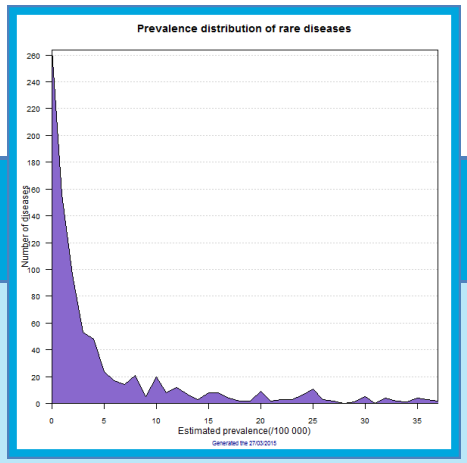


Orphanet Report Series
Rare Diseases collection

Number 1 | January 2019

# Prevalence and incidence of rare diseases: Bibliographic data

Prevalence, incidence or number of published cases listed by diseases (in alphabetical order)

www.orpha.net www.orphadata.org

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# Methodology

Orphanet carries out a systematic survey of literature in order to estimate the prevalence and incidence of rare diseases. This study aims to collect new data regarding point prevalence, birth prevalence and incidence, and to update already published data according to new scientific studies or other available data.

This data is presented in the following reports published biannually:

- Prevalence, incidence or number of published cases listed by diseases (in alphabetical order);
- Diseases listed by decreasing prevalence, incidence or number of published cases;

## Data collection

A number of different sources are used :

- Registries (RARECARE, EUROCAT, etc) ;
- National/international health institutes and agencies (Institut National de Veille Sanitaire (French Institute of Health Surveillance); American Center of Disease Control and Prevention, American National Cancer Institute, European Medicines Agency, World Health Organization etc) ;
- Medline is consulted using the following search algorithm : «Disease names» AND Epidemiology[MeSH:NoExp] OR Incidence[Title/abstract] OR Prevalence[Title/abstract] OR Epidemiology[Title/abstract];
- Medical texts, grey literature and reports from experts;
- Orphanet collaborating experts

## Data characteristics

The data published in this document are worldwide estimations, or European estimations if a worldwide estimation is not available.

The published data is raw collected data or extrapolations of raw data at worldwide or European level when no genetic founder effect is suspected as a cause of a disease.

If a range of national data is available, the average is calculated to estimate the worldwide or European prevalence or incidence.
When a range of data sources is available, the most recent data source that meets a certain number of quality criteria is favoured (registries, meta-analyses, population-based studies, large cohorts studies).

For congenital diseases, the prevalence is estimated, so that:
Prevalence = birth prevalence x (patient life expectancy/general population life expectancy).
When only incidence data is documented, the prevalence is estimated when possible, so that :
Prevalence = incidence x disease mean duration.

When neither prevalence nor incidence data is available, which is the case for very rare diseases, the number of cases or families documented in the medical literature is provided.

## Limitations of the study

The prevalence and incidence data presented in this report are only estimations and cannot be considered to be absolutely correct.
The average values presented in this report do not take into account the heterogeneous nature of the methodologies employed by the studies considered in the literature survey.

The validity and exactitude of raw data sources is taken for granted and have not been verified. Thus, confusion between terms such as incidence and prevalence and/or birth prevalence is possible due to the interchangeable use of these terms in certain sources.
It is possible that prevalence is overestimated in some cases as epidemiological studies are generally based on hospital data in regions with higher prevalence.

## Data presentation

Without specification, published figures are worldwide.
An asterisk * indicates European data.
*P* indicates prevalence data
*I* indicates incidence data.
*BP* indicates birth prevalence

Please note that this is just a selection of Orphanet's rare disease epidemiological data. Currently 5949 rare diseases are annotated with prevalence or incidence

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information in the Orphanet database. To access the complete data sets visit Orphadata (www.orphadata.org).

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*P* indicates prevalence data, *I* indicates incidence data and *BP* indicates birth prevalence

# Prevalence, incidence or number of published cases listed by diseases (in alphabetical order)

| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 79154 | 2-aminoadipic 2-oxoadipic aciduria | | 20 Cases |
| 79157 | 2-methylbutyryl-CoA dehydrogenase deficiency | | 30 Cases |
| 35701 | 3-hydroxy-3-methylglutaryl-CoA synthase deficiency | | 9 Cases |
| 939 | 3-hydroxyisobutyric aciduria | | 13 Cases |
| 6 | 3-methylcrotonyl-CoA carboxylase deficiency | 2.65 *BP* * | |
| 67046 | 3-methylglutaconic aciduria type 1 | | 20 Cases |
| 445038 | 3-methylglutaconic aciduria type 7 | | 22 Cases |
| 505208 | 3-methylglutaconic aciduria type 8 | | 9 Cases |
| 505216 | 3-methylglutaconic aciduria type 9 | | 4 Cases |
| 79351 | 3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form | | 15 Cases |
| 79350 | 3-phosphoserine phosphatase deficiency | | 8 Cases |
| 7 | 3C syndrome | | 25 Cases |
| 2616 | 3M syndrome | | 200 Cases |
| 293843 | 3MC syndrome | | 32 Cases |
| 217064 | 5-fluorouracil poisoning | 2.0 *P* * | |
| 33572 | 5-oxoprolinase deficiency | | 8 Cases |
| 2975 | 46,XX disorder of sex development-skeletal anomalies syndrome | | 2 Cases |
| 444048 | 46,XX ovarian dysgenesis-short stature syndrome | | 3 Cases |
| 2138 | 46,XX ovotesticular disorder of sex development | 2.5 *BP* | |
| 393 | 46,XX testicular disorder of sex development | 2.5 *P* | |
| 753 | 46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency | | 50 Families |
| 90796 | 46,XY disorder of sex development due to isolated 17,20-lyase deficiency | | 15 Cases |
| 443087 | 46,XY disorder of sex development due to testicular 17,20-desmolase deficiency | | 2 Families |
| 168558 | 46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency | | 9 Cases |
| 168563 | 46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome | | 5 Cases |
| 8 | 47,XYY syndrome | 50.0 *BP* * | |
| 96263 | 48,XXXY syndrome | 1.0 *BP* * | |
| 10 | 48,XXYY syndrome | 1.9 *BP* * | |
| 99329 | 48,XYYY syndrome | | 10 Cases |
| 96264 | 49,XXXXY syndrome | 0.55 *BP* * | |
| 261534 | 49,XXXYY syndrome | | 2 Cases |
| 99330 | 49,XYYYY syndrome | | 8 Cases |
| 293948 | 1p21.3 microdeletion syndrome | | 9 Cases |
| 401986 | 1p31p32 microdeletion syndrome | | 5 Cases |
| 456298 | 1p35.2 microdeletion syndrome | | 2 Cases |
| 250994 | 1q21.1 microduplication syndrome | | 46 Cases |
| 238769 | 1q44 microdeletion syndrome | | 100 Cases |
| 363680 | 2p13.2 microdeletion syndrome | | 2 Cases |
| 261349 | 2p15p16.1 microdeletion syndrome | | 11 Cases |
| 163693 | 2p21 microdeletion syndrome | | 7 Cases |
| 369881 | 2p21 microdeletion syndrome without cystinuria | | 2 Cases |
| 228402 | 2q23.1 microdeletion syndrome | | 18 Cases |
| 313947 | 2q23.1 microduplication syndrome | | 2 Cases |
| 1617 | 2q24 microdeletion syndrome | | 23 Cases |
| 251019 | 2q32q33 microdeletion syndrome | | 25 Cases |
| 251028 | 2q33.1 microdeletion syndrome | | 20 Cases |
| 1001 | 2q37 microdeletion syndrome | | 115 Cases |
| 435638 | 3p25.3 microdeletion syndrome | | 8 Cases |
| 1621 | 3q13 microdeletion syndrome | | 42 Cases |
| 96095 | 3q26 microduplication syndrome | | 100 Cases |
| 356947 | 3q26q27 microdeletion syndrome | | 4 Cases |
| 397695 | 3q27.3 microdeletion syndrome | | 7 Cases |
| 238750 | 4q21 microdeletion syndrome | | 14 Cases |
| 502437 | 4q25 proximal deletion syndrome | | 3 Cases |
| 329802 | 5p13 microduplication syndrome | | 7 Cases |
| 228384 | 5q14.3 microdeletion syndrome | | 40 Cases |
| 228415 | 5q35 microduplication syndrome | | 30 Cases |
| 251046 | 6p22 microdeletion syndrome | | 19 Cases |
| 75857 | 6q terminal deletion syndrome | | 19 Cases |
| 171829 | 6q16 deletion syndrome | | 12 Cases |
| 251056 | 6q25 microdeletion syndrome | | 4 Cases |
| 314034 | 7p22.1 microduplication syndrome | | 5 Cases |
| 96121 | 7q11.23 microduplication syndrome | | 163 Cases |
| 251061 | 7q31 microdeletion syndrome | | 20 Cases |
| 96092 | 8p inverted duplication/deletion syndrome | 3.9 *BP* * | |
| 251066 | 8p11.2 deletion syndrome | | 3 Cases |
| 251076 | 8p23.1 duplication syndrome | 1.72 *P* | |

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| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 228399 | 8q12 microduplication syndrome | | 4 Cases |
| 284160 | 8q21.11 microdeletion syndrome | | 13 Cases |
| 178303 | 8q22.1 microdeletion syndrome | | 6 Cases |
| 508488 | 8q24.3 deletion syndrome | | 2 Cases |
| 324313 | 9p13 microdeletion syndrome | | 4 Cases |
| 401923 | 9q31.1q31.3 microdeletion syndrome | | 2 Cases |
| 495818 | 9q33.3q34.11 microdeletion syndrome | | 4 Cases |
| 300305 | 11p15.4 microduplication syndrome | | 1 Family |
| 444002 | 11q22.2q22.3 microdeletion syndrome | | 5 Cases |
| 313884 | 12p12.1 microdeletion syndrome | | 6 Cases |
| 94063 | 12q14 microdeletion syndrome | | 22 Cases |
| 289513 | 12q15q21.1 microdeletion syndrome | | 6 Cases |
| 412035 | 13q12.3 microdeletion syndrome | | 3 Cases |
| 261120 | 14q11.2 microdeletion syndrome | | 3 Cases |
| 261229 | 14q11.2 microduplication syndrome | | 7 Cases |
| 261144 | 14q12 microdeletion syndrome | | 3 Cases |
| 264200 | 14q22q23 microdeletion syndrome | | 5 Cases |
| 401935 | 14q24.1q24.3 microdeletion syndrome | | 3 Cases |
| 488280 | 14q32 duplication syndrome | | 33 Cases |
| 314585 | 15q overgrowth syndrome | | 12 Cases |
| 261183 | 15q11.2 microdeletion syndrome | | 200 Cases |
| 238446 | 15q11q13 microduplication syndrome | | 30 Cases |
| 199318 | 15q13.3 microdeletion syndrome | | 246 Cases |
| 261190 | 15q14 microdeletion syndrome | | 9 Cases |
| 94065 | 15q24 microdeletion syndrome | | 30 Cases |
| 261211 | 16p11.2p12.2 microdeletion syndrome | | 8 Cases |
| 261204 | 16p11.2p12.2 microduplication syndrome | | 7 Cases |
| 485405 | 16p12.1p12.3 triplication syndrome | | 3 Cases |
| 261236 | 16p13.11 microdeletion syndrome | 7.0 *BP* | |
| 261243 | 16p13.11 microduplication syndrome | | 162 Cases |
| 500055 | 16p13.2 microdeletion syndrome | | 6 Cases |
| 96078 | 16p13.3 microduplication syndrome | | 27 Cases |
| 352629 | 16q24.1 microdeletion syndrome | | 42 Cases |
| 261250 | 16q24.3 microdeletion syndrome | | 27 Cases |
| 217385 | 17p13.3 microduplication syndrome | | 50 Cases |
| 97685 | 17q11 microdeletion syndrome | | 170 Cases |
| 139474 | 17q11.2 microduplication syndrome | | 7 Cases |
| 261265 | 17q12 microdeletion syndrome | | 103 Cases |
| 261272 | 17q12 microduplication syndrome | | 118 Cases |
| 363958 | 17q21.31 microdeletion syndrome | 6.25 *P* * | |
| 261279 | 17q23.1q23.2 microdeletion syndrome | | 7 Cases |
| 254346 | 19p13.12 microdeletion syndrome | | 6 Cases |
| 357001 | 19p13.13 microdeletion syndrome | | 7 Cases |
| 447980 | 19p13.3 microduplication syndrome | | 6 Cases |
| 217346 | 19q13.11 microdeletion syndrome | | 12 Cases |
| 261295 | 20p12.3 microdeletion syndrome | | 3 Cases |
| 313781 | 20p13 microdeletion syndrome | | 4 Cases |
| 444051 | 20q11.2 microdeletion syndrome | | 11 Cases |
| 261323 | 21q22.11q22.12 microdeletion syndrome | | 12 Cases |
| 567 | 22q11.2 deletion syndrome | 37.5 *BP* | |
| 439232 | AApoAIV amyloidosis | | 2 Cases |
| 915 | Aarskog-Scott syndrome | 0.5 *BP* * | |
| 916 | Aase-Smith syndrome | | 10 Cases |
| 324723 | ABeta amyloidosis, Arctic type | | 1 Family |
| 100006 | ABeta amyloidosis, Dutch type | | 250 Cases |
| 324708 | ABeta amyloidosis, Iowa type | | 2 Families |
| 324713 | ABeta amyloidosis, Italian type | | 7 Families |
| 324718 | ABetaA21G amyloidosis | | 2 Families |
| 324703 | ABetaL34V amyloidosis | | 1 Family |
| 920 | Ablepharon macrostomia syndrome | | 16 Cases |
| 921 | Abruzzo-Erickson syndrome | | 4 Cases |
| 2310 | Absence deformity of leg-cataract syndrome | | 2 Cases |
| 1658 | Absence of fingerprints-congenital milia syndrome | | 10 Families |
| 980 | Absence of the pulmonary artery | 0.5 *I* * | |
| 3016 | Absent radius-anogenital anomalies syndrome | | 2 Cases |
| 2951 | Absent thumb-short stature-immunodeficiency syndrome | | 3 Cases |
| 3328 | Absent tibia-polydactyly-arachnoid cyst syndrome | | 3 Cases |
| 67043 | Acanthamoeba keratitis | 1.0 *P* * | |
| 90301 | Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome | | 5 Cases |
| 926 | Acatalasemia | 3.2 *P* * | |
| 48818 | Aceruloplasminemia | 0.09 *P* | |
| 929 | Achalasia-microcephaly syndrome | | 7 Cases |

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| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 15 | Achondroplasia | 4.0 *BP* | |
| 49382 | Achromatopsia | 2.7 *P* | |
| 424046 | Acinar cell carcinoma of pancreas | 0.02 *I* * | |
| 40366 | Acitretin/etretinate embryopathy | | 26 Cases |
| 90065 | Acquired aneurysmal subarachnoid hemorrhage | 10.0 *P* * | |
| 91385 | Acquired angioedema | | 200 Cases |
| 46487 | Acquired epidermolysis bullosa | 0.03 *I* * | |
| 79086 | Acquired generalized lipodystrophy | 1.0 *P* * | |
| 73274 | Acquired hemophilia | 0.1 *P* * | |
| 73274 | Acquired hemophilia | 0.08 *I* | |
| 2221 | Acquired hypertrichosis lanuginosa | | 60 Cases |
| 75564 | Acquired idiopathic sideroblastic anemia | 0.09 *I* * | |
| 464453 | Acquired methemoglobinemia | | 242 Cases |
| 91136 | Acquired monoclonal Ig light chain-associated Fanconi syndrome | | 100 Cases |
| 79087 | Acquired partial lipodystrophy | 1.0 *P* * | |
| 228247 | Acquired pseudoxanthoma elasticum | | 20 Cases |
| 99147 | Acquired von Willebrand syndrome | | 300 Cases |
| 158673 | Acral dystrophic epidermolysis bullosa | | 10 Families |
| 263534 | Acral peeling skin syndrome | | 40 Cases |
| 281127 | Acral self-healing collodion baby | | 2 Cases |
| 958 | Acro-renal-mandibular syndrome | | 10 Cases |
| 959 | Acro-renal-ocular syndrome | | 20 Families |
| 36 | Acrocallosal syndrome | | 38 Cases |
| 2008 | Acrocardiofacial syndrome | | 9 Cases |
| 221054 | Acrocephalopolydactyly | | 8 Cases |
| 949 | Acrocraniofacial dysostosis | | 2 Cases |
| 950 | Acrodysostosis | | 80 Cases |
| 280651 | Acrodysostosis with multiple hormone resistance | | 40 Cases |
| 2956 | Acrodysplasia scoliosis | | 2 Cases |
| 1786 | Acrofacial dysostosis, Catania type | | 2 Families |
| 64542 | Acrofacial dysostosis, Kennedy-Teebi type | | 2 Cases |
| 1787 | Acrofacial dysostosis, Palagonia type | | 4 Cases |
| 1788 | Acrofacial dysostosis, Rodríguez type | | 13 Cases |
| 1784 | Acrofrontofacionasal dysostosis | | 12 Cases |
| 965 | Acromegaloid facial appearance syndrome | | 23 Cases |
| 963 | Acromegaly | 5.5 *P* | |
| 963 | Acromegaly | 0.35 *I* | |

| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 39 | Acromelanosis | | 10 Cases |
| 1827 | Acromelic frontonasal dysplasia | | 22 Cases |
| 968 | Acromesomelic dysplasia, Hunter-Thompson type | | 10 Cases |
| 40 | Acromesomelic dysplasia, Maroteaux type | | 50 Cases |
| 969 | Acromicric dysplasia | | 60 Cases |
| 955 | Acroosteolysis dominant type | | 100 Cases |
| 363665 | Acroosteolysis-keloid-like lesions-premature aging syndrome | | 5 Cases |
| 85203 | Acropectoral syndrome | | 25 Cases |
| 957 | Acropectorovertebral dysplasia | | 30 Cases |
| 971 | Acrorenal syndrome | | 20 Cases |
| 99892 | ACTH-dependent Cushing syndrome | 0.55 *I* | |
| 163696 | Action myoclonus-renal failure syndrome | | 38 Cases |
| 397596 | Activated PI3K-delta syndrome | | 18 Cases |
| 284460 | Acute annular outer retinopathy | | 12 Cases |
| 83597 | Acute disseminated encephalomyelitis | 0.6 *I* * | |
| 363549 | Acute encephalopathy with biphasic seizures and late reduced diffusion | | 283 Cases |
| 293173 | Acute generalized exanthematous pustulosis | 0.3 *I* | |
| 217371 | Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins | | 19 Cases |
| 466794 | Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome | | 3 Cases |
| 370088 | Acute infantile liver failure-multisystemic involvement syndrome | | 6 Cases |
| 98916 | Acute inflammatory demyelinating polyradiculoneuropathy | 3.1 *P* * | |
| 79276 | Acute intermittent porphyria | 0.54 *P* * | |
| 79276 | Acute intermittent porphyria | 0.013 *I* * | |
| 79126 | Acute interstitial pneumonia | 3.8 *P* * | |
| 90062 | Acute liver failure | 20.0 *P* * | |
| 178320 | Acute lung injury | 25.0 *I* * | |
| 513 | Acute lymphoblastic leukemia | 11.0 *P* * | |
| 513 | Acute lymphoblastic leukemia | 2.75 *I* * | |
| 488239 | Acute macular neuroretinopathy | | 101 Cases |
| 518 | Acute megakaryoblastic leukemia | 0.02 *I* * | |
| 514 | Acute monoblastic leukemia | 0.13 *I* * | |
| 98834 | Acute myeloblastic leukemia with maturation | 0.02 *I* * | |
| 98833 | Acute myeloblastic leukemia without maturation | 0.01 *I* * | |

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|---|---|---|---|
| 519 | Acute myeloid leukemia | 2.5 *I* | |
| 98832 | Acute myeloid leukemia with minimal differentiation | 0.01 *I* * | |
| 98277 | Acute myeloid leukemia with recurrent genetic anomaly | 0.11 *I* * | |
| 517 | Acute myelomonocytic leukemia | 0.17 *I* * | |
| 86843 | Acute panmyelosis with myelofibrosis | 0.06 *I* * | |
| 90064 | Acute peripheral arterial occlusion | 16.0 *P* * | |
| 520 | Acute promyelocytic leukemia | 0.11 *I* * | |
| 90059 | Acute sensorineural hearing loss by acute acoustic trauma or sudden deafness or surgery induced acoustic trauma | 37.0 *P* * | |
| 139417 | Acute transverse myelitis | 1.6 *I* | |
| 284454 | Acute zonal occult outer retinopathy | | 150 Cases |
| 99901 | Acyl-CoA dehydrogenase 9 deficiency | | 23 Cases |
| 100008 | ACys amyloidosis | | 9 Families |
| 55881 | Adamantinoma | 0.01 *I* * | |
| 974 | Adams-Oliver syndrome | | 398 Cases |
| 85138 | Addison disease | 12.5 *P* * | |
| 2952 | Adducted thumbs-arthrogryposis syndrome, Christian type | | 9 Cases |
| 213504 | Adenocarcinoma of ovary | 5.97 *I* * | |
| 424016 | Adenocarcinoma of the anal canal | 0.26 *I* * | |
| 213772 | Adenocarcinoma of the cervix uteri | 1.01 *I* * | |
| 99976 | Adenocarcinoma of the esophagus | 0.7 *I* | |
| 424991 | Adenocarcinoma of the gallbladder and extrahepatic biliary tract | 2.62 *I* * | |
| 424943 | Adenocarcinoma of the liver and intrahepatic biliary tract | 0.21 *I* * | |
| 104075 | Adenocarcinoma of the small instestine | 0.57 *I* * | |
| 45 | Adenosine monophosphate deaminase deficiency | | 100 Cases |
| 91127 | Adenovirus infection in immunocompromised patients | 18.0 *P* * | |
| 46 | Adenylosuccinate lyase deficiency | | 56 Cases |
| 482601 | Adenylosuccinate synthetase-like 1-related distal myopathy | | 19 Cases |
| 404448 | ADNP syndrome | | 10 Cases |
| 1501 | Adrenocortical carcinoma | 0.75 *P* * | |
| 1501 | Adrenocortical carcinoma | 0.03 *I* * | |
| 977 | Adrenomyodystrophy | | 2 Cases |
| 2666 | Adult familial nephronophthisis-spastic quadriparesia syndrome | | 2 Cases |
| 178487 | Adult intestinal botulism | | 19 Cases |
| 206583 | Adult polyglucosan body disease | | 50 Cases |

| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 978 | ADULT syndrome | | 50 Cases |
| 86875 | Adult T-cell leukemia/lymphoma | 3.0 *P* * | |
| 99027 | Adult-onset autosomal dominant leukodystrophy | | 20 Families |
| 284289 | Adult-onset autosomal recessive cerebellar ataxia | | 14 Cases |
| 255132 | Adult-onset autosomal recessive sideroblastic anemia | | 2 Cases |
| 420492 | Adult-onset cervical dystonia, DYT23 type | | 2 Families |
| 329478 | Adult-onset distal myopathy due to VCP mutation | | 9 Cases |
| 199351 | Adult-onset dystonia-parkinsonism | | 14 Cases |
| 183669 | Agammaglobulinemia | 0.13 *P* * | |
| 83617 | Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome | | 3 Cases |
| 85448 | AGel amyloidosis | | 475 Cases |
| 98850 | Aggressive systemic mastocytosis | 0.33 *P* * | |
| 442582 | AH amyloidosis | | 12 Cases |
| 412069 | AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome | | 4 Cases |
| 250977 | AICA-ribosiduria | | 1 Case |
| 51 | Aicardi-Goutières syndrome | 10.0 *P* * | |
| 90081 | AIDS wasting syndrome | 20.0 *P* * | |
| 79085 | AKT2-related familial partial lipodystrophy | | 1 Family |
| 85443 | AL amyloidosis | 11.0 *P* * | |
| 404454 | Alacrimia-choreoathetosis-liver dysfunction syndrome | | 8 Cases |
| 52 | Alagille syndrome | 0.8 *BP* * | |
| 178333 | Åland Islands eye disease | | 5 Families |
| 2007 | Alar cartilages hypoplasia-coloboma-telecanthus syndrome | | 2 Cases |
| 53 | Albers-Schönberg osteopetrosis | 1.0 *P* | |
| 998 | Albinism-deafness syndrome | | 1 Family |
| 35664 | ALDH18A1-related De Barsy syndrome | | 32 Cases |
| 79327 | ALG1-CDG | | 57 Cases |
| 79326 | ALG2-CDG | | 1 Case |
| 79321 | ALG3-CDG | | 15 Cases |
| 79320 | ALG6-CDG | | 54 Cases |
| 79325 | ALG8-CDG | | 15 Cases |
| 79328 | ALG9-CDG | | 12 Cases |
| 280071 | ALG11-CDG | | 8 Cases |
| 79324 | ALG12-CDG | | 11 Cases |
| 324422 | ALG13-CDG | | 1 Case |

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|---|---|---|---|
| 502444 | Alkaline ceramidase 3 deficiency | | 2 Cases |
| 59 | Allan-Herndon-Dudley syndrome | | 320 Cases |
| 1006 | Alopecia antibody deficiency | | 3 Cases |
| 700 | Alopecia totalis | 10.5 *P* * | |
| 701 | Alopecia universalis | 25.0 *P* * | |
| 1005 | Alopecia-contractures-dwarfism-intellectual disability syndrome | | 5 Cases |
| 1008 | Alopecia-epilepsy-pyorrhea-intellectual disability syndrome | | 12 Cases |
| 2850 | Alopecia-intellectual disability syndrome | | 15 Families |
| 1014 | Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome | | 2 Cases |
| 726 | Alpers-Huttenlocher syndrome | 0.07 *P* * | |
| 726 | Alpers-Huttenlocher syndrome | 0.7 *BP* * | |
| 60 | Alpha-1-antitrypsin deficiency | 20.0 *P* * | |
| 399058 | Alpha-B crystallin-related late-onset myopathy | | 17 Cases |
| 100025 | Alpha-heavy chain disease | | 400 Cases |
| 61 | Alpha-mannosidosis | 0.1 *P* * | |
| 3137 | Alpha-N-acetylgalactosaminidase deficiency | | 20 Cases |
| 79279 | Alpha-N-acetylgalactosaminidase deficiency type 1 | | 10 Cases |
| 79280 | Alpha-N-acetylgalactosaminidase deficiency type 2 | | 10 Cases |
| 79281 | Alpha-N-acetylgalactosaminidase deficiency type 3 | | 10 Cases |
| 98791 | Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16 | | 20 Cases |
| 231401 | Alpha-thalassemia-myelodysplastic syndrome | | 80 Cases |
| 847 | Alpha-thalassemia-X-linked intellectual disability syndrome | | 200 Cases |
| 63 | Alport syndrome | 2.0 *P* * | |
| 86818 | Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome | | 2 Families |
| 64 | Alström syndrome | | 950 Cases |
| 284 | Alveolar echinococcosis | 0.16 *I* * | |
| 169095 | Alymphoid cystic thymic dysgenesis | | 9 Cases |
| 93561 | ALys amyloidosis | | 7 Families |
| 1021 | Amaurosis-hypertrichosis syndrome | | 2 Cases |
| 314422 | Ameloblastic carcinoma | | 40 Cases |
| 1028 | Ameloonychohypohidrotic syndrome | | 2 Cases |
| 1908 | Aminopterin/methotrexate embryofetopathy | | 17 Cases |
| 319635 | Amyloidosis cutis dyschromia | | 27 Cases |
| 803 | Amyotrophic lateral sclerosis | 3.85 *P* | |
| 803 | Amyotrophic lateral sclerosis | 1.35 *I* | |
| 357043 | Amyotrophic lateral sclerosis type 4 | | 70 Cases |
| 228113 | Anal fistula | 23.0 *P* * | |
| 98841 | Anaplastic large cell lymphoma | 2.0 *P* * | |
| 251630 | Anaplastic oligodendroglioma | 0.09 *I* * | |
| 142 | Anaplastic thyroid carcinoma | 0.1 *P* * | |
| 142 | Anaplastic thyroid carcinoma | 0.17 *I* * | |
| 93347 | Anauxetic dysplasia | | 10 Cases |
| 37553 | Andersen-Tawil syndrome | 0.1 *I* * | |
| 157954 | ANE syndrome | | 5 Cases |
| 284984 | Aneurysm-osteoarthritis syndrome | | 45 Cases |
| 63442 | Angel-shaped phalango-epiphyseal dysplasia | | 20 Cases |
| 72 | Angelman syndrome | 7.5 *P* | |
| 72 | Angelman syndrome | 1.3 *BP* * | |
| 251671 | Angiocentric glioma | | 52 Cases |
| 2346 | Angioosteohypertrophic syndrome | 0.8 *BP* * | |
| 370039 | Angora hair nevus | | 2 Cases |
| 69088 | Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome | | 2 Cases |
| 1069 | Aniridia-absent patella syndrome | | 3 Cases |
| 1065 | Aniridia-cerebellar ataxia-intellectual disability syndrome | | 22 Families |
| 1068 | Aniridia-intellectual disability syndrome | | 2 Cases |
| 1067 | Aniridia-ptosis-intellectual disability-familial obesity syndrome | | 3 Cases |
| 1064 | Aniridia-renal agenesis-psychomotor retardation syndrome | | 2 Cases |
| 1070 | Anisakiasis | 0.32 *I* | |
| 356996 | ANK3-related intellectual disability-sleep disturbance syndrome | | 3 Cases |
| 1074 | Ankyloblepharon filiforme adnatum-imperforate anus syndrome | | 3 Families |
| 2206 | Ankylosing vertebral hyperostosis with tylosis | | 8 Cases |
| 254411 | Annular atrophic lichen planus | | 10 Cases |
| 281139 | Annular epidermolytic ichthyosis | | 7 Families |
| 675 | Annular pancreas | 1.8 *BP* * | |
| 69125 | Anonychia with flexural pigmentation | | 3 Cases |
| 1094 | Anonychia-microcephaly syndrome | | 4 Cases |
| 90390 | Anonychia-onychodystrophy syndrome | | 14 Cases |
| 1104 | Anophthalmia plus syndrome | | 17 Cases |

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|---|---|---|---|
| 1101 | Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome | | 3 Cases |
| 77298 | Anophthalmia/microphthalmia-esophageal atresia syndrome | | 30 Cases |
| 93976 | Anotia | 0.028 *BP* * | |
| 2987 | Antecubital pterygium syndrome | | 11 Cases |
| 90079 | Anthracycline extravasation | 0.3 *P* * | |
| 375 | Anti-glomerular basement membrane disease | 0.08 *I* * | |
| 454710 | Anti-p200 pemphigoid | | 50 Cases |
| 81 | Antisynthetase syndrome | 3.5 *P* | |
| 83 | Antley-Bixler syndrome | | 34 Cases |
| 1457 | Aorta coarctation | 35.6 *BP* * | |
| 1110 | Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome | | 4 Cases |
| 2299 | Aortic arch interruption | 0.3 *BP* * | |
| 3400 | Aorto-ventricular tunnel | | 130 Cases |
| 1112 | Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome | | 3 Cases |
| 1113 | Aphalangy-syndactyly-microcephaly syndrome | | 5 Cases |
| 324540 | Aphonia-deafness-retinal dystrophy-bifid halluces-intellectual disability syndrome | | 2 Cases |
| 1114 | Aplasia cutis congenita | 10.0 *BP* | |
| 1116 | Aplasia cutis congenita-intestinal lymphangiectasia syndrome | | 3 Cases |
| 1117 | Aplasia cutis-myopia syndrome | | 4 Cases |
| 99981 | Apnea of prematurity | 8.5 *P* * | |
| 425 | Apolipoprotein A-I deficiency | | 30 Families |
| 1126 | Aprosencephaly cerebellar dysgenesis | | 2 Cases |
| 1129 | Arachnodactyly-abnormal ossification-intellectual disability syndrome | | 5 Cases |
| 1130 | Arachnodactyly-intellectual disability-dysmorphism syndrome | | 3 Cases |
| 1133 | AREDYLD syndrome | | 3 Cases |
| 23 | Argininosuccinic aciduria | 1.0 *P* * | |
| 23 | Argininosuccinic aciduria | 0.46 *BP* | |
| 91 | Aromatase deficiency | | 38 Cases |
| 178345 | Aromatase excess syndrome | | 30 Cases |
| 35708 | Aromatic L-amino acid decarboxylase deficiency | | 100 Cases |
| 1135 | Arrhinia-choanal atresia-microphthalmia syndrome | | 4 Cases |
| 247 | Arrhythmogenic right ventricular cardiomyopathy | 20.0 *P* | |
| 1682 | Arterial dissection-lentiginosis syndrome | | 4 Cases |
| 3342 | Arterial tortuosity syndrome | | 102 Cases |
| 1037 | Arthrogryposis multiplex congenita | 5.7 *BP* * | |
| 1150 | Arthrogryposis multiplex congenita-whistling face syndrome | | 10 Cases |
| 3200 | Arthrogryposis-ectodermal dysplasia-other anomalies syndrome | | 2 Cases |
| 1485 | Arthrogryposis-hyperkeratosis syndrome, lethal form | | 2 Cases |
| 1144 | Arthrogryposis-like hand anomaly-sensorineural deafness syndrome | | 1 Family |
| 2697 | Arthrogryposis-renal dysfunction-cholestasis syndrome | | 100 Cases |
| 65720 | Arthrogryposis-severe scoliosis syndrome | | 2 Families |
| 1253 | Ascher syndrome | | 50 Cases |
| 137686 | Asherman syndrome | 44.0 *P* * | |
| 85175 | Astley-Kendall dysplasia | | 5 Cases |
| 251679 | Astroblastoma | 0.02 *I* * | |
| 94 | Astrocytoma | 2.5 *P* * | |
| 94 | Astrocytoma | 4.8 *I* * | |
| 96 | Ataxia with vitamin E deficiency | 0.33 *P* * | |
| 1188 | Ataxia-deafness-intellectual disability syndrome | | 8 Cases |
| 370022 | Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome | | 7 Cases |
| 459033 | Ataxia-oculomotor apraxia type 4 | | 12 Cases |
| 1184 | Ataxia-photosensitivity-short stature syndrome | | 2 Cases |
| 100 | Ataxia-telangiectasia | 0.49 *P* * | |
| 1190 | Atelosteogenesis type I | | 12 Cases |
| 56304 | Atelosteogenesis type II | | 25 Cases |
| 56305 | Atelosteogenesis type III | | 25 Cases |
| 69739 | Athabaskan brainstem dysgenesis syndrome | | 13 Cases |
| 1192 | Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome | | 2 Cases |
| 95713 | Athyreosis | 3.5 *P* * | |
| 1193 | Atkin-Flaitz syndrome | | 14 Cases |
| 163934 | Atopic keratoconjunctivitis | 15.0 *P* * | |
| 314632 | ATP13A2-related juvenile neuronal ceroid lipofuscinosis | | 4 Cases |
| 1201 | Atresia of small intestine | 16.0 *BP* * | |
| 1479 | Atrial septal defect-atrioventricular conduction defects syndrome | | 11 Cases |
| 1352 | Atrioventricular defect-blepharophimosis-radial and anal defect syndrome | | 2 Cases |

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|---|---|---|---|
| 352723 | Attenuated Chédiak-Higashi syndrome | | 100 Cases |
| 1456 | Atypical coarctation of aorta | 0.17 *BP* * | |
| 314721 | Atypical dentin dysplasia due to SMOC2 deficiency | | 4 Cases |
| 289863 | Atypical glycine encephalopathy | | 20 Cases |
| 2134 | Atypical hemolytic-uremic syndrome | 1.0 *P* * | |
| 357008 | Atypical hemolytic-uremic syndrome with DGKE deficiency | | 13 Cases |
| 238523 | Atypical hypotonia-cystinuria syndrome | | 2 Cases |
| 391411 | Atypical juvenile parkinsonism | | 6 Families |
| 86797 | Atypical lichen myxedematosus | | 20 Cases |
| 314466 | Atypical Meigs syndrome | | 9 Cases |
| 77300 | Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome | | 2 Cases |
| 137888 | Auriculocondylar syndrome | | 50 Cases |
| 114 | Auriculoosteodysplasia | | 2 Families |
| 352490 | Autism spectrum disorder due to AUTS2 deficiency | | 60 Cases |
| 370943 | Autism spectrum disorder-epilepsy-arthrogryposis syndrome | | 8 Cases |
| 308410 | Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency | | 5 Families |
| 137911 | Autism-facial port-wine stain syndrome | | 4 Cases |
| 324636 | Autoerythrocyte sensitization syndrome | | 170 Cases |
| 420789 | Autoimmune encephalopathy with parasomnia and obstructive sleep apnea | | 10 Cases |
| 391487 | Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome | | 5 Cases |
| 98375 | Autoimmune hemolytic anemia | 2.02 *I* * | |
| 444463 | Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome | | 6 Cases |
| 2137 | Autoimmune hepatitis | 23.5 *P* | |
| 2137 | Autoimmune hepatitis | 1.2 *I* | |
| 444092 | Autoimmune interstitial lung disease-arthritis syndrome | | 5 Families |
| 3261 | Autoimmune lymphoproliferative syndrome | | 500 Cases |
| 436159 | Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency | | 17 Cases |
| 275517 | Autoimmune lymphoproliferative syndrome with recurrent viral infections | | 1 Family |
| 747 | Autoimmune pulmonary alveolar proteinosis | 0.5 *P* | |
| 747 | Autoimmune pulmonary alveolar proteinosis | 0.04 *I* | |
| 324530 | Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation | | 2 Cases |
| 329173 | Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis | | 3 Cases |
| 33110 | Autosomal agammaglobulinemia | | 100 Cases |
| 209335 | Autosomal dominant adult-onset proximal spinal muscular atrophy | 0.1 *P* * | |
| 314399 | Autosomal dominant aplasia and myelodysplasia | | 6 Cases |
| 99 | Autosomal dominant cerebellar ataxia | 2.7 *P* | |
| 314404 | Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome | | 24 Cases |
| 487814 | Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation | | 2 Cases |
| 435819 | Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation | | 2 Cases |
| 401964 | Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons | | 2 Families |
| 99946 | Autosomal dominant Charcot-Marie-Tooth disease type 2A1 | | 1 Family |
| 99938 | Autosomal dominant Charcot-Marie-Tooth disease type 2D | | 44 Cases |
| 521414 | Autosomal dominant Charcot-Marie-Tooth disease type 2DD | | 51 Cases |
| 99940 | Autosomal dominant Charcot-Marie-Tooth disease type 2F | | 5 Families |
| 99941 | Autosomal dominant Charcot-Marie-Tooth disease type 2G | | 1 Family |
| 99944 | Autosomal dominant Charcot-Marie-Tooth disease type 2K | | 30 Cases |
| 99945 | Autosomal dominant Charcot-Marie-Tooth disease type 2L | | 1 Family |
| 228179 | Autosomal dominant Charcot-Marie-Tooth disease type 2M | | 20 Cases |
| 228174 | Autosomal dominant Charcot-Marie-Tooth disease type 2N | | 28 Cases |
| 329258 | Autosomal dominant Charcot-Marie-Tooth disease type 2Q | | 8 Cases |
| 397735 | Autosomal dominant Charcot-Marie-Tooth disease type 2U | | 2 Cases |
| 447964 | Autosomal dominant Charcot-Marie-Tooth disease type 2V | | 21 Cases |
| 488333 | Autosomal dominant Charcot-Marie-Tooth disease type 2W | | 24 Cases |
| 435387 | Autosomal dominant Charcot-Marie-Tooth disease type 2Y | | 7 Cases |
| 466768 | Autosomal dominant Charcot-Marie-Tooth disease type 2Z | | 21 Cases |

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|---|---|---|---|
| 90348 | Autosomal dominant cutis laxa | | 50 Cases |
| 79499 | Autosomal dominant deafness-onychodystrophy syndrome | | 22 Cases |
| 476093 | Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome | | 8 Cases |
| 329466 | Autosomal dominant focal dystonia, DYT25 type | | 28 Cases |
| 402003 | Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering | | 21 Cases |
| 2314 | Autosomal dominant hyper-IgE syndrome | 0.1 *I* * | |
| 1810 | Autosomal dominant hypohidrotic ectodermal dysplasia | | 40 Cases |
| 89937 | Autosomal dominant hypophosphatemic rickets | | 100 Cases |
| 457193 | Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome | | 16 Cases |
| 100043 | Autosomal dominant intermediate Charcot-Marie-Tooth disease type A | | 20 Cases |
| 100044 | Autosomal dominant intermediate Charcot-Marie-Tooth disease type B | | 37 Cases |
| 100045 | Autosomal dominant intermediate Charcot-Marie-Tooth disease type C | | 35 Cases |
| 100046 | Autosomal dominant intermediate Charcot-Marie-Tooth disease type D | | 12 Cases |
| 93114 | Autosomal dominant intermediate Charcot-Marie-Tooth disease type E | | 21 Cases |
| 352670 | Autosomal dominant intermediate Charcot-Marie-Tooth disease type F | | 8 Cases |
| 324585 | Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain | | 9 Cases |
| 266 | Autosomal dominant limb-girdle muscular dystrophy type 1A | | 4 Families |
| 34516 | Autosomal dominant limb-girdle muscular dystrophy type 1D | | 6 Families |
| 34517 | Autosomal dominant limb-girdle muscular dystrophy type 1E | | 20 Families |
| 55595 | Autosomal dominant limb-girdle muscular dystrophy type 1F | | 64 Cases |
| 55596 | Autosomal dominant limb-girdle muscular dystrophy type 1G | | 2 Families |
| 238755 | Autosomal dominant limb-girdle muscular dystrophy type 1H | | 11 Cases |
| 140957 | Autosomal dominant macrothrombocytopenia | | 100 Cases |
| 319581 | Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency | | 68 Cases |
| 319589 | Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency | | 2 Cases |
| 457050 | Autosomal dominant mitochondrial myopathy with exercise intolerance | | 15 Cases |
| 65743 | Autosomal dominant multiple pterygium syndrome | | 4 Cases |
| 99846 | Autosomal dominant myoglobinuria | | 2 Families |
| 440354 | Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome | | 1 Family |
| 329211 | Autosomal dominant neovascular inflammatory vitreoretinopathy | | 99 Cases |
| 98784 | Autosomal dominant nocturnal frontal lobe epilepsy | | 100 Families |
| 98672 | Autosomal dominant optic atrophy | 3.3 *P* | |
| 67036 | Autosomal dominant optic atrophy and cataract | | 3 Families |
| 1215 | Autosomal dominant optic atrophy plus syndrome | 0.4 *P* * | |
| 98673 | Autosomal dominant optic atrophy, classic form | 2.0 *P* | |
| 2783 | Autosomal dominant osteopetrosis type 1 | | 33 Cases |
| 1010 | Autosomal dominant palmoplantar keratoderma and congenital alopecia | | 10 Cases |
| 730 | Autosomal dominant polycystic kidney disease | 3.96 *P* * | |
| 88924 | Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis | | 30 Cases |
| 1300 | Autosomal dominant popliteal pterygium syndrome | 0.3 *P* | |
| 476119 | Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome | | 1 Family |
| 34528 | Autosomal dominant primary hypomagnesemia with hypocalciuria | | 28 Cases |
| 88659 | Autosomal dominant progressive nephropathy with hypertension | | 14 Cases |
| 314889 | Autosomal dominant proximal renal tubular acidosis | | 1 Family |
| 209867 | Autosomal dominant rhegmatogenous retinal detachment | | 38 Cases |
| 3107 | Autosomal dominant Robinow syndrome | | 100 Cases |
| 140481 | Autosomal dominant slowed nerve conduction velocity | | 1 Family |
| 251282 | Autosomal dominant spastic ataxia type 1 | | 53 Cases |
| 100988 | Autosomal dominant spastic paraplegia type 6 | | 10 Families |
| 100989 | Autosomal dominant spastic paraplegia type 8 | | 10 Families |
| 447753 | Autosomal dominant spastic paraplegia type 9A | | 2 Families |
| 447757 | Autosomal dominant spastic paraplegia type 9B | | 3 Families |

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|---|---|---|---|
| 100991 | Autosomal dominant spastic paraplegia type 10 | | 10 Families |
| 100993 | Autosomal dominant spastic paraplegia type 12 | | 27 Cases |
| 100994 | Autosomal dominant spastic paraplegia type 13 | | 10 Cases |
| 100998 | Autosomal dominant spastic paraplegia type 17 | | 20 Families |
| 100999 | Autosomal dominant spastic paraplegia type 19 | | 1 Family |
| 101009 | Autosomal dominant spastic paraplegia type 29 | | 1 Family |
| 320365 | Autosomal dominant spastic paraplegia type 36 | | 1 Family |
| 171612 | Autosomal dominant spastic paraplegia type 37 | | 13 Cases |
| 171617 | Autosomal dominant spastic paraplegia type 38 | | 1 Family |
| 320355 | Autosomal dominant spastic paraplegia type 41 | | 7 Cases |
| 171863 | Autosomal dominant spastic paraplegia type 42 | | 1 Family |
| 444099 | Autosomal dominant spastic paraplegia type 73 | | 1 Family |
| 228169 | Autosomal dominant striatal neurodegeneration | | 11 Cases |
| 466806 | Autosomal dominant thrombocytopenia with platelet secretion defect | | 4 Families |
| 34149 | Autosomal dominant tubulointerstitial kidney disease | 0.11 *P* * | |
| 3086 | Autosomal dominant vitreoretinochoroidopathy | | 3 Cases |
| 79278 | Autosomal erythropoietic protoporphyria | 0.92 *P* * | |
| 79278 | Autosomal erythropoietic protoporphyria | 0.012 *I* * | |
| 1027 | Autosomal recessive amelia | | 3 Cases |
| 247815 | Autosomal recessive ataxia due to PEX10 deficiency | | 6 Cases |
| 139485 | Autosomal recessive ataxia due to ubiquinone deficiency | | 31 Cases |
| 88644 | Autosomal recessive ataxia, Beauce type | | 57 Cases |
| 521411 | Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect | | 2 Cases |
| 324442 | Autosomal recessive axonal neuropathy with neuromyotonia | | 33 Families |
| 139455 | Autosomal recessive bestrophinopathy | | 20 Cases |
| 448242 | Autosomal recessive brachyolmia | | 20 Cases |
| 1172 | Autosomal recessive cerebellar ataxia | 3.3 *P* | |
| 453521 | Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency | | 2 Cases |
| 412057 | Autosomal recessive cerebellar ataxia due to STUB1 deficiency | | 10 Families |

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|---|---|---|---|
| 352641 | Autosomal recessive cerebellar ataxia with late-onset spasticity | | 10 Cases |
| 404499 | Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency | | 2 Cases |
| 404493 | Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency | | 3 Cases |
| 363429 | Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome | | 17 Cases |
| 95434 | Autosomal recessive cerebellar ataxia-saccadic intrusion syndrome | | 1 Family |
| 363969 | Autosomal recessive cerebral atrophy | | 4 Cases |
| 466775 | Autosomal recessive Charcot-Marie-Tooth disease type 2X | | 29 Cases |
| 506353 | Autosomal recessive complex spastic paraplegia due to Kennedy pathway dysfunction | | 4 Cases |
| 363432 | Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency | | 7 Cases |
| 324262 | Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency | | 10 Cases |
| 281097 | Autosomal recessive congenital ichthyosis | 0.5 *P* * | |
| 90349 | Autosomal recessive cutis laxa type 1 | | 60 Cases |
| 90350 | Autosomal recessive cutis laxa type 2 | | 40 Cases |
| 101150 | Autosomal recessive dopa-responsive dystonia | | 50 Cases |
| 1974 | Autosomal recessive faciodigitogenital syndrome | | 26 Cases |
| 329329 | Autosomal recessive frontotemporal pachygyria | | 7 Cases |
| 300547 | Autosomal recessive infantile hypercalcemia | | 12 Cases |
| 217055 | Autosomal recessive intermediate Charcot-Marie-Tooth disease type A | | 8 Families |
| 254334 | Autosomal recessive intermediate Charcot-Marie-Tooth disease type B | | 1 Case |
| 369867 | Autosomal recessive intermediate Charcot-Marie-Tooth disease type C | | 3 Cases |
| 435998 | Autosomal recessive intermediate Charcot-Marie-Tooth disease type D | | 4 Cases |
| 98676 | Autosomal recessive isolated optic atrophy | | 5 Cases |
| 314572 | Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome | | 3 Cases |
| 267 | Autosomal recessive limb-girdle muscular dystrophy type 2A | 1.0 *P* * | |
| 353 | Autosomal recessive limb-girdle muscular dystrophy type 2C | 0.2 *P* * | |

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|---|---|---|---|
| 119 | Autosomal recessive limb-girdle muscular dystrophy type 2E | 0.1 *P* * | |
| 219 | Autosomal recessive limb-girdle muscular dystrophy type 2F | 0.3 *P* * | |
| 34514 | Autosomal recessive limb-girdle muscular dystrophy type 2G | | 16 Cases |
| 34515 | Autosomal recessive limb-girdle muscular dystrophy type 2I | 1.0 *P* * | |
| 140922 | Autosomal recessive limb-girdle muscular dystrophy type 2J | | 1 Family |
| 206554 | Autosomal recessive limb-girdle muscular dystrophy type 2M | | 5 Families |
| 206559 | Autosomal recessive limb-girdle muscular dystrophy type 2N | | 1 Case |
| 206564 | Autosomal recessive limb-girdle muscular dystrophy type 2O | | 2 Cases |
| 280333 | Autosomal recessive limb-girdle muscular dystrophy type 2P | | 1 Case |
| 254361 | Autosomal recessive limb-girdle muscular dystrophy type 2Q | | 6 Cases |
| 363543 | Autosomal recessive limb-girdle muscular dystrophy type 2R | | 2 Cases |
| 369840 | Autosomal recessive limb-girdle muscular dystrophy type 2S | | 3 Cases |
| 363623 | Autosomal recessive limb-girdle muscular dystrophy type 2T | | 2 Cases |
| 352479 | Autosomal recessive limb-girdle muscular dystrophy type 2U | | 8 Cases |
| 466801 | Autosomal recessive limb-girdle muscular dystrophy type 2W | | 2 Cases |
| 476084 | Autosomal recessive limb-girdle muscular dystrophy type 2X | | 3 Cases |
| 424261 | Autosomal recessive limb-girdle muscular dystrophy type 2Y | | 3 Cases |
| 480682 | Autosomal recessive limb-girdle muscular dystrophy type 2Z | | 4 Cases |
| 206580 | Autosomal recessive lower motor neuron disease with childhood onset | | 5 Cases |
| 238505 | Autosomal recessive lymphoproliferative disease | | 17 Cases |
| 667 | Autosomal recessive malignant osteopetrosis | 0.75 *BP* * | |
| 477857 | Autosomal recessive mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency | | 7 Cases |
| 319569 | Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency | | 18 Cases |
| 319574 | Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency | | 6 Cases |
| 319332 | Autosomal recessive myogenic arthrogryposis multiplex congenita | | 1 Family |
| 280654 | Autosomal recessive nail dysplasia | | 4 Cases |
| 93329 | Autosomal recessive omodysplasia | | 23 Cases |
| 227976 | Autosomal recessive optic atrophy, OPA7 type | | 17 Cases |
| 1366 | Autosomal recessive palmoplantar keratoderma and congenital alopecia | | 8 Cases |
| 731 | Autosomal recessive polycystic kidney disease | 1.17 *P* * | |
| 437552 | Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity | | 3 Cases |
| 1507 | Autosomal recessive Robinow syndrome | | 100 Cases |
| 420702 | Autosomal recessive severe congenital neutropenia due to CSF3R deficiency | | 4 Cases |
| 420699 | Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency | | 2 Cases |
| 331176 | Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency | | 57 Cases |
| 423384 | Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency | | 14 Cases |
| 314603 | Autosomal recessive spastic ataxia with leukoencephalopathy | | 54 Cases |
| 254343 | Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome | | 6 Cases |
| 447760 | Autosomal recessive spastic paraplegia type 9B | | 2 Families |
| 100995 | Autosomal recessive spastic paraplegia type 14 | | 1 Family |
| 100996 | Autosomal recessive spastic paraplegia type 15 | | 10 Families |
| 209951 | Autosomal recessive spastic paraplegia type 18 | | 9 Cases |
| 101000 | Autosomal recessive spastic paraplegia type 20 | | 36 Cases |
| 101001 | Autosomal recessive spastic paraplegia type 21 | | 35 Cases |
| 101003 | Autosomal recessive spastic paraplegia type 23 | | 5 Families |
| 101004 | Autosomal recessive spastic paraplegia type 24 | | 1 Family |
| 101005 | Autosomal recessive spastic paraplegia type 25 | | 1 Family |
| 101006 | Autosomal recessive spastic paraplegia type 26 | | 10 Families |
| 101007 | Autosomal recessive spastic paraplegia type 27 | | 10 Cases |
| 101008 | Autosomal recessive spastic paraplegia type 28 | | 7 Cases |
| 171622 | Autosomal recessive spastic paraplegia type 32 | | 1 Family |
| 171629 | Autosomal recessive spastic paraplegia type 35 | | 38 Cases |
| 139480 | Autosomal recessive spastic paraplegia type 39 | | 2 Families |

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|---|---|---|---|
| 320370 | Autosomal recessive spastic paraplegia type 43 | | 2 Cases |
| 320401 | Autosomal recessive spastic paraplegia type 44 | | 3 Cases |
| 320396 | Autosomal recessive spastic paraplegia type 45 | | 7 Families |
| 320391 | Autosomal recessive spastic paraplegia type 46 | | 5 Cases |
| 306511 | Autosomal recessive spastic paraplegia type 48 | | 2 Cases |
| 319199 | Autosomal recessive spastic paraplegia type 53 | | 9 Cases |
| 320380 | Autosomal recessive spastic paraplegia type 54 | | 6 Families |
| 320375 | Autosomal recessive spastic paraplegia type 55 | | 14 Cases |
| 320411 | Autosomal recessive spastic paraplegia type 56 | | 5 Families |
| 431329 | Autosomal recessive spastic paraplegia type 57 | | 2 Cases |
| 401795 | Autosomal recessive spastic paraplegia type 59 | | 3 Cases |
| 401800 | Autosomal recessive spastic paraplegia type 60 | | 1 Case |
| 401780 | Autosomal recessive spastic paraplegia type 61 | | 4 Cases |
| 401785 | Autosomal recessive spastic paraplegia type 62 | | 7 Cases |
| 401805 | Autosomal recessive spastic paraplegia type 63 | | 2 Cases |
| 401810 | Autosomal recessive spastic paraplegia type 64 | | 4 Cases |
| 401815 | Autosomal recessive spastic paraplegia type 66 | | 2 Cases |
| 401820 | Autosomal recessive spastic paraplegia type 67 | | 2 Cases |
| 401830 | Autosomal recessive spastic paraplegia type 69 | | 2 Cases |
| 401835 | Autosomal recessive spastic paraplegia type 70 | | 4 Cases |
| 401840 | Autosomal recessive spastic paraplegia type 71 | | 1 Case |
| 468661 | Autosomal recessive spastic paraplegia type 74 | | 11 Cases |
| 459056 | Autosomal recessive spastic paraplegia type 75 | | 5 Cases |
| 488594 | Autosomal recessive spastic paraplegia type 76 | | 7 Families |
| 466722 | Autosomal recessive spastic paraplegia type 77 | | 8 Cases |
| 513436 | Autosomal recessive spastic paraplegia type 78 | | 7 Cases |
| 95433 | Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome | | 3 Families |
| 401979 | Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type | | 4 Cases |
| 250984 | Autosomal recessive Stickler syndrome | | 15 Cases |
| 280365 | Autosomal semi-dominant severe lipodystrophic laminopathy | | 7 Cases |
| 101010 | Autosomal spastic paraplegia type 30 | | 3 Families |
| 397946 | Autosomal spastic paraplegia type 58 | | 19 Cases |
| 401849 | Autosomal spastic paraplegia type 72 | | 14 Cases |
| 300345 | Autosomal systemic lupus erythematosus | | 7 Families |
| 454836 | Avian influenza | | 826 Cases |
| 782 | Axenfeld-Rieger syndrome | 0.5 *P* * | |
| 168549 | Axial spondylometaphyseal dysplasia | | 13 Cases |
| 401911 | AXIN2-related attenuated familial adenomatous polyposis | | 4 Families |
| 1272 | Aymé-Gripp syndrome | | 18 Cases |
| 79332 | B4GALT1-CDG | | 1 Case |
| 67038 | B-cell chronic lymphocytic leukemia | 48.0 *P* * | |
| 171915 | B-cell non-Hodgkin lymphoma | 17.45 *I* * | |
| 86852 | B-cell prolymphocytic leukemia | 0.05 *I* * | |
| 36234 | Bacterial toxic-shock syndrome | 3.0 *P* | |
| 93395 | Ballard syndrome | | 12 Cases |
| 1225 | Baller-Gerold syndrome | | 40 Cases |
| 1226 | Bamforth-Lazarus syndrome | | 8 Cases |
| 1227 | Bangstad syndrome | | 2 Cases |
| 1228 | Banki syndrome | | 1 Family |
| 2995 | Baraitser-Winter cerebrofrontofacial syndrome | | 60 Cases |
| 1231 | Barber-Say syndrome | | 16 Cases |
| 110 | Bardet-Biedl syndrome | 0.7 *P* * | |
| 110 | Bardet-Biedl syndrome | 0.5 *BP* * | |
| 111 | Barth syndrome | 0.22 *P* * | |
| 1234 | Bartsocas-Papas syndrome | | 24 Cases |
| 112 | Bartter syndrome | 0.1 *I* * | |
| 100976 | Bathing suit ichthyosis | | 20 Cases |
| 166113 | Bazex syndrome | | 145 Cases |
| 113 | Bazex-Dupré-Christol syndrome | | 143 Cases |
| 98895 | Becker muscular dystrophy | 1.53 *P* | |
| 98895 | Becker muscular dystrophy | 2.2 *BP* * | |
| 116 | Beckwith-Wiedemann syndrome | 3.5 *BP* * | |
| 1237 | Beemer-Ertbruggen syndrome | | 2 Cases |
| 1241 | Bencze syndrome | | 2 Families |
| 251287 | Benign concentric annular macular dystrophy | | 27 Cases |

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|---|---|---|---|
| 1949 | Benign familial neonatal epilepsy | | 100 Families |
| 140927 | Benign familial neonatal-infantile seizures | | 10 Families |
| 166308 | Benign infantile focal epilepsy with midline spikes and waves during sleep | | 36 Cases |
| 166305 | Benign infantile seizures associated with mild gastroenteritis | | 100 Cases |
| 209973 | Benign nocturnal alternating hemiplegia of childhood | | 12 Cases |
| 1179 | Benign paroxysmal tonic upgaze of childhood with ataxia | | 12 Cases |
| 71518 | Benign paroxysmal torticollis of infancy | | 50 Cases |
| 324581 | Benign Samaritan congenital myopathy | | 4 Cases |
| 252164 | Benign schwannoma | 6.0 *P* * | |
| 464336 | BENTA disease | | 8 Cases |
| 528 | Berardinelli-Seip congenital lipodystrophy | 0.5 *P* * | |
| 274 | Bernard-Soulier syndrome | | 100 Cases |
| 118 | Beta-mannosidosis | 0.14 *BP* * | |
| 1035 | Beta-mercaptolactate cysteine disulfiduria | | 1 Case |
| 329284 | Beta-propeller protein-associated neurodegeneration | | 68 Cases |
| 848 | Beta-thalassemia | 1.0 *I* | |
| 65287 | Beta-ureidopropionase deficiency | | 5 Cases |
| 363454 | BICD2-related autosomal dominant childhood-onset proximal spinal muscular atrophy | | 60 Cases |
| 69736 | Bilateral acute depigmentation of the iris | | 62 Cases |
| 140963 | Bilateral microtia-deafness-cleft palate syndrome | | 3 Families |
| 1980 | Bilateral striopallidodentate calcinosis | | 200 Cases |
| 79241 | Biotinidase deficiency | 1.6 *P* * | |
| 79241 | Biotinidase deficiency | 1.6 *BP* | |
| 364198 | Bipartite talus | | 23 Cases |
| 179 | Birdshot chorioretinopathy | 0.35 *P* | |
| 122 | Birt-Hogg-Dubé syndrome | 0.5 *P* * | |
| 123 | Björnstad syndrome | | 33 Cases |
| 124 | Blackfan-Diamond anemia | 0.67 *BP* * | |
| 93930 | Bladder exstrophy | 3.05 *BP* | |
| 73271 | Bleeding diathesis due to a collagen receptor defect | | 20 Cases |
| 420566 | Bleeding disorder due to CalDAG-GEFI deficiency | | 3 Cases |
| 36355 | Bleeding disorder due to P2Y12 defect | | 14 Cases |
| 1997 | Blepharo-cheilo-odontic syndrome | | 50 Cases |
| 1252 | Blepharonasofacial malformation syndrome | | 3 Families |
| 126 | Blepharophimosis-epicanthus inversus-ptosis syndrome | 2.0 *P* | |
| 293642 | Blepharophimosis-intellectual disability syndrome | | 58 Cases |
| 2728 | Blepharophimosis-intellectual disability syndrome, Ohdo type | | 30 Cases |
| 3047 | Blepharophimosis-intellectual disability syndrome, SBBYS type | | 20 Cases |
| 2057 | Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome | | 6 Cases |
| 1259 | Blepharoptosis-myopia-ectopia lentis syndrome | | 3 Cases |
| 171844 | Blindness-scoliosis-arachnodactyly syndrome | | 4 Cases |
| 50945 | Blomstrand lethal chondrodysplasia | | 13 Cases |
| 125 | Bloom syndrome | | 400 Cases |
| 16 | Blue cone monochromatism | 1.0 *P* | |
| 16 | Blue cone monochromatism | 1.0 *BP* | |
| 1059 | Blue rubber bleb nevus | | 200 Cases |
| 217266 | BNAR syndrome | | 9 Families |
| 217008 | Bockenheimer syndrome | | 40 Cases |
| 91135 | Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency | | 11 Cases |
| 97297 | Bohring-Opitz syndrome | | 30 Cases |
| 1842 | Bone dysplasia, lethal Holmgren type | | 7 Cases |
| 223727 | Bone sarcoma | 9.29 *P* * | |
| 223727 | Bone sarcoma | 0.8 *I* * | |
| 1261 | Bonnemann-Meinecke-Reich syndrome | | 4 Cases |
| 1262 | Böök syndrome | | 26 Cases |
| 1263 | Boomerang dysplasia | | 10 Cases |
| 127 | Borjeson-Forssman-Lehmann syndrome | | 50 Cases |
| 69737 | Bosley-Salih-Alorainy syndrome | | 16 Cases |
| 1267 | Botulism | 0.022 *I* * | |
| 1270 | Bowen-Conradi syndrome | | 60 Cases |
| 93389 | Brachydactyly type A5 | | 2 Families |
| 93382 | Brachydactyly type A6 | | 7 Cases |
| 93397 | Brachydactyly type A7 | | 1 Family |
| 1276 | Brachydactyly-arterial hypertension syndrome | | 10 Families |
| 1275 | Brachydactyly-elbow wrist dysplasia syndrome | | 4 Families |
| 2946 | Brachydactyly-long thumb syndrome | | 4 Cases |

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|---|---|---|---|
| 1277 | Brachydactyly-mesomelia-intellectual disability-heart defects syndrome | | 2 Cases |
| 1246 | Brachydactyly-nystagmus-cerebellar ataxia syndrome | | 1 Family |
| 1278 | Brachydactyly-preaxial hallux varus syndrome | | 8 Cases |
| 166035 | Brachydactyly-short stature-retinitis pigmentosa syndrome | | 12 Cases |
| 93409 | Brachydactyly-syndactyly, Zhao type | | 2 Families |
| 1292 | Brachymorphism-onychodysplasia-dysphalangism syndrome | | 9 Cases |
| 1293 | Brachyolmia | | 100 Cases |
| 93302 | Brachyolmia, Maroteaux type | | 4 Families |
| 1295 | Brachytelephalangy-dysmorphism-Kallmann syndrome | | 2 Cases |
| 52047 | Braddock syndrome | | 2 Cases |
| 75374 | Bradyopsia | | 5 Cases |
| 178506 | Brain calcification, Rajab type | | 8 Cases |
| 168598 | Brain demyelination due to methionine adenosyltransferase deficiency | | 2 Cases |
| 352649 | Brain dopamine-serotonin vesicular transport disease | | 8 Cases |
| 75389 | Brain malformation-congenital heart disease-postaxial polydactyly syndrome | | 2 Cases |
| 500150 | Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome | | 29 Cases |
| 209905 | Brain-lung-thyroid syndrome | | 100 Cases |
| 1297 | Branchio-oculo-facial syndrome | | 150 Cases |
| 50815 | Branchiogenic deafness syndrome | | 5 Cases |
| 1299 | Branchioskeletogenital syndrome | | 7 Cases |
| 85284 | BRESEK syndrome | | 5 Cases |
| 90354 | Brittle cornea syndrome | | 65 Cases |
| 70589 | Bronchopulmonary dysplasia | 13.0 *P* * | |
| 79493 | Brooke-Spiegler syndrome | | 100 Cases |
| 1304 | Brucellosis | 0.1 *I* * | |
| 2771 | Bruck syndrome | | 60 Cases |
| 130 | Brugada syndrome | 20.0 *P* * | |
| 131 | Budd-Chiari syndrome | 1.5 *P* * | |
| 36258 | Buerger disease | 16.0 *P* | |
| 280785 | Bullous diffuse cutaneous mastocytosis | | 40 Cases |
| 703 | Bullous pemphigoid | 26.0 *P* * | |
| 543 | Burkitt lymphoma | 0.17 *I* * | |
| 1306 | Buschke-Ollendorff syndrome | 5.0 *I* | |
| 1308 | C syndrome | 0.11 *P* * | |
| 495844 | C11ORF73-related autosomal recessive hypomyelinating leukodystrophy | | 6 Cases |
| 497623 | C12ORF65-related combined oxidative phosphorylation defect | | 30 Cases |
| 135 | CACH syndrome | | 148 Cases |
| 448010 | CAD-CDG | | 1 Case |
| 136 | CADASIL | 3.0 *P* * | |
| 369942 | CADDS | | 4 Cases |
| 1310 | Caffey disease | | 100 Cases |
| 280062 | Calciphylaxis | 5.0 *P* * | |
| 85192 | Calvarial doughnut lesions-bone fragility syndrome | | 20 Cases |
| 83472 | CAMOS syndrome | | 5 Cases |
| 1318 | Campomelia, Cumming type | | 8 Cases |
| 140 | Campomelic dysplasia | 0.33 *BP* * | |
| 1319 | Camptobrachydactyly | | 1 Family |
| 1327 | Camptodactyly syndrome, Guadalajara type 1 | | 8 Cases |
| 1326 | Camptodactyly syndrome, Guadalajara type 2 | | 2 Cases |
| 488434 | Camptodactyly syndrome, Guadalajara type 3 | | 5 Cases |
| 2848 | Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome | | 30 Families |
| 1321 | Camptodactyly-fibrous tissue hyperplasia-skeletal dysplasia syndrome | | 3 Cases |
| 1323 | Camptodactyly-joint contractures-facial skeletal defects syndrome | | 4 Cases |
| 85164 | Camptodactyly-tall stature-scoliosis-hearing loss syndrome | | 30 Cases |
| 1325 | Camptodactyly-taurinuria syndrome | | 17 Cases |
| 1328 | Camurati-Engelmann disease | | 300 Cases |
| 141 | Canavan disease | 1.0 *BP* | |
| 325004 | CANDLE syndrome | | 30 Cases |
| 171881 | Cap myopathy | | 21 Cases |
| 160148 | Cap polyposis | | 67 Cases |
| 137667 | Capillary malformation-arteriovenous malformation | | 261 Cases |
| 147 | Carbamoyl-phosphate synthetase 1 deficiency | 0.31 *P* | |
| 70482 | Carcinoma of esophagus | 9.8 *P* | |
| 70482 | Carcinoma of esophagus | 7.0 *I* | |
| 418945 | Carcinoma of esophagus, salivary gland type | 0.01 *I* * | |
| 56044 | Carcinoma of gallbladder and extrahepatic biliary tract | 12.0 *I* | |

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|---|---|---|---|
| 423781 | Carcinoma of stomach, salivary gland type | 0.01 *I* * | |
| 137628 | Cardiac anomalies-heterotaxy syndrome | | 9 Cases |
| 2872 | Cardiocranial syndrome, Pfeiffer type | | 7 Cases |
| 1340 | Cardiofaciocutaneous syndrome | | 300 Cases |
| 97292 | Cardiogenic shock | 40.0 *P* * | |
| 1345 | Cardiomyopathy-cataract-hip spine disease syndrome | | 9 Cases |
| 91130 | Cardiomyopathy-hypotonia-lactic acidosis syndrome | | 2 Cases |
| 90022 | Cardiomyopathy-renal anomalies syndrome | | 2 Cases |
| 3238 | Cardiospondylocarpofacial syndrome | | 5 Cases |
| 1358 | Carey-Fineman-Ziter syndrome | | 20 Cases |
| 1359 | Carney complex | | 160 Cases |
| 319340 | Carney complex-trismus-pseudocamptodactyly syndrome | | 3 Families |
| 139411 | Carney triad | | 150 Cases |
| 97286 | Carney-Stratakis syndrome | | 20 Families |
| 156 | Carnitine palmitoyl transferase 1A deficiency | | 60 Cases |
| 228302 | Carnitine palmitoyl transferase II deficiency, myopathic form | | 300 Cases |
| 228308 | Carnitine palmitoyl transferase II deficiency, neonatal form | | 20 Families |
| 228305 | Carnitine palmitoyl transferase II deficiency, severe infantile form | | 30 Families |
| 157 | Carnitine palmitoyltransferase II deficiency | 1.0 *P* * | |
| 159 | Carnitine-acylcarnitine translocase deficiency | | 60 Cases |
| 1361 | Carnosinase deficiency | 0.2 *BP* | |
| 53035 | Caroli disease | 0.1 *I* | |
| 65759 | Carpenter syndrome | | 70 Cases |
| 93973 | Carpenter-Waziri syndrome | | 6 Cases |
| 65282 | Carvajal syndrome | | 7 Cases |
| 195 | Cat-eye syndrome | 1.35 *BP* * | |
| 50839 | Cat-scratch disease | 6.6 *P* * | |
| 1373 | Cataract-aberrant oral frenula-growth delay syndrome | | 3 Cases |
| 1368 | Cataract-ataxia-deafness syndrome | | 2 Cases |
| 314993 | Cataract-congenital heart disease-neural tube defect syndrome | | 2 Cases |
| 1383 | Cataract-deafness-hypogonadism syndrome | | 3 Cases |
| 162 | Cataract-glaucoma syndrome | | 3 Families |
| 436174 | Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome | | 3 Cases |
| 1381 | Cataract-intellectual disability-anal atresia-urinary defects syndrome | | 3 Cases |
| 1387 | Cataract-intellectual disability-hypogonadism syndrome | | 20 Cases |
| 1377 | Cataract-microcornea syndrome | | 8 Families |
| 1380 | Cataract-nephropathy-encephalopathy syndrome | | 2 Cases |
| 3286 | Catecholaminergic polymorphic ventricular tachycardia | 10.0 *P* * | |
| 1388 | Catel-Manzke syndrome | | 33 Cases |
| 1123 | Caudal appendage-deafness syndrome | | 2 Cases |
| 468684 | CCDC115-CDG | | 8 Cases |
| 86870 | CD4+/CD56+ hematodermic neoplasm | 12.0 *P* * | |
| 66631 | CEDNIK syndrome | | 13 Cases |
| 1459 | Celiac disease-epilepsy-cerebral calcification syndrome | | 170 Cases |
| 3258 | Cenani-Lenz syndrome | | 30 Cases |
| 2431 | Central bilateral macrogyria | | 4 Cases |
| 98972 | Central cloudy dystrophy of François | | 24 Cases |
| 178029 | Central diabetes insipidus | 4.0 *P* * | |
| 3240 | Central nervous system calcification-deafness-tubular acidosis-anemia syndrome | | 2 Cases |
| 251870 | Central nervous system primitive neuroectodermal tumor | 0.07 *I* * | |
| 73256 | Central neurocytoma | | 500 Cases |
| 411527 | Central retinal vein occlusion | 28.0 *P* * | |
| 89841 | Centripetalis recessive dystrophic epidermolysis bullosa | | 10 Cases |
| 1171 | Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome | | 10 Cases |
| 2246 | Cerebellar hypoplasia-tapetoretinal degeneration syndrome | | 3 Cases |
| 444072 | Cerebellar-facial-dental syndrome | | 3 Families |
| 46724 | Cerebral arteriovenous malformation | 6.0 *P* * | |
| 2081 | Cerebral gigantism-jaw cysts syndrome | | 9 Cases |
| 329217 | Cerebral sinovenous thrombosis | 0.35 *I* * | |
| 1393 | Cerebrocostomandibular syndrome | | 75 Cases |
| 314679 | Cerebrofacioarticular syndrome | | 9 Cases |
| 1394 | Cerebrofaciothoracic dysplasia | | 20 Cases |
| 66625 | Cerebrooculonasal syndrome | | 21 Cases |
| 169079 | Cernunnos-XLF deficiency | | 5 Cases |
| 2218 | Cervical hypertrichosis-peripheral neuropathy syndrome | | 4 Cases |
| 88642 | Channelopathy-associated congenital insensitivity to pain | | 20 Cases |

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|---|---|---|---|
| 46627 | Char syndrome | | 10 Cases |
| 65753 | Charcot-Marie-Tooth disease type 1 | 17.5 *P* | |
| 101101 | Charcot-Marie-Tooth disease type 2B2 | | 1 Family |
| 228374 | Charcot-Marie-Tooth disease type 2B5 | | 4 Cases |
| 101102 | Charcot-Marie-Tooth disease type 2H | | 13 Cases |
| 300319 | Charcot-Marie-Tooth disease type 2P | | 18 Cases |
| 397968 | Charcot-Marie-Tooth disease type 2R | | 1 Case |
| 443073 | Charcot-Marie-Tooth disease type 2S | | 35 Cases |
| 495274 | Charcot-Marie-Tooth disease type 2T | | 10 Cases |
| 99955 | Charcot-Marie-Tooth disease type 4B1 | | 11 Families |
| 363981 | Charcot-Marie-Tooth disease type 4B3 | | 3 Cases |
| 99954 | Charcot-Marie-Tooth disease type 4H | | 15 Cases |
| 139515 | Charcot-Marie-Tooth disease type 4J | | 18 Cases |
| 90103 | Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome | | 7 Cases |
| 166 | Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy | 25.0 *P* * | |
| 138 | CHARGE syndrome | 6.5 *BP* | |
| 1406 | Charlie M syndrome | | 4 Cases |
| 167 | Chédiak-Higashi syndrome | | 500 Cases |
| 1221 | Cheilitis glandularis | | 100 Cases |
| 184 | Cherubism | | 300 Cases |
| 324625 | Chikungunya | 0.08 *I* * | |
| 90280 | Chilblain lupus | | 70 Cases |
| 139 | CHILD syndrome | | 60 Cases |
| 209908 | Childhood apraxia of speech | | 22 Cases |
| 168782 | Childhood disintegrative disorder | 2.0 *P* * | |
| 293955 | Childhood encephalopathy due to thiamine pyrophosphokinase deficiency | | 5 Cases |
| 363677 | Childhood-onset autosomal recessive myopathy with external ophthalmoplegia | | 22 Cases |
| 497906 | Childhood-onset basal ganglia degeneration syndrome | | 4 Cases |
| 494541 | Childhood-onset benign chorea with striatal involvement | | 3 Cases |
| 500180 | Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder | | 7 Cases |
| 466921 | Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome | | 3 Families |
| 401866 | Childhood-onset spasticity with hyperglycinemia | | 3 Cases |
| 3474 | CHIME syndrome | | 8 Cases |
| 137914 | Choanal atresia | 8.6 *BP* * | |
| 1200 | Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome | | 11 Families |
| 70567 | Cholangiocarcinoma | 2.1 *P* | |
| 70567 | Cholangiocarcinoma | 4.2 *I* | |
| 173 | Cholera | 0.01 *I* * | |
| 1414 | Cholestasis-lymphedema syndrome | | 47 Cases |
| 1415 | Cholestasis-pigmentary retinopathy-cleft palate syndrome | | 5 Cases |
| 79347 | Chondrodysplasia punctata, Toriello type | | 3 Cases |
| 280586 | Chondrodysplasia with joint dislocations, gPAPP type | | 4 Cases |
| 1422 | Chondrodysplasia-disorder of sex development syndrome | | 2 Cases |
| 319195 | Chondroectodermal dysplasia with night blindness | | 4 Cases |
| 404507 | Chondromyxoid fibroma | | 50 Cases |
| 55880 | Chondrosarcoma | 0.24 *I* * | |
| 251899 | Choroid plexus carcinoma | 0.01 *I* * | |
| 1433 | Choroidal atrophy-alopecia syndrome | | 2 Cases |
| 180 | Choroideremia | 2.0 *P* * | |
| 1435 | Choroideremia-deafness-obesity syndrome | | 4 Cases |
| 1434 | Choroideremia-hypopituitarism syndrome | | 2 Cases |
| 85278 | Christianson syndrome | | 30 Cases |
| 319303 | Chromophobe renal cell carcinoma | 0.01 *I* * | |
| 435988 | Chronic atrial and intestinal dysrhythmia syndrome | | 17 Cases |
| 314373 | Chronic diarrhea due to guanylate cyclase 2C overactivity | | 32 Cases |
| 1670 | Chronic diarrhea with villous atrophy | | 2 Cases |
| 468641 | Chronic enteropathy associated with SLCO2A1 gene | | 18 Cases |
| 379 | Chronic granulomatous disease | 0.46 *BP* | |
| 396 | Chronic hiccup | 1.0 *P* * | |
| 2932 | Chronic inflammatory demyelinating polyneuropathy | 3.7 *P* * | |
| 521 | Chronic myeloid leukemia | 6.0 *P* * | |
| 521 | Chronic myeloid leukemia | 1.25 *I* * | |
| 98823 | Chronic myelomonocytic leukemia | 0.29 *I* * | |

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|---|---|---|---|
| 86830 | Chronic myeloproliferative disease, unclassifiable | 0.53 *I* * | |
| 324964 | Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis | 0.3 *P* | |
| 324964 | Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis | 2.5 *I* | |
| 95426 | Chronic pain requiring intraspinal analgesia | 12.0 *P* * | |
| 101959 | Chronic primary adrenal insufficiency | 14.0 *P* * | |
| 101959 | Chronic primary adrenal insufficiency | 0.4 *I* * | |
| 70591 | Chronic thromboembolic pulmonary hypertension | 3.0 *P* * | |
| 263463 | CHST3-related skeletal dysplasia | | 2 Families |
| 93971 | Chudley-Lowry-Hoar syndrome | | 3 Cases |
| 314597 | Chudley-McCullough syndrome | | 25 Cases |
| 71 | Chylomicron retention disease | | 55 Cases |
| 435651 | CIDEC-related familial partial lipodystrophy | | 1 Case |
| 1451 | CINCA syndrome | | 200 Cases |
| 69744 | Circumscribed palmoplantar hypokeratosis | | 17 Cases |
| 309854 | Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome | | 20 Cases |
| 247525 | Citrullinemia type I | 2.4 *P* * | |
| 251383 | CK syndrome | | 24 Cases |
| 168984 | CLAPO syndrome | | 6 Cases |
| 90794 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency | 7.0 *P* * | |
| 90794 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency | 7.0 *BP* | |
| 315306 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form | 7.5 *P* * | |
| 315306 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form | 7.5 *BP* * | |
| 315311 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form | 2.5 *P* * | |
| 79239 | Classic galactosemia | 2.1 *I* * | |
| 58017 | Classic hairy cell leukemia | 0.29 *I* * | |
| 391 | Classic Hodgkin lymphoma | 2.38 *I* * | |
| 98846 | Classic Hodgkin lymphoma, lymphocyte-depleted type | 0.04 *I* * | |
| 98845 | Classic Hodgkin lymphoma, lymphocyte-rich type | 0.1 *I* * | |
| 98844 | Classic Hodgkin lymphoma, mixed cellularity type | 0.42 *I* * | |
| 98843 | Classic Hodgkin lymphoma, nodular sclerosis type | 1.28 *I* * | |
| 394 | Classic homocystinuria | 1.65 *P* * | |
| 394 | Classic homocystinuria | 0.3 *BP* | |
| 2584 | Classic mycosis fungoides | 0.5 *I* * | |
| 329977 | Classic neuroendocrine tumor of appendix | 0.25 *I* | |
| 485350 | CLCN4-related X-linked intellectual disability syndrome | | 38 Cases |
| 398971 | Clear cell adenocarcinoma of the ovary | 0.32 *I* * | |
| 319276 | Clear cell renal carcinoma | 1.99 *I* * | |
| 508476 | Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-hearing loss syndrome | | 7 Cases |
| 1995 | Cleft lip-retinopathy syndrome | | 2 Cases |
| 199306 | Cleft lip/palate | 80.0 *BP* | |
| 2003 | Cleft lip/palate-deafness-sacral lipoma syndrome | | 2 Cases |
| 2001 | Cleft lip/palate-intestinal malrotation-cardiopathy syndrome | | 5 Cases |
| 2014 | Cleft palate | 53.6 *BP* * | |
| 2013 | Cleft palate-large ears-small head syndrome | | 8 Cases |
| 2016 | Cleft palate-lateral synechia syndrome | | 11 Cases |
| 2015 | Cleft palate-short stature-vertebral anomalies syndrome | | 2 Cases |
| 2010 | Cleft palate-stapes fixation-oligodontia syndrome | | 2 Cases |
| 1452 | Cleidocranial dysplasia | 0.1 *P* | |
| 1452 | Cleidocranial dysplasia | 0.4 *BP* * | |
| 1453 | Cleidorhizomelic syndrome | | 2 Cases |
| 284448 | CLIPPERS | | 50 Cases |
| 228357 | CLN9 disease | | 2 Cases |
| 314629 | CLN11 disease | | 2 Cases |
| 352709 | CLN13 disease | | 4 Cases |
| 93929 | Cloacal exstrophy | 0.54 *BP* | |
| 93267 | Cloverleaf skull-multiple congenital anomalies syndrome | | 3 Cases |
| 140944 | CLOVES syndrome | | 100 Cases |
| 397725 | COASY protein-associated neurodegeneration | | 2 Cases |
| 53721 | Cobb syndrome | | 45 Cases |
| 51577 | Cobblestone lissencephaly | 1.0 *BP* * | |
| 352682 | Cobblestone lissencephaly without muscular or ocular involvement | | 4 Cases |
| 90068 | Cocaine intoxication | 1.0 *P* * | |
| 3233 | Cochleosaccular degeneration-cataract syndrome | | 2 Families |

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|---|---|---|---|
| 191 | Cockayne syndrome | 0.2 *BP* * | |
| 191 | Cockayne syndrome | 0.5 *I* * | |
| 1458 | CODAS syndrome | | 12 Cases |
| 192 | Coffin-Lowry syndrome | 1.5 *P* | |
| 1465 | Coffin-Siris syndrome | | 190 Cases |
| 1466 | COFS syndrome | | 20 Cases |
| 263508 | COG1-CDG | | 3 Cases |
| 435934 | COG2-CDG | | 1 Case |
| 263501 | COG4-CDG | | 2 Cases |
| 263487 | COG5-CDG | | 9 Cases |
| 464443 | COG6-CGD | | 10 Cases |
| 79333 | COG7-CDG | | 8 Cases |
| 95428 | COG8-CDG | | 2 Cases |
| 1467 | Cogan syndrome | | 300 Cases |
| 444077 | Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome | | 3 Cases |
| 193 | Cohen syndrome | | 200 Cases |
| 31824 | Colchicine poisoning | 0.1 *P* * | |
| 157820 | Cold-induced sweating syndrome | | 6 Cases |
| 2050 | Cole-Carpenter syndrome | | 3 Cases |
| 1471 | Coloboma of macula-brachydactyly type B syndrome | | 10 Cases |
| 468672 | Colobomatous macrophthalmia-microcornea syndrome | | 21 Cases |
| 424099 | Colobomatous microphthalmia-rhizomelic dysplasia syndrome | | 5 Families |
| 435930 | Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome | | 3 Cases |
| 35909 | Combined deficiency of factor V and factor VIII | 0.5 *P* * | |
| 440727 | Combined hamartoma of the retina and retinal pigment epithelium | | 120 Cases |
| 169090 | Combined immunodeficiency due to CRAC channel dysfunction | | 10 Cases |
| 217390 | Combined immunodeficiency due to DOCK8 deficiency | | 11 Cases |
| 505227 | Combined immunodeficiency due to GINS1 deficiency | | 5 Cases |
| 357329 | Combined immunodeficiency due to IL21R deficiency | | 6 Cases |
| 445018 | Combined immunodeficiency due to LRBA deficiency | | 23 Cases |
| 397964 | Combined immunodeficiency due to MALT1 deficiency | | 3 Cases |
| 504530 | Combined immunodeficiency due to Moesin deficiency | | 7 Cases |
| 317428 | Combined immunodeficiency due to ORAI1 deficiency | | 6 Cases |
| 431149 | Combined immunodeficiency due to OX40 deficiency | | 1 Case |
| 231154 | Combined immunodeficiency due to partial RAG1 deficiency | | 9 Cases |
| 314689 | Combined immunodeficiency due to STK4 deficiency | | 7 Cases |
| 476113 | Combined immunodeficiency due to TFRC deficiency | | 2 Families |
| 221139 | Combined immunodeficiency with facioculoskeletal anomalies | | 2 Cases |
| 254920 | Combined oxidative phosphorylation defect type 2 | | 1 Case |
| 254925 | Combined oxidative phosphorylation defect type 4 | | 2 Cases |
| 254930 | Combined oxidative phosphorylation defect type 7 | | 7 Cases |
| 319504 | Combined oxidative phosphorylation defect type 8 | | 7 Cases |
| 319509 | Combined oxidative phosphorylation defect type 9 | | 4 Cases |
| 324535 | Combined oxidative phosphorylation defect type 11 | | 32 Cases |
| 319514 | Combined oxidative phosphorylation defect type 13 | | 2 Cases |
| 319519 | Combined oxidative phosphorylation defect type 14 | | 5 Cases |
| 319524 | Combined oxidative phosphorylation defect type 15 | | 16 Cases |
| 369913 | Combined oxidative phosphorylation defect type 17 | | 20 Families |
| 420728 | Combined oxidative phosphorylation defect type 20 | | 2 Cases |
| 420733 | Combined oxidative phosphorylation defect type 21 | | 2 Cases |
| 444013 | Combined oxidative phosphorylation defect type 23 | | 11 Cases |
| 444458 | Combined oxidative phosphorylation defect type 24 | | 3 Cases |
| 447954 | Combined oxidative phosphorylation defect type 25 | | 2 Cases |
| 477684 | Combined oxidative phosphorylation defect type 26 | | 2 Cases |
| 477774 | Combined oxidative phosphorylation defect type 27 | | 3 Cases |
| 478029 | Combined oxidative phosphorylation defect type 29 | | 1 Case |
| 478042 | Combined oxidative phosphorylation defect type 30 | | 2 Cases |
| 309111 | Combined pancreatic lipase-colipase deficiency | | 3 Cases |
| 280133 | Complement component 3 deficiency | | 27 Cases |
| 99429 | Complete androgen insensitivity syndrome | 0.83 *P* | |
| 99429 | Complete androgen insensitivity syndrome | 3.0 *I* * | |
| 1329 | Complete atrioventricular canal | 20.0 *BP* * | |
| 98949 | Complete cryptophthalmia | | 15 Cases |

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|---|---|---|---|
| 457378 | Complex lethal osteochondrodysplasia | | 6 Cases |
| 306644 | Complication after organ transplantation | 9.0 *P* * | |
| 268316 | Complication in hemodialysis | 13.0 *I* * | |
| 458758 | Composite hemangioendothelioma | | 39 Cases |
| 168966 | Composite lymphoma | 0.01 *I* * | |
| 3216 | Conductive deafness-malformed external ear syndrome | | 8 Cases |
| 3236 | Conductive deafness-ptosis-skeletal anomalies syndrome | | 3 Cases |
| 209932 | Cone dystrophy with supernormal rod response | | 45 Cases |
| 1872 | Cone rod dystrophy | 2.5 *P* * | |
| 221142 | Confetti-like macular atrophy | | 2 Cases |
| 294975 | Congenital absence of upper arm and forearm with hand present | 0.62 *BP* | |
| 973 | Congenital absence/hypoplasia of fingers excluding thumb, unilateral | | 2 Families |
| 418 | Congenital adrenal hyperplasia | 6.0 *P* * | |
| 418 | Congenital adrenal hyperplasia | 6.7 *BP* * | |
| 418 | Congenital adrenal hyperplasia | 13.35 *I* * | |
| 90791 | Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency | | 68 Cases |
| 90795 | Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency | 0.47 *P* * | |
| 90795 | Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency | 0.75 *BP* * | |
| 90793 | Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency | 0.1 *P* * | |
| 95699 | Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency | 0.75 *BP* * | |
| 495879 | Congenital agenesis of the scrotum | | 6 Cases |
| 79 | Congenital alpha2-antiplasmin deficiency | | 40 Cases |
| 210122 | Congenital alveolar capillary dysplasia | | 40 Cases |
| 3319 | Congenital amegakaryocytic thrombocytopenia | | 100 Cases |
| 86816 | Congenital analbuminemia | | 50 Cases |
| 1195 | Congenital atransferrinemia | | 16 Cases |
| 48 | Congenital bilateral absence of vas deferens | 50.0 *P* * | |
| 79302 | Congenital bile acid synthesis defect type 3 | | 2 Cases |
| 79095 | Congenital bile acid synthesis defect type 4 | | 5 Cases |
| 514352 | Congenital brachyesophagus-intrathoracic stomach-vertebral anomalies syndrome | | 8 Cases |
| 71278 | Congenital brain dysgenesis due to glutamine synthetase deficiency | | 3 Cases |
| 2040 | Congenital bronchobiliary fistula | | 35 Cases |
| 300313 | Congenital cataract-hearing loss-severe developmental delay syndrome | | 5 Cases |
| 1369 | Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome | | 40 Cases |
| 464738 | Congenital cataract-microcephaly-nevus flammeus simplex-severe intellectual disability syndrome | | 7 Cases |
| 330054 | Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome | | 3 Cases |
| 521432 | Congenital cataract-severe neonatal hepatopathy-global developmental delay syndrome | | 2 Cases |
| 48431 | Congenital cataracts-facial dysmorphism-neuropathy syndrome | | 170 Cases |
| 512260 | Congenital cerebellar ataxia due to RNU12 mutation | | 6 Cases |
| 329242 | Congenital chronic diarrhea with protein-losing enteropathy | | 2 Cases |
| 168612 | Congenital deficiency in alpha-fetoprotein | | 22 Cases |
| 2140 | Congenital diaphragmatic hernia | 30.0 *BP* | |
| 137 | Congenital disorder of glycosylation | 1.5 *BP* * | |
| 85 | Congenital dyserythropoietic anemia | 0.16 *BP* * | |
| 98870 | Congenital dyserythropoietic anemia type III | | 60 Cases |
| 293825 | Congenital dyserythropoietic anemia type IV | | 4 Cases |
| 103910 | Congenital enterocyte heparan sulfate deficiency | | 3 Cases |
| 231573 | Congenital erosive and vesicular dermatosis | | 31 Cases |
| 79277 | Congenital erythropoietic porphyria | 0.065 *I* * | |
| 325 | Congenital factor II deficiency | 0.05 *P* * | |
| 326 | Congenital factor V deficiency | 0.1 *P* * | |
| 327 | Congenital factor VII deficiency | 0.33 *P* * | |
| 329 | Congenital factor XI deficiency | 0.1 *P* * | |
| 331 | Congenital factor XIII deficiency | 0.05 *P* * | |
| 331 | Congenital factor XIII deficiency | 0.04 *I* * | |
| 335 | Congenital fibrinogen deficiency | 0.15 *P* * | |
| 476406 | Congenital generalized hypercontractile muscle stiffness syndrome | | 2 Cases |
| 1023 | Congenital generalized hypertrichosis, Ambras type | | 40 Cases |
| 98976 | Congenital glaucoma | 3.6 *BP* * | |
| 60041 | Congenital heart block | 4.54 *BP* | |

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| 1355 | Congenital heart defect-round face-developmental delay syndrome | | 3 Cases |
| 98975 | Congenital hereditary endothelial dystrophy type I | | 68 Cases |
| 306530 | Congenital hereditary facial paralysis-variable hearing loss syndrome | | 13 Cases |
| 2185 | Congenital hydrocephalus | 46.5 *BP* * | |
| 442 | Congenital hypothyroidism | 38.0 *BP* * | |
| 95711 | Congenital hypothyroidism due to developmental anomaly | 21.3 *P* * | |
| 95715 | Congenital hypothyroidism due to transplacental passage of TSH-binding inhibitory antibodies | 1.0 *P* * | |
| 352333 | Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome | | 2 Cases |
| 2271 | Congenital ichthyosis-microcephalus-tetraplegia syndrome | | 2 Cases |
| 217399 | Congenital insensitivity to pain with hyperhidrosis | | 2 Cases |
| 453510 | Congenital insensitivity to pain with severe intellectual disability | | 3 Cases |
| 1229 | Congenital intrauterine infection-like syndrome | | 30 Cases |
| 332 | Congenital intrinsic factor deficiency | | 100 Cases |
| 657 | Congenital isolated hyperinsulinism | 2.0 *BP* | |
| 495875 | Congenital labioscrotal agenesis-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome | | 3 Cases |
| 1954 | Congenital lethal erythroderma | | 17 Cases |
| 210163 | Congenital lethal myopathy, Compton-North type | | 4 Cases |
| 1928 | Congenital lobar emphysema | 4.0 *BP* | |
| 93109 | Congenital megacalycosis | | 25 Cases |
| 69063 | Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization | | 15 Cases |
| 391376 | Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome | | 20 Cases |
| 508512 | Congenital multiple café-au-lait macules-increased sister chromatid exchange syndrome | | 2 Cases |
| 157973 | Congenital muscular dystrophy due to LMNA mutation | | 23 Cases |
| 258 | Congenital muscular dystrophy type 1A | 0.3 *P* * | |
| 98893 | Congenital muscular dystrophy type 1B | | 6 Cases |
| 371007 | Congenital muscular dystrophy with hyperlaxity | | 14 Cases |
| 34520 | Congenital muscular dystrophy with integrin alpha-7 deficiency | 0.03 *P* * | |
| 329178 | Congenital muscular dystrophy with intellectual disability and severe epilepsy | | 3 Cases |
| 1875 | Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome | | 7 Cases |
| 486815 | Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome | | 4 Cases |
| 590 | Congenital myasthenic syndrome | 0.3 *P* * | |
| 319160 | Congenital myopathy with internal nuclei and atypical cores | | 5 Cases |
| 424107 | Congenital myopathy with myasthenic-like onset | | 2 Cases |
| 199329 | Congenital myopathy, Paradas type | | 2 Cases |
| 206973 | Congenital myotonia | 1.0 *P* | |
| 168486 | Congenital neuronal ceroid lipofuscinosis | | 10 Cases |
| 369852 | Congenital neutropenia-myelofibrosis-nephromegaly syndrome | | 16 Cases |
| 79394 | Congenital non-bullous ichthyosiform erythroderma | 0.3 *P* * | |
| 2772 | Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome | | 3 Cases |
| 313906 | Congenital pancreatic cyst | | 10 Cases |
| 139414 | Congenital panfollicular nevus | | 3 Cases |
| 508542 | Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome | | 5 Cases |
| 66630 | Congenital pseudoarthrosis of the clavicle | | 200 Cases |
| 2444 | Congenital pulmonary airway malformation | 8.2 *BP* * | |
| 2414 | Congenital pulmonary lymphangiectasia | | 100 Cases |
| 3189 | Congenital pulmonary valve stenosis | 39.3 *BP* * | |
| 3269 | Congenital radioulnar synostosis | | 350 Cases |
| 281190 | Congenital reticular ichthyosiform erythroderma | | 40 Cases |
| 290 | Congenital rubella syndrome | 0.35 *BP* * | |
| 290 | Congenital rubella syndrome | 0.03 *I* * | |
| 2301 | Congenital short bowel syndrome | | 43 Cases |
| 369861 | Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome | | 16 Cases |
| 103908 | Congenital sodium diarrhea | | 15 Cases |

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| 101068 | Congenital stromal corneal dystrophy | | 6 Families |
| 35122 | Congenital sucrase-isomaltase deficiency | 20.0 *P* * | |
| 499009 | Congenital syphilis | 1.4 *BP* * | |
| 99125 | Congenital total pulmonary venous return anomaly | 9.0 *BP* | |
| 858 | Congenital toxoplasmosis | 33.0 *BP* * | |
| 92050 | Congenital tufting enteropathy | 0.5 *BP* * | |
| 291 | Congenital varicella syndrome | | 130 Cases |
| 521438 | Congenital vertebral-cardiac-renal anomalies syndrome | | 4 Cases |
| 216694 | Congenitally corrected transposition of the great arteries | 3.0 *BP* | |
| 2391 | Congenitally short costocoracoid ligament | | 1 Family |
| 860 | Congenitally uncorrected transposition of the great arteries | 24.25 *BP* * | |
| 300284 | Connective tissue disorder due to lysyl hydroxylase-3 deficiency | | 2 Cases |
| 420794 | Cono-spondylar dysplasia | | 3 Cases |
| 319651 | Constitutional megaloblastic anemia with severe neurologic disease | | 6 Cases |
| 436003 | Contractures-developmental delay-Pierre Robin syndrome | | 6 Cases |
| 1484 | Contractures-ectodermal dysplasia-cleft lip/palate syndrome | | 2 Cases |
| 314002 | Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome | | 2 Cases |
| 1487 | Cooks syndrome | | 12 Cases |
| 1488 | Cooper-Jabs syndrome | | 2 Cases |
| 1490 | Corneal dystrophy-perceptive deafness syndrome | | 24 Cases |
| 352662 | Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome | | 19 Cases |
| 199 | Cornelia de Lange syndrome | 1.9 *P* * | |
| 199 | Cornelia de Lange syndrome | 1.3 *BP* * | |
| 3194 | Corneodermatoosseous syndrome | | 7 Cases |
| 52055 | Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome | | 2 Cases |
| 459074 | Corpus callosum agenesis-macrocephaly-hypertelorism syndrome | | 4 Cases |
| 1389 | Cortical blindness-intellectual disability-polydactyly syndrome | | 3 Cases |
| 300570 | Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation | | 12 Cases |
| 54251 | Corticosteroid-sensitive aseptic abscess syndrome | | 49 Cases |
| 3071 | Costello syndrome | | 300 Cases |
| 201 | Cowden syndrome | 0.5 *P* * | |
| 1508 | Coxoauricular syndrome | | 4 Cases |
| 1509 | Coxopodopatellar syndrome | | 47 Cases |
| 1512 | Crane-Heise syndrome | | 9 Cases |
| 1525 | Cranio-osteoarthropathy | | 30 Cases |
| 1513 | Craniodiaphyseal dysplasia | | 20 Cases |
| 1514 | Craniodigital-intellectual disability syndrome | | 5 Cases |
| 1515 | Cranioectodermal dysplasia | | 60 Cases |
| 85168 | Craniofacial conodysplasia | | 1 Family |
| 314555 | Craniofacial dysplasia-osteopenia syndrome | | 5 Cases |
| 459061 | Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome | | 8 Cases |
| 1516 | Craniofacial dyssynostosis | | 14 Cases |
| 1529 | Craniofacial-deafness-hand syndrome | | 3 Cases |
| 363705 | Craniofaciofrontodigital syndrome | | 4 Cases |
| 1521 | Craniofrontonasal dysplasia-Poland anomaly syndrome | | 2 Cases |
| 50814 | Craniolenticulosutural dysplasia | | 28 Cases |
| 85184 | Craniometadiaphyseal dysplasia, wormian bone type | | 4 Cases |
| 1522 | Craniometaphyseal dysplasia | | 160 Cases |
| 54595 | Craniopharyngioma | 2.0 *P* * | |
| 54595 | Craniopharyngioma | 1.0 *I* | |
| 157832 | Craniorhiny | | 6 Cases |
| 1531 | Craniosynostosis | 24.3 *BP* * | |
| 1541 | Craniosynostosis, Boston type | | 3 Families |
| 2145 | Craniosynostosis, Herrmann-Opitz type | | 2 Cases |
| 1527 | Craniosynostosis, Philadelphia type | | 1 Family |
| 85199 | Craniosynostosis-anal anomalies-porokeratosis syndrome | | 9 Cases |
| 1538 | Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome | | 4 Cases |
| 1533 | Craniosynostosis-fibular aplasia syndrome | | 2 Cases |
| 171839 | Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome | | 2 Cases |
| 52054 | Craniosynostosis-intracranial calcifications syndrome | | 3 Cases |
| 1528 | Craniotelencephalic dysplasia | | 4 Cases |
| 90290 | CREST syndrome | 8.0 *P* * | |
| 205 | Crigler-Najjar syndrome | 1.0 *P* * | |

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|---|---|---|---|
| 205 | Crigler-Najjar syndrome | 0.1 *BP* * | |
| 1545 | Crisponi syndrome | | 30 Cases |
| 1461 | Criss-cross heart | 0.8 *BP* * | |
| 2930 | Cronkhite-Canada syndrome | | 500 Cases |
| 2935 | Crossed polysyndactyly | | 12 Cases |
| 207 | Crouzon disease | 0.9 *BP* * | |
| 93262 | Crouzon syndrome-acanthosis nigricans syndrome | 0.1 *BP* | |
| 1546 | Cryptococcosis | 11.0 *I* * | |
| 468635 | Cryptogenic multifocal ulcerous stenosing enteritis | | 60 Cases |
| 1547 | Cryptomicrotia-brachydactyly-excess fingertip arch syndrome | | 2 Cases |
| 1548 | Cryptorchidism-arachnodactyly-intellectual disability syndrome | | 3 Cases |
| 1549 | Cryptosporidiosis | 1.96 *I* * | |
| 307766 | Curly hair-acral keratoderma-caries syndrome | | 14 Cases |
| 1552 | Currarino syndrome | 1.0 *P* * | |
| 1553 | Curry-Jones syndrome | | 9 Cases |
| 96253 | Cushing disease | 4.0 *P* * | |
| 96253 | Cushing disease | 0.2 *I* * | |
| 553 | Cushing syndrome | 5.9 *P* | |
| 553 | Cushing syndrome | 0.15 *I* * | |
| 189427 | Cushing syndrome due to macronodular adrenal hyperplasia | 0.08 *P* * | |
| 280779 | Cutaneous collagenous vasculopathy | | 20 Cases |
| 79140 | Cutaneous neuroendocrine carcinoma | 4.0 *P* * | |
| 79140 | Cutaneous neuroendocrine carcinoma | 0.27 *I* | |
| 2881 | Cutaneous photosensitivity-lethal colitis syndrome | | 3 Cases |
| 451607 | Cutaneous pseudolymphoma | | 60 Cases |
| 1555 | Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome | | 12 Cases |
| 209 | Cutis laxa | 0.1 *BP* * | |
| 221145 | Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies | | 21 Cases |
| 171719 | Cutis laxa-Marfanoid syndrome | | 18 Cases |
| 1556 | Cutis marmorata telangiectatica congenita | | 300 Cases |
| 2686 | Cyclic neutropenia | 0.1 *P* * | |
| 2674 | Cyprus facial-neuromusculoskeletal syndrome | | 1 Family |
| 400 | Cystic echinococcosis | 1.0 *I* * | |
| 586 | Cystic fibrosis | 7.4 *P* * | |
| 2575 | Cystic fibrosis-gastritis-megaloblastic anemia syndrome | | 2 Cases |
| 2111 | Cystic hamartoma of lung and kidney | | 3 Cases |
| 85136 | Cystic leukoencephalopathy without megalencephaly | | 50 Cases |
| 213 | Cystinosis | 1.5 *P* * | |
| 213 | Cystinosis | 0.75 *BP* | |
| 214 | Cystinuria | 14.0 *P* | |
| 75381 | Cystoid macular dystrophy | | 97 Cases |
| 95702 | Cytomegalic congenital adrenal hypoplasia | 8.0 *BP* | |
| 137698 | Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk | 25.5 *P* * | |
| 94087 | Cytophagic histiocytic panniculitis | | 100 Cases |
| 477787 | Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder | | 2 Cases |
| 137678 | Czech dysplasia, metatarsal type | | 20 Cases |
| 2437 | Czeizel-Losonci syndrome | | 3 Cases |
| 356978 | D,L-2-hydroxyglutaric aciduria | | 13 Cases |
| 79315 | D-2-hydroxyglutaric aciduria | | 80 Cases |
| 1562 | Dacryocystitis-osteopoikilosis syndrome | | 5 Cases |
| 1563 | Dahlberg-Borer-Newcomer syndrome | | 2 Cases |
| 1566 | Dandy-Walker malformation-postaxial polydactyly syndrome | | 5 Cases |
| 218 | Darier disease | 3.4 *P* * | |
| 300536 | DDOST-CDG | | 1 Case |
| 488647 | DDX41-related hematologic malignancy predisposition syndrome | | 3 Families |
| 2962 | De Barsy syndrome | | 40 Cases |
| 3214 | Deaf blind hypopigmentation syndrome, Yemenite type | | 2 Cases |
| 90024 | Deafness with labyrinthine aplasia, microtia, and microdontia | | 56 Cases |
| 3241 | Deafness-craniofacial syndrome | | 2 Cases |
| 3232 | Deafness-ear malformation-facial palsy syndrome | | 4 Cases |
| 3220 | Deafness-enamel hypoplasia-nail defects syndrome | | 15 Families |
| 254898 | Deafness-encephaloneuropathy-obesity-valvulopathy syndrome | | 2 Cases |
| 3218 | Deafness-epiphyseal dysplasia-short stature syndrome | | 2 Cases |
| 3224 | Deafness-genital anomalies-metacarpal and metatarsal synostosis syndrome | | 2 Cases |
| 90646 | Deafness-hypogonadism syndrome | | 5 Cases |
| 94064 | Deafness-infertility syndrome | | 3 Families |

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|---|---|---|---|
| 85321 | Deafness-intellectual disability syndrome, Martin-Probst type | | 3 Cases |
| 3226 | Deafness-lymphedema-leukemia syndrome | | 20 Cases |
| 3230 | Deafness-oligodontia syndrome | | 5 Cases |
| 3231 | Deafness-onychodystrophy syndrome | | 50 Cases |
| 3217 | Deafness-small bowel diverticulosis-neuropathy syndrome | | 5 Cases |
| 3239 | Deafness-vitiligo-achalasia syndrome | | 2 Cases |
| 99970 | Dedifferentiated liposarcoma | 0.27 *I* * | |
| 293978 | Deficiency in anterior pituitary function-variable immunodeficiency syndrome | | 7 Cases |
| 3202 | Dehydrated hereditary stomatocytosis | | 20 Families |
| 3034 | Delayed membranous cranial ossification | | 2 Families |
| 3038 | Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome | | 6 Cases |
| 1627 | Deletion 5q35 | | 10 Cases |
| 79134 | DEND syndrome | | 40 Cases |
| 99828 | Dengue fever | 714.0 *I* | |
| 93571 | Dense deposit disease | 0.25 *P* | |
| 1652 | Dent disease | | 250 Families |
| 99789 | Dentin dysplasia type I | 1.0 *P* * | |
| 99791 | Dentin dysplasia type II | | 19 Families |
| 99792 | Dentin dysplasia-sclerotic bones syndrome | | 1 Family |
| 49042 | Dentinogenesis imperfecta | 14.5 *P* * | |
| 166260 | Dentinogenesis imperfecta type 2 | 14.6 *P* * | |
| 71267 | Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome | | 2 Cases |
| 220 | Denys-Drash syndrome | | 200 Cases |
| 1656 | Dermatitis herpetiformis | 27.0 *P* * | |
| 31112 | Dermatofibrosarcoma protuberans | 10.0 *P* * | |
| 1659 | Dermatoleukodystrophy | | 2 Cases |
| 221 | Dermatomyositis | 6.0 *P* * | |
| 221 | Dermatomyositis | 0.55 *I* * | |
| 1657 | Dermatoosteolysis, Kirghizian type | | 5 Cases |
| 86920 | Dermatopathia pigmentosa reticularis | | 20 Cases |
| 79149 | Dermochondrocorneal dystrophy | | 15 Cases |
| 1660 | Dermoodontodysplasia | | 11 Cases |
| 1425 | Desbuquois syndrome | | 50 Cases |
| 84132 | Desmin-related myopathy with Mallory body-like inclusions | | 5 Cases |
| 873 | Desmoid tumor | 0.3 *I* * | |
| 83469 | Desmoplastic small round cell tumor | | 300 Cases |
| 251863 | Desmoplastic/nodular medulloblastoma | 0.01 *I* * | |
| 35107 | Desmosterolosis | | 9 Cases |
| 313892 | Developmental and speech delay due to SOX5 deficiency | | 12 Cases |
| 329195 | Developmental delay with autism spectrum disorder and gait instability | | 22 Cases |
| 163988 | Developmental delay-deafness syndrome, Hildebrand type | | 1 Family |
| 369891 | Developmental delay-facial dysmorphism syndrome due to MED13L deficiency | | 4 Cases |
| 79107 | Developmental malformations-deafness-dystonia syndrome | | 2 Cases |
| 275523 | Dianzani autoimmune lymphoproliferative disease | | 30 Cases |
| 494444 | DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome | | 8 Cases |
| 66637 | Diaphanospondylodysostosis | | 18 Cases |
| 2141 | Diaphragmatic defect-limb deficiency-skull defect syndrome | | 7 Cases |
| 527468 | Diaphragmatic hernia-short bowel-asplenia syndrome | | 2 Cases |
| 628 | Diastrophic dwarfism | 1.2 *P* * | |
| 628 | Diastrophic dwarfism | 0.3 *BP* * | |
| 370046 | Didymosis aplasticosebacea | | 18 Cases |
| 146 | Differentiated thyroid carcinoma | 5.25 *I* | |
| 90060 | Diffuse alveolar hemorrhage | 1.0 *P* * | |
| 404437 | Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome | | 4 Cases |
| 79456 | Diffuse cutaneous mastocytosis | | 30 Cases |
| 544 | Diffuse large B-cell lymphoma | 16.0 *P* * | |
| 544 | Diffuse large B-cell lymphoma | 2.79 *I* * | |
| 2123 | Diffuse neonatal hemangiomatosis | | 70 Cases |
| 86918 | Diffuse palmoplantar keratoderma-acrocyanosis syndrome | | 10 Cases |
| 352487 | Digital anomalies-intellectual disability-short stature syndrome | | 8 Cases |
| 2926 | Digital extensor muscle aplasia-polyneuropathy | | 3 Cases |
| 1146 | Digitotalar dysmorphism | 10.0 *P* | |
| 226 | Dihydropteridine reductase deficiency | | 150 Cases |
| 2229 | Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome | | 20 Families |
| 243343 | Dimethylglycine dehydrogenase deficiency | | 1 Case |

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| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 227 | Diphallia | 0.02 *BP* | |
| 1681 | Diprosopus | | 33 Cases |
| 2412 | Dislocation of the hip-dysmorphism syndrome | | 4 Cases |
| 79168 | Disorder of bile acid synthesis | 0.6 *P* * | |
| 2983 | Disorder of sex development-intellectual disability syndrome | | 3 Cases |
| 71274 | Disseminated peritoneal leiomyomatosis | | 150 Cases |
| 254351 | Distal 7q11.23 microdeletion syndrome | | 41 Cases |
| 261102 | Distal 7q11.23 microduplication syndrome | | 5 Cases |
| 319171 | Distal 17p13.1 microdeletion syndrome | | 16 Cases |
| 261257 | Distal 17p13.3 microdeletion syndrome | | 16 Cases |
| 399096 | Distal anoctaminopathy | | 24 Cases |
| 329457 | Distal arthrogryposis type 5D | | 33 Cases |
| 251515 | Distal arthrogryposis type 10 | | 53 Cases |
| 139525 | Distal hereditary motor neuropathy type 2 | | 4 Families |
| 139552 | Distal hereditary motor neuropathy, Jerash type | | 30 Cases |
| 1307 | Distal limb deficiencies-micrognathia syndrome | | 6 Cases |
| 1620 | Distal monosomy 3p | | 34 Cases |
| 96125 | Distal monosomy 6p | | 35 Cases |
| 1642 | Distal monosomy 9p | | 89 Cases |
| 96148 | Distal monosomy 10q | | 40 Cases |
| 280325 | Distal monosomy 12p | | 8 Cases |
| 1590 | Distal monosomy 13q | | 150 Cases |
| 1596 | Distal monosomy 15q | | 30 Cases |
| 178400 | Distal myopathy with anterior tibial onset | | 4 Cases |
| 63273 | Distal myopathy with posterior leg and anterior hand involvement | | 16 Cases |
| 488650 | Distal myopathy, Tateyama type | | 7 Cases |
| 399103 | Distal nebulin myopathy | | 13 Cases |
| 139547 | Distal spinal muscular atrophy type 3 | | 28 Cases |
| 3248 | Distal symphalangism | | 8 Families |
| 314588 | Distal tetrasomy 15q | | 23 Cases |
| 1745 | Distal trisomy 6p | | 40 Cases |
| 96102 | Distal trisomy 10q | | 40 Cases |
| 293939 | Distal Xq28 microduplication syndrome | | 9 Cases |
| 404546 | DITRA | | 70 Cases |
| 91131 | DK1-CDG | | 17 Cases |
| 352470 | DNA2-related mitochondrial DNA deletion syndrome | | 4 Cases |
| 443950 | DNAJB2-related Charcot-Marie-Tooth disease type 2 | | 2 Cases |
| 330050 | DNM1L-related encephalopathy due to mitochondrial and peroxisomal fission defect | | 11 Cases |
| 3262 | Dobrow syndrome | | 2 Cases |
| 447737 | DOCK2 deficiency | | 5 Cases |
| 158676 | Dominant dystrophic epidermolysis bullosa, nails only | | 10 Families |
| 244305 | Dominant hypophosphatemia with nephrolithiasis or osteoporosis | | 12 Cases |
| 2143 | Donnai-Barrow syndrome | | 50 Cases |
| 79500 | DOORS syndrome | | 50 Cases |
| 255 | Dopa-responsive dystonia | 0.5 *P* | |
| 70594 | Dopa-responsive dystonia due to sepiapterin reductase deficiency | | 43 Cases |
| 230 | Dopamine beta-hydroxylase deficiency | | 21 Cases |
| 3427 | Double outlet left ventricle | 0.5 *BP* | |
| 3411 | Double uterus-hemivagina-renal agenesis syndrome | | 60 Cases |
| 870 | Down syndrome | 95.0 *BP* | |
| 86309 | DPAGT1-CDG | | 18 Cases |
| 79322 | DPM1-CDG | | 9 Cases |
| 263494 | DPM3-CDG | | 1 Case |
| 33069 | Dravet syndrome | 2.5 *BP* | |
| 50817 | Duane anomaly-myopathy-scoliosis syndrome | | 2 Cases |
| 233 | Duane retraction syndrome | 10.0 *P* * | |
| 235 | Dubowitz syndrome | 0.2 *BP* * | |
| 98896 | Duchenne muscular dystrophy | 4.78 *P* | |
| 98896 | Duchenne muscular dystrophy | 15.1 *BP* * | |
| 1203 | Duodenal atresia | 9.0 *P* * | |
| 1203 | Duodenal atresia | 9.0 *BP* * | |
| 314621 | Duplication of the pituitary gland | | 38 Cases |
| 237 | Duplication of urethra | | 300 Cases |
| 239 | Dyggve-Melchior-Clausen disease | | 60 Cases |
| 464306 | DYRK1A-related intellectual disability syndrome | | 54 Cases |
| 268261 | DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion | | 19 Cases |
| 1765 | Dyschondrosteosis-nephritis syndrome | | 1 Family |
| 41 | Dyschromatosis symmetrica hereditaria | | 300 Cases |
| 1766 | Dysequilibrium syndrome | | 51 Cases |
| 1775 | Dyskeratosis congenita | 0.1 *P* * | |

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|---|---|---|---|
| 2104 | Dysmorphism-pectus carinatum-joint laxity syndrome | | 2 Cases |
| 2282 | Dysmorphism-short stature-deafness-disorder of sex development syndrome | | 2 Cases |
| 1822 | Dysplasia epiphysealis hemimelica | 0.1 *I* | |
| 2204 | Dysplastic cortical hyperostosis | | 2 Cases |
| 2476 | Dysraphism-cleft lip/palate-limb reduction defects syndrome | | 3 Cases |
| 85198 | Dysspondyloenchondromatosis | | 16 Cases |
| 210571 | Dystonia 16 | | 12 Cases |
| 412217 | Dystonia-aphonia syndrome | | 32 Cases |
| 521406 | Dystonia-parkinsonism-hypermanganesemia syndrome | | 11 Cases |
| 303 | Dystrophic epidermolysis bullosa | 0.7 *P* | |
| 89843 | Dystrophic epidermolysis bullosa pruriginosa | | 100 Families |
| 2554 | Ear-patella-short stature syndrome | | 67 Cases |
| 1935 | Early myoclonic encephalopathy | | 80 Cases |
| 488635 | Early-onset epilepsy-intellectual disability-brain anomalies syndrome | | 5 Cases |
| 411986 | Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome | | 3 Cases |
| 494348 | Early-onset familial noncirrhotic portal hypertension | | 3 Cases |
| 256 | Early-onset generalized limb-onset dystonia | 0.4 *P* * | |
| 324290 | Early-onset Lafora body disease | | 3 Cases |
| 439212 | Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome | | 13 Cases |
| 2379 | Early-onset parkinsonism-intellectual disability syndrome | | 2 Families |
| 496641 | Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome | | 22 Cases |
| 1943 | Early-onset progressive encephalopathy with migrant continuous myoclonus | | 3 Cases |
| 500144 | Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome | | 3 Cases |
| 496756 | Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome | | 6 Cases |
| 352654 | Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome | | 6 Cases |
| 505237 | Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome | | 12 Cases |
| 313772 | Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome | | 2 Cases |
| 98890 | Early-onset X-linked optic atrophy | | 4 Families |
| 199343 | EAST syndrome | | 26 Cases |
| 391320 | East Texas bleeding disorder | | 19 Cases |
| 319218 | Ebola hemorrhagic fever | | 28220 Cases |
| 1880 | Ebstein malformation | 1.25 *P* * | |
| 1880 | Ebstein malformation | 3.5 *BP* * | |
| 1818 | Ectodermal dysplasia, trichoodontoonychial type | | 7 Cases |
| 1806 | Ectodermal dysplasia-blindness syndrome | | 2 Cases |
| 247827 | Ectodermal dysplasia-cutaneous syndactyly syndrome | | 4 Cases |
| 1883 | Ectodermal dysplasia-sensorineural deafness syndrome | | 2 Cases |
| 247820 | Ectodermal dysplasia-syndactyly syndrome | | 22 Cases |
| 448270 | Ectopia cordis | 0.67 *BP* | |
| 1884 | Ectopia lentis-chorioretinal dystrophy-myopia syndrome | | 4 Cases |
| 1892 | Ectrodactyly-polydactyly syndrome | | 1 Family |
| 1894 | Ectrodactyly-spina bifida-cardiopathy syndrome | | 1 Case |
| 293936 | EDICT syndrome | | 4 Families |
| 1895 | Edinburgh malformation syndrome | | 2 Families |
| 1896 | EEC syndrome | 1.11 *BP* * | |
| 1897 | EEM syndrome | | 7 Families |
| 98249 | Ehlers-Danlos syndrome | 0.9 *BP* * | |
| 230839 | Ehlers-Danlos syndrome due to tenascin-X deficiency | | 17 Cases |
| 90309 | Ehlers-Danlos syndrome type 1 | 5.0 *P* * | |
| 230851 | Ehlers-Danlos syndrome, cardiac valvular type | | 6 Cases |
| 287 | Ehlers-Danlos syndrome, classic type | 5.0 *P* | |
| 1901 | Ehlers-Danlos syndrome, dermatosparaxis type | | 15 Cases |
| 75501 | Ehlers-Danlos syndrome, fibronectinemic type | | 1 Family |
| 285 | Ehlers-Danlos syndrome, hypermobility type | 12.5 *P* * | |
| 300179 | Ehlers-Danlos syndrome, kyphoscoliotic and deafness type | | 9 Cases |
| 1900 | Ehlers-Danlos syndrome, kyphoscoliotic type | 1.0 *BP* | |
| 2953 | Ehlers-Danlos syndrome, musculocontractural type | | 34 Cases |
| 75392 | Ehlers-Danlos syndrome, periodontitis type | | 62 Cases |
| 157965 | Ehlers-Danlos syndrome, spondylocheirodysplastic type | | 6 Cases |

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|---|---|---|---|
| 286 | Ehlers-Danlos syndrome, vascular type | 1.0 *P* * | |
| 230845 | Ehlers-Danlos syndrome, vascular-like type | | 3 Cases |
| 1902 | Ehrlichiosis | | 50 Cases |
| 79106 | Eiken syndrome | | 6 Cases |
| 228240 | Elastoderma | | 5 Cases |
| 289 | Ellis Van Creveld syndrome | 1.1 *BP* | |
| 96170 | Emanuel syndrome | | 350 Cases |
| 180226 | Embryonal carcinoma | 0.01 *I* * | |
| 251852 | Embryonal tumor of neuroepithelial tissue | 0.22 *I* * | |
| 261 | Emery-Dreifuss muscular dystrophy | 0.3 *P* * | |
| 1927 | Emery-Nelson syndrome | | 2 Cases |
| 485418 | EMILIN-1-related connective tissue disease | | 3 Cases |
| 1031 | Enamel-renal syndrome | | 11 Cases |
| 2396 | Encephalocraniocutaneous lipomatosis | | 77 Cases |
| 527276 | Encephalopathy due to mitochondrial and peroxisomal fission defect | | 15 Cases |
| 139406 | Encephalopathy due to prosaposin deficiency | | 10 Cases |
| 833 | Encephalopathy due to sulfite oxidase deficiency | | 100 Cases |
| 319678 | Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome | | 1 Case |
| 296 | Enchondromatosis | 1.0 *P* * | |
| 199332 | Endocrine-cerebro-osteodysplasia syndrome | | 7 Cases |
| 454723 | Endometrioid carcinoma of ovary | 0.81 *I* * | |
| 2790 | Endosteal hyperostosis, Worth type | | 6 Families |
| 85186 | Endosteal sclerosis-cerebellar hypoplasia syndrome | | 4 Cases |
| 1937 | Eng-Strom syndrome | | 2 Cases |
| 60015 | Enlarged parietal foramina | 3.7 *P* | |
| 83620 | Enteric anendocrinosis | | 7 Cases |
| 85438 | Enthesitis-related juvenile idiopathic arthritis | 5.7 *P* * | |
| 449566 | Eosinophilic angiocentric fibrosis | | 52 Cases |
| 402035 | Eosinophilic colitis | | 196 Cases |
| 73247 | Eosinophilic esophagitis | 40.08 *P* | |
| 3165 | Eosinophilic fasciitis | | 200 Cases |
| 2070 | Eosinophilic gastroenteritis | | 280 Cases |
| 183 | Eosinophilic granulomatosis with polyangiitis | 1.5 *P* | |
| 183 | Eosinophilic granulomatosis with polyangiitis | 0.18 *I* * | |

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|---|---|---|---|
| 301 | Ependymal tumor | 0.2 *I* * | |
| 251636 | Ependymoma | 0.16 *I* * | |
| 231742 | Epibulbar lipodermoid-preauricular appendage-polythelia syndrome | | 1 Family |
| 35125 | Epidermal nevus syndrome | | 400 Cases |
| 302 | Epidermodysplasia verruciformis | | 200 Cases |
| 304 | Epidermolysis bullosa simplex | 1.8 *P* | |
| 304 | Epidermolysis bullosa simplex | 2.2 *BP* * | |
| 412181 | Epidermolysis bullosa simplex due to BP230 deficiency | | 2 Cases |
| 412189 | Epidermolysis bullosa simplex due to exophilin 5 deficiency | | 3 Cases |
| 257 | Epidermolysis bullosa simplex with muscular dystrophy | | 40 Cases |
| 89838 | Epidermolysis bullosa simplex, autosomal recessive K14 | | 19 Cases |
| 79401 | Epidermolysis bullosa simplex, Ogna type | | 6 Families |
| 141077 | Epignathus | 1.68 *BP* | |
| 1948 | Epilepsy-microcephaly-skeletal dysplasia syndrome | | 2 Cases |
| 1951 | Epilepsy-telangiectasia syndrome | | 6 Cases |
| 1819 | Epimetaphyseal skeletal dysplasia | | 4 Cases |
| 1825 | Epiphyseal dysplasia-hearing loss-dysmorphism syndrome | | 2 Cases |
| 79135 | Episodic ataxia type 3 | | 1 Family |
| 79136 | Episodic ataxia type 4 | | 2 Families |
| 211067 | Episodic ataxia type 5 | | 7 Cases |
| 209967 | Episodic ataxia type 6 | | 4 Cases |
| 209970 | Episodic ataxia type 7 | | 7 Cases |
| 401953 | Episodic ataxia with slurred speech | | 13 Cases |
| 293381 | Epithelial recurrent erosion dystrophy | | 186 Cases |
| 313920 | Epstein-Barr virus-associated gastric carcinoma | 1.2 *I* | |
| 35687 | Erdheim-Chester disease | | 500 Cases |
| 999 | Ermine phenotype | | 6 Cases |
| 317 | Erythrokeratodermia variabilis | | 200 Cases |
| 476096 | Erythrokeratodermia-cardiomyopathy syndrome | | 3 Cases |
| 1199 | Esophageal atresia | 24.3 *BP* * | |
| 3318 | Essential thrombocythemia | 0.48 *I* * | |
| 1957 | Esthesioneuroblastoma | 0.02 *I* * | |
| 785 | Estrogen resistance syndrome | | 2 Cases |
| 51188 | Ethylmalonic encephalopathy | | 80 Cases |
| 1959 | Evans syndrome | 0.1 *P* * | |
| 496751 | EVEN-plus syndrome | | 3 Cases |
| 319 | Ewing sarcoma | 0.13 *I* * | |

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|---|---|---|---|
| 1962 | Exostoses-anetodermia-brachydactyly type E syndrome | | 1 Family |
| 3294 | Extensor tendons of finger anomalies | | 2 Cases |
| 3023 | External auditory canal atresia-vertical talus-hypertelorism syndrome | | 10 Cases |
| 363579 | Extragonadal germ cell tumor | 0.13 *I* * | |
| 209916 | Extraskeletal myxoid chondrosarcoma | 0.2 *P* * | |
| 1964 | Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome | | 2 Cases |
| 2725 | Eye defects-arachnodactyly-cardiopathy syndrome | | 6 Cases |
| 3172 | Eyebrow duplication-syndactyly syndrome | | 3 Cases |
| 324 | Fabry disease | 0.22 *BP* * | |
| 1969 | Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome | | 3 Cases |
| 284169 | Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion | | 19 Cases |
| 466950 | Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation | | 10 Cases |
| 352712 | Facial dysmorphism-immunodeficiency-livedo-short stature syndrome | | 11 Cases |
| 412022 | Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome | | 4 Families |
| 1970 | Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome | | 3 Cases |
| 1778 | Facial dysmorphism-shawl scrotum-joint laxity syndrome | | 2 Cases |
| 85162 | Facial onset sensory and motor neuronopathy | | 47 Cases |
| 1973 | Faciocardiorenal syndrome | | 4 Cases |
| 269 | Facioscapulohumeral dystrophy | 4.5 *P* * | |
| 306550 | FADD-related immunodeficiency | | 4 Cases |
| 3304 | Fallot complex-intellectual disability-growth delay syndrome | | 5 Cases |
| 88619 | Familial acute necrotizing encephalopathy | | 11 Cases |
| 733 | Familial adenomatous polyposis | 6.0 *P* * | |
| 95700 | Familial adrenal hypoplasia with absent pituitary luteinizing hormone | | 3 Cases |
| 280397 | Familial Alzheimer-like prion disease | | 2 Cases |
| 228277 | Familial anetoderma | | 12 Families |
| 615 | Familial atrial myxoma | | 17 Families |
| 436242 | Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease | | 7 Cases |
| 1551 | Familial benign copper deficiency | | 1 Family |
| 1416 | Familial calcium pyrophosphate deposition | | 100 Families |
| 1768 | Familial caudal dysgenesis | | 4 Cases |
| 464760 | Familial cavitary optic disc anomaly | | 17 Cases |
| 221061 | Familial cerebral cavernous malformation | 15.0 *P* | |
| 481662 | Familial Chilblain lupus | | 10 Families |
| 1428 | Familial chondromalacia patellae | | 5 Families |
| 444490 | Familial chylomicronemia syndrome | 1.0 *P* * | |
| 238578 | Familial clubfoot due to 17q23.1q23.2 microduplication | | 4 Families |
| 238722 | Familial congenital mirror movements | | 75 Cases |
| 451612 | Familial congenital nasolacrimal duct obstruction | | 4 Cases |
| 91498 | Familial congenital palsy of trochlear nerve | | 6 Cases |
| 319189 | Familial cortical myoclonus | | 11 Cases |
| 53296 | Familial cutaneous collagenoma | | 16 Cases |
| 313846 | Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome | | 24 Cases |
| 1799 | Familial developmental dysphasia | | 6 Families |
| 324588 | Familial dyskinesia and facial myokymia | | 18 Cases |
| 85110 | Familial encephalopathy with neuroserpin inclusion bodies | | 6 Families |
| 391392 | Familial episodic pain syndrome with predominantly lower limb involvement | | 28 Cases |
| 391389 | Familial episodic pain syndrome with predominantly upper body involvement | | 21 Cases |
| 464756 | Familial gastric type 1 neuroendocrine tumor | | 5 Cases |
| 251274 | Familial hyperaldosteronism type III | | 7 Families |
| 238475 | Familial hypercholanemia | | 23 Cases |
| 99764 | Familial hyperreninemic hypoaldosteronism type 2 | | 5 Cases |
| 424 | Familial hyperthyroidism due to mutations in TSH receptor | | 28 Families |
| 93372 | Familial hypocalciuric hypercalcemia type 1 | 5.5 *P* | |
| 300373 | Familial infantile gigantism | | 3 Cases |
| 352582 | Familial infantile myoclonic epilepsy | | 7 Cases |
| 154 | Familial isolated dilated cardiomyopathy | 17.5 *P* * | |

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|---|---|---|---|
| 154 | Familial isolated dilated cardiomyopathy | 2.91 *I* * | |
| 99879 | Familial isolated hyperparathyroidism | | 100 Families |
| 2238 | Familial isolated hypoparathyroidism | | 10 Families |
| 2239 | Familial isolated hypoparathyroidism due to agenesis of parathyroid gland | | 2 Families |
| 314777 | Familial isolated pituitary adenoma | | 150 Cases |
| 75249 | Familial isolated restrictive cardiomyopathy | 2.5 *P* * | |
| 411788 | Familial isolated trichomegaly | | 2 Families |
| 79293 | Familial LCAT deficiency | | 70 Cases |
| 768 | Familial long QT syndrome | 40.0 *BP* * | |
| 401942 | Familial median cleft of the upper and lower lips | | 8 Cases |
| 618 | Familial melanoma | 1.5 *I* * | |
| 165805 | Familial mesial temporal lobe epilepsy with febrile seizures | | 4 Cases |
| 495930 | Familial monosomy 7 syndrome | | 14 Families |
| 338 | Familial multiple fibrofolliculoma | | 7 Cases |
| 922 | Familial nasal acilia | | 8 Cases |
| 280403 | Familial omphalocele syndrome with facial dysmorphism | | 5 Cases |
| 569 | Familial or sporadic hemiplegic migraine | 10.0 *P* * | |
| 2769 | Familial osteodysplasia, Anderson type | | 4 Cases |
| 97290 | Familial papillary thyroid carcinoma with renal papillary neoplasia | | 2 Cases |
| 98306 | Familial partial lipodystrophy | 1.0 *P* * | |
| 79084 | Familial partial lipodystrophy, Köbberling type | | 20 Cases |
| 34526 | Familial primary hypomagnesemia | | 500 Cases |
| 306516 | Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis | | 200 Cases |
| 2196 | Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement | | 72 Cases |
| 31043 | Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement | | 110 Cases |
| 34527 | Familial primary hypomagnesemia with normocalciuria and normocalcemia | | 5 Families |
| 871 | Familial progressive cardiac conduction defect | | 50 Cases |
| 280628 | Familial progressive hyper- and hypopigmentation | | 3 Families |
| 488197 | Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome | | 9 Cases |
| 79147 | Familial reactive perforating collagenosis | | 50 Cases |
| 231108 | Familial rhabdoid tumor | | 5 Families |
| 168624 | Familial scaphocephaly syndrome, McGillivray type | | 11 Cases |
| 166282 | Familial sick sinus syndrome | | 11 Cases |
| 280406 | Familial steroid-resistant nephrotic syndrome with sensorineural deafness | | 13 Cases |
| 91387 | Familial thoracic aortic aneurysm and aortic dissection | | 22 Cases |
| 93953 | Familial thyroglossal duct cyst | | 22 Cases |
| 95716 | Familial thyroid dyshormonogenesis | 4.0 *P* * | |
| 95716 | Familial thyroid dyshormonogenesis | 2.67 *I* | |
| 84 | Fanconi anemia | 0.3 *P* | |
| 84 | Fanconi anemia | 0.62 *BP* * | |
| 333 | Farber disease | | 96 Cases |
| 166105 | FASTKD2-related infantile mitochondrial encephalomyopathy | | 3 Cases |
| 439854 | Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease | | 10 Cases |
| 466 | Fatal familial insomnia | | 27 Cases |
| 280553 | Fatal infantile hypertonic myofibrillar myopathy | | 11 Cases |
| 168566 | Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3 | | 7 Cases |
| 391343 | Fatal post-viral neurodegenerative disorder | | 2 Cases |
| 2492 | FATCO syndrome | | 22 Cases |
| 404451 | FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome | | 3 Cases |
| 163703 | Febrile infection-related epilepsy syndrome | 1.0 *P* * | |
| 1305 | Feingold syndrome | | 123 Cases |
| 391641 | Feingold syndrome type 1 | | 120 Cases |
| 391646 | Feingold syndrome type 2 | | 7 Cases |
| 488191 | Female infertility due to oocyte meiotic arrest | | 16 Cases |
| 404466 | Female infertility due to zona pellucida defect | | 4 Cases |
| 101039 | Female restricted epilepsy with intellectual disability | | 5 Families |
| 1988 | Femoral-facial syndrome | | 62 Cases |
| 2019 | Femur-fibula-ulna complex | 1.5 *BP* * | |
| 397922 | Ferro-cerebro-cutaneous syndrome | | 3 Cases |

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|---|---|---|---|
| 994 | Fetal akinesia deformation sequence | 0.6 *BP* * | |
| 363409 | Fetal akinesia-cerebral and retinal hemorrhage syndrome | | 3 Cases |
| 1915 | Fetal alcohol syndrome | 1.6 *BP* * | |
| 294 | Fetal cytomegalovirus syndrome | 40.0 *P* * | |
| 465824 | Fetal encasement syndrome | | 2 Cases |
| 85212 | Fetal Gaucher disease | | 50 Cases |
| 1917 | Fetal methylmercury syndrome | | 800 Cases |
| 1906 | Fetal valproate syndrome | 1.02 *BP* * | |
| 464724 | Fever-associated acute infantile liver failure syndrome | | 11 Cases |
| 313855 | FGFR2-related bent bone dysplasia | | 11 Cases |
| 477650 | Fibroblastic rheumatism | | 30 Cases |
| 2021 | Fibrochondrogenesis | | 20 Cases |
| 337 | Fibrodysplasia ossificans progressiva | 0.05 *P* | |
| 84090 | Fibronectin glomerulopathy | | 16 Families |
| 2030 | Fibrosarcoma | 0.01 *I* * | |
| 1118 | Fibular aplasia-ectrodactyly syndrome | | 50 Cases |
| 1757 | Fibular dimelia-diplopodia syndrome | | 11 Cases |
| 93323 | Fibular hemimelia | 2.0 *P* * | |
| 2256 | Fibulo-ulnar hypoplasia-renal anomalies syndrome | | 2 Cases |
| 3255 | Filippi syndrome | | 29 Cases |
| 369979 | Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome | | 2 Cases |
| 97232 | Fingerprint body myopathy | | 20 Cases |
| 399086 | Finnish upper limb-onset distal myopathy | | 7 Cases |
| 79292 | Fish-eye disease | | 30 Cases |
| 1968 | Flat face-microstomia-ear anomaly syndrome | | 2 Cases |
| 98970 | Fleck corneal dystrophy | | 30 Cases |
| 2044 | Floating-Harbor syndrome | | 87 Cases |
| 2045 | FLOTCH syndrome | | 6 Families |
| 2047 | Flynn-Aird syndrome | | 10 Cases |
| 2092 | Focal dermal hypoplasia | | 300 Cases |
| 352587 | Focal epilepsy-intellectual disability-cerebro-cerebellar malformation | | 7 Cases |
| 398166 | Focal facial dermal dysplasia | | 147 Cases |
| 79133 | Focal facial dermal dysplasia type I | | 81 Cases |
| 398173 | Focal facial dermal dysplasia type II | | 22 Cases |
| 1807 | Focal facial dermal dysplasia type III | | 20 Cases |
| 398189 | Focal facial dermal dysplasia type IV | | 21 Cases |
| 48918 | Focal myositis | | 115 Cases |
| 1866 | Focal, segmental or multifocal dystonia | 11.7 *P* * | |
| 1866 | Focal, segmental or multifocal dystonia | 2.0 *I* * | |
| 2048 | Foix-Chavany-Marie syndrome | | 150 Cases |
| 300552 | Follicular cholangitis and pancreatitis | | 5 Cases |
| 545 | Follicular lymphoma | 28.0 *P* * | |
| 545 | Follicular lymphoma | 2.12 *I* * | |
| 228371 | Foodborne botulism | 0.1 *I* * | |
| 3219 | Fountain syndrome | | 8 Cases |
| 397618 | Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome | | 7 Families |
| 2253 | Foveal hypoplasia-presenile cataract syndrome | | 11 Cases |
| 221126 | Fowler syndrome | | 44 Cases |
| 908 | Fragile X syndrome | 32.5 *P* | |
| 908 | Fragile X syndrome | 2.4 *BP* * | |
| 137834 | Frank-Ter Haar syndrome | | 30 Cases |
| 2052 | Fraser syndrome | 0.2 *BP* * | |
| 347 | Frasier syndrome | | 88 Cases |
| 834 | Free sialic acid storage disease | | 130 Cases |
| 2053 | Freeman-Sheldon syndrome | | 100 Cases |
| 85335 | Fried syndrome | | 1 Family |
| 95 | Friedreich ataxia | 2.0 *P* * | |
| 99672 | Fried's tooth and nail syndrome | | 12 Cases |
| 1791 | Frontofacionasal dysplasia | | 14 Cases |
| 1826 | Frontometaphyseal dysplasia | | 100 Cases |
| 250 | Frontonasal dysplasia | 0.7 *BP* * | |
| 228390 | Frontonasal dysplasia-alopecia-genital anomalies syndrome | | 5 Cases |
| 521308 | Frontonasal dysplasia-bifid nose-upper limb anomalies syndrome | | 11 Cases |
| 306542 | Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome | | 3 Cases |
| 282 | Frontotemporal dementia | 3.0 *P* * | |
| 293848 | Frontotemporal dementia, right temporal atrophy variant | | 200 Cases |
| 2059 | Fryns syndrome | 7.0 *BP* * | |
| 2058 | Fryns-Smeets-Thiry syndrome | | 2 Cases |
| 247790 | FTH1-related iron overload | | 4 Cases |
| 349 | Fucosidosis | | 100 Cases |
| 2854 | Fuhrmann syndrome | | 11 Cases |

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|---|---|---|---|
| 24 | Fumaric aciduria | | 40 Cases |
| 506358 | Gabriele-de Vries syndrome | | 10 Cases |
| 352 | Galactosemia | 2.0 *BP* * | |
| 352 | Galactosemia | 2.1 *I* * | |
| 351 | Galactosialidosis | | 100 Cases |
| 2065 | Galloway-Mowat syndrome | | 97 Cases |
| 2066 | Gamma-aminobutyric acid transaminase deficiency | | 3 Families |
| 33573 | Gamma-glutamyl transpeptidase deficiency | | 7 Cases |
| 100026 | Gamma-heavy chain disease | | 120 Cases |
| 2067 | GAPO syndrome | | 38 Cases |
| 79665 | Gardner syndrome | 9.1 *BP* | |
| 314022 | Gastric adenocarcinoma and proximal polyposis of the stomach | | 28 Cases |
| 2069 | Gastrocutaneous syndrome | | 24 Cases |
| 44890 | Gastrointestinal stromal tumor | 13.0 *P* * | |
| 44890 | Gastrointestinal stromal tumor | 1.0 *I* | |
| 2368 | Gastroschisis | 23.7 *BP* * | |
| 355 | Gaucher disease | 1.0 *P* * | |
| 355 | Gaucher disease | 1.3 *BP* | |
| 355 | Gaucher disease | 1.7 *I* * | |
| 77259 | Gaucher disease type 1 | 1.0 *P* * | |
| 77260 | Gaucher disease type 2 | 0.01 *P* * | |
| 77261 | Gaucher disease type 3 | 0.05 *P* * | |
| 2072 | Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome | | 10 Cases |
| 438274 | GCGR-related hyperglucagonemia | | 8 Cases |
| 2623 | Geleophysic dysplasia | | 27 Cases |
| 2074 | Gemignani syndrome | | 2 Cases |
| 508529 | Generalized basal epidermolysis bullosa simplex with skin atrophy, scarring and hair loss | | 14 Cases |
| 411777 | Generalized eruptive keratoacanthoma | | 40 Cases |
| 98497 | Genetic peripheral neuropathy | 40.0 *P* | |
| 2075 | Genitopalatocardiac syndrome | | 15 Cases |
| 85201 | Genitopatellar syndrome | | 22 Cases |
| 93398 | Genochondromatosis type 2 | | 10 Cases |
| 2077 | German syndrome | | 5 Cases |
| 2078 | Geroderma osteodysplastica | | 50 Cases |
| 356 | Gerstmann-Straussler-Scheinker syndrome | 0.0055 *I* | |
| 643 | Giant axonal neuropathy | | 50 Families |
| 251579 | Giant cell glioblastoma | 0.02 *I* * | |
| 2025 | Gingival fibromatosis-facial dysmorphism syndrome | | 2 Cases |
| 2027 | Gingival fibromatosis-progressive deafness syndrome | | 2 Families |
| 358 | Gitelman syndrome | 2.5 *P* * | |
| 238763 | Glaucoma secondary to spherophakia/ectopia lentis and megalocornea | | 12 Cases |
| 2084 | Glaucoma-ectopia lentis-microspherophakia-stiff joints-short stature syndrome | | 3 Cases |
| 2085 | Glaucoma-sleep apnea syndrome | | 5 Cases |
| 182067 | Glial tumor | 10.0 *P* * | |
| 182067 | Glial tumor | 5.35 *I* * | |
| 360 | Glioblastoma | 1.0 *P* | |
| 360 | Glioblastoma | 3.0 *I* | |
| 251582 | Gliomatosis cerebri | 0.01 *I* * | |
| 251576 | Gliosarcoma | 0.03 *I* * | |
| 404476 | Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome | | 2 Cases |
| 488613 | Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome | | 26 Cases |
| 73223 | Global developmental delay-osteopenia-ectodermal defect syndrome | | 3 Cases |
| 480898 | Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome | | 6 Cases |
| 141163 | Glossopalatine ankylosis | | 30 Cases |
| 97280 | Glucagonoma | 0.005 *I* * | |
| 33574 | Glutamate-cysteine ligase deficiency | | 10 Cases |
| 25 | Glutaryl-CoA dehydrogenase deficiency | 1.0 *BP* | |
| 32 | Glutathione synthetase deficiency | | 70 Cases |
| 407 | Glycine encephalopathy | 0.17 *P* * | |
| 365 | Glycogen storage disease due to acid maltase deficiency | 0.8 *BP* * | |
| 420429 | Glycogen storage disease due to acid maltase deficiency, late-onset | 1.75 *BP* | |
| 364 | Glycogen storage disease due to glucose-6-phosphatase deficiency | 1.0 *BP* | |
| 79258 | Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia | 1.0 *BP* * | |
| 79259 | Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib | | 150 Cases |
| 2088 | Glycogen storage disease due to GLUT2 deficiency | | 200 Cases |

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|---|---|---|---|
| 367 | Glycogen storage disease due to glycogen branching enzyme deficiency | 0.1 *BP* | |
| 2089 | Glycogen storage disease due to hepatic glycogen synthase deficiency | | 16 Cases |
| 34587 | Glycogen storage disease due to LAMP-2 deficiency | | 84 Cases |
| 264580 | Glycogen storage disease due to liver phosphorylase kinase deficiency | 1.0 *BP* * | |
| 137625 | Glycogen storage disease due to muscle and heart glycogen synthase deficiency | | 4 Cases |
| 99849 | Glycogen storage disease due to muscle beta-enolase deficiency | | 1 Case |
| 371 | Glycogen storage disease due to muscle phosphofructokinase deficiency | | 100 Cases |
| 715 | Glycogen storage disease due to muscle phosphorylase kinase deficiency | | 30 Cases |
| 713 | Glycogen storage disease due to phosphoglycerate kinase 1 deficiency | | 30 Families |
| 97234 | Glycogen storage disease due to phosphoglycerate mutase deficiency | | 50 Cases |
| 370 | Glycogen storage disease due to phosphorylase kinase deficiency | 1.0 *BP* * | |
| 263297 | Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency | | 1 Case |
| 354 | GM1 gangliosidosis | 0.75 *BP* * | |
| 79255 | GM1 gangliosidosis type 1 | | 200 Cases |
| 79256 | GM1 gangliosidosis type 2 | | 50 Cases |
| 79257 | GM1 gangliosidosis type 3 | | 70 Cases |
| 309152 | GM2 gangliosidosis | 5.0 *P* * | |
| 309246 | GM2 gangliosidosis, AB variant | | 10 Cases |
| 2090 | GMS syndrome | | 1 Family |
| 602 | GNE myopathy | 1.0 *P* | |
| 329984 | Goblet cell carcinoma | 0.025 *I* | |
| 66629 | Goldberg-Shprintzen megacolon syndrome | | 8 Families |
| 374 | Goldenhar syndrome | 2.9 *BP* * | |
| 53540 | Goldmann-Favre syndrome | | 50 Cases |
| 1986 | Gollop-Wolfgang complex | | 200 Cases |
| 1532 | Gómez-López-Hernández syndrome | | 36 Cases |
| 169105 | Good syndrome | | 241 Cases |
| 65798 | Goodman syndrome | | 3 Cases |
| 73 | Gorham-Stout disease | | 300 Cases |
| 377 | Gorlin syndrome | 1.1 *P* | |
| 2095 | Gorlin-Chaudhry-Moss syndrome | | 7 Cases |
| 39812 | Graft versus host disease | 3.0 *P* * | |
| 79094 | Grange syndrome | | 7 Cases |
| 2097 | Grant syndrome | | 1 Family |
| 900 | Granulomatosis with polyangiitis | 9.0 *P* * | |
| 900 | Granulomatosis with polyangiitis | 0.85 *I* * | |
| 33111 | Granulomatous slack skin | | 50 Cases |
| 721 | Gray platelet syndrome | | 60 Cases |
| 293375 | Grayson-Wilbrandt corneal dystrophy | | 1 Family |
| 1426 | Greenberg dysplasia | | 10 Cases |
| 381 | Griscelli syndrome | | 150 Cases |
| 79476 | Griscelli syndrome type 1 | | 20 Cases |
| 79477 | Griscelli syndrome type 2 | | 102 Cases |
| 79478 | Griscelli syndrome type 3 | | 13 Cases |
| 391348 | Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome | | 2 Cases |
| 73272 | Growth delay due to insulin-like growth factor type 1 deficiency | | 5 Cases |
| 3035 | Growth delay-hydrocephaly-lung hypoplasia syndrome | | 4 Cases |
| 391366 | Growth retardation-mild developmental delay-chronic hepatitis syndrome | | 2 Cases |
| 2101 | Grubben-de Cock-Borghgraef syndrome | | 3 Cases |
| 2102 | GTP cyclohydrolase I deficiency | | 16 Cases |
| 382 | Guanidinoacetate methyltransferase deficiency | | 80 Cases |
| 2103 | Guillain-Barré syndrome | 3.5 *P* * | |
| 2103 | Guillain-Barré syndrome | 1.45 *I* | |
| 2957 | Guttmacher syndrome | | 3 Cases |
| 414 | Gyrate atrophy of choroid and retina | | 200 Cases |
| 168569 | H syndrome | | 100 Cases |
| 99803 | Haddad syndrome | | 60 Cases |
| 2342 | Haim-Munk syndrome | | 100 Cases |
| 1408 | Hair defect-photosensitivity-intellectual disability syndrome | | 3 Cases |
| 2107 | Hall-Riggs syndrome | | 8 Cases |
| 2108 | Hallermann-Streiff syndrome | | 150 Cases |
| 2109 | Hallermann-Streiff-like syndrome | | 2 Cases |
| 2110 | Hallux varus-preaxial polysyndactyly syndrome | | 2 Cases |
| 93946 | Hamel cerebro-palato-cardiac syndrome | | 4 Cases |
| 73229 | HANAC syndrome | | 6 Families |

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|---|---|---|---|
| 457 | Harlequin ichthyosis | | 200 Cases |
| 199282 | Harlequin syndrome | | 100 Cases |
| 2115 | Harrod syndrome | | 3 Cases |
| 2116 | Hartnup disease | 4.2 *P* | |
| 2117 | Hartsfield syndrome | | 17 Cases |
| 99872 | Hashimoto-Pritzker syndrome | | 50 Cases |
| 2118 | Hawkinsinuria | | 5 Families |
| 3225 | Hearing loss-familial salivary gland insensitivity to aldosterone syndrome | | 2 Cases |
| 1338 | Heart defect-tongue hamartoma-polysyndactyly syndrome | | 4 Cases |
| 1354 | Heart defects-limb shortening syndrome | | 2 Cases |
| 1350 | Heart-hand syndrome type 2 | | 2 Families |
| 1342 | Heart-hand syndrome type 3 | | 3 Cases |
| 168796 | Heart-hand syndrome, Slovenian type | | 14 Cases |
| 2119 | HEC syndrome | | 2 Cases |
| 178330 | Heinz body anemia | | 10 Cases |
| 86813 | Helicoid peripapillary chorioretinal degeneration | | 100 Cases |
| 90053 | Hematopoietic stem cell transplantation | 0.65 *P* * | |
| 306741 | Hemidystonia-hemiatrophy syndrome | | 100 Cases |
| 141148 | Hemifacial myohyperplasia | | 12 Cases |
| 276280 | Hemihyperplasia-multiple lipomatosis syndrome | | 10 Cases |
| 2130 | Hemimelia | 4.15 *P* * | |
| 306669 | Hemiparkinsonism-hemiatrophy syndrome | | 68 Cases |
| 79230 | Hemochromatosis type 2 | | 74 Cases |
| 225123 | Hemochromatosis type 3 | | 33 Cases |
| 139491 | Hemochromatosis type 4 | | 200 Cases |
| 280615 | Hemoglobinopathy Toms River | | 10 Cases |
| 86817 | Hemolytic anemia due to adenylate kinase deficiency | | 7 Families |
| 712 | Hemolytic anemia due to glucophosphate isomerase deficiency | | 50 Cases |
| 90030 | Hemolytic anemia due to glutathione reductase deficiency | | 3 Cases |
| 766 | Hemolytic anemia due to red cell pyruvate kinase deficiency | 5.0 *P* * | |
| 448 | Hemophilia | 7.7 *P* * | |
| 448 | Hemophilia | 6.25 *I* * | |
| 98878 | Hemophilia A | 4.85 *P* | |
| 98878 | Hemophilia A | 11.25 *BP* | |
| 98879 | Hemophilia B | 1.7 *P* * | |
| 98879 | Hemophilia B | 1.665 *BP* | |
| 178396 | Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation | | 4 Cases |
| 340 | Hemorrhagic fever-renal syndrome | 37.0 *P* * | |
| 340 | Hemorrhagic fever-renal syndrome | 0.74 *I* * | |
| 324632 | Hendra virus infection | | 7 Cases |
| 2136 | Hennekam syndrome | | 50 Cases |
| 2135 | Hennekam-Beemer syndrome | | 3 Cases |
| 2031 | Hepatic fibrosis-renal cysts-intellectual disability syndrome | | 4 Cases |
| 890 | Hepatic veno-occlusive disease | 11.0 *P* * | |
| 79124 | Hepatic veno-occlusive disease-immunodeficiency syndrome | | 28 Cases |
| 90073 | Hepatitis B reinfection following liver transplantation | 2.0 *P* * | |
| 402823 | Hepatitis delta | 40.0 *P* * | |
| 449 | Hepatoblastoma | 0.02 *I* * | |
| 88673 | Hepatocellular carcinoma | 15.0 *P* * | |
| 88673 | Hepatocellular carcinoma | 3.09 *I* * | |
| 137681 | Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1 | | 2 Cases |
| 95159 | Hepatoerythropoietic porphyria | | 40 Cases |
| 91378 | Hereditary angioedema | 1.5 *P* * | |
| 289601 | Hereditary arterial and articular multiple calcification syndrome | | 16 Cases |
| 145 | Hereditary breast and ovarian cancer syndrome | 25.0 *P* * | |
| 1867 | Hereditary bullous dystrophy, macular type | | 2 Families |
| 85458 | Hereditary cerebral hemorrhage with amyloidosis | | 350 Cases |
| 676 | Hereditary chronic pancreatitis | 0.43 *P* * | |
| 98434 | Hereditary combined deficiency of vitamin K-dependent clotting factors | | 30 Families |
| 398088 | Hereditary cryohydrocytosis with normal stomatin | | 53 Cases |
| 168577 | Hereditary cryohydrocytosis with reduced stomatin | | 3 Cases |
| 26106 | Hereditary diffuse gastric cancer | 1.5 *I* * | |
| 313808 | Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia | | 27 Cases |
| 221043 | Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome | | 15 Cases |
| 90045 | Hereditary folate malabsorption | | 30 Cases |
| 469 | Hereditary fructose intolerance | 5.0 *P* * | |
| 774 | Hereditary hemorrhagic telangiectasia | 16.0 *P* * | |

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|---|---|---|---|
| 3197 | Hereditary hyperekplexia | | 150 Cases |
| 163 | Hereditary hyperferritinemia-cataract syndrome | | 64 Cases |
| 217407 | Hereditary hypotrichosis with recurrent skin vesicles | | 4 Cases |
| 324381 | Hereditary inclusion body myopathy type 4 | | 17 Cases |
| 79091 | Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome | | 21 Cases |
| 523 | Hereditary leiomyomatosis and renal cell cancer | | 200 Cases |
| 90117 | Hereditary motor and sensory neuropathy, Okinawa type | | 120 Cases |
| 178464 | Hereditary myopathy with early respiratory failure | | 10 Families |
| 43115 | Hereditary myopathy with lactic acidosis due to ISCU deficiency | | 19 Cases |
| 1062 | Hereditary neurocutaneous malformation | | 9 Families |
| 640 | Hereditary neuropathy with liability to pressure palsies | 3.5 *P* * | |
| 279943 | Hereditary neutrophilia | | 16 Cases |
| 168583 | Hereditary North American Indian childhood cirrhosis | | 36 Cases |
| 30 | Hereditary orotic aciduria | | 20 Cases |
| 79141 | Hereditary painful callosities | | 2 Families |
| 476102 | Hereditary pediatric Behçet-like disease | | 13 Cases |
| 168615 | Hereditary persistence of alpha-fetoprotein | | 19 Families |
| 29072 | Hereditary pheochromocytoma-paraganglioma | 0.3 *I* | |
| 158025 | Hereditary progressive mucinous histiocytosis | | 18 Cases |
| 221039 | Hereditary sclerosing poikiloderma, Weary type | | 9 Cases |
| 280598 | Hereditary sensorimotor neuropathy with hyperelastic skin | | 4 Cases |
| 320385 | Hereditary sensory and autonomic neuropathy due to TECPR2 mutation | | 5 Cases |
| 139564 | Hereditary sensory and autonomic neuropathy type 1B | | 2 Families |
| 970 | Hereditary sensory and autonomic neuropathy type 2 | | 35 Cases |
| 314381 | Hereditary sensory and autonomic neuropathy type 6 | | 4 Cases |
| 391397 | Hereditary sensory and autonomic neuropathy type 7 | | 3 Cases |
| 478664 | Hereditary sensory and autonomic neuropathy type 8 | | 11 Families |
| 139573 | Hereditary sensory and autonomic neuropathy with deafness and global delay | | 4 Cases |
| 456318 | Hereditary sensory neuropathy-deafness-dementia syndrome | | 6 Families |

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|---|---|---|---|
| 685 | Hereditary spastic paraplegia | 5.2 *P* | |
| 84093 | Hereditary thermosensitive neuropathy | | 1 Family |
| 480851 | Hereditary thrombocytopenia with early-onset myelofibrosis | | 9 Cases |
| 3467 | Hereditary xanthinuria | 9.05 *I* * | |
| 275777 | Heritable pulmonary arterial hypertension | 0.08 *P* * | |
| 79430 | Hermansky-Pudlak syndrome | 0.15 *P* | |
| 231531 | Hermansky-Pudlak syndrome type 7 | | 2 Cases |
| 231537 | Hermansky-Pudlak syndrome type 8 | | 6 Cases |
| 280663 | Hermansky-Pudlak syndrome type 9 | | 2 Cases |
| 183678 | Hermansky-Pudlak syndrome with neutropenia | | 40 Cases |
| 1930 | Herpes simplex virus encephalitis | 0.3 *I* | |
| 189 | Hidrotic ectodermal dysplasia | 1.0 *P* * | |
| 1808 | Hidrotic ectodermal dysplasia, Christianson-Fourie type | | 6 Cases |
| 1809 | Hidrotic ectodermal dysplasia, Halal type | | 4 Cases |
| 314029 | High bone mass osteogenesis imperfecta | | 2 Cases |
| 363396 | High myopia-sensorineural deafness syndrome | | 7 Cases |
| 231080 | High-grade dysplasia in patients with Barrett esophagus | 36.0 *P* * | |
| 388 | Hirschsprung disease | 10.9 *BP* * | |
| 2155 | Hirschsprung disease-deafness-polydactyly syndrome | | 2 Cases |
| 2153 | Hirschsprung disease-nail hypoplasia-dysmorphism syndrome | | 3 Cases |
| 2150 | Hirschsprung disease-type D brachydactyly syndrome | | 4 Cases |
| 2158 | Histidinuria-renal tubular defect syndrome | | 5 Cases |
| 98287 | Histiocytic and dendritic cell tumor | 0.05 *I* * | |
| 98293 | Hodgkin lymphoma | 2.4 *I* * | |
| 93970 | Holmes-Gang syndrome | | 3 Cases |
| 79242 | Holocarboxylase synthetase deficiency | 0.5 *BP* * | |
| 2162 | Holoprosencephaly | 13.4 *BP* * | |
| 2163 | Holoprosencephaly-craniosynostosis syndrome | | 11 Cases |
| 3186 | Holoprosencephaly-radial heart renal anomalies syndrome | | 4 Cases |
| 392 | Holt-Oram syndrome | 0.7 *BP* * | |
| 2167 | Holzgreve syndrome | | 3 Cases |
| 622 | Homocystinuria without methylmalonic aciduria | | 73 Cases |
| 391665 | Homozygous familial hypercholesterolemia | 0.1 *P* | |

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|---|---|---|---|
| 3322 | Hoyeraal-Hreidarsson syndrome | | 33 Cases |
| 391417 | HSD10 disease | | 37 Cases |
| 85295 | HSD10 disease, atypical type | | 5 Cases |
| 391457 | HSD10 disease, neonatal type | | 3 Cases |
| 482077 | HTRA1-related autosomal dominant cerebral small vessel disease | | 21 Cases |
| 228116 | Hughes-Stovin syndrome | | 30 Cases |
| 56970 | Human prion disease | 0.3 *P* * | |
| 56970 | Human prion disease | 0.15 *I* * | |
| 3265 | Humero-radial synostosis | | 150 Cases |
| 3266 | Humero-radio-ulnar synostosis | | 30 Cases |
| 94056 | Humero-ulnar synostosis | | 5 Cases |
| 3383 | Humerus trochlea aplasia | | 5 Cases |
| 97340 | Hunter-McAlpine craniosynostosis | | 10 Cases |
| 399 | Huntington disease | 2.7 *P* | |
| 399 | Huntington disease | 0.38 *I* | |
| 98934 | Huntington disease-like 2 | | 50 Families |
| 401901 | Huntington disease-like syndrome due to C9ORF72 expansions | | 10 Cases |
| 93473 | Hurler syndrome | 0.5 *P* * | |
| 93473 | Hurler syndrome | 0.7 *BP* * | |
| 740 | Hutchinson-Gilford progeria syndrome | 0.005 *P* | |
| 740 | Hutchinson-Gilford progeria syndrome | 0.025 *BP* | |
| 498474 | Hyaline fibromatosis syndrome | | 150 Cases |
| 2177 | Hydranencephaly | 10.0 *BP* | |
| 2182 | Hydrocephalus with stenosis of the aqueduct of Sylvius | 1.7 *P* | |
| 2182 | Hydrocephalus with stenosis of the aqueduct of Sylvius | 1.7 *BP* | |
| 2186 | Hydrocephalus-blue sclerae-nephropathy syndrome | | 1 Family |
| 2180 | Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome | | 8 Cases |
| 2183 | Hydrocephalus-obesity-hypogonadism syndrome | | 2 Cases |
| 2184 | Hydrocephaly-low insertion umbilicus syndrome | | 2 Cases |
| 2181 | Hydrocephaly-tall stature-joint laxity syndrome | | 2 Cases |
| 528091 | Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome | | 1 Case |
| 79155 | Hydroxykynureninuria | | 30 Cases |
| 309147 | Hyper-beta-alaninemia | | 2 Cases |

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|---|---|---|---|
| 401948 | Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency | | 4 Cases |
| 168588 | Hyperandrogenism due to cortisone reductase deficiency | | 11 Cases |
| 276405 | Hyperbiliverdinemia | | 2 Cases |
| 209902 | Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency | | 24 Cases |
| 83639 | Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency | | 2 Families |
| 1032 | Hyperdibasic aminoaciduria type 1 | | 26 Cases |
| 163985 | Hyperekplexia-epilepsy syndrome | | 2 Cases |
| 168956 | Hypereosinophilic syndrome | 1.5 *P* * | |
| 2410 | Hypergonadotropic hypogonadism-cataract syndrome | | 3 Cases |
| 343 | Hyperimmunoglobulinemia D with periodic fever | | 200 Cases |
| 324575 | Hyperinsulinism due to HNF1A deficiency | | 2 Cases |
| 263458 | Hyperinsulinism due to INSR deficiency | | 10 Cases |
| 71212 | Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency | | 10 Cases |
| 276556 | Hyperinsulinism due to UCP2 deficiency | | 2 Cases |
| 682 | Hyperkalemic periodic paralysis | 0.5 *P* * | |
| 1336 | Hyperkeratosis-hyperpigmentation syndrome | | 10 Cases |
| 412 | Hyperlipoproteinemia type 3 | 10.0 *P* | |
| 415 | Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome | 12.0 *P* * | |
| 3416 | Hyperostosis corticalis generalisata | | 35 Cases |
| 443098 | Hyperostosis cranialis interna | | 13 Cases |
| 99880 | Hyperparathyroidism-jaw tumor syndrome | | 100 Cases |
| 508523 | Hyperphenylalaninemia due to DNAJC12 deficiency | | 6 Cases |
| 238583 | Hyperphenylalaninemia due to tetrahydrobiopterin deficiency | 0.2 *P* | |
| 247262 | Hyperphosphatasia-intellectual disability syndrome | | 24 Cases |
| 157798 | Hyperplastic polyposis syndrome | 1.0 *I* | |
| 1519 | Hypertelorism, Teebi type | | 25 Cases |
| 2211 | Hypertelorism-hypospadias-polysyndactyly syndrome | | 3 Families |
| 2213 | Hypertelorism-microtia-facial clefting syndrome | | 9 Cases |
| 293958 | Hypertelorism-preauricular sinus-punctual pits-deafness syndrome | | 13 Cases |
| 2220 | Hypertrichosis cubiti | | 28 Cases |

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|---|---|---|---|
| 2222 | Hypertrichosis lanuginosa congenita | | 100 Cases |
| 966 | Hypertrichosis-acromegaloid facial appearance syndrome | | 27 Cases |
| 1517 | Hypertrichotic osteochondrodysplasia, Cantu type | | 50 Cases |
| 324525 | Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation | | 3 Cases |
| 2224 | Hypertryptophanemia | | 12 Cases |
| 363694 | Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome | | 4 Families |
| 251523 | Hyperzincemia and hypercalprotectinemia | | 5 Cases |
| 2435 | Hypo- and hypermelanotic cutaneous macules-retarded growth-intellectual disability syndrome | | 14 Cases |
| 429 | Hypochondroplasia | 3.3 *P* * | |
| 36412 | Hypocomplementemic urticarial vasculitis | | 200 Cases |
| 989 | Hypoglossia-hypodactyly syndrome | | 47 Cases |
| 2233 | Hypogonadism-mitral valve prolapse-intellectual disability syndrome | | 2 Cases |
| 2230 | Hypogonadotropic hypogonadism-frontoparietal alopecia syndrome | | 6 Cases |
| 2235 | Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome | | 2 Cases |
| 293967 | Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome | | 4 Cases |
| 528105 | Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome | | 22 Cases |
| 363523 | Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome | | 12 Cases |
| 238468 | Hypohidrotic ectodermal dysplasia | 6.7 *P* * | |
| 98813 | Hypohidrotic ectodermal dysplasia with immunodeficiency | 0.2 *BP* * | |
| 1882 | Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome | | 3 Cases |
| 293964 | Hypoinsulinemic hypoglycemia and body hemihypertrophy | | 5 Cases |
| 681 | Hypokalemic periodic paralysis | 1.0 *P* * | |
| 1790 | Hypomandibular faciocranial dysostosis | | 3 Cases |
| 137639 | Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome | | 8 Cases |
| 2680 | Hypomyelination neuropathy-arthrogryposis syndrome | | 9 Cases |
| 139441 | Hypomyelination with atrophy of basal ganglia and cerebellum | | 19 Cases |
| 363412 | Hypomyelination with brain stem and spinal cord involvement and leg spasticity | | 13 Cases |
| 447893 | Hypomyelination-cerebellar atrophy-hypoplasia of the corpus callosum syndrome | | 4 Cases |
| 85163 | Hypomyelination-congenital cataract syndrome | | 10 Cases |
| 88637 | Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome | | 105 Cases |
| 2237 | Hypoparathyroidism-sensorineural deafness-renal disease syndrome | | 180 Cases |
| 436 | Hypophosphatasia | 0.21 *BP* * | |
| 324561 | Hypopigmentation-punctate palmoplantar keratoderma syndrome | | 6 Families |
| 722 | Hypoplasminogenemia | 0.2 *P* * | |
| 2248 | Hypoplastic left heart syndrome | 24.0 *BP* | |
| 293864 | Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome | | 16 Cases |
| 98723 | Hypoplastic right heart syndrome | 3.3 *BP* * | |
| 2250 | Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome | | 2 Cases |
| 2261 | Hypospadias-intellectual disability, Goldblatt type syndrome | | 3 Cases |
| 137908 | Hypotonia with lactic acidemia and hyperammonemia | | 4 Cases |
| 163690 | Hypotonia-cystinuria syndrome | | 22 Cases |
| 79507 | Hypotonia-failure to thrive-microcephaly syndrome | | 2 Cases |
| 55654 | Hypotrichosis simplex | | 38 Cases |
| 1573 | Hypotrichosis with juvenile macular degeneration | | 50 Cases |
| 330029 | Hypotrichosis-deafness syndrome | | 1 Case |
| 2266 | Hypotrichosis-intellectual disability, Lopes type | | 2 Cases |
| 69735 | Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome | | 4 Cases |
| 307936 | Hypotrichosis-osteolysis-periodontitis-palmoplantar keratoderma syndrome | | 2 Cases |
| 254509 | Iatrogenic botulism | | 180 Cases |
| 2268 | ICF syndrome | | 66 Cases |
| 2273 | Ichthyosis follicularis-alopecia-photophobia syndrome | | 40 Cases |
| 79503 | Ichthyosis hystrix of Curth-Macklin | | 10 Cases |
| 2269 | Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome | | 4 Cases |

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|---|---|---|---|
| 2274 | Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome | | 2 Cases |
| 91132 | Ichthyosis-hypotrichosis syndrome | | 11 Cases |
| 2278 | Ichthyosis-intellectual disability-dwarfism-renal impairment syndrome | | 4 Cases |
| 2272 | Ichthyosis-oral and digital anomalies syndrome | | 2 Cases |
| 88621 | Ichthyosis-prematurity syndrome | | 16 Families |
| 363992 | Ichthyosis-short stature-brachydactyly-microspherophakia syndrome | | 7 Cases |
| 930 | Idiopathic achalasia | 8.0 *P* | |
| 930 | Idiopathic achalasia | 0.77 *I* | |
| 724 | Idiopathic acute eosinophilic pneumonia | | 100 Cases |
| 139423 | Idiopathic acute transverse myelitis | 0.25 *I* * | |
| 422 | Idiopathic and/or familial pulmonary arterial hypertension | 1.0 *P* * | |
| 88 | Idiopathic aplastic anemia | 0.4 *P* * | |
| 33208 | Idiopathic hypersomnia | 30.0 *P* * | |
| 238624 | Idiopathic intracranial hypertension | 14.0 *P* * | |
| 45452 | Idiopathic neonatal atrial flutter | 1.5 *BP* * | |
| 494428 | Idiopathic pleuroparenchymal fibroelastosis | | 37 Cases |
| 275766 | Idiopathic pulmonary arterial hypertension | 1.1 *P* * | |
| 2032 | Idiopathic pulmonary fibrosis | 11.5 *P* * | |
| 2032 | Idiopathic pulmonary fibrosis | 3.81 *I* * | |
| 99931 | Idiopathic pulmonary hemosiderosis | 0.0425 *I* * | |
| 90003 | IgG4-related hepatopathy | | 140 Cases |
| 49041 | IgG4-related retroperitoneal fibrosis | 0.35 *I* * | |
| 477661 | IL21-related infantile inflammatory bowel disease | | 3 Cases |
| 238621 | Ileal pouch anal anastomosis related faecal incontinence | 3.0 *P* * | |
| 85173 | IMAGe syndrome | | 28 Cases |
| 42062 | Iminoglycinuria | 6.68 *P* * | |
| 42062 | Iminoglycinuria | 6.67 *BP* * | |
| 238569 | Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome | | 80 Cases |
| 37042 | Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome | | 150 Cases |
| 3002 | Immune thrombocytopenic purpura | 25.0 *P* * | |
| 3002 | Immune thrombocytopenic purpura | 6.75 *I* * | |
| 206569 | Immune-mediated necrotizing myopathy | | 300 Cases |
| 34592 | Immunodeficiency by defective expression of HLA class 1 | | 30 Cases |
| 572 | Immunodeficiency by defective expression of HLA class 2 | | 179 Cases |
| 169100 | Immunodeficiency due to CD25 deficiency | | 2 Cases |
| 331190 | Immunodeficiency due to ficolin3 deficiency | | 1 Case |
| 70592 | Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency | | 49 Cases |
| 331187 | Immunodeficiency due to MASP-2 deficiency | | 1 Case |
| 70593 | Immunodeficiency due to selective anti-polysaccharide antibody deficiency | | 100 Cases |
| 200418 | Immunodeficiency with factor I anomaly | | 35 Families |
| 2759 | Imperforate oropharynx-costovertebral anomalies syndrome | | 2 Cases |
| 45453 | Incessant infant ventricular tachycardia | 1.5 *BP* * | |
| 52430 | Inclusion body myopathy with Paget disease of bone and frontotemporal dementia | | 26 Families |
| 611 | Inclusion body myositis | 0.5 *P* * | |
| 464 | Incontinentia pigmenti | 1.2 *BP* * | |
| 98848 | Indolent systemic mastocytosis | 3.8 *P* * | |
| 70587 | Infant acute respiratory distress syndrome | 20.0 *P* * | |
| 70587 | Infant acute respiratory distress syndrome | 3.5 *I* * | |
| 178478 | Infant botulism | 0.2 *BP* * | |
| 178478 | Infant botulism | 0.3 *I* * | |
| 313850 | Infantile cerebellar-retinal degeneration | | 11 Cases |
| 402364 | Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly | | 5 Cases |
| 1313 | Infantile choroidocerebral calcification syndrome | | 10 Cases |
| 199267 | Infantile digital fibromatosis | | 200 Cases |
| 238455 | Infantile dystonia-parkinsonism | | 16 Cases |
| 352563 | Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency | | 2 Cases |
| 522077 | Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome | | 11 Cases |
| 456312 | Infantile multisystem neurologic-endocrine-pancreatic disease | | 2 Cases |
| 2591 | Infantile myofibromatosis | 0.67 *BP* * | |
| 35069 | Infantile neuroaxonal dystrophy | | 150 Cases |

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|---|---|---|---|
| 251304 | Infantile onset panniculitis with uveitis and systemic granulomatosis | | 4 Cases |
| 1186 | Infantile onset spinocerebellar ataxia | | 29 Cases |
| 3173 | Infantile spasms-broad thumbs syndrome | | 2 Cases |
| 263410 | Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome | | 4 Cases |
| 293168 | Infantile-onset ascending hereditary spastic paralysis | | 17 Families |
| 457205 | Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome | | 2 Cases |
| 494526 | Infantile-onset generalized dyskinesia with orofacial involvement | | 8 Cases |
| 391316 | Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression | | 3 Cases |
| 500062 | Infantile-onset periodic fever-panniculitis-dermatosis syndrome | | 5 Cases |
| 1145 | Infantile-onset X-linked spinal muscular atrophy | | 14 Families |
| 1849 | Infundibulopelvic stenosis-multicystic kidney syndrome | | 3 Cases |
| 247257 | Inhalational anthrax | 0.1 *P* * | |
| 254504 | Inhalational botulism | | 10 Cases |
| 210141 | Inherited congenital spastic tetraplegia | | 17 Cases |
| 79361 | Inherited epidermolysis bullosa | 0.8 *P* * | |
| 79361 | Inherited epidermolysis bullosa | 1.9 *BP* * | |
| 63259 | Iniencephaly | 50.0 *P* * | |
| 411593 | Insulin autoimmune syndrome | | 404 Cases |
| 97279 | Insulinoma | 0.25 *I* | |
| 464311 | Intellectual disability syndrome due to a DYRK1A point mutation | | 35 Cases |
| 166108 | Intellectual disability, Birk-Barel type | | 1 Family |
| 3079 | Intellectual disability, Buenos-Aires type | | 5 Cases |
| 3080 | Intellectual disability, Wolff type | | 2 Cases |
| 3041 | Intellectual disability-balding-patella luxation-acromicria syndrome | | 3 Cases |
| 364577 | Intellectual disability-brachydactyly-Pierre Robin syndrome | | 4 Cases |
| 508498 | Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome | | 18 Cases |
| 3042 | Intellectual disability-cataracts-calcified pinnae-myopathy syndrome | | 13 Cases |
| 171860 | Intellectual disability-cataracts-kyphosis syndrome | | 3 Cases |
| 397709 | Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome | | 30 Cases |
| 329224 | Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome | | 2 Cases |
| 3454 | Intellectual disability-developmental delay-contractures syndrome | | 5 Families |
| 3044 | Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome | | 4 Cases |
| 468620 | Intellectual disability-epilepsy-extrapyramidal syndrome | | 3 Cases |
| 436151 | Intellectual disability-expressive aphasia-facial dysmorphism syndrome | | 13 Cases |
| 404440 | Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency | | 7 Cases |
| 370010 | Intellectual disability-facial dysmorphism-hand anomalies syndrome | | 3 Cases |
| 363611 | Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome | | 5 Cases |
| 369847 | Intellectual disability-hyperkinetic movement-truncal ataxia syndrome | | 5 Cases |
| 1495 | Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome | | 3 Cases |
| 314575 | Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome | | 2 Cases |
| 457279 | Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome | | 16 Cases |
| 457365 | Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome | | 3 Cases |
| 3068 | Intellectual disability-myopathy-short stature-endocrine defect syndrome | | 2 Cases |
| 352530 | Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome | | 2 Cases |
| 397973 | Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome | | 2 Cases |
| 3082 | Intellectual disability-polydactyly-uncombable hair syndrome | | 2 Cases |
| 513456 | Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome | | 15 Cases |
| 369837 | Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome | | 4 Cases |

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|---|---|---|---|
| 369950 | Intellectual disability-seizures-macrocephaly-obesity syndrome | | 7 Cases |
| 391372 | Intellectual disability-severe speech delay-mild dysmorphism syndrome | | 48 Cases |
| 3074 | Intellectual disability-short stature-hypertelorism syndrome | | 6 Cases |
| 3051 | Intellectual disability-sparse hair-brachydactyly syndrome | | 61 Cases |
| 1891 | Intellectual disability-spasticity-ectrodactyly syndrome | | 3 Cases |
| 363528 | Intellectual disability-strabismus syndrome | | 34 Cases |
| 981 | Internal carotid agenesis | | 100 Cases |
| 79099 | Interstitial granulomatous dermatitis with arthritis | | 53 Cases |
| 182095 | Interstitial lung disease | 5.4 *I* * | |
| 314376 | Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency | | 16 Cases |
| 137622 | Intractable diarrhea-choanal atresia-eye anomalies syndrome | | 3 Cases |
| 436144 | Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome | | 15 Cases |
| 329324 | Inverse Klippel-Trénaunay syndrome | | 15 Cases |
| 209981 | IRIDA syndrome | | 75 Cases |
| 209943 | IRVAN syndrome | | 30 Cases |
| 84142 | Isaac syndrome | | 150 Cases |
| 229717 | Isolated agammaglobulinemia | 0.3 *P* | |
| 1048 | Isolated anencephaly/exencephaly | 35.0 *BP* * | |
| 250923 | Isolated aniridia | 1.31 *I* * | |
| 557 | Isolated anorectal malformation | 20.0 *BP* | |
| 3387 | Isolated anterior cervical hypertrichosis | | 20 Cases |
| 1134 | Isolated arrhinia | | 20 Cases |
| 199326 | Isolated autosomal dominant hypomagnesemia, Glaudemans type | | 21 Cases |
| 30391 | Isolated biliary atresia | 18.5 *BP* | |
| 2343 | Isolated cloverleaf skull syndrome | | 120 Cases |
| 79143 | Isolated congenital anonychia | | 50 Cases |
| 88620 | Isolated congenital anosmia | | 15 Cases |
| 91396 | Isolated cryptophthalmia | | 30 Cases |
| 217 | Isolated Dandy-Walker malformation | 2.1 *P* * | |
| 217 | Isolated Dandy-Walker malformation | 1.0 *BP* * | |
| 1885 | Isolated ectopia lentis | | 90 Cases |
| 93928 | Isolated epispadias | 2.4 *BP* * | |
| 448264 | Isolated focal non-epidermolytic palmoplantar keratoderma | | 2 Cases |
| 468666 | Isolated generalized anhidrosis with normal sweat glands | | 7 Cases |
| 306527 | Isolated hereditary congenital facial paralysis | | 8 Families |
| 2345 | Isolated Klippel-Feil syndrome | 2.0 *P* * | |
| 2345 | Isolated Klippel-Feil syndrome | 0.6 *BP* * | |
| 2542 | Isolated microphthalmia-anophthalmia-coloboma | 5.3 *BP* * | |
| 480556 | Isolated neonatal sclerosing cholangitis | | 4 Cases |
| 718 | Isolated Pierre Robin syndrome | 5.0 *BP* * | |
| 35098 | Isolated plagiocephaly | 3.0 *BP* | |
| 2924 | Isolated polycystic liver disease | 1.0 *P* * | |
| 440713 | Isolated sedoheptulokinase deficiency | | 2 Cases |
| 823 | Isolated spina bifida | 18.6 *BP* * | |
| 457083 | Isolated splenogonadal fusion | | 145 Cases |
| 2440 | Isolated split hand-split foot malformation | 5.4 *BP* * | |
| 3208 | Isolated succinate-CoQ reductase deficiency | | 37 Cases |
| 99731 | Isolated sulfite oxidase deficiency | | 50 Cases |
| 454750 | Isolated tracheoesophageal fistula | 2.2 *BP* | |
| 3366 | Isolated trigonocephaly | 6.7 *BP* * | |
| 2306 | Isotretinoin-like syndrome | | 6 Cases |
| 33 | Isovaleric acidemia | 1.0 *P* * | |
| 439254 | ITM2B amyloidosis | | 2 Families |
| 435 | Ito hypomelanosis | 10.85 *I* * | |
| 457375 | ITPA-related encephalopathy | | 7 Cases |
| 2307 | IVIC syndrome | | 4 Families |
| 1540 | Jackson-Weiss syndrome | | 200 Cases |
| 2308 | Jacobsen syndrome | 1.0 *BP* * | |
| 1873 | Jalili syndrome | | 49 Cases |
| 79139 | Japanese encephalitis | 0.65 *I* * | |
| 313795 | Jawad syndrome | | 4 Cases |
| 90647 | Jervell and Lange-Nielsen syndrome | 0.3 *P* | |
| 474 | Jeune syndrome | 1.4 *BP* * | |
| 324999 | JMP syndrome | | 3 Cases |
| 2315 | Johanson-Blizzard syndrome | 0.4 *BP* * | |
| 475 | Joubert syndrome | 1.125 *BP* | |
| 140874 | Joubert syndrome and related disorders | 1.1 *BP* | |
| 1454 | Joubert syndrome with hepatic defect | | 8 Cases |
| 397715 | Joubert syndrome with Jeune asphyxiating thoracic dystrophy | | 8 Cases |
| 2318 | Joubert syndrome with oculorenal defect | | 17 Cases |

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|---|---|---|---|
| 2319 | Juberg-Hayward syndrome | | 13 Cases |
| 93972 | Juberg-Marsidi syndrome | | 16 Cases |
| 79405 | Junctional epidermolysis bullosa inversa | | 9 Cases |
| 306504 | Junctional epidermolysis bullosa with respiratory and renal involvement | | 3 Cases |
| 79404 | Junctional epidermolysis bullosa, generalized severe | 0.17 *BP* | |
| 79403 | Junctional epidermolysis bullosa-pyloric atresia syndrome | | 100 Cases |
| 2321 | Jung-Wolff-Back-Stahl syndrome | | 2 Cases |
| 1941 | Juvenile absence epilepsy | 7.5 *I* * | |
| 247794 | Juvenile cataract-microcornea-renal glucosuria syndrome | | 12 Cases |
| 93672 | Juvenile dermatomyositis | 0.295 *I* | |
| 248111 | Juvenile Huntington disease | 0.6 *P* * | |
| 248111 | Juvenile Huntington disease | 0.04 *I* * | |
| 2028 | Juvenile hyaline fibromatosis | | 70 Cases |
| 92 | Juvenile idiopathic arthritis | 1.5 *I* * | |
| 86834 | Juvenile myelomonocytic leukemia | 0.1 *P* * | |
| 2801 | Juvenile Paget disease | | 50 Cases |
| 79076 | Juvenile polyposis of infancy | | 11 Cases |
| 2929 | Juvenile polyposis syndrome | 3.85 *I* * | |
| 247604 | Juvenile primary lateral sclerosis | | 4 Cases |
| 26137 | Juvenile temporal arteritis | | 20 Cases |
| 445062 | Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome | | 5 Cases |
| 2322 | Kabuki syndrome | 3.1 *P* * | |
| 254519 | Kagami-Ogata syndrome | | 84 Cases |
| 254534 | Kagami-Ogata syndrome due to maternal 14q32.2 hypermethylation | | 7 Cases |
| 254528 | Kagami-Ogata syndrome due to maternal 14q32.2 microdeletion | | 8 Cases |
| 96334 | Kagami-Ogata syndrome due to paternal uniparental disomy of chromosome 14 | | 37 Cases |
| 478 | Kallmann syndrome | 3.75 *P* * | |
| 2326 | Kallmann syndrome-heart disease syndrome | | 8 Cases |
| 33276 | Kaposi sarcoma | 0.34 *I* * | |
| 2328 | Kapur-Toriello syndrome | | 6 Cases |
| 2329 | Karsch-Neugebauer syndrome | | 11 Cases |
| 401996 | Karyomegalic interstitial nephritis | | 12 Families |
| 2330 | Kasabach-Merritt syndrome | | 300 Cases |
| 2332 | KBG syndrome | | 100 Cases |
| 439218 | KCNQ2-related epileptic encephalopathy | | 11 Families |
| 480 | Kearns-Sayre syndrome | 2.0 *P* * | |
| 2662 | Keipert syndrome | | 12 Cases |
| 2333 | Kenny-Caffey syndrome | | 65 Cases |
| 435628 | Keppen-Lubinsky syndrome | | 3 Cases |
| 494 | Keratoderma hereditarium mutilans | | 50 Cases |
| 79395 | Keratoderma hereditarium mutilans with ichthyosis | | 50 Cases |
| 2339 | Keratosis follicularis-dwarfism-cerebral atrophy syndrome | | 6 Cases |
| 86919 | Keratosis palmaris et plantaris-clinodactyly syndrome | | 20 Cases |
| 293807 | Ketamine-induced biliary dilatation | | 2 Cases |
| 438075 | Ketoacidosis due to monocarboxylate transporter-1 deficiency | | 9 Cases |
| 85202 | Keutel syndrome | | 30 Cases |
| 477 | KID syndrome | | 100 Cases |
| 50918 | Kikuchi-Fujimoto disease | | 1052 Cases |
| 482 | Kimura disease | | 300 Cases |
| 2908 | Kindler syndrome | | 250 Cases |
| 99741 | King-Denborough syndrome | | 18 Cases |
| 261494 | Kleefstra syndrome | | 114 Cases |
| 96147 | Kleefstra syndrome due to 9q34 microdeletion | | 86 Cases |
| 261652 | Kleefstra syndrome due to a point mutation | | 23 Cases |
| 399081 | KLHL9-related early-onset distal myopathy | | 10 Cases |
| 447974 | Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome | | 2 Cases |
| 90308 | Klippel-Trénaunay syndrome | 0.007 *P* * | |
| 96169 | Koolen-De Vries syndrome | 4.0 *P* * | |
| 363965 | Koolen-De Vries syndrome due to a point mutation | | 4 Cases |
| 99749 | Kostmann syndrome | | 45 Cases |
| 2351 | Kousseff syndrome | | 8 Cases |
| 487 | Krabbe disease | 1.0 *P* * | |
| 487 | Krabbe disease | 0.7 *BP* | |
| 306674 | Kufor-Rakeb syndrome | | 16 Cases |
| 454745 | Kuru | | 2700 Cases |
| 1149 | Kuskokwim syndrome | | 8 Families |
| 496689 | Kyphoscoliosis-lateral tongue atrophy-hereditary spastic paraplegia syndrome | | 12 Cases |
| 496686 | Kyphosis-lateral tongue atrophy-myofibrillar myopathy syndrome | | 3 Cases |
| 79314 | L-2-hydroxyglutaric aciduria | | 140 Cases |
| 35704 | L-Arginine:glycine amidinotransferase deficiency | | 9 Cases |

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| 440731 | L-ferritin deficiency | | 2 Cases |
| 2363 | Lacrimoauriculodentodigital syndrome | | 100 Cases |
| 501 | Lafora disease | | 300 Cases |
| 521450 | LAMA5-related multisystemic syndrome | | 11 Cases |
| 306507 | LAMB2-related infantile-onset nephrotic syndrome | | 14 Cases |
| 1296 | Lambert syndrome | | 4 Cases |
| 43393 | Lambert-Eaton myasthenic syndrome | 0.35 *P* | |
| 313 | Lamellar ichthyosis | 0.55 *P* * | |
| 2632 | Langer mesomelic dysplasia | | 50 Cases |
| 389 | Langerhans cell histiocytosis | 1.5 *P* * | |
| 626 | Large congenital melanocytic nevus | 2.75 *P* * | |
| 633 | Laron syndrome | 0.3 *P* * | |
| 220465 | Laron syndrome with immunodeficiency | | 10 Cases |
| 503 | Larsen syndrome | 0.4 *BP* * | |
| 2370 | Larsen-like osseous dysplasia-short stature syndrome | | 3 Cases |
| 284139 | Larsen-like syndrome, B3GAT3 type | | 14 Cases |
| 2808 | Laryngeal abductor paralysis | | 9 Cases |
| 2375 | Laryngeal abductor paralysis-intellectual disability syndrome | | 20 Cases |
| 2004 | Laryngotracheoesophageal cleft | 7.5 *BP* * | |
| 93940 | Laryngotracheoesophageal cleft type 3 | | 30 Cases |
| 93941 | Laryngotracheoesophageal cleft type 4 | | 20 Cases |
| 98912 | Late-onset distal myopathy, Markesbery-Griggs type | | 11 Cases |
| 228227 | Late-onset focal dermal elastosis | | 5 Cases |
| 79406 | Late-onset junctional epidermolysis bullosa | | 37 Cases |
| 231556 | Late-onset localized junctional epidermolysis bullosa-intellectual disability syndrome | | 2 Cases |
| 2789 | Lateral meningocele syndrome | | 14 Cases |
| 46059 | Lathosterolosis | | 4 Cases |
| 2378 | Laurin-Sandrow syndrome | | 14 Cases |
| 650 | LCAT deficiency | | 125 Cases |
| 330015 | Lead poisoning | 2.3 *P* * | |
| 65 | Leber congenital amaurosis | 2.5 *P* | |
| 65 | Leber congenital amaurosis | 2.5 *BP* | |
| 104 | Leber hereditary optic neuropathy | 4.3 *P* | |
| 99718 | Leber plus disease | 0.04 *P* * | |
| 549 | Legionellosis | 1.3 *I* * | |
| 137605 | Legius syndrome | 2.2 *BP* | |
| 506 | Leigh syndrome | 2.0 *P* * | |
| 506 | Leigh syndrome | 2.8 *BP* * | |
| 507 | Leishmaniasis | 0.1 *P* * | |
| 507 | Leishmaniasis | 25.0 *I* | |
| 140936 | Lelis syndrome | | 9 Cases |
| 137839 | Lemierre syndrome | 10.0 *I* * | |
| 2382 | Lennox-Gastaut syndrome | 15.0 *P* * | |
| 2382 | Lennox-Gastaut syndrome | 0.1 *I* * | |
| 2658 | Lenz-Majewski hyperostotic dwarfism | | 10 Cases |
| 548 | Leprosy | 3.7 *I* | |
| 509 | Leptospirosis | 0.12 *I* * | |
| 510 | Lesch-Nyhan syndrome | 0.34 *BP* * | |
| 158687 | Lethal acantholytic epidermolysis bullosa | | 4 Cases |
| 314718 | Lethal arteriopathy syndrome due to fibulin-4 deficiency | | 22 Cases |
| 53696 | Lethal arthrogryposis-anterior horn cell disease syndrome | | 15 Cases |
| 1187 | Lethal ataxia with deafness and optic atrophy | | 4 Families |
| 137776 | Lethal congenital contracture syndrome type 2 | | 1 Family |
| 137783 | Lethal congenital contracture syndrome type 3 | | 14 Cases |
| 1972 | Lethal faciocardiomelic dysplasia | | 3 Cases |
| 444069 | Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome | | 4 Cases |
| 439897 | Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome | | 2 Cases |
| 1046 | Lethal hemolytic anemia-genital anomalies syndrome | | 2 Cases |
| 480528 | Lethal hydranencephaly-diaphragmatic hernia syndrome | | 2 Cases |
| 2570 | Lethal intrauterine growth restriction-cortical malformation-congenital contractures syndrome | | 4 Cases |
| 2347 | Lethal Kniest-like dysplasia | | 2 Cases |
| 2371 | Lethal Larsen-like syndrome | | 8 Cases |
| 478049 | Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome | | 4 Cases |
| 33108 | Lethal multiple pterygium syndrome | | 28 Families |
| 435845 | Lethal neonatal spasticity-epileptic encephalopathy syndrome | | 8 Cases |
| 293925 | Lethal occipital encephalocele-skeletal dysplasia syndrome | | 5 Cases |
| 2736 | Lethal omphalocele-cleft palate syndrome | | 3 Cases |

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|---|---|---|---|
| 1832 | Lethal osteosclerotic bone dysplasia | | 40 Cases |
| 210144 | Lethal polymalformative syndrome, Boissel type | | 10 Cases |
| 1423 | Lethal recessive chondrodysplasia | | 4 Cases |
| 99870 | Letterer-Siwe disease | 0.2 *P* * | |
| 2968 | Leukocyte adhesion deficiency | | 350 Cases |
| 99842 | Leukocyte adhesion deficiency type I | 0.1 *P* * | |
| 99843 | Leukocyte adhesion deficiency type II | | 7 Cases |
| 99844 | Leukocyte adhesion deficiency type III | | 40 Cases |
| 139444 | Leukoencephalopathy with bilateral anterior temporal lobe cysts | | 29 Cases |
| 137898 | Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome | | 127 Cases |
| 363540 | Leukoencephalopathy with mild cerebellar ataxia and white matter edema | | 6 Cases |
| 163684 | Leukoencephalopathy-dystonia-motor neuropathy syndrome | | 2 Cases |
| 83629 | Leukoencephalopathy-metaphyseal chondrodysplasia syndrome | | 4 Cases |
| 2386 | Leukoencephalopathy-palmoplantar keratoderma syndrome | | 4 Cases |
| 314051 | Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome | | 14 Cases |
| 1816 | Leukomelanoderma-infantilism-intellectual disability-hypodontia-hypotrichosis syndrome | | 4 Cases |
| 210133 | Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome | | 11 Cases |
| 48162 | Lewis-Sumner syndrome | 0.9 *P* * | |
| 65285 | Lhermitte-Duclos disease | | 220 Cases |
| 524 | Li-Fraumeni syndrome | 6.0 *P* | |
| 525 | Lichen planopilaris | | 300 Cases |
| 254478 | Lichen planus pemphigoides | | 100 Cases |
| 2390 | Lichtenstein syndrome | | 2 Cases |
| 526 | Liddle syndrome | | 80 Cases |
| 99812 | LIG4 syndrome | | 28 Cases |
| 97231 | Ligneous conjunctivitis | | 200 Cases |
| 263 | Limb-girdle muscular dystrophy | 2.32 *P* | |
| 445110 | Limb-girdle muscular dystrophy due to POMK deficiency | | 2 Cases |
| 69085 | Limb-mammary syndrome | | 38 Cases |
| 171673 | Limbal stem cell deficiency | 3.0 *P* * | |
| 329341 | Limbic encephalitis with DPP6 antibodies | | 4 Cases |
| 498700 | Limbic encephalitis with neurexin-3 antibodies | | 5 Cases |
| 220407 | Limited systemic sclerosis | | 200 Cases |
| 140933 | Linear atrophoderma of Moulin | | 30 Cases |
| 228236 | Linear focal elastosis | | 30 Cases |
| 2612 | Linear nevus sebaceus syndrome | 10.0 *BP* * | |
| 435660 | LIPE-related familial partial lipodystrophy | | 4 Cases |
| 156156 | Lipoatrophy with diabetes, leukomelanodermic papules, liver steatosis, and hypertrophic cardiomyopathy | | 1 Case |
| 1979 | Lipodystrophy due to peptidic growth factors deficiency | | 1 Family |
| 50811 | Lipodystrophy-intellectual disability-deafness syndrome | | 3 Cases |
| 401859 | Lipoic acid synthetase deficiency | | 3 Cases |
| 530 | Lipoid proteinosis | | 500 Cases |
| 69078 | Liposarcoma | 1.0 *I* * | |
| 401862 | Lipoyl transferase 1 deficiency | | 4 Cases |
| 98955 | Lisch epithelial corneal dystrophy | | 36 Cases |
| 171680 | Lissencephaly due to TUBA1A mutation | | 15 Cases |
| 86821 | Lissencephaly type 3-familial fetal akinesia sequence syndrome | | 5 Cases |
| 86822 | Lissencephaly type 3-metacarpal bone dysplasia syndrome | | 2 Cases |
| 100012 | Lissencephaly with cerebellar hypoplasia type B | | 50 Cases |
| 100013 | Lissencephaly with cerebellar hypoplasia type C | | 2 Cases |
| 533 | Listeriosis | 0.337 *I* | |
| 363618 | LMNA-related cardiocutaneous progeria syndrome | | 5 Cases |
| 2407 | LOC syndrome | | 50 Cases |
| 93685 | Localized Castleman disease | 1.0 *P* | |
| 251393 | Localized junctional epidermolysis bullosa, non-Herlitz type | | 20 Cases |
| 90398 | Localized lichen myxedematosus with mixed features of different subtypes | | 10 Cases |
| 90399 | Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms | | 5 Cases |
| 2406 | Locked-in syndrome | | 33 Cases |
| 60030 | Loeys-Dietz syndrome | | 52 Families |
| 5 | Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency | 8.0 *P* * | |
| 5 | Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency | 1.0 *BP* * | |
| 2408 | Lowe-Kohn-Cohen syndrome | | 1 Family |

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|---|---|---|---|
| 2487 | Lower limb malformation-hypospadias syndrome | | 2 Cases |
| 276435 | Lower motor neuron syndrome with late-adult onset | | 17 Families |
| 844 | Lown-Ganong-Levine syndrome | | 12 Cases |
| 2409 | Lowry-MacLean syndrome | | 3 Cases |
| 1824 | Lowry-Wood syndrome | | 8 Cases |
| 83628 | LUMBAR syndrome | | 54 Cases |
| 1120 | Lung agenesis-heart defect-thumb anomalies syndrome | | 9 Cases |
| 137631 | Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome | | 2 Cases |
| 90283 | Lupus erythematosus tumidus | | 250 Cases |
| 91546 | Lyme disease | 21.9 *I* | |
| 538 | Lymphangioleiomyomatosis | 0.15 *P* | |
| 538 | Lymphangioleiomyomatosis | 0.0135 *I* | |
| 86915 | Lymphedema-atrial septal defects-facial changes syndrome | | 5 Cases |
| 86914 | Lymphedema-cerebral arteriovenous anomaly syndrome | | 5 Cases |
| 99141 | Lymphedema-posterior choanal atresia syndrome | | 6 Cases |
| 275761 | Lysosomal acid lipase deficiency | 2.0 *P* * | |
| 397612 | Macrocephaly-developmental delay syndrome | | 9 Cases |
| 210548 | Macrocephaly-intellectual disability-autism syndrome | | 40 Cases |
| 466791 | Macrocephaly-intellectual disability-left ventricular non compaction syndrome | | 6 Cases |
| 457485 | Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome | | 8 Cases |
| 2427 | Macrocephaly-short stature-paraplegia syndrome | | 2 Cases |
| 2432 | Macrosomia-microphthalmia-cleft palate syndrome | | 5 Cases |
| 83619 | Macrostomia-preauricular tags-external ophthalmoplegia syndrome | | 9 Cases |
| 220448 | Macrothrombocytopenia with mitral valve insufficiency | | 2 Cases |
| 487796 | Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome | | 2 Cases |
| 91494 | Macular coloboma-cleft palate-hallux valgus syndrome | | 2 Cases |
| 137867 | Madras motor neuron disease | | 200 Cases |
| 163634 | Maffucci syndrome | | 250 Cases |
| 324972 | MAGIC syndrome | | 21 Cases |
| 77297 | Majeed syndrome | | 4 Families |
| 87503 | Mal de Meleda | 1.0 *P* | |
| 420179 | Malan overgrowth syndrome | | 20 Cases |
| 673 | Malaria | 3.0 *P* * | |
| 673 | Malaria | 73.0 *I* | |
| 2234 | Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome | | 2 Cases |
| 99915 | Maligant granulosa cell tumor of the ovary | 0.12 *I* * | |
| 679 | Malignant atrophic papulosis | | 200 Cases |
| 99912 | Malignant dysgerminomatous germ cell tumor of the ovary | 0.04 *I* * | |
| 398934 | Malignant epithelial tumor of ovary | 9.39 *I* * | |
| 276145 | Malignant epithelial tumor of salivary glands | 0.73 *I* * | |
| 35807 | Malignant germ cell tumor of ovary | 0.08 *I* * | |
| 168999 | Malignant melanoma of the mucosa | 0.26 *I* * | |
| 293181 | Malignant migrating partial seizures of infancy | | 114 Cases |
| 213512 | Malignant mixed Müllerian tumor of the ovary | 0.12 *I* * | |
| 398940 | Malignant non-epithelial tumor of ovary | 0.43 *I* * | |
| 3148 | Malignant peripheral nerve sheath tumor | 1.0 *I* | |
| 168811 | Malignant peritoneal mesothelioma | 1.5 *P* * | |
| 35808 | Malignant sex cord stromal tumor of ovary | 1.85 *P* * | |
| 35808 | Malignant sex cord stromal tumor of ovary | 0.13 *I* * | |
| 398987 | Malignant teratoma of ovary | 0.07 *I* * | |
| 252212 | Malignant triton tumor | | 170 Cases |
| 180242 | Malignant tumor of fallopian tubes | 1.0 *P* * | |
| 398043 | Malignant tumor of penis | 1.075 *I* * | |
| 943 | Malonic aciduria | | 34 Cases |
| 52417 | MALT lymphoma | 4.0 *P* * | |
| 52417 | MALT lymphoma | 0.3 *I* * | |
| 238744 | Mammary-digital-nail syndrome | | 11 Cases |
| 397941 | MAN1B1-CDG | | 25 Cases |
| 363649 | Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome | | 21 Cases |
| 2457 | Mandibuloacral dysplasia | | 40 Cases |
| 443995 | Mandibulofacial dysostosis with alopecia | | 4 Cases |
| 357158 | Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome | | 2 Cases |
| 79113 | Mandibulofacial dysostosis-microcephaly syndrome | | 107 Cases |

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|---|---|---|---|
| 52416 | Mantle cell lymphoma | 3.5 *P* * | |
| 511 | Maple syrup urine disease | 0.67 *BP* | |
| 99826 | Marburg hemorrhagic fever | | 500 Cases |
| 221074 | Marchiafava-Bignami disease | | 250 Cases |
| 2461 | Marden-Walker syndrome | | 50 Cases |
| 558 | Marfan syndrome | 15.0 *P* | |
| 558 | Marfan syndrome | 25.0 *I* * | |
| 2463 | Marfanoid habitus-autosomal recessive intellectual disability syndrome | | 4 Cases |
| 314041 | Marfanoid habitus-inguinal hernia-advanced bone age syndrome | | 2 Cases |
| 2464 | Marfanoid syndrome, De Silva type | | 6 Cases |
| 300912 | Marginal zone lymphoma | 7.0 *P* * | |
| 300912 | Marginal zone lymphoma | 0.3 *I* * | |
| 559 | Marinesco-Sjögren syndrome | | 200 Cases |
| 560 | Marshall syndrome | | 17 Cases |
| 561 | Marshall-Smith syndrome | | 33 Cases |
| 466718 | Martinique crinkled retinal pigment epitheliopathy | | 14 Cases |
| 98292 | Mastocytosis | 9.0 *P* * | |
| 2209 | Maternal phenylketonuria | 10.0 *I* * | |
| 411712 | Maternal riboflavin deficiency | | 2 Cases |
| 96181 | Maternal uniparental disomy of chromosome 6 | | 15 Cases |
| 97678 | Maternal uniparental disomy of chromosome 13 | | 3 Cases |
| 96186 | Maternal uniparental disomy of chromosome 20 | | 12 Cases |
| 96187 | Maternal uniparental disomy of chromosome 21 | | 2 Cases |
| 96188 | Maternal uniparental disomy of chromosome 22 | | 4 Cases |
| 225 | Maternally-inherited diabetes and deafness | 0.1 *P* * | |
| 2470 | Matthew-Wood syndrome | | 43 Cases |
| 3109 | Mayer-Rokitansky-Küster-Hauser syndrome | 11.0 *BP* | |
| 2578 | Mayer-Rokitansky-Küster-Hauser syndrome type 2 | 1.0 *BP* * | |
| 57782 | Mazabraud syndrome | | 54 Cases |
| 562 | McCune-Albright syndrome | 0.55 *P* * | |
| 2471 | McDonough syndrome | | 2 Families |
| 2473 | McKusick-Kaufman syndrome | | 90 Cases |
| 59306 | McLeod neuroacanthocytosis syndrome | | 100 Cases |
| 3097 | Meacham syndrome | | 13 Cases |
| 564 | Meckel syndrome | 4.0 *BP* | |
| 70588 | Meconium aspiration syndrome | 2.44 *P* * | |
| 57196 | Medial condensing osteitis of the clavicle | | 58 Cases |
| 2006 | Median cleft lip/mandibule | | 70 Cases |
| 2699 | Median nodule of the upper lip | | 4 Families |
| 370127 | Medich giant platelet syndrome | | 3 Cases |
| 42 | Medium chain acyl-CoA dehydrogenase deficiency | 6.85 *P* | |
| 42 | Medium chain acyl-CoA dehydrogenase deficiency | 12.0 *BP* * | |
| 171851 | MEDNIK syndrome | | 5 Families |
| 1332 | Medullary thyroid carcinoma | 7.0 *P* * | |
| 1332 | Medullary thyroid carcinoma | 0.22 *I* * | |
| 616 | Medulloblastoma | 1.0 *P* * | |
| 616 | Medulloblastoma | 0.11 *I* * | |
| 98954 | Meesmann corneal dystrophy | | 250 Cases |
| 280671 | Megaconial congenital muscular dystrophy | | 19 Cases |
| 2241 | Megacystis-microcolon-intestinal hypoperistalsis syndrome | | 230 Cases |
| 2478 | Megalencephalic leukoencephalopathy with subcortical cysts | | 100 Cases |
| 60040 | Megalencephaly-capillary malformation-polymicrogyria syndrome | | 170 Cases |
| 83473 | Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome | | 62 Cases |
| 457359 | Megalencephaly-severe kyphoscoliosis-overgrowth syndrome | | 2 Cases |
| 352328 | MEGDEL syndrome | | 67 Cases |
| 85282 | MEHMO syndrome | | 8 Cases |
| 550 | MELAS | 0.6 *P* * | |
| 2482 | Melhem-Fahl syndrome | | 2 Cases |
| 2484 | Melnick-Needles syndrome | | 70 Cases |
| 2485 | Melorheostosis | 0.09 *P* * | |
| 1879 | Melorheostosis with osteopoikilosis | | 5 Families |
| 401973 | MEND syndrome | | 19 Cases |
| 99898 | Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency | | 31 Cases |
| 319547 | Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency | | 13 Cases |
| 319558 | Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency | | 49 Cases |
| 319552 | Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency | | 180 Cases |

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|---|---|---|---|
| 319563 | Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency | | 6 Cases |
| 319600 | Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency | | 2 Cases |
| 319595 | Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency | | 17 Cases |
| 2495 | Meningioma | 0.15 *I* * | |
| 565 | Menkes disease | 0.33 *BP* * | |
| 498251 | Menstrual cycle-dependent periodic fever | | 5 Cases |
| 508093 | MEPAN syndrome | | 7 Cases |
| 157801 | Mesoaxial synostotic syndactyly with phalangeal reduction | | 6 Families |
| 2496 | Mesomelia-synostoses syndrome | | 5 Cases |
| 2631 | Mesomelic dwarfism-cleft palate-camptodactyly syndrome | | 2 Cases |
| 1836 | Mesomelic dysplasia, Kantaputra type | | 5 Families |
| 2499 | Metachondromatosis | | 25 Cases |
| 512 | Metachromatic leukodystrophy | 0.1 *P* * | |
| 512 | Metachromatic leukodystrophy | 1.47 *BP* * | |
| 1240 | Metaphyseal acroscyphodysplasia | | 4 Cases |
| 1040 | Metaphyseal anadysplasia | | 27 Cases |
| 33067 | Metaphyseal chondrodysplasia, Jansen type | | 16 Cases |
| 166038 | Metaphyseal chondrodysplasia, Kaitila type | | 2 Cases |
| 2501 | Metaphyseal chondrodysplasia, Spahr type | | 18 Cases |
| 2502 | Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome | | 3 Cases |
| 2504 | Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome | | 2 Families |
| 213531 | Metaplastic carcinoma of the breast | 0.06 *I* * | |
| 2635 | Metatropic dysplasia | 0.2 *BP* * | |
| 1923 | Methimazole embryofetopathy | | 40 Cases |
| 413690 | Methotrexate toxicity or dose selection | 3.0 *P* * | |
| 2169 | Methylcobalamin deficiency type cblE | | 27 Cases |
| 2170 | Methylcobalamin deficiency type cblG | | 33 Cases |
| 308425 | Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency | | 7 Cases |
| 26 | Methylmalonic acidemia with homocystinuria | | 500 Cases |
| 79284 | Methylmalonic acidemia with homocystinuria type cblF | | 15 Cases |
| 79282 | Methylmalonic acidemia with homocystinuria, type cblC | | 500 Cases |
| 79283 | Methylmalonic acidemia with homocystinuria, type cblD | | 17 Cases |
| 369955 | Methylmalonic acidemia with homocystinuria, type cblJ | | 2 Cases |
| 369962 | Methylmalonic acidemia with homocystinuria, type cblX | | 18 Cases |
| 280183 | Methylmalonic aciduria due to transcobalamin receptor defect | | 5 Cases |
| 502430 | Metopic ridging-ptosis-facial dysmorphism syndrome | | 8 Cases |
| 309025 | Mevalonate kinase deficiency | | 300 Cases |
| 29 | Mevalonic aciduria | | 30 Cases |
| 485421 | MFF-related encephalopathy due to mitochondrial and peroxisomal fission defect | | 4 Cases |
| 79329 | MGAT2-CDG | | 13 Cases |
| 2510 | Micro syndrome | | 203 Cases |
| 2511 | Microbrachycephaly-ptosis-cleft lip syndrome | | 2 Cases |
| 85172 | Microcephalic osteodysplastic dysplasia, Saul-Wilson type | | 4 Cases |
| 2637 | Microcephalic osteodysplastic primordial dwarfism type II | | 150 Cases |
| 2636 | Microcephalic osteodysplastic primordial dwarfism types I and III | | 30 Cases |
| 468631 | Microcephalic primordial dwarfism due to RTTN deficiency | | 12 Cases |
| 329228 | Microcephalic primordial dwarfism due to ZNF335 deficiency | | 10 Cases |
| 319671 | Microcephalic primordial dwarfism, Alazami type | | 10 Cases |
| 319675 | Microcephalic primordial dwarfism, Dauber type | | 2 Cases |
| 2617 | Microcephalic primordial dwarfism, Montreal type | | 3 Cases |
| 2643 | Microcephalic primordial dwarfism, Toriello type | | 2 Cases |
| 436182 | Microcephalic primordial dwarfism-insulin resistance syndrome | | 2 Cases |
| 2513 | Microcephaly-albinism-digital anomalies syndrome | | 2 Cases |
| 3433 | Microcephaly-brachydactyly-kyphoscoliosis syndrome | | 3 Cases |
| 2523 | Microcephaly-brain defect-spasticity-hypernatremia syndrome | | 3 Cases |
| 294016 | Microcephaly-capillary malformation syndrome | | 10 Cases |
| 2516 | Microcephaly-cardiac defect-lung malsegmentation syndrome | | 3 Cases |
| 2515 | Microcephaly-cardiomyopathy syndrome | | 3 Cases |
| 329332 | Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome | | 4 Cases |
| 2522 | Microcephaly-cervical spine fusion anomalies syndrome | | 2 Cases |

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| 2521 | Microcephaly-cleft palate-abnormal retinal pigmentation syndrome | | 3 Cases |
| 423894 | Microcephaly-complex motor and sensory axonal neuropathy syndrome | | 3 Cases |
| 488168 | Microcephaly-congenital cataract-psoriasiform dermatitis syndrome | | 5 Cases |
| 500159 | Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrom | | 4 Cases |
| 457284 | Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome | | 5 Cases |
| 2533 | Microcephaly-deafness-intellectual disability syndrome | | 2 Cases |
| 521445 | Microcephaly-facial dysmorphism-ocular anomalies-multiple congenital anomalies syndrome | | 10 Cases |
| 217026 | Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type | | 5 Cases |
| 2172 | Microcephaly-glomerulonephritis-marfanoid habitus syndrome | | 2 Cases |
| 457351 | Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome | | 14 Cases |
| 2526 | Microcephaly-lymphedema-chorioretinopathy syndrome | | 50 Families |
| 2528 | Microcephaly-microcornea syndrome, Seemanova type | | 2 Cases |
| 171703 | Microcephaly-polymicrogyria-corpus callosum agenesis syndrome | | 4 Cases |
| 2519 | Microcephaly-seizures-intellectual disability-heart disease syndrome | | 2 Cases |
| 423306 | Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome | | 2 Cases |
| 397951 | Microcephaly-thin corpus callosum-intellectual disability syndrome | | 4 Cases |
| 2535 | Microcornea-corectopia-macular hypoplasia syndrome | | 3 Cases |
| 2536 | Microcornea-glaucoma-absent frontal sinuses syndrome | | 4 Cases |
| 369970 | Microcornea-myopic chorioretinal atrophy-telecanthus syndrome | | 14 Cases |
| 231736 | Microcornea-posterior megalolenticonus-persistent fetal vasculature-coloboma syndrome | | 8 Cases |
| 83642 | Microcytic anemia with liver iron overload | | 3 Cases |
| 217377 | Microduplication Xp11.22p11.23 syndrome | | 12 Cases |
| 2538 | Microgastria-limb reduction defect syndrome | | 16 Cases |
| 476126 | Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome | | 4 Cases |
| 50810 | Microlissencephaly-micromelia syndrome | | 2 Cases |
| 139471 | Microphthalmia with brain and digit anomalies | | 2 Families |
| 1106 | Microphthalmia with limb anomalies | | 35 Families |
| 2556 | Microphthalmia with linear skin defects syndrome | | 55 Cases |
| 98555 | Microphthalmia-anophthalmia-coloboma | 8.3 *BP* * | |
| 77299 | Microphthalmia-brain atrophy syndrome | | 3 Cases |
| 2547 | Microphthalmia-microtia-fetal akinesia syndrome | | 2 Cases |
| 251279 | Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome | | 9 Cases |
| 727 | Microscopic polyangiitis | 1.0 *I* * | |
| 83463 | Microtia | 15.5 *BP* | |
| 139450 | Microtia-eye coloboma-imperforation of the nasolacrimal duct syndrome | | 1 Family |
| 289522 | Microtriplication 11q24.1 | | 2 Cases |
| 2290 | Microvillus inclusion disease | | 137 Cases |
| 2557 | Mietens syndrome | | 9 Cases |
| 2558 | Mikati-Najjar-Sahli syndrome | | 5 Cases |
| 169799 | Mild hemophilia B | 0.6 *P* * | |
| 93279 | Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis | | 4 Families |
| 531 | Miller-Dieker syndrome | 1.0 *BP* * | |
| 98919 | Miller-Fisher syndrome | 0.1 *I* * | |
| 352734 | Minimal pigment oculocutaneous albinism type 1 | | 10 Cases |
| 494433 | MIRAGE syndrome | | 19 Cases |
| 3004 | Mirror polydactyly-vertebral segmentation-limbs defects syndrome | 0.3 *P* * | |
| 293822 | MITF-related melanoma and renal cell carcinoma predisposition syndrome | | 30 Families |
| 1933 | Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria | | 2 Cases |
| 255235 | Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy | | 5 Cases |
| 369897 | Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies | | 20 Cases |
| 363534 | Mitochondrial DNA depletion syndrome, hepatocerebrorenal form | | 3 Cases |

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| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 254875 | Mitochondrial DNA depletion syndrome, myopathic form | | 45 Cases |
| 1349 | Mitochondrial DNA-related cardiomyopathy and hearing loss | | 2 Families |
| 314637 | Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency | | 8 Cases |
| 289560 | Mitochondrial membrane protein-associated neurodegeneration | 0.1 *P* | |
| 2598 | Mitochondrial myopathy and sideroblastic anemia | | 7 Cases |
| 502423 | Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome | | 9 Cases |
| 2597 | Mitochondrial myopathy-lactic acidosis-deafness syndrome | | 2 Cases |
| 298 | Mitochondrial neurogastrointestinal encephalomyopathy | 0.1 *P* * | |
| 2443 | Mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies | 9.0 *P* * | |
| 447784 | Mitochondrial pyruvate carrier deficiency | | 4 Cases |
| 746 | Mitochondrial trifunctional protein deficiency | 1.0 *P* * | |
| 180234 | Mixed germ cell tumor | 0.01 *I* * | |
| 324364 | Mixed sclerosing bone dystrophy with extra-skeletal manifestations | | 2 Cases |
| 497757 | MME-related autosomal dominant Charcot Marie Tooth disease type 2 | | 19 Cases |
| 90056 | Moderate and severe traumatic brain injury | 37.8 *P* * | |
| 169796 | Moderately severe hemophilia B | 0.6 *P* * | |
| 570 | Moebius syndrome | | 300 Cases |
| 2560 | Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome | | 7 Cases |
| 79330 | MOGS-CDG | | 3 Cases |
| 52368 | Mohr-Tranebjaerg syndrome | | 91 Cases |
| 2563 | MOMO syndrome | | 8 Cases |
| 228423 | Monocytopenia with susceptibility to infections | | 22 Cases |
| 2565 | Mononen-Karnes-Senac syndrome | | 1 Family |
| 77301 | Monosomy 9q22.3 | | 42 Cases |
| 1598 | Monosomy 18p | 2.0 *BP* * | |
| 1600 | Monosomy 18q | 2.5 *BP* | |
| 574 | Monosomy 21 | | 50 Cases |
| 48652 | Monosomy 22q13 | | 200 Cases |
| 83467 | Morvan syndrome | | 60 Cases |
| 329813 | Mosaic genome-wide paternal uniparental disomy | | 13 Cases |
| 1692 | Mosaic trisomy 1 | | 18 Cases |
| 1723 | Mosaic trisomy 2 | | 22 Cases |
| 100071 | Mosaic trisomy 3 | | 6 Cases |
| 1747 | Mosaic trisomy 7 | | 31 Cases |
| 96061 | Mosaic trisomy 8 | 3.0 *I* * | |
| 99776 | Mosaic trisomy 9 | | 50 Cases |
| 1708 | Mosaic trisomy 16 | | 226 Cases |
| 1711 | Mosaic trisomy 17 | | 31 Cases |
| 1052 | Mosaic variegated aneuploidy syndrome | | 41 Cases |
| 3347 | Mounier-Kühn syndrome | | 300 Cases |
| 2152 | Mowat-Wilson syndrome | 1.7 *BP* * | |
| 280679 | Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome | | 9 Cases |
| 2573 | Moyamoya disease | 0.035 *I* * | |
| 401945 | Moyamoya disease with early-onset achalasia | | 9 Cases |
| 2574 | Moynahan syndrome | | 26 Cases |
| 79323 | MPDU1-CDG | | 8 Cases |
| 79319 | MPI-CDG | | 25 Cases |
| 263347 | MRCS syndrome | | 7 Cases |
| 480536 | MSH3-related attenuated familial adenomatous polyposis | | 4 Cases |
| 320360 | MT-ATP6-related mitochondrial spastic paraplegia | | 5 Cases |
| 100024 | Mu-heavy chain disease | | 35 Cases |
| 398961 | Mucinous adenocarcinoma of ovary | 0.85 *I* * | |
| 424053 | Mucinous cystadenocarcinoma of the pancreas | 0.01 *I* * | |
| 575 | Muckle-Wells syndrome | | 200 Cases |
| 576 | Mucolipidosis type II | 0.84 *BP* * | |
| 577 | Mucolipidosis type III | 1.0 *BP* * | |
| 423461 | Mucolipidosis type III alpha/beta | 13.0 *P* | |
| 579 | Mucopolysaccharidosis type 1 | 0.25 *P* * | |
| 579 | Mucopolysaccharidosis type 1 | 0.82 *BP* | |
| 580 | Mucopolysaccharidosis type 2 | 10.0 *P* * | |
| 580 | Mucopolysaccharidosis type 2 | 0.68 *BP* | |
| 217085 | Mucopolysaccharidosis type 2, severe form | 0.4 *BP* * | |
| 581 | Mucopolysaccharidosis type 3 | 0.3 *P* * | |
| 581 | Mucopolysaccharidosis type 3 | 0.87 *BP* * | |
| 309297 | Mucopolysaccharidosis type 4A | 15.0 *P* * | |
| 583 | Mucopolysaccharidosis type 6 | 0.16 *P* * | |
| 583 | Mucopolysaccharidosis type 6 | 0.16 *BP* * | |
| 584 | Mucopolysaccharidosis type 7 | 0.01 *P* * | |

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|---|---|---|---|
| 505248 | Mucopolysaccharidosis-like syndrome with congenital heart defects and hematopoietic disorders | | 19 Cases |
| 53271 | Muenke syndrome | 3.33 *BP* | |
| 587 | Muir-Torre syndrome | | 205 Cases |
| 2576 | MULIBREY nanism | | 150 Cases |
| 1655 | Müllerian derivatives-lymphangiectasia-polydactyly syndrome | | 8 Cases |
| 2491 | Müllerian duct anomalies-limb anomalies syndrome | | 5 Cases |
| 93686 | Multicentric Castleman disease | | 100 Cases |
| 371428 | Multicentric osteolysis-nodulosis-arthropathy spectrum | | 50 Cases |
| 139436 | Multicentric reticulohistiocytosis | | 200 Cases |
| 1851 | Multicystic dysplastic kidney | 23.26 *BP* | |
| 3282 | Multifocal atrial tachycardia | 0.67 *BP* | |
| 641 | Multifocal motor neuropathy | 1.5 *P* | |
| 2091 | Multinodular goiter-cystic kidney-polydactyly syndrome | | 3 Cases |
| 500135 | Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome | | 3 Cases |
| 280633 | Multiple congenital anomalies-hypotonia-seizures syndrome | | 15 Cases |
| 300496 | Multiple congenital anomalies-hypotonia-seizures syndrome type 2 | | 15 Cases |
| 652 | Multiple endocrine neoplasia type 1 | 3.3 *P* * | |
| 653 | Multiple endocrine neoplasia type 2 | 2.9 *P* * | |
| 251 | Multiple epiphyseal dysplasia | 5.0 *P* * | |
| 93311 | Multiple epiphyseal dysplasia type 5 | | 18 Families |
| 166024 | Multiple epiphyseal dysplasia, Al-Gazali type | | 4 Cases |
| 166011 | Multiple epiphyseal dysplasia, Beighton type | | 1 Family |
| 166016 | Multiple epiphyseal dysplasia, Lowry type | | 2 Cases |
| 166032 | Multiple epiphyseal dysplasia, with miniepiphyses | | 2 Cases |
| 166029 | Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia | | 3 Cases |
| 401869 | Multiple mitochondrial dysfunctions syndrome type 1 | | 21 Cases |
| 401874 | Multiple mitochondrial dysfunctions syndrome type 2 | | 6 Cases |
| 363424 | Multiple mitochondrial dysfunctions syndrome type 3 | | 2 Cases |
| 457406 | Multiple mitochondrial dysfunctions syndrome type 4 | | 8 Cases |
| 29073 | Multiple myeloma | 11.9 *P* * | |
| 29073 | Multiple myeloma | 6.0 *I* | |
| 321 | Multiple osteochondromas | 1.0 *P* * | |
| 324299 | Multiple paragangliomas associated with polycythemia | | 2 Cases |
| 2215 | Multiple pterygium-malignant hyperthermia syndrome | | 4 Cases |
| 3151 | Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome | | 2 Cases |
| 65748 | Multiple self-healing squamous epithelioma | | 100 Cases |
| 585 | Multiple sulfatase deficiency | | 50 Cases |
| 3237 | Multiple synostoses syndrome | | 30 Families |
| 102 | Multiple system atrophy | 3.5 *P* | |
| 102 | Multiple system atrophy | 1.8 *I* | |
| 98933 | Multiple system atrophy, parkinsonian type | 2.4 *P* * | |
| 404463 | Multisystemic smooth muscle dysfunction syndrome | | 7 Cases |
| 370997 | Muscle-eye-brain disease with bilateral multicystic leucodystrophy | | 2 Cases |
| 2579 | Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome | | 10 Cases |
| 199340 | Muscular dystrophy, Selcen type | | 12 Cases |
| 1877 | Muscular dystrophy-white matter spongiosis syndrome | | 2 Cases |
| 324416 | Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome | | 2 Cases |
| 139578 | Mutilating hereditary sensory neuropathy with spastic paraplegia | | 14 Cases |
| 659 | Mutilating palmoplantar keratoderma with periorificial keratotic plaques | | 73 Cases |
| 589 | Myasthenia gravis | 7.77 *P* | |
| 589 | Myasthenia gravis | 0.53 *I* | |
| 498693 | MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome | | 4 Cases |
| 268249 | Mycophenolate mofetil embryopathy | | 25 Cases |
| 178566 | Mycosis fungoides and variants | 0.59 *I* * | |
| 52688 | Myelodysplastic syndrome | 1.5 *I* * | |
| 98275 | Myelodysplastic/myeloproliferative disease | 0.29 *I* * | |
| 86850 | Myeloid sarcoma | 0.02 *I* * | |
| 98274 | Myeloproliferative neoplasm | 3.07 *I* * | |
| 437572 | MYH7-related late-onset scapuloperoneal muscular dystrophy | | 12 Cases |
| 182050 | MYH9-related disease | 0.3 *P* * | |
| 2588 | Myhre syndrome | | 55 Cases |

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|---|---|---|---|
| 480491 | MYO5B-related progressive familial intrahepatic cholestasis | | 5 Cases |
| 86909 | Myoclonic epilepsy of infancy | | 106 Cases |
| 2589 | Myoclonus-cerebellar ataxia-deafness syndrome | | 4 Cases |
| 2601 | Myopathy-growth delay-intellectual disability-hypospadias syndrome | | 1 Case |
| 206647 | Myotonic dystrophy | 6.7 *P* | |
| 99967 | Myxoid/round cell liposarcoma | 0.1 *I* * | |
| 2608 | N syndrome | | 3 Cases |
| 69087 | Naegeli-Franceschetti-Jadassohn syndrome | 0.035 *P* * | |
| 245 | Nager syndrome | | 100 Cases |
| 423454 | Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome | | 6 Cases |
| 2614 | Nail-patella syndrome | 0.2 *BP* * | |
| 2613 | Nail-patella-like renal disease | | 3 Cases |
| 2615 | Nakajo-Nishimura syndrome | | 30 Cases |
| 627 | Nance-Horan syndrome | | 196 Cases |
| 2073 | Narcolepsy type 1 | 25.0 *P* * | |
| 2399 | Nasopalpebral lipoma-coloboma syndrome | | 30 Cases |
| 150 | Nasopharyngeal carcinoma | 2.0 *P* * | |
| 150 | Nasopharyngeal carcinoma | 0.36 *I* * | |
| 2663 | Nathalie syndrome | | 1 Family |
| 255229 | Navajo neurohepatopathy | | 49 Cases |
| 443162 | NDE1-related microhydranencephaly | | 1 Family |
| 391673 | Necrotizing enterocolitis | 45.0 *P* | |
| 464366 | NEK9-related lethal skeletal dysplasia | | 5 Cases |
| 607 | Nemaline myopathy | 2.0 *BP* * | |
| 217563 | Neonatal acute respiratory distress due to SP-B deficiency | 0.067 *BP* | |
| 398097 | Neonatal antiphospholipid syndrome | | 34 Cases |
| 398109 | Neonatal autoimmune hemolytic anemia | | 2 Cases |
| 398117 | Neonatal dermatomyositis | | 3 Cases |
| 224 | Neonatal diabetes mellitus | 1.1 *BP* * | |
| 79118 | Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome | | 3 Cases |
| 457185 | Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome | | 11 Cases |
| 446 | Neonatal hemochromatosis | | 35 Cases |
| 59303 | Neonatal ichthyosis-sclerosing cholangitis syndrome | | 12 Cases |
| 294023 | Neonatal inflammatory skin and bowel disease | | 3 Cases |
| 398127 | Neonatal scleroderma | | 6 Cases |
| 466784 | Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect | | 3 Cases |
| 94058 | Neovascular glaucoma | 24.4 *P* * | |
| 654 | Nephroblastoma | 10.0 *BP* * | |
| 654 | Nephroblastoma | 0.14 *I* * | |
| 223 | Nephrogenic diabetes insipidus | 0.15 *P* * | |
| 3145 | Nephrogenic diabetes insipidus-intracranial calcification syndrome | | 2 Cases |
| 93606 | Nephrogenic syndrome of inappropriate antidiuresis | | 21 Cases |
| 2668 | Nephropathy-deafness-hyperparathyroidism syndrome | | 5 Cases |
| 2669 | Nephrosis-deafness-urinary tract-digital malformations syndrome | | 5 Cases |
| 300333 | Nephrotic syndrome-deafness-pretibial epidermolysis bullosa syndrome | | 3 Cases |
| 280576 | Nestor-Guillermo progeria syndrome | | 2 Cases |
| 634 | Netherton syndrome | 0.5 *P* * | |
| 634 | Netherton syndrome | 0.5 *BP* * | |
| 2671 | Neu-Laxova syndrome | | 91 Cases |
| 2672 | Neuhauser-Eichner-Opitz syndrome | | 5 Cases |
| 3388 | Neural tube defect | 91.05 *BP* * | |
| 635 | Neuroblastoma | 11.0 *P* * | |
| 635 | Neuroblastoma | 5.8 *BP* * | |
| 635 | Neuroblastoma | 1.26 *I* | |
| 2481 | Neurocutaneous melanocytosis | 1.25 *P* * | |
| 88639 | Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency | | 20 Cases |
| 385 | Neurodegeneration with brain iron accumulation | 0.2 *P* * | |
| 217382 | Neurodegenerative syndrome due to cerebral folate transport deficiency | | 3 Cases |
| 453499 | Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome | | 15 Cases |
| 352665 | Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to 9q21 microdeletion | | 13 Cases |
| 453504 | Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to a point mutation | | 2 Cases |

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|---|---|---|---|
| 33445 | Neuroectodermal melanolysosomal disease | | 20 Cases |
| 2676 | Neuroectodermal-endocrine syndrome | | 4 Cases |
| 877 | Neuroendocrine neoplasm | 2.53 *I* * | |
| 97253 | Neuroendocrine tumor of pancreas | 0.21 *I* * | |
| 100075 | Neuroendocrine tumor of stomach | 3.2 *P* * | |
| 2673 | Neurofaciodigitorenal syndrome | | 3 Cases |
| 157846 | Neuroferritinopathy | | 90 Cases |
| 636 | Neurofibromatosis type 1 | 21.3 *P* * | |
| 636 | Neurofibromatosis type 1 | 33.3 *BP* | |
| 637 | Neurofibromatosis type 2 | 1.7 *P* * | |
| 1143 | Neurogenic arthrogryposis multiplex congenita | 4.3 *BP* * | |
| 137754 | Neurological conditions associated with aminoacylase 1 deficiency | | 15 Cases |
| 35705 | Neurometabolic disorder due to serine deficiency | | 30 Cases |
| 71211 | Neuromyelitis optica | 1.5 *P* * | |
| 139512 | Neuropathy with hearing impairment | | 1 Family |
| 137596 | Neurotrophic keratopathy | 4.2 *P* * | |
| 165 | Neutral lipid storage disease | | 50 Cases |
| 98908 | Neutral lipid storage myopathy | | 36 Cases |
| 2690 | Neutropenia-monocytopenia-deafness syndrome | | 3 Cases |
| 183707 | Neutrophil immunodeficiency syndrome | | 2 Cases |
| 263432 | Nevus of Ito | 1.17 *P* * | |
| 77292 | Niemann-Pick disease type A | 0.25 *BP* * | |
| 77293 | Niemann-Pick disease type B | 0.4 *P* * | |
| 646 | Niemann-Pick disease type C | 1.0 *P* * | |
| 1390 | Night blindness-skeletal anomalies-dysmorphism syndrome | | 2 Cases |
| 647 | Nijmegen breakage syndrome | 1.0 *BP* | |
| 240760 | Nijmegen breakage syndrome-like disorder | | 1 Case |
| 447731 | NIK deficiency | | 2 Cases |
| 99825 | Nipah virus disease | | 556 Cases |
| 263665 | NK-cell enteropathy | | 8 Cases |
| 527497 | NKX6-2-related autosomal recessive hypomyelinating leukodystrophy | | 25 Cases |
| 247868 | NLRP12-associated hereditary periodic fever syndrome | | 19 Cases |
| 86867 | Nodal marginal zone B-cell lymphoma | 1.0 *P* * | |
| 86893 | Nodular lymphocyte predominant Hodgkin lymphoma | 0.12 *I* | |
| 467 | Non-acquired combined pituitary hormone deficiency | 29.0 *BP* * | |
| 231720 | Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome | | 13 Cases |
| 631 | Non-acquired isolated growth hormone deficiency | 0.39 *P* | |
| 2337 | Non-epidermolytic palmoplantar keratoderma | 2.5 *P* * | |
| 2972 | Non-eruption of teeth-maxillary hypoplasia-genu valgum syndrome | | 4 Cases |
| 91349 | Non-functioning pituitary adenoma | 1.05 *I* | |
| 357034 | Non-hereditary retinoblastoma | 0.038 *I* * | |
| 547 | Non-Hodgkin lymphoma | 11.6 *I* * | |
| 329883 | Non-hypoproteinemic hypertrophic gastropathy | | 1 Family |
| 363999 | Non-immune hydrops fetalis | 42.0 *BP* | |
| 329918 | Non-immunoglobulin-mediated membranoproliferative glomerulonephritis | 14.0 *P* * | |
| 329918 | Non-immunoglobulin-mediated membranoproliferative glomerulonephritis | 0.15 *I* * | |
| 90061 | Non-infectious posterior uveitis | 18.0 *P* * | |
| 209989 | Non-papillary transitional cell carcinoma of the bladder | 37.0 *P* * | |
| 314647 | Non-progressive cerebellar ataxia with intellectual disability | | 15 Cases |
| 363494 | Non-seminomatous germ cell tumor of testis | 1.21 *I* * | |
| 90031 | Non-spherocytic hemolytic anemia due to hexokinase deficiency | | 17 Families |
| 500 | Noonan syndrome with multiple lentigines | | 296 Cases |
| 2701 | Noonan syndrome-like disorder with loose anagen hair | | 27 Cases |
| 649 | Norrie disease | | 400 Cases |
| 75327 | North Carolina macular dystrophy | | 2 Families |
| 3032 | NPHP3-related Meckel-like syndrome | | 10 Cases |
| 397615 | Obesity due to CEP19 deficiency | | 15 Cases |
| 66628 | Obesity due to congenital leptin deficiency | | 30 Cases |
| 71526 | Obesity due to pro-opiomelanocortin deficiency | | 7 Cases |
| 71528 | Obesity due to prohormone convertase I deficiency | | 16 Cases |
| 88643 | Obesity-colitis-hypothyroidism-cardiac hypertrophy-developmental delay syndrome | | 2 Cases |
| 198 | Occipital horn syndrome | | 20 Cases |
| 280640 | Occipital pachygyria and polymicrogyria | | 3 Cases |
| 2704 | Ochoa syndrome | | 100 Cases |

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|---|---|---|---|
| 352740 | Ocular albinism with congenital sensorineural deafness | | 3 Families |
| 1000 | Ocular albinism with late-onset sensorineural deafness | | 9 Cases |
| 1125 | Ocular motor apraxia, Cogan type | | 50 Cases |
| 2714 | Oculo-palato-cerebral syndrome | | 5 Cases |
| 157962 | Oculoauricular syndrome, Schorderet type | | 5 Cases |
| 398156 | Oculoauriculofrontonasal syndrome | | 41 Cases |
| 2705 | Oculocerebral dysplasia | | 2 Cases |
| 2719 | Oculocerebral hypopigmentation syndrome, Cross type | | 14 Cases |
| 2720 | Oculocerebral hypopigmentation syndrome, Preus type | | 2 Cases |
| 1647 | Oculocerebrocutaneous syndrome | | 38 Cases |
| 2707 | Oculocerebrofacial syndrome, Kaufman type | | 14 Cases |
| 534 | Oculocerebrorenal syndrome of Lowe | 0.2 *P* | |
| 55 | Oculocutaneous albinism | 5.9 *P* | |
| 352731 | Oculocutaneous albinism type 1 | 2.5 *P* | |
| 79431 | Oculocutaneous albinism type 1A | 1.3 *P* | |
| 79434 | Oculocutaneous albinism type 1B | 1.3 *P* | |
| 79432 | Oculocutaneous albinism type 2 | 2.55 *P* | |
| 79435 | Oculocutaneous albinism type 4 | 1.0 *P* | |
| 370091 | Oculocutaneous albinism type 5 | | 1 Family |
| 370097 | Oculocutaneous albinism type 6 | | 1 Case |
| 352745 | Oculocutaneous albinism type 7 | | 9 Cases |
| 2709 | Oculodental syndrome, Rutherfurd type | | 1 Family |
| 2710 | Oculodentodigital dysplasia | | 243 Cases |
| 1876 | Oculogastrointestinal muscular dystrophy | | 1 Family |
| 1794 | Oculomaxillofacial dysostosis | | 4 Cases |
| 2713 | Oculoosteocutaneous syndrome | | 3 Cases |
| 99806 | Oculootodental syndrome | | 1 Family |
| 2715 | Oculorenocerebellar syndrome | | 5 Cases |
| 2717 | Oculotrichoanal syndrome | | 20 Cases |
| 2718 | Oculotrichodysplasia | | 2 Cases |
| 2722 | Odonto-onycho dysplasia-alopecia syndrome | | 2 Cases |
| 2721 | Odonto-onycho-dermal dysplasia | | 30 Cases |
| 69082 | Odonto-tricho-ungual-digito-palmar syndrome | | 21 Cases |
| 166272 | Odontochondrodysplasia | | 11 Cases |
| 77295 | Odontoleukodystrophy | | 4 Cases |
| 2724 | Odontomatosis-aortae esophagus stenosis syndrome | | 3 Cases |

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|---|---|---|---|
| 1811 | Odontomicronychial dysplasia | | 5 Cases |
| 2723 | Odontotrichomelic syndrome | | 4 Cases |
| 391655 | Off-periods in Parkinson disease not responding to oral treatment | 4.15 *P* * | |
| 276432 | Ogden syndrome | | 8 Cases |
| 75382 | Oguchi disease | | 50 Cases |
| 2729 | Okamoto syndrome | | 5 Cases |
| 85410 | Oligoarticular juvenile idiopathic arthritis | 20.5 *P* * | |
| 251651 | Oligoastrocytic tumor | 0.11 *I* * | |
| 75378 | Oligocone trichromacy | | 14 Cases |
| 46484 | Oligodendroglial tumor | 0.35 *I* * | |
| 251627 | Oligodendroglioma | 0.25 *I* * | |
| 300576 | Oligodontia-cancer predisposition syndrome | | 2 Families |
| 2920 | Oliver syndrome | | 7 Cases |
| 39041 | Omenn syndrome | | 25 Cases |
| 2733 | Omodysplasia | | 30 Cases |
| 660 | Omphalocele | 11.7 *BP* * | |
| 3164 | Omphalocele syndrome, Shprintzen-Goldberg type | | 5 Cases |
| 496693 | Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome | | 7 Cases |
| 352540 | Oncogenic osteomalacia | | 400 Cases |
| 300504 | Onychocytic matricoma | | 5 Cases |
| 300512 | Onychomatricoma | | 50 Cases |
| 2741 | Ophthalmomandibulomelic dysplasia | | 3 Cases |
| 2743 | Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome | | 6 Cases |
| 2745 | Opitz G/BBB syndrome | 3.0 *P* * | |
| 2746 | Opsismodysplasia | | 30 Cases |
| 1183 | Opsoclonus-myoclonus syndrome | 0.02 *I* * | |
| 401777 | Optic atrophy-intellectual disability syndrome | | 6 Cases |
| 496790 | Optic atrophy-peripheral neuropathy-developmental delay syndrome | | 8 Cases |
| 313800 | Optic nerve edema-splenomegaly syndrome | | 3 Cases |
| 2086 | Optic pathway glioma | 0.12 *I* | |
| 508501 | Oral-facial-digital syndrome with short stature and brachymesophalangy | | 3 Cases |
| 52994 | Orbital leiomyoma | | 26 Cases |
| 664 | Ornithine transcarbamylase deficiency | 1.4 *P* * | |
| 664 | Ornithine transcarbamylase deficiency | 1.77 *BP* | |

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|---|---|---|---|
| 2750 | Orofaciodigital syndrome type 1 | 1.2 *BP* * | |
| 2751 | Orofaciodigital syndrome type 2 | | 20 Cases |
| 2752 | Orofaciodigital syndrome type 3 | | 5 Cases |
| 2753 | Orofaciodigital syndrome type 4 | | 29 Cases |
| 2919 | Orofaciodigital syndrome type 5 | | 12 Cases |
| 2754 | Orofaciodigital syndrome type 6 | | 2 Families |
| 2755 | Orofaciodigital syndrome type 8 | | 20 Cases |
| 141007 | Orofaciodigital syndrome type 9 | | 10 Cases |
| 141327 | Orofaciodigital syndrome type 12 | | 1 Case |
| 141330 | Orofaciodigital syndrome type 13 | | 1 Case |
| 434179 | Orofaciodigital syndrome type 14 | | 2 Families |
| 2760 | OSLAM syndrome | | 3 Cases |
| 73230 | Ossification anomalies-psychomotor developmental delay syndrome | | 2 Cases |
| 2764 | Osteochondritis dissecans | 35.0 *P* * | |
| 2653 | Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome | | 2 Cases |
| 2763 | Osteocraniostenosis | | 30 Cases |
| 666 | Osteogenesis imperfecta | 10.0 *P* * | |
| 216804 | Osteogenesis imperfecta type 2 | 0.4 *BP* * | |
| 216828 | Osteogenesis imperfecta type 5 | | 47 Cases |
| 2773 | Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome | | 2 Cases |
| 2645 | Osteoglosphonic dysplasia | | 7 Cases |
| 2777 | Osteomesopyknosis | | 35 Cases |
| 2780 | Osteopathia striata-cranial sclerosis syndrome | | 100 Cases |
| 2779 | Osteopathia striata-pigmentary dermopathy-white forelock syndrome | | 3 Cases |
| 2324 | Osteopenia-intellectual disability-sparse hair syndrome | | 2 Cases |
| 91133 | Osteopenia-myopia-hearing loss-intellectual disability-facial dysmorphism syndrome | | 2 Cases |
| 2781 | Osteopetrosis and related disorders | 1.0 *I* * | |
| 2785 | Osteopetrosis with renal tubular acidosis | | 100 Cases |
| 178389 | Osteopetrosis-hypogammaglobulinemia syndrome | | 8 Cases |
| 2786 | Osteoporosis-oculocutaneous hypopigmentation syndrome | | 1 Case |
| 2788 | Osteoporosis-pseudoglioma syndrome | 0.05 *P* * | |
| 668 | Osteosarcoma | 0.23 *I* * | |
| 178377 | Osteosclerosis-developmental delay-craniosynostosis syndrome | | 13 Cases |
| 75325 | Osteosclerosis-ichthyosis-premature ovarian failure syndrome | | 3 Cases |
| 500548 | Osteosclerotic metaphyseal dysplasia | | 7 Cases |
| 2791 | Otodental syndrome | | 10 Families |
| 2793 | Otoonychoperoneal syndrome | | 6 Cases |
| 90652 | Otopalatodigital syndrome type 2 | | 40 Cases |
| 1427 | Otospondylomegaepiphyseal dysplasia | | 30 Cases |
| 213500 | Ovarian cancer | 30.0 *P* * | |
| 99853 | Ovarioleukodystrophy | | 17 Cases |
| 498488 | Overgrowth syndrome with 2q37 translocation | | 4 Cases |
| 137634 | Overgrowth-macrocephaly-facial dysmorphism syndrome | | 6 Families |
| 498485 | Overgrowth-metaphyseal undermodeling-spondylar dysplasia syndrome | | 4 Cases |
| 3203 | Overhydrated hereditary stomatocytosis | | 20 Families |
| 2796 | Pachydermoperiostosis | | 204 Cases |
| 2798 | Pachygyria-intellectual disability-epilepsy syndrome | | 5 Cases |
| 2309 | Pachyonychia congenita | | 1000 Cases |
| 1952 | Pacman dysplasia | | 4 Cases |
| 180275 | Paget disease of the nipple | 0.51 *I* * | |
| 991 | PAGOD syndrome | | 6 Cases |
| 1993 | Pai syndrome | | 37 Cases |
| 300501 | Painful orbital and systemic neurofibromas-marfanoid habitus syndrome | | 4 Cases |
| 477993 | Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome | | 3 Cases |
| 672 | Pallister-Hall syndrome | | 100 Cases |
| 140966 | Palmoplantar keratoderma, Nagashima type | | 40 Cases |
| 2202 | Palmoplantar keratoderma-deafness syndrome | | 10 Families |
| 2198 | Palmoplantar keratoderma-esophageal carcinoma syndrome | | 10 Families |
| 2201 | Palmoplantar keratoderma-spastic paralysis syndrome | | 25 Cases |
| 85112 | Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome | | 5 Cases |
| 309108 | Pancreatic colipase deficiency | | 2 Cases |
| 2255 | Pancreatic hypoplasia-diabetes-congenital heart disease syndrome | | 10 Cases |
| 199337 | Pancreatic insufficiency-anemia-hyperostosis syndrome | | 5 Cases |
| 677 | Pancreatoblastoma | | 60 Cases |

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|---|---|---|---|
| 317473 | Pancytopenia due to IKZF1 mutations | | 39 Cases |
| 401764 | Pancytopenia-developmental delay syndrome | | 3 Cases |
| 157850 | Pantothenate kinase-associated neurodegeneration | 0.15 *P* * | |
| 319298 | Papillary renal cell carcinoma | 0.14 *I* * | |
| 678 | Papillon-Lefèvre syndrome | 0.25 *P* | |
| 2812 | Parana hard skin syndrome | | 8 Cases |
| 63455 | Paraneoplastic pemphigus | | 60 Cases |
| 2824 | Paraplegia-intellectual disability-hyperkeratosis syndrome | | 6 Cases |
| 143 | Parathyroid carcinoma | 0.02 *I* * | |
| 2825 | PARC syndrome | | 2 Cases |
| 251290 | Parietal foramina with clavicular hypoplasia | | 8 Cases |
| 851 | Paris-Trousseau thrombocytopenia | | 50 Cases |
| 53583 | Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity | | 20 Cases |
| 98811 | Paroxysmal exertion-induced dyskinesia | | 50 Cases |
| 46348 | Paroxysmal extreme pain disorder | | 4 Families |
| 157835 | Paroxysmal hemicrania | 2.0 *P* * | |
| 98809 | Paroxysmal kinesigenic dyskinesia | 0.6 *P* | |
| 447 | Paroxysmal nocturnal hemoglobinuria | 2.0 *P* * | |
| 98810 | Paroxysmal non-kinesigenic dyskinesia | 0.1 *P* | |
| 1330 | Partial atrioventricular canal | 30.0 *P* * | |
| 1330 | Partial atrioventricular canal | 20.0 *BP* * | |
| 1646 | Partial chromosome Y deletion | 20.8 *P* | |
| 401959 | Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome | | 2 Cases |
| 90076 | Partial deep dermal and full thickness burns | 10.0 *P* * | |
| 262941 | Partial duplication of the long arm of chromosome 14 | | 50 Cases |
| 2805 | Partial pancreatic agenesis | | 50 Cases |
| 94083 | Partington syndrome | | 2 Families |
| 86789 | Patella aplasia/hypoplasia | | 5 Families |
| 228190 | Patent ductus arteriosus-bicuspid aortic valve-hand anomalies syndrome | | 7 Cases |
| 261304 | Paternal 20q13.2q13.3 microdeletion syndrome | | 2 Cases |
| 96192 | Paternal uniparental disomy of chromosome 7 | | 4 Cases |
| 2439 | Patterson-Stevenson-Fontaine syndrome | | 7 Cases |
| 438134 | PCNA-related progressive neurodegenerative photosensitivity syndrome | | 4 Cases |
| 439822 | PDE4D haploinsufficiency syndrome | | 7 Cases |
| 699 | Pearson syndrome | | 95 Cases |
| 2835 | Pectus excavatum-macrocephaly-dysplastic nails syndrome | | 1 Family |
| 93682 | Pediatric Castleman disease | | 150 Cases |
| 487809 | Pediatric collagenous gastritis | | 24 Cases |
| 33402 | Pediatric hepatocellular carcinoma | 0.15 *I* * | |
| 263548 | Peeling skin syndrome type A | | 40 Families |
| 263553 | Peeling skin syndrome type B | | 30 Families |
| 444138 | Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome | | 4 Cases |
| 99807 | PEHO-like syndrome | | 10 Cases |
| 702 | Pelizaeus-Merzbacher disease | 0.25 *P* * | |
| 280219 | Pelizaeus-Merzbacher disease, classic form | 0.17 *P* * | |
| 280210 | Pelizaeus-Merzbacher disease, connatal form | 0.03 *P* * | |
| 280224 | Pelizaeus-Merzbacher disease, transitional form | 0.03 *P* * | |
| 2840 | Pelvic dysplasia-arthrogryposis of lower limbs syndrome | | 5 Cases |
| 2839 | Pelvis-shoulder dysplasia | | 10 Cases |
| 93333 | Pelviscapular dysplasia | | 4 Cases |
| 704 | Pemphigus vulgaris | 18.0 *P* * | |
| 705 | Pendred syndrome | 7.0 *P* * | |
| 49 | Penile agenesis | | 80 Cases |
| 313936 | PENS syndrome | | 13 Cases |
| 1335 | Pentalogy of Cantrell | 0.67 *BP* | |
| 2847 | Pericardial and diaphragmatic defect | | 20 Cases |
| 65250 | Perineural cyst | 50.0 *I* * | |
| 436166 | Periodic fever-infantile enterocolitis-autoinflammatory syndrome | | 4 Cases |
| 397750 | Periodic paralysis with later-onset distal motor neuropathy | | 9 Cases |
| 397755 | Periodic paralysis with transient compartment-like syndrome | | 4 Cases |
| 139426 | Perioral myoclonia with absences | | 10 Cases |
| 563 | Peripartum cardiomyopathy | 30.0 *BP* | |
| 163746 | Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease | | 40 Cases |
| 1795 | Peripheral dysostosis | | 6 Cases |

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|---|---|---|---|
| 2400 | Peripheral motor neuropathy-dysautonomia syndrome | | 2 Cases |
| 397744 | Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome | | 15 Cases |
| 97927 | Peripheral resistance to thyroid hormones | 2.5 *P* * | |
| 168816 | Peritoneal cystic mesothelioma | | 150 Cases |
| 2849 | Perlman syndrome | | 30 Cases |
| 226292 | Permanent congenital hypothyroidism | 33.3 *BP* * | |
| 99885 | Permanent neonatal diabetes mellitus | 0.38 *BP* * | |
| 65288 | Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome | | 4 Cases |
| 2971 | Peroxisomal acyl-CoA oxidase deficiency | | 40 Cases |
| 2855 | Perrault syndrome | | 61 Cases |
| 178509 | Perry syndrome | | 53 Cases |
| 97341 | Persistent placoid maculopathy | | 5 Cases |
| 300324 | Persistent polyclonal B-cell lymphocytosis | | 154 Cases |
| 708 | Peters anomaly | | 60 Cases |
| 709 | Peters plus syndrome | | 100 Cases |
| 2869 | Peutz-Jeghers syndrome | 0.4 *P* * | |
| 2869 | Peutz-Jeghers syndrome | 2.2 *BP* | |
| 42642 | PFAPA syndrome | | 500 Cases |
| 710 | Pfeiffer syndrome | 1.0 *I* * | |
| 2871 | Pfeiffer-Palm-Teller syndrome | | 2 Cases |
| 319646 | PGM1-CDG | | 46 Cases |
| 443811 | PGM3-CDG | | 20 Cases |
| 42775 | PHACE syndrome | | 300 Cases |
| 2874 | Phakomatosis pigmentokeratotica | | 34 Cases |
| 352636 | Phalangeal microgeodic syndrome | | 50 Cases |
| 2876 | PHAVER syndrome | | 2 Cases |
| 716 | Phenylketonuria | 10.0 *BP* * | |
| 2878 | Phocomelia-ectrodactyly-deafness-sinus arrhythmia syndrome | | 4 Cases |
| 2880 | Phosphoenolpyruvate carboxykinase deficiency | | 10 Cases |
| 3222 | Phosphoribosylpyrophosphate synthetase superactivity | | 30 Families |
| 498228 | Phyllodes tumor of the prostate | | 90 Cases |
| 2885 | Piebald trait-neurologic defects syndrome | | 8 Cases |
| 487825 | Pierpont syndrome | | 7 Cases |
| 2888 | Pierre Robin syndrome-faciodigital anomaly syndrome | | 2 Cases |
| 2670 | Pierson syndrome | | 40 Families |
| 447961 | Pigmentation defects-palmoplantar keratoderma-skin carcinoma syndrome | | 2 Cases |
| 251295 | Pigmented paravenous retinochoroidal atrophy | | 100 Cases |
| 66627 | Pigmented villonodular synovitis | 20.0 *P* * | |
| 2891 | Pili torti-developmental delay-neurological abnormalities syndrome | | 2 Cases |
| 2890 | Pili torti-onychodysplasia syndrome | | 1 Family |
| 2892 | Pilodental dysplasia-refractive errors syndrome | | 2 Cases |
| 251909 | Pineoblastoma | 0.02 *I* * | |
| 2896 | Pitt-Hopkins syndrome | | 500 Cases |
| 221150 | Pitt-Hopkins-like syndrome | | 105 Cases |
| 300385 | Pituitary carcinoma | 0.04 *I* * | |
| 2897 | Pityriasis rubra pilaris | | 48 Cases |
| 521426 | PLAA-associated neurodevelopmental disorder | | 15 Cases |
| 439167 | Placental insufficiency | 33.0 *P* | |
| 99928 | Placental site trophoblastic tumor | 0.02 *I* * | |
| 707 | Plague | 2.2 *I* * | |
| 454714 | Plasma cell leukemia | 0.04 *I* * | |
| 300359 | PLCG2-associated antibody deficiency and immune dysregulation | | 3 Families |
| 99969 | Pleomorphic liposarcoma | 0.05 *I* * | |
| 454821 | Pleomorphic salivary gland adenoma | 2.725 *I* | |
| 251607 | Pleomorphic xanthoastrocytoma | 0.01 *I* * | |
| 449266 | Pleural empyema | 13.0 *P* * | |
| 50251 | Pleural mesothelioma | 3.1 *P* * | |
| 50251 | Pleural mesothelioma | 1.9 *I* * | |
| 64742 | Pleuropulmonary blastoma | 0.5 *BP* * | |
| 284343 | Pleuropulmonary blastoma familial tumor susceptibility syndrome | 0.007 *I* | |
| 280356 | PLIN1-related familial partial lipodystrophy | | 3 Cases |
| 54028 | Plummer-Vinson syndrome | | 25 Cases |
| 476394 | PMP2-related Charcot-Marie-Tooth disease type 1 | | 13 Cases |
| 477817 | PMP22-RAI1 contiguous gene duplication syndrome | | 23 Cases |
| 90066 | Pneumonia caused by Pseudomonas aeruginosa infection | 50.0 *P* * | |
| 221046 | Poikiloderma with neutropenia | | 50 Cases |
| 2911 | Poland syndrome | 1.5 *BP* * | |
| 330009 | Poliomyelitis in patients with immunodeficiencies deemed at risk | 8.0E-4 *P* * | |
| 767 | Polyarteritis nodosa | 3.0 *P* * | |

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| 2795 | Polycystic ovaries-urethral sphincter dysfunction syndrome | | 33 Cases |
| 729 | Polycythemia vera | 30.0 *P* * | |
| 729 | Polycythemia vera | 1.9 *I* * | |
| 2917 | Polydactyly-myopia syndrome | | 1 Family |
| 453533 | Polyendocrine-polyneuropathy syndrome | | 3 Cases |
| 397937 | Polyglucosan body myopathy type 1 | | 11 Cases |
| 456369 | Polyglucosan body myopathy type 2 | | 15 Cases |
| 500533 | Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome | | 17 Cases |
| 183422 | Polymalformative genetic syndrome with increased risk of developing cancer | 10.0 *P* * | |
| 300573 | Polymicrogyria due to TUBB2B mutation | | 36 Cases |
| 250972 | Polymicrogyria with optic nerve hypoplasia | | 4 Cases |
| 732 | Polymyositis | 7.1 *P* * | |
| 732 | Polymyositis | 0.585 *I* * | |
| 171848 | Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome | | 19 Cases |
| 2928 | Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome | | 3 Cases |
| 2934 | Polysyndactyly-cardiac malformation syndrome | | 8 Cases |
| 228410 | Polyvalvular heart disease syndrome | | 19 Cases |
| 477749 | Pontine autosomal dominant microangiopathy with leukoencephalopathy | | 11 Cases |
| 269229 | Pontine tegmental cap dysplasia | | 22 Cases |
| 2254 | Pontocerebellar hypoplasia type 1 | | 40 Families |
| 2524 | Pontocerebellar hypoplasia type 2 | | 81 Families |
| 97249 | Pontocerebellar hypoplasia type 3 | | 3 Families |
| 166063 | Pontocerebellar hypoplasia type 4 | | 10 Families |
| 166068 | Pontocerebellar hypoplasia type 5 | | 3 Cases |
| 166073 | Pontocerebellar hypoplasia type 6 | | 10 Cases |
| 284339 | Pontocerebellar hypoplasia type 7 | | 4 Cases |
| 324569 | Pontocerebellar hypoplasia type 8 | | 6 Cases |
| 369920 | Pontocerebellar hypoplasia type 9 | | 14 Cases |
| 411493 | Pontocerebellar hypoplasia type 10 | | 23 Cases |
| 294963 | Popliteal pterygium syndrome | 0.3 *P* * | |
| 2941 | Porencephaly-cerebellar hypoplasia-internal malformations syndrome | | 2 Cases |
| 306547 | Porencephaly-microcephaly-bilateral congenital cataract syndrome | | 8 Cases |
| 166286 | Porokeratotic eccrine ostial and dermal duct nevus | | 45 Cases |
| 738 | Porphyria | 5.25 *P* | |
| 101330 | Porphyria cutanea tarda | 4.0 *P* * | |
| 101330 | Porphyria cutanea tarda | 0.6 *I* * | |
| 79473 | Porphyria variegata | 0.32 *P* * | |
| 79473 | Porphyria variegata | 0.008 *I* * | |
| 2703 | Port-wine nevi-mega cisterna magna-hydrocephalus syndrome | | 5 Cases |
| 70568 | Post-transplant lymphoproliferative disease | 26.2 *P* * | |
| 246 | Postaxial acrofacial dysostosis | | 30 Cases |
| 420584 | Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome | | 112 Cases |
| 2916 | Postaxial polydactyly-dental and vertebral anomalies syndrome | | 3 Cases |
| 2730 | Postaxial tetramelic oligodactyly | | 4 Cases |
| 98971 | Posterior amorphous corneal dystrophy | | 11 Families |
| 88628 | Posterior column ataxia-retinitis pigmentosa syndrome | | 20 Cases |
| 2064 | Posterior fusion of lumbosacral vertebrae-blepharoptosis syndrome | | 3 Cases |
| 93110 | Posterior urethral valve | 2.0 *P* * | |
| 93110 | Posterior urethral valve | 4.125 *BP* * | |
| 477673 | Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome | | 17 Cases |
| 279947 | Postorgasmic illness syndrome | | 45 Cases |
| 443236 | Postural orthostatic tachycardia syndrome due to NET deficiency | | 2 Cases |
| 52022 | Potocki-Shaffer syndrome | | 40 Cases |
| 217067 | Pouchitis | 22.0 *P* * | |
| 79083 | PPARG-related familial partial lipodystrophy | | 10 Cases |
| 739 | Prader-Willi syndrome | 3.1 *BP* * | |
| 398069 | Prader-Willi syndrome due to a point mutation | | 28 Cases |
| 398073 | Prader-Willi-like syndrome | | 117 Cases |
| 398079 | Prader-Willi-like syndrome due to a point mutation | | 4 Cases |
| 293462 | Pre-Descemet corneal dystrophy | | 5 Cases |
| 2921 | Preaxial polydactyly-colobomata-intellectual disability syndrome | | 2 Cases |
| 275555 | Preeclampsia | 45.0 *P* * | |

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|---|---|---|---|
| 486811 | Prenatal-onset spinal muscular atrophy with congenital bone fractures | | 7 Cases |
| 79410 | Pretibial dystrophic epidermolysis bullosa | | 40 Families |
| 186 | Primary biliary cholangitis | 21.05 *P* | |
| 186 | Primary biliary cholangitis | 3.0 *I* | |
| 169464 | Primary CD59 deficiency | | 6 Cases |
| 244 | Primary ciliary dyskinesia | 5.0 *BP* * | |
| 247522 | Primary ciliary dyskinesia-retinitis pigmentosa syndrome | | 20 Cases |
| 226295 | Primary congenital hypothyroidism | 37.5 *P* * | |
| 541 | Primary cutaneous CD30+ T-cell lymphoproliferative disease | 0.18 *I* * | |
| 542 | Primary cutaneous lymphoma | 0.75 *I* * | |
| 171901 | Primary cutaneous T-cell lymphoma | 24.0 *P* * | |
| 171901 | Primary cutaneous T-cell lymphoma | 5.2 *I* * | |
| 98805 | Primary dystonia, DYT4 type | | 22 Cases |
| 98806 | Primary dystonia, DYT6 type | | 53 Cases |
| 98807 | Primary dystonia, DYT13 type | | 8 Cases |
| 370103 | Primary dystonia, DYT17 type | | 3 Cases |
| 306734 | Primary dystonia, DYT21 type | | 16 Cases |
| 464440 | Primary dystonia, DYT27 type | | 5 Cases |
| 48686 | Primary effusion lymphoma | | 200 Cases |
| 90026 | Primary erythromelalgia | | 30 Families |
| 100085 | Primary hepatic neuroendocrine carcinoma | 0.2 *I* | |
| 369929 | Primary hyperaldosteronism-seizures-neurological abnormalities syndrome | | 2 Cases |
| 2232 | Primary hypergonadotropic hypogonadism-partial alopecia syndrome | | 7 Cases |
| 93599 | Primary hyperoxaluria type 2 | | 10 Cases |
| 93600 | Primary hyperoxaluria type 3 | | 50 Cases |
| 30924 | Primary hypomagnesemia with secondary hypocalcemia | | 100 Cases |
| 90023 | Primary immunodeficiency syndrome due to p14 deficiency | | 4 Cases |
| 75391 | Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency | | 4 Cases |
| 431166 | Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection | | 1 Case |
| 458768 | Primary intralymphatic angioendothelioma | | 30 Cases |
| 35689 | Primary lateral sclerosis | 1.5 *P* * | |
| 77240 | Primary lymphedema | 16.7 *P* * | |
| 98838 | Primary mediastinal large B-cell lymphoma | 3.0 *P* * | |
| 54370 | Primary membranoproliferative glomerulonephritis | 16.0 *P* * | |
| 306558 | Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome | | 8 Cases |
| 391408 | Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome | | 8 Cases |
| 824 | Primary myelofibrosis | 3.0 *P* * | |
| 824 | Primary myelofibrosis | 1.0 *I* * | |
| 238606 | Primary orthostatic tremor | | 390 Cases |
| 189439 | Primary pigmented nodular adrenocortical disease | 0.04 *P* * | |
| 95432 | Primary progressive aphasia | 7.0 *P* | |
| 314566 | Primary progressive apraxia of speech | | 16 Cases |
| 171 | Primary sclerosing cholangitis | 8.1 *P* | |
| 171 | Primary sclerosing cholangitis | 0.65 *I* | |
| 289390 | Primary Sjögren syndrome | 48.99 *P* * | |
| 289390 | Primary Sjögren syndrome | 6.92 *I* | |
| 314701 | Primary systemic amyloidosis | 30.0 *P* * | |
| 412066 | PRKAR1B-related neurodegenerative dementia with intermediate filaments | | 12 Cases |
| 2959 | Progeria-short stature-pigmented nevi syndrome | | 11 Cases |
| 300382 | Progeroid and marfanoid aspect-lipodystrophy syndrome | | 7 Cases |
| 435953 | Progeroid features-hepatocellular carcinoma predisposition syndrome | | 3 Cases |
| 2963 | Progeroid syndrome, Petty type | | 1 Case |
| 448251 | Progressive autosomal recessive ataxia-deafness syndrome | | 13 Cases |
| 75373 | Progressive bifocal chorioretinal atrophy | | 2 Families |
| 139447 | Progressive cavitating leukoencephalopathy | | 19 Cases |
| 247198 | Progressive cerebello-cerebral atrophy | | 7 Cases |
| 431361 | Progressive encephalopathy with leukodystrophy due to DECR deficiency | | 2 Cases |
| 457212 | Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome | | 5 Cases |
| 352447 | Progressive external ophthalmoplegia-myopathy-emaciation syndrome | | 6 Cases |
| 480483 | Progressive familial intrahepatic cholestasis type 4 | | 14 Cases |
| 480476 | Progressive familial intrahepatic cholestasis type 5 | | 4 Cases |

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| 477814 | Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome | | 9 Cases |
| 263516 | Progressive myoclonic epilepsy type 3 | | 9 Families |
| 402082 | Progressive myoclonic epilepsy type 5 | | 3 Cases |
| 280620 | Progressive myoclonic epilepsy type 6 | | 12 Cases |
| 435438 | Progressive myoclonic epilepsy type 7 | | 13 Cases |
| 424027 | Progressive myoclonic epilepsy type 8 | | 4 Cases |
| 457265 | Progressive myoclonic epilepsy type 9 | | 2 Cases |
| 352596 | Progressive myoclonic epilepsy with dystonia | | 5 Cases |
| 100070 | Progressive non-fluent aphasia | 2.5 *P* * | |
| 100070 | Progressive non-fluent aphasia | 0.7 *I* * | |
| 2062 | Progressive non-infectious anterior vertebral fusion | | 67 Cases |
| 217396 | Progressive polyneuropathy with bilateral striatal necrosis | | 4 Cases |
| 352718 | Progressive retinal dystrophy due to retinol transport defect | | 5 Cases |
| 447977 | Progressive scapulohumeroperoneal distal myopathy | | 33 Cases |
| 228012 | Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome | | 4 Families |
| 457395 | Progressive spondyloepimetaphyseal dysplasia-short stature-short fourth metatarsals-intellectual disability syndrome | | 4 Cases |
| 683 | Progressive supranuclear palsy | 6.0 *P* | |
| 683 | Progressive supranuclear palsy | 0.65 *I* | |
| 240103 | Progressive supranuclear palsy-corticobasal syndrome | 0.6 *P* * | |
| 240112 | Progressive supranuclear palsy-progressive non-fluent aphasia syndrome | | 10 Cases |
| 742 | Prolidase deficiency | | 90 Cases |
| 2083 | Prominent glabella-microcephaly-hypogenitalism syndrome | | 2 Cases |
| 35 | Propionic acidemia | 0.2 *P* * | |
| 35 | Propionic acidemia | 1.5 *I* | |
| 324977 | Proteasome disability syndrome | | 40 Cases |
| 251598 | Protoplasmic astrocytoma | 0.01 *I* * | |
| 261197 | Proximal 16p11.2 microdeletion syndrome | 20.0 *P* * | |
| 401768 | Proximal myopathy with extrapyramidal signs | | 15 Cases |
| 521305 | Proximal myopathy with focal depletion of mitochondria | | 4 Cases |
| 606 | Proximal myotonic myopathy | 1.0 *P* * | |
| 70 | Proximal spinal muscular atrophy | 20.0 *BP* * | |
| 70 | Proximal spinal muscular atrophy | 2.6 *I* * | |
| 83330 | Proximal spinal muscular atrophy type 1 | 0.26 *I* * | |
| 83418 | Proximal spinal muscular atrophy type 2 | 1.23 *I* * | |
| 83419 | Proximal spinal muscular atrophy type 3 | 1.1 *I* * | |
| 83420 | Proximal spinal muscular atrophy type 4 | 0.32 *I* * | |
| 3390 | Proximal tubulopathy-diabetes mellitus-cerebellar ataxia syndrome | | 2 Cases |
| 397606 | PrP systemic amyloidosis | | 16 Cases |
| 52530 | Pseudo-von Willebrand disease | | 60 Cases |
| 750 | Pseudoachondroplasia | 3.3 *P* | |
| 221120 | Pseudoaminopterin syndrome | | 11 Cases |
| 85174 | Pseudodiastrophic dysplasia | | 13 Cases |
| 756 | Pseudohypoaldosteronism type 1 | | 107 Cases |
| 757 | Pseudohypoaldosteronism type 2 | | 80 Families |
| 300525 | Pseudohypoaldosteronism type 2D | | 24 Cases |
| 300530 | Pseudohypoaldosteronism type 2E | | 17 Cases |
| 2976 | Pseudoleprechaunism syndrome, Patterson type | | 2 Cases |
| 26790 | Pseudomyxoma peritonei | 0.1 *I* | |
| 2985 | Pseudoprogeria syndrome | | 2 Cases |
| 758 | Pseudoxanthoma elasticum | 2.5 *P* * | |
| 436274 | Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa | | 13 Cases |
| 280794 | Pseudoxanthomatous diffuse cutaneous mastocytosis | | 10 Cases |
| 85436 | Psoriasis-related juvenile idiopathic arthritis | 4.2 *P* * | |
| 505242 | Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome | | 6 Cases |
| 88618 | Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency | | 4 Cases |
| 1578 | Pterin-4 alpha-carbinolamine dehydratase deficiency | | 21 Cases |
| 2988 | Pterygium colli-intellectual disability-digital anomalies syndrome | | 2 Cases |
| 2999 | Ptosis-strabismus-ectopic pupils syndrome | | 1 Family |
| 228396 | Ptosis-upper ocular movement limitation-absence of lacrimal punctum syndrome | | 3 Cases |
| 2997 | Ptosis-vocal cord paralysis syndrome | | 2 Cases |

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| ORPHA Number | Disease or Group of diseases | Estimated prevalence/ incidence (/100,000) | Number of published cases or families |
|---|---|---|---|
| 182090 | Pulmonary arterial hypertension | 3.3 *P* * | |
| 275803 | Pulmonary arterial hypertension associated with congenital heart disease | 0.57 *P* * | |
| 275798 | Pulmonary arterial hypertension associated with connective tissue disease | 0.25 *P* * | |
| 2038 | Pulmonary arteriovenous malformation | 2.5 *I* | |
| 99050 | Pulmonary artery coming from the aorta | | 200 Cases |
| 64741 | Pulmonary blastoma | | 350 Cases |
| 199241 | Pulmonary capillary hemangiomatosis | | 100 Cases |
| 210136 | Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome | | 4 Cases |
| 217080 | Pulmonary fungal infections in patients deemed at risk | 22.0 *P* * | |
| 411703 | Pulmonary non-tuberculous mycobacterial infection | 6.0 *P* * | |
| 31837 | Pulmonary venoocclusive disease | 0.015 *I* * | |
| 79501 | Punctate palmoplantar keratoderma type 1 | | 35 Families |
| 79502 | Punctate palmoplantar keratoderma type 2 | | 13 Cases |
| 438213 | PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome | | 24 Cases |
| 438216 | PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation | | 24 Cases |
| 69084 | Pure hair and nail ectodermal dysplasia | | 20 Cases |
| 760 | Purine nucleoside phosphorylase deficiency | | 70 Cases |
| 763 | Pycnodysostosis | 0.13 *P* | |
| 481152 | PYCR2-related microcephaly-progressive leukoencephalopathy | | 18 Cases |
| 3003 | Pyknoachondrogenesis | | 5 Cases |
| 3005 | Pyle disease | | 30 Cases |
| 48104 | Pyoderma gangrenosum | 0.74 *I* | |
| 69126 | Pyogenic arthritis-pyoderma gangrenosum-acne syndrome | | 34 Cases |
| 183713 | Pyogenic bacterial infections due to MyD88 deficiency | | 24 Cases |
| 2561 | Pyramidal molars-abnormal upper lip syndrome | | 8 Cases |
| 79096 | Pyridoxal phosphate-responsive seizures | 0.2 *P* * | |
| 3006 | Pyridoxine-dependent epilepsy | 0.2 *BP* * | |
| 3008 | Pyruvate carboxylase deficiency | 0.4 *BP* * | |
| 353320 | Pyruvate carboxylase deficiency, benign type | | 5 Cases |
| 2394 | Pyruvate dehydrogenase E3 deficiency | | 20 Cases |
| 781 | Q fever | 0.16 *I* * | |
| 3010 | Qazi-Markouizos syndrome | | 3 Cases |
| 2252 | Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome | | 8 Cases |
| 3026 | Radial ray hypoplasia-choanal atresia syndrome | | 3 Cases |
| 70475 | Radiation proctitis | 35.0 *P* * | |
| 3015 | Radio-renal syndrome | | 4 Cases |
| 71289 | Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome | | 20 Cases |
| 3270 | Radioulnar synostosis-developmental delay-hypotonia syndrome | | 4 Cases |
| 3268 | Radioulnar synostosis-microcephaly-scoliosis syndrome | | 13 Cases |
| 3019 | Ramon syndrome | | 8 Cases |
| 1051 | Ramos-Arroyo syndrome | | 6 Cases |
| 3021 | RAPADILINO syndrome | | 20 Cases |
| 293987 | Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome | | 48 Cases |
| 71517 | Rapid-onset dystonia-parkinsonism | | 100 Cases |
| 213528 | Rare adenocarcinoma of the breast | 3.55 *I* * | |
| 217074 | Rare carcinoma of pancreas | 3.5 *P* | |
| 217074 | Rare carcinoma of pancreas | 3.9 *I* | |
| 88991 | Rare congenital non-syndromic heart malformation | 7.8 *BP* * | |
| 535 | Rare cutaneous lupus erythematosus | 50.0 *P* * | |
| 63443 | Rare epithelial tumor of stomach | 18.6 *I* * | |
| 2415 | Rare lymphatic malformation | 12.5 *P* * | |
| 182114 | Rare urogenital tumor | 0.13 *I* * | |
| 438114 | RARS-related autosomal recessive hypomyelinating leukodystrophy | | 4 Cases |
| 268114 | RAS-associated autoimmune leukoproliferative disease | | 20 Cases |
| 1929 | Rasmussen subacute encephalitis | | 100 Cases |
| 99852 | Ravine syndrome | | 38 Cases |
| 1115 | Recessive aplasia cutis congenita of limbs | | 6 Cases |
| 79409 | Recessive dystrophic epidermolysis bullosa inversa | | 100 Cases |
| 280384 | Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome | | 12 Cases |
| 461 | Recessive X-linked ichthyosis | 16.6 *P* * | |
| 461 | Recessive X-linked ichthyosis | 15.0 *I* * | |
| 64740 | Recurrent acute pancreatitis | 10.0 *P* * | |

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|---|---|---|---|
| 90052 | Recurrent hepatitis C virus induced liver disease in liver transplant recipients | 7.0 *P* * | |
| 480864 | Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome | | 24 Cases |
| 97239 | Reducing body myopathy | | 4 Families |
| 86839 | Refractory anemia with excess blasts | 0.15 *I* * | |
| 168960 | Refractory anemia with excess blasts in transformation | 0.04 *I* * | |
| 773 | Refsum disease | 0.1 *P* * | |
| 83450 | Regional odontodysplasia | | 140 Cases |
| 448267 | Regressive spondylometaphyseal dysplasia | | 2 Cases |
| 98961 | Reis-Bücklers corneal dystrophy | | 81 Cases |
| 728 | Relapsing polychondritis | 0.35 *I* | |
| 217330 | REN-related autosomal dominant tubulointerstitial kidney disease | | 21 Cases |
| 1848 | Renal agenesis, bilateral | 1.7 *BP* * | |
| 93100 | Renal agenesis, unilateral | 50.0 *BP* | |
| 2838 | Renal caliceal diverticuli-deafness syndrome | | 4 Cases |
| 217071 | Renal cell carcinoma | 42.0 *P* * | |
| 217071 | Renal cell carcinoma | 8.35 *I* * | |
| 1475 | Renal coloboma syndrome | | 180 Cases |
| 93108 | Renal dysplasia | 43.5 *BP* * | |
| 93975 | Renier-Gabreels-Jasper syndrome | | 5 Cases |
| 3242 | Renpenning syndrome | | 64 Cases |
| 494344 | RERE-related neurodevelopmental syndrome | | 10 Cases |
| 99832 | Resistance to thyrotropin-releasing hormone syndrome | | 2 Cases |
| 1662 | Restrictive dermopathy | | 30 Cases |
| 33355 | Reticular dysgenesis | 0.03 *I* * | |
| 178307 | Reticulate acropigmentation of Kitamura | | 130 Cases |
| 458763 | Retiform hemangioendothelioma | | 32 Cases |
| 75326 | Retinal arterial tortuosity | | 100 Cases |
| 1574 | Retinal degeneration-nanophthalmos-glaucoma syndrome | | 7 Cases |
| 397758 | Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies | | 14 Cases |
| 3018 | Retinal ischemic syndrome-digestive tract small vessel hyalinosis-diffuse cerebral calcifications syndrome | | 3 Cases |
| 319640 | Retinal macular dystrophy type 2 | | 5 Families |
| 791 | Retinitis pigmentosa | 26.7 *P* | |
| 494439 | Retinitis pigmentosa-hearing loss-premature aging-short stature-facial dysmorphism syndrome | | 3 Cases |
| 3085 | Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome | | 2 Families |
| 436245 | Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome | | 3 Cases |
| 52427 | Retinitis punctata albescens | 0.125 *P* | |
| 790 | Retinoblastoma | 6.0 *BP* | |
| 790 | Retinoblastoma | 0.05 *I* * | |
| 3087 | Retinohepatoendocrinologic syndrome | | 7 Cases |
| 778 | Rett syndrome | 10.0 *P* * | |
| 778 | Rett syndrome | 5.0 *BP* * | |
| 294049 | Reunion Island Larsen-like syndrome | | 30 Cases |
| 3088 | Revesz syndrome | | 4 Cases |
| 244310 | RFT1-CDG | | 8 Cases |
| 69077 | Rhabdoid tumor | | 500 Cases |
| 780 | Rhabdomyosarcoma | 0.59 *I* * | |
| 3099 | Rheumatic fever | 5.0 *I* * | |
| 85408 | Rheumatoid factor-negative juvenile idiopathic arthritis | 8.0 *P* * | |
| 85435 | Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis | 4.2 *P* * | |
| 177 | Rhizomelic chondrodysplasia punctata | 1.0 *P* * | |
| 468717 | Rhizomelic chondrodysplasia punctata type 5 | | 4 Cases |
| 2831 | Rhizomelic dysplasia, Patterson-Lowry type | | 5 Cases |
| 3098 | Rhizomelic syndrome, Urbach type | | 3 Cases |
| 59315 | Rhombencephalosynapsis | | 100 Cases |
| 140976 | RHYNS syndrome | | 4 Cases |
| 97229 | Riboflavin transporter deficiency | | 80 Cases |
| 440706 | Ribose-5-P isomerase deficiency | | 1 Case |
| 3101 | Richieri Costa-da Silva syndrome | | 4 Cases |
| 3102 | Richieri Costa-Pereira syndrome | | 33 Cases |
| 83312 | Rickettsialpox | | 800 Cases |
| 420741 | RIDDLE syndrome | | 2 Cases |
| 217335 | RIN2 syndrome | | 10 Cases |
| 363203 | Ring chromosome | 2.0 *BP* | |
| 1437 | Ring chromosome 1 syndrome | | 35 Cases |
| 96171 | Ring chromosome 2 syndrome | | 18 Cases |

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|---|---|---|---|
| 96172 | Ring chromosome 3 syndrome | | 11 Cases |
| 1447 | Ring chromosome 4 syndrome | | 20 Cases |
| 1448 | Ring chromosome 6 syndrome | | 25 Cases |
| 1449 | Ring chromosome 7 syndrome | | 18 Cases |
| 1450 | Ring chromosome 8 syndrome | | 8 Cases |
| 96173 | Ring chromosome 9 syndrome | | 31 Cases |
| 1438 | Ring chromosome 10 syndrome | | 16 Cases |
| 96175 | Ring chromosome 11 syndrome | | 20 Cases |
| 1439 | Ring chromosome 12 syndrome | | 10 Cases |
| 1440 | Ring chromosome 14 syndrome | | 80 Cases |
| 96177 | Ring chromosome 15 syndrome | | 50 Cases |
| 96178 | Ring chromosome 16 syndrome | | 10 Cases |
| 1441 | Ring chromosome 17 syndrome | | 18 Cases |
| 1442 | Ring chromosome 18 syndrome | | 70 Cases |
| 1443 | Ring chromosome 19 syndrome | | 10 Cases |
| 1444 | Ring chromosome 20 syndrome | | 50 Cases |
| 1446 | Ring chromosome 22 syndrome | | 100 Cases |
| 91481 | Ring dermoid of cornea | | 30 Cases |
| 3103 | Roberts syndrome | | 150 Cases |
| 3104 | Robin sequence-oligodactyly syndrome | | 3 Cases |
| 97360 | Robinow syndrome | | 200 Cases |
| 3105 | Robinow-like syndrome | | 2 Cases |
| 353298 | Roifman syndrome | | 7 Cases |
| 163727 | Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome | | 1 Family |
| 101016 | Romano-Ward syndrome | 40.0 *P* * | |
| 158014 | Rosaï-Dorfman disease | | 1000 Cases |
| 2909 | Rothmund-Thomson syndrome | | 400 Cases |
| 221008 | Rothmund-Thomson syndrome type 1 | | 100 Cases |
| 221016 | Rothmund-Thomson syndrome type 2 | | 200 Cases |
| 3111 | Rotor syndrome | | 50 Cases |
| 83616 | Rubella panencephalitis | | 20 Cases |
| 783 | Rubinstein-Taybi syndrome | 0.7 *BP* * | |
| 353284 | Rubinstein-Taybi syndrome due to EP300 haploinsufficiency | | 34 Cases |
| 397927 | Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome | | 4 Cases |
| 794 | Saethre-Chotzen syndrome | 3.0 *BP* * | |
| 300493 | Sagliker syndrome | | 60 Cases |
| 140969 | Saldino-Mainzer syndrome | | 10 Cases |
| 213557 | Salivary gland type cancer of the breast | 0.05 *I* * | |
| 370938 | Salt-and-pepper syndrome | | 3 Cases |
| 796 | Sandhoff disease | 0.67 *BP* * | |
| 79269 | Sanfilippo syndrome type A | 0.32 *P* * | |
| 79269 | Sanfilippo syndrome type A | 1.4 *BP* | |
| 79270 | Sanfilippo syndrome type B | 0.2 *P* * | |
| 79271 | Sanfilippo syndrome type C | 5.0 *P* * | |
| 797 | Sarcoidosis | 12.5 *P* | |
| 3129 | Sarcosinemia | 2.0 *BP* | |
| 3130 | Satoyoshi syndrome | | 50 Cases |
| 3132 | Say-Barber-Miller syndrome | | 2 Cases |
| 3133 | Say-Field-Coldwell syndrome | | 4 Cases |
| 1003 | Scalp defects-postaxial polydactyly syndrome | | 2 Cases |
| 370052 | SCALP syndrome | | 4 Cases |
| 2036 | Scalp-ear-nipple syndrome | | 30 Cases |
| 431255 | Scapuloperoneal spinal muscular atrophy | | 31 Cases |
| 3134 | SCARF syndrome | | 2 Cases |
| 90080 | Scarring in glaucoma filtration surgical procedures | 22.0 *P* * | |
| 2353 | Schilbach-Rott syndrome | | 18 Cases |
| 1830 | Schimke immuno-osseous dysplasia | | 71 Cases |
| 798 | Schinzel-Giedion syndrome | | 46 Cases |
| 37748 | Schnitzler syndrome | | 150 Cases |
| 98967 | Schnyder corneal dystrophy | | 115 Cases |
| 50944 | Schöpf-Schulz-Passarge syndrome | | 25 Cases |
| 800 | Schwartz-Jampel syndrome | | 129 Cases |
| 185 | Scimitar syndrome | 2.0 *BP* * | |
| 801 | Scleroderma | 42.0 *P* | |
| 801 | Scleroderma | 1.41 *I* | |
| 167635 | Scleromyxedema | | 250 Cases |
| 90400 | Scleromyxedema without monoclonal gammopathy | | 15 Cases |
| 3152 | Sclerosteosis | | 80 Cases |
| 806 | Scott syndrome | | 4 Cases |
| 158029 | Sea-blue histiocytosis | | 60 Cases |
| 168606 | Seborrhea-like dermatitis with psoriasiform elements | | 44 Cases |
| 808 | Seckel syndrome | 0.2 *BP* * | |
| 67039 | Segmental odontomaxillary dysplasia | | 32 Cases |
| 314662 | Segmental progressive overgrowth syndrome with fibroadipose hyperplasia | | 10 Cases |

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|---|---|---|---|
| 79156 | Seizures-intellectual disability due to hydroxylysinuria syndrome | | 6 Cases |
| 466926 | Seizures-scoliosis-macrocephaly syndrome | | 4 Cases |
| 281122 | Self-improving collodion baby | | 25 Cases |
| 3156 | Senior-Loken syndrome | | 150 Cases |
| 217622 | Sensorineural deafness with dilated cardiomyopathy | | 2 Families |
| 66633 | Sensorineural hearing loss-early graying-essential tremor syndrome | | 3 Cases |
| 90051 | Sepsis in premature infants | 32.0 *P* * | |
| 3157 | Septo-optic dysplasia spectrum | 10.0 *BP* * | |
| 139466 | SERKAL syndrome | | 3 Cases |
| 85165 | Severe achondroplasia-developmental delay-acanthosis nigricans syndrome | | 5 Cases |
| 438207 | Severe autosomal recessive macrothrombocytopenia | | 2 Cases |
| 183660 | Severe combined immunodeficiency | 1.65 *BP* * | |
| 277 | Severe combined immunodeficiency due to adenosine deaminase deficiency | 0.2 *P* * | |
| 277 | Severe combined immunodeficiency due to adenosine deaminase deficiency | 0.3 *BP* * | |
| 357237 | Severe combined immunodeficiency due to CARD11 deficiency | | 3 Cases |
| 331206 | Severe combined immunodeficiency due to complete RAG1/2 deficiency | 1.0 *P* * | |
| 228003 | Severe combined immunodeficiency due to CORO1A deficiency | | 6 Cases |
| 420573 | Severe combined immunodeficiency due to CTPS1 deficiency | | 8 Cases |
| 317425 | Severe combined immunodeficiency due to DNA-PKcs deficiency | | 2 Cases |
| 397787 | Severe combined immunodeficiency due to IKK2 deficiency | | 9 Cases |
| 504523 | Severe combined immunodeficiency due to LAT deficiency | | 3 Cases |
| 280142 | Severe combined immunodeficiency due to LCK deficiency | | 4 Cases |
| 300298 | Severe congenital hypochromic anemia with ringed sideroblasts | | 3 Cases |
| 42738 | Severe congenital neutropenia | 0.07 *P* | |
| 42738 | Severe congenital neutropenia | 0.4 *BP* * | |
| 369992 | Severe dermatitis-multiple allergies-metabolic wasting syndrome | | 3 Cases |
| 329249 | Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency | | 13 Cases |
| 352577 | Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome | | 28 Cases |
| 488627 | Severe growth deficiency-strabismus-extensive dermal melanocytosis-intellectual disability syndrome | | 3 Cases |
| 169802 | Severe hemophilia A | 2.8 *P* * | |
| 169793 | Severe hemophilia B | 0.8 *P* * | |
| 745 | Severe hereditary thrombophilia due to congenital protein C deficiency | 0.16 *BP* | |
| 467176 | Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome | | 6 Cases |
| 280763 | Severe intellectual disability and progressive spastic paraplegia | | 15 Cases |
| 466688 | Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome | | 6 Cases |
| 94066 | Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia | | 2 Cases |
| 438178 | Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency | | 3 Cases |
| 436141 | Severe intellectual disability-hypotonia-strabismus-coarse face-planovalgus syndrome | | 6 Cases |
| 363686 | Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome | | 4 Cases |
| 397933 | Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome | | 3 Cases |
| 404473 | Severe intellectual disability-progressive spastic diplegia syndrome | | 4 Cases |
| 391307 | Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome | | 3 Cases |
| 324307 | Severe lateral tibial bowing with short stature | | 2 Cases |
| 1236 | Severe microbrachycephaly-intellectual disability-athetoid cerebral palsy syndrome | | 2 Cases |
| 369939 | Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome | | 7 Cases |
| 527450 | Severe myopia-generalized joint laxity-short stature syndrome | | 5 Cases |

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|---|---|---|---|
| 314655 | Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion | | 7 Cases |
| 397593 | Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency | | 5 Cases |
| 209370 | Severe neonatal-onset encephalopathy with microcephaly | | 30 Cases |
| 363400 | Severe neurodegenerative syndrome with lipodystrophy | | 10 Cases |
| 500545 | Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract | | 6 Cases |
| 3078 | Severe X-linked intellectual disability, Gustavson type | | 7 Cases |
| 238329 | Severe X-linked mitochondrial encephalomyopathy | | 2 Cases |
| 363489 | Sex cord-stromal tumor of testis | 0.02 *I* * | |
| 810 | Shigellosis | 1.68 *I* * | |
| 99063 | Shone complex | | 100 Cases |
| 104008 | Short bowel syndrome | 3.4 *P* * | |
| 66518 | Short fifth metacarpals-insulin resistance syndrome | | 6 Cases |
| 498497 | Short rib-polydactyly syndrome type 5 | | 2 Cases |
| 93269 | Short rib-polydactyly syndrome, Majewski type | | 34 Cases |
| 314811 | Short stature due to GHSR deficiency | | 8 Cases |
| 629 | Short stature due to growth hormone qualitative anomaly | | 3 Cases |
| 2867 | Short stature, Brussels type | | 2 Cases |
| 435804 | Short stature-advanced bone age-early-onset osteoarthritis syndrome | | 3 Families |
| 397623 | Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome | | 4 Cases |
| 464288 | Short stature-brachydactyly-obesity-global developmental delay syndrome | | 6 Cases |
| 2994 | Short stature-craniofacial anomalies-genital hypoplasia syndrome | | 3 Families |
| 2866 | Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome | | 2 Cases |
| 314394 | Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome | | 14 Cases |
| 391677 | Short stature-optic atrophy-Pelger-Huët anomaly syndrome | | 34 Cases |
| 85442 | Short stature-pituitary and cerebellar defects-small sella turcica syndrome | | 5 Families |
| 2868 | Short stature-valvular heart disease-characteristic facies syndrome | | 3 Cases |
| 2865 | Short stature-webbed neck-heart disease syndrome | | 4 Cases |
| 2863 | Short stature-wormian bones-dextrocardia syndrome | | 3 Cases |
| 3163 | SHORT syndrome | | 32 Cases |
| 2832 | Short tarsus-absence of lower eyelashes syndrome | | 11 Cases |
| 357175 | Short ulna-dysmorphism-hypotonia-intellectual disability syndrome | | 4 Cases |
| 935 | Short-limb skeletal dysplasia with severe combined immunodeficiency | | 19 Cases |
| 2462 | Shprintzen-Goldberg syndrome | | 60 Cases |
| 811 | Shwachman-Diamond syndrome | 0.28 *P* | |
| 811 | Shwachman-Diamond syndrome | 0.5 *BP* | |
| 309294 | Sialidosis | 0.05 *BP* * | |
| 3166 | Sialuria | | 5 Cases |
| 232 | Sickle cell anemia | 22.0 *P* * | |
| 3167 | Siegler-Brewer-Carey syndrome | | 2 Cases |
| 71276 | Silent sinus syndrome | | 98 Cases |
| 3168 | Sillence syndrome | | 5 Cases |
| 813 | Silver-Russell syndrome | 0.7 *BP* * | |
| 813 | Silver-Russell syndrome | 15.5 *I* * | |
| 397590 | Silver-Russell syndrome due to a point mutation | | 8 Cases |
| 373 | Simpson-Golabi-Behmel syndrome | | 250 Cases |
| 500163 | SIN3A-related intellectual disability syndrome | | 15 Cases |
| 85191 | Singleton-Merten dysplasia | | 22 Cases |
| 324321 | Sinoatrial node dysfunction and deafness | | 8 Cases |
| 3169 | Sirenomelia | 0.01 *P* | |
| 3169 | Sirenomelia | 0.98 *BP* | |
| 2882 | Sitosterolemia | | 100 Cases |
| 488437 | SIX2-related frontonasal dysplasia | | 1 Family |
| 1858 | Skeletal dysplasia-epilepsy-short stature syndrome | | 7 Cases |
| 508533 | Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome | | 12 Cases |
| 477831 | Skeletal overgrowth-craniofacial dysmorphism-hyperelastic skin-white matter lesions syndrome | | 2 Cases |
| 293165 | Skin fragility-woolly hair-palmoplantar keratoderma syndrome | | 7 Cases |
| 238459 | SLC35A1-CDG | | 3 Cases |
| 356961 | SLC35A2-CDG | | 4 Cases |
| 468699 | SLC39A8-CDG | | 10 Cases |
| 70573 | Small cell lung cancer | 11.2 *P* * | |

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|---|---|---|---|
| 466962 | SMARCA4-deficient sarcoma of thorax | | 19 Cases |
| 93974 | Smith-Fineman-Myers syndrome | | 11 Families |
| 818 | Smith-Lemli-Opitz syndrome | 3.7 *BP* * | |
| 819 | Smith-Magenis syndrome | 4.0 *P* | |
| 178355 | Smith-McCort dysplasia | | 16 Cases |
| 820 | Sneddon syndrome | 0.4 *I* * | |
| 91496 | Snowflake vitreoretinal degeneration | | 50 Cases |
| 306577 | Sodium channelopathy-related small fiber neuropathy | | 8 Cases |
| 3394 | Soft tissue sarcoma | 30.0 *P* * | |
| 3394 | Soft tissue sarcoma | 4.74 *I* * | |
| 97230 | Solar urticaria | 36.0 *P* * | |
| 209964 | Solitary rectal ulcer syndrome | 1.0 *I* * | |
| 97283 | Somatostatinoma | 0.0025 *I* * | |
| 821 | Sotos syndrome | 7.1 *BP* | |
| 79132 | Sparse hair-short stature-skin anomalies syndrome | | 4 Cases |
| 1182 | Spastic ataxia with congenital miosis | | 3 Families |
| 2572 | Spastic ataxia-corneal dystrophy syndrome | | 1 Family |
| 2815 | Spastic paraparesis-deafness syndrome | | 6 Cases |
| 99015 | Spastic paraplegia type 2 | | 100 Cases |
| 99013 | Spastic paraplegia type 7 | 4.0 *P* * | |
| 2819 | Spastic paraplegia-facial-cutaneous lesions syndrome | | 5 Cases |
| 2818 | Spastic paraplegia-glaucoma-intellectual disability syndrome | | 2 Families |
| 521390 | Spastic paraplegia-intellectual disability-nystagmus-obesity syndrome | | 4 Cases |
| 2820 | Spastic paraplegia-nephritis-deafness syndrome | | 4 Cases |
| 2821 | Spastic paraplegia-neuropathy-poikiloderma syndrome | | 1 Family |
| 320406 | Spastic paraplegia-optic atrophy-neuropathy syndrome | | 75 Cases |
| 329475 | Spastic paraplegia-Paget disease of bone syndrome | | 1 Family |
| 2826 | Spastic paraplegia-precocious puberty syndrome | | 2 Cases |
| 464282 | Spastic paraplegia-severe developmental delay-epilepsy syndrome | | 16 Cases |
| 3011 | Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome | | 2 Cases |
| 447997 | Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome | | 15 Cases |
| 352403 | Spectrin-associated autosomal recessive cerebellar ataxia | | 2 Families |
| 99865 | Spermatocytic seminoma | 0.03 *I* * | |
| 314432 | Spigelian hernia-cryptorchidism syndrome | | 15 Cases |
| 1217 | Spinal atrophy-ophthalmoplegia-pyramidal syndrome | | 2 Cases |
| 90058 | Spinal cord injury | 32.0 *P* * | |
| 98920 | Spinal muscular atrophy with respiratory distress type 1 | | 128 Cases |
| 404521 | Spinal muscular atrophy with respiratory distress type 2 | | 1 Case |
| 73245 | Spinal muscular atrophy-Dandy-Walker malformation-cataracts syndrome | | 2 Cases |
| 2590 | Spinal muscular atrophy-progressive myoclonic epilepsy syndrome | | 10 Cases |
| 98755 | Spinocerebellar ataxia type 1 | 1.5 *P* | |
| 98756 | Spinocerebellar ataxia type 2 | 1.5 *P* | |
| 98757 | Spinocerebellar ataxia type 3 | 1.5 *P* | |
| 98766 | Spinocerebellar ataxia type 5 | | 3 Families |
| 98767 | Spinocerebellar ataxia type 11 | | 51 Cases |
| 98762 | Spinocerebellar ataxia type 12 | | 40 Families |
| 98768 | Spinocerebellar ataxia type 13 | | 20 Cases |
| 98763 | Spinocerebellar ataxia type 14 | | 20 Families |
| 98769 | Spinocerebellar ataxia type 15/16 | | 80 Cases |
| 98759 | Spinocerebellar ataxia type 17 | | 100 Families |
| 98771 | Spinocerebellar ataxia type 18 | | 26 Cases |
| 98772 | Spinocerebellar ataxia type 19/22 | | 12 Cases |
| 101110 | Spinocerebellar ataxia type 20 | | 20 Cases |
| 98773 | Spinocerebellar ataxia type 21 | | 35 Cases |
| 101108 | Spinocerebellar ataxia type 23 | | 4 Families |
| 101111 | Spinocerebellar ataxia type 25 | | 10 Cases |
| 101112 | Spinocerebellar ataxia type 26 | | 1 Family |
| 98764 | Spinocerebellar ataxia type 27 | | 30 Cases |
| 208513 | Spinocerebellar ataxia type 29 | | 50 Cases |
| 211017 | Spinocerebellar ataxia type 30 | | 6 Cases |
| 217012 | Spinocerebellar ataxia type 31 | | 30 Families |
| 276183 | Spinocerebellar ataxia type 32 | | 1 Family |
| 1955 | Spinocerebellar ataxia type 34 | | 27 Cases |
| 276193 | Spinocerebellar ataxia type 35 | | 3 Families |
| 276198 | Spinocerebellar ataxia type 36 | | 100 Families |
| 363710 | Spinocerebellar ataxia type 37 | | 9 Cases |
| 423296 | Spinocerebellar ataxia type 38 | | 4 Families |
| 423275 | Spinocerebellar ataxia type 40 | | 5 Cases |

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|---|---|---|---|
| 458798 | Spinocerebellar ataxia type 41 | | 1 Case |
| 458803 | Spinocerebellar ataxia type 42 | | 25 Cases |
| 497764 | Spinocerebellar ataxia type 43 | | 7 Cases |
| 94124 | Spinocerebellar ataxia with axonal neuropathy type 1 | | 9 Cases |
| 1185 | Spinocerebellar ataxia-dysmorphism syndrome | | 3 Cases |
| 3177 | Spinocerebellar degeneration-corneal dystrophy syndrome | | 2 Cases |
| 86854 | Splenic marginal zone lymphoma | 0.5 *P* * | |
| 2063 | Splenogonadal fusion-limb defects-micrognathia syndrome | | 30 Cases |
| 71271 | Split hand-split foot-deafness syndrome | | 22 Cases |
| 488232 | Split-foot malformation-mesoaxial polydactyly syndrome | | 5 Cases |
| 93357 | SPONASTRIME dysplasia | | 16 Cases |
| 228387 | Spondylo-megaepiphyseal-metaphyseal dysplasia | | 19 Cases |
| 85194 | Spondylo-ocular syndrome | | 7 Cases |
| 3180 | Spondylocamptodactyly syndrome | | 5 Cases |
| 3275 | Spondylocarpotarsal synostosis | | 24 Cases |
| 94095 | Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome | | 3 Cases |
| 329252 | Spondylocostal dysostosis-hypospadias-intellectual disability syndrome | | 2 Cases |
| 1855 | Spondyloenchondrodysplasia | | 36 Cases |
| 93346 | Spondyloepimetaphyseal dysplasia congenita, Strudwick type | | 30 Cases |
| 171866 | Spondyloepimetaphyseal dysplasia, aggrecan type | | 3 Cases |
| 168448 | Spondyloepimetaphyseal dysplasia, Bieganski type | | 7 Cases |
| 168454 | Spondyloepimetaphyseal dysplasia, Geneviève type | | 6 Families |
| 99642 | Spondyloepimetaphyseal dysplasia, Handigodu type | | 234 Cases |
| 370015 | Spondyloepimetaphyseal dysplasia, Isidor type | | 2 Cases |
| 156728 | Spondyloepimetaphyseal dysplasia, matrilin-3 type | | 5 Cases |
| 93356 | Spondyloepimetaphyseal dysplasia, Missouri type | | 14 Cases |
| 93282 | Spondyloepimetaphyseal dysplasia, PAPSS2 type | | 17 Cases |
| 93352 | Spondyloepimetaphyseal dysplasia, Shohat type | | 5 Cases |
| 168451 | Spondyloepimetaphyseal dysplasia-abnormal dentition syndrome | | 2 Cases |
| 168443 | Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome | | 5 Cases |
| 93358 | Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome | | 27 Cases |
| 94068 | Spondyloepiphyseal dysplasia congenita | 1.0 *BP* * | |
| 163665 | Spondyloepiphyseal dysplasia tarda, Kohn type | | 3 Cases |
| 163654 | Spondyloepiphyseal dysplasia, Cantu type | | 4 Cases |
| 93283 | Spondyloepiphyseal dysplasia, Kimberley type | | 1 Family |
| 163668 | Spondyloepiphyseal dysplasia, MacDermot type | | 4 Cases |
| 263482 | Spondyloepiphyseal dysplasia, Maroteaux type | | 10 Cases |
| 163649 | Spondyloepiphyseal dysplasia, Nishimura type | | 4 Cases |
| 163662 | Spondyloepiphyseal dysplasia, Reardon type | | 1 Family |
| 459051 | Spondyloepiphyseal dysplasia, Stanescu type | | 7 Cases |
| 254 | Spondylometaphyseal dysplasia | 1.0 *BP* * | |
| 168555 | Spondylometaphyseal dysplasia, A4 type | | 3 Cases |
| 93315 | Spondylometaphyseal dysplasia, 'corner fracture' type | | 30 Cases |
| 370019 | Spondylometaphyseal dysplasia, Czarny-Ratajczak type | | 2 Cases |
| 168544 | Spondylometaphyseal dysplasia, Golden type | | 3 Cases |
| 93316 | Spondylometaphyseal dysplasia, Schmidt type | | 7 Cases |
| 93317 | Spondylometaphyseal dysplasia, Sedaghatian type | | 9 Cases |
| 168552 | Spondylometaphyseal dysplasia-bowed forearms-facial dysmorphism syndrome | | 2 Cases |
| 85167 | Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome | | 18 Cases |
| 1856 | Spondyloperipheral dysplasia-short ulna syndrome | | 10 Families |
| 29822 | Spontaneous periodic hypothermia | | 50 Cases |
| 247234 | Sporadic adult-onset ataxia of unknown etiology | 7.6 *P* * | |
| 204 | Sporadic Creutzfeldt-Jakob disease | 0.1 *P* * | |
| 204 | Sporadic Creutzfeldt-Jakob disease | 0.15 *I* | |
| 424996 | Squamous cell carcinoma of gallbladder and extrahepatic biliary tract | 0.04 *I* * | |
| 424975 | Squamous cell carcinoma of liver and intrahepatic biliary tract | 0.01 *I* * | |
| 424039 | Squamous cell carcinoma of pancreas | 0.03 *I* * | |
| 424019 | Squamous cell carcinoma of the anal canal | 0.73 *I* * | |
| 213767 | Squamous cell carcinoma of the cervix uteri | 4.28 *I* * | |

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|---|---|---|---|
| 423994 | Squamous cell carcinoma of the colon | 0.02 *I* * | |
| 213716 | Squamous cell carcinoma of the corpus uteri | 0.12 *I* * | |
| 99977 | Squamous cell carcinoma of the esophagus | 5.2 *I* | |
| 494547 | Squamous cell carcinoma of the hypopharynx | 1.27 *I* * | |
| 494550 | Squamous cell carcinoma of the larynx | 4.61 *I* * | |
| 502366 | Squamous cell carcinoma of the lip | 1.02 *I* | |
| 500464 | Squamous cell carcinoma of the nasal cavity and paranasal sinuses | 0.35 *I* | |
| 502363 | Squamous cell carcinoma of the oral cavity | 3.51 *I* * | |
| 500478 | Squamous cell carcinoma of the oropharynx | 3.12 *I* * | |
| 398058 | Squamous cell carcinoma of the penis | 0.57 *I* * | |
| 424002 | Squamous cell carcinoma of the rectum | 0.07 *I* * | |
| 423968 | Squamous cell carcinoma of the small intestine | 0.01 *I* * | |
| 418959 | Squamous cell carcinoma of the stomach | 0.13 *I* * | |
| 324737 | SRD5A3-CDG | | 7 Families |
| 370927 | SSR4-CDG | | 9 Cases |
| 83484 | St. Louis encephalitis | 0.38 *I* * | |
| 502434 | STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome | | 17 Cases |
| 140917 | Stapes ankylosis with broad thumbs and toes | | 6 Families |
| 827 | Stargardt disease | 10.0 *P* * | |
| 438159 | STAT3-related early-onset multisystem autoimmune disease | | 19 Cases |
| 438117 | Steel syndrome | | 40 Cases |
| 273 | Steinert myotonic dystrophy | 12.5 *P* | |
| 210115 | Sterile multifocal osteomyelitis with periostitis and pustulosis | | 17 Cases |
| 2017 | Sternal cleft | 2.0 *BP* * | |
| 3196 | Steroid dehydrogenase deficiency-dental anomalies syndrome | | 1 Family |
| 36426 | Stevens-Johnson syndrome | 0.36 *I* * | |
| 95455 | Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum | 0.19 *I* | |
| 828 | Stickler syndrome | 12.2 *BP* | |
| 2833 | Stiff skin syndrome | | 54 Cases |
| 3199 | Stimmler syndrome | | 2 Cases |
| 425120 | STING-associated vasculopathy with onset in infancy | | 9 Cases |
| 3204 | Stormorken-Sjaastad-Langslet syndrome | | 17 Cases |
| 137599 | Stromal keratitis | 16.0 *P* * | |
| 506307 | Stromme syndrome | | 11 Cases |
| 370921 | STT3A-CDG | | 2 Cases |
| 370924 | STT3B-CDG | | 1 Case |
| 3205 | Sturge-Weber syndrome | 3.5 *BP* * | |
| 3206 | Stüve-Wiedemann syndrome | | 56 Cases |
| 3191 | Subaortic stenosis-short stature syndrome | | 1 Family |
| 48377 | Subcorneal pustular dermatosis | | 200 Cases |
| 98959 | Subepithelial mucinous corneal dystrophy | | 1 Family |
| 22 | Succinic semialdehyde dehydrogenase deficiency | | 450 Cases |
| 832 | Succinyl-CoA:3-ketoacid CoA transferase deficiency | | 33 Cases |
| 168593 | Sudden infant death-dysgenesis of the testes syndrome | | 21 Cases |
| 498602 | Sugarman brachydactyly | | 1 Family |
| 3210 | Summitt syndrome | | 3 Cases |
| 57145 | SUNCT syndrome | 6.7 *P* * | |
| 455 | Superficial epidermolytic ichthyosis | | 20 Cases |
| 46485 | Superficial pemphigus | 1.2 *P* * | |
| 247245 | Superficial siderosis | | 300 Cases |
| 141096 | Supernumerary nostril | | 32 Cases |
| 466695 | Supratip dysplasia | | 5 Cases |
| 3193 | Supravalvular aortic stenosis | 13.3 *P* * | |
| 3193 | Supravalvular aortic stenosis | 4.0 *BP* * | |
| 391351 | SURF1-related Charcot-Marie-Tooth disease type 4 | | 3 Cases |
| 838 | Susac syndrome | | 304 Cases |
| 331226 | Susceptibility to infection due to TYK2 deficiency | | 8 Cases |
| 3243 | Sweet syndrome | | 100 Cases |
| 1570 | Symbrachydactyly of hands and feet | | 2 Cases |
| 1314 | Symmetrical thalamic calcifications | | 30 Cases |
| 79098 | Sympathetic ophthalmia | 0.6 *P* * | |
| 3246 | Symphalangism with multiple anomalies of hands and feet | | 6 Cases |
| 93402 | Syndactyly type 1 | 25.0 *BP* * | |
| 93405 | Syndactyly type 4 | | 4 Cases |
| 93406 | Syndactyly type 5 | | 10 Cases |
| 357332 | Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome | | 26 Cases |
| 294026 | Syndactyly-nystagmus syndrome due to 2q31.1 microduplication | | 2 Cases |
| 3259 | Syndactyly-polydactyly-ear lobe syndrome | | 10 Cases |

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|---|---|---|---|
| 140952 | Syndactyly-telecanthus-anogenital and renal malformations syndrome | | 6 Cases |
| 84064 | Syndromic diarrhea | | 44 Cases |
| 178364 | Syndromic microphthalmia type 5 | | 20 Cases |
| 228426 | Syndromic multisystem autoimmune disease due to Itch deficiency | | 10 Cases |
| 98606 | Syndromic orbital border hypoplasia | | 2 Families |
| 281090 | Syndromic recessive X-linked ichthyosis | 1.3 *P* * | |
| 457223 | Syndromic sensorineural deafness due to combined oxidative phosphorylation defect | | 2 Cases |
| 85274 | Syndromic X-linked intellectual disability 7 | | 10 Cases |
| 85279 | Syndromic X-linked intellectual disability due to JARID1C mutation | | 10 Families |
| 840 | Syringocystadenoma papilliferum | | 300 Cases |
| 3280 | Syringomyelia | 8.4 *P* * | |
| 188 | Systemic capillary leak syndrome | | 150 Cases |
| 2467 | Systemic mastocytosis | 3.75 *P* | |
| 2467 | Systemic mastocytosis | 0.9 *I* * | |
| 158 | Systemic primary carnitine deficiency | 3.2 *BP* * | |
| 90291 | Systemic sclerosis | 15.4 *P* * | |
| 85414 | Systemic-onset juvenile idiopathic arthritis | 5.0 *P* * | |
| 169157 | T-B+ severe combined immunodeficiency due to CD45 deficiency | | 3 Cases |
| 324294 | T-cell immunodeficiency with epidermodysplasia verruciformis | | 2 Cases |
| 86872 | T-cell large granular lymphocyte leukemia | 0.4 *I* * | |
| 171918 | T-cell non-Hodgkin lymphoma | 0.99 *I* * | |
| 457077 | TAFRO syndrome | | 28 Cases |
| 3287 | Takayasu arteritis | 1.34 *P* * | |
| 3287 | Takayasu arteritis | 0.084 *I* * | |
| 404443 | Tall stature-intellectual disability-facial dysmorphism syndrome | | 17 Cases |
| 500095 | Tall stature-intellectual disability-renal anomalies syndrome | | 4 Cases |
| 329191 | Tall stature-scoliosis-macrodactyly of the great toes syndrome | | 2 Families |
| 50809 | Talo-patello-scaphoid osteolysis | | 2 Cases |
| 31150 | Tangier disease | | 100 Cases |
| 2886 | TARP syndrome | | 6 Families |
| 1412 | Tarsal-carpal coalition syndrome | | 10 Families |
| 2731 | Taurodontia-absent teeth-sparse hair syndrome | | 15 Cases |
| 845 | Tay-Sachs disease | 0.28 *BP* | |
| 488632 | TBCK-related intellectual disability syndrome | | 25 Cases |
| 397959 | TCR-alpha-beta-positive T-cell deficiency | | 2 Cases |
| 3291 | Teebi-Shaltout syndrome | | 5 Cases |
| 3293 | Telecanthus-hypertelorism-strabismus-pes cavus syndrome | | 2 Cases |
| 488642 | TELO2-related intellectual disability-neurodevelopmental disorder | | 6 Cases |
| 352737 | Temperature-sensitive oculocutaneous albinism type 1 | | 10 Cases |
| 284227 | TEMPI syndrome | | 10 Cases |
| 254516 | Temple syndrome | | 53 Cases |
| 96184 | Temple syndrome due to maternal uniparental disomy of chromosome 14 | | 64 Cases |
| 254531 | Temple syndrome due to paternal 14q32.2 hypomethylation | | 12 Cases |
| 254525 | Temple syndrome due to paternal 14q32.2 microdeletion | | 9 Cases |
| 420561 | Temple-Baraitser syndrome | | 7 Cases |
| 363417 | Temtamy preaxial brachydactyly syndrome | | 18 Cases |
| 1777 | Temtamy syndrome | | 7 Families |
| 141258 | Tessier number 4 facial cleft | | 2 Cases |
| 842 | Testicular seminomatous germ cell tumor | 1.71 *I* * | |
| 3299 | Tetanus | 0.026 *I* * | |
| 3301 | Tetraamelia-multiple malformations syndrome | | 5 Families |
| 3303 | Tetralogy of Fallot | 34.0 *BP* | |
| 3310 | Tetrasomy 9p | | 70 Cases |
| 884 | Tetrasomy 12p | 4.0 *BP* * | |
| 96055 | Tetrasomy 21 | | 13 Cases |
| 9 | Tetrasomy X | | 50 Cases |
| 1780 | Thakker-Donnai syndrome | | 2 Cases |
| 3312 | Thalidomide embryopathy | 0.77 *P* | |
| 2655 | Thanatophoric dysplasia | 3.5 *BP* * | |
| 199348 | Thiamine-responsive encephalopathy | | 2 Cases |
| 49827 | Thiamine-responsive megaloblastic anemia syndrome | | 80 Cases |
| 2405 | Thickened earlobes-conductive deafness syndrome | | 2 Families |
| 98960 | Thiel-Behnke corneal dystrophy | | 173 Cases |
| 1506 | Thin ribs-tubular bones-dysmorphism syndrome | | 2 Cases |
| 363444 | THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome | | 4 Cases |
| 3316 | Thomas syndrome | | 6 Cases |

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| 614 | Thomsen and Becker disease | 1.0 *P* | |
| 1861 | Thoracic dysplasia-hydrocephalus syndrome | | 2 Cases |
| 3317 | Thoracolaryngopelvic dysplasia | | 10 Cases |
| 1803 | Thoracomelic dysplasia | | 2 Cases |
| 329319 | Thrombocythemia with distal limb defects | | 3 Families |
| 67044 | Thrombocytopenia with congenital dyserythropoietic anemia | | 3 Families |
| 3320 | Thrombocytopenia-absent radius syndrome | 0.5 *BP* * | |
| 3323 | Thrombocytopenia-Robin sequence syndrome | | 2 Cases |
| 436169 | Thrombomodulin-related bleeding disorder | | 15 Cases |
| 54057 | Thrombotic thrombocytopenic purpura | 25.5 *P* * | |
| 2251 | Thumb deformity-alopecia-pigmentation anomaly syndrome | | 1 Family |
| 1078 | Thumb stiffness-brachydactyly-intellectual disability syndrome | | 6 Cases |
| 3398 | Thymic epithelial neoplasm | 0.17 *I* * | |
| 3326 | Thymic-renal-anal-lung dysplasia | | 3 Cases |
| 99867 | Thymoma | 0.14 *I* * | |
| 3327 | Thyrocerebrorenal syndrome | | 2 Cases |
| 100088 | Thyroid carcinoma | 12.7 *P* | |
| 100088 | Thyroid carcinoma | 3.1 *I* | |
| 95712 | Thyroid ectopia | 14.3 *P* * | |
| 95719 | Thyroid hemiagenesis | 25.0 *P* | |
| 95720 | Thyroid hypoplasia | 3.5 *P* | |
| 100087 | Thyroid tumor | 3.2 *I* | |
| 3329 | Tibial aplasia-ectrodactyly syndrome | 0.1 *P* * | |
| 93322 | Tibial hemimelia | 0.1 *BP* * | |
| 609 | Tibial muscular dystrophy | 6.0 *P* * | |
| 42665 | Tietz syndrome | | 2 Families |
| 65283 | Timothy syndrome | | 20 Cases |
| 314667 | TMEM165-CDG | | 6 Cases |
| 466703 | TMEM199-CDG | | 7 Cases |
| 3460 | Torg-Winchester syndrome | | 12 Cases |
| 3338 | Toriello-Carey syndrome | | 59 Cases |
| 3339 | Toriello-Lacassie-Droste syndrome | | 19 Cases |
| 3341 | Torticollis-keloids-cryptorchidism-renal dysplasia syndrome | | 7 Cases |
| 227972 | Toxic oil syndrome | | 20000 Cases |
| 3346 | Tracheal agenesis | 2.0 *BP* * | |
| 3348 | Tracheobronchopathia osteochondroplastica | | 400 Cases |
| 101028 | Transaldolase deficiency | | 23 Cases |
| 859 | Transcobalamin deficiency | | 40 Cases |
| 79411 | Transient bullous dermolysis of the newborn | | 30 Cases |
| 300293 | Transient infantile hypertriglyceridemia and hepatosteatosis | | 11 Cases |
| 99886 | Transient neonatal diabetes mellitus | 0.3 *BP* * | |
| 329942 | Transient neonatal multiple acyl-CoA dehydrogenase deficiency | | 1 Case |
| 488618 | Transketolase deficiency | | 5 Cases |
| 216675 | Transposition of the great arteries | 31.7 *BP* * | |
| 861 | Treacher-Collins syndrome | 2.0 *BP* * | |
| 447896 | Tremor-ataxia-central hypomyelination syndrome | | 7 Cases |
| 3350 | Tremor-nystagmus-duodenal ulcer syndrome | | 17 Cases |
| 863 | Trichinellosis | 0.06 *I* * | |
| 3352 | Tricho-dento-osseous syndrome | | 30 Cases |
| 1264 | Tricho-retino-dento-digital syndrome | | 9 Cases |
| 3351 | Trichodental syndrome | | 5 Families |
| 3353 | Trichodermodysplasia-dental alterations syndrome | | 3 Cases |
| 79129 | Trichodysplasia-amelogenesis imperfecta syndrome | | 1 Family |
| 3361 | Trichodysplasia-xeroderma syndrome | | 1 Family |
| 3363 | Trichomegaly-retina pigmentary degeneration-dwarfism syndrome | | 14 Cases |
| 3355 | Trichoodontoonychial dysplasia | | 4 Cases |
| 77258 | Trichorhinophalangeal syndrome type 1 and 3 | | 100 Cases |
| 502 | Trichorhinophalangeal syndrome type 2 | | 100 Cases |
| 33364 | Trichothiodystrophy | 0.12 *BP* * | |
| 1209 | Tricuspid atresia | 4.2 *BP* * | |
| 3368 | Trigonocephaly-bifid nose-acral anomalies syndrome | | 2 Cases |
| 3365 | Trigonocephaly-broad thumbs syndrome | | 2 Cases |
| 3369 | Trigonocephaly-short stature-developmental delay syndrome | | 3 Cases |
| 868 | Triose phosphate-isomerase deficiency | | 50 Cases |
| 2950 | Triphalangeal thumb-polysyndactyly syndrome | | 15 Families |
| 2947 | Triphalangeal thumbs-brachyectrodactyly syndrome | | 4 Families |
| 869 | Triple A syndrome | | 100 Cases |
| 3376 | Triploidy | 12.6 *BP* * | |
| 261344 | Trisomy 1q | | 18 Cases |

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|---|---|---|---|
| 1738 | Trisomy 4p | | 85 Cases |
| 1742 | Trisomy 5p | | 40 Cases |
| 1752 | Trisomy 8q | | 30 Cases |
| 236 | Trisomy 9p | | 150 Cases |
| 171929 | Trisomy 10p | | 50 Cases |
| 1699 | Trisomy 12p | 2.0 *BP* | |
| 3378 | Trisomy 13 | 3.7 *BP* * | |
| 3380 | Trisomy 18 | 16.7 *BP* | |
| 1715 | Trisomy 18p | | 25 Cases |
| 3375 | Trisomy X | 42.5 *P* * | |
| 88629 | Tritanopia | 4.8 *P* * | |
| 3384 | Truncus arteriosus | 4.3 *BP* | |
| 3389 | Tuberculosis | 20.0 *P* * | |
| 3389 | Tuberculosis | 139.0 *I* | |
| 805 | Tuberous sclerosis complex | 12.0 *P* * | |
| 805 | Tuberous sclerosis complex | 10.0 *BP* * | |
| 73224 | Tubular renal disease-cardiomyopathy syndrome | | 2 Cases |
| 467166 | Tubulinopathy-associated dysgyria | | 7 Cases |
| 1063 | Tufted angioma | | 200 Cases |
| 3392 | Tularemia | 2.0 *P* * | |
| 3392 | Tularemia | 0.14 *I* * | |
| 32960 | Tumor necrosis factor receptor 1 associated periodic syndrome | 0.1 *P* * | |
| 182130 | Tumor of endocrine glands | 64.0 *P* * | |
| 182130 | Tumor of endocrine glands | 3.75 *I* * | |
| 363472 | Tumor of testis and paratestis | 3.15 *I* * | |
| 881 | Turner syndrome | 5.5 *BP* * | |
| 99745 | Typhoid | 3.0 *I* * | |
| 882 | Tyrosinemia type 1 | 0.9 *BP* | |
| 28378 | Tyrosinemia type 2 | | 150 Cases |
| 69723 | Tyrosinemia type 3 | | 20 Cases |
| 3403 | Uhl anomaly | 1.0 *BP* | |
| 3404 | Ulbright-Hodes syndrome | | 3 Cases |
| 2249 | Ulna hypoplasia-intellectual disability syndrome | | 2 Cases |
| 1837 | Ulna metaphyseal dysplasia syndrome | | 3 Cases |
| 1122 | Ulnar hypoplasia-split foot syndrome | | 1 Family |
| 3138 | Ulnar-mammary syndrome | | 117 Cases |
| 52056 | Ulnar/fibula ray defect-brachydactyly syndrome | | 1 Family |
| 3405 | Umbilical cord ulceration-intestinal atresia syndrome | | 55 Cases |
| 167714 | Unclassified acute myeloid leukemia | 0.49 *I* * | |
| 418951 | Undifferentiated carcinoma of esophagus | 0.07 *I* * | |
| 424970 | Undifferentiated carcinoma of liver and intrahepatic biliary tract | 0.02 *I* * | |
| 423786 | Undifferentiated carcinoma of stomach | 0.17 *I* * | |
| 2023 | Undifferentiated pleomorphic sarcoma | 0.9 *I* * | |
| 97363 | Unilateral multicystic dysplastic kidney | 23.2 *BP* | |
| 1464 | Univentricular heart | 7.5 *BP* | |
| 99069 | Univentricular heart with single atrio-ventricular valve | | 2 Cases |
| 3408 | Upington disease | | 1 Family |
| 2489 | Upper limb defect-eye and ear abnormalities syndrome | | 2 Cases |
| 2497 | Upper limb mesomelic dysplasia | | 4 Cases |
| 3409 | Urban-Rogers-Meyer syndrome | | 3 Cases |
| 94059 | Uremic pruritus | 35.0 *P* * | |
| 210128 | Urocanic aciduria | | 4 Cases |
| 481665 | USP18 deficiency | | 5 Cases |
| 178338 | UV-sensitive syndrome | | 7 Cases |
| 1473 | Uveal coloboma-cleft lip and palate-intellectual disability | | 12 Cases |
| 39044 | Uveal melanoma | 0.5 *I* * | |
| 98715 | Uveitis | 38.0 *P* * | |
| 98715 | Uveitis | 17.0 *I* * | |
| 3412 | VACTERL with hydrocephalus | | 10 Families |
| 887 | VACTERL/VATER association | 6.25 *BP* * | |
| 88635 | Vacuolar myopathy with sarcoplasmic reticulum protein aggregates | | 4 Cases |
| 3417 | Van den Bosch syndrome | | 1 Family |
| 2460 | Van den Ende-Gupta syndrome | | 29 Cases |
| 3419 | Van Regemorter-Pierquin-Vamos syndrome | | 3 Cases |
| 314652 | Variant ABeta2M amyloidosis | | 5 Cases |
| 52759 | Vasculitis | 6.3 *P* * | |
| 404553 | Vasculitis due to ADA2 deficiency | | 48 Cases |
| 3424 | Velo-facial-skeletal syndrome | | 2 Cases |
| 443988 | Ventriculomegaly-cystic kidney disease | | 11 Cases |
| 3429 | Verloove Vanhorick-Brubakk syndrome | | 2 Cases |
| 70476 | Vernal keratoconjunctivitis | 32.0 *P* * | |
| 493342 | Vibratory urticaria | | 37 Cases |
| 1493 | Vici syndrome | | 50 Cases |

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|---|---|---|---|
| 228379 | Virus-associated trichodysplasia spinulosa | | 7 Cases |
| 73246 | Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome | | 2 Cases |
| 28 | Vitamin B12-responsive methylmalonic acidemia | | 192 Cases |
| 79310 | Vitamin B12-responsive methylmalonic acidemia type cblA | | 60 Cases |
| 79312 | Vitamin B12-unresponsive methylmalonic acidemia type mut- | | 450 Cases |
| 3439 | Von Voss-Cherstvoy syndrome | | 10 Cases |
| 903 | Von Willebrand disease | 12.5 *P* | |
| 466934 | VPS11-related autosomal recessive hypomyelinating leukodystrophy | | 13 Cases |
| 137583 | Vulvar intraepithelial neoplasia | 20.0 *P* * | |
| 83453 | Vulvovaginal gingival syndrome | | 380 Cases |
| 2804 | W syndrome | | 6 Cases |
| 3440 | Waardenburg syndrome | 0.37 *BP* * | |
| 897 | Waardenburg-Shah syndrome | | 100 Cases |
| 466943 | WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome | | 22 Cases |
| 898 | Wagner disease | | 100 Cases |
| 893 | WAGR syndrome | 0.2 *BP* | |
| 33226 | Waldenström macroglobulinemia | 0.81 *I* * | |
| 899 | Walker-Warburg syndrome | 1.65 *BP* * | |
| 280558 | Warsaw breakage syndrome | | 4 Cases |
| 3447 | Weaver syndrome | | 48 Cases |
| 3448 | Weaver-Williams syndrome | | 2 Cases |
| 3449 | Weill-Marchesani syndrome | 1.0 *P* | |
| 3344 | Weismann-Netter syndrome | | 100 Cases |
| 99971 | Well-differentiated liposarcoma | 0.51 *I* * | |
| 901 | Wells syndrome | | 200 Cases |
| 902 | Werner syndrome | 0.5 *P* * | |
| 3451 | West syndrome | 6.0 *P* * | |
| 3451 | West syndrome | 3.7 *BP* | |
| 83476 | West-Nile encephalitis | 0.036 *I* * | |
| 51636 | WHIM syndrome | | 65 Cases |
| 2475 | White forelock with malformations | | 2 Cases |
| 3207 | White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome | | 4 Cases |
| 370131 | White platelet syndrome | | 1 Family |
| 1489 | Whooping cough | 9.32 *I* * | |
| 3455 | Wiedemann-Rautenstrauch syndrome | | 54 Cases |
| 319182 | Wiedemann-Steiner syndrome | | 18 Cases |
| 85446 | Wild type ABeta2M amyloidosis | 4.5 *P* * | |
| 330001 | Wild type ATTR amyloidosis | 30.0 *P* * | |
| 904 | Williams syndrome | 10.8 *BP* | |
| 905 | Wilson disease | 3.3 *P* | |
| 905 | Wilson disease | 2.2 *BP* | |
| 3459 | Wilson-Turner syndrome | | 28 Cases |
| 906 | Wiskott-Aldrich syndrome | 0.1 *P* * | |
| 1667 | Wolcott-Rallison syndrome | | 60 Cases |
| 280 | Wolf-Hirschhorn syndrome | 2.0 *BP* * | |
| 3463 | Wolfram syndrome | 0.13 *P* | |
| 3464 | Woodhouse-Sakati syndrome | | 30 Cases |
| 1409 | Woolly hair-hypotrichosis-everted lower lip-outstanding ears syndrome | | 1 Family |
| 420686 | Woolly hair-palmoplantar keratoderma syndrome | | 8 Cases |
| 166277 | Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia | | 3 Cases |
| 3465 | Worster-Drought syndrome | 3.7 *P* * | |
| 178475 | Wound botulism | 0.1 *I* * | |
| 2834 | Wrinkly skin syndrome | | 30 Cases |
| 3466 | WT limb-blood syndrome | | 3 Families |
| 53719 | Wyburn-Mason syndrome | | 90 Cases |
| 448372 | X-linked acrogigantism due to Xq26 microduplication | | 22 Cases |
| 43 | X-linked adrenoleukodystrophy | 5.0 *BP* | |
| 47 | X-linked agammaglobulinemia | 0.22 *P* | |
| 391327 | X-linked calvarial hyperostosis | | 1 Family |
| 329235 | X-linked central congenital hypothyroidism with late-onset testicular enlargement | | 27 Cases |
| 596 | X-linked centronuclear myopathy | 0.2 *P* * | |
| 163961 | X-linked cerebral-cerebellar-coloboma syndrome | | 3 Cases |
| 64747 | X-linked Charcot-Marie-Tooth disease | 1.6 *P* * | |
| 101076 | X-linked Charcot-Marie-Tooth disease type 2 | | 5 Cases |
| 101077 | X-linked Charcot-Marie-Tooth disease type 3 | | 4 Families |
| 101078 | X-linked Charcot-Marie-Tooth disease type 4 | | 7 Cases |
| 99014 | X-linked Charcot-Marie-Tooth disease type 5 | | 9 Cases |
| 352675 | X-linked Charcot-Marie-Tooth disease type 6 | | 8 Cases |

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|---|---|---|---|
| 431140 | X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome | | 1 Family |
| 1497 | X-linked complicated corpus callosum dysgenesis | | 11 Cases |
| 90001 | X-linked cone dysfunction syndrome with myopia | | 10 Families |
| 1661 | X-linked corneal dermoid | | 6 Cases |
| 52503 | X-linked creatine transporter deficiency | | 150 Cases |
| 139557 | X-linked distal spinal muscular atrophy type 3 | | 2 Families |
| 35173 | X-linked dominant chondrodysplasia punctata | 0.25 *BP* * | |
| 163966 | X-linked dominant chondrodysplasia, Chassaing-Lacombe type | | 10 Cases |
| 363727 | X-linked dyserythropoietic anemia with abnormal platelets and neutropenia | | 1 Family |
| 75497 | X-linked Ehlers-Danlos syndrome | | 2 Families |
| 98863 | X-linked Emery-Dreifuss muscular dystrophy | 1.0 *P* | |
| 98863 | X-linked Emery-Dreifuss muscular dystrophy | 1.0 *BP* | |
| 293621 | X-linked endothelial corneal dystrophy | | 35 Cases |
| 443197 | X-linked erythropoietic protoporphyria | | 50 Cases |
| 500188 | X-linked external auditory canal atresia-dilated internal auditory canal-facial dysmorphism syndrome | | 4 Cases |
| 480880 | X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability | | 17 Cases |
| 139583 | X-linked hereditary sensory and autonomic neuropathy with deafness | | 5 Families |
| 181 | X-linked hypohidrotic ectodermal dysplasia | 0.75 *BP* * | |
| 89936 | X-linked hypophosphatemia | 0.21 *P* * | |
| 317476 | X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia | | 7 Cases |
| 2571 | X-linked immunoneurologic disorder | | 5 Cases |
| 364028 | X-linked intellectual disability due to GRIA3 mutations | | 14 Cases |
| 67045 | X-linked intellectual disability with isolated growth hormone deficiency | | 2 Families |
| 85273 | X-linked intellectual disability, Abidi type | | 8 Cases |
| 85276 | X-linked intellectual disability, Armfield type | | 6 Cases |
| 3056 | X-linked intellectual disability, Brooks type | | 9 Cases |
| 85293 | X-linked intellectual disability, Cabezas type | | 24 Families |
| 85277 | X-linked intellectual disability, Cantagrel type | | 30 Cases |
| 163971 | X-linked intellectual disability, Cilliers type | | 4 Cases |
| 93947 | X-linked intellectual disability, Golabi-Ito-Hall type | | 3 Cases |
| 93952 | X-linked intellectual disability, Hedera type | | 9 Cases |
| 85283 | X-linked intellectual disability, Miles-Carpenter type | | 4 Cases |
| 163937 | X-linked intellectual disability, Najm type | | 35 Families |
| 163956 | X-linked intellectual disability, Nascimento type | | 8 Cases |
| 85322 | X-linked intellectual disability, Pai type | | 1 Family |
| 85285 | X-linked intellectual disability, Schimke type | | 4 Cases |
| 85323 | X-linked intellectual disability, Seemanova type | | 4 Cases |
| 85286 | X-linked intellectual disability, Shashi type | | 9 Cases |
| 85324 | X-linked intellectual disability, Shrimpton type | | 3 Cases |
| 85287 | X-linked intellectual disability, Siderius type | | 2 Families |
| 3063 | X-linked intellectual disability, Snyder type | | 21 Cases |
| 85325 | X-linked intellectual disability, Stevenson type | | 4 Cases |
| 85288 | X-linked intellectual disability, Stocco Dos Santos type | | 1 Family |
| 85326 | X-linked intellectual disability, Stoll type | | 4 Cases |
| 163976 | X-linked intellectual disability, Van Esch type | | 7 Cases |
| 85290 | X-linked intellectual disability, Wilson type | | 3 Cases |
| 85327 | X-linked intellectual disability-acromegaly-hyperactivity syndrome | | 2 Cases |
| 85338 | X-linked intellectual disability-ataxia-apraxia syndrome | | 9 Cases |
| 324410 | X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome | | 2 Cases |
| 137831 | X-linked intellectual disability-cerebellar hypoplasia syndrome | | 14 Families |
| 459070 | X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome | | 2 Cases |
| 85330 | X-linked intellectual disability-corpus callosum agenesis-spastic quadriparesis syndrome | | 4 Cases |
| 163979 | X-linked intellectual disability-craniofacioskeletal syndrome | | 9 Cases |

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|---|---|---|---|
| 85280 | X-linked intellectual disability-cubitus valgus-dysmorphism syndrome | | 5 Cases |
| 1568 | X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome | | 10 Cases |
| 2958 | X-linked intellectual disability-dysmorphism-cerebral atrophy syndrome | | 8 Cases |
| 85319 | X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome | | 2 Cases |
| 480907 | X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome | | 14 Cases |
| 85317 | X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome | | 3 Cases |
| 3055 | X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome | | 4 Cases |
| 85329 | X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome | | 10 Cases |
| 457260 | X-linked intellectual disability-hypotonia-movement disorder syndrome | | 38 Cases |
| 423479 | X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome | | 2 Cases |
| 85320 | X-linked intellectual disability-macrocephaly-macroorchidism syndrome | | 12 Cases |
| 2898 | X-linked intellectual disability-plagiocephaly syndrome | | 2 Cases |
| 3077 | X-linked intellectual disability-psychosis-macroorchidism syndrome | | 6 Cases |
| 3052 | X-linked intellectual disability-seizures-psoriasis syndrome | | 4 Cases |
| 457240 | X-linked intellectual disability-short stature-overweight syndrome | | 20 Cases |
| 482606 | X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome | | 15 Cases |
| 79447 | X-linked lethal multiple pterygium syndrome | | 6 Families |
| 452 | X-linked lissencephaly with abnormal genitalia | | 30 Families |
| 2442 | X-linked lymphoproliferative disease | 0.05 *P* * | |
| 1131 | X-linked mandibulofacial dysostosis | | 7 Cases |
| 319605 | X-linked mendelian susceptibility to mycobacterial diseases | | 13 Cases |
| 319623 | X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency | | 7 Cases |
| 319612 | X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency | | 6 Cases |
| 435938 | X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome | | 3 Cases |
| 25980 | X-linked myopathy with excessive autophagy | | 18 Families |
| 178461 | X-linked myopathy with postural muscle atrophy | | 7 Families |
| 456328 | X-linked myotubular myopathy-abnormal genitalia syndrome | | 4 Cases |
| 85334 | X-linked neurodegenerative syndrome, Bertini type | | 7 Cases |
| 85336 | X-linked neurodegenerative syndrome, Hamel type | | 11 Cases |
| 314978 | X-linked non progressive cerebellar ataxia | | 3 Families |
| 391330 | X-linked osteoporosis with fractures | | 8 Families |
| 363654 | X-linked parkinsonism-spasticity syndrome | | 5 Cases |
| 83648 | X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome | | 1 Family |
| 54 | X-linked recessive ocular albinism | 0.58 *BP* * | |
| 85453 | X-linked reticulate pigmentary disorder | | 6 Families |
| 1852 | X-linked retinal dysplasia | | 8 Cases |
| 792 | X-linked retinoschisis | 5.0 *P* | |
| 431272 | X-linked scapuloperoneal muscular dystrophy | | 22 Cases |
| 86788 | X-linked severe congenital neutropenia | | 45 Cases |
| 75563 | X-linked sideroblastic anemia | | 200 Cases |
| 2802 | X-linked sideroblastic anemia and spinocerebellar ataxia | | 5 Families |
| 1436 | X-linked skeletal dysplasia-intellectual disability syndrome | | 4 Cases |
| 100997 | X-linked spastic paraplegia type 16 | | 1 Family |
| 171607 | X-linked spastic paraplegia type 34 | | 24 Cases |
| 3175 | X-linked spasticity-intellectual disability-epilepsy syndrome | | 6 Cases |
| 85297 | X-linked spinocerebellar ataxia type 3 | | 5 Cases |
| 85292 | X-linked spinocerebellar ataxia type 4 | | 1 Family |
| 910 | Xeroderma pigmentosum | 0.23 *BP* * | |
| 90342 | Xeroderma pigmentosum variant | | 50 Cases |
| 220295 | Xeroderma pigmentosum-Cockayne syndrome complex | | 30 Cases |
| 3469 | XK aprosencephaly syndrome | | 10 Cases |
| 261476 | Xp21 microdeletion syndrome | | 100 Cases |
| 314389 | Xq12-q13.3 duplication syndrome | | 3 Cases |

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| 521258 | Xq25 microduplication syndrome | | 28 Cases |
| 261483 | Xq27.3q28 duplication syndrome | | 8 Cases |
| 1770 | XY type gonadal dysgenesis-associated anomalies syndrome | | 2 Cases |
| 370930 | XYLT1-CDG | | 2 Cases |
| 662 | Yellow nail syndrome | | 400 Cases |
| 314485 | Young adult-onset distal hereditary motor neuropathy | | 3 Cases |
| 2828 | Young-onset Parkinson disease | 15.0 *P* * | |
| 3472 | Yunis-Varon syndrome | | 25 Cases |
| 97240 | Zebra body myopathy | | 10 Cases |
| 217017 | Zechi-Ceide syndrome | | 3 Cases |
| 50812 | Zellweger-like syndrome without peroxisomal anomalies | | 2 Cases |
| 3473 | Zimmermann-Laband syndrome | | 52 Cases |
| 3253 | Zlotogora-Ogur syndrome | | 50 Cases |
| 913 | Zollinger-Ellison syndrome | 0.125 *I* | |

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