

**Prevalence of rare diseases :
Bibliographic data**

Listed in alphabetical order of diseases

Method

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

Search strategy

The search strategy is carried out using several data sources:

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

Collected data

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

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

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

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

Updated Data

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

Limitation of the study

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

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

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
2,8 dihydroxyadenine urolithiasis	1.7	
3C syndrome		25 cases
3-hydroxy 3-methylglutaryl-CoA (HMG) synthase deficiency		6 cases
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency of	1	
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
4-hydroxybutyricaciduria		350 cases
5-oxoprolinase deficiency		8 cases
6-pyruvoyl-tetrahydropterin synthase, deficiency		248 cases
6q terminal deletion		19 cases
Aarskog-Scott syndrome		> 200 cases
Aase-Smith syndrome		< 10 cases
Ablepharon macrostomia syndrome		15 cases
Abruzzo-Erickson syndrome		4 cases
Acalvaria	< 1**	
Acanthamoeba keratitis	1	
Acatlasemia	3.1	
Aceruloplasminemia	0.05	
Achalasia - microcephaly		5 cases
Achalasia, primary	10	
Acheiropodia		< 10 families
Achondrogenesis		> 100 cases
Achondroplasia	4.5	
Achromatopsia	3.33	
Ackerman syndrome		8 cases
Aconitase deficiency		19 cases
Acrocallosal syndrome, Schinzel type		34 cases
Acrocephalosyndactyly	4.6	
Acrocraniofacial dysostosis		2 cases
Acrodermatitis enteropathica, zinc deficiency type	0.2	
Acrofacial dysostosis postaxial, atypical		1 case
Acrofacial dysostosis, autosomal recessive		2 cases
Acrofacial dysostosis, Catania type		6 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Acrofacial dysostosis, Nager type		90 cases
Acrofacial dysostosis, palagonia type		4 cases
Acrofacial dysostosis, postaxial		< 30 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
Acute interstitial pneumonia	3.8	
Acute Respiratory Distress Syndrome, adult	30	
Acyl-CoA dehydrogenase, medium chain, deficiency of	15	
Adactylia unilateral	34	
Adamantinoma		513 cases
Adducted thumbs - arthrogryposis, Christian type		3 families
Adducted thumbs-arthrogryposis, Dunder type		5 cases
Adenosine monophosphate deaminase deficiency		200 cases
Adenylosuccinate lyase deficiency		50 cases
Adrenocortical carcinoma	1	
Adrenoleukodystrophy, X-linked	5	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
ADULT syndrome		14 cases
Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis		3 cases
Agammaglobulinemia, alymphocytotic type	0.35	
Agenesis of the corpus callosum - intellectual deficit - coloboma - micrognathia		2 cases
Agnathia holoprosencephaly situs inversus		30 cases
Agonadism - dextrocardia - diaphragmatic hernia		6 cases
Aicardi syndrome		200 cases
Aicardi-Goutieres syndrome		120 cases
Alagille syndrome	1.4	
Alar cartilages hypoplasia - coloboma - telecanthus		2 cases
Albers-Schonberg osteopetrosis	1	
Albinism ocular - late onset sensorineural deafness		7 cases
Albinism-deafness syndrome		1 family
Albright hereditary osteodystrophy	0.72	
Alexander disease		300 cases
Al-Gazali-Dattani syndrome		3 cases
Alkaptonuria	0.3	
Allan-Herndon-Dudley syndrome		89 cases
Alopecia - congenita keratosis palmoplantaris		1 family
Alopecia - contractures - dwarfism - intellectual deficit		5 cases
Alopecia - hypogonadism - extrapyramidal disorder		2 cases
Alopecia- epilepsy - pyorrhea - mental subnormality		12 cases
Alopecia totalis	10.5	
Alpers syndrome	0.025	
Alpha thalassemia - intellectual deficit, X-linked		168 cases
Alpha-1 antitrypsin deficiency	25	
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
Ambras syndrome		40 cases
Amelia, autosomal recessive		3 cases
Amelo-cerebro-hypohidrotic syndrome		19 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Aminopterin embryofetopathy		17 cases
Amniotic bands	4**	
Amoebiasis due to free-living amoebae	1.75	
Amyotrophic lateral sclerosis	5	
Androgen insensitivity syndrome	13	
ANE syndrome		5 cases
Anemia, sideroblastic, X-linked - ataxia		5 families
Anencephaly	3.2**	
Angelman syndrome	6.5	
Angel-shaped phalango-epiphyseal dysplasia		15 cases
Angioma neurocutaneous, hereditary		< 10 families
Angio-osteohypertrophic syndrome		1000 cases
Aniridia	1.75	
Aniridia - absent patella		3 cases
Aniridia - cerebellar ataxia - mental deficiency		> 10 families
Aniridia - ptosis - intellectual deficit - obesity, familial form		3 cases
Aniridia - renal agenesis - psychomotor retardation		2 cases
Aniridia-intellectual deficit syndrome		2 cases
Anisakiasis	3.8	
Ankyloblepharon - ectodermal defects - cleft lip palate		8 families
Ankyloblepharon filiforme - imperforate anus		2 families
Ankylosing vertebral hyperostosis with tylosis		8 cases
Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges		11 cases
Anonychia microcephaly		5 cases
Anonychia with flexural pigmentation		3 cases
Anophthalmia - hypothalamo-pituitary insufficiency		30 cases
Anophthalmia - megalocornea - cardiopathy - skeletal anomalies		3 cases
Anophthalmia - Microphthalmia, isolated	14	
Anophthalmia plus syndrome		4 cases
Anophthalmia/microphthalmia - esophageal atresia		14 cases
Anorchidia, bilateral	2.5	
Anosmia, isolated, congenital		< 15 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

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or 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
Aplasia cutis congenita of limbs recessive		6 cases
Aplastic anemia	0.4	
Arachnodactyly - intellectual deficit - dysmorphism		3 cases
Arachnodactyly - ossification abnormal - intellectual deficit		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
Arterial tortuosity		< 40 cases
Arthritis, enthesitis-related	5.7	
Arthritis, juvenile psoriatic	4.2	
Arthritis, oligoarticular juvenile	20.5	
Arthritis, systemic-onset, juvenile idiopathic	6.3	
Arthrogryposis - hyperkeratosis, lethal form		2 cases
Arthrogryposis - renal dysfunction - cholestasis		< 100 cases
Arthrogryposis multiplex congenita	30	
Arthrogryposis multiplex congenita - whistling face		10 cases
Arthrogryposis-like hand anomaly - sensorineural deafness		1 family
Ascher syndrome		50 cases
Astley-Kendall dysplasia		< 10 cases
Ataxia - apraxia - intellectual deficit, X-linked		9 cases
Ataxia - deafness - optic atrophy, lethal		12 cases
Ataxia, autosomal recessive, Beauce type		57 cases
Ataxia, cerebellar, autosomal recessive - blindness - deafness		3 families

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Ataxia, cerebellar, autosomal recessive - saccadic intrusion		1 family
Ataxia, episodic, type 3		1 family
Ataxia, episodic, type 4		2 families
Ataxia, spinocerebellar, X-linked, type 3		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
Atkin-Flaitz syndrome		14 cases
Atransferrinemia		9 cases
Atresia of small intestine	20	
Atrial septal defect - atrioventricular conduction defects		11 cases
Atrial tachyarrhythmia with short PR interval		12 cases
Atrioventricular canal, complete	15**	
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
Auriculoosteodysplasia		2 families
Aurocephalosyndactyly		5 cases
Autism	45	
Autism - facial port-wine stain		4 cases
Autoimmune lymphoproliferative syndrome		100 cases
Autosomal dominant cerebellar ataxia	2.15	
Autosomal dominant Charcot-Marie-Tooth disease, type 2F		2 families
Autosomal dominant Charcot-Marie-Tooth disease, type 2G		1 family
Autosomal dominant Charcot-Marie-Tooth disease, type 2K		1 family
Autosomal dominant Charcot-Marie-Tooth disease, type 2L		1 family
Autosomal dominant hyper IgE syndrome		250 cases
Autosomal dominant hypohidrotic ectodermal dysplasia		40 cases
Autosomal dominant limb-girdle muscular dystrophy, type 1A		1 family
Autosomal dominant limb-girdle muscular dystrophy, type 1D		5 families

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or 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 multiple pterygium syndrome		4 cases
Autosomal dominant severe congenital neutropenia	0.4**	
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 6		10 families
Autosomal dominant spastic paraplegia, type 8		< 10 families
Autosomal dominant spastic paraplegia, type 9		1 family
Autosomal recessive cerebellar ataxia	7	
Autosomal recessive limb-girdle muscular dystrophy, type 2G		14 cases
Autosomal recessive limb-girdle muscular dystrophy, type 2I		> 40 families
Autosomal recessive spastic paraplegia, type 14		1 family
Autosomal recessive spastic paraplegia, type 15		< 10 families
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
Autosomal recessive spastic paraplegia, type 39		2 families
Axenfeld-Rieger anomaly - hydrocephaly - skeletal abnormalities		3 cases
Babesiosis		40 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Bacterial toxic-shock syndrome	3	
Bakrania-Ragge syndrome		2 families
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	
Bartsocas-Papas syndrome		24 cases
Bartter syndrome	0.12	
Bazex-Dupre-Christol syndrome		143 cases
Beckwith-Wiedemann syndrome	7.3	
Beemer-Ertbruggen syndrome		2 cases
Behcet disease	2.5	
Bencze syndrome		2 families
Benign exophthalmos syndrome		4 cases
Benign familial neonatal-infantile seizures		10 families
Benign paroxysmal torticollis of infancy		50 cases
Bernard-Soulier syndrome		100 cases
Best disease	4.4	
Beta-mannosidosis		14 cases
Beta-sarcoglycanopathy	0.57	
Beta-thalassemia	0.5	
Beta-ureidopropionase deficiency		5 cases
Bethlem myopathy		100 cases
Bickel-Fanconi glycogenosis		112 cases
Bilateral microtia - deafness - cleft palate		4 cases
Bile acid synthesis defect, congenital, type 4		4 cases
Biliary atresia	5.6	
Biliary tract malformation - renal failure		6 cases
Birt-Hogg-Dube syndrome	0.5	
Björnstad syndrome		33 cases
Blackfan-Diamond disease	0.32	
Blaichman syndrome		1 case
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
Bloom syndrome		> 100 cases
Blue rubber bleb nevus		> 200 cases
Bone dysplasia, Azouz type		1 case
Bone dysplasia, lethal, Holmgren type		4 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Bone fragility - craniosynostosis - proptosis - hydrocephalus		4 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
Brachydactyly - arterial hypertension		> 10 families
Brachydactyly - long thumb		4 cases
Brachydactyly - nystagmus - cerebellar ataxia		1 family
Brachydactyly - preaxial hallux varus		8 cases
Brachydactyly A6		7 cases
Brachydactyly, type A5		2 families
Brachydactyly, type A7		1 family
Brachymorphism - onychodysplasia - dysphalangism		9 cases
Brachytelephalangy - dysmorphism - Kallmann syndrome		2 cases
Braddock syndrome		2 cases
Bradyopsia		5 cases
Brain dysgenesis due to glutamine synthetase deficiency, congenital		2 cases
Brain malformation - congenital heart disease - postaxial polydactyly		2 cases
Branchiogenic deafness syndrome		5 cases
Branchio-oculo-facial syndrome		< 50 cases
Branchio-skeleto-genital syndrome		3 cases
Breast cancer, familial	25	
Bronchobiliary fistula, congenital		23 cases
Bronchopneumopathy, chronic, due to TAP deficiency		< 20 cases
Bronchopulmonary dysplasia	13	
Brown-Vialetto-van Laere syndrome		< 100 cases
Bruck syndrome		< 40 cases
Budd-Chiari syndrome	1.5	
Buerger's disease	12.5	
Bullous dystrophy, macular type		2 families
Bullous pemphigoid	2.5	
Buschke-Ollendorff syndrome	5	
Buttiens-Fryns syndrome		3 cases
CACH syndrome		148 cases
CADASIL		500 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Calvarial doughnut lesions - bone fragility		20 cases
CAMFAK syndrome		3 families
CAMOS syndrome		5 cases
Campomelia, Cumming type		8 cases
Campomelic dysplasia	0.35	
Campptobrachydactyly		1 family
Campptodactyly - fibrous tissue hyperplasia - skeletal dysplasia		3 cases
Campptodactyly - tall stature - scoliosis - hearing loss		30 cases
Campptodactyly - taurinuria		4 families
Campptodactyly syndrome, Guadalajara type 1		8 cases
Campptodactyly syndrome, Guadalajara type 2		2 cases
Camurati-Engelmann disease		200 cases
Cantrell pentalogy	0.55**	
Cantu syndrome		4 cases
Capillary leak syndrome		57 cases
Carbamoylphosphate synthetase deficiency	0.7	
Carcinoma of the gallbladder	6.5	
Cardiac anomalies - heterotaxy		9 cases
Cardiac anomalies - heterotaxy		9 cases
Cardiac conduction disease, dilated cardiomyopathy and brachydactyly		10 cases
Cardiocranial syndrome, Pfeiffer type		< 10 cases
Cardiogenital syndrome		7 cases
Cardiomyopathy - cataract - hip spine disease		9 cases
Cardiomyopathy - renal anomalies		2 cases
Cardiomyopathy, familial dilated	17.5	
Cardiomyopathy-exercise intolerance due to muscle and heart glycogen deficiency		3 cases
Carey-Fineman-Ziter syndrome		< 20 cases
Carney complex		160 cases
Carney triad		100 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 disease		< 250 cases
Carpenter syndrome		40 cases
Carpotarsal osteochondromatosis		< 10 cases
Carpotarsal osteolysis, recessive		< 10 cases
Castleman disease		400 cases
Cataract - deafness - hypogonadism		3 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Cataract - hypertrichosis - intellectual deficit		1 case
Cataract - intellectual deficit - hypogonadism		10 cases
Cataract - microphthalmia - septal defect		2 cases
Cataract - nephropathy - encephalopathy		2 cases
Cataract- ataxia - deafness		2 cases
Cataract cardiomyopathy		30 cases
Cataract-glaucoma		3 families
Cataract-microcornea syndrome		8 families
Catel-Manzke syndrome		27 cases
Cat-eye syndrome	1.35	
Cat-scratch disease	6.6	
Cavitating leukoencephalopathy, progressive		19 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 IIh		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		2 families
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

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
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
Charcot-Marie-Tooth disease, X-linked	1.6	
CHARGE syndrome	0.14	
Chediak-Higashi syndrome		> 10 cases
CHILD syndrome		30 cases
Choanal atresia	8.2	
Choanal atresia - deafness - cardiac defects - dysmorphism		5 cases
Cholangiocarcinoma	10	
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 - situs inversus - imperforate anus - polydactyly		1 case
Chondrodysplasia punctata, rhizomelic type	1	
Chondrodysplasia, Blomstrand type		13 cases
Chondrodysplasia, lethal recessive		4 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
Choroido cerebral 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 myeloid leukemia	6	
Chronic recurrent multifocal osteomyelitis, juvenile		> 260 cases
Churg-Strauss syndrome	1	
Chylomicron retention disease		40 cases
CINCA syndrome		100 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Craniosynostosis - dysmorphism - brachydactyly		5 cases
Craniosynostosis - fibular aplasia		2 cases
Craniosynostosis - intracranial calcifications		3 cases
Craniosynostosis dandy walker hydrocephalus		4 cases
Craniosynostosis, Boston type		19 cases
Craniosynostosis, Philadelphia type		1 family
Craniosynostosis-radial aplasia, Imaizumi type		2 cases
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	
Curry-Jones syndrome		5 cases
Cutaneous albinism, ermine phenotype		3 cases
Cutaneous lymphoma	8.3	
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 - intellectual deficit	1.02	
Cyprus facial-neuromusculoskeletal 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
Czech dysplasia, metatarsal type		< 20 cases
Dacryocystitis - osteopoikilosis		5 cases
Dahlberg-Borer-Newcomer syndrome		2 cases
Dandy-Walker malformation - macrocephaly		2 cases
Dandy-Walker malformation - polydactyly, postaxial		2 cases
Darier disease	1.5	
Deaf blind hypopigmentation syndrome, Yemenite type		2 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Deafness - enamel hypoplasia - nail defects		6 cases
Deafness - genital anomalies - metacarpal and metatarsal synostosis		2 cases
Deafness - lymphedema - 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 - vitiligo - achalasia		2 cases
Deafness-infertility syndrome		3 families
Deafness-intellectual deficit, Martin-Probst type		3 cases
Deafness-intellectual deficit, Martin-Probst type		3 cases
Deafness-tubular acidosis-anemia		2 cases
Dehydratase deficiency		21 cases
Deletion 2q24		23 cases
Deletion 2q37		10 cases
Delta-sarcoglycanopathy	0.57	
DEND syndrome		14 cases
Dentatorubral pallidolusian atrophy	< 0.3	
Dentinogenesis imperfecta - short stature - hearing loss - intellectual deficit		2 cases
Denys-Drash syndrome		150 cases
Depigmentation of the iris, acute, bilateral		5 cases
Dermatitis herpetiformis	20.2	
Dermato-cardio-skeletal syndrome, Borroni type		2 cases
Dermatofibrosarcoma protuberans	10	
Dermatoleukodystrophy		2 cases
Dermatomyositis	14.8	
Dermatoosteolysis, Kirghizian type		5 cases
Dermo-odonto dysplasia		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 mellitus, permanent neonatal - pancreatic and cerebellar agenesis		4 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
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 - Alport syndrome X-linked	0.1	
Diffuse neonatal hemangiomatosis		< 70 cases
Diffuse palmoplantar keratoderma - acrocyanosis		10 cases
Digitorenocerebral syndrome		< 10 cases
Digitotalar dysmorphism	6	
Dihydropteridine reductase deficiency		134 cases
Dihydropyrimidinuria		7 cases
Dincsoy-Salih-Patel syndrome		2 cases
Disorder of sex development - intellectual deficit		3 cases
Distal asymmetrical inflammatory myopathy of the upper limbs		10 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 trisomy 10q		40 cases
Distal trisomy 6p		40 cases
DOOR syndrome		< 50 cases
Dopamine beta-hydroxylase deficiency		12 cases
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
Duchenne and Becker muscular dystrophy	5	
Duker-Weiss-Siber syndrome		4 cases
Duodenal atresia	8.55	
Dyggve-Melchior-Clausen disease		60 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Dyschondrosteosis - nephritis		1 family
Dyserythropoietic anemia, congenital	1	
Dyskeratosis congenita	0.1	
Dysmorphism - short stature - deafness - disorder of sex development		2 cases
Dysplasia, mandibuloacral		37 cases
Dystonia, dopa-responsive	0.3	
Dystonia, mixed		3 families
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 - absent dermatoglyphs		< 30 cases
Ectodermal dysplasia - anhidrotic, with immunodeficiency - osteopetrosis - lymphedema		2 cases
Ectodermal dysplasia - arthrogyriposis - diabetes mellitus		1 case
Ectodermal dysplasia - blindness		2 cases
Ectodermal dysplasia - skin fragility syndrome		10 cases
Ectodermal dysplasia, Berlin type		4 cases
Ectodermal dysplasia, hidrotic, Christianson-Fourie type		6 cases
Ectodermal dysplasia, hidrotic, Halal type		4 cases
Ectodermal dysplasia, hypohidrotic - hypothyroidism - ciliary dyskinesia		3 cases
Ectodermal dysplasia, "pure" hair-nail type		< 20 cases
Ectodermic dysplasia hypothyroidism cleft		3 cases
Ectopia lentis - chorioretinal dystrophy - myopia		4 cases
Ectopia lentis isolated	6.4	
Ectrodactyly - ectodermal dysplasia without clefting		5 cases
EEM syndrome		7 families
Ehlers-Danlos syndrome, spondylocheiro dysplastic type		6 cases
Ehlers-Danlos syndrome, type 10		1 family
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

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Elliptocytosis, hereditary	35	
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 sulphite oxidase deficiency		50 cases
Encephalopathy with neuroserpin inclusion bodies, familial form		> 5 families
Encephalopathy, ethylmalonic		< 40 cases
Enchondromatosis		600 cases
Endosteal sclerosis - cerebellar hypoplasia		4 cases
Eng-Strom syndrome		2 cases
Enterocyte heparan sulphate deficiency, congenital		3 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	
Epithelio-exfoliative colitis - deafness		2 cases
Erdheim-Chester disease		178 cases
Erythroderma, congenital ichthyosiform, bullous	0.4	
Erythroderma, lethal, congenital		17 cases
Erythrokeratoderma ataxia		25 cases
Erythrokeratoderma variabilis, Mendes da Costa type		> 200 cases
Esophageal atresia	25	
Esophageal carcinoma	8	
Esthesioneuroblastoma		< 1000 cases
Evans syndrome	0.1	
Ewing sarcoma	0.1	
Excessive growth - learning disabilities - facial dysmorphism		6 families

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Exostoses, multiple	2	
Eyebrow duplication - syndactyly		3 cases
Fabry disease	1.75	
Facial dysmorphism - macrocephaly - myopia - Dandy-Walker malformation		3 cases
Facial onset sensory and motor neuropathy		4 cases
Faciocardiomeic dysplasia, lethal		3 cases
Facioscapulohumeral muscular dystrophy	7	
Fahr syndrome		< 20 families
Familial adenomatous polyposis	5.25	
Familial caudal dysgenesis		4 cases
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		< 20 families
Familial platelet syndrome with predisposition to acute myelogenous leukemia		13 families
Familial spastic paraplegia	5	
Fanconi anemia	1	
Fanconi ichthyosis dysmorphism		6 cases
Fanconi syndrome		100 cases
Femur bifid - monodactylous - ectrodactyly		200 cases
Femur-fibula-ulna complex	1.5	
Fetal cytomegalovirus syndrome	40	
Fetal methyl mercury syndrome		800 cases
Fetal varicella syndrome		> 100 cases
Fibrochondrogenesis		11 cases
Fibrodysplasia ossificans progressiva	0.08	
Fibrous dysplasia of bone	< 0.05	
Fibular aplasia - ectrodactyly		< 50 cases
Fibular dimelia - diplopodia		11 cases
Filippi syndrome		< 25 cases
Fine-Lubinsky syndrome		5 cases
Fingerprints, absence of - milia, congenital		14 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
Folate malabsorption, hereditary		17 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Follicular lymphoma	36	
Foveal hypoplasia - presenile cataract		11 cases
Fragile X syndrome	14.25	
Frank-Ter Haar syndrome		5 cases
Fraser syndrome		150 cases
Freeman-Sheldon syndrome		100 cases
Fried syndrome		1 family
Friedreich ataxia	2.5	
Frontometaphyseal 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-Abbott syndrome		3 cases
Gastric cancer	20	
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 dwarfism		27 cases
Genitopatellar syndrome		7 cases
German syndrome		5 cases
Geroderma osteodysplastica		30 cases
Giant axonal neuropathy		20 families
Giant cell arteritis	8.9	
Giant pigmented hairy nevus	2	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Gingival fibromatosis - progressive deafness		2 families
Gitelman syndrome	2.5	
Glaucoma - ectopia - microspherophakia - stiff joints - short stature		3 cases
Glaucoma - sleep apnoea		5 cases
Glioblastoma	11	
Global developmental delay - osteopenia - ectodermal defect		3 cases
Glomerulonephritis - sparse hair - telangiectasis		< 10 cases
Glossopalatine ankylosis		30 cases
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		84 cases
Glycogen storage disease type 2	1.1	
Glycogen storage disease type 4	0.6	
Glycogen storage disease type 7		< 30 cases
Glycogen storage, 0 type		16 cases
Goldberg-Shprintzen megacolon syndrome		10 cases
Goldenhar syndrome	3.5	
Goldmann-Favre syndrome		< 50 cases
Gombo syndrome		4 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	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
Greenberg dysplasia		< 10 cases
Greig syndrome		100 cases
Griscelli disease		60 cases
Growth deficiency - brachydactyly - dysmorphism		2 families

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate		4 cases
Growth delay due to insulin-like growth factor I deficiency		4 cases
Growth retardation - microcephaly - digital abnormalities - hypospadias		4 cases
GTP cyclohydrolase I deficiency		17 cases
Guanidinoacetate methyltransferase deficiency		9 cases
Guillain-Barré syndrome	3.45	
Hair defect - photosensitivity - intellectual deficit		3 cases
Hallermand-Streiff-Francois 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
Helicoid peripapillary chorioretinal degeneration		100 cases
Hemicrania, paroxysmal	2	
Hemimelia	4.15	
Hemiplegic migraine, familial or sporadic	10	
Hemochromatosis, neonatal		100 cases
Hemolytic anemia due to adenylate kinase deficiency		12 cases
Hemolytic anemia due to glutathione reductase deficiency		3 cases
Hemolytic anemia due to red cell pyruvate kinase deficiency	5.1	
Hemolytic anemia, lethal - genital anomalies		2 cases
Hemolytic anemia, nonspherocytic, due to hexokinase deficiency		17 families
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
Henoch-Schoenlein purpura	8.5	
Hepatic veno-occlusive disease	11	
Hepatic veno-occlusive disease - immunodeficiency		< 25 cases
Hepatitis, chronic autoimmune	0.75	
Hereditary chronic pancreatitis	0.125	
Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia		19 cases
Hereditary neuropathy with liability to pressure palsies	9	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Hereditary sensory and autonomic neuropathy with deafness and global delay		4 cases
Hereditary sensory and autonomic neuropathy, type 2		35 cases
Hereditary vascular retinopathy		1 family
Hereditary vascular retinopathy		1 family
Hermansky-Pudlak syndrome	0.15	
HERNS syndrome		3 families
Hersh-Podbruch-Weisskopf syndrome		2 cases
Heterotaxia	2.5	
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	
Histiocytosis, progressive mucinous, hereditary		13 cases
Histiocytosis, sea-blue		60 cases
Histiocytosis, sinus, with massive lymphadenopathy		500 cases
Hodgkin lymphoma, classical	10.5	
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	6.2	
Hyaline fibromatosis, juvenile		50 cases
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**	
Hypercoagulability syndrome, due to glycosylphosphatidylinositol deficiency		2 cases
Hyperferritinemia, hereditary, with congenital cataracts		> 64 cases
Hyperglycemia, isolated nonketotic	0.2	
Hypergonadotropic ovarian dysgenesis	12	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Hyperimmunoglobulinemia D with recurrent fever		180 cases
Hyperkalemic periodic paralysis	0.75	
Hyperkeratosis - hyperpigmentation syndrome		10 cases
Hyperlipidemia type 3	7.8	
Hyperlipoproteinemia type 1	0.6	
Hyperostotic dwarfism, Lenz-Majewski type		7 cases
Hyperoxaluria	0.2	
Hyperplastic polyposis syndrome	50	
Hypertrichosis cubiti - short stature		28 cases
Hypertrichosis lanuginosa congenita		< 100 cases
Hypertrichosis lanuginosa, acquired		60 cases
Hypertrichosis, anterior cervical, isolated		< 20 cases
Hypertrichotic osteochondrodysplasia		18 cases
Hypochondroplasia	3.3	
Hypocomplementemic leucocytoclastic vasculitis		< 100 cases
Hypogammaglobulinemia due to CD19 deficiency		4 cases
Hypoglossia - hypodactyly		< 50 cases
Hypogonadism - retinitis pigmentosa		3 cases
Hypokalemic periodic paralysis	1	
Hypomagnesemia with hypocalciuria		3 families
Hypomagnesemia with normocalciuria		2 cases
Hypomandibular faciocranial dysostosis		4 cases
Hypomyelination - congenital cataract		10 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodontia		4 cases
Hypomyelination - hypogonadotropic hypogonadism - hypodontia		4 cases
Hypomyelination with atrophy of basal ganglia and cerebellum		19 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
Hypospadias-hypertelorism-coloboma and deafness		2 cases
Hypotelorism - cleft palate - hypospadias		13 cases
Hypothyroidism - dysmorphism - postaxial polydactyly - intellectual deficit		5 cases
Hypothyroidism, congenital	29	
Hypotonia with lactic acidemia and hyperammonemia		3 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
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 - 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 follicularis - atrichia - photophobia		10 cases
Ichthyosis prematurity syndrome		16 families
Ichthyosis, congenital - microcephalus - quadriplegia		2 cases
Ichthyosis, lamellar	> 0.33	
Ichthyosis, X-linked	16.6	
Idiopathic and/or familial pulmonary arterial hypertension	1.5	
Idiopathic hypereosinophilic syndrome	10	
Idiopathic hypersomnia	4	
IMAGe syndrome		< 20 cases
Iminoglycinuria	6.68	
Immune dysregulation - polyendocrinopathy - enteropathy, X-linked		7 families
Immunodeficiency by defective expression of HLA class 2		100 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	
Infant epilepsy with migrant focal crisis		29 cases
Infantile neuroaxonal dystrophy		> 150 cases
Infantile onset spinocerebellar ataxia		24 cases
Inflammatory pseudotumor of the liver		143 cases
Insomnia, familial fatal		27 cases
Insulin resistance, short fifth metacarpals		1 family
Intellectual deficit - dysmorphism - hypogonadism - diabetes mellitus		4 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Intellectual deficit - hypoplastic corpus callosum - preauricular tag		3 cases
Intellectual deficit - progressive spasticity, X-linked		1 family
Intellectual deficit - sparse hair - brachydactyly		6 cases
Intellectual deficit, severe - epilepsy - anal anomalies - distal phalangeal hypoplasia		2 cases
Intellectual deficit, X-linked - acromegaly - hyperactivity		2 cases
Intellectual deficit, X-linked - choreoathetosis - abnormal behavior		5 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 paraplegia with iron deposits		1 family
Intellectual deficit, X-linked, Abidi type		8 cases
Intellectual deficit, X-linked, Armfield type		6 cases
Intellectual deficit, X-linked, Cabezas type		1 family
Intellectual deficit, X-linked, Cantagrel type		2 cases
Intellectual deficit, X-linked, Lubs type		5 cases
Intellectual deficit, X-linked, Miles-Carpenter type		4 cases
Intellectual deficit, X-linked, Pai type		1 family
Intellectual deficit, X-linked, recessive - macrocephaly - ciliary dysfunction		1 family
Intellectual deficit, X-linked, Reish type		2 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Intellectual deficit, X-linked, Schimke type		4 cases
Intellectual deficit, X-linked, Seemanova type		4 cases
Intellectual deficit, X-linked, severe, Gustavson type		7 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, Stoll type		4 cases
Intellectual deficit, X-linked, syndromic 7		10 cases
Intellectual deficit, X-linked, syndromic, due to JARID1C mutation		< 10 families
Intellectual deficit, X-linked, Vitale type		8 cases
Intellectual deficit, X-linked, Wilson type		3 cases
Intellectual deficit, X-linked, with isolated growth hormone deficiency		3 families
Intellectual deficit, X-linked, with seizures, short stature and midface hypoplasia		17 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
Intrauterine infection-like syndrome, congenital		> 30 cases
Iris coloboma with ptosis - intellectual deficit		10 cases
Iris dysplasia - hypertelorism - deafness		2 cases
Isolated anorectal malformation	24	
Isolated brachycephaly	5	
Isolated cloverleaf skull syndrome		150 cases
Isolated humeroradial synostosis		150 cases
Isolated Klippel-Feil syndrome	2	
Isolated plagiocephaly	10	
Isolated scaphocephaly	20	
Isolated trigonocephaly	6.7	
Isotretinoin-like syndrome		6 cases
Isovaleric acidemia	1	
IVIC syndrome		4 families
Jackson-Weiss syndrome		2 families

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Jacobsen syndrome		150 cases
Jalili syndrome		29 cases
Jeune syndrome	0.2	
Johanson-Blizzard syndrome		23 cases
Joubert syndrome	1	
Juberg-Hayward syndrome		10 cases
Juvenile polyposis of infancy		11 cases
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		2 cases
Kasabach-Merritt syndrome		> 175 cases
KBG syndrome		45 cases
Kearns-Sayre syndrome	2	
Kennedy disease	2.8	
Keratoconjunctivitis, vernal	21	
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 cases
Keratosis palmoplantaris - esophageal carcinoma		< 10 families
Keratosis palmoplantaris - periodontopathia - onychogryposis		< 100 cases
Keratosis, Nagashima-type		20 cases
Ketoacidosis due to betaketothiolase deficiency		60 cases
KID syndrome		> 100 cases
Kimura disease		200 cases
Kniest-like dysplasia, lethal form		2 cases
Kozłowski-Brown-Hardwick syndrome		2 cases
Krabbe disease	0.75**	
Kudo-Tamura-Fuse syndrome		2 cases
Kumar-Levick syndrome		1 family
Lacrimo-auriculo-dento-digital syndrome		20 cases
Lafora disease	< 0.1	
Lambert-Eaton myasthenic syndrome	1	
Laminopathy, type Decadain-Vigouroux		9 cases
Langerhans cell histiocytosis	2	
Larsen syndrome		100 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Larsen-like syndrome, lethal form		< 10 cases
Laryngeal abductor paralysis - intellectual deficit		< 20 cases
Laryngo-tracheo-oesophageal cleft	1.5	
Lateral body wall complex	2**	
Lathosterolosis		< 5 cases
LCAT deficiency		30 families
Leber amaurosis, congenital	10	
Leber hereditary optic neuropathy	6.5	
Legg-Calve-Perthes disease	23	
Leigh syndrome	2.75**	
Leleis syndrome		8 cases
Lemierre syndrome	0.1	
Lennox-Gastaut syndrome	15	
LEOPARD syndrome		200 cases
Leprechaunism	0.1**	
Leptospirosis	0.24	
Lesch-Nyhan syndrome	0.38	
Leucinosi	15.6	
Leukemia, B-cell lymphocytic, chronic	32	
Leukemia, lymphoblastic, acute	6.5	
Leukemia, myeloid, acute	11.8	
Leukemia, promyelocytic, acute	8	
Leukocyte adhesion deficiency		< 350 cases
Leukocyte adhesion deficiency, type II		< 10 cases
Leukocyte adhesion deficiency, type III		17 cases
Leukodystrophy with oligodontia		4 cases
Leukoencephalopathy - ataxia - hypodontia - hypomyelination		8 cases
Leukoencephalopathy - metaphyseal chondrodysplasia		4 cases
Leukoencephalopathy - palmoplantar keratoderma		4 cases
Leukoencephalopathy with bilateral anterior temporal lobe cysts		29 cases
Leukoencephalopathy with brain stem, spinal cord involvement - lactate elevation		39 cases
Lewis-Pashayan syndrome		3 cases
Lewis-Sumner syndrome	0.9	
Lhermitte-Duclos disease		220 cases
Lichenstein syndrome		2 cases
Li-Fraumeni syndrome		400 families
Ligneous conjunctivitis	1.1	
Limb-girdle muscular dystrophy	0.8	
Limb-mammary syndrome		27 cases
Linear atrophoderma of Moulin		< 30 cases
Lipoamide dehydrogenase deficiency		20 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Lipodystrophy - intellectual deficit - deafness		3 cases
Lipodystrophy, acquired generalized		80 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		300 cases
Lipodystrophy, familial partial, Köbberling type		< 20 cases
Lipodystrophy, partial acquired		250 cases
Lipoid proteinosis		> 280 cases
Lissencephaly type 2	0.12	
Lissencephaly, type III - familial fetal akinesia sequence		5 cases
Lissencephaly, type III - metacarpal bone dysplasia		2 cases
Lobar emphysema, congenital	4.5	
Long QT syndrome, familial	25	
Lopez-Hernandez syndrome		11 cases
Low birth weight - dwarfism - dysgammaglobulinemia		2 cases
Lowe syndrome	0.19	
Lowry-Wood syndrome		< 10 cases
Lumbosacral vertebrae, posterior fusion of - blepharoptosis		3 cases
Lung cancer, small cell	10	
Lung fibrosis - immunodeficiency - gonadal dysgenesis		2 cases
Lupus erythematosus, bullous systemic		70 cases
Lupus erythematosus, cutaneous	50	
Lymphangioliomyomatosis	0.1	
Lymphedema - atrial septal defects - facial changes		3 cases
Lymphedema - cerebral arteriovenous anomaly		5 cases
Lymphedema, congenital	8.8	
Macrocephaly - Cutis Marmorata Telangiectatica Congenita		40 cases
Macrocephaly - immune deficiency - anemia		2 cases
Macrocephaly - short stature - paraplegia		2 cases
Macrogyria pseudobulbar palsy		4 cases
Macrophagic myofasciitis	1	
Macrostomia - preauricular tags - external ophthalmoplegia		9 cases
Macrothrombocytopenia with abnormal proplatelet formation, autosomal dominant		5 cases
Macular degeneration, juvenile - hypotrichosis		7 families

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Madras motor neuron disease		154 cases
Malabsorptive diarrhea due to paucity of enteroendocrine cells, congenital		3 cases
Malakoplasiya		500 cases
Malaria	3	
Malignant hyperthermia	33	
Malignant hyperthermia - arthrogryposis - torticollis		4 cases
Malonic aciduria		17 cases
Mandibulofacial dysostosis, X-linked		7 cases
Mantle cell lymphoma	3.9	
Marden-Walker syndrome		30 cases
Marfan syndrome	30	
Marie Unna congenital hypotrichosis		12 families
Marinesco-Sjogren 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, cutaneous	0.75	
Mastocytosis, systemic	3.3	
Maternal hyperphenylalaninemia	1.25	
Matthew-Wood syndrome		5 cases
Mayer-Rokitansky-Küster-Hauser syndrome	9	
Mazabraud syndrome		54 cases
McLeod neuroacanthocytosis syndrome		150 cases
Meacham syndrome		< 15 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, with or without hyperuricemia	0.11	
Medullary cystic kidney disease, autosomal recessive	1.05	
Megacalycosis, congenital		> 50 cases
Megacystis microcolon - intestinal hypoperistalsis - hydronephrosis		89 cases
Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus		6 cases
MEHMO syndrome		7 cases
Melanoma, familial	46.8	
MELAS syndrome	16	
Melorheostosis		300 cases
Mendelian susceptibility to atypical mycobacteria	0.059	
Meniere disease	42.5	
Menkes syndrome	0.7	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
MERRF syndrome	0.9	
Mesoaxial synostotic syndactyly with phalangeal reduction		2 families
Mesomelic dysplasia - skin dimples		2 cases
Metachondromatosis		25 cases
Metachromatic leukodystrophy	0.16	
Metaphyseal acroscyphodysplasia		4 cases
Metaphyseal anadysplasia		< 20 cases
Metaphyseal chondrodysplasia, Jansen type		16 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
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
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 - micropenis - convulsions		4 cases
Microcephaly - seizures - intellectual deficit - heart disease		2 cases
Microcytic anemia with liver iron overload		3 cases
Microdontia - type I microtia - deafness		9 cases
Microgastria - limb reduction defect		16 cases
Microlissencephaly - micromelia		2 cases
Microphthalmia - brain atrophy		3 cases
Microscopic polyangiitis	7.5	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Microtia	15	
Microtia - eye coloboma - imperforation of the nasolacrimal duct		1 family
Midas syndrome		< 50 cases
Midface retraction syndrome, Schinzel-Giedion type		34 cases
Miller-Dieker syndrome	0.3	
Mirhosseini-Holmes-Walton syndrome		< 15 cases
Mitochondrial disease of nuclear origin	9	
Mitochondrial encephalomyopathy aminoacidopathy		2 cases
Mitochondrial myopathy with sideroblastic anemia		7 cases
Mitral regurgitation - deafness - skeletal anomalies		3 cases
Moebius syndrome		300 cases
Mohr-Tranebjaerg syndrome		46 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	
Mucopolipidosis type 2	0.15**	
Mucopolipidosis 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
Mucosulfatidosis		50 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 fibrofolliculoma, familial		7 cases
Multiple pterygium syndrome, lethal form		200 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Multiple sclerosis - ichthyosis - factor VIII deficiency		2 cases
Multiple synostoses		20 families
Multiple system atrophy	4.6	
MURCS association	11.25	
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus		10 cases
Muscular dystrophy, Fukuyama type	0.54	
Muscular dystrophy, limb-girdle, autosomal recessive, type 2A	3.8	
Myasthenia gravis	20	
Myelodysplastic syndromes	25	
Myelofibrosis with myeloid metaplasia	10	
Myeloma, multiple	14.25	
Myhre syndrome		16 cases
Myoclonic epilepsy of infancy		106 cases
Myoclonus - cerebellar ataxia - deafness		4 cases
Myoclonus hereditary - progressive distal muscular atrophy		< 10 cases
Myoneurogastrointestinal encephalopathy syndrome		87 cases
Myopathy due to calsequestrin and SERCA1 protein overload		4 cases
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
Narcolepsy-cataplexy	49	
NARP/MILS syndrome	8.5	
Nasopalpebral lipoma - coloboma - telecanthus		< 30 cases
Necrotizing encephalopathy, acute, autosomal dominant		11 cases
Nemaline myopathy	1	
Neonatal ichthyosis - sclerosing cholangitis		< 10 cases
Nephroblastoma	10.1	
Nephronoptosis familial, adult form - spastic quadriplegia		2 cases
Nephropathy - deafness - hyperparathyroidism		5 cases
Nephrosis - deafness - urinary tract - digital malformations		5 cases
Nephrotic syndrome, idiopathic, steroid-sensitive	18	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Netherton disease	1.35	
Neu-Laxova syndrome		60 cases
Neuroaxonal dystrophy - renal tubular acidosis		3 cases
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 syndrome, Johnson type		< 30 cases
Neuroectodermal-endocrine syndrome		4 cases
Neurofibromatosis type 1	25	
Neurofibromatosis type 2	0.5	
Neurometabolic disorder due to serine deficiency		< 30 cases
Neuromyelitis optica	1.5	
Neuropathy with hearing impairment		1 family
Neutropenia cyclic	0.1	
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	1**	
Nodular regenerative hyperplasia of the liver	3	
Non-distal trisomy 10p		60 cases
Non-distal trisomy 12p	2**	
Non-distal trisomy 8q		> 30 cases
Non-Hodgkin 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 coloboma	1	

** Prevalence at birth

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
Oculocutaneous albinism	7.15	
Oculodental syndrome, Rutherford type		1 family
Oculodentodigital dysplasia		243 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	
Oculotrichodysplasia		2 cases
Odontomicronychia dysplasia		5 cases
Odonto-onycho-dermal dysplasia		< 15 cases
Odontotrichomelic syndrome		4 cases
Odonto-tricho-ungual-digito-palmarn syndrome		21 cases
Okamoto syndrome		2 cases
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
Ophthalmic acromelic syndrome		30 cases
Opitz BBB/G syndrome	3	
Opsismodysplasia		25 cases
Optic atrophy	6	
Optic atrophy and cataract, autosomal dominant		14 cases
Oral-facial-digital syndrome, type 1	1.2	
Oral-facial-digital syndrome, type 10		1 case
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 6		29 cases
Oral-facial-digital syndrome, type 8		2 families
Orbital leiomyoma		16 cases
Ornithine carbamoyltransferase deficiency	1	
Orotic aciduria hereditary		< 20 cases
Ossification anomalies - psychomotor development delay		2 cases
Osteochondritis dissecans	35	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Osteochondrodysplastic 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
Osteogenesis imperfecta, congenital - microcephaly - cataracts		3 cases
Osteopathia striata - cranial sclerosis		100 cases
Osteopetrosis, autosomal dominant, type 1		33 cases
Osteopetrosis, intermediate form		50 cases
Osteopetrosis, malignant, autosomal recessive	0.75**	
Osteopoiikilosis - short stature - intellectual deficit		4 cases
Osteoporosis - pseudoglioma	0.05	
Osteoporosis-oculocutaneous-hypopigmentation syndrome		3 cases
Osteosarcoma	5	
Osteosclerosis - ichthyosis - premature ovarian failure		3 cases
Osteosclerotic bone dysplasia, lethal		8 families
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 of		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 hypokeratosis, circumscribed		17 cases
Palmoplantar keratoderma - amyotrophy		4 cases
Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma		5 cases
Palmoplantar keratoderma, diffuse, Norrbotten dominant type	2.5	
Palmoplantar porokeratosis of Mantoux		< 10 cases
Pancreas agenesis		8 cases
Pancreatic hypoplasia - diabetes - heart disease		< 10 cases
Pancreatitis, acute, recurrent	10	
Pancreatoblastoma		60 cases
Panniculitis, cytophagic histiocytic		< 100 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Papillon-Lefevre syndrome	0.25	
Papulosis, malignant atrophic		> 200 cases
Paraneoplastic pemphigus		> 60 cases
Paraplegia - brachydactyly - cone-shaped epiphysis		5 cases
Paraplegia - intellectual deficit - hyperkeratosis		4 cases
PARC syndrome		2 cases
Parietal foramina	5	
Parkinson disease, genetic type	15	
Parkinsonism, young adult onset	37.5	
Parkinson's disease dementia, familial	41	
Paroxysmal extreme pain disorder		4 families
Paroxysmal nocturnal hemoglobinuria	0.55	
Parsonage-Turner syndrome	3.3	
Partington syndrome		2 families
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
Peritoneal leiomyomatosis, disseminated		100 cases
Perlman syndrome		30 cases
Perrault syndrome		34 cases
Persistent hyperinsulinemic hypoglycemia of infancy	2	
Peters-plus syndrome		50 cases
Peutz-Jeghers syndrome	2.2	
Pfeiffer syndrome	1	
PHACE syndrome		100 cases
Phenylketonuria	4	
Pheochromocytoma and secreting paraganglioma	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 sequence - faciodigital anomaly		2 cases
Pierre Robin sequence, isolated	8.75	

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Pierson syndrome		22 cases
Pili torti - onychodysplasia		1 family
Pilodental dysplasia - refractive errors		2 cases
Pitt-Hopkins syndrome		150 cases
Pityriasis rubra pilaris		48 cases
Plummer-Vinson syndrome		25 cases
Poikiloderma of Kindler		100 cases
Poland anomaly	2	
Pollitt syndrome		10 cases
Polyarteritis nodosa	3.07	
Polyarthritis, juvenile, rheumatoid factor-negative	8	
Polyarthritis, juvenile, rheumatoid factor-positive	4.2	
Polycystic kidney disease, autosomal dominant, type 1, with tuberous sclerosis		30 cases
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, 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
Porphyria, acute intermittent	10	
Porphyria, chronic hepatic	1.5	
Porphyria, congenital erythropoietic		> 200 cases
Posterior column ataxia - retinitis pigmentosa		13 cases
Post-transplant lymphoproliferative disease	26.2	
Potocki-Shaffer syndrome		23 cases
Prader-Willi syndrome	10.7	
Primary biliary cirrhosis	13.5	
Primary ciliary dyskinesia	5	
Primary erythralgia		30 families
Primary lateral sclerosis	1.5	
Primary sclerosing cholangitis	11	
Progeria	0.25**	
Progeria - short stature - pigmented nevi		< 10 cases
Progressive bulbar paralysis of childhood		< 40 cases
Progressive neurodegeneration - joint laxity - cataract		2 cases

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Progressive vertebral fusion, non-infectious, syndromic form		10 cases
Prolidase deficiency		50 cases
Propionic acidemia	3.75	
Proteus syndrome		200 cases
Protoporphyrin, erythropoietic	0.9	
Proximal myotonic myopathy	1	
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
Pseudohypoadosteronism, type 1		70 cases
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
Pterygium popliteal syndrome, autosomal dominant	0.3	
Pterygium syndrome, antecubital		11 cases
Ptosis - strabismus - ectopic pupils		1 family
Ptosis - strabismus - rectus abdominis diastasis		2 cases
Pulmonary alveolar proteinosis	0.1	
Pulmonary fibrosis, idiopathic	27	
Pulmonary hemosiderosis, primary		250 cases
Pulmonary lymphangiectasia, congenital		> 100 cases
Pulmonary valve stenosis, congenital	7.2	
Pure autonomic failure	1	
Pyknoachondrogenesis		5 cases
Pyle disease		< 30 cases
Pyogenic arthritis - pyoderma gangrenosum - acne		34 cases
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

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Recessive hereditary methemoglobinemia type 2		< 100 cases
Recurrent infections - short stature - hypopigmentation - coarse face		4 cases
Reducing body myopathy		4 families
Refsum disease	0.1	
Refsum disease, infantile form	0.005	
Regional odontodysplasia		139 cases
Relapsing polychondritis	0.35	
Renal agenesis, bilateral	17	
Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts		10 cases
Rendu-Osler-Weber disease	3.5	
Renpenning syndrome		10 families
Resistance to thyrotropin-releasing hormone syndrome		2 cases
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 - intellectual deficit - deafness - hypogenitalism		2 families
Retinoblastoma	5.4	
Retinohepatoendocrinologic syndrome		7 cases
Retinoschisis, X-linked	4.5	
Rett syndrome	4.15	
Rhabdoid tumor		500 cases
Rheumatic fever	5	
Rhombencephalosynapsis		50 cases
Rickettsialpox		> 800 cases
Rieger syndrome	0.5	
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/SC phocomelia		100 cases
Robinow syndrome		> 120 cases
Robinow-like syndrome		2 cases
Rolled and spiral hairs - palmoplantar keratoderma		4 cases
Rothmund-Thomson syndrome		300 cases
Rubella panencephalitis		> 20 cases
Rubella syndrome, congenital	1**	
Rubinstein-Taybi syndrome	1	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
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
Scalp defects - postaxial polydactyly		2 cases
Scalp-ear-nipple syndrome		30 cases
SCARF syndrome		2 cases
Schimke immuno-osseous dysplasia		50 cases
Schinzel syndrome		< 10 families
Schizencephaly	1.54	
Schizophrenia - intellectual deficit - deafness - retinitis		1 family
Schnitzler syndrome		50 cases
Schopf-Schulz-Passarge syndrome		19 cases
Schwartz-Jampel syndrome		100 cases
Scleroderma	42	
Sebastian syndrome		< 10 families
Seckel syndrome		100 cases
Segmental odontomaxillary dysplasia		32 cases
Seizures - intellectual deficit due to hydroxylysineuria		3 cases
Senior-Loken syndrome	0.1	
Sensenbrenner syndrome		15 cases
Sensorineural hearing loss, early greying, and essential tremor		3 cases
SERKAL syndrome		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+, X-linked	1.5	
Sezary's syndrome	0.18	
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 resistance	0.2	
Short stature, Brussels type		2 cases
SHORT syndrome		30 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Shprintzen-Goldberg syndrome		< 50 cases
Shwachman-Diamond syndrome		200 cases
Sialidosis type 1	0.02**	
Sialidosis, type 2	0.02**	
Sickle cell anemia	11	
Sideroblastic anemia, X-linked		< 200 cases
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
Sirenomelia	1**	
Sjögren-Larsson syndrome	0.4	
Skeletal dysplasia - intellectual deficit		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 - 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 - epilepsy, X-linked		6 cases
Spherocytosis hereditary	20	
Spinal muscular atrophy - Dandy-Walker complex - cataracts		2 cases
Split hand - split foot	1.1	
Split hand - split foot - deafness		22 cases
Split hand - urinary anomalies - spina bifida		3 cases
Spondyloenchondrodysplasia		36 cases
Spondylometaphyseal dysplasia	0.34	
Spondylometaphyseal dysplasia - cone-rod dystrophy		8 cases
Spondylometaphyseal dysplasia with combined immunodeficiency		4 cases
Spontaneous periodic hypothermia		< 30 cases
Sporadic inclusion body myositis	0.49	
Sporotrichosis		55 cases
Squamous cell carcinoma of head and neck	46	

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Stapes ankylosis with broad thumbs and toes		6 families
Stargardt disease	11.25	
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	
Stiff-man syndrome	0.1	
Still's disease, adult onset	1.23	
Stimmler syndrome		2 cases
Stoll-Alembik-Finck syndrome		2 cases
Stoll-Alembik-Finck syndrome		1 case
Stormorken-Sjaastad-Langslet syndrome		6 cases
Stromal keratitis	16	
Subcorneal pustular dermatosis		200 cases
Succinic acidemia		50 cases
Succinyl-CoA acetoacetate transferase deficiency		10 cases
Sucrase-isomaltase deficiency, congenital	20	
Summitt syndrome		3 cases
Superficial pemphigus	1.2	
Supranuclear palsy, progressive	3.7	
Supravalvar aortic stenosis	12.5	
Susac syndrome		< 100 cases
Symphalangism with multiple anomalies of hands and feet		6 cases
Symphalangism, distal		< 5 families
Syndactyly - telecanthus - anogenital and renal malformations		6 cases
Syndactyly, Cenani-Lenz type		< 30 cases
Syndactyly, type 1	25	
Syndactyly, type 4		4 cases
Syngnathia multiple anomalies		2 cases
Synspondylism		24 cases
Syringomyelia	8.4	
Takayasu arteritis	0.45	
Talo-patello-scapoid osteolysis		2 cases
Tangier disease		> 70 cases
Taurodontia - absent teeth - sparse hair		< 15 cases
Tay-Sachs disease	0.3**	
Teebi-Shaltout syndrome		2 cases
Terminal osseous dysplasia - pigmentary defects		18 cases
Tetraamelia - pulmonary hypoplasia		5 families
Tetralogy of Fallot	45	
Thalamic calcifications, symmetrical		29 cases
Thalidomide embryopathy		5000 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Thanatophoric dwarfism	3,5**	
Thiamine-responsive megaloblastic anemia with diabetes mellitus and sensorineural deafness		30 families
Thomsen and Becker disease	5	
Thoracolyngopelvic dysplasia		< 10 cases
Thrombocythemia, essential	27.5	
Thrombocytopenia - Robin sequence		2 cases
Thrombocytopenic purpura, autoimmune	25	
Thumb absent - short stature - immune deficiency		3 cases
Thumb stiffness - brachydactyly - intellectual deficit		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		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
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
Transmissible spongiform encephalopathies	0.3	
Transposition of the great arteries	32.5	
Treacher-Collins syndrome	6	
Treft-Sanborn-Carey syndrome		23 cases
Trichodontal syndrome		< 5 families
Tricho-dento-osseous syndrome		> 30 cases
Trichodysplasia - amelogenesis imperfecta		1 family
Trichomegaly - cataract - spherocytosis, hereditary		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

** Prevalence at birth

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
Trigonocephaly - broad thumbs		2 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 (HHH) syndrome		50 cases
Triplo-X syndrome	42.5	
Trisomy 13	13**	
Trisomy 18	9**	
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		< 100 cases
Uhl anomaly		84 cases
Ulbricht-Hodes syndrome		3 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	
Vasculitis	6.3	
VATER association	23	
Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay		2 cases
Vitamin B12 responsive methylmalonic acidemia, type cbl A		60 cases
Von Hippel-Lindau disease	0.2	
Von Willebrand disease	12.5	
Von Willebrand syndrome, acquired		300 cases
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		2 cases

Diseases name	Estimated prevalence (/100,000)	Number of published cases or families
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
Winchester disease		12 cases
Wiskott-Aldrich syndrome	0.15	
Wolcott-Rallison syndrome		17 cases
Wolf-Hirschhorn syndrome	2**	
Wolfram syndrome	0.57	
Wolman disease	0.28**	
Woods-Black-Norbury syndrome		5 cases
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 agammaglobulinemia	0.45	
X-linked dominant chondrodysplasia punctata	0.5	
X-linked hereditary sensory and autonomic neuropathy with deafness		1 family
X-linked lymphoproliferative disease	0.1	
X-linked myopathy with excessive autophagy		15 families
X-linked severe congenital neutropenia		7 cases
X-linked spastic paraplegia, type 16		1 family
X-linked spastic paraplegia, type 2		< 100 cases
Zadik-Barak-Levin syndrome		1 case
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

