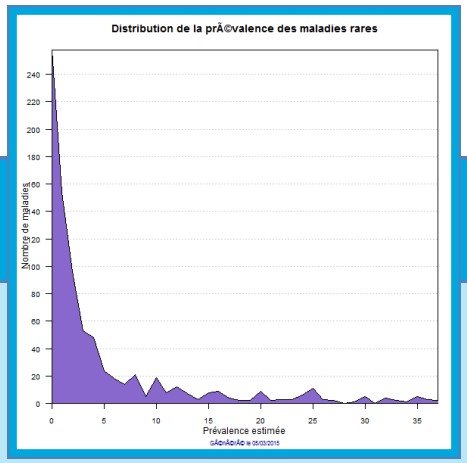


Orphanet Report Series
Rare Diseases collection

Number 2 | June 2018

# Prevalence and incidence of rare diseases: Bibilographic data

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

www.orpha.net www.orphadata.org

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From science to health

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# Methodology

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

This data is presented in the following reports published biannually:

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

## Data collection

A number of different sources are used :

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

## Data characteristics

The data published in this document are worldwide estimations, or European estimations if a worldwide estimation is not available.
The published data is raw collected data or extrapolations of raw data at worldwide or European level when no genetic founder effect is suspected as a cause of a disease.

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

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

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

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

## 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

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Please note that this is just a selection of Orphanet's rare disease epidemiological data. Currently 5880 rare diseases are annotated with prevalence or incidence information in the Orphanet database. To access the complete data sets visit Orphadata (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 * |
| 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 * |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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-cataplexy syndrome | 25.0 * |
| 95719 | Thyroid hemiagenesis | 25.0 |
| 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 * |
| 774 | Hereditary hemorrhagic telangiectasia | 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 * |

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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 * |
| 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 * |
| 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 |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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* |
| 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 * |
| 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* |
| 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 | Anophthalmia-microphthalmia syndrome | 8.3 *BP** |
| 2444 | Congenital pulmonary airway malformation | 8.2 *BP** |
| 171 | Primary sclerosing cholangitis | 8.1 |
| 930 | Idiopathic achalasia | 8.0 |
| 194 | Ocular coloboma | 8.0 *BP** |
| 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** |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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* |
| 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 anophthalmia-microphthalmia syndrome | 5.3 *BP** |
| 738 | Porphyria | 5.25 |
| 685 | Hereditary spastic paraplegia | 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 * |
| 89936 | X-linked hypophosphatemia | 4.45 *BP* |
| 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 |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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 * |
| 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 * |
| 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 * |

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

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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 * |
| 97927 | Peripheral resistance to thyroid hormones | 2.5 * |
| 100070 | Progressive non-fluent aphasia | 2.5 * |
| 315311 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form | 2.5 * |
| 65 | Leber congenital amaurosis | 2.5 |
| 352731 | Oculocutaneous albinism type 1 | 2.5 |
| 1600 | Monosomy 18q | 2.5 *BP* |
| 70588 | Meconium aspiration syndrome | 2.44 * |
| 93928 | Epispadias | 2.4 *BP** |
| 98933 | Multiple system atrophy, parkinsonian type | 2.4 * |
| 247525 | Citrullinemia type I | 2.4 * |
| 908 | Fragile X syndrome | 2.4 *BP** |
| 263 | Limb-girdle muscular dystrophy | 2.32 |
| 330015 | Lead poisoning | 2.3 * |
| 905 | Wilson disease | 2.2 *BP* |
| 2869 | Peutz-Jeghers syndrome | 2.2 *BP* |
| 304 | Epidermolysis bullosa simplex | 2.2 *BP** |
| 137605 | Legius syndrome | 2.2 *BP* |
| 98895 | Becker muscular dystrophy | 2.2 *BP** |
| 454750 | Isolated tracheoesophageal fistula | 2.2 *BP* |
| 217 | Isolated Dandy-Walker malformation | 2.1 * |
| 70567 | Cholangiocarcinoma | 2.1 |
| 95 | Friedreich ataxia | 2.0 * |
| 480 | Kearns-Sayre syndrome | 2.0 * |
| 607 | Nemaline myopathy | 2.0 *BP** |
| 280 | Wolf-Hirschhorn syndrome | 2.0 *BP** |
| 126 | Blepharophimosis-epicanthus inversus-ptosis syndrome | 2.0 |
| 861 | Treacher-Collins syndrome | 2.0 *BP** |
| 352 | Galactosemia | 2.0 *BP** |
| 3346 | Tracheal agenesis | 2.0 *BP** |
| 63 | Alport syndrome | 2.0 * |
| 3129 | Sarcosinemia | 2.0 *BP* |
| 180 | Choroideremia | 2.0 * |
| 1699 | Trisomy 12p | 2.0 *BP* |
| 2017 | Sternal cleft | 2.0 *BP** |
| 185 | Scimitar syndrome | 2.0 *BP** |
| 2345 | Isolated Klippel-Feil syndrome | 2.0 * |

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 54595 | Craniopharyngioma | 2.0 * |
| 93323 | Fibular hemimelia | 2.0 * |
| 98841 | Anaplastic large cell lymphoma | 2.0 * |
| 217064 | 5-fluorouracil poisoning | 2.0 * |
| 157835 | Paroxysmal hemicrania | 2.0 * |
| 168782 | Childhood disintegrative disorder | 2.0 * |
| 657 | Congenital isolated hyperinsulinism | 2.0 *BP* |
| 93110 | Posterior urethral valve | 2.0 * |
| 275761 | Lysosomal acid lipase deficiency | 2.0 * |
| 150 | Nasopharyngeal carcinoma | 2.0 * |
| 506 | Leigh syndrome | 2.0 * |
| 363203 | Ring chromosome | 2.0 *BP* |
| 90073 | Hepatitis B reinfection following liver transplantation | 2.0 * |
| 3392 | Tularemia | 2.0 * |
| 1598 | Monosomy 18p | 2.0 *BP** |
| 447 | Paroxysmal nocturnal hemoglobinuria | 2.0 * |
| 199 | Cornelia de Lange syndrome | 1.9 * |
| 10 | 48,XXYY syndrome | 1.9 *BP** |
| 79361 | Inherited epidermolysis bullosa | 1.9 *BP** |
| 35808 | Malignant sex cord stromal tumor of ovary | 1.85 * |
| 304 | Epidermolysis bullosa simplex | 1.8 |
| 675 | Annular pancreas | 1.8 *BP** |
| 664 | Ornithine transcarbamylase deficiency | 1.77 *BP* |
| 77 | Aniridia | 1.75 |
| 420429 | Glycogen storage disease due to acid maltase deficiency, late-onset | 1.75 *BP* |
| 251076 | 8p23.1 duplication syndrome | 1.72 |
| 2182 | Hydrocephalus with stenosis of the aqueduct of Sylvius | 1.7 *BP* |
| 2182 | Hydrocephalus with stenosis of the aqueduct of Sylvius | 1.7 |
| 637 | Neurofibromatosis type 2 | 1.7 * |
| 2152 | Mowat-Wilson syndrome | 1.7 *BP** |
| 1848 | Renal agenesis, bilateral | 1.7 *BP** |
| 98879 | Hemophilia B | 1.7 * |
| 141077 | Epignathus | 1.68 *BP* |
| 394 | Classic homocystinuria | 1.65 * |
| 899 | Walker-Warburg syndrome | 1.65 *BP** |
| 183660 | Severe combined immunodeficiency | 1.65 *BP** |
| 1915 | Fetal alcohol syndrome | 1.6 *BP** |
| 64747 | X-linked Charcot-Marie-Tooth disease | 1.6 * |
| 79241 | Biotinidase deficiency | 1.6 * |
| 79241 | Biotinidase deficiency | 1.6 *BP* |
| 98895 | Becker muscular dystrophy | 1.53 |
| 192 | Coffin-Lowry syndrome | 1.5 |
| 131 | Budd-Chiari syndrome | 1.5 * |
| 2019 | Femur-fibula-ulna complex | 1.5 *BP** |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 2911 | Poland syndrome | 1.5 *BP** |
| 389 | Langerhans cell histiocytosis | 1.5 * |
| 137 | Congenital disorder of glycosylation | 1.5 *BP** |
| 35689 | Primary lateral sclerosis | 1.5 * |
| 641 | Multifocal motor neuropathy | 1.5 |
| 71211 | Neuromyelitis optica | 1.5 * |
| 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* |
| 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 |
| 487 | Krabbe disease | 1.0 * |
| 614 | Thomsen and Becker disease | 1.0 |
| 16 | Blue cone monochromatism | 1.0 |

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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 * |
| 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 * |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 90060 | Diffuse alveolar hemorrhage | 1.0 * |
| 98863 | X-linked Emery-Dreifuss muscular dystrophy | 1.0 |
| 98863 | X-linked Emery-Dreifuss muscular dystrophy | 1.0 *BP* |
| 96263 | 48,XXXY syndrome | 1.0 *BP** |
| 95715 | Congenital hypothyroidism due to transplacental passage of maternal 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 * |
| 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 * |

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 169793 | Severe hemophilia B | 0.8 * |
| 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 akinesia 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 * |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 313 | Lamellar ichthyosis | 0.55 * |
| 96264 | 49,XXXXY syndrome | 0.55 *BP** |
| 79276 | Acute intermittent porphyria | 0.54 * |
| 93929 | Cloacal exstrophy | 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 * |
| 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 | Autosomal dominant 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 * |
| 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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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 811 | Shwachman-Diamond syndrome | 0.28 |
| 579 | Mucopolysaccharidosis type 1 | 0.25 * |
| 702 | Pelizaeus-Merzbacher disease | 0.25 * |
| 678 | Papillon-Lefèvre syndrome | 0.25 |
| 35173 | X-linked dominant chondrodysplasia punctata | 0.25 *BP** |
| 77292 | Niemann-Pick disease type A | 0.25 *BP** |
| 93571 | Dense deposit disease | 0.25 |
| 275798 | Pulmonary arterial hypertension associated with connective tissue disease | 0.25 * |
| 910 | Xeroderma pigmentosum | 0.23 *BP** |
| 324 | Fabry disease | 0.22 *BP** |
| 47 | X-linked agammaglobulinemia | 0.22 |
| 111 | Barth syndrome | 0.22 * |
| 436 | Hypophosphatasia | 0.21 *BP** |
| 89936 | X-linked hypophosphatemia | 0.21 * |
| 534 | Oculocerebrorenal syndrome of Lowe | 0.2 |
| 893 | WAGR syndrome | 0.2 *BP* |
| 385 | Neurodegeneration with brain iron accumulation | 0.2 * |
| 235 | Dubowitz syndrome | 0.2 *BP** |
| 191 | Cockayne syndrome | 0.2 *BP** |
| 3006 | Pyridoxine-dependent epilepsy | 0.2 *BP** |
| 2052 | Fraser syndrome | 0.2 *BP** |
| 277 | Severe combined immunodeficiency due to adenosine deaminase deficiency | 0.2 * |
| 35 | Propionic acidemia | 0.2 * |
| 722 | Hypoplasminogenemia | 0.2 * |
| 353 | Autosomal recessive limb-girdle muscular dystrophy type 2C | 0.2 * |
| 79270 | Sanfilippo syndrome type B | 0.2 * |
| 98813 | Hypohidrotic ectodermal dysplasia with immunodeficiency | 0.2 *BP** |
| 99870 | Letterer-Siwe disease | 0.2 * |
| 238583 | Hyperphenylalaninemia due to tetrahydrobiopterin deficiency | 0.2 |
| 178478 | Infant botulism | 0.2 *BP** |
| 209916 | Extraskeletal myxoid chondrosarcoma | 0.2 * |
| 596 | X-linked centronuclear myopathy | 0.2 * |
| 79096 | Pyridoxal phosphate-responsive seizures | 0.2 * |
| 1361 | Carnosinemia | 0.2 *BP* |
| 2635 | Metatropic dysplasia | 0.2 *BP** |
| 808 | Seckel syndrome | 0.2 *BP** |
| 2614 | Nail-patella syndrome | 0.2 *BP** |
| 1456 | Atypical coarctation of aorta | 0.17 *BP** |
| 407 | Glycine encephalopathy | 0.17 * |
| 79404 | Junctional epidermolysis bullosa, generalized severe | 0.17 *BP* |
| 280219 | Pelizaeus-Merzbacher disease, classic form | 0.17 * |
| 583 | Mucopolysaccharidosis type 6 | 0.16 *BP** |
| 583 | Mucopolysaccharidosis type 6 | 0.16 * |
| 85 | Congenital dyserythropoietic anemia | 0.16 *BP** |
| 745 | Severe hereditary thrombophilia due to congenital protein C deficiency | 0.16 *BP* |
| 335 | Congenital fibrinogen deficiency | 0.15 * |
| 538 | Lymphangioleiomyomatosis | 0.15 |
| 223 | Nephrogenic diabetes insipidus | 0.15 * |
| 79430 | Hermansky-Pudlak syndrome | 0.15 |
| 157850 | Pantothenate kinase-associated neurodegeneration | 0.15 * |
| 118 | Beta-mannosidosis | 0.14 *BP** |
| 763 | Pycnodysostosis | 0.13 |
| 3463 | Wolfram syndrome | 0.13 |
| 183669 | Agammaglobulinemia | 0.13 * |
| 33364 | Trichothiodystrophy | 0.12 *BP** |
| 1308 | C syndrome | 0.11 * |
| 34149 | Autosomal dominant tubulointerstitial kidney disease | 0.11 * |
| 61 | Alpha-mannosidosis | 0.1 * |
| 367 | Glycogen storage disease due to glycogen branching enzyme deficiency | 0.1 *BP* |
| 512 | Metachromatic leukodystrophy | 0.1 * |
| 906 | Wiskott-Aldrich syndrome | 0.1 * |
| 205 | Crigler-Najjar syndrome | 0.1 *BP** |
| 773 | Refsum disease | 0.1 * |
| 1452 | Cleidocranial dysplasia | 0.1 |
| 1775 | Dyskeratosis congenita | 0.1 * |
| 204 | Sporadic Creutzfeldt-Jakob disease | 0.1 * |
| 1959 | Evans syndrome | 0.1 * |
| 326 | Congenital factor V deficiency | 0.1 * |
| 507 | Leishmaniasis | 0.1 * |
| 3329 | Tibial aplasia-ectrodactyly syndrome | 0.1 * |
| 329 | Congenital factor XI deficiency | 0.1 * |
| 2686 | Cyclic neutropenia | 0.1 * |
| 298 | Mitochondrial neurogastrointestinal encephalomyopathy | 0.1 * |
| 225 | Maternally-inherited diabetes and deafness | 0.1 * |
| 209 | Cutis laxa | 0.1 *BP** |
| 31824 | Colchicine poisoning | 0.1 * |
| 119 | Autosomal recessive limb-girdle muscular dystrophy type 2E | 0.1 * |
| 142 | Anaplastic thyroid carcinoma | 0.1 * |
| 73274 | Acquired hemophilia | 0.1 * |
| 90793 | Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency | 0.1 * |
| 93262 | Crouzon syndrome-acanthosis nigricans syndrome | 0.1 *BP* |
| 93322 | Tibial hemimelia | 0.1 *BP** |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Estimated prevalence (/100,000) |
|---|---|---|
| 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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*BP* indicates birth prevalence

# 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* |
| 3467 | Hereditary xanthinuria | 9.05* |
| 217071 | Renal cell carcinoma | 8.35* |
| 1941 | Juvenile absence epilepsy | 7.5* |
| 70482 | Carcinoma of esophagus | 7.0 |
| 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* |
| 1489 | Whooping cough | 4.37* |
| 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 dyshormonogenesis | 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* |
| 355 | Gaucher disease | 1.7* |

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| ORPHA Number | Disease or Group of diseases | Estimated incidence (/100,000) |
|---|---|---|
| 139417 | Acute transverse myelitis | 1.6 |
| 810 | Shigellosis | 1.59* |
| 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* |
| 77 | Aniridia | 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* |
| 549 | Legionellosis | 0.97* |
| 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* |
| 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* |
| 340 | Hemorrhagic fever-renal syndrome | 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 instestine | 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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*BP* indicates birth prevalence

| 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 | Oligodendroglioma | 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 |
| 781 | Q fever | 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* |
| 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* |
| 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 |
| 2086 | Optic pathway glioma | 0.12 |
| 86893 | Nodular lymphocyte predominant Hodgkin lymphoma | 0.12 |
| 3392 | Tularemia | 0.12* |
| 213716 | Squamous cell carcinoma of the corpus uteri | 0.12* |
| 99915 | Maligant granulosa cell tumor of the ovary | 0.12* |
| 213512 | Malignant mixed Müllerian tumor of the ovary | 0.12* |
| 509 | Leptospirosis | 0.11* |
| 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 | Cardiodysrhythmic potassium-sensitive periodic paralysis | 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* |
| 1822 | Dysplasia epiphysealis hemimelica | 0.1 |
| 98845 | Classic Hodgkin lymphoma, lymphocyte-rich type | 0.1* |
| 251630 | Anaplastic oligodendroglioma | 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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*BP* indicates birth prevalence

| 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 |
| 1304 | Brucellosis | 0.07* |
| 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 |
| 83476 | West-Nile encephalitis | 0.04* |
| 3299 | Tetanus | 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* |
| 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 the pancreas | 0.03* |
| 251576 | Gliosarcoma | 0.03* |
| 329984 | Goblet cell carcinoma | 0.025 |
| 449 | Hepatoblastoma | 0.02* |
| 1267 | Botulism | 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 protoporphyria | 0.012* |
| 2030 | Fibrosarcoma | 0.01* |
| 55881 | Adamantinoma | 0.01* |
| 251899 | Choroid plexus carcinoma | 0.01* |
| 173 | Cholera | 0.01* |
| 324625 | Chikungunya | 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 | Rosaï-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 |
| 69077 | Rhabdoid tumor | 500 Cases |
| 73256 | Central neurocytoma | 500 Cases |
| 79282 | Methylmalonic acidemia with homocystinuria, type cblC | 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 |
| 662 | Yellow nail syndrome | 400 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 |
| 2909 | Rothmund-Thomson syndrome | 300 Cases |
| 1467 | Cogan syndrome | 300 Cases |
| 2330 | Kasabach-Merritt syndrome | 300 Cases |
| 3347 | Mounier-Kühn syndrome | 300 Cases |
| 42775 | PHACE syndrome | 300 Cases |
| 530 | Lipoid proteinosis | 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 | Erythrokeratodermia 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 |
| 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 hypercalciuria 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 |
| 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 |
| 3103 | Roberts syndrome | 150 Cases |
| 2048 | Foix-Chavany-Marie syndrome | 150 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 2108 | Hallermann-Streiff syndrome | 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 |
| 2576 | MULIBREY nanism | 140 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 |
| 2343 | Isolated cloverleaf skull syndrome | 120 Cases |
| 51 | Aicardi-Goutières syndrome | 120 Cases |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 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 |
| 79477 | Griscelli disease 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 |
| 981 | Internal carotid agenesis | 100 Cases |
| 1507 | Autosomal recessive Robinow syndrome | 100 Cases |
| 672 | Pallister-Hall syndrome | 100 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 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 paraplegia type 2 | 100 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 |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 75381 | Cystoid macular dystrophy | 97 Cases |
| 333 | Farber disease | 96 Cases |
| 699 | Pearson syndrome | 95 Cases |
| 2671 | Neu-Laxova syndrome | 91 Cases |
| 52368 | Mohr-Tranebjaerg syndrome | 91 Cases |
| 1885 | Isolated ectopia lentis | 90 Cases |
| 742 | Prolidase deficiency | 90 Cases |
| 276198 | Spinocerebellar ataxia type 36 | 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 1935 | Early myoclonic encephalopathy | 80 Cases |
| 526 | Liddle syndrome | 80 Cases |
| 709 | Peters plus syndrome | 80 Cases |
| 382 | Guanidinoacetate methyltransferase deficiency | 80 Cases |
| 1440 | Ring chromosome 14 syndrome | 80 Cases |
| 3342 | Arterial tortuosity syndrome | 80 Cases |
| 49 | Penile agenesis | 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 | Acrodysostosis | 80 Cases |
| 3152 | Sclerosteosis | 80 Cases |
| 98769 | Spinocerebellar ataxia type 15/16 | 80 Cases |
| 600 | Vocal cord and pharyngeal distal myopathy | 78 Cases |
| 2396 | Encephalocraniocutaneous lipomatosis | 77 Cases |
| 1393 | Cerebrocostomandibular syndrome | 75 Cases |
| 238722 | Familial congenital mirror movements | 75 Cases |
| 320406 | Spastic paraplegia-optic atrophy-neuropathy syndrome | 75 Cases |
| 79230 | Hemochromatosis type 2 | 74 Cases |
| 209981 | IRIDA syndrome | 74 Cases |
| 622 | Homocystinuria without methylmalonic aciduria | 73 Cases |
| 659 | Mutilating palmoplantar keratoderma with periorificial keratotic plaques | 73 Cases |
| 2196 | Familial primary hypomagnesemia with hypercalciuria 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/mandibule | 70 Cases |
| 2028 | Juvenile hyaline fibromatosis | 70 Cases |
| 2484 | Melnick-Needles syndrome | 70 Cases |
| 756 | Pseudohypoaldosteronism type 1 | 70 Cases |
| 65759 | Carpenter syndrome | 70 Cases |
| 79293 | Familial LCAT deficiency | 70 Cases |
| 79257 | GM1 gangliosidosis type 3 | 70 Cases |
| 404546 | DITRA | 70 Cases |
| 357043 | Amyotrophic lateral sclerosis type 4 | 70 Cases |
| 3310 | Tetrasomy 9p | 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 |
| 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 |
| 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 | 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-Clausen disease | 60 Cases |
| 677 | Pancreatoblastoma | 60 Cases |
| 381 | Griscelli disease | 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 |
| 139 | CHILD syndrome | 60 Cases |
| 2065 | Galloway-Mowat 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 79310 | Vitamin B12-responsive methylmalonic acidemia 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 |
| 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 |
| 2332 | KBG syndrome | 59 Cases |
| 3338 | Toriello-Carey syndrome | 59 Cases |
| 293642 | Blepharophimosis-intellectual disability syndrome | 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 |
| 329284 | Beta-propeller protein-associated neurodegeneration | 54 Cases |
| 2833 | Stiff skin syndrome | 54 Cases |
| 464306 | DYRK1A-related intellectual disability syndrome | 54 Cases |
| 79099 | Interstitial granulomatous dermatitis with arthritis | 53 Cases |
| 254516 | Motor developmental delay due to 14q32.2 paternally expressed gene defect | 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 |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 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 |
| 1297 | Branchio-oculo-facial 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 |
| 361 | Familial glucocorticoid deficiency | 50 Cases |
| 1902 | Ehrlichiosis | 50 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 |

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*BP* indicates birth prevalence

| ORPHA Number | Disease or Group of diseases | Number of 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 megalencephaly | 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 |
| 157846 | Neuroferritinopathy | 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 |
| 96177 | Ring chromosome 15 syndrome | 50 Cases |
| 99776 | Mosaic trisomy 9 | 50 Cases |
| 1873 | Jalili syndrome | 49 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 |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 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 |
| 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 |
| 2301 | Congenital short bowel syndrome | 41 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 |
| 1023 | Congenital generalized hypertrichosis, Ambras type | 40 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 1923 | Methimazole embryofetopathy | 40 Cases |
| 859 | Transcobalamin deficiency | 40 Cases |
| 2273 | Ichthyosis follicularis-alopecia-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 |
| 51188 | Ethylmalonic encephalopathy | 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 |
| 140944 | CLOVES syndrome | 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 |
| 217008 | Bockenheimer syndrome | 40 Cases |
| 1515 | Cranioectodermal dysplasia | 39 Cases |
| 458758 | Composite hemangioendothelioma | 39 Cases |
| 36 | Acrocallosal syndrome | 38 Cases |
| 1647 | Oculocerebrocutaneous syndrome | 38 Cases |
| 2067 | GAPO syndrome | 38 Cases |
| 69085 | Limb-mammary syndrome | 38 Cases |
| 55654 | Hypotrichosis simplex | 38 Cases |
| 85162 | Facial onset sensory and motor neuronopathy | 38 Cases |
| 163696 | Action myoclonus-renal failure syndrome | 38 Cases |
| 171629 | Autosomal recessive spastic paraplegia 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 |
| 329457 | Distal arthrogryposis type 5D | 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 | Paternal uniparental disomy of chromosome 14 | 37 Cases |
| 1532 | Gómez-López-Hernández syndrome | 36 Cases |
| 1855 | Spondyloenchondrodysplasia | 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 |
| 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 |
| 91 | Aromatase deficiency | 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 |
| 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 |
| 83 | Antley-Bixler syndrome | 34 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 943 | Malonic aciduria | 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-Huët anomaly syndrome | 34 Cases |
| 398097 | Neonatal antiphospholipid syndrome | 34 Cases |
| 93269 | Short rib-polydactyly syndrome, Majewski type | 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 cblG | 33 Cases |
| 832 | Succinyl-CoA:3-ketoacid CoA transferase deficiency | 33 Cases |
| 3322 | Hoyeraal-Hreidarsson syndrome | 33 Cases |
| 2795 | Polycystic ovaries-urethral sphincter dysfunction syndrome | 33 Cases |
| 123 | Björnstad syndrome | 33 Cases |
| 225123 | Hemochromatosis type 3 | 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 |
| 458763 | Retiform hemangioendothelioma | 32 Cases |
| 139485 | Autosomal recessive ataxia due to ubiquinone deficiency | 31 Cases |
| 231573 | Congenital erosive and vesicular dermatosis | 31 Cases |
| 276435 | Lower motor neuron syndrome with late-adult onset | 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 |
| 1225 | Baller-Gerold syndrome | 30 Cases |
| 1314 | Symmetrical thalamic calcifications | 30 Cases |
| 1662 | Restrictive dermopathy | 30 Cases |
| 1545 | Crisponi syndrome | 30 Cases |
| 1525 | Cranio-osteoarthropathy | 30 Cases |
| 1427 | Otospondylomegaepiphyseal dysplasia | 30 Cases |
| 2036 | Scalp-ear-nipple syndrome | 30 Cases |
| 2763 | Osteocraniostenosis | 30 Cases |
| 2733 | Omodysplasia | 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 | Encephalopathy due to hydroxykynureninuria | 30 Cases |
| 79157 | 2-methylbutyryl-CoA dehydrogenase deficiency | 30 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 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 |
| 497623 | C12ORF65-related combined oxidative phosphorylation defect | 30 Cases |
| 178345 | Aromatase excess syndrome | 30 Cases |
| 1596 | Distal monosomy 15q | 30 Cases |
| 1943 | Infant epilepsy with migrant focal crisis | 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 |
| 101000 | Autosomal recessive spastic paraplegia type 20 | 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 |
| 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 |
| 466 | Fatal familial insomnia | 27 Cases |
| 966 | Hypertrichosis-acromegaloid facial appearance syndrome | 27 Cases |
| 1040 | Metaphyseal anadysplasia | 27 Cases |
| 2169 | Methylcobalamin deficiency type cblE | 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 paraplegia type 12 | 27 Cases |
| 99812 | LIG4 syndrome | 27 Cases |
| 251287 | Benign concentric annular macular dystrophy | 27 Cases |
| 313808 | Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia | 27 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 1186 | Infantile onset spinocerebellar ataxia | 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 |
| 85173 | IMAGe syndrome | 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 |
| 363454 | Autosomal dominant childhood-onset proximal spinal muscular atrophy with contractures | 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 |
| 1234 | Bartsocas-Papas syndrome | 24 Cases |
| 1490 | Corneal dystrophy-perceptive deafness syndrome | 24 Cases |
| 1361 | Carnosinemia | 24 Cases |
| 3275 | Spondylocarpotarsal synostosis | 24 Cases |
| 34521 | Distal myopathy with early respiratory muscle involvement | 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 paraplegia type 34 | 24 Cases |
| 209902 | Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency | 24 Cases |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 438216 | PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation | 24 Cases |
| 480864 | Recurrent metabolic encephalomyopathic 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 |
| 88630 | Terminal osseous dysplasia-pigmentary defects syndrome | 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 |
| 2953 | Ehlers-Danlos syndrome, musculocontractural type | 22 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 |
| 93953 | Familial thyroglossal duct cyst | 22 Cases |
| 228423 | Monocytopenia with susceptibility to infections | 22 Cases |
| 228429 | Generalized congenital lipodystrophy with myopathy | 22 Cases |
| 163690 | Hypotonia-cystinuria syndrome | 22 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 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 |
| 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-digito-palmar syndrome | 21 Cases |
| 66625 | Cerebrooculonasal 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 |
| 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 syndrome | 21 Cases |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 30 | Hereditary orotic aciduria | 20 Cases |
| 3137 | Alpha-N-acetylgalactosaminidase 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 |
| 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-aminoadipic 2-oxoadipic aciduria | 20 Cases |
| 79084 | Familial partial lipodystrophy, Köbberling type | 20 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 352328 | MEGDEL 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 |
| 420179 | Malan overgrowth syndrome | 20 Cases |
| 100043 | Autosomal dominant intermediate Charcot-Marie-Tooth disease type A | 20 Cases |
| 3416 | Hyperostosis corticalis generalisata | 20 Cases |
| 79476 | Griscelli disease 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 |
| 94063 | 12q14 microdeletion syndrome | 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 recessive spastic paraplegia type 58 | 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 |
| 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 86309 | DPAGT1-CDG | 18 Cases |
| 85167 | Spondylometaphyseal 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 cblX | 18 Cases |
| 370046 | Didymosis aplasticosebacea | 18 Cases |
| 363417 | Temtamy preaxial brachydactyly syndrome | 18 Cases |
| 1827 | Acromelic frontonasal dysplasia | 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 SLCO2A1 gene | 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 cblD | 17 Cases |
| 91131 | DK1-CDG | 17 Cases |
| 93282 | Spondyloepimetaphyseal 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 |

| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 324381 | Hereditary inclusion body myopathy type 4 | 17 Cases |
| 319595 | Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency | 17 Cases |
| 436159 | Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency | 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 distal myopathy | 17 Cases |
| 464760 | Familial cavitary 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 |
| 71528 | Obesity due to prohormone convertase I 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 464282 | Spastic paraplegia-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 maternal anti-neutral endopeptidase alloimmunization | 15 Cases |
| 79284 | Methylmalonic acidemia with homocystinuria type cblF | 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 |
| 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 paraplegia | 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 |
| 439212 | Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome | 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 |
| 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 |
| 1193 | Atkin-Flaitz syndrome | 14 Cases |
| 1660 | Dermoodontodysplasia | 14 Cases |
| 1516 | Craniofacial dyssynostosis | 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 | P2Y12 defect | 14 Cases |
| 75378 | Oligocone trichromacy | 14 Cases |
| 90390 | Anonychia-onychodystrophy syndrome | 14 Cases |
| 93356 | Spondyloepimetaphyseal dysplasia, Missouri type | 14 Cases |
| 139578 | Hereditary sensory and autonomic 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 137783 | Lethal congenital contracture syndrome type 3 | 14 Cases |
| 364028 | X-linked intellectual disability due to GRIA3 anomalies | 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 |
| 404443 | Tall stature-intellectual disability-facial dysmorphism 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 |
| 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 |
| 436151 | Intellectual disability-expressive aphasia-facial dysmorphism 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 disease type 3 | 13 Cases |
| 329813 | Mosaic genome-wide paternal uniparental disomy | 13 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 Xp11.22-p11.23 syndrome | 12 Cases |
| 210571 | Dystonia 16 | 12 Cases |
| 238763 | Glaucoma secondary to spherophakia/ectopia lentis and megalocornea | 12 Cases |
| 254531 | Paternal 14q32.2 hypomethylation syndrome | 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 |
| 284169 | Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion | 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 |
| 496689 | Kyphoscoliosis-lateral tongue atrophy-hereditary spastic paraplegia syndrome | 12 Cases |
| 99672 | Fried's tooth and nail syndrome | 12 Cases |
| 141148 | Hemifacial myohyperplasia | 12 Cases |
| 2253 | Foveal hypoplasia-presenile cataract syndrome | 11 Cases |
| 1031 | Amelogenesis imperfecta-nephrocalcinosis syndrome | 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 261349 | 2p15p16.1 microdeletion syndrome | 11 Cases |
| 280553 | Fatal infantile hypertonic myofibrillar myopathy | 11 Cases |
| 397937 | Polyglucosan body myopathy type 1 | 11 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, Markesbery-Griggs type | 11 Cases |
| 464724 | Fever-associated acute infantile liver failure syndrome | 11 Cases |
| 319189 | Familial cortical myoclonus | 11 Cases |
| 468661 | Autosomal recessive spastic paraplegia type 74 | 11 Cases |
| 96172 | Ring chromosome 3 syndrome | 11 Cases |
| 221120 | Pseudoaminopterin 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 choroidocerebral 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 |
| 2072 | Gaucher disease-ophthalmoplegia-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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 85163 | Hypomyelination-congenital cataract syndrome | 10 Cases |
| 85274 | Syndromic X-linked intellectual disability 7 | 10 Cases |
| 97340 | Hunter-McAlpine craniosynostosis | 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 paraplegia type 13 | 10 Cases |
| 101007 | Autosomal recessive spastic paraplegia 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 chondrodysplasia, 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 |
| 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 |
| 280633 | Multiple congenital anomalies-hypotonia-seizures syndrome | 10 Cases |
| 284227 | TEMPI syndrome | 10 Cases |

| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 404448 | ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder | 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 |
| 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 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 | Paternal 14q32.2 microdeletion syndrome | 9 Cases |
| 251279 | Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome | 9 Cases |
| 163979 | X-linked intellectual disability-craniofacioskeletal syndrome | 9 Cases |
| 163982 | X-linked intellectual disability-spastic quadriparesis 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 paraplegia 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 paraplegia type 53 | 9 Cases |
| 438075 | Ketoacidosis due to monocarboxylate transporter-1 deficiency | 9 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 |
| 79022 | Simpson-Golabi-Behmel syndrome type 2 | 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 2561 | Pyramidal molar-glaucoma-upper abnormal lip syndrome | 8 Cases |
| 2371 | Lethal Larsen-like syndrome | 8 Cases |
| 2351 | Kousseff 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 |
| 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 | Maternal 14q32.2 microdeletion syndrome | 8 Cases |
| 251290 | Parietal foramina with clavicular hypoplasia | 8 Cases |
| 178506 | Brain calcification, Rajab type | 8 Cases |
| 178389 | Osteopetrosis-hypogammaglobulinemia syndrome | 8 Cases |
| 171836 | Amelogenesis imperfecta-gingival hyperplasia 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 |
| 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 |
| 399096 | Distal anoctaminopathy | 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 |
| 468684 | CCDC115-CDG | 8 Cases |
| 459061 | Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome | 8 Cases |
| 1450 | Ring chromosome 8 syndrome | 8 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 502430 | Metopic ridging-ptosis-facial dysmorphism 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 |
| 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-glaucoma 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 |
| 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 | Maternal 14q32.2 hypermethylation syndrome | 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 |
| 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 |
| 306558 | Primary microcephaly-epilepsy-permanent neonatal diabetes 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 pachygyria | 7 Cases |
| 329228 | Microcephalic primordial dwarfism due to ZNF335 deficiency | 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 370022 | Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts 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 |
| 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 |
| 1767 | Familial progressive vestibulocochlear dysfunction | 7 Cases |
| 261204 | 16p11.2p12.2 microduplication syndrome | 7 Cases |
| 399086 | Finnish upper limb-onset distal myopathy | 7 Cases |
| 1818 | Ectodermal dysplasia, trichoodontoonychial 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 paraplegia 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 |
| 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 |
| 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 |
| 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 |
| 79106 | Eiken syndrome | 6 Cases |
| 85276 | X-linked intellectual disability, Armfield type | 6 Cases |
| 93973 | Carpenter-Waziri syndrome | 6 Cases |
| 101008 | Autosomal recessive spastic paraplegia type 28 | 6 Cases |
| 211017 | Spinocerebellar ataxia type 30 | 6 Cases |
| 230851 | Ehlers-Danlos syndrome, cardiac valvular type | 6 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 228003 | Severe combined immunodeficiency due to CORO1A deficiency | 6 Cases |
| 244305 | Dominant hypophosphatemia with nephrolithiasis or osteoporosis | 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 |
| 247815 | Autosomal recessive ataxia due to PEX10 deficiency | 6 Cases |
| 247820 | Ectodermal dysplasia-syndactyly syndrome | 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 |
| 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 |
| 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 |
| 3357 | Autosomal dominant trichoodontoonychodysplasia-syndactyly | 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 1113 | Aphalangy-syndactyly-microcephaly syndrome | 5 Cases |
| 1811 | Odontomicronychial 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 |
| 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 |
| 217026 | Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type | 5 Cases |
| 228390 | Frontonasal dysplasia with alopecia and genital anomaly | 5 Cases |
| 228227 | Late-onset focal dermal elastosis | 5 Cases |
| 228240 | Elastoderma | 5 Cases |
| 238766 | Ptosis-syndactyly-learning difficulties syndrome | 5 Cases |
| 251523 | Hyperzincemia and hypercalprotectinemia | 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 264200 | 14q22q23 microdeletion syndrome | 5 Cases |
| 280183 | Methylmalonic aciduria due to transcobalamin receptor defect | 5 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 |
| 357329 | Cryptosporidiosis-chronic cholangitis-liver disease 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 |
| 2816 | Spastic paraplegia-epilepsy-intellectual disability syndrome | 5 Cases |
| 2819 | Spastic paraplegia-facial-cutaneous lesions syndrome | 5 Cases |
| 2432 | Macrosomia-microphthalmia-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 |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 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 | Maternally-inherited spastic paraplegia | 5 Cases |
| 320385 | Autosomal recessive spastic paraplegia type 49 | 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-psoriasiform dermatitis syndrome | 5 Cases |
| 488635 | Early-onset epilepsy-intellectual disability-brain anomalies syndrome | 5 Cases |
| 488618 | Transketolase deficiency | 5 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 1538 | Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome | 4 Cases |
| 1135 | Arrhinia-choanal atresia-microphthalmia syndrome | 4 Cases |
| 2431 | Central bilateral macrogyria | 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-alopecia-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 paraplegia-nephritis-deafness syndrome | 4 Cases |

| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 2168 | Homocarnosinosis | 4 Cases |
| 3355 | Trichoodontoonychial 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 quadriparesis 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 | Myopathy due to calsequestrin and SERCA1 protein overload | 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 |
| 93352 | Spondyloepimetaphyseal dysplasia, Shohat type | 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 137625 | Glycogen storage disease due to muscle and heart glycogen synthase deficiency | 4 Cases |
| 217396 | Progressive polyneuropathy with bilateral striatal necrosis | 4 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-sensorineural 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 | Parkinsonism due to ATP13A2 deficiency | 4 Cases |
| 324313 | 9p13 microdeletion syndrome | 4 Cases |
| 324581 | Benign Samaritan congenital myopathy | 4 Cases |
| 352470 | Mitochondrial DNA deletion syndrome with progressive myopathy | 4 Cases |
| 329332 | Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome | 4 Cases |
| 329341 | Limbic encephalitis with DPP6 antibodies | 4 Cases |
| 263410 | Infantile spams-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, Mégarbané type | 4 Cases |
| 404473 | Severe intellectual disability-progressive spastic diplegia syndrome | 4 Cases |
| 404466 | Female infertility due to zona pellucida defect | 4 Cases |
| 401862 | Lipoyl 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 | Cardiac anomalies-developmental delay-facial dysmorphism syndrome | 4 Cases |
| 370052 | SCALP syndrome | 4 Cases |

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| 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-hypotonia-ophthalmologic-skeletal anomalies syndrome | 4 Cases |
| 363969 | Autosomal recessive cerebral atrophy | 4 Cases |
| 363992 | Ichthyosis-short stature-brachydactyly-microspherophakia syndrome | 4 Cases |
| 363965 | Koolen-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 |
| 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 |
| 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 | Holoprosencephaly-hypokinesia-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 |
| 466722 | Autosomal recessive spastic paraplegia type 77 | 4 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 468717 | Rhizomelic chondrodysplasia punctata type 5 | 4 Cases |
| 466926 | Seizures-scoliosis-macrocephaly syndrome | 4 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 syndrom | 4 Cases |
| 500188 | X-linked external auditory canal atresia-dilated internal auditory canal-facial dysmorphism syndrome | 4 Cases |
| 506353 | Autosomal recessive complex spastic paraplegia due to Kennedy pathway dysfunction | 4 Cases |
| 505216 | 3-methylglutaconic aciduria type 9 | 4 Cases |
| 485421 | Leigh-like basal ganglia disease-optic atrophy-peripheral neuropathy syndrome | 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 |
| 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 |
| 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-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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 79156 | Seizures-intellectual disability due to hydroxylysinuria syndrome | 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 |
| 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 |
| 85318 | X-linked intellectual disability-precocious puberty-obesity 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 |
| 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 |
| 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-pretibial epidermolysis bullosa syndrome | 3 Cases |
| 313800 | Optic nerve edema-splenomegaly syndrome | 3 Cases |
| 306504 | Junctional epidermolysis bullosa with respiratory and renal involvement | 3 Cases |
| 306542 | Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome | 3 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 pachygyria 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 |
| 401795 | Autosomal recessive spastic paraplegia 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 |
| 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 | Intellectual disability-hypotonia-spasticity-sleep disorder 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 |
| 2476 | Dysraphism-cleft lip/palate-limb reduction defects syndrome | 3 Cases |
| 453510 | Congenital insensitivity to pain with severe intellectual disability | 3 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 457365 | Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome | 3 Cases |
| 320401 | Autosomal recessive spastic paraplegia 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 |
| 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 |

| ORPHA Number | Disease or Group of diseases | Number of 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 | Erythrokeratodermia-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-arachnodactyly-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 |
| 468620 | Intellectual disability-epilepsy-extrapyramidal syndrome | 3 Cases |
| 500144 | Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome | 3 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 485418 | EMILIN-1-related connective tissue disease | 3 Cases |
| 494348 | Early-onset familial noncirrhotic portal hypertension | 3 Cases |
| 495875 | Congenital labioscrotal 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 |
| 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 |

| ORPHA Number | Disease or Group of diseases | Number of 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 | Cleidorhizomelic 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 | Corneal-cerebellar syndrome | 2 Cases |
| 3199 | Stimmler 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-faciodigital 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 paraplegia-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 |
| 3368 | Trigonocephaly-bifid nose-acral anomalies syndrome | 2 Cases |
| 3200 | Arthrogryposis-ectodermal dysplasia-other anomalies syndrome | 2 Cases |
| 3167 | Siegler-Brewer-Carey syndrome | 2 Cases |
| 2519 | Microcephaly-seizures-intellectual disability-heart disease syndrome | 2 Cases |
| 2653 | Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome | 2 Cases |

| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 2666 | Adult familial nephronophthisis-spastic quadriparesia 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 166105 | FASTKD2-related infantile mitochondrial encephalomyopathy | 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-radioulnar synostosis syndrome | 2 Cases |
| 183707 | Neutrophil immunodeficiency syndrome | 2 Cases |
| 199348 | Thiamine-responsive encephalopathy | 2 Cases |
| 199329 | Congenital myopathy, Paradas type | 2 Cases |
| 293807 | Ketamine-induced biliary dilatation | 2 Cases |
| 294026 | 2q31.1 microduplication syndrome | 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 epidermodysplasia 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,XXXYY 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 2575 | Cystic fibrosis-gastritis-megaloblastic anemia syndrome | 2 Cases |
| 412217 | Dystonia-aphonia syndrome | 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 |
| 401764 | Pancytopenia-developmental delay 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 acidemia with homocystinuria, type cblJ | 2 Cases |
| 369929 | Aldosterone-producing adenoma with seizures and neurological abnormalities | 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 |
| 363424 | Multiple mitochondrial dysfunctions syndrome type 3 | 2 Cases |
| 352662 | Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome | 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 | Ameloonychohypohidrotic 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 |
| 457359 | Megalencephaly-severe kyphoscoliosis-overgrowth syndrome | 2 Cases |
| 96187 | Maternal uniparental disomy of chromosome 21 | 2 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 paraplegia 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 |
| 435660 | LIPE-related familial partial lipodystrophy | 2 Cases |
| 438207 | Severe autosomal recessive macrothrombocytopenia | 2 Cases |
| 431361 | Progressive encephalopathy with leukodystrophy due to DECR deficiency | 2 Cases |
| 440731 | L-ferritin deficiency | 2 Cases |

| ORPHA Number | Disease or Group of diseases | Number of 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-athetoid 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 | Prata-Liberal-Goncalves syndrome | 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 | Symbrachydactyly 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 |
| 2547 | Microphthalmia-microtia-fetal akinesia syndrome | 2 Cases |

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| ORPHA Number | Disease or Group of diseases | Number of 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 | Microtriplication 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 |
| 1804 | Dyssegmental dysplasia-glaucoma syndrome | 2 Cases |
| 2722 | Odonto-onycho dysplasia-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 |
| 1654 | Natal teeth-intestinal pseudoobstruction-patent ductus 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 |

| 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 | Orthostatic intolerance due to NET deficiency | 2 Cases |
| 254925 | Combined oxidative phosphorylation defect type 4 | 2 Cases |
| 502444 | Alkaline ceramidase 3 deficiency | 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 facioocculoskeletal 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 |
| 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 |
| 238459 | SLC35A1-CDG | 1 Case |
| 254334 | Autosomal recessive intermediate Charcot-Marie-Tooth disease type B | 1 Case |

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| ORPHA Number | Disease or Group of diseases | Number of cases |
|---|---|---|
| 250977 | AICA-ribosiduria | 1 Case |
| 300536 | DDOST-CDG | 1 Case |
| 324422 | ALG13-CDG | 1 Case |
| 330029 | Hypotrichosis-deafness syndrome | 1 Case |
| 330050 | Lethal encephalopathy due to mitochondrial and peroxisomal fission defect | 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 |
| 411712 | Maternal riboflavin deficiency | 1 Case |
| 401800 | Autosomal recessive spastic paraplegia type 60 | 1 Case |
| 401825 | Autosomal recessive spastic paraplegia type 68 | 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 |
| 240760 | Nijmegen breakage syndrome-like disorder | 1 Case |
| 1035 | Beta-mercaptolactate 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 |
| 317473 | Pancytopenia due to IKZF1 mutations | 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 |

## 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 |
| 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of families |
|---|---|---|
| 452 | X-linked lissencephaly with abnormal genitalia | 30 Families |
| 90026 | Primary erythermalgia | 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 paraplegia 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 |
| 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 |
| 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 |

| ORPHA Number | Disease or Group of diseases | Number of 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 |
| 178355 | Smith-McCort dysplasia | 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 | Otodental 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 |
| 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 proximal myopathy with early respiratory failure | 10 Families |
| 412057 | Autosomal recessive cerebellar ataxia due to STUB1 deficiency | 10 Families |
| 481662 | Familial Chilblain lupus | 10 Families |

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| ORPHA Number | Disease or Group of diseases | Number of 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 | Arthrogryposis-like syndrome | 8 Families |
| 1897 | EEM syndrome | 7 Families |
| 1777 | Temtamy syndrome | 7 Families |
| 86817 | Hemolytic anemia due to adenylate kinase deficiency | 7 Families |
| 100990 | Autosomal dominant spastic paraplegia type 9 | 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 |
| 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 | Spondyloepimetaphyseal 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 |
| 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 |
| 1275 | Brachydactyly-elbow wrist dysplasia syndrome | 4 Families |

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| ORPHA Number | Disease or Group of diseases | Number of 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-brachyectrodactyly 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 |
| 399103 | Distal nebulin myopathy | 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 |
| 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 paraplegia 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 complex spastic paraplegia 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-pseudocamptodactyly syndrome | 3 Families |
| 466921 | Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome | 3 Families |
| 1182 | Spastic ataxia with congenital miosis | 3 Families |
| 1349 | Maternally-inherited 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 |
| 114 | Auriculoosteodysplasia | 2 Families |
| 1867 | 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-brachydacty syndrome | 2 Families |
| 2405 | Thickened earlobes-conductive deafness syndrome | 2 Families |
| 2818 | Spastic paraplegia-glaucoma-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 |

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| ORPHA Number | Disease or Group of diseases | Number of 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 paraplegia 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 |
| 1092 | Renal-genital-middle ear anomalies | 2 Families |
| 2471 | McDonough syndrome | 2 Families |
| 420492 | Adult-onset cervical dystonia, DYT23 type | 2 Families |
| 447753 | Autosomal dominant complex spastic paraplegia type 9A | 2 Families |
| 447760 | Autosomal recessive complex 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 |
| 2885 | Piebald trait-neurologic defects 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, Rutherfurd 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 |
| 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 |

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

| ORPHA Number | Disease or Group of diseases | Number of families |
|---|---|---|
| 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 | Oculootodental 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 |
| 156728 | Spondyloepimetaphyseal dysplasia, matrilin-3 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 |
| 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 |

| ORPHA Number | Disease or Group of diseases | Number of families |
|---|---|---|
| 363727 | X-linked dyserythropoetic 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 |
| 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 |

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| ORPHA Number | Disease or Group of diseases | Number of families |
|---|---|---|
| 498602 | Sugarman brachydactyly | 1 Family |

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

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