

# Prevalencia de las enfermedades raras: Una revisión bibliográfica Juli 2007

## Informes Periódicos de Orphanet

### Método

Lista de enfermedades por orden alfabético

Lista de enfermedades por orden de prevalencia decreciente

Por número de casos publicados

## Método

Se está haciendo una revisión sistemática de la literatura para proporcionar una estimación de la prevalencia de las enfermedades raras en Europa. Se publicará de forma regular un informe actualizado que substituirá a la versión anterior. La actualización contendrá nuevos datos epidemiológicos y modificaciones de los datos existentes, elaboradas a partir de nueva información disponible.

### **Estrategia de búsqueda**

Para realizar esta búsqueda se han utilizado varias fuentes de información:

- Páginas Web: Orphanet, e-medicine, GeneClinics, EMEA y OMIM ;

- Medline ha sido consultado utilizando el algoritmo de búsqueda:

«Nombre de enfermedad» AND [Epidemiology[MeSH:NoExp] OR Incidence[Title/abstract]  
OR Prevalence[Title/abstract] OR Epidemiology[Title/abstract] ;

- Libros de Medicina, literatura gris e informes de expertos son también fuentes de datos importantes.

### **Datos obtenidos**

Los valores de prevalencias que se proporcionan son la media entre los valores más altos y los más bajos recolectados. Cuando la prevalencia no estaba documentada la hemos calculado usando la incidencia:

- Para enfermedades congénitas que aparecen al nacer, prevalencia = incidencia al nacer x (esperanza de vida del paciente/esperanza de vida de la población general) ;

- Para las otras enfermedades raras, prevalencia = incidencia duración media de la enfermedad rara.

NB: esperanza de vida de la población francesa (78 años) es la esperanza de vida usada para la población general.

### **Actualización de datos**

Nueva información procedente de las bases de datos disponibles: EMEA, nuevas publicaciones científicas, literatura gris, opinión de expertos.

### **Limitaciones del estudio**

Los valores exactos de prevalencia de cada enfermedad son difíciles de obtener a partir de las bases de datos disponibles. Existe un cierto nivel de inconsistencia entre los estudios, poca documentación sobre el método usado, confusión entre la incidencia y prevalencia, y/o confusión entre incidencia al nacer e incidencia a largo plazo.

La validez de los estudios se da por sentada y no se somete a evaluación. Es probable que exista una sobreestimación para la mayoría de las enfermedades puesto que los pocos informes sobre prevalencia publicados se realizan generalmente en las regiones de mayor prevalencia y están basados en datos hospitalarios. Por ello, estas estimaciones son una indicación de la prevalencia asumida pero puede no ser exacta.

Para cualquier pregunta o comentario, por favor contacte con nosotros: [orphanet@orpha.net](mailto:orphanet@orpha.net)

## Prevalencia por orden alfabético de cada enfermedad

Nombre de enfermedades	Prevalencia estima (/100 000) o número de casos o familias publicados
2,4-dienoyl-CoA reductase deficiency	1 caso
2,8 dihydroxyadenine urolithiasis	1,7
3C syndrome	25 casos
3-hydroxy 3-methylglutaryl-CoA synthase	6 casos
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency	1
3M syndrome	40 casos
3-methylcrotonylglycinuria	2,25
3-methylglutaconic aciduria, type 1	20 casos
46 XX gonadal dysgenesis - epibulbar dermoid	1 caso
46,XX disorders of sex development - skeletal anomalies	2 casos
46,XY disorders of sex development due to 17-beta-hydroxysteroid dehydrogenase deficiency	0,68
4-hydroxybutyricaciduria	350 casos
5-oxoprolinase deficiency	8 casos
6-pyruvoyl-tetrahydropterin synthase deficiency	248 casos
Aarskog-Scott syndrome	>200 casos
Aase-Smith syndrome	<10 casos
Ablepharon-macrostomia syndrome	15 casos
Abruzzo-Erickson syndrome	4 casos
Acanthamoeba keratitis	1
Acalvaria	<1**
Acatasemia	3,1
Aceruloplasminemia	0,05
Achalasia - microcephaly	5 casos
Achalasia, primary	37,5
Acheiropodia	<10 familias
Achondrogenesis	>100 casos
Achondroplasia	4,5
Achromatopsia	2,5
Ackerman syndrome	8 casos
Acquired generalized lipodystrophy	80 casos
Acrocallosal syndrome, Schinzel type	34 casos
Acrocephalosyndactyly	4,6
Acrocraniofacial dysostosis	2 casos
Acrodermatitis enteropathica, zinc deficiency type	0,2
Acrofacial dysostosis autosomal recessive	2 casos
Acro-facial dysostosis postaxial, atypical	1 caso
Acro-facial dysostosis, Catania form	6 casos
Acrofacial dysostosis, Nager type	90 casos
Acro-facial dysostosis, Palagonia type	4 casos
Acro-facial dysostosis, Preis type	1 caso

Acro-facial dysostosis, Rodriguez type	<10 casos
Acro-fronto-facio-nasal dysostosis	5 casos
Acromegaloïd facial appearance syndrome	<20 casos
Acromegaloïd facies - hypertrichosis	<20 casos
Acromegaly	5
Acromegaly - cutis verticis gyrata - corneal leukoma	16 casos
Acromelanosis	<10 casos
Acromesomelic dysplasia Hunter-Thompson type	10 casos
Acromesomelic dysplasia, Brahimi-Bacha type	3 casos
Acromesomelic dysplasia, Maroteaux type	50 casos
Acromicric dysplasia	<40 casos
Acroosteolysis, autosomal dominant	50 casos
Acro-pectoral syndrome	22 casos
Acropectororenal field defect	12 casos
Acropectorovertebral dysplasia	<30 casos
Acrorenal syndrome	20 casos
Acrorenalandibular syndrome	7 casos
Acrorenooocular syndrome	<20 familias
Acute interstitial pneumonia	3,8
Acute lymphoblastic leukemia	7,5
Acute non lymphoblastic leukemia	7
Acute promyelocytic leukemia	8
Acute Respiratory Distress Syndrome, Adult	30
Acyl-CoA dehydrogenase, medium chain, deficiency	15
Adactylia unilateral	34
Adamantinoma	513 casos
Adducted thumbs - arthrogryposis, Christian type	3 familias
Adducted thumbs - arthrogryposis, Dundar type	5 casos
Adenosine monophosphate deaminase deficiency	200 casos
Adenylosuccinate lyase deficiency	50 casos
Adrenal hyperplasia, congenital	10
Adrenal hypoplasia congenital, X-linked	4
Adrenocortical carcinoma	1
Adrenoleukodystrophy, X-linked	3,5
Adult Onset Still's disease	1,23
ADULT syndrome	14 casos
Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis	3 casos
Agammaglobulinemia, X-linked	0,45
Agenesis of the corpus callosum - mental retardation - coloboma - micrognathia	2 casos
Aglossia - adactylia	<50 casos
Agnathia - holoprosencephaly - situs inversus	30 casos
Agonadism - dextrocardia - diaphragmatic hernia	6 casos
Aicardi syndrome	500 casos
Aicardi-Goutieres syndrome	30 casos

\*\* Prevalencia al nacer

Alagille syndrome	1,4
Alar cartilages hypoplasia - coloboma - telecanthus	2 casos
Albers-Schonberg disease	1
Albinism ocular - late onset sensorineural deafness	7 casos
Albinism-deafness syndrome	1 familia
Albright hereditary osteodystrophy	0,72
Albright-like syndrome	10 casos
Alexander disease	300 casos
Alkaptonuria	0,3
Alopecia - congenita keratosis palmoplantaris	1 familia
Alopecia - contractures - dwarfism - mental retardation	5 casos
Alopecia - epilepsy - pyorrhea - mental subnormality	12 casos
Alopecia - hypogonadism - extrapyramidal disorder	2 casos
Alopecia totalis	10,5
Alpers syndrome	0,025
Alpha thalassemia-mental retardation, X-linked	168 casos
Alpha-1 antitrypsin deficiency	20
Alpha-mannosidosis	0,1
Alpha-sarcoglycanopathy	0,57
Alport syndrome	2
Alström syndrome	300 casos
Alveolar echinococcosis	<1000 casos
Alzheimer disease, familial	5,3
Amaurosis - hypertrichosis	2 casos
Amaurosis congenita of Leber	2,5
Ambras syndrome	10 casos
Amelia, autosomal recessive	3 casos
Aminopterin embryofetopathy	17 casos
Amniotic bands	4**
Amoebiasis due to free-living amoebae	1,75
Amyotrophic lateral sclerosis	6
Androgen insensitivity syndrome	13
Anemia, sideroblastic, X-linked - ataxia	5 familias
Anencephaly	3,2**
Angelman syndrome	6,5
Angel-shaped phalango-epiphyseal dysplasia	15 casos
Angioma hereditary neurocutaneous	<10 familias
Angioneurotic edema	1
Angio-osteohypertrophic syndrome	1000 casos
Aniridia	1,75
Aniridia - absent patella	3 casos
Aniridia - cerebellar ataxia - mental deficiency	>10 familias
Aniridia - ptosis - mental retardation - obesity, familial type	3 casos
Aniridia - renal agenesis - psychomotor retardation	2 casos
Aniridia-mental retardation syndrome	2 casos
Anisakiasis	3,8
Ankyloblepharon - ectodermal defects - cleft lip palate	8 familias
Ankyloblepharon filiforme - imperforate anus	2 familias

Ankylosing vertebral hyperostosis - tylosis	8 casos
Anonychia - microcephaly	5 casos
Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges	11 casos
Anonychia with flexural pigmentation	3 casos
Anophthalmia/microphthalmia, isolated	14
Anophthalmia - hypothalamo-pituitary insufficiency	30 casos
Anophthalmia - megalocornea - cardiopathy - skeletal anomalies	3 casos
Anophthalmia - short stature - obesity	1 caso
Anophthalmia plus syndrome	4 casos
Anophthalmia/microphthalmia - esophageal atresia	14 casos
Anorectal malformation	24
Antisynthetase syndrome	1,5
Antley-Bixler syndrome	34 casos
Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis	16 casos
Aortic aneurysm syndrome, Loey-Dietz type	10 familias
Aortic arch anomaly - peculiar facies - mental retardation	4 casos
Aortic dilatation- joint hypermobility- arterial tortuosity	22 casos
Aorto-ventricular tunnel	130 casos
Apert syndrome	1,25
Aphalangia - syndactyly - microcephaly	1 familia
Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis	3 casos
Aplasia cutis - myopia	4 casos
Aplasia cutis congenita - intestinal lymphangiectasia	3 casos
Aplasia cutis congenita of limbs recessive	6 casos
Arachnodactyly - mental retardation - dysmorphism	3 casos
Arachnodactyly - ossification abnormal - mental retardation	5 casos
Aredyld syndrome	3 casos
Argininemia	31 casos
Arhinia - choanal atresia - microphthalmia	4 casos
Aromatase deficiency	13 casos
Arrhinia	20 casos
Arrhythmogenic right ventricular dysplasia	43,5
Arterial dissection - lentiginosis	4 casos
Arthritis-related enthesitis	5,7
Arthrogyposis - hyperkeratosis, lethal form	2 casos
Arthrogyposis - renal dysfunction - cholestasis	<150 casos
Arthrogyposis iugr thoracic dystrophy	1 caso
Arthrogyposis multiplex congenita	30
Arthrogyposis multiplex congenita - whistling face	10 casos
Ascher syndrome	50 casos
Astley-Kendall dysplasia	<10 casos
Ataxia - apraxia - mental retardation, X-linked	9 casos
Ataxia - deafness - optic atrophy, lethal	12 casos
Ataxia telangiectasia	1

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

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

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CADASIL syndrome	500 casos
Calpainopathy	3,8
Calvarial doughnut lesions - bone fragility	20 casos
CAMFAK syndrome	3 familias
CAMOS syndrome	5 casos
Campomelia Cumming type	8 casos
Campomelic dysplasia	0,35
Campptobrachydactyly	1 familia
Campptodactyly - fibrous tissue hyperplasia - skeletal dysplasia	3 casos
Campptodactyly - tall stature - scoliosis - hearing loss	30 casos
Campptodactyly - taurinuria	4 familias
Campptodactyly syndrome, Guadalajara type 1	8 casos
Campptodactyly syndrome, Guadalajara type 2	2 casos
Camurati-Engelmann disease	200 casos
Cantrell pentalogy	0,55**
Capillary leak syndrome	57 casos
Carbamoylphosphate synthetase deficiency	0,7
Carcinoma of the gallbladder	6,5
Cardiac conduction disease, dilated cardiomyopathy and brachydactyly	10 casos
Cardiogenital syndrome	7 casos
Cardiomyopathy - cataract - hip spine disease	9 casos
Cardiomyopathy - renal anomalies	2 casos
Cardiomyopathy, familial dilated	17,5
Carey-Fineman-Ziter syndrome	<20 casos
Carney complex	160 casos
Carnitine palmitoyl transferase 1 deficiency	35 casos
Carnitine palmitoyl transferase 2 deficiency	>100 casos
Carnitine-acylcarnitine translocase deficiency	30 casos
Carnosinemia	30 casos
Caroli's disease	<250 casos
Carpenter syndrome	40 casos
Carpotarsal osteochondromatosis	<10 casos
Carpotarsalosteolysis, autosomal recessive	<10 casos
Castleman disease	400 casos
Cataract - ataxia - deafness	2 casos
Cataract - cardiomyopathy	30 casos
Cataract - deafness - hypogonadism	3 casos
Cataract - hypertrichosis - mental retardation	1 caso
Cataract - mental retardation - hypogonadism	10 casos
Cataract - microphthalmia - septal defect	2 casos
Cataract - nephropathy - encephalopathy	2 casos
Cataract anterior polar	4,4
Cataract congenital, Volkmann type	<100 casos
Cataract, Hutterite type	1 familia
Cataract total, congenital	7,9
Cataract-glaucoma	3 familias
Cataract-microcornea syndrome	8 familias
Catel-Manzke syndrome	27 casos

Cat-eye syndrome	1,35
Cat-scratch disease	6,6
Caudal dysgenesis familial type	4 casos
CDG syndrome	1,5**
CDG syndrome type Ia	> 300 casos
CDG syndrome type Ib	20 casos
CDG syndrome type Ic	>30 casos
CDG syndrome type Id	5 casos
CDG syndrome type Ie	7 casos
CDG syndrome type If	4 casos
CDG syndrome type Ig	6 casos
CDG syndrome type Ih	5 casos
CDG syndrome type Ii	1 caso
CDG syndrome type IIa	4 casos
CDG syndrome type IIb	1 caso
CDG syndrome type IIc	1 caso
CDG syndrome type IId	1 caso
CDG syndrome type IIe	2 casos
CDG syndrome type Ij	1 caso
CDG syndrome type Ik	4 casos
CDG syndrome type IL	2 casos
CEDNIK syndrome	7 casos
Celiac disease - epilepsy - occipital calcifications	170 casos
Central neurocytoma	>100 casos
Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss	1-2 familias
Cerebellar ataxia, autosomal dominant	2,15
Cerebellar ataxia, autosomal recessive	7
Cerebral arteriovenous fistula	6
Cerebral gigantism - jaw cysts	<10 casos
Cerebro-costo-mandibular syndrome	60 casos
Cerebro-oculo-nasal syndrome	10 casos
Cerebretinal vasculopathy	3 familias
Ceroid lipofuscinosis, neuronal	4
Cervical hypertrichosis - peripheral neuropathy	3 casos
CHAND syndrome	>10 casos
Chaotic atrial tachycardia	100 casos
Char syndrome	10 casos
Charcot-Marie-Tooth disease (generic term)	32,5
Charcot-Marie-Tooth disease, X-linked	1,6
CHARGE association	0,14
Chediak-Higashi syndrome	>10 casos
CHILD syndrome	30 casos
Choanal atresia - deafness - cardiac defects dysmorphism	5 casos
Cholangiocarcinoma	10
Cholestasis - lymphoedema, syndrome	20-50 casos
Cholestasis - pigmentary retinopathy - cleft palate	4 casos
Cholesteryl ester storage disease	<50 casos
Chondrodysplasia - disorder of sex development	2 casos
Chondrodysplasia - situs inversus- imperforate anus - polydactyly	1 caso

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Chondrodysplasia lethal, recessive	4 casos
Chondrodysplasia punctata, rhizomelic type	1
Chondrodysplasia, Blomstrand type	13 casos
Chordoma	0,05
Chorioretinal atrophy, progressive bifocal	2 familias
Choroidal atrophy - alopecia	2 casos
Choroidal dystrophy, central areolar	3,33
Choroideremia	2
Choroideremia - deafness - obesity	4 casos
Choroidocerebral calcification syndrome, infantile form	10 casos
Christ-Siemens-Touraine syndrome	0,35
Chromosome Y deletion	42
Chronic hiccup	1
Chronic inflammatory demyelinating polyneuropathy	4,4
Chronic recurrent multifocal osteomyelitis, juvenile	>260 casos
Churg-Strauss syndrome	1
Chylomicron retention disease	40 casos
CINCA syndrome	100 casos
Cleft lip - retinopathy	2 casos
Cleft lip palate - malrotation - cardiopathy	4 casos
Cleft lip palate - mental retardation - corneal opacities	2 casos
Cleft palate - cardiac defect - genital anomalies - ectrodactyly	5 casos
Cleft palate - short stature - vertebral anomalies	2 casos
Cleft palate - stapes fixation - oligodontia	2 casos
Cleft palate-lateral synechia syndrome	7 casos
Cleido-rhizomelic syndrome	2 casos
Clouston syndrome	1
Cloverleaf skull syndrome	150 casos
COACH syndrome	8 casos
Coats disease	2
Cobb syndrome	35 casos
Cockayne syndrome	200 casos
Coffin-Lowry syndrome	0,55
Coffin-Siris syndrome	40 casos
Cogan syndrome	200 casos
Cohen syndrome	100 casos
Collagenous colitis	10,5
Coloboma of macula - brachydactyly type B	12 casos
Coloboma uveal - cleft lip palate - mental retardation	12 casos
Coloboma, ocular	1
Colobomatous - microphthalmia - heart disease - hearing loss	10 casos
Cone rod dystrophy	2,5
Cone rod dystrophy - amelogenesis imperfecta	29 casos
Congenital alopecia, X linked	1 familia
Congenital anosmia, isolated	<15 casos
Congenital brain dysgenesis due to glutamine synthetase deficiency	<30 casos

Congenital bronchobiliary fistula	23 casos
Congenital cataracts - facial dysmorphism - neuropathy	100 casos
Congenital ichthyosis - microcephalus - quadriplegia	2 casos
Congenital indifference to pain	> 20 casos
Congenital lobar emphysema	4,5
Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	3 casos
Congenital myasthenic syndromes	0,75
Congenital rubella syndrome	1**
Contractures - ectodermal dysplasia - cleft lip palate	2 casos
Cooper-Jabs syndrome	2 casos
Corneal anesthesia - deafness - mental retardation	2 casos
Corneal crystals - myopathy - neuropathy	1 caso
Corneal crystals myopathy neuropathy	1 caso
Corneal dystrophy - ichthyosis - microcephaly - mental retardation	1 caso
Corneal dystrophy - perceptive deafness	<10 casos
Corneal-cerebellar syndrome	2 casos
Cornelia de Lange syndrome	5,25
Coronary artery disease - hyperlipidemia - hypertension - diabetes - osteoporosis	1 familia
Corpus callosum agenesis - neuronopathy	19
Corpus callosum dysgenesis, X-linked recessive	11 casos
Corpus callosum, agenesis - cataract - immunodeficiency	8 casos
Cortical blindness - mental retardation - polydactyly	3 casos
Corticobasal degeneration	4
Corticosteroid-sensitive aseptic abscesses	49 casos
Costello syndrome	200 casos
Costocoracoid ligament congenitally short	1 familia
Cowden syndrome	0,45
Coxo-auricular syndrome	4 casos
Coxo-podo-patellar syndrome	47 casos
Craniodiaphyseal dysplasia	>20 casos
Cranio-digital syndrome - mental retardation	4 casos
Cranio-ectodermal dysplasia	15 casos
Craniofacial conodysplasia	1 familia
Cranio-facial dysmorphism - coloboma - corpus callosum agenesis	3 casos
Craniofacial dyssynostosis	0,05
Craniofacial-deafness-hand syndrome	3 casos
Craniofaciocardioskeletal syndrome	4 casos
Craniofrontonasal dysplasia - poland anomaly	3 casos
Craniofrontonasal syndrome, Teebi type	20 casos
Cranioleptocrotaphic dysplasia	21 casos
Cranioleptocrotaphic dysplasia	70 casos
Cranioosteopathy	4 casos
Craniosynostosis - brachydactyly	5 casos
Craniosynostosis - Dandy Walker hydrocephalus	4 casos

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Craniosynostosis - fibular aplasia	2 casos
Craniosynostosis - intracranial calcifications	3 casos
Craniosynostosis radial aplasia, Imaizumi type	2 casos
Craniosynostosis, Boston type	19 casos
Craniosynostosis, Philadelphia type	1 familia
Craniotubular syndrome	2 casos
Creutzfeldt-Jakob disease	0,1
Crigler-Najjar syndrome	200 casos
Crisponi syndrome	18 casos
Criss-cross heart	0,8
Crouzon disease	2
Cryoglobulinemia	1,33*
Cryptomicrotia - brachydactyly - excess fingertip arch	2 casos
Cryptosporidiosis	34
Curry-Jones syndrome	5 casos
Cutaneous albinism, ermine phenotype	3 casos
Cutaneous lymphoma	8,3
Cutaneous mastocytosis	0,75
Cutaneous neuroendocrine carcinoma	0,5
Cutaneous photosensitivity - colitis lethal	3 casos
Cutis gyrata - acanthosis nigricans - craniosynostosis	6 casos
Cutis laxa	>100 casos
Cutis marmorata telangiectatica congenita	300 casos
Cutis verticis gyrata - mental deficiency	1,02
Cutis verticis gyrata - thyroid aplasia - mental retardation	5 casos
Cyprus facial neuromusculoskeletal syndrome	1 familia
Cystathioninuria	7
Cystic fibrosis	12
Cystic hamartoma of lung and kidney	<5 casos
Cystic hygroma lethal - cleft palate	2 casos
Cystinosis	0,5
Cystinuria	14
Cystoid macular dystrophy	6 familias
Dacryocystitis osteopoikilosis	5 casos
Dahlberg-Borer-Newcomer syndrome	2 casos
Dandy Walker - macrocephaly	2 casos
Dandy Walker malformation - postaxial polydactyly	2 casos
Darier disease	1,5
Deaf - blind - hypopigmentation	2 casos
Deafness - enamel hypoplasia - nail defects	6 casos
Deafness - lymphoedema - leukemia	<10 casos
Deafness - opticoacoustic nerve atrophy - dementia	3 casos
Deafness - peripheral neuropathy - arterial disease	4 casos
Deafness - skeletal dysplasia - lip granuloma	8 casos
Deafness - tubular acidosis - anemia	2 casos
Deafness - vitiligo - achalasia	2 casos
Deafness, autosomal dominant, nonsyndromic, sensorineural, type DFNA	8

Deafness, autosomal recessive, nonsyndromic, sensorineural, type DFNB	44
Deafness, X-linked, non syndromic, sensorineural, type DFN	0,6
Deafness-infertility syndrome	3 familias
Deafness-mental retardation, Martin-Probst type	3 casos
Deafness-mental retardation, Martin-Probst type	3 casos
Defective expression of HLA class 2	100 casos
Dehydratase deficiency	21 casos
Deletion 18p	<200 casos
Deletion 2q24	23 casos
Delta-sarcoglycanopathy	0,57
DEND syndrome	14 casos
Dentinogenesis imperfecta - short stature - hearing loss - mental retardation	2 casos
Dentatorubral pallidolusian atrophy	<0,3
Denys-Drash syndrome	150 casos
Depigmentation of the iris, acute, bilateral	5 casos
Dermatitis herpetiformis	20,2
Dermato-cardio-skeletal syndrome, Borrone type	2 casos
Dermatofibrosarcoma protuberans	10
Dermatoleukodystrophy	2 casos
Dermatomyositis	14,8
Dermatoosteolysis, Kirghizian type	5 casos
Dermoodontodysplasia	14 casos
Dermopathy restrictive lethal	30 casos
Desbuquois syndrome	>40 casos
Desmosterolosis	2 casos
Developmental delay due to 2-methylbutyryl-coA dehydrogenase deficiency	<30 casos
Developmental dysphasia familial	6 familias
Developmental malformations - deafness - dystonia	2 casos
Diabetes insipidus, nephrogenic	0,5
Diabetes mellitus, neonatal	0,2
Diabetes, neonatal - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys	2 casos
Diaphanospondylodysostosis	<10 casos
Diaphragmatic defect - limb deficiency - skull defect	4 casos
Diaphragmatic hernia - exomphalos - corpus callosum agenesis	13 casos
Diaphragmatic hernia, congenital	15
Diastrophic dwarfism	3,5
Diffuse leiomyomatosis with Alport syndrome	0,1
Diffuse neonatal haemangiomas	<70 casos
Diffuse palmoplantar keratoderma - acrocyanosis	10 casos
Diffuse palmoplantar keratoderma, Norrbotten dominant type	2,5
Digito-reno-cerebral syndrome	<10 casos
Digitotalar dysmorphism	6
Dihydropteridine reductase deficiency	134 casos
Dihydropyrimidinuria	7 casos

\*\* Prevalencia al nacer

Dincsoy-Salih-Patel syndrome	2 casos
Disorder of sex development - mental retardation	3 casos
Distal monosomy 5q	10 casos
Distal monosomy 8p	20 casos
Distal myopathy with vocal cord weakness	12 casos
Distal myopathy, Nonaka type	0,1
Distal myopathy, Welander type	10
Distal myopathy, with early respiratory muscle involvement	24 casos
Distal myopathy, with posterior leg and anterior upper limb involvement	12 casos
DOOR syndrome	<50 casos
Dopamine beta-hydroxylase, deficiency of	12 casos
Dopa-responsive dystonia	0,3
Double outlet left ventricle	32 casos
Double uterus - hemivagina - renal agenesis	<60 casos
Duane anomaly - myopathy - scoliosis	2 casos
Duane syndrome	10
Dubowitz syndrome	150 casos
Duker-Weiss-Siber syndrome	4 casos
Duodenal atresia	8,55
Duplication 8q	>30 casos
Dyggve-Melchior-Clausen disease	60 casos
Dyschondrosteosis - nephritis	1 familia
Dyserythropoietic anemia, congenital	1
Dyskeratosis congenita	0,1
Dysmorphism - short stature - deafness - pseudohermaphroditism	2 casos
Earlobes, thickened - conductive deafness	2 familias
Early infantile epileptic encephalopathy	88 casos
Early myoclonic encephalopathy	30 casos
Early onset torsion dystonia	0,4
Ear-patella-short stature syndrome	42 casos
Ebstein anomaly	0,75
Ectodermal dysplasia - alopecia - preaxial polydactyly	1 caso
Ectodermal dysplasia - arthrogryposis - diabetes mellitus	1 caso
Ectodermal dysplasia - blindness	2 casos
Ectodermal dysplasia - mental retardation - syndactyly	1 caso
Ectodermal dysplasia - absent dermatoglyphics	<30 casos
Ectodermal dysplasia anhidrotic - immunodeficiency - osteopetrosis - lymphedema	2 casos
Ectodermal dysplasia, «pure» hair-nail type	<20 casos
Ectodermal dysplasia, Berlin type	4 casos
Ectodermal dysplasia, hypohidrotic - hypothyroidism - ciliary dyskinesia	3 casos
Ectodermal dysplasia, hypohidrotic, autosomal dominant	40 casos
Ectodermal dysplasia-skin fragility syndrome	10 casos
Ectodermic dysplasia - hypothyroidism - cleft	3 casos
Ectopia lentis - chorioretinal dystrophy - myopia	4 casos

Ectopia lentis isolated	6,4
Ectrodactyly - ectodermal dysplasia	5 casos
EEM syndrome	7 familias
Ehlers-Danlos syndrome type 10	1 familia
Ehlers-Danlos syndrome, classic type	3,5
Ehlers-Danlos syndrome, type 3	12,5
Ehlers-Danlos syndrome, type 4	1
Ehlers-Danlos syndrome, type 5	2 familias
Ehlers-Danlos syndrome, type 7C	7 casos
Ehrlichiosis	<50 casos
Eiken syndrome	6 casos
Elejalde syndrome	30 casos
Elliptocytosis, hereditary	37,5
Ellis Van Creveld syndrome	150 casos
Emery-Dreifuss muscular dystrophy	1,5
Enamel hypoplasia cataract hydrocephaly	1 caso
Encephalo-cranio-cutaneous lipomatosis	45 casos
Encephalopathy due to hydroxykynureninuria	<30 casos
Encephalopathy with neuroserpin inclusion bodies, familial form	>5 familias
Enchondromatosis	> 600 casos
Eng-Strom syndrome	2 casos
Eosinophilic fasciitis	200 casos
Eosinophilic gastroenteritis	280 casos
Eosinophilic pneumonia, acute idiopathic	>100 casos
Epidermal nevus syndrome	>400 casos
Epidermolysis bullosa simplex - limb girdle muscular dystrophy	<20 casos
Epidermolysis bullosa, acquired	100 casos
Epidermolysis bullosa, dystrophic	0,27
Epidermolysis bullosa, epidermolytic	2,5
Epidermolysis bullosa, junctional	0,06
Epilepsy - microcephaly - skeletal dysplasia	2 casos
Epilepsy - telangiectasia	6 casos
Epilepsy, pyridoxin-dependent	0,15
Epiphyseal dysplasia multiple	5
Episodic ataxia, type 3	1 familia
Episodic ataxia, type 4	2 familias
Erdheim-Chester disease	178 casos
Erythralgia, primary	30 familias
Erythroderma lethal, congenital	17 casos
Erythrokeratoderma - ataxia	25 casos
Erythrokeratoderma variabilis, Mendes da Costa type	>200 casos
Esophageal carcinoma	8
Esthesioneuroblastoma	<1000 casos
Evans syndrome	0,1
Ewing sarcoma	0,1
Exostoses, multiple	4
Eyebrow duplication syndactyly	3 casos
Fabry disease	1,75

\*\* Prevalencia al nacer

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

Fructose intolerance	5
Fructose-1,6-bisphosphatase deficiency	2,5
Fryns syndrome	7**
Fucosidosis	100 casos
Fuhrmann syndrome	11 casos
Fumaric aciduria	>20 casos
Fuqua-Berkovitz syndrome	2 casos
Galactosemia	6,6
Galloway-Mowat syndrome	40 casos
Gamma aminobutyric acid transaminase deficiency	2 casos
Gamma-glutamyl transpeptidase deficiency	7 casos
Gamma-glutamylcysteine synthetase deficiency	9 casos
Gamma-sarcoglycanopathy	1,96
GAP0 syndrome	27 casos
Gardner-Morrison-Abbot syndrome	3 casos
Gastric cancer	20
Gastrointestinal stromal tumor	1,8
Gastroschisis	12
Gaucher - ichthyosis - restrictive dermopathy	4 casos
Gaucher disease	1
Gaucher disease, type 1	0,94
Gaucher disease, type 2	0,01
Gaucher disease, type 3	0,05
Geleophysic dwarfism	27 casos
Gelineau disease	49
Gemss syndrome	3 casos
Genitopatellar syndrome	7 casos
German syndrome	5 casos
Geroderma osteodysplastica	30 casos
Giant cell arteritis	8,9
Giant pigmented hairy nevus	2
Glaucoma-sleep apnea	5 casos
Glioblastoma	11
Global developmental delay - osteopenia - ectodermal defect	3 casos
Glomerulonephritis - sparse hair - telangiectases	<10 casos
Glossopalatine ankylosis - cataracts - digital anomalies	1 caso
Glucocorticoid deficiency, familial	50 casos
Glucose-galactose malabsorption	200 casos
Glucosephosphate isomerase deficiency	50 casos
Glutaryl-CoA dehydrogenase deficiency	0,4
Glutathione synthetase deficiency	65 casos
Glycogen storage disease due to LAMP-2 deficiency	30 casos
Glycogen storage disease, type 2	1,1
Glycogen storage disease, type 4	0,6
Glycogen storage disease, type 7	<30 casos
Glycogen storage, type 0	16 casos
Goldberg-Shprintzen megacolon syndrome	10 casos
Goldenhar syndrome	3,5
Goldmann-Favre syndrome	<50 casos

\*\* Prevalencia al nacer

Gombo syndrome	4 casos
Gonadal dysgenesis, XX type	12
Gonadal dysgenesis, XY type - associated anomalies	2 casos
Goodman syndrome	3 casos
Goodpasture syndrome	0,64
Gorham-Stout disease	200 casos
Gorlin syndrome	1
Gorlin-Chaudhry-Moss, syndrome	4 casos
GRACILE syndrome	2,12**
Graft versus host disease	3,4
Grange syndrome	6 casos
Granulomatous arthritis of childhood	40 familias
Granulomatous disease, chronic	0,2
Granulomatous slack skin	<50 casos
Gräsbeck-Imerslund disease	300 casos
Gray platelet syndrome	20 casos
Great vessels transposition (TGV)	32,5
Greenberg dysplasia	<10 casos
Greig syndrome	100 casos
Griscelli disease	60 casos
Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate	4 casos
Growth retardation - microcephaly - digital abnormalities - hypospadias	4 casos
GTP cyclohydrolase I deficiency	17 casos
Guanidinoacetate methyltransferase deficiency	9 casos
Guillain-Barré syndrome	47,5
Haemolytic anaemia due to glutathione reductase deficiency	3 casos
Haemolytic anaemia, nonspherocytic, due to hexokinase deficiency	17 familias
Hair defect - photosensitivity - mental retardation	3 casos
Hallermann-Streiff-Francois syndrome	<100 casos
Harding ataxia	1
Hartnup syndrome	4
Heart defects - limb shortening	2 casos
Hec syndrome	2 casos
Helicoid peripapillary chorioretinal degeneration	100 casos
Hemimelia	4,15
Hemiplegic migraine, familial	6,5
Hemolytic anemia due to adenylate kinase deficiency	12 casos
Hemolytic anemia, lethal - genital anomalies	2 casos
Hemophilia	7,7
Hemophilia, acquired	0,1
Hemorrhagic disorders due to collagen receptors deficiency	<20 casos
Hennekam syndrome	>50 casos
Hennekam-Beemer syndrome	2 casos
Hepatic veno-occlusive disease	11
Hepatic veno-occlusive disease - immunodeficiency	<25 casos
Hepatitis, chronic autoimmune	0,65

Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia	19 casos
Hereditary sensory and autonomic neuropathy, type 2	35 casos
Hereditary vascular retinopathy	1 familia
Hereditary vascular retinopathy	1 familia
HERNS syndrome	3 familias
Herpes simplex encephalitis	0,021*
Hersh-Podbruch-Weisskopf syndrome	2 casos
Heterotaxia	2,5
Hidrotic ectodermal dysplasia, Christianson-Fourie type	6 casos
Hidrotic ectodermal dysplasia, Halal type	4 casos
Hirschsprung disease	20
Hirschsprung disease - deafness - polydactyly	2 casos
Hirschsprung disease - nail hypoplasia - dysmorphism	3 casos
Histidinemia	4
Hodgkin lymphoma	10,5
Holoprosencephaly	7
Holt-Oram syndrome	1
Homocarnosinosis	4 casos
Homocystinuria due to cystathionine beta-synthase deficiency	0,4
Huntington disease	6,2
Hyaluronidase deficiency	1 caso
Hydrocephalus - blue sclerae - nephropathy	1 familia
Hydrocephalus - costovertebral dysplasia - Sprengel anomaly	8 casos
Hydrocephaly - tall stature - joint laxity	2 casos
Hydrolethalus	5**
Hydrops ectrodactyly syndactyly	1 caso
Hypercoagulability syndrome, due to glycosylphosphatidylinositol deficiency	2 casos
Hyperferritinemia, hereditary, with congenital cataracts	>64 casos
Hyperglycinemia, isolated nonketotic	0,2
Hyper-IGM syndrome, autosomal recessive	0,05**
Hyperimmunoglobulinemia D - recurrent fever	180 casos
Hyperkalemic periodic paralysis	0,75
Hyperkeratosis - hyperpigmentation syndrome	10 casos
Hyperlipidemia type 3	7,8
Hyperlipoproteinemia type 1	0,6
Hyperoxaluria	0,2
Hypertrichosis - brachydactyly - obesity - mental retardation	1 caso
Hypertrichosis cubiti - short stature	28 casos
Hypertrichosis lanuginosa congenita	<100 casos
Hypertrichosis lanuginosa, acquired	60 casos
Hypertrichotic osteochondrodysplasia	18 casos
Hypochondroplasia	3,3
Hypocomplementaemic leucocytoclastic vasculitis	<100 casos
Hypogammaglobulinemia due to CD19 deficiency	4 casos

\*\* Prevalencia al nacer

Hypogonadism - retinitis pigmentosa	3 casos
Hypokalemic periodic paralysis	1
Hypokeratosis, palmo-plantar, circumscribed	17 casos
Hypomagnesemia with hypocalciuria	3 familias
Hypomagnesemia with normocalciuria	2 casos
Hypomandibular facio-cranial dysostosis	4 casos
Hypomyelination - congenital cataract	8 casos
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 casos
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 casos
Hypoparathyroidism - deafness - renal disease	12 casos
Hypoparathyroidism familial isolated	<10 familias
Hypoparathyroidism X-linked	2 familias
Hypopituitarism - microphthalmia	<10 casos
Hypopituitarism - postaxial polydactyly	6 casos
Hypotelorism - cleft palate - hypospadias	13 casos
Hypothyroidism - dermoid cyst - cleft palate	1 caso
Hypothyroidism, congenital	29
Hypotrichosis - lymphedema - telangiectasia	4 casos
Hypotrichosis simplex	38 casos
Hypotrichosis-mental retardation lopes type	1 caso
IBIDS syndrome	15 casos
ICF syndrome	50 casos
Ichthyosis - alopecia - ectropion - mental retardation	4 casos
Ichthyosis - hepatosplenomegaly - cerebellar degeneration	2 casos
Ichthyosis - male hypogonadism	5 casos
Ichthyosis - oral and digital anomalies	2 casos
Ichthyosis bullosa of Siemens	<20 casos
Ichthyosis congenita - biliary atresia	2 casos
Ichthyosis congenita, harlequin type	<100 casos
Ichthyosis - deafness - mental retardation - skeletal anomalies	1 caso
Ichthyosis follicularis-atrichia-photophobia syndrome	10 casos
Ichthyosis prematurity syndrome	16 familias
Ichthyosis, X-linked	16,6
Idiopathic hypereosinophilic syndrome	10
Idiopathic hypersomnia	4
IMAGe syndrome	<20 casos
Iminoglycinuria	6,68
Immune dysregulation - polyendocrinopathy - enteropathy, X linked	7 familias
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	<15 casos
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	100 casos
Immunodeficiency with natural-killer cell deficiency	4 casos
Immunodeficiency, common variable	7,5
Inappropriate antidiuretic hormone secretion syndrome	2 casos

Inclusion body myositis, IBM	0,49
Incontinentia pigmenti	0,2
Infant epilepsy with migrant focal crisis	29 casos
Infantile neuroaxonal dystrophy	>150 casos
Inflammatory pseudotumor of the liver	143 casos
Infundibulopelvic stenosis - multicystic kidney	1 familia
Insomnia, familial fatal	27 casos
Insulin resistance, short fifth metacarpals	1 familia
Intellectual deficit, severe - epilepsy - anal anomalies - distal phalangeal hypoplasia	2 casos
Internal carotid agenesis	100 casos
Interstitial granulomatous dermatitis - arthritis	<20 casos
Intrathoracic kidney vertebral fusion	1 caso
Iris coloboma with ptosis - intellectual deficit	10 casos
Iris dysplasia - hypertelorism - deafness	2 casos
Isotretinoin-like syndrome	6 casos
Isovaleric acidemia	1
IVIC syndrome	4 familias
Jackson-Weiss syndrome	2 familias
Jacobsen syndrome	150 casos
Jeune syndrome	0,2
Job syndrome	250 casos
Johanson-Blizzard syndrome	23 casos
Johnson neuroectodermal syndrome	<30 casos
Joubert syndrome	0,85
Juberg-Hayward syndrome	10 casos
Juvenile arthritis, idiopathic	41,8
Juvenile hyaline fibromatosis	40-50 casos
Juvenile idiopathic arthritis, systemic-onset	6,3
Juvenile macular degeneration, hypotrichosis	7 familias
Juvenile temporal arteritis	20 casos
Kabuki syndrome	1,16
Kaler-Garrity-Stern syndrome	2 casos
Kallmann syndrome	0,66
Kaposi's sarcoma	1,7
Kapur-Toriello syndrome	2 casos
Kartagener syndrome	2,5
Kasabach-Merritt syndrome	>175 casos
KBG syndrome	45 casos
Kearns-Sayre syndrome	223 casos
Kennedy disease	2,8
Keratoderma - epithelioma - dental abnormalities-hypogonadism	5 casos
Keratoderma - hypotrichosis - leukonychia	2 casos
Keratoderma palmoplantar - deafness	<10 familias
Keratoderma palmoplantar - spastic paralysis	25 casos
Keratosis follicularis - dwarfism - cerebral atrophy	6 casos
Keratosis palmaris et plantaris - clinodactyly	<20
Ketoacidosis due to betaketothiolase deficiency	60 casos
KID syndrome	>100 casos
Kimura disease	200 casos

\*\* Prevalencia al nacer

Klippel-Feil syndrome	2
Kniest-like dysplasia, lethal	2 casos
Krabbe disease	0,75**
Kudo-Tamura-Fuse syndrome	2 casos
Lacrimo-auriculo-dento-digital syndrome	20 casos
Lambert-Eaton myasthenic syndrome	1
Lamellar ichthyosis	>0,33
Langerhans cell histiocytosis	2
Larsen syndrome	100 casos
Laryngeal abductor paralysis - mental retardation	<20 casos
Laryngotracheoesophageal cleft	1,5
Lateral body wall complex	2**
Lathosterolosis	<5 casos
LCAT deficiency	80 casos
Leber hereditary optic neuropathy	6,5
Legg-Calve-Perthes disease	23
Leigh disease	2,75**
Lennox-Gastaut syndrome	15
Lenz-Majewski hyperostotic dwarfism	7 casos
LEOPARD syndrome	>70 casos
Leprechaunism	0,1**
Leptospirosis	0,24
Lesch-Nyhan syndrome	0,38
Lethal osteosclerotic bone dysplasia	8 familias
Leucinosis	15,6
Leukemia, chronic myeloid	6
Leukocyte adhesion deficiency	<350 casos
Leukodystrophy with oligodontia	4 casos
Leukoencephalopathy - metaphyseal chondrodysplasia	4 casos
Leukoencephalopathy - palmoplantar keratoderma	4 casos
Lewis-Pashayan syndrome	3 casos
Lewis-Sumner syndrome	0,9
Lhermitte-Duclos disease	220 casos
Lichenstein syndrome	2 casos
Li-Fraumeni syndrome	400 familias
Limb-mammary syndrome	27 casos
Lipoamide dehydrogenase deficiency	20 casos
Lipodystrophy - mental retardation - deafness	3 casos
Lipodystrophy, Berardinelli type	0,25
Lipodystrophy, familial partial, associated with PPARG mutations	10 casos
Lipodystrophy, familial partial, due to AKT2 mutations	1 familia
Lipodystrophy, familial partial, Dunnigan type	200-300 casos
Lipodystrophy, familial partial, Köbberling type	<20 casos
Lipodystrophy, partial acquired	250 casos
Lipoid proteinosis	>280 casos
Lissencephaly - immunodeficiency	1 caso
Lissencephaly type 1, due to LIS 1 anomalies	0,3
Lissencephaly type 2	0,12

Lissencephaly type III - familial fetal akinesia sequence	5 casos
Lissencephaly type III - metacarpal bone dysplasia	2 casos
Liver disease - retinitis pigmentosa - polyneuropathy - epilepsy	4 casos
Long QT syndrome, familial	25
Lopez-Hernandez syndrome	11 casos
Low birth weight - dwarfism - dysgammaglobulinemia	2 casos
Lowe syndrome	0,19
Lumbosacral vertebrae, posterior fusion of - blepharoptosis	3 casos
Lung cancer, small cell	5
Lymphangioliomyomatosis	0,1
Lymphoblastic lymphoma	10
Lymphoedema - atrial septal defects - facial changes	3 casos
Lymphoedema - cerebral arteriovenous anomaly	5 casos
Lymphoedema, congenital	8,8
Macrocephaly - cutis Marmorata Telangiectatica Congenita	40 casos
Macrocephaly - immune deficiency - anemia	2 casos
Macrocephaly - short stature - paraplegia	2 casos
Macroepiphyseal dysplasia, Macalister Coe type	1 caso
Macrogyria - pseudobulbar palsy	4 casos
Macrophagic myofasciitis	1
Macrostomia - preauricular tags - external ophthalmoplegia	9 casos
Malakoplasiya	500 casos
Malignant hyperthermia	33
Malignant hyperthermia - arthrogyrosis - torticollis	4 casos
Malonic aciduria	17 casos
Mandibuloacral dysplasia	37 casos
Mantle cell lymphoma	3,9
Marden-Walker syndrome	30 casos
Marfan syndrome	30
Marie Unna, congenital - hypotrichosis	12 familias
Marinesco-Sjogren syndrome	100-200 casos
Marshall's syndrome with periodic fever	30 casos
Marshall-Smith syndrome	33 casos
Martinez-Monasterio-Pinheiro syndrome	1 caso
MASA syndrome	3,5
Maternal hyperphenylalaninemia	1,25
Matthew-Wood syndrome	5 casos
Mayer-Rokitansky-Küster-Hauser syndrome	9
McCune-Albright syndrome	158 casos
Meckel syndrome	4**
Meconium aspiration syndrome	2,44
Median cleft of the upper lip - corpus callosum lipoma - cutaneous polyps	>10 casos
Medullary cystic kidney disease, autosomal dominant	0,11

\*\* Prevalencia al nacer

Megacystis microcolon - intestinal hypoperistalsis - hydronephrosis	89 casos
Megaepiphyseal dwarfism	1 caso
Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus	6 casos
MEHMO syndrome	7 casos
MEHMO syndrome	7 casos
Melanoma, familial	46,8
MELAS syndrome	16
Melorheostosis	300 casos
Mendelian susceptibility to atypical mycobacteria	0,059
Meniere's disease	42,5
Menkes syndrome	0,7
Mental retardation - cubitus valgus - unusual facies	5 casos
Mental retardation - dysmorphism - hypogonadism - diabetes mellitus	4 casos
Mental retardation - hypoplastic corpus callosum - preauricular tag	3 casos
Mental retardation - microcephaly - phalangeal - facial abnormalities	6 casos
Mental retardation - progressive spasticity, X-linked	1 familia
Mental retardation - sparse hair - brachydactyly	6 casos
Mental retardation multiple nevi	1 caso
Mental retardation X-linked - dysmorphism	8 casos
Mental retardation X-linked - psychosis - macroorchidism	6 casos
Mental retardation X-linked - seizures - short stature - midface hypoplasia	17 casos
Mental retardation, choreoathesis and abnormal behavior	5 casos
Mental retardation, X linked - precocious puberty - obesity	3 casos
Mental retardation, X-linked - acromegaly - hyperactivity	2 casos
Mental retardation, X-linked - Dandy Walker malformation - Basal ganglia disease - Seizures	16 casos
Mental retardation, X-linked - epilepsy - progressive joint contractures - typical face	2 casos
Mental retardation, X-linked - hypogammaglobulinemia - progressive neurological deterioration	3 casos
Mental retardation, X-linked - hypogonadism - ichthyosis - obesity - short stature	4 casos
Mental retardation, X-linked - hypotonia - facial dysmorphism - aggressive behavior	10 casos
Mental retardation, X-linked - macrocephaly - macro-orchidism	12 casos
Mental retardation, X-linked - seizures - psoriasis	4 casos
Mental retardation, X-linked - Spastic paraplegia with iron deposits	1 familia
Mental retardation, X-linked recessive - macrocephaly - ciliary dysfunction	1 familia
Mental retardation, X-linked severe, Gustavson type	7 casos
Mental retardation, X-linked, Abidi type	8 casos
Mental retardation, X-linked, Armfield type	6 casos
Mental retardation, X-linked, Cabezas type	1 familia

Mental retardation, X-linked, Cantagrel type	2 casos
Mental retardation, X-linked, Lubs type	5 casos
Mental retardation, X-linked, Miles-Carpenter type	4 casos
Mental retardation, X-linked, Pai type	1 familia
Mental retardation, X-linked, Reish type	2 casos
Mental retardation, X-linked, Schimke type	4 casos
Mental retardation, X-linked, Seemanova type	4 casos
Mental retardation, X-linked, Shashi type	9 casos
Mental retardation, X-linked, Shrimpton type	3 casos
Mental retardation, X-linked, Siderius type	4 casos
Mental retardation, X-linked, Snyder type	11 casos
Mental retardation, X-linked, South African type	16 casos
Mental retardation, X-linked, Stevenson type	4 casos
Mental retardation, X-linked, Stocco Dos Santos type	4 casos
Mental retardation, X-linked, Stoll type	4 casos
Mental retardation, X-linked, syndromic 7	10 casos
Mental retardation, X-linked, syndromic, due to JARID1C mutation	<10 familias
Mental retardation, X-linked, Vitale type	8 casos
Mental retardation, X-linked, Wilson type	3 casos
Mental retardation, X-linked, with isolated growth hormone deficiency	3 familias
Mental retardation, X-linked, Zorick type	6 casos
MERRF syndrome	0,9
Mesomelic dysplasia - skin dimples	2 casos
Metachromatic leukodystrophy	0,16
Metaphyseal acroschiphodysplasia	4 casos
Metaphyseal anadysplasia	<20 casos
Metaphyseal chondrodysplasia, Jansen type	16 casos
Metaphyseal dysplasia hypertelorism hypospadias	1 caso
Metatropic dwarfism	60 casos
Methimazole embryofetopathy	40 casos
Methylmalonic acidemia - homocystinuria	300 casos
Methylmalonic aciduria - microcephaly - cataract	2 casos
Mevalonicaciduria	30 casos
Michels syndrome	7 casos
Micro syndrome	8 casos
Microbrachycephaly - ptosis - cleft lip	2 casos
Microcephalic osteodysplastic dysplasia, Saul-Wilson type	4 casos
Microcephaly - cardiomyopathy	3 casos
Microcephaly - cleft palate	3 casos
Microcephaly - glomerulonephritis - marfanoid habitus	2 casos
Microcephaly - micropenis - convulsions	4 casos
Microcephaly - seizures - mental retardation - heart disease	2 casos
Microcephaly syndactyly brachymesophalangy	1 caso
Microcoria - congenital nephrosis	22 casos
Microcytic anemia - liver iron overload - low ferritinemia	3 casos
Microdontia - type I microtia - deafness	9 casos

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Microgastria - limb reduction defect	16 casos
Microlissencephaly - micromelia	2 casos
Microphthalmia - brain atrophy	3 casos
Microscopic polyangiitis	7,5
Microtia	15
Midas syndrome	<50 casos
Midline cleft of lower lip	70 casos
Mitochondrial diseases of nuclear origin	9
Mitochondrial encephalomyopathy - aminoacidopathy	2 casos
Mitral regurgitation - deafness - skeletal anomalies	3 casos
Mixed dystonias	3 familias
Moebius syndrome	300 casos
Mohr-Tranebjaerg syndrome	46 casos
Molarization of anterior teeth deafness	1 caso
Molybdenum cofactor deficiency	>100 casos
Monosomy 22q11	20
Monosomy 22q13	>200 casos
Monosomy 5p	4,6
Monosomy 9q22.3	2 casos
Moore-Federman syndrome	6 casos
Mosaic variegated aneuploidy syndrome	29 casos
Mowat-Wilson syndrome	<100 casos
Moya-Moya disease	3,16
Mucopolipidosis type 2	0,15**
Mucopolipidosis type 4	>100 casos
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	<40casos
Mucosulfatidosis	50 casos
Muir-Torre syndrome	205 casos
Mullerian derivatives - lymphangiectasia - polydactyly	3 casos
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 casos
Multiple joint dislocations metaphyseal dysplasia	1 caso
Multiple pterygium syndrome, autosomal dominant	4 casos
Multiple pterygium syndrome, lethal form	200 casos
Multiple sclerosis - ichthyosis - factor VIII deficiency	2 casos
Multiple system atrophy	4,6
MURCS association	11,25
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus	10 casos
Muscular dystrophy congenital, merosin negative	0,3
Muscular dystrophy congenital, merosin-positive	0,15

Muscular dystrophy congenital, with integrin deficiency	0,03
Muscular dystrophy Fukuyama type	0,54
Muscular dystrophy limb-girdle	0,8
Muscular dystrophy, Duchenne and Becker types	5
Muscular dystrophy, limb girdle, autosomal recessive, type 2G	14 casos
Muscular dystrophy, limb girdle, autosomal recessive, type 2I	>40 familias
Muscular dystrophy, limb-girdle, autosomal dominant, type 1A	1 familia
Muscular dystrophy, limb-girdle, autosomal dominant, type 1D	5 familias
Muscular dystrophy, limb-girdle, autosomal dominant, type 1E	5 familias
Muscular dystrophy, limb-girdle, autosomal dominant, type 1F	1 familia
Muscular dystrophy, limb-girdle, autosomal dominant, type 1G	1 familia
Myasthenia gravis	8,5
Myelodysplastic syndromes	25
Myelofibrosis with myeloid metaplasia	10
Myeloma, multiple	14,25
Myhre syndrome	15 casos
Myoclonus - cerebellar ataxia - deafness	4 casos
Myoclonus hereditary - progressive distal muscular atrophy	<10 casos
Myoneurogastrointestinal encephalopathy syndrome	70 casos
Myopathy - lactic acidosis - sideroblastic anemia	7 casos
Myopathy due to casequestrin and SERCA1 protein overload	4 casos
Myopathy, X-linked, with excessive autophagy	15 familias
N syndrome	3 casos
N-acetyl-alpha-D-galactosaminidase deficiency	12 casos
Naegeli-Franceschetti-Jadassohn syndrome	0,035
Nail patella-like renal disease	3 casos
Nail-patella syndrome	2
Nance-Horan syndrome	50 familias
Nanism due to growth hormone qualitative anomaly	3 casos
Nanism due to growth hormone resistance	0,2
NARP syndrome	8,5
Nasopalpebral lipoma - coloboma - telecanthus	<30 casos
Nasopharyngeal teratoma - Dandy Walker - diaphragmatic hernia	1 caso
Necrotizing encephalopathy, acute, autosomal dominant	11 casos
Nemaline myopathy	1
Neonatal death - immune deficiency	5 casos
Neonatal hemochromatosis	100 casos
Nephroblastoma	10,1
Nephronophthisis, autosomal recessive	1,05
Nephronophthisis familial - adult spastic quadriplegia	2 casos

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

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

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Osteopetrosis lethal	2 casos
Osteopetrosis, intermediate form	50 casos
Osteopetrosis, malignant	0,75**
Osteopoikilosis - short stature - intellectual deficit	4 casos
Osteoporosis oculo-cutaneous hypopigmentation syndrome	3 casos
Osteoporosis pseudoglioma syndrome	0,05
Osteosarcoma	5
Osteosclerosis - ichthyosis - premature ovarian failure	3 casos
Otodental syndrome	9 familias
Otopalatodigital syndrome	30 casos
Overhydrated hereditary stomatocytosis	> 20 familias
P2Y12 deficiency	5 casos
Pachydermoperiostosis	204 casos
Pachyonychia congenita	230 casos
Pacman dysplasia	<10 casos
Paget disease, juvenile type	50 casos
Pallister-Hall syndrome	100 casos
Palmoplantar keratoderma - amyotrophy	4 casos
Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma	5 casos
Palmoplantar porokeratosis of Mantoux	>10 casos
Pancreas agenesis	8 casos
Pancreatic and cerebellar agenesis	4 casos
Pancreatic hypoplasia - diabetes - heart disease	<10 casos
Pancreatic lipomatosis - duodenal stenosis	1 caso
Pancreatitis, hereditary	0,125
Pancreatoblastoma	60 casos
Papillon-Lefevre syndrome	0,25
Papulosis, malignant atrophic	>200 casos
Paraplegia - brachydactyly - cone-shaped epiphysis	5 casos
Paraplegia - mental retardation - hyperkeratosis	4 casos
Paraplégie spastique - glaucoma - déficit intellectuel	2 familias
PARC syndrome	2 casos
Parietal foramina	5
Parkinson disease, genetic types	15
Parkinsonism, young adult onset	37,5
Paroxysmal nocturnal hemoglobinuria	0,55
Parsonage-Turner syndrome	3,3
Patella hypoplasia skeletal malformations	1 caso
Pearson syndrome	60 casos
Pelizaeus-Merzbacher disease	0,25
PELVIS syndrome	11 casos
Pemphigus paraneoplastic	>60 casos
Pemphigus superficial	1,2
Pemphigus vulgaris	3,8
Pendred syndrome	5,5
Perinatal-lethal Gaucher disease	0,01
Peritoneal leiomyomatosis, disseminated	100 casos
Perlman syndrome	<20 casos

Persistent hyperinsulinemic hypoglycemia of infancy	2
Persistent Mullerian duct syndrome	<200 casos
Peters-plus syndrome	50 casos
Peutz-Jeghers syndrome	2,2
Pfeiffer syndrome	0,38
Pfeiffer-Singer-Zschesche syndrome	<10 casos
PHACE syndrome	100 casos
Phenylketonuria	4
Pheochromocytoma and paraganglioma, secreting	10
Phosphoenolpyruvate carboxykinase (PEPCK) deficiency	<10 casos
Phosphoglycerate kinase 1 deficiency	23 casos
Phosphoribosylpyrophosphate synthetase superactivity	<30 familias
Phytosterolemia	40 casos
PIBIDS syndrome	20 casos
Piebaldism	0,25
Pierre Robin syndrome	8,75
Pili torti - onychodysplasia	1 familia
Pilodental dysplasia with refractive errors	2 casos
Pityriasis rubra pilaris	48 casos
Plagiocephaly - mental retardation, X-linked	2 casos
Platelet syndrome, familial	<20 familias
Plummer-Vinson syndrome	25 casos
Podder-tolmie syndrome	1 caso
Poikiloderma of Kindler	100 casos
Poland anomaly	2
Pollitt syndrome	10 casos
Polyarteritis nodosa	3,07
Polyarthritis, rheumatic factor-negative	8
Polyarthritis, rheumatoid factor-positive	4,2
Polycystic kidney disease, autosomal recessive	6,5
Polycystic lipomembranous osteodysplasia - sclerosing leukoencephalopathy	0,15
Polycystic ovaries - urethral sphincter dysfunction	33 casos
Polycythemia vera	25
Polydactyly postaxial	50
Polydactyly preaxial	25
Polymorphic catecholergic ventricular tachycardia	10
Polymyositis	14,8
Polysyndactyly - cardiac malformation	6 casos
Pontocerebellar hypoplasia type 1	6 familias
Pontocerebellar hypoplasia type 2	<30 casos
Porencephaly, familial	10 familias
Posterior column ataxia - retinitis pigmentosa	13 casos
Post-transplant lymphoproliferative disease	26,2
Potocki-Shaffer syndrome	23 casos
Prader-Willi syndrome	10,7
Preauricular pits - renal disease	1 caso
Primary biliary cirrhosis	13,5
Primary ciliary dyskinesia	5

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Primary lateral sclerosis	1,5
Primary sclerosing cholangitis	7
Progeria	0,25**
Progeria - short stature - pigmented nevi	<10 casos
Progressive bulbar paralysis of childhood	<40 casos
Progressive neurodegeneration - joint laxity - cataract	2 casos
Progressive vertebral fusion, non-infectious, syndromic form	10 casos
Prolidase deficiency	50 casos
Propionic acidemia	3,75
Proteus syndrome	100-200
Proximal spinal muscular atrophy	3
Proximal spinal muscular atrophy, type 1	0,26
Proximal spinal muscular atrophy, type 2	2,6
Proximal spinal muscular atrophy, type 3	2,6
Proximal spinal muscular atrophy, type 4	0,32
Pseudoachondroplasia	3
Pseudoarthrosis of clavicle, congenital	>200 casos
Pseudodiastrophic dysplasia	10 casos
Pseudo-Gaucher disease	<10 casos
Pseudohypoadosteronism type 1	70 casos
Pseudo-progeria syndrome	2 casos
Pseudoxanthoma elasticum	2,5
Pseudo-Zellweger syndrome	<10 casos
Psoriatic arthritis, juvenile form	4,2
Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	3 casos
Pterygium colli - intellectual deficit - digital anomalies	2 casos
Pterygium popliteal syndrome, autosomal dominant	0,3
Pterygium syndrome, antecubital	11 casos
Ptosis - strabismus - ectopic pupils	1 familia
Ptosis strabismus diastasis	2 casos
Pulmonary alveolar proteinosis	0,1
Pulmonary arterial hypertension	1,5
Pulmonary fibrosis, idiopathic	27
Pulmonary haemosiderosis, primary	250 casos
Pulmonary lymphangiectasia, congenital	>100 casos
Pulmonary valve stenosis, congenital	7,2
Pure autonomic failure	0,3
Pyogenic arthritis - pyoderma gangrenosum - acne	34 casos
Pyruvate kinase deficiency	0,4
Qazi-Markouizos syndrome	3 casos
Radiation proctitis	35
Radio-ulnar synostosis - amegakaryocytic thrombocytopenia	<20 casos
Rambaud-Galian syndrome	3 casos
RAPADILINO syndrome	<20 casos
Rapid-onset dystonia-parkinsonism	3 familias
Rapp-Hodgkin syndrome	72 casos
Rasmussen subacute encephalitis	>100 casos

Recurrent infections - short stature - hypopigmentation - coarse face	4 casos
Refsum disease	0,1
Refsum disease, infantile form	0,005
Regional osteodysplasia	139 casos
Relapsing polychondritis	3,5
Renal adysplasia	26
Renal dysplasia, multicystic	3 casos
Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts	10 casos
Rendu-Osler-Weber disease	3,5
Renpenning syndrome	10 familias
Restrictive cardiomyopathy, idiopathic or familial	2,5
Retinal arteries, tortuosity of	100 casos
Retinal degeneration - nanophthalmos - glaucoma	7 casos
Retinitis pigmentosa	27,5
Retinitis pigmentosa - deafness - hypogonadism	2 familias
Retinoblastoma	5,4
Retino-hepato-endocrinologic syndrome	7 casos
Retinopathy pigmentary - mental retardation	<15 casos
Retinoschisis, X-linked	4,5
Rett syndrome	8,2
Rheumatic fever	5
Rheumatoid purpura	8,5
Rhombencephalosynapsis	50 casos
Richieri costa colletto otto syndrome	1 caso
Rickettsialpox	>800 casos
Rieger syndrome	0,5
Rigid mask like face deafness polydactyly	1 caso
Ring chromosome 10	<20 casos
Ring chromosome 14	50 casos
Ring chromosome 17	14 casos
Ring chromosome 20	>50 casos
Roberts syndrome	100 casos
Robinow like syndrome	2 casos
Robinow syndrome, dominant form	100 casos
Robinow syndrome, recessive form	70 casos
Rolled and spiral hairs - palmoplantar keratoderma	4 casos
Rothmund-Thomson syndrome	300 casos
Rubella panencephalitis	>20 casos
Rubinstein-Taybi like, syndrome	8 casos
Rubinstein-Taybi syndrome	1
Rudiger syndrome	2 casos
Sacral hemangiomas - multiple congenital abnormalities	5 casos
Saethre-Chotzen syndrome	3
Sakati-Nyhan syndrome	<5 casos
Sandhoff disease	0,75
Sarcoidosis	15
Sarcosinemia	2
Say-Barber-Miller syndrome	2 casos

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Scalp defects - postaxial polydactyly	2 casos
Scalp-ear-nipple syndrome	30 casos
SCARF syndrome	2 casos
Schinzel syndrome	<10 familias
Schinzel-Giedion midface retraction syndrome	34 casos
Schizencephaly	1,54
Schizophrenia - mental retardation - deafness - retinitis	1 familia
Schnitzler syndrome	50 casos
Schopf-Schulz-Passarge syndrome	19 casos
Schwartz-Jampel syndrome	100 casos
Scleroderma	42
Sclerosing bone dysplasia - mental retardation	1 caso
Sclerosing bone dysplasia mental retardation	1 caso
Sebastian syndrome	<10 familias
Seckel syndrome	100 casos
Segmental odontomaxillary dysplasia	32 casos
Seizures - intellectual deficit due to hydroxylysineuria	3 casos
Senior-Loken syndrome	0,1
Sensorineural hearing loss - early greying - essential tremor	3 casos
Serpentine fibula - polycystic kidneys	6 casos
Severe achondroplasia - developmental delay - acanthosis nigricans	4 casos
Severe combined immunodeficiency due to adenosine deaminase deficiency	0,22
Severe combined immunodeficiency T- B-	0,35
Severe combined immunodeficiency T- B+, X-linked	1,5
Sezary's syndrome	0,18
Short stature - mental retardation - eye anomalies - cleft lip palat	3 casos
Short stature - pituitary and cerebellar defects - small sella turcica	1 familia
Short stature - webbed neck - heart disease	4 casos
Short stature, Brussels type	2 casos
SHORT syndrome	30 casos
Shprintzen-Goldberg syndrome	<50 casos
Shwachman-Diamond syndrome	200 casos
Sialidosis type 1	0,02**
Sialidosis type 2	0,02**
Sickle cell anaemia	11
Siegler-Brewer-Carey syndrome	2 casos
Silent sinus syndrome	98 casos
Sillence syndrome	5 casos
Silver-Russell dwarfism	400 casos
Simpson-Golabi-Behmel syndrome	>100 casos
Simpson-Golabi-Behmel syndrome, type 2	4 casos
Singleton-Merten dysplasia	<10 casos
Sirenomelia	1**
Sjögren-Larsson syndrome	0,4
Skeletal dysplasia - intellectual deficit	1-2 familias

Small vessel disease of the brain, not NOTCH3-related	2 casos
Smith-Lemli-Opitz syndrome	6,5
Smith-Magenis syndrome	4
Soft tissue sarcomas	13
Sotos syndrome	7**
Sparse hair - short stature - skin anomalies	4 casos
Spastic paraplegia - nephritis - deafness	4 casos
Spastic paraplegia, familial	5
Spastic quadriplegia - retinitis pigmentosa - mental retardation	2 casos
Spasticity - mental retardation - epilepsy, X-linked	6 casos
Spherocytosis hereditary	20
Spinal muscular atrophy - Dandy-Walker complex - cataracts	2 casos
Spinocerebellar ataxia X-linked, type 3	5 casos
Spinocerebellar ataxia, infantile onset	21 casos
Split hand - split foot	1,1
Split hand - split foot - deafness	22 casos
Split hand - urinary anomalies - spina bifida	3 casos
Spondylocarpotarsal synostosis	>20 casos
Spondylo-costal dysostosis - Dandy Walker	1 caso
Spondyloenchondrodysplasia	36 casos
Spondylometaphyseal dysplasia	0,34
Spondylometaphyseal dysplasia - combined immunodeficiency	4 casos
Spondylometaphyseal dysplasia - cone-rod dystrophy	8 casos
Spontaneous periodic hypothermia	<30 casos
Squamous cell carcinoma of head and neck	46
Stargardt disease	11,25
Steinert myotonic dystrophy	4,5
Stern-Lubinsky-Durrie syndrome	7 casos
Sternal cleft	<2
Steroid dehydrogenase deficiency - dental anomalies	1 familia
Stickler syndrome	13,5
Stimmler syndrome	2 casos
Stoll-Alembik-Finck syndrome	1 caso
Subcorneal pustular dermatosis	200 casos
Succinic acidemia	50 casos
Succinyl-CoA acetoacetate transferase deficiency	10 casos
Sulfite oxidase deficiency	50 casos
Summitt syndrome	3 casos
Supranuclear palsy, progressive	5
Susac syndrome	<100 casos
Symmetrical thalamic calcifications	29 casos
Sympathetic ophthalmia	0,6
Symphalangism - short stature - accessory testis	1 caso
Symphalangism distal	<5 familias
Symphalangism short stature accessory testis	1 caso
Symphalangism with multiple anomalies of hands and feet	6 casos

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Synostoses, multiple - brachydactyly	20 familias
Synspondylism	24 casos
Syringomyelia	8,4
Systemic mastocytosis	3,3
Systemic vasculitis	6,3
Takayasu arteritis	0,45
Talo-patello-scaphoid osteolysis	2 casos
Tangier disease	>70 casos
Taurodontia - absent teeth - sparse hair	<15 casos
Tay-Sachs disease	0,3**
Terminal osseous dysplasia - pigmentary defects	18 casos
Tetraamelia - pulmonary hypoplasia	5 familias
Tetralogy of Fallot	45
Thalidomide embryopathy	5000 casos
Thanatophoric dwarfism	3,5**
Thiamine-responsive megaloblastic anemia syndrome	30 familias
Thomsen and Becker disease	5
Thrombocythemia, essential	27,5
Thrombocytopenic purpura, autoimmune	10
Thumb absent - short stature - immune deficiency	3 casos
Thumb stiff - brachydactyly - mental retardation	6 casos
Thymic-renal-anal-lung dysplasia	3 casos
Thyrocerbrorenal syndrome	2 casos
Thyroid carcinoma, anaplastic	0,13
Thyroid carcinoma, medullary	7
Thyrotoxic periodic paralysis	2
Tibial muscular dystrophy	6
Tietz syndrome	1 familia
Toriello-Carey syndrome	60 casos
Toriello-Lacassie-Droste syndrome	10 casos
Torticollis - keloids - cryptorchidism - renal dysplasia	7 casos
Townes-Brocks syndrome	0,42
Tracheal agenesis	1**
Tracheobronchomegaly	<40 casos
Tracheo-bronchomegaly	<100 casos
Transmissible spongiform encephalopathies	0,3
Treacher-Collins syndrome	6
Treft-Sanborn-Carey syndrome	23 casos
Trichinellosis	6500 casos
Trichodental syndrome	<5 familias
Trichodontoosseous syndrome	>30 casos
Trichodysplasia - amelogensis imperfecta	1 familia
Tricho-megaly - retina pigmentary degeneration - dwarfism	11 casos
Trichooculodermovertbral syndrome	1 caso
Trichoodontoonychial dysplasia	4 casos
Trichoonychohypohidrotic dysplasia	1 caso
Tricho-retino-dento-digital syndrome	9 casos
Tricho-rhino-phalangeal syndrome type 1	>100 casos

Trichorhinophalangeal syndrome, type 1 and 3	>100 casos
Tricuspid atresia	5
Trigonocephaly - bifid nose - acral anomalies	2 casos
Trigonocephaly - broad thumbs	2 casos
Trigonocephaly - ptosis coloboma	8 casos
Trigonocephaly - short stature - developmental delay	3 casos
Triose phosphate-isomerase deficiency	30 casos
Triphalangeal thumbs - brachyectrodactyly	4 familias
Triple A syndrome	100 casos
Triple H syndrome	50 casos
Triplo-X syndrome	42,5
Trisomy 13	13**
Trisomy 18	9**
Tritanopia	4,8
True hermaphroditism	>500 casos
Tuberculosis	20
Tuberous sclerosis	8,8
Tubular renal disease - cardiomyopathy	2 casos
Tufted angioma	>200 casos
Turner syndrome	20
Tyrosinemia type 1	0,05
Tyrosinemia type 2	<100 casos
Tyrosinemia type 3	2 casos
Uhl anomaly	84 casos
Ulbright-Hodes syndrome	3 casos
Ullrich, congenital muscular dystrophy	<100 casos
Ulnar / fibula ray defect - brachydactyly	1 familia
Umbilical cord ulceration - intestinal atresia	15 casos
Unverricht-Lundborg disease	0,2
Upington disease	1 familia
Usher syndrome	3,5
VACTERL with hydrocephalus	<10 familias
Van den Bosch syndrome	1 familia
Van Der Woude syndrome	2
VATER association	23
Vernal keratoconjunctivitis	10
Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay	2 casos
Von Hippel-Lindau disease	0,2
Vulvovaginal gingival syndrome	127 casos
W syndrome	6 casos
Waardenburg syndrome	2,4
Waardenburg syndrome type 1	3,75
Waardenburg-Shah syndrome	50 casos
Waldenström macroglobulinemia	2,6
Walker-Warburg syndrome	1,65**
Weaver syndrome	30 casos
Weaver-Williams syndrome	2 casos
Wegener granulomatosis	6,6
Weill-Marchesani syndrome	128 casos

\*\* Prevalencia al nacer

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

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

\*\* Prevalencia al nacer

## Enfermedades por orden decreciente de prevalencia

### *Enfermedades con datos de prevalencia disponibles*

Nombre de enfermedades	Prevalencia estima (/100 000)
Polydactyly postaxial	50
Gelineau disease	49
Guillain-Barré syndrome	47,5
Melanoma, familial	46,8
Squamous cell carcinoma of head and neck	46
Autism	45
Tetralogy of Fallot	45
Deafness, autosomal recessive, nonsyndromic, sensorineural, type DFNB	44
Arrhythmogenic right ventricular dysplasia	43,5
Meniere's disease	42,5
Triplo-X syndrome	42,5
Chromosome Y deletion	42
Scleroderma	42
Juvenile arthritis, idiopathic	41,8
Familial venous malformations	40
Fetal cytomegalovirus syndrome	40
Achalasia, primary	37,5
Elliptocytosis, hereditary	37,5
Parkinsonism, young adult onset	37,5
Follicular lymphoma	36
Non-Hodgkin malignant lymphoma	36
Osteochondritis dissecans	35
Radiation proctitis	35
Adactyly unilateral	34
Cryptosporidiosis	34
Malignant hyperthermia	33
Charcot-Marie-Tooth disease (generic term)	32,5
Great vessels transposition (TGV)	32,5
B-cell chronic lymphocytic leukemia	32
Acute Respiratory Distress Syndrome, Adult	30
Arthrogyposis multiplex congenita	30
Marfan syndrome	30
Hypothyroidism, congenital	29
Retinitis pigmentosa	27,5
Thrombocythemia, essential	27,5
Pulmonary fibrosis, idiopathic	27
Post-transplant lymphoproliferative disease	26,2
Renal adysplasia	26
Long QT syndrome, familial	25
Myelodysplastic syndromes	25
Neurofibromatosis type 1	25

Oesophageal atresia	25
Polycythemia vera	25
Polydactyly preaxial	25
Anorectal malformation	24
Legg-Calve-Perthes disease	23
VATER association	23
Oligoarticular chronic arthritis	20,5
Dermatitis herpetiformis	20,2
Alpha-1 antitrypsin deficiency	20
Atresia of small intestine	20
Atrioventricular canal, partial	20
Gastric cancer	20
Hirschsprung disease	20
Monosomy 22q11	20
Spherocytosis hereditary	20
Tuberculosis	20
Turner syndrome	20
Corpus callosum agenesis - neuronopathy	19
Nephrotic syndrome, steroid-sensitive	18
Cardiomyopathy, familial dilated	17,5
Boutonneuse fever	17
Breast cancer, familial	17
Ichthyosis, X-linked	16,6
MELAS syndrome	16
Leucinosi	15,6
Acyl-CoA dehydrogenase, medium chain, deficiency	15
Diaphragmatic hernia, congenital	15
Lennox-Gastaut syndrome	15
Microtia	15
Parkinson disease, genetic types	15
Sarcoidosis	15
Dermatomyositis	14,8
Polymyositis	14,8
Fragile X syndrome	14,25
Myeloma, multiple	14,25
Anophthalmia/microphthalmia, isolated	14
Cystinuria	14
Primary biliary cirrhosis	13,5
Stickler syndrome	13,5
Williams syndrome	13,3
Androgen insensitivity syndrome	13
Bronchopulmonary dysplasia	13
Soft tissue sarcomas	13
Trisomy 13	13**

\*\* Prevalencia al nacer

Buerger's disease	12,5
Ehlers-Danlos syndrome, type 3	12,5
Willebrand disease	12,5
Cystic fibrosis	12
Gastroschisis	12
Gonadal dysgenesis, XX type	12
Omphalocele	12
Focal dystonia	11,7
MURCS association	11,25
Stargardt disease	11,25
Glioblastoma	11
Hepatic veno-occlusive disease	11
Multiple endocrine neoplasia type 1	11
Sickle cell anaemia	11
Prader-Willi syndrome	10,7
Alopecia totalis	10,5
Collagenous colitis	10,5
Hodgkin lymphoma	10,5
Nephroblastoma	10,1
Adrenal hyperplasia, congenital	10
Bone tumor	10
Cholangiocarcinoma	10
Dermatofibrosarcoma protuberans	10
Distal myopathy, Welander type	10
Duane syndrome	10
Factor II deficiency	10
Idiopathic hypereosinophilic syndrome	10
Lymphoblastic lymphoma	10
Myelofibrosis with myeloid metaplasia	10
Pheochromocytoma and paraganglioma, secreting	10
Polymorphic catecholergic ventricular tachycardia	10
Thrombocytopenic purpura, autoimmune	10
Vernal keratoconjunctivitis	10
Neuroblastoma	10
Mayer-Rokitansky-Küster-Hauser syndrome	9
Mitochondrial diseases of nuclear origin	9
Neuropathy hereditary with liability to pressure palsies	9
Trisomy 18	9**
Giant cell arteritis	8,9
Lymphoedema, congenital	8,8
Tuberous sclerosis	8,8
Pierre Robin syndrome	8,75
Duodenal atresia	8,55
Myasthenia gravis	8,5
NARP syndrome	8,5
Rheumatoid purpura	8,5
Syringomyelia	8,4
Cutaneous lymphoma	8,3
Rett syndrome	8,2
Acute promyelocytic leukemia	8

Deafness, autosomal dominant, nonsyndromic, sensorineural, type DFNA	8
Esophageal carcinoma	8
Polyarthritis, rheumatic factor-negative	8
Cataract, total, congenital	7,9
Hyperlipidemia type 3	7,8
Hemophilia	7,7
Acute lymphoblastic leukemia	7,5
Immunodeficiency, common variable	7,5
Microscopic polyangiitis	7,5
Beckwith-Wiedemann syndrome	7,3
Pulmonary valve stenosis, congenital	7,2
Oculocutaneous albinism	7,15
Acute non lymphoblastic leukemia	7
Cerebellar ataxia, autosomal recessive	7
Cystathioninuria	7
Facio-scapulo-humeral muscular dystrophy	7
Holoprosencephaly	7
Primary sclerosing cholangitis	7
Thyroid carcinoma, medullary	7
Fryns syndrome	7**
Sotos syndrome	7**
Iminoglycinuria	6,68
Cat-scratch disease	6,6
Galactosemia	6,6
Wegener granulomatosis	6,6
Angelman syndrome	6,5
Carcinoma of the gallbladder	6,5
Hemiplegic migraine, familial	6,5
Leber hereditary optic neuropathy	6,5
Osteogenesis imperfecta	6,5
Polycystic kidney disease, autosomal recessive	6,5
Smith-Lemli-Opitz syndrome	6,5
Ectopia lentis isolated	6,4
Juvenile idiopathic arthritis, systemic-onset	6,3
Systemic vasculitis	6,3
Huntington disease	6,2
Amyotrophic lateral sclerosis	6
Cerebral arteriovenous fistula	6
Digitotalar dysmorphism	6
Leukemia, chronic myeloid	6
Optic atrophy	6
Tibial muscular dystrophy	6
Treacher-Collins syndrome	6
Wilson disease	5,84
Arthritis-related enthesitis	5,7
Biliary atresia	5,6
Pendred syndrome	5,5
Retinoblastoma	5,4
Alzheimer disease, familial	5,3
Zollinger-Ellison syndrome	5,3

\*\* Prevalencia al nacer

Cornelia de Lange syndrome	5,25
Familial adenomatous polyposis, autosomal dominant	5,25
Acromegaly	5
Epiphyseal dysplasia multiple	5
Fructose intolerance	5
Lung cancer, small cell	5
Muscular dystrophy, Duchenne and Becker types	5
Osteosarcoma	5
Parietal foramina	5
Primary ciliary dyskinesia	5
Rheumatic fever	5
Spastic paraplegia, familial	5
Supranuclear palsy, progressive	5
Thomsen and Becker disease	5
Tricuspid atresia	5
Hydroletharus	5**
Tritanopia	4,8
Acrocephalosyndactyly	4,6
Monosomy 5p	4,6
Multiple system atrophy	4,6
Achondroplasia	4,5
Congenital lobar emphysema	4,5
Retinoschisis, X-linked	4,5
Steinert myotonic dystrophy	4,5
Best disease	4,4
Cataract anterior polar	4,4
Chronic inflammatory demyelinating polyneuropathy	4,4
Polyarthritis, rheumatoid factor-positive	4,2
Psoriatic arthritis, juvenile form	4,2
Hemimelia	4,15
Adrenal hypoplasia congenital, X-linked	4
Autoimmune polyendocrinopathy, type 1	4
Ceroid lipofuscinosis, neuronal	4
Corticobasal degeneration	4
Exostoses, multiple	4
Hartnup syndrome	4
Histidinemia	4
Idiopathic hypersomnia	4
Phenylketonuria	4
Smith-Magenis syndrome	4
Amniotic bands	4**
Meckel syndrome	4**
Mantle cell lymphoma	3,9
Acute interstitial pneumonia	3,8
Anisakiasis	3,8
Calpainopathy	3,8
Pemphigus vulgaris	3,8
Propionic acidemia	3,75
Waardenburg syndrome type 1	3,75
West syndrome	3,7**

Adrenoleukodystrophy, X-linked	3,5
Diastrophic dwarfism	3,5
Ehlers-Danlos syndrome, classic type	3,5
Goldenhar syndrome	3,5
MASA syndrome	3,5
Relapsing polychondritis	3,5
Rendu-Osler-Weber disease	3,5
Usher syndrome	3,5
Thanatophoric dwarfism	3,5**
Graft versus host disease	3,4
Choroidal dystrophy, central areolar	3,33
Hypochondroplasia	3,3
Multiple endocrine neoplasia, type 2	3,3
Parsonage-Turner syndrome	3,3
Systemic mastocytosis	3,3
Anencephaly	3,2**
Moya-Moya disease	3,16
Acatlasemia	3,1
Polyarteritis nodosa	3,07
Bacterial toxic-shock syndrome	3
Frontotemporal dementia	3
Nodular regenerative hyperplasia of the liver	3
Opitz BBB/G syndrome	3
Proximal spinal muscular atrophy	3
Pseudoachondroplasia	3
Saethre-Chatzen syndrome	3
Kennedy disease	2,8
Leigh disease	2,75**
Proximal spinal muscular atrophy, type 2	2,6
Proximal spinal muscular atrophy, type 3	2,6
Waldenström macroglobulinemia	2,6
Achromatopsia	2,5
Amaurosis congenita of Leber	2,5
Behcet disease	2,5
BOR syndrome	2,5
Bullous pemphigoid	2,5
Cone rod dystrophy	2,5
Diffuse palmoplantar keratoderma, Norrbotten dominant type	2,5
Epidermolysis bullosa, epidermolytic	2,5
Friedreich ataxia	2,5
Fructose-1,6-bisphosphatase deficiency	2,5
Heterotaxia	2,5
Kartagener syndrome	2,5
Pseudoxanthoma elasticum	2,5
Restrictive cardiomyopathy, idiopathic or familial	2,5
Niemann-Pick disease	2,5**
Meconium aspiration syndrome	2,44
Waardenburg syndrome	2,4
3-methylcrotonylglycinuria	2,25
Ondine syndrome	2,25

\*\* Prevalencia al nacer

Peutz-Jeghers syndrome	2,2
Cerebellar ataxia, autosomal dominant	2,15
GRACILE syndrome	2,12**
Alport syndrome	2
Choroideremia	2
Coats disease	2
Crouzon disease	2
Giant pigmented hairy nevus	2
Klippel-Feil syndrome	2
Langerhans cell histiocytosis	2
Nail-patella syndrome	2
Ocular albinism X-linked, recessive	2
Persistent hyperinsulinemic hypoglycemia of infancy	2
Poland anomaly	2
Sarcosinemia	2
Thyrotoxic periodic paralysis	2
Van Der Woude syndrome	2
Sternal cleft	<2
Wolf-Hirschhorn syndrome	2**
Lateral body wall complex	2**
Gamma-sarcoglycanopathy	1,96
Gastrointestinal stromal tumor	1,8
Amoebiasis due to free-living amoebae	1,75
Aniridia	1,75
Fabry disease	1,75
2,8 dihydroxyadenine urolithiasis	1,7
Kaposi's sarcoma	1,7
Walker-Warburg syndrome	1,65**
Charcot-Marie-Tooth disease, X-linked	1,6
Schizencephaly	1,54
Antisynthetase syndrome	1,5
Budd-Chiari syndrome	1,5
Darier disease	1,5
Emery-Dreifuss muscular dystrophy	1,5
Femur-fibula-ulna complex	1,5
Laryngotracheoesophageal cleft	1,5
Multifocal motor neuropathy with conduction block	1,5
Primary lateral sclerosis	1,5
Pulmonary arterial hypertension	1,5
Severe combined immunodeficiency T- B+, X-linked	1,5
CDG syndrome	1,5**
Alagille syndrome	1,4
Cat-eye syndrome	1,35
Netherton disease	1,35
Cryoglobulinemia	1,33*
Mucopolysaccharidosis type 1	1,3
Apert syndrome	1,25
Maternal hyperphenylalaninemia	1,25
Adult Onset Still's disease	1,23
Orofaciodigital syndrome, type 1	1,2

Pemphigus superficial	1,2
Kabuki syndrome	1,16
Glycogen storage disease, type 2	1,1
Mucopolysaccharidosis type 3	1,1
Split hand - split foot	1,1
Zellweger syndrome	1,1
Nephronophthisis, autosomal recessive	1,05
Cutis verticis gyrata - mental deficiency	1,02
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency	1
Acanthamoeba keratitis	1
Adrenocortical carcinoma	1
Albers-Schonberg disease	1
Angioneurotic edema	1
Ataxia telangiectasia	1
Blue cone monochromatism	1
Chondrodysplasia punctata, rhizomelic type	1
Chronic hiccup	1
Churg-Strauss syndrome	1
Clouston syndrome	1
Coloboma, ocular	1
Dyserythropoietic anemia, congenital	1
Ehlers-Danlos syndrome, type 4	1
Fanconi anaemia	1
Gaucher disease	1
Gorlin syndrome	1
Harding ataxia	1
Holt-Oram syndrome	1
Hypokalemic periodic paralysis	1
Isovaleric acidemia	1
Lambert-Eaton myasthenic syndrome	1
Macrophagic myofasciitis	1
Nemaline myopathy	1
Neuroendocrine tumor	1
Oculopharyngeal muscular dystrophy	1
Rubinstein-Taybi syndrome	1
Congenital rubella syndrome	1**
Sirenomelia	1**
Tracheal agenesis	1**
Acalvaria	<1**
Gaucher disease, type 1	0,94
Lewis-Sumner syndrome	0,9
MERRF syndrome	0,9
Joubert syndrome	0,85
Niemann-Pick disease, type C	0,85
Bardet-Biedl syndrome	0,8
Criss-cross heart	0,8
Muscular dystrophy limb-girdle	0,8
Congenital myasthenic syndromes	0,75
Cutaneous mastocytosis	0,75
Ebstein anomaly	0,75

\*\* Prevalencia al nacer

Hyperkalemic periodic paralysis	0,75
Sandhoff disease	0,75
Niemann-Pick disease, type B	0,75**
Osteopetrosis, malignant	0,75**
Krabbe disease	0,75**
Albright hereditary osteodystrophy	0,72
Carbamoylphosphate synthetase deficiency	0,7
Menkes syndrome	0,7
46,XY disorders of sex development due to 17-beta-hydroxysteroid dehydrogenase deficiency	0,68
Kallmann syndrome	0,66
Hepatitis, chronic autoimmune	0,65
Goodpasture syndrome	0,64
Deafness, X-linked, non syndromic, sensorineural, type DFN	0,6
Glycogen storage disease, type 4	0,6
Hyperlipoproteinemia type 1	0,6
Mucopolysaccharidosis type 2	0,6
Sympathetic ophthalmia	0,6
Alpha-sarcoglycanopathy	0,57
Beta-sarcoglycanopathy	0,57
Delta-sarcoglycanopathy	0,57
Wolfram syndrome	0,57
Cantrell pentalogy	0,55**
Coffin-Lowry syndrome	0,55
Paroxysmal nocturnal hemoglobinuria	0,55
Muscular dystrophy Fukuyama type	0,54
Cutaneous neuroendocrine carcinoma	0,5
Cystinosis	0,5
Diabetes insipidus, nephrogenic	0,5
Neurofibromatosis type 2	0,5
Rieger syndrome	0,5
Xeroderma pigmentosum	0,5
X-linked dominant chondrodysplasia punctata	0,5
Inclusion body myositis, IBM	0,49
Agammaglobulinemia, X-linked	0,45
Cowden syndrome	0,45
Takayasu arteritis	0,45
Werner syndrome	0,45
Townes-Brocks syndrome	0,42
Bullous ichthyosiform - erythroderma congenita	0,4
Early onset torsion dystonia	0,4
Glutaryl-CoA dehydrogenase deficiency	0,4
Homocystinuria due to cystathionine beta-synthase deficiency	0,4
Mucopolysaccharidosis type 4	0,4
Pyruvate kinase deficiency	0,4
Sjögren-Larsson syndrome	0,4
Lesch-Nyhan syndrome	0,38
Pfeiffer syndrome	0,38
Christ-Siemens-Touraine syndrome	0,35

Severe combined immunodeficiency T- B-	0,35
Campomelic dysplasia	0,35
Spondylometaphyseal dysplasia	0,34
Sialidosis type 1	>0,33
Neutropenia severe congenital	0,33**
Blackfan-Diamond disease	0,32
Proximal spinal muscular atrophy, type 4	0,32
Alkaptonuria	0,3
Dopa-responsive dystonia	0,3
Lissencephaly type 1, due to LIS 1 anomalies	0,3
Muscular dystrophy congenital, merosin negative	0,3
Pterygium popliteal syndrome, autosomal dominant	0,3
Pure autonomic failure	0,3
Transmissible spongiform encephalopathies	0,3
Dentatorubral pallidoluysian atrophy	<0,3
Tay-Sachs disease	0,3**
Wolman disease	0,28**
Epidermolysis bullosa, dystrophic	0,27
Proximal spinal muscular atrophy, type 1	0,26
Factor VII deficiency	0,25
Lipodystrophy, Berardinelli type	0,25
Papillon-Lefevre syndrome	0,25
Pelizaeus-Merzbacher disease	0,25
Piebaldism	0,25
Niemann-Pick disease, type A	0,25**
Progeria	0,25**
Leptospirosis	0,24
Severe combined immunodeficiency due to adenosine deaminase deficiency	0,22
Von Hippel-Lindau disease	0,2
Acrodermatitis enteropathica, zinc deficiency type	0,2
Diabetes mellitus, neonatal	0,2
Granulomatous disease, chronic	0,2
Hyperglycinemia, isolated nonketotic	0,2
Hyperoxaluria	0,2
Incontinentia pigmenti	0,2
Jeune syndrome	0,2
Nanism due to growth hormone resistance	0,2
Unverricht-Lundborg disease	0,2
Lowe syndrome	0,19
Sezary's syndrome	0,18
Atypical coarctation of aorta	0,17**
Metachromatic leukodystrophy	0,16
Mucopolysaccharidosis type 6	0,16**
Epilepsy, pyridoxin-dependent	0,15
Fibrinogen deficiency, congenital	0,15
Muscular dystrophy congenital, merosin-positive	0,15
Polycystic lipomembranous osteodysplasia - sclerosing leukoencephalopathy	0,15
Wiskott-Aldrich syndrome	0,15
Mucopolipidosis type 2	0,15**

\*\* Prevalencia al nacer

CHARGE association	0,14
Thyroid carcinoma, anaplastic	0,13
Xanthomatosis cerebrotendinous	0,13
Pancreatitis, hereditary	0,125
Bartter syndrome	0,12
Lissencephaly type 2	0,12
Medullary cystic kidney disease, autosomal dominant	0,11
Alpha-mannosidosis	0,1
Creutzfeldt-Jakob disease	0,1
Diffuse leiomyomatosis with Alport syndrome	0,1
Distal myopathy, Nonaka type	0,1
Dyskeratosis congenita	0,1
Evans syndrome	0,1
Ewing sarcoma	0,1
Factor V deficiency	0,1
Familial cold urticaria	0,1
Hemophilia, acquired	0,1
Lymphangioliomyomatosis	0,1
Pulmonary alveolar proteinosis	0,1
Refsum disease	0,1
Senior-Loken syndrome	0,1
X-linked lymphoproliferative disease	0,1

Leprechaunism	0,1**
Fibrodysplasia ossificans progressiva	0,08
Epidermolysis bullosa, junctional	0,06
Mendelian susceptibility to atypical mycobacteria	0,059
Aceruloplasminemia	0,05
Botulism	0,05
Chordoma	0,05
Craniofacial dysynostosis	0,05
Gaucher disease, type 3	0,05
Osteoporosis pseudoglioma syndrome	0,05
Tyrosinemia type 1	0,05
Hyper-IGM syndrome, autosomal recessive	0,05**
Factor XIII deficiency, congenital	0,04
Naegeli-Franceschetti-Jadassohn syndrome	0,035
Muscular dystrophy congenital, with integrin deficiency	0,03
Alpers syndrome	0,025
Herpes simplex encephalitis	0,021*
Sialidosis type 1	0,02**
Sialidosis type 2	0,02**
Perinatal-lethal Gaucher disease	0,01
Gaucher disease, type 2	0,01
Refsum disease, infantile form	0,005

\*\* Prevalencia al nacer

## Enfermedades por número de casos publicados

*Enfermedades sin datos disponibles de prevalencia. Informe del número de casos publicados.*

Nombre de enfermedades	Número de casos o familias publicados
Trichinellosis	6500 casos
Thalidomide embryopathy	5000 casos
Angio-osteohypertrophic syndrome	1000 casos
Whipple disease	1000 casos
Alveolar echinococcosis	<1000 casos
Esthesioneuroblastoma	<1000 casos
Rickettsialpox	>800 casos
Fetal methyl mercury syndrome	800 casos
Familial dysautonomia	550 casos
Western equine encephalitis	>600 casos
Enchondromatosis	> 600 casos
Adamantinoma	513 casos
True hermaphroditism	>500 casos
Aicardi syndrome	500 casos
CADASIL syndrome	500 casos
Malakoplasia	500 casos
Epidermal nevus syndrome	>400 casos
Castleman disease	400 casos
Silver-Russell dwarfism	400 casos
4-hydroxybutyricaciduria	350 casos
Leukocyte adhesion deficiency	<350 casos
CDG syndrome type Ia	> 300 casos
Alexander disease	300 casos
Alström syndrome	300 casos
Cutis marmorata telangiectatica congenita	300 casos
Gräsbeck-Imerslund disease	300 casos
Melorheostosis	300 casos
Methylmalonic acidemia - homocystinuria	300 casos
Moebius syndrome	300 casos
Norrie disease	300 casos
Rothmund-Thomson syndrome	300 casos
Lipoid proteinosis	>280 casos
Chronic recurrent multifocal osteomyelitis, juvenile	>260 casos
Caroli's disease	<250 casos
Focal dermal hypoplasia	200-300 casos
Lipodystrophy, familial partial, Dunnigan type	200-300 casos
Eosinophilic gastroenteritis	280 casos
Job syndrome	250 casos
Lipodystrophy, partial acquired	250 casos
Pulmonary haemosiderosis, primary	250 casos
6-pyruvoyl-tetrahydropterin synthase deficiency	248 casos
Oculo-dento-osseous dysplasia, autosomal dominant	243 casos

Pachyonychia congenita	230 casos
Kearns-Sayre syndrome	223 casos
Lhermitte-Duclos disease	220 casos
Muir-Torre syndrome	205 casos
Pachydermoperiostosis	204 casos
Aarskog-Scott syndrome	>200 casos
Blue rubber bleb nevus	>200 casos
Erythrokeratoderma variabilis, Mendes da Costa type	>200 casos
Monosomy 22q13	>200 casos
Papulosis, malignant atrophic	>200 casos
Pseudoarthrosis of clavicle, congenital	>200 casos
Tufted angioma	>200 casos
Adenosine monophosphate deaminase deficiency	200 casos
Camurati-Engelmann disease	200 casos
Cockayne syndrome	200 casos
Cogan syndrome	200 casos
Costello syndrome	200 casos
Crigler-Najjar syndrome	200 casos
Eosinophilic fasciitis	200 casos
Glucose-galactose malabsorption	200 casos
Gorham-Stout disease	200 casos
Kimura disease	200 casos
Multiple pterygium syndrome, lethal form	200 casos
Shwachman-Diamond syndrome	200 casos
Subcorneal pustular dermatosis	200 casos
Deletion 18p	<200 casos
Persistent Mullerian duct syndrome	<200 casos
Hyperimmunoglobulinemia D with recurrent fever	180 casos
Erdheim-Chester disease	178 casos
Kasabach-Merritt syndrome	>175 casos
Celiac disease - epilepsy - occipital calcifications	170 casos
Alpha thalassemia-mental retardation, X-linked	168 casos
Carney complex	160 casos
McCune-Albright syndrome	158 casos
Infantile neuroaxonal dystrophy	>150 casos
Cloverleaf skull syndrome	150 casos
Denys-Drash syndrome	150 casos
Dubowitz syndrome	150 casos
Ellis Van Creveld syndrome	150 casos
Fraser syndrome	150 casos
Jacobsen syndrome	150 casos
Xanthinuria	150 casos
Arthrogyposis - renal dysfunction - cholestasis	<150 casos
Inflammatory pseudotumor of the liver	143 casos
Regional osteodysplasia	139 casos

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

Triple A syndrome	100 casos
Cataract congenital, Volkmann type	<100 casos
Hallermann-Streiff-Francois syndrome	<100 casos
Hypertrichosis lanuginosa congenita	<100 casos
Hypocomplementaemic leucocytoclastic vasculitis	<100 casos
Ichthyosis congenita, harlequin type	<100 casos
Mowat-Wilson syndrome	<100 casos
Susac syndrome	<100 casos
Tracheo-bronchomegaly	<100 casos
Tyrosinemia type 2	<100 casos
Ullrich, congenital muscular dystrophy	<100 casos
Silent sinus syndrome	98 casos
Acrofacial dysostosis, Nager type	90 casos
Megacystis microcolon - intestinal hypoperistalsis - hydronephrosis	89 casos
Early infantile epileptic encephalopathy	88 casos
Uhl anomaly	84 casos
Acquired generalized lipodystrophy	80 casos
LCAT deficiency	80 casos
Wells syndrome	80 casos
Rapp-Hodgkin syndrome	72 casos
LEOPARD syndrome	>70 casos
Tangier disease	>70 casos
Bullous systemic lupus erythematosus	70 casos
Cranio-metaphyseal dysplasia	70 casos
Midline cleft of lower lip	70 casos
Myoneurogastrointestinal encephalopathy syndrome	70 casos
Pseudohypoaldosteronism type 1	70 casos
Robinow syndrome, recessive form	70 casos
Diffuse neonatal haemangiomas	<70 casos
Glutathione synthetase deficiency	65 casos
Hyperferritinemia, hereditary, with congenital cataracts	>64 casos
Griselli disease	60 casos
Pemphigus paraneoplastic	>60 casos
Cerebro-costo-mandibular syndrome	60 casos
Dygve-Melchior-Clausen disease	60 casos
Hypertrichosis lanuginosa, acquired	60 casos
Neu laxova syndrome	50-60 casos
Ketoacidosis due to betaketothiolase deficiency	60 casos
Metatropic dwarfism	60 casos
Non-distal trisomy 10p	60 casos
Pancreatoblastoma	60 casos
Pearson syndrome	60 casos
Toriello-Carey syndrome	60 casos
Double uterus - hemivagina - renal agenesis	<60 casos
Ataxia, autosomal recessive, Beauce type	57 casos
Capillary leak syndrome	57 casos
Hennekam syndrome	>50 casos
Osteodysplasty, Melnick-Needles type	>50 casos
Ring chromosome 20	>50 casos
Acromesomelic dysplasia, Maroteaux type	50 casos

Acroosteolysis, autosomal dominant	50 casos
Adenylosuccinate lyase deficiency	50 casos
Ascher syndrome	50 casos
Benign paroxysmal torticollis of infancy	50 casos
Focal myositis	50 casos
Fronto-temporal dementia and Parkinsonism linked to chromosome 17 (FTDP-17)	50 casos
Glucocorticoid deficiency, familial	50 casos
Glucosephosphate isomerase deficiency	50 casos
ICF syndrome	50 casos
Mucosulfatidosis	50 casos
Ocular motor apraxia, Cogan type	50 casos
Osteopetrosis, intermediate form	50 casos
Paget disease, juvenile type	50 casos
Peters-plus syndrome	50 casos
Prolidase deficiency	50 casos
Rhombencephalosynapsis	50 casos
Ring chromosome 14	50 casos
Schnitzler syndrome	50 casos
Succinic acidemia	50 casos
Sulfite oxidase deficiency	50 casos
Triple H syndrome	50 casos
Waardenburg-Shah syndrome	50 casos
XY gonadal agenesis	50 casos
Ehrlchiosis	<50 casos
Aglossia - adactylia	<50 casos
Blepharo-cheilo-dontic syndrome	<50 casos
Branchio-oculo-facial syndrome	<50 casos
Cholesteryl ester storage disease	<50 casos
D00R syndrome	<50 casos
Fibular aplasia - ectrodactyly	<50 casos
Goldmann-Favre syndrome	<50 casos
Granulomatous slack skin	<50 casos
Midas syndrome	<50 casos
Oculo-digito-esophageal-duodenal syndrome (ODED)	<50 casos
Shprintzen-Goldberg syndrome	<50 casos
Cholestasis - lymphoedema, syndrome	20-50 casos
Corticosteroid-sensitive aseptic abscesses	49 casos
Pityriasis rubra pilaris	48 casos
Coxo-podo-patellar syndrome	47 casos
Mohr-Tranebjaerg syndrome	46 casos
Encephalo-cranio-cutaneous lipomatosis	45 casos
KBG syndrome	45 casos
Bowen-Conradi syndrome	44 casos
Ear-patella-short stature syndrome	42 casos
Juvenile hyaline fibromatosis	40-50 casos
Desbuquois syndrome	>40 casos
3M syndrome	40 casos
Babesiosis	40 casos
Carpenter syndrome	40 casos
Chylomicron retention disease	40 casos

Coffin-Siris syndrome	40 casos
Ectodermal dysplasia, hypohidrotic, autosomal dominant	40 casos
Galloway-Mowat syndrome	40 casos
Macrocephaly - cutis Marmorata Telangiectatica Congenita	40 casos
Methimazole embryofetopathy	40 casos
Phytosterolemia	40 casos
WHIM syndrome	40 casos
Mucopolysaccharidosis type 7	<40casos
Acromicric dysplasia	<40 casos
Progressive bulbar paralysis of childhood	<40 casos
Tracheobronchomegaly	<40 casos
Hypotrichosis simplex	38 casos
Mandibuloacral dysplasia	37 casos
Oculocerebrocutaneous syndrome	36 casos
Spondyloenchondrodysplasia	36 casos
Carnitine palmitoyl transferase 1 deficiency	35 casos
Cobb syndrome	35 casos
Hereditary sensory and autonomic neuropathy, type 2	35 casos
Acrocallosal syndrome, Schinzel type	34 casos
Antley-Bixler syndrome	34 casos
Pyogenic arthritis - pyoderma gangrenosum - acne	34 casos
Schinzel-Giedion midface retraction syndrome	34 casos
Björnstadt syndrome	33 casos
Marshall-Smith syndrome	33 casos
Osteopetrosis autosomal dominant, type 1	33 casos
Polycystic ovaries - urethral sphincter dysfunction	33 casos
Double outlet left ventricle	32 casos
Olmsted syndrome	32 casos
Segmental odontomaxillary dysplasia	32 casos
Argininemia	31 casos
Duplication 8q	>30 casos
Trichodontoosseous syndrome	>30 casos
CDG syndrome type Ic	>30 casos
Agnathia - holoprosencephaly - situs inversus	30 casos
Aicardi-Goutieres syndrome	30 casos
Anophthalmia - hypothalamo-pituitary insufficiency	30 casos
Camptodactyly - tall stature - scoliosis - hearing loss	30 casos
Carnitine-acylcarnitine translocase deficiency	30 casos
Carnosinemia	30 casos
Cataract - cardiomyopathy	30 casos
CHILD syndrome	30 casos
Dermopathy restrictive lethal	30 casos
Early myoclonic encephalopathy	30 casos
Elejalde syndrome	30 casos
Geroderma osteodysplastica	30 casos
Glycogen storage disease due to LAMP-2 deficiency	30 casos
Marden-Walker syndrome	30 casos
Marshall's syndrome with periodic fever	30 casos
Mevalonicaciduria	30 casos
Omodysplasia	30 casos

Ophthalmo acromelic syndrome	30 casos
Otopalatodigital syndrome	30 casos
Scalp-ear-nipple syndrome	30 casos
SHORT syndrome	30 casos
Triose phosphate-isomerase deficiency	30 casos
Weaver syndrome	30 casos
Acropectorovertebral dysplasia	<30 casos
Congenital brain dysgenesis due to glutamine synthetase deficiency	<30 casos
Developmental delay due to 2-methylbutyryl-coA dehydrogenase deficiency	<30 casos
Encephalopathy due to hydroxykynureninuria	<30 casos
Fronto-metaphyseal dysplasia	<30 casos
Glycogen storage disease, type 7	<30 casos
Johnson neuroectodermal syndrome	<30 casos
Nasopalpebral lipoma - coloboma - telecanthus	<30 casos
Neurometabolic disorder due to serine deficiency	<30 casos
Obesity due to congenital leptin deficiency	<30 casos
Pontocerebellar hypoplasia type 2	<30 casos
Spontaneous periodic hypothermia	<30 casos
Wrinkly skin syndrome	<30 casos
Ectodermal dysplasia absent dermatoglyphics	<30 casos
Cone rod dystrophy - amelogenesis imperfecta	29 casos
Infant epilepsy with migrant focal crisis	29 casos
Mosaic variegated aneuploidy syndrome	29 casos
Orofaciodigital syndrome, type 6	29 casos
Symmetrical thalamic calcifications	29 casos
Hypertrichosis cubiti - short stature	28 casos
Catel-Manzke syndrome	27 casos
GAPO syndrome	27 casos
Geleophysic dwarfism	27 casos
Insomnia, familial fatal	27 casos
Limb-mammary syndrome	27 casos
3C syndrome	25 casos
Atelosteogenesis II	25 casos
Book syndrome	25 casos
Erythrokeratoderma - ataxia	25 casos
Keratoderma palmoplantar - spastic paralysis	25 casos
Opsismodysplasia	25 casos
Plummer-Vinson syndrome	25 casos
Wiedemann-Rautenstrauch syndrome	25 casos
Hepatic veno-occlusive disease - immunodeficiency	<25 casos
Bartsocas-Papas syndrome	24 casos
Distal myopathy, with early respiratory muscle involvement	24 casos
Synspondylism	24 casos
Congenital bronchobiliary fistula	23 casos
Deletion 2q24	23 casos
Johanson-Blizzard syndrome	23 casos
Phosphoglycerate kinase 1 deficiency	23 casos
Potocki-Shaffer syndrome	23 casos
Treft-Sanborn-Carey syndrome	23 casos

Acro-pectoral syndrome	22 casos
Aortic dilatation- joint hypermobility- arterial tortuosity	22 casos
Microcoria - congenital nephrosis	22 casos
Split hand - split foot - deafness	22 casos
Cranioleptoculosutural dysplasia	21 casos
Dehydratase deficiency	21 casos
Odonto-tricho-ungual-digito-palmar syndrome	21 casos
Spinocerebellar ataxia, infantile onset	21 casos
Craniodiaphyseal dysplasia	>20 casos
Fumaric aciduria	>20 casos
Rubella panencephalitis	>20 casos
Spondylocarpotarsal synostosis	>20 casos
Congenital indifference to pain	> 20 casos
3-methylglutaconic aciduria, type 1	20 casos
Acrorenal syndrome	20 casos
Arrhinia	20 casos
Calvarial doughnut lesions - bone fragility	20 casos
Craniofrontonasal syndrome, Teebi type	20 casos
Distal monosomy 8p	20 casos
Gray platelet syndrome	20 casos
Juvenile temporal arteritis	20 casos
Lacrimo-auriculo-dento-digital syndrome	20 casos
Lipoamide dehydrogenase deficiency	20 casos
PIBIDS syndrome	20 casos
CDG syndrome type Ib	20 casos
Acromegaloïd facial appearance syndrome	<20 casos
Acromegaloïd facies - hypertrichosis	<20 casos
Bronchopneumopathy, chronic, due to TAP deficiency	<20 casos
Carey-Fineman-Ziter syndrome	<20 casos
Epidermolysis bullosa simplex - limb girdle muscular dystrophy	<20 casos
Hemorrhagic disorders due to collagen receptors deficiency	<20 casos
Ichthyosis bullosa of Siemens	<20 casos
IMAGE syndrome	<20 casos
Interstitial granulomatous dermatitis - arthritis	<20 casos
Laryngeal abductor paralysis - mental retardation	<20 casos
Lipodystrophy, familial partial, Köbberling type	<20 casos
Metaphyseal anadysplasia	<20 casos
Orotic aciduria hereditary	<20 casos
Perlman syndrome	<20 casos
Radio-ulnar synostosis - amegakaryocytic thrombocytopenia	<20 casos
RAPADILINO syndrome	<20 casos
Ring chromosome 10	<20 casos
Woolly hair - palmoplantar keratoderma - dilated cardiomyopathy	<20 casos
Ectodermal dysplasia, «pure» hair-nail type	<20 casos
Keratosis palmaris et plantaris - clinodactyly	<20
Craniosynostosis, Boston type	19 casos
Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia	19 casos

Schopf-Schulz-Passarge syndrome	19 casos
Crisponi syndrome	18 casos
Hypertrichotic osteochondrodysplasia	18 casos
Terminal osseous dysplasia - pigmentary defects	18 casos
Aminopterin embryofetopathy	17 casos
Erythroderma lethal, congenital	17 casos
Folate malabsorption, hereditary	17 casos
GTP cyclohydrolase I deficiency	17 casos
Hypokeratosis, palmo-plantar, circumscribed	17 casos
Malonic aciduria	17 casos
Mental retardation X-linked - seizures - short stature - midface hypoplasia	17 casos
Wolcott-Rallison syndrome	17 casos
Acromegaly - cutis verticis gyrata - corneal leukoma	16 casos
Antley-Bixler-like syndrome - ambiguous genitalia - disordered steroidogenesis	16 casos
Glycogen storage, type 0	16 casos
Mental retardation, X-linked - Dandy Walker malformation - Basal ganglia disease - Seizures	16 casos
Mental retardation, X-linked, South African type	16 casos
Metaphyseal chondrodysplasia, Jansen type	16 casos
Microgastria - limb reduction defect	16 casos
Orbital leiomyoma	16 casos
Orofaciodigital syndrome, type 4	16 casos
Ablepharon-macrostomia syndrome	15 casos
Angel-shaped phalango-epiphyseal dysplasia	15 casos
Cranio-ectodermal dysplasia	15 casos
IBIDS syndrome	15 casos
Myhre syndrome	15 casos
Umbilical cord ulceration - intestinal atresia	15 casos
Congenital anosmia, isolated	<15 casos
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	<15 casos
Odontoonychodermal dysplasia	<15 casos
Retinopathy pigmentary - mental retardation	<15 casos
Taurodontia - absent teeth - sparse hair	<15 casos
Wilson-Turner syndrome	>14 casos
Muscular dystrophy, limb girdle, autosomal recessive, type 2G	14 casos
ADULT syndrome	14 casos
Anophthalmia/microphthalmia - esophageal atresia	14 casos
Atkin-Flaitz syndrome	14 casos
Beta-mannosidosis	14 casos
DEND syndrome	14 casos
Dermoodontodysplasia	14 casos
Fingerprints absence - syndactyly milia	14 casos
Oligocone trichromacy	14 casos
Optic atrophy and cataract, autosomal dominant	14 casos
Ring chromosome 17	14 casos
Aromatase deficiency	13 casos
Chondrodysplasia, Blomstrand type	13 casos
Diaphragmatic hernia - exomphalos - corpus callosum agenesis	13 casos

Hypotelorism - cleft palate - hypospadias	13 casos
Posterior column ataxia - retinitis pigmentosa	13 casos
Hemolytic anemia due to adenylate kinase deficiency	12 casos
Acropectororenal field defect	12 casos
Alopecia - epilepsy - pyorrhea - mental subnormality	12 casos
Ataxia - deafness - optic atrophy, lethal	12 casos
Atelosteogenesis I	12 casos
Atelosteogenesis III	12 casos
Atrial tachyarrhythmia with short PR interval	12 casos
Coloboma of macula - brachydactyly type B	12 casos
Coloboma uveal - cleft lip palate - mental retardation	12 casos
Distal myopathy with vocal cord weakness	12 casos
Distal myopathy, with posterior leg and anterior upper limb involvement	12 casos
Dopamine beta-hydroxylase, deficiency of	12 casos
Hypoparathyroidism - deafness - renal disease	12 casos
Mental retardation, X-linked - macrocephaly - macro-orchidism	12 casos
N-acetyl-alpha-D-galactosaminidase deficiency	12 casos
Osteocraniostenosis	12 casos
Winchester disease	12 casos
Bickel-Fanconi glycogenosis	112 casos
Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges	11 casos
Atrial septal defect - atrioventricular conduction	11 casos
Corpus callosum dysgenesis, X-linked recessive	11 casos
Fibrochondrogenesis	11 casos
Fibular dimelia - diplopodia	11 casos
Foveal hypoplasia presenile cataract	11 casos
Fuhrmann syndrome	11 casos
Lopez-Hernandez syndrome	11 casos
Mental retardation, X-linked, Snyder type	11 casos
Necrotizing encephalopathy, acute, autosomal dominant	11 casos
PELVIS syndrome	11 casos
Pterygium syndrome, antecubital	11 casos
Tricho-megaly - retina pigmentary degeneration - dwarfism	11 casos
CHAND syndrome	>10 casos
Palmoplantar porokeratosis of Mantoux	>10 casos
Chediak-Higashi syndrome	>10 casos
Median cleft of the upper lip - corpus callosum lipoma - cutaneous polyps	>10 casos
Diffuse palmoplantar keratoderma - acrocyanosis	10 casos
Acromesomic dysplasia Hunter-Thompson type	10 casos
Albright like syndrome	10 casos
Ambras syndrome	10 casos
Arthrogryposis multiplex congenita - whistling face	10 casos
Athabaskan brainstem dysgenesis syndrome	10 casos
Barber-Say syndrome	10 casos
Boomerang dysplasia	10 casos
Cardiac conduction disease, dilated cardiomyopathy and brachydactyly	10 casos

Cataract - mental retardation - hypogonadism	10 casos
Cerebro-oculo-nasal syndrome	10 casos
Char syndrome	10 casos
Choroidocerebral calcification syndrome, infantile form	10 casos
Colobomatous - microphthalmia - heart disease - hearing loss	10 casos
Distal monosomy 5q	10 casos
Ectodermal dysplasia-skin fragility syndrome	10 casos
Flynn-Aird syndrome	10 casos
Goldberg-Shprintzen megacolon syndrome	10 casos
Hyperkeratosis - hyperpigmentation syndrome	10 casos
Ichthyosis follicularis-atrichia-photophobia syndrome	10 casos
Iris coloboma with ptosis - intellectual deficit	10 casos
Juberg-Hayward syndrome	10 casos
Lipodystrophy, familial partial, associated with PPARG mutations	10 casos
Mental retardation, X-linked - hypotonia - facial dysmorphism - aggressive behavior	10 casos
Mental retardation, X-linked, syndromic 7	10 casos
Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus	10 casos
Nevo syndrome	10 casos
Pollitt syndrome	10 casos
Progressive vertebral fusion, non-infectious, syndromic form	10 casos
Pseudodiastrophic dysplasia	10 casos
Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts	10 casos
Succinyl-CoA acetoacetate transferase deficiency	10 casos
Toriello-Lacassie-Droste syndrome	10 casos
Neurodegenerative syndrome, X-linked, Hamel type	10 casos
Aase-Smith syndrome	<10 casos
Acro-facial dysostosis, Rodriguez type	<10 casos
Acromelanosis	<10 casos
Astley-Kendall dysplasia	<10 casos
Carpotarsal osteochondromatosis	<10 casos
Carpotarsalosteolysis, autosomal recessive	<10 casos
Cerebral gigantism - jaw cysts	<10 casos
Corneal dystrophy - perceptive deafness	<10 casos
Deafness - lymphoedema - leukemia	<10 casos
Diaphanospondylodysostosis	<10 casos
Digito-reno-cerebral syndrome	<10 casos
Glomerulonephritis - sparse hair - telangiectases	<10 casos
Greenberg dysplasia	<10 casos
Hypopituitarism - microphthalmia	<10 casos
Myoclonus hereditary - progressive distal muscular atrophy	<10 casos
Pacman dysplasia	<10 casos
Pancreatic hypoplasia - diabetes - heart disease	<10 casos
Pfeiffer-Singer-Zschiesche syndrome	<10 casos
Phosphoenolpyruvate carboxykinase (PEPCK) deficiency	<10 casos
Progeria - short stature - pigmented nevi	<10 casos

Pseudo-Gaucher disease	<10 casos
Pseudo-Zellweger syndrome	<10 casos
Singleton-Merten dysplasia	<10 casos
Xk aprosencephaly	<10 casos
Atransferrinemia	9 casos
Bosley-Salih-Alorainy syndrome	9 casos
Brachymorphism - onychodysplasia - dysphalangism	9 casos
Cardiomyopathy - cataract - hip spine disease	9 casos
Gamma-glutamylcysteine synthetase deficiency	9 casos
Guanidinoacetate methyltransferase deficiency	9 casos
Macrostomia - preauricular tags - external ophthalmoplegia	9 casos
Mental retardation, X-linked, Shashi type	9 casos
Microdontia - type I microtia - deafness	9 casos
Oculocerebrofacial syndrome, Kaufman type	9 casos
Tricho-retino-dento-digital syndrome	9 casos
Ataxia - apraxia - mental retardation, X-linked	9 casos
5-oxoprolinase deficiency	8 casos
Ackerman syndrome	8 casos
Ankylosing vertebral hyperostosis - tylosis	8 casos
Campomelia Cumming type	8 casos
Camptodactyly syndrome, Guadalajara type 1	8 casos
COACH syndrome	8 casos
Corpus callosum, agenesis - cataract - immunodeficiency	8 casos
Deafness - skeletal dysplasia - lip granuloma	8 casos
Familial hematuria, autosomal dominant - retinal arteriolar tortuosity - contractures	8 casos
Hydrocephalus - costovertebral dysplasia - Sprengel anomaly	8 casos
Hypomyelination - congenital cataract	8 casos
Mental retardation X-linked - dysmorphism	8 casos
Mental retardation, X-linked, Abidi type	8 casos
Mental retardation, X-linked, Vitale type	8 casos
Micro syndrome	8 casos
Pancreas agenesis	8 casos
Spondylometaphyseal dysplasia - cone-rod dystrophy	8 casos
Trigonocephaly - ptosis coloboma	8 casos
Rubinstein-Taybi like, syndrome	8 casos
Acrorenalmandibular syndrome	7 casos
Albinism ocular - late onset sensorineural deafness	7 casos
Cardiogenital syndrome	7 casos
CDG syndrome type Ie	7 casos
CEDNIK syndrome	7 casos
Cleft palate-lateral synechia syndrome	7 casos
Dihydropyrimidinuria	7 casos
Ehlers-Danlos syndrome, type 7C	7 casos
Gamma-glutamyl transpeptidase deficiency	7 casos
Genitopatellar syndrome	7 casos
Lenz-Majewski hyperostotic dwarfism	7 casos
MEHMO syndrome	7 casos
Mental retardation, X-linked severe, Gustavson type	7 casos

Michels syndrome	7 casos
Multiple fibrofolliculoma familial	7 casos
Myopathy - lactic acidosis - sideroblastic anemia	7 casos
Neutropenia, severe congenital, X-linked	7 casos
Obesity due to pro-opiomelanocortin deficiency	7 casos
Retinal degeneration - nanophthalmos - glaucoma	7 casos
Retino-hepato-endocrinologic syndrome	7 casos
Stern-Lubinsky-Durrie syndrome	7 casos
Torticollis - keloids - cryptorchidism - renal dysplasia	7 casos
MEHMO syndrome	7 casos
Neurodegenerative syndrome, X-linked, Bertini type	7 casos
Mental retardation - microcephaly - phalangeal - facial abnormalities	6 casos
3-hydroxy 3-methylglutaryl-CoA synthase	6 casos
Acro-facial dysostosis, Catania form	6 casos
Agonadism - dextrocardia - diaphragmatic hernia	6 casos
Aplasia cutis congenita of limbs recessive	6 casos
Biliary malformation - renal tubular insufficiency	6 casos
Blepharophimosis - ptosis - esotropia - syndactyly - short stature	6 casos
CDG syndrome type Ig	6 casos
Cutis gyrata - acanthosis nigricans - craniosynostosis	6 casos
Deafness - enamel hypoplasia - nail defects	6 casos
Eiken syndrome	6 casos
Epilepsy - telangiectasia	6 casos
Fanconi - ichthyosis - dysmorphism	6 casos
Grange syndrome	6 casos
Hidrotic ectodermal dysplasia, Christianson-Fourie type	6 casos
Hypopituitarism - postaxial polydactyly	6 casos
Isotretinoin-like syndrome	6 casos
Keratosis follicularis - dwarfism - cerebral atrophy	6 casos
Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus	6 casos
Mental retardation - sparse hair - brachydactyly	6 casos
Mental retardation X-linked - psychosis - macroorchidism	6 casos
Mental retardation, X-linked, Armfield type	6 casos
Mental retardation, X-linked, Zorick type	6 casos
Moore-Federman syndrome	6 casos
Polysyndactyly - cardiac malformation	6 casos
Serpentine fibula - polycystic kidneys	6 casos
Spasticity - mental retardation - epilepsy, X-linked	6 casos
Symphalangism with multiple anomalies of hands and feet	6 casos
Thumb stiff - brachydactyly - mental retardation	6 casos
W syndrome	6 casos
Wieacker-Wolff syndrome	6 casos
Zunich-Kaye syndrome	6 casos
Branchial arch syndrome, X-linked	5 à 7 casos
Cleft palate - cardiac defect - genital anomalies - ectrodactyly	5 casos
Lissencephaly type III - familial fetal akinesia sequence	5 casos

Lymphoedema - cerebral arteriovenous anomaly	5 casos
Achalasia - microcephaly	5 casos
Acro-fronto-facio-nasal dysostosis	5 casos
Adducted thumbs - arthrogryposis, Dundar type	5 casos
Alopecia - contractures - dwarfism - mental retardation	5 casos
Anonychia - microcephaly	5 casos
Arachnodactyly - ossification abnormal - mental retardation	5 casos
Auro-cephalo-syndactyly	5 casos
Bamforth syndrome	5 casos
Beta-ureidopropionase deficiency	5 casos
Bradyopsia	5 casos
Branchiogenic deafness syndrome	5 casos
CAMOS syndrome	5 casos
CDG syndrome type Ih	5 casos
Choanal atresia - deafness - cardiac defects dysmorphism	5 casos
Craniosynostosis - brachydactyly	5 casos
Curry-Jones syndrome	5 casos
Cutis verticis gyrata - thyroid aplasia - mental retardation	5 casos
Dacryocystitis osteopoikilosis	5 casos
Depigmentation of the iris, acute, bilateral	5 casos
Dermatoosteolysis, Kirghizian type	5 casos
Ectrodactyly - ectodermal dysplasia	5 casos
Fine-Lubinsky syndrome	5 casos
German syndrome	5 casos
Glaucoma-sleep apnea	5 casos
Ichthyosis - male hypogonadism	5 casos
Matthew-Wood syndrome	5 casos
Mental retardation - cubitus valgus - unusual facies	5 casos
Mental retardation, choreoathesis and abnormal behavior	5 casos
Neonatal death - immune deficiency	5 casos
Nephropathy - deafness - hyperparathyroidism	5 casos
Nephrosis - deafness - urinary tract and digital malformations	5 casos
Oculo-dento-osseous dysplasia, autosomal recessive	5 casos
Oculo-palato-cerebral syndrome	5 casos
Odonto-micronychial dysplasia	5 casos
Onycho-tricho-dysplasia - neutropenia	5 casos
P2Y12 deficiency	5 casos
Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma	5 casos
Paraplegia - brachydactyly - cone-shaped epiphysis	5 casos
Sacral hemangiomas - multiple congenital abnormalities	5 casos
Sillence syndrome	5 casos
Spinocerebellar ataxia X-linked, type 3	5 casos
CDG syndrome type Id	5 casos
Keratoderma - epithelioma - dental abnormalities-hypogonadism	5 casos
Mental retardation, X-linked, Lubs type	5 casos

Cystic hamartoma of lung and kidney	<5 casos
Lathosterolosis	<5 casos
Sakati-Nyhan syndrome	<5 casos
Palmoplantar keratoderma - amyotrophy	4 casos
Rolled and spiral hairs - palmoplantar keratoderma	4 casos
Abruzzo-Erickson syndrome	4 casos
Acro-facial dysostosis, Palagonia type	4 casos
Anophthalmia plus syndrome	4 casos
Aortic arch anomaly - peculiar facies - mental retardation	4 casos
Aplasia cutis - myopia	4 casos
Arhinia - choanal atresia - microphthalmia	4 casos
Arterial dissection - lentiginosis	4 casos
Benign exophthalmos syndrome	4 casos
Bone dysplasia lethal, Holmgren type	4 casos
Bone fragility - craniosynostosis - proptosis hydrocephalus	4 casos
Bonnemann-Meinecke-Reich syndrome	4 casos
Brachydactyly, long thumb type	4 casos
Caudal dysgenesis familial type	4 casos
CDG syndrome type If	4 casos
Cholestasis - pigmentary retinopathy - cleft palate	4 casos
Chondrodysplasia lethal, recessive	4 casos
Choroideremia - deafness - obesity	4 casos
Cleft lip palate - malrotation - cardiopathy	4 casos
Coxo-auricular syndrome	4 casos
Craniofaciocardioskeletal syndrome	4 casos
Cranioosteoarthropathy	4 casos
Craniosynostosis - Dandy Walker hydrocephalus	4 casos
Deafness - peripheral neuropathy - arterial disease	4 casos
Diaphragmatic defect - limb deficiency - skull defect	4 casos
Duker-Weiss-Siber syndrome	4 casos
Ectodermal dysplasia, Berlin type	4 casos
Ectopia lentis - chorioretinal dystrophy - myopia	4 casos
Facial onset sensory and motor neuropathy	4 casos
Gaucher - ichthyosis - restrictive dermopathy	4 casos
Gombo syndrome	4 casos
Gorlin-Chaudhry-Moss, syndrome	4 casos
Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate	4 casos
Growth retardation - microcephaly - digital abnormalities - hypospadias	4 casos
Hidrotic ectodermal dysplasia, Halal type	4 casos
Homocarnosinosis	4 casos
Hypogammaglobulinemia due to CD19 deficiency	4 casos
Hypomandibular facio-cranial dysostosis	4 casos
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 casos
Hypomyelination - hypogonadotropic hypogonadism - hypodontia	4 casos
Hypotrichosis - lymphedema - telangiectasia	4 casos
Ichthyosis - alopecia - ectropion - mental retardation	4 casos
Immunodeficiency with natural-killer cell deficiency	4 casos

Leukodystrophy with oligodontia	4 casos
Leukoencephalopathy - metaphyseal chondrodysplasia	4 casos
Leukoencephalopathy - palmoplantar keratoderma	4 casos
Liver disease - retinitis pigmentosa - polyneuropathy - epilepsy	4 casos
Macrogyria - pseudobulbar palsy	4 casos
Malignant hyperthermia arthrogyrosis torticollis	4 casos
Mental retardation - dysmorphism - hypogonadism - diabetes mellitus	4 casos
Mental retardation, X-linked - seizures - psoriasis	4 casos
Mental retardation, X-linked, Miles-Carpenter type	4 casos
Mental retardation, X-linked, Schimke type	4 casos
Mental retardation, X-linked, Seemanova type	4 casos
Mental retardation, X-linked, Siderius type	4 casos
Mental retardation, X-linked, Stevenson type	4 casos
Mental retardation, X-linked, Stocco Dos Santos type	4 casos
Mental retardation, X-linked, Stoll type	4 casos
Metaphyseal acroschiphodysplasia	4 casos
Microcephalic osteodysplastic dysplasia, Saul-Wilson type	4 casos
Microcephaly - micropenis - convulsions	4 casos
Multiple pterygium syndrome, autosomal dominant	4 casos
Myoclonus - cerebellar ataxia - deafness	4 casos
Myopathy due to casequestrin and SERCA1 protein overload	4 casos
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	4 casos
Oculo-oto-facial dysplasia	4 casos
Odontotrichomelic syndrome	4 casos
Orofaciodigital syndrome, type 5	4 casos
Osteopoikilosis - short stature - intellectual deficit	4 casos
Pancreatic and cerebellar agenesis	4 casos
Paraplegia - mental retardation - hyperkeratosis	4 casos
Recurrent infections - short stature - hypopigmentation - coarse face	4 casos
Severe achondroplasia - developmental delay - acanthosis nigricans	4 casos
Short stature - webbed neck - heart disease	4 casos
Simpson-Golabi-Behmel syndrome, type 2	4 casos
Sparse hair - short stature - skin anomalies	4 casos
Spastic paraplegia - nephritis - deafness	4 casos
Spondylometaphyseal dysplasia - combined immunodeficiency	4 casos
Trichoodontoonychial dysplasia	4 casos
CDG syndrome type IIa	4 casos
CDG syndrome type Ik	4 casos
Mental retardation, X-linked - hypogonadism - ichthyosis - obesity - short stature	4 casos
Neuroectodermal endocrine syndrome	4 casos
Cranio-digital syndrome - mental retardation	4 casos
Arachnodactyly - mental retardation - dysmorphism	3 casos
Lymphoedema - atrial septal defects - facial changes	3 casos
Acromesomelic dysplasia, Brahimi-Bacha type	3 casos

Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis	3 casos
Amelia, autosomal recessive	3 casos
Aniridia - absent patella	3 casos
Aniridia - ptosis - mental retardation - obesity, familial type	3 casos
Anonychia with flexural pigmentation	3 casos
Anophthalmia - megalocornea - cardiopathy - skeletal anomalies	3 casos
Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis	3 casos
Aplasia cutis congenita - intestinal lymphangiectasia	3 casos
Aredyld syndrome	3 casos
Axenfeld-Rieger anomaly - hydrocephaly - skeletal abnormalities	3 casos
Blepharoptosis - myopia - ectopia lentis	3 casos
Bone sclerosing - dysplasia - ichthyosis - premature ovarian failure	3 casos
Brachio-skeleto-genital syndrome	3 casos
Buttiens-Fryns syndrome	3 casos
Cataract - deafness - hypogonadism	3 casos
Cervical hypertrichosis - peripheral neuropathy	3 casos
Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	3 casos
Cortical blindness - mental retardation - polydactyly	3 casos
Cranio-facial dysmorphism - coloboma - corpus callosum agenesis	3 casos
Craniofacial-deafness-hand syndrome	3 casos
Craniofrontonasal dysplasia - poland anomaly	3 casos
Craniosynostosis - intracranial calcifications	3 casos
Cutaneous albinism, ermine phenotype	3 casos
Cutaneous photosensitivity - colitis lethal	3 casos
Deafness - opticoacoustic nerve atrophy - dementia	3 casos
Deafness-mental retardation, Martin-Probst type	3 casos
Deafness-mental retardation, Martin-Probst type	3 casos
Disorder of sex development - mental retardation	3 casos
Ectodermal dysplasia, hypohidrotic - hypothyroidism - ciliary dyskinesia	3 casos
Ectodermic dysplasia - hypothyroidism - cleft	3 casos
Eyebrow duplication syndactyly	3 casos
Facial dysmorphism - macrocephaly - myopia - Dandy Walker	3 casos
Faciocardiomeic dysplasia, lethal	3 casos
Gardner-Morrison-Abbot syndrome	3 casos
Gemss syndrome	3 casos
Global developmental delay - osteopenia - ectodermal defect	3 casos
Goodman syndrome	3 casos
Haemolytic anaemia due to glutathione reductase deficiency	3 casos
Hair defect - photosensitivity - mental retardation	3 casos
Hirschsprung disease - nail hypoplasia - dysmorphism	3 casos
Hypogonadism - retinitis pigmentosa	3 casos
Lewis-Pashayan syndrome	3 casos
Lipodystrophy - mental retardation - deafness	3 casos

Lumbosacral vertebrae, posterior fusion of - blepharoptosis	3 casos
Mental retardation - hypoplastic corpus callosum - preauricular tag	3 casos
Mental retardation, X linked - precocious puberty - obesity	3 casos
Mental retardation, X-linked, Shrimpton type	3 casos
Mental retardation, X-linked, Wilson type	3 casos
Microcephaly - cardiomyopathy	3 casos
Microcephaly - cleft palate	3 casos
Microcytic anemia - liver iron overload - low ferritinemia	3 casos
Microphthalmia - brain atrophy	3 casos
Mitral regurgitation - deafness - skeletal anomalies	3 casos
Mullerian derivatives - lymphangiectasia - polydactyly	3 casos
N syndrome	3 casos
Nail patella-like renal disease	3 casos
Nanism due to growth hormone qualitative anomaly	3 casos
Neuroaxonal dystrophy - renal tubular acidosis	3 casos
Oculoosteocutaneous syndrome	3 casos
Omphalocele-cleft palate syndrome, lethal	3 casos
Orofaciadigital syndrome, type 3	3 casos
Osteogenesis imperfecta congenita - microcephaly - cataracts	3 casos
Osteoporosis oculo-cutaneous hypopigmentation syndrome	3 casos
Osteosclerosis - ichthyosis - premature ovarian failure	3 casos
Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency	3 casos
Qazi-Markouizos syndrome	3 casos
Rambaud-Galian syndrome	3 casos
Renal dysplasia, multicystic	3 casos
Seizures - intellectual deficit due to hydroxylysinuria	3 casos
Sensorineural hearing loss - early greying - essential tremor	3 casos
Short stature - mental retardation - eye anomalies - cleft lip palat	3 casos
Split hand - urinary anomalies - spina bifida	3 casos
Summitt syndrome	3 casos
Thumb absent - short stature - immune deficiency	3 casos
Thymic-renal-anal-lung dysplasia	3 casos
Trigonocephaly - short stature - developmental delay	3 casos
Ulbright-Hodes syndrome	3 casos
Wittwer syndrome	3 casos
Mental retardation, X-linked - hypogammaglobulinemia - progressive neurological deterioration	3 casos
Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia	3 casos
Lissencephaly type III - metacarpal bone dysplasia	2 casos
46,XX disorders of sex development - skeletal anomalies	2 casos
Acrocraniofacial dysostosis	2 casos
Acrofacial dysostosis autosomal recessive	2 casos

Agenesis of the corpus callosum - mental retardation - coloboma - micrognathia	2 casos
Alar cartilages hypoplasia - coloboma - telecanthus	2 casos
Alopecia - hypogonadism - extrapyramidal disorder	2 casos
Amaurosis - hypertrichosis	2 casos
Aniridia - renal agenesis - psychomotor retardation	2 casos
Aniridia-mental retardation syndrome	2 casos
Arthrogryposis - hyperkeratosis, lethal form	2 casos
Atherosclerosis- deafness - diabetes - epilepsy - nephropathy	2 casos
Aughton syndrome	2 casos
Auricular abnormalities - cleft lip with or without cleft palate - ocular abnormalities	2 casos
Auriculoocular anomalies - cleft lip	2 casos
Bangstad syndrome	2 casos
Beemer-Ertbruggen syndrome	2 casos
Blepharo-facio-skeletal syndrome	2 casos
Bouwes-Bavinck syndrome	2 casos
Brachytelephalangy - dysmorphism - Kallmann syndrome	2 casos
Braddock syndrome	2 casos
Brain malformation - congenital heart disease - postaxial polydactyly	2 casos
Campodactyly syndrome, Guadalajara type 2	2 casos
Cardiomyopathy - renal anomalies	2 casos
Cataract - ataxia - deafness	2 casos
Cataract - microphthalmia - septal defect	2 casos
Cataract - nephropathy - encephalopathy	2 casos
Chondrodysplasia - disorder of sex development	2 casos
Choroidal atrophy - alopecia	2 casos
Cleft lip - retinopathy	2 casos
Cleft lip palate - mental retardation - corneal opacities	2 casos
Cleft palate - short stature - vertebral anomalies	2 casos
Cleft palate - stapes fixation - oligodontia	2 casos
Cleido-rhizomelic syndrome	2 casos
Congenital ichthyosis - microcephalus - quadriplegia	2 casos
Contractures - ectodermal dysplasia - cleft lip palate	2 casos
Cooper-Jabs syndrome	2 casos
Corneal anesthesia - deafness - mental retardation	2 casos
Corneal-cerebellar syndrome	2 casos
Craniosynostosis - fibular aplasia	2 casos
Craniosynostosis radial aplasia, Imaizumi type	2 casos
Craniotubular syndrome	2 casos
Cryptomicrotia - brachydactyly - excess fingertip arch	2 casos
Cystic hygroma lethal - cleft palate	2 casos
Dahlberg-Borer-Newcomer syndrome	2 casos
Dandy Walker - macrocephaly	2 casos
Dandy Walker malformation - postaxial polydactyly	2 casos
Deaf - blind - hypopigmentation	2 casos
Deafness - tubular acidosis - anemia	2 casos
Deafness - vitiligo - achalasia	2 casos

Dentinogenesis imperfecta - short stature - hearing loss - mental retardation	2 casos
Dermato-cardio-skeletal syndrome, Borrone type	2 casos
Dermatoleukodystrophy	2 casos
Desmosterolosis	2 casos
Developmental malformations - deafness - dystonia	2 casos
Diabetes, neonatal - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys	2 casos
Dincsoy-Salih-Patel syndrome	2 casos
Duane anomaly - myopathy - scoliosis	2 casos
Dysmorphism - short stature - deafness - pseudohermaphroditism	2 casos
Ectodermal dysplasia - blindness	2 casos
Ectodermal dysplasia anhidrotic - immunodeficiency - osteopetrosis - lymphedema	2 casos
Eng-Strom syndrome	2 casos
Epilepsy - microcephaly - skeletal dysplasia	2 casos
Fuqua-Berkovitz syndrome	2 casos
Gamma aminobutyric acid transaminase deficiency	2 casos
Gonadal dysgenesis, XY type - associated anomalies	2 casos
Hec syndrome	2 casos
Hemolytic anemia, lethal - genital anomalies	2 casos
Hennekam-Beemer syndrome	2 casos
Hersh-Podruch-Weisskopf syndrome	2 casos
Hirschsprung disease - deafness - polydactyly	2 casos
Hydrocephaly - tall stature - joint laxity	2 casos
Hypercoagulability syndrome, due to glycosylphosphatidylinositol deficiency	2 casos
Hypomagnesemia with normocalciuria	2 casos
Ichthyosis - hepatosplenomegaly - cerebellar degeneration	2 casos
Ichthyosis - oral and digital anomalies	2 casos
Ichthyosis congenita - biliary atresia	2 casos
Inappropriate antidiuretic hormone secretion syndrome	2 casos
Intellectual deficit, severe - epilepsy - anal anomalies - distal phalangeal hypoplasia	2 casos
Iris dysplasia - hypertelorism - deafness	2 casos
Kaler-Garrity-Stern syndrome	2 casos
Kapur-Toriello syndrome	2 casos
Kniest-like dysplasia, lethal	2 casos
Kudo-Tamura-Fuse syndrome	2 casos
Lichstenstein syndrome	2 casos
Low birth weight - dwarfism - dysgammaglobulinemia	2 casos
Macrocephaly - immune deficiency - anemia	2 casos
Macrocephaly - short stature - paraplegia	2 casos
Mental retardation, X-linked - acromegaly - hyperactivity	2 casos
Mental retardation, X-linked - epilepsy - progressive joint contractures - typical face	2 casos
Mental retardation, X-linked, Cantagrel type	2 casos
Mental retardation, X-linked, Reish type	2 casos
Mesomelic dysplasia - skin dimples	2 casos
Methylmalonic aciduria - microcephaly - cataract	2 casos

Microbrachycephaly - ptosis - cleft lip	2 casos
Microcephaly - glomerulonephritis - marfanoid habitus	2 casos
Microcephaly - seizures - mental retardation - heart disease	2 casos
Microlissencephaly - micromelia	2 casos
Mitochondrial encephalomyopathy - aminoacidopathy	2 casos
Monosomy 9q22.3	2 casos
Multiple sclerosis - ichthyosis - factor VIII deficiency	2 casos
Nephronophtisis familial - adult spastic quadripareisis	2 casos
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	2 casos
Obesity - colitis - hypothyroidism - cardiac hypertrophy - developmental delay	2 casos
Obesity due to prohormone convertase-I deficiency	2 casos
Oculo-tricho-dysplasia	2 casos
Okamoto syndrome	2 casos
Ossification anomalies - psychomotor development delay	2 casos
Osteogenesis imperfecta - retinopathy - seizures - intellectual deficit	2 casos
Osteopetrosis lethal	2 casos
PARC syndrome	2 casos
Pilodental dysplasia with refractive errors	2 casos
Plagiocephaly - mental retardation, X-linked	2 casos
Progressive neurodegeneration - joint laxity - cataract	2 casos
Pseudo-progeria syndrome	2 casos
Pterygium colli - intellectual deficit - digital anomalies	2 casos
Ptosis strabismus diastasis	2 casos
Robinow like syndrome	2 casos
Rudiger syndrome	2 casos
Say-Barber-Miller syndrome	2 casos
Scalp defects - postaxial polydactyly	2 casos
SCARF syndrome	2 casos
Short stature, Brussels type	2 casos
Siegler-Brewer-Carey syndrome	2 casos
Small vessel disease of the brain, not NOTCH3-related	2 casos
Spastic quadriplegia - retinitis pigmentosa - mental retardation	2 casos
Spinal muscular atrophy - Dandy-Walker complex - cataracts	2 casos
Stimmler syndrome	2 casos
Talo-patello-scapoid osteolysis	2 casos
Thyrocerebrorenal syndrome	2 casos
Trigonocephaly - bifid nose - acral anomalies	2 casos
Trigonocephaly - broad thumbs	2 casos
Tubular renal disease - cardiomyopathy	2 casos
Tyrosinemia type 3	2 casos
Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay	2 casos
Weaver-Williams syndrome	2 casos
Xeroderma - talipes - enamel defects	2 casos

Zellweger-like syndrome, without peroxisomal anomalies	2 casos
CDG syndrome type IIe	2 casos
CDG syndrome type IL	2 casos
Heart defects - limb shortening	2 casos
Keratoderma - hypotrichosis - leukonychia	2 casos
Hypotrichosis-mental retardation lopes type	1 caso
2,4-dienoyl-CoA reductase deficiency	1 caso
46 XX gonadal dysgenesis - epibulbar dermoid	1 caso
Acro-facial dysostosis postaxial, atypical	1 caso
Acro-facial dysostosis, Preis type	1 caso
Anophthalmia - short stature - obesity	1 caso
Arthrogryposis - iugr thoracic - dystrophy	1 caso
Blaichman syndrome	1 caso
Bone dysplasia, Azouz type	1 caso
Cataract - hypertrichosis - mental retardation	1 caso
Chondrodysplasia - situs inversus- imperforate anus - polydactyly	1 caso
Corneal crystals - myopathy - neuropathy	1 caso
Corneal crystals myopathy neuropathy	1 caso
Corneal dystrophy - ichthyosis - microcephaly - mental retardation	1 caso
Ectodermal dysplasia - alopecia - preaxial polydactyly	1 caso
Ectodermal dysplasia - arthrogryposis - diabetes mellitus	1 caso
Ectodermal dysplasia - mental retardation - syndactyly	1 caso
Enamel hypoplasia - cataract - hydrocephaly	1 caso
Facial clefting - corpus callosum agenesis	1 caso
Glossopalatine ankylosis - cataracts - digital anomalies	1 caso
Hyaluronidase deficiency	1 caso
Hydrops - ectrodactyly - syndactyly	1 caso
Hypertrichosis - brachydactyly - obesity - mental retardation	1 caso
Hypothyroidism - dermoid cyst - cleft palate	1 caso
Ichthyosis - deafness - mental retardation - skeletal anomalies	1 caso
Intrathoracic kidney vertebral fusion	1 caso
Lissencephaly - immunodeficiency	1 caso
Macroepiphyseal dysplasia, Mcalister coe type	1 caso
Martinez-Monasterio-Pinheiro syndrome	1 caso
Megaepiphyseal dwarfism	1 caso
Mental retardation - multiple nevi	1 caso
Metaphyseal dysplasia - hypertelorism - hypospadias	1 caso
Microcephaly - syndactyly - brachymesophalangy	1 caso
Molarization of anterior teeth - deafness	1 caso
Multiple joint dislocations - metaphyseal dysplasia	1 caso
Nasopharyngeal teratoma - Dandy Walker - diaphragmatic hernia	1 caso
Nevus of ota - retinitis pigmentosa	1 caso
Odonto-onycho-hypohidrotic dysplasia - midline scalp defects	1 caso
Orofacioidigital syndrome, type 10	1 caso

Osteochondrodysplasia thrombocytopenia hydrocephalus	1 caso
Pancreatic lipomatosis - duodenal stenosis	1 caso
Patella hypoplasia - skeletal malformations	1 caso
Podder-Tolmie syndrome	1 caso
Preauricular pits - renal disease	1 caso
Richieri costa colletto otto syndrome	1 caso
Rigid mask like face - deafness - polydactyly	1 caso
Sclerosing bone dysplasia - mental retardation	1 caso
Sclerosing bone dysplasia - mental retardation	1 caso
Spondylo-costal dysostosis - Dandy Walker	1 caso
Stoll-Alembik-Finck syndrome	1 caso
Symphalangism - short stature - accessory testis	1 caso
Symphalangism short stature accessory testis	1 caso
Trichooculodermovertebral syndrome	1 caso
Trichoonychohypohidrotic dysplasia	1 caso
Bone dysplasia - corpus callosum agenesis	1 caso
CDG syndrome type Ii	1 caso
CDG syndrome type IIb	1 caso
CDG syndrome type IIId	1 caso
CDG syndrome type Ij	1 caso
Li-Fraumeni syndrome	400 familias
Birt-Hogg-Dube syndrome	>60 familias
Nephropathy familial with gout	57 familias
Nance-Horan syndrome	50 familias
Muscular dystrophy, limb girdle, autosomal recessive, type 2I	>40 familias
Granulomatous arthritis of childhood	40 familias
Thiamine-responsive megaloblastic anemia syndrome	30 familias
Erythermalgia, primary	30 familias
Phosphoribosylpyrophosphate synthetase superactivity	<30 familias
Neuropathy, giant axonal	> 20 familias
Overhydrated hereditary stomatocytosis	> 20 familias
Synostoses, multiple - brachydactyly	20 familias
Fahr syndrome	<20 familias
Acrorenocular syndrome	<20 familias
Platelet syndrome, familial	<20 familias
Haemolytic anaemia, nonspherocytic, due to hexokinase deficiency	17 familias
Ichthyosis prematurity syndrome	16 familias
Myopathy, X-linked, with excessive autophagy	15 familias
Familial platelet syndrome with predisposition to acute myelogenous leukemia	13 familias
Marie Unna congenital hypotrichosis	12 familias
Aniridia - cerebellar ataxia - mental deficiency	>10 familias
Brachydactyly - arterial hypertension	>10 familias
Aortic aneurysm syndrome, Loeys-Dietz type	10 familias
Porencephaly, familial	10 familias
Renpenning syndrome	10 familias
Acheiropodia	<10 familias
Angioma hereditary neurocutaneous	<10 familias
Focal facial dermal dysplasia	<10 familias

Hypoparathyroidism familial isolated	<10 familias
Keratoderma palmoplantar - deafness	<10 familias
Mental retardation, X-linked, syndromic, due to JARID1C mutation	<10 familias
Schinzel syndrome	<10 familias
Sebastian syndrome	<10 familias
VACTERL with hydrocephalus	<10 familias
Otodental syndrome	9 familias
Ankyloblepharon - ectodermal defects - cleft lip palate	8 familias
Cataract-microcornea syndrome	8 familias
Lethal osteosclerotic bone dysplasia	8 familias
Juvenile macular degeneration, hypotrichosis	7 familias
EEM syndrome	7 familias
Immune dysregulation - polyendocrinopathy - enteropathy, X linked	7 familias
Cystoid macular dystrophy	6 familias
Developmental dysphasia familial	6 familias
Pontocerebellar hypoplasia type 1	6 familias
Encephalopathy with neuroserpin inclusion bodies, familial form	>5 familias
Anemia, sideroblastic, X-linked - ataxia	5 familias
Muscular dystrophy, limb-girdle, autosomal dominant, type 1D	5 familias
Muscular dystrophy, limb-girdle, autosomal dominant, type 1E	5 familias
Tetraamelia - pulmonary hypoplasia	5 familias
Symphalangism distal	<5 familias
Trichodontal syndrome	<5 familias
Camptodactyly - taurinuria	4 familias
Familial rectal pain	4 familias
IVIC syndrome	4 familias
Triphalangeal thumbs - brachyectrodactyly	4 familias
Adducted thumbs - arthrogryposis, Christian type	3 familias
CAMFAK syndrome	3 familias
Cataract-glaucoma	3 familias
Cerebroretinal vasculopathy	3 familias
Deafness-infertility syndrome	3 familias
HERNS syndrome	3 familias
Hypomagnesemia with hypocalciuria	3 familias
Mental retardation, X-linked, with isolated growth hormone deficiency	3 familias
Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss	1-2 familias
Skeletal dysplasia - intellectual deficit	1-2 familias
Mixed dystonias	3 familias
Rapid-onset dystonia-parkinsonism	3 familias
North Carolina macular dystrophy	2 familias
Ankyloblepharon filiforme - imperforate anus	2 familias
Bence syndrome	2 familias
Blepharonasofacial malformation syndrome	2 familias
Bullous dystrophy macular type	2 familias
Chorioretinal atrophy, progressive bifocal	2 familias
Earlobes, thickened - conductive deafness	2 familias

Ehlers-Danlos syndrome, type 5	2 familias
Episodic ataxia, type 4	2 familias
Fibromatosis, gingival - progressive deafness	2 familias
Hypoparathyroidism X-linked	2 familias
Jackson-Weiss syndrome	2 familias
Paraplégie spastique - glaucome - déficit intellectuel	2 familias
Retinitis pigmentosa - deafness - hypogenitalism	2 familias
Orofaciodigital syndrome, type 8	1 ou 2 familias
Albinism-deafness syndrome	1 familia
Alopecia - congenita keratosis palmoplantaris	1 familia
Aphalangia - syndactyly - microcephaly	1 familia
Banki syndrome	1 familia
Camptobrachydactyly	1 familia
Cataract, Hutterite type	1 familia
Congenital alopecia, X linked	1 familia
Coronary artery disease - hyperlipidemia - hypertension - diabetes - osteoporosis	1 familia
Costocoracoid ligament congenitally short	1 familia
Craniofacial conodysplasia	1 familia
Craniosynostosis, Philadelphia type	1 familia
Cyprus facial neuromusculoskeletal syndrome	1 familia
Dyschondrosteosis - nephritis	1 familia
Ehlers-Danlos syndrome type 10	1 familia
Episodic ataxia, type 3	1 familia
Hereditary vascular retinopathy	1 familia
Hereditary vascular retinopathy	1 familia
Hydrocephalus - blue sclerae - nephropathy	1 familia
Infundibulopelvic stenosis - multicystic kidney	1 familia
Insulin resistance, short fifth metacarpals	1 familia

Lipodystrophy, familial partial, due to AKT2 mutations	1 familia
Mental retardation - progressive spasticity, X-linked	1 familia
Mental retardation, X-linked recessive - macrocephaly - ciliary dysfunction	1 familia
Mental retardation, X-linked, Cabezas type	1 familia
Mental retardation, X-linked, Pai type	1 familia
Muscular dystrophy, limb-girdle, autosomal dominant, type 1F	1 familia
Muscular dystrophy, limb-girdle, autosomal dominant, type 1G	1 familia
Oculodental syndrome rutherford syndrome	1 familia
Oculogastrointestinal muscular dystrophy	1 familia
Pili torti - onychodysplasia	1 familia
Ptosis - strabismus - ectopic pupils	1 familia
Schizophrenia - mental retardation - deafness - retinitis	1 familia
Short stature - pituitary and cerebellar defects - small sella turcica	1 familia
Steroid dehydrogenase deficiency - dental anomalies	1 familia
Tietz syndrome	1 familia
Trichodysplasia - amelogensis imperfecta	1 familia
Ulnar / fibula ray defect - brachydactyly	1 familia
Upington disease	1 familia
Van den Bosch syndrome	1 familia
Woolly hair - hypotrichosis - everted lower lip - outstanding ears	1 familia
Muscular dystrophy, limb-girdle, autosomal dominant, type 1A	1 familia
Fried Syndrome	1 familia
Mental retardation, X-linked - Spastic paraplegia with iron deposits	1 familia