Orphanet Report Series
Rare Diseases collection

Prevalence of rare diseases : Bibliographic data

Listed in order of decreasing prevalence or number of published cases

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Method

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, EMEA 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: EMEA, 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)
Hyperplastic polyposis syndrome	50
Lupus erythematosus, cutaneous	50
Narcolepsy-cataplexy	49
Melanoma, familial	46.8
Squamous cell carcinoma of head and neck	46
Autism	45
Tetralogy of Fallot	45
Arrhythmogenic right ventricular dysplasia	43.5
Meniere disease	42.5
Triplo-X syndrome	42.5
Chromosome Y deletion	42
Scleroderma	42
Parkinson's disease dementia, familial	41
Fetal cytomegalovirus syndrome	40
Mucocutaneous venous malformations	40
Parkinsonism, young adult onset	37.5
Follicular lymphoma	36
Non-Hodgkin malignant lymphoma	36
Elliptocytosis, hereditary	35
Osteochondritis dissecans	35
Radiation proctitis	35
Adactylia unilateral	34
Cryptosporidiosis	34
Malignant hyperthermia	33
Charcot-Marie-Tooth disease	32.5
Transposition of the great arteries	32.5
Leukaemia, B-cell lymphocytic, chronic	32
Acute Respiratory Distress Syndrome, adult	30
Arthrogryposis multiplex congenita	30
Marfan syndrome	30
Hypothyroidism, congenital	29
Retinitis pigmentosa	27.5
Thrombocythemia, essential	27.5
Pulmonary fibrosis, idiopathic	27
Post-transplant lymphoproliferative disease	26.2
Alpha-1 antitrypsin deficiency	25
Breast cancer, familial	25
Esophageal atresia	25
Long QT syndrome, familial	25
Myelodysplastic syndromes	25
Neurofibromatosis type 1	25

Diseases name	Estimated prevalence (/100,000)
Polycythemia vera	25
Polydactyly, preaxial	25
Syndactyly, type 1	25
Thrombocytopenic purpura, autoimmune	25
Anorectal malformation	24
Legg-Calve-Perthes disease	23
VATER association	23
Keratoconjunctivitis, vernal	21
Arthritis, oligoarticular juvenile	20.5
Dermatitis herpetiformis	20.2
Atresia of small intestine	20
Atrioventricular canal, partial	20
Gastric cancer	20
Hirschsprung disease	20
Isolated scaphocephaly	20
Monosomy 22q11	20
Spherocytosis hereditary	20
Sucrase-isomaltase deficiency, congenital	20
Tuberculosis	20
Turner syndrome	20
Corpus callosum agenesis neuronopathy	19
Nephrotic syndrome, idiopathic, steroid-sensitive	18
Cardiomyopathy, familial dilated	17.5
Boutonneuse fever	17
Renal agenesis, bilateral	17
Ichthyosis, X-linked	16.6
MELAS syndrome	16
Stromal keratitis	16
Leucinosis	15.6
Acyl-CoA dehydrogenase, medium chain, deficiency of	15
Atrioventricular canal, complete	15**
Diaphragmatic hernia, congenital	15
Lennox-Gastaut syndrome	15
Microtia	15
Parkinson disease, genetic types	15
Sarcoidosis	15
Dermatomyositis	14.8
Polymyositis	14.8
Fragile X syndrome	14.25
Myeloma, multiple	14.25
Anophthalmia - Microphthalmia, isolated	14

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Cystinuria	14
Primary biliary cirrhosis	13.5
Stickler syndrome	13.5
Williams syndrome	13.3
Androgen insensitivity syndrome	13
Bronchopulmonary dysplasia	13
Gastrointestinal stromal tumour	13
Soft tissue sarcomas	13
Trisomy 13	13**
Buerger's disease	12.5
Ehlers-Danlos syndrome, type 3	12.5
Supravalvar aortic stenosis	12.5
Von Willebrand disease	12.5
Cystic fibrosis	12
Gastroschisis	12
Gonadal dysgenesis, XX type	12
Omphalocele	12
Leukaemia, myeloid, acute	11.8
Focal dystonia	11.7
MURCS association	11.25
Stargardt disease	11.25
Glioblastoma	11
Hepatic venoocclusive disease	11
Multiple endocrine neoplasia type 1	11
Primary sclerosing cholangitis	11
Sickle cell anaemia	11
Prader-Willi syndrome	10.7
Alopecia totalis	10.5
Collagenous colitis	10.5
Hodgkin lymphoma, classical	10.5
Nephroblastoma	10.1
3-methylglutaconic aciduria, type 3	10
Achalasia, primary	10
Adrenal hyperplasia, congenital	10
Bone tumour	10
Cholangiocarcinoma	10
Dermatofibrosarcoma protuberans	10
Distal myopathy, Welander type	10
Duane syndrome	10
Factor II deficiency	10
Hemiplegic migraine, familial or sporadic	10
Idiopathic hypereosinophilic syndrome	10
Isolated plagiocephaly	10
Leber amaurosis, congenital	10
Lung cancer, small cell	10
Myelofibrosis with myeloid metaplasia	10
Neuroblastoma	10

Diseases name	Estimated prevalence (/100,000)
Pancreatitis, acute, recurrent	10
Phaeochromocytoma and paraganglioma, secreting	10
Polymorphic catecholergic ventricular tachycardia	10
Mayer-Rokitansky-Küster-Hauser syndrome	9
Mitochondrial diseases of nuclear origin	9
Neuropathy, hereditary, with liability to pressure palsies	9
Trisomy 18	9**
Giant cell arteritis	8.9
Lymphoedema, congenital	8.8
Tuberous sclerosis	8.8
Pierre Robin sequence, isolated	8.75
Duodenal atresia	8.55
Henoch-Schoenlein purpura	8.5
Myasthenia gravis	8.5
NARP syndrome	8.5
Syringomyelia	8.4
Cutaneous lymphomas	8.3
Choanal atresia	8.2
Esophageal carcinoma	8
Leukaemia, promyelocytic, acute	8
Polyarthritis, juvenile, rheumatoid factor-negative	8
Porphyria, acute hepatic	8
Hyperlipidemia type 3	7.8
Hemophilia	7.7
Kallmann syndrome	7.7
Immunodeficiency, common variable	7.5
Microscopic polyangiitis	7.5
Beckwith-Wiedemann syndrome	7.3
Pulmonary valve stenosis, congenital	7.2
Oculocutaneous albinism	7.15
Cerebellar ataxia, autosomal recessive	7
Cystathioninuria	7
Facioscapulohumeral muscular dystrophy	7
Fryns syndrome	7**
Holoprosencephaly	7
Sotos syndrome	7**
Thyroid carcinoma, medullary	7
Isolated trigonocephaly	6.7
Iminoglycinuria	6.68
Cat-scratch disease	6.6
Galactosemia	6.6
Wegener granulomatosis	6.6
Angelman syndrome	6.5
Carcinoma of the gallbladder	6.5
Leber hereditary optic neuropathy	6.5
Leukaemia, lymphoblastic, acute	6.5
Osteogenesis imperfecta	6.5

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Polycystic kidney disease, autosomal recessive	6.5
Smith-Lemli-Opitz syndrome	6.5
Ectopia lentis isolated	6.4
Arthritis, systemic-onset, juvenile idiopathic	6.3
Vasculitis	6.3
Huntington disease	6.2
Amyotrophic lateral sclerosis	6
Cerebral arteriovenous fistula	6
Digitotalar dysmorphism	6
Leukaemia, myeloid, chronic	6
Muscular dystrophy, tibial	6
Optic atrophy	6
Treacher-Collins syndrome	6
Wilson disease	5.84
Arthritis, enthesitis-related	5.7
Biliary atresia	5.6
Pendred syndrome	5.5
Retinoblastoma	5.4
Alzheimer disease, familial	5.3
Zollinger-Ellison syndrome	5.3
Cornelia de Lange syndrome	5.25
Familial adenomatous polyposis	5.25
Acromegaly	5
Adrenoleukodystrophy, X-linked	5
Epiphyseal dysplasia multiple	5
Fructose intolerance	5
Hydrolethalus	5**
Isolated brachycephaly	5
Muscular dystrophy, Duchenne and Becker types	5
Osteosarcoma	5
Parietal foramina	5
Primary ciliary dyskinesia	5
Rheumatic fever	5
Spastic paraplegia, familial	5
Thomsen and Becker disease	5
Tricuspid atresia	5
Tritanopia	4.8
Acrocephalosyndactyly	4.6
Monosomy 5p	4.6
Multiple system atrophy	4.6
Achondroplasia	4.5
Lobar emphysema, congenital	4.5
Retinoschisis, X-linked	4.5
Steinert myotonic dystrophy	4.5
Best disease	4.4
Chronic inflammatory demyelinating polyneuropathy	4.4
Arthritis, juvenile psoriatic	4.2

Diseases name	Estimated prevalence (/100,000)
Polyarthritis, juvenile, rheumatoid factor-positive	4.2
Hemimelia	4.15
Rett syndrome	4.15
Amniotic bands	4**
Autoimmune polyendocrinopathy, type 1	4
Ceroid lipofuscinosis, neuronal	4
Corticobasal degeneration	4
Hartnup syndrome	4
Histidinemia	4
Idiopathic hypersomnia	4
Meckel syndrome	4**
Phenylketonuria	4
Smith-Magenis syndrome	4
Mantle cell lymphoma	3.9
Acute interstitial pneumonia	3.8
Anisakiasis	3.8
Calpainopathy	3.8
Pemphigus vulgaris	3.8
Propionic acidemia	3.75
Supranuclear palsy, progressive	3.7
West syndrome	3.7**
Diastrophic dwarfism	3.5
Ehlers-Danlos syndrome, classic type	3.5
Goldenhar syndrome	3.5
MASA syndrome	3.5
Relapsing polychondritis	3.5
Rendu-Osler-Weber disease	3.5
Thanatophoric dwarfism	3.5**
Usher syndrome	3.5
Guillain-Barré syndrome	3.45
Graft versus host disease	3.4
Achromatopsia	3.33
Choroidal dystrophy, central areolar	3.33
Hypochondroplasia	3.3
Multiple endocrine neoplasia, type 2	3.3
Parsonage-Turner syndrome	3.3
Anencephaly	3.2**
Moya-Moya disease	3.16
Acatalasemia	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
Pseudoachondroplasia	3
Saethre-Chotzen syndrome	3

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Spinal muscular atrophy, proximal	3
Kennedy disease	2.8
Leigh syndrome	2.75**
Spinal muscular atrophy, proximal, type 2	2.6
Spinal muscular atrophy, proximal, type 3	2.6
Waldenström macroglobulinemia	2.6
Behcet disease	2.5
BOR syndrome	2.5
Bullous pemphigoid	2.5
Cone rod dystrophy	2.5
Epidermolysis bullosa, epidermolytic	2.5
Friedreich ataxia	2.5
Fructose-1,6-bisphosphatase deficiency	2.5
Gitelman syndrome	2.5
Heterotaxia	2.5
Niemann-Pick disease	2.5**
Palmoplantar keratoderma, diffuse, Norrbotten dominant type	2.5
Pseudoxanthoma elasticum	2.5
Restrictive cardiomyopathy, idiopathic or familial	2.5
Meconium aspiration syndrome	2.44
Waardenburg syndrome	2.4
3-methylcrotonylglycinuria	2.25
Ondine syndrome	2.25
Peutz-Jeghers syndrome	2.2
Ataxia, cerebellar, autosomal dominant	2.15
GRACILE syndrome	2.12**
Alport syndrome	2
Choroideremia	2
Coats disease	2
Craniopharyngioma	2
Crouzon disease	2
Exostoses, multiple	2
Gaucher disease	2
Giant pigmented hairy nevus	2
Hemicrania, paroxysmal	2
Kearns-Sayre syndrome	2
Klippel-Feil syndrome	2
Langerhans cell histiocytosis	2
Lateral body wall complex	2**
Nail-patella syndrome	2
Non-distal trisomy 12p	2**
Ocular albinism, X-linked recessive	2
Persistent hyperinsulinemic hypoglycemia of infancy	2
Poland anomaly	2
Sarcosinemia	2
Thyrotoxic periodic paralysis	2

Diseases name	Estimated prevalence (/100,000)
Van Der Woude syndrome	2
Wolf-Hirschhorn syndrome	2**
Sternal cleft	< 2
Gamma-sarcoglycanopathy	1.96
Muenke syndrome	1.8**
Amoebiasis due to free-living amoebae	1.75
Aniridia	1.75
Fabry disease	1.75
2,8 dihydroxyadenine urolithiasis	1.7
Kaposi's sarcoma	1.7
Walker-Warburg syndrome	1.65**
Charcot-Marie-Tooth disease, X-linked	1.6
Schizencephaly	1.54
Antisynthetase syndrome	1.5
Budd-Chiari syndrome	1.5
CDG syndrome	1.5**
Darier disease	1.5
Femur-fibula-ulna complex	1.5
Laryngo-tracheo-oesophageal cleft	1.5
Multifocal motor neuropathy with conduction block	1.5
Porphyria, chronic hepatic	1.5
Primary lateral sclerosis	1.5
Pulmonary arterial hypertension	1.5
Severe combined immunodeficiency T- B+, X-linked	1.5
Alagille syndrome	1.4
Cat-eye syndrome	1.35
Netherton disease	1.35
Mucopolysaccharidosis type 1	1.3
Apert syndrome	1.25
Maternal hyperphenylalaninemia	1.25
Still's disease, adult onset	1.23
Oral-facial-digital syndrome syndrome, type 1	1.2
Superficial pemphigus	1.2
Kabuki syndrome	1.16
Glycogen storage disease type 2	1.1
Ligneous conjunctivitis	1.1
Mucopolysaccharidosis type 3	1.1
Split hand - split foot	1.1
Zellweger syndrome	1.1
Medullary cystic kidney disease, autosomal recessive	1.05
Cutis verticis gyrata - intellectual deficit	1.02
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency of	1
Acanthamoeba keratitis	1
Adrenocortical carcinoma	1
Albers-Schonberg osteopetrosis	1
Ataxia telangiectasia	1

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Chondrodysplasia punctata, rhizomelic type	1
Chronic hiccup	1
Churg-Strauss syndrome	1
Clouston syndrome	1
Coloboma, ocular	1
Crigler-Najjar syndrome	1
Dyserythropoietic anaemia, congenital	1
Ehlers-Danlos syndrome, type 4	1
Fanconi anaemia	1
Gorlin syndrome	1
Harding ataxia	1
Holt-Oram syndrome	1
Hypokalemic periodic paralysis	1
Isovaleric acidemia	1
Lambert-Eaton myasthenic syndrome	1
Macrophagic myofasciitis	1
Nemaline myopathy	1
Nijmegen breakage syndrome	1**
Oculopharyngeal muscular dystrophy	1
Ornithine carbamoyltransferase deficiency	1
Proximal myotonic myopathy	1
Rubella syndrome, congenital	1**
Rubinstein-Taybi syndrome	1
Sirenomelia	1**
Tracheal agenesis	1**
Acalvaria	< 1**
Gaucher disease, type 1	0.94
Lewis-Sumner syndrome	0.9
MERRF syndrome	0.9
Protoporphyria, erythropoietic	0.9
Joubert syndrome	0.85
Niemann-Pick disease, type C	0.85
Bardet-Biedl syndrome	0.8
Criss-cross heart	0.8
Muscular dystrophy limb-girdle	0.8
Ebstein anomaly	0.75
Hepatitis, chronic autoimmune	0.75
Hyperkalemic periodic paralysis	0.75
Krabbe disease	0.75**
Mastocytosis, cutaneous	0.75
Myasthenic syndromes, congenital	0.75
Niemann-Pick disease, type B	0.75**
Osteopetrosis, malignant, autosomal recessive	0.75**
Sandhoff disease	0.75
Albright hereditary osteodystrophy	0.72
Carbamoylphosphate synthetase deficiency	0.7
Menkes syndrome	0.7

Diseases name	Estimated prevalence (/100,000)
Goodpasture syndrome	0.64
Glycogen storage disease type 4	0.6
Hyperlipoproteinemia type 1	0.6
Mucopolysaccharidosis type 2	0.6
Sympathetic ophthalmia	0.6
Alpha-sarcoglycanopathy	0.57
Beta-sarcoglycanopathy	0.57
Delta-sarcoglycanopathy	0.57
Wolfram syndrome	0.57
Cantrell pentalogy	0.55**
Coffin-Lowry syndrome	0.55
Paroxysmal nocturnal haemoglobinuria	0.55
Muscular dystrophy, Fukuyama type	0.54
Birt-Hogg-Dube syndrome	0.5
Cutaneous neuroendocrine carcinoma	0.5
Cystinosis	0.5
Diabetes insipidus, nephrogenic	0.5
Neurofibromatosis type 2	0.5
Rieger syndrome	0.5
Xeroderma pigmentosum	0.5
X-linked dominant chondrodysplasia punctata	0.5
Inclusion body myositis, IBM	0.49
Agammaglobulinemia X-linked	0.45
Cowden syndrome	0.45
Takayasu arteritis	0.45
Werner syndrome	0.45
Townes-Brocks syndrome	0.42
Aplastic anaemia	0.4
Early onset torsion dystonia	0.4
Erythroderma, congenital ichthyosiform, bullous	0.4
Glutaryl-CoA dehydrogenase deficiency	0.4
Haemolytic anaemia, due to red cell pyruvate kinase deficiency	0.4
Homocystinuria due to cystathionine beta-synthase deficiency	0.4
Mucopolysaccharidosis type 4	0.4
Neutropenia severe, congenital	0.4**
Sjögren-Larsson syndrome	0.4
Lesch-Nyhan syndrome	0.38
Pfeiffer syndrome	0.38
Campomelic dysplasia	0.35
Christ-Siemens-Touraine syndrome	0.35
Severe combined immunodeficiency T- B-	0.35
Spondylometaphyseal dysplasia	0.34
Ichthyosis, lamellar	> 0.33
Mastocytosis, systemic	0.33
Blackfan-Diamond disease	0.32

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Spinal muscular atrophy, proximal, type 4	0.32
Alkaptonuria	0.3
Aortic arch interruption	0.3**
Dystonia, dopa-responsive	0.3
Emery-Dreifuss muscular dystrophy	0.3
Miller-Dieker syndrome	0.3
Muscular dystrophy, congenital, type 1A	0.3
Pterygium popliteal syndrome, autosomal dominant	0.3
Tay-Sachs disease	0.3**
Transmissible spongiform encephalopathies	0.3
Dentatorubral pallidoluysian atrophy	< 0.3
Wolman disease	0.28**
Epidermolysis bullosa, dystrophic	0.27
Spinal muscular atrophy, proximal, type 1	0.26
Factor VII deficiency	0.25
Lipodystrophy, Berardinelli type	0.25
Niemann-Pick disease, type A	0.25**
Papillon-Lefevre syndrome	0.25
Pelizaeus-Merzbacher disease	0.25
Piebaldism	0.25
Progeria	0.25**
Leptospirosis	0.24
Severe combined immunodeficiency due to adenosine deaminase deficiency	0.22
Acrodermatitis enteropathica, zinc deficiency type	0.2
Diabetes mellitus, neonatal	0.2
Granulomatous disease, chronic	0.2
Hyperglycinemia, isolated nonketotic	0.2
Hyperoxaluria	0.2
Incontinentia pigmenti	0.2
Jeune syndrome	0.2
Short stature due to growth hormone resistance	0.2
Unverricht-Lundborg disease	0.2
Von Hippel-Lindau disease	0.2
Lowe syndrome	0.19
Sezary's syndrome	0.18
Atypical coarctation of aorta	0.17**
Metachromatic leukodystrophy	0.16
Mucopolysaccharidosis type 6	0.16**
Epilepsy, pyridoxin-dependent	0.15
Fibrinogen deficiency, congenital	0.15
Hermansky-Pudlak syndrome	0.15
Mucolipidosis type 2	0.15**
Muscular dystrophy, congenital, non merosin negative	0.15
Polycystic lipomembranous osteodysplasia - sclerosing leukoencephalopathy	0.15
Wiskott-Aldrich syndrome	0.15

Diseases name	Estimated prevalence (/100,000)
CHARGE syndrome	0.14
Thyroid carcinoma, anaplastic	0.13
Xanthomatosis cerebrotendinous	0.13
Pancreatitis, hereditary	0.125
Bartter syndrome	0.12
Lissencephaly, type 2	0.12
Medullary cystic kidney disease, autosomal dominant, with or without hyperuricemia	0.11
Alpha-mannosidosis	0.1
Creutzfeldt-Jakob disease	0.1
Diffuse leiomyomatosis - Alport syndrome X-linked	0.1
Distal myopathy, Nonaka type	0.1
Dyskeratosis congenita	0.1
Evans syndrome	0.1
Ewing sarcoma	0.1
Factor V deficiency	0.1
Factor XI deficiency, congenital	0.1
Familial cold urticaria	0.1
Hemophilia, acquired	0.1
Lemierre syndrome	0.1
Leprechaunism	0.1**
Lymphangioleiomyomatosis	0.1
Neutropenia cyclic	0.1
Pulmonary alveolar proteinosis	0.1
Pure autonomic failure	0.1
Refsum disease	0.1
Senior-Loken syndrome	0.1
Stiff-man syndrome	0.1
Tibial aplasia - ectrodactyly	0.1
Tibial hemimelia	0.1
X-linked lymphoproliferative disease	0.1
Lafora disease	< 0.1
Fibrodysplasia ossificans progressiva	0.08
Epidermolysis bullosa, junctional	0.06
Mendelian susceptibility to atypical mycobacteria	0.059
Aceruloplasminemia	0.05
Botulism	0.05
Chordoma	0.05
Craniofacial dyssynostosis	0.05
Gaucher disease, type 3	0.05
Osteoporosis - pseudoglioma	0.05
Tyrosinemia type 1	0.05
Fibrous dysplasia of bone	< 0.05
Factor XIII deficiency, congenital	0.04
Naegeli-Franceschetti-Jadassohn syndrome	0.035
Muscular dystrophy congenital, with integrin deficiency	0.03
Alpers syndrome	0.025

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)
Sialidosis type 1	0.02**
Sialidosis, type 2	0.02**
Gaucher disease, type 2	0.01

Diseases name	Estimated prevalence (/100,000)
Perinatal-lethal Gaucher disease	0.01
Refsum disease, infantile form	0.005

** Prevalence at birth

List of diseases by decreasing number of published cases

Diseases name	Number of published cases or families
Thalidomide embryopathy	5000 cases
Angio-osteohypertrophic syndrome	1000 cases
Whipple disease	1000 cases
Alveolar echinococcosis	< 1000 cases
Esthesioneuroblastoma	< 1000 cases
Rickettsialpox	> 800 cases
Fetal methyl mercury syndrome	800 cases
Western equine encephalitis	> 600 cases
Enchondromatosis	600 cases
Familial dysautonomia	550 cases
Adamantinoma	513 cases
True hermaphrodism	> 500 cases
CADASIL	500 cases
Histiocytosis, sinus, with massive lymphadenopathy	500 cases
Malakoplasia	500 cases
Rhabdoid tumour	500 cases
Epidermal nevus syndrome	> 400 cases
Castleman disease	400 cases
Silver-Russell dwarfism	400 cases
4-hydroxybutyricaciduria	350 cases
Leukocyte adhesion deficiency (LAD)	< 350 cases
Alexander disease	300 cases
Alström syndrome	300 cases
CDG syndrome, type Ia	300 cases
Cutis marmorata telangiectatica congenita	300 cases
Focal dermal hypoplasia	300 cases
Gräsbeck-Imerslund disease	300 cases
Lipodystrophy, familial partial, Dunnigan type	300 cases
Melorheostosis	300 cases
Methylmalonic acidemia - homocystinuria	300 cases
Methylmalonicacidemia - homocystinuria, type cbl C	300 cases
Moebius syndrome	300 cases
Norrie disease	300 cases
Rothmund-Thomson syndrome	300 cases
Von Willebrand syndrome, acquired	300 cases
Lipoid proteinosis	> 280 cases
Eosinophilic gastroenteritis	280 cases
Chronic recurrent multifocal osteomyelitis, juvenile	> 260 cases
Job syndrome	250 cases

Diseases name	Number of published cases or families
Lipodystrophy, partial acquired	250 cases
Pulmonary haemosiderosis, primary	250 cases
Caroli's disease	< 250 cases
6-pyruvoyl-tetrahydropterin synthase, deficiency	248 cases
Oculodentodigital dysplasia	243 cases
Pachyonychia congenita	230 cases
Lhermitte-Duclos disease	220 cases
Muir-Torre syndrome	205 cases
Pachydermoperiostosis	204 cases
Aarskog-Scott syndrome	> 200 cases
Blue rubber bleb nevus	> 200 cases
Erythrokeratodermia variabilis, Mendes da Costa type	> 200 cases
Monosomy 22q13	> 200 cases
Papulosis, malignant atrophic	> 200 cases
Porphyria, congenital erythropoietic	> 200 cases
Pseudoarthrosis of clavicle, congenital	> 200 cases
Tufted angioma	> 200 cases
Adenosine monophosphate deaminase deficiency	200 cases
Aicardi syndrome	200 cases
Camurati-Engelmann disease	200 cases
Cockayne syndrome	200 cases
Cogan syndrome	200 cases
Costello syndrome	200 cases
Eosinophilic fasciitis	200 cases
Femur bifid - monodactylous - ectrodactyly	200 cases
Glucose-galactose malabsorption	200 cases
Gorham-Stout disease	200 cases
Kimura disease	200 cases
LEOPARD syndrome	200 cases
Marinesco-Sjogren syndrome	200 cases
Multiple pterygium syndrome, lethal form	200 cases
Proteus syndrome	200 cases
Shwachman-Diamond syndrome	200 cases
Subcorneal pustular dermatosis	200 cases
Monosomy 18p	< 200 cases
Mowat-Wilson syndrome	< 200 cases
Multicentric reticulohistiocytosis	< 200 cases
Persistent Mullerian duct syndrome	< 200 cases
Sideroblastic anaemia, X-linked	< 200 cases
Hyperimmunoglobinemia D with recurrent fever	180 cases

Diseases name	Number of published cases or families
Erdheim-Chester disease	178 cases
Kasabach-Merritt syndrome	> 175 cases
Celiac disease - epilepsy - occipital calcifications	170 cases
Alpha thalassemia - mental retrdation, X-linked	168 cases
Carney complex	160 cases
Madras motor neuron disease	154 cases
Infantile neuroaxonal dystrophy	> 150 cases
Cloverleaf skull syndrome	150 cases
Denys-Drash syndrome	150 cases
Dubowitz syndrome	150 cases
Ellis Van Creveld syndrome	150 cases
Fraser syndrome	150 cases
Humeroradial synostosis	150 cases
Jacobsen syndrome	150 cases
McLeod neuroacanthocytosis syndrome	150 cases
Xanthinuria	150 cases
Arthrogryposis - renal dysfunction - cholestasis	< 150 cases
CACH syndrome	148 cases
Inflammatory pseudotumour of the liver	143 cases
Regional odontodysplasia	139 cases
Dihydropteridine reductase deficiency	134 cases
Aorto-ventricular tunnel	130 cases
Weill-Marchesani syndrome	128 cases
Vulvovaginal gingival syndrome	127 cases
Robinow syndrome	> 120 cases
Aicardi-Goutieres syndrome	120 cases
MULIBREY nanism	115 cases
Bickel-Fanconi glycogenosis	112 cases
Myoclonic epilepsy of infancy	106 cases
Achondrogenesis	> 100 cases
Bloom syndrome	> 100 cases
Carnitine palmitoyl transferase 2 deficiency	> 100 cases
Central neurocytoma	> 100 cases
Cutis laxa	> 100 cases
Eosinophilic pneumonia, acute, idiopathic	> 100 cases
Fetal varicella syndrome	> 100 cases
KID/HID syndrome	> 100 cases
Mucolipidosis type 4	> 100 cases
Ochoa syndrome	> 100 cases
Pulmonary lymphangiectasia, congenital	> 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

Diseases name	Number of published cases or families
Cataracts, congenital - facial dysmorphism - neuropathy	100 cases
Chaotic atrial tachycardia	100 cases
CINCA syndrome	100 cases
Cohen syndrome	100 cases
Defective expression of HLA class 2	100 cases
Epidermolysis bullosa, acquired	100 cases
Fanconi syndrome	100 cases
Freeman-Sheldon syndrome	100 cases
Fucosidosis	100 cases
Greig syndrome	100 cases
Helicoid peripapillary chorioretinal degeneration	100 cases
Hemochromatosis, neonatal	100 cases
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	100 cases
Internal carotid agenesis	100 cases
Larsen syndrome	100 cases
Osteopathia striata - cranial sclerosis	100 cases
Pallister-Hall syndrome	100 cases
Peritoneal leiomyomatosis, disseminated	100 cases
PHACE syndrome	100 cases
Poikiloderma of Kindler	100 cases
Retinal arteries, tortuosity of	100 cases
Roberts syndrome	100 cases
Schwartz-Jampel syndrome	100 cases
Seckel syndrome	100 cases
Triple A syndrome	100 cases
Brown-Vialetto-van Laere syndrome	< 100 cases
Hallermann-Streiff-Francois syndrome	< 100 cases
Hypertrichosis lanuginosa congenita	< 100 cases
Hypocomplementaemic leucocytoclasic vasculitis	< 100 cases
Ichthyosis congenita, harlequin type	< 100 cases
Muscular dystrophy, Ullrich type, congenital	< 100 cases
Panniculitis, cytophagic histiocytic	< 100 cases
Susac syndrome	< 100 cases
Tracheobronchomegaly	< 100 cases
Tyrosinemia, type 2	< 100 cases
Silent sinus syndrome	98 cases
Acrofacial dysostosis, Nager type	90 cases
Megacystis microcolon - intestinal hypoperistalsis - hydronephrosis	89 cases
Early infantile epileptic encephalopathy	88 cases
Myoneurogastrointestinal encephalopathy syndrome	87 cases
Glycogen storage disease due to LAMP-2 deficiency	84 cases
Uhl anomaly	84 cases
Lipodystrophy, acquired generalized	80 cases

Diseases name	Number of published cases or families
Metatropic dwarfism	80 cases
Wells syndrome	80 cases
Rapp-Hodgkin syndrome	72 cases
Tangier disease	> 70 cases
Cleft lip/mandibule, median	70 cases
Craniometaphyseal dysplasia	70 cases
Lupus erythematosus, bullous systemic	70 cases
Pseudohypoaldosteronism, type 1	70 cases
Ring chromosome 18	70 cases
Diffuse neonatal haemangiomatosis	< 70 cases
Glutathione synthetase deficiency	65 cases
Hyperferritinemia, hereditary, with congenital cataracts	> 64 cases
Paraneoplastic pemphigus	> 60 cases
Cerebro-costo-mandibular syndrome	60 cases
Dyggve-Melchior-Clausen disease	60 cases
Griscelli disease	60 cases
Histiocytosis, sea-blue	60 cases
Homocystinuria without methylmalonic aciduria	60 cases
Hypertrichosis lanuginosa, acquired	60 cases
Ketoacidosis due to betaketothiolase deficiency	60 cases
Neu-Laxova syndrome	60 cases
Non-distal trisomy 10p	60 cases
Pancreatoblastoma	60 cases
Pearson syndrome	60 cases
Toriello-Carey syndrome	60 cases
Vitamin B12 responsive methylmalonic acidemia, type cbl A	60 cases
Double uterus-hemivagina-renal agenesis	< 60 cases
Ataxia, autosomal recessive, Beauce type	57 cases
Capillary leak syndrome	57 cases
Sporotrichosis	55 cases
Mazabraud syndrome	54 cases
Hennekam syndrome	> 50 cases
Megacalycosis, congenital	> 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
Anorchidia, bilateral	50 cases
Ascher syndrome	50 cases
Benign paroxysmal torticollis of infancy	50 cases
Cholestasis - lymphoedema	50 cases
Encephalopathy due to sulphite oxidase deficiency	50 cases
Focal myositis	50 cases

Diseases name	Number of published cases or families
Fronto-temporal dementia and Parkinsonism linked to chromosome 17 (FTDP-17)	50 cases
Glucocorticoid deficiency, familial	50 cases
Glucosephosphate isomerase deficiency	50 cases
Hyaline fibromatosis, juvenile	50 cases
ICF syndrome	50 cases
Mucosulfatidosis	50 cases
Ocular motor apraxia Cogan type	50 cases
Osteopetrosis, intermediate form	50 cases
Paget disease juvenile type	50 cases
Peters-plus syndrome	50 cases
Prolidase deficiency	50 cases
Rhombencephalosynapsis	50 cases
Ring chromosome 14	50 cases
Schnitzler syndrome	50 cases
Succinic acidemia	50 cases
Triple H (HHH) syndrome	50 cases
Waardenburg-Shah syndrome	50 cases
Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis	< 50 cases
Blepharo-cheilo-dontic syndrome	< 50 cases
Branchio-oculo-facial syndrome	< 50 cases
Cholesteryl ester storage disease	< 50 cases
DOOR syndrome	< 50 cases
Ehrlichiosis	< 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
Oculo-digito-esophageal-duodenal syndrome (ODED)	< 50 cases
Shprintzen-Goldberg syndrome	< 50 cases
Corticosteroid-sensitive aseptic abscesses	49 cases
Pityriasis rubra pilaris	48 cases
Coxo-podo-patellar syndrome	47 cases
Mohr-Tranebjaerg syndrome	46 cases
Encephalocraniocutaneous lipomatosis	45 cases
KBG syndrome	45 cases
Bowen-Conradi syndrome	44 cases
Ear-patella-short stature syndrome	42 cases
Desbuquois syndrome	> 40 cases
3M syndrome	40 cases
Ambras syndrome	40 cases
Babesiosis	40 cases
Carpenter syndrome	40 cases
Chylomicron retention disease	40 cases

Diseases name	Number of published cases or families
Coffin-Siris syndrome	40 cases
Distal monosomy 10q	40 cases
Ectodermal dysplasia, hypohidrotic, autosomal dominant	40 cases
Galloway-Mowat syndrome	40 cases
Macrocephaly - Cutis Marmorata Telangiectatica Congenita	40 cases
Methimazole embryofetopathy	40 cases
Phytosterolemia	40 cases
WHIM syndrome	40 cases
Acromicric dysplasia	< 40 cases
Arterial tortuosity	< 40 cases
Bruck syndrome	< 40 cases
Encephalopathy, ethylmalonic	< 40 cases
Mucopolysaccharidosis type 7	< 40 cases
Progressive bulbar paralysis of childhood	< 40 cases
Leukoencephalopathy with brain stem, spinal cord involvement - lactate elevation	39 cases
Hypotrichosis simplex	38 cases
Dysplasia, mandibuloacral	37 cases
Oculocerebrocutaneous syndrome	36 cases
Spondyloenchondrodysplasia	36 cases
Carnitine palmitoyl transferase 1 deficiency	35 cases
Cobb syndrome	35 cases
Neuropathy, sensory and autonomic , hereditary, 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
Björnstad syndrome	33 cases
Marshall-Smith syndrome	33 cases
Methylcobalamin deficiency, cbl G type	33 cases
Osteopetrosis, autosomal dominant, type 1	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
Intrauterine infection-like syndrome, congenital	> 30 cases
Non-distal trisomy 8q	> 30 cases
Tricho-dento-osseous syndrome	> 30 cases
Agnathia holoprosencephaly situs inversus	30 cases

Diseases name	Number of published cases or families
Anophthalmia - hypothalamo-pituitary insufficiency	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
Dermopathy restrictive, lethal	30 cases
Early myoclonic encephalopathy	30 cases
Elejalde syndrome	30 cases
Geroderma osteodysplastica	30 cases
Humeroradioulnar synostosis	30 cases
Marden-Walker syndrome	30 cases
Marshall's syndrome with periodic fever	30 cases
Mevalonicaciduria	30 cases
Omodysplasia	30 cases
Ophthalmo acromelic syndrome	30 cases
Otopalatodigital syndrome	30 cases
Perlman syndrome	30 cases
Polycystic kidney disease, autosomal dominant, type 1, with tuberous sclerosis	30 cases
Scalp-ear-nipple syndrome	30 cases
SHORT syndrome	30 cases
Triose phosphate-isomerase deficiency	30 cases
Weaver syndrome	30 cases
Acrofacial dysostosis, postaxial	< 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 type 7	< 30 cases
Nasopalpebral lipoma - coloboma - telecanthus	< 30 cases
Neuroectodermal syndrome, Johnson type	< 30 cases
Neurometabolic disorder due to serine deficiency	< 30 cases
Obesity due to congenital leptin deficiency	< 30 cases
Pontocerebellar hypoplasia type 2	< 30 cases
Pyle disease	< 30 cases
Ring dermoid of cornea	< 30 cases
Spontaneous periodic hypothermia	< 30 cases
Syndactyly, Cenani-Lenz type	< 30 cases
Wrinkly skin syndrome	< 30 cases
Cone rod dystrophy - amelogenesis imperfecta	29 cases
Infant epilepsy with migrant focal crisis	29 cases

Diseases name	Number of published cases or families
Leukoencephalopathy with bilateral anterior temporal lobe cysts	29 cases
Mosaic variegated aneuploidy syndrome	29 cases
Oral-facial-digital syndrome, type 6	29 cases
Thalamic calcifications, symmetrical	29 cases
Craniolenticulosutural dysplasia	28 cases
Hypertrichosis cubiti - short stature	28 cases
Catel-Manzke syndrome	27 cases
GAPO syndrome	27 cases
Geleophysic dwarfism	27 cases
Insomnia, familial fatal	27 cases
Limb-mammary syndrome	27 cases
Methylcobalamin deficiency type cbl E	27 cases
3C syndrome	25 cases
Atelosteogenesis, type II	25 cases
Book syndrome	25 cases
Erythrokeratodermia ataxia	25 cases
Keratoderma palmoplantar - spastic paralysis	25 cases
Opsismodysplasia	25 cases
Plummer-Vinson syndrome	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
Synspondylism	24 cases
Bronchobiliary fistula, congenital	23 cases
Deletion 2q24	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
Split hand - split foot - deafness	22 cases
Dehydratase deficiency	21 cases
Odonto-tricho-ungual-digito-palmarn syndrome	21 cases
Spinocerebellar ataxia, infantile onset	21 cases
Craniodiaphyseal dysplasia	> 20 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
CDG syndrome, type Ib	20 cases
Craniofrontonasal dysplasia, Teebi type	20 cases
Distal monosomy 8p	20 cases
Gray platelet syndrome	20 cases
Indifference to pain, congenital	20 cases
Juvenile temporal arteritis	20 cases
Lacrimo-auriculo-dento-digital syndrome	20 cases
Lipoamide dehydrogenase deficiency	20 cases
PIBIDS syndrome	20 cases
Acromegaloid facial appearance syndrome	< 20 cases
Acromegaloid facies - hypertrichosis	< 20 cases
Bronchopneumopathy, chronic, due to TAP deficiency	< 20 cases
Carey-Fineman-Ziter syndrome	< 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
Hemorrhagic disorders due to collagen receptors deficiency	< 20 cases
Hypertrichosis, anterior cervical, isolated	< 20 cases
Ichthyosis bullosa of Siemens	< 20 cases
IMAGe syndrome	< 20 cases
Interstitial granulomatous dermatitis with arthritis	< 20 cases
Keratosis palmaris et plantaris - clinodactyly	< 20 cases
Laryngeal abductor paralysis - mental retardation	< 20 cases
Lipodystrophy, familial partial, Köbberling type	< 20 cases
Metaphyseal anadysplasia	< 20 cases
Orotic aciduria hereditary	< 20 cases
Radio-ulnar synostosis - amegakaryocytic thrombocytopaenia	< 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
Cavitating leukoencephalopathy, progressive	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
Schopf-Schulz-Passarge syndrome	19 cases
Hypertrichotic osteochondrodysplasia	18 cases
Terminal osseous dysplasia - pigmentary defects	18 cases

Diseases name	Number of published cases or families
Aminopterin embryofetopathy	17 cases
Erythroderma, lethal, congenital	17 cases
Folate malabsorption, hereditary	17 cases
GTP cyclohydrolase I deficiency	17 cases
Malonic aciduria	17 cases
Mental retardation, X-linked, with seizures, short stature and midface hypoplasia	17 cases
Palmoplantar hypokeratosis, circumscribed	17 cases
Wolcott-Rallison syndrome	17 cases
Acromegaly - cutis verticis gyrata - corneal leukoma	16 cases
Glycogen storage, 0 type	16 cases
Mental retardation, X-linked - Dandy-Walker malformation - basal ganglia disease - Seizures	16 cases
Mental retardation, X-linked, South African type	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
IBIDS syndrome	15 cases
Laminopathy-related congenital muscular dystrophy	15 cases
Sensenbrenner syndrome	15 cases
Umbilical cord ulceration - intestinal atresia	15 cases
Anosmia, isolated, congenital	< 15 cases
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	< 15 cases
Mirhosseini-Holmes-Walton syndrome	< 15 cases
Odonto-onycho-dermal dysplasia	< 15 cases
Taurodontia - absent teeth - sparse hair	< 15 cases
Wilson-Turner syndrome	> 14 cases
ADULT syndrome	14 cases
Anophthalmia/microphthalmia - esophageal atresia	14 cases
Atkin-Flaitz syndrome	14 cases
Beta-mannosidosis	14 cases
DEND syndrome	14 cases
Dermo-odonto dysplasia	14 cases
Fingerprints, absence of - milia, congenital	14 cases
Muscular dystrophy, limb girdle, autosomal recessive, type 2G	14 cases
Oligocone trichromacy	14 cases
Optic atrophy and cataract, autosomal dominant	14 cases
Ring chromosome 17	14 cases
Aromatase deficiency	13 cases
Chondrodysplasia, Blomstrand type	13 cases

Diseases name	Number of published cases or families
Diaphragmatic hernia - exomphalos - corpus callosum agenesis	13 cases
Histiocytosis, progressive mucinous, hereditary	13 cases
Hypotelorism - cleft palate - hypospadias	13 cases
Posterior column ataxia - retinitis pigmentosa	13 cases
Acro-pectoro-renal field defect	12 cases
Alopecia- epilepsy - pyorrhea - mental subnormality	12 cases
Ataxia - deafness - optic atrophy, lethal	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
Coloboma uveal - cleft lip palate - intellectual deficit	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
Haemolytic anaemia due to adenylate kinase deficiency	12 cases
Hypoparathyroidism - deafness - renal disease	12 cases
Mental retardation, X-linked - macrocephaly - macro-orchidism	12 cases
N-acetyl-alpha-D-galactosaminidase deficiency	12 cases
Osteocraniostenosis	12 cases
Winchester disease	12 cases
Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges	11 cases
Atrial septal defect - atrioventricular conduction defects	11 cases
Corpus callosum dysgenesis, X-linked recessive	11 cases
Fibrochondrogenesis	11 cases
Fibular dimelia - diplopodia	11 cases
Foveal hypoplasia - presenile cataract	11 cases
Fuhrmann syndrome	11 cases
Lopez-Hernandez syndrome	11 cases
Martinez-Frias syndrome	11 cases
Mental retardation, X-linked, Snyder type	11 cases
Necrotizing encephalopathy, acute, autosomal dominant	11 cases
PELVIS syndrome	11 cases
Pterygium syndrome, antecubital	11 cases
Trichomegaly - retina pigmentary degeneration - dwarfism	11 cases
CHAND syndrome	> 10 cases
Chediak-Higashi syndrome	> 10 cases
Median cleft of the upper lip - corpus callosum lipoma - cutaneous polyps	> 10 cases

Diseases name	Number of published cases or families
Acromesomelic dysplasia, Hunter-Thomson type	10 cases
Arthrogryposis multiplex congenita - whistling face	10 cases
Athabaskan brainstem dysgenesis syndrome	10 cases
Barber-Say syndrome	10 cases
Boomerang dysplasia	10 cases
Cardiac conduction disease, dilated cardiomyopathy and brachydactyly	10 cases
Cataract - mental retardation - hypogonadism	10 cases
Cerebro-oculo-nasal syndrome	10 cases
Char syndrome	10 cases
Charcot-Marie-Tooth disease - nephropathy	10 cases
Choroido cerebral calcification syndrome, infantile form	10 cases
Colobomatous - microphthalmia - heart disease - hearing loss	10 cases
Deletion 2q37	10 cases
Diffuse palmoplantar keratoderma - acrocyanosis	10 cases
Distal monosomy 5q	10 cases
Ectodermal dysplasia - skin fragility syndrome	10 cases
Flynn-Aird syndrome	10 cases
Goldberg-Shprintzen megacolon syndrome	10 cases
Hyperkeratosis - hyperpigmentation syndrome	10 cases
Ichthyosis follicularis - atrichia - photophobia	10 cases
Iris coloboma with ptosis - intellectual deficit	10 cases
Juberg-Hayward syndrome	10 cases
Lipodystrophy, familial partial, associated with PPARG mutations	10 cases
Mental retardation, X-linked - hypotonia - facial dysmorphism - aggressive behaviour	10 cases
Mental retardation, X-linked, syndromic 7	10 cases
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus	10 cases
Myopathy, distal inflammatory, of the upper limbs, asymmetric	10 cases
Neurodegenerative syndrome, X-linked, Hamel type	10 cases
Nevo syndrome	10 cases
Pollitt syndrome	10 cases
Progressive vertebral fusion, non-infectious, syndromic form	10 cases
Progressive vertebral fusion, non-infectious, syndromic form	10 cases
Pseudodiastrophic dysplasia	10 cases
Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts	10 cases
Succinyl-CoA acetoacetate transferase deficiency	10 cases
Toriello-Lacassie-Droste syndrome	10 cases
Aase-Smith syndrome	< 10 cases
Acrofacial dysostosis, Rodriguez type	< 10 cases
Acromelanosis	< 10 cases
Astley-Kendall dysplasia	< 10 cases
Cardiocranial syndrome, Pfeiffer type	< 10 cases
Carpotarsal osteochondromatosis	< 10 cases
Carpotarsal osteolysis, recessive	< 10 cases
Cerebral gigantism - jaw cysts	< 10 cases
Corneal dystrophy - perceptive deafness	< 10 cases
Deafness - lymphoedema - leukaemia	< 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
Hypopituitarism - microphthalmia	< 10 cases
Larsen-like syndrome, lethal form	< 10 cases
Myoclonus hereditary - progressive distal muscular atrophy	< 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 (PEPCK) deficiency	< 10 cases
Progeria - short stature - pigmented nevi	< 10 cases
Pseudo-Zellweger syndrome	< 10 cases
Singleton-Merten dysplasia	< 10 cases
XK aprosencephaly	< 10 cases
Ataxia - apraxia - mental retardation, X-linked	9 cases
Atransferrinemia	9 cases
Bosley-Salih-Alorainy syndrome	9 cases
Brachymorphism - onychodysplasia - dysphalangism	9 cases
Cardiac anomalies - heterotaxy	9 cases
Cardiac anomalies - heterotaxy	9 cases
Cardiomyopathy - cataract - hip spine disease	9 cases
Gamma-glutamylcysteine synthetase deficiency	9 cases
Guanidinoacetate methyltransferase deficiency	9 cases
Laminopathy, type Decaudain-Vigouroux	9 cases
Macrostomia - preauricular tags - external ophthalmoplegia	9 cases
Mental retardation, X-linked, Shashi type	9 cases
Methylmalonicaciduria - homocystinuria, type cbl F	9 cases
Microdontia - type I microtia - deafness	9 cases
Oculocerebrofacial syndrome, Kaufman type	9 cases
Tricho-retino-dento-digital syndrome	9 cases
5-oxoprolinase deficiency	8 cases

Diseases name	Number of published cases or families
Ackerman syndrome	8 cases
Ankylosing vertebral hyperostosis with tylosis	8 cases
Ataxia-deafness-retardation syndrome	8 cases
Brachydactyly - preaxial hallux varus	8 cases
Campomelia, Cumming type	8 cases
Camptodactyly syndrome, Guadalajara type 1	8 cases
COACH syndrome	8 cases
Corpus callosum, agenesis - cataract - immunodeficiency	8 cases
Deafness - skeletal dysplasia - lip granuloma	8 cases
Familial hematuria, autosomal dominant - retinal arteriolar tortuosity - contractures	8 cases
Hydrocephalus - costovertebral dysplasia - Sprengel anomaly	8 cases
Hypomyelination - congenital cataract	8 cases
Kallmann syndrome - heart disease	8 cases
Leukoencephalopathy - ataxia - hypodontia - hypomyelination	8 cases
Mental retardation, X-linked - dysmorphism - cerebral atrophy	8 cases
Mental retardation, X-linked, Abidi type	8 cases
Mental retardation, X-linked, Vitale type	8 cases
Micro syndrome	8 cases
Pancreas agenesis	8 cases
Spondylometaphyseal dysplasia - cone-rod dystrophy	8 cases
Acro-renal-mandibular syndrome	7 cases
Albinism ocular - late onset sensorineural deafness	7 cases
Cardiogenital syndrome	7 cases
CDG syndrome, type Ie	7 cases
CEDNIK syndrome	7 cases
Cleft palate-lateral synechia syndrome	7 cases
Dihydropyrimidinuria	7 cases
Ehlers-Danlos syndrome, type 7C	7 cases
Gamma-glutamyl transpeptidase deficiency	7 cases
Genitopatellar syndrome	7 cases
Hyperostotic dwarfism, Lenz-Majewski type	7 cases
Mandibulofacial dysostosis, X-linked	7 cases
MEHMO syndrome	7 cases
MEHMO syndrome	7 cases
Mental retardation, X-linked, severe, Gustavson type	7 cases
Michels syndrome	7 cases
Mitochondrial myopathy with sideroblastic anaemia	7 cases
Multiple fibrofolliculoma, familial	7 cases
Neurodegenerative syndrome, X-linked, Bertini type	7 cases
Neutropenia, severe congenital, X-linked	7 cases

Diseases name	Number of published cases or families
Obesity due to pro-opiomelanocortin deficiency	7 cases
Retinal degeneration - nanophtalmos - glaucoma	7 cases
Retinohepatoendocrinologic syndrome	7 cases
Stern-Lubinsky-Durrie syndrome	7 cases
Torticollis - keloids - cryptorchidism - renal dysplasia	7 cases
3-hydroxy 3-methylglutaryl-CoA (HMG) synthase	6 cases
Acrofacial dysostosis, Catania type	6 cases
Agonadism - dextrocardia - diaphragmatic hernia	6 cases
Aplasia cutis congenita of limbs recessive	6 cases
Biliary tract malformation - renal failure	6 cases
Blepharophimosis - ptosis - esotropia - syndactyly - short stature	6 cases
CDG syndrome, type Ig	6 cases
Cutis gyrata - acanthosis nigricans - craniosynostosis	6 cases
Deafness - enamel hypoplasia - nail defects	6 cases
Ectodermal dysplasia, hidrotic, Christianson-Fourie type	6 cases
Ehlers-Danlos syndrome, spondylocheiro dysplastic form	6 cases
Eiken syndrome	6 cases
Epilepsy telangiectasia	6 cases
Fanconi ichthyosis dysmorphism	6 cases
Grange syndrome	6 cases
Hartsfield-Bixler-Demyer syndrome	6 cases
Hypopituitarism - postaxial polydactyly	6 cases
Isotretinoin-like syndrome	6 cases
Keratosis follicularis - dwarfism - cerebral atrophy	6 cases
Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus	6 cases
Mental retardation - sparse hair - brachydactyly	6 cases
Mental retardation, X-linked - psychosis - macroorchidism	6 cases
Mental retardation, X-linked, Armfield type	6 cases
Mental retardation, X-linked, Zorick type	6 cases
Moore-Federman syndrome	6 cases
Polysyndactyly - cardiac malformation	6 cases
Serpentine fibula - polycystic kidneys	6 cases
Spasticity - mental retardation - epilepsy, X-linked	6 cases
Stormorken-Sjaastad-Langslet syndrome	6 cases
Symphalangism with multiple anomalies of hands and feet	6 cases
Thumb stiffness - brachydactyly - mental retardation	6 cases
W syndrome	6 cases
Wieacker-Wolff syndrome	6 cases
Zunich-Kaye syndrome	6 cases

Diseases name	Number of published cases or families
Achalasia - microcephaly	5 cases
Acro-fronto-facio-nasal dysostosis	5 cases
Adducted thumbs-arthrogryposis, Dundar type	5 cases
Alopecia - contractures - dwarfism - mental retardation	5 cases
ANE syndrome	5 cases
Anonychia microcephaly	5 cases
Arachnodactyly - ossification abnormal - mental retardation	5 cases
Ataxia, spinocerebellar, X-linked, type 3	5 cases
Aurocephalosyndactyly	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
Choanal atresia - deafness - cardiac defects - dysmorphism	5 cases
Cholestasis pigmentary - retinopathy - cleft palate	5 cases
Cleft palate - cardiac defect - genital anomalies - ectrodactyly	5 cases
Craniodigital syndrome - intellectual deficit	5 cases
Craniosynostosis - dysmorphism - brachydactyly	5 cases
Curry-Jones syndrome	5 cases
Dacryocystitis - osteopoikilosis	5 cases
Depigmentation of the iris, acute, bilateral	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 apnoea	5 cases
Humerospinal dysostosis	5 cases
Humeroulnar synostosis	5 cases
Hypothyroidism - dysmorphism - postaxial polydactyly - intellectual deficit	5 cases
Ichthyosis - male hypogonadism	5 cases
Lissencephaly, type III - familial foetal akinesia sequence	5 cases
Lymphoedema - cerebral arteriovenous anomaly	5 cases
Matthew-Wood syndrome	5 cases
Mental retardation, X-linked - choreoathetosis - abnormal behaviour	5 cases
Mental retardation, X-linked - cubitus valgus - dysmorphism	5 cases
Mental retardation, X-linked, Lubs type	5 cases

Diseases name	Number of published cases or families
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
Oculodentoosseous dysplasia recessive	5 cases
Oculo-palato-cerebral syndrome	5 cases
Odontomicronychial dysplasia	5 cases
Onycho-tricho-dysplasia - neutropenia	5 cases
P2Y12, deficiency of	5 cases
Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma	5 cases
Paraplegia - brachydactyly - cone-shaped epiphysis	5 cases
Sacral hemangiomas - multiple congenital abnormalities	5 cases
Sillence syndrome	5 cases
Woods-Black-Norbury syndrome	5 cases
Cystic hamartoma of lung and kidney	< 5 cases
Lathosterolosis	< 5 cases
Sakati-Nyhan syndrome	< 5 cases
Abruzzo-Erickson syndrome	4 cases
Acrofacial dysostosis, palagonia type	4 cases
Anophthalmia plus syndrome	4 cases
Aortic arch anomaly - peculiar facies - mental retardation	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
Benign exophthalmos syndrome	4 cases
Bile acid synthesis defect, congenital, type 4	4 cases
Bone dysplasia, lethal, Holmgren type	4 cases
Bone fragility - craniosynostosis - proptosis - hydrocephalus	4 cases
Bonnemann-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
Charcot-Marie-Tooth disease, type 4J	4 cases
Chondrodysplasia, lethal recessive	4 cases
Choroideremia - deafness - obesity	4 cases
Cleft lip/palate - intestinal malrotation - cardiopathy	4 cases
Coxoauricular syndrome	4 cases
Cranio osteoarthropathy	4 cases
Craniofaciocardioskeletal syndrome	4 cases

Diseases name	Number of published cases or families
Craniosynostosis dandy walker hydrocephalus	4 cases
Deafness - peripheral neuropathy - arterial disease	4 cases
Diabetes mellitus, permanent neonatal - pancreatic and cerebellar agenesis	4 cases
Diaphragmatic defect - limb deficiency - skull defect	4 cases
Duker-Weiss-Siber syndrome	4 cases
Ectodermal dysplasia, Berlin type	4 cases
Ectodermal dysplasia, hidrotic, Halal type	4 cases
Ectopia lentis - chorioretinal dystrophy - myopia	4 cases
Endosteal sclerosis - cerebellar hypoplasia	4 cases
Facial onset sensory and motor neuronopathy	4 cases
Familial caudal dysgenesis	4 cases
Gombo syndrome	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
Growth retardation - microcephaly - digital abnormalities - hypospadias	4 cases
Hirschsprung disease - type D brachydactyly	4 cases
Homocarnosinosis	4 cases
Hypogammaglobulinemia due to CD19 deficiency	4 cases
Hypomandibular faciocranial dysostosis	4 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 cases
Hypotrichosis - lymphoedema - telangiectasia	4 cases
Ichthyosis - alopecia - eclabion - ectropion - mental retardation	4 cases
Immunodeficiency with natural-killer cell deficiency	4 cases
Leukodystrophy with oligodontia	4 cases
Leukoencephalopathy - metaphyseal chondrodysplasia	4 cases
Leukoencephalopathy - palmoplantar keratoderma	4 cases
Macrogyria pseudobulbar palsy	4 cases
Malignant hyperthermia - arthrogryposis - torticollis	4 cases
Mental retardation - dysmorphism - hypogonadism - diabetes mellitus	4 cases
Mental retardation, X-linked - hypogonadism - ichthyosis - obesity - short stature	4 cases
Mental retardation, X-linked - seizures - psoriasis	4 cases
Mental retardation, X-linked, Miles-Carpenter type	4 cases
Mental retardation, X-linked, Schimke type	4 cases
Mental retardation, X-linked, Seemanova type	4 cases
Mental retardation, X-linked, Siderius type	4 cases
Mental retardation, X-linked, Stevenson type	4 cases
Mental retardation, X-linked, Stocco Dos Santos type	4 cases
Mental retardation, X-linked, Stoll type	4 cases
Metaphyseal acroscyphodysplasia	4 cases
Microcephalic osteodysplastic dysplasia, Saul-Wilson type	4 cases
Microcephaly - micropenis - convulsions	4 cases
Multiple pterygium syndrome, autosomal dominant	4 cases
Myoclonus - cerebellar ataxia - deafness	4 cases
Myopathy due to calsequestrin and SERCA1 protein overload	4 cases
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	4 cases
Neuroectodermal-endocrine syndrome	4 cases
Oculo-oto-facial dysplasia	4 cases
Odontotrichomelic syndrome	4 cases
Oral-facial-digital syndrome, type 5	4 cases
Osteopoikilosis - short stature - intellectual deficit	4 cases
Palmoplantar keratoderma - amyotrophy	4 cases
Paraplegia - mental retardation - hyperkeratosis	4 cases
Recurrent infections - short stature - hypopigmentation - coarse face	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
Spondylometaphyseal dysplasia with combined immunodeficiency	4 cases
Syndactyly, type 4	4 cases
Tomé-Brunet-Fardeau syndrome	4 cases
Tricho-odonto-onychial dysplasia	4 cases
Acromesomelic dysplasia brahimi bacha type	3 cases
Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis	3 cases
Al-Gazali-Dattani syndrome	3 cases
Amelia, autosomal recessive	3 cases
Aniridia - absent patella	3 cases
Aniridia - ptosis - mental retardation - obesity, familial form	3 cases
Anonychia with flexural pigmentation	3 cases
Anophthalmia - megalocornea - cardiopathy - skeletal anomalies	3 cases

Diseases name	Number of published cases or families
Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis	3 cases
Aplasia cutis congenita - intestinal lymphangiectasia	3 cases
Arachnodactyly - mental retardation - dysmorphism	3 cases
AREDYLD syndrome	3 cases
Axenfeld-Rieger anomaly - hydrocephaly - skeletal abnormalities	3 cases
Blepharoptosis - myopia - ectopia lentis	3 cases
Branchio-skeleto-genital syndrome	3 cases
Buttiens-Fryns syndrome	3 cases
Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia	3 cases
Cardiomyopathy-exercise intolerance due to muscle and heart glycogen deficiency	3 cases
Cataract - deafness - hypogonadism	3 cases
Cervical hypertrichosis - peripheral neuropathy	3 cases
Cortical blindness - mental retardation - polydactyly	3 cases
Craniofacial dysmorphism - coloboma - corpus callosum agenesis	3 cases
Craniofacial-deafness-hand syndrome	3 cases
Craniofrontonasal dysplasia - Poland anomaly	3 cases
Craniosynostosis - intracranial calcifications	3 cases
Cutaneous albinism, ermine phenotype	3 cases
Cutaneous photosensitivity - colitis, lethal	3 cases
Deafness - opticoacoustic nerve atrophy - dementia	3 cases
Deafness-mental retardation, Martin-Probst type	3 cases
Deafness-mental retardation, Martin-Probst type	3 cases
Ectodermal dysplasia, hypohidrotic - hypothyroidism - ciliary dyskinesia	3 cases
Ectodermic dysplasia hypothyroidism cleft	3 cases
Enterocyte heparan sulphate deficiency, congenital	3 cases
Eyebrow duplication - syndactyly	3 cases
Facial dysmorphism - macrocephaly - myopia - Dandy-Walker malformation	3 cases
Faciocardiomelic dysplasia, lethal	3 cases
Gardner-Morrison-Abbott syndrome	3 cases
Glaucoma - ectopia - microspherophakia - stiff joints - short stature	3 cases
Global developmental delay - osteopenia - ectodermal defect	3 cases
Goodman syndrome	3 cases
Haemolytic anaemia due to glutathione reductase deficiency	3 cases
Hair defect - photosensitivity - mental retardation	3 cases
Hirschsprung disease - nail hypoplasia - dysmorphism	3 cases

Diseases name	Number of published cases or families
Hypogonadism - retinitis pigmentosa	3 cases
Hypotonia with lactic acidaemia and hyperammonaemia	3 cases
Intractable diarrhoea - choanal atresia - eye anomalies	3 cases
Lewis-Pashayan syndrome	3 cases
Lipodystrophy - mental retardation - deafness	3 cases
Lumbosacral vertebrae, posterior fusion of - blepharoptosis	3 cases
Lymphoedema - atrial septal defects - facial changes	3 cases
Malabsorptive diarrhoea due to paucity of enteroendocrine cells, congenital	3 cases
Mental retardation - hypoplastic corpus callosum - preauricular tag	3 cases
Mental retardation, X-linked - hypogammaglobulinemia - progressive neurological deterioration	3 cases
Mental retardation, X-linked - precocious puberty - obesity	3 cases
Mental retardation, X-linked, Shrimpton type	3 cases
Mental retardation, X-linked, Wilson type	3 cases
Mental retardation, X-linked, Wittwer type	3 cases
Microcephaly - brachydactyly - kyphoscoliosis	3 cases
Microcephaly - cardiomyopathy	3 cases
Microcephaly - cleft palate	3 cases
Microcephaly - intellectual deficit - phalangeal and neurological anomalies	3 cases
Microcytic anaemia 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
Nanism due to growth hormone qualitative anomaly	3 cases
Neuroaxonal dystrophy - renal tubular acidosis	3 cases
Oculoosteocutaneous syndrome	3 cases
Omphalocele-cleft palate syndrome, lethal	3 cases
Oral-facial-digital syndrome, type 3	3 cases
Osteogenesis imperfecta, congenital - microcephaly - cataracts	3 cases
Osteoporosis-oculocutaneous-hypopigmentation syndrome	3 cases
Osteosclerosis - ichthyosis - premature ovarian failure	3 cases
Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	3 cases
Qazi-Markouizos syndrome	3 cases
Rambaud-Galian syndrome	3 cases

Diseases name	Number of published cases or families
Seizures - intellectual deficit due to hydroxylysinuria	3 cases
Sensorineural hearing loss, early greying, and essential tremor	3 cases
SERKAL syndrome	3 cases
Sex development disorder - intellectual deficit	3 cases
Short stature - mental retardation - eye anomalies - cleft/lip palate	3 cases
Split hand - urinary anomalies - spina bifida	3 cases
Summitt syndrome	3 cases
Thumb absent - short stature - immune deficiency	3 cases
Thymic-renal-anal-lung dysplasia	3 cases
Trigonocephaly - short stature - developmental delay	3 cases
Ulbright-Hodes syndrome	3 cases
Acrocraniofacial dysostosis	2 cases
Acrofacial dysostosis, autosomal recessive	2 cases
Agenesis of the corpus callosum - mental retardation - coloboma - micrognathia	2 cases
Alar cartilages hypoplasia - coloboma - telecanthus	2 cases
Alopecia - hypogonadism - extrapyramidal disorder	2 cases
Amaurosis - hypertrichosis	2 cases
Aniridia - renal agenesis - psychomotor retardation	2 cases
Aniridia-mental retardation syndrome	2 cases
Arthrogryposis - hyperkeratosis, lethal form	2 cases
Atherosclerosis- deafness - diabetes - epilepsy - nephropathy	2 cases
Aughton syndrome	2 cases
Auricular abnormalities - cleft lip with or without cleft palate - ocular abnormalities	2 cases
Auriculoocular anomalies - cleft lip	2 cases
Bangstad syndrome	2 cases
Beemer-Ertbruggen syndrome	2 cases
Blepharo-facio-skeletal syndrome	2 cases
Bouwes-Bavinck syndrome	2 cases
Brachytelephalangy - dysmorphism - Kallmann syndrome	2 cases
Braddock syndrome	2 cases
Brain dysgenesis due to glutamine synthetase deficiency, congenital	2 cases
Brain malformation - congenital heart disease - postaxial polydactyly	2 cases
Camptodactyly syndrome, Guadalajara type 2	2 cases
Cardiomyopathy - renal anomalies	2 cases
Cataract - microphthalmia - septal defect	2 cases
Cataract - nephropathy - encephalopathy	2 cases
Cataract- ataxia - deafness	2 cases

Diseases name	Number of published cases or families
CDG syndrome, type IIe	2 cases
CDG syndrome, type IIh	2 cases
CDG syndrome, type IL	2 cases
Chondrodysplasia - disorder of sex development	2 cases
Choroidal atrophy - alopecia	2 cases
Cleft lip - retinopathy	2 cases
Cleft lip/palate - mental retardation - corneal opacities	2 cases
Cleft palate - short stature - vertebral anomalies	2 cases
Cleft palate - stapes fixation - oligodontia	2 cases
Cleido rhizomelic syndrome	2 cases
Contractures - ectodermal dysplasia - cleft lip/palate	2 cases
Cooper-Jabs syndrome	2 cases
Corneal anesthesia - deafness - mental retardation	2 cases
Corneal-cerebellar syndrome	2 cases
Craniodiaphyseal dysplasia, autosomal dominant	2 cases
Craniosynostosis - fibular aplasia	2 cases
Craniosynostosis-radial aplasia, Imaizumi type	2 cases
Cryptomicrotia - brachydactyly - excess fingertip arch	2 cases
Cystic hygroma, lethal - cleft palate	2 cases
Dahlberg-Borer-Newcomer syndrome	2 cases
Dandy-Walker malformation - macrocephay	2 cases
Dandy-Walker malformation - polydactyly, postaxial	2 cases
Deaf blind hypopigmentation syndrome, Yemenite type	2 cases
Deafness - genital anomalies - metacarpal and metatarsal synostosis	2 cases
Deafness - vitiligo - achalasia	2 cases
Deafness-tubular acidosis-anaemia	2 cases
Dentinogenesis imperfecta - short stature - hearing loss - mental retardation	2 cases
Dermato-cardio-skeletal syndrome, Borrone type	2 cases
Dermatoleukodystrophy	2 cases
Desmosterolosis	2 cases
Developmental malformations - deafness - dystonia	2 cases
Diabetes, neonatal - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys	2 cases
Dincsoy-Salih-Patel syndrome	2 cases
Duane anomaly - myopathy - scoliosis	2 cases
Dysmorphism - short stature - deafness - pseudohermaphroditism	2 cases
Ectodermal dysplasia - anhidrotic, with immunodeficiency - osteopetrosis - lymphoedema	2 cases
Ectodermal dysplasia - blindness	2 cases

Diseases name	Number of published cases or families
Eng-Strom syndrome	2 cases
Epilepsy - microcephaly - skeletal dysplasia	2 cases
Epithelio-exfoliative colitis - deafness	2 cases
Fuqua-Berkovitz syndrome	2 cases
Gamma aminobutyric acid transaminase deficiency	2 cases
Generalised resistance to thyrotropin-releasing hormone	2 cases
Gonadal dysgenesis, XY type - associated anomalies	2 cases
Haemolytic anaemia, lethal - genital anomalies	2 cases
Heart defects - limb shortening	2 cases
HEC syndrome	2 cases
Hennekam-Beemer syndrome	2 cases
Hersh-Podruch-Weisskopf syndrome	2 cases
Hirschsprung disease - deafness - polydactyly	2 cases
Hydrocephaly - tall stature - joint laxity	2 cases
Hypercoagulability syndrome, due to glycosylphosphatidylinositol deficiency	2 cases
Hypomagnesemia with normocalciuria	2 cases
Hypospadias-hypertelorism-coloboma and deafness	2 cases
Hypotrichosis-mental retardation, Lopes type	2 cases
Ichthyosis - hepatosplenomegaly - cerebellar degeneration	2 cases
Ichthyosis - oral and digital anomalies	2 cases
Ichthyosis congenita - biliary atresia	2 cases
Ichthyosis, congenital - microcephalus - quadriplegia	2 cases
Inappropriate antidiuretic hormone secretion syndrome	2 cases
Intellectual deficit, severe - epilepsy - anal anomalies - distal phalangeal hypoplasia	2 cases
Iris dysplasia - hypertelorism - deafness	2 cases
Kaler-Garrity-Stern syndrome	2 cases
Kapur-Toriello syndrome	2 cases
Keratoderma - hypotrichosis - leukonychia	2 cases
Kniest-like dysplasia, lethal form	2 cases
Kozlowski-Brown-Hardwick syndrome	2 cases
Kudo-Tamura-Fuse syndrome	2 cases
Lichstenstein syndrome	2 cases
Lissencephaly, type III - metacarpal bone dysplasia	2 cases
Low birth weight - dwarfism - dysgammaglobulinemia	2 cases
Lung fibrosis - immunodeficiency - gonadal dysgenesis	2 cases
Macrocephaly - immune deficiency - anemia	2 cases
Macrocephaly - short stature - paraplegia	2 cases
Mental retardation, X-linked - acromegaly - hyperactivity	2 cases
Mental retardation, X-linked - epilepsy - progressive joint contractures - dysmorphism	2 cases
Mental retardation, X-linked - plagiocephaly	2 cases
Mental retardation, X-linked, Cantagrel type	2 cases
Mental retardation, X-linked, Reish type	2 cases
Mesomelic dysplasia - skin dimples	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 - mental retardation - heart disease	2 cases
Microlissencephaly - micromelia	2 cases
Mitochondrial encephalomyopathy aminoacidopathy	2 cases
Monosomy 9q22.3	2 cases
Multiple sclerosis - ichthyosis - factor VIII deficiency	2 cases
Nephronophtisis familial, adult form - spastic quadriparesia	2 cases
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	2 cases
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
Osteochondrodysplatic nanism - deafness - retinitis pigmentosa	2 cases
Osteogenesis imperfecta - retinopathy - seizures - intellectual deficit	2 cases
PARC syndrome	2 cases
Pierre Robin sequence - faciodigital 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
Ptosis - strabismus - rectus abdominis diastasis	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
Sex development disorder, 46,XX - skeletal anomalies	2 cases

Diseases name	Number of published cases or families
Short stature, Brussels type	2 cases
Siegler-Brewer-Carey syndrome	2 cases
Small vessel disease of the brain, not NOTCH3-related	2 cases
Spastic paraplegia - precocious puberty	2 cases
Spastic quadriplegia - retinitis pigmentosa - mental retardation	2 cases
Spinal muscular atrophy - Dandy-Walker complex - cataracts	2 cases
Stimmler syndrome	2 cases
Stoll-Alembik-Finck syndrome	2 cases
Syngnathia multiple anomalies	2 cases
Talo-patello-scaphoid osteolysis	2 cases
Teebi-Shaltout syndrome	2 cases
Thrombocytopenia - Robin sequence	2 cases
Thyrocerebrorenal syndrome	2 cases
Trichomegaly - cataract - spherocytosis, hereditary	2 cases
Trigonocephaly - bifid nose - acral anomalies	2 cases
Trigonocephaly - broad thumbs	2 cases
Tubular renal disease - cardiomyopathy	2 cases
Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay	2 cases
Weaver-Williams syndrome	2 cases
Xeroderma - talipes - enamel defects	2 cases
Zellweger-like syndrome, without peroxisomal anomalies	2 cases
Acrofacial dysostosis postaxial, atypical	1 case
Acrofacial dysostosis, Preis type	1 case
Blaichman syndrome	1 case
Bone dysplasia - corpus callosum agenesis	1 case
Bone dysplasia, Azouz type	1 case
Cataract - hypertrichosis - intellectual deficit	1 case
CDG syndrome, type Ii	1 case
CDG syndrome, type IIb	1 case
CDG syndrome, type IId	1 case
CDG syndrome, type Ij	1 case
Chondrodysplasia - situs inversus - imperforate anus - polydactyly	1 case
Corneal dystrophy - ichthyosis - microcephaly - mental retardation	1 case
Ectodermal dysplasia - arthrogryposis - diabetes mellitus	1 case
Hyaluronidase deficiency	1 case
Lipomatosis, pancreatic - duodenal stenosis	1 case
Lissencephaly - immunodeficiency	1 case
Nevus of ota - retinitis pigmentosa	1 case
Oral-facial-digital syndrome, type 10	1 case
Stoll-Alembik-Finck syndrome	1 case
Zadik-Barak-Levin syndrome	1 case

Diseases name	Number of published cases or families
Li-Fraumeni syndrome	400 families
Nance-Horan syndrome	50 families
Muscular dystrophy, limb girdle, autosomal recessive, type 2I	> 40 families
Granulomatous arthritis of childhood	40 families
Erythermalgia, primary	30 families
LCAT deficiency	30 families
Thiamine-responsive megaloblastic anaemia syndrome	30 families
Pericarditis - arthropathy - camptodactyly	< 30 families
Phosphoribosylpyrophosphate synthetase superactivity	< 30 families
Neuropathy, giant axonal	20 families
Overhydrated hereditary stomatocytosis	20 families
Synostoses, multiple	20 families
Acro-renal-ocular syndrome	< 20 families
Fahr syndrome	< 20 families
Familial platelet syndrome with predisposition to acute myelogenous leukaemia	< 20 families
Haemolytic anaemia, nonspherocytic, due to hexokinase deficiency	17 families
Ichthyosis prematurity syndrome	16 families
Myopathy, X-linked, with excessive autophagy	15 families
Familial platelet syndrome with predisposition to acute myelogenous leukaemia	13 families
Marie Unna congenital hypotrichosis	12 families
Aniridia - cerebellar ataxia - mental deficiency	> 10 families
Brachydactyly - arterial hypertension	> 10 families
Aortic aneurysm syndrome, Loeys-Dietz type	10 families
Renpenning syndrome	10 families
Acheiropodia	< 10 families
Angioma neurocutaneous, hereditary	< 10 families
Focal facial dermal dysplasia	< 10 families
Hypoparathyroidism, familial, isolated	< 10 families
Keratoderma palmoplantar - deafness	< 10 families
Keratosis palmoplantaris - esophageal carcinoma	< 10 families
Mental retardation, X-linked, syndromic, due to JARID1C mutation	< 10 families
Schinzel syndrome	< 10 families
Sebastian syndrome	< 10 families
VACTERL with hydrocephalus	< 10 families
Otodental syndrome	9 families
Ankyloblepharon - ectodermal defects - cleft lip palate	8 families
Cataract-microcornea syndrome	8 families
Osteosclerotic bone dysplasia, lethal	8 families
EEM syndrome	7 families
Immune dysregulation - polyendocrinopathy - enteropathy, X-linked	7 families
Macular degeneration, juvenile - hypotrichosis	7 families

Diseases name	Number of published cases or families
Cystoid macular dystrophy	6 families
Developmental dysphasia familial	6 families
Excessive growth - learning disabilities - facial dysmorphism	6 families
Pontocerebellar hypoplasia type 1	6 families
Encephalopathy with neuroserpin inclusion bodies, familial form	> 5 families
Anaemia, sideroblastic, X-linked - ataxia	5 families
Muscular dystrophy, limb-girdle, autosomal dominant, type 1D	5 families
Muscular dystrophy, limb-girdle, autosomal dominant, type 1E	5 families
Tetraamelia - pulmonary hypoplasia	5 families
Symphalangism, distal	< 5 families
Trichodental syndrome	< 5 families
Camptodactyly - taurinuria	4 families
IVIC syndrome	4 families
Myopathy, reducing body	4 families
Paroxysmal extreme pain disorder	4 families
Triphalangeal thumbs - brachyectrodactyly	4 families
Adducted thumbs - arthrogryposis, Christian type	3 families
Ataxia, cerebellar, autosomal recessive - blindness - deafness	3 families
CAMFAK syndrome	3 families
Cataract-glaucoma	3 families
Cerebroretinal vasculopathy	3 families
Craniorhiny	3 families
Deafness-infertility syndrome	3 families
Dystonia, mixed	3 families
HERNS syndrome	3 families
Hypomagnesemia with hypocalciuria	3 families
Mental retardation, X-linked, with isolated growth hormone deficiency	3 families
Rapid-onset dystonia-parkinsonism	3 families
Ankyloblepharon filiforme - imperforate anus	2 families
Ataxia, episodic, type 4	2 families
Auriculoosteodysplasia	2 families
Bakrania-Ragge syndrome	2 families
Bencze 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
Charcot-Marie-Tooth disease, type 2F, autosomal dominant	2 families
Chorioretinal atrophy, progressive bifocal	2 families
Earlobes, thickened - conductive deafness	2 families
Ehlers-Danlos syndrome, type 5	2 families
Gingival fibromatosis - progressive deafness	2 families
Growth deficiency - brachydactyly - dysmorphism	2 families
Hypoparathyroidism, X-linked	2 families
Jackson-Weiss syndrome	2 families
Mesoaxial synostotic syndactyly with phalangeal reduction	2 families
North Carolina macular dystrophy	2 families
Oral-facial-digital syndrome, type 8	2 families
Partington syndrome	2 families
Retinitis pigmentosa - intellectual deficit - deafness - hypogenitalism	2 families
Skeletal dysplasia - intellectual deficit	2 families
Spastic paraplegia - glaucoma - intellectual deficit	2 families
Albinism-deafness syndrome	1 family
Alopecia - congenita keratosis palmoplantaris	1 family
Aphalangy - syndactyly - microcephaly	1 family
Arthrogryposis-like hand anomaly - sensorineural deafness	1 family
Ataxia, cerebellar, autosomal recessive - saccadic intrusion	1 family
Ataxia, episodic, type 3	1 family
Banki syndrome	1 family
Brachydactyly - nystagmus - cerebellar ataxia	1 family
Brachydactyly, type A7	1 family
Camptobrachydactyly	1 family
Charcot-Marie-Tooth disease, type 2B2	1 family
Charcot-Marie-Tooth disease, type 2G, autosomal dominant	1 family
Charcot-Marie-Tooth disease, type 2K, autosomal dominant	1 family
Charcot-Marie-Tooth disease, type 2L, autosomal dominant	1 family
Coronary artery disease - hyperlipidemia - hypertension - diabetes - osteoporosis	1 family
Costocoracoid ligament, congenitally short	1 family
Craniofacial conodysplasia	1 family
Craniosynostosis, Philadelphia type	1 family
Cyprus facial-neuromusculoskeletal syndrome	1 family
Dyschondrosteosis - nephritis	1 family
Ehlers-Danlos syndrome, type 10	1 family
Fried syndrome	1 family
Hereditary vascular retinopathy	1 family
Hereditary vascular retinopathy	1 family
Hydrocephalus - blue sclerae - nephropathy	1 family
Insulin resistance, short fifth metacarpals	1 family
Kumar-Levick syndrome	1 family
Lipodystrophy, familial partial, due to AKT2 mutations	1 family

Diseases name	Number of published cases or families
Mental retardation - progressive spasticity, X-linked	1 family
Mental retardation, X-linked - Spastic paraplegia with iron deposits	1 family
Mental retardation, X-linked, Cabezas type	1 family
Mental retardation, X-linked, Pai type	1 family
Mental retardation, X-linked, recessive - macrocephaly - ciliary dysfunction	1 family
Microtia - eye coloboma - imperforation of the nasolacrimal duct	1 family
Muscular dystrophy, limb-girdle, autosomal dominant, type 1A	1 family
Muscular dystrophy, limb-girdle, autosomal dominant, type 1F	1 family
Muscular dystrophy, limb-girdle, autosomal dominant, type 1G	1 family
Neuropathy, sensory and autonomic, hereditary, with deafness and global delay	1 family
Neuropathy, sensory and autonomic, hereditary, with deafness, X-linked	1 family
Oculodental syndrome, Rutherfurd 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 stature - pituitary and cerebellar defects - small sella turcica	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


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