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## Prevalence and incidence of rare diseases: Bibliographic data

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

[www.orpha.net](http://www.orpha.net)

[www.orphadata.org](http://www.orphadata.org)

# 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

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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 cohorts studies).

For congenital diseases, the prevalence is estimated, so that:

$$\text{Prevalence} = \text{birth prevalence} \times (\text{patient life expectancy}/\text{general population life expectancy}).$$

When only incidence data is documented, the prevalence is estimated when possible, so that :  
$$\text{Prevalence} = \text{incidence} \times \text{disease mean duration}.$$

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.

## 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

Without specification, published figures are worldwide. An asterisk \* indicates European data. BP indicates birth prevalence

Please note that this is just a selection of Orphanet's rare disease epidemiological data. Currently 5949 rare diseases are annotated with prevalence or incidence information in the Orphanet database. To access the complete data sets visit Orphadata ([www.orphadata.org](http://www.orphadata.org)).

# List of diseases or groups of diseases by decreasing prevalence

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
870	Down syndrome	95.0 BP
3388	Neural tube defect	91.05 BP*
199306	Cleft lip/palate	80.0 BP
182130	Tumor of endocrine glands	64.0 *
2014	Cleft palate	53.6 BP*
535	Rare cutaneous lupus erythematosus	50.0 *
8	47,XYY syndrome	50.0 BP*
48	Congenital bilateral absence of vas deferens	50.0 *
63259	Iniencephaly	50.0 *
90066	Pneumonia caused by Pseudomonas aeruginosa infection	50.0 *
93100	Renal agenesis, unilateral	50.0 BP
289390	Primary Sjögren syndrome	48.99 *
67038	B-cell chronic lymphocytic leukemia	48.0 *
2185	Congenital hydrocephalus	46.5 BP*
391673	Necrotizing enterocolitis	45.0
275555	Preeclampsia	45.0 *
137686	Asherman syndrome	44.0 *
93108	Renal dysplasia	43.5 BP*
3375	Trisomy X	42.5 *
801	Scleroderma	42.0
217071	Renal cell carcinoma	42.0 *
363999	Non-immune hydrops fetalis	42.0 BP
73247	Eosinophilic esophagitis	40.08
768	Familial long QT syndrome	40.0 BP*
294	Fetal cytomegalovirus syndrome	40.0 *
98497	Genetic peripheral neuropathy	40.0
97292	Cardiogenic shock	40.0 *
101016	Romano-Ward syndrome	40.0 *
402823	Hepatitis delta	40.0 *
3189	Congenital pulmonary valve stenosis	39.3 BP*
442	Congenital hypothyroidism	38.0 BP*
98715	Uveitis	38.0 *
90056	Moderate and severe traumatic brain injury	37.8 *
567	22q11.2 deletion syndrome	37.5 BP
226295	Primary congenital hypothyroidism	37.5 *
90059	Acute sensorineural hearing loss by acute acoustic trauma or sudden deafness or surgery induced acoustic trauma	37.0 *
209989	Non-papillary transitional cell carcinoma of the bladder	37.0 *

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
340	Hemorrhagic fever-renal syndrome	37.0 *
97230	Solar urticaria	36.0 *
231080	High-grade dysplasia in patients with Barrett esophagus	36.0 *
1457	Aorta coarctation	35.6 BP*
2764	Osteochondritis dissecans	35.0 *
1048	Isolated anencephaly/exencephaly	35.0 BP*
70475	Radiation proctitis	35.0 *
94059	Uremic pruritus	35.0 *
3303	Tetralogy of Fallot	34.0 BP
636	Neurofibromatosis type 1	33.3 BP
226292	Permanent congenital hypothyroidism	33.3 BP*
858	Congenital toxoplasmosis	33.0 BP*
439167	Placental insufficiency	33.0
908	Fragile X syndrome	32.5
70476	Vernal keratoconjunctivitis	32.0 *
90051	Sepsis in premature infants	32.0 *
90058	Spinal cord injury	32.0 *
216675	Transposition of the great arteries	31.7 BP*
2140	Congenital diaphragmatic hernia	30.0 BP
3394	Soft tissue sarcoma	30.0 *
563	Peripartum cardiomyopathy	30.0 BP
1330	Partial atrioventricular canal	30.0 *
729	Polycythemia vera	30.0 *
213500	Ovarian cancer	30.0 *
330001	Wild type ATTR amyloidosis	30.0 *
33208	Idiopathic hypersomnia	30.0 *
314701	Primary systemic amyloidosis	30.0 *
467	Non-acquired combined pituitary hormone deficiency	29.0 BP*
545	Follicular lymphoma	28.0 *
411527	Central retinal vein occlusion	28.0 *
1656	Dermatitis herpetiformis	27.0 *
791	Retinitis pigmentosa	26.7
70568	Post-transplant lymphoproliferative disease	26.2 *
703	Bullous pemphigoid	26.0 *
54057	Thrombotic thrombocytopenic purpura	25.5 *
137698	Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	25.5 *
166	Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy	25.0 *
3002	Immune thrombocytopenic purpura	25.0 *
701	Alopecia universalis	25.0 *
145	Hereditary breast and ovarian cancer syndrome	25.0 *
2073	Narcolepsy type 1	25.0 *
95719	Thyroid hemiagenesis	25.0

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93402	Syndactyly type 1	25.0 BP*
94058	Neovascular glaucoma	24.4 *
1199	Esophageal atresia	24.3 BP*
1531	Craniosynostosis	24.3 BP*
860	Congenitally uncorrected transposition of the great arteries	24.25 BP*
2248	Hypoplastic left heart syndrome	24.0 BP
171901	Primary cutaneous T-cell lymphoma	24.0 *
2368	Gastroschisis	23.7 BP*
2137	Autoimmune hepatitis	23.5
1851	Multicystic dysplastic kidney	23.26 BP
97363	Unilateral multicystic dysplastic kidney	23.2 BP
228113	Anal fistula	23.0 *
232	Sickle cell anemia	22.0 *
90080	Scarring in glaucoma filtration surgical procedures	22.0 *
217067	Pouchitis	22.0 *
217080	Pulmonary fungal infections in patients deemed at risk	22.0 *
636	Neurofibromatosis type 1	21.3 *
95711	Congenital hypothyroidism due to developmental anomaly	21.3 *
186	Primary biliary cholangitis	21.05
1646	Partial chromosome Y deletion	20.8
85410	Oligoarticular juvenile idiopathic arthritis	20.5 *
60	Alpha-1-antitrypsin deficiency	20.0 *
70	Proximal spinal muscular atrophy	20.0 BP*
1329	Complete atrioventricular canal	20.0 BP*
557	Isolated anorectal malformation	20.0 BP
130	Brugada syndrome	20.0 *
35122	Congenital sucrase-isomaltase deficiency	20.0 *
1330	Partial atrioventricular canal	20.0 BP*
247	Arrhythmogenic right ventricular cardiomyopathy	20.0
70587	Infant acute respiratory distress syndrome	20.0 *
90062	Acute liver failure	20.0 *
90081	AIDS wasting syndrome	20.0 *
261197	Proximal 16p11.2 microdeletion syndrome	20.0 *
137583	Vulvar intraepithelial neoplasia	20.0 *
66627	Pigmented villonodular synovitis	20.0 *
3389	Tuberculosis	20.0 *
823	Isolated spina bifida	18.6 BP*
30391	Isolated biliary atresia	18.5 BP
704	Pemphigus vulgaris	18.0 *
90061	Non-infectious posterior uveitis	18.0 *
91127	Adenovirus infection in immunocompromised patients	18.0 *
154	Familial isolated dilated cardiomyopathy	17.5 *

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
65753	Charcot-Marie-Tooth disease type 1	17.5
3380	Trisomy 18	16.7 BP
77240	Primary lymphedema	16.7 *
461	Recessive X-linked ichthyosis	16.6 *
774	Hereditary hemorrhagic telangiectasia	16.0 *
1201	Atresia of small intestine	16.0 BP*
36258	Buerger disease	16.0
544	Diffuse large B-cell lymphoma	16.0 *
54370	Primary membranoproliferative glomerulonephritis	16.0 *
90064	Acute peripheral arterial occlusion	16.0 *
137599	Stromal keratitis	16.0 *
83463	Microtia	15.5 BP
90291	Systemic sclerosis	15.4 *
98896	Duchenne muscular dystrophy	15.1 BP*
558	Marfan syndrome	15.0
2382	Lennox-Gastaut syndrome	15.0 *
2828	Young-onset Parkinson disease	15.0 *
88673	Hepatocellular carcinoma	15.0 *
221061	Familial cerebral cavernous malformation	15.0
163934	Atopic keratoconjunctivitis	15.0 *
309297	Mucopolysaccharidosis type 4A	15.0 *
166260	Dentinogenesis imperfecta type 2	14.6 *
49042	Dentinogenesis imperfecta	14.5 *
95712	Thyroid ectopia	14.3 *
214	Cystinuria	14.0
101959	Chronic primary adrenal insufficiency	14.0 *
238624	Idiopathic intracranial hypertension	14.0 *
329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis	14.0 *
2162	Holoprosencephaly	13.4 BP*
3193	Supravalvular aortic stenosis	13.3 *
44890	Gastrointestinal stromal tumor	13.0 *
70589	Bronchopulmonary dysplasia	13.0 *
449266	Pleural empyema	13.0 *
423461	Mucolipidosis type III alpha/beta	13.0
100088	Thyroid carcinoma	12.7
3376	Triploidy	12.6 BP*
273	Steinert myotonic dystrophy	12.5
797	Sarcoidosis	12.5
285	Ehlers-Danlos syndrome, hypermobility type	12.5 *
903	Von Willebrand disease	12.5
2415	Rare lymphatic malformation	12.5 *
85138	Addison disease	12.5 *
828	Stickler syndrome	12.2 BP

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42	Medium chain acyl-CoA dehydrogenase deficiency	12.0 BP*
95426	Chronic pain requiring intraspinal analgesia	12.0 *
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	12.0 *
805	Tuberous sclerosis complex	12.0 *
86870	CD4+/CD56+ hematodermic neoplasm	12.0 *
29073	Multiple myeloma	11.9 *
660	Omphalocele	11.7 BP*
1866	Focal, segmental or multifocal dystonia	11.7 *
2032	Idiopathic pulmonary fibrosis	11.5 *
98878	Hemophilia A	11.25 BP
70573	Small cell lung cancer	11.2 *
635	Neuroblastoma	11.0 *
3109	Mayer-Rokitansky-Küster-Hauser syndrome	11.0 BP
513	Acute lymphoblastic leukemia	11.0 *
890	Hepatic veno-occlusive disease	11.0 *
85443	AL amyloidosis	11.0 *
388	Hirschsprung disease	10.9 BP*
904	Williams syndrome	10.8 BP
700	Alopecia totalis	10.5 *
778	Rett syndrome	10.0 *
580	Mucopolysaccharidosis type 2	10.0 *
827	Stargardt disease	10.0 *
1146	Digitotalar dysmorphism	10.0
2612	Linear nevus sebaceus syndrome	10.0 BP*
716	Phenylketonuria	10.0 BP*
805	Tuberous sclerosis complex	10.0 BP*
666	Osteogenesis imperfecta	10.0 *
654	Nephroblastoma	10.0 BP*
569	Familial or sporadic hemiplegic migraine	10.0 *
233	Duane retraction syndrome	10.0 *
51	Aicardi-Goutières syndrome	10.0 *
3157	Septo-optic dysplasia spectrum	10.0 BP*
412	Hyperlipoproteinemia type 3	10.0
3286	Catecholaminergic polymorphic ventricular tachycardia	10.0 *
31112	Dermatofibrosarcoma protuberans	10.0 *
64740	Recurrent acute pancreatitis	10.0 *
90065	Acquired aneurysmal subarachnoid hemorrhage	10.0 *
90076	Partial deep dermal and full thickness burns	10.0 *
183422	Polymalformative genetic syndrome with increased risk of developing cancer	10.0 *
182067	Glial tumor	10.0 *
1114	Aplasia cutis congenita	10.0 BP
2177	Hydranencephaly	10.0 BP

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
70482	Carcinoma of esophagus	9.8
223727	Bone sarcoma	9.29 *
79665	Gardner syndrome	9.1 BP
2443	Mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies	9.0 *
900	Granulomatosis with polyangiitis	9.0 *
1203	Duodenal atresia	9.0 *
1203	Duodenal atresia	9.0 BP*
98292	Mastocytosis	9.0 *
99125	Congenital total pulmonary venous return anomaly	9.0 BP
306644	Complication after organ transplantation	9.0 *
137914	Choanal atresia	8.6 BP*
99981	Apnea of prematurity	8.5 *
3280	Syringomyelia	8.4 *
98555	Microphthalmia-anophthalmia-coloboma	8.3 BP*
2444	Congenital pulmonary airway malformation	8.2 BP*
171	Primary sclerosing cholangitis	8.1
930	Idiopathic achalasia	8.0
90290	CREST syndrome	8.0 *
85408	Rheumatoid factor-negative juvenile idiopathic arthritis	8.0 *
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	8.0 *
95702	Cytomegalic congenital adrenal hypoplasia	8.0 BP
88991	Rare congenital non-syndromic heart malformation	7.8 BP*
589	Myasthenia gravis	7.77
448	Hemophilia	7.7 *
247234	Sporadic adult-onset ataxia of unknown etiology	7.6 *
72	Angelman syndrome	7.5
2004	Laryngotracheoesophageal cleft	7.5 BP*
1464	Univentricular heart	7.5 BP
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	7.5 *
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	7.5 BP*
586	Cystic fibrosis	7.4 *
821	Sotos syndrome	7.1 BP
732	Polymyositis	7.1 *
705	Pendred syndrome	7.0 *
1332	Medullary thyroid carcinoma	7.0 *
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	7.0 BP

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90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	7.0 *
90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients	7.0 *
95432	Primary progressive aphasia	7.0
261236	16p13.11 microdeletion syndrome	7.0 BP
2059	Fryns syndrome	7.0 BP*
300912	Marginal zone lymphoma	7.0 *
42	Medium chain acyl-CoA dehydrogenase deficiency	6.85
418	Congenital adrenal hyperplasia	6.7 BP*
3366	Isolated trigonocephaly	6.7 BP*
57145	SUNCT syndrome	6.7 *
238468	Hypohidrotic ectodermal dysplasia	6.7 *
206647	Myotonic dystrophy	6.7
42062	Iminoglycinuria	6.68 *
42062	Iminoglycinuria	6.67 BP*
50839	Cat-scratch disease	6.6 *
138	CHARGE syndrome	6.5 BP
52759	Vasculitis	6.3 *
887	VACTERL/VATER association	6.25 BP*
363958	17q21.31 microdeletion syndrome	6.25 *
733	Familial adenomatous polyposis	6.0 *
790	Retinoblastoma	6.0 BP
418	Congenital adrenal hyperplasia	6.0 *
221	Dermatomyositis	6.0 *
683	Progressive supranuclear palsy	6.0
3451	West syndrome	6.0 *
521	Chronic myeloid leukemia	6.0 *
609	Tibial muscular dystrophy	6.0 *
46724	Cerebral arteriovenous malformation	6.0 *
252164	Benign schwannoma	6.0 *
524	Li-Fraumeni syndrome	6.0
411703	Pulmonary non-tuberculous mycobacterial infection	6.0 *
55	Oculocutaneous albinism	5.9
553	Cushing syndrome	5.9
635	Neuroblastoma	5.8 BP*
1037	Arthrogryposis multiplex congenita	5.7 BP*
85438	Enthesitis-related juvenile idiopathic arthritis	5.7 *
881	Turner syndrome	5.5 BP*
963	Acromegaly	5.5
93372	Familial hypocalciuric hypercalcemia type 1	5.5
2440	Isolated split hand-split foot malformation	5.4 BP*
2542	Isolated microphthalmia-anophthalmia-coloboma	5.3 BP*
738	Porphyria	5.25

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
685	Hereditary spastic paraparesis	5.2
778	Rett syndrome	5.0 BP*
792	X-linked retinoschisis	5.0
469	Hereditary fructose intolerance	5.0 *
718	Isolated Pierre Robin syndrome	5.0 BP*
287	Ehlers-Danlos syndrome, classic type	5.0
244	Primary ciliary dyskinesia	5.0 BP*
43	X-linked adrenoleukodystrophy	5.0 BP
251	Multiple epiphyseal dysplasia	5.0 *
766	Hemolytic anemia due to red cell pyruvate kinase deficiency	5.0 *
90309	Ehlers-Danlos syndrome type 1	5.0 *
85414	Systemic-onset juvenile idiopathic arthritis	5.0 *
280062	Calciphylaxis	5.0 *
79271	Sanfilippo syndrome type C	5.0 *
309152	GM2 gangliosidosis	5.0 *
98878	Hemophilia A	4.85
88629	Tritanopia	4.8 *
98896	Duchenne muscular dystrophy	4.78
60041	Congenital heart block	4.54 BP
269	Facioscapulohumeral dystrophy	4.5 *
85446	Wild type ABeta2M amyloidosis	4.5 *
104	Leber hereditary optic neuropathy	4.3
1143	Neurogenic arthrogryposis multiplex congenita	4.3 BP*
3384	Truncus arteriosus	4.3 BP
2116	Hartnup disease	4.2
1209	Tricuspid atresia	4.2 BP*
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	4.2 *
85436	Psoriasis-related juvenile idiopathic arthritis	4.2 *
137596	Neurotrophic keratopathy	4.2 *
2130	Hemimelia	4.15 *
391655	Off-periods in Parkinson disease not responding to oral treatment	4.15 *
93110	Posterior urethral valve	4.125 BP*
15	Achondroplasia	4.0 BP
564	Meckel syndrome	4.0 BP
819	Smith-Magenis syndrome	4.0
884	Tetrasomy 12p	4.0 BP*
1928	Congenital lobar emphysema	4.0 BP
3193	Supravalvular aortic stenosis	4.0 BP*
52417	MALT lymphoma	4.0 *
79140	Cutaneous neuroendocrine carcinoma	4.0 *
96169	Koolen-De Vries syndrome	4.0 *
96253	Cushing disease	4.0 *
95716	Familial thyroid dyshormonogenesis	4.0 *

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101330	Porphyria cutanea tarda	4.0 *
99013	Spastic paraplegia type 7	4.0 *
178029	Central diabetes insipidus	4.0 *
730	Autosomal dominant polycystic kidney disease	3.96 *
96092	8p inverted duplication/deletion syndrome	3.9 BP*
803	Amyotrophic lateral sclerosis	3.85
79126	Acute interstitial pneumonia	3.8 *
98848	Indolent systemic mastocytosis	3.8 *
2467	Systemic mastocytosis	3.75
478	Kallmann syndrome	3.75 *
3378	Trisomy 13	3.7 BP*
3451	West syndrome	3.7 BP
2932	Chronic inflammatory demyelinating polyneuropathy	3.7 *
3465	Worster-Drought syndrome	3.7 *
818	Smith-Lemli-Opitz syndrome	3.7 BP*
60015	Enlarged parietal foramina	3.7
98976	Congenital glaucoma	3.6 BP*
640	Hereditary neuropathy with liability to pressure palsies	3.5 *
116	Beckwith-Wiedemann syndrome	3.5 BP*
1880	Ebstein malformation	3.5 BP*
2655	Thanatophoric dysplasia	3.5 BP*
3205	Sturge-Weber syndrome	3.5 BP*
2103	Guillain-Barré syndrome	3.5 *
81	Antisynthetase syndrome	3.5
102	Multiple system atrophy	3.5
52416	Mantle cell lymphoma	3.5 *
95713	Athyreosis	3.5 *
95720	Thyroid hypoplasia	3.5
217074	Rare carcinoma of pancreas	3.5
104008	Short bowel syndrome	3.4 *
218	Darier disease	3.4 *
53271	Muenke syndrome	3.33 BP
652	Multiple endocrine neoplasia type 1	3.3 *
905	Wilson disease	3.3
429	Hypochondroplasia	3.3 *
1172	Autosomal recessive cerebellar ataxia	3.3
98672	Autosomal dominant optic atrophy	3.3
98723	Hypoplastic right heart syndrome	3.3 BP*
182090	Pulmonary arterial hypertension	3.3 *
750	Pseudoachondroplasia	3.3
926	Acatalasemia	3.2 *
158	Systemic primary carnitine deficiency	3.2 BP*
100075	Neuroendocrine tumor of stomach	3.2 *
739	Prader-Willi syndrome	3.1 BP*
2322	Kabuki syndrome	3.1 *

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
50251	Pleural mesothelioma	3.1 *
98916	Acute inflammatory demyelinating polyradiculoneuropathy	3.1 *
93930	Bladder exstrophy	3.05 BP
673	Malaria	3.0 *
794	Saethre-Chotzen syndrome	3.0 BP*
136	CADASIL	3.0 *
282	Frontotemporal dementia	3.0 *
767	Polyarteritis nodosa	3.0 *
2745	Opitz G/BBB syndrome	3.0 *
39812	Graft versus host disease	3.0 *
36234	Bacterial toxic-shock syndrome	3.0
824	Primary myelofibrosis	3.0 *
70591	Chronic thromboembolic pulmonary hypertension	3.0 *
216694	Congenitally corrected transposition of the great arteries	3.0 BP
238621	Ileal pouch anal anastomosis related faecal incontinence	3.0 *
171673	Limbal stem cell deficiency	3.0 *
98838	Primary mediastinal large B-cell lymphoma	3.0 *
35098	Isolated plagiocephaly	3.0 BP
413690	Methotrexate toxicity or dose selection	3.0 *
86875	Adult T-cell leukemia/lymphoma	3.0 *
374	Goldenhar syndrome	2.9 BP*
653	Multiple endocrine neoplasia type 2	2.9 *
506	Leigh syndrome	2.8 BP*
169802	Severe hemophilia A	2.8 *
626	Large congenital melanocytic nevus	2.75 *
399	Huntington disease	2.7
99	Autosomal dominant cerebellar ataxia	2.7
49382	Achromatopsia	2.7
6	3-methylcrotonyl-CoA carboxylase deficiency	2.65 BP*
79432	Oculocutaneous albinism type 2	2.55
358	Gitelman syndrome	2.5 *
1872	Cone rod dystrophy	2.5 *
2138	46,XX ovotesticular disorder of sex development	2.5 BP
2337	Non-epidermolytic palmoplantar keratoderma	2.5 *
65	Leber congenital amaurosis	2.5 BP
758	Pseudoxanthoma elasticum	2.5 *
94	Astrocytoma	2.5 *
393	46,XX testicular disorder of sex development	2.5
33069	Dravet syndrome	2.5 BP
75249	Familial isolated restrictive cardiomyopathy	2.5 *

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97927	Peripheral resistance to thyroid hormones	2.5 *	275761	Lysosomal acid lipase deficiency	2.0 *
100070	Progressive non-fluent aphasia	2.5 *	150	Nasopharyngeal carcinoma	2.0 *
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	2.5 *	506	Leigh syndrome	2.0 *
65	Leber congenital amaurosis	2.5	363203	Ring chromosome	2.0 BP
352731	Oculocutaneous albinism type 1	2.5	90073	Hepatitis B reinfection following liver transplantation	2.0 *
1600	Monosomy 18q	2.5 BP	3392	Tularemia	2.0 *
70588	Meconium aspiration syndrome	2.44 *	1598	Monosomy 18p	2.0 BP*
93928	Isolated epispadias	2.4 BP*	98673	Autosomal dominant optic atrophy, classic form	2.0
98933	Multiple system atrophy, parkinsonian type	2.4 *	447	Paroxysmal nocturnal hemoglobinuria	2.0 *
247525	Citrullinemia type I	2.4 *	199	Cornelia de Lange syndrome	1.9 *
908	Fragile X syndrome	2.4 BP*	10	48,XXYY syndrome	1.9 BP*
263	Limb-girdle muscular dystrophy	2.32	79361	Inherited epidermolysis bullosa	1.9 BP*
330015	Lead poisoning	2.3 *	35808	Malignant sex cord stromal tumor of ovary	1.85 *
905	Wilson disease	2.2 BP	304	Epidermolysis bullosa simplex	1.8
2869	Peutz-Jeghers syndrome	2.2 BP	675	Annular pancreas	1.8 BP*
304	Epidermolysis bullosa simplex	2.2 BP*	664	Ornithine transcarbamylase deficiency	1.77 BP
137605	Legius syndrome	2.2 BP	420429	Glycogen storage disease due to acid maltase deficiency, late-onset	1.75 BP
98895	Becker muscular dystrophy	2.2 BP*	251076	8p23.1 duplication syndrome	1.72
454750	Isolated tracheoesophageal fistula	2.2 BP	2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	1.7 BP
217	Isolated Dandy-Walker malformation	2.1 *	2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	1.7
70567	Cholangiocarcinoma	2.1	637	Neurofibromatosis type 2	1.7 *
95	Friedreich ataxia	2.0 *	2152	Mowat-Wilson syndrome	1.7 BP*
480	Kearns-Sayre syndrome	2.0 *	1848	Renal agenesis, bilateral	1.7 BP*
607	Nemaline myopathy	2.0 BP*	98879	Hemophilia B	1.7 *
280	Wolf-Hirschhorn syndrome	2.0 BP*	141077	Epignathus	1.68 BP
126	Blepharophimosis-epicanthus inversus-ptosis syndrome	2.0	98879	Hemophilia B	1.665 BP
861	Treacher-Collins syndrome	2.0 BP*	394	Classic homocystinuria	1.65 *
352	Galactosemia	2.0 BP*	899	Walker-Warburg syndrome	1.65 BP*
3346	Tracheal agenesis	2.0 BP*	183660	Severe combined immunodeficiency	1.65 BP*
63	Alport syndrome	2.0 *	1915	Fetal alcohol syndrome	1.6 BP*
3129	Sarcosinemia	2.0 BP	64747	X-linked Charcot-Marie-Tooth disease	1.6 *
180	Choroideremia	2.0 *	79241	Biotinidase deficiency	1.6 *
1699	Trisomy 12p	2.0 BP	79241	Biotinidase deficiency	1.6 BP
2017	Sternal cleft	2.0 BP*	98895	Becker muscular dystrophy	1.53
185	Scimitar syndrome	2.0 BP*	192	Coffin-Lowry syndrome	1.5
2345	Isolated Klippel-Feil syndrome	2.0 *	131	Budd-Chiari syndrome	1.5 *
54595	Craniopharyngioma	2.0 *	2019	Femur-fibula-ulna complex	1.5 BP*
93323	Fibular hemimelia	2.0 *	2911	Poland syndrome	1.5 BP*
98841	Anaplastic large cell lymphoma	2.0 *	389	Langerhans cell histiocytosis	1.5 *
217064	5-fluorouracil poisoning	2.0 *	137	Congenital disorder of glycosylation	1.5 BP*
157835	Paroxysmal hemicrania	2.0 *	35689	Primary lateral sclerosis	1.5 *
168782	Childhood disintegrative disorder	2.0 *	641	Multifocal motor neuropathy	1.5
657	Congenital isolated hyperinsulinism	2.0 BP	71211	Neuromyelitis optica	1.5 *
93110	Posterior urethral valve	2.0 *			

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ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
45453	Incessant infant ventricular tachycardia	1.5 BP*
45452	Idiopathic neonatal atrial flutter	1.5 BP*
91378	Hereditary angioedema	1.5 *
98757	Spinocerebellar ataxia type 3	1.5
98756	Spinocerebellar ataxia type 2	1.5
98755	Spinocerebellar ataxia type 1	1.5
168811	Malignant peritoneal mesothelioma	1.5 *
168956	Hypereosinophilic syndrome	1.5 *
183	Eosinophilic granulomatosis with polyangiitis	1.5
213	Cystinosis	1.5 *
512	Metachromatic leukodystrophy	1.47 BP*
664	Ornithine transcarbamylase deficiency	1.4 *
474	Jeune syndrome	1.4 BP*
79269	Sanfilippo syndrome type A	1.4 BP
499009	Congenital syphilis	1.4 BP*
195	Cat-eye syndrome	1.35 BP*
3287	Takayasu arteritis	1.34 *
72	Angelman syndrome	1.3 BP*
199	Cornelia de Lange syndrome	1.3 BP*
355	Gaucher disease	1.3 BP
79434	Oculocutaneous albinism type 1B	1.3
79431	Oculocutaneous albinism type 1A	1.3
281090	Syndromic recessive X-linked ichthyosis	1.3 *
1880	Ebstein malformation	1.25 *
2481	Neurocutaneous melanocytosis	1.25 *
628	Diastrophic dwarfism	1.2 *
464	Incontinentia pigmenti	1.2 BP*
2750	Orofaciodigital syndrome type 1	1.2 BP*
46485	Superficial pemphigus	1.2 *
731	Autosomal recessive polycystic kidney disease	1.17 *
263432	Nevus of Ito	1.17 *
475	Joubert syndrome	1.125 BP
1896	EEC syndrome	1.11 BP*
289	Ellis Van Creveld syndrome	1.1 BP
224	Neonatal diabetes mellitus	1.1 BP*
140874	Joubert syndrome and related disorders	1.1 BP
275766	Idiopathic pulmonary arterial hypertension	1.1 *
377	Gorlin syndrome	1.1
1906	Fetal valproate syndrome	1.02 BP*
487	Krabbe disease	1.0 *
614	Thomsen and Becker disease	1.0
16	Blue cone monochromatism	1.0
16	Blue cone monochromatism	1.0 BP
681	Hypokalemic periodic paralysis	1.0 *
53	Albers-Schönberg osteopetrosis	1.0
205	Crigler-Najjar syndrome	1.0 *

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
370	Glycogen storage disease due to phosphorylase kinase deficiency	1.0 BP*
217	Isolated Dandy-Walker malformation	1.0 BP*
23	Argininosuccinic aciduria	1.0 *
33	Isovaleric acidemia	1.0 *
2308	Jacobsen syndrome	1.0 BP*
606	Proximal myotonic myopathy	1.0 *
355	Gaucher disease	1.0 *
364	Glycogen storage disease due to glucose-6-phosphatase deficiency	1.0 BP
646	Niemann-Pick disease type C	1.0 *
2134	Atypical hemolytic-uremic syndrome	1.0 *
267	Autosomal recessive limb-girdle muscular dystrophy type 2A	1.0 *
1552	Currarino syndrome	1.0 *
189	Hidrotic ectodermal dysplasia	1.0 *
254	Spondylometaphyseal dysplasia	1.0 BP*
2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2	1.0 BP*
296	Enchondromatosis	1.0 *
647	Nijmegen breakage syndrome	1.0 BP
2924	Isolated polycystic liver disease	1.0 *
422	Idiopathic and/or familial pulmonary arterial hypertension	1.0 *
3403	Uhl anomaly	1.0 BP
3449	Weill-Marchesani syndrome	1.0
616	Medulloblastoma	1.0 *
360	Glioblastoma	1.0
1900	Ehlers-Danlos syndrome, kyphoscoliotic type	1.0 BP
286	Ehlers-Danlos syndrome, vascular type	1.0 *
531	Miller-Dieker syndrome	1.0 BP*
396	Chronic hiccup	1.0 *
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	1.0 BP*
25	Glutaryl-CoA dehydrogenase deficiency	1.0 BP
177	Rhizomelic chondrodysplasia punctata	1.0 *
34515	Autosomal recessive limb-girdle muscular dystrophy type 2I	1.0 *
602	GNE myopathy	1.0
67043	Acanthamoeba keratitis	1.0 *
51577	Cobblestone lissencephaly	1.0 BP*
79435	Oculocutaneous albinism type 4	1.0
79258	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia	1.0 BP*
77259	Gaucher disease type 1	1.0 *
90060	Diffuse alveolar hemorrhage	1.0 *
98863	X-linked Emery-Dreifuss muscular dystrophy	1.0

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ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
98863	X-linked Emery-Dreifuss muscular dystrophy	1.0 BP
96263	48,XXX syndrome	1.0 BP*
95715	Congenital hypothyroidism due to transplacental passage of TSH-binding inhibitory antibodies	1.0 *
94068	Spondyloepiphyseal dysplasia congenita	1.0 BP*
93685	Localized Castleman disease	1.0
99789	Dentin dysplasia type I	1.0 *
163703	Febrile infection-related epilepsy syndrome	1.0 *
180242	Malignant tumor of fallopian tubes	1.0 *
264580	Glycogen storage disease due to liver phosphorylase kinase deficiency	1.0 BP*
141	Canavan disease	1.0 BP
577	Mucolipidosis type III	1.0 BP*
157	Carnitine palmitoyltransferase II deficiency	1.0 *
79087	Acquired partial lipodystrophy	1.0 *
746	Mitochondrial trifunctional protein deficiency	1.0 *
98306	Familial partial lipodystrophy	1.0 *
79086	Acquired generalized lipodystrophy	1.0 *
321	Multiple osteochondromas	1.0 *
444490	Familial chylomicronemia syndrome	1.0 *
87503	Mal de Meleda	1.0
90068	Cocaine intoxication	1.0 *
331206	Severe combined immunodeficiency due to complete RAG1/2 deficiency	1.0 *
206973	Congenital myotonia	1.0
86867	Nodal marginal zone B-cell lymphoma	1.0 *
3169	Sirenomelia	0.98 BP
79278	Autosomal erythropoietic protoporphyria	0.92 *
207	Crouzon disease	0.9 BP*
882	Tyrosinemia type 1	0.9 BP
48162	Lewis-Sumner syndrome	0.9 *
98249	Ehlers-Danlos syndrome	0.9 BP*
581	Mucopolysaccharidosis type 3	0.87 BP*
576	Mucolipidosis type II	0.84 BP*
99429	Complete androgen insensitivity syndrome	0.83
579	Mucopolysaccharidosis type 1	0.82 BP
365	Glycogen storage disease due to acid maltase deficiency	0.8 BP*
52	Alagille syndrome	0.8 BP*
2346	Angioosteohypertrophic syndrome	0.8 BP*
1461	Criss-cross heart	0.8 BP*
79361	Inherited epidermolysis bullosa	0.8 *
169793	Severe hemophilia B	0.8 *

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
3312	Thalidomide embryopathy	0.77
213	Cystinosis	0.75 BP
181	X-linked hypohidrotic ectodermal dysplasia	0.75 BP*
354	GM1 gangliosidosis	0.75 BP*
667	Autosomal recessive malignant osteopetrosis	0.75 BP*
1501	Adrenocortical carcinoma	0.75 *
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	0.75 BP*
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	0.75 BP*
487	Krabbe disease	0.7 BP
783	Rubinstein-Taybi syndrome	0.7 BP*
813	Silver-Russell syndrome	0.7 BP*
392	Holt-Oram syndrome	0.7 BP*
726	Alpers-Huttenlocher syndrome	0.7 BP*
110	Bardet-Biedl syndrome	0.7 *
303	Dystrophic epidermolysis bullosa	0.7
93473	Hurler syndrome	0.7 BP*
250	Frontonasal dysplasia	0.7 BP*
580	Mucopolysaccharidosis type 2	0.68 BP
796	Sandhoff disease	0.67 BP*
124	Blackfan-Diamond anemia	0.67 BP*
511	Maple syrup urine disease	0.67 BP
2591	Infantile myofibromatosis	0.67 BP*
3282	Multifocal atrial tachycardia	0.67 BP
1335	Pentalogy of Cantrell	0.67 BP
448270	Ectopia cordis	0.67 BP
90053	Hematopoietic stem cell transplantation	0.65 *
84	Fanconi anemia	0.62 BP*
294975	Congenital absence of upper arm and forearm with hand present	0.62 BP
994	Fetal aknesia deformation sequence	0.6 BP*
2345	Isolated Klippel-Feil syndrome	0.6 BP*
79168	Disorder of bile acid synthesis	0.6 *
79098	Sympathetic ophthalmia	0.6 *
98809	Paroxysmal kinesigenic dyskinesia	0.6
240103	Progressive supranuclear palsy-corticobasal syndrome	0.6 *
248111	Juvenile Huntington disease	0.6 *
169796	Moderately severe hemophilia B	0.6 *
169799	Mild hemophilia B	0.6 *
550	MELAS	0.6 *
54	X-linked recessive ocular albinism	0.58 BP*
275803	Pulmonary arterial hypertension associated with congenital heart disease	0.57 *
562	McCune-Albright syndrome	0.55 *
313	Lamellar ichthyosis	0.55 *

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ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
96264	49,XXXXY syndrome	0.55 BP*
79276	Acute intermittent porphyria	0.54 *
93929	Cloacal extrophy	0.54 BP
682	Hyperkalemic periodic paralysis	0.5 *
915	Aarskog-Scott syndrome	0.5 BP*
255	Dopa-responsive dystonia	0.5
3320	Thrombocytopenia-absent radius syndrome	0.5 BP*
611	Inclusion body myositis	0.5 *
634	Netherton syndrome	0.5 BP*
634	Netherton syndrome	0.5 *
902	Werner syndrome	0.5 *
528	Berardinelli-Seip congenital lipodystrophy	0.5 *
110	Bardet-Biedl syndrome	0.5 BP*
3427	Double outlet left ventricle	0.5 BP
811	Shwachman-Diamond syndrome	0.5 BP
747	Autoimmune pulmonary alveolar proteinosis	0.5
782	Axenfeld-Rieger syndrome	0.5 *
35909	Combined deficiency of factor V and factor VIII	0.5 *
122	Birt-Hogg-Dubé syndrome	0.5 *
64742	Pleuropulmonary blastoma	0.5 BP*
79242	Holocarboxylase synthetase deficiency	0.5 BP*
92050	Congenital tufting enteropathy	0.5 BP*
281097	Autosomal recessive congenital ichthyosis	0.5 *
93473	Hurler syndrome	0.5 *
201	Cowden syndrome	0.5 *
86854	Splenic marginal zone lymphoma	0.5 *
100	Ataxia-telangiectasia	0.49 *
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	0.47 *
379	Chronic granulomatous disease	0.46 BP
23	Argininosuccinic aciduria	0.46 BP
676	Hereditary chronic pancreatitis	0.43 *
2869	Peutz-Jeghers syndrome	0.4 *
1452	Cleidocranial dysplasia	0.4 BP*
1215	Autosomal dominant optic atrophy plus syndrome	0.4 *
2315	Johanson-Blizzard syndrome	0.4 BP*
3008	Pyruvate carboxylase deficiency	0.4 BP*
256	Early-onset generalized limb-onset dystonia	0.4 *
503	Larsen syndrome	0.4 BP*
42738	Severe congenital neutropenia	0.4 BP*
88	Idiopathic aplastic anemia	0.4 *
77293	Niemann-Pick disease type B	0.4 *

ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
217085	Mucopolysaccharidosis type 2, severe form	0.4 BP*
216804	Osteogenesis imperfecta type 2	0.4 BP*
631	Non-acquired isolated growth hormone deficiency	0.39
99885	Permanent neonatal diabetes mellitus	0.38 BP*
3440	Waardenburg syndrome	0.37 BP*
290	Congenital rubella syndrome	0.35 BP*
179	Birdshot chorioretinopathy	0.35
43393	Lambert-Eaton myasthenic syndrome	0.35
510	Lesch-Nyhan syndrome	0.34 BP*
96	Ataxia with vitamin E deficiency	0.33 *
565	Menkes disease	0.33 BP*
327	Congenital factor VII deficiency	0.33 *
140	Campomelic dysplasia	0.33 BP*
98850	Aggressive systemic mastocytosis	0.33 *
79473	Porphyria variegata	0.32 *
79269	Sanfilippo syndrome type A	0.32 *
147	Carbamoyl-phosphate synthetase 1 deficiency	0.31
261	Emery-Dreifuss muscular dystrophy	0.3 *
394	Classic homocystinuria	0.3 BP
628	Diastrophic dwarfism	0.3 BP*
258	Congenital muscular dystrophy type 1A	0.3 *
84	Fanconi anemia	0.3
581	Mucopolysaccharidosis type 3	0.3 *
1300	Autosomal dominant popliteal pterygium syndrome	0.3
3004	Mirror polydactyly-vertebral segmentation-limbs defects syndrome	0.3 *
633	Laron syndrome	0.3 *
2299	Aortic arch interruption	0.3 BP*
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	0.3 BP*
590	Congenital myasthenic syndrome	0.3 *
219	Autosomal recessive limb-girdle muscular dystrophy type 2F	0.3 *
56970	Human prion disease	0.3 *
79394	Congenital non-bullous ichthyosiform erythroderma	0.3 *
90647	Jervell and Lange-Nielsen syndrome	0.3
90079	Anthracycline extravasation	0.3 *
229717	Isolated agammaglobulinemia	0.3
182050	MYH9-related disease	0.3 *
294963	Popliteal pterygium syndrome	0.3 *
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	0.3
99886	Transient neonatal diabetes mellitus	0.3 BP*
845	Tay-Sachs disease	0.28 BP

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ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)	ORPHA Number	Disease or Group of diseases	Estimated prevalence (/100,000)
811	Shwachman-Diamond syndrome	0.28	583	Mucopolysaccharidosis type 6	0.16 BP*
579	Mucopolysaccharidosis type 1	0.25 *	583	Mucopolysaccharidosis type 6	0.16 *
702	Pelizaeus-Merzbacher disease	0.25 *	85	Congenital dyserythropoietic anemia	0.16 BP*
678	Papillon-Lefèvre syndrome	0.25	745	Severe hereditary thrombophilia due to congenital protein C deficiency	0.16 BP
35173	X-linked dominant chondrodysplasia punctata	0.25 BP*	335	Congenital fibrinogen deficiency	0.15 *
77292	Niemann-Pick disease type A	0.25 BP*	538	Lymphangiomyomatosis	0.15
93571	Dense deposit disease	0.25	223	Nephrogenic diabetes insipidus	0.15 *
275798	Pulmonary arterial hypertension associated with connective tissue disease	0.25 *	79430	Hermansky-Pudlak syndrome	0.15
910	Xeroderma pigmentosum	0.23 BP*	157850	Pantothenate kinase-associated neurodegeneration	0.15 *
324	Fabry disease	0.22 BP*	118	Beta-mannosidosis	0.14 BP*
47	X-linked agammaglobulinemia	0.22	763	Pycnodynatosis	0.13
111	Barth syndrome	0.22 *	3463	Wolfram syndrome	0.13
436	Hypophosphatasia	0.21 BP*	183669	Agammaglobulinemia	0.13 *
89936	X-linked hypophosphatemia	0.21 *	52427	Retinitis punctata albescens	0.125
534	Oculocerebral syndrome of Lowe	0.2	33364	Trichothiodystrophy	0.12 BP*
893	WAGR syndrome	0.2 BP	1308	C syndrome	0.11 *
385	Neurodegeneration with brain iron accumulation	0.2 *	34149	Autosomal dominant tubulointerstitial kidney disease	0.11 *
235	Dubowitz syndrome	0.2 BP*	61	Alpha-mannosidosis	0.1 *
191	Cockayne syndrome	0.2 BP*	367	Glycogen storage disease due to glycogen branching enzyme deficiency	0.1 BP
3006	Pyridoxine-dependent epilepsy	0.2 BP*	512	Metachromatic leukodystrophy	0.1 *
2052	Fraser syndrome	0.2 BP*	906	Wiskott-Aldrich syndrome	0.1 *
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	0.2 *	205	Crigler-Najjar syndrome	0.1 BP*
35	Propionic acidemia	0.2 *	773	Refsum disease	0.1 *
722	Hypoplasmminogenemia	0.2 *	1452	Cleidocranial dysplasia	0.1
353	Autosomal recessive limb-girdle muscular dystrophy type 2C	0.2 *	1775	Dyskeratosis congenita	0.1 *
79270	Sanfilippo syndrome type B	0.2 *	204	Sporadic Creutzfeldt-Jakob disease	0.1 *
98813	Hypohidrotic ectodermal dysplasia with immunodeficiency	0.2 BP*	1959	Evans syndrome	0.1 *
99870	Letterer-Siwe disease	0.2 *	326	Congenital factor V deficiency	0.1 *
238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	0.2	507	Leishmaniasis	0.1 *
178478	Infant botulism	0.2 BP*	3329	Tibial aplasia-ectrodactyly syndrome	0.1 *
209916	Extraskeletal myxoid chondrosarcoma	0.2 *	329	Congenital factor XI deficiency	0.1 *
596	X-linked centronuclear myopathy	0.2 *	2686	Cyclic neutropenia	0.1 *
79096	Pyridoxal phosphate-responsive seizures	0.2 *	298	Mitochondrial neurogastrointestinal encephalomyopathy	0.1 *
1361	Carnosinase deficiency	0.2 BP	225	Maternally-inherited diabetes and deafness	0.1 *
2635	Metatropic dysplasia	0.2 BP*	209	Cutis laxa	0.1 BP*
808	Seckel syndrome	0.2 BP*	31824	Colchicine poisoning	0.1 *
2614	Nail-patella syndrome	0.2 BP*	119	Autosomal recessive limb-girdle muscular dystrophy type 2E	0.1 *
1456	Atypical coarctation of aorta	0.17 BP*	142	Anaplastic thyroid carcinoma	0.1 *
407	Glycine encephalopathy	0.17 *	73274	Acquired hemophilia	0.1 *
79404	Junctional epidermolysis bullosa, generalized severe	0.17 BP	90793	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	0.1 *
280219	Pelizaeus-Merzbacher disease, classic form	0.17 *	93262	Crouzon syndrome-acanthosis nigricans syndrome	0.1 BP

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93322	Tibial hemimelia	0.1 BP*
86834	Juvenile myelomonocytic leukemia	0.1 *
98810	Paroxysmal non-kinesigenic dyskinesia	0.1
99842	Leukocyte adhesion deficiency type I	0.1 *
247257	Inhalational anthrax	0.1 *
209335	Autosomal dominant adult-onset proximal spinal muscular atrophy	0.1 *
289560	Mitochondrial membrane protein-associated neurodegeneration	0.1
391665	Homozygous familial hypercholesterolemia	0.1
32960	Tumor necrosis factor receptor 1 associated periodic syndrome	0.1 *
2485	Melorheostosis	0.09 *
48818	Aceruloplasminemia	0.09
189427	Cushing syndrome due to macronodular adrenal hyperplasia	0.08 *
275777	Heritable pulmonary arterial hypertension	0.08 *
726	Alpers-Huttenlocher syndrome	0.07 *
42738	Severe congenital neutropenia	0.07
217563	Neonatal acute respiratory distress due to SP-B deficiency	0.067 BP
337	Fibrodysplasia ossificans progressiva	0.05
2442	X-linked lymphoproliferative disease	0.05 *
2788	Osteoporosis-pseudoglioma syndrome	0.05 *
325	Congenital factor II deficiency	0.05 *
331	Congenital factor XIII deficiency	0.05 *
77261	Gaucher disease type 3	0.05 *
309294	Sialidosis	0.05 BP*
99718	Leber plus disease	0.04 *
189439	Primary pigmented nodular adrenocortical disease	0.04 *
69087	Naegeli-Franceschetti-Jadassohn syndrome	0.035 *
34520	Congenital muscular dystrophy with integrin alpha-7 deficiency	0.03 *
280210	Pelizaeus-Merzbacher disease, connatal form	0.03 *
280224	Pelizaeus-Merzbacher disease, transitional form	0.03 *
93976	Anotia	0.028 BP*
740	Hutchinson-Gilford progeria syndrome	0.025 BP
227	Diphallia	0.02 BP
584	Mucopolysaccharidosis type 7	0.01 *
3169	Sirenomelia	0.01
77260	Gaucher disease type 2	0.01 *
90308	Klippel-Trénaunay syndrome	0.007 *
740	Hutchinson-Gilford progeria syndrome	0.005
330009	Poliomyelitis in patients with immunodeficiencies deemed at risk	8.0E-4 *

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# List of diseases or groups of diseases by decreasing incidence

ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
99828	Dengue fever	714.0
3389	Tuberculosis	139.0
673	Malaria	73.0
65250	Perineural cyst	50.0*
558	Marfan syndrome	25.0*
507	Leishmaniasis	25.0
178320	Acute lung injury	25.0*
91546	Lyme disease	21.9
63443	Rare epithelial tumor of stomach	18.6*
171915	B-cell non-Hodgkin lymphoma	17.45*
98715	Uveitis	17.0*
813	Silver-Russell syndrome	15.5*
461	Recessive X-linked ichthyosis	15.0*
418	Congenital adrenal hyperplasia	13.35*
268316	Complication in hemodialysis	13.0*
56044	Carcinoma of gallbladder and extrahepatic biliary tract	12.0
547	Non-Hodgkin lymphoma	11.6*
1546	Cryptococcosis	11.0*
435	Ito hypomelanosis	10.85*
137839	Lemierre syndrome	10.0*
2209	Maternal phenylketonuria	10.0*
398934	Malignant epithelial tumor of ovary	9.39*
1489	Whooping cough	9.32*
3467	Hereditary xanthinuria	9.05*
217071	Renal cell carcinoma	8.35*
1941	Juvenile absence epilepsy	7.5*
70482	Carcinoma of esophagus	7.0
289390	Primary Sjögren syndrome	6.92
3002	Immune thrombocytopenic purpura	6.75*
448	Hemophilia	6.25*
29073	Multiple myeloma	6.0
213504	Adenocarcinoma of ovary	5.97*
182095	Interstitial lung disease	5.4*
182067	Glial tumor	5.35*
146	Differentiated thyroid carcinoma	5.25
171901	Primary cutaneous T-cell lymphoma	5.2*
99977	Squamous cell carcinoma of the esophagus	5.2
3099	Rheumatic fever	5.0*
1306	Buschke-Ollendorff syndrome	5.0
94	Astrocytoma	4.8*
3394	Soft tissue sarcoma	4.74*
494550	Squamous cell carcinoma of the larynx	4.61*

ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
213767	Squamous cell carcinoma of the cervix uteri	4.28*
70567	Cholangiocarcinoma	4.2
217074	Rare carcinoma of pancreas	3.9
2929	Juvenile polyposis syndrome	3.85*
2032	Idiopathic pulmonary fibrosis	3.81*
182130	Tumor of endocrine glands	3.75*
548	Leprosy	3.7
213528	Rare adenocarcinoma of the breast	3.55*
502363	Squamous cell carcinoma of the oral cavity	3.51*
70587	Infant acute respiratory distress syndrome	3.5*
100087	Thyroid tumor	3.2
363472	Tumor of testis and paratestis	3.15*
500478	Squamous cell carcinoma of the oropharynx	3.12*
100088	Thyroid carcinoma	3.1
88673	Hepatocellular carcinoma	3.09*
98274	Myeloproliferative neoplasm	3.07*
186	Primary biliary cholangitis	3.0
360	Glioblastoma	3.0
96061	Mosaic trisomy 8	3.0*
99745	Typhoid	3.0*
99429	Complete androgen insensitivity syndrome	3.0*
154	Familial isolated dilated cardiomyopathy	2.91*
544	Diffuse large B-cell lymphoma	2.79*
513	Acute lymphoblastic leukemia	2.75*
454821	Pleomorphic salivary gland adenoma	2.725
95716	Familial thyroid dys hormonogenesis	2.67
424991	Adenocarcinoma of the gallbladder and extrahepatic biliary tract	2.62*
70	Proximal spinal muscular atrophy	2.6*
877	Neuroendocrine neoplasm	2.53*
2038	Pulmonary arteriovenous malformation	2.5
519	Acute myeloid leukemia	2.5
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	2.5
98293	Hodgkin lymphoma	2.4*
391	Classic Hodgkin lymphoma	2.38*
707	Plague	2.2*
545	Follicular lymphoma	2.12*
352	Galactosemia	2.1*
79239	Classic galactosemia	2.1*
98375	Autoimmune hemolytic anemia	2.02*
1866	Focal, segmental or multifocal dystonia	2.0*
319276	Clear cell renal carcinoma	1.99*
1549	Cryptosporidiosis	1.96*
729	Polycythemia vera	1.9*
50251	Pleural mesothelioma	1.9*
102	Multiple system atrophy	1.8
842	Testicular seminomatous germ cell tumor	1.71*

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ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
355	Gaucher disease	1.7*
810	Shigellosis	1.68*
139417	Acute transverse myelitis	1.6
92	Juvenile idiopathic arthritis	1.5*
35	Propionic acidemia	1.5
618	Familial melanoma	1.5*
26106	Hereditary diffuse gastric cancer	1.5*
52688	Myelodysplastic syndrome	1.5*
2103	Guillain-Barré syndrome	1.45
801	Scleroderma	1.41
803	Amyotrophic lateral sclerosis	1.35
250923	Isolated aniridia	1.31*
549	Legionellosis	1.3*
98843	Classic Hodgkin lymphoma, nodular sclerosis type	1.28*
494547	Squamous cell carcinoma of the hypopharynx	1.27*
635	Neuroblastoma	1.26
521	Chronic myeloid leukemia	1.25*
83418	Proximal spinal muscular atrophy type 2	1.23*
363494	Non-seminomatous germ cell tumor of testis	1.21*
2137	Autoimmune hepatitis	1.2
313920	Epstein-Barr virus-associated gastric carcinoma	1.2
83419	Proximal spinal muscular atrophy type 3	1.1*
398043	Malignant tumor of penis	1.075*
91349	Non-functioning pituitary adenoma	1.05
502366	Squamous cell carcinoma of the lip	1.02
213772	Adenocarcinoma of the cervix uteri	1.01*
848	Beta-thalassemia	1.0
710	Pfeiffer syndrome	1.0*
727	Microscopic polyangiitis	1.0*
3148	Malignant peripheral nerve sheath tumor	1.0
400	Cystic echinococcosis	1.0*
2781	Osteopetrosis and related disorders	1.0*
44890	Gastrointestinal stromal tumor	1.0
824	Primary myelofibrosis	1.0*
69078	Liposarcoma	1.0*
54595	Craniopharyngioma	1.0
157798	Hyperplastic polyposis syndrome	1.0
209964	Solitary rectal ulcer syndrome	1.0*
171918	T-cell non-Hodgkin lymphoma	0.99*
2023	Undifferentiated pleomorphic sarcoma	0.9*
2467	Systemic mastocytosis	0.9*
900	Granulomatosis with polyangiitis	0.85*
398961	Mucinous adenocarcinoma of ovary	0.85*
33226	Waldenström macroglobulinemia	0.81*
454723	Endometrioid carcinoma of ovary	0.81*
223727	Bone sarcoma	0.8*

ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
930	Idiopathic achalasia	0.77
542	Primary cutaneous lymphoma	0.75*
340	Hemorrhagic fever-renal syndrome	0.74*
48104	Pyoderma gangrenosum	0.74
424019	Squamous cell carcinoma of the anal canal	0.73*
276145	Malignant epithelial tumor of salivary glands	0.73*
100070	Progressive non-fluent aphasia	0.7*
99976	Adenocarcinoma of the esophagus	0.7
683	Progressive supranuclear palsy	0.65
171	Primary sclerosing cholangitis	0.65
79139	Japanese encephalitis	0.65*
83597	Acute disseminated encephalomyelitis	0.6*
101330	Porphyria cutanea tarda	0.6*
780	Rhabdomyosarcoma	0.59*
178566	Mycosis fungoides and variants	0.59*
732	Polymyositis	0.585*
398058	Squamous cell carcinoma of the penis	0.57*
104075	Adenocarcinoma of the small intestine	0.57*
221	Dermatomyositis	0.55*
99892	ACTH-dependent Cushing syndrome	0.55
589	Myasthenia gravis	0.53
86830	Chronic myeloproliferative disease, unclassifiable	0.53*
99971	Well-differentiated liposarcoma	0.51*
180275	Paget disease of the nipple	0.51*
191	Cockayne syndrome	0.5*
2584	Classic mycosis fungoides	0.5*
980	Absence of the pulmonary artery	0.5*
39044	Uveal melanoma	0.5*
167714	Unclassified acute myeloid leukemia	0.49*
3318	Essential thrombocythemia	0.48*
398940	Malignant non-epithelial tumor of ovary	0.43*
98844	Classic Hodgkin lymphoma, mixed cellularity type	0.42*
820	Sneddon syndrome	0.4*
86872	T-cell large granular lymphocyte leukemia	0.4*
101959	Chronic primary adrenal insufficiency	0.4*
399	Huntington disease	0.38
83484	St. Louis encephalitis	0.38*
36426	Stevens-Johnson syndrome	0.36*
150	Nasopharyngeal carcinoma	0.36*
963	Acromegaly	0.35
728	Relapsing polychondritis	0.35
46484	Oligodendroglial tumor	0.35*
49041	IgG4-related retroperitoneal fibrosis	0.35*
329217	Cerebral sinovenous thrombosis	0.35*
500464	Squamous cell carcinoma of the nasal cavity and paranasal sinuses	0.35
33276	Kaposi sarcoma	0.34*

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ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
533	Listeriosis	0.337
1070	Anisakiasis	0.32
83420	Proximal spinal muscular atrophy type 4	0.32*
398971	Clear cell adenocarcinoma of the ovary	0.32*
1930	Herpes simplex virus encephalitis	0.3
29072	Hereditary pheochromocytoma-paraganglioma	0.3
873	Desmoid tumor	0.3*
52417	MALT lymphoma	0.3*
178478	Infant botulism	0.3*
293173	Acute generalized exanthematous pustulosis	0.3
300912	Marginal zone lymphoma	0.3*
93672	Juvenile dermatomyositis	0.295
58017	Classic hairy cell leukemia	0.29*
98275	Myelodysplastic/myeloproliferative disease	0.29*
98823	Chronic myelomonocytic leukemia	0.29*
79140	Cutaneous neuroendocrine carcinoma	0.27
99970	Dedifferentiated liposarcoma	0.27*
83330	Proximal spinal muscular atrophy type 1	0.26*
168999	Malignant melanoma of the mucosa	0.26*
424016	Adenocarcinoma of the anal canal	0.26*
97279	Insulinoma	0.25
139423	Idiopathic acute transverse myelitis	0.25*
329977	Classic neuroendocrine tumor of appendix	0.25
251627	Oligodendrogloma	0.25*
55880	Chondrosarcoma	0.24*
668	Osteosarcoma	0.23*
1332	Medullary thyroid carcinoma	0.22*
251852	Embryonal tumor of neuroepithelial tissue	0.22*
97253	Neuroendocrine tumor of pancreas	0.21*
424943	Adenocarcinoma of the liver and intrahepatic biliary tract	0.21*
301	Ependymal tumor	0.2*
96253	Cushing disease	0.2*
100085	Primary hepatic neuroendocrine carcinoma	0.2
95455	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	0.19
541	Primary cutaneous CD30+ T-cell lymphoproliferative disease	0.18*
183	Eosinophilic granulomatosis with polyangiitis	0.18*
543	Burkitt lymphoma	0.17*
3398	Thymic epithelial neoplasm	0.17*
142	Anaplastic thyroid carcinoma	0.17*
423786	Undifferentiated carcinoma of stomach	0.17*
517	Acute myelomonocytic leukemia	0.17*
284	Alveolar echinococcosis	0.16*
251636	Ependymoma	0.16*
781	Q fever	0.16*
204	Sporadic Creutzfeldt-Jakob disease	0.15

ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
2495	Meningioma	0.15*
553	Cushing syndrome	0.15*
56970	Human prion disease	0.15*
33402	Pediatric hepatocellular carcinoma	0.15*
329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis	0.15*
86839	Refractory anemia with excess blasts	0.15*
654	Nephroblastoma	0.14*
99867	Thymoma	0.14*
3392	Tularemia	0.14*
319298	Papillary renal cell carcinoma	0.14*
319	Ewing sarcoma	0.13*
514	Acute monoblastic leukemia	0.13*
35808	Malignant sex cord stromal tumor of ovary	0.13*
182114	Rare urogenital tumor	0.13*
418959	Squamous cell carcinoma of the stomach	0.13*
363579	Extragonadal germ cell tumor	0.13*
913	Zollinger-Ellison syndrome	0.125
509	Leptospirosis	0.12*
2086	Optic pathway glioma	0.12
86893	Nodular lymphocyte predominant Hodgkin lymphoma	0.12
213716	Squamous cell carcinoma of the corpus uteri	0.12*
99915	Malignant granulosa cell tumor of the ovary	0.12*
213512	Malignant mixed Müllerian tumor of the ovary	0.12*
616	Medulloblastoma	0.11*
520	Acute promyelocytic leukemia	0.11*
251651	Oligoastrocytic tumor	0.11*
98277	Acute myeloid leukemia with recurrent genetic anomaly	0.11*
112	Bartter syndrome	0.1*
2314	Autosomal dominant hyper-IgE syndrome	0.1*
2382	Lennox-Gastaut syndrome	0.1*
37553	Andersen-Tawil syndrome	0.1*
26790	Pseudomyxoma peritonei	0.1
53035	Caroli disease	0.1
99967	Myxoid/round cell liposarcoma	0.1*
98919	Miller-Fisher syndrome	0.1*
228371	Foodborne botulism	0.1*
178475	Wound botulism	0.1*
1304	Brucellosis	0.1*
1822	Dysplasia epiphysealis hemimelica	0.1
98845	Classic Hodgkin lymphoma, lymphocyte-rich type	0.1*
251630	Anaplastic oligodendrogloma	0.09*
75564	Acquired idiopathic sideroblastic anemia	0.09*
3287	Takayasu arteritis	0.084*
375	Anti-glomerular basement membrane disease	0.08*

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ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
35807	Malignant germ cell tumor of ovary	0.08*
73274	Acquired hemophilia	0.08
324625	Chikungunya	0.08*
424002	Squamous cell carcinoma of the rectum	0.07*
418951	Undifferentiated carcinoma of esophagus	0.07*
251870	Central nervous system primitive neuroectodermal tumor	0.07*
398987	Malignant teratoma of ovary	0.07*
79277	Congenital erythropoietic porphyria	0.065*
863	Trichinellosis	0.06*
213531	Metaplastic carcinoma of the breast	0.06*
86843	Acute panmyelosis with myelofibrosis	0.06*
790	Retinoblastoma	0.05*
99969	Pleomorphic liposarcoma	0.05*
213557	Salivary gland type cancer of the breast	0.05*
98287	Histiocytic and dendritic cell tumor	0.05*
86852	B-cell prolymphocytic leukemia	0.05*
99931	Idiopathic pulmonary hemosiderosis	0.0425*
331	Congenital factor XIII deficiency	0.04*
248111	Juvenile Huntington disease	0.04*
300385	Pituitary carcinoma	0.04*
747	Autoimmune pulmonary alveolar proteinosis	0.04
454714	Plasma cell leukemia	0.04*
424996	Squamous cell carcinoma of gallbladder and extrahepatic biliary tract	0.04*
98846	Classic Hodgkin lymphoma, lymphocyte-depleted type	0.04*
99912	Malignant dysgerminomatous germ cell tumor of the ovary	0.04*
168960	Refractory anemia with excess blasts in transformation	0.04*
357034	Non-hereditary retinoblastoma	0.038*
83476	West-Nile encephalitis	0.036*
2573	Moyamoya disease	0.035*
290	Congenital rubella syndrome	0.03*
1501	Adrenocortical carcinoma	0.03*
33355	Reticular dysgenesis	0.03*
46487	Acquired epidermolysis bullosa	0.03*
99865	Spermatocytic seminoma	0.03*
424039	Squamous cell carcinoma of pancreas	0.03*
251576	Gliosarcoma	0.03*
3299	Tetanus	0.026*
329984	Goblet cell carcinoma	0.025
1267	Botulism	0.022*
449	Hepatoblastoma	0.02*
1957	Esthesioneuroblastoma	0.02*
1183	Opsoclonus-myoclonus syndrome	0.02*
143	Parathyroid carcinoma	0.02*
99928	Placental site trophoblastic tumor	0.02*

ORPHA Number	Disease or Group of diseases	Estimated incidence (/100,000)
251909	Pineoblastoma	0.02*
251679	Astroblastoma	0.02*
363489	Sex cord-stromal tumor of testis	0.02*
424970	Undifferentiated carcinoma of liver and intrahepatic biliary tract	0.02*
424046	Acinar cell carcinoma of pancreas	0.02*
423994	Squamous cell carcinoma of the colon	0.02*
86850	Myeloid sarcoma	0.02*
518	Acute megakaryoblastic leukemia	0.02*
98834	Acute myeloblastic leukemia with maturation	0.02*
251579	Giant cell glioblastoma	0.02*
31837	Pulmonary venoocclusive disease	0.015*
538	Lymphangioleiomyomatosis	0.0135
79276	Acute intermittent porphyria	0.013*
79278	Autosomal erythropoietic protoporphyrina	0.012*
2030	Fibrosarcoma	0.01*
55881	Adamantinoma	0.01*
251899	Choroid plexus carcinoma	0.01*
173	Cholera	0.01*
424975	Squamous cell carcinoma of liver and intrahepatic biliary tract	0.01*
424053	Mucinous cystadenocarcinoma of the pancreas	0.01*
423968	Squamous cell carcinoma of the small intestine	0.01*
423781	Carcinoma of stomach, salivary gland type	0.01*
418945	Carcinoma of esophagus, salivary gland type	0.01*
168966	Composite lymphoma	0.01*
98833	Acute myeloblastic leukemia without maturation	0.01*
98832	Acute myeloid leukemia with minimal differentiation	0.01*
251863	Desmoplastic/nodular medulloblastoma	0.01*
251607	Pleomorphic xanthoastrocytoma	0.01*
251598	Protoplasmic astrocytoma	0.01*
251582	Gliomatosis cerebri	0.01*
180234	Mixed germ cell tumor	0.01*
180226	Embryonal carcinoma	0.01*
319303	Chromophobe renal cell carcinoma	0.01*
79473	Porphyria variegata	0.008*
284343	Pleuropulmonary blastoma familial tumor susceptibility syndrome	0.007
356	Gerstmann-Straussler-Scheinker syndrome	0.0055
97280	Glucagonoma	0.005*
97283	Somatostatinoma	0.0025*

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# List of diseases or groups of diseases by decreasing number of published cases or families

## Number of published cases

ORPHA Number	Disease or Group of diseases	Number of cases
319218	Ebola hemorrhagic fever	28220 Cases
227972	Toxic oil syndrome	20000 Cases
454745	Kuru	2700 Cases
50918	Kikuchi-Fujimoto disease	1052 Cases
2309	Pachyonychia congenita	1000 Cases
158014	Rosai-Dorfman disease	1000 Cases
64	Alström syndrome	950 Cases
454836	Avian influenza	826 Cases
1917	Fetal methylmercury syndrome	800 Cases
83312	Rickettsialpox	800 Cases
85	Congenital dyserythropoietic anemia	740 Cases
99825	Nipah virus disease	556 Cases
167	Chédiak-Higashi syndrome	500 Cases
26	Methylmalonic acidemia with homocystinuria	500 Cases
2138	46,XX ovotesticular disorder of sex development	500 Cases
2930	Cronkhite-Canada syndrome	500 Cases
2896	Pitt-Hopkins syndrome	500 Cases
3261	Autoimmune lymphoproliferative syndrome	500 Cases
34526	Familial primary hypomagnesemia	500 Cases
42642	PFAPA syndrome	500 Cases
35687	Erdheim-Chester disease	500 Cases
530	Lipoid proteinosis	500 Cases
69077	Rhabdoid tumor	500 Cases
73256	Central neurocytoma	500 Cases
79282	Methylmalonic acidemia with homocystinuria, type cbIC	500 Cases
99826	Marburg hemorrhagic fever	500 Cases
85448	AGel amyloidosis	475 Cases
22	Succinic semialdehyde dehydrogenase deficiency	450 Cases
79312	Vitamin B12-unresponsive methylmalonic acidemia type mut-	450 Cases
411593	Insulin autoimmune syndrome	404 Cases
649	Norrie disease	400 Cases
125	Bloom syndrome	400 Cases
2909	Rothmund-Thomson syndrome	400 Cases
662	Yellow nail syndrome	400 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
35125	Epidermal nevus syndrome	400 Cases
100025	Alpha-heavy chain disease	400 Cases
3348	Tracheobronchopathia osteochondroplastica	400 Cases
352540	Oncogenic osteomalacia	400 Cases
974	Adams-Oliver syndrome	398 Cases
238606	Primary orthostatic tremor	390 Cases
83453	Vulvovaginal gingival syndrome	380 Cases
2968	Leukocyte adhesion deficiency	350 Cases
64741	Pulmonary blastoma	350 Cases
85458	Hereditary cerebral hemorrhage with amyloidosis	350 Cases
3269	Congenital radioulnar synostosis	350 Cases
96170	Emanuel syndrome	350 Cases
59	Allan-Herndon-Dudley syndrome	320 Cases
838	Susac syndrome	304 Cases
1556	Cutis marmorata telangiectatica congenita	300 Cases
570	Moebius syndrome	300 Cases
3071	Costello syndrome	300 Cases
157	Carnitine palmitoyltransferase II deficiency	300 Cases
184	Cherubism	300 Cases
1340	Cardiofaciocutaneous syndrome	300 Cases
1328	Camurati-Engelmann disease	300 Cases
2092	Focal dermal hypoplasia	300 Cases
1467	Cogan syndrome	300 Cases
2330	Kasabach-Merritt syndrome	300 Cases
3347	Mounier-Kuhn syndrome	300 Cases
42775	PHACE syndrome	300 Cases
41	Dyschromatosis symmetrica hereditaria	300 Cases
482	Kimura disease	300 Cases
525	Lichen planopilaris	300 Cases
840	Syringocystadenoma papilliferum	300 Cases
73	Gorham-Stout disease	300 Cases
83469	Desmoplastic small round cell tumor	300 Cases
99147	Acquired von Willebrand syndrome	300 Cases
228302	Carnitine palmitoyl transferase II deficiency, myopathic form	300 Cases
247245	Superficial siderosis	300 Cases
206569	Immune-mediated necrotizing myopathy	300 Cases
237	Duplication of urethra	300 Cases
501	Lafora disease	300 Cases
309025	Mevalonate kinase deficiency	300 Cases
500	Noonan syndrome with multiple lentigines	296 Cases
363549	Acute encephalopathy with biphasic seizures and late reduced diffusion	283 Cases
2070	Eosinophilic gastroenteritis	280 Cases
137667	Capillary malformation-arteriovenous malformation	261 Cases
2908	Kindler syndrome	250 Cases
79087	Acquired partial lipodystrophy	250 Cases
100006	ABeta amyloidosis, Dutch type	250 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
98954	Meesmann corneal dystrophy	250 Cases
163634	Maffucci syndrome	250 Cases
167635	Scleromyxedema	250 Cases
373	Simpson-Golabi-Behmel syndrome	250 Cases
90283	Lupus erythematosus tumidus	250 Cases
221074	Marchiafava-Bignami disease	250 Cases
199318	15q13.3 microdeletion syndrome	246 Cases
2710	Oculodentodigital dysplasia	243 Cases
464453	Acquired methemoglobinemia	242 Cases
169105	Good syndrome	241 Cases
99642	Spondyloepimetaphyseal dysplasia, Handigodu type	234 Cases
2241	Megacystis-microcolon-intestinal hypoperistalsis syndrome	230 Cases
1708	Mosaic trisomy 16	226 Cases
65285	Lhermitte-Duclos disease	220 Cases
587	Muir-Torre syndrome	205 Cases
2796	Pachydermoperiostosis	204 Cases
2510	Micro syndrome	203 Cases
33364	Trichothiodystrophy	201 Cases
193	Cohen syndrome	200 Cases
1059	Blue rubber bleb nevus	200 Cases
3165	Eosinophilic fasciitis	200 Cases
847	Alpha-thalassemia-X-linked intellectual disability syndrome	200 Cases
1986	Gollop-Wolfgang complex	200 Cases
1980	Bilateral striopallidodentate calcinosis	200 Cases
2616	3M syndrome	200 Cases
575	Muckle-Wells syndrome	200 Cases
559	Marinesco-Sjögren syndrome	200 Cases
343	Hyperimmunoglobulinemia D with periodic fever	200 Cases
414	Gyrate atrophy of choroid and retina	200 Cases
1451	CINCA syndrome	200 Cases
317	Erythrokeratoderma variabilis	200 Cases
302	Epidermodysplasia verruciformis	200 Cases
2088	Glycogen storage disease due to GLUT2 deficiency	200 Cases
1063	Tufted angioma	200 Cases
220	Denys-Drash syndrome	200 Cases
36412	Hypocomplementemic urticarial vasculitis	200 Cases
523	Hereditary leiomyomatosis and renal cell cancer	200 Cases
901	Wells syndrome	200 Cases
679	Malignant atrophic papulosis	200 Cases
66630	Congenital pseudoarthrosis of the clavicle	200 Cases
48377	Subcorneal pustular dermatosis	200 Cases
48686	Primary effusion lymphoma	200 Cases
48652	Monosomy 22q13	200 Cases
79277	Congenital erythropoietic porphyria	200 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
79255	GM1 gangliosidosis type 1	200 Cases
75563	X-linked sideroblastic anemia	200 Cases
91385	Acquired angioedema	200 Cases
97360	Robinow syndrome	200 Cases
97231	Ligneous conjunctivitis	200 Cases
139436	Multicentric reticulohistiocytosis	200 Cases
139491	Hemochromatosis type 4	200 Cases
137867	Madras motor neuron disease	200 Cases
99050	Pulmonary artery coming from the aorta	200 Cases
220407	Limited systemic sclerosis	200 Cases
221016	Rothmund-Thomson syndrome type 2	200 Cases
199267	Infantile digital fibromatosis	200 Cases
293848	Frontotemporal dementia, right temporal atrophy variant	200 Cases
306516	Familial primary hypomagnesemia with hypercalcioruria and nephrocalcinosis	200 Cases
261183	15q11.2 microdeletion syndrome	200 Cases
1540	Jackson-Weiss syndrome	200 Cases
457	Harlequin ichthyosis	200 Cases
627	Nance-Horan syndrome	196 Cases
402035	Eosinophilic colitis	196 Cases
28	Vitamin B12-responsive methylmalonic acidemia	192 Cases
1465	Coffin-Siris syndrome	190 Cases
293381	Epithelial recurrent erosion dystrophy	186 Cases
1475	Renal coloboma syndrome	180 Cases
254509	Iatrogenic botulism	180 Cases
319552	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	180 Cases
2237	Hypoparathyroidism-sensorineural deafness-renal disease syndrome	180 Cases
572	Immunodeficiency by defective expression of HLA class 2	179 Cases
98960	Thiel-Behnke corneal dystrophy	173 Cases
1459	Celiac disease-epilepsy-cerebral calcification syndrome	170 Cases
48431	Congenital cataracts-facial dysmorphism-neuropathy syndrome	170 Cases
60040	Megalencephaly-capillary malformation-polymicrogyria syndrome	170 Cases
97685	17q11 microdeletion syndrome	170 Cases
252212	Malignant triton tumor	170 Cases
324636	Autoerythrocyte sensitization syndrome	170 Cases
96121	7q11.23 microduplication syndrome	163 Cases
261243	16p13.11 microduplication syndrome	162 Cases
1522	Craniometaphyseal dysplasia	160 Cases
1359	Carney complex	160 Cases
300324	Persistent polyclonal B-cell lymphocytosis	154 Cases
226	Dihydropteridine reductase deficiency	150 Cases
3467	Hereditary xanthinuria	150 Cases
381	Griselli syndrome	150 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
3103	Roberts syndrome	150 Cases
1297	Branchio-oculo-facial syndrome	150 Cases
2048	Foix-Chavany-Marie syndrome	150 Cases
2108	Hallermann-Streiff syndrome	150 Cases
2576	MULIBREY nanism	150 Cases
3197	Hereditary hyperekplexia	150 Cases
3265	Humero-radial synostosis	150 Cases
2637	Microcephalic osteodysplastic primordial dwarfism type II	150 Cases
35069	Infantile neuroaxonal dystrophy	150 Cases
28378	Tyrosinemia type 2	150 Cases
37042	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	150 Cases
37748	Schnitzler syndrome	150 Cases
188	Systemic capillary leak syndrome	150 Cases
71274	Disseminated peritoneal leiomyomatosis	150 Cases
52503	X-linked creatine transporter deficiency	150 Cases
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	150 Cases
84142	Isaac syndrome	150 Cases
93682	Pediatric Castleman disease	150 Cases
139411	Carney triad	150 Cases
168816	Peritoneal cystic mesothelioma	150 Cases
314777	Familial isolated pituitary adenoma	150 Cases
284454	Acute zonal occult outer retinopathy	150 Cases
3156	Senior-Loken syndrome	150 Cases
236	Trisomy 9p	150 Cases
498474	Hyaline fibromatosis syndrome	150 Cases
1590	Distal monosomy 13q	150 Cases
135	CACH syndrome	148 Cases
398166	Focal facial dermal dysplasia	147 Cases
166113	Bazex syndrome	145 Cases
457083	Isolated splenogonadal fusion	145 Cases
113	Bazex-Dupré-Christol syndrome	143 Cases
83450	Regional odontodysplasia	140 Cases
79314	L-2-hydroxyglutaric aciduria	140 Cases
90003	IgG4-related hepatopathy	140 Cases
2290	Microvillus inclusion disease	137 Cases
291	Congenital varicella syndrome	130 Cases
3400	Aorto-ventricular tunnel	130 Cases
178307	Reticulate acropigmentation of Kitamura	130 Cases
834	Free sialic acid storage disease	130 Cases
800	Schwartz-Jampel syndrome	129 Cases
98920	Spinal muscular atrophy with respiratory distress type 1	128 Cases
137898	Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	127 Cases
650	LCAT deficiency	125 Cases
1305	Feingold syndrome	123 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
2343	Isolated cloverleaf skull syndrome	120 Cases
90117	Hereditary motor and sensory neuropathy, Okinawa type	120 Cases
100026	Gamma-heavy chain disease	120 Cases
391641	Feingold syndrome type 1	120 Cases
440727	Combined hamartoma of the retina and retinal pigment epithelium	120 Cases
261272	17q12 microduplication syndrome	118 Cases
3138	Ulnar-mammary syndrome	117 Cases
398073	Prader-Willi-like syndrome	117 Cases
1001	2q37 microdeletion syndrome	115 Cases
48918	Focal myositis	115 Cases
98967	Schnyder corneal dystrophy	115 Cases
293181	Malignant migrating partial seizures of infancy	114 Cases
261494	Kleefstra syndrome	114 Cases
420584	Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome	112 Cases
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	111 Cases
31043	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement	110 Cases
756	Pseudohypoaldosteronism type 1	107 Cases
79113	Mandibulofacial dysostosis-microcephaly syndrome	107 Cases
86909	Myoclonic epilepsy of infancy	106 Cases
88637	Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome	105 Cases
221150	Pitt-Hopkins-like syndrome	105 Cases
261265	17q12 microdeletion syndrome	103 Cases
3342	Arterial tortuosity syndrome	102 Cases
79477	Griselli syndrome type 2	102 Cases
488239	Acute macular neuroretinopathy	101 Cases
349	Fucosidosis	100 Cases
371	Glycogen storage disease due to muscle phosphofructokinase deficiency	100 Cases
45	Adenosine monophosphate deaminase deficiency	100 Cases
833	Encephalopathy due to sulfite oxidase deficiency	100 Cases
245	Nager syndrome	100 Cases
2053	Freeman-Sheldon syndrome	100 Cases
2785	Osteopetrosis with renal tubular acidosis	100 Cases
2414	Congenital pulmonary lymphangiectasia	100 Cases
477	KID syndrome	100 Cases
2882	Sitosterolemia	100 Cases
274	Bernard-Soulier syndrome	100 Cases
897	Waardenburg-Shah syndrome	100 Cases
955	Acroosteolysis dominant type	100 Cases
869	Triple A syndrome	100 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
709	Peters plus syndrome	100 Cases
981	Internal carotid agenesis	100 Cases
1507	Autosomal recessive Robinow syndrome	100 Cases
672	Pallister-Hall syndrome	100 Cases
2222	Hypertrichosis lanuginosa congenita	100 Cases
1826	Frontometaphyseal dysplasia	100 Cases
2780	Osteopathia striata-cranial sclerosis syndrome	100 Cases
2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	100 Cases
2704	Ochoa syndrome	100 Cases
2363	Lacrimoauriculodentodigital syndrome	100 Cases
2342	Haim-Munk syndrome	100 Cases
2332	KBG syndrome	100 Cases
3107	Autosomal dominant Robinow syndrome	100 Cases
746	Mitochondrial trifunctional protein deficiency	100 Cases
332	Congenital intrinsic factor deficiency	100 Cases
3243	Sweet syndrome	100 Cases
3319	Congenital amegakaryocytic thrombocytopenia	100 Cases
1929	Rasmussen subacute encephalitis	100 Cases
2478	Megalencephalic leukoencephalopathy with subcortical cysts	100 Cases
33110	Autosomal agammaglobulinemia	100 Cases
31150	Tangier disease	100 Cases
30924	Primary hypomagnesemia with secondary hypocalcemia	100 Cases
35708	Aromatic L-amino acid decarboxylase deficiency	100 Cases
898	Wagner disease	100 Cases
724	Idiopathic acute eosinophilic pneumonia	100 Cases
71517	Rapid-onset dystonia-parkinsonism	100 Cases
70593	Immunodeficiency due to selective anti-polysaccharide antibody deficiency	100 Cases
59306	McLeod neuroacanthocytosis syndrome	100 Cases
59315	Rhombencephalosynapsis	100 Cases
79403	Junctional epidermolysis bullosa-pyloric atresia syndrome	100 Cases
79409	Recessive dystrophic epidermolysis bullosa inversa	100 Cases
77258	Trichorhinophalangeal syndrome type 1 and 3	100 Cases
75326	Retinal arterial tortuosity	100 Cases
91136	Acquired monoclonal Ig light chain-associated Fanconi syndrome	100 Cases
89937	Autosomal dominant hypophosphatemic rickets	100 Cases
86813	Helicoid peripapillary chorioretinal degeneration	100 Cases
94087	Cytophagic histiocytic panniculitis	100 Cases
93686	Multicentric Castleman disease	100 Cases
99015	Spastic paraparesis type 2	100 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
221008	Rothmund-Thomson syndrome type 1	100 Cases
238769	1q44 microdeletion syndrome	100 Cases
254478	Lichen planus pemphigoides	100 Cases
251295	Pigmented paravenous retinochoroidal atrophy	100 Cases
140944	CLOVES syndrome	100 Cases
140957	Autosomal dominant macrothrombocytopenia	100 Cases
168569	H syndrome	100 Cases
209905	Brain-lung-thyroid syndrome	100 Cases
199241	Pulmonary capillary hemangiomatosis	100 Cases
306741	Hemidystonia-hemiatrophy syndrome	100 Cases
1293	Brachyolmia	100 Cases
1310	Caffey disease	100 Cases
1221	Cheilitis glandularis	100 Cases
351	Galactosialidosis	100 Cases
352723	Attenuated Chédiak-Higashi syndrome	100 Cases
99880	Hyperparathyroidism-jaw tumor syndrome	100 Cases
65748	Multiple self-healing squamous epithelioma	100 Cases
502	Trichorhinophalangeal syndrome type 2	100 Cases
79493	Brooke-Spiegler syndrome	100 Cases
261476	Xp21 microdeletion syndrome	100 Cases
1446	Ring chromosome 22 syndrome	100 Cases
166305	Benign infantile seizures associated with mild gastroenteritis	100 Cases
3344	Weismann-Netter syndrome	100 Cases
96095	3q26 microduplication syndrome	100 Cases
99063	Shone complex	100 Cases
199282	Harlequin syndrome	100 Cases
329211	Autosomal dominant neovascular inflammatory vitreoretinopathy	99 Cases
71276	Silent sinus syndrome	98 Cases
2065	Galloway-Mowat syndrome	97 Cases
75381	Cystoid macular dystrophy	97 Cases
333	Farber disease	96 Cases
699	Pearson syndrome	95 Cases
2671	Neu-Laxova syndrome	91 Cases
52368	Mohr-Tranebjærg syndrome	91 Cases
1885	Isolated ectopia lentis	90 Cases
742	Prolidase deficiency	90 Cases
157846	Neuroferritinopathy	90 Cases
53719	Wyburn-Mason syndrome	90 Cases
2473	McKusick-Kaufman syndrome	90 Cases
498228	Phyllodes tumor of the prostate	90 Cases
1642	Distal monosomy 9p	89 Cases
347	Frasier syndrome	88 Cases
2044	Floating-Harbor syndrome	87 Cases
96147	Kleefstra syndrome due to 9q34 microdeletion	86 Cases
1738	Trisomy 4p	85 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
3403	Uhl anomaly	84 Cases
34587	Glycogen storage disease due to LAMP-2 deficiency	84 Cases
254519	Kagami-Ogata syndrome	84 Cases
2635	Metatropic dysplasia	81 Cases
79133	Focal facial dermal dysplasia type I	81 Cases
98961	Reis-Bücklers corneal dystrophy	81 Cases
1935	Early myoclonic encephalopathy	80 Cases
526	Liddle syndrome	80 Cases
382	Guanidinoacetate methyltransferase deficiency	80 Cases
1440	Ring chromosome 14 syndrome	80 Cases
49	Penile agenesis	80 Cases
51188	Ethylmalonic encephalopathy	80 Cases
49827	Thiamine-responsive megaloblastic anemia syndrome	80 Cases
79315	D-2-hydroxyglutaric aciduria	80 Cases
97229	Riboflavin transporter deficiency	80 Cases
238569	Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome	80 Cases
231401	Alpha-thalassemia-myelodysplastic syndrome	80 Cases
950	Acrodyostosis	80 Cases
3152	Sclerosteosis	80 Cases
98769	Spinocerebellar ataxia type 15/16	80 Cases
2396	Encephalocranioscutaneous lipomatosis	77 Cases
1393	Cerebrocostomandibular syndrome	75 Cases
238722	Familial congenital mirror movements	75 Cases
209981	IRIDA syndrome	75 Cases
320406	Spastic paraparesis-optic atrophy-neuropathy syndrome	75 Cases
79230	Hemochromatosis type 2	74 Cases
622	Homocystinuria without methylmalonic aciduria	73 Cases
659	Mutilating palmoplantar keratoderma with periorificial keratotic plaques	73 Cases
2196	Familial primary hypomagnesemia with hypercalcuria and nephrocalcinosis with severe ocular involvement	72 Cases
1830	Schimke immuno-osseous dysplasia	71 Cases
1442	Ring chromosome 18 syndrome	70 Cases
760	Purine nucleoside phosphorylase deficiency	70 Cases
32	Glutathione synthetase deficiency	70 Cases
2123	Diffuse neonatal hemangiomatosis	70 Cases
2006	Median cleft lip/mandible	70 Cases
2028	Juvenile hyaline fibromatosis	70 Cases
2484	Melnick-Needles syndrome	70 Cases
65759	Carpenter syndrome	70 Cases
79293	Familial LCAT deficiency	70 Cases
79257	GM1 gangliosidosis type 3	70 Cases
404546	DITRA	70 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
357043	Amyotrophic lateral sclerosis type 4	70 Cases
3310	Tetrasomy 9p	70 Cases
90280	Chilblain lupus	70 Cases
90791	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	68 Cases
98975	Congenital hereditary endothelial dystrophy type I	68 Cases
319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	68 Cases
329284	Beta-propeller protein-associated neurodegeneration	68 Cases
306669	Hemiparkinsonism-hemiatrophy syndrome	68 Cases
2554	Ear-patella-short stature syndrome	67 Cases
2062	Progressive non-infectious anterior vertebral fusion	67 Cases
160148	Cap polyposis	67 Cases
352328	MEGDEL syndrome	67 Cases
2268	ICF syndrome	66 Cases
2333	Kenny-Caffey syndrome	65 Cases
51636	WHIM syndrome	65 Cases
90354	Brittle cornea syndrome	65 Cases
163	Hereditary hyperferritinemia-cataract syndrome	64 Cases
3242	Renpenning syndrome	64 Cases
55595	Autosomal dominant limb-girdle muscular dystrophy type 1F	64 Cases
96184	Temple syndrome due to maternal uniparental disomy of chromosome 14	64 Cases
69736	Bilateral acute depigmentation of the iris	62 Cases
83473	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	62 Cases
75392	Ehlers-Danlos syndrome, periodontitis type	62 Cases
1988	Femoral-facial syndrome	62 Cases
2855	Perrault syndrome	61 Cases
3051	Intellectual disability-sparse hair-brachydactyly syndrome	61 Cases
2771	Bruck syndrome	60 Cases
239	Dyggve-Melchior-Claussen disease	60 Cases
677	Pancreatoblastoma	60 Cases
969	Acromicric dysplasia	60 Cases
156	Carnitine palmitoyl transferase 1A deficiency	60 Cases
1270	Bowen-Conradi syndrome	60 Cases
1667	Wolcott-Rallison syndrome	60 Cases
1515	Cranioectodermal dysplasia	60 Cases
139	CHILD syndrome	60 Cases
2462	Shprintzen-Goldberg syndrome	60 Cases
708	Peters anomaly	60 Cases
159	Carnitine-acylcarnitine translocase deficiency	60 Cases
2995	Baraitser-Winter cerebrofrontofacial syndrome	60 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
3411	Double uterus-hemivagina-renal agenesis syndrome	60 Cases
2221	Acquired hypertrichosis lanuginosa	60 Cases
721	Gray platelet syndrome	60 Cases
52530	Pseudo-von Willebrand disease	60 Cases
63455	Paraneoplastic pemphigus	60 Cases
79310	Vitamin B12-responsive methylmalonic aciduria type cblA	60 Cases
90349	Autosomal recessive cutis laxa type 1	60 Cases
98870	Congenital dyserythropoietic anemia type III	60 Cases
99803	Haddad syndrome	60 Cases
158029	Sea-blue histiocytosis	60 Cases
300493	Sagliker syndrome	60 Cases
773	Refsum disease	60 Cases
363454	BICD2-related autosomal dominant childhood-onset proximal spinal muscular atrophy	60 Cases
352490	Autism spectrum disorder due to AUTS2 deficiency	60 Cases
83467	Morvan syndrome	60 Cases
468635	Cryptogenic multifocal ulcerous stenosing enteritis	60 Cases
451607	Cutaneous pseudolymphoma	60 Cases
3338	Toriello-Carey syndrome	59 Cases
293642	Blepharophimosis-intellectual disability syndrome	58 Cases
57196	Medial condensing osteitis of the clavicle	58 Cases
79327	ALG1-CDG	57 Cases
88644	Autosomal recessive ataxia, Beauce type	57 Cases
331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	57 Cases
46	Adenylosuccinate lyase deficiency	56 Cases
90024	Deafness with labyrinthine aplasia, microtia, and microdontia	56 Cases
3206	Stüve-Wiedemann syndrome	56 Cases
71	Chylomicron retention disease	55 Cases
2588	Myhre syndrome	55 Cases
2556	Microphthalmia with linear skin defects syndrome	55 Cases
3405	Umbilical cord ulceration-intestinal atresia syndrome	55 Cases
3455	Wiedemann-Rautenstrauch syndrome	54 Cases
57782	Mazabraud syndrome	54 Cases
79320	ALG6-CDG	54 Cases
83628	LUMBAR syndrome	54 Cases
314603	Autosomal recessive spastic ataxia with leukoencephalopathy	54 Cases
2833	Stiff skin syndrome	54 Cases
464306	DYRK1A-related intellectual disability syndrome	54 Cases
79099	Interstitial granulomatous dermatitis with arthritis	53 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
254516	Temple syndrome	53 Cases
251515	Distal arthrogryposis type 10	53 Cases
178509	Perry syndrome	53 Cases
398088	Hereditary cryohydrocytosis with normal stomatin	53 Cases
98806	Primary dystonia, DYT6 type	53 Cases
251282	Autosomal dominant spastic ataxia type 1	53 Cases
3473	Zimmermann-Laband syndrome	52 Cases
251671	Angiocentric glioma	52 Cases
449566	Eosinophilic angiocentric fibrosis	52 Cases
98767	Spinocerebellar ataxia type 11	51 Cases
1766	Dysequilibrium syndrome	51 Cases
521414	Autosomal dominant Charcot-Marie-Tooth disease type 2DD	51 Cases
585	Multiple sulfatase deficiency	50 Cases
868	Triose phosphate-isomerase deficiency	50 Cases
9	Tetrasomy X	50 Cases
2801	Juvenile Paget disease	50 Cases
3111	Rotor syndrome	50 Cases
574	Monosomy 21	50 Cases
808	Seckel syndrome	50 Cases
871	Familial progressive cardiac conduction defect	50 Cases
1444	Ring chromosome 20 syndrome	50 Cases
40	Acromesomelic dysplasia, Maroteaux type	50 Cases
1253	Ascher syndrome	50 Cases
127	Borjeson-Forssman-Lehmann syndrome	50 Cases
978	ADULT syndrome	50 Cases
1125	Ocular motor apraxia, Cogan type	50 Cases
1118	Fibular aplasia-ectrodactyly syndrome	50 Cases
1573	Hypotrichosis with juvenile macular degeneration	50 Cases
1517	Hypertrichotic osteochondrodysplasia, Cantu type	50 Cases
1493	Vici syndrome	50 Cases
1425	Desbuquois syndrome	50 Cases
165	Neutral lipid storage disease	50 Cases
2078	Geroderma osteodysplastica	50 Cases
2143	Donnai-Barrow syndrome	50 Cases
2136	Hennekam syndrome	50 Cases
1997	Blepharo-cheilo-odontic syndrome	50 Cases
2632	Langer mesomelic dysplasia	50 Cases
2461	Marden-Walker syndrome	50 Cases
2407	LOC syndrome	50 Cases
2805	Partial pancreatic agenesis	50 Cases
712	Hemolytic anemia due to glucophosphate isomerase deficiency	50 Cases
851	Paris-Trousseau thrombocytopenia	50 Cases
3231	Deafness-onychodystrophy syndrome	50 Cases
3253	Zlotogora-Ogur syndrome	50 Cases
1902	Ehrlichiosis	50 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
33111	Granulomatous slack skin	50 Cases
29822	Spontaneous periodic hypothermia	50 Cases
71518	Benign paroxysmal torticollis of infancy	50 Cases
53540	Goldmann-Favre syndrome	50 Cases
79500	DOORS syndrome	50 Cases
79143	Isolated congenital anonychia	50 Cases
79256	GM1 gangliosidosis type 2	50 Cases
75382	Oguchi disease	50 Cases
91496	Snowflake vitreoretinal degeneration	50 Cases
90342	Xeroderma pigmentosum variant	50 Cases
90348	Autosomal dominant cutis laxa	50 Cases
86816	Congenital analbuminemia	50 Cases
85136	Cystic leukoencephalopathy without megalecephaly	50 Cases
85212	Fetal Gaucher disease	50 Cases
98811	Paroxysmal exertion-induced dyskinesia	50 Cases
97234	Glycogen storage disease due to phosphoglycerate mutase deficiency	50 Cases
93600	Primary hyperoxaluria type 3	50 Cases
137888	Auriculocondylar syndrome	50 Cases
101150	Autosomal recessive dopa-responsive dystonia	50 Cases
100012	Lissencephaly with cerebellar hypoplasia type B	50 Cases
99872	Hashimoto-Pritzker syndrome	50 Cases
217385	17p13.3 microduplication syndrome	50 Cases
221046	Poikiloderma with neutropenia	50 Cases
171929	Trisomy 10p	50 Cases
208513	Spinocerebellar ataxia type 29	50 Cases
206583	Adult polyglucosan body disease	50 Cases
300512	Onychomatricoma	50 Cases
284448	CLIPPERS	50 Cases
371428	Multicentric osteolysis-nodulosis-arthropathy spectrum	50 Cases
404507	Chondromyxoid fibroma	50 Cases
352636	Phalangeal microgeodic syndrome	50 Cases
3130	Satoyoshi syndrome	50 Cases
454710	Anti-p200 pemphigoid	50 Cases
79147	Familial reactive perforating collagenosis	50 Cases
79395	Keratoderma hereditarium mutilans with ichthyosis	50 Cases
262941	Partial duplication of the long arm of chromosome 14	50 Cases
494	Keratoderma hereditarium mutilans	50 Cases
443197	X-linked erythropoietic protoporphyria	50 Cases
99731	Isolated sulfite oxidase deficiency	50 Cases
96177	Ring chromosome 15 syndrome	50 Cases
99776	Mosaic trisomy 9	50 Cases
1873	Jalili syndrome	49 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	49 Cases
54251	Corticosteroid-sensitive aseptic abscess syndrome	49 Cases
319558	Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency	49 Cases
255229	Navajo neurohepatopathy	49 Cases
3447	Weaver syndrome	48 Cases
2897	Pityriasis rubra pilaris	48 Cases
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	48 Cases
391372	Intellectual disability-severe speech delay-mild dysmorphism syndrome	48 Cases
404553	Vasculitis due to ADA2 deficiency	48 Cases
1414	Cholestasis-lymphedema syndrome	47 Cases
989	Hypoglossia-hypodactyly syndrome	47 Cases
1509	Coxopodopatellar syndrome	47 Cases
85162	Facial onset sensory and motor neuronopathy	47 Cases
216828	Osteogenesis imperfecta type 5	47 Cases
250994	1q21.1 microduplication syndrome	46 Cases
798	Schinzel-Giedion syndrome	46 Cases
319646	PGM1-CDG	46 Cases
53721	Cobb syndrome	45 Cases
86788	X-linked severe congenital neutropenia	45 Cases
99749	Kostmann syndrome	45 Cases
166286	Porokeratotic eccrine ostial and dermal duct nevus	45 Cases
209932	Cone dystrophy with supernormal rod response	45 Cases
279947	Postorgasmic illness syndrome	45 Cases
284984	Aneurysm-osteoarthritis syndrome	45 Cases
254875	Mitochondrial DNA depletion syndrome, myopathic form	45 Cases
84064	Syndromic diarrhea	44 Cases
168606	Seborrhea-like dermatitis with psoriasiform elements	44 Cases
99938	Autosomal dominant Charcot-Marie-Tooth disease type 2D	44 Cases
221126	Fowler syndrome	44 Cases
2470	Matthew-Wood syndrome	43 Cases
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency	43 Cases
2301	Congenital short bowel syndrome	43 Cases
77301	Monosomy 9q22.3	42 Cases
352629	16q24.1 microdeletion syndrome	42 Cases
1621	3q13 microdeletion syndrome	42 Cases
1052	Mosaic variegated aneuploidy syndrome	41 Cases
254351	Distal 7q11.23 microdeletion syndrome	41 Cases
398156	Oculoauriculofrontonasal syndrome	41 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
1369	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome	40 Cases
2971	Peroxisomal acyl-CoA oxidase deficiency	40 Cases
1745	Distal trisomy 6p	40 Cases
1742	Trisomy 5p	40 Cases
1699	Trisomy 12p	40 Cases
1225	Baller-Gerold syndrome	40 Cases
1023	Congenital generalized hypertrichosis, Ambras type	40 Cases
1923	Methimazole embryofetopathy	40 Cases
859	Transcobalamin deficiency	40 Cases
2273	Ichthyosis follicularis-alopexia-photophobia syndrome	40 Cases
2457	Mandibuloacral dysplasia	40 Cases
1832	Lethal osteosclerotic bone dysplasia	40 Cases
2962	De Barsy syndrome	40 Cases
24	Fumaric aciduria	40 Cases
257	Epidermolysis bullosa simplex with muscular dystrophy	40 Cases
1810	Autosomal dominant hypohidrotic ectodermal dysplasia	40 Cases
52022	Potocki-Shaffer syndrome	40 Cases
79134	DEND syndrome	40 Cases
90350	Autosomal recessive cutis laxa type 2	40 Cases
96102	Distal trisomy 10q	40 Cases
96148	Distal monosomy 10q	40 Cases
95159	Hepatoerythropoietic porphyria	40 Cases
99844	Leukocyte adhesion deficiency type III	40 Cases
210548	Macrocephaly-intellectual disability-autism syndrome	40 Cases
228384	5q14.3 microdeletion syndrome	40 Cases
163746	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	40 Cases
140966	Palmoplantar keratoderma, Nagashima type	40 Cases
210122	Congenital alveolar capillary dysplasia	40 Cases
183678	Hermansky-Pudlak syndrome with neutropenia	40 Cases
314422	Ameloblastic carcinoma	40 Cases
263534	Acral peeling skin syndrome	40 Cases
280785	Bullous diffuse cutaneous mastocytosis	40 Cases
280651	Acrodysostosis with multiple hormone resistance	40 Cases
281190	Congenital reticular ichthyosiform erythroderma	40 Cases
411777	Generalized eruptive keratoacanthoma	40 Cases
438117	Steel syndrome	40 Cases
324977	Proteasome disability syndrome	40 Cases
79	Congenital alpha2-antiplasmin deficiency	40 Cases
90652	Otopalatodigital syndrome type 2	40 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
217008	Bockenheimer syndrome	40 Cases
317473	Pancytopenia due to IKZF1 mutations	39 Cases
458758	Composite hemangioendothelioma	39 Cases
36	Acrocallosal syndrome	38 Cases
1647	Oculocerebrocutaneous syndrome	38 Cases
2067	GAPO syndrome	38 Cases
91	Aromatase deficiency	38 Cases
69085	Limb-mammary syndrome	38 Cases
55654	Hypotrichosis simplex	38 Cases
163696	Action myoclonus-renal failure syndrome	38 Cases
171629	Autosomal recessive spastic paraparesis type 35	38 Cases
209867	Autosomal dominant rhegmatogenous retinal detachment	38 Cases
314621	Duplication of the pituitary gland	38 Cases
99852	Ravine syndrome	38 Cases
457260	X-linked intellectual disability-hypotonia-movement disorder syndrome	38 Cases
485350	CLCN4-related X-linked intellectual disability syndrome	38 Cases
1993	Pai syndrome	37 Cases
3208	Isolated succinate-CoQ reductase deficiency	37 Cases
79406	Late-onset junctional epidermolysis bullosa	37 Cases
391417	HSD10 disease	37 Cases
100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	37 Cases
493342	Vibratory urticaria	37 Cases
494428	Idiopathic pleuroparenchymal fibroelastosis	37 Cases
96334	Kagami-Ogata syndrome due to paternal uniparental disomy of chromosome 14	37 Cases
1532	Gómez-López-Hernández syndrome	36 Cases
1855	Spondyloenchondroplasia	36 Cases
98955	Lisch epithelial corneal dystrophy	36 Cases
98908	Neutral lipid storage myopathy	36 Cases
168583	Hereditary North American Indian childhood cirrhosis	36 Cases
300573	Polymicrogyria due to TUBB2B mutation	36 Cases
101000	Autosomal recessive spastic paraparesis type 20	36 Cases
166308	Benign infantile focal epilepsy with midline spikes and waves during sleep	36 Cases
446	Neonatal hemochromatosis	35 Cases
2040	Congenital bronchobiliary fistula	35 Cases
1437	Ring chromosome 1 syndrome	35 Cases
970	Hereditary sensory and autonomic neuropathy type 2	35 Cases
98773	Spinocerebellar ataxia type 21	35 Cases
96125	Distal monosomy 6p	35 Cases
100024	Mu-heavy chain disease	35 Cases
293621	X-linked endothelial corneal dystrophy	35 Cases
2777	Osteomesopyknosis	35 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
443073	Charcot-Marie-Tooth disease type 2S	35 Cases
464311	Intellectual disability syndrome due to a DYRK1A point mutation	35 Cases
100045	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C	35 Cases
101001	Autosomal recessive spastic paraplegia type 21	35 Cases
3416	Hyperostosis corticalis generalisata	35 Cases
83	Antley-Bixler syndrome	34 Cases
943	Malonic aciduria	34 Cases
2953	Ehlers-Danlos syndrome, musculocontractural type	34 Cases
69126	Pyogenic arthritis-pyoderma gangrenosum-acne syndrome	34 Cases
2874	Phakomatosis pigmentokeratotica	34 Cases
363528	Intellectual disability-strabismus syndrome	34 Cases
391677	Short stature-optic atrophy-Pelger-Hüet anomaly syndrome	34 Cases
398097	Neonatal antiphospholipid syndrome	34 Cases
93269	Short rib-polydactyly syndrome, Majewski type	34 Cases
353284	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency	34 Cases
1620	Distal monosomy 3p	34 Cases
2406	Locked-in syndrome	33 Cases
1388	Catel-Manzke syndrome	33 Cases
2783	Autosomal dominant osteopetrosis type 1	33 Cases
561	Marshall-Smith syndrome	33 Cases
3102	Richieri Costa-Pereira syndrome	33 Cases
2170	Methylcobalamin deficiency type cbIG	33 Cases
832	Succinyl-CoA:3-ketoacid CoA transferase deficiency	33 Cases
3322	Hoyer-Hreidarsson syndrome	33 Cases
2795	Polycystic ovaries-urethral sphincter dysfunction syndrome	33 Cases
123	Björnstad syndrome	33 Cases
225123	Hemochromatosis type 3	33 Cases
329457	Distal arthrogryposis type 5D	33 Cases
447977	Progressive scapulohumeroperoneal distal myopathy	33 Cases
1681	Diprosopus	33 Cases
488280	14q32 duplication syndrome	33 Cases
3163	SHORT syndrome	32 Cases
35664	ALDH18A1-related De Barsy syndrome	32 Cases
67039	Segmental odontomaxillary dysplasia	32 Cases
141096	Supernumerary nostril	32 Cases
293843	3MC syndrome	32 Cases
314373	Chronic diarrhea due to guanylate cyclase 2C overactivity	32 Cases
324535	Combined oxidative phosphorylation defect type 11	32 Cases
412217	Dystonia-aphonia syndrome	32 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
458763	Retiform hemangioendothelioma	32 Cases
139485	Autosomal recessive ataxia due to ubiquinone deficiency	31 Cases
231573	Congenital erosive and vesicular dermatosis	31 Cases
431255	Scapuloperoneal spinal muscular atrophy	31 Cases
96173	Ring chromosome 9 syndrome	31 Cases
99898	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency	31 Cases
1711	Mosaic trisomy 17	31 Cases
1747	Mosaic trisomy 7	31 Cases
246	Postaxial acrofacial dysostosis	30 Cases
29	Mevalonic aciduria	30 Cases
2746	Opsismodysplasia	30 Cases
715	Glycogen storage disease due to muscle phosphorylase kinase deficiency	30 Cases
1752	Trisomy 8q	30 Cases
957	Acropectorovertebral dysplasia	30 Cases
2615	Nakajo-Nishimura syndrome	30 Cases
1229	Congenital intrauterine infection-like syndrome	30 Cases
1314	Symmetrical thalamic calcifications	30 Cases
1662	Restrictive dermopathy	30 Cases
1545	Crisponi syndrome	30 Cases
1525	Cranio-osteopathia	30 Cases
1427	Otospondylomegaepiphyseal dysplasia	30 Cases
2036	Scalp-ear-nipple syndrome	30 Cases
2763	Osteocraniostenosis	30 Cases
2733	Omodyplasia	30 Cases
2721	Odonto-onycho-dermal dysplasia	30 Cases
2728	Blepharophimosis-intellectual disability syndrome, Ohdo type	30 Cases
2636	Microcephalic osteodysplastic primordial dwarfism types I and III	30 Cases
2399	Nasopalpebral lipoma-coloboma syndrome	30 Cases
3005	Pyle disease	30 Cases
2834	Wrinkly skin syndrome	30 Cases
3266	Humero-radio-ulnar synostosis	30 Cases
3258	Cenani-Lenz syndrome	30 Cases
3352	Tricho-dento-osseous syndrome	30 Cases
3464	Woodhouse-Sakati syndrome	30 Cases
2849	Perlman syndrome	30 Cases
35705	Neurometabolic disorder due to serine deficiency	30 Cases
66628	Obesity due to congenital leptin deficiency	30 Cases
79411	Transient bullous dermolysis of the newborn	30 Cases
79456	Diffuse cutaneous mastocytosis	30 Cases
79292	Fish-eye disease	30 Cases
79155	Hydroxykynureninuria	30 Cases
79157	2-methylbutyryl-CoA dehydrogenase deficiency	30 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
77298	Anophthalmia/microphthalmia-esophageal atresia syndrome	30 Cases
91481	Ring dermoid of cornea	30 Cases
93315	Spondylometaphyseal dysplasia, 'corner fracture' type	30 Cases
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type	30 Cases
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	30 Cases
90045	Hereditary folate malabsorption	30 Cases
85164	Camptodactyly-tall stature-scoliosis-hearing loss syndrome	30 Cases
85277	X-linked intellectual disability, Cantagrel type	30 Cases
85278	Christianson syndrome	30 Cases
85202	Keutel syndrome	30 Cases
98764	Spinocerebellar ataxia type 27	30 Cases
97297	Bohring-Opitz syndrome	30 Cases
94065	15q24 microdeletion syndrome	30 Cases
93940	Laryngotracheoesophageal cleft type 3	30 Cases
140933	Linear atrophoderma of Moulin	30 Cases
137834	Frank-Ter Haar syndrome	30 Cases
99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K	30 Cases
98970	Fleck corneal dystrophy	30 Cases
220295	Xeroderma pigmentosum-Cockayne syndrome complex	30 Cases
228415	5q35 microduplication syndrome	30 Cases
228116	Hughes-Stovin syndrome	30 Cases
228236	Linear focal elastosis	30 Cases
238446	15q11q13 microduplication syndrome	30 Cases
141163	Glossopalatine ankylosis	30 Cases
209370	Severe neonatal-onset encephalopathy with microcephaly	30 Cases
209943	IRVAN syndrome	30 Cases
294049	Reunion Island Larsen-like syndrome	30 Cases
325004	CANDLE syndrome	30 Cases
275523	Dianzani autoimmune lymphoproliferative disease	30 Cases
397709	Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	30 Cases
2063	Splenogonadal fusion-limb defects-micrognathia syndrome	30 Cases
91396	Isolated cryptophthalmia	30 Cases
477650	Fibroblastic rheumatism	30 Cases
139552	Distal hereditary motor neuropathy, Jerash type	30 Cases
458768	Primary intralymphatic angioendothelioma	30 Cases
34592	Immunodeficiency by defective expression of HLA class 1	30 Cases
497623	C12ORF65-related combined oxidative phosphorylation defect	30 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
178345	Aromatase excess syndrome	30 Cases
1596	Distal monosomy 15q	30 Cases
1186	Infantile onset spinocerebellar ataxia	29 Cases
2753	Orofaciodigital syndrome type 4	29 Cases
3255	Filippi syndrome	29 Cases
2460	Van den Ende-Gupta syndrome	29 Cases
139444	Leukoencephalopathy with bilateral anterior temporal lobe cysts	29 Cases
466775	Autosomal recessive Charcot-Marie-Tooth disease type 2X	29 Cases
500150	Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	29 Cases
2220	Hypertrichosis cubiti	28 Cases
3459	Wilson-Turner syndrome	28 Cases
34528	Autosomal dominant primary hypomagnesemia with hypocalciuria	28 Cases
50814	Craniolenticulosutural dysplasia	28 Cases
79124	Hepatic veno-occlusive disease-immunodeficiency syndrome	28 Cases
85173	IMAGe syndrome	28 Cases
99812	LIG4 syndrome	28 Cases
228174	Autosomal dominant Charcot-Marie-Tooth disease type 2N	28 Cases
314022	Gastric adenocarcinoma and proximal polyposis of the stomach	28 Cases
329466	Autosomal dominant focal dystonia, DYT25 type	28 Cases
398069	Prader-Willi syndrome due to a point mutation	28 Cases
352577	Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome	28 Cases
391392	Familial episodic pain syndrome with predominantly lower limb involvement	28 Cases
457077	TAFRO syndrome	28 Cases
139547	Distal spinal muscular atrophy type 3	28 Cases
521258	Xq25 microduplication syndrome	28 Cases
466	Fatal familial insomnia	27 Cases
966	Hypertrichosis-acromegaloid facial appearance syndrome	27 Cases
1040	Metaphyseal adysplasia	27 Cases
2169	Methylcobalamin deficiency type cbIE	27 Cases
1955	Spinocerebellar ataxia type 34	27 Cases
2701	Noonan syndrome-like disorder with loose anagen hair	27 Cases
2623	Geleophysic dysplasia	27 Cases
100993	Autosomal dominant spastic paraparesis type 12	27 Cases
251287	Benign concentric annular macular dystrophy	27 Cases
313808	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia	27 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement	27 Cases
261250	16q24.3 microdeletion syndrome	27 Cases
280133	Complement component 3 deficiency	27 Cases
93358	Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome	27 Cases
96078	16p13.3 microduplication syndrome	27 Cases
319635	Amyloidosis cutis dyschromia	27 Cases
1262	Böök syndrome	26 Cases
1974	Autosomal recessive faciodigitogenital syndrome	26 Cases
40366	Acitretin/etretinate embryopathy	26 Cases
52994	Orbital leiomyoma	26 Cases
98771	Spinocerebellar ataxia type 18	26 Cases
199343	EAST syndrome	26 Cases
357332	Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome	26 Cases
1032	Hyperdibasic aminoaciduria type 1	26 Cases
2574	Moynahan syndrome	26 Cases
488613	Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome	26 Cases
7	3C syndrome	25 Cases
1519	Hypertelorism, Teebi type	25 Cases
2201	Palmoplantar keratoderma-spastic paralysis syndrome	25 Cases
2499	Metachondromatosis	25 Cases
3472	Yunis-Varon syndrome	25 Cases
39041	Omenn syndrome	25 Cases
50944	Schöpf-Schulz-Passarge syndrome	25 Cases
54028	Plummer-Vinson syndrome	25 Cases
56305	Atelosteogenesis type III	25 Cases
56304	Atelosteogenesis type II	25 Cases
79319	MPI-CDG	25 Cases
93109	Congenital megacalycosis	25 Cases
85203	Acropectoral syndrome	25 Cases
251019	2q32q33 microdeletion syndrome	25 Cases
314597	Chudley-McCullough syndrome	25 Cases
268249	Mycophenolate mofetil embryopathy	25 Cases
281122	Self-improving collodion baby	25 Cases
397941	MAN1B1-CDG	25 Cases
1715	Trisomy 18p	25 Cases
458803	Spinocerebellar ataxia type 42	25 Cases
488632	TBCK-related intellectual disability syndrome	25 Cases
1448	Ring chromosome 6 syndrome	25 Cases
527497	NKX6-2-related autosomal recessive hypomyelinating leukodystrophy	25 Cases
1234	Bartsocas-Papas syndrome	24 Cases
1490	Corneal dystrophy-perceptive deafness syndrome	24 Cases
1361	Carnosinase deficiency	24 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
3275	Spondylocarpotarsal synostosis	24 Cases
98972	Central cloudy dystrophy of François	24 Cases
247262	Hyperphosphatasia-intellectual disability syndrome	24 Cases
251383	CK syndrome	24 Cases
171607	X-linked spastic paraparesis type 34	24 Cases
209902	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	24 Cases
183713	Pyogenic bacterial infections due to MyD88 deficiency	24 Cases
300525	Pseudohypoaldosteronism type 2D	24 Cases
314404	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	24 Cases
313846	Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome	24 Cases
399096	Distal anoctaminopathy	24 Cases
438216	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation	24 Cases
480864	Recurrent metabolic encephalopathy crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome	24 Cases
2069	Gastrocutaneous syndrome	24 Cases
438213	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	24 Cases
487809	Pediatric collagenous gastritis	24 Cases
488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W	24 Cases
965	Acromegaloid facial appearance syndrome	23 Cases
1617	2q24 microdeletion syndrome	23 Cases
93329	Autosomal recessive omodysplasia	23 Cases
101028	Transaldolase deficiency	23 Cases
99901	Acyl-CoA dehydrogenase 9 deficiency	23 Cases
238475	Familial hypercholanemia	23 Cases
157973	Congenital muscular dystrophy due to LMNA mutation	23 Cases
314588	Distal tetrasomy 15q	23 Cases
261652	Kleefstra syndrome due to a point mutation	23 Cases
411493	Pontocerebellar hypoplasia type 10	23 Cases
445018	Combined immunodeficiency due to LRBA deficiency	23 Cases
477817	PMP22-RAI1 contiguous gene duplication syndrome	23 Cases
364198	Bipartite talus	23 Cases
71271	Split hand-split foot-deafness syndrome	22 Cases
79499	Autosomal dominant deafness-onychodystrophy syndrome	22 Cases
91387	Familial thoracic aortic aneurysm and aortic dissection	22 Cases
85191	Singleton-Merten dysplasia	22 Cases
85201	Genitopatellar syndrome	22 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
94063	12q14 microdeletion syndrome	22 Cases
93953	Familial thyroglossal duct cyst	22 Cases
228423	Monocytopenia with susceptibility to infections	22 Cases
247820	Ectodermal dysplasia-syndactyly syndrome	22 Cases
163690	Hypotonia-cystinuria syndrome	22 Cases
209908	Childhood apraxia of speech	22 Cases
314718	Lethal arteriopathy syndrome due to fibulin-4 deficiency	22 Cases
329195	Developmental delay with autism spectrum disorder and gait instability	22 Cases
269229	Pontine tegmental cap dysplasia	22 Cases
363677	Childhood-onset autosomal recessive myopathy with external ophthalmoplegia	22 Cases
398173	Focal facial dermal dysplasia type II	22 Cases
1827	Acromelic frontonasal dysplasia	22 Cases
98805	Primary dystonia, DYT4 type	22 Cases
448372	X-linked acrogigantism due to Xq26 microduplication	22 Cases
431272	X-linked scapuloperoneal muscular dystrophy	22 Cases
445038	3-methylglutaconic aciduria type 7	22 Cases
168612	Congenital deficiency in alpha-fetoprotein	22 Cases
2492	FATCO syndrome	22 Cases
466943	WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	22 Cases
496641	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	22 Cases
1723	Mosaic trisomy 2	22 Cases
528105	Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome	22 Cases
3063	X-linked intellectual disability, Snyder type	21 Cases
1578	Pterin-4 alpha-carbinolamine dehydratase deficiency	21 Cases
230	Dopamine beta-hydroxylase deficiency	21 Cases
69082	Odonto-tricho-ungual-digit-palmar syndrome	21 Cases
66625	Cerebrooculonatal syndrome	21 Cases
79091	Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome	21 Cases
93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	21 Cases
93606	Nephrogenic syndrome of inappropriate antidiuresis	21 Cases
217330	REN-related autosomal dominant tubulointerstitial kidney disease	21 Cases
221145	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies	21 Cases
168593	Sudden infant death-dysgenesis of the testes syndrome	21 Cases
171881	Cap myopathy	21 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
199326	Isolated autosomal dominant hypomagnesemia, Glaudemans type	21 Cases
402003	Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering	21 Cases
401869	Multiple mitochondrial dysfunctions syndrome type 1	21 Cases
363649	Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome	21 Cases
391389	Familial episodic pain syndrome with predominantly upper body involvement	21 Cases
398189	Focal facial dermal dysplasia type IV	21 Cases
447964	Autosomal dominant Charcot-Marie-Tooth disease type 2V	21 Cases
482077	HTRA1-related autosomal dominant cerebral small vessel disease	21 Cases
466768	Autosomal dominant Charcot-Marie-Tooth disease type 2Z	21 Cases
324972	MAGIC syndrome	21 Cases
468672	Colobomatous macrophtalmia-microcornea syndrome	21 Cases
30	Hereditary orotic aciduria	20 Cases
3137	Alpha-N-acetylglucosaminidase deficiency	20 Cases
1358	Carey-Fineman-Ziter syndrome	20 Cases
971	Acrorenal syndrome	20 Cases
1807	Focal facial dermal dysplasia type III	20 Cases
1513	Craniodiaphyseal dysplasia	20 Cases
1394	Cerebrofaciothoracic dysplasia	20 Cases
1387	Cataract-intellectual disability-hypogonadism syndrome	20 Cases
1466	COFS syndrome	20 Cases
455	Superficial epidermolytic ichthyosis	20 Cases
2021	Fibrochondrogenesis	20 Cases
2755	Orofaciodigital syndrome type 8	20 Cases
2751	Orofaciodigital syndrome type 2	20 Cases
2375	Laryngeal abductor paralysis-intellectual disability syndrome	20 Cases
3021	RAPADILINO syndrome	20 Cases
3047	Blepharophimosis-intellectual disability syndrome, SBBYS type	20 Cases
2847	Pericardial and diaphragmatic defect	20 Cases
3226	Deafness-lymphedema-leukemia syndrome	20 Cases
3387	Isolated anterior cervical hypertrichosis	20 Cases
1134	Isolated arrhinia	20 Cases
2394	Pyruvate dehydrogenase E3 deficiency	20 Cases
198	Occipital horn syndrome	20 Cases
33445	Neuroectodermal melanolysosomal disease	20 Cases
26137	Juvenile temporal arteritis	20 Cases
69084	Pure hair and nail ectodermal dysplasia	20 Cases
67046	3-methylglutaconic aciduria type 1	20 Cases
69723	Tyrosinemia type 3	20 Cases
65283	Timothy syndrome	20 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
71289	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome	20 Cases
73271	Bleeding diathesis due to a collagen receptor defect	20 Cases
63442	Angel-shaped phalango-epiphyseal dysplasia	20 Cases
83616	Rubella panencephalitis	20 Cases
79154	2-aminoacidic 2-oxoadipic aciduria	20 Cases
79084	Familial partial lipodystrophy, Körberling type	20 Cases
88642	Channelopathy-associated congenital insensitivity to pain	20 Cases
88639	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	20 Cases
86797	Atypical lichen myxedematosus	20 Cases
88628	Posterior column ataxia-retinitis pigmentosa syndrome	20 Cases
86919	Keratosis palmaris et plantaris-clinodactyly syndrome	20 Cases
85192	Calvarial doughnut lesions-bone fragility syndrome	20 Cases
98791	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	20 Cases
98768	Spinocerebellar ataxia type 13	20 Cases
97232	Fingerprint body myopathy	20 Cases
93941	Laryngotracheoesophageal cleft type 4	20 Cases
139455	Autosomal recessive bestrophinopathy	20 Cases
137678	Czech dysplasia, metatarsal type	20 Cases
101110	Spinocerebellar ataxia type 20	20 Cases
100976	Bathing suit ichthyosis	20 Cases
228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M	20 Cases
228247	Acquired pseudoxanthoma elasticum	20 Cases
247522	Primary ciliary dyskinesia-retinitis pigmentosa syndrome	20 Cases
251393	Localized junctional epidermolysis bullosa, non-Herlitz type	20 Cases
251061	7q31 microdeletion syndrome	20 Cases
251028	2q33.1 microdeletion syndrome	20 Cases
178364	Syndromic microphthalmia type 5	20 Cases
309854	Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome	20 Cases
268114	RAS-associated autoimmune leukoproliferative disease	20 Cases
280779	Cutaneous collagenous vasculopathy	20 Cases
289863	Atypical glycine encephalopathy	20 Cases
369897	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	20 Cases
391376	Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome	20 Cases
457240	X-linked intellectual disability-short stature-overweight syndrome	20 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
420179	Malan overgrowth syndrome	20 Cases
100043	Autosomal dominant intermediate Charcot-Marie-Tooth disease type A	20 Cases
79476	Griselli syndrome type 1	20 Cases
448242	Autosomal recessive brachyolmia	20 Cases
443811	PGM3-CDG	20 Cases
86920	Dermatopathia pigmentosa reticularis	20 Cases
1447	Ring chromosome 4 syndrome	20 Cases
96175	Ring chromosome 11 syndrome	20 Cases
53583	Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	20 Cases
2717	Oculotrichoanal syndrome	20 Cases
3339	Toriello-Lacassie-Droste syndrome	19 Cases
43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency	19 Cases
75857	6q terminal deletion syndrome	19 Cases
89838	Epidermolysis bullosa simplex, autosomal recessive K14	19 Cases
139441	Hypomyelination with atrophy of basal ganglia and cerebellum	19 Cases
139447	Progressive cavitating leukoencephalopathy	19 Cases
217371	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	19 Cases
228410	Polyvalvular heart disease syndrome	19 Cases
228387	Spondylo-megaepiphyseal-metaphyseal dysplasia	19 Cases
251046	6p22 microdeletion syndrome	19 Cases
178487	Adult intestinal botulism	19 Cases
171848	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	19 Cases
268261	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion	19 Cases
280671	Megaconial congenital muscular dystrophy	19 Cases
401973	MEND syndrome	19 Cases
397946	Autosomal spastic paraparesis type 58	19 Cases
352662	Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome	19 Cases
391320	East Texas bleeding disorder	19 Cases
438159	STAT3-related early-onset multisystem autoimmune disease	19 Cases
247868	NLRP12-associated hereditary periodic fever syndrome	19 Cases
284169	Facial dysmorphisms-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion	19 Cases
466962	SMARCA4-deficient sarcoma of thorax	19 Cases
505248	Mucopolysaccharidosis-like syndrome with congenital heart defects and hematopoietic disorders	19 Cases
497757	MME-related autosomal dominant Charcot-Marie Tooth disease type 2	19 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
494433	MIRAGE syndrome	19 Cases
482601	Adenylosuccinate synthetase-like 1-related distal myopathy	19 Cases
935	Short-limb skeletal dysplasia with severe combined immunodeficiency	19 Cases
2353	Schilbach-Rott syndrome	18 Cases
1272	Aymé-Gripp syndrome	18 Cases
1441	Ring chromosome 17 syndrome	18 Cases
66637	Diaphanospondylodysostosis	18 Cases
86309	DPAGT1-CDG	18 Cases
85167	Spondyloepiphyseal dysplasia-cone-rod dystrophy syndrome	18 Cases
139515	Charcot-Marie-Tooth disease type 4J	18 Cases
228402	2q23.1 microdeletion syndrome	18 Cases
158025	Hereditary progressive mucinous histiocytosis	18 Cases
300319	Charcot-Marie-Tooth disease type 2P	18 Cases
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	18 Cases
324588	Familial dyskinesia and facial myokymia	18 Cases
397596	Activated PI3K-delta syndrome	18 Cases
369962	Methylmalonic acidemia with homocystinuria, type cbIX	18 Cases
370046	Didymosis aplasticosebacea	18 Cases
363417	Temptamy preaxial brachydactyly syndrome	18 Cases
481152	PYCR2-related microcephaly-progressive leukoencephalopathy	18 Cases
319182	Wiedemann-Steiner syndrome	18 Cases
1449	Ring chromosome 7 syndrome	18 Cases
2501	Metaphyseal chondrodysplasia, Spahr type	18 Cases
468641	Chronic enteropathy associated with SLC2A1 gene	18 Cases
508498	Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome	18 Cases
171719	Cutis laxa-Marfanoid syndrome	18 Cases
99741	King-Denborough syndrome	18 Cases
261344	Trisomy 1q	18 Cases
96171	Ring chromosome 2 syndrome	18 Cases
1692	Mosaic trisomy 1	18 Cases
2318	Joubert syndrome with oculorenal defect	17 Cases
560	Marshall syndrome	17 Cases
3204	Stormorken-Sjaastad-Langslet syndrome	17 Cases
1325	Camptodactyly-taurinuria syndrome	17 Cases
1104	Anophthalmia plus syndrome	17 Cases
1954	Congenital lethal erythroderma	17 Cases
1908	Aminopterin/methotrexate embryofetopathy	17 Cases
2117	Hartsfield syndrome	17 Cases
69744	Circumscribed palmoplantar hypokeratosis	17 Cases
79283	Methylmalonic acidemia with homocystinuria, type cbID	17 Cases
91131	DK1-CDG	17 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
93282	Spondyloepiphyseal dysplasia, PAPSS2 type	17 Cases
210141	Inherited congenital spastic tetraplegia	17 Cases
230839	Ehlers-Danlos syndrome due to tenascin-X deficiency	17 Cases
238505	Autosomal recessive lymphoproliferative disease	17 Cases
210115	Sterile multifocal osteomyelitis with periostitis and pustulosis	17 Cases
300530	Pseudohypoaldosteronism type 2E	17 Cases
324381	Hereditary inclusion body myopathy type 4	17 Cases
319595	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency	17 Cases
404443	Tall stature-intellectual disability-facial dysmorphism syndrome	17 Cases
436159	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsufficiency	17 Cases
435988	Chronic atrial and intestinal dysrhythmia syndrome	17 Cases
99853	Ovarioleukodystrophy	17 Cases
480880	X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	17 Cases
477673	Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	17 Cases
3350	Tremor-nystagmus-duodenal ulcer syndrome	17 Cases
399058	Alpha-B crystallin-related late-onset myopathy	17 Cases
464760	Familial cavitory optic disc anomaly	17 Cases
502434	STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome	17 Cases
500533	Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	17 Cases
363429	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome	17 Cases
227976	Autosomal recessive optic atrophy, OPA7 type	17 Cases
1195	Congenital atransferrinemia	16 Cases
2102	GTP cyclohydrolase I deficiency	16 Cases
1438	Ring chromosome 10 syndrome	16 Cases
920	Ablepharon macrostomia syndrome	16 Cases
1231	Barber-Say syndrome	16 Cases
2089	Glycogen storage disease due to hepatic glycogen synthase deficiency	16 Cases
2538	Microgastria-limb reduction defect syndrome	16 Cases
34514	Autosomal recessive limb-girdle muscular dystrophy type 2G	16 Cases
33067	Metaphyseal chondrodysplasia, Jansen type	16 Cases
69737	Bosley-Salih-Alorainy syndrome	16 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
71528	Obesity due to prohormone convertase 1 deficiency	16 Cases
63273	Distal myopathy with posterior leg and anterior hand involvement	16 Cases
85198	Dysspondyloenchondromatosis	16 Cases
93972	Juberg-Marsidi syndrome	16 Cases
238455	Infantile dystonia-parkinsonism	16 Cases
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	16 Cases
314376	Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency	16 Cases
314566	Primary progressive apraxia of speech	16 Cases
319171	Distal 17p13.1 microdeletion syndrome	16 Cases
261257	Distal 17p13.3 microdeletion syndrome	16 Cases
279943	Hereditary neutrophilia	16 Cases
369852	Congenital neutropenia-myelofibrosis-nephromegaly syndrome	16 Cases
369861	Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome	16 Cases
397606	PrP systemic amyloidosis	16 Cases
289601	Hereditary arterial and articular multiple calcification syndrome	16 Cases
457193	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	16 Cases
457279	Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome	16 Cases
306734	Primary dystonia, DYT21 type	16 Cases
306674	Kufor-Rakeb syndrome	16 Cases
53296	Familial cutaneous collagenoma	16 Cases
93357	SPONASTRIME dysplasia	16 Cases
178355	Smith-McCort dysplasia	16 Cases
464282	Spastic paraparesis-severe developmental delay-epilepsy syndrome	16 Cases
319524	Combined oxidative phosphorylation defect type 15	16 Cases
488191	Female infertility due to oocyte meiotic arrest	16 Cases
2731	Taurodontia-absent teeth-sparse hair syndrome	15 Cases
1901	Ehlers-Danlos syndrome, dermatosparaxis type	15 Cases
69063	Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	15 Cases
79284	Methylmalonic aciduria with homocystinuria type cbf	15 Cases
79321	ALG3-CDG	15 Cases
79325	ALG8-CDG	15 Cases
90400	Scleromyxedema without monoclonal gammopathy	15 Cases
88620	Isolated congenital anosmia	15 Cases
103908	Congenital sodium diarrhea	15 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
99954	Charcot-Marie-Tooth disease type 4H	15 Cases
221043	Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome	15 Cases
250984	Autosomal recessive Stickler syndrome	15 Cases
171680	Lissencephaly due to TUBA1A mutation	15 Cases
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	15 Cases
314432	Spigelian hernia-cryptorchidism syndrome	15 Cases
314647	Non-progressive cerebellar ataxia with intellectual disability	15 Cases
329324	Inverse Klippel-Trénaunay syndrome	15 Cases
280763	Severe intellectual disability and progressive spastic paraparesis	15 Cases
280633	Multiple congenital anomalies-hypotonia-seizures syndrome	15 Cases
397744	Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome	15 Cases
397615	Obesity due to CEP19 deficiency	15 Cases
401768	Proximal myopathy with extrapyramidal signs	15 Cases
2075	Genitopalatocardiac syndrome	15 Cases
453499	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome	15 Cases
457050	Autosomal dominant mitochondrial myopathy with exercise intolerance	15 Cases
79149	Dermochondrocorneal dystrophy	15 Cases
456369	Polyglucosan body myopathy type 2	15 Cases
447997	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	15 Cases
436144	Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	15 Cases
436169	Thrombomodulin-related bleeding disorder	15 Cases
98949	Complete cryptophthalmia	15 Cases
79351	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form	15 Cases
90796	46,XY disorder of sex development due to isolated 17,20-lyase deficiency	15 Cases
500163	SIN3A-related intellectual disability syndrome	15 Cases
137754	Neurological conditions associated with aminoacylase 1 deficiency	15 Cases
513456	Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome	15 Cases
482606	X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome	15 Cases
53696	Lethal arthrogryposis-anterior horn cell disease syndrome	15 Cases
96181	Maternal uniparental disomy of chromosome 6	15 Cases
527276	Encephalopathy due to mitochondrial and peroxisomal fission defect	15 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
521426	PLAA-associated neurodevelopmental disorder	15 Cases
1193	Atkin-Flaitz syndrome	14 Cases
1516	Craniofacial dysostosis	14 Cases
2707	Oculocerebrofacial syndrome, Kaufman type	14 Cases
2789	Lateral meningocele syndrome	14 Cases
3363	Trichomegaly-retina pigmentary degeneration-dwarfism syndrome	14 Cases
36355	Bleeding disorder due to P2Y12 defect	14 Cases
75378	Oligocone trichromacy	14 Cases
90390	Anonychia-onychodystrophy syndrome	14 Cases
93356	Spondyloepimetaphyseal dysplasia, Missouri type	14 Cases
139578	Mutilating hereditary sensory neuropathy with spastic paraplegia	14 Cases
238750	4q21 microdeletion syndrome	14 Cases
168796	Heart-hand syndrome, Slovenian type	14 Cases
199351	Adult-onset dystonia-parkinsonism	14 Cases
306507	LAMB2-related infantile-onset nephrotic syndrome	14 Cases
314394	Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome	14 Cases
314051	Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome	14 Cases
397758	Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies	14 Cases
401849	Autosomal spastic paraplegia type 72	14 Cases
369970	Microcornea-myopic chorioretinal atrophy-telecanthus syndrome	14 Cases
369920	Pontocerebellar hypoplasia type 9	14 Cases
1791	Frontofacionasal dysplasia	14 Cases
457351	Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome	14 Cases
2435	Hypo- and hypermelanotic cutaneous macules-retarded growth-intellectual disability syndrome	14 Cases
423384	Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency	14 Cases
480907	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	14 Cases
480483	Progressive familial intrahepatic cholestasis type 4	14 Cases
88659	Autosomal dominant progressive nephropathy with hypertension	14 Cases
307766	Curly hair-acral keratoderma-caries syndrome	14 Cases
320375	Autosomal recessive spastic paraplegia type 55	14 Cases
466718	Martinique crinkled retinal pigment epitheliopathy	14 Cases
2719	Oculocerebral hypopigmentation syndrome, Cross type	14 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
508529	Generalized basal epidermolysis bullosa simplex with skin atrophy, scarring and hair loss	14 Cases
137783	Lethal congenital contracture syndrome type 3	14 Cases
364028	X-linked intellectual disability due to GRIA3 mutations	14 Cases
2378	Laurin-Sandrow syndrome	14 Cases
284289	Adult-onset autosomal recessive cerebellar ataxia	14 Cases
284139	Larsen-like syndrome, B3GAT3 type	14 Cases
371007	Congenital muscular dystrophy with hyperlaxity	14 Cases
1788	Acrofacial dysostosis, Rodríguez type	13 Cases
2319	Juberg-Hayward syndrome	13 Cases
3097	Meacham syndrome	13 Cases
69739	Athabaskan brainstem dysgenesis syndrome	13 Cases
66631	CEDNIK syndrome	13 Cases
50945	Blomstrand lethal chondrodysplasia	13 Cases
79329	MGAT2-CDG	13 Cases
85174	Pseudodiastrophic dysplasia	13 Cases
101102	Charcot-Marie-Tooth disease type 2H	13 Cases
231720	Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome	13 Cases
168549	Axial spondylometaphyseal dysplasia	13 Cases
178377	Osteosclerosis-developmental delay-craniosynostosis syndrome	13 Cases
171612	Autosomal dominant spastic paraplegia type 37	13 Cases
293958	Hypertelorism-preauricular sinus-punctual pits-deafness syndrome	13 Cases
306530	Congenital hereditary facial paralysis-variable hearing loss syndrome	13 Cases
313936	PENS syndrome	13 Cases
319605	X-linked mendelian susceptibility to mycobacterial diseases	13 Cases
319547	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency	13 Cases
329249	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency	13 Cases
280406	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	13 Cases
284160	8q21.11 microdeletion syndrome	13 Cases
401953	Episodic ataxia with slurred speech	13 Cases
356978	D,L-2-hydroxyglutaric aciduria	13 Cases
357008	Atypical hemolytic-uremic syndrome with DGKE deficiency	13 Cases
363412	Hypomyelination with brain stem and spinal cord involvement and leg spasticity	13 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
352665	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to 9q21 microdeletion	13 Cases
3268	Radioulnar synostosis-microcephaly-scoliosis syndrome	13 Cases
448251	Progressive autosomal recessive ataxia-deafness syndrome	13 Cases
436274	Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa	13 Cases
435438	Progressive myoclonic epilepsy type 7	13 Cases
443098	Hyperostosis cranialis interna	13 Cases
399103	Distal nebulin myopathy	13 Cases
436151	Intellectual disability-expressive aphasia-facial dysmorphism syndrome	13 Cases
439212	Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome	13 Cases
476102	Hereditary pediatric Behçet-like disease	13 Cases
79502	Punctate palmoplantar keratoderma type 2	13 Cases
939	3-hydroxyisobutyric aciduria	13 Cases
476394	PMP2-related Charcot-Marie-Tooth disease type 1	13 Cases
3042	Intellectual disability-cataracts-calcified pinnae-myopathy syndrome	13 Cases
466934	VPS11-related autosomal recessive hypomyelinating leukodystrophy	13 Cases
79478	Griscelli syndrome type 3	13 Cases
329813	Mosaic genome-wide paternal uniparental disomy	13 Cases
96055	Tetrasomy 21	13 Cases
844	Lown-Ganong-Levine syndrome	12 Cases
1473	Uveal coloboma-cleft lip and palate-intellectual disability	12 Cases
1008	Alopecia-epilepsy-pyorrhea-intellectual disability syndrome	12 Cases
1190	Atelosteogenesis type I	12 Cases
1784	Acrofrontofacionasal dysostosis	12 Cases
1487	Cooks syndrome	12 Cases
1555	Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome	12 Cases
1458	CODAS syndrome	12 Cases
2224	Hypertryptophanemia	12 Cases
2919	Orofaciodigital syndrome type 5	12 Cases
3460	Torg-Winchester syndrome	12 Cases
59303	Neonatal ichthyosis-sclerosing cholangitis syndrome	12 Cases
79328	ALG9-CDG	12 Cases
85320	X-linked intellectual disability-macrocephaly-macroorchidism syndrome	12 Cases
98772	Spinocerebellar ataxia type 19/22	12 Cases
93395	Ballard syndrome	12 Cases
217346	19q13.11 microdeletion syndrome	12 Cases
217377	Microduplication Xp11.22p11.23 syndrome	12 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
210571	Dystonia 16	12 Cases
244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis	12 Cases
238763	Glaucoma secondary to spherophakia/ectopia lentis and megalocornea	12 Cases
254531	Temple syndrome due to paternal 14q32.2 hypomethylation	12 Cases
247794	Juvenile cataract-microcornea-renal glucosuria syndrome	12 Cases
166035	Brachydactyly-short stature-retinitis pigmentosa syndrome	12 Cases
171829	6q16 deletion syndrome	12 Cases
209973	Benign nocturnal alternating hemiplegia of childhood	12 Cases
199340	Muscular dystrophy, Selcen type	12 Cases
300570	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation	12 Cases
300547	Autosomal recessive infantile hypercalcemia	12 Cases
313892	Developmental and speech delay due to SOX5 deficiency	12 Cases
314585	15q overgrowth syndrome	12 Cases
261323	21q22.11q22.12 microdeletion syndrome	12 Cases
280384	Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome	12 Cases
280620	Progressive myoclonic epilepsy type 6	12 Cases
284460	Acute annular outer retinopathy	12 Cases
412066	PRKAR1B-related neurodegenerative dementia with intermediate filaments	12 Cases
363523	Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome	12 Cases
96186	Maternal uniparental disomy of chromosome 20	12 Cases
437572	MYH7-related late-onset scapuloperoneal muscular dystrophy	12 Cases
442582	AH amyloidosis	12 Cases
1179	Benign paroxysmal tonic upgaze of childhood with ataxia	12 Cases
2662	Keipert syndrome	12 Cases
100046	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	12 Cases
2935	Crossed polysyndactyly	12 Cases
468631	Microcephalic primordial dwarfism due to RTTN deficiency	12 Cases
459033	Ataxia-oculomotor apraxia type 4	12 Cases
505237	Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome	12 Cases
508533	Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome	12 Cases
496689	Kyphoscoliosis-lateral tongue atrophy-hereditary spastic paraparesis syndrome	12 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
99672	Fried's tooth and nail syndrome	12 Cases
141148	Hemifacial myohyperplasia	12 Cases
2253	Foveal hypoplasia-presenile cataract syndrome	11 Cases
1031	Enamel-renal syndrome	11 Cases
1660	Dermoodontodysplasia	11 Cases
1757	Fibular dimelia-diplopodia syndrome	11 Cases
1497	X-linked complicated corpus callosum dysgenesis	11 Cases
2016	Cleft palate-lateral synechia syndrome	11 Cases
2329	Karsch-Neugebauer syndrome	11 Cases
1479	Atrial septal defect-atrioventricular conduction defects syndrome	11 Cases
2959	Progeria-short stature-pigmented nevi syndrome	11 Cases
2987	Antecubital pterygium syndrome	11 Cases
2832	Short tarsus-absence of lower eyelashes syndrome	11 Cases
2854	Fuhrmann syndrome	11 Cases
79324	ALG12-CDG	11 Cases
79076	Juvenile polyposis of infancy	11 Cases
91135	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	11 Cases
91132	Ichthyosis-hypotrichosis syndrome	11 Cases
85336	X-linked neurodegenerative syndrome, Hamel type	11 Cases
88619	Familial acute necrotizing encephalopathy	11 Cases
217390	Combined immunodeficiency due to DOCK8 deficiency	11 Cases
228169	Autosomal dominant striatal neurodegeneration	11 Cases
238744	Mammary-digital-nail syndrome	11 Cases
238755	Autosomal dominant limb-girdle muscular dystrophy type 1H	11 Cases
168588	Hyperandrogenism due to cortisone reductase deficiency	11 Cases
168624	Familial scaphocephaly syndrome, McGillivray type	11 Cases
166272	Odontochondrodysplasia	11 Cases
166282	Familial sick sinus syndrome	11 Cases
210133	Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome	11 Cases
300293	Transient infantile hypertriglyceridemia and hepatosteatosis	11 Cases
313855	FGFR2-related bent bone dysplasia	11 Cases
313850	Infantile cerebellar-retinal degeneration	11 Cases
330050	DNM1L-related encephalopathy due to mitochondrial and peroxisomal fission defect	11 Cases
261349	2p15p16.1 microdeletion syndrome	11 Cases
280553	Fatal infantile hypertonic myofibrillar myopathy	11 Cases
397937	Polyglucosan body myopathy type 1	11 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
352712	Facial dysmorphism-immunodeficiency-livedo-short stature syndrome	11 Cases
2163	Holoprosencephaly-craniosynostosis syndrome	11 Cases
457185	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome	11 Cases
444013	Combined oxidative phosphorylation defect type 23	11 Cases
443988	Ventriculomegaly-cystic kidney disease	11 Cases
444051	20q11.2 microdeletion syndrome	11 Cases
477749	Pontine autosomal dominant microangiopathy with leukoencephalopathy	11 Cases
98912	Late-onset distal myopathy, Markesberry-Griggs type	11 Cases
464724	Fever-associated acute infantile liver failure syndrome	11 Cases
319189	Familial cortical myoclonus	11 Cases
468661	Autosomal recessive spastic paraparesis type 74	11 Cases
506307	Stromme syndrome	11 Cases
96172	Ring chromosome 3 syndrome	11 Cases
221120	Pseudoaminopterin syndrome	11 Cases
521406	Dystonia-parkinsonism-hpermangansemia syndrome	11 Cases
521308	Frontonasal dysplasia-bifid nose-upper limb anomalies syndrome	11 Cases
521450	LAMA5-related multisystemic syndrome	11 Cases
522077	Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome	11 Cases
1627	Deletion 5q35	10 Cases
968	Acromesomelic dysplasia, Hunter-Thompson type	10 Cases
958	Acro-renal-mandibular syndrome	10 Cases
916	Aase-Smith syndrome	10 Cases
1313	Infantile choriodocerebral calcification syndrome	10 Cases
1336	Hyperkeratosis-hyperpigmentation syndrome	10 Cases
1263	Boomerang dysplasia	10 Cases
1010	Autosomal dominant palmoplantar keratoderma and congenital alopecia	10 Cases
1150	Arthrogryposis multiplex congenita-whistling face syndrome	10 Cases
1568	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	10 Cases
1471	Coloboma of macula-brachydactyly type B syndrome	10 Cases
1426	Greenberg dysplasia	10 Cases
2255	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome	10 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
2072	Gaucher disease-opthalmoplegia-cardiovascular calcification syndrome	10 Cases
2047	Flynn-Aird syndrome	10 Cases
2658	Lenz-Majewski hyperostotic dwarfism	10 Cases
2579	Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome	10 Cases
2590	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	10 Cases
3032	NPHP3-related Meckel-like syndrome	10 Cases
2839	Pelvis-shoulder dysplasia	10 Cases
2880	Phosphoenolpyruvate carboxykinase deficiency	10 Cases
1171	Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome	10 Cases
3317	Thoracolaryngopelvic dysplasia	10 Cases
3469	XK aprosencephaly syndrome	10 Cases
3439	Von Voss-Cherstvoy syndrome	10 Cases
33574	Glutamate-cysteine ligase deficiency	10 Cases
39	Acromelanosis	10 Cases
71212	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	10 Cases
46627	Char syndrome	10 Cases
79503	Ichthyosis hystrix of Curth-Macklin	10 Cases
79279	Alpha-N-acetylgalactosaminidase deficiency type 1	10 Cases
79280	Alpha-N-acetylgalactosaminidase deficiency type 2	10 Cases
79281	Alpha-N-acetylgalactosaminidase deficiency type 3	10 Cases
79083	PPARG-related familial partial lipodystrophy	10 Cases
90398	Localized lichen myxedematosus with mixed features of different subtypes	10 Cases
89841	Centripetalis recessive dystrophic epidermolysis bullosa	10 Cases
85329	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	10 Cases
86918	Diffuse palmoplantar keratoderma-acrocyanosis syndrome	10 Cases
85163	Hypomyelination-congenital cataract syndrome	10 Cases
85274	Syndromic X-linked intellectual disability 7	10 Cases
97340	Hunter-McAlpine craniostenosis	10 Cases
97240	Zebra body myopathy	10 Cases
93599	Primary hyperoxaluria type 2	10 Cases
93406	Syndactyly type 5	10 Cases
139406	Encephalopathy due to prosaposin deficiency	10 Cases
139426	Perioral myoclonia with absences	10 Cases
101111	Spinocerebellar ataxia type 25	10 Cases
100994	Autosomal dominant spastic paraparesis type 13	10 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
101007	Autosomal recessive spastic paraparesis type 27	10 Cases
220465	Laron syndrome with immunodeficiency	10 Cases
217335	RIN2 syndrome	10 Cases
210144	Lethal polymalformative syndrome, Boissel type	10 Cases
228426	Syndromic multisystem autoimmune disease due to Itch deficiency	10 Cases
240112	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome	10 Cases
254504	Inhalational botulism	10 Cases
254411	Annular atrophic lichen planus	10 Cases
163966	X-linked dominant chondrodyplasia, Chassaing-Lacombe type	10 Cases
140969	Saldino-Mainzer syndrome	10 Cases
141007	Orofaciodigital syndrome type 9	10 Cases
169090	Combined immunodeficiency due to CRAC channel dysfunction	10 Cases
166073	Pontocerebellar hypoplasia type 6	10 Cases
168486	Congenital neuronal ceroid lipofuscinosis	10 Cases
178330	Heinz body anemia	10 Cases
294016	Microcephaly-capillary malformation syndrome	10 Cases
313906	Congenital pancreatic cyst	10 Cases
314662	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	10 Cases
324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency	10 Cases
329228	Microcephalic primordial dwarfism due to ZNF335 deficiency	10 Cases
263458	Hyperinsulinism due to INSR deficiency	10 Cases
263482	Spondyloepiphyseal dysplasia, Maroteaux type	10 Cases
276280	Hemihyperplasia-multiple lipomatosis syndrome	10 Cases
280615	Hemoglobinopathy Toms River	10 Cases
280794	Pseudoxanthomatous diffuse cutaneous mastocytosis	10 Cases
284227	TEMPI syndrome	10 Cases
404448	ADNP syndrome	10 Cases
401901	Huntington disease-like syndrome due to C9ORF72 expansions	10 Cases
363400	Severe neurodegenerative syndrome with lipodystrophy	10 Cases
352641	Autosomal recessive cerebellar ataxia with late-onset spasticity	10 Cases
352734	Minimal pigment oculocutaneous albinism type 1	10 Cases
352737	Temperature-sensitive oculocutaneous albinism type 1	10 Cases
99807	PEHO-like syndrome	10 Cases
420789	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	10 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
309246	GM2 gangliosidosis, AB variant	10 Cases
439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease	10 Cases
93398	Genochondromatosis type 2	10 Cases
399081	KLHL9-related early-onset distal myopathy	10 Cases
3259	Syndactyly-polydactyly-ear lobe syndrome	10 Cases
3023	External auditory canal atresia-vertical talus-hypertelorism syndrome	10 Cases
464443	COG6-CGD	10 Cases
468699	SLC39A8-CDG	10 Cases
466950	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation	10 Cases
319671	Microcephalic primordial dwarfism, Alazami type	10 Cases
93347	Anauxetic dysplasia	10 Cases
506358	Gabriele-de Vries syndrome	10 Cases
494344	RERE-related neurodevelopmental syndrome	10 Cases
495274	Charcot-Marie-Tooth disease type 2T	10 Cases
1439	Ring chromosome 12 syndrome	10 Cases
96178	Ring chromosome 16 syndrome	10 Cases
99329	48,XYYY syndrome	10 Cases
1443	Ring chromosome 19 syndrome	10 Cases
521445	Microcephaly-facial dysmorphism-ocular anomalies-multiple congenital anomalies syndrome	10 Cases
1000	Ocular albinism with late-onset sensorineural deafness	9 Cases
1345	Cardiomyopathy-cataract-hip spine disease syndrome	9 Cases
1264	Tricho-retino-dento-digital syndrome	9 Cases
1292	Brachymorphism-onychodysplasia-dysphalangism syndrome	9 Cases
1553	Curry-Jones syndrome	9 Cases
2213	Hypertelorism-microtia-facial clefting syndrome	9 Cases
2081	Cerebral gigantism-jaw cysts syndrome	9 Cases
2008	Acrocardiofacial syndrome	9 Cases
2557	Mietens syndrome	9 Cases
2952	Adducted thumbs-arthrogryposis syndrome, Christian type	9 Cases
35701	3-hydroxy-3-methylglutaryl-CoA synthase deficiency	9 Cases
35704	L-Arginine:glycine amidinotransferase deficiency	9 Cases
35107	Desmosterolosis	9 Cases
79405	Junctional epidermolysis bullosa inversa	9 Cases
79322	DPM1-CDG	9 Cases
93317	Spondylometaphyseal dysplasia, Sedaghatian type	9 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
85338	X-linked intellectual disability-ataxia-apraxia syndrome	9 Cases
83619	Macrostomia-preauricular tags-external ophthalmoplegia syndrome	9 Cases
85199	Craniosynostosis-anal anomalies-porokeratosis syndrome	9 Cases
85286	X-linked intellectual disability, Shashi type	9 Cases
137628	Cardiac anomalies-heterotaxy syndrome	9 Cases
231154	Combined immunodeficiency due to partial RAG1 deficiency	9 Cases
254525	Temple syndrome due to paternal 14q32.2 microdeletion	9 Cases
251279	Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	9 Cases
163979	X-linked intellectual disability-craniofacioskeletal syndrome	9 Cases
140936	Lelis syndrome	9 Cases
168558	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	9 Cases
209951	Autosomal recessive spastic paraparesis type 18	9 Cases
293939	Distal Xq28 microduplication syndrome	9 Cases
293948	1p21.3 microdeletion syndrome	9 Cases
300179	Ehlers-Danlos syndrome, kyphoscoliotic and deafness type	9 Cases
314466	Atypical Meigs syndrome	9 Cases
314679	Cerebrofacioarticular syndrome	9 Cases
324585	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	9 Cases
329478	Adult-onset distal myopathy due to VCP mutation	9 Cases
261190	15q14 microdeletion syndrome	9 Cases
263487	COG5-CDG	9 Cases
280679	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	9 Cases
397750	Periodic paralysis with later-onset distal motor neuropathy	9 Cases
397787	Severe combined immunodeficiency due to IKK2 deficiency	9 Cases
397612	Macrocephaly-developmental delay syndrome	9 Cases
1120	Lung agenesis-heart defect-thumb anomalies syndrome	9 Cases
401945	Moyamoya disease with early-onset achalasia	9 Cases
363710	Spinocerebellar ataxia type 37	9 Cases
352745	Oculocutaneous albinism type 7	9 Cases
370927	SSR4-CDG	9 Cases
3056	X-linked intellectual disability, Brooks type	9 Cases
319199	Autosomal recessive spastic paraparesis type 53	9 Cases
438075	Ketoacidosis due to monocarboxylate transporter-1 deficiency	9 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
425120	STING-associated vasculopathy with onset in infancy	9 Cases
1512	Crane-Heise syndrome	9 Cases
480851	Hereditary thrombocytopenia with early-onset myelofibrosis	9 Cases
2680	Hypomyelination neuropathy-arthrogryposis syndrome	9 Cases
477814	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome	9 Cases
169095	Alymphoid cystic thymic dysgenesis	9 Cases
94124	Spinocerebellar ataxia with axonal neuropathy type 1	9 Cases
93952	X-linked intellectual disability, Hedera type	9 Cases
2808	Laryngeal abductor paralysis	9 Cases
502423	Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	9 Cases
505208	3-methylglutaconic aciduria type 8	9 Cases
488197	Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome	9 Cases
221039	Hereditary sclerosing poikiloderma, Weary type	9 Cases
99014	X-linked Charcot-Marie-Tooth disease type 5	9 Cases
1226	Bamforth-Lazarus syndrome	8 Cases
1318	Campomelia, Cumming type	8 Cases
1327	Camptodactyly syndrome, Guadalajara type 1	8 Cases
1278	Brachydactyly-preaxial hallux varus syndrome	8 Cases
1188	Ataxia-deafness-intellectual disability syndrome	8 Cases
1824	Lowry-Wood syndrome	8 Cases
1366	Autosomal recessive palmoplantar keratoderma and congenital alopecia	8 Cases
1454	Joubert syndrome with hepatic defect	8 Cases
2180	Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome	8 Cases
2206	Ankylosing vertebral hyperostosis with tylosis	8 Cases
2107	Hall-Riggs syndrome	8 Cases
2563	MOMO syndrome	8 Cases
2561	Pyramidal molars-abnormal upper lip syndrome	8 Cases
2371	Lethal Larsen-like syndrome	8 Cases
2351	Koushoeff syndrome	8 Cases
2958	X-linked intellectual disability-dysmorphism-cerebral atrophy syndrome	8 Cases
2934	Polysyndactyly-cardiac malformation syndrome	8 Cases
3219	Fountain syndrome	8 Cases
1655	Müllerian derivatives-lymphangiectasia-polydactyly syndrome	8 Cases
2326	Kallmann syndrome-heart disease syndrome	8 Cases
3474	CHIME syndrome	8 Cases
33572	5-oxoprolinase deficiency	8 Cases
79323	MPDU1-CDG	8 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
79333	COG7-CDG	8 Cases
85282	MEHMO syndrome	8 Cases
85273	X-linked intellectual disability, Abidi type	8 Cases
137639	Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome	8 Cases
221054	Acrocephalopolydactyly	8 Cases
244310	RFT1-CDG	8 Cases
231736	Microcornea-posterior megalolenticonus-persistent fetal vasculature-coloboma syndrome	8 Cases
254528	Kagami-Ogata syndrome due to maternal 14q32.2 microdeletion	8 Cases
251290	Parietal foramina with clavicular hypoplasia	8 Cases
178506	Brain calcification, Rajab type	8 Cases
178389	Osteopetrosis-hypogammaglobulinemia syndrome	8 Cases
306558	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome	8 Cases
306577	Sodium channelopathy-related small fiber neuropathy	8 Cases
306547	Porencephaly-microcephaly-bilateral congenital cataract syndrome	8 Cases
314811	Short stature due to GHSR deficiency	8 Cases
314637	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	8 Cases
324321	Sinoatrial node dysfunction and deafness	8 Cases
329258	Autosomal dominant Charcot-Marie-Tooth disease type 2Q	8 Cases
352479	Autosomal recessive limb-girdle muscular dystrophy type 2U	8 Cases
261211	16p11.2p12.2 microdeletion syndrome	8 Cases
261483	Xq27.3q28 duplication syndrome	8 Cases
263665	NK-cell enteropathy	8 Cases
276432	Ogden syndrome	8 Cases
280325	Distal monosomy 12p	8 Cases
280071	ALG11-CDG	8 Cases
397715	Joubert syndrome with Jeune asphyxiating thoracic dystrophy	8 Cases
404454	Alacrimia-choreoathetosis-liver dysfunction syndrome	8 Cases
401942	Familial median cleft of the upper and lower lips	8 Cases
397590	Silver-Russell syndrome due to a point mutation	8 Cases
352649	Brain dopamine-serotonin vesicular transport disease	8 Cases
352487	Digital anomalies-intellectual disability-short stature syndrome	8 Cases
352670	Autosomal dominant intermediate Charcot-Marie-Tooth disease type F	8 Cases
352675	X-linked Charcot-Marie-Tooth disease type 6	8 Cases
370943	Autism spectrum disorder-epilepsy-arthrogryposis syndrome	8 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
391408	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	8 Cases
922	Familial nasal acilia	8 Cases
2013	Cleft palate-large ears-small head syndrome	8 Cases
457485	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	8 Cases
457406	Multiple mitochondrial dysfunctions syndrome type 4	8 Cases
98807	Primary dystonia, DYT13 type	8 Cases
420573	Severe combined immunodeficiency due to CTPS1 deficiency	8 Cases
435845	Lethal neonatal spasticity-epileptic encephalopathy syndrome	8 Cases
420686	Woolly hair-palmoplantar keratoderma syndrome	8 Cases
435638	3p25.3 microdeletion syndrome	8 Cases
438274	GCGR-related hyperglucagonemia	8 Cases
3216	Conductive deafness-malformed external ear syndrome	8 Cases
1852	X-linked retinal dysplasia	8 Cases
79350	3-phosphoserine phosphatase deficiency	8 Cases
476093	Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome	8 Cases
3019	Ramon syndrome	8 Cases
2812	Parana hard skin syndrome	8 Cases
2252	Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome	8 Cases
464336	BENTA disease	8 Cases
163956	X-linked intellectual disability, Nascimento type	8 Cases
2885	Piebald trait-neurologic defects syndrome	8 Cases
466722	Autosomal recessive spastic paraplegia type 77	8 Cases
468684	CCDC115-CDG	8 Cases
459061	Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome	8 Cases
1450	Ring chromosome 8 syndrome	8 Cases
502430	Metopic ridging-ptosis-facial dysmorphism syndrome	8 Cases
514352	Congenital brachyesophagus-intrathoracic stomach-vertebral anomalies syndrome	8 Cases
494444	DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	8 Cases
494526	Infantile-onset generalized dyskinesia with orofacial involvement	8 Cases
496790	Optic atrophy-peripheral neuropathy-developmental delay syndrome	8 Cases
331226	Susceptibility to infection due to TYK2 deficiency	8 Cases
99330	49,XYYYY syndrome	8 Cases
929	Achalasia-microcephaly syndrome	7 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
2598	Mitochondrial myopathy and sideroblastic anemia	7 Cases
1299	Branchioskeletogenital syndrome	7 Cases
1131	X-linked mandibulofacial dysostosis	7 Cases
1842	Bone dysplasia, lethal Holmgren type	7 Cases
1574	Retinal degeneration-nanophthalmos-glucoma syndrome	7 Cases
2232	Primary hypergonadotropic hypogonadism-partial alopecia syndrome	7 Cases
2095	Gorlin-Chaudhry-Moss syndrome	7 Cases
2141	Diaphragmatic defect-limb deficiency-skull defect syndrome	7 Cases
2560	Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome	7 Cases
2439	Patterson-Stevenson-Fontaine syndrome	7 Cases
3194	Corneodermatoosseous syndrome	7 Cases
3087	Retinohepatoendocrinologic syndrome	7 Cases
3078	Severe X-linked intellectual disability, Gustavson type	7 Cases
2872	Cardiocranial syndrome, Pfeiffer type	7 Cases
2920	Oliver syndrome	7 Cases
3341	Torticollis-keloids-cryptorchidism-renal dysplasia syndrome	7 Cases
33573	Gamma-glutamyl transpeptidase deficiency	7 Cases
338	Familial multiple fibrofolliculoma	7 Cases
65282	Carvajal syndrome	7 Cases
71526	Obesity due to pro-opiomelanocortin deficiency	7 Cases
79094	Grange syndrome	7 Cases
93316	Spondylometaphyseal dysplasia, Schmidt type	7 Cases
85334	X-linked neurodegenerative syndrome, Bertini type	7 Cases
83620	Enteric anendocrinosis	7 Cases
93382	Brachydactyly type A6	7 Cases
139474	17q11.2 microduplication syndrome	7 Cases
101078	X-linked Charcot-Marie-Tooth disease type 4	7 Cases
101008	Autosomal recessive spastic paraplegia type 28	7 Cases
99843	Leukocyte adhesion deficiency type II	7 Cases
211067	Episodic ataxia type 5	7 Cases
228379	Virus-associated trichodysplasia spinulosa	7 Cases
228190	Patent ductus arteriosus-bicuspid aortic valve-hand anomalies syndrome	7 Cases
247198	Progressive cerebello-cerebral atrophy	7 Cases
254534	Kagami-Ogata syndrome due to maternal 14q32.2 hypermethylation	7 Cases
163693	2p21 microdeletion syndrome	7 Cases
163976	X-linked intellectual disability, Van Esch type	7 Cases
168566	Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	7 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
168448	Spondyloepimetaphyseal dysplasia, Bieganski type	7 Cases
178338	UV-sensitive syndrome	7 Cases
209970	Episodic ataxia type 7	7 Cases
199332	Endocrine-cerebro-osteodysplasia syndrome	7 Cases
293978	Deficiency in anterior pituitary function-variable immunodeficiency syndrome	7 Cases
300382	Progeroid and marfanoid aspect-lipodystrophy syndrome	7 Cases
314689	Combined immunodeficiency due to STK4 deficiency	7 Cases
314655	Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion	7 Cases
317476	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia	7 Cases
329329	Autosomal recessive frontotemporal pachygryria	7 Cases
324632	Hendra virus infection	7 Cases
329802	5p13 microduplication syndrome	7 Cases
261229	14q11.2 microduplication syndrome	7 Cases
261279	17q23.1q23.2 microdeletion syndrome	7 Cases
263347	MRCS syndrome	7 Cases
280365	Autosomal semi-dominant severe lipodystrophic laminopathy	7 Cases
293165	Skin fragility-woolly hair-palmoplantar keratoderma syndrome	7 Cases
397695	3q27.3 microdeletion syndrome	7 Cases
404440	Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	7 Cases
404463	Multisystemic smooth muscle dysfunction syndrome	7 Cases
401785	Autosomal recessive spastic paraplegia type 62	7 Cases
369950	Intellectual disability-seizures-macrocephaly-obesity syndrome	7 Cases
369939	Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome	7 Cases
370022	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome	7 Cases
363992	Ichthyosis-short stature-brachydactyly-microspherophakia syndrome	7 Cases
363396	High myopia-sensorineural deafness syndrome	7 Cases
357001	19p13.13 microdeletion syndrome	7 Cases
363432	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency	7 Cases
352587	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	7 Cases
352582	Familial infantile myoclonic epilepsy	7 Cases
353298	Roifman syndrome	7 Cases
391646	Feingold syndrome type 2	7 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
1875	Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome	7 Cases
457375	ITPA-related encephalopathy	7 Cases
2645	Osteoglosphonic dysplasia	7 Cases
85194	Spondylo-ocular syndrome	7 Cases
447896	Tremor-ataxia-central hypomyelination syndrome	7 Cases
420561	Temple-Baraitser syndrome	7 Cases
439822	PDE4D haploinsufficiency syndrome	7 Cases
436242	Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease	7 Cases
435387	Autosomal dominant Charcot-Marie-Tooth disease type 2Y	7 Cases
477857	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency	7 Cases
459051	Spondyloepiphyseal dysplasia, Stanescu type	7 Cases
261204	16p11.2p12.2 microduplication syndrome	7 Cases
399086	Finnish upper limb-onset distal myopathy	7 Cases
1818	Ectodermal dysplasia, trichodontoonychia type	7 Cases
1858	Skeletal dysplasia-epilepsy-short stature syndrome	7 Cases
464738	Congenital cataract-microcephaly-nevus flammeus simplex-severe intellectual disability syndrome	7 Cases
468666	Isolated generalized anhidrosis with normal sweat glands	7 Cases
466703	TMEM199-CDG	7 Cases
467166	Tubulinopathy-associated dysgyria	7 Cases
320355	Autosomal dominant spastic paraparesis type 41	7 Cases
319504	Combined oxidative phosphorylation defect type 8	7 Cases
254930	Combined oxidative phosphorylation defect type 7	7 Cases
500180	Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	7 Cases
504530	Combined immunodeficiency due to Moesin deficiency	7 Cases
513436	Autosomal recessive spastic paraparesis type 7B	7 Cases
508476	Cleft lip and palate-craniofacial dysmorphisms-congenital heart defect-hearing loss syndrome	7 Cases
508093	MEPAN syndrome	7 Cases
497764	Spinocerebellar ataxia type 43	7 Cases
500548	Osteosclerotic metaphyseal dysplasia	7 Cases
496693	Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome	7 Cases
90103	Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome	7 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
308425	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	7 Cases
319623	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency	7 Cases
487825	Pierpont syndrome	7 Cases
488650	Distal myopathy, Tateyama type	7 Cases
486811	Prenatal-onset spinal muscular atrophy with congenital bone fractures	7 Cases
999	Ermine phenotype	6 Cases
1951	Epilepsy-telangiectasia syndrome	6 Cases
1307	Distal limb deficiencies-micrognathia syndrome	6 Cases
991	PAGOD syndrome	6 Cases
1078	Thumb stiffness-brachydactyly-intellectual disability syndrome	6 Cases
1115	Recessive aplasia cutis congenita of limbs	6 Cases
1808	Hidrotic ectodermal dysplasia, Christianson-Fourie type	6 Cases
1661	X-linked corneal dermoid	6 Cases
2306	Isotretinoin-like syndrome	6 Cases
1051	Ramos-Arroyo syndrome	6 Cases
2230	Hypogonadotropic hypogonadism-frontoparietal alopecia syndrome	6 Cases
2057	Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome	6 Cases
2824	Paraplegia-intellectual disability-hyperkeratosis syndrome	6 Cases
2339	Keratosis follicularis-dwarfism-cerebral atrophy syndrome	6 Cases
2328	Kapur-Toriello syndrome	6 Cases
3175	X-linked spasticity-intellectual disability-epilepsy syndrome	6 Cases
3077	X-linked intellectual disability-psychosis-macroorchidism syndrome	6 Cases
2804	W syndrome	6 Cases
3246	Symphalangism with multiple anomalies of hands and feet	6 Cases
66518	Short fifth metacarpals-insulin resistance syndrome	6 Cases
79156	Seizures-intellectual disability due to hydroxylysineuria syndrome	6 Cases
79106	Eiken syndrome	6 Cases
85276	X-linked intellectual disability, Armfield type	6 Cases
93973	Carpenter-Waziri syndrome	6 Cases
211017	Spinocerebellar ataxia type 30	6 Cases
230851	Ehlers-Danlos syndrome, cardiac valvular type	6 Cases
228003	Severe combined immunodeficiency due to CORO1A deficiency	6 Cases
254343	Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome	6 Cases
254346	19p13.12 microdeletion syndrome	6 Cases
254361	Autosomal recessive limb-girdle muscular dystrophy type 2Q	6 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
247815	Autosomal recessive ataxia due to PEX10 deficiency	6 Cases
140952	Syndactyly-telecanthus-anogenital and renal malformations syndrome	6 Cases
157820	Cold-induced sweating syndrome	6 Cases
157832	Craniorhiny	6 Cases
157965	Ehlers-Danlos syndrome, spondylocheirodysplastic type	6 Cases
168984	CLAPO syndrome	6 Cases
169464	Primary CD59 deficiency	6 Cases
178303	8q22.1 microdeletion syndrome	6 Cases
314399	Autosomal dominant aplasia and myelodysplasia	6 Cases
313884	12p12.1 microdeletion syndrome	6 Cases
317428	Combined immunodeficiency due to ORAI1 deficiency	6 Cases
314667	TMEM165-CDG	6 Cases
319612	X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency	6 Cases
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	6 Cases
319563	Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency	6 Cases
324569	Pontocerebellar hypoplasia type 8	6 Cases
352447	Progressive external ophthalmoplegia-myopathy-emaciation syndrome	6 Cases
231537	Hermansky-Pudlak syndrome type 8	6 Cases
401874	Multiple mitochondrial dysfunctions syndrome type 2	6 Cases
401777	Optic atrophy-intellectual disability syndrome	6 Cases
370088	Acute infantile liver failure-multisystemic involvement syndrome	6 Cases
363540	Leukoencephalopathy with mild cerebellar ataxia and white matter edema	6 Cases
357329	Combined immunodeficiency due to IL21R deficiency	6 Cases
352654	Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	6 Cases
3038	Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome	6 Cases
3316	Thomas syndrome	6 Cases
2464	Marfanoid syndrome, De Silva type	6 Cases
457378	Complex lethal osteochondrodysplasia	6 Cases
447980	19p13.3 microduplication syndrome	6 Cases
423454	Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome	6 Cases
398127	Neonatal scleroderma	6 Cases
444463	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome	6 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
436141	Severe intellectual disability-hypotonia-strabismus-coarse face-planovalgus syndrome	6 Cases
436003	Contractures-developmental delay-Pierre Robin syndrome	6 Cases
480898	Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	6 Cases
98893	Congenital muscular dystrophy type 1B	6 Cases
91498	Familial congenital palsy of trochlear nerve	6 Cases
3074	Intellectual disability-short stature-hypertelorism syndrome	6 Cases
2725	Eye defects-arachnodactyly-cardiopathy syndrome	6 Cases
2743	Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome	6 Cases
2815	Spastic paraparesis-deafness syndrome	6 Cases
2793	Otoonychoperoneal syndrome	6 Cases
464288	Short stature-brachydactyly-obesity-global developmental delay syndrome	6 Cases
466688	Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome	6 Cases
466791	Macrocephaly-intellectual disability-left ventricular non compaction syndrome	6 Cases
467176	Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome	6 Cases
319651	Constitutional megaloblastic anemia with severe neurologic disease	6 Cases
99141	Lymphedema-posterior choanal atresia syndrome	6 Cases
500055	16p13.2 microdeletion syndrome	6 Cases
505242	Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome	6 Cases
512260	Congenital cerebellar ataxia due to RNU12 mutation	6 Cases
508523	Hyperphenylalaninemia due to DNAJC12 deficiency	6 Cases
1795	Peripheral dysostosis	6 Cases
488642	TELO2-related intellectual disability-neurodevelopmental disorder	6 Cases
495844	C11ORF73-related autosomal recessive hypomyelinating leukodystrophy	6 Cases
495879	Congenital agenesis of the scrotum	6 Cases
500545	Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract	6 Cases
496756	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome	6 Cases
289513	12q15q21.1 microdeletion syndrome	6 Cases
100071	Mosaic trisomy 3	6 Cases
1005	Alopecia-contractures-dwarfism-intellectual disability syndrome	5 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
1113	Aphalangy-syndactyly-microcephaly syndrome	5 Cases
1811	Odontomericnychial dysplasia	5 Cases
1657	Dermatoosteolysis, Kirghizian type	5 Cases
1514	Craniodigital-intellectual disability syndrome	5 Cases
1566	Dandy-Walker malformation-postaxial polydactyly syndrome	5 Cases
1562	Dacryocystitis-osteopoikilosis syndrome	5 Cases
2085	Glaucoma-sleep apnea syndrome	5 Cases
2077	German syndrome	5 Cases
2001	Cleft lip/palate-intestinal malrotation-cardiopathy syndrome	5 Cases
2752	Orofaciodigital syndrome type 3	5 Cases
2714	Oculo-palato-cerebral syndrome	5 Cases
2669	Nephrosis-deafness-urinary tract-digital malformations syndrome	5 Cases
2668	Nephropathy-deafness-hyperparathyroidism syndrome	5 Cases
2571	X-linked immunoneurologic disorder	5 Cases
2558	Mikati-Najjar-Sahli syndrome	5 Cases
2496	Mesomelia-synostoses syndrome	5 Cases
3168	Sillence syndrome	5 Cases
3003	Pyknoachondrogenesis	5 Cases
3291	Teebi-Shaltout syndrome	5 Cases
3304	Fallot complex-intellectual disability-growth delay syndrome	5 Cases
3238	Cardiospondylocarpofacial syndrome	5 Cases
1415	Cholestasis-pigmentary retinopathy-cleft palate syndrome	5 Cases
2729	Okamoto syndrome	5 Cases
1129	Arachnodactyly-abnormal ossification-intellectual disability syndrome	5 Cases
65287	Beta-ureidopropionase deficiency	5 Cases
73272	Growth delay due to insulin-like growth factor type 1 deficiency	5 Cases
50815	Branchiogenic deafness syndrome	5 Cases
83472	CAMOS syndrome	5 Cases
75374	Bradyopsia	5 Cases
79095	Congenital bile acid synthesis defect type 4	5 Cases
90646	Deafness-hypogonadism syndrome	5 Cases
90399	Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms	5 Cases
90301	Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome	5 Cases
86821	Lissencephaly type 3-familial fetal akinesia sequence syndrome	5 Cases
86915	Lymphedema-atrial septal defects-facial changes syndrome	5 Cases
86914	Lymphedema-cerebral arteriovenous anomaly syndrome	5 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
85112	Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome	5 Cases
85175	Astley-Kendall dysplasia	5 Cases
85165	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome	5 Cases
85280	X-linked intellectual disability-cubitus valgus-dysmorphism syndrome	5 Cases
85295	HSD10 disease, atypical type	5 Cases
85297	X-linked spinocerebellar ataxia type 3	5 Cases
85284	BRESEK syndrome	5 Cases
97341	Persistent placoid maculopathy	5 Cases
94056	Humero-ulnar synostosis	5 Cases
93975	Renier-Gabreels-Jasper syndrome	5 Cases
93352	Spondyloepimetaphyseal dysplasia, Shohat type	5 Cases
217026	Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type	5 Cases
228390	Frontonasal dysplasia-alopecia-genital anomalies syndrome	5 Cases
228227	Late-onset focal dermal elastosis	5 Cases
228240	Elastodermia	5 Cases
251523	Hyperzinemia and hypercalprotectinemia	5 Cases
156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type	5 Cases
157954	ANE syndrome	5 Cases
168563	46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome	5 Cases
169079	Cernunnos-XLF deficiency	5 Cases
168443	Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome	5 Cases
206580	Autosomal recessive lower motor neuron disease with childhood onset	5 Cases
199337	Pancreatic insufficiency-anemia-hyperostosis syndrome	5 Cases
293925	Lethal occipital encephalocele-skeletal dysplasia syndrome	5 Cases
293964	Hypoinsulinemic hypoglycemia and body hemihypertrophy	5 Cases
293955	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency	5 Cases
293462	Pre-Descemet corneal dystrophy	5 Cases
300504	Onychocytic matricoma	5 Cases
300552	Follicular cholangitis and pancreatitis	5 Cases
300313	Congenital cataract-hearing loss-severe developmental delay syndrome	5 Cases
314555	Craniofacial dysplasia-osteopenia syndrome	5 Cases
314652	Variant ABeta2M amyloidosis	5 Cases
261102	Distal 7q11.23 microduplication syndrome	5 Cases
264200	14q22q23 microdeletion syndrome	5 Cases
280183	Methylmalonic aciduria due to transcobalamin receptor defect	5 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
280403	Familial omphalocele syndrome with facial dysmorphism	5 Cases
397593	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	5 Cases
402364	Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	5 Cases
401986	1p31p32 microdeletion syndrome	5 Cases
98676	Autosomal recessive isolated optic atrophy	5 Cases
363654	X-linked parkinsonism-spasticity syndrome	5 Cases
363665	Acroosteolysis-keloid-like lesions-premature aging syndrome	5 Cases
363611	Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome	5 Cases
363618	LMNA-related cardiocutaneous progeria syndrome	5 Cases
369847	Intellectual disability-hyperkinetic movement-truncal ataxia syndrome	5 Cases
352596	Progressive myoclonic epilepsy with dystonia	5 Cases
353320	Pyruvate carboxylase deficiency, benign type	5 Cases
352718	Progressive retinal dystrophy due to retinol transport defect	5 Cases
391487	Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome	5 Cases
2715	Oculorenocerebellar syndrome	5 Cases
3180	Spondylocamptodactyly syndrome	5 Cases
314034	7p22.1 microduplication syndrome	5 Cases
157962	Oculoauricular syndrome, Schorderet type	5 Cases
2491	Müllerian duct anomalies-limb anomalies syndrome	5 Cases
2819	Spastic paraplegia-facial-cutaneous lesions syndrome	5 Cases
2432	Macrosomia-microphtalmia-cleft palate syndrome	5 Cases
3166	Sialuria	5 Cases
457212	Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	5 Cases
457284	Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome	5 Cases
447737	DOCK2 deficiency	5 Cases
423275	Spinocerebellar ataxia type 40	5 Cases
444002	11q22.2q22.3 microdeletion syndrome	5 Cases
99764	Familial hyperreninemic hypoaldosteronism type 2	5 Cases
445062	Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	5 Cases
480491	MYO5B-related progressive familial intrahepatic cholestasis	5 Cases
481665	USP18 deficiency	5 Cases
3079	Intellectual disability, Buenos-Aires type	5 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
3164	Omphalocele syndrome, Shprintzen-Goldberg type	5 Cases
84132	Desmin-related myopathy with Mallory body-like inclusions	5 Cases
2158	Histidinuria-renal tubular defect syndrome	5 Cases
101076	X-linked Charcot-Marie-Tooth disease type 2	5 Cases
3230	Deafness-oligodontia syndrome	5 Cases
2831	Rhizomelic dysplasia, Patterson-Lowry type	5 Cases
2672	Neuhauser-Eichner-Opitz syndrome	5 Cases
2703	Port-wine nevi-mega cisterna magna-hydrocephalus syndrome	5 Cases
2798	Pachygyria-intellectual disability-epilepsy syndrome	5 Cases
3217	Deafness-small bowel diverticulosis-neuropathy syndrome	5 Cases
319160	Congenital myopathy with internal nuclei and atypical cores	5 Cases
464366	NEK9-related lethal skeletal dysplasia	5 Cases
464440	Primary dystonia, DYT27 type	5 Cases
464756	Familial gastric type 1 neuroendocrine tumor	5 Cases
3383	Humerus trochlea aplasia	5 Cases
2840	Pelvic dysplasia-arthrogryposis of lower limbs syndrome	5 Cases
466695	Supratip dysplasia	5 Cases
319519	Combined oxidative phosphorylation defect type 14	5 Cases
320360	MT-ATP6-related mitochondrial spastic paraplegia	5 Cases
320385	Hereditary sensory and autonomic neuropathy due to TECPR2 mutation	5 Cases
320391	Autosomal recessive spastic paraplegia type 46	5 Cases
459056	Autosomal recessive spastic paraplegia type 75	5 Cases
255235	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy	5 Cases
500062	Infantile-onset periodic fever-panniculitis-dermatosis syndrome	5 Cases
505227	Combined immunodeficiency due to GINS1 deficiency	5 Cases
488232	Split-foot malformation-mesoaxial polydactyly syndrome	5 Cases
488168	Microcephaly-congenital cataract-psoriasisform dermatitis syndrome	5 Cases
488635	Early-onset epilepsy-intellectual disability-brain anomalies syndrome	5 Cases
488618	Transketolase deficiency	5 Cases
498251	Menstrual cycle-dependent periodic fever	5 Cases
498700	Limbic encephalitis with neurexin-3 antibodies	5 Cases
488434	Camptodactyly syndrome, Guadalajara type 3	5 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
527450	Severe myopia-generalized joint laxity-short stature syndrome	5 Cases
508542	Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome	5 Cases
1538	Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome	4 Cases
1135	Arrhinia-choanal atresia-microphthalmia syndrome	4 Cases
2431	Central bilateral macrogryria	4 Cases
921	Abruzzo-Erickson syndrome	4 Cases
1240	Metaphyseal acroscyphodysplasia	4 Cases
1261	Bonnemann-Meinecke-Reich syndrome	4 Cases
1094	Anonychia-microcephaly syndrome	4 Cases
1110	Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome	4 Cases
1117	Aplasia cutis-myopia syndrome	4 Cases
1816	Leukomelanoderma-infantilism-intellectual disability-hypodontia-hypotrichosis syndrome	4 Cases
1809	Hidrotic ectodermal dysplasia, Halal type	4 Cases
1952	Pacman dysplasia	4 Cases
1682	Arterial dissection-lentiginosis syndrome	4 Cases
1508	Coxoauricular syndrome	4 Cases
1436	X-linked skeletal dysplasia-intellectual disability syndrome	4 Cases
1435	Choroideremia-deafness-obesity syndrome	4 Cases
2269	Ichthyosis-aloppecia-eclabion-ectropion-intellectual disability syndrome	4 Cases
2150	Hirschsprung disease-type D brachydactyly syndrome	4 Cases
2215	Multiple pterygium-malignant hyperthermia syndrome	4 Cases
2218	Cervical hypertrichosis-peripheral neuropathy syndrome	4 Cases
1973	Faciocardiorenal syndrome	4 Cases
2723	Odontotrichomelic syndrome	4 Cases
2676	Neuroectodermal-endocrine syndrome	4 Cases
2589	Myoclonus-cerebellar ataxia-deafness syndrome	4 Cases
2386	Leukoencephalopathy-palmoplantar keratoderma syndrome	4 Cases
3088	Revesz syndrome	4 Cases
3044	Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome	4 Cases
3052	X-linked intellectual disability-seizures-psoriasis syndrome	4 Cases
2972	Non-eruption of teeth-maxillary hypoplasia-genu valgum syndrome	4 Cases
2946	Brachydactyly-long thumb syndrome	4 Cases
2865	Short stature-webbed neck-heart disease syndrome	4 Cases
2820	Spastic paraparesis-nephritis-deafness syndrome	4 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
3355	Trichodontoonychial dysplasia	4 Cases
1884	Ectopia lentis-chorioretinal dystrophy-myopia syndrome	4 Cases
1768	Familial caudal dysgenesis	4 Cases
1423	Lethal recessive chondrodysplasia	4 Cases
806	Scott syndrome	4 Cases
1787	Acrofacial dysostosis, Palagonia type	4 Cases
69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	4 Cases
65743	Autosomal dominant multiple pterygium syndrome	4 Cases
65288	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome	4 Cases
46059	Lathosterolosis	4 Cases
79132	Sparse hair-short stature-skin anomalies syndrome	4 Cases
75391	Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency	4 Cases
77295	Odontoleukodystrophy	4 Cases
93333	Pelviscapular dysplasia	4 Cases
90023	Primary immunodeficiency syndrome due to p14 deficiency	4 Cases
85330	X-linked intellectual disability-corpus callosum agenesis-spastic quadripareisis syndrome	4 Cases
85326	X-linked intellectual disability, Stoll type	4 Cases
85325	X-linked intellectual disability, Stevenson type	4 Cases
85323	X-linked intellectual disability, Seemanova type	4 Cases
88635	Vacuolar myopathy with sarcoplasmic reticulum protein aggregates	4 Cases
88618	Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	4 Cases
83629	Leukoencephalopathy-metaphyseal chondrodysplasia syndrome	4 Cases
85172	Microcephalic osteodysplastic dysplasia, Saul-Wilson type	4 Cases
85186	Endosteal sclerosis-cerebellar hypoplasia syndrome	4 Cases
85283	X-linked intellectual disability, Miles-Carpenter type	4 Cases
85285	X-linked intellectual disability, Schimke type	4 Cases
93946	Hamel cerebro-palato-cardiac syndrome	4 Cases
93405	Syndactyly type 4	4 Cases
139573	Hereditary sensory and autonomic neuropathy with deafness and global delay	4 Cases
137908	Hypotonia with lactic acidemia and hyperammonemia	4 Cases
137911	Autism-facial port-wine stain syndrome	4 Cases
137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency	4 Cases
217396	Progressive polyneuropathy with bilateral striatal necrosis	4 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
217407	Hereditary hypotrichosis with recurrent skin vesicles	4 Cases
210163	Congenital lethal myopathy, Compton-North type	4 Cases
210136	Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome	4 Cases
228374	Charcot-Marie-Tooth disease type 2B5	4 Cases
228399	8q12 microduplication syndrome	4 Cases
247604	Juvenile primary lateral sclerosis	4 Cases
251304	Infantile onset panniculitis with uveitis and systemic granulomatosis	4 Cases
251056	6q25 microdeletion syndrome	4 Cases
250972	Polymicrogyria with optic nerve hypoplasia	4 Cases
247827	Ectodermal dysplasia-cutaneous syndactyly syndrome	4 Cases
247790	FTH1-related iron overload	4 Cases
158687	Lethal acantholytic epidermolysis bullosa	4 Cases
163649	Spondyloepiphyseal dysplasia, Nishimura type	4 Cases
163668	Spondyloepiphyseal dysplasia, MacDermot type	4 Cases
163654	Spondyloepiphyseal dysplasia, Cantu type	4 Cases
163971	X-linked intellectual disability, Cilliers type	4 Cases
140976	RHYNS syndrome	4 Cases
166024	Multiple epiphyseal dysplasia, Al-Gazali type	4 Cases
178396	Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation	4 Cases
171703	Microcephaly-polymicrogyria-corpus callosum agenesis syndrome	4 Cases
171844	Blindness-scoliosis-arachnodactyly syndrome	4 Cases
210128	Urocanic aciduria	4 Cases
209967	Episodic ataxia type 6	4 Cases
293825	Congenital dyserythropoietic anemia type IV	4 Cases
293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineuronal hearing loss-dysmorphism syndrome	4 Cases
300501	Painful orbital and systemic neurofibromas-marfanoid habitus syndrome	4 Cases
306550	FADD-related immunodeficiency	4 Cases
313781	20p13 microdeletion syndrome	4 Cases
313795	Jawad syndrome	4 Cases
314381	Hereditary sensory and autonomic neuropathy type 6	4 Cases
314721	Atypical dentin dysplasia due to SMOC2 deficiency	4 Cases
314632	ATP13A2-related juvenile neuronal ceroid lipofuscinosis	4 Cases
324313	9p13 microdeletion syndrome	4 Cases
324581	Benign Samaritan congenital myopathy	4 Cases
352470	DNA2-related mitochondrial DNA deletion syndrome	4 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
329332	Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome	4 Cases
329341	Limbic encephalitis with DPP6 antibodies	4 Cases
263410	Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome	4 Cases
280142	Severe combined immunodeficiency due to LCK deficiency	4 Cases
280598	Hereditary sensorimotor neuropathy with hyperelastic skin	4 Cases
280586	Chondrodysplasia with joint dislocations, gPAPP type	4 Cases
280558	Warsaw breakage syndrome	4 Cases
280654	Autosomal recessive nail dysplasia	4 Cases
284339	Pontocerebellar hypoplasia type 7	4 Cases
397755	Periodic paralysis with transient compartment-like syndrome	4 Cases
397623	Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome	4 Cases
404437	Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	4 Cases
412069	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	4 Cases
401979	Autosomal recessive spondylometaphyseal dysplasia, M��garban�� type	4 Cases
404473	Severe intellectual disability-progressive spastic diplegia syndrome	4 Cases
404466	Female infertility due to zona pellucida defect	4 Cases
401862	Lipoil transferase 1 deficiency	4 Cases
401948	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	4 Cases
401780	Autosomal recessive spastic paraplegia type 61	4 Cases
401810	Autosomal recessive spastic paraplegia type 64	4 Cases
401835	Autosomal recessive spastic paraplegia type 70	4 Cases
1819	Epimetaphyseal skeletal dysplasia	4 Cases
398079	Prader-Willi-like syndrome due to a point mutation	4 Cases
397927	Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome	4 Cases
397951	Microcephaly-thin corpus callosum-intellectual disability syndrome	4 Cases
369942	CADDS	4 Cases
369891	Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	4 Cases
370052	SCALP syndrome	4 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
363686	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	4 Cases
369837	Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	4 Cases
363969	Autosomal recessive cerebral atrophy	4 Cases
363965	Koelen-De Vries syndrome due to a point mutation	4 Cases
357175	Short ulna-dysmorphism-hypotonia-intellectual disability syndrome	4 Cases
356961	SLC35A2-CDG	4 Cases
356947	3q26q27 microdeletion syndrome	4 Cases
363444	THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	4 Cases
352682	Cobblestone lissencephaly without muscular or ocular involvement	4 Cases
352709	CLN13 disease	4 Cases
3270	Radioulnar synostosis-developmental delay-hypotonia syndrome	4 Cases
85184	Craniometadiaphyseal dysplasia, wormian bone type	4 Cases
2536	Microcornea-glaucoma-absent frontal sinuses syndrome	4 Cases
2412	Dislocation of the hip-dysmorphism syndrome	4 Cases
2031	Hepatic fibrosis-renal cysts-intellectual disability syndrome	4 Cases
1296	Lambert syndrome	4 Cases
2497	Upper limb mesomelic dysplasia	4 Cases
2463	Marfanoid habitus-autosomal recessive intellectual disability syndrome	4 Cases
3055	X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome	4 Cases
451612	Familial congenital nasolacrimal duct obstruction	4 Cases
456328	X-linked myotubular myopathy-abnormal genitalia syndrome	4 Cases
1338	Heart defect-tongue hamartoma-polysyndactyly syndrome	4 Cases
457395	Progressive spondyloepimetaphyseal dysplasia-short stature-short fourth metatarsals-intellectual disability syndrome	4 Cases
447784	Mitochondrial pyruvate carrier deficiency	4 Cases
96188	Maternal uniparental disomy of chromosome 22	4 Cases
447893	Hypomyelination-cerebellar atrophy-hypoplasia of the corpus callosum syndrome	4 Cases
420702	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency	4 Cases
438134	PCNA-related progressive neurodegenerative photosensitivity syndrome	4 Cases
435998	Autosomal recessive intermediate Charcot-Marie-Tooth disease type D	4 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
424027	Progressive myoclonic epilepsy type 8	4 Cases
444069	Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome	4 Cases
444138	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	4 Cases
443995	Mandibulofacial dysostosis with alopecia	4 Cases
1528	Craniotelencephalic dysplasia	4 Cases
178400	Distal myopathy with anterior tibial onset	4 Cases
436166	Periodic fever-infantile enterocolitis-autoinflammatory syndrome	4 Cases
435660	LIPE-related familial partial lipodystrophy	4 Cases
438114	RARS-related autosomal recessive hypomyelinating leukodystrophy	4 Cases
480536	MSH3-related attenuated familial adenomatous polyposis	4 Cases
478049	Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome	4 Cases
480556	Isolated neonatal sclerosing cholangitis	4 Cases
480682	Autosomal recessive limb-girdle muscular dystrophy type 2Z	4 Cases
480476	Progressive familial intrahepatic cholestasis type 5	4 Cases
3207	White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome	4 Cases
3015	Radio-renal syndrome	4 Cases
3232	Deafness-ear malformation-facial palsy syndrome	4 Cases
364577	Intellectual disability-brachydactyly-Pierre Robin syndrome	4 Cases
476126	Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	4 Cases
2278	Ichthyosis-intellectual disability-dwarfism-renal impairment syndrome	4 Cases
2769	Familial osteodysplasia, Anderson type	4 Cases
2730	Postaxial tetramelic oligodactyly	4 Cases
3035	Growth delay-hydrocephaly-lung hypoplasia syndrome	4 Cases
2570	Lethal intrauterine growth restriction-cortical malformation-congenital contractures syndrome	4 Cases
1794	Oculomaxillofacial dysostosis	4 Cases
165805	Familial mesial temporal lobe epilepsy with febrile seizures	4 Cases
3186	Holoprosencephaly-radial heart renal anomalies syndrome	4 Cases
2878	Phocomelia-ectrodactyly-deafness-sinus arrhythmia syndrome	4 Cases
3133	Say-Field-Coldwell syndrome	4 Cases
3101	Richieri Costa-da Silva syndrome	4 Cases
468717	Rhizomelic chondrodysplasia punctata type 5	4 Cases
466926	Seizures-scoliosis-macrocephaly syndrome	4 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
319509	Combined oxidative phosphorylation defect type 9	4 Cases
459074	Corpus callosum agenesis-macrocephaly-hypertelorism syndrome	4 Cases
1323	Camptodactyly-joint contractures-facial skeletal defects syndrome	4 Cases
500095	Tall stature-intellectual disability-renal anomalies syndrome	4 Cases
500159	Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrome	4 Cases
500188	X-linked external auditory canal atresia-dilated internal auditory canal-facial dysmorphism syndrome	4 Cases
506353	Autosomal recessive complex spastic paraparesis due to Kennedy pathway dysfunction	4 Cases
505216	3-methylglutaconic aciduria type 9	4 Cases
485421	MFF-related encephalopathy due to mitochondrial and peroxisomal fission defect	4 Cases
495818	9q33.3q34.11 microdeletion syndrome	4 Cases
498488	Overgrowth syndrome with 2q37 translocation	4 Cases
498485	Overgrowth-metaphyseal undermodeling-spondylar dysplasia syndrome	4 Cases
498693	MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome	4 Cases
497906	Childhood-onset basal ganglia degeneration syndrome	4 Cases
363705	Craniofaciofrontodigital syndrome	4 Cases
486815	Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome	4 Cases
2838	Renal caliceal diverticuli-deafness syndrome	4 Cases
1406	Charlie M syndrome	4 Cases
319195	Chondroectodermal dysplasia with night blindness	4 Cases
96192	Paternal uniparental disomy of chromosome 7	4 Cases
521305	Proximal myopathy with focal depletion of mitochondria	4 Cases
521390	Spastic paraparesis-intellectual disability-nystagmus-obesity syndrome	4 Cases
521438	Congenital vertebral-cardiac-renal anomalies syndrome	4 Cases
2772	Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome	3 Cases
2135	Hennekam-Beemer syndrome	3 Cases
2983	Disorder of sex development-intellectual disability syndrome	3 Cases
1943	Early-onset progressive encephalopathy with migrant continuous myoclonus	3 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
1259	Blepharoptosis-myopia-ectopia lentis syndrome	3 Cases
1321	Camptodactyly-fibrous tissue hyperplasia-skeletal dysplasia syndrome	3 Cases
1342	Heart-hand syndrome type 3	3 Cases
1027	Autosomal recessive amelia	3 Cases
1067	Aniridia-ptosis-intellectual disability-familial obesity syndrome	3 Cases
1133	AREDYLD syndrome	3 Cases
1069	Aniridia-absent patella syndrome	3 Cases
1112	Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome	3 Cases
1116	Aplasia cutis congenita-intestinal lymphangiectasia syndrome	3 Cases
1882	Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome	3 Cases
1790	Hypomandibular faciocranial dysostosis	3 Cases
1495	Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome	3 Cases
1529	Craniofacial-deafness-hand syndrome	3 Cases
1408	Hair defect-photosensitivity-intellectual disability syndrome	3 Cases
1389	Cortical blindness-intellectual disability-polydactyly syndrome	3 Cases
2153	Hirschsprung disease-nail hypoplasia-dysmorphism syndrome	3 Cases
2064	Posterior fusion of lumbosacral vertebrae-blepharoptosis syndrome	3 Cases
2050	Cole-Carpenter syndrome	3 Cases
2091	Multinodular goiter-cystic kidney-polydactyly syndrome	3 Cases
2084	Glaucoma-ectopia lentis-microspherophakia-stiff joints-short stature syndrome	3 Cases
2111	Cystic hamartoma of lung and kidney	3 Cases
1972	Lethal faciocardiomelic dysplasia	3 Cases
1970	Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome	3 Cases
2736	Lethal omphalocele-cleft palate syndrome	3 Cases
2713	Oculoosteocutaneous syndrome	3 Cases
2613	Nail-patella-like renal disease	3 Cases
2608	N syndrome	3 Cases
2515	Microcephaly-cardiomyopathy syndrome	3 Cases
2521	Microcephaly-cleft palate-abnormal retinal pigmentation syndrome	3 Cases
2516	Microcephaly-cardiac defect-lung malsegmentation syndrome	3 Cases
2437	Czeizel-Losonci syndrome	3 Cases
2410	Hypergonadotropic hypogonadism-cataract syndrome	3 Cases
2409	Lowry-MacLean syndrome	3 Cases
3172	Eyebrow duplication-syndactyly syndrome	3 Cases
3210	Summitt syndrome	3 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
3086	Autosomal dominant vitreoretinochoroidopathy	3 Cases
3010	Qazi-Markouizos syndrome	3 Cases
3018	Retinal ischemic syndrome-digestive tract small vessel hyalinosis-diffuse cerebral calcifications syndrome	3 Cases
3041	Intellectual disability-balding-patella luxation-acromicria syndrome	3 Cases
2951	Absent thumb-short stature-immunodeficiency syndrome	3 Cases
2881	Cutaneous photosensitivity-lethal colitis syndrome	3 Cases
3326	Thymic-renal-anal-lung dysplasia	3 Cases
3328	Absent tibia-polydactyly-arachnoid cyst syndrome	3 Cases
3404	Ulbright-Hodes syndrome	3 Cases
3369	Trigonocephaly-short stature-developmental delay syndrome	3 Cases
3433	Microcephaly-brachydactyly-kyphoscoliosis syndrome	3 Cases
1101	Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome	3 Cases
1383	Cataract-deafness-hypogonadism syndrome	3 Cases
1130	Arachnodactyly-intellectual disability-dysmorphism syndrome	3 Cases
629	Short stature due to growth hormone qualitative anomaly	3 Cases
69125	Anonychia with flexural pigmentation	3 Cases
66633	Sensorineural hearing loss-early graying-essential tremor syndrome	3 Cases
65798	Goodman syndrome	3 Cases
71278	Congenital brain dysgenesis due to glutamine synthetase deficiency	3 Cases
73223	Global developmental delay-osteopenia-ectodermal defect syndrome	3 Cases
50811	Lipodystrophy-intellectual disability-deafness syndrome	3 Cases
52054	Craniosynostosis-intracranial calcifications syndrome	3 Cases
83617	Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	3 Cases
79330	MOGS-CDG	3 Cases
75325	Osteosclerosis-ichthyosis-premature ovarian failure syndrome	3 Cases
79118	Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome	3 Cases
77299	Microphthalmia-brain atrophy syndrome	3 Cases
93267	Cloverleaf skull-multiple congenital anomalies syndrome	3 Cases
90030	Hemolytic anemia due to glutathione reductase deficiency	3 Cases
85324	X-linked intellectual disability, Shrimpton type	3 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
85321	Deafness-intellectual disability syndrome, Martin-Probst type	3 Cases
83642	Microcytic anemia with liver iron overload	3 Cases
85317	X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome	3 Cases
85290	X-linked intellectual disability, Wilson type	3 Cases
94095	Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome	3 Cases
93970	Holmes-Gang syndrome	3 Cases
93971	Chudley-Lowry-Hoar syndrome	3 Cases
93947	X-linked intellectual disability, Golabi-Ito-Hall type	3 Cases
139414	Congenital panfollicular nevus	3 Cases
139466	SERKAL syndrome	3 Cases
103910	Congenital enterocyte heparan sulfate deficiency	3 Cases
137622	Intractable diarrhea-choanal atresia-eye anomalies syndrome	3 Cases
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency	3 Cases
217017	Zechi-Ceide syndrome	3 Cases
230845	Ehlers-Danlos syndrome, vascular-like type	3 Cases
228396	Ptosis-upper ocular movement limitation-absence of lacrimal punctum syndrome	3 Cases
238459	SLC35A1-CDG	3 Cases
251066	8p11.2 deletion syndrome	3 Cases
163665	Spondyloepiphyseal dysplasia tarda, Kohn type	3 Cases
163961	X-linked cerebral-cerebellar-coloboma syndrome	3 Cases
168577	Hereditary cryohydrocytosis with reduced stomatin	3 Cases
168555	Spondylometaphyseal dysplasia, A4 type	3 Cases
166068	Pontocerebellar hypoplasia type 5	3 Cases
166277	Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia	3 Cases
166105	FASTKD2-related infantile mitochondrial encephalomyopathy	3 Cases
168544	Spondylometaphyseal dysplasia, Golden type	3 Cases
171860	Intellectual disability-cataracts-kyphosis syndrome	3 Cases
171866	Spondyloepimetaphyseal dysplasia, aggrecan type	3 Cases
300373	Familial infantile gigantism	3 Cases
300298	Severe congenital hypochromic anemia with ringed sideroblasts	3 Cases
294023	Neonatal inflammatory skin and bowel disease	3 Cases
300333	Nephrotic syndrome-deafness-pretribial epidermolysis bullosa syndrome	3 Cases
313800	Optic nerve edema-splenomegaly syndrome	3 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
306504	Junctional epidermolysis bullosa with respiratory and renal involvement	3 Cases
306542	Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome	3 Cases
314389	Xq12-q13.3 duplication syndrome	3 Cases
314485	Young adult-onset distal hereditary motor neuropathy	3 Cases
314572	Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome	3 Cases
324525	Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation	3 Cases
324290	Early-onset Lafora body disease	3 Cases
329173	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis	3 Cases
329178	Congenital muscular dystrophy with intellectual disability and severe epilepsy	3 Cases
324999	JMP syndrome	3 Cases
330054	Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome	3 Cases
261120	14q11.2 microdeletion syndrome	3 Cases
261144	14q12 microdeletion syndrome	3 Cases
261295	20p12.3 microdeletion syndrome	3 Cases
263508	COG1-CDG	3 Cases
280356	PLIN1-related familial partial lipodystrophy	3 Cases
280640	Occipital pachygryria and polymicrogyria	3 Cases
412035	13q12.3 microdeletion syndrome	3 Cases
411986	Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	3 Cases
412189	Epidermolysis bullosa simplex due to exophilin 5 deficiency	3 Cases
404493	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency	3 Cases
402082	Progressive myoclonic epilepsy type 5	3 Cases
404451	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome	3 Cases
401859	Lipoic acid synthetase deficiency	3 Cases
401866	Childhood-onset spasticity with hyperglycinemia	3 Cases
401935	14q24.1q24.3 microdeletion syndrome	3 Cases
401764	Pancytopenia-developmental delay syndrome	3 Cases
401795	Autosomal recessive spastic paraparesis type 59	3 Cases
397933	Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	3 Cases
397922	Ferro-cerebro-cutaneous syndrome	3 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
397964	Combined immunodeficiency due to MALT1 deficiency	3 Cases
369992	Severe dermatitis-multiple allergies-metabolic wasting syndrome	3 Cases
370127	Medich giant platelet syndrome	3 Cases
370010	Intellectual disability-facial dysmorphism-hand anomalies syndrome	3 Cases
369867	Autosomal recessive intermediate Charcot-Marie-Tooth disease type C	3 Cases
369840	Autosomal recessive limb-girdle muscular dystrophy type 2S	3 Cases
363981	Charcot-Marie-Tooth disease type 4B3	3 Cases
357237	Severe combined immunodeficiency due to CARD11 deficiency	3 Cases
356996	ANK3-related intellectual disability-sleep disturbance syndrome	3 Cases
363534	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form	3 Cases
363409	Fetal akinesia-cerebral and retinal hemorrhage syndrome	3 Cases
370938	Salt-and-pepper syndrome	3 Cases
391307	Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome	3 Cases
391316	Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression	3 Cases
391351	SURF1-related Charcot-Marie-Tooth disease type 4	3 Cases
391397	Hereditary sensory and autonomic neuropathy type 7	3 Cases
391457	HSD10 disease, neonatal type	3 Cases
2760	OSLAM syndrome	3 Cases
1006	Alopecia antibody deficiency	3 Cases
2617	Microcephalic primordial dwarfism, Montreal type	3 Cases
2535	Microcornea-corectopia-macular hypoplasia syndrome	3 Cases
2957	Guttmacher syndrome	3 Cases
2724	Odontomatosis-aortae esophagus stenosis syndrome	3 Cases
2741	Ophthalmomandibulomelic dysplasia	3 Cases
2101	Grubben-de Cock-Borghgraef syndrome	3 Cases
2115	Harrod syndrome	3 Cases
2502	Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome	3 Cases
3409	Urban-Rogers-Meyer syndrome	3 Cases
2523	Microcephaly-brain defect-spasticity-hypernatremia syndrome	3 Cases
2261	Hypospadias-intellectual disability, Goldblatt type syndrome	3 Cases
2246	Cerebellar hypoplasia-tapetoretinal degeneration syndrome	3 Cases
2167	Holzgreve syndrome	3 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
2476	Dysraphism-cleft lip/palate-limb reduction defects syndrome	3 Cases
453510	Congenital insensitivity to pain with severe intellectual disability	3 Cases
457365	Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome	3 Cases
320401	Autosomal recessive spastic paraparesis type 44	3 Cases
97678	Maternal uniparental disomy of chromosome 13	3 Cases
370103	Primary dystonia, DYT17 type	3 Cases
453533	Polyendocrine-polyneuropathy syndrome	3 Cases
2690	Neutropenia-monocytopenia-deafness syndrome	3 Cases
420566	Bleeding disorder due to CalDAG-GEFI deficiency	3 Cases
420794	Cono-spondylar dysplasia	3 Cases
424261	Autosomal recessive limb-girdle muscular dystrophy type 2Y	3 Cases
436245	Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome	3 Cases
423894	Microcephaly-complex motor and sensory axonal neuropathy syndrome	3 Cases
398117	Neonatal dermatomyositis	3 Cases
444048	46,XX ovarian dysgenesis-short stature syndrome	3 Cases
444458	Combined oxidative phosphorylation defect type 24	3 Cases
444077	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	3 Cases
1891	Intellectual disability-spasticity-ectrodactyly syndrome	3 Cases
436174	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	3 Cases
435930	Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	3 Cases
435938	X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	3 Cases
435953	Progeroid features-hepatocellular carcinoma predisposition syndrome	3 Cases
438178	Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency	3 Cases
437552	Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity	3 Cases
166029	Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia	3 Cases
477993	Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	3 Cases
79347	Chondrodysplasia punctata, Toriello type	3 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
477774	Combined oxidative phosphorylation defect type 27	3 Cases
477661	IL21-related infantile inflammatory bowel disease	3 Cases
1837	Ulna metaphyseal dysplasia syndrome	3 Cases
1185	Spinocerebellar ataxia-dysmorphism syndrome	3 Cases
169157	T-B+ severe combined immunodeficiency due to CD45 deficiency	3 Cases
476084	Autosomal recessive limb-girdle muscular dystrophy type 2X	3 Cases
476096	Erythrokeratoderma-cardiomyopathy syndrome	3 Cases
3236	Conductive deafness-ptosis-skeletal anomalies syndrome	3 Cases
1849	Infundibulopelvic stenosis-multicystic kidney syndrome	3 Cases
2673	Neurofaciodigitorenal syndrome	3 Cases
2779	Osteopathia striata-pigmentary dermopathy-white forelock syndrome	3 Cases
3419	Van Regemorter-Pierquin-Vamos syndrome	3 Cases
1969	Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome	3 Cases
2370	Larsen-like osseous dysplasia-short stature syndrome	3 Cases
1548	Cryptorchidism-arachnodactyl-intellectual disability syndrome	3 Cases
1381	Cataract-intellectual disability-anal atresia-urinary defects syndrome	3 Cases
1355	Congenital heart defect-round face-developmental delay syndrome	3 Cases
1373	Cataract-aberrant oral frenula-growth delay syndrome	3 Cases
435628	Keppen-Lubinsky syndrome	3 Cases
3353	Trichodermodysplasia-dental alterations syndrome	3 Cases
2916	Postaxial polydactyly-dental and vertebral anomalies syndrome	3 Cases
2928	Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome	3 Cases
2926	Digital extensor muscle aplasia-polyneuropathy	3 Cases
2868	Short stature-valvular heart disease-characteristic facies syndrome	3 Cases
2863	Short stature-wormian bones-dextrocardia syndrome	3 Cases
3098	Rhizomelic syndrome, Urbach type	3 Cases
3104	Robin sequence-oligodactyly syndrome	3 Cases
466784	Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect	3 Cases
466794	Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome	3 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
468620	Intellectual disability-epilepsy-extrapyramidal syndrome	3 Cases
500144	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	3 Cases
500135	Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome	3 Cases
502437	4q25 proximal deletion syndrome	3 Cases
504523	Severe combined immunodeficiency due to LAT deficiency	3 Cases
508501	Oral-facial-digital syndrome with short stature and brachymesophalangy	3 Cases
485418	EMILIN-1-related connective tissue disease	3 Cases
494348	Early-onset familial noncirrhotic portal hypertension	3 Cases
495875	Congenital labioscrotaal agenesis-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome	3 Cases
494439	Retinitis pigmentosa-hearing loss-premature aging-short stature-facial dysmorphism syndrome	3 Cases
494541	Childhood-onset benign chorea with striatal involvement	3 Cases
496751	EVEN-plus syndrome	3 Cases
496686	Kyphosis-lateral tongue atrophy-myofibrillar myopathy syndrome	3 Cases
485405	16p12.1p12.3 triplication syndrome	3 Cases
309111	Combined pancreatic lipase-colipase deficiency	3 Cases
95700	Familial adrenal hypoplasia with absent pituitary luteinizing hormone	3 Cases
3026	Radial ray hypoplasia-choanal atresia syndrome	3 Cases
488627	Severe growth deficiency-strabismus-extensive dermal melanocytosis-intellectual disability syndrome	3 Cases
2773	Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome	2 Cases
1488	Cooper-Jabs syndrome	2 Cases
1770	XY type gonadal dysgenesis-associated anomalies syndrome	2 Cases
1046	Lethal hemolytic anemia-genital anomalies syndrome	2 Cases
2233	Hypogonadism-mitral valve prolapse-intellectual disability syndrome	2 Cases
1933	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	2 Cases
1422	Chondrodysplasia-disorder of sex development syndrome	2 Cases
3151	Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome	2 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
1948	Epilepsy-microcephaly-skeletal dysplasia syndrome	2 Cases
1354	Heart defects-limb shortening syndrome	2 Cases
949	Acrocraniofacial dysostosis	2 Cases
1227	Bangstad syndrome	2 Cases
1237	Beemer-Ertbruggen syndrome	2 Cases
1326	Camptodactyly syndrome, Guadalajara type 2	2 Cases
1295	Brachytelephalangy-dysmorphism-Kallmann syndrome	2 Cases
1021	Amaurosis-hypertrichosis syndrome	2 Cases
1068	Aniridia-intellectual disability syndrome	2 Cases
1064	Aniridia-renal agenesis-psychomotor retardation syndrome	2 Cases
1003	Scalp defects-postaxial polydactyly syndrome	2 Cases
1014	Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome	2 Cases
1659	Dermatoleukodystrophy	2 Cases
1806	Ectodermal dysplasia-blindness syndrome	2 Cases
1563	Dahlberg-Borer-Newcomer syndrome	2 Cases
1547	Cryptomicrotia-brachydactyly-excess fingertip arch syndrome	2 Cases
1533	Craniosynostosis-fibular aplasia syndrome	2 Cases
1521	Craniofrontonasal dysplasia-Poland anomaly syndrome	2 Cases
1380	Cataract-nephropathy-encephalopathy syndrome	2 Cases
1368	Cataract-ataxia-deafness syndrome	2 Cases
1484	Contractures-ectodermal dysplasia-cleft lip/palate syndrome	2 Cases
1453	Cleidorrhomelic syndrome	2 Cases
1433	Choroidal atrophy-alopecia syndrome	2 Cases
2235	Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome	2 Cases
2234	Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome	2 Cases
2250	Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome	2 Cases
2249	Ulna hypoplasia-intellectual disability syndrome	2 Cases
2266	Hypotrichosis-intellectual disability, Lopes type	2 Cases
2272	Ichthyosis-oral and digital anomalies syndrome	2 Cases
2271	Congenital ichthyosis-microcephalus-tetraplegia syndrome	2 Cases
2274	Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome	2 Cases
2282	Dysmorphism-short stature-deafness-disorder of sex development syndrome	2 Cases
2155	Hirschsprung disease-deafness-polydactyly syndrome	2 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
2172	Microcephaly-glomerulonephritis-marfanoid habitus syndrome	2 Cases
2181	Hydrocephaly-tall stature-joint laxity syndrome	2 Cases
2119	HEC syndrome	2 Cases
1995	Cleft lip-retinopathy syndrome	2 Cases
2007	Alar cartilages hypoplasia-coloboma-telecanthus syndrome	2 Cases
2010	Cleft palate-stapes fixation-oligodontia syndrome	2 Cases
2025	Gingival fibromatosis-facial dysmorphism syndrome	2 Cases
2718	Oculotrichodysplasia	2 Cases
2513	Microcephaly-albinism-digital anomalies syndrome	2 Cases
2511	Microbrachycephaly-ptosis-cleft lip syndrome	2 Cases
2390	Lichtenstein syndrome	2 Cases
2347	Lethal Kniest-like dysplasia	2 Cases
2324	Osteopenia-intellectual disability-sparse hair syndrome	2 Cases
3177	Spinocerebellar degeneration-corneal dystrophy syndrome	2 Cases
3199	Stimmller syndrome	2 Cases
3214	Deaf blind hypopigmentation syndrome, Yemenite type	2 Cases
3105	Robinow-like syndrome	2 Cases
3132	Say-Barber-Miller syndrome	2 Cases
3134	SCARF syndrome	2 Cases
3011	Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome	2 Cases
2975	46,XX disorder of sex development-skeletal anomalies syndrome	2 Cases
2988	Pterygium colli-intellectual disability-digital anomalies syndrome	2 Cases
2985	Pseudoprogeria syndrome	2 Cases
2888	Pierre Robin syndrome-faciocutaneous anomaly syndrome	2 Cases
2867	Short stature, Brussels type	2 Cases
2876	PHAVER syndrome	2 Cases
2892	Pilodental dysplasia-refractive errors syndrome	2 Cases
2825	PARC syndrome	2 Cases
2826	Spastic paraparesis-precocious puberty syndrome	2 Cases
3262	Dobrow syndrome	2 Cases
3323	Thrombocytopenia-Robin sequence syndrome	2 Cases
3327	Thyrocerebrorenal syndrome	2 Cases
3224	Deafness-genital anomalies-metacarpal and metatarsal synostosis syndrome	2 Cases
3239	Deafness-vitiligo-achalasia syndrome	2 Cases
3365	Trigonocephaly-broad thumbs syndrome	2 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
3368	Trigonocephaly-bifid nose-acral anomalies syndrome	2 Cases
3200	Arthrogryposis-ectodermal dysplasia-other anomalies syndrome	2 Cases
3167	Sieger-Brewer-Carey syndrome	2 Cases
2519	Microcephaly-seizures-intellectual disability-heart disease syndrome	2 Cases
2653	Osteochondrodysplastic nanism-deafness-retinitis pigmentosa syndrome	2 Cases
2666	Adult familial nephronophthisis-spastic quadripareis syndrome	2 Cases
3448	Weaver-Williams syndrome	2 Cases
1485	Arthrogryposis-hyperkeratosis syndrome, lethal form	2 Cases
1192	Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome	2 Cases
2015	Cleft palate-short stature-vertebral anomalies syndrome	2 Cases
2427	Macrocephaly-short stature-paraplegia syndrome	2 Cases
2898	X-linked intellectual disability-plagiocephaly syndrome	2 Cases
2183	Hydrocephalus-obesity-hypogonadism syndrome	2 Cases
3240	Central nervous system calcification-deafness-tubular acidosis-anemia syndrome	2 Cases
69088	Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome	2 Cases
73224	Tubular renal disease-cardiomyopathy syndrome	2 Cases
73230	Ossification anomalies-psychomotor developmental delay syndrome	2 Cases
73245	Spinal muscular atrophy-Dandy-Walker malformation-cataracts syndrome	2 Cases
73246	Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome	2 Cases
71267	Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome	2 Cases
50812	Zellweger-like syndrome without peroxisomal anomalies	2 Cases
50817	Duane anomaly-myopathy-scoliosis syndrome	2 Cases
50809	Talo-patello-scaphoid osteolysis	2 Cases
50810	Microlissencephaly-micromelia syndrome	2 Cases
52047	Braddock syndrome	2 Cases
52055	Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome	2 Cases
64542	Acrofacial dysostosis, Kennedy-Teebi type	2 Cases
79302	Congenital bile acid synthesis defect type 3	2 Cases
75389	Brain malformation-congenital heart disease-postaxial polydactyly syndrome	2 Cases
79107	Developmental malformations-deafness-dystonia syndrome	2 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
77300	Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome	2 Cases
91133	Osteopenia-myopia-hearing loss-intellectual disability-facial dysmorphism syndrome	2 Cases
91130	Cardiomyopathy-hypotonia-lactic acidosis syndrome	2 Cases
91494	Macular coloboma-cleft palate-hallux valgus syndrome	2 Cases
88643	Obesity-colitis-hypothyroidism-cardiac hypertrophy-developmental delay syndrome	2 Cases
90022	Cardiomyopathy-renal anomalies syndrome	2 Cases
85327	X-linked intellectual disability-acromegaly-hyperactivity syndrome	2 Cases
86822	Lissencephaly type 3-metacarpal bone dysplasia syndrome	2 Cases
85319	X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome	2 Cases
97290	Familial papillary thyroid carcinoma with renal papillary neoplasia	2 Cases
95428	COG8-CDG	2 Cases
94066	Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia	2 Cases
137631	Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome	2 Cases
100013	Lissencephaly with cerebellar hypoplasia type C	2 Cases
99832	Resistance to thyrotropin-releasing hormone syndrome	2 Cases
99069	Univentricular heart with single atrio-ventricular valve	2 Cases
217399	Congenital insensitivity to pain with hyperhidrosis	2 Cases
238523	Atypical hypotonia-cystinuria syndrome	2 Cases
238329	Severe X-linked mitochondrial encephalomyopathy	2 Cases
231556	Late-onset localized junctional epidermolysis bullosa-intellectual disability syndrome	2 Cases
163684	Leukoencephalopathy-dystonia-motor neuropathy syndrome	2 Cases
163985	Hyperekplexia-epilepsy syndrome	2 Cases
168598	Brain demyelination due to methionine adenosyltransferase deficiency	2 Cases
169100	Immunodeficiency due to CD25 deficiency	2 Cases
166038	Metaphyseal chondrodysplasia, Kaitila type	2 Cases
168451	Spondyloepimetaphyseal dysplasia-abnormal dentition syndrome	2 Cases
168552	Spondylometaphyseal dysplasia-bowed forearms-facial dysmorphism syndrome	2 Cases
171839	Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radio-ulnar synostosis syndrome	2 Cases
183707	Neutrophil immunodeficiency syndrome	2 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
199348	Thiamine-responsive encephalopathy	2 Cases
199329	Congenital myopathy, Paradas type	2 Cases
293807	Ketamine-induced biliary dilatation	2 Cases
294026	Syndactyly-nystagmus syndrome due to 2q31.1 microduplication	2 Cases
313772	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	2 Cases
306511	Autosomal recessive spastic paraplegia type 48	2 Cases
313947	2q23.1 microduplication syndrome	2 Cases
314002	Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome	2 Cases
314029	High bone mass osteogenesis imperfecta	2 Cases
314041	Marfanoid habitus-inguinal hernia-advanced bone age syndrome	2 Cases
314629	CLN11 disease	2 Cases
314575	Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome	2 Cases
324530	Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation	2 Cases
324540	Aphonia-deafness-retinal dystrophy-bifid halluces-intellectual disability syndrome	2 Cases
324416	Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome	2 Cases
324410	X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome	2 Cases
324307	Severe lateral tibial bowing with short stature	2 Cases
324299	Multiple paragangliomas associated with polycythemia	2 Cases
324294	T-cell immunodeficiency with epidermolyticus verruciformis	2 Cases
319600	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency	2 Cases
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	2 Cases
329252	Spondylocostal dysostosis-hypospadias-intellectual disability syndrome	2 Cases
329242	Congenital chronic diarrhea with protein-losing enteropathy	2 Cases
329224	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	2 Cases
324575	Hyperinsulinism due to HNF1A deficiency	2 Cases
352333	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome	2 Cases
261304	Paternal 20q13.2q13.3 microdeletion syndrome	2 Cases
261534	49,XXXXY syndrome	2 Cases
263501	COG4-CDG	2 Cases
276556	Hyperinsulinism due to UCP2 deficiency	2 Cases
276405	Hyperbiliverdinemia	2 Cases
280576	Nestor-Guillermo progeria syndrome	2 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
280397	Familial Alzheimer-like prion disease	2 Cases
280663	Hermansky-Pudlak syndrome type 9	2 Cases
281127	Acral self-healing collodion baby	2 Cases
397725	COASY protein-associated neurodegeneration	2 Cases
397735	Autosomal dominant Charcot-Marie-Tooth disease type 2U	2 Cases
231531	Hermansky-Pudlak syndrome type 7	2 Cases
2575	Cystic fibrosis-gastritis-megaloblastic anemia syndrome	2 Cases
411712	Maternal riboflavin deficiency	2 Cases
412181	Epidermolysis bullosa simplex due to BP230 deficiency	2 Cases
404499	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency	2 Cases
404476	Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome	2 Cases
401923	9q31.1q31.3 microdeletion syndrome	2 Cases
401959	Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome	2 Cases
401805	Autosomal recessive spastic paraplegia type 63	2 Cases
401815	Autosomal recessive spastic paraplegia type 66	2 Cases
401820	Autosomal recessive spastic paraplegia type 67	2 Cases
401830	Autosomal recessive spastic paraplegia type 69	2 Cases
397959	TCR-alpha-beta-positive T-cell deficiency	2 Cases
397973	Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome	2 Cases
369979	Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome	2 Cases
369955	Methylmalonic aciduria with homocystinuria, type cblJ	2 Cases
369929	Primary hyperaldosteronism-seizures-neurological abnormalities syndrome	2 Cases
369881	2p21 microdeletion syndrome without cystinuria	2 Cases
370921	STT3A-CDG	2 Cases
370039	Angora hair nevus	2 Cases
370015	Spondyloepimetaphyseal dysplasia, Isidor type	2 Cases
370019	Spondylometaphyseal dysplasia, Czarny-Ratajczak type	2 Cases
363680	2p13.2 microdeletion syndrome	2 Cases
363623	Autosomal recessive limb-girdle muscular dystrophy type 2T	2 Cases
363543	Autosomal recessive limb-girdle muscular dystrophy type 2R	2 Cases
357158	Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome	2 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
363424	Multiple mitochondrial dysfunctions syndrome type 3	2 Cases
352530	Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome	2 Cases
352563	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	2 Cases
370930	XYLT1-CDG	2 Cases
370997	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	2 Cases
391343	Fatal post-viral neurodegenerative disorder	2 Cases
391348	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	2 Cases
391366	Growth retardation-mild developmental delay-chronic hepatitis syndrome	2 Cases
977	Adrenomyodystrophy	2 Cases
2631	Mesomelic dwarfism-cleft palate-camptodactyly syndrome	2 Cases
2643	Microcephalic primordial dwarfism, Toriello type	2 Cases
1780	Thakker-Donnai syndrome	2 Cases
2321	Jung-Wolff-Back-Stahl syndrome	2 Cases
2003	Cleft lip/palate-deafness-sacral lipoma syndrome	2 Cases
2083	Prominent glabella-microcephaly-hypogenitalism syndrome	2 Cases
2074	Gemignani syndrome	2 Cases
2104	Dysmorphism-pectus carinatum-joint laxity syndrome	2 Cases
1184	Ataxia-photosensitivity-short stature syndrome	2 Cases
1028	Amelonychohypohidrotic syndrome	2 Cases
2522	Microcephaly-cervical spine fusion anomalies syndrome	2 Cases
2528	Microcephaly-microcornea syndrome, Seemanova type	2 Cases
2533	Microcephaly-deafness-intellectual disability syndrome	2 Cases
2310	Absence deformity of leg-cataract syndrome	2 Cases
2256	Fibulo-ulnar hypoplasia-renal anomalies syndrome	2 Cases
2475	White forelock with malformations	2 Cases
2482	Melhem-Fahl syndrome	2 Cases
2487	Lower limb malformation-hypospadias syndrome	2 Cases
2489	Upper limb defect-eye and ear abnormalities syndrome	2 Cases
456298	1p35.2 microdeletion syndrome	2 Cases
457205	Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome	2 Cases
457223	Syndromic sensorineural deafness due to combined oxidative phosphorylation defect	2 Cases
457265	Progressive myoclonic epilepsy type 9	2 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
457359	Megalencephaly-severe kyphoscoliosis-overgrowth syndrome	2 Cases
96187	Maternal uniparental disomy of chromosome 21	2 Cases
453521	Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	2 Cases
456312	Infantile multisystem neurologic-endocrine-pancreatic disease	2 Cases
448264	Isolated focal non-epidermolytic palmoplantar keratoderma	2 Cases
447961	Pigmentation defects-palmoplantar keratoderma-skin carcinoma syndrome	2 Cases
448267	Regressive spondylometaphyseal dysplasia	2 Cases
447954	Combined oxidative phosphorylation defect type 25	2 Cases
447974	Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome	2 Cases
453504	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to a point mutation	2 Cases
447731	NIK deficiency	2 Cases
420741	RIDDLE syndrome	2 Cases
420728	Combined oxidative phosphorylation defect type 20	2 Cases
420699	Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency	2 Cases
439232	AApoAIV amyloidosis	2 Cases
420733	Combined oxidative phosphorylation defect type 21	2 Cases
424107	Congenital myopathy with myasthenic-like onset	2 Cases
423306	Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome	2 Cases
423479	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	2 Cases
435819	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation	2 Cases
431329	Autosomal recessive spastic paraparesis type 57	2 Cases
398109	Neonatal autoimmune hemolytic anemia	2 Cases
440713	Isolated sedoheptulokinase deficiency	2 Cases
166016	Multiple epiphyseal dysplasia, Lowry type	2 Cases
1927	Emery-Nelson syndrome	2 Cases
1964	Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome	2 Cases
1968	Flat face-microstomia-ear anomaly syndrome	2 Cases
436182	Microcephalic primordial dwarfism-insulin resistance syndrome	2 Cases
439897	Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome	2 Cases
438207	Severe autosomal recessive macrothrombocytopenia	2 Cases
431361	Progressive encephalopathy with leukodystrophy due to DECR deficiency	2 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
440731	L-ferritin deficiency	2 Cases
443950	DNAJB2-related Charcot-Marie-Tooth disease type 2	2 Cases
445110	Limb-girdle muscular dystrophy due to POMK deficiency	2 Cases
166032	Multiple epiphyseal dysplasia, with miniepiphyses	2 Cases
480528	Lethal hydranencephaly-diaphragmatic hernia syndrome	2 Cases
478042	Combined oxidative phosphorylation defect type 30	2 Cases
137681	Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	2 Cases
1126	Aprosencephaly cerebellar dysgenesis	2 Cases
1236	Severe microbrachycephaly-intellectual disability-atetoid cerebral palsy syndrome	2 Cases
3294	Extensor tendons of finger anomalies	2 Cases
2997	Ptosis-vocal cord paralysis syndrome	2 Cases
3016	Absent radius-anogenital anomalies syndrome	2 Cases
3218	Deafness-epiphyseal dysplasia-short stature syndrome	2 Cases
2956	Acrodysplasia scoliosis	2 Cases
477684	Combined oxidative phosphorylation defect type 26	2 Cases
477831	Skeletal overgrowth-craniofacial dysmorphism-hyperelastic skin-white matter lesions syndrome	2 Cases
477787	Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder	2 Cases
2597	Mitochondrial myopathy-lactic acidosis-deafness syndrome	2 Cases
1670	Chronic diarrhea with villous atrophy	2 Cases
1803	Thoracomelic dysplasia	2 Cases
785	Estrogen resistance syndrome	2 Cases
317425	Severe combined immunodeficiency due to DNA-PKcs deficiency	2 Cases
1352	Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	2 Cases
476406	Congenital generalized hypercontractile muscle stiffness syndrome	2 Cases
1570	Sybrachydactyly of hands and feet	2 Cases
3225	Hearing loss-familial salivary gland insensitivity to aldosterone syndrome	2 Cases
3293	Telecanthus-hypertelorism-strabismus-pes cavus syndrome	2 Cases
3241	Deafness-craniofacial syndrome	2 Cases
1277	Brachydactyly-mesomelia-intellectual disability-heart defects syndrome	2 Cases
2204	Dysplastic cortical hyperostosis	2 Cases
1778	Facial dysmorphism-shawl scrotum-joint laxity syndrome	2 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
2547	Microphthalmia-microtia-fetal akinesia syndrome	2 Cases
2184	Hydrocephaly-low insertion umbilicus syndrome	2 Cases
2058	Fryns-Smeets-Thiry syndrome	2 Cases
3082	Intellectual disability-polydactyly-uncombable hair syndrome	2 Cases
289522	Microtripllication 11q24.1	2 Cases
1123	Caudal appendage-deafness syndrome	2 Cases
465824	Fetal encasement syndrome	2 Cases
314993	Cataract-congenital heart disease-neural tube defect syndrome	2 Cases
1825	Epiphyseal dysplasia-hearing loss-dysmorphism syndrome	2 Cases
2722	Odonto-onychodysplasia-alopecia syndrome	2 Cases
2705	Oculocerebral dysplasia	2 Cases
2720	Oculocerebral hypopigmentation syndrome, Preus type	2 Cases
1217	Spinal atrophy-ophthalmoplegia-pyramidal syndrome	2 Cases
254898	Deafness-encephaloneuropathy-obesity-valvulopathy syndrome	2 Cases
1506	Thin ribs-tubular bones-dysmorphism syndrome	2 Cases
1390	Night blindness-skeletal anomalies-dysmorphism syndrome	2 Cases
2110	Hallux varus-preaxial polysyndactyly syndrome	2 Cases
2109	Hallermann-Streiff-like syndrome	2 Cases
2400	Peripheral motor neuropathy-dysautonomia syndrome	2 Cases
1861	Thoracic dysplasia-hydrocephalus syndrome	2 Cases
3145	Nephrogenic diabetes insipidus-intracranial calcification syndrome	2 Cases
3068	Intellectual disability-myopathy-short stature-endocrine defect syndrome	2 Cases
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar keratoderma syndrome	2 Cases
79507	Hypotonia-failure to thrive-microcephaly syndrome	2 Cases
3429	Verloove Vanhorick-Brubakk syndrome	2 Cases
3424	Velo-facial-skeletal syndrome	2 Cases
2941	Porencephaly-cerebellar hypoplasia-internal malformations syndrome	2 Cases
2976	Pseudoleprechaunism syndrome, Patterson type	2 Cases
2871	Pfeiffer-Palm-Teller syndrome	2 Cases
2891	Pili torti-developmental delay-neurological abnormalities syndrome	2 Cases
2866	Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome	2 Cases
3080	Intellectual disability, Wolff type	2 Cases

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ORPHA Number	Disease or Group of diseases	Number of cases
466801	Autosomal recessive limb-girdle muscular dystrophy type 2W	2 Cases
220448	Macrothrombocytopenia with mitral valve insufficiency	2 Cases
319514	Combined oxidative phosphorylation defect type 13	2 Cases
319675	Microcephalic primordial dwarfism, Dauber type	2 Cases
320370	Autosomal recessive spastic paraplegia type 43	2 Cases
141258	Tessier number 4 facial cleft	2 Cases
206564	Autosomal recessive limb-girdle muscular dystrophy type 2O	2 Cases
459070	X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome	2 Cases
443236	Postural orthostatic tachycardia syndrome due to NET deficiency	2 Cases
254925	Combined oxidative phosphorylation defect type 4	2 Cases
502444	Alkaline ceramidase 3 deficiency	2 Cases
309147	Hyper-beta-alaninemia	2 Cases
309108	Pancreatic colipase deficiency	2 Cases
508512	Congenital multiple café-au-lait macules-increased sister chromatid exchange syndrome	2 Cases
508488	8q24.3 deletion syndrome	2 Cases
255132	Adult-onset autosomal recessive sideroblastic anemia	2 Cases
487796	Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome	2 Cases
498497	Short rib-polydactyly syndrome type 5	2 Cases
221139	Combined immunodeficiency with faciooculoskeletal anomalies	2 Cases
324364	Mixed sclerosing bone dystrophy with extra-skeletal manifestations	2 Cases
300284	Connective tissue disorder due to lysyl hydroxylase-3 deficiency	2 Cases
221142	Confetti-like macular atrophy	2 Cases
3173	Infantile spasms-broad thumbs syndrome	2 Cases
2145	Craniosynostosis, Herrmann-Opitz type	2 Cases
2759	Imperforate oropharynx-costovertebral anomalies syndrome	2 Cases
1937	Eng-Strom syndrome	2 Cases
1883	Ectodermal dysplasia-sensorineural deafness syndrome	2 Cases
2921	Preaxial polydactyly-colobomata-intellectual disability syndrome	2 Cases
487814	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation	2 Cases
228357	CLN9 disease	2 Cases
1434	Choroideremia-hypopituitarism syndrome	2 Cases

ORPHA Number	Disease or Group of diseases	Number of cases
1877	Muscular dystrophy-white matter spongiosis syndrome	2 Cases
3390	Proximal tubulopathy-diabetes mellitus-cerebellar ataxia syndrome	2 Cases
527468	Diaphragmatic hernia-short bowel-asplenia syndrome	2 Cases
521432	Congenital cataract-severe neonatal hepatopathy-global developmental delay syndrome	2 Cases
521411	Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect	2 Cases
2786	Osteoporosis-oculocutaneous hypopigmentation syndrome	1 Case
79326	ALG2-CDG	1 Case
79332	B4GALT1-CDG	1 Case
99849	Glycogen storage disease due to muscle beta-enolase deficiency	1 Case
243343	Dimethylglycine dehydrogenase deficiency	1 Case
254334	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B	1 Case
250977	AICA-ribosiduria	1 Case
300536	DDOST-CDG	1 Case
324422	ALG13-CDG	1 Case
330029	Hypotrichosis-deafness syndrome	1 Case
329942	Transient neonatal multiple acyl-CoA dehydrogenase deficiency	1 Case
263297	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	1 Case
263494	DPM3-CDG	1 Case
280333	Autosomal recessive limb-girdle muscular dystrophy type 2P	1 Case
404521	Spinal muscular atrophy with respiratory distress type 2	1 Case
401800	Autosomal recessive spastic paraplegia type 60	1 Case
401840	Autosomal recessive spastic paraplegia type 71	1 Case
397968	Charcot-Marie-Tooth disease type 2R	1 Case
370924	STT3B-CDG	1 Case
370097	Oculocutaneous albinism type 6	1 Case
448010	CAD-CDG	1 Case
440706	Ribose-5-P isomerase deficiency	1 Case
435934	COG2-CDG	1 Case
431166	Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection	1 Case
431149	Combined immunodeficiency due to OX40 deficiency	1 Case
435651	CIDEC-related familial partial lipodystrophy	1 Case
478029	Combined oxidative phosphorylation defect type 29	1 Case
2601	Myopathy-growth delay-intellectual disability-hypospadias syndrome	1 Case

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ORPHA Number	Disease or Group of diseases	Number of cases
240760	Nijmegen breakage syndrome-like disorder	1 Case
1035	Beta-mercaptopolactate cysteine disulfiduria	1 Case
156156	Lipoatrophy with diabetes, leukomelanodermic papules, liver steatosis, and hypertrophic cardiomyopathy	1 Case
206559	Autosomal recessive limb-girdle muscular dystrophy type 2N	1 Case
2963	Progeroid syndrome, Petty type	1 Case
1894	Ectrodactyly-spina bifida-cardiopathy syndrome	1 Case
319678	Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	1 Case
458798	Spinocerebellar ataxia type 41	1 Case
254920	Combined oxidative phosphorylation defect type 2	1 Case
141327	Orofaciodigital syndrome type 12	1 Case
141330	Orofaciodigital syndrome type 13	1 Case
331190	Immunodeficiency due to ficolin3 deficiency	1 Case
331187	Immunodeficiency due to MASP-2 deficiency	1 Case
528091	Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome	1 Case

## Number of published families

ORPHA Number	Disease or Group of diseases	Number of families
1652	Dent disease	250 Families
1949	Benign familial neonatal epilepsy	100 Families
1416	Familial calcium pyrophosphate deposition	100 Families
89843	Dystrophic epidermolysis bullosa pruriginosa	100 Families
98759	Spinocerebellar ataxia type 17	100 Families
98784	Autosomal dominant nocturnal frontal lobe epilepsy	100 Families
276198	Spinocerebellar ataxia type 36	100 Families
99879	Familial isolated hyperparathyroidism	100 Families
2524	Pontocerebellar hypoplasia type 2	81 Families
757	Pseudohypoaldosteronism type 2	80 Families
60030	Loeys-Dietz syndrome	52 Families
643	Giant axonal neuropathy	50 Families
2526	Microcephaly-lymphedema-chorioretinopathy syndrome	50 Families
98934	Huntington disease-like 2	50 Families
753	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency	50 Families
2670	Pierson syndrome	40 Families
2254	Pontocerebellar hypoplasia type 1	40 Families

ORPHA Number	Disease or Group of diseases	Number of families
79410	Pretibial dystrophic epidermolysis bullosa	40 Families
98762	Spinocerebellar ataxia type 12	40 Families
263548	Peeling skin syndrome type A	40 Families
1106	Microphthalmia with limb anomalies	35 Families
79501	Punctate palmoplantar keratoderma type 1	35 Families
163937	X-linked intellectual disability, Najm type	35 Families
200418	Immunodeficiency with factor I anomaly	35 Families
324442	Autosomal recessive axonal neuropathy with neuromyotonia	33 Families
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	30 Families
2848	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	30 Families
425	Apolipoprotein A-I deficiency	30 Families
3222	Phosphoribosylpyrophosphate synthetase superactivity	30 Families
3237	Multiple synostoses syndrome	30 Families
452	X-linked lissencephaly with abnormal genitalia	30 Families
90026	Primary erythromelalgia	30 Families
98434	Hereditary combined deficiency of vitamin K-dependent clotting factors	30 Families
217012	Spinocerebellar ataxia type 31	30 Families
228305	Carnitine palmitoyl transferase II deficiency, severe infantile form	30 Families
293822	MITF-related melanoma and renal cell carcinoma predisposition syndrome	30 Families
263553	Peeling skin syndrome type B	30 Families
33108	Lethal multiple pterygium syndrome	28 Families
424	Familial hyperthyroidism due to mutations in TSH receptor	28 Families
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	26 Families
85293	X-linked intellectual disability, Cabezas type	24 Families
1065	Aniridia-cerebellar ataxia-intellectual disability syndrome	22 Families
959	Acro-renal-ocular syndrome	20 Families
2229	Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome	20 Families
3203	Overhydrated hereditary stomatocytosis	20 Families
34517	Autosomal dominant limb-girdle muscular dystrophy type 1E	20 Families
98763	Spinocerebellar ataxia type 14	20 Families
97286	Carney-Stratakis syndrome	20 Families
100998	Autosomal dominant spastic paraparesis type 17	20 Families
228308	Carnitine palmitoyl transferase II deficiency, neonatal form	20 Families
99027	Adult-onset autosomal dominant leukodystrophy	20 Families
3202	Dehydrated hereditary stomatocytosis	20 Families

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ORPHA Number	Disease or Group of diseases	Number of families
369913	Combined oxidative phosphorylation defect type 17	20 Families
99791	Dentin dysplasia type II	19 Families
168615	Hereditary persistence of alpha-fetoprotein	19 Families
25980	X-linked myopathy with excessive autophagy	18 Families
93311	Multiple epiphyseal dysplasia type 5	18 Families
90031	Non-spherocytic hemolytic anemia due to hexokinase deficiency	17 Families
276435	Lower motor neuron syndrome with late-adult onset	17 Families
293168	Infantile-onset ascending hereditary spastic paralysis	17 Families
615	Familial atrial myxoma	17 Families
88621	Ichthyosis-prematurity syndrome	16 Families
84090	Fibronectin glomerulopathy	16 Families
2950	Triphalangeal thumb-polysyndactyly syndrome	15 Families
3220	Deafness-enamel hypoplasia-nail defects syndrome	15 Families
2850	Alopecia-intellectual disability syndrome	15 Families
137831	X-linked intellectual disability-cerebellar hypoplasia syndrome	14 Families
1145	Infantile-onset X-linked spinal muscular atrophy	14 Families
495930	Familial monosomy 7 syndrome	14 Families
228277	Familial anetoderma	12 Families
401996	Karyomegalic interstitial nephritis	12 Families
1200	Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome	11 Families
93974	Smith-Fineman-Myers syndrome	11 Families
99955	Charcot-Marie-Tooth disease type 4B1	11 Families
98971	Posterior amorphous corneal dystrophy	11 Families
439218	KCNQ2-related epileptic encephalopathy	11 Families
478664	Hereditary sensory and autonomic neuropathy type 8	11 Families
1276	Brachydactyly-arterial hypertension syndrome	10 Families
1658	Absence of fingerprints-congenital milia syndrome	10 Families
1412	Tarsal-carpal coalition syndrome	10 Families
2238	Familial isolated hypoparathyroidism	10 Families
2198	Palmoplantar keratoderma-esophageal carcinoma syndrome	10 Families
2202	Palmoplantar keratoderma-deafness syndrome	10 Families
2791	Otodontal syndrome	10 Families
3412	VACTERL with hydrocephalus	10 Families
90001	X-linked cone dysfunction syndrome with myopia	10 Families
85279	Syndromic X-linked intellectual disability due to JARID1C mutation	10 Families

ORPHA Number	Disease or Group of diseases	Number of families
140927	Benign familial neonatal-infantile seizures	10 Families
100991	Autosomal dominant spastic paraplegia type 10	10 Families
100989	Autosomal dominant spastic paraplegia type 8	10 Families
100988	Autosomal dominant spastic paraplegia type 6	10 Families
101006	Autosomal recessive spastic paraplegia type 26	10 Families
100996	Autosomal recessive spastic paraplegia type 15	10 Families
158673	Acral dystrophic epidermolysis bullosa	10 Families
158676	Dominant dystrophic epidermolysis bullosa, nails only	10 Families
166063	Pontocerebellar hypoplasia type 4	10 Families
178464	Hereditary myopathy with early respiratory failure	10 Families
412057	Autosomal recessive cerebellar ataxia due to STUB1 deficiency	10 Families
481662	Familial Chilblain lupus	10 Families
1856	Spondyloperipheral dysplasia-short ulna syndrome	10 Families
1062	Hereditary neurocutaneous malformation	9 Families
100008	ACys amyloidosis	9 Families
217266	BNAR syndrome	9 Families
263516	Progressive myoclonic epilepsy type 3	9 Families
1377	Cataract-microcornea syndrome	8 Families
3248	Distal symphalangism	8 Families
66629	Goldberg-Shprintzen megacolon syndrome	8 Families
217055	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	8 Families
306527	Isolated hereditary congenital facial paralysis	8 Families
391330	X-linked osteoporosis with fractures	8 Families
1149	Kuskokwim syndrome	8 Families
1897	EEM syndrome	7 Families
1777	Temptamy syndrome	7 Families
86817	Hemolytic anemia due to adenylate kinase deficiency	7 Families
251274	Familial hyperaldosteronism type III	7 Families
178461	X-linked myopathy with postural muscle atrophy	7 Families
300345	Autosomal systemic lupus erythematosus	7 Families
281139	Annular epidermolytic ichthyosis	7 Families
397618	Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome	7 Families
93561	ALys amyloidosis	7 Families
324713	ABeta amyloidosis, Italian type	7 Families
324737	SRD5A3-CDG	7 Families
320396	Autosomal recessive spastic paraplegia type 45	7 Families

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ORPHA Number	Disease or Group of diseases	Number of families
488594	Autosomal recessive spastic paraplegia type 76	7 Families
1799	Familial developmental dysphasia	6 Families
34516	Autosomal dominant limb-girdle muscular dystrophy type 1D	6 Families
73229	HANAC syndrome	6 Families
79401	Epidermolysis bullosa simplex, Ogna type	6 Families
85453	X-linked reticulate pigmentary disorder	6 Families
85110	Familial encephalopathy with neuroserpin inclusion bodies	6 Families
140917	Stapes ankylosis with broad thumbs and toes	6 Families
137634	Overgrowth-macrocephaly-facial dysmorphism syndrome	6 Families
101068	Congenital stromal corneal dystrophy	6 Families
157801	Mesoaxial synostotic syndactyly with phalangeal reduction	6 Families
168454	Spondyloepiphyseal dysplasia, Geneviève type	6 Families
324561	Hypopigmentation-punctate palmoplantar keratoderma syndrome	6 Families
391411	Atypical juvenile parkinsonism	6 Families
456318	Hereditary sensory neuropathy-deafness-dementia syndrome	6 Families
2045	FLOTCH syndrome	6 Families
79447	X-linked lethal multiple pterygium syndrome	6 Families
2790	Endosteal hyperostosis, Worth type	6 Families
2886	TARP syndrome	6 Families
320380	Autosomal recessive spastic paraplegia type 54	6 Families
2118	Hawkinsinuria	5 Families
1428	Familial chondromalacia patellae	5 Families
1836	Mesomelic dysplasia, Kantaputra type	5 Families
2802	X-linked sideroblastic anemia and spinocerebellar ataxia	5 Families
3351	Trichodental syndrome	5 Families
3454	Intellectual disability-developmental delay-contractures syndrome	5 Families
34527	Familial primary hypomagnesemia with normocalciuria and normocalcemia	5 Families
85442	Short stature-pituitary and cerebellar defects-small sella turcica syndrome	5 Families
139583	X-linked hereditary sensory and autonomic neuropathy with deafness	5 Families
101003	Autosomal recessive spastic paraplegia type 23	5 Families
101039	Female restricted epilepsy with intellectual disability	5 Families
99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F	5 Families
231108	Familial rhabdoid tumor	5 Families
171851	MEDNIK syndrome	5 Families

ORPHA Number	Disease or Group of diseases	Number of families
178333	Åland Islands eye disease	5 Families
206554	Autosomal recessive limb-girdle muscular dystrophy type 2M	5 Families
308410	Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency	5 Families
1879	Melorheostosis with osteopoikilosis	5 Families
424099	Colobomatous microphthalmia-rhizomelic dysplasia syndrome	5 Families
444092	Autoimmune interstitial lung disease-arthritis syndrome	5 Families
86789	Patella aplasia/hypoplasia	5 Families
319640	Retinal macular dystrophy type 2	5 Families
320411	Autosomal recessive spastic paraplegia type 56	5 Families
3301	Tetraamelia-multiple malformations syndrome	5 Families
1275	Brachydactyly-elbow wrist dysplasia syndrome	4 Families
1187	Lethal ataxia with deafness and optic atrophy	4 Families
2307	IVIC syndrome	4 Families
2699	Median nodule of the upper lip	4 Families
2947	Triphalangeal thumbs-brachiectrodactyly syndrome	4 Families
266	Autosomal dominant limb-girdle muscular dystrophy type 1A	4 Families
46348	Paroxysmal extreme pain disorder	4 Families
77297	Majeed syndrome	4 Families
97239	Reducing body myopathy	4 Families
101108	Spinocerebellar ataxia type 23	4 Families
98890	Early-onset X-linked optic atrophy	4 Families
228012	Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	4 Families
238578	Familial clubfoot due to 17q23.1q23.2 microduplication	4 Families
293936	EDICT syndrome	4 Families
412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome	4 Families
401911	AXIN2-related attenuated familial adenomatous polyposis	4 Families
363694	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome	4 Families
423296	Spinocerebellar ataxia type 38	4 Families
93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis	4 Families
101077	X-linked Charcot-Marie-Tooth disease type 3	4 Families
93302	Brachyolmia, Maroteaux type	4 Families
466806	Autosomal dominant thrombocytopenia with platelet secretion defect	4 Families
139525	Distal hereditary motor neuropathy type 2	4 Families

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ORPHA Number	Disease or Group of diseases	Number of families
1252	Blepharonasofacial malformation syndrome	3 Families
1074	Ankyloblepharon filiforme adnatum-imperforate anus syndrome	3 Families
2211	Hypertelorism-hypospadias-polysyndactyly syndrome	3 Families
3466	WT limb-blood syndrome	3 Families
1541	Craniosynostosis, Boston type	3 Families
2066	Gamma-aminobutyric acid transaminase deficiency	3 Families
162	Cataract-glaucoma syndrome	3 Families
67044	Thrombocytopenia with congenital dyserythropoietic anemia	3 Families
67036	Autosomal dominant optic atrophy and cataract	3 Families
98766	Spinocerebellar ataxia type 5	3 Families
97249	Pontocerebellar hypoplasia type 3	3 Families
95433	Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome	3 Families
94064	Deafness-infertility syndrome	3 Families
101010	Autosomal spastic paraparesis type 30	3 Families
140963	Bilateral microtia-deafness-cleft palate syndrome	3 Families
300359	PLCG2-associated antibody deficiency and immune dysregulation	3 Families
314978	X-linked non progressive cerebellar ataxia	3 Families
329319	Thrombocythemia with distal limb defects	3 Families
276193	Spinocerebellar ataxia type 35	3 Families
280628	Familial progressive hyper- and hypopigmentation	3 Families
352740	Ocular albinism with congenital sensorineural deafness	3 Families
2994	Short stature-craniofacial anomalies-genital hypoplasia syndrome	3 Families
447757	Autosomal dominant spastic paraparesis type 9B	3 Families
435804	Short stature-advanced bone age-early-onset osteoarthritis syndrome	3 Families
444072	Cerebellar-facial-dental syndrome	3 Families
319340	Carney complex-trismus-pseudocampodactyly syndrome	3 Families
466921	Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome	3 Families
1182	Spastic ataxia with congenital miosis	3 Families
488647	DDX41-related hematologic malignancy predisposition syndrome	3 Families
1349	Mitochondrial DNA-related cardiomyopathy and hearing loss	2 Families
3085	Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome	2 Families
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral	2 Families
1241	Bencze syndrome	2 Families

ORPHA Number	Disease or Group of diseases	Number of families
114	Auriculosteodysplasia	2 Families
1867	Hereditary bullous dystrophy, macular type	2 Families
1786	Acrofacial dysostosis, Catania type	2 Families
1350	Heart-hand syndrome type 2	2 Families
2239	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	2 Families
2027	Gingival fibromatosis-progressive deafness syndrome	2 Families
2754	Orofaciodigital syndrome type 6	2 Families
2504	Metaphyseal dysplasia-maxillary hypoplasia-brachydactyly syndrome	2 Families
2405	Thickened earlobes-conductive deafness syndrome	2 Families
2818	Spastic paraparesis-glucoma-intellectual disability syndrome	2 Families
42665	Tietz syndrome	2 Families
67045	X-linked intellectual disability with isolated growth hormone deficiency	2 Families
55596	Autosomal dominant limb-girdle muscular dystrophy type 1G	2 Families
79141	Hereditary painful callosities	2 Families
79136	Episodic ataxia type 4	2 Families
75497	X-linked Ehlers-Danlos syndrome	2 Families
75327	North Carolina macular dystrophy	2 Families
75373	Progressive bifocal chorioretinal atrophy	2 Families
86818	Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	2 Families
83639	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency	2 Families
85287	X-linked intellectual disability, Siderius type	2 Families
94083	Partington syndrome	2 Families
93409	Brachydactyly-syndactyly, Zhao type	2 Families
93389	Brachydactyly type A5	2 Families
139471	Microphthalmia with brain and digit anomalies	2 Families
139480	Autosomal recessive spastic paraparesis type 39	2 Families
139564	Hereditary sensory and autonomic neuropathy type 1B	2 Families
217622	Sensorineural deafness with dilated cardiomyopathy	2 Families
300576	Oligodontia-cancer predisposition syndrome	2 Families
329191	Tall stature-scoliosis-macrodactyly of the great toes syndrome	2 Families
352403	Spectrin-associated autosomal recessive cerebellar ataxia	2 Families
263463	CHST3-related skeletal dysplasia	2 Families
401964	Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons	2 Families
2471	McDonough syndrome	2 Families
420492	Adult-onset cervical dystonia, DYT23 type	2 Families

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ORPHA Number	Disease or Group of diseases	Number of families
447753	Autosomal dominant spastic paraplegia type 9A	2 Families
447760	Autosomal recessive spastic paraplegia type 9B	2 Families
439254	ITM2B amyloidosis	2 Families
99846	Autosomal dominant myoglobinuria	2 Families
1895	Edinburgh malformation syndrome	2 Families
434179	Orofaciodigital syndrome type 14	2 Families
411788	Familial isolated trichomegaly	2 Families
139557	X-linked distal spinal muscular atrophy type 3	2 Families
476113	Combined immunodeficiency due to TFRC deficiency	2 Families
3233	Cochleosaccular degeneration-cataract syndrome	2 Families
65720	Arthrogryposis-severe scoliosis syndrome	2 Families
3034	Delayed membranous cranial ossification	2 Families
443087	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	2 Families
2379	Early-onset parkinsonism-intellectual disability syndrome	2 Families
324708	ABeta amyloidosis, Iowa type	2 Families
324718	ABetaA21G amyloidosis	2 Families
98606	Syndromic orbital border hypoplasia	2 Families
998	Albinism-deafness syndrome	1 Family
1876	Oculogastrointestinal muscular dystrophy	1 Family
1228	Banki syndrome	1 Family
1319	Camptobrachydactyly	1 Family
1144	Arthrogryposis-like hand anomaly-sensorineural deafness syndrome	1 Family
1979	Lipodystrophy due to peptidic growth factors deficiency	1 Family
3196	Steroid dehydrogenase deficiency-dental anomalies syndrome	1 Family
1765	Dyschondrosteosis-nephritis syndrome	1 Family
1527	Craniosynostosis, Philadelphia type	1 Family
1409	Woolly hair-hypotrichosis-everted lower lip-outstanding ears syndrome	1 Family
2186	Hydrocephalus-blue sclerae-nephropathy syndrome	1 Family
2709	Oculodental syndrome, Rutherford type	1 Family
2674	Cyprus facial-neuromusculoskeletal syndrome	1 Family
2565	Mononen-Karnes-Senac syndrome	1 Family
2391	Congenitally short costocoracoid ligament	1 Family
2999	Ptosis-strabismus-ectopic pupils syndrome	1 Family
2890	Pili torti-onychodysplasia syndrome	1 Family
2917	Polydactyly-myopia syndrome	1 Family
3408	Upington disease	1 Family
3417	Van den Bosch syndrome	1 Family
1246	Brachydactyly-nystagmus-cerebellar ataxia syndrome	1 Family

ORPHA Number	Disease or Group of diseases	Number of families
52056	Ulnar/fibula ray defect-brachydactyly syndrome	1 Family
79135	Episodic ataxia type 3	1 Family
79129	Trichodysplasia-amelogenesis imperfecta syndrome	1 Family
75501	Ehlers-Danlos syndrome, fibronectinemic type	1 Family
79085	AKT2-related familial partial lipodystrophy	1 Family
93283	Spondyloepiphyseal dysplasia, Kimberley type	1 Family
85335	Fried syndrome	1 Family
85322	X-linked intellectual disability, Pai type	1 Family
83648	X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome	1 Family
85168	Craniofacial conodysplasia	1 Family
85292	X-linked spinocerebellar ataxia type 4	1 Family
85288	X-linked intellectual disability, Stocco Dos Santos type	1 Family
95434	Autosomal recessive cerebellar ataxia-saccadic intrusion syndrome	1 Family
93397	Brachydactyly type A7	1 Family
139450	Microtia-eye coloboma-imperforation of the nasolacrimal duct syndrome	1 Family
139512	Neuropathy with hearing impairment	1 Family
101101	Charcot-Marie-Tooth disease type 2B2	1 Family
101112	Spinocerebellar ataxia type 26	1 Family
100995	Autosomal recessive spastic paraplegia type 14	1 Family
101005	Autosomal recessive spastic paraplegia type 25	1 Family
101004	Autosomal recessive spastic paraplegia type 24	1 Family
100999	Autosomal dominant spastic paraplegia type 19	1 Family
100997	X-linked spastic paraplegia type 16	1 Family
101009	Autosomal dominant spastic paraplegia type 29	1 Family
99941	Autosomal dominant Charcot-Marie-Tooth disease type 2G	1 Family
99945	Autosomal dominant Charcot-Marie-Tooth disease type 2L	1 Family
99806	Oculoodontal syndrome	1 Family
98959	Subepithelial mucinous corneal dystrophy	1 Family
231742	Epibulbar lipodermoid-preauricular appendage-polythelia syndrome	1 Family
163662	Spondyloepiphyseal dysplasia, Reardon type	1 Family
163988	Developmental delay-deafness syndrome, Hildebrand type	1 Family
166011	Multiple epiphyseal dysplasia, Beighton type	1 Family
166108	Intellectual disability, Birk-Barel type	1 Family
171617	Autosomal dominant spastic paraplegia type 38	1 Family

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ORPHA Number	Disease or Group of diseases	Number of families
171622	Autosomal recessive spastic paraplegia type 32	1 Family
293375	Grayson-Wilbrandt corneal dystrophy	1 Family
300305	11p15.4 microduplication syndrome	1 Family
314889	Autosomal dominant proximal renal tubular acidosis	1 Family
329883	Non-hypoproteinemic hypertrophic gastropathy	1 Family
329475	Spastic paraplegia-Paget disease of bone syndrome	1 Family
275517	Autoimmune lymphoproliferative syndrome with recurrent viral infections	1 Family
276183	Spinocerebellar ataxia type 32	1 Family
370131	White platelet syndrome	1 Family
370091	Oculocutaneous albinism type 5	1 Family
363727	X-linked dyserythropoietic anemia with abnormal platelets and neutropenia	1 Family
391327	X-linked calvarial hyperostosis	1 Family
324723	ABeta amyloidosis, Arctic type	1 Family
2408	Lowe-Kohn-Cohen syndrome	1 Family
2097	Grant syndrome	1 Family
2090	GMS syndrome	1 Family
137776	Lethal congenital contracture syndrome type 2	1 Family
1122	Ulnar hypoplasia-split foot syndrome	1 Family
2251	Thumb deformity-alopecia-pigmentation anomaly syndrome	1 Family
440354	Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome	1 Family
444099	Autosomal dominant spastic paraplegia type 73	1 Family
163727	Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	1 Family
2821	Spastic paraplegia-neuropathy-poikiloderma syndrome	1 Family
99946	Autosomal dominant Charcot-Marie-Tooth disease type 2A1	1 Family
99792	Dentin dysplasia-sclerotic bones syndrome	1 Family
1962	Exostoses-anetodermia-brachydactyly type E syndrome	1 Family
2663	Nathalie syndrome	1 Family
1892	Ectrodactyly-polydactyly syndrome	1 Family
431140	X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome	1 Family
84093	Hereditary thermosensitive neuropathy	1 Family
1551	Familial benign copper deficiency	1 Family
476119	Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome	1 Family
319332	Autosomal recessive myogenic arthrogryposis multiplex congenita	1 Family
3191	Subaortic stenosis-short stature syndrome	1 Family

ORPHA Number	Disease or Group of diseases	Number of families
3361	Trichodysplasia-xeroderma syndrome	1 Family
320365	Autosomal dominant spastic paraplegia type 36	1 Family
171863	Autosomal dominant spastic paraplegia type 42	1 Family
2835	Pectus excavatum-macrocephaly-dysplastic nails syndrome	1 Family
140481	Autosomal dominant slowed nerve conduction velocity	1 Family
140922	Autosomal recessive limb-girdle muscular dystrophy type 2J	1 Family
443162	NDE1-related microhydranencephaly	1 Family
2572	Spastic ataxia-corneal dystrophy syndrome	1 Family
324703	ABetaL34V amyloidosis	1 Family
488437	SIX2-related frontonasal dysplasia	1 Family
498602	Sugarman brachydactyly	1 Family

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To access the complete Orphanet epidemiological data sets visit Orphadata ([www.orphadata.org](http://www.orphadata.org)).

For any questions or comments, please contact us: [contact.orphanet@inserm.fr](mailto:contact.orphanet@inserm.fr)

Editor-in-chief : Ana Rath – Editor of the report : Stéphanie NGUENGANG WAKAP - Technical support : Samuel Demarest  
and Valérie Lanneau

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