	Disorder	0.01 *
2030	Fibrosarcoma	Disorder	0.01 *
180234	Mixed germ cell tumor	Disorder	0.01 *
180226	Embryonal carcinoma	Disorder	0.01 *
168966	Composite lymphoma	Disorder	0.01 *
424975	Squamous cell carcinoma of liver and intrahepatic biliary tract	Disorder	0.009 *
79473	Variegate porphyria	Disorder	0.008 *

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423968	Squamous cell carcinoma of the small intestine	Disorder	0.008 *
284343	DICER1 tumor-predisposition syndrome	Disorder	0.007
356	Gerstmann-Straussler-Scheinker syndrome	Disorder	0.0055
97280	Glucagonoma	Disorder	0.005 *
418945	Carcinoma of esophagus, salivary gland type	Disorder	0.004 *
424065	Solid pseudopapillary carcinoma of pancreas	Disorder	0.003 *
97283	Somatostatinoma	Disorder	0.0025 *
424982	Biliary cystadenocarcinoma	Disorder	0.002 *
424080	Undifferentiated carcinoma with osteoclast-like giant cells of pancreas	Disorder	0.001 *

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List of diseases by decreasing number of published cases or families

Number of published cases

ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
319218	Ebola hemorrhagic fever	Disorder	28220 Case(s)
227972	Toxic oil syndrome	Disorder	20000 Case(s)
454745	Kuru	Disorder	2700 Case(s)
50918	Kikuchi-Fujimoto disease	Disorder	1052 Case(s)
2309	Pachyonychia congenita	Disorder	1000 Case(s)
158014	Rosaï-Dorfman disease	Disorder	1000 Case(s)
64	Alström syndrome	Disorder	950 Case(s)
454836	Avian influenza	Disorder	826 Case(s)
83312	Rickettsialpox	Disorder	800 Case(s)
1359	Carney complex	Disorder	750 Case(s)
840	Syringocystadenoma papilliferum	Disorder	730 Case(s)
71276	Silent sinus syndrome	Disorder	558 Case(s)
99825	Nipah virus disease	Disorder	556 Case(s)
99826	Marburg hemorrhagic fever	Disorder	500 Case(s)
79282	Methylmalonic acidemia with homocystinuria, type cblC	Subtype of disorder	500 Case(s)
73256	Central neurocytoma	Disorder	500 Case(s)
69077	Rhabdoid tumor	Disorder	500 Case(s)
530	Lipoid proteinosis	Disorder	500 Case(s)
42642	PFAPA syndrome	Disorder	500 Case(s)
35687	Erdheim-Chester disease	Disorder	500 Case(s)
3261	Autoimmune lymphoproliferative syndrome	Disorder	500 Case(s)
2930	Cronkhite-Canada syndrome	Disorder	500 Case(s)
26	Methylmalonic acidemia with homocystinuria	Disorder	500 Case(s)
2138	46,XX ovotesticular difference of sex development	Disorder	500 Case(s)
167	Chédiak-Higashi syndrome	Disorder	500 Case(s)
85448	AGel amyloidosis	Disorder	475 Case(s)
79312	Vitamin B12-unresponsive methylmalonic acidemia type mut-	Subtype of disorder	450 Case(s)
22	Succinic semialdehyde dehydrogenase deficiency	Disorder	450 Case(s)
79501	Punctate palmoplantar keratoderma type 1	Disorder	437 Case(s)
411593	Insulin autoimmune syndrome	Disorder	404 Case(s)
662	Yellow nail syndrome	Disorder	400 Case(s)
649	Norrie disease	Disorder	400 Case(s)
352540	Oncogenic osteomalacia	Disorder	400 Case(s)
35125	Epidermal nevus syndrome	Disorder	400 Case(s)
3348	Tracheobronchopathia osteochondroplastica	Disorder	400 Case(s)
2909	Rothmund-Thomson syndrome	Disorder	400 Case(s)
100025	Alpha-heavy chain disease	Subtype of disorder	400 Case(s)
974	Adams-Oliver syndrome	Disorder	398 Case(s)
238606	Primary orthostatic tremor	Disorder	390 Case(s)
83453	Vulvovaginal gingival syndrome	Disorder	380 Case(s)
96170	Emanuel syndrome	Disorder	350 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
85458	Cerebral Amyloid Angiopathy	Disorder	350 Case(s)
64741	Pulmonary blastoma	Disorder	350 Case(s)
3269	Isolated radio-ulnar synostosis	Disorder	350 Case(s)
2968	Leukocyte adhesion deficiency	Disorder	350 Case(s)
59	Allan-Herndon-Dudley syndrome	Disorder	320 Case(s)
838	Susac syndrome	Disorder	304 Case(s)
99147	Acquired von Willebrand syndrome	Disorder	300 Case(s)
83469	Desmoplastic small round cell tumor	Disorder	300 Case(s)
73	Gorham-Stout disease	Disorder	300 Case(s)
570	Moebius syndrome	Disorder	300 Case(s)
525	Lichen planopilaris	Disorder	300 Case(s)
51608	Generalized arterial calcification of infancy	Disorder	300 Case(s)
501	Lafora disease	Disorder	300 Case(s)
482	Kimura disease	Disorder	300 Case(s)
42775	PHACE syndrome	Disorder	300 Case(s)
41	Dyschromatosis symmetrica hereditaria	Disorder	300 Case(s)
3347	Mounier-Kühn syndrome	Disorder	300 Case(s)
309025	Mevalonate kinase deficiency	Disorder	300 Case(s)
3071	Costello syndrome	Disorder	300 Case(s)
247245	Superficial siderosis	Disorder	300 Case(s)
237	Duplication of urethra	Disorder	300 Case(s)
2330	Kasabach-Merritt phenomenon	Disorder	300 Case(s)
228302	Carnitine palmitoyl transferase II deficiency, myopathic form	Subtype of disorder	300 Case(s)
220	Denys-Drash syndrome	Disorder	300 Case(s)
2092	Focal dermal hypoplasia	Disorder	300 Case(s)
206569	Immune-mediated necrotizing myopathy	Disorder	300 Case(s)
184	Cherubism	Disorder	300 Case(s)
157	Carnitine palmitoyltransferase II deficiency	Disorder	300 Case(s)
1556	Cutis marmorata telangiectatica congenita	Disorder	300 Case(s)
1467	Cogan syndrome	Disorder	300 Case(s)
1340	Cardiofaciocutaneous syndrome	Disorder	300 Case(s)
1328	Camurati-Engelmann disease	Disorder	300 Case(s)
125	Bloom syndrome	Disorder	300 Case(s)
500	Noonan syndrome with multiple lentigines	Disorder	296 Case(s)
363549	Acute encephalopathy with biphasic seizures and late reduced diffusion	Disorder	283 Case(s)
599373	STXBP1-related encephalopathy	Disorder	282 Case(s)
2070	Eosinophilic gastroenteritis	Disorder	280 Case(s)
566943	Mueller-Weiss syndrome	Disorder	277 Case(s)
137667	Capillary malformation-arteriovenous malformation	Disorder	261 Case(s)
98954	Meesmann corneal dystrophy	Disorder	250 Case(s)
90283	Lupus erythematosus tumidus	Disorder	250 Case(s)
77258	Trichorhinophalangeal syndrome type 1	Disorder	250 Case(s)
397596	Activated PI3K-delta syndrome	Disorder	250 Case(s)
373	Simpson-Golabi-Behmel syndrome	Disorder	250 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
2908	Kindler epidermolysis bullosa	Disorder	250 Case(s)
221074	Marchiafava-Bignami disease	Disorder	250 Case(s)
167635	Scleromyxedema	Disorder	250 Case(s)
163634	Maffucci syndrome	Disorder	250 Case(s)
100006	ABeta amyloidosis, Dutch type	Subtype of disorder	250 Case(s)
199318	15q13.3 microdeletion syndrome	Disorder	246 Case(s)
2710	Oculodentodigital dysplasia	Disorder	243 Case(s)
464453	Acquired methemoglobinemia	Disorder	242 Case(s)
169105	Good syndrome	Disorder	241 Case(s)
99642	Spondyloepimetaphyseal dysplasia, Handigodu type	Disorder	234 Case(s)
2241	Megacystis-microcolon-intestinal hypoperistalsis syndrome	Disorder	230 Case(s)
1708	Mosaic trisomy 16	Disorder	226 Case(s)
65285	Lhermitte-Duclos disease	Disorder	220 Case(s)
1727	22q11.2 duplication syndrome	Disorder	216 Case(s)
2796	Pachydermoperiostosis	Disorder	204 Case(s)
2510	Micro syndrome	Disorder	203 Case(s)
33364	Trichothiodystrophy	Disorder	201 Case(s)
99050	Abnormal origin of right or left pulmonary artery from the aorta	Disorder	200 Case(s)
97360	Robinow syndrome	Disorder	200 Case(s)
901	Wells syndrome	Disorder	200 Case(s)
847	X-linked alpha-thalassemia-intellectual disability syndrome	Disorder	200 Case(s)
79277	Congenital erythropoietic porphyria	Disorder	200 Case(s)
79255	GM1 gangliosidosis type 1	Subtype of disorder	200 Case(s)
75563	X-linked sideroblastic anemia	Disorder	200 Case(s)
679	Malignant atrophic papulosis	Subtype of disorder	200 Case(s)
66630	Congenital pseudoarthrosis of the clavicle	Disorder	200 Case(s)
599490	Acquired factor V deficiency	Disorder	200 Case(s)
575	Muckle-Wells syndrome	Disorder	200 Case(s)
565612	Primary triglyceride deposit cardiomyovasculopathy	Disorder	200 Case(s)
559	Marinesco-Sjögren syndrome	Disorder	200 Case(s)
523	Hereditary leiomyomatosis and renal cell cancer	Disorder	200 Case(s)
48686	Primary effusion lymphoma	Disorder	200 Case(s)
48652	Phelan-McDermid syndrome	Disorder	200 Case(s)
48377	Subcorneal pustular dermatosis	Disorder	200 Case(s)
457	Harlequin ichthyosis	Disorder	200 Case(s)
414	Gyrate atrophy of choroid and retina	Disorder	200 Case(s)
343	Hyperimmunoglobulinemia D with periodic fever	Subtype of disorder	200 Case(s)
317	Erythrokeratodermia variabilis	Disorder	200 Case(s)
306516	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis	Disorder	200 Case(s)
302	Inherited epidermodysplasia verruciformis	Disorder	200 Case(s)
289494	4H leukodystrophy	Disorder	200 Case(s)
2616	3M syndrome	Disorder	200 Case(s)
261183	15q11.2 microdeletion syndrome	Disorder	200 Case(s)
221016	Rothmund-Thomson syndrome type 2	Subtype of disorder	200 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
220407	Limited systemic sclerosis	Subtype of disorder	200 Case(s)
2088	Fanconi-Bickel syndrome	Disorder	200 Case(s)
199267	Infantile digital fibromatosis	Disorder	200 Case(s)
1986	Gollop-Wolfgang complex	Disorder	200 Case(s)
1980	Bilateral striopallidodentate calcinosis	Disorder	200 Case(s)
193	Cohen syndrome	Disorder	200 Case(s)
1540	Jackson-Weiss syndrome	Disorder	200 Case(s)
139436	Multicentric reticulohistiocytosis	Disorder	200 Case(s)
137867	Madras motor neuron disease	Disorder	200 Case(s)
1300	Autosomal dominant popliteal pterygium syndrome	Disorder	200 Case(s)
1063	Tufted angioma	Disorder	200 Case(s)
1059	Blue rubber bleb nevus	Disorder	200 Case(s)
627	Nance-Horan syndrome	Disorder	196 Case(s)
402035	Eosinophilic colitis	Disorder	196 Case(s)
37042	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	Disorder	195 Case(s)
699	Pearson syndrome	Disorder	194 Case(s)
28	Vitamin B12-responsive methylmalonic acidemia	Disorder	192 Case(s)
1465	Coffin-Siris syndrome	Disorder	190 Case(s)
293381	Epithelial recurrent erosion dystrophy	Disorder	186 Case(s)
31150	Tangier disease	Disorder	185 Case(s)
757	Pseudohypoaldosteronism type 2	Disorder	180 Case(s)
319552	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	Disorder	180 Case(s)
254509	Iatrogenic botulism	Subtype of disorder	180 Case(s)
2237	Hypoparathyroidism-sensorineural deafness-renal disease syndrome	Disorder	180 Case(s)
1475	Renal coloboma syndrome	Disorder	180 Case(s)
572	Immunodeficiency by defective expression of MHC class II	Disorder	179 Case(s)
98960	Thiel-Behnke corneal dystrophy	Disorder	173 Case(s)
576278	SATB2-associated syndrome	Disorder	171 Case(s)
97685	17q11 microdeletion syndrome	Subtype of disorder	170 Case(s)
60040	Megalencephaly-capillary malformation-polymicrogyria syndrome	Disorder	170 Case(s)
48431	Congenital cataracts-facial dysmorphism-neuropathy syndrome	Disorder	170 Case(s)
324636	Autoerythrocyte sensitization syndrome	Disorder	170 Case(s)
252212	Malignant triton tumor	Subtype of disorder	170 Case(s)
1713	17p11.2 microduplication syndrome	Disorder	170 Case(s)
1459	Celiac disease-epilepsy-cerebral calcification syndrome	Disorder	170 Case(s)
2332	KBG syndrome	Disorder	164 Case(s)
96121	7q11.23 microduplication syndrome	Disorder	163 Case(s)
261243	16p13.11 microduplication syndrome	Disorder	162 Case(s)
349	Fucosidosis	Disorder	161 Case(s)
589618	Dystonia 28	Disorder	160 Case(s)
1522	Craniometaphyseal dysplasia	Disorder	160 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
2065	Galloway-Mowat syndrome	Disorder	159 Case(s)
585	Multiple sulfatase deficiency	Disorder	154 Case(s)
300324	Persistent polyclonal B-cell lymphocytosis	Disorder	154 Case(s)
93164	Transient pseudohypoaldosteronism	Disorder	152 Case(s)
84142	Isaacs syndrome	Disorder	150 Case(s)
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	Subtype of disorder	150 Case(s)
71518	Benign paroxysmal torticollis of infancy	Disorder	150 Case(s)
71274	Disseminated peritoneal leiomyomatosis	Disorder	150 Case(s)
52503	X-linked creatine transporter deficiency	Disorder	150 Case(s)
498474	Hyaline fibromatosis syndrome	Disorder	150 Case(s)
381	Griscelli syndrome	Disorder	150 Case(s)
37748	Schnitzler syndrome	Disorder	150 Case(s)
35069	Infantile neuroaxonal dystrophy	Disorder	150 Case(s)
347	Frasier syndrome	Disorder	150 Case(s)
3467	Hereditary xanthinuria	Disorder	150 Case(s)
329481	Lipoprotein glomerulopathy	Disorder	150 Case(s)
3265	Isolated humero-radial synostosis	Disorder	150 Case(s)
3197	Hereditary hyperekplexia	Disorder	150 Case(s)
314777	Familial isolated pituitary adenoma	Disorder	150 Case(s)
3103	Roberts syndrome	Disorder	150 Case(s)
284454	Acute zonal occult outer retinopathy	Disorder	150 Case(s)
28378	Tyrosinemia type 2	Disorder	150 Case(s)
2637	Microcephalic osteodysplastic primordial dwarfism type II	Disorder	150 Case(s)
2576	Mulibrey nanism	Disorder	150 Case(s)
236	Trisomy 9p	Disorder	150 Case(s)
226	Dihydropteridine reductase deficiency	Subtype of disorder	150 Case(s)
2108	Hallermann-Streiff syndrome	Disorder	150 Case(s)
2048	Foix-Chavany-Marie syndrome	Disorder	150 Case(s)
188	Systemic capillary leak syndrome	Disorder	150 Case(s)
168816	Peritoneal inclusion cyst	Disorder	150 Case(s)
1590	Distal deletion 13q	Disorder	150 Case(s)
140944	CLOVES syndrome	Disorder	150 Case(s)
139411	Carney triad	Disorder	150 Case(s)
1297	Branchio-oculo-facial syndrome	Disorder	150 Case(s)
135	CACH syndrome	Disorder	148 Case(s)
398166	Focal facial dermal dysplasia	Disorder	147 Case(s)
457083	Isolated splenogonadal fusion	Disorder	145 Case(s)
166113	Bazex syndrome	Disorder	145 Case(s)
113	Bazex-Dupré-Christol syndrome	Disorder	143 Case(s)
90003	Inflammatory pseudotumor of the liver	Disorder	140 Case(s)
83450	Regional odontodysplasia	Disorder	140 Case(s)
79314	L-2-hydroxyglutaric aciduria	Disorder	140 Case(s)
35708	Aromatic L-amino acid decarboxylase deficiency	Disorder	140 Case(s)
2290	Microvillus inclusion disease	Disorder	137 Case(s)
1830	Schimke immuno-osseous dysplasia	Disorder	133 Case(s)

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834	Free sialic acid storage disease	Disorder	130 Case(s)
3400	Aorto-ventricular tunnel	Disorder	130 Case(s)
291	Congenital varicella syndrome	Disorder	130 Case(s)
178307	Reticulate acropigmentation of Kitamura	Disorder	130 Case(s)
800	Schwartz-Jampel syndrome	Disorder	129 Case(s)
98920	Spinal muscular atrophy with respiratory distress type 1	Disorder	128 Case(s)
3138	Ulnar-mammary syndrome	Disorder	128 Case(s)
137898	Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	Disorder	127 Case(s)
650	LCAT deficiency	Disorder	125 Case(s)
2855	Perrault syndrome	Disorder	124 Case(s)
93583	Congenital thrombotic thrombocytopenic purpura	Subtype of disorder	123 Case(s)
1305	Feingold syndrome	Disorder	123 Case(s)
597746	Blepharophimosis-intellectual disability syndrome/genitopatellar overlap syndrome	Disorder	122 Case(s)
3047	Blepharophimosis-intellectual disability syndrome, SBBYS type	Disorder	122 Case(s)
90117	Hereditary motor and sensory neuropathy, Okinawa type	Disorder	120 Case(s)
440727	Combined hamartoma of the retina and retinal pigment epithelium	Disorder	120 Case(s)
391641	Feingold syndrome type 1	Subtype of disorder	120 Case(s)
163	Hereditary hyperferritinemia-cataract syndrome	Disorder	120 Case(s)
100026	Gamma-heavy chain disease	Subtype of disorder	120 Case(s)
1571	Knobloch syndrome	Disorder	119 Case(s)
261272	17q12 microduplication syndrome	Disorder	118 Case(s)
84064	Syndromic diarrhea	Disorder	116 Case(s)
98967	Schnyder corneal dystrophy	Disorder	115 Case(s)
48918	Focal myositis	Disorder	115 Case(s)
1001	2q37 microdeletion syndrome	Disorder	115 Case(s)
293181	Malignant migrating focal seizures of infancy	Disorder	114 Case(s)
261494	Kleefstra syndrome	Disorder	114 Case(s)
420584	Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome	Disorder	112 Case(s)
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	Disorder	111 Case(s)
31043	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement	Subtype of disorder	110 Case(s)
97229	Riboflavin transporter deficiency	Disorder	109 Case(s)
46627	Char syndrome	Disorder	109 Case(s)
79113	Mandibulofacial dysostosis-microcephaly syndrome	Disorder	107 Case(s)
86909	Myoclonic epilepsy of infancy	Disorder	106 Case(s)
537072	PLG-related hereditary angioedema with normal C1Inh	Subtype of disorder	105 Case(s)
261265	17q12 microdeletion syndrome	Disorder	103 Case(s)
79477	Griscelli syndrome type 2	Subtype of disorder	102 Case(s)
3342	Arterial tortuosity syndrome	Disorder	102 Case(s)
488239	Acute macular neuroretinopathy	Disorder	101 Case(s)
99880	Hyperparathyroidism-jaw tumor syndrome	Disorder	100 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
99063	Shone complex	Disorder	100 Case(s)
99015	Spastic paraplegia type 2	Disorder	100 Case(s)
981	Internal carotid absence	Disorder	100 Case(s)
96095	3q26 microduplication syndrome	Disorder	100 Case(s)
955	Hajdu-Cheney syndrome	Disorder	100 Case(s)
94087	Cytophagic histiocytic panniculitis	Disorder	100 Case(s)
91136	Acquired monoclonal Ig light chain-associated Fanconi syndrome	Disorder	100 Case(s)
89937	Autosomal dominant hypophosphatemic rickets	Disorder	100 Case(s)
898	Wagner disease	Disorder	100 Case(s)
897	Waardenburg-Shah syndrome	Disorder	100 Case(s)
869	Triple A syndrome	Disorder	100 Case(s)
86813	Helicoid peripapillary chorioretinal degeneration	Disorder	100 Case(s)
833	Encephalopathy due to sulfite oxidase deficiency	Disorder	100 Case(s)
79493	Brooke-Spiegler syndrome	Disorder	100 Case(s)
79409	Recessive dystrophic epidermolysis bullosa inversa	Disorder	100 Case(s)
79403	Junctional epidermolysis bullosa with pyloric atresia	Disorder	100 Case(s)
75326	Familial isolated retinal arteriolar tortuosity	Disorder	100 Case(s)
746	Mitochondrial trifunctional protein deficiency	Disorder	100 Case(s)
724	Idiopathic acute eosinophilic pneumonia	Disorder	100 Case(s)
71517	Rapid-onset dystonia-parkinsonism	Disorder	100 Case(s)
71279	CANOMAD syndrome	Disorder	100 Case(s)
709	Peters plus syndrome	Disorder	100 Case(s)
672	Pallister-Hall syndrome	Disorder	100 Case(s)
65748	Multiple self-healing squamous epithelioma	Disorder	100 Case(s)
617916	Diffuse idiopathic pulmonary neuroendocrine cell hyperplasia	Disorder	100 Case(s)
604680	Symptomatic form of X-linked centronuclear myopathy in female carriers	Disorder	100 Case(s)
59315	Rhombencephalosynapsis	Disorder	100 Case(s)
59306	McLeod neuroacanthocytosis syndrome	Disorder	100 Case(s)
538934	X-linked lymphoproliferative disease due to XIAP deficiency	Disorder	100 Case(s)
538931	X-linked lymphoproliferative disease due to SAP deficiency	Disorder	100 Case(s)
504476	Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome	Disorder	100 Case(s)
502	Trichorhinophalangeal syndrome type 2	Disorder	100 Case(s)
477	KID syndrome	Disorder	100 Case(s)
45	Adenosine monophosphate deaminase deficiency	Disorder	100 Case(s)
371	Glycogen storage disease due to muscle phosphofructokinase deficiency	Disorder	100 Case(s)
352723	Attenuated Chédiak-Higashi syndrome	Disorder	100 Case(s)
351	Galactosialidosis	Disorder	100 Case(s)
3344	Weismann-Netter syndrome	Disorder	100 Case(s)
332	Congenital intrinsic factor deficiency	Disorder	100 Case(s)
3319	Congenital amegakaryocytic thrombocytopenia	Disorder	100 Case(s)
33110	Autosomal agammaglobulinemia	Subtype of disorder	100 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
3107	Autosomal dominant Robinow syndrome	Subtype of disorder	100 Case(s)
30924	Primary hypomagnesemia with secondary hypocalcemia	Disorder	100 Case(s)
306741	Hemidystonia-hemiatrophy syndrome	Disorder	100 Case(s)
2882	Sitosterolemia	Disorder	100 Case(s)
279934	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency	Disorder	100 Case(s)
2785	Osteopetrosis with renal tubular acidosis	Disorder	100 Case(s)
2780	Osteopathia striata-cranial sclerosis syndrome	Disorder	100 Case(s)
274	Bernard-Soulier syndrome	Disorder	100 Case(s)
2704	Urofacial syndrome	Disorder	100 Case(s)
2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	Disorder	100 Case(s)
2632	Langer mesomelic dysplasia	Disorder	100 Case(s)
261476	Xp21 deletion syndrome	Disorder	100 Case(s)
254478	Lichen planus pemphigoides	Disorder	100 Case(s)
251295	Pigmented paravenous retinochoroidal atrophy	Disorder	100 Case(s)
2478	Megalencephalic leukoencephalopathy with subcortical cysts	Disorder	100 Case(s)
245	Nager syndrome	Disorder	100 Case(s)
2414	Congenital pulmonary lymphangiectasia	Disorder	100 Case(s)
239	Dyggve-Melchior-Clausen disease	Disorder	100 Case(s)
238769	1q44 microdeletion syndrome	Disorder	100 Case(s)
2363	Lacrimoauriculodentodigital syndrome	Disorder	100 Case(s)
2342	Haim-Munk syndrome	Disorder	100 Case(s)
2222	Hypertrichosis lanuginosa congenita	Disorder	100 Case(s)
221008	Rothmund-Thomson syndrome type 1	Subtype of disorder	100 Case(s)
209905	Brain-lung-thyroid syndrome	Disorder	100 Case(s)
2053	Freeman-Sheldon syndrome	Disorder	100 Case(s)
199282	Harlequin syndrome	Disorder	100 Case(s)
199241	Pulmonary capillary hemangiomatosis	Disorder	100 Case(s)
1929	Rasmussen subacute encephalitis	Disorder	100 Case(s)
1826	Frontometaphyseal dysplasia	Disorder	100 Case(s)
168569	H syndrome	Disorder	100 Case(s)
166305	Benign infantile seizures associated with mild gastroenteritis	Disorder	100 Case(s)
1507	Autosomal recessive Robinow syndrome	Subtype of disorder	100 Case(s)
1446	Ring chromosome 22 syndrome	Disorder	100 Case(s)
140957	Autosomal dominant macrothrombocytopenia	Disorder	100 Case(s)
14	Abetalipoproteinemia	Disorder	100 Case(s)
137675	Histiocytoid cardiomyopathy	Disorder	100 Case(s)
1310	Caffey disease	Disorder	100 Case(s)
1221	Cheilitis glandularis	Disorder	100 Case(s)
927	Hyperammonemia due to N-acetylglutamate synthase deficiency	Disorder	99 Case(s)
329211	Autosomal dominant neovascular inflammatory vitreoretinopathy	Disorder	99 Case(s)
589547	GRIN2B-related developmental delay, intellectual disability and autism spectrum disorder	Disorder	98 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
2670	Pierson syndrome	Disorder	98 Case(s)
75381	Cystoid macular dystrophy	Disorder	97 Case(s)
363447	Autosomal dominant childhood-onset proximal spinal muscular atrophy	Disorder	97 Case(s)
333	Farber disease	Disorder	96 Case(s)
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	Disorder	96 Case(s)
599513	Acquired factor XIII deficiency	Disorder	95 Case(s)
52368	Mohr-Tranebjaerg syndrome	Disorder	91 Case(s)
2671	Neu-Laxova syndrome	Disorder	91 Case(s)
742	Prolidase deficiency	Disorder	90 Case(s)
53719	Wyburn-Mason syndrome	Disorder	90 Case(s)
498228	Phyllodes tumor of the prostate	Disorder	90 Case(s)
2473	McKusick-Kaufman syndrome	Disorder	90 Case(s)
1885	Isolated ectopia lentis	Disorder	90 Case(s)
157846	Neuroferritinopathy	Disorder	90 Case(s)
1642	Distal deletion 9p	Disorder	89 Case(s)
96147	Kleefstra syndrome due to 9q34 microdeletion	Subtype of disorder	86 Case(s)
1738	Trisomy 4p	Disorder	85 Case(s)
34587	Danon disease	Disorder	84 Case(s)
3403	Uhl anomaly	Disorder	84 Case(s)
319182	Wiedemann-Steiner syndrome	Disorder	84 Case(s)
254519	Kagami-Ogata syndrome	Disorder	84 Case(s)
599495	Acquired factor VII deficiency	Disorder	83 Case(s)
98961	Reis-Bücklers corneal dystrophy	Disorder	81 Case(s)
79133	Focal facial dermal dysplasia type I	Subtype of disorder	81 Case(s)
2635	Metatropic dysplasia	Disorder	81 Case(s)
98769	Spinocerebellar ataxia type 15/16	Disorder	80 Case(s)
950	Acrodysostosis	Disorder	80 Case(s)
79315	D-2-hydroxyglutaric aciduria	Disorder	80 Case(s)
51188	Ethylmalonic encephalopathy	Disorder	80 Case(s)
49827	Thiamine-responsive megaloblastic anemia syndrome	Disorder	80 Case(s)
49	Penile agenesis	Disorder	80 Case(s)
382	Guanidinoacetate methyltransferase deficiency	Disorder	80 Case(s)
3152	Sclerosteosis	Disorder	80 Case(s)
314404	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	Disorder	80 Case(s)
238569	Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome	Disorder	80 Case(s)
231401	Alpha-thalassemia-myelodysplastic syndrome	Disorder	80 Case(s)
1935	Early myoclonic encephalopathy	Disorder	80 Case(s)
1440	Ring chromosome 14 syndrome	Disorder	80 Case(s)
599501	Acquired factor X deficiency	Disorder	77 Case(s)
352577	Bainbridge-Ropers syndrome	Disorder	77 Case(s)
2396	Encephalocraniocutaneous lipomatosis	Disorder	77 Case(s)
457193	Autosomal dominant intellectual disability-craniofacial	Disorder	76 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	anomalies-cardiac defects syndrome		
592564	GNAO1-related developmental delay-seizures-movement disorder spectrum	Disorder	75 Case(s)
320406	Spastic paraplegia-optic atrophy-neuropathy syndrome	Disorder	75 Case(s)
238722	Familial congenital mirror movements	Disorder	75 Case(s)
209981	IRIDA syndrome	Disorder	75 Case(s)
1393	Cerebrocostomandibular syndrome	Disorder	75 Case(s)
79230	HJV or HAMP-related hemochromatosis	Disorder	74 Case(s)
561	Marshall-Smith syndrome	Disorder	74 Case(s)
659	Mutilating palmoplantar keratoderma with periorificial keratotic plaques	Disorder	73 Case(s)
622	Homocystinuria without methylmalonic aciduria	Disorder	73 Case(s)
760	Purine nucleoside phosphorylase deficiency	Disorder	72 Case(s)
2196	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement	Subtype of disorder	72 Case(s)
90280	Chilblain lupus	Disorder	70 Case(s)
79293	Familial LCAT deficiency	Subtype of disorder	70 Case(s)
79257	GM1 gangliosidosis type 3	Subtype of disorder	70 Case(s)
65759	Carpenter syndrome	Disorder	70 Case(s)
530983	Lamb-Shaffer syndrome	Disorder	70 Case(s)
404546	DITRA	Disorder	70 Case(s)
369891	Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	Disorder	70 Case(s)
357043	Amyotrophic lateral sclerosis type 4	Disorder	70 Case(s)
3310	Tetrasomy 9p	Disorder	70 Case(s)
32	Glutathione synthetase deficiency	Disorder	70 Case(s)
2701	Noonan syndrome-like disorder with loose anagen hair	Disorder	70 Case(s)
2484	Melnick-Needles syndrome	Disorder	70 Case(s)
2123	Multifocal infantile hemangioma with extracutenous involvement	Disorder	70 Case(s)
2028	Juvenile hyaline fibromatosis	Subtype of disorder	70 Case(s)
2006	Median cleft lip/mandible	Disorder	70 Case(s)
1442	Ring chromosome 18 syndrome	Disorder	70 Case(s)
90791	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	Disorder	68 Case(s)
329284	Beta-propeller protein-associated neurodegeneration	Disorder	68 Case(s)
319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	Disorder	68 Case(s)
306669	Hemiparkinsonism-hemiatrophy syndrome	Disorder	68 Case(s)
352328	MEGDEL syndrome	Disorder	67 Case(s)
2554	Ear-patella-short stature syndrome	Disorder	67 Case(s)
2062	Progressive non-infectious anterior vertebral fusion	Disorder	67 Case(s)
1993	Pai syndrome	Disorder	67 Case(s)
160148	Cap polyposis	Disorder	67 Case(s)
3405	Umbilical cord ulceration-intestinal atresia syndrome	Disorder	66 Case(s)
2268	ICF syndrome	Disorder	66 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
90354	Brittle cornea syndrome	Disorder	65 Case(s)
51636	WHIM syndrome	Disorder	65 Case(s)
2333	Kenny-Caffey syndrome	Disorder	65 Case(s)
96184	Temple syndrome due to maternal uniparental disomy of chromosome 14	Subtype of disorder	64 Case(s)
55595	TNP03-related limb-girdle muscular dystrophy D2	Disorder	64 Case(s)
3242	Renpenning syndrome	Disorder	64 Case(s)
2990	Autosomal recessive multiple pterygium syndrome	Disorder	64 Case(s)
83473	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	Disorder	62 Case(s)
75392	Periodontal Ehlers-Danlos syndrome	Disorder	62 Case(s)
69736	Bilateral acute depigmentation of the iris	Disorder	62 Case(s)
1988	Femoral-facial syndrome	Disorder	62 Case(s)
641368	Autosomal recessive hyper-IgE syndrome due to ZNF341 deficiency	Disorder	61 Case(s)
3051	Nicolaides-Baraitser syndrome	Disorder	61 Case(s)
99803	Haddad syndrome	Disorder	60 Case(s)
98870	Congenital dyserythropoietic anemia type III	Disorder	60 Case(s)
969	Acromicric dysplasia	Disorder	60 Case(s)
96092	8p inverted duplication/deletion syndrome	Disorder	60 Case(s)
90349	Autosomal recessive cutis laxa type 1	Disorder	60 Case(s)
83467	Morvan syndrome	Disorder	60 Case(s)
79310	Vitamin B12-responsive methylmalonic acidemia type cblA	Subtype of disorder	60 Case(s)
773	Refsum disease	Disorder	60 Case(s)
721	Gray platelet syndrome	Disorder	60 Case(s)
708	Peters anomaly	Disorder	60 Case(s)
677	Pancreatoblastoma	Disorder	60 Case(s)
641829	Neonatal compartment syndrome	Disorder	60 Case(s)
599082	CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	Disorder	60 Case(s)
52530	Pseudo-von Willebrand disease	Disorder	60 Case(s)
468635	Cryptogenic multifocal ulcerous stenosing enteritis	Disorder	60 Case(s)
451607	Cutaneous pseudolymphoma	Disorder	60 Case(s)
363454	BICD2-related autosomal dominant childhood-onset proximal spinal muscular atrophy	Subtype of disorder	60 Case(s)
352490	Autism spectrum disorder due to AUTS2 deficiency	Disorder	60 Case(s)
3411	Double uterus-hemivagina-renal agenesis syndrome	Disorder	60 Case(s)
300493	Sagliker syndrome	Disorder	60 Case(s)
2995	Baraitser-Winter cerebrofrontofacial syndrome	Disorder	60 Case(s)
2771	Bruck syndrome	Disorder	60 Case(s)
2462	Shprintzen-Goldberg syndrome	Disorder	60 Case(s)
2221	Acquired hypertrichosis lanuginosa	Disorder	60 Case(s)
2067	GAPO syndrome	Disorder	60 Case(s)
1667	Wolcott-Rallison syndrome	Disorder	60 Case(s)
159	Carnitine-acylcarnitine translocase deficiency	Disorder	60 Case(s)
158029	Sea-blue histiocytosis	Disorder	60 Case(s)

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156	Carnitine palmitoyl transferase 1A deficiency	Disorder	60 Case(s)
1515	Cranioectodermal dysplasia	Disorder	60 Case(s)
139	CHILD syndrome	Disorder	60 Case(s)
1270	Bowen-Conradi syndrome	Disorder	60 Case(s)
583097	Congenital infiltrating lipomatosis of the face	Disorder	59 Case(s)
3338	Toriello-Carey syndrome	Disorder	59 Case(s)
57196	Medial condensing osteitis of the clavicle	Disorder	58 Case(s)
88644	Autosomal recessive ataxia, Beauce type	Disorder	57 Case(s)
79327	ALG1-CDG	Disorder	57 Case(s)
544254	SYNGAP1-related developmental and epileptic encephalopathy	Disorder	57 Case(s)
331176	Severe congenital neutropenia due to G6PC3 deficiency	Disorder	57 Case(s)
90024	Deafness with labyrinthine aplasia, microtia, and microdontia	Disorder	56 Case(s)
65283	Timothy syndrome	Disorder	56 Case(s)
46	Adenylosuccinate lyase deficiency	Disorder	56 Case(s)
3206	Stüve-Wiedemann syndrome	Disorder	56 Case(s)
1777	Temtamy syndrome	Disorder	56 Case(s)
71	Chylomicron retention disease	Disorder	55 Case(s)
592570	TRAF7-associated heart defect-digital anomalies-facial dysmorphism-motor and speech delay syndrome	Disorder	55 Case(s)
276435	Lower motor neuron syndrome with late-adult onset	Disorder	55 Case(s)
2556	Microphthalmia with linear skin defects syndrome	Disorder	55 Case(s)
1997	Blepharo-cheilo-odontic syndrome	Disorder	55 Case(s)
83628	LUMBAR syndrome	Disorder	54 Case(s)
79320	ALG6-CDG	Disorder	54 Case(s)
57782	Mazabraud syndrome	Disorder	54 Case(s)
464306	DYRK1A-related intellectual disability syndrome	Disorder	54 Case(s)
314603	Autosomal recessive spastic ataxia with leukoencephalopathy	Disorder	54 Case(s)
2833	Stiff skin syndrome	Disorder	54 Case(s)
98806	Primary dystonia, DYT6 type	Disorder	53 Case(s)
79099	Interstitial granulomatous dermatitis with arthritis	Disorder	53 Case(s)
69126	PAPA syndrome	Disorder	53 Case(s)
398088	Hereditary cryohydrocytosis with normal stomatin	Disorder	53 Case(s)
2636	Microcephalic osteodysplastic primordial dwarfism types I and III	Disorder	53 Case(s)
254516	Temple syndrome	Disorder	53 Case(s)
251515	Distal arthrogryposis type 10	Disorder	53 Case(s)
251282	Autosomal dominant spastic ataxia type 1	Disorder	53 Case(s)
178509	Perry syndrome	Disorder	53 Case(s)
79411	Self-improving dystrophic epidermolysis bullosa	Disorder	52 Case(s)
449566	Eosinophilic angiocentric fibrosis	Disorder	52 Case(s)
3473	Zimmermann-Laband syndrome	Disorder	52 Case(s)
251671	Angiocentric glioma	Disorder	52 Case(s)
98767	Spinocerebellar ataxia type 11	Disorder	51 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
572761	DONSON-related microcephaly-short stature-limb abnormalities spectrum	Disorder	51 Case(s)
521414	Autosomal dominant Charcot-Marie-Tooth disease type 2DD	Disorder	51 Case(s)
1766	Dysequilibrium syndrome	Disorder	51 Case(s)
99776	Mosaic trisomy 9	Disorder	50 Case(s)
99731	Isolated sulfite oxidase deficiency	Subtype of disorder	50 Case(s)
98811	Paroxysmal exertion-induced dyskinesia	Disorder	50 Case(s)
978	ADULT syndrome	Disorder	50 Case(s)
96177	Ring chromosome 15 syndrome	Disorder	50 Case(s)
93600	Primary hyperoxaluria type 3	Subtype of disorder	50 Case(s)
91496	Snowflake vitreoretinal degeneration	Disorder	50 Case(s)
90348	Autosomal dominant cutis laxa	Disorder	50 Case(s)
90342	Xeroderma pigmentosum variant	Disorder	50 Case(s)
9	Tetrasomy X	Disorder	50 Case(s)
871	Familial progressive cardiac conduction defect	Disorder	50 Case(s)
86816	Congenital analbuminemia	Disorder	50 Case(s)
868	Triose phosphate-isomerase deficiency	Disorder	50 Case(s)
85212	Fetal Gaucher disease	Subtype of disorder	50 Case(s)
85136	Cystic leukoencephalopathy without megalencephaly	Disorder	50 Case(s)
851	Paris-Trousseau thrombocytopenia	Disorder	50 Case(s)
808	Seckel syndrome	Disorder	50 Case(s)
79500	DOORS syndrome	Disorder	50 Case(s)
79395	Keratoderma hereditarium mutilans with ichthyosis	Disorder	50 Case(s)
79256	GM1 gangliosidosis type 2	Subtype of disorder	50 Case(s)
79147	Familial reactive perforating collagenosis	Disorder	50 Case(s)
79143	Isolated congenital anonychia	Disorder	50 Case(s)
75382	Oguchi disease	Disorder	50 Case(s)
712	Hemolytic anemia due to glucophosphate isomerase deficiency	Disorder	50 Case(s)
631103	Spinocerebellar ataxia type 48	Disorder	50 Case(s)
574	21q deletion syndrome	Disorder	50 Case(s)
542310	Leukoencephalopathy with calcifications and cysts	Disorder	50 Case(s)
53540	Goldmann-Favre syndrome	Disorder	50 Case(s)
494	Keratoderma hereditarium mutilans	Disorder	50 Case(s)
454710	Anti-p200 pemphigoid	Disorder	50 Case(s)
443197	X-linked erythropoietic protoporphyria	Disorder	50 Case(s)
404507	Chondromyxoid fibroma	Disorder	50 Case(s)
40	Acromesomelic dysplasia, Maroteaux type	Disorder	50 Case(s)
371428	Multicentric osteolysis-nodulosis-arthropathy spectrum	Disorder	50 Case(s)
352636	Phalangeal microgeodic syndrome	Disorder	50 Case(s)
33111	Granulomatous slack skin	Disorder	50 Case(s)
3253	Cleft lip/palate-ectodermal dysplasia syndrome	Disorder	50 Case(s)
3130	Satoyoshi syndrome	Disorder	50 Case(s)
3111	Rotor syndrome	Disorder	50 Case(s)
300512	Onychomatricoma	Disorder	50 Case(s)

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29822	Spontaneous periodic hypothermia	Disorder	50 Case(s)
284448	CLIPPERS	Disorder	50 Case(s)
2805	Partial pancreatic agenesis	Disorder	50 Case(s)
2801	Juvenile Paget disease	Disorder	50 Case(s)
2461	Marden-Walker syndrome	Disorder	50 Case(s)
2407	Laryngo-onycho-cutaneous syndrome	Disorder	50 Case(s)
221046	Poikiloderma with neutropenia	Disorder	50 Case(s)
217385	17p13.3 microduplication syndrome	Disorder	50 Case(s)
2143	Donnai-Barrow syndrome	Disorder	50 Case(s)
2136	Hennekam syndrome	Disorder	50 Case(s)
208513	Spinocerebellar ataxia type 29	Disorder	50 Case(s)
2078	Geroderma osteodysplastica	Disorder	50 Case(s)
206583	Adult polyglucosan body disease	Subtype of disorder	50 Case(s)
1902	Ehrlichiosis	Disorder	50 Case(s)
171929	Trisomy 10p	Disorder	50 Case(s)
1573	Hypotrichosis with juvenile macular degeneration	Disorder	50 Case(s)
1517	Cantú syndrome	Disorder	50 Case(s)
1493	Vici syndrome	Disorder	50 Case(s)
1444	Ring chromosome 20 syndrome	Disorder	50 Case(s)
1425	Desbuquois syndrome	Disorder	50 Case(s)
137888	Auriculocondylar syndrome	Disorder	50 Case(s)
127	Borjeson-Forssman-Lehmann syndrome	Disorder	50 Case(s)
1253	Ascher syndrome	Disorder	50 Case(s)
1125	Ocular motor apraxia, Cogan type	Disorder	50 Case(s)
1118	Fibular aplasia-ectrodactyly syndrome	Disorder	50 Case(s)
103908	Congenital sodium diarrhea	Disorder	50 Case(s)
101150	Autosomal recessive dopa-responsive dystonia	Disorder	50 Case(s)
100012	Lissencephaly with cerebellar hypoplasia type B	Disorder	50 Case(s)
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	Disorder	49 Case(s)
54251	Aseptic abscess syndrome	Disorder	49 Case(s)
319558	Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency	Disorder	49 Case(s)
255229	Navajo neurohepatopathy	Disorder	49 Case(s)
1873	Jalili syndrome	Disorder	49 Case(s)
544469	PRUNE1-related neurological syndrome	Disorder	48 Case(s)
404553	Adenosine deaminase 2 deficiency	Disorder	48 Case(s)
391372	FOXP1 Syndrome	Disorder	48 Case(s)
3447	Weaver syndrome	Disorder	48 Case(s)
2897	Pityriasis rubra pilaris	Disorder	48 Case(s)
989	Hypoglossia-hypodactyly syndrome	Disorder	47 Case(s)
85162	Facial onset sensory and motor neuronopathy	Disorder	47 Case(s)
565909	Calpain-3-related limb-girdle muscular dystrophy D4	Disorder	47 Case(s)
357008	Hemolytic uremic syndrome with DGKE deficiency	Disorder	47 Case(s)
216828	Osteogenesis imperfecta type 5	Subtype of disorder	47 Case(s)
1509	Coxopodopatellar syndrome	Disorder	47 Case(s)

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1414	Cholestasis-lymphedema syndrome	Disorder	47 Case(s)
97297	Bohring-Opitz syndrome	Disorder	46 Case(s)
798	Schinzel-Giedion syndrome	Disorder	46 Case(s)
319646	PGM1-CDG	Disorder	46 Case(s)
250994	1q21.1 microduplication syndrome	Disorder	46 Case(s)
99749	Kostmann syndrome	Disorder	45 Case(s)
86788	X-linked severe congenital neutropenia	Disorder	45 Case(s)
53721	Spinal arteriovenous metameric syndrome	Disorder	45 Case(s)
284984	Aneurysm-osteoarthritis syndrome	Disorder	45 Case(s)
279947	Postorgasmic illness syndrome	Disorder	45 Case(s)
254875	Mitochondrial DNA depletion syndrome, myopathic form	Disorder	45 Case(s)
209932	Cone dystrophy with supernormal rod response	Disorder	45 Case(s)
1955	Spinocerebellar ataxia type 34	Disorder	45 Case(s)
166286	Porokeratotic eccrine ostial and dermal duct nevus	Disorder	45 Case(s)
99938	Autosomal dominant Charcot-Marie-Tooth disease type 2D	Disorder	44 Case(s)
538756	Familial multiple discoid fibromas	Disorder	44 Case(s)
221126	Fowler vasculopathy	Disorder	44 Case(s)
168606	Seborrhea-like dermatitis with psoriasiform elements	Disorder	44 Case(s)
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency	Disorder	43 Case(s)
2470	Matthew-Wood syndrome	Disorder	43 Case(s)
2301	Congenital short bowel syndrome	Disorder	43 Case(s)
77301	Monosomy 9q22.3	Disorder	42 Case(s)
352629	16q24.1 microdeletion syndrome	Disorder	42 Case(s)
1621	3q13 microdeletion syndrome	Disorder	42 Case(s)
398156	Oculoauriculofrontonasal syndrome	Disorder	41 Case(s)
2907	Hereditary acrokeratotic poikiloderma	Disorder	41 Case(s)
254351	Distal 7q11.23 microdeletion syndrome	Disorder	41 Case(s)
1052	Mosaic variegated aneuploidy syndrome	Disorder	41 Case(s)
99844	Leukocyte adhesion deficiency type III	Subtype of disorder	40 Case(s)
96148	Distal deletion 10q	Disorder	40 Case(s)
96102	Distal duplication 10q	Disorder	40 Case(s)
95159	Hepatoerythropoietic porphyria	Disorder	40 Case(s)
90652	Otopalatodigital syndrome type 2	Disorder	40 Case(s)
859	Transcobalamin deficiency	Disorder	40 Case(s)
79134	DEND syndrome	Disorder	40 Case(s)
79	Congenital alpha2-antiplasmin deficiency	Disorder	40 Case(s)
52022	Potocki-Shaffer syndrome	Disorder	40 Case(s)
500163	Witteveen-Kolk syndrome	Disorder	40 Case(s)
438117	Steel syndrome	Disorder	40 Case(s)
411777	Generalized eruptive keratoacanthoma	Disorder	40 Case(s)
324977	Proteasome-associated autoinflammatory syndrome	Disorder	40 Case(s)
314422	Ameloblastic carcinoma	Disorder	40 Case(s)
2971	Peroxisomal acyl-CoA oxidase deficiency	Disorder	40 Case(s)
2962	De Barsy syndrome	Disorder	40 Case(s)
281190	Congenital reticular ichthyosiform erythroderma	Disorder	40 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
280785	Bullous diffuse cutaneous mastocytosis	Subtype of disorder	40 Case(s)
263534	Acral peeling skin syndrome	Disorder	40 Case(s)
257	Epidermolysis bullosa simplex with muscular dystrophy	Disorder	40 Case(s)
2457	Mandibuloacral dysplasia	Disorder	40 Case(s)
24	Fumaric aciduria	Disorder	40 Case(s)
228384	5q14.3 microdeletion syndrome	Subtype of disorder	40 Case(s)
2273	Ichthyosis follicularis-alopecia-photophobia syndrome	Disorder	40 Case(s)
217008	Bockenheimer syndrome	Disorder	40 Case(s)
210548	Macrocephaly-intellectual disability-autism syndrome	Disorder	40 Case(s)
210122	Congenital alveolar capillary dysplasia	Disorder	40 Case(s)
1923	Methimazole embryofetopathy	Disorder	40 Case(s)
183678	Hermansky-Pudlak syndrome due to AP-3 deficiency	Subtype of disorder	40 Case(s)
1832	Osteosclerotic bone dysplasia	Disorder	40 Case(s)
1810	Autosomal dominant hypohidrotic ectodermal dysplasia	Subtype of disorder	40 Case(s)
1745	Distal duplication 6p	Disorder	40 Case(s)
1742	Trisomy 5p	Disorder	40 Case(s)
1699	Trisomy 12p	Disorder	40 Case(s)
163746	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	Disorder	40 Case(s)
140966	Palmoplantar keratoderma, Nagashima type	Disorder	40 Case(s)
1369	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome	Disorder	40 Case(s)
1225	Baller-Gerold syndrome	Disorder	40 Case(s)
1023	Congenital generalized hypertrichosis, Ambras type	Subtype of disorder	40 Case(s)
496641	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	Disorder	39 Case(s)
458758	Composite hemangioendothelioma	Disorder	39 Case(s)
391677	Short stature-optic atrophy-Pelger-Huët anomaly syndrome	Disorder	39 Case(s)
317473	Pancytopenia due to IKZF1 mutations	Disorder	39 Case(s)
99852	Ravine syndrome	Disorder	38 Case(s)
91	Aromatase deficiency	Disorder	38 Case(s)
69085	Limb-mammary syndrome	Disorder	38 Case(s)
55654	Hypotrichosis simplex	Disorder	38 Case(s)
485350	CLCN4-related X-linked intellectual disability syndrome	Disorder	38 Case(s)
457260	X-linked intellectual disability-hypotonia-movement disorder syndrome	Disorder	38 Case(s)
36	Acrocallosal syndrome	Disorder	38 Case(s)
314621	Duplication of the pituitary gland	Disorder	38 Case(s)
209867	Autosomal dominant rhegmatogenous retinal detachment	Disorder	38 Case(s)
171629	Autosomal recessive spastic paraplegia type 35	Disorder	38 Case(s)
1647	Oculocerebrocutaneous syndrome	Disorder	38 Case(s)
163696	Action myoclonus-renal failure syndrome	Disorder	38 Case(s)
96334	Kagami-Ogata syndrome due to paternal uniparental disomy of chromosome 14	Subtype of disorder	37 Case(s)
79406	Late-onset junctional epidermolysis bullosa	Disorder	37 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
596753	VEXAS syndrome	Disorder	37 Case(s)
494428	Idiopathic pleuroparenchymal fibroelastosis	Disorder	37 Case(s)
493342	Vibratory urticaria	Disorder	37 Case(s)
391417	HSD10 disease	Disorder	37 Case(s)
3455	Wiedemann-Rautenstrauch syndrome	Disorder	37 Case(s)
3208	Isolated succinate-CoQ reductase deficiency	Disorder	37 Case(s)
209341	DYNC1H1-related autosomal dominant childhood-onset proximal spinal muscular atrophy	Subtype of disorder	37 Case(s)
100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	Disorder	37 Case(s)
98955	Lisch epithelial corneal dystrophy	Disorder	36 Case(s)
98908	Neutral lipid storage disease with myopathy	Disorder	36 Case(s)
300573	Polymicrogyria due to TUBB2B mutation	Disorder	36 Case(s)
289478	PASH syndrome	Disorder	36 Case(s)
1855	Spondyloenchondrodysplasia	Disorder	36 Case(s)
168583	Hereditary North American Indian childhood cirrhosis	Subtype of disorder	36 Case(s)
166308	Benign infantile focal epilepsy with midline spikes and waves during sleep	Disorder	36 Case(s)
1532	Gómez-López-Hernández syndrome	Disorder	36 Case(s)
101000	Autosomal recessive spastic paraplegia type 20	Disorder	36 Case(s)
98773	Spinocerebellar ataxia type 21	Disorder	35 Case(s)
970	Hereditary sensory and autonomic neuropathy type 2	Disorder	35 Case(s)
96125	Distal deletion 6p	Disorder	35 Case(s)
589905	PHIP-related behavioral problems-intellectual disability-obesity-dysmorphic features syndrome	Disorder	35 Case(s)
566231	Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha	Disorder	35 Case(s)
464311	Intellectual disability syndrome due to a DYRK1A point mutation	Subtype of disorder	35 Case(s)
446	Neonatal hemochromatosis	Disorder	35 Case(s)
443073	Charcot-Marie-Tooth disease type 2S	Disorder	35 Case(s)
3416	Hyperostosis corticalis generalisata	Disorder	35 Case(s)
3275	Spondylocarpotarsal synostosis	Disorder	35 Case(s)
293621	X-linked endothelial corneal dystrophy	Disorder	35 Case(s)
2777	Osteomesopyknosis	Disorder	35 Case(s)
2117	Hartsfield syndrome	Disorder	35 Case(s)
2040	Congenital respiratory-biliary fistula	Disorder	35 Case(s)
198	Occipital horn syndrome	Disorder	35 Case(s)
1437	Ring chromosome 1 syndrome	Disorder	35 Case(s)
101001	Autosomal recessive spastic paraplegia type 21	Disorder	35 Case(s)
100045	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C	Disorder	35 Case(s)
100024	Mu-heavy chain disease	Subtype of disorder	35 Case(s)
943	Malonic aciduria	Disorder	34 Case(s)
93269	Short rib-polydactyly syndrome, Majewski type	Disorder	34 Case(s)
75496	B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome	Subtype of disorder	34 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
398097	Neonatal antiphospholipid syndrome	Disorder	34 Case(s)
363528	Intellectual disability-strabismus syndrome	Disorder	34 Case(s)
353284	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency	Subtype of disorder	34 Case(s)
2953	Musculocontractural Ehlers-Danlos syndrome	Disorder	34 Case(s)
2874	Phakomatosis pigmentokeratotica	Disorder	34 Case(s)
1620	Distal deletion 3p	Disorder	34 Case(s)
500150	Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	Disorder	33 Case(s)
488280	14q32 duplication syndrome	Disorder	33 Case(s)
447977	Progressive scapulohumeroperoneal distal myopathy	Disorder	33 Case(s)
411543	Severe phosphoribosylpyrophosphate synthetase superactivity	Subtype of disorder	33 Case(s)
3322	Hoyeraal-Hreidarsson syndrome	Disorder	33 Case(s)
3314	Thiemann disease, familial form	Disorder	33 Case(s)
329457	Distal arthrogryposis type 5D	Disorder	33 Case(s)
3102	Richieri Costa-Pereira syndrome	Disorder	33 Case(s)
300373	X-linked acrogigantism	Disorder	33 Case(s)
2795	Fowler urethral sphincter dysfunction syndrome	Disorder	33 Case(s)
2783	Autosomal dominant osteopetrosis type 1	Disorder	33 Case(s)
2406	Locked-in syndrome	Disorder	33 Case(s)
225123	TFR2-related hemochromatosis	Disorder	33 Case(s)
2170	Methylcobalamin deficiency type cblG	Subtype of disorder	33 Case(s)
1681	Diprosopus	Disorder	33 Case(s)
1388	Catel-Manzke syndrome	Disorder	33 Case(s)
123	Björnstad syndrome	Disorder	33 Case(s)
832	Succinyl-CoA:3-oxoacid CoA transferase deficiency	Disorder	32 Case(s)
67039	Segmental odontomaxillary dysplasia	Disorder	32 Case(s)
641353	Infantile neurodegeneration-progressive spasticity-intellectual disability-white matter lesions syndrome	Disorder	32 Case(s)
622925	X-linked severe syndromic thoracic aortic aneurysm and dissection	Disorder	32 Case(s)
617910	Conjunctival malignant melanoma	Disorder	32 Case(s)
572768	Microcephaly-micromelia syndrome	Subtype of disorder	32 Case(s)
458763	Retiform hemangioendothelioma	Disorder	32 Case(s)
412217	Dystonia-aphonia syndrome	Disorder	32 Case(s)
35664	ALDH18A1-related De Barsy syndrome	Subtype of disorder	32 Case(s)
324535	Combined oxidative phosphorylation defect type 11	Disorder	32 Case(s)
3163	SHORT syndrome	Disorder	32 Case(s)
314373	Chronic infantile diarrhea due to guanylate cyclase 2C overactivity	Disorder	32 Case(s)
293843	3MC syndrome	Disorder	32 Case(s)
217371	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	Disorder	32 Case(s)
141096	Supernumerary nostril	Disorder	32 Case(s)
99898	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency	Disorder	31 Case(s)

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96173	Ring chromosome 9 syndrome	Disorder	31 Case(s)
431255	Scapuloperoneal spinal muscular atrophy	Disorder	31 Case(s)
231573	Congenital erosive and vesicular dermatosis	Disorder	31 Case(s)
1747	Mosaic trisomy 7	Disorder	31 Case(s)
1711	Mosaic trisomy 17	Disorder	31 Case(s)
139485	Autosomal recessive ataxia due to ubiquinone deficiency	Disorder	31 Case(s)
99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K	Disorder	30 Case(s)
98970	Fleck corneal dystrophy	Disorder	30 Case(s)
98764	Spinocerebellar ataxia type 27A	Disorder	30 Case(s)
957	Acropectorovertebral dysplasia	Disorder	30 Case(s)
94065	15q24 microdeletion syndrome	Subtype of disorder	30 Case(s)
93940	Laryngotracheoesophageal cleft type 3	Subtype of disorder	30 Case(s)
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type	Disorder	30 Case(s)
93315	Spondylometaphyseal dysplasia, 'corner fracture' type	Disorder	30 Case(s)
91481	Ring dermoid of cornea	Disorder	30 Case(s)
91396	Isolated cryptophthalmia	Disorder	30 Case(s)
90045	Hereditary folate malabsorption	Disorder	30 Case(s)
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	Disorder	30 Case(s)
85277	X-linked intellectual disability, Cantagrel type	Disorder	30 Case(s)
85202	Keutel syndrome	Disorder	30 Case(s)
85164	Camptodactyly-tall stature-scoliosis-hearing loss syndrome	Disorder	30 Case(s)
79456	Diffuse cutaneous mastocytosis	Disorder	30 Case(s)
79292	Fish-eye disease	Subtype of disorder	30 Case(s)
79157	2-methylbutyryl-CoA dehydrogenase deficiency	Disorder	30 Case(s)
79155	Hydroxykynureninuria	Disorder	30 Case(s)
77298	Anophthalmia/microphthalmia-esophageal atresia syndrome	Disorder	30 Case(s)
715	Glycogen storage disease due to muscle phosphorylase kinase deficiency	Disorder	30 Case(s)
66628	Obesity due to congenital leptin deficiency	Subtype of disorder	30 Case(s)
642099	Spondyloepimetaphyseal dysplasia with joint laxity, Beighton type	Disorder	30 Case(s)
477650	Fibroblastic rheumatism	Disorder	30 Case(s)
458768	Papillary intralymphatic angioendothelioma	Disorder	30 Case(s)
397709	Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	Disorder	30 Case(s)
34592	Immunodeficiency by defective expression of MHC class I	Disorder	30 Case(s)
3352	Tricho-dento-osseous syndrome	Disorder	30 Case(s)
3266	Isolated humero-radio-ulnar synostosis	Disorder	30 Case(s)
3258	Cenani-Lenz syndrome	Disorder	30 Case(s)
3005	Pyle disease	Disorder	30 Case(s)
29	Mevalonic aciduria	Subtype of disorder	30 Case(s)
2849	Perlman syndrome	Disorder	30 Case(s)
2834	Wrinkly skin syndrome	Subtype of disorder	30 Case(s)
2763	Osteocraniostenosis	Disorder	30 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
275523	Dianzani autoimmune lymphoproliferative disease	Disorder	30 Case(s)
2746	Opsismodysplasia	Disorder	30 Case(s)
2733	Omodysplasia	Disorder	30 Case(s)
2728	Blepharophimosis-intellectual disability syndrome, Ohdo type	Disorder	30 Case(s)
2721	Odonto-onycho-dermal dysplasia	Disorder	30 Case(s)
246	Postaxial acrofacial dysostosis	Disorder	30 Case(s)
238446	15q11q13 microduplication syndrome	Disorder	30 Case(s)
228415	5q35 microduplication syndrome	Disorder	30 Case(s)
228236	Linear focal elastosis	Disorder	30 Case(s)
228116	Hughes-Stovin syndrome	Disorder	30 Case(s)
220295	Xeroderma pigmentosum-Cockayne syndrome complex	Disorder	30 Case(s)
209943	IRVAN syndrome	Disorder	30 Case(s)
209370	Severe neonatal-onset encephalopathy with microcephaly	Disorder	30 Case(s)
2063	Splenogonadal fusion-limb defects-micrognathia syndrome	Disorder	30 Case(s)
2036	Scalp-ear-nipple syndrome	Disorder	30 Case(s)
178345	Aromatase excess syndrome	Disorder	30 Case(s)
1752	Trisomy 8q	Disorder	30 Case(s)
1662	Restrictive dermopathy	Disorder	30 Case(s)
1596	Distal deletion 15q	Disorder	30 Case(s)
1545	Crisponi syndrome	Disorder	30 Case(s)
1525	Cranio-osteoarthropathy	Disorder	30 Case(s)
1427	Otospondylomegaepiphyseal dysplasia	Disorder	30 Case(s)
141163	Glossopalatine ankylosis	Disorder	30 Case(s)
140933	Linear atrophoderma of Moulin	Disorder	30 Case(s)
139552	Distal hereditary motor neuropathy, Jerash type	Disorder	30 Case(s)
137834	Frank-Ter Haar syndrome	Disorder	30 Case(s)
1314	Symmetrical thalamic calcifications	Disorder	30 Case(s)
1229	Congenital intrauterine infection-like syndrome	Disorder	30 Case(s)
572773	Microcephaly-short stature-limb abnormalities syndrome	Subtype of disorder	29 Case(s)
466775	Autosomal recessive Charcot-Marie-Tooth disease type 2X	Disorder	29 Case(s)
3255	Filippi syndrome	Disorder	29 Case(s)
2753	Orofaciodigital syndrome type 4	Disorder	29 Case(s)
2460	Van den Ende-Gupta syndrome	Disorder	29 Case(s)
139444	Leukoencephalopathy with bilateral anterior temporal lobe cysts	Disorder	29 Case(s)
1186	Infantile-onset spinocerebellar ataxia	Disorder	29 Case(s)
99812	LIG4 syndrome	Disorder	28 Case(s)
85173	IMAGe syndrome	Disorder	28 Case(s)
79124	Hepatic veno-occlusive disease-immunodeficiency syndrome	Disorder	28 Case(s)
521258	Xq25 microduplication syndrome	Disorder	28 Case(s)
50814	Craniolenticulosutural dysplasia	Disorder	28 Case(s)
468631	Microcephalic cortical malformations-short stature due to RTTN deficiency	Disorder	28 Case(s)
457077	TAFRO syndrome	Disorder	28 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
398069	MAGEL2-related Prader-Willi-like syndrome	Disorder	28 Case(s)
391392	Familial episodic pain syndrome with predominantly lower limb involvement	Subtype of disorder	28 Case(s)
3459	Wilson-Turner syndrome	Disorder	28 Case(s)
34528	Autosomal dominant primary hypomagnesemia with hypocalciuria	Disorder	28 Case(s)
329466	Autosomal dominant focal dystonia, DYT25 type	Disorder	28 Case(s)
314022	Gastric adenocarcinoma and proximal polyposis of the stomach	Disorder	28 Case(s)
276193	Spinocerebellar ataxia type 35	Disorder	28 Case(s)
228174	Autosomal dominant Charcot-Marie-Tooth disease type 2N	Disorder	28 Case(s)
2220	Hypertrichosis cubiti	Disorder	28 Case(s)
163681	CNTNAP2-related developmental and epileptic encephalopathy	Disorder	28 Case(s)
139547	Distal spinal muscular atrophy type 3	Disorder	28 Case(s)
96078	16p13.3 microduplication syndrome	Disorder	27 Case(s)
95434	Autosomal recessive cerebellar ataxia-movement disorder syndrome	Disorder	27 Case(s)
93358	Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome	Disorder	27 Case(s)
592574	Menke-Hennekam syndrome	Disorder	27 Case(s)
586130	Sporadic fatal insomnia	Disorder	27 Case(s)
466	Fatal familial insomnia	Disorder	27 Case(s)
329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement	Disorder	27 Case(s)
319635	Amyloidosis cutis dyschromia	Disorder	27 Case(s)
313808	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	Disorder	27 Case(s)
280133	Complement component 3 deficiency	Disorder	27 Case(s)
2623	Geleophysic dysplasia	Disorder	27 Case(s)
261250	16q24.3 microdeletion syndrome	Disorder	27 Case(s)
251287	Benign concentric annular macular dystrophy	Disorder	27 Case(s)
2169	Methylcobalamin deficiency type cblE	Subtype of disorder	27 Case(s)
1040	Metaphyseal anadysplasia	Disorder	27 Case(s)
100993	Autosomal dominant spastic paraplegia type 12	Disorder	27 Case(s)
98771	Spinocerebellar ataxia type 18	Disorder	26 Case(s)
52994	Orbital leiomyoma	Disorder	26 Case(s)
488613	Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome	Disorder	26 Case(s)
40366	Acitretin/etretinate embryopathy	Disorder	26 Case(s)
357332	Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome	Disorder	26 Case(s)
2574	Moynahan syndrome	Disorder	26 Case(s)
199343	EAST syndrome	Disorder	26 Case(s)
1974	Autosomal recessive faciodigitogenital syndrome	Disorder	26 Case(s)
1262	Böök syndrome	Disorder	26 Case(s)
93109	Congenital megacalycosis	Disorder	25 Case(s)

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85203	Acropectoral syndrome	Disorder	25 Case(s)
79319	MPI-CDG	Disorder	25 Case(s)
7	3C syndrome	Disorder	25 Case(s)
637061	Isolated optic nerve hypoplasia	Disorder	25 Case(s)
56305	Atelosteogenesis type III	Disorder	25 Case(s)
56304	Atelosteogenesis type II	Disorder	25 Case(s)
54028	Plummer-Vinson syndrome	Disorder	25 Case(s)
527497	NKX6-2-related autosomal recessive hypomyelinating leukodystrophy	Disorder	25 Case(s)
50944	Schöpf-Schulz-Passarge syndrome	Disorder	25 Case(s)
488632	TBCK-related intellectual disability syndrome	Disorder	25 Case(s)
458803	Spinocerebellar ataxia type 42	Disorder	25 Case(s)
453499	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome	Disorder	25 Case(s)
397941	MAN1B1-CDG	Disorder	25 Case(s)
39041	Omenn syndrome	Disorder	25 Case(s)
3472	Yunis-Varon syndrome	Disorder	25 Case(s)
314597	Chudley-McCullough syndrome	Disorder	25 Case(s)
281122	Self-improving collodion baby	Disorder	25 Case(s)
268249	Mycophenolate mofetil embryopathy	Disorder	25 Case(s)
251019	2q32q33 microdeletion syndrome	Disorder	25 Case(s)
2499	Metachondromatosis	Disorder	25 Case(s)
230	Dopamine beta-hydroxylase deficiency	Disorder	25 Case(s)
1715	Trisomy 18p	Disorder	25 Case(s)
1519	SPECC1L-related hypertelorism syndrome	Disorder	25 Case(s)
1448	Ring chromosome 6 syndrome	Disorder	25 Case(s)
98972	Central cloudy dystrophy of François	Disorder	24 Case(s)
97234	Glycogen storage disease due to phosphoglycerate mutase deficiency	Disorder	24 Case(s)
66629	Goldberg-Shprintzen megacolon syndrome	Disorder	24 Case(s)
488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W	Disorder	24 Case(s)
487809	Pediatric collagenous gastritis	Disorder	24 Case(s)
480864	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome	Disorder	24 Case(s)
438216	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation	Subtype of disorder	24 Case(s)
438213	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	Disorder	24 Case(s)
401973	MEND syndrome	Disorder	24 Case(s)
399096	Distal anoctaminopathy	Disorder	24 Case(s)
313846	Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome	Disorder	24 Case(s)
300525	Pseudohypoaldosteronism type 2D	Subtype of disorder	24 Case(s)
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	Disorder	24 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
251383	CK syndrome	Disorder	24 Case(s)
247262	Hyperphosphatasia-intellectual disability syndrome	Disorder	24 Case(s)
209902	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	Disorder	24 Case(s)
2069	Gastrocutaneous syndrome	Disorder	24 Case(s)
183713	Bacterial susceptibility due to TLR signaling pathway deficiency	Disorder	24 Case(s)
171607	X-linked spastic paraplegia type 34	Disorder	24 Case(s)
1490	Corneal dystrophy-perceptive deafness syndrome	Disorder	24 Case(s)
1361	Carnosinase deficiency	Disorder	24 Case(s)
1234	Bartsocas-Papas syndrome	Disorder	24 Case(s)
99901	Acyl-CoA dehydrogenase 9 deficiency	Disorder	23 Case(s)
93329	Autosomal recessive omodysplasia	Subtype of disorder	23 Case(s)
597939	Euthyroid dysprealbuminemic hyperthyroxinemia	Disorder	23 Case(s)
569821	Congenital primary lymphedema of Gordon	Disorder	23 Case(s)
538574	Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome	Disorder	23 Case(s)
477817	PMP22-RAI1 contiguous gene duplication syndrome	Disorder	23 Case(s)
445018	Combined immunodeficiency due to LRBA deficiency	Disorder	23 Case(s)
411493	Pontocerebellar hypoplasia type 10	Disorder	23 Case(s)
364198	Bipartite talus	Disorder	23 Case(s)
314588	Distal triplication 15q	Subtype of disorder	23 Case(s)
261652	Kleefstra syndrome due to a point mutation	Subtype of disorder	23 Case(s)
238475	Familial hypercholanemia	Disorder	23 Case(s)
1782	Dysosteosclerosis	Disorder	23 Case(s)
1617	Developmental delay-language impairment-dopa responsive dystonia-parkinsonism syndrome due to 2q24 microdeletion	Subtype of disorder	23 Case(s)
157973	Congenital muscular dystrophy due to LMNA mutation	Disorder	23 Case(s)
101028	Transaldolase deficiency	Disorder	23 Case(s)
98805	Primary dystonia, DYT4 type	Disorder	22 Case(s)
94063	12q14 microdeletion syndrome	Disorder	22 Case(s)
93953	Familial thyroglossal duct cyst	Disorder	22 Case(s)
91387	Familial thoracic aortic aneurysm and aortic dissection	Disorder	22 Case(s)
85282	MEHMO syndrome	Disorder	22 Case(s)
85201	Genitopatellar syndrome	Disorder	22 Case(s)
85191	Singleton-Merten dysplasia	Disorder	22 Case(s)
79499	Autosomal dominant deafness-onychodystrophy syndrome	Disorder	22 Case(s)
71271	Split hand-split foot-deafness syndrome	Disorder	22 Case(s)
542306	GNB5-related intellectual disability-cardiac arrhythmia syndrome	Disorder	22 Case(s)
528105	Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome	Disorder	22 Case(s)
466943	WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	Disorder	22 Case(s)
464738	Basel-Vanagaite-Smirin-Yosef syndrome	Disorder	22 Case(s)
445038	3-methylglutaconic aciduria-neonatal cataract-neurologic	Disorder	22 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	involvement-congenital neutropenia syndrome		
431272	X-linked scapuloperoneal muscular dystrophy	Disorder	22 Case(s)
398173	Focal facial dermal dysplasia type II	Subtype of disorder	22 Case(s)
363677	Childhood-onset autosomal recessive myopathy with external ophthalmoplegia	Disorder	22 Case(s)
329195	Developmental delay with autism spectrum disorder and gait instability	Disorder	22 Case(s)
314718	Lethal arteriopathy syndrome due to fibulin-4 deficiency	Disorder	22 Case(s)
269229	Pontine tegmental cap dysplasia	Disorder	22 Case(s)
2492	FATCO syndrome	Disorder	22 Case(s)
247820	Ectodermal dysplasia-pili torti-cutaneous syndactyly syndrome	Disorder	22 Case(s)
228423	GATA2 deficiency spectrum	Disorder	22 Case(s)
209908	Isolated childhood apraxia of speech	Disorder	22 Case(s)
1827	Acromelic frontonasal dysplasia	Disorder	22 Case(s)
1723	Mosaic trisomy 2	Disorder	22 Case(s)
168612	Congenital deficiency in alpha-fetoprotein	Disorder	22 Case(s)
163690	Hypotonia-cystinuria syndrome	Disorder	22 Case(s)
93606	Nephrogenic syndrome of inappropriate antidiuresis	Disorder	21 Case(s)
93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	Disorder	21 Case(s)
79091	Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome	Disorder	21 Case(s)
69082	Odonto-tricho-ungual-digito-palmar syndrome	Disorder	21 Case(s)
66625	Cerebrooculonasal syndrome	Disorder	21 Case(s)
542301	Combined immunodeficiency due to CARMIL2 deficiency	Disorder	21 Case(s)
482077	HTRA1-related autosomal dominant cerebral small vessel disease	Disorder	21 Case(s)
468672	Colobomatous macrophthalmia-microcornea syndrome	Disorder	21 Case(s)
466768	Autosomal dominant Charcot-Marie-Tooth disease type 2Z	Disorder	21 Case(s)
447964	Autosomal dominant Charcot-Marie-Tooth disease type 2V	Disorder	21 Case(s)
402003	Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering	Disorder	21 Case(s)
401869	Multiple mitochondrial dysfunctions syndrome type 1	Disorder	21 Case(s)
398189	Focal facial dermal dysplasia type IV	Subtype of disorder	21 Case(s)
391389	Familial episodic pain syndrome with predominantly upper body involvement	Subtype of disorder	21 Case(s)
363649	Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome	Disorder	21 Case(s)
324972	MAGIC syndrome	Disorder	21 Case(s)
3063	X-linked intellectual disability, Snyder type	Disorder	21 Case(s)
221145	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies	Disorder	21 Case(s)
199326	Isolated autosomal dominant hypomagnesemia, Glaudemans type	Disorder	21 Case(s)
171881	Cap myopathy	Disorder	21 Case(s)
168593	Sudden infant death-dysgenesis of the testes syndrome	Disorder	21 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
1578	Pterin-4 alpha-carbinolamine dehydratase deficiency	Subtype of disorder	21 Case(s)
98791	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	Disorder	20 Case(s)
98768	Spinocerebellar ataxia type 13	Disorder	20 Case(s)
97232	Fingerprint body myopathy	Disorder	20 Case(s)
971	Acrorenal syndrome	Disorder	20 Case(s)
96175	Ring chromosome 11 syndrome	Disorder	20 Case(s)
93941	Laryngotracheoesophageal cleft type 4	Subtype of disorder	20 Case(s)
88642	Congenital insensitivity to pain-anosmia-neuropathic arthropathy	Disorder	20 Case(s)
88639	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	Disorder	20 Case(s)
88628	Posterior column ataxia-retinitis pigmentosa syndrome	Disorder	20 Case(s)
86920	Dermatopathia pigmentosa reticularis	Disorder	20 Case(s)
86919	Keratosis palmaris et plantaris-clinodactyly syndrome	Disorder	20 Case(s)
86797	Atypical lichen myxedematosus	Disorder	20 Case(s)
85192	Calvarial doughnut lesions-bone fragility syndrome	Disorder	20 Case(s)
83616	Rubella panencephalitis	Disorder	20 Case(s)
79476	Griscelli syndrome type 1	Subtype of disorder	20 Case(s)
79154	2-aminoadipic 2-oxoadipic aciduria	Disorder	20 Case(s)
79084	Familial partial lipodystrophy, Köbberling type	Disorder	20 Case(s)
73271	Bleeding diathesis due to a collagen receptor defect	Disorder	20 Case(s)
71289	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome	Disorder	20 Case(s)
69723	Tyrosinemia type 3	Disorder	20 Case(s)
69084	Pure hair and nail ectodermal dysplasia	Disorder	20 Case(s)
67046	3-methylglutaconic aciduria type 1	Disorder	20 Case(s)
641380	PAPASH syndrome	Disorder	20 Case(s)
63442	Angel-shaped phalango-epiphyseal dysplasia	Disorder	20 Case(s)
599376	Hypomyelination of early myelinating structures	Disorder	20 Case(s)
53583	Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	Disorder	20 Case(s)
457240	X-linked intellectual disability-short stature-overweight syndrome	Disorder	20 Case(s)
455	Superficial epidermolytic ichthyosis	Disorder	20 Case(s)
448242	Autosomal recessive brachyolmia	Disorder	20 Case(s)
443811	PGM3-CDG	Disorder	20 Case(s)
420179	Malan overgrowth syndrome	Disorder	20 Case(s)
391376	Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome	Disorder	20 Case(s)
369897	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	Disorder	20 Case(s)
3387	Isolated anterior cervical hypertrichosis	Disorder	20 Case(s)
33445	Neuroectodermal melanolysosomal disease	Disorder	20 Case(s)
3137	Alpha-N-acetylgalactosaminidase deficiency	Disorder	20 Case(s)
309854	Cirrhosis-dystonia-polycythemia-hypermanganesemia	Disorder	20 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	syndrome		
3021	RAPADILINO syndrome	Disorder	20 Case(s)
30	Hereditary orotic aciduria	Disorder	20 Case(s)
289863	Atypical glycine encephalopathy	Subtype of disorder	20 Case(s)
2847	Pericardial and diaphragmatic defect	Disorder	20 Case(s)
280779	Cutaneous collagenous vasculopathy	Disorder	20 Case(s)
2755	Orofaciodigital syndrome type 8	Disorder	20 Case(s)
2751	Orofaciodigital syndrome type 2	Disorder	20 Case(s)
2717	Oculotrichoanal syndrome	Disorder	20 Case(s)
268114	RAS-associated autoimmune leukoproliferative disease	Disorder	20 Case(s)
26137	Juvenile temporal arteritis	Disorder	20 Case(s)
251393	Localized junctional epidermolysis bullosa	Disorder	20 Case(s)
251061	7q31 microdeletion syndrome	Disorder	20 Case(s)
251028	SATB2-associated syndrome due to a chromosomal rearrangement	Subtype of disorder	20 Case(s)
247522	Primary ciliary dyskinesia-retinitis pigmentosa syndrome	Disorder	20 Case(s)
2394	Pyruvate dehydrogenase E3 deficiency	Subtype of disorder	20 Case(s)
2375	Laryngeal abductor paralysis-intellectual disability syndrome	Disorder	20 Case(s)
228247	Acquired pseudoxanthoma elasticum	Disorder	20 Case(s)
228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M	Disorder	20 Case(s)
2021	Fibrochondrogenesis	Disorder	20 Case(s)
1807	Focal facial dermal dysplasia type III	Subtype of disorder	20 Case(s)
178364	Syndromic microphthalmia type 5	Disorder	20 Case(s)
1513	Craniodiaphyseal dysplasia	Disorder	20 Case(s)
1466	COFS syndrome	Subtype of disorder	20 Case(s)
1447	Ring chromosome 4 syndrome	Disorder	20 Case(s)
139455	Autosomal recessive bestrophinopathy	Disorder	20 Case(s)
1394	Cerebrofaciothoracic dysplasia	Disorder	20 Case(s)
1387	Cataract-intellectual disability-hypogonadism syndrome	Disorder	20 Case(s)
1358	Carey-Fineman-Ziter syndrome	Disorder	20 Case(s)
1134	Isolated arrhinia	Disorder	20 Case(s)
101110	Spinocerebellar ataxia type 20	Disorder	20 Case(s)
100976	Bathing suit ichthyosis	Disorder	20 Case(s)
100043	Autosomal dominant intermediate Charcot-Marie-Tooth disease type A	Disorder	20 Case(s)
935	Short-limb skeletal dysplasia with severe combined immunodeficiency	Disorder	19 Case(s)
89838	Autosomal recessive generalized epidermolysis bullosa simplex	Disorder	19 Case(s)
75857	6q terminal deletion syndrome	Disorder	19 Case(s)
597623	IRF2BPL-related regressive neurodevelopmental disorder-dystonia-seizures syndrome	Disorder	19 Case(s)
529962	17q24.2 microdeletion syndrome	Disorder	19 Case(s)
505248	Mucopolysaccharidosis-like syndrome with congenital heart defects and hematopoietic disorders	Disorder	19 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
497757	MME-related autosomal dominant Charcot Marie Tooth disease type 2	Disorder	19 Case(s)
494433	MIRAGE syndrome	Disorder	19 Case(s)
482601	Adenylosuccinate synthetase-like 1-related distal myopathy	Disorder	19 Case(s)
466962	SMARCA4-deficient sarcoma of thorax	Disorder	19 Case(s)
438159	STAT3-related early-onset multisystem autoimmune disease	Disorder	19 Case(s)
43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency	Disorder	19 Case(s)
397946	Autosomal spastic paraplegia type 58	Disorder	19 Case(s)
391320	East Texas bleeding disorder	Subtype of disorder	19 Case(s)
352662	Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome	Disorder	19 Case(s)
3339	Oculoectodermal syndrome	Disorder	19 Case(s)
3145	Arginine vasopressin resistance-intracranial calcification-short stature-facial dysmorphism syndrome	Disorder	19 Case(s)
284169	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion	Subtype of disorder	19 Case(s)
280671	Megaconial congenital muscular dystrophy	Disorder	19 Case(s)
2707	Oculocerebrofacial syndrome, Kaufman type	Disorder	19 Case(s)
268261	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion	Subtype of disorder	19 Case(s)
251046	6p22 microdeletion syndrome	Disorder	19 Case(s)
247868	NLRP12-associated hereditary periodic fever syndrome	Disorder	19 Case(s)
2399	Nasopalpebral lipoma-coloboma syndrome	Disorder	19 Case(s)
228410	Cardiac anomalies-short stature-joint hypermobility-facial dysmorphism syndrome	Disorder	19 Case(s)
228387	Spondylo-megaepiphyseal-metaphyseal dysplasia	Disorder	19 Case(s)
178487	Adult intestinal botulism	Subtype of disorder	19 Case(s)
171848	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	Disorder	19 Case(s)
139447	Progressive cavitating leukoencephalopathy	Disorder	19 Case(s)
139441	Hypomyelination with atrophy of basal ganglia and cerebellum	Disorder	19 Case(s)
99741	King-Denborough syndrome	Disorder	18 Case(s)
96171	Ring chromosome 2 syndrome	Disorder	18 Case(s)
86309	DPAGT1-CDG	Disorder	18 Case(s)
85167	Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	Disorder	18 Case(s)
66637	Diaphanospondylodysostosis	Disorder	18 Case(s)
643549	Hao-Fountain syndrome	Disorder	18 Case(s)
637051	Borna virus encephalitis	Disorder	18 Case(s)
613274	Pontocerebellar hypoplasia type 14	Disorder	18 Case(s)
589856	Choanal atresia-athelia-hypothyroidism-delayed puberty-short stature syndrome	Disorder	18 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
508498	Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome	Disorder	18 Case(s)
481152	PYCR2-related microcephaly-progressive leukoencephalopathy	Disorder	18 Case(s)
468641	Chronic enteropathy associated with SLCO2A1 gene	Disorder	18 Case(s)
370046	Didymosis aplasticosebacea	Disorder	18 Case(s)
369962	Methylmalonic acidemia with homocystinuria, type cblX	Subtype of disorder	18 Case(s)
363417	Temtamy preaxial brachydactyly syndrome	Disorder	18 Case(s)
324588	Familial dyskinesia and facial myokymia	Disorder	18 Case(s)
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	Disorder	18 Case(s)
300319	Charcot-Marie-Tooth disease type 2P	Disorder	18 Case(s)
261344	Trisomy 1q	Disorder	18 Case(s)
251523	Hyperzincemia and hypercalprotectinemia	Disorder	18 Case(s)
2501	Metaphyseal chondrodysplasia, Spahr type	Disorder	18 Case(s)
238505	Combined immunodeficiency due to CD27 deficiency	Disorder	18 Case(s)
2353	Schilbach-Rott syndrome	Disorder	18 Case(s)
228402	2q23.1 microdeletion syndrome	Disorder	18 Case(s)
171719	Cutis laxa-Marfanoid syndrome	Disorder	18 Case(s)
158025	Hereditary progressive mucinous histiocytosis	Disorder	18 Case(s)
1449	Ring chromosome 7 syndrome	Disorder	18 Case(s)
1441	Ring chromosome 17 syndrome	Disorder	18 Case(s)
139515	Charcot-Marie-Tooth disease type 4J	Disorder	18 Case(s)
1272	Aymé-Gripp syndrome	Disorder	18 Case(s)
99853	Ovarioleukodystrophy	Subtype of disorder	17 Case(s)
93282	Spondyloepimetaphyseal dysplasia, PAPSS2 type	Disorder	17 Case(s)
91131	DK1-CDG	Disorder	17 Case(s)
79283	Methylmalonic acidemia with homocystinuria, type cblD	Subtype of disorder	17 Case(s)
69744	Circumscribed palmoplantar hypokeratosis	Disorder	17 Case(s)
633004	KDM3B-related intellectual disability-facial dysmorphism-short stature syndrome	Disorder	17 Case(s)
631085	Autosomal recessive spastic paraplegia type 86	Disorder	17 Case(s)
560	Marshall syndrome	Disorder	17 Case(s)
543470	Optic atrophy-ataxia-peripheral neuropathy-global developmental delay syndrome	Disorder	17 Case(s)
502434	STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome	Disorder	17 Case(s)
500533	Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	Disorder	17 Case(s)
480880	X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	Disorder	17 Case(s)
477673	Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	Disorder	17 Case(s)
464760	Familial cavitary optic disc anomaly	Disorder	17 Case(s)
436159	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency	Disorder	17 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
435988	Chronic atrial and intestinal dysrhythmia syndrome	Disorder	17 Case(s)
404443	Tatton-Brown-Rahman syndrome	Disorder	17 Case(s)
399058	Alpha-B crystallin-related late-onset myopathy	Disorder	17 Case(s)
363429	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome	Disorder	17 Case(s)
353298	Roifman syndrome	Disorder	17 Case(s)
3350	Tremor-nystagmus-duodenal ulcer syndrome	Disorder	17 Case(s)
324381	Hereditary inclusion body myopathy type 4	Disorder	17 Case(s)
3204	Stormorken-Sjaastad-Langslet syndrome	Disorder	17 Case(s)
319595	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency	Disorder	17 Case(s)
300530	Pseudohypoaldosteronism type 2E	Subtype of disorder	17 Case(s)
2318	Joubert syndrome with oculorenal defect	Disorder	17 Case(s)
230839	Classical-like Ehlers-Danlos syndrome type 1	Disorder	17 Case(s)
227976	Autosomal recessive optic atrophy, OPA7 type	Disorder	17 Case(s)
210141	Inherited congenital spastic tetraplegia	Disorder	17 Case(s)
210115	Sterile multifocal osteomyelitis with periostitis and pustulosis	Disorder	17 Case(s)
1954	Congenital lethal erythroderma	Disorder	17 Case(s)
1908	Aminopterin/methotrexate embryofetopathy	Disorder	17 Case(s)
1325	Camptodactyly-taurinuria syndrome	Disorder	17 Case(s)
1104	Anophthalmia plus syndrome	Disorder	17 Case(s)
93357	SPONASTRIME dysplasia	Disorder	16 Case(s)
920	Ablepharon macrostomia syndrome	Disorder	16 Case(s)
85198	Dysspondyloenchondromatosis	Disorder	16 Case(s)
71528	Obesity due to prohormone convertase I deficiency	Subtype of disorder	16 Case(s)
69737	Bosley-Salih-Alorainy syndrome	Disorder	16 Case(s)
641385	PASS syndrome	Disorder	16 Case(s)
63273	Distal myopathy with posterior leg and anterior hand involvement	Disorder	16 Case(s)
631076	Autosomal recessive spastic paraplegia type 83	Disorder	16 Case(s)
53296	Familial cutaneous collagenoma	Disorder	16 Case(s)
488191	Female infertility due to oocyte meiotic arrest	Disorder	16 Case(s)
464282	Spastic paraplegia-severe developmental delay-epilepsy syndrome	Disorder	16 Case(s)
457279	Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome	Disorder	16 Case(s)
397606	PrP systemic amyloidosis	Disorder	16 Case(s)
369861	Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome	Disorder	16 Case(s)
369852	Congenital neutropenia-myelofibrosis-nephromegaly syndrome	Disorder	16 Case(s)
34514	Telethonin-related limb-girdle muscular dystrophy R7	Disorder	16 Case(s)
33067	Metaphyseal chondrodysplasia, Jansen type	Disorder	16 Case(s)
319524	Combined oxidative phosphorylation defect type 15	Disorder	16 Case(s)
319171	Distal 17p13.1 microdeletion syndrome	Disorder	16 Case(s)
314566	Primary progressive apraxia of speech	Disorder	16 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
314376	Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency	Disorder	16 Case(s)
306734	Primary dystonia, DYT21 type	Disorder	16 Case(s)
306674	Kufor-Rakeb syndrome	Disorder	16 Case(s)
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	Disorder	16 Case(s)
289601	Hereditary arterial and articular multiple calcification syndrome	Disorder	16 Case(s)
279943	Hereditary neutrophilia	Disorder	16 Case(s)
261257	Distal 17p13.3 microdeletion syndrome	Disorder	16 Case(s)
2538	Microgastria-limb reduction defect syndrome	Disorder	16 Case(s)
238455	Infantile dystonia-parkinsonism	Disorder	16 Case(s)
2102	GTP cyclohydrolase I deficiency	Subtype of disorder	16 Case(s)
2089	Glycogen storage disease due to hepatic glycogen synthase deficiency	Disorder	16 Case(s)
178355	Smith-McCort dysplasia	Disorder	16 Case(s)
1438	Ring chromosome 10 syndrome	Disorder	16 Case(s)
1231	Barber-Say syndrome	Disorder	16 Case(s)
1195	Congenital atransferrinemia	Disorder	16 Case(s)
99954	Charcot-Marie-Tooth disease type 4H	Disorder	15 Case(s)
98949	Complete cryptophthalmia	Subtype of disorder	15 Case(s)
96181	Maternal uniparental disomy of chromosome 6	Disorder	15 Case(s)
90796	46,XY difference of sex development due to isolated 17,20-lyase deficiency	Disorder	15 Case(s)
90400	Scleromyxedema without monoclonal gammopathy	Subtype of disorder	15 Case(s)
88620	Isolated congenital anosmia	Disorder	15 Case(s)
88618	S-adenosylhomocysteine hydrolase deficiency	Disorder	15 Case(s)
85146	Neurogenic scapuloperoneal syndrome, Kaeser type	Disorder	15 Case(s)
79351	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form	Subtype of disorder	15 Case(s)
79325	ALG8-CDG	Disorder	15 Case(s)
79321	ALG3-CDG	Disorder	15 Case(s)
79284	Methylmalonic acidemia with homocystinuria type cblF	Subtype of disorder	15 Case(s)
79149	Dermochondrocorneal dystrophy	Disorder	15 Case(s)
69063	Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	Disorder	15 Case(s)
631248	Mitchell Syndrome	Disorder	15 Case(s)
619363	NOCARH syndrome	Disorder	15 Case(s)
599507	Acquired factor XI deficiency	Disorder	15 Case(s)
570371	Bartter syndrome type 5	Subtype of disorder	15 Case(s)
53696	Arthrogryposis-anterior horn cell disease syndrome	Disorder	15 Case(s)
527276	Encephalopathy due to mitochondrial and peroxisomal fission defect	Disorder	15 Case(s)
521426	PLAA-associated neurodevelopmental disorder	Disorder	15 Case(s)
513456	Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome	Disorder	15 Case(s)
482606	X-linked keloid scarring-reduced joint mobility-increased	Disorder	15 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	optic cup-to-disc ratio syndrome		
457050	Autosomal dominant mitochondrial myopathy with exercise intolerance	Disorder	15 Case(s)
456369	Polyglucosan body myopathy type 2	Disorder	15 Case(s)
447997	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	Disorder	15 Case(s)
436169	Thrombomodulin-related bleeding disorder	Disorder	15 Case(s)
436144	Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	Disorder	15 Case(s)
401768	Proximal myopathy with extrapyramidal signs	Disorder	15 Case(s)
397744	Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome	Disorder	15 Case(s)
397615	Obesity due to CEP19 deficiency	Subtype of disorder	15 Case(s)
329324	Inverse Klippel-Trénaunay syndrome	Disorder	15 Case(s)
314647	Non-progressive cerebellar ataxia with intellectual disability	Disorder	15 Case(s)
314432	Spigelian hernia-cryptorchidism syndrome	Disorder	15 Case(s)
280763	Severe intellectual disability and progressive spastic paraplegia	Disorder	15 Case(s)
280633	Multiple congenital anomalies-hypotonia-seizures syndrome	Disorder	15 Case(s)
250984	Autosomal recessive Stickler syndrome	Subtype of disorder	15 Case(s)
221043	Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome	Disorder	15 Case(s)
2075	Genitopalatocardiac syndrome	Disorder	15 Case(s)
1901	Dermatosparaxis Ehlers-Danlos syndrome	Disorder	15 Case(s)
171680	Lissencephaly due to TUBA1A mutation	Disorder	15 Case(s)
137754	Aminoacylase 1 deficiency	Disorder	15 Case(s)
93356	Spondyloepimetaphyseal dysplasia, Missouri type	Disorder	14 Case(s)
90390	Anonychia-onychodystrophy syndrome	Subtype of disorder	14 Case(s)
88659	Autosomal dominant progressive nephropathy with hypertension	Disorder	14 Case(s)
75378	Oligocone trichromacy	Disorder	14 Case(s)
589515	PUM1-associated developmental disability-ataxia-seizure syndrome	Disorder	14 Case(s)
562528	Congenital limbs-face contractures-hypotonia-developmental delay syndrome	Disorder	14 Case(s)
508529	Intermediate epidermolysis bullosa simplex with cardiomyopathy	Disorder	14 Case(s)
480907	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	Disorder	14 Case(s)
480483	Progressive familial intrahepatic cholestasis type 4	Subtype of disorder	14 Case(s)
466718	Martinique crinkled retinal pigment epitheliopathy	Disorder	14 Case(s)
457351	Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome	Disorder	14 Case(s)
423384	Severe congenital neutropenia due to JAGN1 deficiency	Disorder	14 Case(s)
401849	Autosomal spastic paraplegia type 72	Disorder	14 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
397758	Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies	Disorder	14 Case(s)
371007	Congenital muscular dystrophy with hyperlaxity	Disorder	14 Case(s)
369970	Microcornea-myopic chorioretinal atrophy-telecanthus syndrome	Disorder	14 Case(s)
369920	Pontocerebellar hypoplasia type 9	Disorder	14 Case(s)
364028	X-linked intellectual disability due to GRIA3 mutations	Disorder	14 Case(s)
36355	Bleeding disorder due to P2Y12 defect	Disorder	14 Case(s)
3363	Trichomegaly-retina pigmentary degeneration-dwarfism syndrome	Disorder	14 Case(s)
320375	Autosomal recessive spastic paraplegia type 55	Disorder	14 Case(s)
314394	Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome	Disorder	14 Case(s)
314051	Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome	Disorder	14 Case(s)
313892	Developmental and speech delay due to SOX5 deficiency	Subtype of disorder	14 Case(s)
307766	Curly hair-acral keratoderma-caries syndrome	Disorder	14 Case(s)
284289	Adult-onset autosomal recessive cerebellar ataxia	Disorder	14 Case(s)
284139	Larsen-like syndrome, B3GAT3 type	Disorder	14 Case(s)
2789	Lateral meningocele syndrome	Disorder	14 Case(s)
2719	Oculocerebral hypopigmentation syndrome, Cross type	Disorder	14 Case(s)
261323	21q22.11q22.12 microdeletion syndrome	Disorder	14 Case(s)
2435	Hypo- and hypermelanotic cutaneous macules-retarded growth-intellectual disability syndrome	Disorder	14 Case(s)
238750	4q21 microdeletion syndrome	Disorder	14 Case(s)
2378	Laurin-Sandrow syndrome	Disorder	14 Case(s)
199351	Adult-onset dystonia-parkinsonism	Disorder	14 Case(s)
1791	Frontofacionasal dysplasia	Disorder	14 Case(s)
168796	Heart-hand syndrome, Slovenian type	Disorder	14 Case(s)
1516	Non-syndromic bilambdoid and sagittal craniosynostosis	Disorder	14 Case(s)
139578	Mutilating hereditary sensory neuropathy with spastic paraplegia	Disorder	14 Case(s)
137783	Lethal congenital contracture syndrome type 3	Disorder	14 Case(s)
1193	Atkin-Flaitz syndrome	Disorder	14 Case(s)
96055	Tetrasomy 21	Disorder	13 Case(s)
939	3-hydroxyisobutyric aciduria	Disorder	13 Case(s)
85174	Pseudodiastrophic dysplasia	Disorder	13 Case(s)
79502	Punctate palmoplantar keratoderma type 2	Disorder	13 Case(s)
79478	Griscelli syndrome type 3	Subtype of disorder	13 Case(s)
79329	MGAT2-CDG	Disorder	13 Case(s)
69739	Athabaskan brainstem dysgenesis syndrome	Disorder	13 Case(s)
66631	CEDNIK syndrome	Disorder	13 Case(s)
633014	SLC12A2-related developmental delay-intellectual disability-sensorineural deafness syndrome	Disorder	13 Case(s)
631068	Autosomal dominant spastic paraplegia type 80	Disorder	13 Case(s)
611247	Pontocerebellar hypoplasia type 11	Disorder	13 Case(s)
610569	KIAA1109-related early lethal congenital brain	Disorder	13 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	malformations-arthrogryposis syndrome		
556985	Early-onset calcifying leukoencephalopathy-skeletal dysplasia	Disorder	13 Case(s)
542657	Isolated hyperchlorhidrosis	Disorder	13 Case(s)
538963	Combined immunodeficiency due to ITK deficiency	Disorder	13 Case(s)
538096	Autosomal recessive lethal neonatal axonal sensorimotor polyneuropathy	Disorder	13 Case(s)
50945	Blomstrand lethal chondrodysplasia	Disorder	13 Case(s)
476394	PMP2-related Charcot-Marie-Tooth disease type 1	Disorder	13 Case(s)
466934	VPS11-related autosomal recessive hypomyelinating leukodystrophy	Disorder	13 Case(s)
448251	Progressive autosomal recessive ataxia-deafness syndrome	Disorder	13 Case(s)
443098	Hyperostosis cranialis interna	Disorder	13 Case(s)
439212	Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome	Disorder	13 Case(s)
436274	Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa	Disorder	13 Case(s)
436151	Intellectual disability-expressive aphasia-facial dysmorphism syndrome	Disorder	13 Case(s)
435438	Progressive myoclonic epilepsy type 7	Disorder	13 Case(s)
401953	Episodic ataxia with slurred speech	Disorder	13 Case(s)
399103	Distal nebulin myopathy	Disorder	13 Case(s)
363412	Hypomyelination with brain stem and spinal cord involvement and leg spasticity	Disorder	13 Case(s)
356978	D,L-2-hydroxyglutaric aciduria	Disorder	13 Case(s)
329813	Mosaic genome-wide paternal uniparental disomy	Disorder	13 Case(s)
329249	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency	Subtype of disorder	13 Case(s)
3268	Radioulnar synostosis-microcephaly-scoliosis syndrome	Disorder	13 Case(s)
319605	X-linked mendelian susceptibility to mycobacterial diseases	Disorder	13 Case(s)
319547	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency	Disorder	13 Case(s)
313936	PENS syndrome	Disorder	13 Case(s)
3097	Meacham syndrome	Disorder	13 Case(s)
306530	Congenital hereditary facial paralysis-variable hearing loss syndrome	Disorder	13 Case(s)
3042	Intellectual disability-cataracts-calcified pinnae-myopathy syndrome	Disorder	13 Case(s)
293958	Hypertelorism-preauricular sinus-punctual pits-deafness syndrome	Disorder	13 Case(s)
284160	8q21.11 microdeletion syndrome	Disorder	13 Case(s)
280406	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	Disorder	13 Case(s)
2802	X-linked sideroblastic anemia and spinocerebellar ataxia	Disorder	13 Case(s)
2319	Juberg-Hayward syndrome	Disorder	13 Case(s)
231720	Non-acquired combined pituitary hormone deficiency-	Disorder	13 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	sensorineural hearing loss-spine abnormalities syndrome		
1788	Acrofacial dysostosis, Rodríguez type	Disorder	13 Case(s)
178377	Osteosclerosis-developmental delay-craniosynostosis syndrome	Disorder	13 Case(s)
171612	Autosomal dominant spastic paraplegia type 37	Disorder	13 Case(s)
168549	Axial spondylometaphyseal dysplasia	Disorder	13 Case(s)
1435	Xq21 microdeletion syndrome	Disorder	13 Case(s)
140969	Saldino-Mainzer syndrome	Disorder	13 Case(s)
101102	Charcot-Marie-Tooth disease type 2H	Disorder	13 Case(s)
99672	Fried's tooth and nail syndrome	Disorder	12 Case(s)
98772	Spinocerebellar ataxia type 19/22	Disorder	12 Case(s)
96186	Maternal uniparental disomy of chromosome 20	Disorder	12 Case(s)
85320	X-linked intellectual disability-macrocephaly-macroorchidism syndrome	Disorder	12 Case(s)
844	Lown-Ganong-Levine syndrome	Disorder	12 Case(s)
79328	ALG9-CDG	Disorder	12 Case(s)
603689	KLHL7-related Bohring-Opitz-like syndrome	Disorder	12 Case(s)
597743	SETD2-related microcephaly-severe intellectual disability-multiple congenital anomalies syndrome	Disorder	12 Case(s)
59303	Neonatal ichthyosis-sclerosing cholangitis syndrome	Disorder	12 Case(s)
508533	Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome	Disorder	12 Case(s)
505237	Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome	Disorder	12 Case(s)
496689	Kyphoscoliosis-lateral tongue atrophy-hereditary spastic paraplegia syndrome	Disorder	12 Case(s)
459033	Ataxia-oculomotor apraxia type 4	Disorder	12 Case(s)
442582	AH amyloidosis	Disorder	12 Case(s)
420573	Severe combined immunodeficiency due to CTPS1 deficiency	Disorder	12 Case(s)
412066	PRKAR1B-related neurodegenerative dementia with intermediate filaments	Disorder	12 Case(s)
363523	Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome	Disorder	12 Case(s)
314585	15q overgrowth syndrome	Disorder	12 Case(s)
300570	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation	Disorder	12 Case(s)
300547	Autosomal recessive infantile hypercalcemia	Disorder	12 Case(s)
2935	Crossed polysyndactyly	Disorder	12 Case(s)
2919	Orofaciodigital syndrome type 5	Disorder	12 Case(s)
284460	Acute annular outer retinopathy	Disorder	12 Case(s)
280620	Progressive myoclonic epilepsy type 6	Disorder	12 Case(s)
280384	Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome	Disorder	12 Case(s)
2662	Keipert syndrome	Disorder	12 Case(s)
2579	Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome	Disorder	12 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
254531	Temple syndrome due to paternal 14q32.2 hypomethylation	Subtype of disorder	12 Case(s)
247794	Juvenile cataract-microcornea-renal glucosuria syndrome	Disorder	12 Case(s)
244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis	Disorder	12 Case(s)
238763	Glaucoma secondary to spherophakia/ectopia lentis and megalocornea	Disorder	12 Case(s)
2224	Hypertryptophanemia	Disorder	12 Case(s)
217377	Microduplication Xp11.22p11.23 syndrome	Disorder	12 Case(s)
217346	19q13.11 microdeletion syndrome	Disorder	12 Case(s)
210571	Dystonia 16	Disorder	12 Case(s)
209973	Benign nocturnal alternating hemiplegia of childhood	Disorder	12 Case(s)
199340	Muscular dystrophy, Selcen type	Disorder	12 Case(s)
1784	Acrofrontofacionasal dysostosis	Disorder	12 Case(s)
171829	6q16 microdeletion syndrome	Disorder	12 Case(s)
166035	Brachydactyly-short stature-retinitis pigmentosa syndrome	Disorder	12 Case(s)
1555	Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome	Disorder	12 Case(s)
1487	Cooks syndrome	Disorder	12 Case(s)
1473	Uveal coloboma-cleft lip and palate-intellectual disability	Disorder	12 Case(s)
1458	CODAS syndrome	Disorder	12 Case(s)
141148	Hemifacial myohyperplasia	Disorder	12 Case(s)
1179	Benign paroxysmal tonic upgaze of childhood with ataxia	Disorder	12 Case(s)
1008	Alopecia-epilepsy-pyorrhea-intellectual disability syndrome	Disorder	12 Case(s)
100046	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	Disorder	12 Case(s)
98912	Late-onset distal myopathy, Markesbery-Griggs type	Disorder	11 Case(s)
96172	Ring chromosome 3 syndrome	Disorder	11 Case(s)
91135	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	Disorder	11 Case(s)
91132	Ichthyosis-hypotrichosis syndrome	Disorder	11 Case(s)
85336	X-linked neurodegenerative syndrome, Hamel type	Disorder	11 Case(s)
83629	Leukoencephalopathy-spondyloepimetaphyseal dysplasia syndrome	Disorder	11 Case(s)
79324	ALG12-CDG	Disorder	11 Case(s)
79076	Juvenile polyposis of infancy	Subtype of disorder	11 Case(s)
620368	EGF-related primary hypomagnesemia with intellectual disability	Disorder	11 Case(s)
620363	Primary hypomagnesemia-generalized seizures-intellectual disability-obesity syndrome	Disorder	11 Case(s)
600663	NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	Disorder	11 Case(s)
597738	Luscan-Lumish syndrome	Disorder	11 Case(s)
572798	WARS2-related combined oxidative phosphorylation defect	Disorder	11 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
569816	CELSR1-related late-onset primary lymphedema	Disorder	11 Case(s)
555407	NAD(P)HX epimerase deficiency	Disorder	11 Case(s)
522077	Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome	Disorder	11 Case(s)
521450	LAMA5-related multisystemic syndrome	Disorder	11 Case(s)
521406	Dystonia-parkinsonism-hypermanganesemia syndrome	Disorder	11 Case(s)
521308	Frontonasal dysplasia-bifid nose-upper limb anomalies syndrome	Disorder	11 Case(s)
506307	Stromme syndrome	Disorder	11 Case(s)
477749	Pontine autosomal dominant microangiopathy with leukoencephalopathy	Disorder	11 Case(s)
468661	Autosomal recessive spastic paraplegia type 74	Disorder	11 Case(s)
464724	Fever-associated acute infantile liver failure syndrome	Disorder	11 Case(s)
457185	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome	Disorder	11 Case(s)
444077	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	Disorder	11 Case(s)
444051	20q11.2 microdeletion syndrome	Disorder	11 Case(s)
444013	Combined oxidative phosphorylation defect type 23	Disorder	11 Case(s)
443988	Ventriculomegaly-cystic kidney disease	Disorder	11 Case(s)
397937	Polyglucosan body myopathy type 1	Disorder	11 Case(s)
352712	Facial dysmorphism-immunodeficiency-livedo-short stature syndrome	Disorder	11 Case(s)
330050	DNM1L-related encephalopathy due to mitochondrial and peroxisomal fission defect	Subtype of disorder	11 Case(s)
319189	Familial cortical myoclonus	Disorder	11 Case(s)
313884	12p12.1 microdeletion syndrome	Subtype of disorder	11 Case(s)
313855	FGFR2-related bent bone dysplasia	Disorder	11 Case(s)
313850	Infantile cerebellar-retinal degeneration	Disorder	11 Case(s)
300293	Transient infantile hypertriglyceridemia and hepatosteatosis	Disorder	11 Case(s)
2987	Antecubital pterygium syndrome	Disorder	11 Case(s)
2959	Progeria-short stature-pigmented nevi syndrome	Disorder	11 Case(s)
2854	Fuhrmann syndrome	Disorder	11 Case(s)
2832	Short tarsus-absence of lower eyelashes syndrome	Disorder	11 Case(s)
280553	Fatal infantile hypertonic myofibrillar myopathy	Disorder	11 Case(s)
261349	2p15p16.1 microdeletion syndrome	Disorder	11 Case(s)
238744	Mammary-digital-nail syndrome	Disorder	11 Case(s)
2329	Karsch-Neugebauer syndrome	Disorder	11 Case(s)
228169	Autosomal dominant striatal neurodegeneration	Disorder	11 Case(s)
2253	Foveal hypoplasia-presenile cataract syndrome	Disorder	11 Case(s)
221120	Pseudoaminopterin syndrome	Disorder	11 Case(s)
217390	Combined immunodeficiency due to DOCK8 deficiency	Disorder	11 Case(s)
2163	Holoprosencephaly-craniosynostosis syndrome	Disorder	11 Case(s)
210133	Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome	Disorder	11 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
2016	Cleft palate-lateral synechia syndrome	Disorder	11 Case(s)
1757	Fibular dimelia-diplopodia syndrome	Disorder	11 Case(s)
168624	Familial scaphocephaly syndrome, McGillivray type	Disorder	11 Case(s)
168588	Hyperandrogenism due to cortisone reductase deficiency	Disorder	11 Case(s)
166282	Familial sick sinus syndrome	Disorder	11 Case(s)
166272	Odontochondrodysplasia	Disorder	11 Case(s)
1660	Dermoodontodysplasia	Disorder	11 Case(s)
1497	X-linked complicated corpus callosum dysgenesis	Subtype of disorder	11 Case(s)
1479	Atrial septal defect-atrioventricular conduction defects syndrome	Disorder	11 Case(s)
1031	Enamel-renal syndrome	Disorder	11 Case(s)
99807	PEHO-like syndrome	Disorder	10 Case(s)
99329	48,XYYY syndrome	Disorder	10 Case(s)
97340	Hunter-McAlpine syndrome	Disorder	10 Case(s)
97240	Zebra body myopathy	Disorder	10 Case(s)
968	Acromesomelic dysplasia, Hunter-Thompson type	Disorder	10 Case(s)
96178	Ring chromosome 16 syndrome	Disorder	10 Case(s)
958	Acro-renal-mandibular syndrome	Disorder	10 Case(s)
93599	Primary hyperoxaluria type 2	Subtype of disorder	10 Case(s)
93406	Syndactyly type 5	Disorder	10 Case(s)
93398	Genochondromatosis type 2	Disorder	10 Case(s)
93347	Anauxetic dysplasia	Disorder	10 Case(s)
916	Aase-Smith syndrome	Disorder	10 Case(s)
90398	Localized lichen myxedematosus with mixed features of different subtypes	Subtype of disorder	10 Case(s)
86918	Diffuse palmoplantar keratoderma-acrocyanosis syndrome	Disorder	10 Case(s)
85329	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	Disorder	10 Case(s)
85274	Syndromic X-linked intellectual disability 7	Disorder	10 Case(s)
85163	Hypomyelination-congenital cataract syndrome	Disorder	10 Case(s)
79503	Ichthyosis hystrix of Curth-Macklin	Disorder	10 Case(s)
79281	Alpha-N-acetylgalactosaminidase deficiency type 3	Subtype of disorder	10 Case(s)
79280	Alpha-N-acetylgalactosaminidase deficiency type 2	Subtype of disorder	10 Case(s)
79279	Alpha-N-acetylgalactosaminidase deficiency type 1	Subtype of disorder	10 Case(s)
79083	PPARG-related familial partial lipodystrophy	Disorder	10 Case(s)
71212	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	Disorder	10 Case(s)
641390	PsAPASH syndrome	Disorder	10 Case(s)
641361	Neurodevelopmental delay-hypotonia-cerebellar ataxia-cardiac conduction defects syndrome	Disorder	10 Case(s)
621758	Fibrosis-neurodegeneration-cerebral angiomatosis syndrome	Disorder	10 Case(s)
619948	Early-onset autoimmunity-autoinflammation-immunodeficiency syndrome due to SOCS1 haploinsufficiency	Disorder	10 Case(s)
611216	Aplastic anemia-intellectual disability-dwarfism syndrome	Disorder	10 Case(s)
603448	Cerebellar hypoplasia-intellectual disability-congenital	Disorder	10 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	microcephaly-dystonia-anemia-growth retardation syndrome		
568062	PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis	Disorder	10 Case(s)
567502	B-cell immunodeficiency-limb anomaly-urogenital malformation syndrome	Disorder	10 Case(s)
562569	TMEM94-associated congenital heart defect-facial dysmorphism-developmental delay syndrome	Disorder	10 Case(s)
531151	9q21.13 microdeletion syndrome	Disorder	10 Case(s)
529665	Neurodevelopmental delay-seizures-ophthalmic anomalies-osteopenia-cerebellar atrophy syndrome	Disorder	10 Case(s)
521445	Microcephaly-facial dysmorphism-ocular anomalies-multiple congenital anomalies syndrome	Disorder	10 Case(s)
506358	Gabriele-de Vries syndrome	Disorder	10 Case(s)
495274	Charcot-Marie-Tooth disease type 2T	Disorder	10 Case(s)
494344	RERE-related neurodevelopmental syndrome	Disorder	10 Case(s)
468699	SLC39A8-CDG	Disorder	10 Case(s)
466950	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation	Subtype of disorder	10 Case(s)
466926	Seizures-scoliosis-macrocephaly syndrome	Disorder	10 Case(s)
464443	COG6-CGD	Disorder	10 Case(s)
453504	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome due to a point mutation	Subtype of disorder	10 Case(s)
439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease	Disorder	10 Case(s)
420789	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	Disorder	10 Case(s)
401901	Huntington disease-like syndrome due to C9ORF72 expansions	Disorder	10 Case(s)
399081	KLHL9-related early-onset distal myopathy	Disorder	10 Case(s)
39	Acromelanosis	Disorder	10 Case(s)
363400	Severe neurodegenerative syndrome with lipodystrophy	Disorder	10 Case(s)
352737	Temperature-sensitive oculocutaneous albinism type 1	Subtype of disorder	10 Case(s)
352734	Minimal pigment oculocutaneous albinism type 1	Subtype of disorder	10 Case(s)
352641	Autosomal recessive cerebellar ataxia with late-onset spasticity	Disorder	10 Case(s)
35107	Desmosterolosis	Disorder	10 Case(s)
3469	XK aprosencephaly syndrome	Disorder	10 Case(s)
3439	Von Voss-Cherstvoy syndrome	Disorder	10 Case(s)
33574	Glutamate-cysteine ligase deficiency	Disorder	10 Case(s)
3317	Thoracolaryngopelvic dysplasia	Disorder	10 Case(s)
329228	Microcephalic primordial dwarfism due to ZNF335 deficiency	Disorder	10 Case(s)
3259	Syndactyly-polydactyly-ear lobe syndrome	Disorder	10 Case(s)
324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency	Subtype of disorder	10 Case(s)

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319671	Alazami syndrome	Disorder	10 Case(s)
314662	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	Disorder	10 Case(s)
313906	Congenital pancreatic cyst	Disorder	10 Case(s)
309246	GM2 gangliosidosis, AB variant	Disorder	10 Case(s)
3032	NPHP3-related Meckel-like syndrome	Disorder	10 Case(s)
3023	External auditory canal atresia-vertical talus-hypertelorism syndrome	Disorder	10 Case(s)
294016	Microcephaly-capillary malformation syndrome	Disorder	10 Case(s)
2880	Phosphoenolpyruvate carboxykinase deficiency	Disorder	10 Case(s)
284227	TEMPI syndrome	Disorder	10 Case(s)
2839	Pelvis-shoulder dysplasia	Disorder	10 Case(s)
280794	Pseudoxanthomatous diffuse cutaneous mastocytosis	Subtype of disorder	10 Case(s)
280615	Hemoglobinopathy Toms River	Disorder	10 Case(s)
276280	Hemihyperplasia-multiple lipomatosis syndrome	Disorder	10 Case(s)
2658	Lenz-Majewski hyperostotic dwarfism	Disorder	10 Case(s)
263482	Spondyloepimetaphyseal dysplasia, Maroteaux type	Disorder	10 Case(s)
263458	Hyperinsulinism due to INSR deficiency	Disorder	10 Case(s)
2590	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	Disorder	10 Case(s)
254504	Inhalational botulism	Subtype of disorder	10 Case(s)
254411	Annular atrophic lichen planus	Disorder	10 Case(s)
2496	Mesomelia-synostoses syndrome	Disorder	10 Case(s)
240112	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome	Subtype of disorder	10 Case(s)
228426	Syndromic multisystem autoimmune disease due to Itch deficiency	Disorder	10 Case(s)
2255	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome	Disorder	10 Case(s)
220465	Laron syndrome with immunodeficiency	Disorder	10 Case(s)
217335	RIN2 syndrome	Disorder	10 Case(s)
210144	Lethal polymalformative syndrome, Boissel type	Disorder	10 Case(s)
2072	Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome	Subtype of disorder	10 Case(s)
2047	Flynn-Aird syndrome	Disorder	10 Case(s)
2008	Acrocardiofacial syndrome	Disorder	10 Case(s)
169090	Combined immunodeficiency due to CRAC channel dysfunction	Disorder	10 Case(s)
166073	Pontocerebellar hypoplasia type 6	Disorder	10 Case(s)
163966	X-linked dominant chondrodysplasia, Chassaing-Lacombe type	Disorder	10 Case(s)
1627	Deletion 5q35	Disorder	10 Case(s)
1568	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	Disorder	10 Case(s)
1471	Coloboma of macula-brachydactyly type B syndrome	Disorder	10 Case(s)
1443	Ring chromosome 19 syndrome	Disorder	10 Case(s)
1439	Ring chromosome 12 syndrome	Disorder	10 Case(s)

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1426	Greenberg dysplasia	Disorder	10 Case(s)
141007	Orofaciodigital syndrome type 9	Disorder	10 Case(s)
139426	Perioral myoclonia with absences	Disorder	10 Case(s)
139406	Encephalopathy due to prosaposin deficiency	Disorder	10 Case(s)
1336	Hyperkeratosis-hyperpigmentation syndrome	Disorder	10 Case(s)
1313	Infantile choroidocerebral calcification syndrome	Disorder	10 Case(s)
1263	Boomerang dysplasia	Disorder	10 Case(s)
1171	Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome	Disorder	10 Case(s)
1150	Arthrogryposis multiplex congenita-whistling face syndrome	Disorder	10 Case(s)
101111	Spinocerebellar ataxia type 25	Disorder	10 Case(s)
101007	Autosomal recessive spastic paraplegia type 27	Disorder	10 Case(s)
1010	Autosomal dominant palmoplantar keratoderma and congenital alopecia	Disorder	10 Case(s)
100994	Autosomal dominant spastic paraplegia type 13	Disorder	10 Case(s)
99014	X-linked Charcot-Marie-Tooth disease type 5	Disorder	9 Case(s)
94124	Spinocerebellar ataxia with axonal neuropathy type 1	Disorder	9 Case(s)
93952	X-linked intellectual disability, Hedera type	Disorder	9 Case(s)
93317	Spondylometaphyseal dysplasia, Sedaghatian type	Disorder	9 Case(s)
85338	X-linked intellectual disability-ataxia-apraxia syndrome	Disorder	9 Case(s)
85286	X-linked intellectual disability, Shashi type	Disorder	9 Case(s)
85199	Craniosynostosis-anal anomalies-porokeratosis syndrome	Disorder	9 Case(s)
83619	Macrostomia-preauricular tags-external ophthalmoplegia syndrome	Disorder	9 Case(s)
79405	Junctional epidermolysis bullosa inversa	Disorder	9 Case(s)
79322	DPM1-CDG	Disorder	9 Case(s)
636941	Vascular Ehlers-Danlos-polymicrogyria syndrome	Disorder	9 Case(s)
631106	Spinocerebellar ataxia type 49	Disorder	9 Case(s)
631082	Autosomal recessive spastic paraplegia type 85	Disorder	9 Case(s)
619233	Hereditary persistence of fetal hemoglobin-intellectual disability syndrome	Disorder	9 Case(s)
505208	3-methylglutaconic aciduria type 8	Disorder	9 Case(s)
502423	Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	Disorder	9 Case(s)
488197	Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome	Disorder	9 Case(s)
480851	Hereditary thrombocytopenia with early-onset myelofibrosis	Disorder	9 Case(s)
477814	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome	Disorder	9 Case(s)
438075	Ketoacidosis due to monocarboxylate transporter-1 deficiency	Disorder	9 Case(s)
425120	STING-associated vasculopathy with onset in infancy	Disorder	9 Case(s)
420561	Temple-Baraitser syndrome	Disorder	9 Case(s)
401945	Moyamoya disease with early-onset achalasia	Disorder	9 Case(s)
397787	Severe combined immunodeficiency due to IKK2 deficiency	Disorder	9 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
397750	Periodic paralysis with later-onset distal motor neuropathy	Disorder	9 Case(s)
397612	Macrocephaly-developmental delay syndrome	Disorder	9 Case(s)
370927	SSR4-CDG	Disorder	9 Case(s)
363710	Spinocerebellar ataxia type 37	Disorder	9 Case(s)
35704	L-Arginine:glycine amidinotransferase deficiency	Disorder	9 Case(s)
35701	3-hydroxy-3-methylglutaryl-CoA synthase deficiency	Disorder	9 Case(s)
352745	Oculocutaneous albinism type 7	Disorder	9 Case(s)
329478	Adult-onset distal myopathy due to VCP mutation	Disorder	9 Case(s)
324585	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	Disorder	9 Case(s)
319199	Autosomal recessive spastic paraplegia type 53	Disorder	9 Case(s)
314679	Cerebrofacioarticular syndrome	Disorder	9 Case(s)
314466	Atypical Meigs syndrome	Disorder	9 Case(s)
300179	Kyphoscoliotic Ehlers-Danlos syndrome due to FKBP22 deficiency	Subtype of disorder	9 Case(s)
2952	Adducted thumbs-arthrogryposis syndrome, Christian type	Disorder	9 Case(s)
293948	1p21.3 microdeletion syndrome	Disorder	9 Case(s)
293939	Distal Xq28 microduplication syndrome	Disorder	9 Case(s)
2808	Laryngeal abductor paralysis	Disorder	9 Case(s)
280679	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	Disorder	9 Case(s)
2680	Hypomyelination neuropathy-arthrogryposis syndrome	Disorder	9 Case(s)
263487	COG5-CDG	Disorder	9 Case(s)
261190	Cleft palate-congenital heart defect-intellectual disability syndrome due to 15q14 microdeletion	Subtype of disorder	9 Case(s)
2557	Mietens syndrome	Disorder	9 Case(s)
254525	Temple syndrome due to paternal 14q32.2 microdeletion	Subtype of disorder	9 Case(s)
251279	Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	Disorder	9 Case(s)
231531	Hermansky-Pudlak syndrome due to BLOC-1 deficiency	Subtype of disorder	9 Case(s)
231154	Combined immunodeficiency due to partial RAG1 deficiency	Disorder	9 Case(s)
228003	Severe combined immunodeficiency due to CORO1A deficiency	Disorder	9 Case(s)
2213	Hypertelorism-microtia-facial clefting syndrome	Disorder	9 Case(s)
221039	Hereditary sclerosing poikiloderma, Weary type	Disorder	9 Case(s)
209951	Autosomal spastic paraplegia type 18	Disorder	9 Case(s)
169095	Severe combined immunodeficiency due to FOXN1 deficiency	Disorder	9 Case(s)
168558	46,XY difference of sex development-adrenal insufficiency due to CYP11A1 deficiency	Disorder	9 Case(s)
163979	X-linked intellectual disability-craniofacioskeletal syndrome	Disorder	9 Case(s)
1553	Curry-Jones syndrome	Disorder	9 Case(s)
1512	Crane-Heise syndrome	Disorder	9 Case(s)
140936	Lelis syndrome	Disorder	9 Case(s)
137628	Cardiac anomalies-heterotaxy syndrome	Disorder	9 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
1345	Cardiomyopathy-cataract-hip spine disease syndrome	Disorder	9 Case(s)
1292	Brachymorphism-onychodysplasia-dysphalangism syndrome	Disorder	9 Case(s)
1264	Tricho-retino-dento-digital syndrome	Disorder	9 Case(s)
1120	Lung agenesis-heart defect-thumb anomalies syndrome	Disorder	9 Case(s)
1000	Ocular albinism with late-onset sensorineural deafness	Disorder	9 Case(s)
99330	49,XYYYY syndrome	Disorder	8 Case(s)
98807	Primary dystonia, DYT13 type	Disorder	8 Case(s)
922	Familial nasal acilia	Disorder	8 Case(s)
85273	X-linked intellectual disability, Abidi type	Disorder	8 Case(s)
79350	3-phosphoserine phosphatase deficiency, infantile/juvenile form	Subtype of disorder	8 Case(s)
79333	COG7-CDG	Disorder	8 Case(s)
79323	MPDU1-CDG	Disorder	8 Case(s)
633028	CPE-related Prader-Willi-like syndrome	Disorder	8 Case(s)
600731	Clark-Baraitser syndrome	Disorder	8 Case(s)
597201	TRIM22-related inflammatory bowel disease	Disorder	8 Case(s)
572013	Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome	Disorder	8 Case(s)
542585	Auditory neuropathy-optic atrophy syndrome	Disorder	8 Case(s)
536516	Myopathic Ehlers-Danlos syndrome	Disorder	8 Case(s)
519388	Autosomal recessive anterior segment dysgenesis	Disorder	8 Case(s)
514352	Congenital brachyesophagus-intrathoracic stomach-vertebral anomalies syndrome	Disorder	8 Case(s)
502430	Weiss-Kruszka Syndrome	Disorder	8 Case(s)
496790	Ocular anomalies-axonal neuropathy-developmental delay syndrome	Disorder	8 Case(s)
494526	Infantile-onset generalized dyskinesia with orofacial involvement	Disorder	8 Case(s)
494444	DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	Disorder	8 Case(s)
476093	Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome	Disorder	8 Case(s)
468684	CCDC115-CDG	Disorder	8 Case(s)
466722	Autosomal recessive spastic paraplegia type 77	Disorder	8 Case(s)
464336	BENTA disease	Disorder	8 Case(s)
459061	Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome	Disorder	8 Case(s)
457485	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	Disorder	8 Case(s)
457406	Multiple mitochondrial dysfunctions syndrome type 4	Disorder	8 Case(s)
438274	GCGR-related hyperglucagonemia	Disorder	8 Case(s)
435845	Lethal neonatal spasticity-epileptic encephalopathy syndrome	Disorder	8 Case(s)
435638	3p25.3 microdeletion syndrome	Disorder	8 Case(s)
420686	Woolly hair-palmoplantar keratoderma syndrome	Disorder	8 Case(s)
404454	Alacrimia-choreoathetosis-liver dysfunction syndrome	Disorder	8 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
401942	Familial median cleft of the upper and lower lips	Disorder	8 Case(s)
397715	Joubert syndrome with Jeune asphyxiating thoracic dystrophy	Disorder	8 Case(s)
397590	Silver-Russell syndrome due to a point mutation	Subtype of disorder	8 Case(s)
391408	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	Disorder	8 Case(s)
370943	Autism spectrum disorder-epilepsy-arthrogryposis syndrome	Disorder	8 Case(s)
352675	X-linked Charcot-Marie-Tooth disease type 6	Disorder	8 Case(s)
352670	Autosomal dominant intermediate Charcot-Marie-Tooth disease type F	Disorder	8 Case(s)
352649	Brain dopamine-serotonin vesicular transport disease	Disorder	8 Case(s)
352479	ISPD-related limb-girdle muscular dystrophy R20	Disorder	8 Case(s)
3474	CHIME syndrome	Disorder	8 Case(s)
33572	5-oxoprolinase deficiency	Disorder	8 Case(s)
331226	Susceptibility to infection due to TYK2 deficiency	Disorder	8 Case(s)
329258	Autosomal dominant Charcot-Marie-Tooth disease type 2Q	Disorder	8 Case(s)
324321	Sinoatrial node dysfunction and deafness	Disorder	8 Case(s)
3219	Fountain syndrome	Disorder	8 Case(s)
3216	Conductive deafness-malformed external ear syndrome	Disorder	8 Case(s)
314811	Short stature due to GHSR deficiency	Disorder	8 Case(s)
314637	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	Disorder	8 Case(s)
306577	Hereditary sodium channelopathy-related small fibers neuropathy	Disorder	8 Case(s)
306558	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome	Disorder	8 Case(s)
306547	Porencephaly-microcephaly-bilateral congenital cataract syndrome	Disorder	8 Case(s)
3019	Ramon syndrome	Disorder	8 Case(s)
2958	X-linked intellectual disability-dysmorphism-cerebral atrophy syndrome	Disorder	8 Case(s)
2934	Polysyndactyly-cardiac malformation syndrome	Disorder	8 Case(s)
2885	Piebald trait-neurologic defects syndrome	Disorder	8 Case(s)
2812	Parana hard skin syndrome	Disorder	8 Case(s)
280325	Distal deletion 12p	Disorder	8 Case(s)
280071	ALG11-CDG	Disorder	8 Case(s)
276432	Ogden syndrome	Disorder	8 Case(s)
263665	NK-cell enteropathy	Disorder	8 Case(s)
261483	Xq27.3q28 duplication syndrome	Disorder	8 Case(s)
261211	16p11.2p12.2 microdeletion syndrome	Disorder	8 Case(s)
2563	MOMO syndrome	Disorder	8 Case(s)
2561	Pyramidal molars-abnormal upper lip syndrome	Disorder	8 Case(s)
254528	Kagami-Ogata syndrome due to maternal 14q32.2 microdeletion	Subtype of disorder	8 Case(s)
251290	Parietal foramina with clavicular hypoplasia	Disorder	8 Case(s)
244310	RFT1-CDG	Disorder	8 Case(s)

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2371	Lethal Larsen-like syndrome	Disorder	8 Case(s)
2351	Kousseff syndrome	Disorder	8 Case(s)
2326	Kallmann syndrome-heart disease syndrome	Disorder	8 Case(s)
231736	Microcornea-posterior megalolenticonus-persistent fetal vasculature-coloboma syndrome	Disorder	8 Case(s)
2252	Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome	Disorder	8 Case(s)
221054	Acrocephalopolydactyly	Disorder	8 Case(s)
2206	Ankylosing vertebral hyperostosis with tylosis	Disorder	8 Case(s)
2180	Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome	Disorder	8 Case(s)
2107	Hall-Riggs syndrome	Disorder	8 Case(s)
2013	Cleft palate-large ears-small head syndrome	Disorder	8 Case(s)
1884	Ectopia lentis-chorioretinal dystrophy-myopia syndrome	Disorder	8 Case(s)
1852	X-linked retinal dysplasia	Disorder	8 Case(s)
1824	Lowry-Wood syndrome	Disorder	8 Case(s)
178506	Brain calcification, Rajab type	Disorder	8 Case(s)
178389	Osteopetrosis-hypogammaglobulinemia syndrome	Disorder	8 Case(s)
1655	Müllerian derivatives-lymphangiectasia-polydactyly syndrome	Disorder	8 Case(s)
163956	X-linked intellectual disability, Nascimento type	Disorder	8 Case(s)
157965	SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome	Subtype of disorder	8 Case(s)
1454	Joubert syndrome with hepatic defect	Disorder	8 Case(s)
1450	Ring chromosome 8 syndrome	Disorder	8 Case(s)
137639	Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome	Subtype of disorder	8 Case(s)
1366	Autosomal recessive palmoplantar keratoderma and congenital alopecia	Disorder	8 Case(s)
1327	Camptodactyly syndrome, Guadalajara type 1	Disorder	8 Case(s)
1318	Campomelia, Cumming type	Disorder	8 Case(s)
1278	Brachydactyly-preaxial hallux varus syndrome	Disorder	8 Case(s)
1226	Bamforth-Lazarus syndrome	Disorder	8 Case(s)
1188	Ataxia-deafness-intellectual disability syndrome	Disorder	8 Case(s)
99843	Leukocyte adhesion deficiency type II	Subtype of disorder	7 Case(s)
99710	Punctate acrokeratoderma freckle-like pigmentation	Disorder	7 Case(s)
93382	Brachydactyly type A6	Disorder	7 Case(s)
93316	Spondylometaphyseal dysplasia, Schmidt type	Disorder	7 Case(s)
929	Achalasia-microcephaly syndrome	Disorder	7 Case(s)
90103	Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome	Disorder	7 Case(s)
85334	X-linked neurodegenerative syndrome, Bertini type	Disorder	7 Case(s)
85194	Spondylo-ocular syndrome	Disorder	7 Case(s)
83620	Enteric anendocrinosis	Disorder	7 Case(s)
79094	Grange syndrome	Disorder	7 Case(s)
71526	Obesity due to pro-opiomelanocortin deficiency	Subtype of disorder	7 Case(s)
65282	Carvajal syndrome	Disorder	7 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
631095	Spinocerebellar ataxia type 44	Disorder	7 Case(s)
631088	Autosomal recessive spastic paraplegia type 87	Disorder	7 Case(s)
619953	Familial hyperinflammatory lymphoproliferative immunodeficiency	Disorder	7 Case(s)
611201	Oculogastrointestinal-neurodevelopmental syndrome	Disorder	7 Case(s)
600691	Combined deficiency of factor VII and factor X	Disorder	7 Case(s)
589608	Linear hypopigmentation and craniofacial asymmetry with acral, ocular and brain anomalies	Disorder	7 Case(s)
589527	Spinocerebellar ataxia type 45	Disorder	7 Case(s)
589442	Short stature-skeletal dysplasia-retinal degeneration-intellectual disability-sensorineural hearing loss syndrome	Disorder	7 Case(s)
562559	Anterior maxillary protrusion-strabismus-intellectual disability syndrome	Disorder	7 Case(s)
544628	Atypical Fanconi syndrome-neonatal hyperinsulinism syndrome	Disorder	7 Case(s)
538101	Congenital axonal neuropathy with encephalopathy	Disorder	7 Case(s)
536532	Classical-like Ehlers-Danlos syndrome type 2	Disorder	7 Case(s)
529977	Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections-lymphopenia syndrome	Disorder	7 Case(s)
513436	Autosomal recessive spastic paraplegia type 78	Disorder	7 Case(s)
508476	Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-hearing loss syndrome	Disorder	7 Case(s)
508093	MEPAN syndrome	Disorder	7 Case(s)
504530	Combined immunodeficiency due to Moesin deficiency	Disorder	7 Case(s)
500548	Osteosclerotic metaphyseal dysplasia	Disorder	7 Case(s)
500180	Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	Disorder	7 Case(s)
497764	Spinocerebellar ataxia type 43	Disorder	7 Case(s)
496693	Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome	Disorder	7 Case(s)
488650	Distal myopathy, Tateyama type	Disorder	7 Case(s)
487825	Pierpont syndrome	Disorder	7 Case(s)
486811	Prenatal-onset spinal muscular atrophy with congenital bone fractures	Disorder	7 Case(s)
477857	Mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency	Disorder	7 Case(s)
468666	Isolated generalized anhidrosis with normal sweat glands	Disorder	7 Case(s)
467166	Tubulinopathy-associated dysgyria	Disorder	7 Case(s)
466703	TMEM199-CDG	Disorder	7 Case(s)
459051	Spondyloepiphyseal dysplasia, Stanescu type	Disorder	7 Case(s)
457375	ITPA-related lethal infantile neurological disorder with cataract and cardiac involvement	Disorder	7 Case(s)
447896	Tremor-ataxia-central hypomyelination syndrome	Subtype of disorder	7 Case(s)
439822	PDE4D haploinsufficiency syndrome	Disorder	7 Case(s)
436242	Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease	Disorder	7 Case(s)
435387	Autosomal dominant Charcot-Marie-Tooth disease type 2Y	Disorder	7 Case(s)

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404463	Multisystemic smooth muscle dysfunction syndrome	Disorder	7 Case(s)
404440	Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	Disorder	7 Case(s)
401785	Autosomal recessive spastic paraplegia type 62	Disorder	7 Case(s)
399086	Finnish upper limb-onset distal myopathy	Disorder	7 Case(s)
397695	3q27.3 microdeletion syndrome	Disorder	7 Case(s)
391646	Feingold syndrome type 2	Subtype of disorder	7 Case(s)
370022	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome	Disorder	7 Case(s)
369950	Intellectual disability-seizures-macrocephaly-obesity syndrome	Disorder	7 Case(s)
369939	Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome	Disorder	7 Case(s)
363992	Ichthyosis-short stature-brachydactyly-microspherophakia syndrome	Disorder	7 Case(s)
363432	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency	Subtype of disorder	7 Case(s)
363396	High myopia-sensorineural deafness syndrome	Disorder	7 Case(s)
357001	19p13.13 microdeletion syndrome	Disorder	7 Case(s)
352587	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	Disorder	7 Case(s)
352582	Familial infantile myoclonic epilepsy	Disorder	7 Case(s)
33573	Gamma-glutamyl transpeptidase deficiency	Disorder	7 Case(s)
3341	Torticollis-keloids-cryptorchidism-renal dysplasia syndrome	Disorder	7 Case(s)
329802	5p13 microduplication syndrome	Disorder	7 Case(s)
329329	Autosomal recessive frontotemporal pachygyria	Disorder	7 Case(s)
324632	Hendra virus infection	Disorder	7 Case(s)
320355	Autosomal dominant spastic paraplegia type 41	Disorder	7 Case(s)
319623	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency	Subtype of disorder	7 Case(s)
319504	Combined oxidative phosphorylation defect type 8	Disorder	7 Case(s)
3194	Corneodermatoosseous syndrome	Disorder	7 Case(s)
317476	XMEN	Disorder	7 Case(s)
314689	Combined immunodeficiency due to STK4 deficiency	Disorder	7 Case(s)
314655	Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion	Subtype of disorder	7 Case(s)
308425	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	Disorder	7 Case(s)
3078	Severe X-linked intellectual disability, Gustavson type	Disorder	7 Case(s)
300382	Progeroid and marfanoid aspect-lipodystrophy syndrome	Disorder	7 Case(s)
293978	Deficiency in anterior pituitary function-variable immunodeficiency syndrome	Disorder	7 Case(s)
293165	Skin fragility-woolly hair-palmoplantar keratoderma syndrome	Disorder	7 Case(s)
2920	Oliver syndrome	Disorder	7 Case(s)
2872	Cardiocranial syndrome, Pfeiffer type	Disorder	7 Case(s)

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280365	Autosomal semi-dominant severe lipodystrophic laminopathy	Disorder	7 Case(s)
2645	Osteoglosphonic dysplasia	Disorder	7 Case(s)
263347	MRCS syndrome	Disorder	7 Case(s)
261279	17q23.1q23.2 microdeletion syndrome	Disorder	7 Case(s)
261229	14q11.2 microduplication syndrome	Disorder	7 Case(s)
261204	16p11.2p12.2 microduplication syndrome	Disorder	7 Case(s)
2598	Mitochondrial myopathy and sideroblastic anemia	Disorder	7 Case(s)
2560	Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome	Disorder	7 Case(s)
254930	Combined oxidative phosphorylation defect type 7	Disorder	7 Case(s)
254534	Kagami-Ogata syndrome due to maternal 14q32.2 hypermethylation	Subtype of disorder	7 Case(s)
247198	Progressive cerebello-cerebral atrophy	Disorder	7 Case(s)
2439	Patterson-Stevenson-Fontaine syndrome	Disorder	7 Case(s)
228379	Virus-associated trichodysplasia spinulosa	Disorder	7 Case(s)
228190	Patent ductus arteriosus-bicuspid aortic valve-hand anomalies syndrome	Disorder	7 Case(s)
2232	Primary hypergonadotropic hypogonadism-partial alopecia syndrome	Disorder	7 Case(s)
2141	Diaphragmatic defect-limb deficiency-skull defect syndrome	Disorder	7 Case(s)
211067	Episodic ataxia type 5	Disorder	7 Case(s)
209970	Episodic ataxia type 7	Disorder	7 Case(s)
2095	Gorlin-Chaudhry-Moss syndrome	Disorder	7 Case(s)
199332	Endocrine-cerebro-osteodysplasia syndrome	Disorder	7 Case(s)
1875	Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome	Disorder	7 Case(s)
1858	Skeletal dysplasia-epilepsy-short stature syndrome	Disorder	7 Case(s)
1842	Bone dysplasia, lethal Holmgren type	Disorder	7 Case(s)
1818	Ectodermal dysplasia, trichoodontoonychial type	Disorder	7 Case(s)
178338	UV-sensitive syndrome	Disorder	7 Case(s)
168566	Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	Disorder	7 Case(s)
163976	X-linked intellectual disability, Van Esch type	Disorder	7 Case(s)
163693	2p21 microdeletion syndrome	Disorder	7 Case(s)
1574	Retinal degeneration-nanophthalmos-glaucoma syndrome	Disorder	7 Case(s)
140952	Syndactyly-telecanthus-anogenital and renal malformations syndrome	Disorder	7 Case(s)
139474	17q11.2 microduplication syndrome	Disorder	7 Case(s)
1299	Branchioskeletogenital syndrome	Disorder	7 Case(s)
1131	X-linked mandibulofacial dysostosis	Disorder	7 Case(s)
1078	Thumb stiffness-brachydactyly-intellectual disability syndrome	Disorder	7 Case(s)
101078	X-linked Charcot-Marie-Tooth disease type 4	Disorder	7 Case(s)
101008	Autosomal recessive spastic paraplegia type 28	Disorder	7 Case(s)
999	Ermine phenotype	Disorder	6 Case(s)

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99141	Lymphedema-posterior choanal atresia syndrome	Disorder	6 Case(s)
991	PAGOD syndrome	Disorder	6 Case(s)
98893	Congenital muscular dystrophy type 1B	Disorder	6 Case(s)
91498	Familial congenital palsy of trochlear nerve	Disorder	6 Case(s)
85276	X-linked intellectual disability, Armfield type	Disorder	6 Case(s)
79156	Seizures-intellectual disability due to hydroxylysinuria syndrome	Disorder	6 Case(s)
79106	Eiken syndrome	Disorder	6 Case(s)
66518	Short fifth metacarpals-insulin resistance syndrome	Disorder	6 Case(s)
642085	EXOC6B-related spondyloepimetaphyseal dysplasia with joint laxity	Disorder	6 Case(s)
619367	SAMD9L-associated autoinflammatory syndrome	Disorder	6 Case(s)
569274	Multiple mitochondrial dysfunctions syndrome type 5	Disorder	6 Case(s)
565624	Combined oxidative phosphorylation defect type 39	Disorder	6 Case(s)
555402	NAD(P)HX dehydratase deficiency	Disorder	6 Case(s)
541423	Growth delay-intellectual disability-hepatopathy syndrome	Disorder	6 Case(s)
538958	Combined immunodeficiency due to CD70 deficiency	Disorder	6 Case(s)
512260	Congenital cerebellar ataxia due to RNU12 mutation	Disorder	6 Case(s)
508523	Hyperphenylalaninemia due to DNAJC12 deficiency	Disorder	6 Case(s)
505242	Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome	Disorder	6 Case(s)
500545	Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract	Disorder	6 Case(s)
500055	Hao-Fountain syndrome due to 16p13.2 microdeletion	Subtype of disorder	6 Case(s)
496756	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome	Disorder	6 Case(s)
495879	Congenital agenesis of the scrotum	Disorder	6 Case(s)
495844	C11ORF73-related autosomal recessive hypomyelinating leukodystrophy	Disorder	6 Case(s)
488642	TELO2-related intellectual disability-neurodevelopmental disorder	Disorder	6 Case(s)
480898	Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	Disorder	6 Case(s)
467176	Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome	Disorder	6 Case(s)
466791	Macrocephaly-intellectual disability-left ventricular non compaction syndrome	Disorder	6 Case(s)
466688	Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome	Disorder	6 Case(s)
464288	Short stature-brachydactyly-obesity-global developmental delay syndrome	Disorder	6 Case(s)
457378	Complex lethal osteochondrodysplasia	Disorder	6 Case(s)
447980	19p13.3 microduplication syndrome	Disorder	6 Case(s)
444463	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome due to TPP2 deficiency	Disorder	6 Case(s)
436141	HIDEA syndrome	Disorder	6 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
436003	Contractures-developmental delay-Pierre Robin syndrome	Disorder	6 Case(s)
423454	Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome	Disorder	6 Case(s)
401874	Multiple mitochondrial dysfunctions syndrome type 2	Disorder	6 Case(s)
401777	Optic atrophy-intellectual disability syndrome	Disorder	6 Case(s)
398127	Neonatal scleroderma	Disorder	6 Case(s)
370088	Acute infantile liver failure-multisystemic involvement syndrome	Disorder	6 Case(s)
363540	Leukoencephalopathy with mild cerebellar ataxia and white matter edema	Disorder	6 Case(s)
357329	Combined immunodeficiency due to IL21R deficiency	Disorder	6 Case(s)
352682	Cobblestone lissencephaly without muscular or ocular involvement	Disorder	6 Case(s)
352654	Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	Disorder	6 Case(s)
352447	Progressive external ophthalmoplegia-myopathy-emaciation syndrome	Disorder	6 Case(s)
3316	Thomas syndrome	Disorder	6 Case(s)
3246	Symphalangism with multiple anomalies of hands and feet	Disorder	6 Case(s)
324569	Pontocerebellar hypoplasia type 8	Disorder	6 Case(s)
319651	Constitutional megaloblastic anemia with severe neurologic disease	Disorder	6 Case(s)
319612	X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency	Subtype of disorder	6 Case(s)
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	Disorder	6 Case(s)
319563	Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency	Disorder	6 Case(s)
3175	X-linked spasticity-intellectual disability-epilepsy syndrome	Disorder	6 Case(s)
317428	Combined immunodeficiency due to ORAI1 deficiency	Subtype of disorder	6 Case(s)
314667	TMEM165-CDG	Disorder	6 Case(s)
314399	Autosomal dominant aplasia and myelodysplasia	Disorder	6 Case(s)
3077	X-linked intellectual disability-psychosis-macroorchidism syndrome	Disorder	6 Case(s)
3074	Intellectual disability-short stature-hypertelorism syndrome	Disorder	6 Case(s)
3038	Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome	Disorder	6 Case(s)
289513	12q15q21.1 microdeletion syndrome	Disorder	6 Case(s)
2824	Paraplegia-intellectual disability-hyperkeratosis syndrome	Disorder	6 Case(s)
2815	Spastic paraparesis-deafness syndrome	Disorder	6 Case(s)
2804	W syndrome	Disorder	6 Case(s)
2793	Otoonychoperoneal syndrome	Disorder	6 Case(s)
2743	Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome	Disorder	6 Case(s)
254361	Plectin-related limb-girdle muscular dystrophy R17	Disorder	6 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
254346	19p13.12 microdeletion syndrome	Disorder	6 Case(s)
254343	Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome	Disorder	6 Case(s)
247815	Autosomal recessive ataxia due to PEX10 deficiency	Disorder	6 Case(s)
2464	Marfanoid syndrome, De Silva type	Disorder	6 Case(s)
2339	Keratosis follicularis-dwarfism-cerebral atrophy syndrome	Disorder	6 Case(s)
2328	Kapur-Toriello syndrome	Disorder	6 Case(s)
230851	Cardiac-valvular Ehlers-Danlos syndrome	Disorder	6 Case(s)
2306	Isotretinoin-like syndrome	Disorder	6 Case(s)
2230	Hypogonadotropic hypogonadism-frontoparietal alopecia syndrome	Disorder	6 Case(s)
211017	Spinocerebellar ataxia type 30	Disorder	6 Case(s)
2057	Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome	Disorder	6 Case(s)
1951	Epilepsy-telangiectasia syndrome	Disorder	6 Case(s)
1808	Hidrotic ectodermal dysplasia, Christianson-Fourie type	Disorder	6 Case(s)
178303	8q22.1 microdeletion syndrome	Disorder	6 Case(s)
169464	Primary CD59 deficiency	Disorder	6 Case(s)
168984	CLAPO syndrome	Disorder	6 Case(s)
1661	X-linked corneal dermoid	Disorder	6 Case(s)
157820	Cold-induced sweating syndrome	Disorder	6 Case(s)
1307	Distal limb deficiencies-micrognathia syndrome	Disorder	6 Case(s)
1051	Ramos-Arroyo syndrome	Disorder	6 Case(s)
100071	Mosaic trisomy 3	Disorder	6 Case(s)
98676	Autosomal recessive isolated optic atrophy	Disorder	5 Case(s)
97341	Persistent placoid maculopathy	Disorder	5 Case(s)
94056	Isolated humero-ulnar synostosis	Disorder	5 Case(s)
93352	Spondyloepimetaphyseal dysplasia, Shohat type	Disorder	5 Case(s)
90399	Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms	Subtype of disorder	5 Case(s)
90301	Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome	Disorder	5 Case(s)
86915	Lymphedema-atrial septal defects-facial changes syndrome	Disorder	5 Case(s)
86914	Lymphedema-cerebral arteriovenous anomaly-primary pulmonary hypertension syndrome	Disorder	5 Case(s)
86821	Lissencephaly type 3-familial fetal akinesia sequence syndrome	Disorder	5 Case(s)
85297	X-linked spinocerebellar ataxia type 3	Disorder	5 Case(s)
85295	HSD10 disease, atypical type	Subtype of disorder	5 Case(s)
85284	BRESEK syndrome	Disorder	5 Case(s)
85280	X-linked intellectual disability-cubitus valgus-dysmorphism syndrome	Disorder	5 Case(s)
85175	Astley-Kendall dysplasia	Disorder	5 Case(s)
85165	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome	Disorder	5 Case(s)
85112	Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome	Disorder	5 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
84132	Desmin-related myopathy with Mallory body-like inclusions	Disorder	5 Case(s)
83472	CAMOS syndrome	Disorder	5 Case(s)
79095	Congenital bile acid synthesis defect type 4	Disorder	5 Case(s)
75374	Bradyopsia	Disorder	5 Case(s)
73272	Growth delay due to insulin-like growth factor type 1 deficiency	Disorder	5 Case(s)
65287	Beta-ureidopropionase deficiency	Disorder	5 Case(s)
631073	Autosomal recessive spastic paraplegia type 82	Disorder	5 Case(s)
596759	Combined immunodeficiency due to RELA haploinsufficiency	Disorder	5 Case(s)
572428	Infantile-onset pulmonary alveolar proteinosis-hypogammaglobulinemia	Disorder	5 Case(s)
569290	Multiple mitochondrial dysfunctions syndrome type 6	Disorder	5 Case(s)
566192	Congenital autosomal recessive small-platelet thrombocytopenia	Disorder	5 Case(s)
557056	Spastic ataxia-dysarthria due to glutaminase deficiency	Disorder	5 Case(s)
557003	Oculoskeletodental syndrome	Disorder	5 Case(s)
544488	Global developmental delay-alopecia-macrocephaly-facial dysmorphism-structural brain anomalies syndrome	Disorder	5 Case(s)
529965	Intellectual disability-autism-speech apraxia-craniofacial dysmorphism syndrome	Disorder	5 Case(s)
527450	Severe myopia-generalized joint laxity-short stature syndrome	Disorder	5 Case(s)
508542	Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome	Disorder	5 Case(s)
50815	Branchiogenic deafness syndrome	Disorder	5 Case(s)
505227	Combined immunodeficiency due to GINS1 deficiency	Disorder	5 Case(s)
500144	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	Disorder	5 Case(s)
500062	Infantile-onset periodic fever-panniculitis-dermatosis syndrome	Disorder	5 Case(s)
498251	Menstrual cycle-dependent periodic fever	Disorder	5 Case(s)
488635	Early-onset epilepsy-intellectual disability-brain anomalies syndrome	Disorder	5 Case(s)
488618	Transketolase deficiency	Disorder	5 Case(s)
488434	Camptodactyly syndrome, Guadalajara type 3	Disorder	5 Case(s)
488232	Split-foot malformation-mesoaxial polydactyly syndrome	Disorder	5 Case(s)
488168	Microcephaly-congenital cataract-psoriasiform dermatitis syndrome	Disorder	5 Case(s)
481665	USP18 deficiency	Disorder	5 Case(s)
480491	MYO5B-related progressive familial intrahepatic cholestasis	Subtype of disorder	5 Case(s)
466695	Supratip dysplasia	Disorder	5 Case(s)
464756	Familial gastric type 1 neuroendocrine tumor	Disorder	5 Case(s)
464440	Primary dystonia, DYT27 type	Disorder	5 Case(s)
464366	NEK9-related lethal skeletal dysplasia	Disorder	5 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
459056	Autosomal recessive spastic paraplegia type 75	Disorder	5 Case(s)
457284	Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome	Disorder	5 Case(s)
457212	Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	Disorder	5 Case(s)
447737	Combined immunodeficiency due to DOCK2 deficiency	Disorder	5 Case(s)
445062	Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	Disorder	5 Case(s)
444002	11q22.2q22.3 microdeletion syndrome	Disorder	5 Case(s)
423275	Spinocerebellar ataxia type 40	Disorder	5 Case(s)
402364	Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	Disorder	5 Case(s)
401986	1p31p32 microdeletion syndrome	Disorder	5 Case(s)
397593	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	Disorder	5 Case(s)
391487	STAT1-related autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome	Disorder	5 Case(s)
369847	Intellectual disability-hyperkinetic movement-truncal ataxia syndrome	Disorder	5 Case(s)
363665	Acroosteolysis-keloid-like lesions-premature aging syndrome	Disorder	5 Case(s)
363654	X-linked parkinsonism-spasticity syndrome	Disorder	5 Case(s)
363618	LMNA-related cardiocutaneous progeria syndrome	Disorder	5 Case(s)
363611	CTCF-related neurodevelopmental disorder	Disorder	5 Case(s)
356996	ANK3-related intellectual disability-sleep disturbance syndrome	Disorder	5 Case(s)
353320	Pyruvate carboxylase deficiency, benign type	Subtype of disorder	5 Case(s)
352718	Progressive retinal dystrophy due to retinol transport defect	Disorder	5 Case(s)
352596	Progressive myoclonic epilepsy with dystonia	Disorder	5 Case(s)
3383	Humerus trochlea aplasia	Disorder	5 Case(s)
3304	Fallot complex-intellectual disability-growth delay syndrome	Disorder	5 Case(s)
329173	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis	Disorder	5 Case(s)
3291	Teebi-Shaltout syndrome	Disorder	5 Case(s)
3238	Cardiospondylocarpofacial syndrome	Disorder	5 Case(s)
3230	Deafness-oligodontia syndrome	Disorder	5 Case(s)
3217	Deafness-small bowel diverticulosis-neuropathy syndrome	Disorder	5 Case(s)
320391	Autosomal recessive spastic paraplegia type 46	Disorder	5 Case(s)
320385	Hereditary sensory and autonomic neuropathy due to TECPR2 mutation	Disorder	5 Case(s)
320360	MT-ATP6-related mitochondrial spastic paraplegia	Disorder	5 Case(s)
319519	Combined oxidative phosphorylation defect type 14	Disorder	5 Case(s)
319160	Congenital myopathy with internal nuclei and atypical	Disorder	5 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	cores		
3180	Spondylocamptodactyly syndrome	Disorder	5 Case(s)
3168	Sillence syndrome	Disorder	5 Case(s)
3166	Sialuria	Disorder	5 Case(s)
3164	Omphalocele syndrome, Shprintzen-Goldberg type	Disorder	5 Case(s)
314652	Variant ABeta2M amyloidosis	Disorder	5 Case(s)
314555	Facial dysmorphism-ocular anomalies-osteopenia-intellectual disability-dental anomalies syndrome	Disorder	5 Case(s)
314034	7p22.1 microduplication syndrome	Disorder	5 Case(s)
3079	Intellectual disability, Buenos-Aires type	Disorder	5 Case(s)
300552	Follicular cholangitis and pancreatitis	Disorder	5 Case(s)
300504	Onychocytic matricoma	Disorder	5 Case(s)
300313	Congenital cataract-hearing loss-severe developmental delay syndrome	Disorder	5 Case(s)
3003	Pyknoachondrogenesis	Disorder	5 Case(s)
293964	Hypoinsulinemic hypoglycemia and body hemihypertrophy	Disorder	5 Case(s)
293955	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency	Disorder	5 Case(s)
293925	Lethal occipital encephalocele-skeletal dysplasia syndrome	Disorder	5 Case(s)
293462	Pre-Descemet corneal dystrophy	Disorder	5 Case(s)
2840	Pelvic dysplasia-arthrogryposis of lower limbs syndrome	Disorder	5 Case(s)
2831	Rhizomelic dysplasia, Patterson-Lowry type	Disorder	5 Case(s)
2819	Spastic paraplegia-facial-cutaneous lesions syndrome	Disorder	5 Case(s)
280403	Familial omphalocele syndrome with facial dysmorphism	Disorder	5 Case(s)
280183	Methylmalonic aciduria due to transcobalamin receptor defect	Disorder	5 Case(s)
2798	Pachygyria-intellectual disability-epilepsy syndrome	Disorder	5 Case(s)
2736	Lethal omphalocele-cleft palate syndrome	Disorder	5 Case(s)
2715	Severe oculo-renal-cerebellar syndrome	Disorder	5 Case(s)
2714	Oculo-palato-cerebral syndrome	Disorder	5 Case(s)
2703	Port-wine nevi-mega cisterna magna-hydrocephalus syndrome	Disorder	5 Case(s)
2672	Neuhauser-Eichner-Opitz syndrome	Disorder	5 Case(s)
2669	Nephrosis-deafness-urinary tract-digital malformations syndrome	Disorder	5 Case(s)
2668	Nephropathy-deafness-hyperparathyroidism syndrome	Disorder	5 Case(s)
264200	14q22q23 microdeletion syndrome	Disorder	5 Case(s)
261102	Distal 7q11.23 microduplication syndrome	Disorder	5 Case(s)
2571	X-linked immunoneurologic disorder	Disorder	5 Case(s)
2558	Mikati-Najjar-Sahli syndrome	Disorder	5 Case(s)
255235	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy	Disorder	5 Case(s)
2536	Microcornea-glaucoma-absent frontal sinuses syndrome	Disorder	5 Case(s)
2491	Müllerian duct anomalies-limb anomalies syndrome	Disorder	5 Case(s)
2432	Macrosomia-microphthalmia-cleft palate syndrome	Disorder	5 Case(s)
2325	Epidermolysis bullosa simplex with anodontia/hypodontia	Disorder	5 Case(s)
228390	Frontonasal dysplasia-alopecia-genital anomalies	Disorder	5 Case(s)

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	syndrome		
228240	Elastoderma	Disorder	5 Case(s)
228227	Late-onset focal dermal elastosis	Disorder	5 Case(s)
217026	Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type	Disorder	5 Case(s)
2158	Histidinuria-renal tubular defect syndrome	Disorder	5 Case(s)
2085	Glaucoma-sleep apnea syndrome	Disorder	5 Case(s)
2077	German syndrome	Disorder	5 Case(s)
206580	Autosomal recessive lower motor neuron disease with childhood onset	Disorder	5 Case(s)
2001	Cleft lip/palate-intestinal malrotation-cardiopathy syndrome	Disorder	5 Case(s)
199337	Pancreatic insufficiency-anemia-hyperostosis syndrome	Disorder	5 Case(s)
1811	Odontomicronychial dysplasia	Disorder	5 Case(s)
169079	Cernunnos-XLF deficiency	Disorder	5 Case(s)
168563	46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome	Disorder	5 Case(s)
168443	Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome	Disorder	5 Case(s)
1657	Dermatoosteolysis, Kirghizian type	Disorder	5 Case(s)
157962	Oculoauricular syndrome, Schorderet type	Disorder	5 Case(s)
157954	ANE syndrome	Disorder	5 Case(s)
156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type	Disorder	5 Case(s)
1566	Dandy-Walker malformation-postaxial polydactyly syndrome	Disorder	5 Case(s)
1514	Craniodigital-intellectual disability syndrome	Disorder	5 Case(s)
1415	Hardikar syndrome	Disorder	5 Case(s)
1129	Arachnodactyly-abnormal ossification-intellectual disability syndrome	Disorder	5 Case(s)
1113	Aphalangy-syndactyly-microcephaly syndrome	Disorder	5 Case(s)
101076	X-linked Charcot-Marie-Tooth disease type 2	Disorder	5 Case(s)
1005	Alopecia-contractures-dwarfism-intellectual disability syndrome	Disorder	5 Case(s)
96192	Paternal uniparental disomy of chromosome 7	Disorder	4 Case(s)
96188	Maternal uniparental disomy of chromosome 22	Disorder	4 Case(s)
93946	Hamel cerebro-palato-cardiac syndrome	Subtype of disorder	4 Case(s)
93405	Syndactyly type 4	Disorder	4 Case(s)
93333	Pelviscapular dysplasia	Disorder	4 Case(s)
921	Abruzzo-Erickson syndrome	Disorder	4 Case(s)
90023	Primary immunodeficiency syndrome due to P14/LAMTOR2 deficiency	Disorder	4 Case(s)
88635	Vacuolar myopathy with sarcoplasmic reticulum protein aggregates	Disorder	4 Case(s)
85326	X-linked intellectual disability, Stoll type	Disorder	4 Case(s)
85325	X-linked intellectual disability, Stevenson type	Disorder	4 Case(s)
85323	X-linked intellectual disability, Seemanova type	Disorder	4 Case(s)
85285	X-linked intellectual disability, Schimke type	Disorder	4 Case(s)

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85283	X-linked intellectual disability, Miles-Carpenter type	Disorder	4 Case(s)
85186	Endosteal sclerosis-cerebellar hypoplasia syndrome	Disorder	4 Case(s)
85184	Craniometadiaphyseal dysplasia, wormian bone type	Disorder	4 Case(s)
85172	Microcephalic osteodysplastic dysplasia, Saul-Wilson type	Disorder	4 Case(s)
806	Scott syndrome	Disorder	4 Case(s)
77295	Odontoleukodystrophy	Subtype of disorder	4 Case(s)
75391	Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency	Disorder	4 Case(s)
69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	Disorder	4 Case(s)
65743	Autosomal dominant multiple pterygium syndrome	Disorder	4 Case(s)
65288	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome	Disorder	4 Case(s)
619979	Developmental delay-immunodeficiency-leukoencephalopathy-hypohomocysteinemia syndrome	Disorder	4 Case(s)
617919	F12-associated cold autoinflammatory syndrome	Disorder	4 Case(s)
611256	Pontocerebellar hypoplasia type 12	Disorder	4 Case(s)
611237	Parkinsonism with polyneuropathy	Disorder	4 Case(s)
611223	EN1-related dorsoventral syndrome	Disorder	4 Case(s)
600668	CCNK-related neurodevelopmental disorder-severe intellectual disability-facial dysmorphism syndrome	Disorder	4 Case(s)
598603	Facial dysmorphism-hypertrichosis-epilepsy-intellectual disability/developmental delay-gingival overgrowth syndrome	Disorder	4 Case(s)
580933	Lethal brain and heart developmental defects	Disorder	4 Case(s)
566067	CEBPE-associated autoinflammation-immunodeficiency-neutrophil dysfunction syndrome	Disorder	4 Case(s)
557064	Neonatal epileptic encephalopathy due to glutaminase deficiency	Disorder	4 Case(s)
556955	Pancreatic agenesis-holoprosencephaly syndrome	Disorder	4 Case(s)
529574	Duane retraction syndrome with congenital deafness	Disorder	4 Case(s)
521438	Congenital vertebral-cardiac-renal anomalies syndrome	Disorder	4 Case(s)
521390	Spastic paraplegia-intellectual disability-nystagmus-obesity syndrome	Disorder	4 Case(s)
521305	Proximal myopathy with focal depletion of mitochondria	Disorder	4 Case(s)
506353	Autosomal recessive complex spastic paraplegia due to Kennedy pathway dysfunction	Disorder	4 Case(s)
505216	3-methylglutaconic aciduria type 9	Disorder	4 Case(s)
500188	X-linked external auditory canal atresia-dilated internal auditory canal-facial dysmorphism syndrome	Disorder	4 Case(s)
500159	Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrom	Disorder	4 Case(s)
500095	Tall stature-intellectual disability-renal anomalies syndrome	Disorder	4 Case(s)
498693	MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome	Disorder	4 Case(s)

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498488	Overgrowth syndrome with 2q37 translocation	Disorder	4 Case(s)
498485	Overgrowth-metaphyseal undermodeling-spondylar dysplasia syndrome	Disorder	4 Case(s)
497906	Childhood-onset basal ganglia degeneration syndrome	Disorder	4 Case(s)
495818	9q33.3q34.11 microdeletion syndrome	Disorder	4 Case(s)
486815	Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome	Disorder	4 Case(s)
485421	MFF-related encephalopathy due to mitochondrial and peroxisomal fission defect	Subtype of disorder	4 Case(s)
480682	POGLUT1-related limb-girdle muscular dystrophy R21	Disorder	4 Case(s)
480556	Isolated neonatal sclerosing cholangitis	Disorder	4 Case(s)
480536	MSH3-related attenuated familial adenomatous polyposis	Subtype of disorder	4 Case(s)
480476	Progressive familial intrahepatic cholestasis type 5	Subtype of disorder	4 Case(s)
478049	Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome	Disorder	4 Case(s)
476126	Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	Disorder	4 Case(s)
468717	Rhizomelic chondrodysplasia punctata type 5	Subtype of disorder	4 Case(s)
46059	Lathosterolosis	Disorder	4 Case(s)
459074	Corpus callosum agenesis-macrocephaly-hypertelorism syndrome	Disorder	4 Case(s)
457395	Progressive spondyloepimetaphyseal dysplasia-short stature-short fourth metatarsals-intellectual disability syndrome	Disorder	4 Case(s)
456328	X-linked myotubular myopathy-abnormal genitalia syndrome	Disorder	4 Case(s)
451612	Familial congenital nasolacrimal duct obstruction	Disorder	4 Case(s)
447893	Hypomyelination-cerebellar atrophy-hypoplasia of the corpus callosum syndrome	Subtype of disorder	4 Case(s)
447784	Mitochondrial pyruvate carrier deficiency	Disorder	4 Case(s)
444138	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	Disorder	4 Case(s)
444069	Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome	Disorder	4 Case(s)
443995	Mandibulofacial dysostosis with alopecia	Disorder	4 Case(s)
438134	PCNA-related progressive neurodegenerative photosensitivity syndrome	Disorder	4 Case(s)
438114	RARS-related autosomal recessive hypomyelinating leukodystrophy	Disorder	4 Case(s)
436166	Periodic fever-infantile enterocolitis-autoinflammatory syndrome	Disorder	4 Case(s)
435998	Autosomal recessive intermediate Charcot-Marie-Tooth disease type D	Disorder	4 Case(s)
435660	LIPE-related familial partial lipodystrophy	Disorder	4 Case(s)
424027	Progressive myoclonic epilepsy type 8	Disorder	4 Case(s)
420702	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency	Disorder	4 Case(s)

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412069	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	Disorder	4 Case(s)
404473	Severe intellectual disability-progressive spastic diplegia syndrome	Disorder	4 Case(s)
404466	Female infertility due to zona pellucida defect	Disorder	4 Case(s)
404437	Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	Disorder	4 Case(s)
401979	Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type	Disorder	4 Case(s)
401948	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	Disorder	4 Case(s)
401862	Lipoyl transferase 1 deficiency	Disorder	4 Case(s)
401835	Autosomal recessive spastic paraplegia type 70	Disorder	4 Case(s)
401810	Autosomal recessive spastic paraplegia type 64	Disorder	4 Case(s)
401780	Autosomal recessive spastic paraplegia type 61	Disorder	4 Case(s)
398079	SIM1-related Prader-Willi-like syndrome	Disorder	4 Case(s)
397951	Microcephaly-thin corpus callosum-intellectual disability syndrome	Disorder	4 Case(s)
397927	Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome	Disorder	4 Case(s)
397755	Periodic paralysis with transient compartment-like syndrome	Disorder	4 Case(s)
397623	Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome	Disorder	4 Case(s)
370052	SCALP syndrome	Disorder	4 Case(s)
369942	CADDS	Disorder	4 Case(s)
369837	Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	Disorder	4 Case(s)
364577	Intellectual disability-brachydactyly-Pierre Robin syndrome	Disorder	4 Case(s)
363969	Autosomal recessive cerebral atrophy	Disorder	4 Case(s)
363965	Koolen-De Vries syndrome due to a point mutation	Subtype of disorder	4 Case(s)
363705	Craniofaciofrontodigital syndrome	Disorder	4 Case(s)
363686	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	Disorder	4 Case(s)
363444	THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	Disorder	4 Case(s)
357175	Short ulna-dysmorphism-hypotonia-intellectual disability syndrome	Disorder	4 Case(s)
356961	SLC35A2-CDG	Disorder	4 Case(s)
356947	3q26q27 microdeletion syndrome	Disorder	4 Case(s)
352470	DNA2-related mitochondrial DNA deletion syndrome	Disorder	4 Case(s)
3355	Trichoodontoonychial dysplasia	Disorder	4 Case(s)
329332	Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome	Disorder	4 Case(s)
3270	Radioulnar synostosis-developmental delay-hypotonia syndrome	Disorder	4 Case(s)
324581	Benign Samaritan congenital myopathy	Disorder	4 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
324313	9p13 microdeletion syndrome	Disorder	4 Case(s)
3232	Deafness-ear malformation-facial palsy syndrome	Disorder	4 Case(s)
3207	White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome	Disorder	4 Case(s)
319509	Combined oxidative phosphorylation defect type 9	Disorder	4 Case(s)
319195	Chondroectodermal dysplasia with night blindness	Disorder	4 Case(s)
3186	Holoprosencephaly-radial heart renal anomalies syndrome	Disorder	4 Case(s)
314721	Atypical dentin dysplasia due to SMOC2 deficiency	Subtype of disorder	4 Case(s)
314632	CLN12 disease	Disorder	4 Case(s)
314381	Hereditary sensory and autonomic neuropathy type 6	Disorder	4 Case(s)
313795	Jawad syndrome	Disorder	4 Case(s)
313781	20p13 microdeletion syndrome	Disorder	4 Case(s)
3132	Say-Barber-Miller syndrome	Disorder	4 Case(s)
3101	Richieri Costa-da Silva syndrome	Disorder	4 Case(s)
3088	Revesz syndrome	Disorder	4 Case(s)
306550	FADD-related immunodeficiency	Disorder	4 Case(s)
3055	X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome	Disorder	4 Case(s)
3052	X-linked intellectual disability-seizures-psoriasis syndrome	Disorder	4 Case(s)
3044	Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome	Disorder	4 Case(s)
3035	Growth delay-hydrocephaly-lung hypoplasia syndrome	Disorder	4 Case(s)
3015	Radio-renal syndrome	Disorder	4 Case(s)
300501	Painful orbital and systemic neurofibromas-marfanoid habitus syndrome	Disorder	4 Case(s)
2972	Non-eruption of teeth-maxillary hypoplasia-genu valgum syndrome	Disorder	4 Case(s)
2946	Brachydactyly-long thumb syndrome	Disorder	4 Case(s)
293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome	Disorder	4 Case(s)
293825	Congenital dyserythropoietic anemia type IV	Disorder	4 Case(s)
2865	Short stature-webbed neck-heart disease syndrome	Disorder	4 Case(s)
284339	Pontocerebellar hypoplasia type 7	Disorder	4 Case(s)
2838	Renal caliceal diverticuli-deafness syndrome	Disorder	4 Case(s)
2820	Spastic paraplegia-nephritis-deafness syndrome	Disorder	4 Case(s)
280654	Autosomal recessive nail dysplasia	Disorder	4 Case(s)
280598	Hereditary sensorimotor neuropathy with hyperelastic skin	Disorder	4 Case(s)
280586	Chondrodysplasia with joint dislocations, gPAPP type	Disorder	4 Case(s)
280558	Warsaw breakage syndrome	Disorder	4 Case(s)
280142	Severe combined immunodeficiency due to LCK deficiency	Disorder	4 Case(s)
2769	Familial osteodysplasia, Anderson type	Disorder	4 Case(s)
2730	Postaxial tetramelic oligodactyly	Disorder	4 Case(s)
2723	Odontotrichomelic syndrome	Disorder	4 Case(s)
263410	Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome	Disorder	4 Case(s)
2589	Myoclonus-cerebellar ataxia-deafness syndrome	Disorder	4 Case(s)
2570	Lethal intrauterine growth restriction-cortical	Disorder	4 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	malformation-congenital contractures syndrome		
251304	Infantile onset panniculitis with uveitis and systemic granulomatosis	Disorder	4 Case(s)
251056	6q25.2q25.3 microdeletion syndrome	Disorder	4 Case(s)
250977	AICA-ribosiduria	Disorder	4 Case(s)
250972	Polymicrogyria with optic nerve hypoplasia	Disorder	4 Case(s)
2497	Upper limb mesomelic dysplasia, type Fryns	Disorder	4 Case(s)
247827	Ectodermal dysplasia-hyperhidrosis-cutaneous syndactyly syndrome	Disorder	4 Case(s)
247790	FTH1-related iron overload	Disorder	4 Case(s)
247604	Juvenile primary lateral sclerosis	Disorder	4 Case(s)
2463	Marfanoid habitus-autosomal recessive intellectual disability syndrome	Disorder	4 Case(s)
2412	Dislocation of the hip-dysmorphism syndrome	Disorder	4 Case(s)
2386	Leukoencephalopathy-palmoplantar keratoderma syndrome	Disorder	4 Case(s)
228399	8q12 microduplication syndrome	Disorder	4 Case(s)
228374	Charcot-Marie-Tooth disease type 2B5	Disorder	4 Case(s)
2278	Ichthyosis-intellectual disability-dwarfism-renal impairment syndrome	Disorder	4 Case(s)
2269	Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome	Disorder	4 Case(s)
2218	Cervical hypertrichosis-peripheral neuropathy syndrome	Disorder	4 Case(s)
2215	Multiple pterygium-malignant hyperthermia syndrome	Disorder	4 Case(s)
217407	Hereditary hypotrichosis with recurrent skin vesicles	Disorder	4 Case(s)
217396	Progressive polyneuropathy with bilateral striatal necrosis	Disorder	4 Case(s)
2150	Hirschsprung disease-type D brachydactyly syndrome	Disorder	4 Case(s)
210163	Congenital lethal myopathy, Compton-North type	Disorder	4 Case(s)
210136	Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome	Disorder	4 Case(s)
210128	Urocanic aciduria	Disorder	4 Case(s)
209967	Episodic ataxia type 6	Disorder	4 Case(s)
2031	Hepatic fibrosis-renal cysts-intellectual disability syndrome	Disorder	4 Case(s)
1973	Faciocardiorenal syndrome	Disorder	4 Case(s)
1952	Epiphyseal stippling-osteoclastic hyperplasia syndrome	Disorder	4 Case(s)
1816	Leukomelanoderma-infantilism-intellectual disability-hypodontia-hypotrichosis syndrome	Disorder	4 Case(s)
1809	Hidrotic ectodermal dysplasia, Halal type	Disorder	4 Case(s)
1794	Oculomaxillofacial dysostosis	Disorder	4 Case(s)
1787	Acrofacial dysostosis, Palagonia type	Disorder	4 Case(s)
178400	Distal myopathy with anterior tibial onset	Disorder	4 Case(s)
178396	Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation	Disorder	4 Case(s)
1768	Familial caudal dysgenesis	Disorder	4 Case(s)
171844	Blindness-scoliosis-arachnodactyly syndrome	Disorder	4 Case(s)
171703	Microcephaly-polymicrogyria-corpus callosum agenesis syndrome	Disorder	4 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
1682	Arterial dissection-lentiginosis syndrome	Disorder	4 Case(s)
166024	Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome	Disorder	4 Case(s)
165805	Familial mesial temporal lobe epilepsy with febrile seizures	Disorder	4 Case(s)
163985	Hyperekplexia-epilepsy syndrome	Disorder	4 Case(s)
163971	X-linked intellectual disability, Cilliers type	Disorder	4 Case(s)
163668	Spondyloepiphyseal dysplasia, MacDermot type	Disorder	4 Case(s)
163654	Spondyloepiphyseal dysplasia-brachydactyly-speech disorder syndrome	Disorder	4 Case(s)
163649	Spondyloepiphyseal dysplasia-craniosynostosis-cleft palate-cataracts-intellectual disability syndrome	Disorder	4 Case(s)
158687	Lethal acantholytic erosive disorder	Disorder	4 Case(s)
157832	Craniorhiny	Disorder	4 Case(s)
1538	Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome	Disorder	4 Case(s)
1528	Craniotelencephalic dysplasia	Disorder	4 Case(s)
1508	Coxoauricular syndrome	Disorder	4 Case(s)
1436	X-linked skeletal dysplasia-intellectual disability syndrome	Disorder	4 Case(s)
1423	Lethal recessive chondrodysplasia	Disorder	4 Case(s)
140976	RHYNS syndrome	Disorder	4 Case(s)
1406	Charlie M syndrome	Disorder	4 Case(s)
139573	Hereditary sensory and autonomic neuropathy with deafness and global delay	Disorder	4 Case(s)
137908	Hypotonia with lactic acidemia and hyperammonemia	Disorder	4 Case(s)
137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency	Disorder	4 Case(s)
1338	Heart defect-tongue hamartoma-polysyndactyly syndrome	Disorder	4 Case(s)
1323	Camptodactyly-joint contractures-facial skeletal defects syndrome	Disorder	4 Case(s)
1296	Lambert syndrome	Disorder	4 Case(s)
1261	Bonnemann-Meinecke-Reich syndrome	Disorder	4 Case(s)
1135	Arrhinia-choanal atresia-microphthalmia syndrome	Disorder	4 Case(s)
1117	Aplasia cutis-myopia syndrome	Disorder	4 Case(s)
1110	Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome	Disorder	4 Case(s)
1094	Anonychia-microcephaly syndrome	Disorder	4 Case(s)
97678	Maternal uniparental disomy of chromosome 13	Disorder	3 Case(s)
95700	Familial adrenal hypoplasia with absent pituitary luteinizing hormone	Disorder	3 Case(s)
93947	X-linked intellectual disability, Golabi-Ito-Hall type	Subtype of disorder	3 Case(s)
93267	Cloverleaf skull-multiple congenital anomalies syndrome	Disorder	3 Case(s)
90030	Hemolytic anemia due to glutathione reductase deficiency	Disorder	3 Case(s)
85324	X-linked intellectual disability, Shrimpton type	Disorder	3 Case(s)
85321	Deafness-intellectual disability syndrome, Martin-Probst type	Disorder	3 Case(s)
85317	X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome	Disorder	3 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
85290	X-linked intellectual disability, Wilson type	Disorder	3 Case(s)
83642	Microcytic anemia with liver iron overload	Disorder	3 Case(s)
83617	Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	Disorder	3 Case(s)
79347	Chondrodysplasia punctata, Toriello type	Disorder	3 Case(s)
79330	MOGS-CDG	Disorder	3 Case(s)
79118	Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome	Disorder	3 Case(s)
77299	Microphthalmia-brain atrophy syndrome	Disorder	3 Case(s)
75325	Osteosclerosis-ichthyosis-premature ovarian failure syndrome	Disorder	3 Case(s)
73223	Global developmental delay-osteopenia-ectodermal defect syndrome	Disorder	3 Case(s)
71278	Congenital brain dysgenesis due to glutamine synthetase deficiency	Disorder	3 Case(s)
69125	Anonychia with flexural pigmentation	Disorder	3 Case(s)
66633	Sensorineural hearing loss-early graying-essential tremor syndrome	Disorder	3 Case(s)
637064	Isolated optic nerve aplasia	Disorder	3 Case(s)
629	Short stature due to growth hormone qualitative anomaly	Subtype of disorder	3 Case(s)
619941	Congenital neutropenia-combined immunodeficiency due to MKL1 deficiency	Disorder	3 Case(s)
613267	Pontocerebellar hypoplasia type 13	Disorder	3 Case(s)
610573	CLCN6-related childhood-onset progressive neurodegeneration-peripheral neuropathy syndrome	Disorder	3 Case(s)
603684	KLHL7-related Bohring-Opitz-like and Crisponi/Cold-induced sweating-like overlap syndrome	Disorder	3 Case(s)
599519	Factor V short isoforms-related bleeding disorder	Disorder	3 Case(s)
597874	MTHFS-related developmental delay-microcephaly-short stature-epilepsy syndrome	Disorder	3 Case(s)
565899	POMGNT2-related limb-girdle muscular dystrophy R24	Disorder	3 Case(s)
565858	Craniosynostosis-microretrognathia-severe intellectual disability syndrome	Disorder	3 Case(s)
565788	Infantile inflammatory bowel disease with neurological involvement	Disorder	3 Case(s)
564178	Primary hypomagnesemia-refractory seizures-intellectual disability syndrome	Disorder	3 Case(s)
562509	Heme oxygenase-1 deficiency	Disorder	3 Case(s)
544503	RNF13-related severe early-onset epileptic encephalopathy	Disorder	3 Case(s)
52054	Craniosynostosis-intracranial calcifications syndrome	Disorder	3 Case(s)
508501	Oral-facial-digital syndrome with short stature and brachymesophalangy	Disorder	3 Case(s)
50811	Lipodystrophy-intellectual disability-deafness syndrome	Disorder	3 Case(s)
504523	Severe combined immunodeficiency due to LAT deficiency	Disorder	3 Case(s)
502437	4q25 proximal deletion syndrome	Disorder	3 Case(s)
500135	Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome	Disorder	3 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
496751	EVEN-plus syndrome	Disorder	3 Case(s)
496686	Kyphosis-lateral tongue atrophy-myofibrillar myopathy syndrome	Disorder	3 Case(s)
495875	Congenital labioscrotal agenesis-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome	Disorder	3 Case(s)
494541	Childhood-onset benign chorea with striatal involvement	Disorder	3 Case(s)
494439	Retinitis pigmentosa-hearing loss-premature aging-short stature-facial dysmorphism syndrome	Disorder	3 Case(s)
488627	Severe growth deficiency-strabismus-extensive dermal melanocytosis-intellectual disability syndrome	Disorder	3 Case(s)
485418	EMILIN-1-related connective tissue disease	Disorder	3 Case(s)
485405	16p12.1p12.3 triplication syndrome	Disorder	3 Case(s)
477993	Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	Disorder	3 Case(s)
477774	Combined oxidative phosphorylation defect type 27	Disorder	3 Case(s)
477661	IL21-related infantile inflammatory bowel disease	Disorder	3 Case(s)
476096	Erythrokeratodermia-cardiomyopathy syndrome	Disorder	3 Case(s)
476084	BVES-related limb-girdle muscular dystrophy	Disorder	3 Case(s)
468620	Intellectual disability-epilepsy-extrapyramidal syndrome	Disorder	3 Case(s)
466794	Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome	Disorder	3 Case(s)
466784	Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect	Disorder	3 Case(s)
457365	Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome	Disorder	3 Case(s)
453533	Polyendocrine-polyneuropathy syndrome	Disorder	3 Case(s)
453510	Congenital insensitivity to pain with severe intellectual disability	Disorder	3 Case(s)
444458	Combined oxidative phosphorylation defect type 24	Disorder	3 Case(s)
444048	46,XX ovarian dysgenesis-short stature syndrome	Disorder	3 Case(s)
438178	Fatty acyl-CoA reductase 1 deficiency	Disorder	3 Case(s)
437552	Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity	Disorder	3 Case(s)
436245	Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome	Disorder	3 Case(s)
436174	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	Disorder	3 Case(s)
435953	Progeroid features-hepatocellular carcinoma predisposition syndrome	Disorder	3 Case(s)
435938	X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	Disorder	3 Case(s)
435930	Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	Disorder	3 Case(s)
435628	Keppen-Lubinsky syndrome	Disorder	3 Case(s)
424261	TOR1AIP1-related limb-girdle muscular dystrophy	Disorder	3 Case(s)
423894	Microcephaly-complex motor and sensory axonal neuropathy syndrome	Disorder	3 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
420794	Cono-spondylar dysplasia	Disorder	3 Case(s)
420566	Bleeding disorder due to CalDAG-GEFI deficiency	Disorder	3 Case(s)
412189	Epidermolysis bullosa simplex due to exophilin 5 deficiency	Disorder	3 Case(s)
412035	13q12.3 microdeletion syndrome	Disorder	3 Case(s)
411986	Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	Disorder	3 Case(s)
404493	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency	Disorder	3 Case(s)
404451	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome	Disorder	3 Case(s)
402082	Progressive myoclonic epilepsy type 5	Disorder	3 Case(s)
401935	14q24.1q24.3 microdeletion syndrome	Disorder	3 Case(s)
401866	Childhood-onset spasticity with hyperglycinemia	Disorder	3 Case(s)
401859	Lipoic acid synthetase deficiency	Disorder	3 Case(s)
401795	Autosomal recessive spastic paraplegia type 59	Disorder	3 Case(s)
401764	Pancytopenia-developmental delay syndrome	Disorder	3 Case(s)
398117	Neonatal dermatomyositis	Disorder	3 Case(s)
397964	Combined immunodeficiency due to MALT1 deficiency	Disorder	3 Case(s)
397933	Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	Disorder	3 Case(s)
397922	Ferro-cerebro-cutaneous syndrome	Disorder	3 Case(s)
391457	HSD10 disease, neonatal type	Subtype of disorder	3 Case(s)
391397	Hereditary sensory and autonomic neuropathy type 7	Disorder	3 Case(s)
391351	SURF1-related Charcot-Marie-Tooth disease type 4	Disorder	3 Case(s)
391316	Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression	Disorder	3 Case(s)
391307	Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome	Disorder	3 Case(s)
370127	Medich giant platelet syndrome	Disorder	3 Case(s)
370103	Primary dystonia, DYT17 type	Disorder	3 Case(s)
370010	Intellectual disability-facial dysmorphism-hand anomalies syndrome	Disorder	3 Case(s)
369992	Severe dermatitis-multiple allergies-metabolic wasting syndrome	Disorder	3 Case(s)
369867	Autosomal recessive intermediate Charcot-Marie-Tooth disease type C	Disorder	3 Case(s)
369840	TRAPPC11-related limb-girdle muscular dystrophy R18	Disorder	3 Case(s)
363981	Charcot-Marie-Tooth disease type 4B3	Disorder	3 Case(s)
363534	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form	Disorder	3 Case(s)
363409	Fetal akinesia-cerebral and retinal hemorrhage syndrome	Disorder	3 Case(s)
357237	Combined immunodeficiency due to CARD11 deficiency	Disorder	3 Case(s)
3433	Microcephaly-brachydactyly-kyphoscoliosis syndrome	Disorder	3 Case(s)
3409	Urban-Rogers-Meyer syndrome	Disorder	3 Case(s)
3404	Ulbright-Hodes syndrome	Disorder	3 Case(s)
3369	Trigonocephaly-short stature-developmental delay	Disorder	3 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	syndrome		
3353	Trichodermodysplasia-dental alterations syndrome	Disorder	3 Case(s)
3328	Absent tibia-polydactyly-arachnoid cyst syndrome	Disorder	3 Case(s)
3326	Thymic-renal-anal-lung dysplasia	Disorder	3 Case(s)
330054	Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome	Disorder	3 Case(s)
329178	Congenital muscular dystrophy with intellectual disability and severe epilepsy	Disorder	3 Case(s)
324525	Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation	Disorder	3 Case(s)
324290	Early-onset Lafora body disease	Disorder	3 Case(s)
3236	Conductive deafness-ptosis-skeletal anomalies syndrome	Disorder	3 Case(s)
320401	Autosomal recessive spastic paraplegia type 44	Disorder	3 Case(s)
3172	Eyebrow duplication-syndactyly syndrome	Disorder	3 Case(s)
314572	Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome	Disorder	3 Case(s)
314485	Young adult-onset distal hereditary motor neuropathy	Disorder	3 Case(s)
314389	Xq12-q13.3 duplication syndrome	Disorder	3 Case(s)
313800	Retinal dystrophy-optic nerve edema-splenomegaly-anhidrosis-migraine headache syndrome	Disorder	3 Case(s)
3104	Robin sequence-oligodactyly syndrome	Disorder	3 Case(s)
3098	Rhizomelic syndrome, Urbach type	Disorder	3 Case(s)
309147	Hyper-beta-alaninemia	Disorder	3 Case(s)
309111	Combined pancreatic lipase-colipase deficiency	Disorder	3 Case(s)
3086	Autosomal dominant vitreoretinochoroidopathy	Disorder	3 Case(s)
306542	Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome	Disorder	3 Case(s)
306504	Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome	Disorder	3 Case(s)
3041	Intellectual disability-balding-patella luxation-acromicria syndrome	Disorder	3 Case(s)
3026	Radial ray hypoplasia-choanal atresia syndrome	Disorder	3 Case(s)
3018	Retinal ischemic syndrome-digestive tract small vessel hyalinosis-diffuse cerebral calcifications syndrome	Disorder	3 Case(s)
3010	Qazi-Markouizos syndrome	Disorder	3 Case(s)
300333	Nephrotic syndrome-epidermolysis bullosa-sensorineural deafness syndrome	Disorder	3 Case(s)
300298	Severe congenital hypochromic anemia with ringed sideroblasts	Disorder	3 Case(s)
2983	Difference of sex development-intellectual disability syndrome	Disorder	3 Case(s)
2957	Guttmacher syndrome	Disorder	3 Case(s)
2951	Absent thumb-short stature-immunodeficiency syndrome	Disorder	3 Case(s)
294023	Neonatal inflammatory skin and bowel disease	Disorder	3 Case(s)
2928	Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome	Disorder	3 Case(s)
2926	Digital extensor muscle aplasia-polyneuropathy	Disorder	3 Case(s)

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2916	Postaxial polydactyly-dental and vertebral anomalies syndrome	Disorder	3 Case(s)
2881	Cutaneous photosensitivity-lethal colitis syndrome	Disorder	3 Case(s)
2868	Short stature-valvular heart disease-characteristic facies syndrome	Disorder	3 Case(s)
2863	Short stature-wormian bones-dextrocardia syndrome	Disorder	3 Case(s)
280640	Occipital pachygyria and polymicrogyria	Disorder	3 Case(s)
280356	PLIN1-related familial partial lipodystrophy	Disorder	3 Case(s)
2779	Osteopathia striata-pigmentary dermopathy-white forelock syndrome	Disorder	3 Case(s)
2772	Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome	Disorder	3 Case(s)
2760	OSLAM syndrome	Disorder	3 Case(s)
2741	Ophthalmomandibulomelic dysplasia	Disorder	3 Case(s)
2724	Odontomatosis-aortae esophagus stenosis syndrome	Disorder	3 Case(s)
2713	Oculoosteocutaneous syndrome	Disorder	3 Case(s)
2690	Neutropenia-monocytopenia-deafness syndrome	Disorder	3 Case(s)
2673	Neurofaciodigitorenal syndrome	Disorder	3 Case(s)
263508	COG1-CDG	Disorder	3 Case(s)
2617	Microcephalic primordial dwarfism, Montreal type	Disorder	3 Case(s)
2613	Nail-patella-like renal disease	Disorder	3 Case(s)
261295	20p12.3 microdeletion syndrome	Disorder	3 Case(s)
261144	FOXG1 syndrome due to 14q12 microdeletion	Subtype of disorder	3 Case(s)
261120	14q11.2 microdeletion syndrome	Disorder	3 Case(s)
2608	N syndrome	Disorder	3 Case(s)
2523	Microcephaly-brain defect-spasticity-hypernatremia syndrome	Disorder	3 Case(s)
2521	Microcephaly-cleft palate-abnormal retinal pigmentation syndrome	Disorder	3 Case(s)
2516	Microcephaly-cardiac defect-lung malsegmentation syndrome	Disorder	3 Case(s)
2515	Microcephaly-cardiomyopathy syndrome	Disorder	3 Case(s)
251066	8p11.2 deletion syndrome	Disorder	3 Case(s)
2502	Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome	Disorder	3 Case(s)
2476	Dysraphism-cleft lip/palate-limb reduction defects syndrome	Disorder	3 Case(s)
2437	Czeizel-Losonci syndrome	Disorder	3 Case(s)
2410	Hypergonadotropic hypogonadism-cataract syndrome	Disorder	3 Case(s)
2409	Lowry-MacLean syndrome	Disorder	3 Case(s)
238459	SLC35A1-CDG	Disorder	3 Case(s)
2370	Larsen-like osseous dysplasia-short stature syndrome	Disorder	3 Case(s)
228396	Ptosis-upper ocular movement limitation-absence of lacrimal punctum syndrome	Disorder	3 Case(s)
2261	Hypospadias-intellectual disability, Goldblatt type syndrome	Disorder	3 Case(s)
2246	Cerebellar hypoplasia-tapetoretinal degeneration	Disorder	3 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	syndrome		
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency	Disorder	3 Case(s)
217017	Zechi-Ceide syndrome	Disorder	3 Case(s)
2167	Holzgreve syndrome	Disorder	3 Case(s)
2153	Hirschsprung disease-nail hypoplasia-dysmorphism syndrome	Disorder	3 Case(s)
2135	Cutaneous mastocytosis-deafness-microtia syndrome	Disorder	3 Case(s)
2115	Harrod syndrome	Disorder	3 Case(s)
2111	Cystic hamartoma of lung and kidney	Disorder	3 Case(s)
2101	Grubben-de Cock-Borghgraef syndrome	Disorder	3 Case(s)
2091	Multinodular goiter-cystic kidney-polydactyly syndrome	Disorder	3 Case(s)
2084	Glaucoma-ectopia lentis-microspherophakia-stiff joints-short stature syndrome	Disorder	3 Case(s)
2064	Posterior fusion of lumbosacral vertebrae-blepharoptosis syndrome	Disorder	3 Case(s)
2050	Cole-Carpenter syndrome	Disorder	3 Case(s)
1972	Lethal faciocardiomelic dysplasia	Disorder	3 Case(s)
1970	Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome	Disorder	3 Case(s)
1969	Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome	Disorder	3 Case(s)
1943	Early-onset progressive encephalopathy with migrant continuous myoclonus	Disorder	3 Case(s)
1891	Intellectual disability-spasticity-ectrodactyly syndrome	Disorder	3 Case(s)
1882	Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome	Disorder	3 Case(s)
1837	Metaphyseal chondrodysplasia, Rosenberg type	Disorder	3 Case(s)
1790	Hypomandibular faciocranial dysostosis	Disorder	3 Case(s)
171866	Spondyloepimetaphyseal dysplasia, aggrecan type	Disorder	3 Case(s)
169157	T-B+ severe combined immunodeficiency due to CD45 deficiency	Disorder	3 Case(s)
168577	Hereditary cryohydrocytosis with reduced stomatin	Disorder	3 Case(s)
168555	Spondylometaphyseal dysplasia, A4 type	Disorder	3 Case(s)
168544	Spondylometaphyseal dysplasia, Golden type	Disorder	3 Case(s)
166277	Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia	Disorder	3 Case(s)
166105	FASTKD2-related infantile mitochondrial encephalomyopathy	Disorder	3 Case(s)
166029	Multiple epiphyseal dysplasia-severe proximal femoral dysplasia syndrome	Disorder	3 Case(s)
163961	X-linked cerebral-cerebellar-coloboma syndrome	Disorder	3 Case(s)
163665	Spondyloepiphyseal dysplasia tarda, Kohn type	Disorder	3 Case(s)
1548	Cryptorchidism-arachnodactyly-intellectual disability syndrome	Disorder	3 Case(s)
1529	Craniofacial-deafness-hand syndrome	Disorder	3 Case(s)
1495	Intellectual disability-hypoplastic corpus callosum-	Disorder	3 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	preauricular tag syndrome		
139466	SERKAL syndrome	Disorder	3 Case(s)
139414	Congenital panfollicular nevus	Disorder	3 Case(s)
1389	Cortical blindness-intellectual disability-polydactyly syndrome	Disorder	3 Case(s)
1383	Cataract-deafness-hypogonadism syndrome	Disorder	3 Case(s)
1381	Cataract-intellectual disability-anal atresia-urinary defects syndrome	Disorder	3 Case(s)
137622	Intractable diarrhea-choanal atresia-eye anomalies syndrome	Disorder	3 Case(s)
1373	Cataract-aberrant oral frenula-growth delay syndrome	Disorder	3 Case(s)
1355	Congenital heart defect-round face-developmental delay syndrome	Disorder	3 Case(s)
1259	Blepharoptosis-myopia-ectopia lentis syndrome	Disorder	3 Case(s)
1185	Spinocerebellar ataxia-dysmorphism syndrome	Disorder	3 Case(s)
1133	AREDYLD syndrome	Disorder	3 Case(s)
1130	Arachnodactyly-intellectual disability-dysmorphism syndrome	Disorder	3 Case(s)
1116	Aplasia cutis congenita-intestinal lymphangiectasia syndrome	Disorder	3 Case(s)
1112	Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome	Disorder	3 Case(s)
1101	Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome	Disorder	3 Case(s)
1069	Aniridia-absent patella syndrome	Disorder	3 Case(s)
1067	Aniridia-ptosis-intellectual disability-familial obesity syndrome	Disorder	3 Case(s)
103910	Congenital enterocyte heparan sulfate deficiency	Disorder	3 Case(s)
1006	Alopecia antibody deficiency	Disorder	3 Case(s)
99832	Resistance to thyrotropin-releasing hormone syndrome	Disorder	2 Case(s)
977	Adrenomyodystrophy	Disorder	2 Case(s)
97290	Familial papillary thyroid carcinoma with renal papillary neoplasia	Disorder	2 Case(s)
96187	Maternal uniparental disomy of chromosome 21	Disorder	2 Case(s)
95428	COG8-CDG	Disorder	2 Case(s)
949	Acrocraniofacial dysostosis	Disorder	2 Case(s)
94066	Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia	Disorder	2 Case(s)
91494	Macular coloboma-cleft palate-hallux valgus syndrome	Disorder	2 Case(s)
91130	Cardiomyopathy-hypotonia-lactic acidosis syndrome	Disorder	2 Case(s)
88643	Obesity-colitis-hypothyroidism-cardiac hypertrophy-developmental delay syndrome	Disorder	2 Case(s)
86822	Lissencephaly type 3-metacarpal bone dysplasia syndrome	Disorder	2 Case(s)
85327	X-linked intellectual disability-acromegaly-hyperactivity syndrome	Disorder	2 Case(s)
85319	X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
79507	Hypotonia-failure to thrive-microcephaly syndrome	Disorder	2 Case(s)
79302	Congenital bile acid synthesis defect type 3	Disorder	2 Case(s)
79107	Developmental malformations-deafness-dystonia syndrome	Disorder	2 Case(s)
785	Estrogen resistance syndrome	Disorder	2 Case(s)
77300	Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome	Disorder	2 Case(s)
75389	Brain malformation-congenital heart disease-postaxial polydactyly syndrome	Disorder	2 Case(s)
73246	Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome	Disorder	2 Case(s)
73245	Spinal muscular atrophy-Dandy-Walker malformation-cataracts syndrome	Disorder	2 Case(s)
73230	Ossification anomalies-psychomotor developmental delay syndrome	Disorder	2 Case(s)
73224	Kidney tubulopathy-dilated cardiomyopathy syndrome	Disorder	2 Case(s)
71267	Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome	Disorder	2 Case(s)
69088	Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome	Disorder	2 Case(s)
64542	Acrofacial dysostosis, Kennedy-Teebi type	Disorder	2 Case(s)
631079	Autosomal recessive spastic paraplegia type 84	Disorder	2 Case(s)
603494	Coloboma-osteopetrosis-microphthalmia-macrocephaly-albinism-deafness syndrome	Disorder	2 Case(s)
599579	Factor V Amsterdam bleeding disorder	Subtype of disorder	2 Case(s)
597887	ALPI-related inflammatory bowel disease	Disorder	2 Case(s)
597733	Oculocutaneous albinism type 8	Disorder	2 Case(s)
589435	Spondylometaphyseal dysplasia-corneal dystrophy syndrome	Disorder	2 Case(s)
568056	Warts-immunodeficiency-lymphedema-anogenital dysplasia syndrome	Disorder	2 Case(s)
544602	Congenital myopathy with reduced type 2 muscle fibers	Disorder	2 Case(s)
527468	Diaphragmatic hernia-short bowel-asplenia syndrome	Disorder	2 Case(s)
521432	Congenital cataract-severe neonatal hepatopathy-global developmental delay syndrome	Disorder	2 Case(s)
521411	Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect	Disorder	2 Case(s)
52055	Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome	Disorder	2 Case(s)
52047	Braddock syndrome	Disorder	2 Case(s)
508512	Intrauterine growth restriction-congenital multiple café-au-lait macules-increased sister chromatid exchange syndrome	Disorder	2 Case(s)
508488	8q24.3 microdeletion syndrome	Disorder	2 Case(s)
50817	Duane anomaly-myopathy-scoliosis syndrome	Disorder	2 Case(s)
50812	Zellweger-like syndrome without peroxisomal anomalies	Disorder	2 Case(s)
50810	Microlissencephaly-micromelia syndrome	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
50809	Talo-patello-scaphoid osteolysis	Disorder	2 Case(s)
502444	Alkaline ceramidase 3 deficiency	Disorder	2 Case(s)
498497	Short rib-polydactyly syndrome type 5	Disorder	2 Case(s)
487814	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation	Disorder	2 Case(s)
487796	Takenouchi-Kosaki syndrome	Disorder	2 Case(s)
480528	Lethal hydranencephaly-diaphragmatic hernia syndrome	Disorder	2 Case(s)
478042	Combined oxidative phosphorylation defect type 30	Disorder	2 Case(s)
477831	Kosaki overgrowth syndrome	Disorder	2 Case(s)
477787	Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder	Disorder	2 Case(s)
477684	Combined oxidative phosphorylation defect type 26	Disorder	2 Case(s)
476406	Congenital generalized hypercontractile muscle stiffness syndrome	Disorder	2 Case(s)
465824	Fetal encasement syndrome	Disorder	2 Case(s)
459070	X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome	Disorder	2 Case(s)
457359	Megalencephaly-severe kyphoscoliosis-overgrowth syndrome	Disorder	2 Case(s)
457265	Progressive myoclonic epilepsy type 9	Disorder	2 Case(s)
457223	Syndromic sensorineural deafness due to combined oxidative phosphorylation defect	Disorder	2 Case(s)
457205	Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome	Disorder	2 Case(s)
456312	Infantile multisystem neurologic-endocrine-pancreatic disease	Disorder	2 Case(s)
456298	1p35.2 microdeletion syndrome	Disorder	2 Case(s)
453521	Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	Disorder	2 Case(s)
448267	Regressive spondylometaphyseal dysplasia	Disorder	2 Case(s)
448264	Isolated focal non-epidermolytic palmoplantar keratoderma	Disorder	2 Case(s)
447974	Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome	Disorder	2 Case(s)
447961	Pigmentation defects-palmoplantar keratoderma-skin carcinoma syndrome	Disorder	2 Case(s)
447954	Combined oxidative phosphorylation defect type 25	Disorder	2 Case(s)
447731	NIK deficiency	Disorder	2 Case(s)
445110	Limb-girdle muscular dystrophy due to POMK deficiency	Disorder	2 Case(s)
443950	DNAJB2-related Charcot-Marie-Tooth disease type 2	Disorder	2 Case(s)
443236	Postural orthostatic tachycardia syndrome due to NET deficiency	Disorder	2 Case(s)
440731	L-ferritin deficiency	Disorder	2 Case(s)
440713	Isolated sedoheptulokinase deficiency	Disorder	2 Case(s)
439897	Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome	Disorder	2 Case(s)
439232	AApoAIV amyloidosis	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
438207	Severe autosomal recessive macrothrombocytopenia	Disorder	2 Case(s)
436182	Microcephalic primordial dwarfism-insulin resistance syndrome	Disorder	2 Case(s)
435819	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation	Disorder	2 Case(s)
431361	Progressive encephalopathy with leukodystrophy due to DECR deficiency	Disorder	2 Case(s)
431329	Autosomal recessive spastic paraplegia type 57	Disorder	2 Case(s)
424107	Congenital myopathy with myasthenic-like onset	Disorder	2 Case(s)
423479	X-linked intellectual disability-limb spasticity-retinal dystrophy-arginine vasopressin deficiency	Disorder	2 Case(s)
423306	Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome	Disorder	2 Case(s)
420741	RIDDLE syndrome	Disorder	2 Case(s)
420733	Combined oxidative phosphorylation defect type 21	Disorder	2 Case(s)
420728	Combined oxidative phosphorylation defect type 20	Disorder	2 Case(s)
420699	Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency	Disorder	2 Case(s)
412181	Epidermolysis bullosa simplex due to BP230 deficiency	Disorder	2 Case(s)
411712	Maternal riboflavin deficiency	Disorder	2 Case(s)
404499	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency	Disorder	2 Case(s)
404476	Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome	Disorder	2 Case(s)
401959	Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome	Disorder	2 Case(s)
401923	9q31.1q31.3 microdeletion syndrome	Disorder	2 Case(s)
401830	Autosomal recessive spastic paraplegia type 69	Disorder	2 Case(s)
401820	Autosomal recessive spastic paraplegia type 67	Disorder	2 Case(s)
401815	Autosomal recessive spastic paraplegia type 66	Disorder	2 Case(s)
401805	Autosomal recessive spastic paraplegia type 63	Disorder	2 Case(s)
398109	Neonatal autoimmune hemolytic anemia	Disorder	2 Case(s)
397973	Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome	Disorder	2 Case(s)
397959	TCR-alpha-beta-positive T-cell deficiency	Disorder	2 Case(s)
397735	Autosomal dominant Charcot-Marie-Tooth disease type 2U	Disorder	2 Case(s)
397725	COASY protein-associated neurodegeneration	Disorder	2 Case(s)
391366	Growth retardation-mild developmental delay-chronic hepatitis syndrome	Disorder	2 Case(s)
391348	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	Disorder	2 Case(s)
391343	Fatal post-viral neurodegenerative disorder	Disorder	2 Case(s)
370997	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	Disorder	2 Case(s)
370930	XYLT1-CDG	Disorder	2 Case(s)
370921	STT3A-CDG	Disorder	2 Case(s)
370039	Angora hair nevus	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
370015	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type	Disorder	2 Case(s)
369979	Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome	Disorder	2 Case(s)
369955	Methylmalonic acidemia with homocystinuria, type cblJ	Subtype of disorder	2 Case(s)
369929	Primary hyperaldosteronism-seizures-neurological abnormalities syndrome	Disorder	2 Case(s)
369881	2p21 microdeletion syndrome without cystinuria	Disorder	2 Case(s)
363680	2p13.2 microdeletion syndrome	Disorder	2 Case(s)
363623	GMPPB-related limb-girdle muscular dystrophy R19	Disorder	2 Case(s)
363424	Multiple mitochondrial dysfunctions syndrome type 3	Disorder	2 Case(s)
357158	Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome	Disorder	2 Case(s)
352665	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome due to 9q21.3 microdeletion	Subtype of disorder	2 Case(s)
352563	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	Disorder	2 Case(s)
352530	Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome	Disorder	2 Case(s)
352333	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome	Disorder	2 Case(s)
3448	Weaver-Williams syndrome	Disorder	2 Case(s)
3429	Verloove Vanhorick-Brubakk syndrome	Disorder	2 Case(s)
3424	Velo-facial-skeletal syndrome	Disorder	2 Case(s)
3368	Trigonocephaly-bifid nose-acral anomalies syndrome	Disorder	2 Case(s)
3365	Trigonocephaly-broad thumbs syndrome	Disorder	2 Case(s)
3327	Thyrocerebrorenal syndrome	Disorder	2 Case(s)
3294	Extensor tendons of finger anomalies	Disorder	2 Case(s)
3293	Telecanthus-hypertelorism-strabismus-pes cavus syndrome	Disorder	2 Case(s)
329242	Congenital chronic diarrhea with protein-losing enteropathy	Disorder	2 Case(s)
329224	Schuurs-Hoeijmakers syndrome	Disorder	2 Case(s)
3262	Dobrow syndrome	Disorder	2 Case(s)
324575	Hyperinsulinism due to HNF1A deficiency	Disorder	2 Case(s)
324540	Aphonia-deafness-retinal dystrophy-bifid halluces-intellectual disability syndrome	Disorder	2 Case(s)
324530	Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation	Disorder	2 Case(s)
324416	Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome	Disorder	2 Case(s)
324410	X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome	Disorder	2 Case(s)
324364	Mixed sclerosing bone dystrophy with extra-skeletal manifestations	Disorder	2 Case(s)
324307	Severe lateral tibial bowing-short stature-mild winged scapula-mild facial dysmorphism syndrome	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
324299	Multiple paragangliomas associated with polycythemia	Disorder	2 Case(s)
324294	T-cell immunodeficiency with epidermodysplasia verruciformis	Disorder	2 Case(s)
3241	Deafness-craniofacial syndrome	Disorder	2 Case(s)
3240	Early-onset progressive leukoencephalopathy-central nervous system calcification-deafness-visual impairment syndrome	Disorder	2 Case(s)
3239	Deafness-vitiligo-achalasia syndrome	Disorder	2 Case(s)
3224	Deafness-genital anomalies-metacarpal and metatarsal synostosis syndrome	Disorder	2 Case(s)
3218	Deafness-epiphyseal dysplasia-short stature syndrome	Disorder	2 Case(s)
3214	Deaf blind hypopigmentation syndrome, Yemenite type	Disorder	2 Case(s)
320370	Autosomal recessive spastic paraplegia type 43	Disorder	2 Case(s)
3200	Arthrogryposis-ectodermal dysplasia syndrome	Disorder	2 Case(s)
3199	Stimmler syndrome	Disorder	2 Case(s)
319675	Microcephalic primordial dwarfism, Dauber type	Disorder	2 Case(s)
319600	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency	Disorder	2 Case(s)
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	Disorder	2 Case(s)
319514	Combined oxidative phosphorylation defect type 13	Disorder	2 Case(s)
3177	Spinocerebellar degeneration-corneal dystrophy syndrome	Disorder	2 Case(s)
317425	Severe combined immunodeficiency due to DNA-PKcs deficiency	Disorder	2 Case(s)
3173	Infantile spasms-broad thumbs syndrome	Disorder	2 Case(s)
3167	Siegler-Brewer-Carey syndrome	Disorder	2 Case(s)
3151	Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome	Disorder	2 Case(s)
314993	Cataract-congenital heart disease-neural tube defect syndrome	Disorder	2 Case(s)
314575	Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome	Disorder	2 Case(s)
314041	Marfanoid habitus-inguinal hernia-advanced bone age syndrome	Disorder	2 Case(s)
314029	High bone mass osteogenesis imperfecta	Disorder	2 Case(s)
314002	Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome	Disorder	2 Case(s)
313947	2q23.1 microduplication syndrome	Disorder	2 Case(s)
313772	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	Disorder	2 Case(s)
3134	SCARF syndrome	Disorder	2 Case(s)
309108	Pancreatic colipase deficiency	Disorder	2 Case(s)
3082	Intellectual disability-polydactyly-uncombable hair syndrome	Disorder	2 Case(s)
3080	Intellectual disability, Wolff type	Disorder	2 Case(s)
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
	keratoderma syndrome		
3068	Intellectual disability-myopathy-short stature-endocrine defect syndrome	Disorder	2 Case(s)
306511	Autosomal recessive spastic paraplegia type 48	Disorder	2 Case(s)
3016	Absent radius-anogenital anomalies syndrome	Disorder	2 Case(s)
3011	Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome	Disorder	2 Case(s)
300284	Connective tissue disorder due to lysyl hydroxylase-3 deficiency	Disorder	2 Case(s)
2997	Ptosis-vocal cord paralysis syndrome	Disorder	2 Case(s)
2988	Pterygium colli-intellectual disability-digital anomalies syndrome	Disorder	2 Case(s)
2985	Pseudoprogeria syndrome	Disorder	2 Case(s)
2976	Pseudoleprechaunism syndrome, Patterson type	Disorder	2 Case(s)
2975	46,XX difference of sex development-skeletal anomalies syndrome	Disorder	2 Case(s)
2956	Acrodysplasia scoliosis	Disorder	2 Case(s)
2941	Porencephaly-cerebellar hypoplasia-internal malformations syndrome	Disorder	2 Case(s)
294026	Syndactyly-nystagmus syndrome due to 2q31.1 microduplication	Disorder	2 Case(s)
293807	Ketamine-induced biliary dilatation	Disorder	2 Case(s)
2921	Preaxial polydactyly-colobomata-intellectual disability syndrome	Disorder	2 Case(s)
2898	X-linked intellectual disability-plagiocephaly syndrome	Disorder	2 Case(s)
289522	Microtriplication 11q24.1	Disorder	2 Case(s)
2892	Pilodental dysplasia-refractive errors syndrome	Disorder	2 Case(s)
2891	Pili torti-developmental delay-neurological abnormalities syndrome	Disorder	2 Case(s)
2888	Pierre Robin syndrome-faciodigital anomaly syndrome	Disorder	2 Case(s)
2876	PHAVER syndrome	Disorder	2 Case(s)
2871	Pfeiffer-Palm-Teller syndrome	Disorder	2 Case(s)
2867	Short stature, Brussels type	Disorder	2 Case(s)
2866	Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome	Disorder	2 Case(s)
2826	Spastic paraplegia-precocious puberty syndrome	Disorder	2 Case(s)
2825	PARC syndrome	Disorder	2 Case(s)
281127	Acral self-healing collodion baby	Disorder	2 Case(s)
280576	Nestor-Guillermo progeria syndrome	Disorder	2 Case(s)
280397	Familial Alzheimer-like prion disease	Disorder	2 Case(s)
2773	Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome	Disorder	2 Case(s)
276556	Hyperinsulinism due to UCP2 deficiency	Disorder	2 Case(s)
276405	Hyperbiliverdinemia	Disorder	2 Case(s)
2759	Imperforate oropharynx-costovertebral anomalies syndrome	Disorder	2 Case(s)
2722	Odonto-onycho dysplasia-alopecia syndrome	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
2720	Oculocerebral hypopigmentation syndrome, Preus type	Disorder	2 Case(s)
2718	Oculotrichodysplasia	Disorder	2 Case(s)
2666	Adult familial nephronophthisis-spastic quadriparesia syndrome	Disorder	2 Case(s)
2643	Microcephalic primordial dwarfism, Toriello type	Disorder	2 Case(s)
263501	COG4-CDG	Disorder	2 Case(s)
2631	Mesomelic dwarfism-cleft palate-camptodactyly syndrome	Disorder	2 Case(s)
261534	49,XXXYY syndrome	Disorder	2 Case(s)
261304	Paternal 20q13.2q13.3 microdeletion syndrome	Disorder	2 Case(s)
2597	Mitochondrial myopathy-lactic acidosis-deafness syndrome	Disorder	2 Case(s)
2575	Cystic fibrosis-gastritis-megaloblastic anemia syndrome	Disorder	2 Case(s)
255132	Adult-onset autosomal recessive sideroblastic anemia	Disorder	2 Case(s)
254925	Combined oxidative phosphorylation defect type 4	Disorder	2 Case(s)
254898	Deafness-encephaloneuropathy-obesity-valvulopathy syndrome	Disorder	2 Case(s)
2547	Microphthalmia-microtia-fetal akinesia syndrome	Disorder	2 Case(s)
2533	Microcephaly-deafness-intellectual disability syndrome	Disorder	2 Case(s)
2528	Microcephaly-microcornea syndrome, Seemanova type	Disorder	2 Case(s)
2522	Microcephaly-cervical spine fusion anomalies syndrome	Disorder	2 Case(s)
2519	Microcephaly-seizures-intellectual disability-heart disease syndrome	Disorder	2 Case(s)
2513	Microcephaly-albinism-digital anomalies syndrome	Disorder	2 Case(s)
2511	Microbrachycephaly-ptosis-cleft lip syndrome	Disorder	2 Case(s)
2489	Upper limb defect-eye and ear abnormalities syndrome	Disorder	2 Case(s)
2487	Lower limb malformation-hypospadias syndrome	Disorder	2 Case(s)
2482	Melhem-Fahl syndrome	Disorder	2 Case(s)
2475	White forelock with malformations	Disorder	2 Case(s)
2427	Macrocephaly-short stature-paraplegia syndrome	Disorder	2 Case(s)
2400	Peripheral motor neuropathy-dysautonomia syndrome	Disorder	2 Case(s)
2390	Lichtenstein syndrome	Disorder	2 Case(s)
238523	Atypical hypotonia-cystinuria syndrome	Disorder	2 Case(s)
238329	Severe X-linked mitochondrial encephalomyopathy	Disorder	2 Case(s)
2347	Lethal Kniest-like dysplasia	Disorder	2 Case(s)
2324	Osteopenia-intellectual disability-sparse hair syndrome	Disorder	2 Case(s)
2321	Jung syndrome	Disorder	2 Case(s)
231556	Late-onset localized junctional epidermolysis bullosa-intellectual disability syndrome	Disorder	2 Case(s)
2310	Absence deformity of leg-cataract syndrome	Disorder	2 Case(s)
2282	Dysmorphism-short stature-deafness-difference of sex development syndrome	Disorder	2 Case(s)
2274	Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome	Disorder	2 Case(s)
2272	Ichthyosis-oral and digital anomalies syndrome	Disorder	2 Case(s)
2271	Congenital ichthyosis-microcephalus-tetraplegia syndrome	Disorder	2 Case(s)
2266	Hypotrichosis-intellectual disability, Lopes type	Disorder	2 Case(s)
2256	Fibulo-ulnar hypoplasia-renal anomalies syndrome	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
2250	Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome	Disorder	2 Case(s)
2249	Ulna hypoplasia-intellectual disability syndrome	Disorder	2 Case(s)
2235	Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome	Disorder	2 Case(s)
2234	Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome	Disorder	2 Case(s)
2233	Hypogonadism-mitral valve prolapse-intellectual disability syndrome	Disorder	2 Case(s)
221142	Confetti-like macular atrophy	Disorder	2 Case(s)
221139	Combined immunodeficiency with facio-oculo-skeletal anomalies	Disorder	2 Case(s)
220448	Macrothrombocytopenia with mitral valve insufficiency	Disorder	2 Case(s)
2204	Dysplastic cortical hyperostosis, Kozlowski-Tsuruta type	Subtype of disorder	2 Case(s)
2184	Hydrocephaly-low insertion umbilicus syndrome	Disorder	2 Case(s)
2183	Hydrocephalus-obesity-hypogonadism syndrome	Disorder	2 Case(s)
2181	Hydrocephaly-tall stature-joint laxity syndrome	Disorder	2 Case(s)
217399	Congenital insensitivity to pain-hyperhidrosis-absence of cutaneous sensory innervation	Disorder	2 Case(s)
2172	Microcephaly-glomerulonephritis-marfanoid habitus syndrome	Disorder	2 Case(s)
2155	Hirschsprung disease-deafness-polydactyly syndrome	Disorder	2 Case(s)
2145	Craniosynostosis, Herrmann-Opitz type	Disorder	2 Case(s)
2119	HEC syndrome	Disorder	2 Case(s)
2110	Hallux varus-preaxial polysyndactyly syndrome	Disorder	2 Case(s)
2109	Hallermann-Streiff-like syndrome	Disorder	2 Case(s)
2104	Dysmorphism-pectus carinatum-joint laxity syndrome	Disorder	2 Case(s)
2083	Prominent glabella-microcephaly-hypogenitalism syndrome	Disorder	2 Case(s)
2074	Gemignani syndrome	Disorder	2 Case(s)
206564	POMGNT1-related limb-girdle muscular dystrophy R15	Disorder	2 Case(s)
2058	Fryns-Smeets-Thiry syndrome	Disorder	2 Case(s)
2025	Gingival fibromatosis-facial dysmorphism syndrome	Disorder	2 Case(s)
2015	Cleft palate-short stature-vertebral anomalies syndrome	Disorder	2 Case(s)
2010	Cleft palate-stapes fixation-oligodontia syndrome	Disorder	2 Case(s)
2007	Alar cartilages hypoplasia-coloboma-telecanthus syndrome	Disorder	2 Case(s)
2003	Cleft lip/palate-deafness-sacral lipoma syndrome	Disorder	2 Case(s)
1995	Cleft lip-retinopathy syndrome	Disorder	2 Case(s)
199348	Thiamine-responsive encephalopathy	Disorder	2 Case(s)
199329	Congenital myopathy, Paradas type	Disorder	2 Case(s)
1968	Flat face-microstomia-ear anomaly syndrome	Disorder	2 Case(s)
1964	Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome	Disorder	2 Case(s)
1948	Epilepsy-microcephaly-skeletal dysplasia syndrome	Disorder	2 Case(s)
1937	Eng-Strom syndrome	Disorder	2 Case(s)
1933	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	Disorder	2 Case(s)

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1927	Emery-Nelson syndrome	Disorder	2 Case(s)
1883	Ectodermal dysplasia-sensorineural deafness syndrome	Disorder	2 Case(s)
1861	Thoracic dysplasia-hydrocephalus syndrome	Disorder	2 Case(s)
183707	Infantile LAD-like disease due to RAC2 deficiency	Disorder	2 Case(s)
1825	Epiphyseal dysplasia-hearing loss-dysmorphism syndrome	Disorder	2 Case(s)
1806	Ectodermal dysplasia-blindness syndrome	Disorder	2 Case(s)
1803	Thoracomelic dysplasia	Disorder	2 Case(s)
1780	Thakker-Donnai syndrome	Disorder	2 Case(s)
1778	Facial dysmorphism-shawl scrotum-joint laxity syndrome	Disorder	2 Case(s)
1770	XY type gonadal dysgenesis-associated anomalies syndrome	Disorder	2 Case(s)
171839	Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome	Disorder	2 Case(s)
169100	Immunodeficiency due to CD25 deficiency	Disorder	2 Case(s)
168598	Methionine adenosyltransferase I/III deficiency	Disorder	2 Case(s)
168552	Spondylometaphyseal dysplasia-bowed forearms-facial dysmorphism syndrome	Disorder	2 Case(s)
168451	Spondyloepimetaphyseal dysplasia-abnormal dentition syndrome	Disorder	2 Case(s)
1670	Chronic diarrhea with villous atrophy	Disorder	2 Case(s)
166038	Metaphyseal chondrodysplasia, Kaitila type	Disorder	2 Case(s)
166032	Multiple epiphyseal dysplasia-miniepiphyses syndrome	Disorder	2 Case(s)
166016	Multiple epiphyseal dysplasia, Lowry type	Disorder	2 Case(s)
1659	Dermatoleukodystrophy	Disorder	2 Case(s)
163684	Leukoencephalopathy-dystonia-motor neuropathy syndrome	Disorder	2 Case(s)
1570	Symbrachydactyly of hands and feet	Disorder	2 Case(s)
1563	Dahlberg-Borer-Newcomer syndrome	Disorder	2 Case(s)
1547	Cryptomicrotia-brachydactyly-excess fingertip arch syndrome	Disorder	2 Case(s)
1521	Craniofrontonasal dysplasia-Poland anomaly syndrome	Disorder	2 Case(s)
1506	Thin ribs-tubular bones-dysmorphism syndrome	Disorder	2 Case(s)
1488	Cooper-Jabs syndrome	Disorder	2 Case(s)
1485	Arthrogryposis-hyperkeratosis syndrome, lethal form	Disorder	2 Case(s)
1484	Contractures-ectodermal dysplasia-cleft lip/palate syndrome	Disorder	2 Case(s)
1453	Cleidorhizomelic syndrome	Disorder	2 Case(s)
1433	Choroidal atrophy-alopecia syndrome	Disorder	2 Case(s)
1422	Chondrodysplasia-difference of sex development syndrome	Disorder	2 Case(s)
141258	Tessier number 4 facial cleft	Disorder	2 Case(s)
1397	Hydrocephaly-cerebellar agenesis syndrome	Disorder	2 Case(s)
1390	Night blindness-skeletal anomalies-dysmorphism syndrome	Disorder	2 Case(s)
1380	Cataract-nephropathy-encephalopathy syndrome	Disorder	2 Case(s)
137681	Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	Disorder	2 Case(s)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of cases
137631	Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome	Disorder	2 Case(s)
1368	Cataract-ataxia-deafness syndrome	Disorder	2 Case(s)
1354	Heart defects-limb shortening syndrome	Disorder	2 Case(s)
1352	Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	Disorder	2 Case(s)
1326	Camptodactyly syndrome, Guadalajara type 2	Disorder	2 Case(s)
1295	Brachytelephalangy-dysmorphism-Kallmann syndrome	Disorder	2 Case(s)
1277	Brachydactyly-mesomelia-intellectual disability-heart defects syndrome	Disorder	2 Case(s)
1237	Beemer-Ertbruggen syndrome	Disorder	2 Case(s)
1236	Severe microbrachycephaly-intellectual disability-athetoid cerebral palsy syndrome	Disorder	2 Case(s)
1227	Bangstad syndrome	Disorder	2 Case(s)
1217	Spinal atrophy-ophthalmoplegia-pyramidal syndrome	Disorder	2 Case(s)
1192	Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome	Disorder	2 Case(s)
1184	Ataxia-photosensitivity-short stature syndrome	Disorder	2 Case(s)
1126	Aprosencephaly cerebellar dysgenesis	Disorder	2 Case(s)
1123	Caudal appendage-deafness syndrome	Disorder	2 Case(s)
1068	Aniridia-intellectual disability syndrome	Disorder	2 Case(s)
1064	Aniridia-renal agenesis-psychomotor retardation syndrome	Disorder	2 Case(s)
1046	Lethal hemolytic anemia-genital anomalies syndrome	Disorder	2 Case(s)
1028	Amelo-onycho-hypohidrotic syndrome	Disorder	2 Case(s)
1021	Amaurosis-hypertrichosis syndrome	Disorder	2 Case(s)
1014	Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome	Disorder	2 Case(s)
1003	Scalp defects-postaxial polydactyly syndrome	Disorder	2 Case(s)
100013	Lissencephaly with cerebellar hypoplasia type C	Disorder	2 Case(s)
99849	Glycogen storage disease due to muscle beta-enolase deficiency	Disorder	1 Case(s)
79332	B4GALT1-CDG	Disorder	1 Case(s)
79326	ALG2-CDG	Disorder	1 Case(s)
600194	Factor V Atlanta bleeding disorder	Subtype of disorder	1 Case(s)
529980	Inflammatory bowel disease-recurrent sinopulmonary infections syndrome	Disorder	1 Case(s)
528091	Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome	Disorder	1 Case(s)
478029	Combined oxidative phosphorylation defect type 29	Disorder	1 Case(s)
458798	Spinocerebellar ataxia type 41	Disorder	1 Case(s)
448010	CAD-CDG	Disorder	1 Case(s)
440706	Ribose-5-P isomerase deficiency	Disorder	1 Case(s)
435934	COG2-CDG	Disorder	1 Case(s)
435651	CIDEC-related familial partial lipodystrophy	Disorder	1 Case(s)
431166	Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection	Disorder	1 Case(s)
431149	Combined immunodeficiency due to OX40 deficiency	Disorder	1 Case(s)

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404521	Spinal muscular atrophy with respiratory distress type 2	Disorder	1 Case(s)
401840	Autosomal recessive spastic paraplegia type 71	Disorder	1 Case(s)
401800	Autosomal recessive spastic paraplegia type 60	Disorder	1 Case(s)
397968	Charcot-Marie-Tooth disease type 2R	Disorder	1 Case(s)
370924	STT3B-CDG	Disorder	1 Case(s)
370097	Oculocutaneous albinism type 6	Disorder	1 Case(s)
331190	Immunodeficiency due to ficolin3 deficiency	Disorder	1 Case(s)
331187	Immunodeficiency due to MASP-2 deficiency	Disorder	1 Case(s)
330029	Hypotrichosis-deafness syndrome	Disorder	1 Case(s)
329942	Transient neonatal multiple acyl-CoA dehydrogenase deficiency	Disorder	1 Case(s)
324422	ALG13-CDG	Disorder	1 Case(s)
319678	Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	Disorder	1 Case(s)
300536	DDOST-CDG	Disorder	1 Case(s)
2963	Progeroid syndrome, Petty type	Disorder	1 Case(s)
280333	Alpha-dystroglycan-related limb-girdle muscular dystrophy R16	Disorder	1 Case(s)
2786	Osteoporosis-oculocutaneous hypopigmentation syndrome	Disorder	1 Case(s)
263494	DPM3-CDG	Disorder	1 Case(s)
263297	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	Disorder	1 Case(s)
254920	Combined oxidative phosphorylation defect type 2	Disorder	1 Case(s)
254334	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B	Disorder	1 Case(s)
243343	Dimethylglycine dehydrogenase deficiency	Disorder	1 Case(s)
240760	Nijmegen breakage syndrome-like disorder	Disorder	1 Case(s)
206559	POMT2-related limb-girdle muscular dystrophy R14	Disorder	1 Case(s)
1692	Mosaic trisomy 1	Disorder	1 Case(s)
1035	Beta-mercaptolactate cysteine disulfiduria	Disorder	1 Case(s)

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Number of published families

ORPHAcode	Disease or Subtype of disease	Classification Level	Number of families
1652	Dent disease	Disorder	250 Family(ies)
99879	Familial isolated hyperparathyroidism	Disorder	100 Family(ies)
98784	Autosomal dominant nocturnal frontal lobe epilepsy	Disorder	100 Family(ies)
98759	Spinocerebellar ataxia type 17	Disorder	100 Family(ies)
89843	Dystrophic epidermolysis bullosa pruriginosa	Disorder	100 Family(ies)
276198	Spinocerebellar ataxia type 36	Disorder	100 Family(ies)
1949	Self-limited neonatal epilepsy	Disorder	100 Family(ies)
1416	Familial calcium pyrophosphate deposition	Disorder	100 Family(ies)
2524	Pontocerebellar hypoplasia type 2	Disorder	81 Family(ies)
51083	Familial short QT syndrome	Disorder	80 Family(ies)
526	Liddle syndrome	Disorder	72 Family(ies)
60030	Loeys-Dietz syndrome	Disorder	52 Family(ies)
98934	Huntington disease-like 2	Disorder	50 Family(ies)
643	Giant axonal neuropathy	Disorder	50 Family(ies)
2526	Microcephaly-lymphedema-chorioretinopathy syndrome	Disorder	50 Family(ies)
536467	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	Subtype of disorder	41 Family(ies)
98762	Spinocerebellar ataxia type 12	Disorder	40 Family(ies)
79410	Localized dystrophic epidermolysis bullosa, pretibial form	Subtype of disorder	40 Family(ies)
263548	Peeling skin syndrome type A	Subtype of disorder	40 Family(ies)
2254	Pontocerebellar hypoplasia type 1	Disorder	40 Family(ies)
217330	REN-related autosomal dominant tubulointerstitial kidney disease	Subtype of disorder	35 Family(ies)
200418	Immunodeficiency with factor I anomaly	Disorder	35 Family(ies)
163937	X-linked intellectual disability, Najm type	Disorder	35 Family(ies)
1106	Microphthalmia with limb anomalies	Disorder	35 Family(ies)
324442	Autosomal recessive axonal neuropathy with neuromyotonia	Disorder	33 Family(ies)
98434	Hereditary combined deficiency of vitamin K-dependent clotting factors	Disorder	30 Family(ies)
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	Disorder	30 Family(ies)
452	X-linked lissencephaly with abnormal genitalia	Disorder	30 Family(ies)
425	Apolipoprotein A-I deficiency	Disorder	30 Family(ies)
3237	Multiple synostoses syndrome	Disorder	30 Family(ies)
3222	Phosphoribosylpyrophosphate synthetase superactivity	Disorder	30 Family(ies)
293822	MITF-related melanoma and renal cell carcinoma predisposition syndrome	Disorder	30 Family(ies)
2848	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	Disorder	30 Family(ies)
263553	Peeling skin syndrome type B	Subtype of disorder	30 Family(ies)
228305	Carnitine palmitoyl transferase II deficiency, severe infantile form	Subtype of disorder	30 Family(ies)
217012	Spinocerebellar ataxia type 31	Disorder	30 Family(ies)

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424	Familial hyperthyroidism due to mutations in TSH receptor	Disorder	28 Family(ies)
33108	Lethal multiple pterygium syndrome	Disorder	28 Family(ies)
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	Disorder	26 Family(ies)
3464	Woodhouse-Sakati syndrome	Disorder	25 Family(ies)
85293	X-linked intellectual disability, Cabezas type	Disorder	24 Family(ies)
536471	Spondylodysplastic Ehlers-Danlos syndrome	Disorder	24 Family(ies)
1065	Aniridia-cerebellar ataxia-intellectual disability syndrome	Disorder	22 Family(ies)
99027	Adult-onset autosomal dominant leukodystrophy	Disorder	20 Family(ies)
98763	Spinocerebellar ataxia type 14	Disorder	20 Family(ies)
97286	Carney-Stratakis syndrome	Disorder	20 Family(ies)
959	Acro-renal-ocular syndrome	Disorder	20 Family(ies)
369913	Combined oxidative phosphorylation defect type 17	Disorder	20 Family(ies)
3203	Overhydrated hereditary stomatocytosis	Disorder	20 Family(ies)
3202	Dehydrated hereditary stomatocytosis	Disorder	20 Family(ies)
228308	Carnitine palmitoyl transferase II deficiency, neonatal form	Subtype of disorder	20 Family(ies)
2229	Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome	Disorder	20 Family(ies)
1200	Burn-McKeown syndrome	Disorder	20 Family(ies)
100998	Autosomal dominant spastic paraplegia type 17	Disorder	20 Family(ies)
99791	Dentin dysplasia type II	Subtype of disorder	19 Family(ies)
168615	Hereditary persistence of alpha-fetoprotein	Disorder	19 Family(ies)
93311	Multiple epiphyseal dysplasia type 5	Disorder	18 Family(ies)
25980	X-linked myopathy with excessive autophagy	Disorder	18 Family(ies)
90031	Non-spherocytic hemolytic anemia due to hexokinase deficiency	Disorder	17 Family(ies)
615	Familial atrial myxoma	Disorder	17 Family(ies)
293168	Infantile-onset ascending hereditary spastic paralysis	Disorder	17 Family(ies)
88621	Ichthyosis-prematurity syndrome	Disorder	16 Family(ies)
84090	Fibronectin glomerulopathy	Disorder	16 Family(ies)
3220	Deafness-enamel hypoplasia-nail defects syndrome	Disorder	15 Family(ies)
2850	Alopecia-intellectual disability syndrome	Disorder	15 Family(ies)
88619	Familial acute necrotizing encephalopathy	Disorder	14 Family(ies)
620371	Gitelman-like kidney tubulopathy due to mitochondrial DNA mutation	Disorder	14 Family(ies)
495930	Familial monosomy 7 syndrome	Disorder	14 Family(ies)
137831	X-linked intellectual disability-cerebellar hypoplasia syndrome	Disorder	14 Family(ies)
1145	Infantile-onset X-linked spinal muscular atrophy	Disorder	14 Family(ies)
137678	Spondyloepiphyseal dysplasia with metatarsal shortening	Disorder	13 Family(ies)
401996	Karyomegalic interstitial nephritis	Disorder	12 Family(ies)
228277	Familial anetoderma	Disorder	12 Family(ies)
99955	Charcot-Marie-Tooth disease type 4B1	Disorder	11 Family(ies)
98971	Posterior amorphous corneal dystrophy	Disorder	11 Family(ies)
478664	Hereditary sensory and autonomic neuropathy type 8	Disorder	11 Family(ies)

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439218	KCNQ2-related epileptic encephalopathy	Disorder	11 Family(ies)
90001	X-linked cone dysfunction syndrome with myopia	Disorder	10 Family(ies)
85279	KDM5C-related syndromic X-linked intellectual disability	Disorder	10 Family(ies)
535458	Familial GPIHBP1 deficiency	Subtype of disorder	10 Family(ies)
481662	Familial Chilblain lupus	Disorder	10 Family(ies)
412057	Autosomal recessive cerebellar ataxia due to STUB1 deficiency	Disorder	10 Family(ies)
3412	VACTERL with hydrocephalus	Disorder	10 Family(ies)
309020	Familial apolipoprotein C-II deficiency	Subtype of disorder	10 Family(ies)
2791	Otodental syndrome	Disorder	10 Family(ies)
2238	Familial isolated hypoparathyroidism	Disorder	10 Family(ies)
2202	Palmoplantar keratoderma-deafness syndrome	Disorder	10 Family(ies)
2198	Palmoplantar keratoderma-esophageal carcinoma syndrome	Disorder	10 Family(ies)
1856	Spondyloperipheral dysplasia-short ulna syndrome	Disorder	10 Family(ies)
178464	Hereditary myopathy with early respiratory failure	Disorder	10 Family(ies)
166063	Pontocerebellar hypoplasia type 4	Disorder	10 Family(ies)
1658	Absence of fingerprints-congenital milia syndrome	Disorder	10 Family(ies)
158676	Localized dystrophic epidermolysis bullosa, nails only	Subtype of disorder	10 Family(ies)
158673	Localized dystrophic epidermolysis bullosa, acral form	Subtype of disorder	10 Family(ies)
1412	Tarsal-carpal coalition syndrome	Disorder	10 Family(ies)
140927	Self-limited neonatal-infantile epilepsy	Disorder	10 Family(ies)
1276	Brachydactyly-arterial hypertension syndrome	Disorder	10 Family(ies)
101006	Autosomal recessive spastic paraplegia type 26	Disorder	10 Family(ies)
100996	Autosomal recessive spastic paraplegia type 15	Disorder	10 Family(ies)
100991	Autosomal dominant spastic paraplegia type 10	Disorder	10 Family(ies)
100989	Autosomal dominant spastic paraplegia type 8	Disorder	10 Family(ies)
100988	Autosomal dominant spastic paraplegia type 6	Disorder	10 Family(ies)
263516	Progressive myoclonic epilepsy type 3	Disorder	9 Family(ies)
217266	BNAR syndrome	Disorder	9 Family(ies)
1062	Hereditary neurocutaneous malformation	Disorder	9 Family(ies)
100008	ACys amyloidosis	Subtype of disorder	9 Family(ies)
391330	X-linked osteoporosis with fractures	Disorder	8 Family(ies)
3248	Isolated distal symphalangism	Disorder	8 Family(ies)
306527	Isolated hereditary congenital facial paralysis	Disorder	8 Family(ies)
217055	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	Disorder	8 Family(ies)
1377	Cataract-microcornea syndrome	Disorder	8 Family(ies)
1149	Kuskokwim syndrome	Disorder	8 Family(ies)
93561	ALys amyloidosis	Subtype of disorder	7 Family(ies)
86817	Hemolytic anemia due to adenylate kinase deficiency	Disorder	7 Family(ies)
488594	Autosomal recessive spastic paraplegia type 76	Disorder	7 Family(ies)
397618	Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome	Disorder	7 Family(ies)
324737	SRD5A3-CDG	Disorder	7 Family(ies)
324713	ABeta amyloidosis, Italian type	Subtype of disorder	7 Family(ies)

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320396	Autosomal recessive spastic paraplegia type 45	Disorder	7 Family(ies)
300345	Autosomal systemic lupus erythematosus	Disorder	7 Family(ies)
281139	Annular epidermolytic ichthyosis	Disorder	7 Family(ies)
251274	Familial hyperaldosteronism type III	Disorder	7 Family(ies)
1897	EEM syndrome	Disorder	7 Family(ies)
178461	X-linked myopathy with postural muscle atrophy	Disorder	7 Family(ies)
85453	X-linked reticulate pigmentary disorder	Disorder	6 Family(ies)
85110	Familial encephalopathy with neuroserpin inclusion bodies	Disorder	6 Family(ies)
79447	X-linked lethal multiple pterygium syndrome	Disorder	6 Family(ies)
79401	PLEC-related intermediate epidermolysis bullosa simplex without extracutaneous involvement	Disorder	6 Family(ies)
73229	HANAC syndrome	Disorder	6 Family(ies)
456318	Hereditary sensory neuropathy-deafness-dementia syndrome	Disorder	6 Family(ies)
391411	Atypical juvenile parkinsonism	Disorder	6 Family(ies)
34516	DNAJB6-related limb-girdle muscular dystrophy D1	Disorder	6 Family(ies)
324561	Hypopigmentation-punctate palmoplantar keratoderma syndrome	Disorder	6 Family(ies)
320380	Autosomal recessive spastic paraplegia type 54	Disorder	6 Family(ies)
2886	TARP syndrome	Disorder	6 Family(ies)
2790	Endosteal hyperostosis, Worth type	Disorder	6 Family(ies)
2045	FLOTCH syndrome	Disorder	6 Family(ies)
1799	Familial developmental dysphasia	Disorder	6 Family(ies)
168454	Spondyloepimetaphyseal dysplasia, Geneviève type	Disorder	6 Family(ies)
157801	Mesoaxial synostotic syndactyly with phalangeal reduction	Disorder	6 Family(ies)
140917	Stapes ankylosis with broad thumbs and toes	Disorder	6 Family(ies)
137634	Overgrowth-macrocephaly-facial dysmorphism syndrome	Disorder	6 Family(ies)
101068	Congenital stromal corneal dystrophy	Disorder	6 Family(ies)
99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F	Disorder	5 Family(ies)
98766	Spinocerebellar ataxia type 5	Disorder	5 Family(ies)
86789	Isolated patella aplasia/hypoplasia	Disorder	5 Family(ies)
85442	Short stature-pituitary and cerebellar defects-small sella turcica syndrome	Disorder	5 Family(ies)
444092	Autoimmune interstitial lung disease-arthritis syndrome	Disorder	5 Family(ies)
424099	Colobomatous microphthalmia-rhizomelic dysplasia syndrome	Disorder	5 Family(ies)
3454	Intellectual disability-developmental delay-contractures syndrome	Disorder	5 Family(ies)
3351	Trichodental syndrome	Disorder	5 Family(ies)
3301	Tetraamelia-multiple malformations syndrome	Disorder	5 Family(ies)
320411	Autosomal recessive spastic paraplegia type 56	Disorder	5 Family(ies)
319640	Retinal macular dystrophy type 2	Disorder	5 Family(ies)
308410	Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency	Disorder	5 Family(ies)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of families
231108	Rhabdoid tumor predisposition syndrome	Disorder	5 Family(ies)
2118	Hawkinsinuria	Disorder	5 Family(ies)
206554	Fukutin-related limb-girdle muscular dystrophy R13	Disorder	5 Family(ies)
1879	Melorheostosis with osteopoikilosis	Disorder	5 Family(ies)
1836	Mesomelic dysplasia, Kantaputra type	Disorder	5 Family(ies)
178333	Åland Islands eye disease	Disorder	5 Family(ies)
171851	MEDNIK syndrome	Disorder	5 Family(ies)
139583	X-linked hereditary sensory and autonomic neuropathy with deafness	Disorder	5 Family(ies)
101039	Female restricted epilepsy with intellectual disability	Disorder	5 Family(ies)
101003	Autosomal recessive spastic paraplegia type 23	Disorder	5 Family(ies)
98890	Early-onset X-linked optic atrophy	Disorder	4 Family(ies)
97239	Reducing body myopathy	Disorder	4 Family(ies)
93302	Brachyolmia, Maroteaux type	Disorder	4 Family(ies)
93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis	Disorder	4 Family(ies)
77297	Majeed syndrome	Disorder	4 Family(ies)
466806	Autosomal dominant thrombocytopenia with platelet secretion defect	Disorder	4 Family(ies)
46348	Paroxysmal extreme pain disorder	Disorder	4 Family(ies)
423296	Spinocerebellar ataxia type 38	Disorder	4 Family(ies)
412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome	Disorder	4 Family(ies)
401911	AXIN2-related attenuated familial adenomatous polyposis	Subtype of disorder	4 Family(ies)
363694	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome	Disorder	4 Family(ies)
2947	Triphalangeal thumbs-brachyectrodactyly syndrome	Disorder	4 Family(ies)
293936	EDICT syndrome	Disorder	4 Family(ies)
2699	Median nodule of the upper lip	Disorder	4 Family(ies)
266	Autosomal dominant limb-girdle muscular dystrophy type 1A	Disorder	4 Family(ies)
238578	Familial clubfoot due to 17q23.1q23.2 microduplication	Subtype of disorder	4 Family(ies)
2307	IVIC syndrome	Disorder	4 Family(ies)
228012	Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	Disorder	4 Family(ies)
139525	Distal hereditary motor neuropathy type 2	Disorder	4 Family(ies)
1275	Brachydactyly-elbow wrist dysplasia syndrome	Disorder	4 Family(ies)
1187	Lethal ataxia with deafness and optic atrophy	Disorder	4 Family(ies)
101108	Spinocerebellar ataxia type 23	Disorder	4 Family(ies)
101077	X-linked Charcot-Marie-Tooth disease type 3	Disorder	4 Family(ies)
97249	Pontocerebellar hypoplasia type 3	Disorder	3 Family(ies)
95433	Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome	Disorder	3 Family(ies)
94064	Deafness-infertility syndrome	Disorder	3 Family(ies)
895	Waardenburg syndrome type 2	Subtype of disorder	3 Family(ies)
67044	Thrombocytopenia with congenital dyserythropoietic	Disorder	3 Family(ies)

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ORPHAcode	Disease or Subtype of disease	Classification Level	Number of families
	anemia		
67036	Autosomal dominant optic atrophy and cataract	Disorder	3 Family(ies)
530849	Familial apolipoprotein A5 deficiency	Subtype of disorder	3 Family(ies)
488647	DDX41-related hematologic malignancy predisposition syndrome	Disorder	3 Family(ies)
466921	Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome	Disorder	3 Family(ies)
447757	Autosomal dominant spastic paraplegia type 9B	Disorder	3 Family(ies)
444072	Cerebellar-facial-dental syndrome	Disorder	3 Family(ies)
435804	Short stature-advanced bone age-early-onset osteoarthritis syndrome	Disorder	3 Family(ies)
3466	WT limb-blood syndrome	Disorder	3 Family(ies)
329319	Thrombocythemia with distal limb defects	Disorder	3 Family(ies)
319340	Carney complex-trismus-pseudocamptodactyly syndrome	Disorder	3 Family(ies)
314978	X-linked non progressive cerebellar ataxia	Disorder	3 Family(ies)
300359	PLCG2-associated antibody deficiency and immune dysregulation	Disorder	3 Family(ies)
2994	Short stature-craniofacial anomalies-genital hypoplasia syndrome	Disorder	3 Family(ies)
280628	Familial progressive hyper- and hypopigmentation	Disorder	3 Family(ies)
2211	Hypertelorism-hypospadias-polysyndactyly syndrome	Disorder	3 Family(ies)
2066	Gamma-aminobutyric acid transaminase deficiency	Disorder	3 Family(ies)
162	Congenital cataract-anterior segment dysgenesis syndrome	Disorder	3 Family(ies)
1541	Craniosynostosis, Boston type	Disorder	3 Family(ies)
140963	Bilateral microtia-deafness-cleft palate syndrome	Disorder	3 Family(ies)
1252	Blepharonasofacial malformation syndrome	Disorder	3 Family(ies)
1182	Spastic ataxia with congenital miosis	Disorder	3 Family(ies)
1074	Ankyloblepharon filiforme adnatum-imperforate anus syndrome	Subtype of disorder	3 Family(ies)
101010	Autosomal spastic paraplegia type 30	Disorder	3 Family(ies)
99846	Autosomal dominant myoglobinuria	Disorder	2 Family(ies)
98606	Syndromic orbital border hypoplasia	Disorder	2 Family(ies)
973	Isolated absence/hypoplasia of fingers excluding thumb, unilateral	Disorder	2 Family(ies)
94083	Partington syndrome	Disorder	2 Family(ies)
93409	Brachydactyly-syndactyly, Zhao type	Disorder	2 Family(ies)
86818	Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	Disorder	2 Family(ies)
85287	X-linked intellectual disability, Siderius type	Disorder	2 Family(ies)
83639	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency	Disorder	2 Family(ies)
79141	Hereditary painful callosities	Disorder	2 Family(ies)
79136	Episodic ataxia type 4	Disorder	2 Family(ies)
75497	X-linked Ehlers-Danlos syndrome	Disorder	2 Family(ies)
75373	Progressive bifocal chorioretinal atrophy	Disorder	2 Family(ies)
75327	North Carolina macular dystrophy	Disorder	2 Family(ies)

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67045	X-linked intellectual disability with isolated growth hormone deficiency	Subtype of disorder	2 Family(ies)
65720	Arthrogryposis-severe scoliosis syndrome	Disorder	2 Family(ies)
568065	EPHB4-related lymphatic-related hydrops fetalis	Disorder	2 Family(ies)
55596	HNRNPDL-related limb-girdle muscular dystrophy D3	Disorder	2 Family(ies)
535453	Familial lipase maturation factor 1 deficiency	Subtype of disorder	2 Family(ies)
476113	Combined immunodeficiency due to TFRC deficiency	Disorder	2 Family(ies)
447760	Autosomal recessive spastic paraplegia type 9B	Disorder	2 Family(ies)
447753	Autosomal dominant spastic paraplegia type 9A	Disorder	2 Family(ies)
443087	46,XY difference of sex development due to testicular 17,20-desmolase deficiency	Disorder	2 Family(ies)
439254	ITM2B amyloidosis	Disorder	2 Family(ies)
434179	Orofaciodigital syndrome type 14	Disorder	2 Family(ies)
42665	Tietz syndrome	Disorder	2 Family(ies)
420492	Adult-onset cervical dystonia, DYT23 type	Disorder	2 Family(ies)
411788	Familial isolated trichomegaly	Disorder	2 Family(ies)
401964	Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons	Disorder	2 Family(ies)
352403	Spectrin-associated autosomal recessive cerebellar ataxia	Disorder	2 Family(ies)
329191	Tall stature-long halluces-multiple extra-epiphyses syndrome	Disorder	2 Family(ies)
324718	ABetaA21G amyloidosis	Subtype of disorder	2 Family(ies)
324708	ABeta amyloidosis, Iowa type	Subtype of disorder	2 Family(ies)
3233	Cochleosaccular degeneration-cataract syndrome	Disorder	2 Family(ies)
3085	Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome	Disorder	2 Family(ies)
3034	Delayed membranous cranial ossification	Disorder	2 Family(ies)
300576	Oligodontia-cancer predisposition syndrome	Disorder	2 Family(ies)
2818	Spastic paraplegia-glaucoma-intellectual disability syndrome	Disorder	2 Family(ies)
2754	Orofaciodigital syndrome type 6	Disorder	2 Family(ies)
263463	CHST3-related skeletal dysplasia	Disorder	2 Family(ies)
2504	Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome	Disorder	2 Family(ies)
2471	McDonough syndrome	Disorder	2 Family(ies)
2405	Thickened earlobes-conductive deafness syndrome	Disorder	2 Family(ies)
2379	Early-onset parkinsonism-intellectual disability syndrome	Disorder	2 Family(ies)
2251	Thumb deformity-alopecia-pigmentation anomaly syndrome	Disorder	2 Family(ies)
2239	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	Subtype of disorder	2 Family(ies)
217622	Sensorineural deafness with dilated cardiomyopathy	Disorder	2 Family(ies)
2027	Gingival fibromatosis-progressive deafness syndrome	Disorder	2 Family(ies)
1895	Edinburgh malformation syndrome	Disorder	2 Family(ies)
1867	Hereditary bullous dystrophy, macular type	Disorder	2 Family(ies)
1786	Acrofacial dysostosis, Catania type	Disorder	2 Family(ies)
139564	Hereditary sensory and autonomic neuropathy type 1B	Disorder	2 Family(ies)

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139557	X-linked distal spinal muscular atrophy type 3	Disorder	2 Family(ies)
139480	Autosomal recessive spastic paraplegia type 39	Disorder	2 Family(ies)
139471	Microphthalmia with brain and digit anomalies	Disorder	2 Family(ies)
1350	Heart-hand syndrome type 2	Disorder	2 Family(ies)
1349	Mitochondrial DNA-related cardiomyopathy and hearing loss	Disorder	2 Family(ies)
1241	Bencze syndrome	Disorder	2 Family(ies)
114	Auriculoosteodysplasia	Disorder	2 Family(ies)
99946	Autosomal dominant Charcot-Marie-Tooth disease type 2A1	Disorder	1 Family(ies)
99945	Autosomal dominant Charcot-Marie-Tooth disease type 2L	Disorder	1 Family(ies)
99941	Autosomal dominant Charcot-Marie-Tooth disease type 2G	Disorder	1 Family(ies)
99806	Oculootodental syndrome	Disorder	1 Family(ies)
998	Albinism-deafness syndrome	Disorder	1 Family(ies)
99792	Dentin dysplasia-sclerotic bones syndrome	Disorder	1 Family(ies)
98959	Subepithelial mucinous corneal dystrophy	Disorder	1 Family(ies)
93397	Brachydactyly type A7	Disorder	1 Family(ies)
93283	Spondyloepiphyseal dysplasia, Kimberley type	Disorder	1 Family(ies)
85335	Fried syndrome	Disorder	1 Family(ies)
85322	X-linked intellectual disability, Pai type	Disorder	1 Family(ies)
85292	X-linked spinocerebellar ataxia type 4	Disorder	1 Family(ies)
85288	X-linked intellectual disability, Stocco Dos Santos type	Disorder	1 Family(ies)
85168	Craniofacial conodysplasia	Disorder	1 Family(ies)
84093	Hereditary thermosensitive neuropathy	Disorder	1 Family(ies)
79135	Episodic ataxia type 3	Disorder	1 Family(ies)
79129	Trichodysplasia-amelogenesis imperfecta syndrome	Disorder	1 Family(ies)
79085	AKT2-related familial partial lipodystrophy	Disorder	1 Family(ies)
69083	Ectodermal dysplasia with natal teeth, Turnpenny type	Disorder	1 Family(ies)
589522	Spinocerebellar ataxia type 46	Disorder	1 Family(ies)
574918	Predisposition to severe viral infection due to IRF7 deficiency	Disorder	1 Family(ies)
52056	Ulnar/fibula ray defect-brachydactyly syndrome	Disorder	1 Family(ies)
498602	Sugarman brachydactyly	Disorder	1 Family(ies)
488437	SIX2-related frontonasal dysplasia	Disorder	1 Family(ies)
476119	Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome	Disorder	1 Family(ies)
444099	Autosomal dominant spastic paraplegia type 73	Disorder	1 Family(ies)
443162	NDE1-related microhydranencephaly	Disorder	1 Family(ies)
440354	Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome	Disorder	1 Family(ies)
431140	X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome	Disorder	1 Family(ies)
391327	X-linked calvarial hyperostosis	Disorder	1 Family(ies)
370131	White platelet syndrome	Disorder	1 Family(ies)
370091	Oculocutaneous albinism type 5	Disorder	1 Family(ies)

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363727	X-linked dyserythropoietic anemia with abnormal platelets and neutropenia	Disorder	1 Family(ies)
3417	Van den Bosch syndrome	Disorder	1 Family(ies)
3408	Upington disease	Disorder	1 Family(ies)
3361	Trichodysplasia-xeroderma syndrome	Disorder	1 Family(ies)
329883	Non-hypoproteinemic hypertrophic gastropathy	Disorder	1 Family(ies)
329475	Spastic paraplegia-Paget disease of bone syndrome	Disorder	1 Family(ies)
324723	ABeta amyloidosis, Arctic type	Subtype of disorder	1 Family(ies)
324703	ABetaL34V amyloidosis	Subtype of disorder	1 Family(ies)
320365	Autosomal dominant spastic paraplegia type 36	Disorder	1 Family(ies)
3196	Steroid dehydrogenase deficiency-dental anomalies syndrome	Disorder	1 Family(ies)
319332	Autosomal recessive myogenic arthrogryposis multiplex congenita	Disorder	1 Family(ies)
3191	Subaortic stenosis-short stature syndrome	Disorder	1 Family(ies)
314889	Autosomal dominant proximal renal tubular acidosis	Subtype of disorder	1 Family(ies)
300305	11p15.4 microduplication syndrome	Disorder	1 Family(ies)
2999	Ptosis-strabismus-ectopic pupils syndrome	Disorder	1 Family(ies)
293375	Grayson-Wilbrandt corneal dystrophy	Disorder	1 Family(ies)
2917	Polydactyly-myopia syndrome	Disorder	1 Family(ies)
2890	Pili torti-onychodysplasia syndrome	Disorder	1 Family(ies)
2835	Pectus excavatum-macrocephaly-dysplastic nails syndrome	Disorder	1 Family(ies)
2821	Spastic paraplegia-neuropathy-poikiloderma syndrome	Disorder	1 Family(ies)
276183	Spinocerebellar ataxia type 32	Disorder	1 Family(ies)
275517	Autoimmune lymphoproliferative syndrome-recurrent viral infections due to CASP8 deficiency	Disorder	1 Family(ies)
2709	Oculodental syndrome, Rutherfurd type	Disorder	1 Family(ies)
2674	Cyprus facial-neuromusculoskeletal syndrome	Disorder	1 Family(ies)
2663	Nathalie syndrome	Disorder	1 Family(ies)
2572	Spastic ataxia-corneal dystrophy syndrome	Disorder	1 Family(ies)
2565	Mononen-Karnes-Senac syndrome	Disorder	1 Family(ies)
2408	Lowe-Kohn-Cohen syndrome	Disorder	1 Family(ies)
2391	Congenitally short costocoracoid ligament	Disorder	1 Family(ies)
231742	Epibulbar lipodermoid-preauricular appendage-polythelia syndrome	Disorder	1 Family(ies)
2201	Palmoplantar keratoderma-spastic paralysis syndrome	Disorder	1 Family(ies)
2186	Hydrocephalus-blue sclerae-nephropathy syndrome	Disorder	1 Family(ies)
2097	Grant syndrome	Disorder	1 Family(ies)
2090	GMS syndrome	Disorder	1 Family(ies)
1979	Lipodystrophy due to peptidic growth factors deficiency	Disorder	1 Family(ies)
1962	Exostoses-anetodermia-brachydactyly type E syndrome	Disorder	1 Family(ies)
1892	Ectrodactyly-polydactyly syndrome	Disorder	1 Family(ies)
1876	Oculogastrointestinal muscular dystrophy	Disorder	1 Family(ies)
171863	Autosomal dominant spastic paraplegia type 42	Disorder	1 Family(ies)
171622	Autosomal recessive spastic paraplegia type 32	Disorder	1 Family(ies)
171617	Autosomal dominant spastic paraplegia type 38	Disorder	1 Family(ies)

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166108	Intellectual disability, Birk-Barel type	Disorder	1 Family(ies)
163727	Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	Disorder	1 Family(ies)
163662	Spondyloepiphyseal dysplasia, Reardon type	Disorder	1 Family(ies)
1551	Familial benign copper deficiency	Disorder	1 Family(ies)
1527	Craniosynostosis, Philadelphia type	Disorder	1 Family(ies)
140922	Titin-related limb-girdle muscular dystrophy R10	Disorder	1 Family(ies)
140481	Autosomal dominant slowed nerve conduction velocity	Disorder	1 Family(ies)
139512	Neuropathy with hearing impairment	Disorder	1 Family(ies)
139450	Microtia-eye coloboma-imperforation of the nasolacrimal duct syndrome	Disorder	1 Family(ies)
137776	Lethal congenital contracture syndrome type 2	Disorder	1 Family(ies)
1319	Camptobrachydactyly	Disorder	1 Family(ies)
1246	Brachydactyly-nystagmus-cerebellar ataxia syndrome	Disorder	1 Family(ies)
1228	Banki syndrome	Disorder	1 Family(ies)
1144	Arthrogryposis-like hand anomaly-sensorineural deafness syndrome	Disorder	1 Family(ies)
1122	Ulnar hypoplasia-split foot syndrome	Disorder	1 Family(ies)
101112	Spinocerebellar ataxia type 26	Disorder	1 Family(ies)
101101	Charcot-Marie-Tooth disease type 2B2	Disorder	1 Family(ies)
101009	Autosomal dominant spastic paraplegia type 29	Disorder	1 Family(ies)
101005	Autosomal recessive spastic paraplegia type 25	Disorder	1 Family(ies)
101004	Autosomal recessive spastic paraplegia type 24	Disorder	1 Family(ies)
100999	Autosomal dominant spastic paraplegia type 19	Disorder	1 Family(ies)
100997	X-linked spastic paraplegia type 16	Disorder	1 Family(ies)
100995	Autosomal recessive spastic paraplegia type 14	Disorder	1 Family(ies)

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To access the complete Orphanet epidemiological data sets visit Orphadata (www.orphadata.com).
For any questions or comments, please contact us: contact.orphanet@inserm.fr

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