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Prevalence of rare diseases: Bibliographic data

Listed in order of decreasing prevalence
or number of published cases

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A systematic survey of the literature is being performed in order to provide an estimate of the prevalence of rare diseases in Europe. An updated report will be published regularly and will replace the previous version. This update contains new epidemiological data and modifications to existing data for which new information has been made available.

Search strategy

The search strategy is carried out using several data sources:

- Websites: Orphanet, e-medicine, GeneClinics, EMA and OMIM ;
- Medline is consulted using the search algorithm:
«Disease names» AND Epidemiology[MeSH:NoExp]
OR Incidence[Title/abstract] OR Prevalence[Title/abstract] OR Epidemiology[Title/abstract] ;
- Medical books, grey literature and reports from experts are also important sources of data.

Collected data

Prevalence values provided are the mean of the highest and lowest values collected. When prevalence is not documented we calculate it using incidence:

- For congenital diseases with birth-onset, prevalence = incidence at birth x (patient life expectancy / general population life expectancy) ;
- For the other rare diseases, prevalence = incidence x rare disease mean duration ;

When no prevalence or incidence data are available, the number of cases reported in the literature is provided.

NB: Life expectancy of the French population (78 years) is used as the general population life expectancy.

Updated Data

New information from available data sources: EMA, new scientific publications, grey literature, expert opinion.

Limitation of the study

The exact prevalence rate of each rare disease is difficult to assess from the available data sources. There is a low level of consistency between studies, a poor documentation of methods used, confusion between incidence and prevalence, and/or confusion between incidence at birth and life-long incidence. The validity of the published studies is taken for granted and not assessed. It is likely that there is an overestimation for most diseases as the few published prevalence surveys are usually done in regions of higher prevalence and are usually based on hospital data. Therefore, these estimates are an indication of the assumed prevalence but may not be accurate.

List of diseases by decreasing prevalence

Diseases name	Estimated prevalence (/100,000)
Cleft palate	50
Congenital bilateral absence of vas deferens	50
Congenital toxoplasmosis	50
Cutaneous lupus erythematosus	50
Down syndrome	50
Hyperplastic polyposis syndrome	50
Isolated spina bifida	50
Noonan syndrome	50
Obesity due to melanocortin-4 receptor deficiency	50
Patent arterial duct	50
Fibrous dysplasia of bone	< 50
Familial melanoma	46.8
Congenital isolated thyroxine-binding globulin deficiency	46
Tetralogy of Fallot	45
Arrhythmogenic right ventricular dysplasia	43.5
Meniere disease	42.5
Trisomy X	42.5
Partial chromosome Y deletion	42
Scleroderma	42
Familial Parkinson's disease dementia	41
Familial long QT syndrome	40
Fetal cytomegalovirus syndrome	40
Romano-Ward syndrome	40
Squamous cell carcinoma of head and neck	< 40
Nonacquired combined pituitary hormone deficiency	37.7
Primary congenital hypothyroidism	37.5
Non-papillary transitional cell carcinoma of the bladder	37
Follicular lymphoma	36
Renal cell carcinoma	35.8
Hereditary elliptocytosis	35
Osteochondritis dissecans	35
Radiation proctitis	35
Cryptosporidiosis	34
Unilateral adactylia	34
Permanent congenital hypothyroidism	33.3
Alpha-1 antitrypsin deficiency	33
Malignant hyperthermia	33
Charcot-Marie-Tooth disease	32.5
Transposition of the great arteries	32.5
Retinitis pigmentosa	30.2

Diseases name	Estimated prevalence (/100,000)
Adult acute respiratory distress syndrome	30
Amyloidosis	30
Chronic B-cell lymphocytic leukemia	30
Polycythemia vera	30
Congenital hypothyroidism	29
Fragile X syndrome	28
Gastric cancer	28
Rare ovarian cancer	27.8
Post-transplant lymphoproliferative disease	26.2
Narcolepsy-cataplexy	26
Alopecia universalis	25
Esophageal atresia	25
Hereditary breast and ovarian cancer syndrome	25
Immune thrombocytopenic purpura	25
Neurofibromatosis type 1	25
Preaxial polydactyly	25
Syndactyly type 1	25
Thyroid hemiagenesis	25
Thrombotic thrombocytopenic purpura	24.6
Neovascular glaucoma	24.4
Essential thrombocythemia	24
Isolated anorectal malformation	24
Legg-Calve-Perthes disease	23
Myelodysplastic syndromes	22.8
Systemic sclerosis	21.5
Congenital hypothyroidism due to developmental anomaly	21.3
Vernal keratoconjunctivitis	21
Anal fistula	20.5
Oligoarticular juvenile arthritis	20.5
Dermatitis herpetiformis	20.2
Atresia of small intestine	20
Brugada syndrome	20
Congenital hypogonadotropic hypogonadism	20
Congenital sucrase-isomaltase deficiency	20
Diffuse large B-cell lymphoma	20
Hereditary spherocytosis	20
Hirschsprung disease	20
Isolated scaphocephaly	20
Marfan syndrome	20
Monosomy 22q11	20

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Myasthenia gravis	20
Partial atrioventricular canal	20
Soft tissue sarcomas	20
Tuberculosis	20
Turner syndrome	20
Corpus callosum agenesis - neuropathy	19
Idiopathic steroid-sensitive nephrotic syndrome	18
Pneumonia caused by serotype O1 Pseudomonas Aeruginosa	18
Familial isolated dilated cardiomyopathy	17.5
Multiple myeloma	17.5
Bilateral renal agenesis	17
Boutonneuse fever	17
Secondary amyloidosis	17
Idiopathic pulmonary fibrosis	16.7
X-linked ichthyosis	16.6
Rendu-Osler-Weber disease	16.25
Arthrogryposis multiplex congenita	16.1
Acute myeloid leukemia	16
Buerger's disease	16
MELAS syndrome	16
Stromal keratitis	16
Maple syrup urine disease	15.6
Atopic keratoconjunctivitis	15.1
Complete atrioventricular canal	15**
Congenital diaphragmatic hernia	15
Frontotemporal dementia with tau inclusions	15
Lennox-Gastaut syndrome	15
Medium chain acyl-CoA dehydrogenase deficiency	15
Microtia	15
Neuroleptic malignant syndrome	15
Sarcoidosis	15
Sickle cell anemia	15
Young adult-onset Parkinsonism	15
Cutaneous T-cell lymphoma	14.9
Dermatomyositis	14.8
Polymyositis	14.8
Citrullinemia	14.4
Thyroid ectopia	14.2
Cystinuria	14
Enteropancreatic endocrine tumor	14
Isolated anophthalmia - microphthalmia	14
Primary biliary cirrhosis	13.5
Stickler syndrome	13.5
Williams syndrome	13.3
Androgen insensitivity syndrome	13
Bronchopulmonary dysplasia	13

Diseases name	Estimated prevalence (/100,000)
Endocrine tumor	13
Gastrointestinal stromal tumor	13
Trisomy 13	13**
Cystic fibrosis	12.6
Ehlers-Danlos syndrome, hypermobile type	12.5
Lymphatic malformation	12.5
Supravalvular aortic stenosis	12.5
Von Willebrand disease	12.5
Gastroschisis	12
Omphalocele	12
Triple H syndrome	12
Pancreatic carcinoma	11.9
Focal dystonia	11.7
Neuroblastoma	11.3
MURCS association	11.25
Small cell lung cancer	11.2
Hepatic veno-occlusive disease	11
Multiple endocrine neoplasia type 1	11
Primary sclerosing cholangitis	11
Prader-Willi syndrome	10.7
Alopecia totalis	10.5
Collagenous colitis	10.5
Glial tumor	10.4
Stargardt disease	10.4
Hodgkin lymphoma, classical	10.2
Acute intermittent porphyria	10.1
Nephroblastoma	10.1
3-methylglutaconic aciduria type 3	10
Catecholaminergic polymorphic ventricular tachycardia	10
Catecholamine-producing tumor	10
Congenital adrenal hyperplasia	10
Congenital Leber amaurosis	10
Dermatofibrosarcoma protuberans	10
Distal myopathy, Welander type	10
Duane syndrome	10
Familial or sporadic hemiplegic migraine	10
Hairy cell leukemia	10
Hemophilia A	10
Hodgkin lymphoma	10
Idiopathic achalasia	10
Idiopathic hypereosinophilic syndrome	10
Isolated plagiocephaly	10
Mastocytosis	10
Meningococcal meningitis	10
Rare bone tumor	10
Recurrent acute pancreatitis	10

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
46,XX gonadal dysgenesis	< 10
Hereditary neuropathy with liability to pressure palsies	9
Mayer-Rokitansky-Küster-Hauser syndrome	9
Mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies	9
Neonatal hypoxic and ischemic brain injury	9
Trisomy 18	9**
Giant cell arteritis	8.9
Congenital lymphedema	8.8
Tuberous sclerosis	8.8
Isolated Pierre Robin syndrome	8.75
Duodenal atresia	8.55
Apnea of prematurity	8.5
Henoch-Schönlein purpura	8.5
NARP syndrome	8.5
Syringomyelia	8.4
Cutaneous lymphoma	8.3
Choanal atresia	8.2
Acute promyelocytic leukemia	8
CREST syndrome	8
Juvenile rheumatoid factor-negative polyarthritis	8
Limited cutaneous systemic sclerosis	8
Hyperlipidemia type 3	7.8
Hemophilia	7.7
Kallmann syndrome	7.7
Angelman syndrome	7.5
Beckwith-Wiedemann syndrome	7.3
Congenital pulmonary valve stenosis	7.2
Oculocutaneous albinism	7.15
Autosomal recessive cerebellar ataxia	7
Cystathioninuria	7
Facioscapulohumeral dystrophy	7
Fryns syndrome	7**
Holoprosencephaly	7
Huntington disease	7
Medullary thyroid carcinoma	7
Sotos syndrome	7**
Isolated trigonocephaly	6.7
Iminoglycinuria	6.68
Cat-scratch disease	6.6
Galactosemia	6.6
Wegener granulomatosis	6.6
Acute lymphoblastic leukemia	6.5
Autosomal recessive polycystic kidney disease	6.5
Carcinoma of the gallbladder	6.5
Leber hereditary optic neuropathy	6.5
Osteogenesis imperfecta	6.5

Diseases name	Estimated prevalence (/100,000)
Isolated ectopia lentis	6.4
Vasculitis	6.3
ACTH-dependent Cushing syndrome	6
Cerebral arteriovenous fistula	6
Chronic myeloid leukemia	6
Digitotalar dysmorphism	6
Optic atrophy	6
Progressive supranuclear palsy	6
Tibial muscular dystrophy	6
Treacher-Collins syndrome	6
Cushing syndrome	5.9
Wilson disease	5.84
Enthesitis-related arthritis	5.7
Biliary atresia	5.6
Familial adenomatous polyposis	5.5
Pendred syndrome	5.5
Retinoblastoma	5.4
Early-onset autosomal dominant Alzheimer disease	5.3
Smith-Magenis syndrome	5.3
Zollinger-Ellison syndrome	5.3
Amyotrophic lateral sclerosis	5.2
Hemolytic anemia due to red cell pyruvate kinase deficiency	5.1
Acromegaly	5
Buschke-Ollendorff syndrome	5
Congenital muscular dystrophy	5
Craniorachischisis	5
Duchenne and Becker muscular dystrophy	5
Ehlers-Danlos syndrome type 1	5
Familial spastic paraplegia	5
Fructose intolerance	5
Hydrolethalus	5**
Idiopathic hypersomnia	5
Idiopathic juvenile-onset systemic arthritis	5
Isolated brachycephaly	5
Multiple epiphyseal dysplasia	5
Osteosarcoma	5
Parietal foramina	5
Primary ciliary dyskinesia	5
Rheumatic fever	5
Thomsen and Becker disease	5
Tricuspid atresia	5
X-linked adrenoleukodystrophy	5
Tritanopia	4.8
Acrocephalosyndactyly	4.6
Monosomy 5p	4.6
Multiple system atrophy	4.6

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Achondroplasia	4.5
Congenital lobar emphysema	4.5
Steinert myotonic dystrophy	4.5
X-linked retinoschisis	4.5
Best disease	4.4
Chronic inflammatory demyelinating polyneuropathy	4.4
Juvenile psoriatic arthritis	4.2
Juvenile rheumatoid factor-positive polyarthritis	4.2
Hemimelia	4.15
Rett syndrome	4.15
Amniotic bands	4**
Common variable immunodeficiency	4
Corticobasal degeneration	4
Cushing disease	4
Cutaneous neuroendocrine carcinoma	4
Diffuse cutaneous systemic sclerosis	4
Esophageal carcinoma	4
Hartnup syndrome	4
Histidinemia	4
Mantle cell lymphoma	4
Meckel syndrome	4**
Phenylketonuria	4
Porphyria cutanea tarda	4
Acute interstitial pneumonia	3.8
Anisakiasis	3.8
Autosomal recessive limb girdle muscular dystrophy type 2A	3.8
Indolent systemic mastocytosis	3.8
Pemphigus vulgaris	3.8
Propionic acidemia	3.75
Duchenne muscular dystrophy	3.7
West syndrome	3.7**
Worster-Drought syndrome	3.7
Solar urticaria	3.6
Athyreosis	3.5
Autosomal dominant cerebellar ataxia	3.5
Ehlers-Danlos syndrome, classic type	3.5
Goldenhar syndrome	3.5
MASA syndrome	3.5
Thanatophoric dwarfism	3.5**
Thyroid hypoplasia	3.5
Usher syndrome	3.5
Guillain-Barré syndrome	3.45
Behcet disease	3.4
Short bowel syndrome	3.4
Achromatopsia	3.33
Central areolar choroidal dystrophy	3.33

Diseases name	Estimated prevalence (/100,000)
Congenital muscular dystrophy type 1A	3.3
Hypochondroplasia	3.3
Kennedy disease	3.3
Multiple endocrine neoplasia type 2	3.3
Parsonage-Turner syndrome	3.3
Smith-Lemli-Opitz syndrome	3.3
Systemic mastocytosis	3.3
Isolated anencephaly/exencephaly	3.2**
Mixed connective tissue disease	3.2
Moyamoya disease	3.16
Acatlasemia	3.1
Acute inflammatory demyelinating polyradiculoneuropathy	3.1
Mesothelioma	3.1
Polyarteritis nodosa	3.07
Bacterial toxic-shock syndrome	3
Frontotemporal dementia	3
Malaria	3
Nodular regenerative hyperplasia of the liver	3
Opitz BBB/G syndrome	3
Primary peritoneal tumor	3
Proximal spinal muscular atrophy	3
Saethre-Chotzen syndrome	3
Graft versus host disease	2.76
Leigh syndrome	2.75**
Myelofibrosis with myeloid metaplasia	2.7
Waldenström macroglobulinemia	2.6
Autosomal dominant diffuse palmoplantar keratoderma, Norrbotten type	2.5
Bilateral anorchia	2.5
BOR syndrome	2.5
Bullous pemphigoid	2.5
Cone rod dystrophy	2.5
Epidermolytic epidermolysis bullosa	2.5
Familial isolated restrictive cardiomyopathy	2.5
Fructose-1,6-bisphosphatase deficiency	2.5
Gitelman syndrome	2.5
Heterotaxia	2.5
Niemann-Pick disease	2.5**
Peripheral resistance to thyroid hormones	2.5
Progressive non-fluent aphasia	2.5
Pseudoxanthoma elasticum	2.5
Meconium aspiration syndrome	2.44
Waardenburg syndrome	2.4
3-methylcrotonylglycinuria	2.25
Ondine syndrome	2.25
Atypical Rett syndrome	2.22

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Diseases name	Estimated prevalence (/100,000)
Peutz-Jeghers syndrome	2.2	X-linked Charcot-Marie-Tooth disease	1.6
GRACILE syndrome	2.12**	Schizencephaly	1.54
Cholangiocarcinoma	2.1	Antisynthetase syndrome	1.5
Alport syndrome	2	Budd-Chiari syndrome	1.5
Anaplastic large cell lymphoma	2	CDG syndrome	1.5**
Cerebrotendinous xanthomatosis	2	Chronic hepatic porphyria	1.5
Childhood disintegrative disorder	2	Coffin-Lowry syndrome	1.5
Choroideremia	2	Darier disease	1.5
Coats disease	2	Femur-fibula-ulna complex	1.5
Congenital isolated hyperinsulinism	2	Hypereosinophilic syndromes	1.5
Craniopharyngioma	2	Idiopathic and/or familial pulmonary arterial hypertension	1.5
Crouzon disease	2	Laryngo-tracheo-esophageal cleft	1.5
Fibular hemimelia	2	Malignant peritoneal mesothelioma	1.5
Friedreich ataxia	2	Multifocal motor neuropathy with conduction block	1.5
Gaucher disease	2	Neuromyelitis optica	1.5
Hemophilia B	2	Pantothenate-kinase-associated neurodegeneration	1.5
Isolated Klippel-Feil syndrome	2	Primary lateral sclerosis	1.5
Kearns-Sayre syndrome	2	Spinocerebellar ataxia type 1	1.5
Langerhans cell histiocytosis	2	Spinocerebellar ataxia type 2	1.5
Large congenital melanocytic nevus	2	Proximal spinal muscular atrophy type 2	1.42
Limb body wall complex	2**	Alagille syndrome	1.4
Multiple osteochondromas	2	Ornithine transcarbamylase deficiency	1.4
Nail-patella syndrome	2	Cat-eye syndrome	1.35
Neurodegeneration with brain iron accumulation	2	Netherton disease	1.35
Non-distal trisomy 12p	2**	Late infantile neuronal ceroid lipofuscinosis	1.3
Paroxysmal hemicrania	2	Mucopolysaccharidosis type 1	1.3
Poland syndrome	2	Usher syndrome type 1	1.3
Sarcosinemia	2	Apert syndrome	1.25
Scimitar syndrome	2	Ebstein malformation	1.25
Usher syndrome type 2	2	Maternal hyperphenylalaninemia	1.25
Van Der Woude syndrome	2	Proximal spinal muscular atrophy type 1	1.25
Wolf-Hirschhorn syndrome	2**	Adult Still's disease	1.23
X-linked recessive ocular albinism	2	Diastrophic dwarfism	1.2
Sternal cleft	< 2	Oral-facial-digital syndrome type 1	1.2
Autosomal recessive limb-girdle muscular dystrophy type 2C	1.96	Superficial pemphigus	1.2
Cornelia de Lange syndrome	1.9	Kabuki syndrome	1.16
Von Hippel-Lindau disease	1.9	Glycogen storage disease type 2	1.1
Muenke syndrome	1.8**	Ligneous conjunctivitis	1.1
Amoebiasis due to free-living amoebae	1.75	Mucopolysaccharidosis type 3	1.1
Aniridia	1.75	Split hand - split foot	1.1
Fabry disease	1.75	Zellweger syndrome	1.1
2,8 dihydroxyadenine urolithiasis	1.7	Autosomal recessive medullary cystic kidney disease	1.05
Kaposi's sarcoma	1.7	Cutis verticis gyrata - intellectual deficit	1.02
Walker-Warburg syndrome	1.65**	Acanthamoeba keratitis	1
Biotinidase deficiency	1.6	Adrenocortical carcinoma	1
Pseudoachondroplasia	1.6	Albers-Schönberg osteopetrosis	1

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Ataxia-telangiectasia	1
Atypical hemolytic uremic syndrome	1
Chondrodysplasia punctata, rhizomelic type	1
Chronic hiccup	1
Churg-Strauss syndrome	1
Clouston syndrome	1
Congenital dyserythropoietic anemia	1
Congenital hypothyroidism due to transplacental passage of maternal TSH-binding inhibitory antibodies	1
Congenital Rubella syndrome	1**
Crigler-Najjar syndrome	1
Currarino triad	1
Ehlers-Danlos syndrome, vascular type	1
Glioblastoma	1
Gorlin syndrome	1
Harding ataxia	1
Hepatocellular carcinoma	1
Hereditary angioedema	1
Holt-Oram syndrome	1
Hypokalemic periodic paralysis	1
Isovaleric acidemia	1
Joubert syndrome	1
Lambert-Eaton myasthenic syndrome	1
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	1
Macrophagic myofasciitis	1
Malignant tumor of fallopian tube	1
Mixed cryoglobulinemia	1
Nemaline myopathy	1
Nijmegen breakage syndrome	1**
Ocular coloboma	1
Oculopharyngeal muscular dystrophy	1
Pfeiffer syndrome	1
Proximal myotonic myopathy	1
Pseudomyxoma peritonei	1
Pure autonomic failure	1
Rubinstein-Taybi syndrome	1
Sirenomelia	1**
Spinocerebellar ataxia type 3	1
Spondylometaphyseal dysplasia	1
Tracheal agenesis	1**
Acalvaria	< 1**
Familial amyloid polyneuropathy	< 1
Hyperlipoproteinemia type 1	< 1
Gaucher disease type 1	0.94
Erythropoietic protoporphyria	0.9
Lewis-Sumner syndrome	0.9
MERRF syndrome	0.9

Diseases name	Estimated prevalence (/100,000)
Niemann-Pick disease type C	0.85
Bardet-Biedl syndrome	0.8
Carbamoylphosphate synthetase deficiency	0.8
Criss-cross heart	0.8
Hereditary epidermolysis bullosa	0.8
Limb-girdle muscular dystrophy	0.8
Severe hemophilia B	0.8
Autosomal recessive malignant osteopetrosis	0.75**
Chronic autoimmune hepatitis	0.75
Congenital myasthenic syndromes	0.75
Cutaneous mastocytosis	0.75
Krabbe disease	0.75**
Sandhoff disease	0.75
Albright hereditary osteodystrophy	0.72
Dystrophic epidermolysis bullosa	0.7
46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	0.68
Goodpasture syndrome	0.64
Anomaly of bile acid synthesis	0.6
Glycogen branching enzyme deficiency	0.6
Mild hemophilia B	0.6
Moderately severe hemophilia B	0.6
Mucopolysaccharidosis type 2	0.6
Progressive supranuclear palsy - corticobasal syndrome	< 0.6
Autosomal recessive limb-girdle muscular dystrophy type 2D	0.57
Autosomal recessive limb-girdle muscular dystrophy type 2E	0.57
Autosomal recessive limb-girdle muscular dystrophy type 2F	0.57
Hurler syndrome	0.57
Wolfram syndrome	0.57
Lymphangioliomyomatosis	0.56
Cantrell pentalogy	0.55**
McCune-Albright syndrome	0.55
Paroxysmal nocturnal hemoglobinuria	0.55
Spondyloepiphyseal dysplasia tarda	0.55
Congenital muscular dystrophy, Fukuyama type	0.54
Alkaptonuria	0.5
Axenfeld-Rieger syndrome	0.5
Beta-thalassemia	0.5
Birt-Hogg-Dube syndrome	0.5
Combined deficiency of factor V and factor VIII	0.5
Cystinosis	0.5
Hyperkalemic periodic paralysis	0.5
Nephrogenic diabetes insipidus	0.5
Neurofibromatosis type 2	0.5
X-linked dominant chondrodysplasia punctata	0.5

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Sporadic inclusion body myositis	0.49
Dentatorubral-pallidolusian atrophy	0.48
Juvenile neuronal ceroid lipofuscinosis	0.46
Argininosuccinic aciduria	0.45
Cowden syndrome	0.45
Takayasu arteritis	0.45
Werner syndrome	0.45
X-linked agammaglobulinemia	0.45
Mild hemophilia A	0.44
Severe hemophilia A	0.44
Townes-Brocks syndrome	0.42
Autosomal dominant severe congenital neutropenia	0.4**
Congenital bullous ichthyosiform erythroderma	0.4
Early onset torsion dystonia	0.4
Glutaric acidemia type 1	0.4
Homocystinuria due to cystathionine beta-synthase deficiency	0.4
Idiopathic aplastic anemia	0.4
Mucopolysaccharidosis type 4	0.4
Niemann-Pick disease type B	0.4
Severe congenital neutropenia	0.4
Sjögren-Larsson syndrome	0.4
Lesch-Nyhan syndrome	0.38
Campomelic dysplasia	0.35
Christ-Siemens-Touraine syndrome	0.35
Relapsing polychondritis	0.35
Spondyloepiphyseal dysplasia congenita	0.34
Lamellar ichthyosis	> 0.33
Blackfan-Diamond disease	0.32
Proximal spinal muscular atrophy type 4	0.32
Aortic arch interruption	0.3**
Autosomal dominant popliteal pterygium syndrome	0.3
Dopa-responsive dystonia	0.3
Emery-Dreifuss muscular dystrophy	0.3
Fanconi anemia	0.3
Frontotemporal dementia and parkinsonism linked to chromosome 17	0.3
Jervell and Lange-Nielsen syndrome	0.3
Miller-Dieker syndrome	0.3
Mirror polydactyly - vertebral segmentation - limbs defects	0.3
Sanfilippo syndrome type A	0.3
Tay-Sachs disease	0.3**
Transmissible spongiform encephalopathies	0.3
Wolman disease	0.28**
Proximal spinal muscular atrophy type 3	0.26
Berardinelli-Seip congenital lipodystrophy	0.25

Diseases name	Estimated prevalence (/100,000)
Congenital factor VII deficiency	0.25
Niemann-Pick disease type A	0.25**
Papillon-Lefèvre syndrome	0.25
Pelizaeus-Merzbacher disease	0.25
Piebaldism	0.25
Leptospirosis	0.24
Hurler-Scheie syndrome	0.23
Barth syndrome	0.22
Moderately severe hemophilia A	0.22
Severe combined immunodeficiency due to adenosine deaminase deficiency	0.22
Acrodermatitis enteropathica, zinc deficiency type	0.2
Aggressive systemic mastocytosis	0.2
Chronic granulomatous disease	0.2
Congenital factor X deficiency	0.2
Ehlers-Danlos syndrome	0.2
Extraskelatal myxoid chondrosarcoma	0.2
Hereditary thrombophilia due to congenital protein C deficiency	0.2
Hereditary thrombophilia due to congenital protein S deficiency	0.2
Hyperoxaluria	0.2
Incontinentia pigmenti	0.2
Isolated nonketotic hyperglycinemia	0.2
Jeune syndrome	0.2
Laron syndrome	0.2
Letterer-Siwe disease	0.2
Neonatal diabetes mellitus	0.2
Primary hyperoxaluria type 1	0.2
Scheie syndrome	0.2
Unverricht-Lundborg disease	0.2
Oculocerebrorenal syndrome	0.19
Sezary's syndrome	0.18
Atypical coarctation of aorta	0.17**
Hyperargininemia	0.17
Metachromatic leukodystrophy	0.16
Mucopolysaccharidosis type 6	0.16**
Congenital fibrinogen deficiency	0.15
Hermansky-Pudlak syndrome	0.15
Mucopolipidosis type 2	0.15**
Polycystic lipomembranous osteodysplasia - sclerosing leukoencephalopathy	0.15
Pyridoxine-dependent epilepsy	0.15
Wiskott-Aldrich syndrome	0.15
Alström syndrome	0.14
CHARGE syndrome	0.14
Pycnodysostosis	0.13
Hereditary chronic pancreatitis	0.125

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Bartter syndrome	0.12
Lissencephaly type 2	0.12
Autosomal dominant medullary cystic kidney disease with or without hyperuricemia	0.11
Aceruloplasminemia	0.1
Acquired hemophilia	0.1
Acute motor axonal neuropathy	0.1
Acute motor-sensory axonal neuropathy	0.1
Adult-onset proximal spinal muscular atrophy, autosomal dominant	0.1
Alpha-mannosidosis	0.1
Anaplastic thyroid carcinoma	0.1
Childhood-onset proximal spinal muscular atrophy, autosomal dominant	0.1
Colchicine poisoning	0.1
Congenital factor V deficiency	0.1
Congenital factor XI deficiency	0.1
Creutzfeldt-Jakob disease	0.1
Cyclic neutropenia	0.1
Distal myopathy, Nonaka type	0.1
Dyskeratosis congenita	0.1
Evans syndrome	0.1
Ewing sarcoma	0.1
Familial cold urticaria	0.1
Geroderma osteodysplastica	0.1
Idiopathic pulmonary alveolar proteinosis	0.1
Juvenile myelomonocytic leukemia	0.1
Lemierre syndrome	0.1
Leprechaunism	0.1**
Maternally inherited diabetes and deafness	0.1
Refsum disease	0.1
Senior-Loken syndrome	0.1

Diseases name	Estimated prevalence (/100,000)
Spondylometaphyseal dysplasia, Kozlowski type	0.1
Stiff-man syndrome	0.1
Tibial aplasia - ectrodactyly	0.1
Tibial hemimelia	0.1
Xeroderma pigmentosum	0.1
X-linked diffuse leiomyomatosis - Alport syndrome	0.1
X-linked lymphoproliferative disease	0.1
Lafora disease	< 0.1
Aicardi syndrome	0.06
Fibrodysplasia ossificans progressiva	0.06
Junctional epidermolysis bullosa	0.06
Mendelian susceptibility to atypical mycobacteria	0.059
Botulism	0.05
Chordoma	0.05
Congenital factor II deficiency	0.05
Congenital factor XIII deficiency	0.05
Craniofacial dysynostosis	0.05
Gaucher disease type 3	0.05
Osteoporosis - pseudoglioma	0.05
Tyrosinemia type 1	0.05
Naegeli-Franceschetti-Jadassohn syndrome	0.035
Congenital muscular dystrophy with integrin deficiency	0.03
Alpers syndrome	0.025
Paroxysmal non-kinesigenic dyskinesia	0.02
Sialidosis type 1	0.02**
Sialidosis type 2	0.02**
Gaucher disease type 2	0.01
Perinatal-lethal Gaucher disease	0.01
Hutchinson-Gilford progeria syndrome	0.005
Infantile Refsum disease	0.005

** Prevalence at birth

List of diseases by decreasing number of published cases

Diseases name	Number of published cases or families	Diseases name	Number of published cases or families
Thalidomide embryopathy	5000 cases	Rothmund-Thomson syndrome	300 cases
Angio-osteohypertrophic syndrome	1000 cases	Lipoid proteinosis	> 280 cases
Whipple disease	1000 cases	Eosinophilic gastroenteritis	280 cases
Alveolar echinococcosis	< 1000 cases	Juvenile chronic recurrent multifocal osteomyelitis	> 260 cases
Esthesioneuroblastoma	< 1000 cases	Autosomal dominant hyper IgE syndrome	250 cases
Rickettsialpox	> 800 cases	Dent disease	250 cases
Fetal methylmercury syndrome	800 cases	Maffucci syndrome	250 cases
Malakoplasia	> 700 cases	Partial acquired lipodystrophy	250 cases
Western equine encephalitis	> 600 cases	Caroli disease	< 250 cases
Enchondromatosis	600 cases	6-pyruvoyl-tetrahydropterin synthase deficiency	248 cases
Familial dysautonomia	550 cases	Oculodentodigital dysplasia	243 cases
Adamantinoma	513 cases	Pachyonychia congenita	230 cases
Ovotesticular disorder of sex development	> 500 cases	Lhermitte-Duclos disease	220 cases
CADASIL syndrome	500 cases	Muir-Torre syndrome	205 cases
Cronkhite-Canada syndrome	500 cases	Pachydermoperiostosis	204 cases
Rhabdoid tumor	500 cases	Aarskog-Scott syndrome	> 200 cases
Sinus histiocytosis with massive lymphadenopathy	500 cases	Blue rubber bleb nevus	> 200 cases
Epidermal nevus syndrome	> 400 cases	Congenital erythropoietic porphyria	> 200 cases
Castleman disease	400 cases	Congenital pseudoarthrosis of clavicle	> 200 cases
Silver-Russell syndrome	400 cases	Erythrokeratoderma variabilis, Mendes da Costa type	> 200 cases
4-hydroxybutyricaciduria	350 cases	Malignant atrophic papulosis	> 200 cases
Erdheim-Chester disease	350 cases	Monosomy 22q13	> 200 cases
Leukocyte adhesion deficiency	< 350 cases	Tufted angioma	> 200 cases
Carnitine palmitoyl transferase II deficiency	> 300 cases	Adenosine monophosphate deaminase deficiency	200 cases
Acquired Von Willebrand syndrome	300 cases	Camurati-Engelmann disease	200 cases
Alexander disease	300 cases	Cockayne syndrome	200 cases
CDG syndrome type Ia	300 cases	Cogan syndrome	200 cases
Cutis marmorata telangiectatica congenita	300 cases	Costello syndrome	200 cases
Familial partial lipodystrophy, Dunnigan type	300 cases	Eosinophilic fasciitis	200 cases
Focal dermal hypoplasia	300 cases	Glucose-galactose malabsorption	200 cases
Gräsbeck-Imerslund disease	300 cases	Gollop-Wolfgang complex	200 cases
Melorheostosis	300 cases	Gorham-Stout disease	200 cases
Methylmalonic acidemia - homocystinuria	300 cases	Kimura disease	200 cases
Methylmalonicacidemia - homocystinuria, type cbl C	300 cases	LEOPARD syndrome	200 cases
Moebius syndrome	300 cases	Lethal multiple pterygium syndrome	200 cases
Norrie disease	300 cases	Marinesco-Sjögren syndrome	200 cases

Diseases name	Number of published cases or families
Proteus syndrome	200 cases
Robinow syndrome	200 cases
Shwachman-Diamond syndrome	200 cases
Subcorneal pustular dermatosis	200 cases
Bilateral striopallidodentate calcinosis	< 200 cases
Monosomy 18p	< 200 cases
Mowat-Wilson syndrome	< 200 cases
Multicentric reticulohistiocytosis	< 200 cases
X-linked sideroblastic anemia	< 200 cases
Hyperimmunoglobulinemia D with recurrent fever	180 cases
Kasabach-Merritt syndrome	> 175 cases
Celiac disease - epilepsy - occipital calcifications	170 cases
Alpha thalassemia - X-linked intellectual deficit	168 cases
Carney complex	160 cases
Madras motor neuron disease	154 cases
Infantile neuroaxonal dystrophy	> 150 cases
Denys-Drash syndrome	150 cases
Dubowitz syndrome	150 cases
Ellis Van Creveld syndrome	150 cases
Fraser syndrome	150 cases
Isolated cloverleaf skull syndrome	150 cases
Isolated humeroradial synostosis	150 cases
Jacobsen syndrome	150 cases
McLeod neuroacanthocytosis syndrome	150 cases
Xanthinuria	150 cases
Roberts syndrome	< 150 cases
Tyrosinemia type 2	< 150 cases
CACH syndrome	148 cases
Bazex syndrome	145 cases
Bazex-Dupre-Christol syndrome	143 cases
Inflammatory pseudotumor of the liver	143 cases
Regional odontodysplasia	139 cases
Thyrotoxic periodic paralysis	139 cases
Dihydropteridine reductase deficiency	134 cases
Aorto-ventricular tunnel	130 cases
Weill-Marchesani syndrome	128 cases
Vulvovaginal gingival syndrome	127 cases
Aicardi-Goutieres syndrome	120 cases
Macrocephaly - capillary malformation	116 cases
MULIBREY nanism	115 cases
Bickel-Fanconi glycogenosis	112 cases
Myoclonic epilepsy of infancy	106 cases
Achondrogenesis	> 100 cases
Acquired epidermolysis bullosa	> 100 cases
Acquired generalized lipodystrophy	> 100 cases
Bloom syndrome	> 100 cases

Diseases name	Number of published cases or families
Central neurocytoma	> 100 cases
Congenital pulmonary lymphangiectasia	> 100 cases
Cutis laxa	> 100 cases
Fetal varicella syndrome	> 100 cases
Idiopathic acute eosinophilic pneumonia	> 100 cases
KID syndrome	> 100 cases
Mucopolipidosis type 4	> 100 cases
Ochoa syndrome	> 100 cases
Rasmussen subacute encephalitis	> 100 cases
Simpson-Golabi-Behmel syndrome	> 100 cases
Trichorhinophalangeal syndrome type 1 and 3	> 100 cases
Autoimmune lymphoproliferative syndrome	100 cases
Bernard-Soulier syndrome	100 cases
Bethlem myopathy	100 cases
Carney triad	100 cases
Chaotic atrial tachycardia	100 cases
CINCA syndrome	100 cases
Cohen syndrome	100 cases
Congenital cataracts - facial dysmorphism - neuropathy	100 cases
Freeman-Sheldon syndrome	100 cases
Fucosidosis	100 cases
Greig cephalopolysyndactyly syndrome	100 cases
Helicoid peripapillary chorioretinal degeneration	100 cases
Immunodeficiency by defective expression of HLA class 2	100 cases
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	100 cases
Internal carotid agenesis	100 cases
Larsen syndrome	100 cases
Monoclonal Ig light chain-associated Fanconi syndrome	100 cases
Neonatal hemochromatosis	100 cases
Osteopathia striata - cranial sclerosis	100 cases
Pallister-Hall syndrome	100 cases
PHACE syndrome	100 cases
Poikiloderma of Kindler	100 cases
Retinal arteries tortuosity	100 cases
Schnitzler syndrome	100 cases
Schwartz-Jampel syndrome	100 cases
Seckel syndrome	100 cases
Triple A syndrome	100 cases
Arthrogryposis - renal dysfunction - cholestasis	< 100 cases
Brown-Vialetto-van Laere syndrome	< 100 cases
Congenital muscular dystrophy, Ullrich type	< 100 cases
Cytophagic histiocytic panniculitis	< 100 cases
Haim-Munk syndrome	< 100 cases

Diseases name	Number of published cases or families
Hallermann-Streiff-François syndrome	< 100 cases
Hypertrichosis lanuginosa congenita	< 100 cases
Hypocomplementemic leucocytoclastic vasculitis	< 100 cases
Ichthyosis congenita, harlequin type	< 100 cases
Recessive hereditary methemoglobinemia type 2	< 100 cases
Susac syndrome	< 100 cases
Tracheobronchomegaly	< 100 cases
X-linked spastic paraplegia type 2	< 100 cases
Silent sinus syndrome	98 cases
Acrofacial dysostosis, Nager type	90 cases
Allan-Herndon-Dudley syndrome	89 cases
Megacystis - microcolon - intestinal hypoperistalsis - hydronephrosis	89 cases
Early infantile epileptic encephalopathy	88 cases
Myoneurogastrointestinal encephalopathy syndrome	87 cases
Encephalopathy due to GLUT1 deficiency	84 cases
Glycogen storage disease due to LAMP-2 deficiency	84 cases
Uhl anomaly	84 cases
Liddle syndrome	80 cases
Metatropic dwarfism	80 cases
Wells syndrome	80 cases
Arterial tortuosity syndrome	< 80 cases
Cerebro-costo-mandibular syndrome	75 cases
Rapp-Hodgkin syndrome	72 cases
Tangier disease	> 70 cases
Bullous systemic lupus erythematosus	70 cases
Cranio metaphyseal dysplasia	70 cases
Median cleft lip/mandibule	70 cases
Pseudohypoaldosteronism type 1	70 cases
Ring chromosome 18	70 cases
Diffuse neonatal hemangiomatosis	< 70 cases
Glutathione synthetase deficiency	65 cases
Hereditary hyperferritinemia with congenital cataracts	> 64 cases
Paraneoplastic pemphigus	> 60 cases
Acquired hypertrichosis lanuginosa	60 cases
Dyggve-Melchior-Clausen disease	60 cases
Griselli disease	60 cases
Homocystinuria without methylmalonic aciduria	60 cases
Ketoacidosis due to betaketothiolase deficiency	60 cases
Neu-Laxova syndrome	60 cases
Pancreatoblastoma	60 cases
Pearson syndrome	60 cases
Sea-blue histiocytosis	60 cases
Toriello-Carey syndrome	60 cases
Vitamin B12 responsive methylmalonic acidemia type cbl A	60 cases

Diseases name	Number of published cases or families
Congenital alveolar capillary dysplasia	< 60 cases
Double uterus - hemivagina - renal agenesis	< 60 cases
Wolcott-Rallison syndrome	< 60 cases
Autosomal recessive ataxia, Beauce type	57 cases
Capillary leak syndrome	57 cases
Sporotrichosis	55 cases
Mazabraud syndrome	54 cases
Congenital megacalycosis	> 50 cases
Frasier syndrome	> 50 cases
Hennekam syndrome	> 50 cases
Osteodysplasty, Melnick-Needles type	> 50 cases
Ring chromosome 20	> 50 cases
Acromesomelic dysplasia, Maroteaux type	50 cases
Acroosteolysis dominant type	50 cases
Adenylosuccinate lyase deficiency	50 cases
Ascher syndrome	50 cases
Benign paroxysmal torticollis of infancy	50 cases
Cholestasis - lymphedema	50 cases
Encephalopathy due to sulfite oxidase deficiency	50 cases
Familial glucocorticoid deficiency	50 cases
Focal myositis	50 cases
Hemolytic anemia due to glucophosphate isomerase deficiency	50 cases
ICF syndrome	50 cases
Juvenile hyaline fibromatosis	50 cases
Juvenile macular degeneration - hypotrichosis	50 cases
Juvenile Paget's disease	50 cases
Multiple sulfatase deficiency	50 cases
Neutral lipid storage disease	50 cases
Ocular motor apraxia, Cogan type	50 cases
Osteopetrosis with renal tubular acidosis	50 cases
Partial pancreas agenesis	50 cases
Peters-plus syndrome	50 cases
Pitt-Hopkins syndrome	50 cases
Prolidase deficiency	50 cases
Rhombencephalosynapsis	50 cases
Ring chromosome 14	50 cases
Schimke immuno-osseous dysplasia	50 cases
Succinic acidemia	50 cases
Waardenburg-Shah syndrome	50 cases
Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis	< 50 cases
Blepharo-cheilo-odontic syndrome	< 50 cases
Branchio-oculo-facial syndrome	< 50 cases
Cholesteryl ester storage disease	< 50 cases
Congenital analbuminemia	< 50 cases

Diseases name	Number of published cases or families
DOOR syndrome	< 50 cases
Ehrlichiosis	< 50 cases
Feingold syndrome	< 50 cases
Fibular aplasia - ectrodactyly	< 50 cases
Floating-Harbor syndrome	< 50 cases
Goldmann-Favre syndrome	< 50 cases
Granulomatous slack skin	< 50 cases
Hypoglossia - hypodactyly	< 50 cases
MIDAS syndrome	< 50 cases
Shprintzen-Goldberg syndrome	< 50 cases
Spinocerebellar ataxia type 29	< 50 cases
Corticosteroid-sensitive aseptic abscesses	49 cases
Pityriasis rubra pilaris	48 cases
Coxo-podo-patellar syndrome	47 cases
Mohr-Tranebjaerg syndrome	46 cases
Cone dystrophy with supernormal rod response	45 cases
Encephalocraniocutaneous lipomatosis	45 cases
KBG syndrome	45 cases
Bowen-Conradi syndrome	44 cases
Seborrhea-like dermatitis with psoriasiform elements	44 cases
Ear-patella-short stature syndrome	42 cases
Desbuquois syndrome	> 40 cases
3M syndrome	40 cases
Ambras syndrome	40 cases
Autosomal dominant hypohidrotic ectodermal dysplasia	40 cases
Babesiosis	40 cases
Carpenter syndrome	40 cases
Coffin-Siris syndrome	40 cases
Distal monosomy 10q	40 cases
Distal trisomy 10q	40 cases
Distal trisomy 6p	40 cases
Galloway-Mowat syndrome	40 cases
Methimazole embryofetopathy	40 cases
Sitosterolemia	40 cases
WHIM syndrome	40 cases
Acromicric dysplasia	< 40 cases
Bruck syndrome	< 40 cases
Ethylmalonic encephalopathy	< 40 cases
Macrocephaly-autism syndrome	< 40 cases
Mucopolysaccharidosis type 7	< 40 cases
Progressive bulbar paralysis of childhood	< 40 cases
Leukoencephalopathy with brain stem and spinal cord involvement - lactate elevation	39 cases
Autosomal dominant rhegmatogenous retinal detachment	38 cases
Hypotrichosis simplex	38 cases

Diseases name	Number of published cases or families
Mandibuloacral dysplasia	37 cases
Hereditary North American Indian childhood cirrhosis	36 cases
Oculocerebrocutaneous syndrome	36 cases
Spondyloenchondrodysplasia	36 cases
Cobb syndrome	35 cases
Hereditary sensory and autonomic neuropathy type 2	35 cases
Acrocallosal syndrome, Schinzel type	34 cases
Antley-Bixler syndrome	34 cases
Midface retraction syndrome, Schinzel-Giedion type	34 cases
Perrault syndrome	34 cases
Pyogenic arthritis - pyoderma gangrenosum - acne	34 cases
Ring chromosome 1	34 cases
Autosomal dominant osteopetrosis type 1	33 cases
Björnstad syndrome	33 cases
Marshall-Smith syndrome	33 cases
Methylcobalamin deficiency, cbl G type	33 cases
Polycystic ovaries - urethral sphincter dysfunction	33 cases
Double outlet left ventricle	32 cases
Olmsted syndrome	32 cases
Segmental odontomaxillary dysplasia	32 cases
Argininemia	31 cases
CDG syndrome type Ic	> 30 cases
Congenital intrauterine infection-like syndrome	> 30 cases
Tricho-dento-osseous syndrome	> 30 cases
Trisomy 8q	> 30 cases
Weaver-Williams syndrome	> 30 cases
Agnathia - holoprosencephaly - situs inversus	30 cases
Anophthalmia - hypothalamo-pituitary insufficiency	30 cases
Anophthalmia/microphthalmia - esophageal atresia	30 cases
Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	30 cases
Camptodactyly - tall stature - scoliosis - hearing loss	30 cases
Carnitine-acylcarnitine translocase deficiency	30 cases
Carnosinemia	30 cases
Cataract - cardiomyopathy	30 cases
CHILD syndrome	30 cases
Cranio-osteoarthropathy	30 cases
Early myoclonic encephalopathy	30 cases
Elejalde disease	30 cases
Glossopalatine ankylosis	30 cases
Humeroradioulnar synostosis	30 cases
Lethal restrictive dermopathy	30 cases
Marden-Walker syndrome	30 cases
Marshall's syndrome with periodic fever	30 cases
Mevalonic aciduria	30 cases
Microphthalmia with limb anomalies	30 cases

Diseases name	Number of published cases or families
Omodysplasia	30 cases
Otopalatodigital syndrome	30 cases
Perlman syndrome	30 cases
Scalp-ear-nipple syndrome	30 cases
SHORT syndrome	30 cases
Triose phosphate-isomerase deficiency	30 cases
Weaver syndrome	30 cases
Acropectorovertebral dysplasia	< 30 cases
Crisponi syndrome	< 30 cases
Developmental delay due to 2-methylbutyryl-CoA dehydrogenase deficiency	< 30 cases
Ectodermal dysplasia - absent dermatoglyphs	< 30 cases
Encephalopathy due to hydroxykynureninuria	< 30 cases
Frontometaphyseal dysplasia	< 30 cases
Glycogen storage disease due to muscle phosphorylase kinase deficiency	< 30 cases
IRVAN syndrome	< 30 cases
Linear atrophoderma of Moulin	< 30 cases
Microcephalic osteodysplastic primordial dwarfism types 1 and 3	< 30 cases
Muscle phosphofructokinase deficiency	< 30 cases
Nasopalpebral lipoma - coloboma - telecanthus	< 30 cases
Neuroectodermal syndrome, Johnson type	< 30 cases
Neurologic Waardenburg-Shah syndrome	< 30 cases
Neurometabolic disorder due to serine deficiency	< 30 cases
Obesity due to congenital leptin deficiency	< 30 cases
Otospondylomegaepiphyseal dysplasia	< 30 cases
Pontocerebellar hypoplasia type 2	< 30 cases
Postaxial acrofacial dysostosis	< 30 cases
Pyle disease	< 30 cases
Ring dermoid of cornea	< 30 cases
Severe neonatal-onset encephalopathy with microcephaly	< 30 cases
Spontaneous periodic hypothermia	< 30 cases
Syndactyly, Cenani-Lenz type	< 30 cases
Wrinkly skin syndrome	< 30 cases
Infant epilepsy with migrant focal crisis	29 cases
Jalili syndrome	29 cases
Joubert syndrome with orofaciadigital defect	29 cases
Leukoencephalopathy with bilateral anterior temporal lobe cysts	29 cases
Mosaic variegated aneuploidy syndrome	29 cases
Symmetrical thalamic calcifications	29 cases
Craniofaciosutural dysplasia	28 cases
Hypertrichosis cubiti - short stature	28 cases
Catel-Manzke syndrome	27 cases
Fatal familial insomnia	27 cases

Diseases name	Number of published cases or families
GAP0 syndrome	27 cases
Geleophysic dysplasia	27 cases
Limb-mammary syndrome	27 cases
Metaphyseal anadysplasia	27 cases
Methylcobalamin deficiency type cbl E	27 cases
3C syndrome	25 cases
Atelosteogenesis type II	25 cases
Book syndrome	25 cases
Dirofilariasis	25 cases
Erythrokeratoderma - ataxia	25 cases
Metachondromatosis	25 cases
Opsismodysplasia	25 cases
Palmoplantar keratoderma - spastic paralysis	25 cases
Plummer-Vinson syndrome	25 cases
Porokeratotic eccrine ostial and dermal duct nevus	25 cases
Wiedemann-Rautenstrauch syndrome	25 cases
Filippi syndrome	< 25 cases
Hepatic veno-occlusive disease - immunodeficiency	< 25 cases
Bartsocas-Papas syndrome	24 cases
Distal myopathy with early respiratory muscle involvement	24 cases
Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	24 cases
Infantile onset spinocerebellar ataxia	24 cases
Synspondylism	24 cases
X-linked spastic paraplegia type 34	24 cases
2q24 microdeletion syndrome	23 cases
Cantu syndrome	23 cases
Congenital bronchobiliary fistula	23 cases
Johanson-Blizzard syndrome	23 cases
Phosphoglycerate kinase 1 deficiency	23 cases
Potocki-Shaffer syndrome	23 cases
Treft-Sanborn-Carey syndrome	23 cases
Acro-pectoral syndrome	22 cases
Aortic dilatation - joint hypermobility - arterial tortuosity	22 cases
Pierson syndrome	22 cases
Speech-language disorder type 1	22 cases
Split hand - split foot - deafness	22 cases
Dehydratase deficiency	21 cases
Odonto-tricho-ungual-digito-palmar syndrome	21 cases
Sudden infant death - dysgenesis of the testes	21 cases
Fumaric aciduria	> 20 cases
Rubella panencephalitis	> 20 cases
3-methylglutaconic aciduria type 1	20 cases
Acrorenal syndrome	20 cases
Arrhinia	20 cases

Diseases name	Number of published cases or families
Calvarial doughnut lesions - bone fragility	20 cases
Cap polyposis	20 cases
CDG syndrome type Ib	20 cases
Congenital insensitivity to pain	20 cases
Dihydrolipoyl dehydrogenase deficiency	20 cases
Distal monosomy 8p	20 cases
Hypertelorism, Teebi Type	20 cases
Juvenile temporal arteritis	20 cases
Keratitis, Nagashima-type	20 cases
Lacrimo-auriculo-dento-digital syndrome	20 cases
PIBIDS syndrome	20 cases
Syndromic microphthalmia type 5	20 cases
Acromegaly facial appearance syndrome	< 20 cases
Acromegaly facies - hypertrichosis	< 20 cases
Bleeding diathesis due to a collagen receptor defect	< 20 cases
Brain-lung-thyroid syndrome	< 20 cases
Carey-Fineman-Ziter syndrome	< 20 cases
Cataract - intellectual deficit - hypogonadism	< 20 cases
COFS syndrome	< 20 cases
Craniodiaphyseal dysplasia	< 20 cases
Czech dysplasia, metatarsal type	< 20 cases
Ectodermal dysplasia, "pure" hair-nail type	< 20 cases
Epidermolysis bullosa simplex - limb girdle muscular dystrophy	< 20 cases
Familial partial lipodystrophy, Köbberling type	< 20 cases
Hereditary orotic aciduria	< 20 cases
Ichthyosis bullosa of Siemens	< 20 cases
IMAGe syndrome	< 20 cases
Interstitial granulomatous dermatitis with arthritis	< 20 cases
Isolated anterior cervical hypertrichosis	< 20 cases
Keratitis palmaris et plantaris - clinodactyly	< 20 cases
Laryngeal abductor paralysis - intellectual deficit	< 20 cases
Myoclonic dystonia 15	< 20 cases
Radio-ulnar synostosis - amegakaryocytic thrombocytopenia	< 20 cases
RAPADILINO syndrome	< 20 cases
Ring chromosome 10	< 20 cases
Woolly hair - palmoplantar keratoderma - dilated cardiomyopathy	< 20 cases
6q terminal deletion	19 cases
Aconitase deficiency	19 cases
Amelo-cerebro-hypohidrotic syndrome	19 cases
Craniosynostosis, Boston type	19 cases
Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia	19 cases
Hypomyelination with atrophy of basal ganglia and cerebellum	19 cases
Progressive cavitating leukoencephalopathy	19 cases

Diseases name	Number of published cases or families
Schopf-Schulz-Passarge syndrome	19 cases
Hypertrichotic osteochondrodysplasia	18 cases
Terminal osseous dysplasia - pigmentary defects	18 cases
Action myoclonus - renal failure syndrome	17 cases
Aminopterin embryofetopathy	17 cases
Circumscribed palmoplantar hypokeratosis	17 cases
Congenital lethal erythroderma	17 cases
GTP cyclohydrolase I deficiency	17 cases
H syndrome	17 cases
Hereditary folate malabsorption	17 cases
Leukocyte adhesion deficiency type III	17 cases
Malonic aciduria	17 cases
X-linked creatine transporter deficiency	17 cases
Acromegaly - cutis verticis gyrata - corneal leukoma	16 cases
Hepatic glycogen synthase deficiency	16 cases
Intellectual deficit, X-linked - Dandy-Walker malformation - basal ganglia disease - Seizures	16 cases
Intellectual deficit, X-linked, South African type	16 cases
IRIDA syndrome	16 cases
Metaphyseal chondrodysplasia, Jansen type	16 cases
Microgastria - limb reduction defect	16 cases
Myhre syndrome	16 cases
Oral-facial-digital syndrome type 4	16 cases
Orbital leiomyoma	16 cases
Ablepharon macrostomia syndrome	15 cases
Angel-shaped phalango-epiphyseal dysplasia	15 cases
Congenital muscular dystrophy due to lamina A/C deficiency	15 cases
IBIDS syndrome	15 cases
Sensenbrenner syndrome	15 cases
Umbilical cord ulceration - intestinal atresia	15 cases
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	< 15 cases
Isolated congenital anosmia	< 15 cases
Lissencephaly due to TUBA1A mutation	< 15 cases
Meacham syndrome	< 15 cases
Odonto-onycho-dermal dysplasia	< 15 cases
Taurodontia - absent teeth - sparse hair	< 15 cases
Wilson-Turner syndrome	> 14 cases
Absence of fingerprints - congenital milia	14 cases
ADULT syndrome	14 cases
Atkin-Flaitz syndrome	14 cases
Autosomal dominant optic atrophy and cataract	14 cases
Autosomal recessive limb-girdle muscular dystrophy type 2G	14 cases
Autosomal recessive limb-girdle muscular dystrophy type 2L	14 cases
Beta-mannosidosis	14 cases

Diseases name	Number of published cases or families
DEND syndrome	14 cases
Dermo-odonto dysplasia	14 cases
Oligocone trichromacy	14 cases
Ring chromosome 17	14 cases
Aromatase deficiency	13 cases
Autosomal dominant spastic paraplegia type 37	13 cases
Charcot-Marie-Tooth disease type 2H	13 cases
Chondrodysplasia, Blomstrand type	13 cases
Donnai-Barrow syndrome	13 cases
Hereditary progressive mucinous histiocytosis	13 cases
Schilbach-Rott syndrome	13 cases
Acro-pectoro-renal field defect	12 cases
Alopecia - epilepsy - pyorrhea - intellectual deficit	12 cases
Alpha-N-acetylgalactosaminidase deficiency	12 cases
Atelosteogenesis I	12 cases
Atelosteogenesis type III	12 cases
Atrial tachyarrhythmia with short PR interval	12 cases
Ballard syndrome	12 cases
Coloboma of macula - brachydactyly type B	12 cases
Distal myopathy with posterior leg and anterior upper limb involvement	12 cases
Distal myopathy with vocal cord weakness	12 cases
Dopamine beta-hydroxylase deficiency	12 cases
Hemolytic anemia due to adenylate kinase deficiency	12 cases
Hypoparathyroidism - deafness - renal disease	12 cases
Intellectual deficit, X-linked - macrocephaly - macroorchidism	12 cases
Lethal ataxia with deafness and optic atrophy	12 cases
Osteocraniostenosis	12 cases
Torg-Winchester syndrome	12 cases
Uveal coloboma - cleft lip and palate - intellectual deficit	12 cases
Antecubital pterygium syndrome	11 cases
Atrial septal defect - atrioventricular conduction defects	11 cases
Cooks syndrome	11 cases
Familial acute necrotizing encephalopathy	11 cases
Familial scaphocephaly syndrome, McGillivray type	11 cases
Fibrochondrogenesis	11 cases
Fibular dimelia - diplopodia	11 cases
Foveal hypoplasia - presenile cataract	11 cases
Fuhrmann syndrome	11 cases
Goldblatt syndrome	11 cases
Hyperandrogenism due to cortisone reductase deficiency	11 cases
Intellectual deficit, X-linked, Snyder type	11 cases
Juvenile polyposis of infancy	11 cases

Diseases name	Number of published cases or families
Leukonychia totalis - acanthosis-nigrans-like lesions - abnormal hair	11 cases
Lopez-Hernandez syndrome	11 cases
Martinez-Frias syndrome	11 cases
PELVIS syndrome	11 cases
Sick sinus syndrome	11 cases
Trichomegaly - retina pigmentary degeneration - dwarfism	11 cases
X-linked complicated corpus callosum dysgenesis	11 cases
CHAND syndrome	> 10 cases
Chediak-Higashi syndrome	> 10 cases
Pai syndrome	> 10 cases
2q37 microdeletion syndrome	10 cases
Acromesomelic dysplasia, Hunter-Thomson type	10 cases
Arthrogryposis multiplex congenita - whistling face	10 cases
Athabaskan brainstem dysgenesis syndrome	10 cases
Autoinflammatory disease due to interleukin-1 receptor antagonist deficiency	10 cases
Barber-Say syndrome	10 cases
Boomerang dysplasia	10 cases
Cerebro-oculo-nasal syndrome	10 cases
Char syndrome	10 cases
Charcot-Marie-Tooth disease - nephropathy	10 cases
Charcot-Marie-Tooth disease type 4H	10 cases
Colobomatous - microphthalmia - heart disease - hearing loss	10 cases
Diffuse palmoplantar keratoderma - acrocyanosis	10 cases
Distal monosomy 5q	10 cases
Ectodermal dysplasia - skin fragility syndrome	10 cases
Familial partial lipodystrophy associated with PPARG mutations	10 cases
Flynn-Aird syndrome	10 cases
Goldberg-Shprintzen megacolon syndrome	10 cases
Hyperkeratosis - hyperpigmentation syndrome	10 cases
Hypomyelination - congenital cataract	10 cases
Infantile choroido cerebral calcification syndrome	10 cases
Intellectual deficit, X-linked - hypotonia - facial dysmorphism - aggressive behavior	10 cases
Iris coloboma with ptosis - intellectual deficit	10 cases
Juberg-Hayward syndrome	10 cases
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus	10 cases
Nevo syndrome	10 cases
Pollitt syndrome	10 cases
Progressive non-infectious anterior vertebral fusion	10 cases
Pseudodiastrophic dysplasia	10 cases
Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts	10 cases
Saldino-Mainzer syndrome	10 cases

Diseases name	Number of published cases or families
Succinyl-CoA acetoacetate transferase deficiency	10 cases
Syndromic X-linked intellectual deficit 7	10 cases
Toriello-Lacassie-Droste syndrome	10 cases
X-linked neurodegenerative syndrome, Hamel type	10 cases
Aase-Smith syndrome	< 10 cases
Acrofacial dysostosis, Rodriguez type	< 10 cases
Acromelanosis	< 10 cases
Benign familial nocturnal alternating hemiplegia of childhood	< 10 cases
Cap myopathy	< 10 cases
Cardiocranial syndrome, Pfeiffer type	< 10 cases
Carpotarsal osteochondromatosis	< 10 cases
Cerebral gigantism - jaw cysts	< 10 cases
Corneal dystrophy - perceptive deafness	< 10 cases
Deafness - lymphedema - leukemia	< 10 cases
Diaphanospondylodysostosis	< 10 cases
Digitorenocerebral syndrome	< 10 cases
Encephalopathy due to prosaposin deficiency	< 10 cases
Gaucher disease - ophthalmoplegia - cardiovascular calcification	< 10 cases
Glomerulonephritis - sparse hair - telangiectasis	< 10 cases
Greenberg dysplasia	< 10 cases
Heinz body anemia	< 10 cases
Hereditary myoclonus - progressive distal muscular atrophy	< 10 cases
Hypopituitarism - microphthalmia	< 10 cases
Laron syndrome with immunodeficiency	< 10 cases
Lethal Larsen-like syndrome	< 10 cases
Leukocyte adhesion deficiency type II	< 10 cases
Lowry-Wood syndrome	< 10 cases
Neonatal ichthyosis - sclerosing cholangitis	< 10 cases
Pacman dysplasia	< 10 cases
Palmoplantar porokeratosis of Mantoux	< 10 cases
Pancreatic hypoplasia - diabetes - heart disease	< 10 cases
Perioral myoclonia with absences	< 10 cases
Phosphoenolpyruvate carboxykinase deficiency	< 10 cases
Progeria - short stature - pigmented nevi	< 10 cases
Pseudo-Zellweger syndrome	< 10 cases
Singleton-Merten dysplasia	< 10 cases
Thoracolumbar pelvic dysplasia	< 10 cases
XK aprosencephaly	< 10 cases
Acro-cardio-facial syndrome	9 cases
Atransferrinemia	9 cases
Autosomal recessive spastic paraplegia type 18	9 cases
Bosley-Salih-Alorainy syndrome	9 cases
Brachymorphism - onychodysplasia - dysphalangism	9 cases
Cardiac anomalies - heterotaxy	9 cases

Diseases name	Number of published cases or families
Cardiomyopathy - cataract - hip spine disease	9 cases
Gamma-glutamylcysteine synthetase deficiency	9 cases
Guanidinoacetate methyltransferase deficiency	9 cases
Intellectual deficit, X-linked - spastic quadriplegia	9 cases
Intellectual deficit, X-linked, Shashi type	9 cases
Laminopathy type Decaudain-Vigouroux	9 cases
Lenz-Majewski hyperostotic dwarfism	9 cases
Leukodystrophy - spastic paraplegia - dystonia	9 cases
Macrostomia - preauricular tags - external ophthalmoplegia	9 cases
Methylmalonicacidemia - homocystinuria, type cbl F	9 cases
Oculocerebrofacial syndrome, Kaufman type	9 cases
Tricho-retino-dento-digital syndrome	9 cases
X-linked intellectual deficit - ataxia - apraxia	9 cases
5-oxoprolinase deficiency	8 cases
Ackerman syndrome	8 cases
Ankylosing vertebral hyperostosis with tylosis	8 cases
Ataxia-deafness-retardation syndrome	8 cases
Autosomal dominant familial hematuria - retinal arteriolar tortuosity - contractures	8 cases
Brachydactyly - preaxial hallux varus	8 cases
Campomelia, Cumming type	8 cases
Camptodactyly syndrome, Guadalajara type 1	8 cases
Fountain syndrome	8 cases
Hydrocephalus - costovertebral dysplasia - Sprengel anomaly	8 cases
Intellectual deficit, X-linked - dysmorphism - cerebral atrophy	8 cases
Intellectual deficit, X-linked, Abidi type	8 cases
Intellectual deficit, X-linked, Vitale type	8 cases
Joubert syndrome with hepatic defect	8 cases
Kallmann syndrome - heart disease	8 cases
Lelis syndrome	8 cases
Lethal polymalformative syndrome, Boissel type	8 cases
Leukoencephalopathy - ataxia - hypodontia - hypomyelination	8 cases
Micro syndrome	8 cases
Osteopetrosis - hypogammaglobulinemia	8 cases
Spondylometaphyseal dysplasia - cone-rod dystrophy	8 cases
Vici syndrome	8 cases
2p21 microdeletion syndrome	7 cases
Acro-renal-mandibular syndrome	7 cases
Brachydactyly type A6	7 cases
CDG syndrome type Ie	7 cases
CEDNIK syndrome	7 cases
Cleft palate-lateral synechia syndrome	7 cases
Deletion 6q16 syndrome	7 cases
Dihydropyrimidinuria	7 cases

Diseases name	Number of published cases or families
Dystonia 16	7 cases
Ehlers-Danlos syndrome, dermatosparaxis type	7 cases
Episodic ataxia type 5	7 cases
Episodic ataxia type 7	7 cases
Familial multiple fibrofolliculoma	7 cases
Gamma-glutamyl transpeptidase deficiency	7 cases
Genitopatellar syndrome	7 cases
Intellectual deficit, X-linked - craniofacioskeletal syndrome	7 cases
Intellectual deficit, X-linked, Van Esch type	7 cases
MEHMO syndrome	7 cases
Michels syndrome	7 cases
Mitochondrial myopathy and sideroblastic anemia	7 cases
Obesity due to pro-opiomelanocortin deficiency	7 cases
Ocular albinism - late-onset sensorineural deafness	7 cases
Retinal degeneration - nanophthalmos - glaucoma	7 cases
Retinohepatoendocrinologic syndrome	7 cases
Severe X-linked intellectual deficit, Gustavson type	7 cases
Stern-Lubinsky-Durrie syndrome	7 cases
Torticollis - keloids - cryptorchidism - renal dysplasia	7 cases
Transaldolase deficiency	7 cases
X-linked mandibulofacial dysostosis	7 cases
X-linked neurodegenerative syndrome, Bertini type	7 cases
X-linked severe congenital neutropenia	7 cases
3-hydroxy 3-methylglutaryl-CoA synthase deficiency	6 cases
46,XY gonadal dysgenesis - motor and sensory neuropathy	6 cases
Acrofacial dysostosis, Catania type	6 cases
Blepharophimosis - ptosis - esotropia - syndactyly - short stature	6 cases
CDG syndrome type Ig	6 cases
CLAPO syndrome	6 cases
Cold-induced sweating syndrome	6 cases
Cutis gyrata - acanthosis nigricans - craniosynostosis	6 cases
Deafness - enamel hypoplasia - nail defects	6 cases
Ehlers-Danlos syndrome, spondylocheiro dysplastic type	6 cases
Eiken syndrome	6 cases
Epilepsy telangiectasia	6 cases
Grange syndrome	6 cases
Hartsfield-Bixler-Demyer syndrome	6 cases
Hidrotic ectodermal dysplasia, Christianson-Fourie type	6 cases
Hypopituitarism - postaxial polydactyly	6 cases
Intellectual deficit - sparse hair - brachydactyly	6 cases
Intellectual deficit, X-linked - psychosis - macroorchidism	6 cases
Intellectual deficit, X-linked, Armfield type	6 cases

Diseases name	Number of published cases or families
Intellectual deficit, X-linked, Zorick type	6 cases
Isotretinoin-like syndrome	6 cases
Keratosis follicularis - dwarfism - cerebral atrophy	6 cases
Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus	6 cases
Moore-Federman syndrome	6 cases
PAGOD syndrome	6 cases
Polysyndactyly - cardiac malformation	6 cases
Recessive aplasia cutis congenita of limbs	6 cases
Spasticity - intellectual deficit - X-linked epilepsy	6 cases
Spinocerebellar ataxia type 30	6 cases
Stormorken-Sjaastad-Langslet syndrome	6 cases
Symphalangism with multiple anomalies of hands and feet	6 cases
Syndactyly - telecanthus - anogenital and renal malformations	6 cases
Thumb stiffness - brachydactyly - intellectual deficit	6 cases
UV-sensitive syndrome	6 cases
W syndrome	6 cases
Wieacker-Wolff syndrome	6 cases
Zunich-Kaye syndrome	6 cases
Achalasia - microcephaly	5 cases
Acro-fronto-facio-nasal dysostosis	5 cases
Acute bilateral depigmentation of the iris	5 cases
Adducted thumbs-arthrogyrosis, Dundar type	5 cases
Alopecia - contractures - dwarfism - intellectual deficit	5 cases
ANE syndrome	5 cases
Anonychia - microcephaly	5 cases
Arachnodactyly - abnormal ossification - intellectual deficit	5 cases
Astley-Kendall dysplasia	5 cases
Autosomal dominant macrothrombocytopenia with abnormal proplatelet formation	5 cases
Autosomal recessive lower motor neuron disease with childhood onset	5 cases
Bamforth syndrome	5 cases
Beta-ureidopropionase deficiency	5 cases
Bradyopsia	5 cases
Branchiogenic deafness syndrome	5 cases
CAMOS syndrome	5 cases
CDG syndrome type Id	5 cases
CDG syndrome type Ih	5 cases
Charcot-Marie-Tooth disease type 4J	5 cases
Choanal atresia - deafness - cardiac defects - dysmorphism	5 cases
Cholestasis - pigmentary retinopathy - cleft palate	5 cases
Congenital bile acid synthesis defect type 4	5 cases
Congenital spastic tetraplegia	5 cases

Diseases name	Number of published cases or families
Craniodigital syndrome - intellectual deficit	5 cases
Craniosynostosis - dysmorphism - brachydactyly	5 cases
Curry-Jones syndrome	5 cases
Dacryocystitis - osteopoikilosis	5 cases
Dermatoosteolysis, Kirghizian type	5 cases
Ectrodactyly - ectodermal dysplasia without clefting	5 cases
Fine-Lubinsky syndrome	5 cases
Frank-Ter Haar syndrome	5 cases
German syndrome	5 cases
Glaucoma - sleep apnea	5 cases
Humerospinal dysostosis	5 cases
Humeroulnar synostosis	5 cases
Intellectual deficit, X-linked - choreoathetosis - abnormal behavior	5 cases
Intellectual deficit, X-linked - cubitus valgus - dysmorphism	5 cases
Lissencephaly type 3 - familial fetal akinesia sequence	5 cases
Lymphedema - cerebral arteriovenous anomaly	5 cases
Matthew-Wood syndrome	5 cases
Methylmalonicacidemia - homocystinuria, type cbl D	5 cases
Mononen-Karnes-Senac syndrome	5 cases
Nephropathy - deafness - hyperparathyroidism	5 cases
Nephrosis - deafness - urinary tract - digital malformations	5 cases
Oculo-palato-cerebral syndrome	5 cases
Odontomicronychial dysplasia	5 cases
Onycho-tricho-dysplasia - neutropenia	5 cases
P2Y12 deficiency	5 cases
Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma	5 cases
Paraplegia - brachydactyly - cone-shaped epiphysis	5 cases
Pyknoachondrogenesis	5 cases
Sillence syndrome	5 cases
Spondyloepimetaphyseal dysplasia - hypotrichosis	5 cases
X-linked immunoneurologic disorder	5 cases
X-linked intellectual deficit, Najm type	5 cases
X-linked spinocerebellar ataxia type 3	5 cases
Young-Simpson syndrome	5 cases
Cystic hamartoma of lung and kidney	< 5 cases
Lathosterolosis	< 5 cases
Sakati-Nyhan syndrome	< 5 cases
12q14 microdeletion syndrome	4 cases
15q24 microdeletion syndrome	4 cases
8q22.1 microdeletion syndrome	4 cases
Abruzzo-Erickson syndrome	4 cases
Acrofacial dysostosis, Palagonia type	4 cases

Diseases name	Number of published cases or families
Amelogenesis imperfecta and gingival hyperplasia syndrome	4 cases
Anophthalmia plus syndrome	4 cases
Aortic arch anomaly - peculiar facies - intellectual deficit	4 cases
Aplasia cutis - myopia	4 cases
Arhinia - choanal atresia - microphthalmia	4 cases
Arterial dissection - lentiginosis	4 cases
Autism - facial port-wine stain	4 cases
Autosomal dominant multiple pterygium syndrome	4 cases
Benign exophthalmos syndrome	4 cases
Bilateral microtia - deafness - cleft palate	4 cases
Blindness - scoliosis - arachnodactyly	4 cases
Bonnamann-Meinecke-Reich syndrome	4 cases
Brachydactyly - long thumb	4 cases
CDG syndrome type If	4 cases
CDG syndrome type IIa	4 cases
CDG syndrome type Ik	4 cases
Central bilateral macrogyria	4 cases
Choroideremia - deafness - obesity	4 cases
Cleft lip/palate - intestinal malrotation - cardiopathy	4 cases
Cole-Carpenter syndrome	4 cases
Congenital lethal myopathy, Compton-North type	4 cases
Coxoauricular syndrome	4 cases
Craniosynostosis - Dandy-Walker - hydrocephalus	4 cases
Craniosynostosis - hydrocephalus - Chiari I malformation - radioulnar synostosis	4 cases
Deafness - peripheral neuropathy - arterial disease	4 cases
Diaphragmatic defect - limb deficiency - skull defect	4 cases
Distal limb deficiencies - micrognathia syndrome	4 cases
Ectodermal dysplasia, Berlin type	4 cases
Ectopia lentis - chorioretinal dystrophy - myopia	4 cases
Encephalopathy due to urocanase deficiency	4 cases
Endosteal sclerosis - cerebellar hypoplasia	4 cases
Episodic ataxia type 6	4 cases
Facial onset sensory and motor neuronopathy	4 cases
Familial caudal dysgenesis	4 cases
Gorlin-Chaudhry-Moss syndrome	4 cases
Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate	4 cases
Growth delay due to insulin-like growth factor I deficiency	4 cases
Hereditary sensory and autonomic neuropathy with deafness and global delay	4 cases
Hidrotic ectodermal dysplasia, Halal type	4 cases
Hirschsprung disease - type D brachydactyly	4 cases
Homocarnosinosis	4 cases
Hypomandibular faciocranial dysostosis	4 cases

Diseases name	Number of published cases or families
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 cases
Hypotrichosis - lymphedema - telangiectasia	4 cases
Ichthyosis - alopecia - eclabion - ectropion - intellectual deficit	4 cases
Immunodeficiency with natural-killer cell deficiency	4 cases
Intellectual deficit - dysmorphism - hypogonadism - diabetes mellitus	4 cases
Intellectual deficit, X-linked - hypogonadism - ichthyosis - obesity - short stature	4 cases
Intellectual deficit, X-linked - seizures - psoriasis	4 cases
Intellectual deficit, X-linked, Cilliers type	4 cases
Intellectual deficit, X-linked, Miles-Carpenter type	4 cases
Intellectual deficit, X-linked, Schimke type	4 cases
Intellectual deficit, X-linked, Seemanova type	4 cases
Intellectual deficit, X-linked, Siderius type	4 cases
Intellectual deficit, X-linked, Stevenson type	4 cases
Intellectual deficit, X-linked, Stocco Dos Santos type	4 cases
Kapur-Toriello syndrome	4 cases
Lethal bone dysplasia, Holmgren type	4 cases
Lethal recessive chondrodysplasia	4 cases
Leukoencephalopathy - metaphyseal chondrodysplasia	4 cases
Leukoencephalopathy - palmoplantar keratoderma	4 cases
Malignant hyperthermia - arthrogryposis - torticollis	4 cases
Metaphyseal acroscaphodysplasia	4 cases
Microcephalic osteodysplastic dysplasia, Saul-Wilson type	4 cases
Microcephaly - polymicrogyria - corpus callosum agenesis	4 cases
Myoclonus - cerebellar ataxia - deafness	4 cases
Myopathy due to calsequestrin and SERCA1 protein overload	4 cases
Neurocutaneous syndrome, Bicknell type	4 cases
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	4 cases
Neuroectodermal-endocrine syndrome	4 cases
Oculo-oto-facial dysplasia	4 cases
Odontoleukodystrophy	4 cases
Odontotrichomelic syndrome	4 cases
Oral-facial-digital syndrome type 5	4 cases
Palmoplantar keratoderma - amyotrophy	4 cases
Paraplegia - intellectual deficit - hyperkeratosis	4 cases
Pelviscapular dysplasia	4 cases
Permanent neonatal diabetes mellitus - pancreatic and cerebellar agenesis	4 cases
Primary immunodeficiency syndrome due to p14 deficiency	4 cases
Pulmonary fibrosis - hepatic hyperplasia - bone marrow hypoplasia	4 cases

Diseases name	Number of published cases or families
RHYS syndrome	4 cases
Rolled and spiral hairs - palmoplantar keratoderma	4 cases
Severe achondroplasia - developmental delay - acanthosis nigricans	4 cases
Short stature - webbed neck - heart disease	4 cases
Simpson-Golabi-Behmel syndrome type 2	4 cases
Sparse hair - short stature - skin anomalies	4 cases
Spastic paraplegia - nephritis - deafness	4 cases
Spondyloepiphyseal dysplasia, Byers type	4 cases
Spondyloepiphyseal dysplasia, Cantu type	4 cases
Spondyloepiphyseal dysplasia, MacDermot type	4 cases
Spondyloepiphyseal dysplasia, Nishimura type	4 cases
Spondylometaphyseal dysplasia with combined immunodeficiency	4 cases
Syndactyly type 4	4 cases
Tomé-Brunet-Fardeau syndrome	4 cases
Tricho-odonto-onychial dysplasia	4 cases
X-linked intellectual deficit, Stoll type	4 cases
Absent thumb - short stature - immunodeficiency	3 cases
Acromesomelic dysplasia, Brahim-Bacha type	3 cases
Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis	3 cases
Al-Gazali-Dattani syndrome	3 cases
Aniridia - absent patella	3 cases
Aniridia - ptosis - intellectual deficit - familial obesity	3 cases
Anonychia with flexural pigmentation	3 cases
Anophthalmia - megalocornea - cardiopathy - skeletal anomalies	3 cases
Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis	3 cases
Aplasia cutis congenita - intestinal lymphangiectasia	3 cases
Arachnodactyly - intellectual deficit - dysmorphism	3 cases
AREDYLD syndrome	3 cases
Autosomal recessive amelia	3 cases
Autosomal recessive limb-girdle muscular dystrophy type 2M	3 cases
Axenfeld-Rieger anomaly - hydrocephaly - skeletal abnormalities	3 cases
Axial spondylometaphyseal dysplasia	3 cases
Blepharoptosis - myopia - ectopia lentis	3 cases
Branchio-skeleto-genital syndrome	3 cases
Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia	3 cases
Cardiomyopathy-exercise intolerance due to muscle and heart glycogen deficiency	3 cases
Cervical hypertrichosis - peripheral neuropathy	3 cases
CODAS syndrome	3 cases
Congenital enterocyte heparan sulfate deficiency	3 cases

Diseases name	Number of published cases or families
Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	3 cases
Congenital osteogenesis imperfecta - microcephaly - cataracts	3 cases
Cortical blindness - intellectual deficit - polydactyly	3 cases
Craniofacial-deafness-hand syndrome	3 cases
Craniofrontonasal dysplasia - Poland anomaly	3 cases
Craniosynostosis - intracranial calcifications	3 cases
Cutaneous photosensitivity - lethal colitis	3 cases
Deafness - intellectual deficit, Martin-Probst type	3 cases
Disorder of sex development - intellectual deficit	3 cases
Ermine phenotype	3 cases
Eyebrow duplication - syndactyly	3 cases
Facial dysmorphism - macrocephaly - myopia - Dandy-Walker malformation	3 cases
Glaucoma - ectopia - microspherophakia - stiff joints - short stature	3 cases
Global developmental delay - osteopenia - ectodermal defect	3 cases
Goodman syndrome	3 cases
Hair defect - photosensitivity - intellectual deficit	3 cases
Hemolytic anemia due to glutathione reductase deficiency	3 cases
Hemorrhagic disease due to alpha-1 antitrypsin Pittsburgh mutation	3 cases
Hirschsprung disease - nail hypoplasia - dysmorphism	3 cases
Hypohidrotic ectodermal dysplasia - hypothyroidism - ciliary dyskinesia	3 cases
Hypotonia with lactic acidemia and hyperammonemia	3 cases
Intellectual deficit - cataracts - kyphosis	3 cases
Intellectual deficit - hypoplastic corpus callosum - preauricular tag	3 cases
Intellectual deficit, Kahrizi type	3 cases
Intellectual deficit, X-linked - hypogammaglobulinemia - progressive neurological deterioration	3 cases
Intellectual deficit, X-linked - precocious puberty - obesity	3 cases
Intellectual deficit, X-linked, Kroes type	3 cases
Intellectual deficit, X-linked, Shrimpton type	3 cases
Intellectual deficit, X-linked, Wilson type	3 cases
Intellectual deficit, X-linked, Wittwer type	3 cases
Intractable diarrhea - choanal atresia - eye anomalies	3 cases
Lethal faciocardiomelic dysplasia	3 cases
Lethal omphalocele-cleft palate syndrome	3 cases
Lewis-Pashayan syndrome	3 cases
Lipodystrophy - intellectual deficit - deafness	3 cases
Lymphedema - atrial septal defects - facial changes	3 cases

Diseases name	Number of published cases or families
Microcephaly - brachydactyly - kyphoscoliosis	3 cases
Microcephaly - cardiomyopathy	3 cases
Microcephaly - cleft palate	3 cases
Microcephaly - intellectual deficit - phalangeal and neurological anomalies	3 cases
Microcytic anemia with liver iron overload	3 cases
Microphthalmia - brain atrophy	3 cases
Mitral regurgitation - deafness - skeletal anomalies	3 cases
Mullerian derivatives - lymphangiectasia - polydactyly	3 cases
N syndrome	3 cases
Nail patella-like - renal disease	3 cases
Neuroaxonal dystrophy - renal tubular acidosis	3 cases
Oculoosteocutaneous syndrome	3 cases
Oral-facial-digital syndrome type 3	3 cases
Osteoporosis-oculocutaneous-hypopigmentation syndrome	3 cases
Osteosclerosis - ichthyosis - premature ovarian failure	3 cases
Peripheral neuropathy, Fiskerstrand type	3 cases
Pontocerebellar hypoplasia type 4	3 cases
Pontocerebellar hypoplasia type 5	3 cases
Pontocerebellar hypoplasia type 6	3 cases
Posterior fusion of lumbosacral vertebrae - blepharoptosis	3 cases
Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	3 cases
Qazi-Markouizos syndrome	3 cases
Rambaud-Galian syndrome	3 cases
Seizures - intellectual deficit due to hydroxylysineuria	3 cases
Sensorineural hearing loss - early greying - essential tremor	3 cases
SERKAL syndrome	3 cases
Short stature - intellectual deficit - eye anomalies - cleft lip/palate	3 cases
Short stature due to growth hormone qualitative anomaly	3 cases
Split hand - urinary anomalies - spina bifida	3 cases
Spondyloepimetaphyseal dysplasia, aggrecan type	3 cases
Spondyloepimetaphyseal dysplasia, Bieganski type	3 cases
Spondyloepiphyseal dysplasia tarda, Kohn type	3 cases
Spondylometaphyseal dysplasia, Golden type	3 cases
Suarez-Stickler syndrome	3 cases
Summitt syndrome	3 cases
Temtamy syndrome	3 cases
Thymic-renal-anal-lung dysplasia	3 cases
Trigonocephaly - short stature - developmental delay	3 cases
Ulbright-Hodes syndrome	3 cases

Diseases name	Number of published cases or families
46,XX disorder of sex development - skeletal anomalies	2 cases
46,XY disorder of sex development - adrenal insufficiency	2 cases
Acrocraniofacial dysostosis	2 cases
Adult familial nephronophthisis - spastic quadripareisia	2 cases
Agenesis of the corpus callosum - intellectual deficit - coloboma - micrognathia	2 cases
Alar cartilages hypoplasia - coloboma - telecanthus	2 cases
Amaurosis - hypertrichosis	2 cases
Anhidrotic ectodermal dysplasia - immunodeficiency - osteopetrosis - lymphedema	2 cases
Aniridia - renal agenesis - psychomotor retardation	2 cases
Aniridia-intellectual deficit syndrome	2 cases
Arthrogyposis - hyperkeratosis, lethal form	2 cases
Atherosclerosis- deafness - diabetes - epilepsy - nephropathy	2 cases
Auricular abnormalities - cleft lip with or without cleft palate - ocular abnormalities	2 cases
Auriculoocular anomalies - cleft lip	2 cases
Autosomal recessive acrofacial dysostosis	2 cases
Bangstad syndrome	2 cases
Beemer-Ertbruggen syndrome	2 cases
Blepharo-facio-skeletal syndrome	2 cases
Brachytelephalangy - dysmorphism - Kallmann syndrome	2 cases
Braddock syndrome	2 cases
Brain demyelination due to methionine adenosyltransferase deficiency	2 cases
Brain malformation - congenital heart disease - postaxial polydactyly	2 cases
Camptodactyly syndrome, Guadalajara type 2	2 cases
Cardiomyopathy - renal anomalies	2 cases
Carnevale syndrome	2 cases
Cataract - ataxia - deafness	2 cases
Cataract - nephropathy - encephalopathy	2 cases
CDG syndrome type IIe	2 cases
CDG syndrome type IIh	2 cases
CDG syndrome type IL	2 cases
Central nervous system calcification - deafness - tubular acidosis - anemia	2 cases
Chondrodysplasia - disorder of sex development	2 cases
Choroidal atrophy - alopecia	2 cases
Cleft lip - retinopathy	2 cases
Cleft palate - short stature - vertebral anomalies	2 cases
Cleft palate - stapes fixation - oligodontia	2 cases
Cleido rhizomelic syndrome	2 cases
Congenital brain dysgenesis due to glutamine synthetase deficiency	2 cases
Congenital ichthyosis - microcephalus - quadriplegia	2 cases

Diseases name	Number of published cases or families
Contractures - ectodermal dysplasia - cleft lip/palate	2 cases
Cooper-Jabs syndrome	2 cases
Corneal anesthesia - deafness - intellectual deficit	2 cases
Corneal-cerebellar syndrome	2 cases
Craniosynostosis - fibular aplasia	2 cases
Craniosynostosis-radial aplasia, Imaizumi type	2 cases
Cryptomicrotia - brachydactyly - excess fingertip arch	2 cases
Dahlberg-Borer-Newcomer syndrome	2 cases
Dandy-Walker malformation - postaxial polydactyly	2 cases
Deaf blind hypopigmentation syndrome, Yemenite type	2 cases
Deafness - genital anomalies - metacarpal and metatarsal synostosis	2 cases
Deafness - vitiligo - achalasia	2 cases
Dentinogenesis imperfecta - short stature - hearing loss - intellectual deficit	2 cases
Dermato-cardio-skeletal syndrome, Borrone type	2 cases
Dermatoleukodystrophy	2 cases
Desmosterolosis	2 cases
Developmental malformations - deafness - dystonia	2 cases
Dincsoy-Salih-Patel syndrome	2 cases
Duane anomaly - myopathy - scoliosis	2 cases
Dursun syndrome	2 cases
Dysmorphism - short stature - deafness - disorder of sex development	2 cases
Ectodermal dysplasia - blindness	2 cases
Eng-Strom syndrome	2 cases
Epilepsy - microcephaly - skeletal dysplasia	2 cases
Epithelio-exfoliative colitis - deafness	2 cases
FASTKD2-related infantile mitochondrial encephalomyopathy	2 cases
Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3	2 cases
Fuqua-Berkovitz syndrome	2 cases
Gamma aminobutyric acid transaminase deficiency	2 cases
Gonadal dysgenesis, XY type - associated anomalies	2 cases
Heart defects - limb shortening	2 cases
HEC syndrome	2 cases
Hennekam-Beemer syndrome	2 cases
Hereditary cryohydrocytosis with reduced stomatin	2 cases
Hirschsprung disease - deafness - polydactyly	2 cases
Hydrocephaly - tall stature - joint laxity	2 cases
Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency	2 cases
Hyperekplexia - epilepsy	2 cases
Hypogonadotropic hypogonadism - retinitis pigmentosa	2 cases
Hypomagnesemia with normocalciuria	2 cases
Hypospadias-hypertelorism-coloboma and deafness	2 cases

Diseases name	Number of published cases or families
Hypotrichosis-intellectual deficit, Lopes type	2 cases
Ichthyosis - hepatosplenomegaly - cerebellar degeneration	2 cases
Ichthyosis - oral and digital anomalies	2 cases
Ichthyosis congenita - biliary atresia	2 cases
Immunodeficiency due to CD25 deficiency	2 cases
Inappropriate antidiuretic hormone secretion syndrome	2 cases
Intellectual deficit, X-linked - acromegaly - hyperactivity	2 cases
Intellectual deficit, X-linked - epilepsy - progressive joint contractures - dysmorphism	2 cases
Intellectual deficit, X-linked - plagiocephaly	2 cases
Intellectual deficit, X-linked, Cantagrel type	2 cases
Intellectual deficit, X-linked, Reish type	2 cases
Iris dysplasia - hypertelorism - deafness	2 cases
Kaler-Garrity-Stern syndrome	2 cases
Keratoderma - hypotrichosis - leukonychia	2 cases
Kozłowski-Brown-Hardwick syndrome	2 cases
Lethal hemolytic anemia - genital anomalies	2 cases
Lethal Kniest-like dysplasia	2 cases
Leukoencephalopathy - dystonia - motor neuropathy	2 cases
Lichenstein syndrome	2 cases
Lissencephaly type 3 - metacarpal bone dysplasia	2 cases
Low birth weight - dwarfism - dysgammaglobulinemia	2 cases
Lung fibrosis - immunodeficiency - 46,XX gonadal dysgenesis	2 cases
Macrocephaly - immune deficiency - anemia	2 cases
Macrocephaly - short stature - paraplegia	2 cases
Mesomelic dysplasia - skin dimples	2 cases
Metaphyseal chondrodysplasia - retinitis pigmentosa	2 cases
Metaphyseal chondrodysplasia, Kaitila type	2 cases
Methylmalonic aciduria - microcephaly - cataract	2 cases
Microbrachycephaly - ptosis - cleft lip	2 cases
Microcephaly - digital anomalies - intellectual deficit	2 cases
Microcephaly - glomerulonephritis - marfanoid habitus	2 cases
Microcephaly - seizures - intellectual deficit - heart disease	2 cases
Microlissencephaly - micromelia	2 cases
Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	2 cases
Monosomy 9q22.3	2 cases
Multiple sclerosis - ichthyosis - factor VIII deficiency	2 cases
Neonatal diabetes - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys	2 cases
Not NOTCH3-related small vessel disease of the brain	2 cases

Diseases name	Number of published cases or families
Obesity - colitis - hypothyroidism - cardiac hypertrophy - developmental delay	2 cases
Obesity due to prohormone convertase-I deficiency	2 cases
Oculotrichodysplasia	2 cases
Okamoto syndrome	2 cases
Ossification anomalies - psychomotor development delay	2 cases
Osteochondrodysplastic nanism - deafness - retinitis pigmentosa	2 cases
Osteogenesis imperfecta - retinopathy - seizures - intellectual deficit	2 cases
PARC syndrome	2 cases
Pierre Robin syndrome - facioidigital anomaly	2 cases
Pilodental dysplasia - refractive errors	2 cases
Progressive neurodegeneration - joint laxity - cataract	2 cases
Pseudoprogeria syndrome	2 cases
Pterygium colli - intellectual deficit - digital anomalies	2 cases
Resistance to thyrotropin-releasing hormone syndrome	2 cases
Robinow-like syndrome	2 cases
Rudiger syndrome	2 cases
Say-Barber-Miller syndrome	2 cases
Scalp defects - postaxial polydactyly	2 cases
SCARF syndrome	2 cases
Severe intellectual deficit - epilepsy - anal anomalies - distal phalangeal hypoplasia	2 cases
Short stature, Brussels type	2 cases
Siegler-Brewer-Carey syndrome	2 cases
Spastic paraplegia - precocious puberty	2 cases
Spastic quadriplegia - retinitis pigmentosa - intellectual deficit	2 cases
Spinal muscular atrophy - Dandy-Walker complex - cataracts	2 cases
Spondyloepimetaphyseal dysplasia - abnormal dentition	2 cases
Spondyloepimetaphyseal dysplasia, Genevieve type	2 cases
Spondylometaphyseal dysplasia - bowed forearms - facial dysmorphism	2 cases
Spondylometaphyseal dysplasia, A4 type	2 cases
Stimmler syndrome	2 cases
Stoll-Alembik-Finck syndrome	2 cases
Syngnathia multiple anomalies	2 cases
Talo-patello-scapoid osteolysis	2 cases
Teebi-Shaltout syndrome	2 cases
Thrombocytopenia - Robin sequence	2 cases
Thyrocerebrorenal syndrome	2 cases
Trichomegaly - cataract - hereditary spherocytosis	2 cases
Trigonocephaly - bifid nose - acral anomalies	2 cases

Diseases name	Number of published cases or families
Trigonocephaly - broad thumbs	2 cases
Tubular renal disease - cardiomyopathy	2 cases
Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay	2 cases
Xeroderma - talipes - enamel defects	2 cases
Zellweger-like syndrome without peroxisomal anomalies	2 cases
Li-Fraumeni syndrome	400 families
Nance-Horan syndrome	50 families
Autosomal recessive limb-girdle muscular dystrophy type 2I	> 40 families
Autoinflammatory granulomatosis of childhood	40 families
LCAT deficiency	30 families
Primary erythralgia	30 families
Thiamine-responsive megaloblastic anemia syndrome	30 families
Pericarditis - arthropathy - camptodactyly	< 30 families
Phosphoribosylpyrophosphate synthetase superactivity	< 30 families
Giant axonal neuropathy	20 families
Multiple synostoses	20 families
Overhydrated hereditary stomatocytosis	20 families
Acro-renal-ocular syndrome	< 20 families
Autosomal dominant spastic paraplegia type 17	< 20 families
Familial platelet syndrome with predisposition to acute myelogenous leukemia	< 20 families
Nonspherocytic hemolytic anemia due to hexokinase deficiency	17 families
Ichthyosis prematurity syndrome	16 families
X-linked myopathy with excessive autophagy	15 families
Hypotonia - cystinuria syndrome	14 families
Marie Unna congenital hypotrichosis	12 families
Charcot-Marie-Tooth disease type 4B1	11 families
Aniridia - cerebellar ataxia - intellectual deficit	> 10 families
Brachydactyly - arterial hypertension	> 10 families
Aortic aneurysm syndrome, Loews-Dietz type	10 families
Autosomal dominant spastic paraplegia type 6	10 families
Benign familial neonatal-infantile seizures	10 families
Renpenning syndrome	10 families
Acheiropodia	< 10 families
Autosomal dominant spastic paraplegia type 10	< 10 families
Autosomal dominant spastic paraplegia type 12	< 10 families
Autosomal dominant spastic paraplegia type 13	< 10 families
Autosomal dominant spastic paraplegia type 8	< 10 families
Autosomal recessive spastic paraplegia type 15	< 10 families
Familial isolated hypoparathyroidism	< 10 families
Focal facial dermal dysplasia	< 10 families
Hereditary myopathy with early respiratory failure	< 10 families
Hereditary neurocutaneous angioma	< 10 families

Diseases name	Number of published cases or families
Keratosis palmoplantaris - esophageal carcinoma	< 10 families
Palmoplantar keratoderma - deafness	< 10 families
Sebastian syndrome	< 10 families
Syndromic X-linked intellectual deficit due to JARID1C mutation	< 10 families
Ulnar-mammary syndrome	< 10 families
VACTERL with hydrocephalus	< 10 families
Otodental syndrome	9 families
Perry syndrome	9 families
Cataract-microcornea syndrome	8 families
Lethal osteosclerotic bone dysplasia	8 families
EEM syndrome	7 families
X-linked immune dysregulation - polyendocrinopathy - enteropathy	7 families
Cystoid macular dystrophy	6 families
Deafness with labyrinthine aplasia, microtia, and microdontia	6 families
Excessive growth - learning disabilities - facial dysmorphism	6 families
Familial developmental dysphasia	6 families
Pontocerebellar hypoplasia type 1	6 families
Stapes ankylosis with broad thumbs and toes	6 families
Familial encephalopathy with neuroserpin inclusion bodies	> 5 families
Autosomal dominant limb-girdle muscular dystrophy type 1D	5 families
Autosomal dominant limb-girdle muscular dystrophy type 1E	5 families
X-linked sideroblastic anemia - ataxia	5 families
Distal symphalangism	< 5 families
Trichodontal syndrome	< 5 families
Camptodactyly - taurinuria	4 families
IVIC syndrome	4 families
MEDNIK syndrome	4 families
Paroxysmal extreme pain disorder	4 families
Reducing body myopathy	4 families
Triphalangeal thumbs - brachyectrodactyly	4 families
Adducted thumbs - arthrogryposis, Christian type	3 families
Autosomal dominant Charcot-Marie-Tooth disease type 2K	3 families
Autosomal recessive cerebellar ataxia - blindness - deafness	3 families
Cataract-glaucoma	3 families
Cerebroretinal vasculopathy	3 families
Craniorhiny	3 families
Deafness-infertility syndrome	3 families
HERNS syndrome	3 families
Isolated dominant hypomagnesemia	3 families
Mixed dystonia	3 families

Diseases name	Number of published cases or families
Rapid-onset dystonia-parkinsonism	3 families
Ankyloblepharon filiforme - imperforate anus	2 families
Auriculoosteodysplasia	2 families
Autosomal dominant limb-girdle muscular dystrophy type 1A	2 families
Autosomal recessive spastic paraplegia type 26	2 families
Autosomal recessive spastic paraplegia type 27	2 families
Autosomal recessive spastic paraplegia type 39	2 families
Bence syndrome	2 families
Blepharonasofacial malformation syndrome	2 families
Brachydactyly type A5	2 families
Bullous dystrophy, macular type	2 families
Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss	2 families
Episodic ataxia type 4	2 families
Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	2 families
Gingival fibromatosis - progressive deafness	2 families
Growth deficiency - brachydactyly - dysmorphism	2 families
Jackson-Weiss syndrome	2 families
Mesoaxial synostotic syndactyly with phalangeal reduction	2 families
Microphthalmia with brain and digit anomalies	2 families
North Carolina macular dystrophy	2 families
Oral-facial-digital syndrome type 8	2 families
Partington syndrome	2 families
Progressive bifocal chorioretinal atrophy	2 families
Retinitis pigmentosa - intellectual deficit - deafness - hypogenitalism	2 families
Skeletal dysplasia - intellectual deficit	2 families
Spastic paraplegia - glaucoma - intellectual deficit	2 families
Thickened earlobes - conductive deafness	2 families
X-linked Ehlers-Danlos syndrome	2 families
Albinism-deafness syndrome	1 family
Aphalangy - syndactyly - microcephaly	1 family
Autosomal dominant Charcot-Marie-Tooth disease type 2F	1 family
Autosomal dominant Charcot-Marie-Tooth disease type 2G	1 family
Autosomal dominant Charcot-Marie-Tooth disease type 2L	1 family
Autosomal dominant limb-girdle muscular dystrophy type 1F	1 family
Autosomal dominant limb-girdle muscular dystrophy type 1G	1 family
Autosomal dominant palmoplantar keratoderma and congenital alopecia	1 family
Autosomal dominant spastic paraplegia type 29	1 family
Autosomal dominant spastic paraplegia type 38	1 family
Autosomal dominant spastic paraplegia type 9	1 family

Diseases name	Number of published cases or families
Autosomal recessive cerebellar ataxia - saccadic intrusion	1 family
Autosomal recessive spastic paraplegia type 14	1 family
Autosomal recessive spastic paraplegia type 23	1 family
Autosomal recessive spastic paraplegia type 24	1 family
Autosomal recessive spastic paraplegia type 25	1 family
Autosomal recessive spastic paraplegia type 28	1 family
Autosomal recessive spastic paraplegia type 30	1 family
Autosomal recessive spastic paraplegia type 32	1 family
Autosomal recessive spastic paraplegia type 35	1 family
Banki syndrome	1 family
Berant syndrome	1 family
Brachydactyly - nystagmus - cerebellar ataxia	1 family
Brachydactyly type A7	1 family
Cabezas syndrome	1 family
Camptobrachydactyly	1 family
Charcot-Marie-Tooth disease type 2B2	1 family
Congenitally short costocoracoid ligament	1 family
Coronary artery disease - hyperlipidemia - hypertension - diabetes - osteoporosis	1 family
Craniofacial conodysplasia	1 family
Craniosynostosis, Philadelphia type	1 family
Cyprus facial-neuromusculoskeletal syndrome	1 family
Developmental delay - deafness, Hildebrand type	1 family
Distal arthrogyrosis type 6	1 family
Dyschondrosteosis - nephritis	1 family
Ehlers-Danlos syndrome type 10	1 family
Episodic ataxia type 3	1 family
Familial partial lipodystrophy due to AKT2 mutations	1 family
Fried syndrome	1 family
Hereditary vascular retinopathy	1 family
Hydrocephalus - blue sclerae - nephropathy	1 family
Intellectual deficit, Birk-Barel type	1 family
Intellectual deficit, X-linked, Pai type	1 family
Kumar-Levick syndrome	1 family
Microtia - eye coloboma - imperforation of the nasolacrimal duct	1 family
Neuropathy with hearing impairment	1 family
Oculodental syndrome, Rutherford type	1 family
Oculogastrointestinal muscular dystrophy	1 family
Pili torti - onychodysplasia	1 family
Ptosis - strabismus - ectopic pupils	1 family
Schizophrenia - intellectual deficit - deafness - retinitis	1 family
Short fifth metacarpals - insulin resistance	1 family
Short stature - pituitary and cerebellar defects - small sella turcica	1 family
Spondyloepiphyseal dysplasia, Reardon type	1 family

Diseases name	Number of published cases or families
Steroid dehydrogenase deficiency - dental anomalies	1 family
Tietz syndrome	1 family
Trichodysplasia - amelogenesis imperfecta	1 family
Ulnar/fibula ray defect - brachydactyly	1 family
Upington disease	1 family
Van den Bosch syndrome	1 family
Woolly hair - hypotrichosis - everted lower lip - outstanding ears	1 family
X-linked hereditary sensory and autonomic neuropathy with deafness	1 family
X-linked myopathy with postural muscle atrophy	1 family
X-linked recessive intellectual deficit - macrocephaly - ciliary dysfunction	1 family
X-linked spastic paraplegia type 16	1 family
Short fifth metacarpals - insulin resistance	1 family
Short stature - pituitary and cerebellar defects - small sella turcica	1 family

Diseases name	Number of published cases or families
Spondyloepiphyseal dysplasia, Reardon type	1 family
Steroid dehydrogenase deficiency - dental anomalies	1 family
Tietz syndrome	1 family
Trichodysplasia - amelogenesis imperfecta	1 family
Ulnar/fibula ray defect - brachydactyly	1 family
Upington disease	1 family
Van den Bosch syndrome	1 family
Woolly hair - hypotrichosis - everted lower lip - outstanding ears	1 family
X-linked hereditary sensory and autonomic neuropathy with deafness	1 family
X-linked myopathy with postural muscle atrophy	1 family
X-linked recessive intellectual deficit - macrocephaly - ciliary dysfunction	1 family
X-linked spastic paraplegia type 16	1 family

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

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