Prevalence of rare diseases : A bibliographic survey July 2007

Orphanet Report Series

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.

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.

For any questions or comments, please contact us: *orphanet@orpha.net*

Prevalences by alphabetical list of diseases

Disease name	Estimated prevalence (/100,000) or number of published cases or families
2,4-dienoyl-CoA reductase deficiency	1 case
2,8 dihydroxyadenine urolithiasis	1,7
3C syndrome	25 cases
3-hydroxy 3-methylglutaryl-CoA synthase	6 cases
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency	1
3M syndrome	40 cases
3-methylcrotonylglycinuria	2,25
3-methylglutaconic aciduria, type 1	20 cases
46 XX gonadal dysgenesis - epibulbar dermoid	1 case
46,XX disorders of sex development - skeletal anomalies	2 cases
46,XY disorders of sex development due to 17-beta-hydroxysteroid dehydrogenase deficiency	0,68
4-hydroxybutyricaciduria	350 cases
5-oxoprolinase deficiency	8 cases
6-pyruvoyl-tetrahydropterin synthase deficiency	248 cases
Aarskog-Scott syndrome	>200 cases
Aase-Smith syndrome	<10 cases
Ablepharon-macrostomia syndrome	15 cases
Abruzzo-Erickson syndrome	4 cases
Acanthamoeba keratitis	1
Acalvaria	<1**
Acatalasemia	3,1
Aceruloplasminemia	0,05
Achalasia - microcephaly	5 cases
Achalasia, primary	37,5
Acheiropodia	<10 families
Achondrogenesis	>100 cases
Achondroplasia	4,5
Achromatopsia	2,5
Ackerman syndrome	8 cases
Acquired generalized lipodystrophy	80 cases
Acrocallosal syndrome, Schinzel type	34 cases
Acrocephalosyndactyly	4,6
Acrocraniofacial dysostosis	2 cases
Acrodermatitis enteropathica, zinc deficiency type	0,2
Acrofacial dysostosis autosomal recessive	2 cases
Acro-facial dysostosis postaxial, atypical	1 case
Acro-facial dysostosis, Catania form	6 cases
Acrofacial dysostosis, Nager type	90 cases
Acro-facial dysostosis, Palagonia type	4 cases
Acro-facial dysostosis, Preis type	1 case
Acro-facial dysostosis, Rodriguez type	<10 cases
Acro-fronto-facio-nasal dysostosis	5 cases
Acromegaloid facial appearance syndrome	<20 cases
Acromegaloid facies - hypertrichosis	<20 cases
Acromegaly	5
Acromegaly - cutis verticis gyrata - corneal leukoma	16 cases
Acromelanosis	<10 cases
Acromesomelic dysplasia Hunter-Thompson type	10 cases
Acromesomelic dysplasia, Brahimi-Bacha type	3 cases
Acromesomelic dysplasia, Maroteaux type	50 cases
Acromicric dysplasia	<40 cases
Acroosteolysis, autosomal dominant	50 cases
Acro-pectoral syndrome	22 cases
Acropectororenal field defect	12 cases
Acropectorovertebral dysplasia	<30 cases
Acrorenal syndrome	20 cases
Acrorenalmandibular syndrome	7 cases
Acrorenoocular syndrome	<20 families
Acute interstitial pneumonia	3,8
Acute lymphoblastic leukemia	7,5
Acute non lymphoblastic leukemia	7
Acute promyelocytic leukemia	8
Acute Respiratory Distress Syndrome, Adult	30
Acyl-CoA dehydrogenase, medium chain, deficiency	15
Adactylia unilateral	34
Adamantinoma	513 cases
Adducted thumbs - arthrogryposis, Christian type	3 families
Adducted thumbs - arthrogryposis, Dundar type	5 cases
Adenosine monophosphate deaminase deficiency	200 cases
Adenylosuccinate lyase deficiency	50 cases
Adrenal hyperplasia, congenital	10
Adrenal hypoplasia congenital, X-linked	4
Adrenocortical carcinoma	1
Adrenoleukodystrophy, X-linked	3,5
Adult Onset Still's disease	1,23
ADULT syndrome	14 cases
Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis	3 cases
Agammaglobulinemia, X-linked	0,45
Agenesis of the corpus callosum - mental retardation - coloboma - micrognathia	2 cases
Aglossia - adactylia	<50 cases
Agnathia - holoprosencephaly - situs inversus	30 cases
Agonadism - dextrocardia - diaphragmatic hernia	6 cases
Aicardi syndrome	500 cases
Aicardi-Goutieres syndrome	30 cases

** Prevalence at birth

Alagille syndrome	1,4
Alar cartilages hypoplasia - coloboma - telecanthus	2 cases
Albers-Schonberg disease	1
Albinism ocular - late onset sensorineural deafness	7 cases
Albinism-deafness syndrome	1 family
Albright hereditary osteodystrophy	0,72
Albright-like syndrome	10 cases
Alexander disease	300 cases
Alkaptonuria	0,3
Alopecia - congenita keratosis palmoplantaris	1 family
Alopecia - contractures - dwarfism - mental retardation	5 cases
Alopecia - epilepsy - pyorrhea - mental subnormality	12 cases
Alopecia - hypogonadism - extrapyramidal disorder	2 cases
Alopecia totalis	10,5
Alpers syndrome	0,025
Alpha thalassemia-mental retardation, X-linked	168 cases
Alpha-1 antitrypsin deficiency	20
Alpha-mannosidosis	0,1
Alpha-sarcoglycanopathy	0,57
Alport syndrome	2
Alström syndrome	300 cases
Alveolar echinococcosis	<1000 cases
Alzheimer disease, familial	5,3
Amaurosis - hypertrichosis	2 cases
Amaurosis congenita of Leber	2,5
Ambras syndrome	10 cases
Amelia, autosomal recessive	3 cases
Aminopterin embryofetopathy	17 cases
Amniotic bands	4**
Amoebiasis due to free-living amoebae	1,75
Amyotrophic lateral sclerosis	6
Androgen insensitivity syndrome	13
Anemia, sideroblastic, X-linked - ataxia	5 families
Anencephaly	3,2**
Angelman syndrome	6,5
Angel-shaped phalango-epiphyseal dysplasia	15 cases
Angioma hereditary neurocutaneous	<10 families
Angioneurotic edema	1
Angio-osteohypertrophic syndrome	1000 cases
Aniridia	1,75
Aniridia - absent patella	3 cases
Aniridia - cerebellar ataxia - mental deficiency	>10 families
Aniridia - ptosis - mental retardation - obesity, familial type	3 cases
Aniridia - renal agenesis - psychomotor retardation	2 cases
Aniridia-mental retardation syndrome	2 cases
Anisakiasis	3,8
Ankyloblepharon - ectodermal defects - cleft lip palate	8 families
Ankyloblepharon filiforme - imperforate anus	2 families
Ankylosing vertebral hyperostosis - tylosis	8 cases
Anonychia - microcephaly	5 cases
Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges	11 cases
Anonychia with flexural pigmentation	3 cases
Anophtalmia/microphthalmia, isolated	14
Anophthalmia - hypothalamo-pituitary insufficiency	30 cases
Anophthalmia - megalocornea - cardiopathy - skeletal anomalies	3 cases
Anophthalmia - short stature - obesity	1 case
Anophthalmia plus syndrome	4 cases
Anophthalmia/microphthalmia - esophageal atresia	14 cases
Anorectal malformation	24
Antisynthetase syndrome	1,5
Antley-Bixler syndrome	34 cases
Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis	16 cases
Aortic aneurysm syndrome, Loeys-Dietz type	10 families
Aortic arch anomaly - peculiar facies - mental retardation	4 cases
Aortic dilatation- joint hypermobility- arterial tortuosity	22 cases
Aorto-ventricular tunnel	130 cases
Apert syndrome	1,25
Aphalangia - syndactyly - microcephaly	1 family
Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis	3 cases
Aplasia cutis - myopia	4 cases
Aplasia cutis congenita - intestinal lymphangiectasia	3 cases
Aplasia cutis congenita of limbs recessive	6 cases
Arachnodactyly - mental retardation - dysmorphism	3 cases
Arachnodactyly - ossification abnormal - mental retardation	5 cases
Aredyld syndrome	3 cases
Argininemia	31 cases
Arhinia - choanal atresia - microphthalmia	4 cases
Aromatase deficiency	13 cases
Arrhinia	20 cases
Arrhythmogenic right ventricular dysplasia	43,5
Arterial dissection - lentiginosis	4 cases
Arthritis-related enthesitis	5,7
Arthrogryposis - hyperkeratosis, lethal form	2 cases
Arthrogryposis - renal dysfunction - cholestasis	<150 cases
Arthrogryposis iugr thoracic dystrophy	1 case
Arthrogryposis multiplex congenita	30
Arthrogryposis multiplex congenita - whistling face	10 cases
Ascher syndrome	50 cases
Astley-Kendall dysplasia	<10 cases
Ataxia - apraxia - mental retardation, X-linked	9 cases
Ataxia - deafness - optic atrophy, lethal	12 cases
Ataxia telangiectasia	1

** Prevalence at birth

Ataxia, autosomal recessive, Beauce type	57 cases
Atelosteogenesis I	12 cases
Atelosteogenesis II	25 cases
Atelosteogenesis III	12 cases
Athabaskan brainstem dysgenesis syndrome	10 cases
Atherosclerosis- deafness - diabetes - epilepsy - nephropathy	2 cases
Atkin-Flaitz syndrome	14 cases
Atransferrinemia	9 cases
Atresia of small intestine	20
Atrial septal defect - atrioventricular conduction	11 cases
Atrial tachyarrhythmia with short PR interval	12 cases
Atrioventricular canal, partial	20
Atypical coarctation of aorta	0,17**
Aughton syndrome	2 cases
Auricular abnormalities - cleft lip with or without cleft palate - ocular abnormalities	2 cases
Auriculoocular anomalies - cleft lip	2 cases
Auro-cephalo-syndactyly	5 cases
Autism	45
Autoimmune lymphoproliferative syndrome	100 cases
Autoimmune polyendocrinopathy, type 1	4
Axenfeld-Rieger anomaly - hydrocephaly - skeletal abnormalities	3 cases
Babesiosis	40 cases
Bacterial toxic-shock syndrome	3
Bamforth syndrome	5 cases
Bangstad syndrome	2 cases
Banki syndrome	1 family
Barber-Say syndrome	10 cases
Bardet-Biedl syndrome	0,8
Bartsocas-Papas syndrome	24 cases
Bartter syndrome	0,12
B-cell chronic lymphocytic leukemia	32
Beckwith-Wiedemann syndrome	7,3
Beemer-Ertbruggen syndrome	2 cases
Behcet disease	2,5
Bencze syndrome	2 families
Benign exophthalmos syndrome	4 cases
Benign paroxysmal torticolis of infancy	50 cases
Bernard-Soulier syndrome	100 cases
Best disease	4,4
Beta-mannosidosis	14 cases
Beta-sarcoglycanopathy	0,57
Beta-ureidopropionase deficiency	5 cases
Bethlem myopathy	100 cases
Bickel-Fanconi glycogenosis	112 cases
Biliary atresia	5,6
Biliary malformation - renal tubular insufficiency	6 cases
Birt-Hogg-Dube syndrome	>60 families
Björnstadt syndrome	33 cases

Blackfan-Diamond disease	0,32
Blaichman syndrome	1 case
Blepharo-cheilo-dontic syndrome	<50 cases
Blepharo-facio-skeletal syndrome	2 cases
Blepharonasofacial malformation syndrome	2 families
Blepharophimosis - ptosis - esotropia - syndactyly - short stature	6 cases
Blepharoptosis - myopia - ectopia lentis	3 cases
Bloom syndrome	>100 cases
Blue cone monochromatism	1
Blue rubber bleb nevus	>200 cases
Bone dysplasia - corpus callosum agenesis	1 case
Bone dysplasia lethal, Holmgren type	4 cases
Bone dysplasia, Azouz type	1 case
Bone fragility - craniosynostosis - proptosis hydrocephalus	4 cases
Bone sclerosing - dysplasia - ichthyosis - premature ovarian failure	3 cases
Bone tumor	10
Bonnemann-Meinecke-Reich syndrome	4 cases
Book syndrome	25 cases
Boomerang dysplasia	10 cases
BOR syndrome	2,5
Bosley-Salih-Alorainy syndrome	9 cases
Botulism	0,05
Boutonneuse fever	17
Bouwes-Bavinck syndrome	2 cases
Bowen-Conradi syndrome	44 cases
Brachio-skeleto-genital syndrome	3 cases
Brachydactyly - arterial hypertension	>10 families
Brachydactyly, long thumb type	4 cases
Brachymorphism - onychodysplasia - dysphalangism	9 cases
Brachytelephalangy - dysmorphism - Kallmann syndrome	2 cases
Braddock syndrome	2 cases
Bradyopsia	5 cases
Brain malformation - congenital heart disease - postaxial polydactyly	2 cases
Branchial arch syndrome, X-linked	5 à 7 cases
Branchiogenic deafness syndrome	5 cases
Branchio-oculo-facial syndrome	<50 cases
Breast cancer, familial	17
Bronchopneumopathy, chronic, due to TAP deficiency	<20 cases
Bronchopulmonary dysplasia	13
Budd-Chiari syndrome	1,5
Buerger's disease	12,5
Bullous dystrophy macular type	2 families
Bullous ichthyosiform - erythroderma congenita	0,4
Bullous pemphigoid	2,5
Bullous systemic lupus erythematosus	70 cases
Buttiens-Fryns syndrome	3 cases

** Prevalence at birth

CADASIL syndrome	500 cases
Calpainopathy	3,8
Calvarial doughnut lesions - bone fragility	20 cases
CAMFAK syndrome	3 families
CAMOS syndrome	5 cases
Campomelia Cumming type	8 cases
Campomelic dysplasia	0,35
Camptobrachydactyly	1 family
Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia	3 cases
Camptodactyly - tall stature - scoliosis - hearing loss	30 cases
Camptodactyly - taurinuria	4 families
Camptodactyly syndrome, Guadalajara type 1	8 cases
Camptodactyly syndrome, Guadalajara type 2	2 cases
Camurati-Engelmann disease	200 cases
Cantrell pentalogy	0,55**
Capillary leak syndrome	57 cases
Carbamoylphosphate synthetase deficiency	0,7
Carcinoma of the gallbladder	6,5
Cardiac conduction disease, dilated cardiomyopathy and brachydactyly	10 cases
Cardiogenital syndrome	7 cases
Cardiomyopathy - cataract - hip spine disease	9 cases
Cardiomyopathy - renal anomalies	2 cases
Cardiomyopathy, familial dilated	17,5
Carey-Fineman-Ziter syndrome	<20 cases
Carney complex	160 cases
Carnitine palmitoyl transferase 1 deficiency	35 cases
Carnitine palmitoyl transferase 2 deficiency	>100 cases
Carnitine-acylcarnitine translocase deficiency	30 cases
Carnosinemia	30 cases
Caroli's disease	<250 cases
Carpenter syndrome	40 cases
Carpotarsal osteochondromatosis	<10 cases
Carpotarsalosteolysis, autosomal recessive	<10 cases
Castleman disease	400 cases
Cataract - ataxia - deafness	2 cases
Cataract - cardiomyopathy	30 cases
Cataract - deafness - hypogonadism	3 cases
Cataract - hypertrichosis - mental retardation	1 case
Cataract - mental retardation - hypogonadism	10 cases
Cataract - microphthalmia - septal defect	2 cases
Cataract - nephropathy - encephalopathy	2 cases
Cataract anterior polar	4,4
Cataract congenital, Volkmann type	<100 cases
Cataract, Hutterite type	1 family
Cataract total, congenital	7,9
Cataract-glaucoma	3 families
Cataract-microcornea syndrome	8 families
Catel-Manzke syndrome	27 cases
Cat-eye syndrome	1,35
Cat-scratch disease	6,6
Caudal dysgenesis familial type	4 cases
CDG syndrome	1,5**
CDG syndrome type Ia	> 300 cases
CDG syndrome type Ib	20 cases
CDG syndrome type Ic	>30 cases
CDG syndrome type Id	5 cases
CDG syndrome type Ie	7 cases
CDG syndrome type If	4 cases
CDG syndrome type Ig	6 cases
CDG syndrome type Ih	5 cases
CDG syndrome type Ii	1 case
CDG syndrome type IIa	4 cases
CDG syndrome type IIb	1 case
CDG syndrome type IId	1 case
CDG syndrome type IIe	2 cases
CDG syndrome type Ij	1 case
CDG syndrome type Ik	4 cases
CDG syndrome type IL	2 cases
CEDNIK syndrome	7 cases
Celiac disease - epilepsy - occipital calcifications	170 cases
Central neurocytoma	>100 cases
Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss	1-2 families
Cerebellar ataxia, autosomal dominant	2,15
Cerebellar ataxia, autosomal recessive	7
Cerebral arteriovenous fistula	6
Cerebral gigantism - jaw cysts	<10 cases
Cerebro-costo-mandibular syndrome	60 cases
Cerebro-oculo-nasal syndrome	10 cases
Cerebroretinal vasculopathy	3 families
Ceroid lipofuscinosis, neuronal	4
Cervical hypertrichosis - peripheral neuropathy	3 cases
CHAND syndrome	>10 cases
Chaotic atrial tachycardia	100 cases
Char syndrome	10 cases
Charcot-Marie-Tooth disease (generic term)	32,5
Charcot-Marie-Tooth disease, X-linked	1,6
CHARGE association	0,14
Chediak-Higashi syndrome	>10 cases
CHILD syndrome	30 cases
Choanal atresia - deafness - cardiac defects dysmorphism	5 cases
Cholangiocarcinoma	10
Cholestasis - lymphoedema, syndrome	20-50 cases
Cholestasis - pigmentary retinopathy - cleft palate	4 cases
Cholesteryl ester storage disease	<50 cases
Chondrodysplasia - disorder of sex development	2 cases
Chondrodysplasia - situs inversus- imperforate anus - polydactyly	1 case

** Prevalence at birth

Chondrodysplasia lethal, recessive	4 cases
Chondrodysplasia punctata, rhizomelic type	1
Chondrodysplasia, Blomstrand type	13 cases
Chordoma	0,05
Chorioretinal atrophy, progressive bifocal	2 families
Choroidal atrophy - alopecia	2 cases
Choroidal dystrophy, central areolar	3,33
Choroideremia	2
Choroideremia - deafness - obesity	4 cases
Choroidocerebral calcification syndrome, infantile form	10 cases
Christ-Siemens-Touraine syndrome	0,35
Chromosome Y deletion	42
Chronic hiccup	1
Chronic inflammatory demyelinating polyneuropathy	4,4
Chronic recurrent multifocal osteomyelitis, juvenile	>260 cases
Churg-Strauss syndrome	1
Chylomicron retention disease	40 cases
CINCA syndrome	100 cases
Cleft lip - retinopathy	2 cases
Cleft lip palate - malrotation - cardiopathy	4 cases
Cleft lip palate - mental retardation - corneal opacities	2 cases
Cleft palate - cardiac defect - genital anomalies - ectrodactyly	5 cases
Cleft palate - short stature - vertebral anomalies	2 cases
Cleft palate - stapes fixation - oligodontia	2 cases
Cleft palate-lateral synechia syndrome	7 cases
Cleido-rhizomelic syndrome	2 cases
Clouston syndrome	1
Cloverleaf skull syndrome	150 cases
COACH syndrome	8 cases
Coats disease	2
Cobb syndrome	35 cases
Cockayne syndrome	200 cases
Coffin-Lowry syndrome	0,55
Coffin-Siris syndrome	40 cases
Cogan syndrome	200 cases
Cohen syndrome	100 cases
Collagenous colitis	10,5
Coloboma of macula - brachydactyly type B	12 cases
Coloboma uveal - cleft lip palate - mental retardation	12 cases
Coloboma, ocular	1
Colobomatous - microphthalmia - heart disease - hearing loss	10 cases
Cone rod dystrophy	2,5
Cone rod dystrophy - amelogenesis imperfecta	29 cases
Congenital alopecia, X linked	1 family
Congenital anosmia, isolated	<15 cases
Congenital brain dysgenesis due to glutamine synthetase deficiency	<30 cases
Congenital bronchobiliary fistula	23 cases
Congenital cataracts - facial dysmorphism - neuropathy	100 cases
Congenital ichthyosis - microcephalus - quadriplegia	2 cases
Congenital indifference to pain	> 20 cases
Congenital lobar emphysema	4,5
Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	3 cases
Congenital myasthenic syndromes	0,75
Congenital rubella syndrome	1**
Contractures - ectodermal dysplasia - cleft lip palate	2 cases
Cooper-Jabs syndrome	2 cases
Corneal anesthesia - deafness - mental retardation	2 cases
Corneal crystals - myopathy - neuropathy	1 case
Corneal crystals myopathy neuropathy	1 case
Corneal dystrophy - ichthyosis - microcephaly - mental retardation	1 case
Corneal dystrophy - perceptive deafness	<10 cases
Corneal-cerebellar syndrome	2 cases
Cornelia de Lange syndrome	5,25
Coronary artery disease - hyperlipidemia - hypertension - diabetes - osteoporosis	1 family
Corpus callosum agenesis - neuronopathy	19
Corpus callosum dysgenesis, X-linked recessive	11 cases
Corpus callosum, agenesis - cataract - immunodeficiency	8 cases
Cortical blindness - mental retardation - polydactyly	3 cases
Corticobasal degeneration	4
Corticosteroid-sensitive aseptic abscesses	49 cases
Costello syndrome	200 cases
Costocoracoid ligament congenitally short	1 family
Cowden syndrome	0,45
Coxo-auricular syndrome	4 cases
Coxo-podo-patellar syndrome	47 cases
Craniodiaphyseal dysplasia	>20 cases
Cranio-digital syndrome - mental retardation	4 cases
Cranio-ectodermal dysplasia	15 cases
Craniofacial conodysplasia	1 family
Cranio-facial dysmorphism - coloboma - corpus callosum agenesis	3 cases
Craniofacial dyssynostosis	0,05
Craniofacial-deafness-hand syndrome	3 cases
Craniofaciocardioskeletal syndrome	4 cases
Craniofrontonasal dysplasia - poland anomaly	3 cases
Craniofrontonasal syndrome, Teebi type	20 cases
Craniolenticulosutural dysplasia	21 cases
Craniometaphyseal dysplasia	70 cases
Cranioosteoarthropathy	4 cases
Craniosynostosis - brachydactyly	5 cases
Craniosynostosis - Dandy Walker hydrocephalus	4 cases

** Prevalence at birth

Craniosynostosis - fibular aplasia	2 cases
Craniosynostosis - intracranial calcifications	3 cases
Craniosynostosis radial aplasia, Imaizumi type	2 cases
Craniosynostosis, Boston type	19 cases
Craniosynostosis, Philadelphia type	1 family
Craniotubular syndrome	2 cases
Creutzfeldt-Jakob disease	0,1
Crigler-Najjar syndrome	200 cases
Crisponi syndrome	18 cases
Criss-cross heart	0,8
Crouzon disease	2
Cryoglobulinemia	1,33*
Cryptomicrotia - brachydactyly - excess fingertip arch	2 cases
Cryptosporidiosis	34
Curry-Jones syndrome	5 cases
Cutaneous albinism, ermine phenotype	3 cases
Cutaneous lymphoma	8,3
Cutaneous mastocytosis	0,75
Cutaneous neuroendocrine carcinoma	0,5
Cutaneous photosensitivity - colitis lethal	3 cases
Cutis gyrata - acanthosis nigricans - craniosynostosis	6 cases
Cutis laxa	>100 cases
Cutis marmorata telangiectatica congenita	300 cases
Cutis verticis gyrata - mental deficiency	1,02
Cutis verticis gyrata - thyroid aplasia - mental retardation	5 cases
Cyprus facial neuromusculosqueletal syndrome	1 family
Cystathioninuria	7
Cystic fibrosis	12
Cystic hamartoma of lung and kidney	<5 cases
Cystic hygroma lethal - cleft palate	2 cases
Cystinosis	0,5
Cystinuria	14
Cystoid macular dystrophy	6 families
Dacryocystitis osteopoikilosis	5 cases
Dahlberg-Borer-Newcomer syndrome	2 cases
Dandy Walker - macrocephaly	2 cases
Dandy Walker malformation - postaxial polydactyly	2 cases
Darier disease	1,5
Deaf - blind - hypopigmentation	2 cases
Deafness - enamel hypoplasia - nail defects	6 cases
Deafness - lymphoedema - leukemia	<10 cases
Deafness - opticoacoustic nerve atrophy - dementia	3 cases
Deafness - peripheral neuropathy - arterial disease	4 cases
Deafness - skeletal dysplasia - lip granuloma	8 cases
Deafness - tubular acidosis - anemia	2 cases
Deafness - vitiligo - achalasia	2 cases
Deafness, autosomal dominant, nonsyndromic, sensorineural, type DFNA	8
Deafness, autosomal recessive, nonsyndromic, sensorineural, type DFNB	44
Deafness, X-linked, non syndromic, sensorineural, type DFN	0,6
Deafness-infertility syndrome	3 families
Deafness-mental retardation, Martin-Probst type	3 cases
Deafness-mental retardation, Martin-Probst type	3 cases
Defective expression of HLA class 2	100 cases
Dehydratase deficiency	21 cases
Deletion 18p	<200 cases
Deletion 2q24	23 cases
Delta-sarcoglycanopathy	0,57
DEND syndrome	14 cases
Dentinogenesis imperfecta - short stature - hearing loss - mental retardation	2 cases
Dentatorubral pallidoluysian atrophy	<0,3
Denys-Drash syndrome	150 cases
Depigmentation of the iris, acute, bilateral	5 cases
Dermatitis herpetiformis	20,2
Dermato-cardio-skeletal syndrome, Borrone type	2 cases
Dermatofibrosarcoma protuberans	10
Dermatoleukodystrophy	2 cases
Dermatomyositis	14,8
Dermatoosteolysis, Kirghizian type	5 cases
Dermoodontodysplasia	14 cases
Dermopathy restrictive lethal	30 cases
Desbuquois syndrome	>40 cases
Desmosterolosis	2 cases
Developmental delay due to 2-methylbutyryl-coA dehydrogenase deficiency	<30 cases
Developmental dysphasia familial	6 families
Developmental malformations - deafness - dystonia	2 cases
Diabetes insipidus, nephrogenic	0,5
Diabetes mellitus, neonatal	0,2
Diabetes, neonatal - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys	2 cases
Diaphanospondylodysostosis	<10 cases
Diaphragmatic defect - limb deficiency - skull defect	4 cases
Diaphragmatic hernia - exomphalos - corpus callosum agenesis	13 cases
Diaphragmatic hernia, congenital	15
Diastrophic dwarfism	3,5
Diffuse leiomyomatosis with Alport syndrome	0,1
Diffuse neonatal haemangiomatosis	<70 cases
Diffuse palmoplantar keratoderma - acrocyanosis	10 cases
Diffuse palmoplantar keratoderma, Norrbotten dominant type	2,5
Digito-reno-cerebral syndrome	<10 cases
Digitotalar dysmorphism	6
Dihydropteridine reductase deficiency	134 cases
Dihydropyrimidinuria	7 cases

** Prevalence at birth

Dincsoy-Salih-Patel syndrome	2 cases
Disorder of sex development - mental retardation	3 cases
Distal monosomy 5q	10 cases
Distal monosomy 8p	20 cases
Distal myopathy with vocal cord weakness	12 cases
Distal myopathy, Nonaka type	0,1
Distal myopathy, Welander type	10
Distal myopathy, with early respiratory muscle involvement	24 cases
Distal myopathy, with posterior leg and anterior upper limb involvement	12 cases
DOOR syndrome	<50 cases
Dopamine beta-hydroxylase, deficiency of	12 cases
Dopa-responsive dystonia	0,3
Double outlet left ventricle	32 cases
Double uterus - hemivagina - renal agenesis	<60 cases
Duane anomaly - myopathy - scoliosis	2 cases
Duane syndrome	10
Dubowitz syndrome	150 cases
Duker-Weiss-Siber syndrome	4 cases
Duodenal atresia	8,55
Duplication 8q	>30 cases
Dyggve-Melchior-Clausen disease	60 cases
Dyschondrosteosis - nephritis	1 family
Dyserythropoietic anemia, congenital	1
Dyskeratosis congenita	0,1
Dysmorphism - short stature - deafness - pseudohermaphroditism	2 cases
Earlobes, thickened - conductive deafness	2 families
Early infantile epileptic encephalopathy	88 cases
Early myoclonic encephalopathy	30 cases
Early onset torsion dystonia	0,4
Ear-patella-short stature syndrome	42 cases
Ebstein anomaly	0,75
Ectodermal dysplasia - alopecia - preaxial polydactyly	1 case
Ectodermal dysplasia - arthrogryposis - diabetes mellitus	1 case
Ectodermal dysplasia - blindness	2 cases
Ectodermal dysplasia - mental retardation - syndactyly	1 case
Ectodermal dysplasia - absent dermatoglyphics	<30 cases
Ectodermal dysplasia anhidrotic - immunodeficiency - osteopetrosis - lymphedema	2 cases
Ectodermal dysplasia, «pure» hair-nail type	<20 cases
Ectodermal dysplasia, Berlin type	4 cases
Ectodermal dysplasia, hypohidrotic - hypothyroidism - ciliary dyskinesia	3 cases
Ectodermal dysplasia, hypohidrotic, autosomal dominant	40 cases
Ectodermal dysplasia-skin fragility syndrome	10 cases
Ectodermic dysplasia - hypothyroidism - cleft	3 cases
Ectopia lentis - chorioretinal dystrophy - myopia	4 cases
Ectopia lentis isolated	6,4
Ectrodactyly - ectodermal dysplasia	5 cases
EEM syndrome	7 families
Ehlers-Danlos syndrome type 10	1 family
Ehlers-Danlos syndrome, classic type	3,5
Ehlers-Danlos syndrome, type 3	12,5
Ehlers-Danlos syndrome, type 4	1
Ehlers-Danlos syndrome, type 5	2 families
Ehlers-Danlos syndrome, type 7C	7 cases
Ehrlichiosis	<50 cases
Eiken syndrome	6 cases
Elejalde syndrome	30 cases
Elliptocytosis, hereditary	37,5
Ellis Van Creveld syndrome	150 cases
Emery-Dreifuss muscular dystrophy	1,5
Enamel hypoplasia cataract hydrocephaly	1 case
Encephalo-cranio-cutaneous lipomatosis	45 cases
Encephalopathy due to hydroxykynureninuria	<30 cases
Encephalopathy with neuroserpin inclusion bodies, familial form	>5 families
Enchondromatosis	> 600 cases
Eng-Strom syndrome	2 cases
Eosinophilic fasciitis	200 cases
Eosinophilic gastroenteritis	280 cases
Eosinophilic pneumonia, acute idiopathic	>100 cases
Epidermal nevus syndrome	>400 cases
Epidermolysis bullosa simplex - limb girdle muscular dystrophy	<20 cases
Epidermolysis bullosa, acquired	100 cases
Epidermolysis bullosa, dystrophic	0,27
Epidermolysis bullosa, epidermolytic	2,5
Epidermolysis bullosa, junctional	0,06
Epilepsy - microcephaly - skeletal dysplasia	2 cases
Epilepsy - telangiectasia	6 cases
Epilepsy, pyridoxin-dependent	0,15
Epiphyseal dysplasia multiple	5
Episodic ataxia, type 3	1 family
Episodic ataxia, type 4	2 families
Erdheim-Chester disease	178 cases
Erythermalgia, primary	30 families
Erythroderma lethal, congenital	17 cases
Erythrokeratodermia - ataxia	25 cases
Erythrokeratodermia variabilis, Mendes da Costa type	>200 cases
Esophageal carcinoma	8
Esthesioneuroblastoma	<1000 cases
Evans syndrome	0,1
Ewing sarcoma	0,1
Exostoses, multiple	4
Eyebrow duplication syndactyly	3 cases
Fabry disease	1,75

** Prevalence at birth

Facial clefting - corpus callosum - agenesis	1 case
Facial dysmorphism - macrocephaly - myopia - Dandy Walker	3 cases
Facial onset sensory - motor neuronopathy	4 cases
Faciocardiomelic dysplasia, lethal	3 cases
Facio-scapulo-humeral muscular dystrophy	7
Factor II deficiency	10
Factor V deficiency	0,1
Factor VII deficiency	0,25
Factor XIII deficiency, congenital	0,04
Fahr syndrome	<20 families
Familial adenomatous polyposis, autosomal dominant	5,25
Familial cold urticaria	0,1
Familial dysautonomia	550 cases
Familial hematuria, autosomal dominant - retinal arteriolar tortuosity - contractures	8 cases
Familial platelet syndrome with predisposition to acute myelogenous leukemia	13 families
Familial rectal pain	4 families
Familial venous malformations	40
Fanconi - ichthyosis - dysmorphism	6 cases
Fanconi anaemia	1
Femur-fibula-ulna complex	1,5
Fetal cytomegalovirus syndrome	40
Fetal methyl-mercury syndrome	800 cases
Fetal varicella syndrome	>100 cases
Fibrinogen deficiency, congenital	0,15
Fibrochondrogenesis	11 cases
Fibrodysplasia ossificans progressiva	0,08
Fibromatosis, gingival - progressive deafness	2 families
Fibular aplasia - ectrodactyly	<50 cases
Fibular dimelia - diplopodia	11 cases
Fine-Lubinsky syndrome	5 cases
Fingerprints absence - syndactyly milia	14 cases
Flynn-Aird syndrome	10 cases
Focal dermal hypoplasia	200-300 cases
Focal dystonia	11,7
Focal facial dermal dysplasia	<10 families
Focal myositis	50 cases
Folate malabsorption, hereditary	17 cases
Follicular lymphoma	36
Foveal hypoplasia presenile cataract	11 cases
Fragile X syndrome	14,25
Fraser syndrome	150 cases
Freeman-Sheldon syndrome	100 cases
Fried Syndrome	1 family
Friedreich ataxia	2,5
Fronto-metaphyseal dysplasia	<30 cases
Frontotemporal dementia	3
Fronto-temporal dementia and Parkinsonism linked to chromosome 17 (FTDP-17)	50 cases

Fructose intolerance	5
Fructose-1,6-bisphosphatase deficiency	2,5
Fryns syndrome	7**
Fucosidosis	100 cases
Fuhrmann syndrome	11 cases
Fumaric aciduria	>20 cases
Fuqua-Berkovitz syndrome	2 cases
Galactosemia	6,6
Galloway-Mowat syndrome	40 cases
Gamma aminobutyric acid transaminase deficiency	2 cases
Gamma-glutamyl transpeptidase deficiency	7 cases
Gamma-glutamylcysteine synthetase deficiency	9 cases
Gamma-sarcoglycanopathy	1,96
GAPO syndrome	27 cases
Gardner-Morrison-Abbot syndrome	3 cases
Gastric cancer	20
Gastrointestinal stromal tumor	1,8
Gastroschisis	12
Gaucher - ichthyosis - restrictive dermopathy	4 cases
Gaucher disease	1
Gaucher disease, type 1	0,94
Gaucher disease, type 2	0,01
Gaucher disease, type 3	0,05
Geleophysic dwarfism	27 cases
Gelineau disease	49
Gemss syndrome	3 cases
Genitopatellar syndrome	7 cases
German syndrome	5 cases
Geroderma osteodysplastica	30 cases
Giant cell arteritis	8,9
Giant pigmented hairy nevus	2
Glaucoma-sleep apnea	5 cases
Glioblastoma	11
Global developmental delay - osteopenia - ectodermal defect	3 cases
Glomerulonephritis - sparse hair - telangiectases	<10 cases
Glossopalatine ankylosis - cataracts - digital anomalies	1 case
Glucocorticoid deficiency, familial	50 cases
Glucose-galactose malabsorption	200 cases
Glucosephosphate isomerase deficiency	50 cases
Glutaryl-CoA dehydrogenase deficiency	0,4
Glutathione synthetase deficiency	65 cases
Glycogen storage disease due to LAMP-2 deficiency	30 cases
Glycogen storage disease, type 2	1,1
Glycogen storage disease, type 4	0,6
Glycogen storage disease, type 7	<30 cases
Glycogen storage, type 0	16 cases
Goldberg-Shprintzen megacolon syndrome	10 cases
Goldenhar syndrome	3,5
Goldmann-Favre syndrome	<50 cases

** Prevalence at birth

Gombo syndrome	4 cases
Gonadal dysgenesis, XX type	12
Gonadal dysgenesis, XY type - associated anomalies	2 cases
Goodman syndrome	3 cases
Goodpasture syndrome	0,64
Gorham-Stout disease	200 cases
Gorlin syndrome	1
Gorlin-Chaudhry-Moss, syndrome	4 cases
GRACILE syndrome	2,12**
Graft versus host disease	3,4
Grange syndrome	6 cases
Granulomatous arthritis of childhood	40 families
Granulomatous disease, chronic	0,2
Granulomatous slack skin	<50 cases
Gräsbeck-Imerslund disease	300 cases
Gray platelet syndrome	20 cases
Great vessels transposition (TGV)	32,5
Greenberg dysplasia	<10 cases
Greig syndrome	100 cases
Griscelli disease	60 cases
Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate	4 cases
Growth retardation - microcephaly - digital abnormalities - hypospadias	4 cases
GTP cyclohydrolase I deficiency	17 cases
Guanidinoacetate methyltransferase deficiency	9 cases
Guillain-Barré syndrome	47,5
Haemolytic anaemia due to glutathione reductase deficiency	3 cases
Haemolytic anaemia, nonspherocytic, due to hexokinase deficiency	17 families
Hair defect - photosensitivity - mental retardation	3 cases
Hallermann-Streiff-Francois syndrome	<100 cases
Harding ataxia	1
Hartnup syndrome	4
Heart defects - limb shortening	2 cases
Hec syndrome	2 cases
Helicoid peripapillary chorioretinal degeneration	100 cases
Hemimelia	4,15
Hemiplegic migraine, familial	6,5
Hemolytic anemia due to adenylate kinase deficiency	12 cases
Hemolytic anemia, lethal - genital anomalies	2 cases
Hemophilia	7,7
Hemophilia, acquired	0,1
Hemorrhagic disorders due to collagen receptors deficiency	<20 cases
Hennekam syndrome	>50 cases
Hennekam-Beemer syndrome	2 cases
Hepatic veno-occlusive disease	11
Hepatic veno-occlusive disease - immunodeficiency	<25 cases
Hepatitis, chronic autoimmune	0,65
Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia	19 cases
Hereditary sensory and autonomic neuropathy, type 2	35 cases
Hereditary vascular retinopathy	1 family
Hereditary vascular retinopathy	1 family
HERNS syndrome	3 families
Herpes simplex encephalitis	0,021*
Hersh-Podruch-Weisskopf syndrome	2 cases
Heterotaxia	2,5
Hidrotic ectodermal dysplasia, Christianson-Fourie type	6 cases
Hidrotic ectodermal dysplasia, Halal type	4 cases
Hirschsprung disease	20
Hirschsprung disease - deafness - polydactyly	2 cases
Hirschsprung disease - nail hypoplasia - dysmorphism	3 cases
Histidinemia	4
Hodgkin lymphoma	10,5
Holoprosencephaly	7
Holt-Oram syndrome	1
Homocarnosinosis	4 cases
Homocystinuria due to cystathionine beta-synthase deficiency	0,4
Huntington disease	6,2
Hyaluronidase deficiency	1 case
Hydrocephalus - blue sclerae - nephropathy	1 family
Hydrocephalus - costovertebral dysplasia - Sprengel anomaly	8 cases
Hydrocephaly - tall stature - joint laxity	2 cases
Hydrolethalus	5**
Hydrops ectrodactyly syndactyly	1 case
Hypercoagulability syndrome, due to glycosylphosphatidylinositol deficiency	2 cases
Hyperferritinemia, hereditary, with congenital cataracts	>64 cases
Hyperglycinemia, isolated nonketotic	0,2
Hyper-IGM syndrome, autosomal recessive	0,05**
Hyperimmunoglobinemia D - recurrent fever	180 cases
Hyperkalemic periodic paralysis	0,75
Hyperkeratosis - hyperpigmentation syndrome	10 cases
Hyperlipidemia type 3	7,8
Hyperlipoproteinemia type 1	0,6
Hyperoxaluria	0,2
Hypertrichosis - brachydactyly - obesity - mental retardation	1 case
Hypertrichosis cubiti - short stature	28 cases
Hypertrichosis lanuginosa congenita	<100 cases
Hypertrichosis lanuginosa, acquired	60 cases
Hypertrichotic osteochondrodysplasia	18 cases
Hypochondroplasia	3,3
Hypocomplementaemic leucocytoclasic vasculitis	<100 cases
Hypogammaglobulinemia due to CD19 deficiency	4 cases

** Prevalence at birth

Hypogonadism - retinitis pigmentosa	3 cases
Hypokalemic periodic paralysis	1
Hypokeratosis, palmo-plantar, circumscribed	17 cases
Hypomagnesemia with hypocalciuria	3 families
Hypomagnesemia with normocalciuria	2 cases
Hypomandibular facio-cranial dysostosis	4 cases
Hypomyelination - congenital cataract	8 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodonti	4 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 cases
Hypoparathyroidism - deafness - renal disease	12 cases
Hypoparathyroidism familial isolated	<10 families
Hypoparathyroidism X-linked	2 families
Hypopituitarism - microphthalmia	<10 cases
Hypopituitarism - postaxial polydactyly	6 cases
Hypotelorism - cleft palate - hypospadias	13 cases
Hypothyroidism - dermoid cyst - cleft palate	1 case
Hypothyroidism, congenital	29
Hypotrichosis - lymphedema - telangiectasia	4 cases
Hypotrichosis simplex	38 cases
Hypotrichosis-mental retardation lopes type	1 case
IBIDS syndrome	15 cases
ICF syndrome	50 cases
Ichthyosis - alopecia - ectropion - mental retardation	4 cases
Ichthyosis - hepatosplenomegaly - cerebellar degeneration	2 cases
Ichthyosis - male hypogonadism	5 cases
Ichthyosis - oral and digital anomalies	2 cases
Ichthyosis bullosa of Siemens	<20 cases
Ichthyosis congenita - biliary atresia	2 cases
Ichthyosis congenita, harlequin type	<100 cases
Ichthyosis - deafness - mental retardation - skeletal anomalies	1 case
Ichthyosis follicularis-atrichia-photophobia syndrome	10 cases
Ichthyosis prematurity syndrome	16 families
Ichthyosis, X-linked	16,6
Idiopathic hypereosinophilic syndrome	10
Idiopathic hypersomnia	4
IMAGe syndrome	<20 cases
Iminoglycinuria	6,68
Immune dysregulation - polyendocrinopathy - enteropathy, X linked	7 families
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	<15 cases
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	100 cases
Immunodeficiency with natural-killer cell deficiency	4 cases
Immunodeficiency, common variable	7,5
Inappropriate antidiuretic hormone secretion syndrome	2 cases
Inclusion body myositis, IBM	0,49
Incontinentia pigmenti	0,2
Infant epilepsy with migrant focal crisis	29 cases
Infantile neuroaxonal dystrophy	>150 cases
Inflammatory pseudotumor of the liver	143 cases
Infundibulopelvic stenosis - multicystic kidney	1 family
Insomnia, familial fatal	27 cases
Insulin resistance, short fifth metacarpals	1 family
Intellectual deficit, severe - epilepsy - anal anomalies - distal phalangeal hypoplasia	2 cases
Internal carotid agenesis	100 cases
Interstitial granulomatous dermatitis - arthritis	<20 cases
Intrathoracic kidney vertebral fusion	1 case
Iris coloboma with ptosis - intellectual deficit	10 cases
Iris dysplasia - hypertelorism - deafness	2 cases
Isotretinoin-like syndrome	6 cases
Isovaleric acidemia	1
IVIC syndrome	4 families
Jackson-Weiss syndrome	2 families
Jacobsen syndrome	150 cases
Jeune syndrome	0,2
Job syndrome	250 cases
Johanson-Blizzard syndrome	23 cases
Johnson neuroectodermal syndrome	<30 cases
Joubert syndrome	0,85
Juberg-Hayward syndrome	10 cases
Juvenile arthritis, idiopathic	41,8
Juvenile hyaline fibromatosis	40-50 cases
Juvenile idiopathic arthritis, systemic-onset	6,3
Juvenile macular degeneration, hypotrichosis	7 families
Juvenile temporal arteritis	20 cases
Kabuki syndrome	1,16
Kaler-Garrity-Stern syndrome	2 cases
Kallmann syndrome	0,66
Kaposi's sarcoma	1,7
Kapur-Toriello syndrome	2 cases
Kartagener syndrome	2,5
Kasabach-Merritt syndrome	>175 cases
KBG syndrome	45 cases
Kearns-Sayre syndrome	223 cases
Kennedy disease	2,8
Keratoderma - epithelioma - dental abnormalities-hypogonadism	5 cases
Keratoderma - hypotrichosis - leukonychia	2 cases
Keratoderma palmoplantar - deafness	<10 families
Keratoderma palmoplantar - spastic paralysis	25 cases
Keratosis follicularis - dwarfism - cerebral atrophy	6 cases
Keratosis palmaris et plantaris - clinodactyly	<20
Ketoacidosis due to betaketothiolase deficiency	60 cases
KID syndrome	>100 cases
Kimura disease	200 cases

** Prevalence at birth

Klippel-Feil syndrome	2
Kniest-like dysplasia, lethal	2 cases
Krabbe disease	0,75**
Kudo-Tamura-Fuse syndrome	2 cases
Lacrimo-auriculo-dento-digital syndrome	20 cases
Lambert-Eaton myasthenic syndrome	1
Lamellar ichthyosis	>0,33
Langerhans cell histiocytosis	2
Larsen syndrome	100 cases
Laryngeal abductor paralysis - mental retardation	<20 cases
Laryngotracheoesophageal cleft	1,5
Lateral body wall complex	2**
Lathosterolosis	<5 cases
LCAT deficiency	80 cases
Leber hereditary optic neuropathy	6,5
Legg-Calve-Perthes disease	23
Leigh disease	2,75**
Lennox-Gastaut syndrome	15
Lenz-Majewski hyperostotic dwarfism	7 cases
LEOPARD syndrome	>70 cases
Leprechaunism	0,1**
Leptospirosis	0,24
Lesch-Nyhan syndrome	0,38
Lethal osteosclerotic bone dysplasia	8 families
Leucinosis	15,6
Leukemia, chronic myeloid	6
Leukocyte adhesion deficiency	<350 cases
Leukodystrophy with oligodontia	4 cases
Leukoencephalopathy - metaphyseal chondrodysplasia	4 cases
Leukoencephalopathy - palmoplantar keratoderma	4 cases
Lewis-Pashayan syndrome	3 cases
Lewis-Sumner syndrome	0,9
Lhermitte-Duclos disease	220 cases
Lichstenstein syndrome	2 cases
Li-Fraumeni syndrome	400 families
Limb-mammary syndrome	27 cases
Lipoamide dehydrogenase deficiency	20 cases
Lipodystrophy - mental retardation - deafness	3 cases
Lipodystrophy, Berardinelli type	0,25
Lipodystrophy, familial partial, associated with PPARG mutations	10 cases
Lipodystrophy, familial partial, due to AKT2 mutations	1 family
Lipodystrophy, familial partial, Dunnigan type	200-300 cases
Lipodystrophy, familial partial, Köbberling type	<20 cases
Lipodystrophy, partial acquired	250 cases
Lipoid proteinosis	>280 cases
Lissencephaly - immunodeficiency	1 case
Lissencephaly type 1, due to LIS 1 anomalies	0,3
Lissencephaly type 2	0,12
Lissencephaly type III - familial fetal akinesia sequence	5 cases
Lissencephaly type III - metacarpal bone dysplasia	2 cases
Liver disease - retitinitis pigmentosa - polyneuropathy - epilepsy	4 cases
Long QT syndrome, familial	25
Lopez-Hernandez syndrome	11 cases
Low birth weight - dwarfism - dysgammaglobulinemia	2 cases
Lowe syndrome	0,19
Lumbosacral vertebrae, posterior fusion of - blepharoptosis	3 cases
Lung cancer, small cell	5
Lymphangioleiomyomatosis	0,1
Lymphoblastic lymphoma	10
Lymphoedema - atrial septal defects - facial changes	3 cases
Lymphoedema - cerebral arteriovenous anomaly	5 cases
Lymphoedema, congenital	8,8
Macrocephaly - cutis Marmorata Telangiectatica Congenita	40 cases
Macrocephaly - immune deficiency - anemia	2 cases
Macrocephaly - short stature - paraplegia	2 cases
Macroepiphyseal dysplasia, Macalister Coe type	1 case
Macrogyria - pseudobulbar palsy	4 cases
Macrophagic myofasciitis	1
Macrostomia - preauricular tags - external ophthalmoplegia	9 cases
Malakoplasia	500 cases
Malignant hyperthermia	33
Malignant hyperthermia - arthrogryposis - torticollis	4 cases
Malonic aciduria	17 cases
Mandibuloacral dysplasia	37 cases
Mantle cell lymphoma	3,9
Marden-Walker syndrome	30 cases
Marfan syndrome	30
Marie Unna, congenital - hypotrichosis	12 families
Marinesco-Sjogren syndrome	100-200 cases
Marshall's syndrome with periodic fever	30 cases
Marshall-Smith syndrome	33 cases
Martinez-Monasterio-Pinheiro syndrome	1 case
MASA syndrome	3,5
Maternal hyperphenylalaninemia	1,25
Matthew-Wood syndrome	5 cases
Mayer-Rokitansky-Küster-Hauser syndrome	9
McCune-Albright syndrome	158 cases
Meckel syndrome	4**
Meconium aspiration syndrome	2,44
Median cleft of the upper lip - corpus callosum lipoma - cutaneous polyps	>10 cases
Medullary cystic kidney disease, autosomal dominant	0,11

** Prevalence at birth

Megacystis microcolon - intestinal hypoperistalsis - hydronephrosis	89 cases
Megaepiphyseal dwarfism	1 case
Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus	6 cases
MEHMO syndrome	7 cases
MEHMO syndrome	7 cases
Melanoma, familial	46,8
MELAS syndrome	16
Melorheostosis	300 cases
Mendelian susceptibility to atypical mycobacteria	0,059
Meniere's disease	42,5
Menkes syndrome	0,7
Mental retardation - cubitus valgus - unusual facies	5 cases
Mental retardation - dysmorphism - hypogonadism - diabetes mellitus	4 cases
Mental retardation - hypoplastic corpus callosum - preauricular tag	3 cases
Mental retardation - microcephaly - phalangeal - facial abnormalities	6 cases
Mental retardation - progressive spasticity, X-linked	1 family
Mental retardation - sparse hair - brachydactyly	6 cases
Mental retardation multiple nevi	1 case
Mental retardation X-linked - dysmorphism	8 cases
Mental retardation X-linked - psychosis - macroorchidism	6 cases
Mental retardation X-linked - seizures - short stature - midface hypoplasia	17 cases
Mental retardation, choreoathesis and abnormal behavior	5 cases
Mental retardation, X linked - precocious puberty - obesity	3 cases
Mental retardation, X-linked - acromegaly - hyperactivity	2 cases
Mental retardation, X-linked - Dandy Walker malformation - Basal ganglia disease - Seizures	16 cases
Mental retardation, X-linked - epilepsy - progressive joint contractures - typical face	2 cases
Mental retardation, X-linked - hypogammaglobulinemia - progressive neurological deterioration	3 cases
Mental retardation, X-linked - hypogonadism - ichthyosis - obesity - short stature	4 cases
Mental retardation, X-linked - hypotonia - facial dysmorphism - aggressive behavior	10 cases
Mental retardation, X-linked - macrocephaly - macro-orchidism	12 cases
Mental retardation, X-linked - seizures - psoriasis	4 cases
Mental retardation, X-linked - Spastic paraplegia with iron deposits	1 family
Mental retardation, X-linked recessive - macrocephaly - ciliary dysfunction	1 family
Mental retardation, X-linked severe, Gustavson type	7 cases
Mental retardation, X-linked, Abidi type	8 cases
Mental retardation, X-linked, Armfield type	6 cases
Mental retardation, X-linked, Cabezas type	1 family
Mental retardation, X-linked, Cantagrel type	2 cases
Mental retardation, X-linked, Lubs type	5 cases
Mental retardation, X-linked, Miles-Carpenter type	4 cases
Mental retardation, X-linked, Pai type	1 family
Mental retardation, X-linked, Reish type	2 cases
Mental retardation, X-linked, Schimke type	4 cases
Mental retardation, X-linked, Seemanova type	4 cases
Mental retardation, X-linked, Shashi type	9 cases
Mental retardation, X-linked, Shrimpton type	3 cases
Mental retardation, X-linked, Siderius type	4 cases
Mental retardation, X-linked, Snyder type	11 cases
Mental retardation, X-linked, South African type	16 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
Mental retardation, X-linked, syndromic 7	10 cases
Mental retardation, X-linked, syndromic, due to JARID1C mutation	<10 families
Mental retardation, X-linked, Vitale type	8 cases
Mental retardation, X-linked, Wilson type	3 cases
Mental retardation, X-linked, with isolated growth hormone deficiency	3 families
Mental retardation, X-linked, Zorick type	6 cases
MERRF syndrome	0,9
Mesomelic dysplasia - skin dimples	2 cases
Metachromatic leukodystrophy	0,16
Metaphyseal acroschyphodysplasia	4 cases
Metaphyseal anadysplasia	<20 cases
Metaphyseal chondrodysplasia, Jansen type	16 cases
Metaphyseal dysplasia hypertelorism hypospadias	1 case
Metatropic dwarfism	60 cases
Methimazole embryofetopathy	40 cases
Methylmalonic acidemia - homocystinuria	300 cases
Methylmalonic aciduria - microcephaly - cataract	2 cases
Mevalonicaciduria	30 cases
Michels syndrome	7 cases
Micro syndrome	8 cases
Microbrachycephaly - ptosis - cleft lip	2 cases
Microcephalic osteodysplastic dysplasia, Saul-Wilson type	4 cases
Microcephaly - cardiomyopathy	3 cases
Microcephaly - cleft palate	3 cases
Microcephaly - glomerulonephritis - marfanoid habitus	2 cases
Microcephaly - micropenis - convulsions	4 cases
Microcephaly - seizures - mental retardation - heart disease	2 cases
Microcephaly syndactyly brachymesophalangy	1 case
Microcoria - congenital nephrosis	22 cases
Microcytic anemia - liver iron overload - low ferritinemia	3 cases
Microdontia - type I microtia - deafness	9 cases

** Prevalence at birth

Microgastria - limb reduction defect	16 cases
Microlissencephaly - micromelia	2 cases
Microphthalmia - brain atrophy	3 cases
Microscopic polyangiitis	7,5
Microtia	15
Midas syndrome	<50 cases
Midline cleft of lower lip	70 cases
Mitochondrial diseases of nuclear origin	9
Mitochondrial encephalomyopathy - aminoacidopathy	2 cases
Mitral regurgitation - deafness - skeletal anomalies	3 cases
Mixed dystonias	3 families
Moebius syndrome	300 cases
Mohr-Tranebjaerg syndrome	46 cases
Molarization of anterior teeth deafness	1 case
Molybdenum cofactor deficiency	>100 cases
Monosomy 22q11	20
Monosomy 22q13	>200 cases
Monosomy 5p	4,6
Monosomy 9q22.3	2 cases
Moore-Federman syndrome	6 cases
Mosaic variegated aneuploidy syndrome	29 cases
Mowat-Wilson syndrome	<100 cases
Moya-Moya disease	3,16
Mucolipidosis type 2	0,15**
Mucolipidosis type 4	>100 cases
Mucopolysaccharidosis type 1	1,3
Mucopolysaccharidosis type 2	0,6
Mucopolysaccharidosis type 3	1,1
Mucopolysaccharidosis type 4	0,4
Mucopolysaccharidosis type 6	0,16**
Mucopolysaccharidosis type 7	<40cases
Mucosulfatidosis	50 cases
Muir-Torre syndrome	205 cases
Mullerian derivatives - lymphangiectasia - polydactyly	3 cases
Multifocal motor neuropathy with conduction block	1,5
Multiple endocrine neoplasia, type 1	11
Multiple endocrine neoplasia, type 2	3,3
Multiple fibrofolliculoma familial	7 cases
Multiple joint dislocations metaphyseal dysplasia	1 case
Multiple pterygium syndrome, autosomal dominant	4 cases
Multiple pterygium syndrome, lethal form	200 cases
Multiple sclerosis - ichthyosis - factor VIII deficiency	2 cases
Multiple system atrophy	4,6
MURCS association	11,25
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus	10 cases
Muscular dystrophy congenital, merosin negative	0,3
Muscular dystrophy congenital, merosin-positive	0,15
Muscular dystrophy congenital, with integrin deficiency	0,03
Muscular dystrophy Fukuyama type	0,54
Muscular dystrophy limb-girdle	0,8
Muscular dystrophy, Duchenne and Becker types	5
Muscular dystrophy, limb girdle, autosomal recessive, type 2G	14 cases
Muscular dystrophy, limb girdle, autosomal recessive, type 2I	>40 families
Muscular dystrophy, limb-girdle, autosomal dominant, type 1A	1 family
Muscular dystrophy, limb-girdle, autosomal dominant, type 1D	5 families
Muscular dystrophy, limb-girdle, autosomal dominant, type 1E	5 families
Muscular dystrophy, limb-girdle, autosomal dominant, type 1F	1 family
Muscular dystrophy, limb-girdle, autosomal dominant, type 1G	1 family
Myasthenia gravis	8,5
Myelodysplastic syndromes	25
Myelofibrosis with myeloid metaplasia	10
Myeloma, multiple	14,25
Myhre syndrome	15 cases
Myoclonus - cerebellar ataxia - deafness	4 cases
Myoclonus hereditary - progressive distal muscular atrophy	<10 cases
Myoneurogastrointestinal encephalopathy syndrome	70 cases
Myopathy - lactic acidosis - sideroblastic anemia	7 cases
Myopathy due to calsequestrin and SERCA1 protein overload	4 cases
Myopathy, X-linked, with excessive autophagy	15 families
N syndrome	3 cases
N-acetyl-alpha-D-galactosaminidase deficiency	12 cases
Naegeli-Franceschetti-Jadassohn syndrome	0,035
Nail patella-like renal disease	3 cases
Nail-patella syndrome	2
Nance-Horan syndrome	50 families
Nanism due to growth hormone qualitative anomaly	3 cases
Nanism due to growth hormone resistance	0,2
NARP syndrome	8,5
Nasopalpebral lipoma - coloboma - telecanthus	<30 cases
Nasopharyngeal teratoma - Dandy Walker - diaphragmatic hernia	1 case
Necrotizing encephalopathy, acute, autosomal dominant	11 cases
Nemaline myopathy	1
Neonatal death - immune deficiency	5 cases
Neonatal hemochromatosis	100 cases
Nephroblastoma	10,1
Nephronophthisis, autosomal recessive	1,05
Nephronophtisis familial - adult spastic quadriparesis	2 cases

** Prevalence at birth

Nephropathy - deafness - hyperparathyroidism	5 cases
Nephropathy familial with gout	57 families
Nephrosis - deafness - urinary tract and digital malformations	5 cases
Nephrotic syndrome, steroid-sensitive	18
Netherton disease	1,35
Neu laxova syndrome	50-60 cases
Neuroaxonal dystrophy - renal tubular acidosis	3 cases
Neuroblastoma	10
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	4 cases
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	2 cases
Neurodegenerative syndrome, X-linked, Bertini type	7 cases
Neurodegenerative syndrome, X-linked, Hamel type	10 cases
Neuroectodermal endocrine syndrome	4 cases
Neuroendocrine tumor	1
Neurofibromatosis, type 1	25
Neurofibromatosis, type 2	0,5
Neurometabolic disorder due to serine deficiency	<30 cases
Neuropathy hereditary with liability to pressure palsies	9
Neuropathy, giant axonal	> 20 families
Neutropenia severe congenital	0,33**
Neutropenia, severe congenital, X-linked	7 cases
Nevo syndrome	10 cases
Nevus of ota - retinitis pigmentosa	1 case
Niemann-Pick disease	2,5**
Niemann-Pick disease, type A	0,25**
Niemann-Pick disease, type B	0,75**
Niemann-Pick disease, type C	0,85
Nijmegen-breakage syndrome	130 cases
Nodular regenerative hyperplasia of the liver	3
Non-distal trisomy 10p	60 cases
Non-Hodgkin malignant lymphoma	36
Norrie disease	300 cases
North Carolina macular dystrophy	2 families
Obesity - colitis - hypothyroidism - cardiac hypertrophy - developmental delay	2 cases
Obesity due to congenital leptin deficiency	<30 cases
Obesity due to prohormone convertase-I deficiency	2 cases
Obesity due to pro-opiomelanocortin deficiency	7 cases
Ochoa syndrome	>100 cases
Ocular albinism X-linked, recessive	2
Ocular motor apraxia, Cogan type	50 cases
Oculocerebrocutaneous syndrome	36 cases
Oculocerebrofacial syndrome, Kaufman type	9 cases
Oculocutaneous albinism	7,15
Oculodental syndrome rutherfurd syndrome	1 family
Oculo-dento-osseous dysplasia, autosomal dominant	243 cases
Oculo-dento-osseous dysplasia, autosomal recessive	5 cases

Oculo-digito-esophageal-duodenal syndrome (ODED)	<50 cases
Oculogastrointestinal muscular dystrophy	1 family
Oculoosteocutaneous syndrome	3 cases
Oculo-oto-facial dysplasia	4 cases
Oculo-palato-cerebral syndrome	5 cases
Oculopharyngeal muscular dystrophy	1
Oculo-tricho-dysplasia	2 cases
Odonto-micronychial dysplasia	5 cases
Odontoonychodermal dysplasia	<15 cases
Odonto-onycho-hypohidrotic dysplasia - midline scalp defects	1 case
Odontotrichomelic syndrome	4 cases
Odonto-tricho-ungual-digito-palmarn syndrome	21 cases
Oesophageal atresia	25
Okamoto syndrome	2 cases
Oligoarticular chronic arthritis	20,5
Oligocone trichromacy	14 cases
Olmsted syndrome	32 cases
Omodysplasia	30 cases
Omphalocele	12
Omphalocele-cleft palate syndrome, lethal	3 cases
Ondine syndrome	2,25
Onycho-tricho-dysplasia - neutropenia	5 cases
Ophthalmo acromelic syndrome	30 cases
Opitz BBB/G syndrome	3
Opsismodysplasia	25 cases
Optic atrophy	6
Optic atrophy and cataract, autosomal dominant	14 cases
Orbital leiomyoma	16 cases
Orofaciodigital syndrome, type 1	1,2
Orofaciodigital syndrome, type 10	1 case
Orofaciodigital syndrome, type 3	3 cases
Orofaciodigital syndrome, type 4	16 cases
Orofaciodigital syndrome, type 5	4 cases
Orofaciodigital syndrome, type 6	29 cases
Orofaciodigital syndrome, type 8	1 ou 2 families
Orotic aciduria hereditary	<20 cases
Ossification anomalies - psychomotor development delay	2 cases
Osteochondritis dissecans	35
Osteochondrodysplasia thrombocytopenia hydrocephalus	1 case
Osteocraniostenosis	12 cases
Osteodysplasty, Melnick-Needles type	>50 cases
Osteogenesis imperfecta	6,5
Osteogenesis imperfecta - retinopathy - seizures - intellectual deficit	2 cases
Osteogenesis imperfecta congenita - microcephaly - cataracts	3 cases
Osteopathia striata - cranial sclerosis	100 cases
Osteopetrosis autosomal dominant, type 1	33 cases

** Prevalence at birth

Osteopetrosis lethal	2 cases
Osteopetrosis, intermediate form	50 cases
Osteopetrosis, malignant	0,75**
Osteopoikilosis - short stature - intellectual deficit	4 cases
Osteoporosis oculo-cutaneous hypopigmentation syndrome	3 cases
Osteoporosis pseudoglioma syndrome	0,05
Osteosarcoma	5
Osteosclerosis - ichthyosis - premature ovarian failure	3 cases
Otodental syndrome	9 families
Otopalatodigital syndrome	30 cases
Overhydrated hereditary stomatocytosis	> 20 families
P2Y12 deficiency	5 cases
Pachydermoperiostosis	204 cases
Pachyonychia congenita	230 cases
Pacman dysplasia	<10 cases
Paget disease, juvenile type	50 cases
Pallister-Hall syndrome	100 cases
Palmoplantar keratoderma - amyotrophy	4 cases
Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma	5 cases
Palmoplantar porokeratosis of Mantoux	>10 cases
Pancreas agenesis	8 cases
Pancreatic and cerebellar agenesis	4 cases
Pancreatic hypoplasia - diabetes - heart disease	<10 cases
Pancreatic lipomatosis - duodenal stenosis	1 case
Pancreatitis, hereditary	0,125
Pancreatoblastoma	60 cases
Papillon-Lefevre syndrome	0,25
Papulosis, malignant atrophic	>200 cases
Paraplegia - brachydactyly - cone-shaped epiphysis	5 cases
Paraplegia - mental retardation - hyperkeratosis	4 cases
Paraplégie spastique - glaucome - déficit intellectuel	2 families
PARC syndrome	2 cases
Parietal foramina	5
Parkinson disease, genetic types	15
Parkinsonism, young adult onset	37,5
Paroxysmal nocturnal hemoglobinuria	0,55
Parsonage-Turner syndrome	3,3
Patella hypoplasia skeletal malformations	1 case
Pearson syndrome	60 cases
Pelizaeus-Merzbacher disease	0,25
PELVIS syndrome	11 cases
Pemphigus paraneoplastic	>60 cases
Pemphigus superficial	1,2
Pemphigus vulgaris	3,8
Pendred syndrome	5,5
Perinatal-lethal Gaucher disease	0,01
Peritoneal leiomyomatosis, disseminated	100 cases
Perlman syndrome	<20 cases
Persistent hyperinsulinemic hypoglycemia of infancy	2
Persistent Mullerian duct syndrome	<200 cases
Peters-plus syndrome	50 cases
Peutz-Jeghers syndrome	2,2
Pfeiffer syndrome	0,38
Pfeiffer-Singer-Zschiesche syndrome	<10 cases
PHACE syndrome	100 cases
Phenylketonuria	4
Pheochromocytoma and paraganglioma, secreting	10
Phosphoenolpyruvate carboxykinase (PEPCK) deficiency	<10 cases
Phosphoglycerate kinase 1 deficiency	23 cases
Phosphoribosylpyrophosphate synthetase superactivity	<30 families
Phytosterolemia	40 cases
PIBIDS syndrome	20 cases
Piebaldism	0,25
Pierre Robin syndrome	8,75
Pili torti - onychodysplasia	1 family
Pilodental dysplasia with refractive errors	2 cases
Pityriasis rubra pilaris	48 cases
Plagiocephaly - mental retardation, X-linked	2 cases
Platelet syndrome, familial	<20 families
Plummer-Vinson syndrome	25 cases
Podder-tolmie syndrome	1 case
Poikiloderma of Kindler	100 cases
Poland anomaly	2
Pollitt syndrome	10 cases
Polyarteritis nodosa	3,07
Polyarthritis, rheumatic factor-negative	8
Polyarthritis, rheumatoid factor-positive	4,2
Polycystic kidney disease, autosomal recessive	6,5
Polycystic lipomembranous osteodysplasia - sclerosing leukoencephalopathy	0,15
Polycystic ovaries - urethral sphincter dysfunction	33 cases
Polycythemia vera	25
Polydactyly postaxial	50
Polydactyly preaxial	25
Polymorphic catecholergic ventricular tachycardia	10
Polymyositis	14,8
Polysyndactyly - cardiac malformation	6 cases
Pontocerebellar hypoplasia type 1	6 families
Pontocerebellar hypoplasia type 2	<30 cases
Porencephaly, familial	10 families
Posterior column ataxia - retinitis pigmentosa	13 cases
Post-transplant lymphoproliferative disease	26,2
Potocki-Shaffer syndrome	23 cases
Prader-Willi syndrome	10,7
Preauricular pits - renal disease	1 case
Primary biliary cirrhosis	13,5
Primary ciliary dyskinesia	5

** Prevalence at birth

Primary lateral sclerosis	1,5
Primary sclerosing cholangitis	7
Progeria	0,25**
Progeria - short stature - pigmented nevi	<10 cases
Progressive bulbar paralysis of childhood	<40 cases
Progressive neurodegeneration - joint laxity - cataract	2 cases
Progressive vertebral fusion, non-infectious, syndromic form	10 cases
Prolidase deficiency	50 cases
Propionic acidemia	3,75
Proteus syndrome	100-200
Proximal spinal muscular atrophy	3
Proximal spinal muscular atrophy, type 1	0,26
Proximal spinal muscular atrophy, type 2	2,6
Proximal spinal muscular atrophy, type 3	2,6
Proximal spinal muscular atrophy, type 4	0,32
Pseudoachondroplasia	3
Pseudoarthrosis of clavicle, congenital	>200 cases
Pseudodiastrophic dysplasia	10 cases
Pseudo-Gaucher disease	<10 cases
Pseudohypoaldosteronism type 1	70 cases
Pseudo-progeria syndrome	2 cases
Pseudoxanthoma elasticum	2,5
Pseudo-Zellweger syndrome	<10 cases
Psoriatic arthritis, juvenile form	4,2
Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	3 cases
Pterygium colli - intellectual deficit - digital anomalies	2 cases
Pterygium popliteal syndrome, autosomal dominant	0,3
Pterygium syndrome, antecubital	11 cases
Ptosis - strabismus - ectopic pupils	1 family
Ptosis strabismus diastasis	2 cases
Pulmonary alveolar proteinosis	0,1
Pulmonary arterial hypertension	1,5
Pulmonary fibrosis, idiopathic	27
Pulmonary haemosiderosis, primary	250 cases
Pulmonary lymphangiectasia, congenital	>100 cases
Pulmonary valve stenosis, congenital	7,2
Pure autonomic failure	0,3
Pyogenic arthritis - pyoderma gangrenosum - acne	34 cases
Pyruvate kinase deficiency	0,4
Qazi-Markouizos syndrome	3 cases
Radiation proctitis	35
Radio-ulnar synostosis - amegakaryocytic thrombocytopenia	<20 cases
Rambaud-Galian syndrome	3 cases
RAPADILINO syndrome	<20 cases
Rapid-onset dystonia-parkinsonism	3 families
Rapp-Hodgkin syndrome	72 cases
Rasmussen subacute encephalitis	>100 cases
Recurrent infections - short stature - hypopigmentation - coarse face	4 cases
Refsum disease	0,1
Refsum disease, infantile form	0,005
Regional odeontodysplasia	139 cases
Relapsing polychondritis	3,5
Renal adysplasia	26
Renal dysplasia, multicystic	3 cases
Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts	10 cases
Rendu-Osler-Weber disease	3,5
Renpenning syndrome	10 families
Restrictive cardiomyopathy, idiopathic or familial	2,5
Retinal arteries, tortuosity of	100 cases
Retinal degeneration - nanophthalmos - glaucoma	7 cases
Retinitis pigmentosa	27,5
Retinitis pigmentosa - deafness - hypogenitalism	2 families
Retinoblastoma	5,4
Retino-hepato-endocrinologic syndrome	7 cases
Retinopathy pigmentary - mental retardation	<15 cases
Retinoschisis, X-linked	4,5
Rett syndrome	8,2
Rheumatic fever	5
Rheumatoid purpura	8,5
Rhombencephalosynapsis	50 cases
Richieri costa colletto otto syndrome	1 case
Rickettsialpox	>800 cases
Rieger syndrome	0,5
Rigid mask like face deafness polydactyly	1 case
Ring chromosome 10	<20 cases
Ring chromosome 14	50 cases
Ring chromosome 17	14 cases
Ring chromosome 20	>50 cases
Roberts syndrome	100 cases
Robinow like syndrome	2 cases
Robinow syndrome, dominant form	100 cases
Robinow syndrome, recessive form	70 cases
Rolled and spiral hairs - palmoplantar keratoderma	4 cases
Rothmund-Thomson syndrome	300 cases
Rubella panencephalitis	>20 cases
Rubinstein-Taybi like, syndrome	8 cases
Rubinstein-Taybi syndrome	1
Rudiger syndrome	2 cases
Sacral hemangiomas - multiple congenital abnormalities	5 cases
Saethre-Chotzen syndrome	3
Sakati-Nyhan syndrome	<5 cases
Sandhoff disease	0,75
Sarcoidosis	15
Sarcosinemia	2
Say-Barber-Miller syndrome	2 cases

** Prevalence at birth

Scalp defects - postaxial polydactyly	2 cases
Scalp-ear-nipple syndrome	30 cases
SCARF syndrome	2 cases
Schinzel syndrome	<10 families
Schinzel-Giedion midface retraction syndrome	34 cases
Schizencephaly	1,54
Schizophrenia - mental retardation - deafness - retinitis	1 family
Schnitzler syndrome	50 cases
Schopf-Schulz-Passarge syndrome	19 cases
Schwartz-Jampel syndrome	100 cases
Scleroderma	42
Sclerosing bone dysplasia - mental retardation	1 case
Sclerosing bone dysplasia mental retardation	1 case
Sebastian syndrome	<10 families
Seckel syndrome	100 cases
Segmental odontomaxillary dysplasia	32 cases
Seizures - intellectual deficit due to hydroxylysinuria	3 cases
Senior-Loken syndrome	0,1
Sensorineural hearing loss - early greying - essential tremor	3 cases
Serpentine fibula - polycystic kidneys	6 cases
Severe achondroplasia - developmental delay - acanthosis nigricans	4 cases
Severe combined immunodeficiency due to adenosine deaminase deficiency	0,22
Severe combined immunodeficiency T- B-	0,35
Severe combined immunodeficiency T- B+, X-linked	1,5
Sezary's syndrome	0,18
Short stature - mental retardation - eye anomalies - cleft lip palat	3 cases
Short stature - pituitary and cerebellar defects - small sella turcica	1 family
Short stature - webbed neck - heart disease	4 cases
Short stature, Brussels type	2 cases
SHORT syndrome	30 cases
Shprintzen-Goldberg syndrome	<50 cases
Shwachman-Diamond syndrome	200 cases
Sialidosis type 1	0,02**
Sialidosis type 2	0,02**
Sickle cell anaemia	11
Siegler-Brewer-Carey syndrome	2 cases
Silent sinus syndrome	98 cases
Sillence syndrome	5 cases
Silver-Russell dwarfism	400 cases
Simpson-Golabi-Behmel syndrome	>100 cases
Simpson-Golabi-Behmel syndrome, type 2	4 cases
Singleton-Merten dysplasia	<10 cases
Sirenomelia	1**
Sjögren-Larsson syndrome	0,4
Skeletal dysplasia - intellectual deficit	1-2 families
Small vessel disease of the brain, not NOTCH3-related	2 cases
Smith-Lemli-Opitz syndrome	6,5
Smith-Magenis syndrome	4
Soft tissue sarcomas	13
Sotos syndrome	7**
Sparse hair - short stature - skin anomalies	4 cases
Spastic paraplegia - nephritis - deafness	4 cases
Spastic paraplegia, familial	5
Spastic quadriplegia - retinitis pigmentosa - mental retardation	2 cases
Spasticity - mental retardation - epilepsy, X-linked	6 cases
Spherocytosis hereditary	20
Spinal muscular atrophy - Dandy-Walker complex - cataracts	2 cases
Spinocerebellar ataxia X-linked, type 3	5 cases
Spinocerebellar ataxia, infantile onset	21 cases
Split hand - split foot	1,1
Split hand - split foot - deafness	22 cases
Split hand - urinary anomalies - spina bifida	3 cases
Spondylocarpotarsal synostosis	>20 cases
Spondylo-costal dysostosis - Dandy Walker	1 case
Spondyloenchondrodysplasia	36 cases
Spondylometaphyseal dysplasia	0,34
Spondylometaphyseal dysplasia - combined immunodeficiency	4 cases
Spondylometaphyseal dysplasia - cone-rod dystrophy	8 cases
Spontaneous periodic hypothermia	<30 cases
Squamous cell carcinoma of head and neck	46
Stargardt disease	11,25
Steinert myotonic dystrophy	4,5
Stern-Lubinsky-Durrie syndrome	7 cases
Sternal cleft	<2
Steroid dehydrogenase deficiency - dental anomalies	1 family
Stickler syndrome	13,5
Stimmler syndrome	2 cases
Stoll-Alembik-Finck syndrome	1 case
Subcorneal pustular dermatosis	200 cases
Succinic acidemia	50 cases
Succinyl-CoA acetoacetate transferase deficiency	10 cases
Sulfite oxidase deficiency	50 cases
Summitt syndrome	3 cases
Supranuclear palsy, progressive	5
Susac syndrome	<100 cases
Symmetrical thalamic calcifications	29 cases
Sympathetic ophthalmia	0,6
Symphalangism - short stature - accessory testis	1 case
Symphalangism distal	<5 families
Symphalangism short stature accessory testis	1 case
Symphalangism with multiple anomalies of hands and feet	6 cases

** Prevalence at birth

Synostoses, multiple - brachydactyly	20 families
Synspondylism	24 cases
Syringomyelia	8,4
Systemic mastocytosis	3,3
Systemic vasculitis	6,3
Takayasu arteritis	0,45
Talo-patello-scaphoid osteolysis	2 cases
Tangier disease	>70 cases
Taurodontia - absent teeth - sparse hair	<15 cases
Tay-Sachs disease	0,3**
Terminal osseous dysplasia - pigmentary defects	18 cases
Tetraamelia - pulmonary hypoplasia	5 families
Tetralogy of Fallot	45
Thalidomide embryopathy	5000 cases
Thanatophoric dwarfism	3,5**
Thiamine-responsive megaloblastic anemia syndrome	30 families
Thomsen and Becker disease	5
Thrombocythemia, essential	27,5
Thrombocytopenic purpura, autoimmune	10
Thumb absent - short stature - immune deficiency	3 cases
Thumb stiff - brachydactyly - mental retardation	6 cases
Thymic-renal-anal-lung dysplasia	3 cases
Thyrocerebrorenal syndrome	2 cases
Thyroid carcinoma, anaplastic	0,13
Thyroid carcinoma, medullary	7
Thyrotoxic periodic paralysis	2
Tibial muscular dystrophy	6
Tietz syndrome	1 family
Toriello-Carey syndrome	60 cases
Toriello-Lacassie-Droste syndrome	10 cases
Torticollis - keloids - cryptorchidism - renal dysplasia	7 cases
Townes-Brocks syndrome	0,42
Tracheal agenesis	1**
Tracheobronchomegaly	<40 cases
Tracheo-bronchomegaly	<100 cases
Transmissible spongiform encephalopathies	0,3
Treacher-Collins syndrome	6
Treft-Sanborn-Carey syndrome	23 cases
Trichinellosis	6500 cases
Trichodental syndrome	<5 families
Trichodentoosseous syndrome	>30 cases
Trichodysplasia - amelogenesis imperfecta	1 family
Tricho-megaly - retina pigmentary degeneration - dwarfism	11 cases
Trichooculodermovertebral syndrome	1 case
Trichoodontoonychial dysplasia	4 cases
Trichoonychohypohidrotic dysplasia	1 case
Tricho-retino-dento-digital syndrome	9 cases
Tricho-rhino-phalangeal syndrome type 1	>100 cases
Trichorhinophalangeal syndrome, type 1 and 3	>100 cases
Tricuspid atresia	5
Trigonocephaly - bifid nose - acral anomalies	2 cases
Trigonocephaly - broad thumbs	2 cases
Trigonocephaly - ptosis coloboma	8 cases
Trigonocephaly - short stature - developmental delay	3 cases
Triose phosphate-isomerase deficiency	30 cases
Triphalangeal thumbs - brachyectrodactyly	4 families
Triple A syndrome	100 cases
Triple H syndrome	50 cases
Triplo-X syndrome	42,5
Trisomy 13	13**
Trisomy 18	9**
Tritanopia	4,8
True hermaphrodism	>500 cases
Tuberculosis	20
Tuberous sclerosis	8,8
Tubular renal disease - cardiomyopathy	2 cases
Tufted angioma	>200 cases
Turner syndrome	20
Tyrosinemia type 1	0,05
Tyrosinemia type 2	<100 cases
Tyrosinemia type 3	2 cases
Uhl anomaly	84 cases
Ulbright-Hodes syndrome	3 cases
Ullrich, congenital muscular dystrophy	<100 cases
Ulnar / fibula ray defect - brachydactyly	1 family
Umbilical cord ulceration - intestinal atresia	15 cases
Unverricht-Lundborg disease	0,2
Upington disease	1 family
Usher syndrome	3,5
VACTERL with hydrocephalus	<10 families
Van den Bosch syndrome	1 family
Van Der Woude syndrome	2
VATER association	23
Vernal keratoconjunctivitis	10
Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay	2 cases
Von Hippel-Lindau disease	0,2
Vulvovaginal gingival syndrome	127 cases
W syndrome	6 cases
Waardenburg syndrome	2,4
Waardenburg syndrome type 1	3,75
Waardenburg-Shah syndrome	50 cases
Waldenström macroglobulinemia	2,6
Walker-Warburg syndrome	1,65**
Weaver syndrome	30 cases
Weaver-Williams syndrome	2 cases
Wegener granulomatosis	6,6
Weill-Marchesani syndrome	128 cases

** Prevalence at birth

Wells syndrome	80 cases
Werner syndrome	0,45
West syndrome	3,7**
Western equine encephalitis	>600 cases
WHIM syndrome	40 cases
Whipple disease	1000 cases
Wieacker-Wolff syndrome	6 cases
Wiedemann-Rautenstrauch syndrome	25 cases
Willebrand disease	12,5
Williams syndrome	13,3
Wilson disease	5,84
Wilson-Turner syndrome	>14 cases
Winchester disease	12 cases
Wiskott-Aldrich syndrome	0,15
Wittwer syndrome	3 cases
Wolcott-Rallison syndrome	17 cases
Wolf-Hirschhorn syndrome	2**
Wolfram syndrome	0,57

Wolman disease	0,28**
Woolly hair - hypotrichosis - everted lower lip - outstanding ears	1 family
Woolly hair - palmoplantar keratoderma - dilated cardiomyopathy	<20 cases
Wrinkly skin syndrome	<30 cases
Xanthinuria	150 cases
Xanthomatosis cerebrotendinous	0,13
Xeroderma - talipes - enamel defects	2 cases
Xeroderma pigmentosum	0,5
Xk aprosencephaly	<10 cases
X-linked dominant chondrodysplasia punctata	0,5
X-linked lymphoproliferative disease	0,1
XY gonadal agenesis	50 cases
Zellweger syndrome	1,1
Zellweger-like syndrome, without peroxisomal anomalies	2 cases
Zollinger-Ellison syndrome	5,3
Zunich-Kaye syndrome	6 cases

** Prevalence at birth

List of diseases by decreasing prevalence

Diseases with prevalence data available

Disease name	Estimated prevalence (/100,000)
Polydactyly postaxial	50
Gelineau disease	49
Guillain-Barré syndrome	47,5
Melanoma, familial	46,8
Squamous cell carcinoma of head and neck	46
Autism	45
Tetralogy of Fallot	45
Deafness, autosomal recessive, nonsyndromic, sensorineural, type DFNB	44
Arrhythmogenic right ventricular dysplasia	43,5
Meniere's disease	42,5
Triplo-X syndrome	42,5
Chromosome Y deletion	42
Scleroderma	42
Juvenile arthritis, idiopathic	41,8
Familial venous malformations	40
Fetal cytomegalovirus syndrome	40
Achalasia, primary	37,5
Elliptocytosis, hereditary	37,5
Parkinsonism, young adult onset	37,5
Follicular lymphoma	36
Non-Hodgkin malignant lymphoma	36
Osteochondritis dissecans	35
Radiation proctitis	35
Adactylia unilateral	34
Cryptosporidiosis	34
Malignant hyperthermia	33
Charcot-Marie-Tooth disease (generic term)	32,5
Great vessels transposition (TGV)	32,5
B-cell chronic lymphocytic leukemia	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
Renal adysplasia	26
Long QT syndrome, familial	25
Myelodysplastic syndromes	25
Neurofibromatosis type 1	25
Oesophageal atresia	25
Polycythemia vera	25
Polydactyly preaxial	25
Anorectal malformation	24
Legg-Calve-Perthes disease	23
VATER association	23
Oligoarticular chronic arthritis	20,5
Dermatitis herpetiformis	20,2
Alpha-1 antitrypsin deficiency	20
Atresia of small intestine	20
Atrioventricular canal, partial	20
Gastric cancer	20
Hirschsprung disease	20
Monosomy 22q11	20
Spherocytosis hereditary	20
Tuberculosis	20
Turner syndrome	20
Corpus callosum agenesis - neuronopathy	19
Nephrotic syndrome, steroid-sensitive	18
Cardiomyopathy, familial dilated	17,5
Boutonneuse fever	17
Breast cancer, familial	17
Ichthyosis, X-linked	16,6
MELAS syndrome	16
Leucinosis	15,6
Acyl-CoA dehydrogenase, medium chain, deficiency	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
Anophtalmia/microphthalmia, isolated	14
Cystinuria	14
Primary biliary cirrhosis	13,5
Stickler syndrome	13,5
Williams syndrome	13,3
Androgen insensitivity syndrome	13
Bronchopulmonary dysplasia	13
Soft tissue sarcomas	13
Trisomy 13	13**

** Prevalence at birth

Buerger's disease	12,5
Ehlers-Danlos syndrome, type 3	12,5
Willebrand disease	12,5
Cystic fibrosis	12
Gastroschisis	12
Gonadal dysgenesis, XX type	12
Omphalocele	12
Focal dystonia	11,7
MURCS association	11,25
Stargardt disease	11,25
Glioblastoma	11
Hepatic veno-occlusive disease	11
Multiple endocrine neoplasia type 1	11
Sickle cell anaemia	11
Prader-Willi syndrome	10,7
Alopecia totalis	10,5
Collagenous colitis	10,5
Hodgkin lymphoma	10,5
Nephroblastoma	10,1
Adrenal hyperplasia, congenital	10
Bone tumor	10
Cholangiocarcinoma	10
Dermatofibrosarcoma protuberans	10
Distal myopathy, Welander type	10
Duane syndrome	10
Factor II deficiency	10
Idiopathic hypereosinophilic syndrome	10
Lymphoblastic lymphoma	10
Myelofibrosis with myeloid metaplasia	10
Pheochromocytoma and paraganglioma, secreting	10
Polymorphic catecholergic ventricular tachycardia	10
Thrombocytopenic purpura, autoimmune	10
Vernal keratoconjunctivitis	10
Neuroblastoma	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 syndrome	8,75
Duodenal atresia	8,55
Myasthenia gravis	8,5
NARP syndrome	8,5
Rheumatoid purpura	8,5
Syringomyelia	8,4
Cutaneous lymphoma	8,3
Rett syndrome	8,2
Acute promyelocytic leukemia	8
Deafness, autosomal dominant, nonsyndromic, sensorineural, type DFNA	8
Esophageal carcinoma	8
Polyarthritis, rheumatic factor-negative	8
Cataract, total, congenital	7,9
Hyperlipidemia type 3	7,8
Hemophilia	7,7
Acute lymphoblastic leukemia	7,5
Immunodeficiency, common variable	7,5
Microscopic polyangiitis	7,5
Beckwith-Wiedemann syndrome	7,3
Pulmonary valve stenosis, congenital	7,2
Oculocutaneous albinism	7,15
Acute non lymphoblastic leukemia	7
Cerebellar ataxia, autosomal recessive	7
Cystathioninuria	7
Facio-scapulo-humeral muscular dystrophy	7
Holoprosencephaly	7
Primary sclerosing cholangitis	7
Thyroid carcinoma, medullary	7
Fryns syndrome	7**
Sotos syndrome	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
Hemiplegic migraine, familial	6,5
Leber hereditary optic neuropathy	6,5
Osteogenesis imperfecta	6,5
Polycystic kidney disease, autosomal recessive	6,5
Smith-Lemli-Opitz syndrome	6,5
Ectopia lentis isolated	6,4
Juvenile idiopathic arthritis, systemic-onset	6,3
Systemic vasculitis	6,3
Huntington disease	6,2
Amyotrophic lateral sclerosis	6
Cerebral arteriovenous fistula	6
Digitotalar dysmorphism	6
Leukemia, chronic myeloid	6
Optic atrophy	6
Tibial muscular dystrophy	6
Treacher-Collins syndrome	6
Wilson disease	5,84
Arthritis-related enthesitis	5,7
Biliary atresia	5,6
Pendred syndrome	5,5
Retinoblastoma	5,4
Alzheimer disease, familial	5,3
Zollinger-Ellison syndrome	5,3

** Prevalence at birth

Cornelia de Lange syndrome	5,25
Familial adenomatous polyposis, autosomal dominant	5,25
Acromegaly	5
Epiphyseal dysplasia multiple	5
Fructose intolerance	5
Lung cancer, small cell	5
Muscular dystrophy, Duchenne and Becker types	5
Osteosarcoma	5
Parietal foramina	5
Primary ciliary dyskinesia	5
Rheumatic fever	5
Spastic paraplegia, familial	5
Supranuclear palsy, progressive	5
Thomsen and Becker disease	5
Tricuspid atresia	5
Hydrolethalus	5**
Tritanopia	4,8
Acrocephalosyndactyly	4,6
Monosomy 5p	4,6
Multiple system atrophy	4,6
Achondroplasia	4,5
Congenital lobar emphysema	4,5
Retinoschisis, X-linked	4,5
Steinert myotonic dystrophy	4,5
Best disease	4,4
Cataract anterior polar	4,4
Chronic inflammatory demyelinating polyneuropathy	4,4
Polyarthritis, rheumatoid factor-positive	4,2
Psoriatic arthritis, juvenile form	4,2
Hemimelia	4,15
Adrenal hypoplasia congenital, X-linked	4
Autoimmune polyendocrinopathy, type 1	4
Ceroid lipofuscinosis, neuronal	4
Corticobasal degeneration	4
Exostoses, multiple	4
Hartnup syndrome	4
Histidinemia	4
Idiopathic hypersomnia	4
Phenylketonuria	4
Smith-Magenis syndrome	4
Amniotic bands	4**
Meckel 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
Waardenburg syndrome type 1	3,75
West syndrome	3,7**

Adrenoleukodystrophy, X-linked	3,5
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
Usher syndrome	3,5
Thanatophoric dwarfism	3,5**
Graft versus host disease	3,4
Choroidal dystrophy, central areolar	3,33
Hypochondroplasia	3,3
Multiple endocrine neoplasia, type 2	3,3
Parsonage-Turner syndrome	3,3
Systemic mastocytosis	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
Nodular regenerative hyperplasia of the liver	3
Opitz BBB/G syndrome	3
Proximal spinal muscular atrophy	3
Pseudoachondroplasia	3
Saethre-Chotzen syndrome	3
Kennedy disease	2,8
Leigh disease	2,75**
Proximal spinal muscular atrophy, type 2	2,6
Proximal spinal muscular atrophy, type 3	2,6
Waldenström macroglobulinemia	2,6
Achromatopsia	2,5
Amaurosis congenita of Leber	2,5
Behcet disease	2,5
BOR syndrome	2,5
Bullous pemphigoid	2,5
Cone rod dystrophy	2,5
Diffuse palmoplantar keratoderma, Norrbotten dominant type	2,5
Epidermolysis bullosa, epidermolytic	2,5
Friedreich ataxia	2,5
Fructose-1,6-bisphosphatase deficiency	2,5
Heterotaxia	2,5
Kartagener syndrome	2,5
Pseudoxanthoma elasticum	2,5
Restrictive cardiomyopathy, idiopathic or familial	2,5
Niemann-Pick disease	2,5**
Meconium aspiration syndrome	2,44
Waardenburg syndrome	2,4
3-methylcrotonylglycinuria	2,25
Ondine syndrome	2,25

** Prevalence at birth

Peutz-Jeghers syndrome	2,2
Cerebellar ataxia, autosomal dominant	2,15
GRACILE syndrome	2,12**
Alport syndrome	2
Choroideremia	2
Coats disease	2
Crouzon disease	2
Giant pigmented hairy nevus	2
Klippel-Feil syndrome	2
Langerhans cell histiocytosis	2
Nail-patella syndrome	2
Ocular albinism X-linked, recessive	2
Persistent hyperinsulinemic hypoglycemia of infancy	2
Poland anomaly	2
Sarcosinemia	2
Thyrotoxic periodic paralysis	2
Van Der Woude syndrome	2
Sternal cleft	<2
Wolf-Hirschhorn syndrome	2**
Lateral body wall complex	2**
Gamma-sarcoglycanopathy	1,96
Gastrointestinal stromal tumor	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
Darier disease	1,5
Emery-Dreifuss muscular dystrophy	1,5
Femur-fibula-ulna complex	1,5
Laryngotracheoesophageal cleft	1,5
Multifocal motor neuropathy with conduction block	1,5
Primary lateral sclerosis	1,5
Pulmonary arterial hypertension	1,5
Severe combined immunodeficiency T- B+, X-linked	1,5
CDG syndrome	1,5**
Alagille syndrome	1,4
Cat-eye syndrome	1,35
Netherton disease	1,35
Cryoglobulinemia	1,33*
Mucopolysaccharidosis type 1	1,3
Apert syndrome	1,25
Maternal hyperphenylalaninemia	1,25
Adult Onset Still's disease	1,23
Orofaciodigital syndrome, type 1	1,2
Pemphigus superficial	1,2
Kabuki syndrome	1,16
Glycogen storage disease, type 2	1,1
Mucopolysaccharidosis type 3	1,1
Split hand - split foot	1,1
Zellweger syndrome	1,1
Nephronophthisis, autosomal recessive	1,05
Cutis verticis gyrata - mental deficiency	1,02
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency	1
Acanthamoeba keratitis	1
Adrenocortical carcinoma	1
Albers-Schonberg disease	1
Angioneurotic edema	1
Ataxia telangiectasia	1
Blue cone monochromatism	1
Chondrodysplasia punctata, rhizomelic type	1
Chronic hiccup	1
Churg-Strauss syndrome	1
Clouston syndrome	1
Coloboma, ocular	1
Dyserythropoietic anemia, congenital	1
Ehlers-Danlos syndrome, type 4	1
Fanconi anaemia	1
Gaucher disease	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
Neuroendocrine tumor	1
Oculopharyngeal muscular dystrophy	1
Rubinstein-Taybi syndrome	1
Congenital rubella syndrome	1**
Sirenomelia	1**
Tracheal agenesis	1**
Acalvaria	<1**
Gaucher disease, type 1	0,94
Lewis-Sumner syndrome	0,9
MERRF syndrome	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
Congenital myasthenic syndromes	0,75
Cutaneous mastocytosis	0,75
Ebstein anomaly	0,75

** Prevalence at birth

Hyperkalemic periodic paralysis	0,75
Sandhoff disease	0,75
Niemann-Pick disease, type B	0,75**
Osteopetrosis, malignant	0,75**
Krabbe disease	0,75**
Albright hereditary osteodystrophy	0,72
Carbamoylphosphate synthetase deficiency	0,7
Menkes syndrome	0,7
46,XY disorders of sex development due to 17-beta-hydroxysteroid dehydrogenase deficiency	0,68
Kallmann syndrome	0,66
Hepatitis, chronic autoimmune	0,65
Goodpasture syndrome	0,64
Deafness, X-linked, non syndromic, sensorineural, type DFN	0,6
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 hemoglobinuria	0,55
Muscular dystrophy Fukuyama type	0,54
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
Bullous ichthyosiform - erythroderma congenita	0,4
Early onset torsion dystonia	0,4
Glutaryl-CoA dehydrogenase deficiency	0,4
Homocystinuria due to cystathionine beta-synthase deficiency	0,4
Mucopolysaccharidosis type 4	0,4
Pyruvate kinase deficiency	0,4
Sjögren-Larsson syndrome	0,4
Lesch-Nyhan syndrome	0,38
Pfeiffer syndrome	0,38
Christ-Siemens-Touraine syndrome	0,35
Severe combined immunodeficiency T- B-	0,35
Campomelic dysplasia	0,35
Spondylometaphyseal dysplasia	0,34
Sialidosis type 1	>0,33
Neutropenia severe congenital	0,33**
Blackfan-Diamond disease	0,32
Proximal spinal muscular atrophy, type 4	0,32
Alkaptonuria	0,3
Dopa-responsive dystonia	0,3
Lissencephaly type 1, due to LIS 1 anomalies	0,3
Muscular dystrophy congenital, merosin negative	0,3
Pterygium popliteal syndrome, autosomal dominant	0,3
Pure autonomic failure	0,3
Transmissible spongiform encephalopathies	0,3
Dentatorubral pallidoluysian atrophy	<0,3
Tay-Sachs disease	0,3**
Wolman disease	0,28**
Epidermolysis bullosa, dystrophic	0,27
Proximal spinal muscular atrophy, type 1	0,26
Factor VII deficiency	0,25
Lipodystrophy, Berardinelli type	0,25
Papillon-Lefevre syndrome	0,25
Pelizaeus-Merzbacher disease	0,25
Piebaldism	0,25
Niemann-Pick disease, type A	0,25**
Progeria	0,25**
Leptospirosis	0,24
Severe combined immunodeficiency due to adenosine deaminase deficiency	0,22
Von Hippel-Lindau disease	0,2
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
Nanism due to growth hormone resistance	0,2
Unverricht-Lundborg 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
Muscular dystrophy congenital, merosin-positive	0,15
Polycystic lipomembranous osteodysplasia - sclerosing leukoencephalopathy	0,15
Wiskott-Aldrich syndrome	0,15
Mucolipidosis type 2	0,15**

** Prevalence at birth

CHARGE association	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	0,11
Alpha-mannosidosis	0,1
Creutzfeldt-Jakob disease	0,1
Diffuse leiomyomatosis with Alport syndrome	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
Familial cold urticaria	0,1
Hemophilia, acquired	0,1
Lymphangioleiomyomatosis	0,1
Pulmonary alveolar proteinosis	0,1
Refsum disease	0,1
Senior-Loken syndrome	0,1
X-linked lymphoproliferative disease	0,1

Leprechaunism	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 syndrome	0,05
Tyrosinemia type 1	0,05
Hyper-IGM syndrome, autosomal recessive	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
Herpes simplex encephalitis	0,021*
Sialidosis type 1	0,02**
Sialidosis type 2	0,02**
Perinatal-lethal Gaucher disease	0,01
Gaucher disease, type 2	0,01
Refsum disease, infantile form	0,005

** Prevalence at birth

List of diseases by reported number of published cases

Diseases without prevalence data available. Report on number of published cases

Disease name	Number of published cases or families
Trichinellosis	6500 cases
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
Familial dysautonomia	550 cases
Western equine encephalitis	>600 cases
Enchondromatosis	> 600 cases
Adamantinoma	513 cases
True hermaphrodism	>500 cases
Aicardi syndrome	500 cases
CADASIL syndrome	500 cases
Malakoplasia	500 cases
Epidermal nevus syndrome	>400 cases
Castleman disease	400 cases
Silver-Russell dwarfism	400 cases
4-hydroxybutyricaciduria	350 cases
Leukocyte adhesion deficiency	<350 cases
CDG syndrome type Ia	> 300 cases
Alexander disease	300 cases
Alström syndrome	300 cases
Cutis marmorata telangiectatica congenita	300 cases
Gräsbeck-Imerslund disease	300 cases
Melorheostosis	300 cases
Methylmalonic acidemia - homocystinuria	300 cases
Moebius syndrome	300 cases
Norrie disease	300 cases
Rothmund-Thomson syndrome	300 cases
Lipoid proteinosis	>280 cases
Chronic recurrent multifocal osteomyelitis, juvenile	>260 cases
Caroli's disease	<250 cases
Focal dermal hypoplasia	200-300 cases
Lipodystrophy, familial partial, Dunnigan type	200-300 cases
Eosinophilic gastroenteritis	280 cases
Job syndrome	250 cases
Lipodystrophy, partial acquired	250 cases
Pulmonary haemosiderosis, primary	250 cases
6-pyruvoyl-tetrahydropterin synthase deficiency	248 cases
Oculo-dento-osseous dysplasia, autosomal dominant	243 cases
Pachyonychia congenita	230 cases
Kearns-Sayre syndrome	223 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
Pseudoarthrosis of clavicle, congenital	>200 cases
Tufted angioma	>200 cases
Adenosine monophosphate deaminase deficiency	200 cases
Camurati-Engelmann disease	200 cases
Cockayne syndrome	200 cases
Cogan syndrome	200 cases
Costello syndrome	200 cases
Crigler-Najjar syndrome	200 cases
Eosinophilic fasciitis	200 cases
Glucose-galactose malabsorption	200 cases
Gorham-Stout disease	200 cases
Kimura disease	200 cases
Multiple pterygium syndrome, lethal form	200 cases
Shwachman-Diamond syndrome	200 cases
Subcorneal pustular dermatosis	200 cases
Deletion 18p	<200 cases
Persistent Mullerian duct syndrome	<200 cases
Hyperimmunoglobinemia D with recurrent fever	180 cases
Erdheim-Chester disease	178 cases
Kasabach-Merritt syndrome	>175 cases
Celiac disease - epilepsy - occipital calcifications	170 cases
Alpha thalassemia-mental retardation, X-linked	168 cases
Carney complex	160 cases
McCune-Albright syndrome	158 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
Jacobsen syndrome	150 cases
Xanthinuria	150 cases
Arthrogryposis - renal dysfunction - cholestasis	<150 cases
Inflammatory pseudotumor of the liver	143 cases
Regional odeontodysplasia	139 cases

Dihydropteridine reductase deficiency	134 cases
Aorto-ventricular tunnel	130 cases
Nijmegen-breakage syndrome	130 cases
Weill-Marchesani syndrome	128 cases
Vulvovaginal gingival syndrome	127 cases
Marinesco-Sjogren syndrome	100-200 cases
Proteus syndrome	100-200
Eosinophilic pneumonia, acute idiopathic	>100 cases
Achondrogenesis	>100 cases
Bloom syndrome	>100 cases
Carnitine palmitoyl transferase 2 deficiency	>100 cases
Central neurocytoma	>100 cases
Cutis laxa	>100 cases
Fetal varicella syndrome	>100 cases
KID syndrome	>100 cases
Molybdenum cofactor deficiency	>100 cases
Mucolipidosis type 4	>100 cases
Ochoa syndrome	>100 cases
Simpson-Golabi-Behmel syndrome	>100 cases
Tricho-rhino-phalangeal syndrome type 1	>100 cases
Trichorhinophalangeal syndrome, type 1 and 3	>100 cases
Pulmonary lymphangiectasia, congenital	>100 cases
Rasmussen subacute encephalitis	>100 cases
Helicoid peripapillary chorioretinal degeneration	100 cases
Autoimmune lymphoproliferative syndrome	100 cases
Bernard-Soulier syndrome	100 cases
Bethlem myopathy	100 cases
Chaotic atrial tachycardia	100 cases
CINCA syndrome	100 cases
Cohen syndrome	100 cases
Congenital cataracts - facial dysmorphism - neuropathy	100 cases
Defective expression of HLA class 2	100 cases
Epidermolysis bullosa, acquired	100 cases
Freeman-Sheldon syndrome	100 cases
Fucosidosis	100 cases
Greig syndrome	100 cases
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	100 cases
Internal carotid agenesis	100 cases
Larsen syndrome	100 cases
Neonatal hemochromatosis	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
Robinow syndrome, dominant form	100 cases
Schwartz-Jampel syndrome	100 cases
Seckel syndrome	100 cases

Triple A syndrome	100 cases
Cataract congenital, Volkmann type	<100 cases
Hallermann-Streiff-Francois syndrome	<100 cases
Hypertrichosis lanuginosa congenita	<100 cases
Hypocomplementaemic leucocytoclasic vasculitis	<100 cases
Ichthyosis congenita, harlequin type	<100 cases
Mowat-Wilson syndrome	<100 cases
Susac syndrome	<100 cases
Tracheo-bronchomegaly	<100 cases
Tyrosinemia type 2	<100 cases
Ullrich, congenital muscular dystrophy	<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
Uhl anomaly	84 cases
Acquired generalized lipodystrophy	80 cases
LCAT deficiency	80 cases
Wells syndrome	80 cases
Rapp-Hodgkin syndrome	72 cases
LEOPARD syndrome	>70 cases
Tangier disease	>70 cases
Bullous systemic lupus erythematosus	70 cases
Craniometaphyseal dysplasia	70 cases
Midline cleft of lower lip	70 cases
Myoneurogastrointestinal encephalopathy syndrome	70 cases
Pseudohypoaldosteronism type 1	70 cases
Robinow syndrome, recessive form	70 cases
Diffuse neonatal haemangiomatosis	<70 cases
Glutathione synthetase deficiency	65 cases
Hyperferritinemia, hereditary, with congenital cataracts	>64 cases
Griscelli disease	60 cases
Pemphigus paraneoplastic	>60 cases
Cerebro-costo-mandibular syndrome	60 cases
Dyggve-Melchior-Clausen disease	60 cases
Hypertrichosis lanuginosa, acquired	60 cases
Neu laxova syndrome	50-60 cases
Ketoacidosis due to betaketothiolase deficiency	60 cases
Metatropic dwarfism	60 cases
Non-distal trisomy 10p	60 cases
Pancreatoblastoma	60 cases
Pearson syndrome	60 cases
Toriello-Carey syndrome	60 cases
Double uterus - hemivagina - renal agenesis	<60 cases
Ataxia, autosomal recessive, Beauce type	57 cases
Capillary leak syndrome	57 cases
Hennekam syndrome	>50 cases
Osteodysplasty, Melnick-Needles type	>50 cases
Ring chromosome 20	>50 cases
Acromesomelic dysplasia, Maroteaux type	50 cases

Acroosteolysis, autosomal dominant	50 cases
Adenylosuccinate lyase deficiency	50 cases
Ascher syndrome	50 cases
Benign paroxysmal torticolis of infancy	50 cases
Focal myositis	50 cases
Fronto-temporal dementia and Parkinsonism linked to chromosome 17 (FTDP-17)	50 cases
Glucocorticoid deficiency, familial	50 cases
Glucosephosphate isomerase deficiency	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
Sulfite oxidase deficiency	50 cases
Triple H syndrome	50 cases
Waardenburg-Shah syndrome	50 cases
XY gonadal agenesis	50 cases
Ehrlichiosis	<50 cases
Aglossia - adactylia	<50 cases
Blepharo-cheilo-dontic syndrome	<50 cases
Branchio-oculo-facial syndrome	<50 cases
Cholesteryl ester storage disease	<50 cases
DOOR syndrome	<50 cases
Fibular aplasia - ectrodactyly	<50 cases
Goldmann-Favre syndrome	<50 cases
Granulomatous slack skin	<50 cases
Midas syndrome	<50 cases
Oculo-digito-esophageal-duodenal syndrome (ODED)	<50 cases
Shprintzen-Goldberg syndrome	<50 cases
Cholestasis - lymphoedema, syndrome	20-50 cases
Corticosteroid-sensitive aseptic abscesses	49 cases
Pityriasis rubra pilaris	48 cases
Coxo-podo-patellar syndrome	47 cases
Mohr-Tranebjaerg syndrome	46 cases
Encephalo-cranio-cutaneous lipomatosis	45 cases
KBG syndrome	45 cases
Bowen-Conradi syndrome	44 cases
Ear-patella-short stature syndrome	42 cases
Juvenile hyaline fibromatosis	40-50 cases
Desbuquois syndrome	>40 cases
3M syndrome	40 cases
Babesiosis	40 cases
Carpenter syndrome	40 cases
Chylomicron retention disease	40 cases
Coffin-Siris syndrome	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
Mucopolysaccharidosis type 7	<40cases
Acromicric dysplasia	<40 cases
Progressive bulbar paralysis of childhood	<40 cases
Tracheobronchomegaly	<40 cases
Hypotrichosis simplex	38 cases
Mandibuloacral dysplasia	37 cases
Oculocerebrocutaneous syndrome	36 cases
Spondyloenchondrodysplasia	36 cases
Carnitine palmitoyl transferase 1 deficiency	35 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
Pyogenic arthritis - pyoderma gangrenosum - acne	34 cases
Schinzel-Giedion midface retraction syndrome	34 cases
Björnstadt syndrome	33 cases
Marshall-Smith syndrome	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
Duplication 8q	>30 cases
Trichodentoosseous syndrome	>30 cases
CDG syndrome type Ic	>30 cases
Agnathia - holoprosencephaly - situs inversus	30 cases
Aicardi-Goutieres syndrome	30 cases
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
Glycogen storage disease due to LAMP-2 deficiency	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
Scalp-ear-nipple syndrome	30 cases
SHORT syndrome	30 cases
Triose phosphate-isomerase deficiency	30 cases
Weaver syndrome	30 cases
Acropectorovertebral dysplasia	<30 cases
Congenital brain dysgenesis due to glutamine synthetase deficiency	<30 cases
Developmental delay due to 2-methylbutyryl-coA dehydrogenase deficiency	<30 cases
Encephalopathy due to hydroxykynureninuria	<30 cases
Fronto-metaphyseal dysplasia	<30 cases
Glycogen storage disease, type 7	<30 cases
Johnson neuroectodermal syndrome	<30 cases
Nasopalpebral lipoma - coloboma - telecanthus	<30 cases
Neurometabolic disorder due to serine deficiency	<30 cases
Obesity due to congenital leptin deficiency	<30 cases
Pontocerebellar hypoplasia type 2	<30 cases
Spontaneous periodic hypothermia	<30 cases
Wrinkly skin syndrome	<30 cases
Ectodermal dysplasia absent dermatoglyphics	<30 cases
Cone rod dystrophy - amelogenesis imperfecta	29 cases
Infant epilepsy with migrant focal crisis	29 cases
Mosaic variegated aneuploidy syndrome	29 cases
Orofaciodigital syndrome, type 6	29 cases
Symmetrical thalamic calcifications	29 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
3C syndrome	25 cases
Atelosteogenesis 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
Hepatic veno-occlusive disease - immunodeficiency	<25 cases
Bartsocas-Papas syndrome	24 cases
Distal myopathy, with early respiratory muscle involvement	24 cases
Synspondylism	24 cases
Congenital bronchobiliary fistula	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
Microcoria - congenital nephrosis	22 cases
Split hand - split foot - deafness	22 cases
Craniolenticulosutural dysplasia	21 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
Spondylocarpotarsal synostosis	>20 cases
Congenital indifference to pain	> 20 cases
3-methylglutaconic aciduria, type 1	20 cases
Acrorenal syndrome	20 cases
Arrhinia	20 cases
Calvarial doughnut lesions - bone fragility	20 cases
Craniofrontonasal syndrome, Teebi type	20 cases
Distal monosomy 8p	20 cases
Gray platelet syndrome	20 cases
Juvenile temporal arteritis	20 cases
Lacrimo-auriculo-dento-digital syndrome	20 cases
Lipoamide dehydrogenase deficiency	20 cases
PIBIDS syndrome	20 cases
CDG syndrome type Ib	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
Epidermolysis bullosa simplex - limb girdle muscular dystrophy	<20 cases
Hemorrhagic disorders due to collagen receptors deficiency	<20 cases
Ichthyosis bullosa of Siemens	<20 cases
IMAGe syndrome	<20 cases
Interstitial granulomatous dermatitis - arthritis	<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
Perlman syndrome	<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
Ectodermal dysplasia, «pure» hair-nail type	<20 cases
Keratosis palmaris et plantaris - clinodactyly	<20
Craniosynostosis, Boston type	19 cases
Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia	19 cases

Schopf-Schulz-Passarge syndrome	19 cases
Crisponi syndrome	18 cases
Hypertrichotic osteochondrodysplasia	18 cases
Terminal osseous dysplasia - pigmentary defects	18 cases
Aminopterin embryofetopathy	17 cases
Erythroderma lethal, congenital	17 cases
Folate malabsorption, hereditary	17 cases
GTP cyclohydrolase I deficiency	17 cases
Hypokeratosis, palmo-plantar, circumscribed	17 cases
Malonic aciduria	17 cases
Mental retardation X-linked - seizures - short stature - midface hypoplasia	17 cases
Wolcott-Rallison syndrome	17 cases
Acromegaly - cutis verticis gyrata - corneal leukoma	16 cases
Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis	16 cases
Glycogen storage, type 0	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
Orbital leiomyoma	16 cases
Orofaciodigital syndrome, type 4	16 cases
Ablepharon-macrostomia syndrome	15 cases
Angel-shaped phalango-epiphyseal dysplasia	15 cases
Cranio-ectodermal dysplasia	15 cases
IBIDS syndrome	15 cases
Myhre syndrome	15 cases
Umbilical cord ulceration - intestinal atresia	15 cases
Congenital anosmia, isolated	<15 cases
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	<15 cases
Odontoonychodermal dysplasia	<15 cases
Retinopathy pigmentary - mental retardation	<15 cases
Taurodontia - absent teeth - sparse hair	<15 cases
Wilson-Turner syndrome	>14 cases
Muscular dystrophy, limb girdle, autosomal recessive, type 2G	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
Dermoodontodysplasia	14 cases
Fingerprints absence - syndactyly milia	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
Diaphragmatic hernia - exomphalos - corpus callosum agenesis	13 cases
Hypotelorism - cleft palate - hypospadias	13 cases
Posterior column ataxia - retinitis pigmentosa	13 cases
Hemolytic anemia due to adenylate kinase deficiency	12 cases
Acropectororenal field defect	12 cases
Alopecia - epilepsy - pyorrhea - mental subnormality	12 cases
Ataxia - deafness - optic atrophy, lethal	12 cases
Atelosteogenesis I	12 cases
Atelosteogenesis III	12 cases
Atrial tachyarrhythmia with short PR interval	12 cases
Coloboma of macula - brachydactyly type B	12 cases
Coloboma uveal - cleft lip palate - mental retardation	12 cases
Distal myopathy with vocal cord weakness	12 cases
Distal myopathy, with posterior leg and anterior upper limb involvement	12 cases
Dopamine beta-hydroxylase, deficiency of	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
Bickel-Fanconi glycogenosis	112 cases
Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges	11 cases
Atrial septal defect - atrioventricular conduction	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
Mental retardation, X-linked, Snyder type	11 cases
Necrotizing encephalopathy, acute, autosomal dominant	11 cases
PELVIS syndrome	11 cases
Pterygium syndrome, antecubital	11 cases
Tricho-megaly - retina pigmentary degeneration - dwarfism	11 cases
CHAND syndrome	>10 cases
Palmoplantar porokeratosis of Mantoux	>10 cases
Chediak-Higashi syndrome	>10 cases
Median cleft of the upper lip - corpus callosum lipoma - cutaneous polyps	>10 cases
Diffuse palmoplantar keratoderma - acrocyanosis	10 cases
Acromesomelic dysplasia Hunter-Thompson type	10 cases
Albright like syndrome	10 cases
Ambras syndrome	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
Choroidocerebral calcification syndrome, infantile form	10 cases
Colobomatous - microphthalmia - heart disease - hearing loss	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 syndrome	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 behavior	10 cases
Mental retardation, X-linked, syndromic 7	10 cases
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus	10 cases
Nevo syndrome	10 cases
Pollitt syndrome	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
Neurodegenerative syndrome, X-linked, Hamel type	10 cases
Aase-Smith syndrome	<10 cases
Acro-facial dysostosis, Rodriguez type	<10 cases
Acromelanosis	<10 cases
Astley-Kendall dysplasia	<10 cases
Carpotarsal osteochondromatosis	<10 cases
Carpotarsalosteolysis, autosomal recessive	<10 cases
Cerebral gigantism - jaw cysts	<10 cases
Corneal dystrophy - perceptive deafness	<10 cases
Deafness - lymphoedema - leukemia	<10 cases
Diaphanospondylodysostosis	<10 cases
Digito-reno-cerebral syndrome	<10 cases
Glomerulonephritis - sparse hair - telangiectases	<10 cases
Greenberg dysplasia	<10 cases
Hypopituitarism - microphthalmia	<10 cases
Myoclonus hereditary - progressive distal muscular atrophy	<10 cases
Pacman dysplasia	<10 cases
Pancreatic hypoplasia - diabetes - heart disease	<10 cases
Pfeiffer-Singer-Zschiesche syndrome	<10 cases
Phosphoenolpyruvate carboxykinase (PEPCK) deficiency	<10 cases
Progeria - short stature - pigmented nevi	<10 cases
Pseudo-Gaucher disease	<10 cases
Pseudo-Zellweger syndrome	<10 cases
Singleton-Merten dysplasia	<10 cases
Xk aprosencephaly	<10 cases
Atransferrinemia	9 cases
Bosley-Salih-Alorainy syndrome	9 cases
Brachymorphism - onychodysplasia - dysphalangism	9 cases
Cardiomyopathy - cataract - hip spine disease	9 cases
Gamma-glutamylcysteine synthetase deficiency	9 cases
Guanidinoacetate methyltransferase deficiency	9 cases
Macrostomia - preauricular tags - external ophthalmoplegia	9 cases
Mental retardation, X-linked, Shashi type	9 cases
Microdontia - type I microtia - deafness	9 cases
Oculocerebrofacial syndrome, Kaufman type	9 cases
Tricho-retino-dento-digital syndrome	9 cases
Ataxia - apraxia - mental retardation, X-linked	9 cases
5-oxoprolinase deficiency	8 cases
Ackerman syndrome	8 cases
Ankylosing vertebral hyperostosis - tylosis	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
Mental retardation X-linked - dysmorphism	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
Trigonocephaly - ptosis coloboma	8 cases
Rubinstein-Taybi like, syndrome	8 cases
Acrorenalmandibular 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
Lenz-Majewski hyperostotic dwarfism	7 cases
MEHMO syndrome	7 cases
Mental retardation, X-linked severe, Gustavson type	7 cases

Michels syndrome	7 cases
Multiple fibrofolliculoma familial	7 cases
Myopathy - lactic acidosis - sideroblastic anemia	7 cases
Neutropenia, severe congenital, X-linked	7 cases
Obesity due to pro-opiomelanocortin deficiency	7 cases
Retinal degeneration - nanophthalmos - glaucoma	7 cases
Retino-hepato-endocrinologic syndrome	7 cases
Stern-Lubinsky-Durrie syndrome	7 cases
Torticollis - keloids - cryptorchidism - renal dysplasia	7 cases
MEHMO syndrome	7 cases
Neurodegenerative syndrome, X-linked, Bertini type	7 cases
Mental retardation - microcephaly - phalangeal - facial abnormalities	6 cases
3-hydroxy 3-methylglutaryl-CoA synthase	6 cases
Acro-facial dysostosis, Catania form	6 cases
Agonadism - dextrocardia - diaphragmatic hernia	6 cases
Aplasia cutis congenita of limbs recessive	6 cases
Biliary malformation - renal tubular insufficiency	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
Eiken syndrome	6 cases
Epilepsy - telangiectasia	6 cases
Fanconi - ichthyosis - dysmorphism	6 cases
Grange syndrome	6 cases
Hidrotic ectodermal dysplasia, Christianson-Fourie type	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
Symphalangism with multiple anomalies of hands and feet	6 cases
Thumb stiff - brachydactyly - mental retardation	6 cases
W syndrome	6 cases
Wieacker-Wolff syndrome	6 cases
Zunich-Kaye syndrome	6 cases
Branchial arch syndrome, X-linked	5 à 7 cases
Cleft palate - cardiac defect - genital anomalies - ectrodactyly	5 cases
Lissencephaly type III - familial fetal akinesia sequence	5 cases
Lymphoedema - cerebral arteriovenous anomaly	5 cases
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
Anonychia - microcephaly	5 cases
Arachnodactyly - ossification abnormal - mental retardation	5 cases
Auro-cephalo-syndactyly	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 Ih	5 cases
Choanal atresia - deafness - cardiac defects dysmorphism	5 cases
Craniosynostosis - brachydactyly	5 cases
Curry-Jones syndrome	5 cases
Cutis verticis gyrata - thyroid aplasia - mental retardation	5 cases
Dacryocystitis osteopoikilosis	5 cases
Depigmentation of the iris, acute, bilateral	5 cases
Dermatoosteolysis, Kirghizian type	5 cases
Ectrodactyly - ectodermal dysplasia	5 cases
Fine-Lubinsky syndrome	5 cases
German syndrome	5 cases
Glaucoma-sleep apnea	5 cases
Ichthyosis - male hypogonadism	5 cases
Matthew-Wood syndrome	5 cases
Mental retardation - cubitus valgus - unusual facies	5 cases
Mental retardation, choreoathesis and abnormal behavior	5 cases
Neonatal death - immune deficiency	5 cases
Nephropathy - deafness - hyperparathyroidism	5 cases
Nephrosis - deafness - urinary tract and digital malformations	5 cases
Oculo-dento-osseous dysplasia, autosomal recessive	5 cases
Oculo-palato-cerebral syndrome	5 cases
Odonto-micronychial 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
Sacral hemangiomas - multiple congenital abnormalities	5 cases
Sillence syndrome	5 cases
Spinocerebellar ataxia X-linked, type 3	5 cases
CDG syndrome type Id	5 cases
Keratoderma - epithelioma - dental abnormalities-hypogonadism	5 cases
Mental retardation, X-linked, Lubs type	5 cases

Cystic hamartoma of lung and kidney	<5 cases
Lathosterolosis	<5 cases
Sakati-Nyhan syndrome	<5 cases
Palmoplantar keratoderma - amyotrophy	4 cases
Rolled and spiral hairs - palmoplantar keratoderma	4 cases
Abruzzo-Erickson syndrome	4 cases
Acro-facial 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
Benign exophthalmos syndrome	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 type	4 cases
Caudal dysgenesis familial type	4 cases
CDG syndrome type If	4 cases
Cholestasis - pigmentary retinopathy - cleft palate	4 cases
Chondrodysplasia lethal, recessive	4 cases
Choroideremia - deafness - obesity	4 cases
Cleft lip palate - malrotation - cardiopathy	4 cases
Coxo-auricular syndrome	4 cases
Craniofaciocardioskeletal syndrome	4 cases
Cranioosteoarthropathy	4 cases
Craniosynostosis - Dandy Walker hydrocephalus	4 cases
Deafness - peripheral neuropathy - arterial disease	4 cases
Diaphragmatic defect - limb deficiency - skull defect	4 cases
Duker-Weiss-Siber syndrome	4 cases
Ectodermal dysplasia, Berlin type	4 cases
Ectopia lentis - chorioretinal dystrophy - myopia	4 cases
Facial onset sensory and motor neuronopathy	4 cases
Gaucher - ichthyosis - restrictive dermopathy	4 cases
Gombo syndrome	4 cases
Gorlin-Chaudhry-Moss, syndrome	4 cases
Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate	4 cases
Growth retardation - microcephaly - digital abnormalities - hypospadias	4 cases
Hidrotic ectodermal dysplasia, Halal type	4 cases
Homocarnosinosis	4 cases
Hypogammaglobulinemia due to CD19 deficiency	4 cases
Hypomandibular facio-cranial dysostosis	4 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodonti	4 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 cases
Hypotrichosis - lymphedema - telangiectasia	4 cases
Ichthyosis - alopecia - 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
Liver disease - retitinitis pigmentosa - polyneuropathy - epilepsy	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 - 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 acroschyphodysplasia	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
Oculo-oto-facial dysplasia	4 cases
Odontotrichomelic syndrome	4 cases
Orofaciodigital syndrome, type 5	4 cases
Osteopoikilosis - short stature - intellectual deficit	4 cases
Pancreatic and cerebellar agenesis	4 cases
Paraplegia - mental retardation - hyperkeratosis	4 cases
Recurrent infections - short stature - hypopigmentation - coarse face	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 - combined immunodeficiency	4 cases
Trichoodontoonychial dysplasia	4 cases
CDG syndrome type IIa	4 cases
CDG syndrome type Ik	4 cases
Mental retardation, X-linked - hypogonadism - ichthyosis - obesity - short stature	4 cases
Neuroectodermal endocrine syndrome	4 cases
Cranio-digital syndrome - mental retardation	4 cases
Arachnodactyly - mental retardation - dysmorphism	3 cases
Lymphoedema - atrial septal defects - facial changes	3 cases
Acromesomelic dysplasia, Brahimi-Bacha type	3 cases

Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis	3 cases
Amelia, autosomal recessive	3 cases
Aniridia - absent patella	3 cases
Aniridia - ptosis - mental retardation - obesity, familial type	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
Aredyld syndrome	3 cases
Axenfeld-Rieger anomaly - hydrocephaly - skeletal abnormalities	3 cases
Blepharoptosis - myopia - ectopia lentis	3 cases
Bone sclerosing - dysplasia - ichthyosis - premature ovarian failure	3 cases
Brachio-skeleto-genital syndrome	3 cases
Buttiens-Fryns syndrome	3 cases
Cataract - deafness - hypogonadism	3 cases
Cervical hypertrichosis - peripheral neuropathy	3 cases
Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	3 cases
Cortical blindness - mental retardation - polydactyly	3 cases
Cranio-facial 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
Disorder of sex development - mental retardation	3 cases
Ectodermal dysplasia, hypohidrotic - hypothyroidism - ciliary dyskinesia	3 cases
Ectodermic dysplasia - hypothyroidism - cleft	3 cases
Eyebrow duplication syndactyly	3 cases
Facial dysmorphism - macrocephaly - myopia - Dandy Walker	3 cases
Faciocardiomelic dysplasia, lethal	3 cases
Gardner-Morrison-Abbot syndrome	3 cases
Gemss syndrome	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
Hypogonadism - retinitis pigmentosa	3 cases
Lewis-Pashayan syndrome	3 cases
Lipodystrophy - mental retardation - deafness	3 cases
Lumbosacral vertebrae, posterior fusion of - blepharoptosis	3 cases
Mental retardation - hypoplastic corpus callosum - preauricular tag	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
Microcephaly - cardiomyopathy	3 cases
Microcephaly - cleft palate	3 cases
Microcytic anemia - liver iron overload - low ferritinemia	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
Orofaciodigital syndrome, type 3	3 cases
Osteogenesis imperfecta congenita - microcephaly - cataracts	3 cases
Osteoporosis oculo-cutaneous 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
Renal dysplasia, multicystic	3 cases
Seizures - intellectual deficit due to hydroxylysinuria	3 cases
Sensorineural hearing loss - early greying - essential tremor	3 cases
Short stature - mental retardation - eye anomalies - cleft lip palat	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
Wittwer syndrome	3 cases
Mental retardation, X-linked - hypogammaglobulinemia - progressive neurological deterioration	3 cases
Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia	3 cases
Lissencephaly type III - metacarpal bone dysplasia	2 cases
46,XX disorders of sex development - skeletal anomalies	2 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 malformation - congenital heart disease - postaxial polydactyly	2 cases
Camptodactyly syndrome, Guadalajara type 2	2 cases
Cardiomyopathy - renal anomalies	2 cases
Cataract - ataxia - deafness	2 cases
Cataract - microphthalmia - septal defect	2 cases
Cataract - nephropathy - encephalopathy	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
Congenital ichthyosis - microcephalus - quadriplegia	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
Craniosynostosis - fibular aplasia	2 cases
Craniosynostosis radial aplasia, Imaizumi type	2 cases
Craniotubular syndrome	2 cases
Cryptomicrotia - brachydactyly - excess fingertip arch	2 cases
Cystic hygroma lethal - cleft palate	2 cases
Dahlberg-Borer-Newcomer syndrome	2 cases
Dandy Walker - macrocephaly	2 cases
Dandy Walker malformation - postaxial polydactyly	2 cases
Deaf - blind - hypopigmentation	2 cases
Deafness - tubular acidosis - anemia	2 cases
Deafness - vitiligo - achalasia	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 - blindness	2 cases
Ectodermal dysplasia anhidrotic - immunodeficiency - osteopetrosis - lymphedema	2 cases
Eng-Strom syndrome	2 cases
Epilepsy - microcephaly - skeletal dysplasia	2 cases
Fuqua-Berkovitz syndrome	2 cases
Gamma aminobutyric acid transaminase deficiency	2 cases
Gonadal dysgenesis, XY type - associated anomalies	2 cases
Hec syndrome	2 cases
Hemolytic anemia, lethal - genital anomalies	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
Ichthyosis - hepatosplenomegaly - cerebellar degeneration	2 cases
Ichthyosis - oral and digital anomalies	2 cases
Ichthyosis congenita - biliary atresia	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
Kniest-like dysplasia, lethal	2 cases
Kudo-Tamura-Fuse syndrome	2 cases
Lichstenstein syndrome	2 cases
Low birth weight - dwarfism - dysgammaglobulinemia	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 - typical face	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 - 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 spastic quadriparesis	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
Oculo-tricho-dysplasia	2 cases
Okamoto syndrome	2 cases
Ossification anomalies - psychomotor development delay	2 cases
Osteogenesis imperfecta - retinopathy - seizures - intellectual deficit	2 cases
Osteopetrosis lethal	2 cases
PARC syndrome	2 cases
Pilodental dysplasia with refractive errors	2 cases
Plagiocephaly - mental retardation, X-linked	2 cases
Progressive neurodegeneration - joint laxity - cataract	2 cases
Pseudo-progeria syndrome	2 cases
Pterygium colli - intellectual deficit - digital anomalies	2 cases
Ptosis strabismus 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
Short stature, Brussels type	2 cases
Siegler-Brewer-Carey syndrome	2 cases
Small vessel disease of the brain, not NOTCH3-related	2 cases
Spastic quadriplegia - retinitis pigmentosa - mental retardation	2 cases
Spinal muscular atrophy - Dandy-Walker complex - cataracts	2 cases
Stimmler syndrome	2 cases
Talo-patello-scaphoid osteolysis	2 cases
Thyrocerebrorenal syndrome	2 cases
Trigonocephaly - bifid nose - acral anomalies	2 cases
Trigonocephaly - broad thumbs	2 cases
Tubular renal disease - cardiomyopathy	2 cases
Tyrosinemia type 3	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
CDG syndrome type IIe	2 cases
CDG syndrome type IL	2 cases
Heart defects - limb shortening	2 cases
Keratoderma - hypotrichosis - leukonychia	2 cases
Hypotrichosis-mental retardation lopes type	1 case
2,4-dienoyl-CoA reductase deficiency	1 case
46 XX gonadal dysgenesis - epibulbar dermoid	1 case
Acro-facial dysostosis postaxial, atypical	1 case
Acro-facial dysostosis, Preis type	1 case
Anophthalmia - short stature - obesity	1 case
Arthrogryposis - iugr thoracic - dystrophy	1 case
Blaichman syndrome	1 case
Bone dysplasia, Azouz type	1 case
Cataract - hypertrichosis - mental retardation	1 case
Chondrodysplasia - situs inversus- imperforate anus - polydactyly	1 case
Corneal crystals - myopathy - neuropathy	1 case
Corneal crystals myopathy neuropathy	1 case
Corneal dystrophy - ichthyosis - microcephaly - mental retardation	1 case
Ectodermal dysplasia - alopecia - preaxial polydactyly	1 case
Ectodermal dysplasia - arthrogryposis - diabetes mellitus	1 case
Ectodermal dysplasia - mental retardation - syndactyly	1 case
Enamel hypoplasia - cataract - hydrocephaly	1 case
Facial clefting - corpus callosum agenesis	1 case
Glossopalatine ankylosis - cataracts - digital anomalies	1 case
Hyaluronidase deficiency	1 case
Hydrops - ectrodactyly - syndactyly	1 case
Hypertrichosis - brachydactyly - obesity - mental retardation	1 case
Hypothyroidism - dermoid cyst - cleft palate	1 case
Ichthyosis - deafness - mental retardation - skeletal anomalies	1 case
Intrathoracic kidney vertebral fusion	1 case
Lissencephaly - immunodeficiency	1 case
Macroepiphyseal dysplasia, Mcalister coe type	1 case
Martinez-Monasterio-Pinheiro syndrome	1 case
Megaepiphyseal dwarfism	1 case
Mental retardation - multiple nevi	1 case
Metaphyseal dysplasia - hypertelorism - hypospadias	1 case
Microcephaly - syndactyly - brachymesophalangy	1 case
Molarization of anterior teeth - deafness	1 case
Multiple joint dislocations - metaphyseal dysplasia	1 case
Nasopharyngeal teratoma - Dandy Walker - diaphragmatic hernia	1 case
Nevus of ota - retinitis pigmentosa	1 case
Odonto-onycho-hypohidrotic dysplasia - midline scalp defects	1 case
Orofaciodigital syndrome, type 10	1 case

Osteochondrodysplasia thrombocytopenia hydrocephalus	1 case
Pancreatic lipomatosis - duodenal stenosis	1 case
Patella hypoplasia - skeletal malformations	1 case
Podder-Tolmie syndrome	1 case
Preauricular pits - renal disease	1 case
Richieri costa colletto otto syndrome	1 case
Rigid mask like face - deafness - polydactyly	1 case
Sclerosing bone dysplasia - mental retardation	1 case
Sclerosing bone dysplasia - mental retardation	1 case
Spondylo-costal dysostosis - Dandy Walker	1 case
Stoll-Alembik-Finck syndrome	1 case
Symphalangism - short stature - accessory testis	1 case
Symphalangism short stature accessory testis	1 case
Trichooculodermovertebral syndrome	1 case
Trichoonychohypohidrotic dysplasia	1 case
Bone dysplasia - corpus callosum agenesis	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
Li-Fraumeni syndrome	400 families
Birt-Hogg-Dube syndrome	>60 families
Nephropathy familial with gout	57 families
Nance-Horan syndrome	50 families
Muscular dystrophy, limb girdle, autosomal recessive, type 2I	>40 families
Granulomatous arthritis of childhood	40 families
Thiamine-responsive megaloblastic anemia syndrome	30 families
Erythermalgia, primary	30 families
Phosphoribosylpyrophosphate synthetase superactivity	<30 families
Neuropathy, giant axonal	> 20 families
Overhydrated hereditary stomatocytosis	> 20 families
Synostoses, multiple - brachydactyly	20 families
Fahr syndrome	<20 families
Acrorenoocular syndrome	<20 families
Platelet syndrome, familial	<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 leukemia	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
Porencephaly, familial	10 families
Renpenning syndrome	10 families
Acheiropodia	<10 families
Angioma hereditary neurocutaneous	<10 families
Focal facial dermal dysplasia	<10 families

Hypoparathyroidism familial isolated	<10 families
Keratoderma palmoplantar - deafness	<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
Lethal osteosclerotic bone dysplasia	8 families
Juvenile macular degeneration, hypotrichosis	7 families
EEM syndrome	7 families
Immune dysregulation - polyendocrinopathy - enteropathy, X linked	7 families
Cystoid macular dystrophy	6 families
Developmental dysphasia familial	6 families
Pontocerebellar hypoplasia type 1	6 families
Encephalopathy with neuroserpin inclusion bodies, familial form	>5 families
Anemia, 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
Familial rectal pain	4 families
IVIC syndrome	4 families
Triphalangeal thumbs - brachyectrodactyly	4 families
Adducted thumbs - arthrogryposis, Christian type	3 families
CAMFAK syndrome	3 families
Cataract-glaucoma	3 families
Cerebroretinal vasculopathy	3 families
Deafness-infertility syndrome	3 families
HERNS syndrome	3 families
Hypomagnesemia with hypocalciuria	3 families
Mental retardation, X-linked, with isolated growth hormone deficiency	3 families
Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss	1-2 families
Skeletal dysplasia - intellectual deficit	1-2 families
Mixed dystonias	3 families
Rapid-onset dystonia-parkinsonism	3 families
North Carolina macular dystrophy	2 families
Ankyloblepharon filiforme - imperforate anus	2 families
Bencze syndrome	2 families
Blepharonasofacial malformation syndrome	2 families
Bullous dystrophy macular type	2 families
Chorioretinal atrophy, progressive bifocal	2 families
Earlobes, thickened - conductive deafness	2 families

Ehlers-Danlos syndrome, type 5	2 families
Episodic ataxia, type 4	2 families
Fibromatosis, gingival - progressive deafness	2 families
Hypoparathyroidism X-linked	2 families
Jackson-Weiss syndrome	2 families
Paraplégie spastique - glaucome - déficit intellectuel	2 families
Retinitis pigmentosa - deafness - hypogenitalism	2 families
Orofaciodigital syndrome, type 8	1 ou 2 families
Albinism-deafness syndrome	1 family
Alopecia - congenita keratosis palmoplantaris	1 family
Aphalangia - syndactyly - microcephaly	1 family
Banki syndrome	1 family
Camptobrachydactyly	1 family
Cataract, Hutterite type	1 family
Congenital alopecia, X linked	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 neuromusculosqueletal syndrome	1 family
Dyschondrosteosis - nephritis	1 family
Ehlers-Danlos syndrome type 10	1 family
Episodic ataxia, type 3	1 family
Hereditary vascular retinopathy	1 family
Hereditary vascular retinopathy	1 family
Hydrocephalus - blue sclerae - nephropathy	1 family
Infundibulopelvic stenosis - multicystic kidney	1 family
Insulin resistance, short fifth metacarpals	1 family

Lipodystrophy, familial partial, due to AKT2 mutations	1 family
Mental retardation - progressive spasticity, X-linked	1 family
Mental retardation, X-linked recessive - macrocephaly - ciliary dysfunction	1 family
Mental retardation, X-linked, Cabezas type	1 family
Mental retardation, X-linked, Pai type	1 family
Muscular dystrophy, limb-girdle, autosomal dominant, type 1F	1 family
Muscular dystrophy, limb-girdle, autosomal dominant, type 1G	1 family
Oculodental syndrome rutherfurd syndrome	1 family
Oculogastrointestinal muscular dystrophy	1 family
Pili torti - onychodysplasia	1 family
Ptosis - strabismus - ectopic pupils	1 family
Schizophrenia - mental retardation - 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
Muscular dystrophy, limb-girdle, autosomal dominant, type 1A	1 family
Fried Syndrome	1 family
Mental retardation, X-linked - Spastic paraplegia with iron deposits	1 family