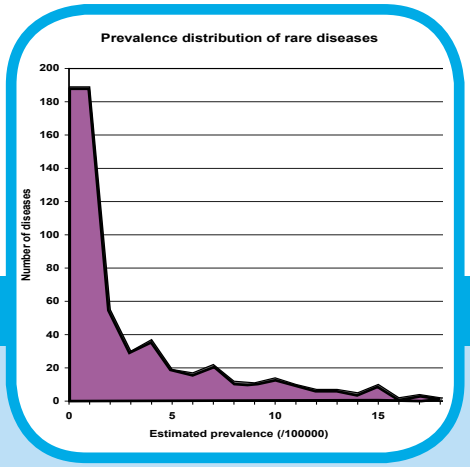

Orphanet Report Series

Rare Diseases collection

November 2011 | Number 1

Prevalence of rare diseases: Bibliographic data

Listed in alphabetical order of diseases

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Liberté • Égalité • Fraternité
RÉPUBLIQUE FRANÇAISE
MINISTÈRE DU TRAVAIL, DE L'EMPLOI ET DE LA SANTÉ

20102206

Methodology

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

Search strategy

The search strategy is carried out using several data sources:

- Websites: Orphanet, e-medicine, GeneClinics, EMA and OMIM ;

- Medline is consulted using the search algorithm:

«Disease names» AND Epidemiology[MeSH:NoExp] OR Incidence[Title/abstract] OR Prevalence[Title/abstract] OR Epidemiology[Title/abstract] ;

- Medical books, grey literature and reports from experts are also important sources of data.

Collected data

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

- For congenital diseases with birth-onset, prevalence = incidence at birth x (patient life expectancy/ general population life expectancy) ;

- For the other rare diseases, prevalence = incidence x rare disease mean duration ;

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

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

Updated Data

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

Limitation of the study

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

Prevalence or reported number of published cases listed in alphabetical order of diseases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
12q14 microdeletion syndrome		4 cases
15q24 microdeletion syndrome		4 cases
2,8 dihydroxyadenine urolithiasis	1.7	
2p21 microdeletion syndrome		7 cases
2q24 microdeletion syndrome		23 cases
2q37 microdeletion syndrome		10 cases
3C syndrome		25 cases
3-hydroxy 3-methylglutaryl-CoA synthase deficiency		6 cases
3M syndrome		40 cases
3-methylcrotonylglycinuria	2.25	
3-methylglutaconic aciduria type 1		20 cases
3-methylglutaconic aciduria type 3	10	
46,XX disorder of sex development - skeletal anomalies		2 cases
46,XX gonadal dysgenesis	< 10	
46,XY disorder of sex development - adrenal insufficiency		2 cases
46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	0.68	
46,XY gonadal dysgenesis - motor and sensory neuropathy		6 cases
4-hydroxybutyricaciduria		350 cases
5-oxoprolinase deficiency		8 cases
6-pyruvoyl-tetrahydropterin synthase deficiency		248 cases
6q terminal deletion		19 cases
8q22.1 microdeletion syndrome		4 cases
Aarskog-Scott syndrome		> 200 cases
Aase-Smith syndrome		< 10 cases
Ablepharon macrostomia syndrome		15 cases
Abruzzo-Erickson syndrome		4 cases
Absence of fingerprints - congenital milia		14 cases
Absent thumb - short stature - immunodeficiency		3 cases
Acalvaria	< 1**	
Acanthamoeba keratitis	1	
Acatalasemia	3.1	
Aceruloplasminemia	0.1	
Achalasia - microcephaly		5 cases
Acheiropodia		< 10 families

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Achondrogenesis		> 100 cases
Achondroplasia	4.5	
Achromatopsia	3.33	
Ackerman syndrome		8 cases
Aconitase deficiency		19 cases
Acquired epidermolysis bullosa		> 100 cases
Acquired generalized lipodystrophy		> 100 cases
Acquired hemophilia	0.1	
Acquired hypertrichosis lanuginosa		60 cases
Acquired Von Willebrand syndrome		300 cases
Acrocallosal syndrome, Schinzel type		34 cases
Acro-cardio-facial syndrome		9 cases
Acrocephalosyndactyly	4.6	
Acrocraniofacial dysostosis		2 cases
Acrodermatitis enteropathica, zinc deficiency type	0.2	
Acrofacial dysostosis, Catania type		6 cases
Acrofacial dysostosis, Nager type		90 cases
Acrofacial dysostosis, Palagonia type		4 cases
Acrofacial 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, Brahimi-Bacha type		3 cases
Acromesomelic dysplasia, Hunter-Thomson type		10 cases
Acromesomelic dysplasia, Maroteaux type		50 cases
Acromicric dysplasia		< 40 cases
Acroosteolysis dominant type		50 cases
Acro-pectoral syndrome		22 cases
Acro-pectoro-renal field defect		12 cases
Acropectorovertebral dysplasia		< 30 cases
Acrorenal syndrome		20 cases
Acro-renal-mandibular syndrome		7 cases
Acro-renal-ocular syndrome		< 20 families
ACTH-dependent Cushing syndrome	6	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Action myoclonus - renal failure syndrome		17 cases
Acute bilateral depigmentation of the iris		5 cases
Acute inflammatory demyelinating polyradiculoneuropathy	3.1	
Acute intermittent porphyria	10.1	
Acute interstitial pneumonia	3.8	
Acute lymphoblastic leukemia	6.5	
Acute motor axonal neuropathy	0.1	
Acute motor-sensory axonal neuropathy	0.1	
Acute myeloid leukemia	16	
Acute promyelocytic leukemia	8	
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
Adrenocortical carcinoma	1	
Adult acute respiratory distress syndrome	30	
Adult familial nephronophtisis - spastic quadriparesia		2 cases
Adult Still's disease	1.23	
ADULT syndrome		14 cases
Adult-onset proximal spinal muscular atrophy, autosomal dominant	0.1	
Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis		3 cases
Agenesis of the corpus callosum - intellectual deficit - coloboma - micrognathia		2 cases
Aggressive systemic mastocytosis	0.2	
Agnathia - holoprosencephaly - situs inversus		30 cases
Aicardi syndrome	0.06	
Aicardi-Goutieres syndrome		120 cases
Alagille syndrome	1.4	
Alar cartilages hypoplasia - coloboma - telecanthus		2 cases
Albers-Schönberg osteopetrosis	1	
Albinism-deafness syndrome		1 family
Albright hereditary osteodystrophy	0.72	
Alexander disease		300 cases
Al-Gazali-Dattani syndrome		3 cases
Alkaptonuria	0.5	
Allan-Herndon-Dudley syndrome		89 cases
Alopecia - contractures - dwarfism - intellectual deficit		5 cases
Alopecia - epilepsy - pyorrhea - intellectual deficit		12 cases
Alopecia totalis	10.5	
Alopecia universalis	25	
Alpers syndrome	0.025	
Alpha thalassemia - X-linked intellectual deficit		168 cases
Alpha-1 antitrypsin deficiency	33	
Alpha-mannosidosis	0.1	
Alpha-N-acetylgalactosaminidase deficiency		12 cases
Alport syndrome	2	
Alström syndrome	0.14	
Alveolar echinococcosis		< 1000 cases
Amaurosis - hypertrichosis		2 cases
Ambras syndrome		40 cases
Amelo-cerebro-hypohidrotic syndrome		19 cases
Amelogenesis imperfecta and gingival hyperplasia syndrome		4 cases
Aminopterin embryofetopathy		17 cases
Amniotic bands	4**	
Amoebiasis due to free-living amoebae	1.75	
Amyloidosis	30	
Amyotrophic lateral sclerosis	5.2	
Anal fistula	20.5	
Anaplastic large cell lymphoma	2	
Anaplastic thyroid carcinoma	0.1	
Androgen insensitivity syndrome	13	
ANE syndrome		5 cases
Angelman syndrome	7.5	
Angel-shaped phalango-epiphyseal dysplasia		15 cases
Angio-osteohypertrophic syndrome		1000 cases
Anhidrotic ectodermal dysplasia - immunodeficiency - osteopetrosis - lymphedema		2 cases
Aniridia	1.75	
Aniridia - absent patella		3 cases
Aniridia - cerebellar ataxia - intellectual deficit		> 10 families
Aniridia - ptosis - intellectual deficit - familial obesity		3 cases
Aniridia - renal agenesis - psychomotor retardation		2 cases
Aniridia-intellectual deficit syndrome		2 cases
Anisakiasis	3.8	
Ankyloblepharon filiforme - imperforate anus		2 families
Ankylosing vertebral hyperostosis with tylosis		8 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Anomaly of bile acid synthesis	0.6	
Anonychia - microcephaly		5 cases
Anonychia with flexural pigmentation		3 cases
Anophthalmia - hypothalamo-pituitary insufficiency		30 cases
Anophthalmia - megalocornea - cardiopathy - skeletal anomalies		3 cases
Anophthalmia plus syndrome		4 cases
Anophthalmia/microphthalmia - esophageal atresia		30 cases
Antecubital pterygium syndrome		11 cases
Antisynthetase syndrome	1.5	
Antley-Bixler syndrome		34 cases
Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis		< 50 cases
Aortic aneurysm syndrome, Loeys-Dietz type		10 families
Aortic arch anomaly - peculiar facies - intellectual deficit		4 cases
Aortic arch interruption	0.3**	
Aortic dilatation - joint hypermobility - arterial tortuosity		22 cases
Aorto-ventricular tunnel		130 cases
Apert syndrome	1.25	
Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis		3 cases
Aphalangy - syndactyly - microcephaly		1 family
Aplasia cutis - myopia		4 cases
Aplasia cutis congenita - intestinal lymphangiectasia		3 cases
Apnea of prematurity	8.5	
Arachnodactyly - abnormal ossification - intellectual deficit		5 cases
Arachnodactyly - intellectual deficit - dysmorphism		3 cases
AREDYLD syndrome		3 cases
Argininemia		31 cases
Argininosuccinic aciduria	0.45	
Arhinia - choanal atresia - microphthalmia		4 cases
Aromatase deficiency		13 cases
Arrhinia		20 cases
Arrhythmogenic right ventricular dysplasia	43.5	
Arterial dissection - lentiginosis		4 cases
Arterial tortuosity syndrome		< 80 cases
Arthrogryposis - hyperkeratosis, lethal form		2 cases
Arthrogryposis - renal dysfunction - cholestasis		< 100 cases
Arthrogryposis multiplex congenita	16.1	
Arthrogryposis multiplex congenita - whistling face		10 cases
Ascher syndrome		50 cases
Astley-Kendall dysplasia		5 cases
Ataxia-deafness-retardation syndrome		8 cases
Ataxia-telangiectasia	1	
Atelosteogenesis I		12 cases
Atelosteogenesis type II		25 cases
Atelosteogenesis type III		12 cases
Athabaskan brainstem dysgenesis syndrome		10 cases
Atherosclerosis- deafness - diabetes - epilepsy - nephropathy		2 cases
Athyreosis	3.5	
Atkin-Flaitz syndrome		14 cases
Atopic keratoconjunctivitis	15.1	
Atransferrinemia		9 cases
Atresia of small intestine	20	
Atrial septal defect - atrioventricular conduction defects		11 cases
Atrial tachyarrhythmia with short PR interval		12 cases
Atypical coarctation of aorta	0.17**	
Atypical hemolytic uremic syndrome	1	
Atypical Rett syndrome	2.22	
Auricular abnormalities - cleft lip with or without cleft palate - ocular abnormalities		2 cases
Auriculoocular anomalies - cleft lip		2 cases
Auriculoosteodysplasia		2 families
Autism - facial port-wine stain		4 cases
Autoimmune lymphoproliferative syndrome		100 cases
Autoinflammatory disease due to interleukin-1 receptor antagonist deficiency		10 cases
Autoinflammatory granulomatosis of childhood		40 families
Autosomal dominant cerebellar ataxia	3.5	
Autosomal dominant Charcot-Marie-Tooth disease type 2F		1 family
Autosomal dominant Charcot-Marie-Tooth disease type 2G		1 family
Autosomal dominant Charcot-Marie-Tooth disease type 2K		3 families
Autosomal dominant Charcot-Marie-Tooth disease type 2L		1 family
Autosomal dominant diffuse palmoplantar keratoderma, Norrbotten type	2.5	
Autosomal dominant familial hematuria - retinal arteriolar tortuosity - contractures		8 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Autosomal dominant hyper IgE syndrome		250 cases
Autosomal dominant hypohidrotic ectodermal dysplasia		40 cases
Autosomal dominant limb-girdle muscular dystrophy type 1A		2 families
Autosomal dominant limb-girdle muscular dystrophy type 1D		5 families
Autosomal dominant limb-girdle muscular dystrophy type 1E		5 families
Autosomal dominant limb-girdle muscular dystrophy type 1F		1 family
Autosomal dominant limb-girdle muscular dystrophy type 1G		1 family
Autosomal dominant macrothrombocytopenia with abnormal proplatelet formation		5 cases
Autosomal dominant medullary cystic kidney disease with or without hyperuricemia	0.11	
Autosomal dominant multiple pterygium syndrome		4 cases
Autosomal dominant optic atrophy and cataract		14 cases
Autosomal dominant osteopetrosis type 1		33 cases
Autosomal dominant palmoplantar keratoderma and congenital alopecia		1 family
Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis		30 cases
Autosomal dominant popliteal pterygium syndrome	0.3	
Autosomal dominant rhegmatogenous retinal detachment		38 cases
Autosomal dominant severe congenital neutropenia	0.4**	
Autosomal dominant spastic paraplegia type 6		10 families
Autosomal dominant spastic paraplegia type 8		< 10 families
Autosomal dominant spastic paraplegia type 9		1 family
Autosomal dominant spastic paraplegia type 10		< 10 families
Autosomal dominant spastic paraplegia type 12		< 10 families
Autosomal dominant spastic paraplegia type 13		< 10 families
Autosomal dominant spastic paraplegia type 17		< 20 families
Autosomal dominant spastic paraplegia type 29		1 family
Autosomal dominant spastic paraplegia type 37		13 cases
Autosomal dominant spastic paraplegia type 38		1 family
Autosomal recessive acrofacial dysostosis		2 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Autosomal recessive amelia		3 cases
Autosomal recessive ataxia, Beauce type		57 cases
Autosomal recessive cerebellar ataxia	7	
Autosomal recessive cerebellar ataxia - blindness - deafness		3 families
Autosomal recessive cerebellar ataxia - saccadic intrusion		1 family
Autosomal recessive limb girdle muscular dystrophy type 2A	3.8	
Autosomal recessive limb-girdle muscular dystrophy type 2C	1.96	
Autosomal recessive limb-girdle muscular dystrophy type 2D	0.57	
Autosomal recessive limb-girdle muscular dystrophy type 2E	0.57	
Autosomal recessive limb-girdle muscular dystrophy type 2F	0.57	
Autosomal recessive limb-girdle muscular dystrophy type 2G		14 cases
Autosomal recessive limb-girdle muscular dystrophy type 2I		> 40 families
Autosomal recessive limb-girdle muscular dystrophy type 2L		14 cases
Autosomal recessive limb-girdle muscular dystrophy type 2M		3 cases
Autosomal recessive lower motor neuron disease with childhood onset		5 cases
Autosomal recessive malignant osteopetrosis	0.75**	
Autosomal recessive medullary cystic kidney disease	1.05	
Autosomal recessive polycystic kidney disease	6.5	
Autosomal recessive spastic paraplegia type 14		1 family
Autosomal recessive spastic paraplegia type 15		< 10 families
Autosomal recessive spastic paraplegia type 18		9 cases
Autosomal recessive spastic paraplegia type 23		1 family
Autosomal recessive spastic paraplegia type 24		1 family
Autosomal recessive spastic paraplegia type 25		1 family
Autosomal recessive spastic paraplegia type 26		2 families
Autosomal recessive spastic paraplegia type 27		2 families
Autosomal recessive spastic paraplegia type 28		1 family
Autosomal recessive spastic paraplegia type 30		1 family

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Autosomal recessive spastic paraplegia type 32		1 family
Autosomal recessive spastic paraplegia type 35		1 family
Autosomal recessive spastic paraplegia type 39		2 families
Axenfeld-Rieger anomaly - hydrocephaly - skeletal abnormalities		3 cases
Axenfeld-Rieger syndrome	0.5	
Axial spondylometaphyseal dysplasia		3 cases
Babesiosis		40 cases
Bacterial toxic-shock syndrome	3	
Ballard syndrome		12 cases
Bamforth syndrome		5 cases
Bangstad syndrome		2 cases
Banki syndrome		1 family
Barber-Say syndrome		10 cases
Bardet-Biedl syndrome	0.8	
Barth syndrome	0.22	
Bartsocas-Papas syndrome		24 cases
Bartter syndrome	0.12	
Bazex syndrome		145 cases
Bazex-Dupre-Christol syndrome		143 cases
Beckwith-Wiedemann syndrome	7.3	
Beemer-Ertbruggen syndrome		2 cases
Behcet disease	3.4	
Bencze syndrome		2 families
Benign exophthalmos syndrome		4 cases
Benign familial neonatal-infantile seizures		10 families
Benign familial nocturnal alternating hemiplegia of childhood		< 10 cases
Benign paroxysmal torticollis of infancy		50 cases
Berant syndrome		1 family
Berardinelli-Seip congenital lipodystrophy	0.25	
Bernard-Soulier syndrome		100 cases
Best disease	4.4	
Beta-mannosidosis		14 cases
Beta-thalassemia	0.5	
Beta-ureidopropionase deficiency		5 cases
Bethlem myopathy		100 cases
Bickel-Fanconi glycogenosis		112 cases
Bilateral anorchia	2.5	
Bilateral microtia - deafness - cleft palate		4 cases
Bilateral renal agenesis	17	
Bilateral striopallidodentate calcinosis		< 200 cases
Biliary atresia	5.6	
Biotinidase deficiency	1.6	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Birt-Hogg-Dube syndrome	0.5	
Björnstad syndrome		33 cases
Blackfan-Diamond disease	0.32	
Bleeding diathesis due to a collagen receptor defect		< 20 cases
Blepharo-cheilo-odontic 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
Blindness - scoliosis - arachnodactyly		4 cases
Bloom syndrome		> 100 cases
Blue rubber bleb nevus		> 200 cases
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	
Bowen-Conradi syndrome		44 cases
Brachydactyly - arterial hypertension		> 10 families
Brachydactyly - long thumb		4 cases
Brachydactyly - nystagmus - cerebellar ataxia		1 family
Brachydactyly - preaxial hallux varus		8 cases
Brachydactyly type A5		2 families
Brachydactyly type A6		7 cases
Brachydactyly type A7		1 family
Brachymorphism - onychodysplasia - dysphalangism		9 cases
Brachytelephalangy - dysmorphism - Kallmann syndrome		2 cases
Braddock syndrome		2 cases
Bradyopsia		5 cases
Brain demyelination due to methionine adenosyltransferase deficiency		2 cases
Brain malformation - congenital heart disease - postaxial polydactyly		2 cases
Brain-lung-thyroid syndrome		< 20 cases
Branchiogenic deafness syndrome		5 cases
Branchio-oculo-facial syndrome		< 50 cases
Branchio-skeleto-genital syndrome		3 cases
Bronchopulmonary dysplasia	13	
Brown-Vialetto-van Laere syndrome		< 100 cases
Bruck syndrome		< 40 cases
Brugada syndrome	20	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Budd-Chiari syndrome	1.5	
Buerger's disease	16	
Bullous dystrophy, macular type		2 families
Bullous pemphigoid	2.5	
Bullous systemic lupus erythematosus		70 cases
Buschke-Ollendorff syndrome	5	
Cabezas syndrome		1 family
CACH syndrome		148 cases
CADASIL syndrome		500 cases
Calvarial doughnut lesions - bone fragility		20 cases
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**	
Cantu syndrome		23 cases
Cap myopathy		< 10 cases
Cap polyposis		20 cases
Capillary leak syndrome		57 cases
Carbamoylphosphate synthetase deficiency	0.8	
Carcinoma of the gallbladder	6.5	
Cardiac anomalies - heterotaxy		9 cases
Cardiocranial syndrome, Pfeiffer type		< 10 cases
Cardiomyopathy - cataract - hip spine disease		9 cases
Cardiomyopathy - renal anomalies		2 cases
Cardiomyopathy-exercise intolerance due to muscle and heart glycogen deficiency		3 cases
Carey-Fineman-Ziter syndrome		< 20 cases
Carnevale syndrome		2 cases
Carney complex		160 cases
Carney triad		100 cases
Carnitine palmitoyl transferase II deficiency		> 300 cases
Carnitine-acylcarnitine translocase deficiency		30 cases
Carnosinemia		30 cases
Caroli disease		< 250 cases
Carpenter syndrome		40 cases
Carpotarsal osteochondromatosis		< 10 cases
Castleman disease		400 cases
Cataract - ataxia - deafness		2 cases
Cataract - cardiomyopathy		30 cases
Cataract - intellectual deficit - hypogonadism		< 20 cases
Cataract - nephropathy - encephalopathy		2 cases
Cataract-glaucoma		3 families
Cataract-microcornea syndrome		8 families
Catecholinergic polymorphic ventricular tachycardia	10	
Catel-Manzke syndrome		27 cases
Cat-eye syndrome	1.35	
Cathecolamine-producing tumor	10	
Cat-scratch disease	6.6	
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 Ik		4 cases
CDG syndrome type IL		2 cases
CDG syndrome type IIa		4 cases
CDG syndrome type IIe		2 cases
CDG syndrome type IIh		2 cases
CEDNIK syndrome		7 cases
Celiac disease - epilepsy - occipital calcifications		170 cases
Central areolar choroidal dystrophy	3.33	
Central bilateral macrogyria		4 cases
Central nervous system calcification - deafness - tubular acidosis - anemia		2 cases
Central neurocytoma		> 100 cases
Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss		2 families
Cerebral arteriovenous fistula	6	
Cerebral gigantism - jaw cysts		< 10 cases
Cerebro-costo-mandibular syndrome		75 cases
Cerebro-oculo-nasal syndrome		10 cases
Cerebroretinal vasculopathy		3 families
Cerebrotendinous xanthomatosis	2	
Cervical hypertrichosis - peripheral neuropathy		3 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
CHAND syndrome		> 10 cases
Chaotic atrial tachycardia		100 cases
Char syndrome		10 cases
Charcot-Marie-Tooth disease	32.5	
Charcot-Marie-Tooth disease - nephropathy		10 cases
Charcot-Marie-Tooth disease type 2B2		1 family
Charcot-Marie-Tooth disease type 2H		13 cases
Charcot-Marie-Tooth disease type 4B1		11 families
Charcot-Marie-Tooth disease type 4H		10 cases
Charcot-Marie-Tooth disease type 4J		5 cases
CHARGE syndrome	0.14	
Chediak-Higashi syndrome		> 10 cases
CHILD syndrome		30 cases
Childhood disintegrative disorder	2	
Childhood-onset proximal spinal muscular atrophy, autosomal dominant	0.1	
Choanal atresia	8.2	
Choanal atresia - deafness - cardiac defects - dysmorphism		5 cases
Cholangiocarcinoma	2.1	
Cholestasis - lymphedema		50 cases
Cholestasis - pigmentary retinopathy - cleft palate		5 cases
Cholesteryl ester storage disease		< 50 cases
Chondrodysplasia - disorder of sex development		2 cases
Chondrodysplasia punctata, rhizomelic type	1	
Chondrodysplasia, Blomstrand type		13 cases
Chordoma	0.05	
Choroidal atrophy - alopecia		2 cases
Choroideremia	2	
Choroideremia - deafness - obesity		4 cases
Christ-Siemens-Touraine syndrome	0.35	
Chronic autoimmune hepatitis	0.75	
Chronic B-cell lymphocytic leukemia	30	
Chronic granulomatous disease	0.2	
Chronic hepatic porphyria	1.5	
Chronic hiccup	1	
Chronic inflammatory demyelinating polyneuropathy	4.4	
Chronic myeloid leukemia	6	
Churg-Strauss syndrome	1	
CINCA syndrome		100 cases
Circumscribed palmoplantar hypokeratosis		17 cases
Citrullinemia	14.4	
CLAPO syndrome		6 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Cleft lip - retinopathy		2 cases
Cleft lip/palate - intestinal malrotation - cardiopathy		4 cases
Cleft palate	50	
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	
Coats disease	2	
Cobb syndrome		35 cases
Cockayne syndrome		200 cases
CODAS syndrome		3 cases
Coffin-Lowry syndrome	1.5	
Coffin-Siris syndrome		40 cases
COFS syndrome		< 20 cases
Cogan syndrome		200 cases
Cohen syndrome		100 cases
Colchicine poisoning	0.1	
Cold-induced sweating syndrome		6 cases
Cole-Carpenter syndrome		4 cases
Collagenous colitis	10.5	
Coloboma of macula - brachydactyly type B		12 cases
Colobomatous - microphthalmia - heart disease - hearing loss		10 cases
Combined deficiency of factor V and factor VIII	0.5	
Common variable immunodeficiency	4	
Complete atrioventricular canal	15**	
Cone dystrophy with supernormal rod response		45 cases
Cone rod dystrophy	2.5	
Congenital adrenal hyperplasia	10	
Congenital alveolar capillary dysplasia		< 60 cases
Congenital analbuminemia		< 50 cases
Congenital bilateral absence of vas deferens	50	
Congenital bile acid synthesis defect type 4		5 cases
Congenital brain dysgenesis due to glutamine synthetase deficiency		2 cases
Congenital bronchobiliary fistula		23 cases
Congenital bullous ichthyosiform erythroderma	0.4	
Congenital cataracts - facial dysmorphism - neuropathy		100 cases
Congenital diaphragmatic hernia	15	
Congenital dyserythropoietic anemia	1	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Congenital enterocyte heparan sulfate deficiency		3 cases
Congenital erythropoietic porphyria		> 200 cases
Congenital factor II deficiency	0.05	
Congenital factor V deficiency	0.1	
Congenital factor VII deficiency	0.25	
Congenital factor X deficiency	0.2	
Congenital factor XI deficiency	0.1	
Congenital factor XIII deficiency	0.05	
Congenital fibrinogen deficiency	0.15	
Congenital hypogonadotropic hypogonadism	20	
Congenital hypothyroidism	29	
Congenital hypothyroidism due to developmental anomaly	21.3	
Congenital hypothyroidism due to transplacental passage of maternal TSH-binding inhibitory antibodies	1	
Congenital ichthyosis - microcephalus - quadriplegia		2 cases
Congenital insensitivity to pain		20 cases
Congenital intrauterine infection-like syndrome		> 30 cases
Congenital isolated hyperinsulinism	2	
Congenital isolated thyroxine-binding globulin deficiency	46	
Congenital Leber amaurosis	10	
Congenital lethal erythroderma		17 cases
Congenital lethal myopathy, Compton-North type		4 cases
Congenital lobar emphysema	4.5	
Congenital lymphedema	8.8	
Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells		3 cases
Congenital megacalycosis		> 50 cases
Congenital muscular dystrophy	5	
Congenital muscular dystrophy due to lamine A/C deficiency		15 cases
Congenital muscular dystrophy type 1A	3.3	
Congenital muscular dystrophy with integrin deficiency	0.03	
Congenital muscular dystrophy, Fukuyama type	0.54	
Congenital muscular dystrophy, Ullrich type		< 100 cases
Congenital myasthenic syndromes	0.75	
Congenital osteogenesis imperfecta - microcephaly - cataracts		3 cases
Congenital pseudoarthrosis of clavicle		> 200 cases
Congenital pulmonary lymphangiectasia		> 100 cases
Congenital pulmonary valve stenosis	7.2	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Congenital Rubella syndrome	1**	
Congenital spastic tetraplegia		5 cases
Congenital sucrase-isomaltase deficiency	20	
Congenital toxoplasmosis	50	
Congenitally short costocoracoid ligament		1 family
Contractures - ectodermal dysplasia - cleft lip/palate		2 cases
Cooks syndrome		11 cases
Cooper-Jabs syndrome		2 cases
Corneal anesthesia - deafness - intellectual deficit		2 cases
Corneal dystrophy - perceptive deafness		< 10 cases
Corneal-cerebellar syndrome		2 cases
Cornelia de Lange syndrome	1.9	
Coronary artery disease - hyperlipidemia - hypertension - diabetes - osteoporosis		1 family
Corpus callosum agenesis - neuronopathy	19	
Cortical blindness - intellectual deficit - polydactyly		3 cases
Corticobasal degeneration	4	
Corticosteroid-sensitive aseptic abscesses		49 cases
Costello syndrome		200 cases
Cowden syndrome	0.45	
Coxoauricular syndrome		4 cases
Coxo-podo-patellar syndrome		47 cases
Craniodiaphyseal dysplasia		< 20 cases
Craniodigital syndrome - intellectual deficit		5 cases
Craniofacial conodysplasia		1 family
Craniofacial dyssynostosis	0.05	
Craniofacial-deafness-hand syndrome		3 cases
Craniofrontonasal dysplasia - Poland anomaly		3 cases
Craniolenticulosutural dysplasia		28 cases
Craniometaphyseal dysplasia		70 cases
Cranio-osteoarthropathy		30 cases
Craniopharyngioma	2	
Craniorachischisis	5	
Craniorhiny		3 families
Craniosynostosis - Dandy-Walker - hydrocephalus		4 cases
Craniosynostosis - dysmorphism - brachydactyly		5 cases
Craniosynostosis - fibular aplasia		2 cases
Craniosynostosis - hydrocephalus - Chiari I malformation - radioulnar synostosis		4 cases
Craniosynostosis - intracranial calcifications		3 cases
Craniosynostosis, Boston type		19 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Craniosynostosis, Philadelphia type		1 family
Craniosynostosis-radial aplasia, Imaizumi type		2 cases
CREST syndrome	8	
Creutzfeldt-Jakob disease	0.1	
Crigler-Najjar syndrome	1	
Crisponi syndrome		< 30 cases
Criss-cross heart	0.8	
Cronkhite-Canada syndrome		500 cases
Crouzon disease	2	
Cryptomicrotia - brachydactyly - excess fingertip arch		2 cases
Cryptosporidiosis	34	
Currarino triad	1	
Curry-Jones syndrome		5 cases
Cushing disease	4	
Cushing syndrome	5.9	
Cutaneous lupus erythematosus	50	
Cutaneous lymphoma	8.3	
Cutaneous mastocytosis	0.75	
Cutaneous neuroendocrine carcinoma	4	
Cutaneous photosensitivity - lethal colitis		3 cases
Cutaneous T-cell lymphoma	14.9	
Cutis gyrata - acanthosis nigricans - craniosynostosis		6 cases
Cutis laxa		> 100 cases
Cutis marmorata telangiectatica congenita		300 cases
Cutis verticis gyrata - intellectual deficit	1.02	
Cyclic neutropenia	0.1	
Cyprus facial-neuromusculoskeletal syndrome		1 family
Cystathioninuria	7	
Cystic fibrosis	12.6	
Cystic hamartoma of lung and kidney		< 5 cases
Cystinosis	0.5	
Cystinuria	14	
Cystoid macular dystrophy		6 families
Cytophagic histiocytic panniculitis		< 100 cases
Czech dysplasia, metatarsal type		< 20 cases
Dacryocystitis - osteopoikilosis		5 cases
Dahlberg-Borer-Newcomer syndrome		2 cases
Dandy-Walker malformation - postaxial polydactyly		2 cases
Darier disease	1.5	
Deaf blind hypopigmentation syndrome, Yemenite type		2 cases
Deafness - enamel hypoplasia - nail defects		6 cases
Deafness - genital anomalies - metacarpal and metatarsal synostosis		2 cases
Deafness - intellectual deficit, Martin-Probst type		3 cases
Deafness - lymphedema - leukemia		< 10 cases
Deafness - peripheral neuropathy - arterial disease		4 cases
Deafness - vitiligo - achalasia		2 cases
Deafness with labyrinthine aplasia, microtia, and microdontia		6 families
Deafness-infertility syndrome		3 families
Dehydratase deficiency		21 cases
Deletion 6q16 syndrome		7 cases
DEND syndrome		14 cases
Dent disease		250 cases
Dentatorubral-pallidoluysian atrophy	0.48	
Dentinogenesis imperfecta - short stature - hearing loss - intellectual deficit		2 cases
Denys-Drash syndrome		150 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
Dermo-odonto dysplasia		14 cases
Desbuquois syndrome		> 40 cases
Desmosterolosis		2 cases
Developmental delay - deafness, Hildebrand type		1 family
Developmental delay due to 2-methylbutyryl-CoA dehydrogenase deficiency		< 30 cases
Developmental malformations - deafness - dystonia		2 cases
Diaphanospondylodysostosis		< 10 cases
Diaphragmatic defect - limb deficiency - skull defect		4 cases
Diastrophic dwarfism	1.2	
Diffuse cutaneous systemic sclerosis	4	
Diffuse large B-cell lymphoma	20	
Diffuse neonatal hemangiomatosis		< 70 cases
Diffuse palmoplantar keratoderma - acrocyanosis		10 cases
Digitorenocerebral syndrome		< 10 cases
Digitotalar dysmorphism	6	
Dihydrolipoyl dehydrogenasee deficiency		20 cases
Dihydropteridine reductase deficiency		134 cases
Dihydropyrimidinuria		7 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Dincsoy-Salih-Patel syndrome		2 cases
Dirofilariasis		25 cases
Disorder of sex development - intellectual deficit		3 cases
Distal arthrogryposis type 6		1 family
Distal limb deficiencies - micrognathia syndrome		4 cases
Distal monosomy 10q		40 cases
Distal monosomy 5q		10 cases
Distal monosomy 8p		20 cases
Distal myopathy with early respiratory muscle involvement		24 cases
Distal myopathy with posterior leg and anterior upper limb involvement		12 cases
Distal myopathy with vocal cord weakness		12 cases
Distal myopathy, Nonaka type	0.1	
Distal myopathy, Welander type	10	
Distal symphalangism		< 5 families
Distal trisomy 10q		40 cases
Distal trisomy 6p		40 cases
Donnai-Barrow syndrome		13 cases
DOOR syndrome		< 50 cases
Dopamine beta-hydroxylase deficiency		12 cases
Dopa-responsive dystonia	0.3	
Double outlet left ventricle		32 cases
Double uterus - hemivagina - renal agenesis		< 60 cases
Down syndrome	50	
Duane anomaly - myopathy - scoliosis		2 cases
Duane syndrome	10	
Dubowitz syndrome		150 cases
Duchenne and Becker muscular dystrophy	5	
Duchenne muscular dystrophy	3.7	
Duodenal atresia	8.55	
Dursun syndrome		2 cases
Dyggve-Melchior-Clausen disease		60 cases
Dyschondrosteosis - nephritis		1 family
Dyskeratosis congenita	0.1	
Dysmorphism - short stature - deafness - disorder of sex development		2 cases
Dystonia 16		7 cases
Dystrophic epidermolysis bullosa	0.7	
Early infantile epileptic encephalopathy		88 cases
Early myoclonic encephalopathy		30 cases
Early onset torsion dystonia	0.4	
Early-onset autosomal dominant Alzheimer disease	5.3	
Ear-patella-short stature syndrome		42 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Ebstein malformation	1.25	
Ectodermal dysplasia - absent dermatoglyphs		< 30 cases
Ectodermal dysplasia - blindness		2 cases
Ectodermal dysplasia - skin fragility syndrome		10 cases
Ectodermal dysplasia, Berlin type		4 cases
Ectodermal dysplasia, "pure" hair-nail type		< 20 cases
Ectopia lentis - chorioretinal dystrophy - myopia		4 cases
Ectrodactyly - ectodermal dysplasia without clefting		5 cases
EEM syndrome		7 families
Ehlers-Danlos syndrome	0.2	
Ehlers-Danlos syndrome type 1	5	
Ehlers-Danlos syndrome type 10		1 family
Ehlers-Danlos syndrome, classic type	3.5	
Ehlers-Danlos syndrome, dermatosparaxis type		7 cases
Ehlers-Danlos syndrome, hypermobile type	12.5	
Ehlers-Danlos syndrome, spondylocheiro dysplastic type		6 cases
Ehlers-Danlos syndrome, vascular type	1	
Ehrlichiosis		< 50 cases
Eiken syndrome		6 cases
Elejalde disease		30 cases
Ellis Van Creveld syndrome		150 cases
Emery-Dreifuss muscular dystrophy	0.3	
Encephalocraniocutaneous lipomatosis		45 cases
Encephalopathy due to GLUT1 deficiency		84 cases
Encephalopathy due to hydroxykynureninuria		< 30 cases
Encephalopathy due to prosaposin deficiency		< 10 cases
Encephalopathy due to sulfite oxidase deficiency		50 cases
Encephalopathy due to urocanase deficiency		4 cases
Enchondromatosis		600 cases
Endocrine tumor	13	
Endosteal sclerosis - cerebellar hypoplasia		4 cases
Eng-Strom syndrome		2 cases
Enteropancreatic endocrine tumor	14	
Enthesitis-related arthritis	5.7	
Eosinophilic fasciitis		200 cases
Eosinophilic gastroenteritis		280 cases
Epidermal nevus syndrome		> 400 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Epidermolysis bullosa simplex - limb girdle muscular dystrophy		< 20 cases
Epidermolytic epidermolysis bullosa	2.5	
Epilepsy - microcephaly - skeletal dysplasia		2 cases
Epilepsy telangiectasia		6 cases
Episodic ataxia type 3		1 family
Episodic ataxia type 4		2 families
Episodic ataxia type 5		7 cases
Episodic ataxia type 6		4 cases
Episodic ataxia type 7		7 cases
Epithelio-exfoliative colitis - deafness		2 cases
Erdheim-Chester disease		350 cases
Ermine phenotype		3 cases
Erythrokeratodermia - ataxia		25 cases
Erythrokeratodermia variabilis, Mendes da Costa type		> 200 cases
Erythropoietic protoporphyria	0.9	
Esophageal atresia	25	
Esophageal carcinoma	4	
Essential thrombocythemia	24	
Esthesioneuroblastoma		< 1000 cases
Ethylmalonic encephalopathy		< 40 cases
Evans syndrome	0.1	
Ewing sarcoma	0.1	
Excessive growth - learning disabilities - facial dysmorphism		6 families
Extraskeletal myxoid chondrosarcoma	0.2	
Eyebrow duplication - syndactyly		3 cases
Fabry disease	1.75	
Facial dysmorphism - macrocephaly - myopia - Dandy-Walker malformation		3 cases
Facial onset sensory and motor neuronopathy		4 cases
Facioscapulohumeral dystrophy	7	
Familial acute necrotizing encephalopathy		11 cases
Familial adenomatous polyposis	5.5	
Familial amyloid polyneuropathy	< 1	
Familial caudal dysgenesis		4 cases
Familial cold urticaria	0.1	
Familial developmental dysphasia		6 families
Familial dysautonomia		550 cases
Familial encephalopathy with neuroserpin inclusion bodies		> 5 families
Familial glucocorticoid deficiency		50 cases
Familial isolated dilated cardiomyopathy	17.5	
Familial isolated hypoparathyroidism		< 10 families

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Familial isolated hypoparathyroidism due to agenesis of parathyroid gland		2 families
Familial isolated restrictive cardiomyopathy	2.5	
Familial long QT syndrome	40	
Familial melanoma	46.8	
Familial multiple fibrofolliculoma		7 cases
Familial or sporadic hemiplegic migraine	10	
Familial Parkinson's disease dementia	41	
Familial partial lipodystrophy associated with PPARG mutations		10 cases
Familial partial lipodystrophy due to AKT2 mutations		1 family
Familial partial lipodystrophy, Dunnigan type		300 cases
Familial partial lipodystrophy, Köbberling type		< 20 cases
Familial platelet syndrome with predisposition to acute myelogenous leukemia		< 20 families
Familial scaphocephaly syndrome, McGillivray type		11 cases
Familial spastic paraplegia	5	
Fanconi anemia	0.3	
FASTKD2-related infantile mitochondrial encephalomyopathy		2 cases
Fatal familial insomnia		27 cases
Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3		2 cases
Feingold syndrome		< 50 cases
Femur-fibula-ulna complex	1.5	
Fetal cytomegalovirus syndrome	40	
Fetal methylmercury syndrome		800 cases
Fetal varicella syndrome		> 100 cases
Fibrochondrogenesis		11 cases
Fibrodysplasia ossificans progressiva	0.06	
Fibrous dysplasia of bone	< 50	
Fibular aplasia - ectrodactyly		< 50 cases
Fibular dimelia - diplopodia		11 cases
Fibular hemimelia	2	
Filippi syndrome		< 25 cases
Fine-Lubinsky syndrome		5 cases
Floating-Harbor syndrome		< 50 cases
Flynn-Aird syndrome		10 cases
Focal dermal hypoplasia		300 cases
Focal dystonia	11.7	
Focal facial dermal dysplasia		< 10 families
Focal myositis		50 cases
Follicular lymphoma	36	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Fountain syndrome		8 cases
Foveal hypoplasia - presenile cataract		11 cases
Fragile X syndrome	28	
Frank-Ter Haar syndrome		5 cases
Fraser syndrome		150 cases
Frasier syndrome		> 50 cases
Freeman-Sheldon syndrome		100 cases
Fried syndrome		1 family
Friedreich ataxia	2	
Frontometaphyseal dysplasia		< 30 cases
Frontotemporal dementia	3	
Frontotemporal dementia and parkinsonism linked to chromosome 17	0.3	
Frontotemporal dementia with tau inclusions	15	
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
GAPO syndrome		27 cases
Gastric cancer	28	
Gastrointestinal stromal tumor	13	
Gastroschisis	12	
Gaucher disease	2	
Gaucher disease - ophthalmoplegia - cardiovascular calcification		< 10 cases
Gaucher disease type 1	0.94	
Gaucher disease type 2	0.01	
Gaucher disease type 3	0.05	
Geleophysic dysplasia		27 cases
Genitopatellar syndrome		7 cases
German syndrome		5 cases
Geroderma osteodysplastica	0.1	
Giant axonal neuropathy		20 families
Giant cell arteritis	8.9	
Gingival fibromatosis - progressive deafness		2 families
Gitelman syndrome	2.5	
Glaucoma - ectopia - microspherophakia - stiff joints - short stature		3 cases
Glaucoma - sleep apnea		5 cases
Glial tumor	10.4	
Glioblastoma	1	
Global developmental delay - osteopenia - ectodermal defect		3 cases
Glomerulonephritis - sparse hair - telangiectasis		< 10 cases
Glossopalatine ankylosis		30 cases
Glucose-galactose malabsorption		200 cases
Glutaric acidemia type 1	0.4	
Glutathione synthetase deficiency		65 cases
Glycogen branching enzyme deficiency	0.6	
Glycogen storage disease due to LAMP-2 deficiency		84 cases
Glycogen storage disease due to muscle phosphorylase kinase deficiency		< 30 cases
Glycogen storage disease type 2	1.1	
Goldberg-Shprintzen megacolon syndrome		10 cases
Goldblatt syndrome		11 cases
Goldenhar syndrome	3.5	
Goldmann-Favre syndrome		< 50 cases
Gollop-Wolfgang complex		200 cases
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	2.76	
Grange syndrome		6 cases
Granulomatous slack skin		< 50 cases
Gräsbeck-Imerslund disease		300 cases
Greenberg dysplasia		< 10 cases
Greig cephalopolysyndactyly syndrome		100 cases
Griscelli disease		60 cases
Growth deficiency - brachydactyly - dysmorphism		2 families
Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate		4 cases
Growth delay due to insulin-like growth factor I deficiency		4 cases
GTP cyclohydrolase I deficiency		17 cases
Guanidinoacetate methyltransferase deficiency		9 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Guillain-Barré syndrome	3.45	
H syndrome		17 cases
Haim-Munk syndrome		< 100 cases
Hair defect - photosensitivity - intellectual deficit		3 cases
Hairy cell leukemia	10	
Hallermann-Streiff-François syndrome		< 100 cases
Harding ataxia	1	
Hartnup syndrome	4	
Hartsfield-Bixler-Demyer syndrome		6 cases
Heart defects - limb shortening		2 cases
HEC syndrome		2 cases
Heinz body anemia		< 10 cases
Helicoid peripapillary chorioretinal degeneration		100 cases
Hemimelia	4.15	
Hemolytic anemia due to adenylate kinase deficiency		12 cases
Hemolytic anemia due to glucophosphate isomerase deficiency		50 cases
Hemolytic anemia due to glutathione reductase deficiency		3 cases
Hemolytic anemia due to red cell pyruvate kinase deficiency	5.1	
Hemophilia	7.7	
Hemophilia A	10	
Hemophilia B	2	
Hemorrhagic disease due to alpha-1 antitrypsin Pittsburgh mutation		3 cases
Hennekam syndrome		> 50 cases
Hennekam-Beemer syndrome		2 cases
Henoch-Schönlein purpura	8.5	
Hepatic glycogen synthase deficiency		16 cases
Hepatic veno-occlusive disease	11	
Hepatic veno-occlusive disease - immunodeficiency		< 25 cases
Hepatocellular carcinoma	1	
Hereditary angioedema	1	
Hereditary breast and ovarian cancer syndrome	25	
Hereditary chronic pancreatitis	0.125	
Hereditary cryohydrocytosis with reduced stomatin		2 cases
Hereditary elliptocytosis	35	
Hereditary epidermolysis bullosa	0.8	
Hereditary folate malabsorption		17 cases
Hereditary hyperferritinemia with congenital cataracts		> 64 cases
Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia		19 cases
Hereditary myoclonus - progressive distal muscular atrophy		< 10 cases
Hereditary myopathy with early respiratory failure		< 10 families
Hereditary neurocutaneous angioma		< 10 families
Hereditary neuropathy with liability to pressure palsies	9	
Hereditary North American Indian childhood cirrhosis		36 cases
Hereditary orotic aciduria		< 20 cases
Hereditary progressive mucinous histiocytosis		13 cases
Hereditary sensory and autonomic neuropathy type 2		35 cases
Hereditary sensory and autonomic neuropathy with deafness and global delay		4 cases
Hereditary spherocytosis	20	
Hereditary thrombophilia due to congenital protein C deficiency	0.2	
Hereditary thrombophilia due to congenital protein S deficiency	0.2	
Hereditary vascular retinopathy		1 family
Hermansky-Pudlak syndrome	0.15	
HERNS syndrome		3 families
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
Hirschsprung disease - type D brachydactyly		4 cases
Histidinemia	4	
Hodgkin lymphoma	10	
Hodgkin lymphoma, classical	10.2	
Holoprosencephaly	7	
Holt-Oram syndrome	1	
Homocarnosinosis		4 cases
Homocystinuria due to cystathionine beta-synthase deficiency	0.4	
Homocystinuria without methylmalonic aciduria		60 cases
Humeroradioulnar synostosis		30 cases
Humerospinal dysostosis		5 cases
Humeroulnar synostosis		5 cases
Huntington disease	7	
Hurler syndrome	0.57	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Hurler-Scheie syndrome	0.23	
Hutchinson-Gilford progeria syndrome	0.005	
Hydrocephalus - blue sclerae - nephropathy		1 family
Hydrocephalus - costovertebral dysplasia - Sprengel anomaly		8 cases
Hydrocephaly - tall stature - joint laxity		2 cases
Hydrolethalus	5**	
Hyperandrogenism due to cortisone reductase deficiency		11 cases
Hyperargininemia	0.17	
Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency		24 cases
Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency		2 cases
Hyperekplexia - epilepsy		2 cases
Hypereosinophilic syndromes	1.5	
Hyperimmunoglobinemia D with recurrent fever		180 cases
Hyperkalemic periodic paralysis	0.5	
Hyperkeratosis - hyperpigmentation syndrome		10 cases
Hyperlipidemia type 3	7.8	
Hyperlipoproteinemia type 1	< 1	
Hyperoxaluria	0.2	
Hyperplastic polyposis syndrome	50	
Hypertelorism, Teebi Type		20 cases
Hypertrichosis cubiti - short stature		28 cases
Hypertrichosis lanuginosa congenita		< 100 cases
Hypertrichotic osteochondrodysplasia		18 cases
Hypochondroplasia	3.3	
Hypocomplementemic leucocytoclasic vasculitis		< 100 cases
Hypoglossia - hypodactyly		< 50 cases
Hypogonadotropic hypogonadism - retinitis pigmentosa		2 cases
Hypohidrotic ectodermal dysplasia - hypothyroidism - ciliary dyskinesia		3 cases
Hypokalemic periodic paralysis	1	
Hypomagnesemia with normocalciuria		2 cases
Hypomandibular faciocranial dysostosis		4 cases
Hypomyelination - congenital cataract		10 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodontia		4 cases
Hypomyelination with atrophy of basal ganglia and cerebellum		19 cases
Hypoparathyroidism - deafness - renal disease		12 cases
Hypopituitarism - microphthalmia		< 10 cases
Hypopituitarism - postaxial polydactyly		6 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Hypospadias-hypertelorism-coloboma and deafness		2 cases
Hypotonia - cystinuria syndrome		14 families
Hypotonia with lactic acidemia and hyperammonemia		3 cases
Hypotrichosis - lymphedema - telangiectasia		4 cases
Hypotrichosis simplex		38 cases
Hypotrichosis-intellectual deficit, Lopes type		2 cases
IBIDS syndrome		15 cases
ICF syndrome		50 cases
Ichthyosis - alopecia - eclabion - ectropion - intellectual deficit		4 cases
Ichthyosis - hepatosplenomegaly - cerebellar degeneration		2 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 prematurity syndrome		16 families
Idiopathic achalasia	10	
Idiopathic acute eosinophilic pneumonia		> 100 cases
Idiopathic and/or familial pulmonary arterial hypertension	1.5	
Idiopathic aplastic anemia	0.4	
Idiopathic hypereosinophilic syndrome	10	
Idiopathic hypersomnia	5	
Idiopathic juvenile-onset systemic arthritis	5	
Idiopathic pulmonary alveolar proteinosis	0.1	
Idiopathic pulmonary fibrosis	16.7	
Idiopathic steroid-sensitive nephrotic syndrome	18	
IMAGe syndrome		< 20 cases
Iminoglycinuria	6.68	
Immune thrombocytopenic purpura	25	
Immunodeficiency by defective expression of HLA class 2		100 cases
Immunodeficiency due to CD25 deficiency		2 cases
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
Inappropriate antidiuretic hormone secretion syndrome		2 cases
Incontinentia pigmenti	0.2	
Indolent systemic mastocytosis	3.8	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Infant epilepsy with migrant focal crisis		29 cases
Infantile choroido cerebral calcification syndrome		10 cases
Infantile neuroaxonal dystrophy		> 150 cases
Infantile onset spinocerebellar ataxia		24 cases
Infantile Refsum disease	0.005	
Inflammatory pseudotumor of the liver		143 cases
Intellectual deficit - cataracts - kyphosis		3 cases
Intellectual deficit - dysmorphism - hypogonadism - diabetes mellitus		4 cases
Intellectual deficit - hypoplastic corpus callosum - preauricular tag		3 cases
Intellectual deficit - sparse hair - brachydactyly		6 cases
Intellectual deficit, Birk-Barel type		1 family
Intellectual deficit, Kahrizi type		3 cases
Intellectual deficit, X-linked - acromegaly - hyperactivity		2 cases
Intellectual deficit, X-linked - choreoathetosis - abnormal behavior		5 cases
Intellectual deficit, X-linked - craniofacioskeletal syndrome		7 cases
Intellectual deficit, X-linked - cubitus valgus - dysmorphism		5 cases
Intellectual deficit, X-linked - Dandy-Walker malformation - basal ganglia disease - Seizures		16 cases
Intellectual deficit, X-linked - dysmorphism - cerebral atrophy		8 cases
Intellectual deficit, X-linked - epilepsy - progressive joint contractures - dysmorphism		2 cases
Intellectual deficit, X-linked - hypogammaglobulinemia - progressive neurological deterioration		3 cases
Intellectual deficit, X-linked - hypogonadism - ichthyosis - obesity - short stature		4 cases
Intellectual deficit, X-linked - hypotonia - facial dysmorphism - aggressive behavior		10 cases
Intellectual deficit, X-linked - macrocephaly - macro-orchidism		12 cases
Intellectual deficit, X-linked - plagiocephaly		2 cases
Intellectual deficit, X-linked - precocious puberty - obesity		3 cases
Intellectual deficit, X-linked - psychosis - macroorchidism		6 cases
Intellectual deficit, X-linked - seizures - psoriasis		4 cases
Intellectual deficit, X-linked - spastic quadriparesis		9 cases
Intellectual deficit, X-linked, Abidi type		8 cases
Intellectual deficit, X-linked, Armfield type		6 cases
Intellectual deficit, X-linked, Cantagrel type		2 cases
Intellectual deficit, X-linked, Cilliers type		4 cases
Intellectual deficit, X-linked, Kroes type		3 cases
Intellectual deficit, X-linked, Miles-Carpenter type		4 cases
Intellectual deficit, X-linked, Pai type		1 family
Intellectual deficit, X-linked, Reish type		2 cases
Intellectual deficit, X-linked, Schimke type		4 cases
Intellectual deficit, X-linked, Seemanova type		4 cases
Intellectual deficit, X-linked, Shashi type		9 cases
Intellectual deficit, X-linked, Shrimpton type		3 cases
Intellectual deficit, X-linked, Siderius type		4 cases
Intellectual deficit, X-linked, Snyder type		11 cases
Intellectual deficit, X-linked, South African type		16 cases
Intellectual deficit, X-linked, Stevenson type		4 cases
Intellectual deficit, X-linked, Stocco Dos Santos type		4 cases
Intellectual deficit, X-linked, Van Esch type		7 cases
Intellectual deficit, X-linked, Vitale type		8 cases
Intellectual deficit, X-linked, Wilson type		3 cases
Intellectual deficit, X-linked, Wittwer type		3 cases
Intellectual deficit, X-linked, Zorick type		6 cases
Internal carotid agenesis		100 cases
Interstitial granulomatous dermatitis with arthritis		< 20 cases
Intractable diarrhea - choanal atresia - eye anomalies		3 cases
IRIDA syndrome		16 cases
Iris coloboma with ptosis - intellectual deficit		10 cases
Iris dysplasia - hypertelorism - deafness		2 cases
IRVAN syndrome		< 30 cases
Isolated anencephaly/exencephaly	3.2**	
Isolated anophthalmia - microphthalmia	14	
Isolated anorectal malformation	24	
Isolated anterior cervical hypertrichosis		< 20 cases
Isolated brachycephaly	5	
Isolated cloverleaf skull syndrome		150 cases
Isolated congenital anosmia		< 15 cases
Isolated dominant hypomagnesemia		3 families

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Isolated ectopia lentis	6.4	
Isolated humeroradial synostosis		150 cases
Isolated Klippel-Feil syndrome	2	
Isolated nonketotic hyperglycinemia	0.2	
Isolated Pierre Robin syndrome	8.75	
Isolated plagiocephaly	10	
Isolated scaphocephaly	20	
Isolated spina bifida	50	
Isolated trigonocephaly	6.7	
Isotretinoin-like syndrome		6 cases
Isovaleric acidemia	1	
IVIC syndrome		4 families
Jackson-Weiss syndrome		2 families
Jacobsen syndrome		150 cases
Jalili syndrome		29 cases
Jervell and Lange-Nielsen syndrome	0.3	
Jeune syndrome	0.2	
Johanson-Blizzard syndrome		23 cases
Joubert syndrome	1	
Joubert syndrome with hepatic defect		8 cases
Joubert syndrome with orofaciodigital defect		29 cases
Juberg-Hayward syndrome		10 cases
Junctional epidermolysis bullosa	0.06	
Juvenile chronic recurrent multifocal osteomyelitis		> 260 cases
Juvenile hyaline fibromatosis		50 cases
Juvenile macular degeneration - hypotrichosis		50 cases
Juvenile myelomonocytic leukemia	0.1	
Juvenile neuronal ceroid lipofuscinosis	0.46	
Juvenile Paget's disease		50 cases
Juvenile polyposis of infancy		11 cases
Juvenile psoriatic arthritis	4.2	
Juvenile rheumatoid factor-negative polyarthritis	8	
Juvenile rheumatoid factor-positive polyarthritis	4.2	
Juvenile temporal arteritis		20 cases
Kabuki syndrome	1.16	
Kaler-Garrity-Stern syndrome		2 cases
Kallmann syndrome	7.7	
Kallmann syndrome - heart disease		8 cases
Kaposi's sarcoma	1.7	
Kapur-Toriello syndrome		4 cases
Kasabach-Merritt syndrome		> 175 cases
KBG syndrome		45 cases
Kearns-Sayre syndrome	2	
Kennedy disease	3.3	
Keratoderma - hypotrichosis - leukonychia		2 cases
Keratosis follicularis - dwarfism - cerebral atrophy		6 cases
Keratosis palmaris et plantaris - clinodactyly		< 20 cases
Keratosis palmoplantaris - esophageal carcinoma		< 10 families
Keratosis, Nagashima-type		20 cases
Ketoacidosis due to betaketothiolase deficiency		60 cases
KID syndrome		> 100 cases
Kimura disease		200 cases
Kozlowski-Brown-Hardwick syndrome		2 cases
Krabbe disease	0.75**	
Kumar-Levick syndrome		1 family
Lacrimo-auriculo-dento-digital syndrome		20 cases
Lafora disease	< 0.1	
Lambert-Eaton myasthenic syndrome	1	
Lamellar ichthyosis	> 0.33	
Laminopathy type Decaudain-Vigouroux		9 cases
Langerhans cell histiocytosis	2	
Large congenital melanocytic nevus	2	
Laron syndrome	0.2	
Laron syndrome with immunodeficiency		< 10 cases
Larsen syndrome		100 cases
Laryngeal abductor paralysis - intellectual deficit		< 20 cases
Laryngo-tracheo-esophageal cleft	1.5	
Late infantile neuronal ceroid lipofuscinosis	1.3	
Lathosterolosis		< 5 cases
LCAT deficiency		30 families
Leber hereditary optic neuropathy	6.5	
Legg-Calve-Perthes disease	23	
Leigh syndrome	2.75**	
Lelis syndrome		8 cases
Lemierre syndrome	0.1	
Lennox-Gastaut syndrome	15	
Lenz-Majewski hyperostotic dwarfism		9 cases
LEOPARD syndrome		200 cases
Leprechaunism	0.1**	
Leptospirosis	0.24	
Lesch-Nyhan syndrome	0.38	
Lethal ataxia with deafness and optic atrophy		12 cases
Lethal bone dysplasia, Holmgren type		4 cases
Lethal faciocardiomelic dysplasia		3 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Lethal hemolytic anemia - genital anomalies		2 cases
Lethal Kniest-like dysplasia		2 cases
Lethal Larsen-like syndrome		< 10 cases
Lethal multiple pterygium syndrome		200 cases
Lethal omphalocele-cleft palate syndrome		3 cases
Lethal osteosclerotic bone dysplasia		8 families
Lethal polymalformative syndrome, Boissel type		8 cases
Lethal recessive chondrodysplasia		4 cases
Lethal restrictive dermopathy		30 cases
Letterer-Siwe disease	0.2	
Leukocyte adhesion deficiency		< 350 cases
Leukocyte adhesion deficiency type II		< 10 cases
Leukocyte adhesion deficiency type III		17 cases
Leukodystrophy - spastic paraplegia - dystonia		9 cases
Leukoencephalopathy - ataxia - hypodontia - hypomyelination		8 cases
Leukoencephalopathy - dystonia - motor neuropathy		2 cases
Leukoencephalopathy - metaphyseal chondrodysplasia		4 cases
Leukoencephalopathy - palmoplantar keratoderma		4 cases
Leukoencephalopathy with bilateral anterior temporal lobe cysts		29 cases
Leukoencephalopathy with brain stem and spinal cord involvement - lactate elevation		39 cases
Leukonychia totalis - acanthosis-nigricans-like lesions - abnormal hair		11 cases
Lewis-Pashayan syndrome		3 cases
Lewis-Sumner syndrome	0.9	
Lhermitte-Duclos disease		220 cases
Lichstenstein syndrome		2 cases
Liddle syndrome		80 cases
Li-Fraumeni syndrome		400 families
Ligneous conjunctivitis	1.1	
Limb body wall complex	2**	
Limb-girdle muscular dystrophy	0.8	
Limb-mammary syndrome		27 cases
Limited cutaneous systemic sclerosis	8	
Linear atrophoderma of Moulin		< 30 cases
Lipodystrophy - intellectual deficit - deafness		3 cases
Lipoid proteinosis		> 280 cases
Lissencephaly due to TUBA1A mutation		< 15 cases
Lissencephaly type 2	0.12	
Lissencephaly type 3 - familial fetal akinesia sequence		5 cases
Lissencephaly type 3 - metacarpal bone dysplasia		2 cases
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	1	
Lopez-Hernandez syndrome		11 cases
Low birth weight - dwarfism - dysgammaglobulinemia		2 cases
Lowry-Wood syndrome		< 10 cases
Lung fibrosis - immunodeficiency - 46,XX gonadal dysgenesis		2 cases
Lymphangioleiomyomatosis	0.56	
Lymphatic malformation	12.5	
Lymphedema - atrial septal defects - facial changes		3 cases
Lymphedema - cerebral arteriovenous anomaly		5 cases
Macrocephaly - capillary malformation		116 cases
Macrocephaly - immune deficiency - anemia		2 cases
Macrocephaly - short stature - paraplegia		2 cases
Macrocephaly-autism syndrome		< 40 cases
Macrophagic myofasciitis	1	
Macrostomia - preauricular tags - external ophthalmoplegia		9 cases
Madras motor neuron disease		154 cases
Maffucci syndrome		250 cases
Malakoplasia		> 700 cases
Malaria	3	
Malignant atrophic papulosis		> 200 cases
Malignant hyperthermia	33	
Malignant hyperthermia - arthrogryposis - torticollis		4 cases
Malignant peritoneal mesothelioma	1.5	
Malignant tumor of fallopian tube	1	
Malonic aciduria		17 cases
Mandibuloacral dysplasia		37 cases
Mantle cell lymphoma	4	
Maple syrup urine disease	15.6	
Marden-Walker syndrome		30 cases
Marfan syndrome	20	
Marie Unna congenital hypotrichosis		12 families
Marinesco-Sjögren syndrome		200 cases
Marshall's syndrome with periodic fever		30 cases
Marshall-Smith syndrome		33 cases
Martinez-Frias syndrome		11 cases
MASA syndrome	3.5	
Mastocytosis	10	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Maternal hyperphenylalaninemia	1.25	
Maternally inherited diabetes and deafness	0.1	
Matthew-Wood syndrome		5 cases
Mayer-Rokitansky-Küster-Hauser syndrome	9	
Mazabraud syndrome		54 cases
McCune-Albright syndrome	0.55	
McLeod neuroacanthocytosis syndrome		150 cases
Meacham syndrome		< 15 cases
Meckel syndrome	4**	
Meconium aspiration syndrome	2.44	
Median cleft lip/mandibule		70 cases
Medium chain acyl-CoA dehydrogenase deficiency	15	
MEDNIK syndrome		4 families
Medullary thyroid carcinoma	7	
Megacystis - microcolon - intestinal hypoperistalsis - hydronephrosis		89 cases
Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus		6 cases
MEHMO syndrome		7 cases
MELAS syndrome	16	
Melorheostosis		300 cases
Mendelian susceptibility to atypical mycobacteria	0.059	
Meniere disease	42.5	
Meningococcal meningitis	10	
MERRF syndrome	0.9	
Mesoaxial synostotic syndactyly with phalangeal reduction		2 families
Mesomelic dysplasia - skin dimples		2 cases
Mesothelioma	3.1	
Metachondromatosis		25 cases
Metachromatic leukodystrophy	0.16	
Metaphyseal acroscyphodysplasia		4 cases
Metaphyseal anadysplasia		27 cases
Metaphyseal chondrodysplasia - retinitis pigmentosa		2 cases
Metaphyseal chondrodysplasia, Jansen type		16 cases
Metaphyseal chondrodysplasia, Kaitila type		2 cases
Metatropic dwarfism		80 cases
Methimazole embryofetopathy		40 cases
Methylcobalamin deficiency type cbl E		27 cases
Methylcobalamin deficiency, cbl G type		33 cases
Methylmalonic acidemia - homocystinuria		300 cases
Methylmalonic aciduria - microcephaly - cataract		2 cases
Methylmalonicacidemia - homocystinuria, type cbl C		300 cases
Methylmalonicacidemia - homocystinuria, type cbl D		5 cases
Methylmalonicacidemia - homocystinuria, type cbl F		9 cases
Mevalonic aciduria		30 cases
Michels syndrome		7 cases
Micro syndrome		8 cases
Microbrachycephaly - ptosis - cleft lip		2 cases
Microcephalic osteodysplastic dysplasia, Saul-Wilson type		4 cases
Microcephalic osteodysplastic primordial dwarfism types 1 and 3		< 30 cases
Microcephaly - brachydactyly - kyphoscoliosis		3 cases
Microcephaly - cardiomyopathy		3 cases
Microcephaly - cleft palate		3 cases
Microcephaly - digital anomalies - intellectual deficit		2 cases
Microcephaly - glomerulonephritis - marfanoid habitus		2 cases
Microcephaly - intellectual deficit - phalangeal and neurological anomalies		3 cases
Microcephaly - polymicrogyria - corpus callosum agenesis		4 cases
Microcephaly - seizures - intellectual deficit - heart disease		2 cases
Microcytic anemia with liver iron overload		3 cases
Microgastria - limb reduction defect		16 cases
Microlissencephaly - micromelia		2 cases
Microphthalmia - brain atrophy		3 cases
Microphthalmia with brain and digit anomalies		2 families
Microphthalmia with limb anomalies		30 cases
Microtia	15	
Microtia - eye coloboma - imperforation of the nasolacrimal duct		1 family
MIDAS syndrome		< 50 cases
Midface retraction syndrome, Schinzel-Giedion type		34 cases
Mild hemophilia A	0.44	
Mild hemophilia B	0.6	
Miller-Dieker syndrome	0.3	
Mirror polydactyly - vertebral segmentation - limbs defects	0.3	
Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria		2 cases
Mitochondrial myopathy and sideroblastic anemia		7 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies	9	
Mitral regurgitation - deafness - skeletal anomalies		3 cases
Mixed connective tissue disease	3.2	
Mixed cryoglobulinemia	1	
Mixed dystonia		3 families
Moderately severe hemophilia A	0.22	
Moderately severe hemophilia B	0.6	
Moebius syndrome		300 cases
Mohr-Tranebjaerg syndrome		46 cases
Monoclonal Ig light chain-associated Fanconi syndrome		100 cases
Mononen-Karnes-Senac syndrome		5 cases
Monosomy 18p		< 200 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		< 200 cases
Moyamoya 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		< 40 cases
Muenke syndrome	1.8**	
Muir-Torre syndrome		205 cases
MULIBREY nanism		115 cases
Mullerian derivatives - lymphangiectasia - polydactyly		3 cases
Multicentric reticulohistiocytosis		< 200 cases
Multifocal motor neuropathy with conduction block	1.5	
Multiple endocrine neoplasia type 1	11	
Multiple endocrine neoplasia type 2	3.3	
Multiple epiphyseal dysplasia	5	
Multiple myeloma	17.5	
Multiple osteochondromas	2	
Multiple sclerosis - ichthyosis - factor VIII deficiency		2 cases
Multiple sulfatase deficiency		50 cases
Multiple synostoses		20 families
Multiple system atrophy	4.6	
MURCS association	11.25	
Muscle phosphofructokinase deficiency		< 30 cases
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus		10 cases
Myasthenia gravis	20	
Myelodysplastic syndromes	22.8	
Myelofibrosis with myeloid metaplasia	2.7	
Myhre syndrome		16 cases
Myoclonic dystonia 15		< 20 cases
Myoclonic epilepsy of infancy		106 cases
Myoclonus - cerebellar ataxia - deafness		4 cases
Myoneurogastrointestinal encephalopathy syndrome		87 cases
Myopathy due to calsequestrin and SERCA1 protein overload		4 cases
N syndrome		3 cases
Naegeli-Franceschetti-Jadassohn syndrome	0.035	
Nail patella-like - renal disease		3 cases
Nail-patella syndrome	2	
Nance-Horan syndrome		50 families
Narcolepsy-cataplexy	26	
NARP syndrome	8.5	
Nasopalpebral lipoma - coloboma - telecanthus		< 30 cases
Nemaline myopathy	1	
Neonatal diabetes - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys		2 cases
Neonatal diabetes mellitus	0.2	
Neonatal hemochromatosis		100 cases
Neonatal hypoxic and ischemic brain injury	9	
Neonatal ichthyosis - sclerosing cholangitis		< 10 cases
Neovascular glaucoma	24.4	
Nephroblastoma	10.1	
Nephrogenic diabetes insipidus	0.5	
Nephropathy - deafness - hyperparathyroidism		5 cases
Nephrosis - deafness - urinary tract - digital malformations		5 cases
Netherton disease	1.35	
Neu-Laxova syndrome		60 cases
Neuroaxonal dystrophy - renal tubular acidosis		3 cases
Neuroblastoma	11.3	
Neurocutaneous syndrome, Bicknell type		4 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency		4 cases
Neurodegeneration with brain iron accumulation	2	
Neuroectodermal syndrome, Johnson type		< 30 cases
Neuroectodermal-endocrine syndrome		4 cases
Neurofibromatosis type 1	25	
Neurofibromatosis type 2	0.5	
Neuroleptic malignant syndrome	15	
Neurologic Waardenburg-Shah syndrome		< 30 cases
Neurometabolic disorder due to serine deficiency		< 30 cases
Neuromyelitis optica	1.5	
Neuropathy with hearing impairment		1 family
Neutral lipid storage disease		50 cases
Nevo syndrome		10 cases
Niemann-Pick disease	2.5**	
Niemann-Pick disease type A	0.25**	
Niemann-Pick disease type B	0.4	
Niemann-Pick disease type C	0.85	
Nijmegen breakage syndrome	1**	
Nodular regenerative hyperplasia of the liver	3	
Nonacquired combined pituitary hormone deficiency	37.7	
Non-distal trisomy 12p	2**	
Non-papillary transitional cell carcinoma of the bladder	37	
Nonspherocytic hemolytic anemia due to hexokinase deficiency		17 families
Noonan syndrome	50	
Norrie disease		300 cases
North Carolina macular dystrophy		2 families
Not NOTCH3-related small vessel disease of the brain		2 cases
Obesity - colitis - hypothyroidism - cardiac hypertrophy - developmental delay		2 cases
Obesity due to congenital leptin deficiency		< 30 cases
Obesity due to melanocortin-4 receptor deficiency	50	
Obesity due to prohormone convertase-I deficiency		2 cases
Obesity due to pro-opiomelanocortin deficiency		7 cases
Ochoa syndrome		> 100 cases
Ocular albinism - late-onset sensorineural deafness		7 cases
Ocular coloboma	1	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Ocular motor apraxia, Cogan type		50 cases
Oculocerebrocutaneous syndrome		36 cases
Oculocerebrofacial syndrome, Kaufman type		9 cases
Oculocerebrorenal syndrome	0.19	
Oculocutaneous albinism	7.15	
Oculodental syndrome, Rutherfurd type		1 family
Oculodentodigital dysplasia		243 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	
Oculotrichodysplasia		2 cases
Odontoleukodystrophy		4 cases
Odontomicronychial dysplasia		5 cases
Odonto-onycho-dermal dysplasia		< 15 cases
Odontotrichomelic syndrome		4 cases
Odonto-tricho-ungual-digito-palmar syndrome		21 cases
Okamoto syndrome		2 cases
Oligoarticular juvenile arthritis	20.5	
Oligocone trichromacy		14 cases
Olmsted syndrome		32 cases
Omodysplasia		30 cases
Omphalocele	12	
Ondine syndrome	2.25	
Onycho-tricho-dysplasia - neutropenia		5 cases
Opitz BBB/G syndrome	3	
Opsismodysplasia		25 cases
Optic atrophy	6	
Oral-facial-digital syndrome type 1	1.2	
Oral-facial-digital syndrome type 3		3 cases
Oral-facial-digital syndrome type 4		16 cases
Oral-facial-digital syndrome type 5		4 cases
Oral-facial-digital syndrome type 8		2 families
Orbital leiomyoma		16 cases
Ornithine transcarbamylase deficiency	1.4	
Ossification anomalies - psychomotor development delay		2 cases
Osteochondritis dissecans	35	
Osteochondrodysplatic nanism - deafness - retinitis pigmentosa		2 cases
Osteocraniostenosis		12 cases
Osteodysplasty, Melnick-Needles type		> 50 cases
Osteogenesis imperfecta	6.5	
Osteogenesis imperfecta - retinopathy - seizures - intellectual deficit		2 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Osteopathia striata - cranial sclerosis		100 cases
Osteopetrosis - hypogammaglobulinemia		8 cases
Osteopetrosis with renal tubular acidosis		50 cases
Osteoporosis - pseudoglioma	0.05	
Osteoporosis-oculocutaneous-hypopigmentation syndrome		3 cases
Osteosarcoma	5	
Osteosclerosis - ichthyosis - premature ovarian failure		3 cases
Otodental syndrome		9 families
Otopalatodigital syndrome		30 cases
Otospondylomegaepiphyseal dysplasia		< 30 cases
Overhydrated hereditary stomatocytosis		20 families
Ovotesticular disorder of sex development		> 500 cases
P2Y12 deficiency		5 cases
Pachydermoperiostosis		204 cases
Pachyonychia congenita		230 cases
Pacman dysplasia		< 10 cases
PAGOD syndrome		6 cases
Pai syndrome		> 10 cases
Pallister-Hall syndrome		100 cases
Palmoplantar keratoderma - amyotrophy		4 cases
Palmoplantar keratoderma - deafness		< 10 families
Palmoplantar keratoderma - spastic paralysis		25 cases
Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma		5 cases
Palmoplantar porokeratosis of Mantoux		< 10 cases
Pancreatic carcinoma	11.9	
Pancreatic hypoplasia - diabetes - heart disease		< 10 cases
Pancreatoblastoma		60 cases
Pantothenate-kinase-associated neurodegeneration	1.5	
Papillon-Lefèvre syndrome	0.25	
Paraneoplastic pemphigus		> 60 cases
Paraplegia - brachydactyly - cone-shaped epiphysis		5 cases
Paraplegia - intellectual deficit - hyperkeratosis		4 cases
PARC syndrome		2 cases
Parietal foramina	5	
Paroxysmal extreme pain disorder		4 families
Paroxysmal hemicrania	2	
Paroxysmal nocturnal hemoglobinuria	0.55	
Paroxysmal non-kinesigenic dyskinesia	0.02	
Parsonage-Turner syndrome	3.3	
Partial acquired lipodystrophy		250 cases
Partial atrioventricular canal	20	
Partial chromosome Y deletion	42	
Partial pancreas agenesis		50 cases
Partington syndrome		2 families
Patent arterial duct	50	
Pearson syndrome		60 cases
Pelizaeus-Merzbacher disease	0.25	
PELVIS syndrome		11 cases
Pelviscapular dysplasia		4 cases
Pemphigus vulgaris	3.8	
Pendred syndrome	5.5	
Pericarditis - arthropathy - camptodactyly		< 30 families
Perinatal-lethal Gaucher disease	0.01	
Perioral myoclonia with absences		< 10 cases
Peripheral neuropathy, Fiskerstrand type		3 cases
Peripheral resistance to thyroid hormones	2.5	
Perlman syndrome		30 cases
Permanent congenital hypothyroidism	33.3	
Permanent neonatal diabetes mellitus - pancreatic and cerebellar agenesis		4 cases
Perrault syndrome		34 cases
Perry syndrome		9 families
Peters-plus syndrome		50 cases
Peutz-Jeghers syndrome	2.2	
Pfeiffer syndrome	1	
PHACE syndrome		100 cases
Phenylketonuria	4	
Phosphoenolpyruvate carboxykinase deficiency		< 10 cases
Phosphoglycerate kinase 1 deficiency		23 cases
Phosphoribosylpyrophosphate synthetase superactivity		< 30 families
PIBIDS syndrome		20 cases
Piebaldism	0.25	
Pierre Robin syndrome - faciodigital anomaly		2 cases
Pierson syndrome		22 cases
Pili torti - onychodysplasia		1 family
Pilodental dysplasia - refractive errors		2 cases
Pitt-Hopkins syndrome		50 cases
Pityriasis rubra pilaris		48 cases
Plummer-Vinson syndrome		25 cases
Pneumonia caused by serotype 01 Pseudomonas Aeruginosa	18	
Poikiloderma of Kindler		100 cases
Poland syndrome	2	
Pollitt syndrome		10 cases
Polyarteritis nodosa	3.07	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Polycystic lipomembranous osteodysplasia - sclerosing leukoencephalopathy	0.15	
Polycystic ovaries - urethral sphincter dysfunction		33 cases
Polycythemia vera	30	
Polymyositis	14.8	
Polysyndactyly - cardiac malformation		6 cases
Pontocerebellar hypoplasia type 1		6 families
Pontocerebellar hypoplasia type 2		< 30 cases
Pontocerebellar hypoplasia type 4		3 cases
Pontocerebellar hypoplasia type 5		3 cases
Pontocerebellar hypoplasia type 6		3 cases
Porokeratotic eccrine ostial and dermal duct nevus		25 cases
Porphyria cutanea tarda	4	
Postaxial acrofacial dysostosis		< 30 cases
Posterior fusion of lumbosacral vertebrae - blepharoptosis		3 cases
Post-transplant lymphoproliferative disease	26.2	
Potocki-Shaffer syndrome		23 cases
Prader-Willi syndrome	10.7	
Preaxial polydactyly	25	
Primary biliary cirrhosis	13.5	
Primary ciliary dyskinesia	5	
Primary congenital hypothyroidism	37.5	
Primary erythermalgia		30 families
Primary hyperoxaluria type 1	0.2	
Primary immunodeficiency syndrome due to p14 deficiency		4 cases
Primary lateral sclerosis	1.5	
Primary peritoneal tumor	3	
Primary sclerosing cholangitis	11	
Progeria - short stature - pigmented nevi		< 10 cases
Progressive bifocal chorioretinal atrophy		2 families
Progressive bulbar paralysis of childhood		< 40 cases
Progressive cavitating leukoencephalopathy		19 cases
Progressive neurodegeneration - joint laxity - cataract		2 cases
Progressive non-fluent aphasia	2.5	
Progressive non-infectious anterior vertebral fusion		10 cases
Progressive supranuclear palsy	6	
Progressive supranuclear palsy - corticobasal syndrome	< 0.6	
Prolidase deficiency		50 cases
Propionic acidemia	3.75	
Proteus syndrome		200 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Proximal myotonic myopathy	1	
Proximal spinal muscular atrophy	3	
Proximal spinal muscular atrophy type 1	1.25	
Proximal spinal muscular atrophy type 2	1.42	
Proximal spinal muscular atrophy type 3	0.26	
Proximal spinal muscular atrophy type 4	0.32	
Pseudoachondroplasia	1.6	
Pseudodiastrophic dysplasia		10 cases
Pseudohypoaldosteronism type 1		70 cases
Pseudomyxoma peritonei	1	
Pseudoprogeria syndrome		2 cases
Pseudoxanthoma elasticum	2.5	
Pseudo-Zellweger syndrome		< 10 cases
Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency		3 cases
Pterygium colli - intellectual deficit - digital anomalies		2 cases
Ptosis - strabismus - ectopic pupils		1 family
Pulmonary fibrosis - hepatic hyperplasia - bone marrow hypoplasia		4 cases
Pure autonomic failure	1	
Pycnodysostosis	0.13	
Pyknoachondrogenesis		5 cases
Pyle disease		< 30 cases
Pyogenic arthritis - pyoderma gangrenosum - acne		34 cases
Pyridoxine-dependent epilepsy	0.15	
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
Rare bone tumor	10	
Rare ovarian cancer	27.8	
Rasmussen subacute encephalitis		> 100 cases
Recessive aplasia cutis congenita of limbs		6 cases
Recessive hereditary methemoglobinemia type 2		< 100 cases
Recurrent acute pancreatitis	10	
Reducing body myopathy		4 families
Refsum disease	0.1	
Regional odontodysplasia		139 cases
Relapsing polychondritis	0.35	
Renal cell carcinoma	35.8	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts		10 cases
Rendu-Osler-Weber disease	16.25	
Renpenning syndrome		10 families
Resistance to thyrotropin-releasing hormone syndrome		2 cases
Retinal arteries tortuosity		100 cases
Retinal degeneration - nanophthalmos - glaucoma		7 cases
Retinitis pigmentosa	30.2	
Retinitis pigmentosa - intellectual deficit - deafness - hypogenitalism		2 families
Retinoblastoma	5.4	
Retinohepatoendocrinologic syndrome		7 cases
Rett syndrome	4.15	
Rhabdoid tumor		500 cases
Rheumatic fever	5	
Rhombencephalosynapsis		50 cases
RHYNS syndrome		4 cases
Rickettsialpox		> 800 cases
Ring chromosome 1		34 cases
Ring chromosome 10		< 20 cases
Ring chromosome 14		50 cases
Ring chromosome 17		14 cases
Ring chromosome 18		70 cases
Ring chromosome 20		> 50 cases
Ring dermoid of cornea		< 30 cases
Roberts syndrome		< 150 cases
Robinow syndrome		200 cases
Robinow-like syndrome		2 cases
Rolled and spiral hairs - palmoplantar keratoderma		4 cases
Romano-Ward syndrome	40	
Rothmund-Thomson syndrome		300 cases
Rubella panencephalitis		> 20 cases
Rubinstein-Taybi syndrome	1	
Rudiger syndrome		2 cases
Saethre-Chotzen syndrome	3	
Sakati-Nyhan syndrome		< 5 cases
Saldino-Mainzer syndrome		10 cases
Sandhoff disease	0.75	
Sanfilippo syndrome type A	0.3	
Sarcoidosis	15	
Sarcosinemia	2	
Say-Barber-Miller syndrome		2 cases
Scalp defects - postaxial polydactyly		2 cases
Scalp-ear-nipple syndrome		30 cases
SCARF syndrome		2 cases
Scheie syndrome	0.2	
Schilbach-Rott syndrome		13 cases
Schimke immuno-osseous dysplasia		50 cases
Schizencephaly	1.54	
Schizophrenia - intellectual deficit - deafness - retinitis		1 family
Schnitzler syndrome		100 cases
Schopf-Schulz-Passarge syndrome		19 cases
Schwartz-Jampel syndrome		100 cases
Scimitar syndrome	2	
Scleroderma	42	
Sea-blue histiocytosis		60 cases
Sebastian syndrome		< 10 families
Seborrhea-like dermatitis with psoriasiform elements		44 cases
Seckel syndrome		100 cases
Secondary amyloidosis	17	
Segmental odontomaxillary dysplasia		32 cases
Seizures - intellectual deficit due to hydroxylysinuria		3 cases
Senior-Loken syndrome	0.1	
Sensenbrenner syndrome		15 cases
Sensorineural hearing loss - early greying - essential tremor		3 cases
SERKAL syndrome		3 cases
Severe achondroplasia - developmental delay - acanthosis nigricans		4 cases
Severe combined immunodeficiency due to adenosine deaminase deficiency	0.22	
Severe congenital neutropenia	0.4	
Severe hemophilia A	0.44	
Severe hemophilia B	0.8	
Severe intellectual deficit - epilepsy - anal anomalies - distal phalangeal hypoplasia		2 cases
Severe neonatal-onset encephalopathy with microcephaly		< 30 cases
Severe X-linked intellectual deficit, Gustavson type		7 cases
Sezary's syndrome	0.18	
Short bowel syndrome	3.4	
Short fifth metacarpals - insulin resistance		1 family
Short stature - intellectual deficit - eye anomalies - cleft lip/palate		3 cases
Short stature - pituitary and cerebellar defects - small sella turcica		1 family
Short stature - webbed neck - heart disease		4 cases
Short stature due to growth hormone qualitative anomaly		3 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
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**	
Sick sinus syndrome		11 cases
Sickle cell anemia	15	
Siegler-Brewer-Carey syndrome		2 cases
Silent sinus syndrome		98 cases
Sillence syndrome		5 cases
Silver-Russell syndrome		400 cases
Simpson-Golabi-Behmel syndrome		> 100 cases
Simpson-Golabi-Behmel syndrome type 2		4 cases
Singleton-Merten dysplasia		< 10 cases
Sinus histiocytosis with massive lymphadenopathy		500 cases
Sirenomelia	1**	
Sitosterolemia		40 cases
Sjögren-Larsson syndrome	0.4	
Skeletal dysplasia - intellectual deficit		2 families
Small cell lung cancer	11.2	
Smith-Lemli-Opitz syndrome	3.3	
Smith-Magenis syndrome	5.3	
Soft tissue sarcomas	20	
Solar urticaria	3.6	
Sotos syndrome	7**	
Sparse hair - short stature - skin anomalies		4 cases
Spastic paraplegia - glaucoma - intellectual deficit		2 families
Spastic paraplegia - nephritis - deafness		4 cases
Spastic paraplegia - precocious puberty		2 cases
Spastic quadriplegia - retinitis pigmentosa - intellectual deficit		2 cases
Spasticity - intellectual deficit - X-linked epilepsy		6 cases
Speech-language disorder type 1		22 cases
Spinal muscular atrophy - Dandy-Walker complex - cataracts		2 cases
Spinocerebellar ataxia type 1	1.5	
Spinocerebellar ataxia type 2	1.5	
Spinocerebellar ataxia type 29		< 50 cases
Spinocerebellar ataxia type 3	1	
Spinocerebellar ataxia type 30		6 cases
Split hand - split foot	1.1	
Split hand - split foot - deafness		22 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Split hand - urinary anomalies - spina bifida		3 cases
Spondyloenchondrodysplasia		36 cases
Spondyloepimetaphyseal dysplasia - abnormal dentition		2 cases
Spondyloepimetaphyseal dysplasia - hypotrichosis		5 cases
Spondyloepimetaphyseal dysplasia, aggrecan type		3 cases
Spondyloepimetaphyseal dysplasia, Bieganski type		3 cases
Spondyloepimetaphyseal dysplasia, Genevieve type		2 cases
Spondyloepiphyseal dysplasia congenita	0.34	
Spondyloepiphyseal dysplasia tarda	0.55	
Spondyloepiphyseal dysplasia tarda, Kohn type		3 cases
Spondyloepiphyseal dysplasia, Byers type		4 cases
Spondyloepiphyseal dysplasia, Cantu type		4 cases
Spondyloepiphyseal dysplasia, MacDermot type		4 cases
Spondyloepiphyseal dysplasia, Nishimura type		4 cases
Spondyloepiphyseal dysplasia, Reardon type		1 family
Spondylometaphyseal dysplasia	1	
Spondylometaphyseal dysplasia - bowed forearms - facial dysmorphism		2 cases
Spondylometaphyseal dysplasia - cone-rod dystrophy		8 cases
Spondylometaphyseal dysplasia with combined immunodeficiency		4 cases
Spondylometaphyseal dysplasia, A4 type		2 cases
Spondylometaphyseal dysplasia, Golden type		3 cases
Spondylometaphyseal dysplasia, Kozlowski type	0.1	
Spontaneous periodic hypothermia		< 30 cases
Sporadic inclusion body myositis	0.49	
Sporotrichosis		55 cases
Squamous cell carcinoma of head and neck	< 40	
Stapes ankylosis with broad thumbs and toes		6 families
Stargardt disease	10.4	
Steinert myotonic dystrophy	4.5	
Sternal cleft	< 2	
Stern-Lubinsky-Durrie syndrome		7 cases
Steroid dehydrogenase deficiency - dental anomalies		1 family
Stickler syndrome	13.5	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Stiff-man syndrome	0.1	
Stimmler syndrome		2 cases
Stoll-Alembik-Finck syndrome		2 cases
Stormorken-Sjaastad-Langslet syndrome		6 cases
Stromal keratitis	16	
Suarez-Stickler syndrome		3 cases
Subcorneal pustular dermatosis		200 cases
Succinic acidemia		50 cases
Succinyl-CoA acetoacetate transferase deficiency		10 cases
Sudden infant death - dysgenesis of the testes		21 cases
Summitt syndrome		3 cases
Superficial pemphigus	1.2	
Supravalvular aortic stenosis	12.5	
Susac syndrome		< 100 cases
Symmetrical thalamic calcifications		29 cases
Symphalangism with multiple anomalies of hands and feet		6 cases
Syndactyly - telecanthus - anogenital and renal malformations		6 cases
Syndactyly type 1	25	
Syndactyly type 4		4 cases
Syndactyly, Cenani-Lenz type		< 30 cases
Syndromic microphthalmia type 5		20 cases
Syndromic X-linked intellectual deficit 7		10 cases
Syndromic X-linked intellectual deficit due to JARID1C mutation		< 10 families
Syngnathia multiple anomalies		2 cases
Synspondylism		24 cases
Syringomyelia	8.4	
Systemic mastocytosis	3.3	
Systemic sclerosis	21.5	
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**	
Teebi-Shaltout syndrome		2 cases
Temtamy syndrome		3 cases
Terminal osseous dysplasia - pigmentary defects		18 cases
Tetralogy of Fallot	45	
Thalidomide embryopathy		5000 cases
Thanatophoric dwarfism	3.5**	
Thiamine-responsive megaloblastic anemia syndrome		30 families
Thickened earlobes - conductive deafness		2 families
Thomsen and Becker disease	5	
Thoracolaryngopelvic dysplasia		< 10 cases
Thrombocytopenia - Robin sequence		2 cases
Thrombotic thrombocytopenic purpura	24.6	
Thumb stiffness - brachydactyly - intellectual deficit		6 cases
Thymic-renal-anal-lung dysplasia		3 cases
Thyrocerebrorenal syndrome		2 cases
Thyroid ectopia	14.2	
Thyroid hemiagenesis	25	
Thyroid hypoplasia	3.5	
Thyrotoxic periodic paralysis		139 cases
Tibial aplasia - ectrodactyly	0.1	
Tibial hemimelia	0.1	
Tibial muscular dystrophy	6	
Tietz syndrome		1 family
Tomé-Brunet-Fardeau syndrome		4 cases
Torg-Winchester syndrome		12 cases
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		< 100 cases
Transaldolase deficiency		7 cases
Transmissible spongiform encephalopathies	0.3	
Transposition of the great arteries	32.5	
Treacher-Collins syndrome	6	
Treft-Sanborn-Carey syndrome		23 cases
Trichodental syndrome		< 5 families
Tricho-dento-osseous syndrome		> 30 cases
Trichodysplasia - amelogenesis imperfecta		1 family
Trichomegaly - cataract - hereditary spherocytosis		2 cases
Trichomegaly - retina pigmentary degeneration - dwarfism		11 cases
Tricho-odonto-onychial dysplasia		4 cases
Tricho-retino-dento-digital syndrome		9 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 - short stature - developmental delay		3 cases
Triose phosphate-isomerase deficiency		30 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Triphalangeal thumbs - brachyectrodactyly		4 families
Triple A syndrome		100 cases
Triple H syndrome	12	
Trisomy 13	13**	
Trisomy 18	9**	
Trisomy 8q		> 30 cases
Trisomy X	42.5	
Tritanopia	4.8	
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		< 150 cases
Uhl anomaly		84 cases
Ulbright-Hodes syndrome		3 cases
Ulnar/fibula ray defect - brachydactyly		1 family
Ulnar-mammary syndrome		< 10 families
Umbilical cord ulceration - intestinal atresia		15 cases
Unilateral adactylia	34	
Unverricht-Lundborg disease	0.2	
Upington disease		1 family
Usher syndrome	3.5	
Usher syndrome type 1	1.3	
Usher syndrome type 2	2	
Uveal coloboma - cleft lip and palate - intellectual deficit		12 cases
UV-sensitive syndrome		6 cases
VACTERL with hydrocephalus		< 10 families
Van den Bosch syndrome		1 family
Van Der Woude syndrome	2	
Vasculitis	6.3	
Vernal keratoconjunctivitis	21	
Vici syndrome		8 cases
Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay		2 cases
Vitamin B12 responsive methylmalonic acidemia type cbl A		60 cases
Von Hippel-Lindau disease	1.9	
Von Willebrand disease	12.5	
Vulvovaginal gingival syndrome		127 cases
W syndrome		6 cases
Waardenburg syndrome	2.4	
Waardenburg-Shah syndrome		50 cases
Waldenström macroglobulinemia	2.6	
Walker-Warburg syndrome	1.65**	
Weaver syndrome		30 cases
Weaver-Williams syndrome		> 30 cases
Wegener granulomatosis	6.6	
Weill-Marchesani syndrome		128 cases
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
Williams syndrome	13.3	
Wilson disease	5.84	
Wilson-Turner syndrome		> 14 cases
Wiskott-Aldrich syndrome	0.15	
Wolcott-Rallison syndrome		< 60 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
Worster-Drought syndrome	3.7	
Wrinkly skin syndrome		< 30 cases
Xanthinuria		150 cases
Xeroderma - talipes - enamel defects		2 cases
Xeroderma pigmentosum	0.1	
XK aprosencephaly		< 10 cases
X-linked adrenoleukodystrophy	5	
X-linked agammaglobulinemia	0.45	
X-linked Charcot-Marie-Tooth disease	1.6	
X-linked complicated corpus callosum dysgenesis		11 cases
X-linked creatine transporter deficiency		17 cases
X-linked diffuse leiomyomatosis - Alport syndrome	0.1	
X-linked dominant chondrodysplasia punctata	0.5	
X-linked Ehlers-Danlos syndrome		2 families
X-linked hereditary sensory and autonomic neuropathy with deafness		1 family
X-linked ichthyosis	16.6	
X-linked immune dysregulation - polyendocrinopathy - enteropathy		7 families
X-linked immunoneurologic disorder		5 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
X-linked intellectual deficit - ataxia - apraxia		9 cases
X-linked intellectual deficit, Najm type		5 cases
X-linked intellectual deficit, Stoll type		4 cases
X-linked lymphoproliferative disease	0.1	
X-linked mandibulofacial dysostosis		7 cases
X-linked myopathy with excessive autophagy		15 families
X-linked myopathy with postural muscle atrophy		1 family
X-linked neurodegenerative syndrome, Bertini type		7 cases
X-linked neurodegenerative syndrome, Hamel type		10 cases
X-linked recessive intellectual deficit - macrocephaly - ciliary dysfunction		1 family
X-linked recessive ocular albinism	2	
X-linked retinoschisis	4.5	
X-linked severe congenital neutropenia		7 cases
X-linked sideroblastic anemia		< 200 cases
X-linked sideroblastic anemia - ataxia		5 families
X-linked spastic paraplegia type 2		< 100 cases
X-linked spastic paraplegia type 16		1 family
X-linked spastic paraplegia type 34		24 cases
X-linked spinocerebellar ataxia type 3		5 cases
Young adult-onset Parkinsonism	15	
Young-Simpson syndrome		5 cases
Zellweger syndrome	1.1	
Zellweger-like syndrome without peroxisomal anomalies		2 cases
Zollinger-Ellison syndrome	5.3	
Zunich-Kaye syndrome		6 cases

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

Editor-in-chief: Ségolène Aymé ● Editor of the report : Catherine Gonthier ● Visual design : Céline Angin

The correct form when quoting this document is :

« Prevalence of rare diseases: Bibliographic data », Orphanet Report Series, *Rare Diseases collection*, November 2011, Number 1 : Listed in alphabetical order of diseases, http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_alphabetical_list.pdf

** Prevalence at birth