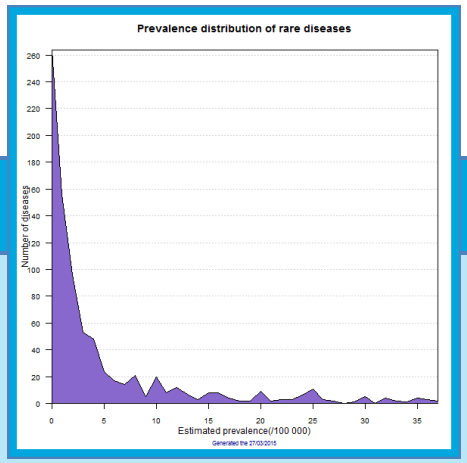

Orphanet Report Series
Rare Diseases collection

Number 1 | November 2023

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.com

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Co-funded by the Health Programme of the European Union

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 favored (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.

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

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

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Please note that this is just a selection of Orphanet's rare disease epidemiological data. Currently 6089 rare diseases are annotated with prevalence or incidence information in the Orphanet database. To access the complete data sets visit Orphadata (www.orphadata.com).

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

ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
300305	11p15.4 microduplication syndrome	Disorder		1 *Family*
444002	11q22.2q22.3 microdeletion syndrome	Disorder		5 *Cases*
313884	12p12.1 microdeletion syndrome	Subtype of disorder		11 *Cases*
94063	12q14 microdeletion syndrome	Disorder		22 *Cases*
289513	12q15q21.1 microdeletion syndrome	Disorder		6 *Cases*
412035	13q12.3 microdeletion syndrome	Disorder		3 *Cases*
261120	14q11.2 microdeletion syndrome	Disorder		3 *Cases*
261229	14q11.2 microduplication syndrome	Disorder		7 *Cases*
264200	14q22q23 microdeletion syndrome	Disorder		5 *Cases*
401935	14q24.1q24.3 microdeletion syndrome	Disorder		3 *Cases*
488280	14q32 duplication syndrome	Disorder		33 *Cases*
314585	15q overgrowth syndrome	Disorder		12 *Cases*
261183	15q11.2 microdeletion syndrome	Disorder		200 *Cases*
238446	15q11q13 microduplication syndrome	Disorder		30 *Cases*
199318	15q13.3 microdeletion syndrome	Disorder		246 *Cases*
94065	15q24 microdeletion syndrome	Subtype of disorder		30 *Cases*
261211	16p11.2p12.2 microdeletion syndrome	Disorder		8 *Cases*
261204	16p11.2p12.2 microduplication syndrome	Disorder		7 *Cases*
485405	16p12.1p12.3 triplication syndrome	Disorder		3 *Cases*
261236	16p13.11 microdeletion syndrome	Disorder	7.0 *BP*	
261243	16p13.11 microduplication syndrome	Disorder		162 *Cases*
96078	16p13.3 microduplication syndrome	Disorder		27 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
352629	16q24.1 microdeletion syndrome	Disorder		42 *Cases*
261250	16q24.3 microdeletion syndrome	Disorder		27 *Cases*
1713	17p11.2 microduplication syndrome	Disorder		170 *Cases*
217385	17p13.3 microduplication syndrome	Disorder		50 *Cases*
97685	17q11 microdeletion syndrome	Subtype of disorder		170 *Cases*
139474	17q11.2 microduplication syndrome	Disorder		7 *Cases*
261265	17q12 microdeletion syndrome	Disorder		103 *Cases*
261272	17q12 microduplication syndrome	Disorder		118 *Cases*
363958	17q21.31 microdeletion syndrome	Subtype of disorder	1.82 *P**	
261279	17q23.1q23.2 microdeletion syndrome	Disorder		7 *Cases*
529962	17q24.2 microdeletion syndrome	Disorder		19 *Cases*
254346	19p13.12 microdeletion syndrome	Disorder		6 *Cases*
357001	19p13.13 microdeletion syndrome	Disorder		7 *Cases*
447980	19p13.3 microduplication syndrome	Disorder		6 *Cases*
217346	19q13.11 microdeletion syndrome	Disorder		12 *Cases*
293948	1p21.3 microdeletion syndrome	Disorder		9 *Cases*
401986	1p31p32 microdeletion syndrome	Disorder		5 *Cases*
456298	1p35.2 microdeletion syndrome	Disorder		2 *Cases*
250994	1q21.1 microduplication syndrome	Disorder		46 *Cases*
238769	1q44 microdeletion syndrome	Disorder		100 *Cases*
79154	2-aminoadipic 2-oxoadipic aciduria	Disorder		20 *Cases*
79157	2-methylbutyryl-CoA dehydrogenase deficiency	Disorder		30 *Cases*
261295	20p12.3 microdeletion syndrome	Disorder		3 *Cases*
313781	20p13 microdeletion syndrome	Disorder		4 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
444051	20q11.2 microdeletion syndrome	Disorder		11 *Cases*
574	21q deletion syndrome	Disorder		50 *Cases*
261323	21q22.11q22.12 microdeletion syndrome	Disorder		14 *Cases*
567	22q11.2 deletion syndrome	Disorder	9.6 *BP**	
567	22q11.2 deletion syndrome	Disorder	37.5 *BP*	
1727	22q11.2 duplication syndrome	Disorder		216 *Cases*
363680	2p13.2 microdeletion syndrome	Disorder		2 *Cases*
261349	2p15p16.1 microdeletion syndrome	Disorder		11 *Cases*
163693	2p21 microdeletion syndrome	Disorder		7 *Cases*
369881	2p21 microdeletion syndrome without cystinuria	Disorder		2 *Cases*
228402	2q23.1 microdeletion syndrome	Disorder		18 *Cases*
313947	2q23.1 microduplication syndrome	Disorder		2 *Cases*
1617	2q24 microdeletion syndrome	Disorder		23 *Cases*
251019	2q32q33 microdeletion syndrome	Disorder		25 *Cases*
1001	2q37 microdeletion syndrome	Disorder		115 *Cases*
35701	3-hydroxy-3-methylglutaryl-CoA synthase deficiency	Disorder		9 *Cases*
939	3-hydroxyisobutyric aciduria	Disorder		13 *Cases*
6	3-methylcrotonyl-CoA carboxylase deficiency	Disorder	2.65 *BP**	
67046	3-methylglutaconic aciduria type 1	Disorder		20 *Cases*
445038	3-methylglutaconic aciduria type 7	Disorder		22 *Cases*
505208	3-methylglutaconic aciduria type 8	Disorder		9 *Cases*
505216	3-methylglutaconic aciduria type 9	Disorder		4 *Cases*
79351	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form	Subtype of disorder		15 *Cases*
79350	3-phosphoserine phosphatase deficiency, infantile/juvenile form	Subtype of disorder		8 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
7	3C syndrome	Disorder		25 *Cases*
2616	3M syndrome	Disorder		200 *Cases*
293843	3MC syndrome	Disorder		32 *Cases*
435638	3p25.3 microdeletion syndrome	Disorder		8 *Cases*
1621	3q13 microdeletion syndrome	Disorder		42 *Cases*
96095	3q26 microduplication syndrome	Disorder		100 *Cases*
356947	3q26q27 microdeletion syndrome	Disorder		4 *Cases*
397695	3q27.3 microdeletion syndrome	Disorder		7 *Cases*
2975	46,XX difference of sex development-skeletal anomalies syndrome	Disorder		2 *Cases*
444048	46,XX ovarian dysgenesis-short stature syndrome	Disorder		3 *Cases*
2138	46,XX ovotesticular difference of sex development	Disorder		500 *Cases*
2138	46,XX ovotesticular difference of sex development	Disorder	2.5 *BP*	
393	46,XX testicular difference of sex development	Disorder	2.5 *P*	
90796	46,XY difference of sex development due to isolated 17,20-lyase deficiency	Disorder		15 *Cases*
443087	46,XY difference of sex development due to testicular 17,20-desmolase deficiency	Disorder		2 *Families*
168558	46,XY difference of sex development-adrenal insufficiency due to CYP11A1 deficiency	Disorder		9 *Cases*
168563	46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome	Disorder		5 *Cases*
8	47,XYY syndrome	Disorder	50.0 *BP**	
96263	48,XXXY syndrome	Disorder	1.0 *BP**	
10	48,XXYY syndrome	Disorder	1.9 *BP**	
99329	48,XYYY syndrome	Disorder		10 *Cases*
96264	49,XXXXY syndrome	Disorder	0.55 *BP**	
261534	49,XXXYY syndrome	Disorder		2 *Cases*
99330	49,XYYYY syndrome	Disorder		8 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
289494	4H leukodystrophy	Disorder		200 *Cases*
238750	4q21 microdeletion syndrome	Disorder		14 *Cases*
502437	4q25 proximal deletion syndrome	Disorder		3 *Cases*
217064	5-fluorouracil poisoning	Disorder	2.0 *P**	
33572	5-oxoprolinase deficiency	Disorder		8 *Cases*
329802	5p13 microduplication syndrome	Disorder		7 *Cases*
228384	5q14.3 microdeletion syndrome	Disorder		40 *Cases*
228415	5q35 microduplication syndrome	Disorder		30 *Cases*
251046	6p22 microdeletion syndrome	Disorder		19 *Cases*
75857	6q terminal deletion syndrome	Disorder		19 *Cases*
171829	6q16 microdeletion syndrome	Disorder		12 *Cases*
251056	6q25 microdeletion syndrome	Disorder		4 *Cases*
314034	7p22.1 microduplication syndrome	Disorder		5 *Cases*
96121	7q11.23 microduplication syndrome	Disorder		163 *Cases*
251061	7q31 microdeletion syndrome	Disorder		20 *Cases*
96092	8p inverted duplication/deletion syndrome	Disorder		60 *Cases*
251066	8p11.2 deletion syndrome	Disorder		3 *Cases*
251076	8p23.1 duplication syndrome	Disorder	1.72 *P*	
228399	8q12 microduplication syndrome	Disorder		4 *Cases*
284160	8q21.11 microdeletion syndrome	Disorder		13 *Cases*
178303	8q22.1 microdeletion syndrome	Disorder		6 *Cases*
508488	8q24.3 microdeletion syndrome	Disorder		2 *Cases*
324313	9p13 microdeletion syndrome	Disorder		4 *Cases*
531151	9q21.13 microdeletion syndrome	Disorder		10 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
401923	9q31.1q31.3 microdeletion syndrome	Disorder		2 *Cases*
495818	9q33.3q34.11 microdeletion syndrome	Disorder		4 *Cases*
439232	AApoAIV amyloidosis	Disorder		2 *Cases*
324723	ABeta amyloidosis, Arctic type	Subtype of disorder		1 *Family*
100006	ABeta amyloidosis, Dutch type	Subtype of disorder		250 *Cases*
324708	ABeta amyloidosis, Iowa type	Subtype of disorder		2 *Families*
324713	ABeta amyloidosis, Italian type	Subtype of disorder		7 *Families*
324718	ABetaA21G amyloidosis	Subtype of disorder		2 *Families*
324703	ABetaL34V amyloidosis	Subtype of disorder		1 *Family*
100008	ACys amyloidosis	Subtype of disorder		9 *Families*
978	ADULT syndrome	Disorder		50 *Cases*
85448	AGel amyloidosis	Disorder		475 *Cases*
442582	AH amyloidosis	Disorder		12 *Cases*
412069	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	Disorder		4 *Cases*
250977	AICA-ribosiduria	Disorder		4 *Cases*
90081	AIDS wasting syndrome	Disorder	20.0 *P**	
79085	AKT2-related familial partial lipodystrophy	Disorder		1 *Family*
85443	AL amyloidosis	Disorder	5.127 *P*	
85443	AL amyloidosis	Disorder	5.5311 *P**	
85443	AL amyloidosis	Disorder	1.044 *I*	
85443	AL amyloidosis	Disorder	1.1177 *I**	
35664	ALDH18A1-related De Barsy syndrome	Subtype of disorder		32 *Cases*
79327	ALG1-CDG	Disorder		57 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
280071	ALG11-CDG	Disorder		8 *Cases*
79324	ALG12-CDG	Disorder		11 *Cases*
324422	ALG13-CDG	Disorder		1 *Case*
79326	ALG2-CDG	Disorder		1 *Case*
79321	ALG3-CDG	Disorder		15 *Cases*
79320	ALG6-CDG	Disorder		54 *Cases*
79325	ALG8-CDG	Disorder		15 *Cases*
79328	ALG9-CDG	Disorder		12 *Cases*
597887	ALPI-related inflammatory bowel disease	Disorder		2 *Cases*
93561	ALys amyloidosis	Subtype of disorder		7 *Families*
157954	ANE syndrome	Disorder		5 *Cases*
356996	ANK3-related intellectual disability-sleep disturbance syndrome	Disorder		5 *Cases*
1133	AREDYLD syndrome	Disorder		3 *Cases*
314632	ATP13A2-related juvenile neuronal ceroid lipofuscinosis	Disorder		4 *Cases*
401911	AXIN2-related attenuated familial adenomatous polyposis	Subtype of disorder		4 *Families*
915	Aarskog-Scott syndrome	Disorder	0.5 *BP**	
916	Aase-Smith syndrome	Disorder		10 *Cases*
14	Abetalipoproteinemia	Disorder		100 *Cases*
920	Ablepharon macrostomia syndrome	Disorder		16 *Cases*
99050	Abnormal origin of right or left pulmonary artery from the aorta	Disorder		200 *Cases*
921	Abruzzo-Erickson syndrome	Disorder		4 *Cases*
2310	Absence deformity of leg-cataract syndrome	Disorder		2 *Cases*
1658	Absence of fingerprints-congenital milia syndrome	Disorder		10 *Families*
980	Absence of the pulmonary artery	Disorder	0.5 *I**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
3016	Absent radius-anogenital anomalies syndrome	Disorder		2 *Cases*
2951	Absent thumb-short stature-immunodeficiency syndrome	Disorder		3 *Cases*
3328	Absent tibia-polydactyly-arachnoid cyst syndrome	Disorder		3 *Cases*
90301	Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome	Disorder		5 *Cases*
926	Acatalasemia	Disorder	3.2 *P**	
48818	Aceruloplasminemia	Disorder	0.09 *P*	
929	Achalasia-microcephaly syndrome	Disorder		7 *Cases*
15	Achondroplasia	Disorder	4.73 *BP*	
15	Achondroplasia	Disorder	3.62 *BP**	
49382	Achromatopsia	Disorder	2.7 *P*	
424046	Acinar cell carcinoma of pancreas	Disorder	0.029 *I**	
40366	Acitretin/etretinate embryopathy	Disorder		26 *Cases*
90065	Acquired aneurysmal subarachnoid hemorrhage	Disorder	10.0 *P**	
599490	Acquired factor V deficiency	Disorder		200 *Cases*
599495	Acquired factor VII deficiency	Disorder		83 *Cases*
599501	Acquired factor X deficiency	Disorder		77 *Cases*
599501	Acquired factor X deficiency	Disorder	0.0 *P*	
79086	Acquired generalized lipodystrophy	Disorder	1.0 *P**	
599480	Acquired hemophilia A	Disorder	0.1505 *I**	
2221	Acquired hypertrichosis lanuginosa	Disorder		60 *Cases*
75564	Acquired idiopathic sideroblastic anemia	Disorder	0.09 *I**	
464453	Acquired methemoglobinemia	Disorder		242 *Cases*
91136	Acquired monoclonal Ig light chain-associated Fanconi syndrome	Disorder		100 *Cases*
228247	Acquired pseudoxanthoma elasticum	Disorder		20 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
99147	Acquired von Willebrand syndrome	Disorder		300 *Cases*
263534	Acral peeling skin syndrome	Disorder		40 *Cases*
281127	Acral self-healing collodion baby	Disorder		2 *Cases*
958	Acro-renal-mandibular syndrome	Disorder		10 *Cases*
959	Acro-renal-ocular syndrome	Disorder		20 *Families*
36	Acrocallosal syndrome	Disorder		38 *Cases*
2008	Acrocardiofacial syndrome	Disorder		10 *Cases*
221054	Acrocephalopolydactyly	Disorder		8 *Cases*
949	Acrocraniofacial dysostosis	Disorder		2 *Cases*
950	Acrodysostosis	Disorder		80 *Cases*
2956	Acrodysplasia scoliosis	Disorder		2 *Cases*
1786	Acrofacial dysostosis, Catania type	Disorder		2 *Families*
64542	Acrofacial dysostosis, Kennedy-Teebi type	Disorder		2 *Cases*
1787	Acrofacial dysostosis, Palagonia type	Disorder		4 *Cases*
1788	Acrofacial dysostosis, Rodríguez type	Disorder		13 *Cases*
1784	Acrofrontofacionasal dysostosis	Disorder		12 *Cases*
963	Acromegaly	Disorder	0.47 *I*	
39	Acromelanosis	Disorder		10 *Cases*
1827	Acromelic frontonasal dysplasia	Disorder		22 *Cases*
968	Acromesomelic dysplasia, Hunter-Thompson type	Disorder		10 *Cases*
40	Acromesomelic dysplasia, Maroteaux type	Disorder		50 *Cases*
969	Acromicric dysplasia	Disorder		60 *Cases*
363665	Acroosteolysis-keloid-like lesions-premature aging syndrome	Disorder		5 *Cases*
85203	Acropectoral syndrome	Disorder		25 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
957	Acropectorovertebral dysplasia	Disorder		30 *Cases*
971	Acrorenal syndrome	Disorder		20 *Cases*
163696	Action myoclonus-renal failure syndrome	Disorder		38 *Cases*
397596	Activated PI3K-delta syndrome	Disorder		250 *Cases*
284460	Acute annular outer retinopathy	Disorder		12 *Cases*
83597	Acute disseminated encephalomyelitis	Disorder	0.6 *I**	
363549	Acute encephalopathy with biphasic seizures and late reduced diffusion	Disorder		283 *Cases*
293173	Acute generalized exanthematous pustulosis	Disorder	0.3 *I*	
217371	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	Disorder		32 *Cases*
466794	Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome	Disorder		3 *Cases*
370088	Acute infantile liver failure-multisystemic involvement syndrome	Disorder		6 *Cases*
98916	Acute inflammatory demyelinating polyradiculoneuropathy	Disorder	3.1 *P**	
79276	Acute intermittent porphyria	Disorder	0.013 *I**	
79276	Acute intermittent porphyria	Disorder	0.54 *P**	
79126	Acute interstitial pneumonia	Disorder	3.8 *P**	
90062	Acute liver failure	Disorder	20.0 *P**	
178320	Acute lung injury	Disorder	25.0 *I**	
488239	Acute macular neuroretinopathy	Disorder		101 *Cases*
518	Acute megakaryoblastic leukemia	Disorder	0.02 *I**	
514	Acute monoblastic/monocytic leukemia	Disorder	0.13 *I**	
98834	Acute myeloblastic leukemia with maturation	Disorder	0.02 *I**	
98833	Acute myeloblastic leukemia without maturation	Disorder	0.01 *I**	
98832	Acute myeloid leukemia with minimal	Disorder	0.01 *I**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	differentiation			
585867	Acute myeloid leukemia with t(9;22)(q34.1;q11.2)	Disorder	4.0 *I*	
517	Acute myelomonocytic leukemia	Disorder	0.17 *I**	
86843	Acute panmyelosis with myelofibrosis	Disorder	0.06 *I**	
90064	Acute peripheral arterial occlusion	Disorder	16.0 *P**	
520	Acute promyelocytic leukemia	Disorder	0.11 *I**	
139417	Acute transverse myelitis	Disorder	4.72 *I*	
284454	Acute zonal occult outer retinopathy	Disorder		150 *Cases*
99901	Acyl-CoA dehydrogenase 9 deficiency	Disorder		23 *Cases*
55881	Adamantinoma	Disorder	0.01 *I**	
55881	Adamantinoma	Disorder	0.11	
974	Adams-Oliver syndrome	Disorder		398 *Cases*
85138	Addison disease	Disorder	12.5 *P**	
2952	Adducted thumbs-arthrogryposis syndrome, Christian type	Disorder		9 *Cases*
213504	Adenocarcinoma of ovary	Disorder	5.97 *I**	
424016	Adenocarcinoma of the anal canal	Disorder	0.253 *I**	
213772	Adenocarcinoma of the cervix uteri	Disorder	1.01 *I**	
99976	Adenocarcinoma of the esophagus	Disorder	3.264 *I**	
99976	Adenocarcinoma of the esophagus	Disorder	0.7 *I*	
99976	Adenocarcinoma of the esophagus	Disorder	5.55	
424991	Adenocarcinoma of the gallbladder and extrahepatic biliary tract	Disorder	2.62 *I**	
424943	Adenocarcinoma of the liver and intrahepatic biliary tract	Disorder	0.412 *I**	
104075	Adenocarcinoma of the small intestine	Disorder	0.588 *I**	
45	Adenosine monophosphate deaminase deficiency	Disorder		100 *Cases*
91127	Adenovirus infection in	Disorder	18.0 *P**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	immunocompromised patients			
46	Adenylosuccinate lyase deficiency	Disorder		56 *Cases*
482601	Adenylosuccinate synthetase-like 1-related distal myopathy	Disorder		19 *Cases*
1501	Adrenocortical carcinoma	Disorder	0.75 *P**	
1501	Adrenocortical carcinoma	Disorder	0.03 *I**	
977	Adrenomyodystrophy	Disorder		2 *Cases*
86875	Adult T-cell leukemia/lymphoma	Disorder	3.0 *P**	
2666	Adult familial nephronophthisis-spastic quadriparesia syndrome	Disorder		2 *Cases*
210159	Adult hepatocellular carcinoma	Disorder	3.22 *I**	
178487	Adult intestinal botulism	Subtype of disorder		19 *Cases*
206583	Adult polyglucosan body disease	Subtype of disorder		50 *Cases*
99027	Adult-onset autosomal dominant leukodystrophy	Disorder		20 *Families*
284289	Adult-onset autosomal recessive cerebellar ataxia	Disorder		14 *Cases*
255132	Adult-onset autosomal recessive sideroblastic anemia	Disorder		2 *Cases*
420492	Adult-onset cervical dystonia, DYT23 type	Disorder		2 *Families*
329478	Adult-onset distal myopathy due to VCP mutation	Disorder		9 *Cases*
199351	Adult-onset dystonia-parkinsonism	Disorder		14 *Cases*
83617	Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	Disorder		3 *Cases*
51	Aicardi-Goutières syndrome	Disorder	10.0 *P**	
404454	Alacrimia-choreoathetosis-liver dysfunction syndrome	Disorder		8 *Cases*
52	Alagille syndrome	Disorder	0.8 *BP**	
2007	Alar cartilages hypoplasia-coloboma-telecanthus syndrome	Disorder		2 *Cases*
319671	Alazami syndrome	Disorder		10 *Cases*
53	Albers-Schönberg osteopetrosis	Disorder	1.0 *P*	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
53	Albers-Schönberg osteopetrosis	Disorder	5.0 *P**	
998	Albinism-deafness syndrome	Disorder		1 *Family*
502444	Alkaline ceramidase 3 deficiency	Disorder		2 *Cases*
59	Allan-Herndon-Dudley syndrome	Disorder		320 *Cases*
1006	Alopecia antibody deficiency	Disorder		3 *Cases*
700	Alopecia totalis	Disorder	10.5 *P**	
701	Alopecia universalis	Disorder	25.0 *P**	
1005	Alopecia-contractures-dwarfism-intellectual disability syndrome	Disorder		5 *Cases*
1008	Alopecia-epilepsy-pyorrhea-intellectual disability syndrome	Disorder		12 *Cases*
2850	Alopecia-intellectual disability syndrome	Disorder		15 *Families*
1014	Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome	Disorder		2 *Cases*
726	Alpers-Huttenlocher syndrome	Disorder	0.7 *BP**	
726	Alpers-Huttenlocher syndrome	Disorder	0.07 *P**	
60	Alpha-1-antitrypsin deficiency	Disorder	20.0 *P**	
399058	Alpha-B crystallin-related late-onset myopathy	Disorder		17 *Cases*
3137	Alpha-N-acetylgalactosaminidase deficiency	Disorder		20 *Cases*
79279	Alpha-N-acetylgalactosaminidase deficiency type 1	Subtype of disorder		10 *Cases*
79280	Alpha-N-acetylgalactosaminidase deficiency type 2	Subtype of disorder		10 *Cases*
79281	Alpha-N-acetylgalactosaminidase deficiency type 3	Subtype of disorder		10 *Cases*
280333	Alpha-dystroglycan-related limb-girdle muscular dystrophy R16	Disorder		1 *Case*
100025	Alpha-heavy chain disease	Subtype of disorder		400 *Cases*
61	Alpha-mannosidosis	Disorder	0.1 *P**	
847	Alpha-thalassemia-X-linked intellectual disability syndrome	Disorder		200 *Cases*
98791	Alpha-thalassemia-intellectual disability	Disorder		20 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	syndrome linked to chromosome 16			
231401	Alpha-thalassemia-myelodysplastic syndrome	Disorder		80 *Cases*
86818	Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	Disorder		2 *Families*
64	Alström syndrome	Disorder		950 *Cases*
284	Alveolar echinococcosis	Disorder	0.16 *I**	
1021	Amaurosis-hypertrichosis syndrome	Disorder		2 *Cases*
1028	Amelo-onycho-hypohidrotic syndrome	Disorder		2 *Cases*
314422	Ameloblastic carcinoma	Disorder		40 *Cases*
1908	Aminopterin/methotrexate embryofetopathy	Disorder		17 *Cases*
67043	Amoebic keratitis	Disorder	1.0 *P**	
319635	Amyloidosis cutis dyschromia	Disorder		27 *Cases*
803	Amyotrophic lateral sclerosis	Disorder	2.2 *I**	
803	Amyotrophic lateral sclerosis	Disorder	3.85 *P*	
803	Amyotrophic lateral sclerosis	Disorder	5.2 *P**	
803	Amyotrophic lateral sclerosis	Disorder	1.35 *I*	
357043	Amyotrophic lateral sclerosis type 4	Disorder		70 *Cases*
228113	Anal fistula	Disorder	18.3 *P**	
98841	Anaplastic large cell lymphoma	Disorder	2.0 *P**	
251630	Anaplastic oligodendroglioma	Disorder	0.09 *I**	
142	Anaplastic thyroid carcinoma	Disorder	0.17 *I**	
142	Anaplastic thyroid carcinoma	Disorder	0.1 *P**	
93347	Anauxetic dysplasia	Disorder		10 *Cases*
37553	Andersen-Tawil syndrome	Disorder	0.1 *I**	
284984	Aneurysm-osteoarthritis syndrome	Disorder		45 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
63442	Angel-shaped phalango-epiphyseal dysplasia	Disorder		20 *Cases*
72	Angelman syndrome	Disorder	7.5 *P*	
72	Angelman syndrome	Disorder	1.3 *BP**	
251671	Angiocentric glioma	Disorder		52 *Cases*
263413	Angiosarcoma	Disorder	0.02	
370039	Angora hair nevus	Disorder		2 *Cases*
69088	Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome	Disorder		2 *Cases*
1069	Aniridia-absent patella syndrome	Disorder		3 *Cases*
1065	Aniridia-cerebellar ataxia-intellectual disability syndrome	Disorder		22 *Families*
1068	Aniridia-intellectual disability syndrome	Disorder		2 *Cases*
1067	Aniridia-ptosis-intellectual disability-familial obesity syndrome	Disorder		3 *Cases*
1064	Aniridia-renal agenesis-psychomotor retardation syndrome	Disorder		2 *Cases*
1070	Anisakiasis	Disorder	0.32 *I*	
1074	Ankyloblepharon filiforme adnatum-imperforate anus syndrome	Subtype of disorder		3 *Families*
2206	Ankylosing vertebral hyperostosis with tylosis	Disorder		8 *Cases*
254411	Annular atrophic lichen planus	Disorder		10 *Cases*
281139	Annular epidermolytic ichthyosis	Disorder		7 *Families*
675	Annular pancreas	Disorder	1.8 *BP**	
69125	Anonychia with flexural pigmentation	Disorder		3 *Cases*
1094	Anonychia-microcephaly syndrome	Disorder		4 *Cases*
90390	Anonychia-onychodystrophy syndrome	Subtype of disorder		14 *Cases*
1104	Anophthalmia plus syndrome	Disorder		17 *Cases*
1101	Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome	Disorder		3 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
77298	Anophthalmia/microphthalmia-esophageal atresia syndrome	Disorder		30 *Cases*
93976	Anotia	Disorder	0.028 *BP**	
2987	Antecubital pterygium syndrome	Disorder		11 *Cases*
562559	Anterior maxillary protrusion-strabismus-intellectual disability syndrome	Disorder		7 *Cases*
375	Anti-glomerular basement membrane disease	Disorder	0.08 *I**	
375	Anti-glomerular basement membrane disease	Disorder	0.2 *P**	
454710	Anti-p200 pemphigoid	Disorder		50 *Cases*
81	Antisynthetase syndrome	Disorder	3.5 *P*	
1457	Aorta coarctation	Disorder	35.6 *BP**	
1110	Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome	Disorder		4 *Cases*
2299	Aortic arch interruption	Disorder	0.3 *BP**	
3400	Aorto-ventricular tunnel	Disorder		130 *Cases*
1112	Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome	Disorder		3 *Cases*
1113	Aphalangy-syndactyly-microcephaly syndrome	Disorder		5 *Cases*
324540	Aphonia-deafness-retinal dystrophy-bifid halluces-intellectual disability syndrome	Disorder		2 *Cases*
1114	Aplasia cutis congenita	Disorder	10.0 *BP*	
1116	Aplasia cutis congenita-intestinal lymphangiectasia syndrome	Disorder		3 *Cases*
1117	Aplasia cutis-myopia syndrome	Disorder		4 *Cases*
99981	Apnea of prematurity	Disorder	8.5 *P**	
425	Apolipoprotein A-I deficiency	Disorder		30 *Families*
1126	Aprosencephaly cerebellar dysgenesis	Disorder		2 *Cases*
1129	Arachnodactyly-abnormal ossification-intellectual disability syndrome	Disorder		5 *Cases*
1130	Arachnodactyly-intellectual disability-dysmorphism syndrome	Disorder		3 *Cases*
101096	Aregenerative anemia	Disorder	0.3312 *I*	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
23	Argininosuccinic aciduria	Disorder	1.0 *P**	
91	Aromatase deficiency	Disorder		38 *Cases*
178345	Aromatase excess syndrome	Disorder		30 *Cases*
35708	Aromatic L-amino acid decarboxylase deficiency	Disorder		140 *Cases*
1135	Arrhinia-choanal atresia-microphthalmia syndrome	Disorder		4 *Cases*
1682	Arterial dissection-lentiginosis syndrome	Disorder		4 *Cases*
3342	Arterial tortuosity syndrome	Disorder		102 *Cases*
1150	Arthrogryposis multiplex congenita-whistling face syndrome	Disorder		10 *Cases*
53696	Arthrogryposis-anterior horn cell disease syndrome	Disorder		15 *Cases*
3200	Arthrogryposis-ectodermal dysplasia syndrome	Disorder		2 *Cases*
1485	Arthrogryposis-hyperkeratosis syndrome, lethal form	Disorder		2 *Cases*
1144	Arthrogryposis-like hand anomaly-sensorineural deafness syndrome	Disorder		1 *Family*
2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	Disorder		100 *Cases*
65720	Arthrogryposis-severe scoliosis syndrome	Disorder		2 *Families*
1253	Ascher syndrome	Disorder		50 *Cases*
137686	Asherman syndrome	Disorder	44.0 *P**	
85175	Astley-Kendall dysplasia	Disorder		5 *Cases*
251679	Astroblastoma	Disorder	0.02 *I**	
96	Ataxia with vitamin E deficiency	Disorder	0.33 *P**	
1188	Ataxia-deafness-intellectual disability syndrome	Disorder		8 *Cases*
370022	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome	Disorder		7 *Cases*
459033	Ataxia-oculomotor apraxia type 4	Disorder		12 *Cases*
1184	Ataxia-photosensitivity-short stature syndrome	Disorder		2 *Cases*
100	Ataxia-telangiectasia	Disorder	0.49 *P**	

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56304	Atelosteogenesis type II	Disorder		25 *Cases*
56305	Atelosteogenesis type III	Disorder		25 *Cases*
69739	Athabaskan brainstem dysgenesis syndrome	Disorder		13 *Cases*
1192	Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome	Disorder		2 *Cases*
95713	Athyreosis	Disorder	3.5 *P**	
1193	Atkin-Flaitz syndrome	Disorder		14 *Cases*
163934	Atopic keratoconjunctivitis	Disorder	15.0 *P**	
1479	Atrial septal defect-atrioventricular conduction defects syndrome	Disorder		11 *Cases*
1352	Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	Disorder		2 *Cases*
352723	Attenuated Chédiak-Higashi syndrome	Disorder		100 *Cases*
544628	Atypical Fanconi syndrome-neonatal hyperinsulinism syndrome	Disorder		7 *Cases*
314466	Atypical Meigs syndrome	Disorder		9 *Cases*
1456	Atypical coarctation of aorta	Subtype of disorder	0.17 *BP**	
314721	Atypical dentin dysplasia due to SMOC2 deficiency	Subtype of disorder		4 *Cases*
289863	Atypical glycine encephalopathy	Subtype of disorder		20 *Cases*
2134	Atypical hemolytic uremic syndrome	Disorder	1.0 *P**	
238523	Atypical hypotonia-cystinuria syndrome	Disorder		2 *Cases*
391411	Atypical juvenile parkinsonism	Disorder		6 *Families*
86797	Atypical lichen myxedematosus	Disorder		20 *Cases*
542585	Auditory neuropathy-optic atrophy syndrome	Disorder		8 *Cases*
77300	Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome	Disorder		2 *Cases*
137888	Auriculocondylar syndrome	Disorder		50 *Cases*
114	Auriculoosteodysplasia	Disorder		2 *Families*

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352490	Autism spectrum disorder due to AUTS2 deficiency	Disorder		60 *Cases*
370943	Autism spectrum disorder-epilepsy-arthrogryposis syndrome	Disorder		8 *Cases*
308410	Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency	Disorder		5 *Families*
324636	Autoerythrocyte sensitization syndrome	Disorder		170 *Cases*
420789	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	Disorder		10 *Cases*
391487	Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome	Disorder		5 *Cases*
444463	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome	Disorder		6 *Cases*
2137	Autoimmune hepatitis	Disorder	1.2 *I*	
2137	Autoimmune hepatitis	Disorder	0.75 *I**	
2137	Autoimmune hepatitis	Disorder	23.5 *P*	
444092	Autoimmune interstitial lung disease-arthritis syndrome	Disorder		5 *Families*
3261	Autoimmune lymphoproliferative syndrome	Disorder		500 *Cases*
436159	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency	Disorder		17 *Cases*
275517	Autoimmune lymphoproliferative syndrome with recurrent viral infections	Disorder		1 *Family*
747	Autoimmune pulmonary alveolar proteinosis	Disorder	2.66 *P*	
324530	Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation	Disorder		2 *Cases*
329173	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis	Disorder		5 *Cases*
33110	Autosomal agammaglobulinemia	Subtype of disorder		100 *Cases*
487814	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation	Disorder		2 *Cases*
435819	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation	Disorder		2 *Cases*
401964	Autosomal dominant Charcot-Marie-	Disorder		2 *Families*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	Tooth disease type 2 with giant axons			
99946	Autosomal dominant Charcot-Marie-Tooth disease type 2A1	Disorder		1 *Family*
99938	Autosomal dominant Charcot-Marie-Tooth disease type 2D	Disorder		44 *Cases*
521414	Autosomal dominant Charcot-Marie-Tooth disease type 2DD	Disorder		51 *Cases*
99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F	Disorder		5 *Families*
99941	Autosomal dominant Charcot-Marie-Tooth disease type 2G	Disorder		1 *Family*
99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K	Disorder		30 *Cases*
99945	Autosomal dominant Charcot-Marie-Tooth disease type 2L	Disorder		1 *Family*
228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M	Disorder		20 *Cases*
228174	Autosomal dominant Charcot-Marie-Tooth disease type 2N	Disorder		28 *Cases*
329258	Autosomal dominant Charcot-Marie-Tooth disease type 2Q	Disorder		8 *Cases*
397735	Autosomal dominant Charcot-Marie-Tooth disease type 2U	Disorder		2 *Cases*
447964	Autosomal dominant Charcot-Marie-Tooth disease type 2V	Disorder		21 *Cases*
488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W	Disorder		24 *Cases*
435387	Autosomal dominant Charcot-Marie-Tooth disease type 2Y	Disorder		7 *Cases*
466768	Autosomal dominant Charcot-Marie-Tooth disease type 2Z	Disorder		21 *Cases*
3107	Autosomal dominant Robinow syndrome	Subtype of disorder		100 *Cases*
209335	Autosomal dominant adult-onset proximal spinal muscular atrophy	Disorder	0.1 *P**	
314399	Autosomal dominant aplasia and myelodysplasia	Disorder		6 *Cases*
314404	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	Disorder		80 *Cases*
363447	Autosomal dominant childhood-onset proximal spinal muscular atrophy	Disorder		97 *Cases*
90348	Autosomal dominant cutis laxa	Disorder		50 *Cases*
79499	Autosomal dominant deafness-onychodystrophy syndrome	Disorder		22 *Cases*
476093	Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome	Disorder		8 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
329466	Autosomal dominant focal dystonia, DYT25 type	Disorder		28 *Cases*
402003	Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering	Disorder		21 *Cases*
2314	Autosomal dominant hyper-IgE syndrome due to STAT3 deficiency	Disorder	0.1 *I**	
1810	Autosomal dominant hypohidrotic ectodermal dysplasia	Subtype of disorder		40 *Cases*
89937	Autosomal dominant hypophosphatemic rickets	Disorder		100 *Cases*
457193	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	Disorder		76 *Cases*
100043	Autosomal dominant intermediate Charcot-Marie-Tooth disease type A	Disorder		20 *Cases*
100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	Disorder		37 *Cases*
100045	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C	Disorder		35 *Cases*
100046	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	Disorder		12 *Cases*
93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	Disorder		21 *Cases*
352670	Autosomal dominant intermediate Charcot-Marie-Tooth disease type F	Disorder		8 *Cases*
324585	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	Disorder		9 *Cases*
266	Autosomal dominant limb-girdle muscular dystrophy type 1A	Disorder		4 *Families*
140957	Autosomal dominant macrothrombocytopenia	Disorder		100 *Cases*
319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	Disorder		68 *Cases*
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	Disorder		2 *Cases*
457050	Autosomal dominant mitochondrial myopathy with exercise intolerance	Disorder		15 *Cases*
65743	Autosomal dominant multiple pterygium syndrome	Disorder		4 *Cases*
99846	Autosomal dominant myoglobinuria	Disorder		2 *Families*
440354	Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome	Disorder		1 *Family*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
329211	Autosomal dominant neovascular inflammatory vitreoretinopathy	Disorder		99 *Cases*
98784	Autosomal dominant nocturnal frontal lobe epilepsy	Disorder		100 *Families*
67036	Autosomal dominant optic atrophy and cataract	Disorder		3 *Families*
1215	Autosomal dominant optic atrophy plus syndrome	Disorder	0.5 *P**	
98673	Autosomal dominant optic atrophy, classic form	Disorder	2.0 *P*	
2783	Autosomal dominant osteopetrosis type 1	Disorder		33 *Cases*
1010	Autosomal dominant palmoplantar keratoderma and congenital alopecia	Disorder		10 *Cases*
730	Autosomal dominant polycystic kidney disease	Disorder	39.6 *P**	
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	Disorder		30 *Cases*
1300	Autosomal dominant popliteal pterygium syndrome	Disorder		200 *Cases*
476119	Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome	Disorder		1 *Family*
34528	Autosomal dominant primary hypomagnesemia with hypocalciuria	Disorder		28 *Cases*
88659	Autosomal dominant progressive nephropathy with hypertension	Disorder		14 *Cases*
314889	Autosomal dominant proximal renal tubular acidosis	Subtype of disorder		1 *Family*
209867	Autosomal dominant rhegmatogenous retinal detachment	Disorder		38 *Cases*
140481	Autosomal dominant slowed nerve conduction velocity	Disorder		1 *Family*
251282	Autosomal dominant spastic ataxia type 1	Disorder		53 *Cases*
100991	Autosomal dominant spastic paraplegia type 10	Disorder		10 *Families*
100993	Autosomal dominant spastic paraplegia type 12	Disorder		27 *Cases*
100994	Autosomal dominant spastic paraplegia type 13	Disorder		10 *Cases*
100998	Autosomal dominant spastic paraplegia type 17	Disorder		20 *Families*
100999	Autosomal dominant spastic paraplegia type 19	Disorder		1 *Family*
101009	Autosomal dominant spastic paraplegia type 29	Disorder		1 *Family*
320365	Autosomal dominant spastic paraplegia	Disorder		1 *Family*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	type 36			
171612	Autosomal dominant spastic paraplegia type 37	Disorder		13 *Cases*
171617	Autosomal dominant spastic paraplegia type 38	Disorder		1 *Family*
320355	Autosomal dominant spastic paraplegia type 41	Disorder		7 *Cases*
171863	Autosomal dominant spastic paraplegia type 42	Disorder		1 *Family*
100988	Autosomal dominant spastic paraplegia type 6	Disorder		10 *Families*
444099	Autosomal dominant spastic paraplegia type 73	Disorder		1 *Family*
100989	Autosomal dominant spastic paraplegia type 8	Disorder		10 *Families*
447753	Autosomal dominant spastic paraplegia type 9A	Disorder		2 *Families*
447757	Autosomal dominant spastic paraplegia type 9B	Disorder		3 *Families*
228169	Autosomal dominant striatal neurodegeneration	Disorder		11 *Cases*
466806	Autosomal dominant thrombocytopenia with platelet secretion defect	Disorder		4 *Families*
3086	Autosomal dominant vitreoretinochoroidopathy	Disorder		3 *Cases*
79278	Autosomal erythropoietic protoporphyria	Disorder	0.012 *I**	
79278	Autosomal erythropoietic protoporphyria	Disorder	0.92 *P**	
466775	Autosomal recessive Charcot-Marie-Tooth disease type 2X	Disorder		29 *Cases*
1507	Autosomal recessive Robinow syndrome	Subtype of disorder		100 *Cases*
250984	Autosomal recessive Stickler syndrome	Subtype of disorder		15 *Cases*
1027	Autosomal recessive amelia	Disorder		3 *Cases*
519388	Autosomal recessive anterior segment dysgenesis	Disorder		8 *Cases*
247815	Autosomal recessive ataxia due to PEX10 deficiency	Disorder		6 *Cases*
139485	Autosomal recessive ataxia due to ubiquinone deficiency	Disorder		31 *Cases*
88644	Autosomal recessive ataxia, Beauce type	Disorder		57 *Cases*
521411	Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect	Disorder		2 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
324442	Autosomal recessive axonal neuropathy with neuromyotonia	Disorder		33 *Families*
139455	Autosomal recessive bestrophinopathy	Disorder		20 *Cases*
448242	Autosomal recessive brachyolmia	Disorder		20 *Cases*
453521	Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	Disorder		2 *Cases*
412057	Autosomal recessive cerebellar ataxia due to STUB1 deficiency	Disorder		10 *Families*
352641	Autosomal recessive cerebellar ataxia with late-onset spasticity	Disorder		10 *Cases*
404499	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency	Disorder		2 *Cases*
404493	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency	Disorder		3 *Cases*
95434	Autosomal recessive cerebellar ataxia-movement disorder syndrome	Disorder		27 *Cases*
363429	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome	Disorder		17 *Cases*
363969	Autosomal recessive cerebral atrophy	Disorder		4 *Cases*
506353	Autosomal recessive complex spastic paraplegia due to Kennedy pathway dysfunction	Disorder		4 *Cases*
363432	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency	Subtype of disorder		7 *Cases*
324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency	Subtype of disorder		10 *Cases*
90349	Autosomal recessive cutis laxa type 1	Disorder		60 *Cases*
101150	Autosomal recessive dopa-responsive dystonia	Disorder		50 *Cases*
1974	Autosomal recessive faciodigitogenital syndrome	Disorder		26 *Cases*
329329	Autosomal recessive frontotemporal pachygyria	Disorder		7 *Cases*
79408	Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	Disorder	1.3 *BP**	
89838	Autosomal recessive generalized epidermolysis bullosa simplex	Disorder		19 *Cases*
300547	Autosomal recessive infantile hypercalcemia	Disorder		12 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
217055	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	Disorder		8 *Families*
254334	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B	Disorder		1 *Case*
369867	Autosomal recessive intermediate Charcot-Marie-Tooth disease type C	Disorder		3 *Cases*
435998	Autosomal recessive intermediate Charcot-Marie-Tooth disease type D	Disorder		4 *Cases*
98676	Autosomal recessive isolated optic atrophy	Disorder		5 *Cases*
538096	Autosomal recessive lethal neonatal axonal sensorimotor polyneuropathy	Disorder		13 *Cases*
314572	Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome	Disorder		3 *Cases*
206580	Autosomal recessive lower motor neuron disease with childhood onset	Disorder		5 *Cases*
667	Autosomal recessive malignant osteopetrosis	Disorder	0.75 *BP**	
477857	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency	Disorder		7 *Cases*
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	Disorder		18 *Cases*
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency	Disorder		6 *Cases*
2990	Autosomal recessive multiple pterygium syndrome	Disorder		64 *Cases*
319332	Autosomal recessive myogenic arthrogryposis multiplex congenita	Disorder		1 *Family*
280654	Autosomal recessive nail dysplasia	Disorder		4 *Cases*
93329	Autosomal recessive omodysplasia	Subtype of disorder		23 *Cases*
227976	Autosomal recessive optic atrophy, OPA7 type	Disorder		17 *Cases*
1366	Autosomal recessive palmoplantar keratoderma and congenital alopecia	Disorder		8 *Cases*
437552	Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity	Disorder		3 *Cases*
420702	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency	Disorder		4 *Cases*
420699	Autosomal recessive severe congenital	Disorder		2 *Cases*

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	neutropenia due to CXCR2 deficiency			
331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	Disorder		57 *Cases*
423384	Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency	Disorder		14 *Cases*
314603	Autosomal recessive spastic ataxia with leukoencephalopathy	Disorder		54 *Cases*
254343	Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome	Disorder		6 *Cases*
100995	Autosomal recessive spastic paraplegia type 14	Disorder		1 *Family*
100996	Autosomal recessive spastic paraplegia type 15	Disorder		10 *Families*
101000	Autosomal recessive spastic paraplegia type 20	Disorder		36 *Cases*
101001	Autosomal recessive spastic paraplegia type 21	Disorder		35 *Cases*
101003	Autosomal recessive spastic paraplegia type 23	Disorder		5 *Families*
101004	Autosomal recessive spastic paraplegia type 24	Disorder		1 *Family*
101005	Autosomal recessive spastic paraplegia type 25	Disorder		1 *Family*
101006	Autosomal recessive spastic paraplegia type 26	Disorder		10 *Families*
101007	Autosomal recessive spastic paraplegia type 27	Disorder		10 *Cases*
101008	Autosomal recessive spastic paraplegia type 28	Disorder		7 *Cases*
171622	Autosomal recessive spastic paraplegia type 32	Disorder		1 *Family*
171629	Autosomal recessive spastic paraplegia type 35	Disorder		38 *Cases*
139480	Autosomal recessive spastic paraplegia type 39	Disorder		2 *Families*
320370	Autosomal recessive spastic paraplegia type 43	Disorder		2 *Cases*
320401	Autosomal recessive spastic paraplegia type 44	Disorder		3 *Cases*
320396	Autosomal recessive spastic paraplegia type 45	Disorder		7 *Families*
320391	Autosomal recessive spastic paraplegia type 46	Disorder		5 *Cases*
306511	Autosomal recessive spastic paraplegia type 48	Disorder		2 *Cases*
319199	Autosomal recessive spastic paraplegia type 53	Disorder		9 *Cases*
320380	Autosomal recessive spastic paraplegia	Disorder		6 *Families*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	type 54			
320375	Autosomal recessive spastic paraplegia type 55	Disorder		14 *Cases*
320411	Autosomal recessive spastic paraplegia type 56	Disorder		5 *Families*
431329	Autosomal recessive spastic paraplegia type 57	Disorder		2 *Cases*
401795	Autosomal recessive spastic paraplegia type 59	Disorder		3 *Cases*
401800	Autosomal recessive spastic paraplegia type 60	Disorder		1 *Case*
401780	Autosomal recessive spastic paraplegia type 61	Disorder		4 *Cases*
401785	Autosomal recessive spastic paraplegia type 62	Disorder		7 *Cases*
401805	Autosomal recessive spastic paraplegia type 63	Disorder		2 *Cases*
401810	Autosomal recessive spastic paraplegia type 64	Disorder		4 *Cases*
401815	Autosomal recessive spastic paraplegia type 66	Disorder		2 *Cases*
401820	Autosomal recessive spastic paraplegia type 67	Disorder		2 *Cases*
401830	Autosomal recessive spastic paraplegia type 69	Disorder		2 *Cases*
401835	Autosomal recessive spastic paraplegia type 70	Disorder		4 *Cases*
401840	Autosomal recessive spastic paraplegia type 71	Disorder		1 *Case*
468661	Autosomal recessive spastic paraplegia type 74	Disorder		11 *Cases*
459056	Autosomal recessive spastic paraplegia type 75	Disorder		5 *Cases*
488594	Autosomal recessive spastic paraplegia type 76	Disorder		7 *Families*
466722	Autosomal recessive spastic paraplegia type 77	Disorder		8 *Cases*
513436	Autosomal recessive spastic paraplegia type 78	Disorder		7 *Cases*
447760	Autosomal recessive spastic paraplegia type 9B	Disorder		2 *Families*
95433	Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome	Disorder		3 *Families*
401979	Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type	Disorder		4 *Cases*
280365	Autosomal semi-dominant severe lipodystrophic laminopathy	Disorder		7 *Cases*

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209951	Autosomal spastic paraplegia type 18	Disorder		9 *Cases*
101010	Autosomal spastic paraplegia type 30	Disorder		3 *Families*
397946	Autosomal spastic paraplegia type 58	Disorder		19 *Cases*
401849	Autosomal spastic paraplegia type 72	Disorder		14 *Cases*
300345	Autosomal systemic lupus erythematosus	Disorder		7 *Families*
454836	Avian influenza	Disorder		826 *Cases*
782	Axenfeld-Rieger syndrome	Disorder	0.5 *P**	
168549	Axial spondylometaphyseal dysplasia	Disorder		13 *Cases*
1272	Aymé-Gripp syndrome	Disorder		18 *Cases*
67038	B-cell chronic lymphocytic leukemia	Disorder	48.0 *P**	
567502	B-cell immunodeficiency-limb anomaly-urogenital malformation syndrome	Disorder		10 *Cases*
86852	B-cell prolymphocytic leukemia	Disorder	0.05 *I**	
536467	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	Subtype of disorder		41 *Families*
79332	B4GALT1-CDG	Disorder		1 *Case*
75496	B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome	Subtype of disorder		34 *Cases*
464336	BENTA disease	Disorder		8 *Cases*
363454	BICD2-related autosomal dominant childhood-onset proximal spinal muscular atrophy	Subtype of disorder		60 *Cases*
217266	BNAR syndrome	Disorder		9 *Families*
107	BOR syndrome	Disorder	2.5 *P*	
85284	BRESEK syndrome	Disorder		5 *Cases*
476084	BVES-related limb-girdle muscular dystrophy	Disorder		3 *Cases*
183713	Bacterial susceptibility due to TLR signaling pathway deficiency	Disorder		24 *Cases*
36234	Bacterial toxic-shock syndrome	Disorder	3.0 *P*	
352577	Bainbridge-Ropers syndrome	Disorder		77 *Cases*

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1225	Baller-Gerold syndrome	Disorder		40 *Cases*
1226	Bamforth-Lazarus syndrome	Disorder		8 *Cases*
1227	Bangstad syndrome	Disorder		2 *Cases*
1228	Banki syndrome	Disorder		1 *Family*
2995	Baraitser-Winter cerebrofrontofacial syndrome	Disorder		60 *Cases*
1231	Barber-Say syndrome	Disorder		16 *Cases*
110	Bardet-Biedl syndrome	Disorder	0.5 *BP**	
111	Barth syndrome	Disorder	0.22 *P**	
1234	Bartsocas-Papas syndrome	Disorder		24 *Cases*
112	Bartter syndrome	Disorder	0.1 *I**	
570371	Bartter syndrome type 5	Subtype of disorder		15 *Cases*
464738	Basel-Vanagaite-Smirin-Yosef syndrome	Disorder		22 *Cases*
100976	Bathing suit ichthyosis	Disorder		20 *Cases*
166113	Bazex syndrome	Disorder		145 *Cases*
113	Bazex-Dupré-Christol syndrome	Disorder		143 *Cases*
98895	Becker muscular dystrophy	Disorder	2.0 *P**	
98895	Becker muscular dystrophy	Disorder	1.53 *P*	
98895	Becker muscular dystrophy	Disorder	2.2 *BP**	
116	Beckwith-Wiedemann syndrome	Disorder	3.5 *BP**	
1237	Beemer-Ertbruggen syndrome	Disorder		2 *Cases*
1241	Bencze syndrome	Disorder		2 *Families*
324581	Benign Samaritan congenital myopathy	Disorder		4 *Cases*
251287	Benign concentric annular macular dystrophy	Disorder		27 *Cases*
1949	Benign familial neonatal epilepsy	Disorder		100 *Families*

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140927	Benign familial neonatal-infantile seizures	Disorder		10 *Families*
166308	Benign infantile focal epilepsy with midline spikes and waves during sleep	Disorder		36 *Cases*
166305	Benign infantile seizures associated with mild gastroenteritis	Disorder		100 *Cases*
209973	Benign nocturnal alternating hemiplegia of childhood	Disorder		12 *Cases*
1179	Benign paroxysmal tonic upgaze of childhood with ataxia	Disorder		12 *Cases*
71518	Benign paroxysmal torticollis of infancy	Disorder		150 *Cases*
252164	Benign schwannoma	Disorder	6.0 *P**	
274	Bernard-Soulier syndrome	Disorder		100 *Cases*
118	Beta-mannosidosis	Disorder	0.14 *BP**	
1035	Beta-mercaptolactate cysteine disulfiduria	Disorder		1 *Case*
329284	Beta-propeller protein-associated neurodegeneration	Disorder		68 *Cases*
119	Beta-sarcoglycan-related limb-girdle muscular dystrophy R4	Disorder	0.1 *P**	
848	Beta-thalassemia	Disorder	1.0 *I*	
848	Beta-thalassemia	Disorder	10.0 *I**	
65287	Beta-ureidopropionase deficiency	Disorder		5 *Cases*
69736	Bilateral acute depigmentation of the iris	Disorder		62 *Cases*
140963	Bilateral microtia-deafness-cleft palate syndrome	Disorder		3 *Families*
1980	Bilateral striopallidodentate calcinosis	Disorder		200 *Cases*
424982	Biliary cystadenocarcinoma	Disorder	0.002 *I**	
79241	Biotinidase deficiency	Disorder	1.6 *BP*	
79241	Biotinidase deficiency	Disorder	1.6 *P**	
364198	Bipartite talus	Disorder		23 *Cases*
179	Birdshot chorioretinopathy	Disorder	0.35 *P*	
122	Birt-Hogg-Dubé syndrome	Disorder	0.5 *P**	

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123	Björnstad syndrome	Disorder		33 *Cases*
93930	Bladder exstrophy	Subtype of disorder	3.05 *BP*	
86870	Blastic plasmacytoid dendritic cell neoplasm	Disorder	12.0 *P**	
73271	Bleeding diathesis due to a collagen receptor defect	Disorder		20 *Cases*
420566	Bleeding disorder due to CalDAG-GEFI deficiency	Disorder		3 *Cases*
36355	Bleeding disorder due to P2Y12 defect	Disorder		14 *Cases*
1997	Blepharo-cheilo-odontic syndrome	Disorder		55 *Cases*
1252	Blepharonasofacial malformation syndrome	Disorder		3 *Families*
2728	Blepharophimosis-intellectual disability syndrome, Ohdo type	Disorder		30 *Cases*
3047	Blepharophimosis-intellectual disability syndrome, SBBYS type	Disorder		122 *Cases*
597746	Blepharophimosis-intellectual disability syndrome/genitopatellar overlap syndrome	Disorder		122 *Cases*
2057	Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome	Disorder		6 *Cases*
1259	Blepharoptosis-myopia-ectopia lentis syndrome	Disorder		3 *Cases*
171844	Blindness-scoliosis-arachnodactyly syndrome	Disorder		4 *Cases*
50945	Blomstrand lethal chondrodysplasia	Disorder		13 *Cases*
125	Bloom syndrome	Disorder		300 *Cases*
16	Blue cone monochromatism	Disorder	1.0 *BP*	
16	Blue cone monochromatism	Disorder	1.0 *P*	
1059	Blue rubber bleb nevus	Disorder		200 *Cases*
217008	Bockenheimer syndrome	Disorder		40 *Cases*
91135	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	Disorder		11 *Cases*
97297	Bohring-Opitz syndrome	Disorder		46 *Cases*
1842	Bone dysplasia, lethal Holmgren type	Disorder		7 *Cases*

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1261	Bonnemann-Meinecke-Reich syndrome	Disorder		4 *Cases*
1263	Boomerang dysplasia	Disorder		10 *Cases*
127	Borjeson-Forssman-Lehmann syndrome	Disorder		50 *Cases*
69737	Bosley-Salih-Alorainy syndrome	Disorder		16 *Cases*
1267	Botulism	Disorder	0.022 *I**	
1270	Bowen-Conradi syndrome	Disorder		60 *Cases*
93382	Brachydactyly type A6	Disorder		7 *Cases*
93397	Brachydactyly type A7	Disorder		1 *Family*
1276	Brachydactyly-arterial hypertension syndrome	Disorder		10 *Families*
1275	Brachydactyly-elbow wrist dysplasia syndrome	Disorder		4 *Families*
2946	Brachydactyly-long thumb syndrome	Disorder		4 *Cases*
1277	Brachydactyly-mesomelia-intellectual disability-heart defects syndrome	Disorder		2 *Cases*
1246	Brachydactyly-nystagmus-cerebellar ataxia syndrome	Disorder		1 *Family*
1278	Brachydactyly-preaxial hallux varus syndrome	Disorder		8 *Cases*
166035	Brachydactyly-short stature-retinitis pigmentosa syndrome	Disorder		12 *Cases*
93409	Brachydactyly-syndactyly, Zhao type	Disorder		2 *Families*
1292	Brachymorphism-onychodysplasia-dysphalangism syndrome	Disorder		9 *Cases*
93302	Brachyolmia, Maroteaux type	Disorder		4 *Families*
1295	Brachytelephalangy-dysmorphism-Kallmann syndrome	Disorder		2 *Cases*
52047	Braddock syndrome	Disorder		2 *Cases*
75374	Bradyopsia	Disorder		5 *Cases*
178506	Brain calcification, Rajab type	Disorder		8 *Cases*
352649	Brain dopamine-serotonin vesicular transport disease	Disorder		8 *Cases*
75389	Brain malformation-congenital heart disease-postaxial polydactyly syndrome	Disorder		2 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
500150	Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	Disorder		33 *Cases*
209905	Brain-lung-thyroid syndrome	Disorder		100 *Cases*
1297	Branchio-oculo-facial syndrome	Disorder		150 *Cases*
50815	Branchiogenic deafness syndrome	Disorder		5 *Cases*
1299	Branchioskeletogenital syndrome	Disorder		7 *Cases*
90354	Brittle cornea syndrome	Disorder		65 *Cases*
70589	Bronchopulmonary dysplasia	Disorder	13.0 *P**	
79493	Brooke-Spiegler syndrome	Disorder		100 *Cases*
1304	Brucellosis	Disorder	0.09 *I**	
2771	Bruck syndrome	Disorder		60 *Cases*
130	Brugada syndrome	Disorder	20.0 *P**	
131	Budd-Chiari syndrome	Disorder	1.5 *P**	
131	Budd-Chiari syndrome	Disorder	1.1 *P*	
131	Budd-Chiari syndrome	Disorder	0.1 *I*	
36258	Buerger disease	Disorder	16.0 *P*	
36258	Buerger disease	Disorder	10.0 *P**	
280785	Bullous diffuse cutaneous mastocytosis	Subtype of disorder		40 *Cases*
703	Bullous pemphigoid	Disorder	25.0 *P**	
543	Burkitt lymphoma	Disorder	0.17 *I**	
1200	Burn-McKeown syndrome	Disorder		20 *Families*
1262	Böök syndrome	Disorder		26 *Cases*
1308	C syndrome	Disorder	0.11 *P**	
495844	C11ORF73-related autosomal recessive hypomyelinating leukodystrophy	Disorder		6 *Cases*
329918	C3 glomerulopathy	Subtype of	0.15 *I**	

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		disorder		
135	CACH syndrome	Disorder		148 *Cases*
448010	CAD-CDG	Disorder		1 *Case*
369942	CADDS	Disorder		4 *Cases*
83472	CAMOS syndrome	Disorder		5 *Cases*
71279	CANOMAD syndrome	Disorder		100 *Cases*
468684	CCDC115-CDG	Disorder		8 *Cases*
600668	CCNK-related neurodevelopmental disorder-severe intellectual disability-facial dysmorphism syndrome	Disorder		4 *Cases*
566067	CEBPE-associated autoinflammation-immunodeficiency-neutrophil dysfunction syndrome	Disorder		4 *Cases*
66631	CEDNIK syndrome	Disorder		13 *Cases*
569816	CELSR1-related late-onset primary lymphedema	Disorder		11 *Cases*
138	CHARGE syndrome	Disorder	6.5 *BP*	
138	CHARGE syndrome	Disorder	9.0 *P**	
599082	CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	Disorder		60 *Cases*
139	CHILD syndrome	Disorder		60 *Cases*
3474	CHIME syndrome	Disorder		8 *Cases*
263463	CHST3-related skeletal dysplasia	Disorder		2 *Families*
435651	CIDEC-related familial partial lipodystrophy	Disorder		1 *Case*
251383	CK syndrome	Disorder		24 *Cases*
168984	CLAPO syndrome	Disorder		6 *Cases*
485350	CLCN4-related X-linked intellectual disability syndrome	Disorder		38 *Cases*
284448	CLIPPERS	Disorder		50 *Cases*
140944	CLOVES syndrome	Disorder		150 *Cases*

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163681	CNTNAP2-related developmental and epileptic encephalopathy	Disorder		28 *Cases*
397725	COASY protein-associated neurodegeneration	Disorder		2 *Cases*
1458	CODAS syndrome	Disorder		12 *Cases*
1466	COFS syndrome	Subtype of disorder		20 *Cases*
263508	COG1-CDG	Disorder		3 *Cases*
435934	COG2-CDG	Disorder		1 *Case*
263501	COG4-CDG	Disorder		2 *Cases*
263487	COG5-CDG	Disorder		9 *Cases*
464443	COG6-CGD	Disorder		10 *Cases*
79333	COG7-CDG	Disorder		8 *Cases*
95428	COG8-CDG	Disorder		2 *Cases*
363611	CTCF-related neurodevelopmental disorder	Disorder		5 *Cases*
1310	Caffey disease	Disorder		100 *Cases*
565909	Calpain-3-related limb-girdle muscular dystrophy D4	Disorder		47 *Cases*
267	Calpain-3-related limb-girdle muscular dystrophy R1	Disorder	1.0 *P**	
85192	Calvarial doughnut lesions-bone fragility syndrome	Disorder		20 *Cases*
1318	Campomelia, Cumming type	Disorder		8 *Cases*
140	Campomelic dysplasia	Disorder	1.875 *BP*	
1319	Camptobrachydactyly	Disorder		1 *Family*
1327	Camptodactyly syndrome, Guadalajara type 1	Disorder		8 *Cases*
1326	Camptodactyly syndrome, Guadalajara type 2	Disorder		2 *Cases*
488434	Camptodactyly syndrome, Guadalajara type 3	Disorder		5 *Cases*
2848	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	Disorder		30 *Families*
1323	Camptodactyly-joint contractures-facial	Disorder		4 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	skeletal defects syndrome			
85164	Camptodactyly-tall stature-scoliosis-hearing loss syndrome	Disorder		30 *Cases*
1325	Camptodactyly-taurinuria syndrome	Disorder		17 *Cases*
1328	Camurati-Engelmann disease	Disorder		300 *Cases*
141	Canavan disease	Disorder	1.0 *BP*	
1517	Cantú syndrome	Disorder		50 *Cases*
171881	Cap myopathy	Disorder		21 *Cases*
160148	Cap polyposis	Disorder		67 *Cases*
137667	Capillary malformation-arteriovenous malformation	Disorder		261 *Cases*
418945	Carcinoma of esophagus, salivary gland type	Disorder	0.004 *I**	
137628	Cardiac anomalies-heterotaxy syndrome	Disorder		9 *Cases*
230851	Cardiac-valvular Ehlers-Danlos syndrome	Disorder		6 *Cases*
2872	Cardiocranial syndrome, Pfeiffer type	Disorder		7 *Cases*
1340	Cardiofaciocutaneous syndrome	Disorder		300 *Cases*
97292	Cardiogenic shock	Disorder	40.0 *P**	
1345	Cardiomyopathy-cataract-hip spine disease syndrome	Disorder		9 *Cases*
91130	Cardiomyopathy-hypotonia-lactic acidosis syndrome	Disorder		2 *Cases*
3238	Cardiospondylocarpofacial syndrome	Disorder		5 *Cases*
1358	Carey-Fineman-Ziter syndrome	Disorder		20 *Cases*
1359	Carney complex	Disorder		750 *Cases*
319340	Carney complex-trismus-pseudocamptodactyly syndrome	Disorder		3 *Families*
139411	Carney triad	Disorder		150 *Cases*
97286	Carney-Stratakis syndrome	Disorder		20 *Families*
156	Carnitine palmitoyl transferase 1A deficiency	Disorder		60 *Cases*
228302	Carnitine palmitoyl transferase II	Subtype of		300 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	deficiency, myopathic form	disorder		
228308	Carnitine palmitoyl transferase II deficiency, neonatal form	Subtype of disorder		20 *Families*
228305	Carnitine palmitoyl transferase II deficiency, severe infantile form	Subtype of disorder		30 *Families*
157	Carnitine palmitoyltransferase II deficiency	Disorder		300 *Cases*
157	Carnitine palmitoyltransferase II deficiency	Disorder	1.0 *P**	
159	Carnitine-acylcarnitine translocase deficiency	Disorder		60 *Cases*
1361	Carnosinase deficiency	Disorder		24 *Cases*
1361	Carnosinase deficiency	Disorder	0.2 *BP*	
53035	Caroli disease	Disorder	0.1 *I*	
65759	Carpenter syndrome	Disorder		70 *Cases*
65282	Carvajal syndrome	Disorder		7 *Cases*
195	Cat-eye syndrome	Disorder	1.35 *BP**	
50839	Cat-scratch disease	Disorder	6.6 *P**	
1373	Cataract-aberrant oral frenula-growth delay syndrome	Disorder		3 *Cases*
1368	Cataract-ataxia-deafness syndrome	Disorder		2 *Cases*
314993	Cataract-congenital heart disease-neural tube defect syndrome	Disorder		2 *Cases*
1383	Cataract-deafness-hypogonadism syndrome	Disorder		3 *Cases*
162	Cataract-glaucoma syndrome	Disorder		3 *Families*
436174	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	Disorder		3 *Cases*
1381	Cataract-intellectual disability-anal atresia-urinary defects syndrome	Disorder		3 *Cases*
1387	Cataract-intellectual disability-hypogonadism syndrome	Disorder		20 *Cases*
1377	Cataract-microcornea syndrome	Disorder		8 *Families*
1380	Cataract-nephropathy-encephalopathy syndrome	Disorder		2 *Cases*
3286	Catecholaminergic polymorphic ventricular tachycardia	Disorder	10.0 *P**	

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1388	Catel-Manzke syndrome	Disorder		33 *Cases*
1123	Caudal appendage-deafness syndrome	Disorder		2 *Cases*
1459	Celiac disease-epilepsy-cerebral calcification syndrome	Disorder		170 *Cases*
3258	Cenani-Lenz syndrome	Disorder		30 *Cases*
98972	Central cloudy dystrophy of François	Disorder		24 *Cases*
178029	Central diabetes insipidus	Disorder	4.0 *P**	
73256	Central neurocytoma	Disorder		500 *Cases*
411527	Central retinal vein occlusion	Disorder	28.0 *P**	
504476	Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome	Disorder		100 *Cases*
1171	Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome	Disorder		10 *Cases*
2246	Cerebellar hypoplasia-tapetoretinal degeneration syndrome	Disorder		3 *Cases*
444072	Cerebellar-facial-dental syndrome	Disorder		3 *Families*
46724	Cerebral arteriovenous malformation	Disorder	6.0 *P**	
136	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	Disorder	3.0 *P**	
1393	Cerebrocostomandibular syndrome	Disorder		75 *Cases*
314679	Cerebrofacioarticular syndrome	Disorder		9 *Cases*
1394	Cerebrofaciothoracic dysplasia	Disorder		20 *Cases*
66625	Cerebrooculonasal syndrome	Disorder		21 *Cases*
169079	Cernunnos-XLF deficiency	Disorder		5 *Cases*
2218	Cervical hypertrichosis-peripheral neuropathy syndrome	Disorder		4 *Cases*
46627	Char syndrome	Disorder		109 *Cases*
101101	Charcot-Marie-Tooth disease type 2B2	Disorder		1 *Family*
228374	Charcot-Marie-Tooth disease type 2B5	Disorder		4 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
101102	Charcot-Marie-Tooth disease type 2H	Disorder		13 *Cases*
300319	Charcot-Marie-Tooth disease type 2P	Disorder		18 *Cases*
397968	Charcot-Marie-Tooth disease type 2R	Disorder		1 *Case*
443073	Charcot-Marie-Tooth disease type 2S	Disorder		35 *Cases*
495274	Charcot-Marie-Tooth disease type 2T	Disorder		10 *Cases*
99955	Charcot-Marie-Tooth disease type 4B1	Disorder		11 *Families*
363981	Charcot-Marie-Tooth disease type 4B3	Disorder		3 *Cases*
99954	Charcot-Marie-Tooth disease type 4H	Disorder		15 *Cases*
139515	Charcot-Marie-Tooth disease type 4J	Disorder		18 *Cases*
90103	Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome	Disorder		7 *Cases*
1406	Charlie M syndrome	Disorder		4 *Cases*
1221	Cheilitis glandularis	Disorder		100 *Cases*
184	Cherubism	Disorder		300 *Cases*
324625	Chikungunya	Disorder	0.12 *I**	
90280	Chilblain lupus	Disorder		70 *Cases*
168782	Childhood disintegrative disorder	Disorder	2.0 *P**	
293955	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency	Disorder		5 *Cases*
363677	Childhood-onset autosomal recessive myopathy with external ophthalmoplegia	Disorder		22 *Cases*
497906	Childhood-onset basal ganglia degeneration syndrome	Disorder		4 *Cases*
494541	Childhood-onset benign chorea with striatal involvement	Disorder		3 *Cases*
500180	Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	Disorder		7 *Cases*
466921	Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome	Disorder		3 *Families*
401866	Childhood-onset spasticity with hyperglycinemia	Disorder		3 *Cases*

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137914	Choanal atresia	Disorder	8.6 *BP**	
589856	Choanal atresia-athelia-hypothyroidism-delayed puberty-short stature syndrome	Disorder		18 *Cases*
70567	Cholangiocarcinoma	Disorder	4.2 *I*	
70567	Cholangiocarcinoma	Disorder	4.0 *I**	
70567	Cholangiocarcinoma	Disorder	2.1 *P*	
1414	Cholestasis-lymphedema syndrome	Disorder		47 *Cases*
1415	Cholestasis-pigmentary retinopathy-cleft palate syndrome	Disorder		5 *Cases*
79347	Chondrodysplasia punctata, Toriello type	Disorder		3 *Cases*
280586	Chondrodysplasia with joint dislocations, gPAPP type	Disorder		4 *Cases*
1422	Chondrodysplasia-difference of sex development syndrome	Disorder		2 *Cases*
319195	Chondroectodermal dysplasia with night blindness	Disorder		4 *Cases*
404507	Chondromyxoid fibroma	Disorder		50 *Cases*
55880	Chondrosarcoma	Disorder	0.24 *I**	
55880	Chondrosarcoma	Disorder	3.55	
251899	Choroid plexus carcinoma	Disorder	0.01 *I**	
251899	Choroid plexus carcinoma	Disorder	0.35	
1433	Choroidal atrophy-alopecia syndrome	Disorder		2 *Cases*
180	Choroideremia	Disorder	2.0 *P**	
319303	Chromophobe renal cell carcinoma	Disorder	0.01 *I**	
435988	Chronic atrial and intestinal dysrhythmia syndrome	Disorder		17 *Cases*
1670	Chronic diarrhea with villous atrophy	Disorder		2 *Cases*
468641	Chronic enteropathy associated with SLCO2A1 gene	Disorder		18 *Cases*
379	Chronic granulomatous disease	Disorder	0.46 *BP*	
379	Chronic granulomatous disease	Disorder	0.5 *BP**	

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396	Chronic hiccup	Disorder	1.0 *P**	
314373	Chronic infantile diarrhea due to guanylate cyclase 2C overactivity	Disorder		32 *Cases*
2932	Chronic inflammatory demyelinating polyneuropathy	Disorder	3.7 *P**	
521	Chronic myeloid leukemia	Disorder	1.25 *I**	
521	Chronic myeloid leukemia	Disorder	5.63	
521	Chronic myeloid leukemia	Disorder	6.0 *P**	
98823	Chronic myelomonocytic leukemia	Disorder	0.68 *I*	
86830	Chronic myeloproliferative disease, unclassifiable	Disorder	0.53 *I**	
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	Disorder	0.3 *P*	
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	Disorder	2.5 *I*	
70591	Chronic thromboembolic pulmonary hypertension	Disorder	4.2 *I**	
77293	Chronic visceral acid sphingomyelinase deficiency	Disorder	0.4 *BP**	
314597	Chudley-McCullough syndrome	Disorder		25 *Cases*
71	Chylomicron retention disease	Disorder		55 *Cases*
167	Chédiak-Higashi syndrome	Disorder		500 *Cases*
69744	Circumscribed palmoplantar hypokeratosis	Disorder		17 *Cases*
309854	Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome	Disorder		20 *Cases*
247525	Citrullinemia type I	Disorder	2.4 *P**	
600731	Clark-Baraitser syndrome	Disorder		8 *Cases*
391	Classic Hodgkin lymphoma	Disorder	2.463 *I**	
391	Classic Hodgkin lymphoma	Disorder	22.9	
98846	Classic Hodgkin lymphoma, lymphocyte-depleted type	Subtype of disorder	0.04 *I**	
98845	Classic Hodgkin lymphoma, lymphocyte-rich type	Subtype of disorder	0.1 *I**	

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98844	Classic Hodgkin lymphoma, mixed cellularity type	Subtype of disorder	0.42 *I**	
98843	Classic Hodgkin lymphoma, nodular sclerosis type	Subtype of disorder	1.28 *I**	
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Disorder	7.0 *P**	
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Disorder	7.0 *BP*	
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	Subtype of disorder	7.5 *BP**	
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	Subtype of disorder	7.5 *P**	
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	Subtype of disorder	2.5 *P**	
79239	Classic galactosemia	Disorder	2.1 *I**	
71277	Classic glucose transporter type 1 deficiency syndrome	Disorder	0.538 *P*	
58017	Classic hairy cell leukemia	Disorder	0.29 *I**	
58017	Classic hairy cell leukemia	Disorder	3.12	
2584	Classic mycosis fungoides	Disorder	0.5 *I**	
329977	Classic neuroendocrine tumor of appendix	Subtype of disorder	0.25 *I*	
79254	Classic phenylketonuria	Subtype of disorder	6.0 *BP*	
79254	Classic phenylketonuria	Subtype of disorder	6.34 *BP**	
287	Classical Ehlers-Danlos syndrome	Disorder	5.0 *P*	
230839	Classical-like Ehlers-Danlos syndrome type 1	Disorder		17 *Cases*
536532	Classical-like Ehlers-Danlos syndrome type 2	Disorder		7 *Cases*
398971	Clear cell adenocarcinoma of the ovary	Disorder	0.32 *I**	
319276	Clear cell renal carcinoma	Disorder	1.99 *I**	
508476	Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-hearing loss syndrome	Disorder		7 *Cases*
1995	Cleft lip-retinopathy syndrome	Disorder		2 *Cases*

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199306	Cleft lip/palate	Disorder	80.0 *BP*	
2003	Cleft lip/palate-deafness-sacral lipoma syndrome	Disorder		2 *Cases*
3253	Cleft lip/palate-ectodermal dysplasia syndrome	Disorder		50 *Cases*
2001	Cleft lip/palate-intestinal malrotation-cardiopathy syndrome	Disorder		5 *Cases*
261190	Cleft palate-congenital heart defect-intellectual disability syndrome due to 15q14 microdeletion	Subtype of disorder		9 *Cases*
2013	Cleft palate-large ears-small head syndrome	Disorder		8 *Cases*
2016	Cleft palate-lateral synechia syndrome	Disorder		11 *Cases*
2015	Cleft palate-short stature-vertebral anomalies syndrome	Disorder		2 *Cases*
2010	Cleft palate-stapes fixation-oligodontia syndrome	Disorder		2 *Cases*
1452	Cleidocranial dysplasia	Disorder	0.1 *P*	
1452	Cleidocranial dysplasia	Disorder	0.4 *BP**	
1453	Cleidorhizomelic syndrome	Disorder		2 *Cases*
93929	Cloacal exstrophy	Subtype of disorder	0.75 *BP**	
93929	Cloacal exstrophy	Subtype of disorder	0.54 *BP*	
93267	Cloverleaf skull-multiple congenital anomalies syndrome	Disorder		3 *Cases*
352682	Cobblestone lissencephaly without muscular or ocular involvement	Disorder		6 *Cases*
90068	Cocaine intoxication	Disorder	1.0 *P**	
3233	Cochleosaccular degeneration-cataract syndrome	Disorder		2 *Families*
191	Cockayne syndrome	Disorder	0.5 *I**	
191	Cockayne syndrome	Disorder	0.2 *BP**	
192	Coffin-Lowry syndrome	Disorder	1.5 *P*	
192	Coffin-Lowry syndrome	Disorder	1.5 *P**	
1465	Coffin-Siris syndrome	Disorder		190 *Cases*
1467	Cogan syndrome	Disorder		300 *Cases*

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444077	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	Disorder		11 *Cases*
193	Cohen syndrome	Disorder		200 *Cases*
31824	Colchicine poisoning	Disorder	0.1 *P**	
157820	Cold-induced sweating syndrome	Disorder		6 *Cases*
2050	Cole-Carpenter syndrome	Disorder		3 *Cases*
1471	Coloboma of macula-brachydactyly type B syndrome	Disorder		10 *Cases*
468672	Colobomatous macrophthalmia-microcornea syndrome	Disorder		21 *Cases*
424099	Colobomatous microphthalmia-rhizomelic dysplasia syndrome	Disorder		5 *Families*
435930	Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	Disorder		3 *Cases*
1198	Colonic atresia	Disorder	5.0 *BP*	
35909	Combined deficiency of factor V and factor VIII	Disorder	0.5 *P**	
440727	Combined hamartoma of the retina and retinal pigment epithelium	Disorder		120 *Cases*
357237	Combined immunodeficiency due to CARD11 deficiency	Disorder		3 *Cases*
542301	Combined immunodeficiency due to CARMIL2 deficiency	Disorder		21 *Cases*
238505	Combined immunodeficiency due to CD27 deficiency	Disorder		18 *Cases*
538958	Combined immunodeficiency due to CD70 deficiency	Disorder		6 *Cases*
169090	Combined immunodeficiency due to CRAC channel dysfunction	Disorder		10 *Cases*
447737	Combined immunodeficiency due to DOCK2 deficiency	Disorder		5 *Cases*
217390	Combined immunodeficiency due to DOCK8 deficiency	Disorder		11 *Cases*
505227	Combined immunodeficiency due to GINS1 deficiency	Disorder		5 *Cases*
357329	Combined immunodeficiency due to IL21R deficiency	Disorder		6 *Cases*
538963	Combined immunodeficiency due to ITK deficiency	Disorder		13 *Cases*
445018	Combined immunodeficiency due to LRBA deficiency	Disorder		23 *Cases*

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397964	Combined immunodeficiency due to MALT1 deficiency	Disorder		3 *Cases*
504530	Combined immunodeficiency due to Moesin deficiency	Disorder		7 *Cases*
317428	Combined immunodeficiency due to ORAI1 deficiency	Subtype of disorder		6 *Cases*
431149	Combined immunodeficiency due to OX40 deficiency	Disorder		1 *Case*
596759	Combined immunodeficiency due to RELA haploinsufficiency	Disorder		5 *Cases*
314689	Combined immunodeficiency due to STK4 deficiency	Disorder		7 *Cases*
476113	Combined immunodeficiency due to TFRC deficiency	Disorder		2 *Families*
231154	Combined immunodeficiency due to partial RAG1 deficiency	Disorder		9 *Cases*
221139	Combined immunodeficiency with facio-oculo-skeletal anomalies	Disorder		2 *Cases*
324535	Combined oxidative phosphorylation defect type 11	Disorder		32 *Cases*
319514	Combined oxidative phosphorylation defect type 13	Disorder		2 *Cases*
319519	Combined oxidative phosphorylation defect type 14	Disorder		5 *Cases*
319524	Combined oxidative phosphorylation defect type 15	Disorder		16 *Cases*
369913	Combined oxidative phosphorylation defect type 17	Disorder		20 *Families*
254920	Combined oxidative phosphorylation defect type 2	Disorder		1 *Case*
420728	Combined oxidative phosphorylation defect type 20	Disorder		2 *Cases*
420733	Combined oxidative phosphorylation defect type 21	Disorder		2 *Cases*
444013	Combined oxidative phosphorylation defect type 23	Disorder		11 *Cases*
444458	Combined oxidative phosphorylation defect type 24	Disorder		3 *Cases*
447954	Combined oxidative phosphorylation defect type 25	Disorder		2 *Cases*
477684	Combined oxidative phosphorylation defect type 26	Disorder		2 *Cases*
477774	Combined oxidative phosphorylation defect type 27	Disorder		3 *Cases*
478029	Combined oxidative phosphorylation defect type 29	Disorder		1 *Case*
478042	Combined oxidative phosphorylation defect type 30	Disorder		2 *Cases*

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565624	Combined oxidative phosphorylation defect type 39	Disorder		6 *Cases*
254925	Combined oxidative phosphorylation defect type 4	Disorder		2 *Cases*
254930	Combined oxidative phosphorylation defect type 7	Disorder		7 *Cases*
319504	Combined oxidative phosphorylation defect type 8	Disorder		7 *Cases*
319509	Combined oxidative phosphorylation defect type 9	Disorder		4 *Cases*
309111	Combined pancreatic lipase-colipase deficiency	Disorder		3 *Cases*
280133	Complement component 3 deficiency	Disorder		27 *Cases*
99429	Complete androgen insensitivity syndrome	Disorder	3.0 *I**	
99429	Complete androgen insensitivity syndrome	Disorder	0.83 *P*	
1329	Complete atrioventricular septal defect	Disorder	20.0 *BP**	
98949	Complete cryptophthalmia	Subtype of disorder		15 *Cases*
457378	Complex lethal osteochondrodysplasia	Disorder		6 *Cases*
306644	Complication after organ transplantation	Disorder	9.0 *P**	
268316	Complication in hemodialysis	Disorder	13.0 *I**	
90053	Complications after hematopoietic stem cell transplantation	Disorder	0.65 *P**	
458758	Composite hemangioendothelioma	Disorder		39 *Cases*
168966	Composite lymphoma	Disorder	0.01 *I**	
3216	Conductive deafness-malformed external ear syndrome	Disorder		8 *Cases*
3236	Conductive deafness-ptosis-skeletal anomalies syndrome	Disorder		3 *Cases*
209932	Cone dystrophy with supernormal rod response	Disorder		45 *Cases*
1872	Cone rod dystrophy	Disorder	2.5 *P**	
221142	Confetti-like macular atrophy	Disorder		2 *Cases*
294975	Congenital absence of upper arm and forearm with hand present	Disorder	0.62 *BP*	
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral	Disorder		2 *Families*

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90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	Disorder	0.75 *BP**	
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	Disorder	0.47 *P**	
90793	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	Disorder	0.1 *P**	
90791	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	Disorder		68 *Cases*
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	Disorder	0.75 *BP**	
495879	Congenital agenesis of the scrotum	Disorder		6 *Cases*
79	Congenital alpha2-antiplasmin deficiency	Disorder		40 *Cases*
210122	Congenital alveolar capillary dysplasia	Disorder		40 *Cases*
3319	Congenital amegakaryocytic thrombocytopenia	Disorder		100 *Cases*
86816	Congenital analbuminemia	Disorder		50 *Cases*
1195	Congenital atransferrinemia	Disorder		16 *Cases*
566192	Congenital autosomal recessive small-platelet thrombocytopenia	Disorder		5 *Cases*
538101	Congenital axonal neuropathy with encephalopathy	Disorder		7 *Cases*
48	Congenital bilateral absence of vas deferens	Disorder	50.0 *P**	
79302	Congenital bile acid synthesis defect type 3	Disorder		2 *Cases*
79095	Congenital bile acid synthesis defect type 4	Disorder		5 *Cases*
514352	Congenital brachyesophagus-intrathoracic stomach-vertebral anomalies syndrome	Disorder		8 *Cases*
71278	Congenital brain dysgenesis due to glutamine synthetase deficiency	Disorder		3 *Cases*
300313	Congenital cataract-hearing loss-severe developmental delay syndrome	Disorder		5 *Cases*
1369	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome	Disorder		40 *Cases*
330054	Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome	Disorder		3 *Cases*
521432	Congenital cataract-severe neonatal	Disorder		2 *Cases*

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	hepatopathy-global developmental delay syndrome			
48431	Congenital cataracts-facial dysmorphism-neuropathy syndrome	Disorder		170 *Cases*
512260	Congenital cerebellar ataxia due to RNU12 mutation	Disorder		6 *Cases*
329242	Congenital chronic diarrhea with protein-losing enteropathy	Disorder		2 *Cases*
168612	Congenital deficiency in alpha-fetoprotein	Disorder		22 *Cases*
2140	Congenital diaphragmatic hernia	Disorder	30.0 *BP*	
2140	Congenital diaphragmatic hernia	Disorder	21.2 *BP**	
98870	Congenital dyserythropoietic anemia type III	Disorder		60 *Cases*
293825	Congenital dyserythropoietic anemia type IV	Disorder		4 *Cases*
103910	Congenital enterocyte heparan sulfate deficiency	Disorder		3 *Cases*
231573	Congenital erosive and vesicular dermatosis	Disorder		31 *Cases*
79277	Congenital erythropoietic porphyria	Disorder		200 *Cases*
79277	Congenital erythropoietic porphyria	Disorder	0.065 *I**	
325	Congenital factor II deficiency	Disorder	0.05 *P**	
326	Congenital factor V deficiency	Disorder	0.1 *P**	
327	Congenital factor VII deficiency	Disorder	0.33 *P**	
329	Congenital factor XI deficiency	Disorder	0.1 *P**	
331	Congenital factor XIII deficiency	Disorder	0.04 *I**	
331	Congenital factor XIII deficiency	Disorder	0.05 *P**	
335	Congenital fibrinogen deficiency	Disorder	0.15 *P**	
476406	Congenital generalized hypercontractile muscle stiffness syndrome	Disorder		2 *Cases*
1023	Congenital generalized hypertrichosis, Ambras type	Subtype of disorder		40 *Cases*
528	Congenital generalized lipodystrophy	Disorder	0.5 *P**	
98976	Congenital glaucoma	Disorder	2.2 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
60041	Congenital heart block	Disorder	4.54 *BP*	
1355	Congenital heart defect-round face-developmental delay syndrome	Disorder		3 *Cases*
98975	Congenital hereditary endothelial dystrophy type I	Disorder		68 *Cases*
306530	Congenital hereditary facial paralysis-variable hearing loss syndrome	Disorder		13 *Cases*
2185	Congenital hydrocephalus	Disorder	46.5 *BP**	
95715	Congenital hypothyroidism due to transplacental passage of TSH-binding inhibitory antibodies	Disorder	1.0 *P**	
79394	Congenital ichthyosiform erythroderma	Disorder	0.3 *P**	
352333	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome	Disorder		2 *Cases*
2271	Congenital ichthyosis-microcephalus-tetraplegia syndrome	Disorder		2 *Cases*
583097	Congenital infiltrating lipomatosis of the face	Disorder		59 *Cases*
453510	Congenital insensitivity to pain with severe intellectual disability	Disorder		3 *Cases*
88642	Congenital insensitivity to pain-anosmia-neuropathic arthropathy	Disorder		20 *Cases*
217399	Congenital insensitivity to pain-hyperhidrosis-absence of cutaneous sensory innervation	Disorder		2 *Cases*
1229	Congenital intrauterine infection-like syndrome	Disorder		30 *Cases*
332	Congenital intrinsic factor deficiency	Disorder		100 *Cases*
495875	Congenital labioscrotal agenesis-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome	Disorder		3 *Cases*
1954	Congenital lethal erythroderma	Disorder		17 *Cases*
210163	Congenital lethal myopathy, Compton-North type	Disorder		4 *Cases*
562528	Congenital limbs-face contractures-hypotonia-developmental delay syndrome	Disorder		14 *Cases*
1928	Congenital lobar emphysema	Disorder	4.0 *BP*	
93109	Congenital megacalycosis	Disorder		25 *Cases*
69063	Congenital membranous nephropathy due to fetomaternal anti-neutral	Disorder		15 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	endopeptidase alloimmunization			
391376	Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome	Disorder		20 *Cases*
157973	Congenital muscular dystrophy due to LMNA mutation	Disorder		23 *Cases*
98893	Congenital muscular dystrophy type 1B	Disorder		6 *Cases*
371007	Congenital muscular dystrophy with hyperlaxity	Disorder		14 *Cases*
34520	Congenital muscular dystrophy with integrin alpha-7 deficiency	Disorder	0.03 *P**	
329178	Congenital muscular dystrophy with intellectual disability and severe epilepsy	Disorder		3 *Cases*
1875	Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome	Disorder		7 *Cases*
486815	Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome	Disorder		4 *Cases*
590	Congenital myasthenic syndrome	Disorder	0.3 *P**	
319160	Congenital myopathy with internal nuclei and atypical cores	Disorder		5 *Cases*
424107	Congenital myopathy with myasthenic-like onset	Disorder		2 *Cases*
544602	Congenital myopathy with reduced type 2 muscle fibers	Disorder		2 *Cases*
199329	Congenital myopathy, Paradas type	Disorder		2 *Cases*
168486	Congenital neuronal ceroid lipofuscinosis	Disorder		10 *Cases*
369852	Congenital neutropenia-myelofibrosis-nephromegaly syndrome	Disorder		16 *Cases*
2772	Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome	Disorder		3 *Cases*
313906	Congenital pancreatic cyst	Disorder		10 *Cases*
139414	Congenital panfollicular nevus	Disorder		3 *Cases*
569821	Congenital primary lymphedema of Gordon	Disorder		23 *Cases*
508542	Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome	Disorder		5 *Cases*
66630	Congenital pseudoarthrosis of the clavicle	Disorder		200 *Cases*
2444	Congenital pulmonary airway malformation	Disorder	8.2 *BP**	

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2414	Congenital pulmonary lymphangiectasia	Disorder		100 *Cases*
3269	Congenital radioulnar synostosis	Disorder		350 *Cases*
2040	Congenital respiratory-biliary fistula	Disorder		35 *Cases*
281190	Congenital reticular ichthyosiform erythroderma	Disorder		40 *Cases*
290	Congenital rubella syndrome	Disorder	0.03 *I**	
290	Congenital rubella syndrome	Disorder	0.35 *BP**	
2301	Congenital short bowel syndrome	Disorder		43 *Cases*
369861	Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome	Disorder		16 *Cases*
103908	Congenital sodium diarrhea	Disorder		50 *Cases*
101068	Congenital stromal corneal dystrophy	Disorder		6 *Families*
35122	Congenital sucrase-isomaltase deficiency	Disorder	20.0 *P**	
499009	Congenital syphilis	Disorder	1.3 *BP**	
93583	Congenital thrombotic thrombocytopenic purpura	Subtype of disorder		123 *Cases*
99125	Congenital total pulmonary venous return anomaly	Disorder	9.0 *BP*	
858	Congenital toxoplasmosis	Disorder	33.0 *BP**	
92050	Congenital tufting enteropathy	Disorder	0.5 *BP**	
291	Congenital varicella syndrome	Disorder		130 *Cases*
521438	Congenital vertebral-cardiac-renal anomalies syndrome	Disorder		4 *Cases*
216694	Congenitally corrected transposition of the great arteries	Disorder	3.0 *BP*	
2391	Congenitally short costocoracoid ligament	Disorder		1 *Family*
860	Congenitally uncorrected transposition of the great arteries	Disorder	24.25 *BP**	
300284	Connective tissue disorder due to lysyl hydroxylase-3 deficiency	Disorder		2 *Cases*
420794	Cono-spondylar dysplasia	Disorder		3 *Cases*
319651	Constitutional megaloblastic anemia with	Disorder		6 *Cases*

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	severe neurologic disease			
436003	Contractures-developmental delay-Pierre Robin syndrome	Disorder		6 *Cases*
1484	Contractures-ectodermal dysplasia-cleft lip/palate syndrome	Disorder		2 *Cases*
314002	Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome	Disorder		2 *Cases*
1487	Cooks syndrome	Disorder		12 *Cases*
1488	Cooper-Jabs syndrome	Disorder		2 *Cases*
1490	Corneal dystrophy-perceptive deafness syndrome	Disorder		24 *Cases*
352662	Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome	Disorder		19 *Cases*
199	Cornelia de Lange syndrome	Disorder	1.24 *BP**	
3194	Corneodermatoosseous syndrome	Disorder		7 *Cases*
52055	Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome	Disorder		2 *Cases*
459074	Corpus callosum agenesis-macrocephaly-hypertelorism syndrome	Disorder		4 *Cases*
1389	Cortical blindness-intellectual disability-polydactyly syndrome	Disorder		3 *Cases*
300570	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation	Disorder		12 *Cases*
54251	Corticosteroid-sensitive aseptic abscess syndrome	Disorder		49 *Cases*
3071	Costello syndrome	Disorder		300 *Cases*
1508	Coxoauricular syndrome	Disorder		4 *Cases*
1509	Coxopodopatellar syndrome	Disorder		47 *Cases*
1512	Crane-Heise syndrome	Disorder		9 *Cases*
1525	Cranio-osteoarthropathy	Disorder		30 *Cases*
1513	Craniodiaphyseal dysplasia	Disorder		20 *Cases*
1514	Craniodigital-intellectual disability syndrome	Disorder		5 *Cases*
1515	Cranioectodermal dysplasia	Disorder		60 *Cases*
85168	Craniofacial conodysplasia	Disorder		1 *Family*

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459061	Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome	Disorder		8 *Cases*
1529	Craniofacial-deafness-hand syndrome	Disorder		3 *Cases*
363705	Craniofaciofrontodigital syndrome	Disorder		4 *Cases*
1521	Craniofrontonasal dysplasia-Poland anomaly syndrome	Disorder		2 *Cases*
50814	Craniolenticulosutural dysplasia	Disorder		28 *Cases*
85184	Craniometadiaphyseal dysplasia, wormian bone type	Disorder		4 *Cases*
1522	Craniometaphyseal dysplasia	Disorder		160 *Cases*
54595	Craniopharyngioma	Disorder	1.0 *I*	
54595	Craniopharyngioma	Disorder	2.0 *P**	
157832	Craniorhiny	Disorder		4 *Cases*
1541	Craniosynostosis, Boston type	Disorder		3 *Families*
2145	Craniosynostosis, Herrmann-Opitz type	Disorder		2 *Cases*
1527	Craniosynostosis, Philadelphia type	Disorder		1 *Family*
1538	Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome	Disorder		4 *Cases*
85199	Craniosynostosis-anal anomalies-porokeratosis syndrome	Disorder		9 *Cases*
171839	Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome	Disorder		2 *Cases*
52054	Craniosynostosis-intracranial calcifications syndrome	Disorder		3 *Cases*
565858	Craniosynostosis-microretrognathia-severe intellectual disability syndrome	Disorder		3 *Cases*
1528	Craniotelencephalic dysplasia	Disorder		4 *Cases*
205	Crigler-Najjar syndrome	Disorder	0.1 *BP**	
205	Crigler-Najjar syndrome	Disorder	1.0 *P**	
1545	Crisponi syndrome	Disorder		30 *Cases*
1461	Criss-cross heart	Disorder	0.8 *BP**	

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2930	Cronkhite-Canada syndrome	Disorder		500 *Cases*
2935	Crossed polysyndactyly	Disorder		12 *Cases*
207	Crouzon syndrome	Disorder	0.9 *BP**	
93262	Crouzon syndrome-acanthosis nigricans syndrome	Disorder	0.1 *BP*	
1546	Cryptococcosis	Disorder	11.0 *I**	
468635	Cryptogenic multifocal ulcerous stenosing enteritis	Disorder		60 *Cases*
1547	Cryptomicrotia-brachydactyly-excess fingertip arch syndrome	Disorder		2 *Cases*
1548	Cryptorchidism-arachnodactyly-intellectual disability syndrome	Disorder		3 *Cases*
307766	Curly hair-acral keratoderma-caries syndrome	Disorder		14 *Cases*
1552	Currarino syndrome	Disorder	1.0 *P**	
1553	Curry-Jones syndrome	Disorder		9 *Cases*
96253	Cushing disease	Disorder	4.0 *P**	
96253	Cushing disease	Disorder	0.2 *I**	
189427	Cushing syndrome due to bilateral macronodular adrenocortical disease	Disorder	0.08 *P**	
280779	Cutaneous collagenous vasculopathy	Disorder		20 *Cases*
79140	Cutaneous neuroendocrine carcinoma	Disorder	0.27 *I*	
79140	Cutaneous neuroendocrine carcinoma	Disorder	0.13 *I**	
79140	Cutaneous neuroendocrine carcinoma	Disorder	4.0 *P**	
79140	Cutaneous neuroendocrine carcinoma	Disorder	0.86	
2881	Cutaneous photosensitivity-lethal colitis syndrome	Disorder		3 *Cases*
451607	Cutaneous pseudolymphoma	Disorder		60 *Cases*
1555	Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome	Disorder		12 *Cases*
221145	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies	Disorder		21 *Cases*
171719	Cutis laxa-Marfanoid syndrome	Disorder		18 *Cases*

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1556	Cutis marmorata telangiectatica congenita	Disorder		300 *Cases*
2686	Cyclic neutropenia	Disorder	0.1 *P**	
2674	Cyprus facial-neuromusculoskeletal syndrome	Disorder		1 *Family*
400	Cystic echinococcosis	Disorder	1.0 *I**	
586	Cystic fibrosis	Disorder	19.3912 *BP**	
586	Cystic fibrosis	Disorder	11.1319 *P**	
2575	Cystic fibrosis-gastritis-megaloblastic anemia syndrome	Disorder		2 *Cases*
2111	Cystic hamartoma of lung and kidney	Disorder		3 *Cases*
85136	Cystic leukoencephalopathy without megalencephaly	Disorder		50 *Cases*
213	Cystinosis	Disorder	0.75 *BP*	
213	Cystinosis	Disorder	1.5 *P**	
213	Cystinosis	Disorder	0.5 *BP**	
214	Cystinuria	Disorder	14.0 *P*	
214	Cystinuria	Disorder	5.0 *P**	
75381	Cystoid macular dystrophy	Disorder		97 *Cases*
137698	Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	Disorder	25.5 *P**	
94087	Cytophagic histiocytic panniculitis	Disorder		100 *Cases*
477787	Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder	Disorder		2 *Cases*
2437	Czeizel-Losonci syndrome	Disorder		3 *Cases*
356978	D,L-2-hydroxyglutaric aciduria	Disorder		13 *Cases*
79315	D-2-hydroxyglutaric aciduria	Disorder		80 *Cases*
300536	DDOST-CDG	Disorder		1 *Case*
488647	DDX41-related hematologic malignancy predisposition syndrome	Disorder		3 *Families*

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79134	DEND syndrome	Disorder		40 *Cases*
494444	DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	Disorder		8 *Cases*
284343	DICER1 tumor-predisposition syndrome	Disorder	0.007 *I*	
404546	DITRA	Disorder		70 *Cases*
91131	DK1-CDG	Disorder		17 *Cases*
352470	DNA2-related mitochondrial DNA deletion syndrome	Disorder		4 *Cases*
443950	DNAJB2-related Charcot-Marie-Tooth disease type 2	Disorder		2 *Cases*
34516	DNAJB6-related limb-girdle muscular dystrophy D1	Disorder		6 *Families*
330050	DNM1L-related encephalopathy due to mitochondrial and peroxisomal fission defect	Subtype of disorder		11 *Cases*
572761	DONSON-related microcephaly-short stature-limb abnormalities spectrum	Disorder		51 *Cases*
79500	DOORS syndrome	Disorder		50 *Cases*
86309	DPAGT1-CDG	Disorder		18 *Cases*
79322	DPM1-CDG	Disorder		9 *Cases*
263494	DPM3-CDG	Disorder		1 *Case*
209341	DYNC1H1-related autosomal dominant childhood-onset proximal spinal muscular atrophy	Subtype of disorder		37 *Cases*
464306	DYRK1A-related intellectual disability syndrome	Disorder		54 *Cases*
268261	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion	Subtype of disorder		19 *Cases*
1562	Dacryocystitis-osteopoikilosis syndrome	Disorder		5 *Cases*
1563	Dahlberg-Borer-Newcomer syndrome	Disorder		2 *Cases*
1566	Dandy-Walker malformation-postaxial polydactyly syndrome	Disorder		5 *Cases*
218	Darier disease	Disorder	3.4 *P**	
2962	De Barsy syndrome	Disorder		40 *Cases*
3214	Deaf blind hypopigmentation syndrome,	Disorder		2 *Cases*

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	Yemenite type			
90024	Deafness with labyrinthine aplasia, microtia, and microdontia	Disorder		56 *Cases*
3241	Deafness-craniofacial syndrome	Disorder		2 *Cases*
3232	Deafness-ear malformation-facial palsy syndrome	Disorder		4 *Cases*
3220	Deafness-enamel hypoplasia-nail defects syndrome	Disorder		15 *Families*
254898	Deafness-encephaloneuropathy-obesity-valvulopathy syndrome	Disorder		2 *Cases*
3218	Deafness-epiphyseal dysplasia-short stature syndrome	Disorder		2 *Cases*
3224	Deafness-genital anomalies-metacarpal and metatarsal synostosis syndrome	Disorder		2 *Cases*
94064	Deafness-infertility syndrome	Disorder		3 *Families*
85321	Deafness-intellectual disability syndrome, Martin-Probst type	Disorder		3 *Cases*
3226	Deafness-lymphedema-leukemia syndrome	Disorder		20 *Cases*
3230	Deafness-oligodontia syndrome	Disorder		5 *Cases*
3217	Deafness-small bowel diverticulosis-neuropathy syndrome	Disorder		5 *Cases*
3239	Deafness-vitiligo-achalasia syndrome	Disorder		2 *Cases*
99970	Dedifferentiated liposarcoma	Subtype of disorder	0.27 *I**	
293978	Deficiency in anterior pituitary function-variable immunodeficiency syndrome	Disorder		7 *Cases*
3202	Dehydrated hereditary stomatocytosis	Disorder		20 *Families*
3034	Delayed membranous cranial ossification	Disorder		2 *Families*
3038	Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome	Disorder		6 *Cases*
1627	Deletion 5q35	Disorder		10 *Cases*
219	Delta-sarcoglycan-related limb-girdle muscular dystrophy R6	Disorder	0.3 *P**	
99828	Dengue fever	Disorder	714.0 *I*	
99828	Dengue fever	Disorder	0.5 *I**	
93571	Dense deposit disease	Subtype of disorder	0.25 *P*	
1652	Dent disease	Disorder		250 *Families*

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99789	Dentin dysplasia type I	Subtype of disorder	1.0 *P**	
99791	Dentin dysplasia type II	Subtype of disorder		19 *Families*
99792	Dentin dysplasia-sclerotic bones syndrome	Disorder		1 *Family*
49042	Dentinogenesis imperfecta	Disorder	14.5 *P**	
166260	Dentinogenesis imperfecta type 2	Subtype of disorder	14.6 *P**	
71267	Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome	Disorder		2 *Cases*
220	Denys-Drash syndrome	Disorder		300 *Cases*
1656	Dermatitis herpetiformis	Disorder	27.0 *P**	
31112	Dermatofibrosarcoma protuberans	Disorder	10.0 *P**	
1659	Dermatoleukodystrophy	Disorder		2 *Cases*
221	Dermatomyositis	Disorder	0.9704 *I*	
221	Dermatomyositis	Disorder	7.5312 *P*	
1657	Dermatoosteolysis, Kirghizian type	Disorder		5 *Cases*
86920	Dermatopathia pigmentosa reticularis	Disorder		20 *Cases*
1901	Dermatosparaxis Ehlers-Danlos syndrome	Disorder		15 *Cases*
79149	Dermochondrocorneal dystrophy	Disorder		15 *Cases*
1660	Dermoodontodysplasia	Disorder		11 *Cases*
1425	Desbuquois syndrome	Disorder		50 *Cases*
84132	Desmin-related myopathy with Mallory body-like inclusions	Disorder		5 *Cases*
873	Desmoid tumor	Disorder	0.3 *I**	
83469	Desmoplastic small round cell tumor	Disorder		300 *Cases*
251863	Desmoplastic/nodular medulloblastoma	Subtype of disorder	0.01 *I**	
35107	Desmosterolosis	Disorder		10 *Cases*

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313892	Developmental and speech delay due to SOX5 deficiency	Subtype of disorder		14 *Cases*
329195	Developmental delay with autism spectrum disorder and gait instability	Disorder		22 *Cases*
369891	Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	Disorder		70 *Cases*
79107	Developmental malformations-deafness-dystonia syndrome	Disorder		2 *Cases*
124	Diamond-Blackfan anemia	Disorder	0.67 *BP**	
275523	Dianzani autoimmune lymphoproliferative disease	Disorder		30 *Cases*
66637	Diaphanospondylodysostosis	Disorder		18 *Cases*
2141	Diaphragmatic defect-limb deficiency-skull defect syndrome	Disorder		7 *Cases*
527468	Diaphragmatic hernia-short bowel-asplenia syndrome	Disorder		2 *Cases*
628	Diastrophic dysplasia	Disorder	1.2 *P**	
628	Diastrophic dysplasia	Disorder	0.3 *BP**	
370046	Didymosis aplasticosebacea	Disorder		18 *Cases*
2983	Difference of sex development-intellectual disability syndrome	Disorder		3 *Cases*
146	Differentiated thyroid carcinoma	Disorder	5.25 *I*	
146	Differentiated thyroid carcinoma	Disorder	2.0 *I**	
90060	Diffuse alveolar hemorrhage	Disorder	1.0 *P**	
404437	Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	Disorder		4 *Cases*
79456	Diffuse cutaneous mastocytosis	Disorder		30 *Cases*
2123	Diffuse neonatal hemangiomatosis	Disorder		70 *Cases*
2337	Diffuse palmoplantar keratoderma, Bothnian type	Disorder	2.5 *P**	
86918	Diffuse palmoplantar keratoderma-acrocyanosis syndrome	Disorder		10 *Cases*
2926	Digital extensor muscle aplasia-polyneuropathy	Disorder		3 *Cases*
226	Dihydropteridine reductase deficiency	Subtype of disorder		150 *Cases*
2229	Dilated cardiomyopathy-	Disorder		20 *Families*

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	hypergonadotropic hypogonadism syndrome			
243343	Dimethylglycine dehydrogenase deficiency	Disorder		1 *Case*
227	Diphallia	Disorder	0.02 *BP*	
1681	Diprosopus	Disorder		33 *Cases*
2412	Dislocation of the hip-dysmorphism syndrome	Disorder		4 *Cases*
71274	Disseminated peritoneal leiomyomatosis	Disorder		150 *Cases*
319171	Distal 17p13.1 microdeletion syndrome	Disorder		16 *Cases*
261257	Distal 17p13.3 microdeletion syndrome	Disorder		16 *Cases*
254351	Distal 7q11.23 microdeletion syndrome	Disorder		41 *Cases*
261102	Distal 7q11.23 microduplication syndrome	Disorder		5 *Cases*
293939	Distal Xq28 microduplication syndrome	Disorder		9 *Cases*
399096	Distal anoctaminopathy	Disorder		24 *Cases*
251515	Distal arthrogryposis type 10	Disorder		53 *Cases*
329457	Distal arthrogryposis type 5D	Disorder		33 *Cases*
96148	Distal deletion 10q	Disorder		40 *Cases*
280325	Distal deletion 12p	Disorder		8 *Cases*
1590	Distal deletion 13q	Disorder		150 *Cases*
1596	Distal deletion 15q	Disorder		30 *Cases*
1620	Distal deletion 3p	Disorder		34 *Cases*
96125	Distal deletion 6p	Disorder		35 *Cases*
1642	Distal deletion 9p	Disorder		89 *Cases*
96102	Distal duplication 10q	Disorder		40 *Cases*
1745	Distal duplication 6p	Disorder		40 *Cases*
139525	Distal hereditary motor neuropathy type 2	Disorder		4 *Families*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
139552	Distal hereditary motor neuropathy, Jerash type	Disorder		30 *Cases*
1307	Distal limb deficiencies-micrognathia syndrome	Disorder		6 *Cases*
178400	Distal myopathy with anterior tibial onset	Disorder		4 *Cases*
63273	Distal myopathy with posterior leg and anterior hand involvement	Disorder		16 *Cases*
488650	Distal myopathy, Tateyama type	Disorder		7 *Cases*
399103	Distal nebulin myopathy	Disorder		13 *Cases*
139547	Distal spinal muscular atrophy type 3	Disorder		28 *Cases*
3248	Distal symphalangism	Disorder		8 *Families*
314588	Distal triplication 15q	Subtype of disorder		23 *Cases*
3262	Dobrow syndrome	Disorder		2 *Cases*
244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis	Disorder		12 *Cases*
2143	Donnai-Barrow syndrome	Disorder		50 *Cases*
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency	Disorder		43 *Cases*
230	Dopamine beta-hydroxylase deficiency	Disorder		25 *Cases*
3427	Double outlet left ventricle	Disorder	0.5 *BP*	
3411	Double uterus-hemivagina-renal agenesis syndrome	Disorder		60 *Cases*
870	Down syndrome	Disorder	95.0 *BP*	
870	Down syndrome	Disorder	57.0 *P**	
870	Down syndrome	Disorder	101.0 *BP**	
33069	Dravet syndrome	Disorder	3.3 *BP**	
50817	Duane anomaly-myopathy-scoliosis syndrome	Disorder		2 *Cases*
233	Duane retraction syndrome	Disorder	10.0 *P**	
529574	Duane retraction syndrome with congenital deafness	Disorder		4 *Cases*
235	Dubowitz syndrome	Disorder	0.2 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
98896	Duchenne muscular dystrophy	Disorder	9.9 *BP*	
98896	Duchenne muscular dystrophy	Disorder	2.8 *P*	
1203	Duodenal atresia	Disorder	9.0 *BP**	
1203	Duodenal atresia	Disorder	9.0 *P**	
314621	Duplication of the pituitary gland	Disorder		38 *Cases*
237	Duplication of urethra	Disorder		300 *Cases*
239	Dyggve-Melchior-Clausen disease	Disorder		100 *Cases*
412	Dysbetalipoproteinemia	Disorder	7.8 *P**	
412	Dysbetalipoproteinemia	Disorder	10.0 *P*	
1765	Dyschondrosteosis-nephritis syndrome	Disorder		1 *Family*
41	Dyschromatosis symmetrica hereditaria	Disorder		300 *Cases*
1766	Dysequilibrium syndrome	Disorder		51 *Cases*
1775	Dyskeratosis congenita	Disorder	0.1 *P**	
2104	Dysmorphism-pectus carinatum-joint laxity syndrome	Disorder		2 *Cases*
2282	Dysmorphism-short stature-deafness-difference of sex development syndrome	Disorder		2 *Cases*
1782	Dysosteosclerosis	Disorder		23 *Cases*
1822	Dysplasia epiphysealis hemimelica	Disorder	0.1 *I*	
2204	Dysplastic cortical hyperostosis, Kozlowski-Tsuruta type	Subtype of disorder		2 *Cases*
2476	Dysraphism-cleft lip/palate-limb reduction defects syndrome	Disorder		3 *Cases*
85198	Dysspondyloenchondromatosis	Disorder		16 *Cases*
210571	Dystonia 16	Disorder		12 *Cases*
589618	Dystonia 28	Disorder		160 *Cases*
412217	Dystonia-aphonia syndrome	Disorder		32 *Cases*
521406	Dystonia-parkinsonism-hypermanganesemia syndrome	Disorder		11 *Cases*

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89843	Dystrophic epidermolysis bullosa pruriginosa	Disorder		100 *Families*
199343	EAST syndrome	Disorder		26 *Cases*
293936	EDICT syndrome	Disorder		4 *Families*
1896	EEC syndrome	Disorder	1.11 *BP**	
1897	EEM syndrome	Disorder		7 *Families*
485418	EMILIN-1-related connective tissue disease	Disorder		3 *Cases*
568065	EPHB4-related lymphatic-related hydrops fetalis	Disorder		2 *Families*
496751	EVEN-plus syndrome	Disorder		3 *Cases*
2554	Ear-patella-short stature syndrome	Disorder		67 *Cases*
1935	Early myoclonic encephalopathy	Disorder		80 *Cases*
324290	Early-onset Lafora body disease	Disorder		3 *Cases*
98890	Early-onset X-linked optic atrophy	Disorder		4 *Families*
556985	Early-onset calcifying leukoencephalopathy-skeletal dysplasia	Disorder		13 *Cases*
488635	Early-onset epilepsy-intellectual disability-brain anomalies syndrome	Disorder		5 *Cases*
411986	Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	Disorder		3 *Cases*
256	Early-onset generalized limb-onset dystonia	Disorder	0.4 *P**	
439212	Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome	Disorder		13 *Cases*
2379	Early-onset parkinsonism-intellectual disability syndrome	Disorder		2 *Families*
496641	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	Disorder		39 *Cases*
1943	Early-onset progressive encephalopathy with migrant continuous myoclonus	Disorder		3 *Cases*
500144	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	Disorder		5 *Cases*
496756	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome	Disorder		6 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
3240	Early-onset progressive leukoencephalopathy-central nervous system calcification-deafness-visual impairment syndrome	Disorder		2 *Cases*
352654	Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	Disorder		6 *Cases*
505237	Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome	Disorder		12 *Cases*
313772	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	Disorder		2 *Cases*
391320	East Texas bleeding disorder	Subtype of disorder		19 *Cases*
319218	Ebola hemorrhagic fever	Disorder		28220 *Cases*
1880	Ebstein malformation of the tricuspid valve	Disorder	1.25 *P**	
1880	Ebstein malformation of the tricuspid valve	Disorder	3.5 *BP**	
69083	Ectodermal dysplasia with natal teeth, Turnpenny type	Disorder		1 *Family*
1818	Ectodermal dysplasia, trichoodontoonychial type	Disorder		7 *Cases*
1806	Ectodermal dysplasia-blindness syndrome	Disorder		2 *Cases*
247827	Ectodermal dysplasia-hyperhidrosis-cutaneous syndactyly syndrome	Disorder		4 *Cases*
247820	Ectodermal dysplasia-pili torti-cutaneous syndactyly syndrome	Disorder		22 *Cases*
1883	Ectodermal dysplasia-sensorineural deafness syndrome	Disorder		2 *Cases*
448270	Ectopia cordis	Disorder	0.67 *BP*	
1884	Ectopia lentis-chorioretinal dystrophy-myopia syndrome	Disorder		8 *Cases*
1892	Ectrodactyly-polydactyly syndrome	Disorder		1 *Family*
1895	Edinburgh malformation syndrome	Disorder		2 *Families*
1902	Ehrlichiosis	Disorder		50 *Cases*
79106	Eiken syndrome	Disorder		6 *Cases*
228240	Elastoderma	Disorder		5 *Cases*
289	Ellis Van Creveld syndrome	Disorder	0.4 *BP**	

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289	Ellis Van Creveld syndrome	Disorder	1.1 *BP*	
96170	Emanuel syndrome	Disorder		350 *Cases*
180226	Embryonal carcinoma	Disorder	0.01 *I**	
261	Emery-Dreifuss muscular dystrophy	Disorder	0.3 *P**	
1927	Emery-Nelson syndrome	Disorder		2 *Cases*
1031	Enamel-renal syndrome	Disorder		11 *Cases*
2396	Encephalocraniocutaneous lipomatosis	Disorder		77 *Cases*
527276	Encephalopathy due to mitochondrial and peroxisomal fission defect	Disorder		15 *Cases*
139406	Encephalopathy due to prosaposin deficiency	Disorder		10 *Cases*
833	Encephalopathy due to sulfite oxidase deficiency	Disorder		100 *Cases*
319678	Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	Disorder		1 *Case*
199332	Endocrine-cerebro-osteodysplasia syndrome	Disorder		7 *Cases*
454723	Endometrioid carcinoma of ovary	Disorder	0.81 *I**	
2790	Endosteal hyperostosis, Worth type	Disorder		6 *Families*
85186	Endosteal sclerosis-cerebellar hypoplasia syndrome	Disorder		4 *Cases*
1937	Eng-Strom syndrome	Disorder		2 *Cases*
60015	Enlarged parietal foramina	Disorder	4.3 *P**	
60015	Enlarged parietal foramina	Disorder	3.7 *P*	
83620	Enteric anendocrinosis	Disorder		7 *Cases*
85438	Enthesitis-related juvenile idiopathic arthritis	Disorder	5.7 *P**	
449566	Eosinophilic angiocentric fibrosis	Disorder		52 *Cases*
402035	Eosinophilic colitis	Disorder		196 *Cases*
2070	Eosinophilic gastroenteritis	Disorder		280 *Cases*
183	Eosinophilic granulomatosis with	Disorder	1.56 *P**	

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	polyangiitis			
183	Eosinophilic granulomatosis with polyangiitis	Disorder	1.5 *P*	
183	Eosinophilic granulomatosis with polyangiitis	Disorder	0.18 *I**	
251636	Ependymoma	Disorder	0.16 *I**	
231742	Epibulbar lipodermoid-preauricular appendage-polythelia syndrome	Disorder		1 *Family*
35125	Epidermal nevus syndrome	Disorder		400 *Cases*
302	Epidermodysplasia verruciformis	Disorder		200 *Cases*
46487	Epidermolysis bullosa acquisita	Disorder	0.03 *I**	
412181	Epidermolysis bullosa simplex due to BP230 deficiency	Disorder		2 *Cases*
412189	Epidermolysis bullosa simplex due to exophilin 5 deficiency	Disorder		3 *Cases*
2325	Epidermolysis bullosa simplex with anodontia/hypodontia	Disorder		5 *Cases*
257	Epidermolysis bullosa simplex with muscular dystrophy	Disorder		40 *Cases*
141077	Epignathus	Subtype of disorder	1.68 *BP*	
1948	Epilepsy-microcephaly-skeletal dysplasia syndrome	Disorder		2 *Cases*
1951	Epilepsy-telangiectasia syndrome	Disorder		6 *Cases*
1825	Epiphyseal dysplasia-hearing loss-dysmorphism syndrome	Disorder		2 *Cases*
1952	Epiphyseal stippling-osteoclastic hyperplasia syndrome	Disorder		4 *Cases*
79135	Episodic ataxia type 3	Disorder		1 *Family*
79136	Episodic ataxia type 4	Disorder		2 *Families*
211067	Episodic ataxia type 5	Disorder		7 *Cases*
209967	Episodic ataxia type 6	Disorder		4 *Cases*
209970	Episodic ataxia type 7	Disorder		7 *Cases*
401953	Episodic ataxia with slurred speech	Disorder		13 *Cases*
293381	Epithelial recurrent erosion dystrophy	Disorder		186 *Cases*
313920	Epstein-Barr virus-associated gastric	Disorder	1.2 *I*	

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	carcinoma			
35687	Erdheim-Chester disease	Disorder		500 *Cases*
999	Ermine phenotype	Disorder		6 *Cases*
317	Erythrokeratodermia variabilis	Disorder		200 *Cases*
476096	Erythrokeratodermia-cardiomyopathy syndrome	Disorder		3 *Cases*
1199	Esophageal atresia	Disorder	24.3 *BP**	
3318	Essential thrombocythemia	Disorder	0.48 *I**	
1957	Esthesioneuroblastoma	Disorder	0.02 *I**	
785	Estrogen resistance syndrome	Disorder		2 *Cases*
51188	Ethylmalonic encephalopathy	Disorder		80 *Cases*
597939	Euthyroid dysprealbuminemic hyperthyroxinemia	Disorder		23 *Cases*
1959	Evans syndrome	Disorder	0.1 *P**	
1962	Exostoses-anetodermia-brachydactyly type E syndrome	Disorder		1 *Family*
3294	Extensor tendons of finger anomalies	Disorder		2 *Cases*
3023	External auditory canal atresia-vertical talus-hypertelorism syndrome	Disorder		10 *Cases*
209916	Extraskeletal myxoid chondrosarcoma	Disorder	0.2 *P**	
1964	Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome	Disorder		2 *Cases*
3172	Eyebrow duplication-syndactyly syndrome	Disorder		3 *Cases*
306550	FADD-related immunodeficiency	Disorder		4 *Cases*
166105	FASTKD2-related infantile mitochondrial encephalomyopathy	Disorder		3 *Cases*
2492	FATCO syndrome	Disorder		22 *Cases*
404451	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome	Disorder		3 *Cases*
313855	FGFR2-related bent bone dysplasia	Disorder		11 *Cases*
2045	FLOTCH syndrome	Disorder		6 *Families*

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261144	FOXG1 syndrome due to 14q12 microdeletion	Subtype of disorder		3 *Cases*
391372	FOXP1 Syndrome	Disorder		48 *Cases*
247790	FTH1-related iron overload	Disorder		4 *Cases*
324	Fabry disease	Disorder	6.66 *BP*	
1969	Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome	Disorder		3 *Cases*
284169	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion	Subtype of disorder		19 *Cases*
466950	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation	Subtype of disorder		10 *Cases*
598603	Facial dysmorphism-hypertrichosis-epilepsy-intellectual disability/developmental delay-gingival overgrowth syndrome	Disorder		4 *Cases*
352712	Facial dysmorphism-immunodeficiency-livedo-short stature syndrome	Disorder		11 *Cases*
412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome	Disorder		4 *Families*
1970	Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome	Disorder		3 *Cases*
314555	Facial dysmorphism-ocular anomalies-osteopenia-intellectual disability-dental anomalies syndrome	Disorder		5 *Cases*
1778	Facial dysmorphism-shawl scrotum-joint laxity syndrome	Disorder		2 *Cases*
85162	Facial onset sensory and motor neuronopathy	Disorder		47 *Cases*
1973	Faciocardiorenal syndrome	Disorder		4 *Cases*
269	Facioscapulohumeral dystrophy	Disorder	4.5 *P**	
3304	Fallot complex-intellectual disability-growth delay syndrome	Disorder		5 *Cases*
280397	Familial Alzheimer-like prion disease	Disorder		2 *Cases*
481662	Familial Chilblain lupus	Disorder		10 *Families*
535458	Familial GPIHBP1 deficiency	Subtype of		10 *Families*

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		disorder		
79293	Familial LCAT deficiency	Subtype of disorder		70 *Cases*
88619	Familial acute necrotizing encephalopathy	Disorder		14 *Families*
733	Familial adenomatous polyposis	Disorder	6.0 *P**	
95700	Familial adrenal hypoplasia with absent pituitary luteinizing hormone	Disorder		3 *Cases*
228277	Familial anetoderma	Disorder		12 *Families*
530849	Familial apolipoprotein A5 deficiency	Subtype of disorder		3 *Families*
309020	Familial apolipoprotein C-II deficiency	Subtype of disorder		10 *Families*
615	Familial atrial myxoma	Disorder		17 *Families*
436242	Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease	Disorder		7 *Cases*
1551	Familial benign copper deficiency	Disorder		1 *Family*
1416	Familial calcium pyrophosphate deposition	Disorder		100 *Families*
1768	Familial caudal dysgenesis	Disorder		4 *Cases*
464760	Familial cavitary optic disc anomaly	Disorder		17 *Cases*
221061	Familial cerebral cavernous malformation	Disorder	15.0 *P*	
444490	Familial chylomicronemia syndrome	Disorder	0.97 *P**	
238578	Familial clubfoot due to 17q23.1q23.2 microduplication	Subtype of disorder		4 *Families*
238722	Familial congenital mirror movements	Disorder		75 *Cases*
451612	Familial congenital nasolacrimal duct obstruction	Disorder		4 *Cases*
91498	Familial congenital palsy of trochlear nerve	Disorder		6 *Cases*
319189	Familial cortical myoclonus	Disorder		11 *Cases*
53296	Familial cutaneous collagenoma	Disorder		16 *Cases*
313846	Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome	Disorder		24 *Cases*
1799	Familial developmental dysphasia	Disorder		6 *Families*

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324588	Familial dyskinesia and facial myokymia	Disorder		18 *Cases*
85110	Familial encephalopathy with neuroserpin inclusion bodies	Disorder		6 *Families*
391392	Familial episodic pain syndrome with predominantly lower limb involvement	Subtype of disorder		28 *Cases*
391389	Familial episodic pain syndrome with predominantly upper body involvement	Subtype of disorder		21 *Cases*
464756	Familial gastric type 1 neuroendocrine tumor	Disorder		5 *Cases*
251274	Familial hyperaldosteronism type III	Disorder		7 *Families*
238475	Familial hypercholanemia	Disorder		23 *Cases*
424	Familial hyperthyroidism due to mutations in TSH receptor	Disorder		28 *Families*
93372	Familial hypocalciuric hypercalcemia type 1	Subtype of disorder	5.5 *P*	
352582	Familial infantile myoclonic epilepsy	Disorder		7 *Cases*
154	Familial isolated dilated cardiomyopathy	Disorder	2.91 *I**	
154	Familial isolated dilated cardiomyopathy	Disorder	17.5 *P**	
99879	Familial isolated hyperparathyroidism	Disorder		100 *Families*
2238	Familial isolated hypoparathyroidism	Disorder		10 *Families*
2239	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	Subtype of disorder		2 *Families*
314777	Familial isolated pituitary adenoma	Disorder		150 *Cases*
411788	Familial isolated trichomegaly	Disorder		2 *Families*
535453	Familial lipase maturation factor 1 deficiency	Subtype of disorder		2 *Families*
401942	Familial median cleft of the upper and lower lips	Disorder		8 *Cases*
618	Familial melanoma	Disorder	1.5 *I**	
165805	Familial mesial temporal lobe epilepsy with febrile seizures	Disorder		4 *Cases*
495930	Familial monosomy 7 syndrome	Disorder		14 *Families*
538756	Familial multiple discoid fibromas	Disorder		44 *Cases*
922	Familial nasal acilia	Disorder		8 *Cases*

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280403	Familial omphalocele syndrome with facial dysmorphism	Disorder		5 *Cases*
569	Familial or sporadic hemiplegic migraine	Disorder	10.0 *P**	
2769	Familial osteodysplasia, Anderson type	Disorder		4 *Cases*
97290	Familial papillary thyroid carcinoma with renal papillary neoplasia	Disorder		2 *Cases*
79084	Familial partial lipodystrophy, Köbberling type	Disorder		20 *Cases*
871	Familial progressive cardiac conduction defect	Disorder		50 *Cases*
280628	Familial progressive hyper- and hypopigmentation	Disorder		3 *Families*
488197	Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome	Disorder		9 *Cases*
79147	Familial reactive perforating collagenosis	Disorder		50 *Cases*
168624	Familial scaphocephaly syndrome, McGillivray type	Disorder		11 *Cases*
51083	Familial short QT syndrome	Disorder		80 *Families*
166282	Familial sick sinus syndrome	Disorder		11 *Cases*
280406	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	Disorder		13 *Cases*
91387	Familial thoracic aortic aneurysm and aortic dissection	Disorder		22 *Cases*
93953	Familial thyroglossal duct cyst	Disorder		22 *Cases*
95716	Familial thyroid dyshormonogenesis	Disorder	2.67 *I*	
95716	Familial thyroid dyshormonogenesis	Disorder	4.0 *P**	
84	Fanconi anemia	Disorder	0.3 *P**	
84	Fanconi anemia	Disorder	0.62 *BP**	
2088	Fanconi-Bickel syndrome	Disorder		200 *Cases*
333	Farber disease	Disorder		96 *Cases*
439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease	Disorder		10 *Cases*
466	Fatal familial insomnia	Disorder		27 *Cases*
280553	Fatal infantile hypertonic myofibrillar	Disorder		11 *Cases*

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	myopathy			
168566	Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	Disorder		7 *Cases*
391343	Fatal post-viral neurodegenerative disorder	Disorder		2 *Cases*
438178	Fatty acyl-CoA reductase 1 deficiency	Disorder		3 *Cases*
1305	Feingold syndrome	Disorder		123 *Cases*
391641	Feingold syndrome type 1	Subtype of disorder		120 *Cases*
391646	Feingold syndrome type 2	Subtype of disorder		7 *Cases*
488191	Female infertility due to oocyte meiotic arrest	Disorder		16 *Cases*
404466	Female infertility due to zona pellucida defect	Disorder		4 *Cases*
101039	Female restricted epilepsy with intellectual disability	Disorder		5 *Families*
1988	Femoral-facial syndrome	Disorder		62 *Cases*
2019	Femur-fibula-ulna complex	Disorder	1.5 *BP**	
397922	Ferro-cerebro-cutaneous syndrome	Disorder		3 *Cases*
85212	Fetal Gaucher disease	Subtype of disorder		50 *Cases*
994	Fetal akinesia deformation sequence	Disorder	0.6 *BP**	
363409	Fetal akinesia-cerebral and retinal hemorrhage syndrome	Disorder		3 *Cases*
1915	Fetal alcohol syndrome	Disorder	1.6 *BP**	
853	Fetal and neonatal alloimmune thrombocytopenia	Disorder	39.6307 *P*	
853	Fetal and neonatal alloimmune thrombocytopenia	Disorder	66.6667 *BP*	
294	Fetal cytomegalovirus syndrome	Disorder	40.0 *P**	
465824	Fetal encasement syndrome	Disorder		2 *Cases*
1906	Fetal valproate spectrum disorder	Disorder	1.02 *BP**	
464724	Fever-associated acute infantile liver failure syndrome	Disorder		11 *Cases*
477650	Fibroblastic rheumatism	Disorder		30 *Cases*

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2021	Fibrochondrogenesis	Disorder		20 *Cases*
337	Fibrodysplasia ossificans progressiva	Disorder	0.05 *P*	
337	Fibrodysplasia ossificans progressiva	Disorder	0.078 *P**	
401920	Fibrolamellar hepatocellular carcinoma	Disorder	0.025 *I**	
84090	Fibronectin glomerulopathy	Disorder		16 *Families*
2030	Fibrosarcoma	Disorder	0.01 *I**	
1118	Fibular aplasia-ectrodactyly syndrome	Disorder		50 *Cases*
1757	Fibular dimelia-diplopodia syndrome	Disorder		11 *Cases*
93323	Fibular hemimelia	Disorder	1.1033 *BP*	
93323	Fibular hemimelia	Disorder	1.1033 *P*	
2256	Fibulo-ulnar hypoplasia-renal anomalies syndrome	Disorder		2 *Cases*
3255	Filippi syndrome	Disorder		29 *Cases*
369979	Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome	Disorder		2 *Cases*
97232	Fingerprint body myopathy	Disorder		20 *Cases*
399086	Finnish upper limb-onset distal myopathy	Disorder		7 *Cases*
79292	Fish-eye disease	Subtype of disorder		30 *Cases*
1968	Flat face-microstomia-ear anomaly syndrome	Disorder		2 *Cases*
98970	Fleck corneal dystrophy	Disorder		30 *Cases*
2047	Flynn-Aird syndrome	Disorder		10 *Cases*
2092	Focal dermal hypoplasia	Disorder		300 *Cases*
352587	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	Disorder		7 *Cases*
398166	Focal facial dermal dysplasia	Disorder		147 *Cases*
79133	Focal facial dermal dysplasia type I	Subtype of disorder		81 *Cases*
398173	Focal facial dermal dysplasia type II	Subtype of disorder		22 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
1807	Focal facial dermal dysplasia type III	Subtype of disorder		20 *Cases*
398189	Focal facial dermal dysplasia type IV	Subtype of disorder		21 *Cases*
48918	Focal myositis	Disorder		115 *Cases*
2048	Foix-Chavany-Marie syndrome	Disorder		150 *Cases*
300552	Follicular cholangitis and pancreatitis	Disorder		5 *Cases*
545	Follicular lymphoma	Disorder	37.0 *P**	
545	Follicular lymphoma	Disorder	2.192 *I**	
228371	Foodborne botulism	Subtype of disorder	0.1 *I**	
3219	Fountain syndrome	Disorder		8 *Cases*
397618	Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome	Disorder		7 *Families*
2253	Foveal hypoplasia-presenile cataract syndrome	Disorder		11 *Cases*
2795	Fowler urethral sphincter dysfunction syndrome	Disorder		33 *Cases*
221126	Fowler vasculopathy	Disorder		44 *Cases*
908	Fragile X syndrome	Disorder	32.5 *P*	
908	Fragile X syndrome	Disorder	2.4 *BP**	
908	Fragile X syndrome	Disorder	20.0 *P**	
137834	Frank-Ter Haar syndrome	Disorder		30 *Cases*
2052	Fraser syndrome	Disorder	0.2 *BP**	
347	Frasier syndrome	Disorder		150 *Cases*
834	Free sialic acid storage disease	Disorder		130 *Cases*
2053	Freeman-Sheldon syndrome	Disorder		100 *Cases*
85335	Fried syndrome	Disorder		1 *Family*
99672	Fried's tooth and nail syndrome	Disorder		12 *Cases*
95	Friedreich ataxia	Disorder	2.0 *P**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
1791	Frontofacionasal dysplasia	Disorder		14 *Cases*
1826	Frontometaphyseal dysplasia	Disorder		100 *Cases*
228390	Frontonasal dysplasia-alopecia-genital anomalies syndrome	Disorder		5 *Cases*
521308	Frontonasal dysplasia-bifid nose-upper limb anomalies syndrome	Disorder		11 *Cases*
306542	Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome	Disorder		3 *Cases*
2059	Fryns syndrome	Disorder	7.0 *BP**	
2058	Fryns-Smeets-Thiry syndrome	Disorder		2 *Cases*
349	Fucosidosis	Disorder		161 *Cases*
2854	Fuhrmann syndrome	Disorder		11 *Cases*
206554	Fukutin-related limb-girdle muscular dystrophy R13	Disorder		5 *Families*
637	Full NF2-related schwannomatosis	Disorder	1.7 *P**	
24	Fumaric aciduria	Disorder		40 *Cases*
2067	GAPO syndrome	Disorder		60 *Cases*
438274	GCGR-related hyperglucagonemia	Disorder		8 *Cases*
354	GM1 gangliosidosis	Disorder	0.75 *BP**	
79255	GM1 gangliosidosis type 1	Subtype of disorder		200 *Cases*
79256	GM1 gangliosidosis type 2	Subtype of disorder		50 *Cases*
79257	GM1 gangliosidosis type 3	Subtype of disorder		70 *Cases*
309246	GM2 gangliosidosis, AB variant	Disorder		10 *Cases*
363623	GMPPB-related limb-girdle muscular dystrophy R19	Disorder		2 *Cases*
2090	GMS syndrome	Disorder		1 *Family*
592564	GNAO1-related developmental delay-seizures-movement disorder spectrum	Disorder		75 *Cases*
542306	GNB5-related intellectual disability-cardiac arrhythmia syndrome	Disorder		22 *Cases*

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602	GNE myopathy	Disorder	1.0 *P*	
589547	GRIN2B-related developmental delay, intellectual disability and autism spectrum disorder	Disorder		98 *Cases*
2102	GTP cyclohydrolase I deficiency	Subtype of disorder		16 *Cases*
506358	Gabriele-de Vries syndrome	Disorder		10 *Cases*
570422	Galactose mutarotase deficiency	Disorder	0.4 *I*	
351	Galactosialidosis	Disorder		100 *Cases*
2065	Galloway-Mowat syndrome	Disorder		159 *Cases*
2066	Gamma-aminobutyric acid transaminase deficiency	Disorder		3 *Families*
33573	Gamma-glutamyl transpeptidase deficiency	Disorder		7 *Cases*
100026	Gamma-heavy chain disease	Subtype of disorder		120 *Cases*
353	Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5	Disorder	0.2 *P**	
79665	Gardner syndrome	Subtype of disorder	9.1 *BP*	
314022	Gastric adenocarcinoma and proximal polyposis of the stomach	Disorder		28 *Cases*
2069	Gastrocutaneous syndrome	Disorder		24 *Cases*
44890	Gastrointestinal stromal tumor	Disorder	13.0 *P**	
44890	Gastrointestinal stromal tumor	Disorder	1.0 *I*	
44890	Gastrointestinal stromal tumor	Disorder	1.0 *I**	
2368	Gastroschisis	Disorder	16.9 *BP**	
355	Gaucher disease	Disorder	1.7 *I**	
355	Gaucher disease	Disorder	1.3 *BP*	
355	Gaucher disease	Disorder	1.0 *P**	
77259	Gaucher disease type 1	Subtype of disorder	1.0 *P**	
77260	Gaucher disease type 2	Subtype of disorder	0.01 *P**	
77261	Gaucher disease type 3	Subtype of	0.05 *P**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
		disorder		
2072	Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome	Subtype of disorder		10 *Cases*
2623	Geleophysic dysplasia	Disorder		27 *Cases*
2074	Gemignani syndrome	Disorder		2 *Cases*
51608	Generalized arterial calcification of infancy	Disorder		300 *Cases*
411777	Generalized eruptive keratoacanthoma	Disorder		40 *Cases*
2075	Genitopalatocardiac syndrome	Disorder		15 *Cases*
85201	Genitopatellar syndrome	Disorder		22 *Cases*
93398	Genochondromatosis type 2	Disorder		10 *Cases*
2077	German syndrome	Disorder		5 *Cases*
2078	Geroderma osteodysplastica	Disorder		50 *Cases*
356	Gerstmann-Straussler-Scheinker syndrome	Disorder	0.0055 *I*	
643	Giant axonal neuropathy	Disorder		50 *Families*
251579	Giant cell glioblastoma	Subtype of disorder	0.02 *I**	
363976	Giant cell tumor of bone	Disorder	0.1404 *I*	
2025	Gingival fibromatosis-facial dysmorphism syndrome	Disorder		2 *Cases*
2027	Gingival fibromatosis-progressive deafness syndrome	Disorder		2 *Families*
358	Gitelman syndrome	Disorder	2.5 *P**	
238763	Glaucoma secondary to spherophakia/ectopia lentis and megalocornea	Disorder		12 *Cases*
2084	Glaucoma-ectopia lentis-microspherophakia-stiff joints-short stature syndrome	Disorder		3 *Cases*
2085	Glaucoma-sleep apnea syndrome	Disorder		5 *Cases*
360	Glioblastoma	Disorder	3.0 *I*	
360	Glioblastoma	Disorder	2.52 *I**	
360	Glioblastoma	Disorder	1.0 *P*	

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251582	Gliomatosis cerebri	Disorder	0.01 *I**	
251576	Gliosarcoma	Subtype of disorder	0.03 *I**	
544488	Global developmental delay-alopecia-macrocephaly-facial dysmorphism-structural brain anomalies syndrome	Disorder		5 *Cases*
404476	Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome	Disorder		2 *Cases*
488613	Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome	Disorder		26 *Cases*
73223	Global developmental delay-osteopenia-ectodermal defect syndrome	Disorder		3 *Cases*
480898	Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	Disorder		6 *Cases*
141163	Glossopalatine ankylosis	Disorder		30 *Cases*
97280	Glucagonoma	Disorder	0.005 *I**	
33574	Glutamate-cysteine ligase deficiency	Disorder		10 *Cases*
25	Glutaryl-CoA dehydrogenase deficiency	Disorder	1.0 *BP*	
32	Glutathione synthetase deficiency	Disorder		70 *Cases*
407	Glycine encephalopathy	Disorder	0.17 *P**	
34587	Glycogen storage disease due to LAMP-2 deficiency	Disorder		84 *Cases*
365	Glycogen storage disease due to acid maltase deficiency	Disorder	0.8 *BP**	
365	Glycogen storage disease due to acid maltase deficiency	Disorder	3.0 *P**	
420429	Glycogen storage disease due to acid maltase deficiency, late-onset	Subtype of disorder	1.75 *BP*	
364	Glycogen storage disease due to glucose-6-phosphatase deficiency	Disorder	1.0 *BP*	
79258	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia	Subtype of disorder	1.0 *BP**	
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	Subtype of disorder		150 *Cases*
367	Glycogen storage disease due to glycogen branching enzyme deficiency	Disorder	0.1 *BP*	
2089	Glycogen storage disease due to hepatic glycogen synthase deficiency	Disorder		16 *Cases*

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264580	Glycogen storage disease due to liver phosphorylase kinase deficiency	Disorder	1.0 *BP**	
137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency	Disorder		4 *Cases*
99849	Glycogen storage disease due to muscle beta-enolase deficiency	Disorder		1 *Case*
371	Glycogen storage disease due to muscle phosphofructokinase deficiency	Disorder		100 *Cases*
715	Glycogen storage disease due to muscle phosphorylase kinase deficiency	Disorder		30 *Cases*
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	Disorder		30 *Families*
97234	Glycogen storage disease due to phosphoglycerate mutase deficiency	Disorder		50 *Cases*
263297	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	Disorder		1 *Case*
329984	Goblet cell carcinoma	Subtype of disorder	0.025 *I*	
66629	Goldberg-Shprintzen megacolon syndrome	Disorder		24 *Cases*
53540	Goldmann-Favre syndrome	Disorder		50 *Cases*
1986	Gollop-Wolfgang complex	Disorder		200 *Cases*
169105	Good syndrome	Disorder		241 *Cases*
65798	Goodman syndrome	Disorder		3 *Cases*
73	Gorham-Stout disease	Disorder		300 *Cases*
377	Gorlin syndrome	Disorder	2.0 *P**	
377	Gorlin syndrome	Disorder	1.1 *P*	
2095	Gorlin-Chaudhry-Moss syndrome	Disorder		7 *Cases*
39812	Graft versus host disease	Disorder	5.0 *P**	
79094	Grange syndrome	Disorder		7 *Cases*
2097	Grant syndrome	Disorder		1 *Family*
900	Granulomatosis with polyangiitis	Disorder	0.85 *I**	
900	Granulomatosis with polyangiitis	Disorder	9.0 *P**	
33111	Granulomatous slack skin	Disorder		50 *Cases*

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721	Gray platelet syndrome	Disorder		60 *Cases*
293375	Grayson-Wilbrandt corneal dystrophy	Disorder		1 *Family*
1426	Greenberg dysplasia	Disorder		10 *Cases*
381	Griscelli syndrome	Disorder		150 *Cases*
79476	Griscelli syndrome type 1	Subtype of disorder		20 *Cases*
79477	Griscelli syndrome type 2	Subtype of disorder		102 *Cases*
79478	Griscelli syndrome type 3	Subtype of disorder		13 *Cases*
391348	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	Disorder		2 *Cases*
73272	Growth delay due to insulin-like growth factor type 1 deficiency	Disorder		5 *Cases*
3035	Growth delay-hydrocephaly-lung hypoplasia syndrome	Disorder		4 *Cases*
541423	Growth delay-intellectual disability-hepatopathy syndrome	Disorder		6 *Cases*
391366	Growth retardation-mild developmental delay-chronic hepatitis syndrome	Disorder		2 *Cases*
2101	Grubben-de Cock-Borghgraef syndrome	Disorder		3 *Cases*
382	Guanidinoacetate methyltransferase deficiency	Disorder		80 *Cases*
2957	Guttmacher syndrome	Disorder		3 *Cases*
414	Gyrate atrophy of choroid and retina	Disorder		200 *Cases*
1532	Gómez-López-Hernández syndrome	Disorder		36 *Cases*
168569	H syndrome	Disorder		100 *Cases*
73229	HANAC syndrome	Disorder		6 *Families*
2119	HEC syndrome	Disorder		2 *Cases*
79230	HJV or HAMP-related hemochromatosis	Disorder		74 *Cases*
55596	HNRNPDL-related limb-girdle muscular dystrophy D3	Disorder		2 *Families*
391417	HSD10 disease	Disorder		37 *Cases*

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85295	HSD10 disease, atypical type	Subtype of disorder		5 *Cases*
391457	HSD10 disease, neonatal type	Subtype of disorder		3 *Cases*
482077	HTRA1-related autosomal dominant cerebral small vessel disease	Disorder		21 *Cases*
99803	Haddad syndrome	Disorder		60 *Cases*
2342	Haim-Munk syndrome	Disorder		100 *Cases*
955	Hajdu-Cheney syndrome	Disorder		100 *Cases*
2107	Hall-Riggs syndrome	Disorder		8 *Cases*
2108	Hallermann-Streiff syndrome	Disorder		150 *Cases*
2109	Hallermann-Streiff-like syndrome	Disorder		2 *Cases*
2110	Hallux varus-preaxial polysyndactyly syndrome	Disorder		2 *Cases*
93946	Hamel cerebro-palato-cardiac syndrome	Subtype of disorder		4 *Cases*
500055	Hao-Fountain syndrome due to 16p13.2 microdeletion	Subtype of disorder		6 *Cases*
457	Harlequin ichthyosis	Disorder		200 *Cases*
199282	Harlequin syndrome	Disorder		100 *Cases*
2115	Harrod syndrome	Disorder		3 *Cases*
2116	Hartnup disease	Disorder	4.2 *P*	
2117	Hartsfield syndrome	Disorder		35 *Cases*
2118	Hawkinsinuria	Disorder		5 *Families*
1338	Heart defect-tongue hamartoma-polysyndactyly syndrome	Disorder		4 *Cases*
1354	Heart defects-limb shortening syndrome	Disorder		2 *Cases*
1350	Heart-hand syndrome type 2	Disorder		2 *Families*
168796	Heart-hand syndrome, Slovenian type	Disorder		14 *Cases*
86813	Helicoid peripapillary chorioretinal degeneration	Disorder		100 *Cases*
562509	Heme oxygenase-1 deficiency	Disorder		3 *Cases*

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306741	Hemidystonia-hemiatrophy syndrome	Disorder		100 *Cases*
141148	Hemifacial myohyperplasia	Disorder		12 *Cases*
276280	Hemihyperplasia-multiple lipomatosis syndrome	Disorder		10 *Cases*
306669	Hemiparkinsonism-hemiatrophy syndrome	Disorder		68 *Cases*
280615	Hemoglobinopathy Toms River	Disorder		10 *Cases*
86817	Hemolytic anemia due to adenylate kinase deficiency	Disorder		7 *Families*
712	Hemolytic anemia due to glucophosphate isomerase deficiency	Disorder		50 *Cases*
90030	Hemolytic anemia due to glutathione reductase deficiency	Disorder		3 *Cases*
766	Hemolytic anemia due to red cell pyruvate kinase deficiency	Disorder	5.0 *P**	
357008	Hemolytic uremic syndrome with DGKE deficiency	Disorder		47 *Cases*
98878	Hemophilia A	Disorder	4.85 *P*	
98878	Hemophilia A	Disorder	8.0 *P**	
98878	Hemophilia A	Disorder	11.25 *BP*	
98879	Hemophilia B	Disorder	3.0 *P**	
98879	Hemophilia B	Disorder	1.665 *BP*	
178396	Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation	Disorder		4 *Cases*
340	Hemorrhagic fever-renal syndrome	Disorder	0.74 *I**	
340	Hemorrhagic fever-renal syndrome	Disorder	37.0 *P**	
324632	Hendra virus infection	Disorder		7 *Cases*
2136	Hennekam syndrome	Disorder		50 *Cases*
2135	Hennekam-Beemer syndrome	Disorder		3 *Cases*
2031	Hepatic fibrosis-renal cysts-intellectual disability syndrome	Disorder		4 *Cases*
79124	Hepatic veno-occlusive disease-immunodeficiency syndrome	Disorder		28 *Cases*
90073	Hepatitis B reinfection following liver transplantation	Disorder	2.0 *P**	

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402823	Hepatitis delta	Disorder	40.0 *P**	
449	Hepatoblastoma	Disorder	0.02 *I**	
137681	Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	Disorder		2 *Cases*
95159	Hepatoerythropoietic porphyria	Disorder		40 *Cases*
168583	Hereditary North American Indian childhood cirrhosis	Subtype of disorder		36 *Cases*
2907	Hereditary acrokeratotic poikiloderma	Disorder		41 *Cases*
289601	Hereditary arterial and articular multiple calcification syndrome	Disorder		16 *Cases*
1867	Hereditary bullous dystrophy, macular type	Disorder		2 *Families*
85458	Hereditary cerebral hemorrhage with amyloidosis	Disorder		350 *Cases*
676	Hereditary chronic pancreatitis	Disorder	0.43 *P**	
98434	Hereditary combined deficiency of vitamin K-dependent clotting factors	Disorder		30 *Families*
398088	Hereditary cryohydrocytosis with normal stomatin	Disorder		53 *Cases*
168577	Hereditary cryohydrocytosis with reduced stomatin	Disorder		3 *Cases*
26106	Hereditary diffuse gastric cancer	Disorder	1.5 *I**	
313808	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia	Disorder		27 *Cases*
221043	Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome	Disorder		15 *Cases*
90045	Hereditary folate malabsorption	Disorder		30 *Cases*
469	Hereditary fructose intolerance	Disorder	5.0 *P**	
774	Hereditary hemorrhagic telangiectasia	Disorder	16.0 *P**	
3197	Hereditary hyperekplexia	Disorder		150 *Cases*
163	Hereditary hyperferritinemia-cataract syndrome	Disorder		120 *Cases*
217407	Hereditary hypotrichosis with recurrent skin vesicles	Disorder		4 *Cases*
324381	Hereditary inclusion body myopathy type 4	Disorder		17 *Cases*

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

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456318	Hereditary sensory neuropathy-deafness-dementia syndrome	Disorder		6 *Families*
306577	Hereditary sodium channelopathy-related small fibers neuropathy	Disorder		8 *Cases*
84093	Hereditary thermosensitive neuropathy	Disorder		1 *Family*
480851	Hereditary thrombocytopenia with early-onset myelofibrosis	Disorder		9 *Cases*
3467	Hereditary xanthinuria	Disorder		150 *Cases*
3467	Hereditary xanthinuria	Disorder	9.05 *I**	
275777	Heritable pulmonary arterial hypertension	Subtype of disorder	0.08 *P**	
79430	Hermansky-Pudlak syndrome	Disorder	0.15 *P*	
183678	Hermansky-Pudlak syndrome due to AP-3 deficiency	Subtype of disorder		40 *Cases*
231531	Hermansky-Pudlak syndrome due to BLOC-1 deficiency	Subtype of disorder		9 *Cases*
1930	Herpes simplex virus encephalitis	Disorder	0.3 *I*	
137599	Herpes simplex virus stromal keratitis	Disorder	16.0 *P**	
189	Hidrotic ectodermal dysplasia	Disorder	1.0 *P**	
1808	Hidrotic ectodermal dysplasia, Christianson-Fourie type	Disorder		6 *Cases*
1809	Hidrotic ectodermal dysplasia, Halal type	Disorder		4 *Cases*
314029	High bone mass osteogenesis imperfecta	Disorder		2 *Cases*
363396	High myopia-sensorineural deafness syndrome	Disorder		7 *Cases*
231080	High-grade dysplasia in patients with Barrett esophagus	Disorder	36.0 *P**	
388	Hirschsprung disease	Disorder	13.2 *BP**	
388	Hirschsprung disease	Disorder	15.0 *P*	
388	Hirschsprung disease	Disorder	13.2 *P**	
388	Hirschsprung disease	Disorder	15.0 *BP*	
2155	Hirschsprung disease-deafness-polydactyly syndrome	Disorder		2 *Cases*
2153	Hirschsprung disease-nail hypoplasia-dysmorphism syndrome	Disorder		3 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
2150	Hirschsprung disease-type D brachydactyly syndrome	Disorder		4 *Cases*
2158	Histidinuria-renal tubular defect syndrome	Disorder		5 *Cases*
137675	Histiocytoid cardiomyopathy	Disorder		100 *Cases*
79242	Holocarboxylase synthetase deficiency	Disorder	0.5 *BP**	
2162	Holoprosencephaly	Disorder	13.4 *BP**	
2163	Holoprosencephaly-craniosynostosis syndrome	Disorder		11 *Cases*
3186	Holoprosencephaly-radial heart renal anomalies syndrome	Disorder		4 *Cases*
392	Holt-Oram syndrome	Disorder	0.7 *BP**	
2167	Holzgreve syndrome	Disorder		3 *Cases*
394	Homocystinuria due to cystathionine beta-synthase deficiency	Disorder	0.3 *BP*	
394	Homocystinuria due to cystathionine beta-synthase deficiency	Disorder	1.65 *P**	
622	Homocystinuria without methylmalonic aciduria	Disorder		73 *Cases*
391665	Homozygous familial hypercholesterolemia	Disorder	0.3194 *P*	
3322	Hoyeraal-Hreidarsson syndrome	Disorder		33 *Cases*
228116	Hughes-Stovin syndrome	Disorder		30 *Cases*
3265	Humero-radial synostosis	Disorder		150 *Cases*
3266	Humero-radio-ulnar synostosis	Disorder		30 *Cases*
94056	Humero-ulnar synostosis	Disorder		5 *Cases*
3383	Humerus trochlea aplasia	Disorder		5 *Cases*
97340	Hunter-McAlpine syndrome	Disorder		10 *Cases*
399	Huntington disease	Disorder	0.38 *I*	
399	Huntington disease	Disorder	12.0 *P**	
399	Huntington disease	Disorder	2.7 *P*	
98934	Huntington disease-like 2	Disorder		50 *Families*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
401901	Huntington disease-like syndrome due to C9ORF72 expansions	Disorder		10 *Cases*
93473	Hurler syndrome	Subtype of disorder	0.5 *P**	
93473	Hurler syndrome	Subtype of disorder	0.7 *BP**	
740	Hutchinson-Gilford progeria syndrome	Disorder	0.025 *BP*	
740	Hutchinson-Gilford progeria syndrome	Disorder	0.005 *P*	
498474	Hyaline fibromatosis syndrome	Disorder		150 *Cases*
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	Subtype of disorder	1.7 *BP*	
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	Subtype of disorder	1.7 *P*	
2186	Hydrocephalus-blue sclerae-nephropathy syndrome	Disorder		1 *Family*
2180	Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome	Disorder		8 *Cases*
2183	Hydrocephalus-obesity-hypogonadism syndrome	Disorder		2 *Cases*
1397	Hydrocephaly-cerebellar agenesis syndrome	Disorder		2 *Cases*
2184	Hydrocephaly-low insertion umbilicus syndrome	Disorder		2 *Cases*
2181	Hydrocephaly-tall stature-joint laxity syndrome	Disorder		2 *Cases*
528091	Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome	Disorder		1 *Case*
79155	Hydroxykynureninuria	Disorder		30 *Cases*
309147	Hyper-beta-alaninemia	Disorder		3 *Cases*
927	Hyperammonemia due to N-acetylglutamate synthase deficiency	Disorder		99 *Cases*
401948	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	Disorder		4 *Cases*
168588	Hyperandrogenism due to cortisone reductase deficiency	Disorder		11 *Cases*
276405	Hyperbiliverdinemia	Disorder		2 *Cases*
209902	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	Disorder		24 *Cases*
83639	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency	Disorder		2 *Families*
163985	Hyperekplexia-epilepsy syndrome	Disorder		4 *Cases*

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2410	Hypergonadotropic hypogonadism-cataract syndrome	Disorder		3 *Cases*
343	Hyperimmunoglobulinemia D with periodic fever	Subtype of disorder		200 *Cases*
324575	Hyperinsulinism due to HNF1A deficiency	Disorder		2 *Cases*
263458	Hyperinsulinism due to INSR deficiency	Disorder		10 *Cases*
276556	Hyperinsulinism due to UCP2 deficiency	Disorder		2 *Cases*
71212	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	Disorder		10 *Cases*
682	Hyperkalemic periodic paralysis	Disorder	0.5 *P**	
1336	Hyperkeratosis-hyperpigmentation syndrome	Disorder		10 *Cases*
285	Hypermobile Ehlers-Danlos syndrome	Disorder	12.5 *P**	
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	Disorder		111 *Cases*
3416	Hyperostosis corticalis generalisata	Disorder		35 *Cases*
443098	Hyperostosis cranialis interna	Disorder		13 *Cases*
99880	Hyperparathyroidism-jaw tumor syndrome	Disorder		100 *Cases*
508523	Hyperphenylalaninemia due to DNAJC12 deficiency	Disorder		6 *Cases*
238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	Disorder	0.2 *P*	
247262	Hyperphosphatasia-intellectual disability syndrome	Disorder		24 *Cases*
2211	Hypertelorism-hypospadias-polysyndactyly syndrome	Disorder		3 *Families*
2213	Hypertelorism-microtia-facial clefting syndrome	Disorder		9 *Cases*
293958	Hypertelorism-preauricular sinus-punctual pits-deafness syndrome	Disorder		13 *Cases*
2220	Hypertrichosis cubiti	Disorder		28 *Cases*
2222	Hypertrichosis lanuginosa congenita	Disorder		100 *Cases*
324525	Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation	Disorder		3 *Cases*
2224	Hypertryptophanemia	Disorder		12 *Cases*

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363694	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome	Disorder		4 *Families*
251523	Hyperzincemia and hypercalprotectinemia	Disorder		18 *Cases*
2435	Hypo- and hypermelanotic cutaneous macules-retarded growth-intellectual disability syndrome	Disorder		14 *Cases*
429	Hypochondroplasia	Disorder	3.0303 *BP*	
429	Hypochondroplasia	Disorder	3.0303 *P*	
989	Hypoglossia-hypodactyly syndrome	Disorder		47 *Cases*
2233	Hypogonadism-mitral valve prolapse-intellectual disability syndrome	Disorder		2 *Cases*
2230	Hypogonadotropic hypogonadism-frontoparietal alopecia syndrome	Disorder		6 *Cases*
2235	Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome	Disorder		2 *Cases*
293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome	Disorder		4 *Cases*
528105	Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome	Disorder		22 *Cases*
363523	Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome	Disorder		12 *Cases*
238468	Hypohidrotic ectodermal dysplasia	Disorder	6.7 *P**	
98813	Hypohidrotic ectodermal dysplasia with immunodeficiency	Disorder	0.2 *BP**	
1882	Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome	Disorder		3 *Cases*
293964	Hypoinsulinemic hypoglycemia and body hemihypertrophy	Disorder		5 *Cases*
681	Hypokalemic periodic paralysis	Disorder	1.0 *P**	
1790	Hypomandibular faciocranial dysostosis	Disorder		3 *Cases*
137639	Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome	Subtype of disorder		8 *Cases*
2680	Hypomyelination neuropathy-arthrogryposis syndrome	Disorder		9 *Cases*
599376	Hypomyelination of early myelinating structures	Disorder		20 *Cases*
139441	Hypomyelination with atrophy of basal	Disorder		19 *Cases*

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	ganglia and cerebellum			
363412	Hypomyelination with brain stem and spinal cord involvement and leg spasticity	Disorder		13 *Cases*
447893	Hypomyelination-cerebellar atrophy-hypoplasia of the corpus callosum syndrome	Subtype of disorder		4 *Cases*
85163	Hypomyelination-congenital cataract syndrome	Disorder		10 *Cases*
2237	Hypoparathyroidism-sensorineural deafness-renal disease syndrome	Disorder		180 *Cases*
324561	Hypopigmentation-punctate palmoplantar keratoderma syndrome	Disorder		6 *Families*
722	Hypoplasminogenemia	Disorder	0.16 *P**	
2248	Hypoplastic left heart syndrome	Disorder	18.0 *BP*	
2248	Hypoplastic left heart syndrome	Disorder	15.1 *BP**	
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	Disorder		16 *Cases*
2250	Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome	Disorder		2 *Cases*
2261	Hypospadias-intellectual disability, Goldblatt type syndrome	Disorder		3 *Cases*
137908	Hypotonia with lactic acidemia and hyperammonemia	Disorder		4 *Cases*
163690	Hypotonia-cystinuria syndrome	Disorder		22 *Cases*
79507	Hypotonia-failure to thrive-microcephaly syndrome	Disorder		2 *Cases*
55654	Hypotrichosis simplex	Disorder		38 *Cases*
1573	Hypotrichosis with juvenile macular degeneration	Disorder		50 *Cases*
330029	Hypotrichosis-deafness syndrome	Disorder		1 *Case*
2266	Hypotrichosis-intellectual disability, Lopes type	Disorder		2 *Cases*
69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	Disorder		4 *Cases*
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar keratoderma syndrome	Disorder		2 *Cases*
2268	ICF syndrome	Disorder		66 *Cases*
477661	IL21-related infantile inflammatory bowel disease	Disorder		3 *Cases*

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85173	IMAGe syndrome	Disorder		28 *Cases*
597623	IRF2BPL-related regressive neurodevelopmental disorder-dystonia-seizures syndrome	Disorder		19 *Cases*
209981	IRIDA syndrome	Disorder		75 *Cases*
209943	IRVAN syndrome	Disorder		30 *Cases*
352479	ISPD-related limb-girdle muscular dystrophy R20	Disorder		8 *Cases*
439254	ITM2B amyloidosis	Disorder		2 *Families*
457375	ITPA-related lethal infantile neurological disorder with cataract and cardiac involvement	Disorder		7 *Cases*
2307	IVIC syndrome	Disorder		4 *Families*
254509	Iatrogenic botulism	Subtype of disorder		180 *Cases*
2273	Ichthyosis follicularis-alopecia-photophobia syndrome	Disorder		40 *Cases*
79503	Ichthyosis hystrix of Curth-Macklin	Disorder		10 *Cases*
2269	Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome	Disorder		4 *Cases*
2274	Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome	Disorder		2 *Cases*
91132	Ichthyosis-hypotrichosis syndrome	Disorder		11 *Cases*
2278	Ichthyosis-intellectual disability-dwarfism-renal impairment syndrome	Disorder		4 *Cases*
2272	Ichthyosis-oral and digital anomalies syndrome	Disorder		2 *Cases*
88621	Ichthyosis-prematurity syndrome	Disorder		16 *Families*
363992	Ichthyosis-short stature-brachydactyly-microspherophakia syndrome	Disorder		7 *Cases*
930	Idiopathic achalasia	Disorder	8.0 *P*	
930	Idiopathic achalasia	Disorder	0.77 *I*	
724	Idiopathic acute eosinophilic pneumonia	Disorder		100 *Cases*
139423	Idiopathic acute transverse myelitis	Subtype of disorder	0.25 *I**	
88	Idiopathic aplastic anemia	Disorder	0.4 *P**	

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33208	Idiopathic hypersomnia	Disorder	30.0 *P**	
238624	Idiopathic intracranial hypertension	Disorder	14.0 *P**	
45452	Idiopathic neonatal atrial flutter	Disorder	1.5 *BP**	
280921	Idiopathic panuveitis	Disorder	2.0194 *P**	
280921	Idiopathic panuveitis	Disorder	0.5051 *I**	
494428	Idiopathic pleuroparenchymal fibroelastosis	Disorder		37 *Cases*
275766	Idiopathic pulmonary arterial hypertension	Subtype of disorder	1.1 *P**	
2032	Idiopathic pulmonary fibrosis	Disorder	16.125 *P*	
2032	Idiopathic pulmonary fibrosis	Disorder	5.55 *I*	
99931	Idiopathic pulmonary hemosiderosis	Disorder	0.0425 *I**	
567548	Idiopathic steroid-resistant nephrotic syndrome	Disorder	0.2582 *I*	
422	Idiopathic/heritable pulmonary arterial hypertension	Disorder	1.0 *P**	
49041	IgG4-related retroperitoneal fibrosis	Subtype of disorder	0.35 *I**	
238621	Ileal pouch anal anastomosis related faecal incontinence	Disorder	3.0 *P**	
42062	Iminoglycinuria	Disorder	6.67 *BP**	
42062	Iminoglycinuria	Disorder	6.68 *P**	
238569	Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome	Disorder		80 *Cases*
529977	Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections-lymphopenia syndrome	Disorder		7 *Cases*
37042	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	Disorder		195 *Cases*
3002	Immune thrombocytopenia	Disorder	25.0 *P**	
3002	Immune thrombocytopenia	Disorder	6.75 *I**	
206569	Immune-mediated necrotizing myopathy	Disorder		300 *Cases*
34592	Immunodeficiency by defective	Disorder		30 *Cases*

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	expression of MHC class I			
572	Immunodeficiency by defective expression of MHC class II	Disorder		179 *Cases*
169100	Immunodeficiency due to CD25 deficiency	Disorder		2 *Cases*
331187	Immunodeficiency due to MASP-2 deficiency	Disorder		1 *Case*
331190	Immunodeficiency due to ficolin3 deficiency	Disorder		1 *Case*
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	Disorder		49 *Cases*
200418	Immunodeficiency with factor I anomaly	Disorder		35 *Families*
2759	Imperforate oropharynx-costovertebral anomalies syndrome	Disorder		2 *Cases*
45453	Incessant infant ventricular tachycardia	Disorder	1.5 *BP**	
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	Disorder		26 *Families*
611	Inclusion body myositis	Disorder	0.5 *P**	
464	Incontinentia pigmenti	Disorder	1.2 *BP**	
70587	Infant acute respiratory distress syndrome	Disorder	3.5 *I**	
70587	Infant acute respiratory distress syndrome	Disorder	20.0 *P**	
178478	Infant botulism	Subtype of disorder	0.2 *BP**	
178478	Infant botulism	Subtype of disorder	0.3 *I**	
313850	Infantile cerebellar-retinal degeneration	Disorder		11 *Cases*
402364	Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	Disorder		5 *Cases*
1313	Infantile choroidocerebral calcification syndrome	Disorder		10 *Cases*
199267	Infantile digital fibromatosis	Disorder		200 *Cases*
238455	Infantile dystonia-parkinsonism	Disorder		16 *Cases*
352563	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	Disorder		2 *Cases*
522077	Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome	Disorder		11 *Cases*
565788	Infantile inflammatory bowel disease	Disorder		3 *Cases*

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	with neurological involvement			
456312	Infantile multisystem neurologic-endocrine-pancreatic disease	Disorder		2 *Cases*
2591	Infantile myofibromatosis	Disorder	0.67 *BP**	
35069	Infantile neuroaxonal dystrophy	Disorder		150 *Cases*
77292	Infantile neurovisceral acid sphingomyelinase deficiency	Disorder	0.25 *BP**	
251304	Infantile onset panniculitis with uveitis and systemic granulomatosis	Disorder		4 *Cases*
3451	Infantile spasms syndrome	Disorder	6.0 *P**	
3451	Infantile spasms syndrome	Disorder	3.7 *BP*	
3451	Infantile spasms syndrome	Disorder	3.5 *BP**	
3173	Infantile spasms-broad thumbs syndrome	Disorder		2 *Cases*
263410	Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome	Disorder		4 *Cases*
1145	Infantile-onset X-linked spinal muscular atrophy	Disorder		14 *Families*
293168	Infantile-onset ascending hereditary spastic paralysis	Disorder		17 *Families*
457205	Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome	Disorder		2 *Cases*
494526	Infantile-onset generalized dyskinesia with orofacial involvement	Disorder		8 *Cases*
391316	Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression	Disorder		3 *Cases*
500062	Infantile-onset periodic fever-panniculitis-dermatosis syndrome	Disorder		5 *Cases*
572428	Infantile-onset pulmonary alveolar proteinosis-hypogammaglobulinemia	Disorder		5 *Cases*
1186	Infantile-onset spinocerebellar ataxia	Disorder		29 *Cases*
529980	Inflammatory bowel disease-recurrent sinopulmonary infections syndrome	Disorder		1 *Case*
90003	Inflammatory pseudotumor of the liver	Disorder		140 *Cases*
254504	Inhalational botulism	Subtype of disorder		10 *Cases*
210141	Inherited congenital spastic tetraplegia	Disorder		17 *Cases*
63259	Iniencephaly	Disorder	50.0 *BP**	

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411593	Insulin autoimmune syndrome	Disorder		404 *Cases*
97279	Insulinoma	Disorder	0.25 *I*	
464311	Intellectual disability syndrome due to a DYRK1A point mutation	Subtype of disorder		35 *Cases*
166108	Intellectual disability, Birk-Barel type	Disorder		1 *Family*
3079	Intellectual disability, Buenos-Aires type	Disorder		5 *Cases*
3080	Intellectual disability, Wolff type	Disorder		2 *Cases*
529965	Intellectual disability-autism-speech apraxia-craniofacial dysmorphism syndrome	Disorder		5 *Cases*
3041	Intellectual disability-balding-patella luxation-acromicria syndrome	Disorder		3 *Cases*
364577	Intellectual disability-brachydactyly-Pierre Robin syndrome	Disorder		4 *Cases*
508498	Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome	Disorder		18 *Cases*
3042	Intellectual disability-cataracts-calcified pinnae-myopathy syndrome	Disorder		13 *Cases*
397709	Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	Disorder		30 *Cases*
329224	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	Disorder		2 *Cases*
3454	Intellectual disability-developmental delay-contractures syndrome	Disorder		5 *Families*
3044	Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome	Disorder		4 *Cases*
468620	Intellectual disability-epilepsy-extrapyramidal syndrome	Disorder		3 *Cases*
436151	Intellectual disability-expressive aphasia-facial dysmorphism syndrome	Disorder		13 *Cases*
404440	Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	Disorder		7 *Cases*
370010	Intellectual disability-facial dysmorphism-hand anomalies syndrome	Disorder		3 *Cases*
369847	Intellectual disability-hyperkinetic movement-truncal ataxia syndrome	Disorder		5 *Cases*
1495	Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome	Disorder		3 *Cases*
314575	Intellectual disability-hypotonia-	Disorder		2 *Cases*

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	brachycephaly-pyloric stenosis-cryptorchidism syndrome			
457279	Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome	Disorder		16 *Cases*
457365	Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome	Disorder		3 *Cases*
3068	Intellectual disability-myopathy-short stature-endocrine defect syndrome	Disorder		2 *Cases*
352530	Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome	Disorder		2 *Cases*
397973	Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome	Disorder		2 *Cases*
3082	Intellectual disability-polydactyly-uncombable hair syndrome	Disorder		2 *Cases*
513456	Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome	Disorder		15 *Cases*
369837	Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	Disorder		4 *Cases*
369950	Intellectual disability-seizures-macrocephaly-obesity syndrome	Disorder		7 *Cases*
3074	Intellectual disability-short stature-hypertelorism syndrome	Disorder		6 *Cases*
1891	Intellectual disability-spasticity-ectrodactyly syndrome	Disorder		3 *Cases*
363528	Intellectual disability-strabismus syndrome	Disorder		34 *Cases*
508529	Intermediate epidermolysis bullosa simplex with cardiomyopathy	Disorder		14 *Cases*
981	Internal carotid absence	Disorder		100 *Cases*
79099	Interstitial granulomatous dermatitis with arthritis	Disorder		53 *Cases*
306504	Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome	Disorder		3 *Cases*
314376	Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency	Disorder		16 *Cases*
137622	Intractable diarrhea-choanal atresia-eye anomalies syndrome	Disorder		3 *Cases*
424058	Intraductal papillary mucinous carcinoma of pancreas	Disorder	0.011 *I**	
508512	Intrauterine growth restriction-congenital multiple café-au-lait macules-	Disorder		2 *Cases*

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	increased sister chromatid exchange syndrome			
436144	Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	Disorder		15 *Cases*
329324	Inverse Klippel-Trénaunay syndrome	Disorder		15 *Cases*
3306	Inverted duplicated chromosome 15 syndrome	Disorder	3.33 *BP*	
84142	Isaacs syndrome	Disorder		150 *Cases*
217	Isolated Dandy-Walker malformation	Disorder	1.0 *BP**	
217	Isolated Dandy-Walker malformation	Disorder	2.1 *P**	
2345	Isolated Klippel-Feil syndrome	Disorder	2.0 *P**	
2345	Isolated Klippel-Feil syndrome	Disorder	0.6 *BP**	
718	Isolated Pierre Robin syndrome	Disorder	5.0 *BP**	
229717	Isolated agammaglobulinemia	Disorder	0.3 *P*	
1048	Isolated anencephaly/exencephaly	Disorder	35.0 *BP**	
250923	Isolated aniridia	Disorder	1.31 *I**	
3387	Isolated anterior cervical hypertrichosis	Disorder		20 *Cases*
1134	Isolated arrhinia	Disorder		20 *Cases*
199326	Isolated autosomal dominant hypomagnesemia, Glaudemans type	Disorder		21 *Cases*
30391	Isolated biliary atresia	Disorder	2.9 *BP**	
30391	Isolated biliary atresia	Disorder	18.5 *BP*	
209908	Isolated childhood apraxia of speech	Disorder		22 *Cases*
79143	Isolated congenital anonychia	Disorder		50 *Cases*
88620	Isolated congenital anosmia	Disorder		15 *Cases*
91396	Isolated cryptophthalmia	Disorder		30 *Cases*
1885	Isolated ectopia lentis	Disorder		90 *Cases*
93928	Isolated epispadias	Subtype of	2.4 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
		disorder		
448264	Isolated focal non-epidermolytic palmoplantar keratoderma	Disorder		2 *Cases*
468666	Isolated generalized anhidrosis with normal sweat glands	Disorder		7 *Cases*
306527	Isolated hereditary congenital facial paralysis	Disorder		8 *Families*
542657	Isolated hyperchlorhidrosis	Disorder		13 *Cases*
583861	Isolated mesenteric vein thrombosis	Disorder	1.6 *I**	
480556	Isolated neonatal sclerosing cholangitis	Disorder		4 *Cases*
166119	Isolated osteopoikilosis	Disorder	2.0 *P*	
166119	Isolated osteopoikilosis	Disorder	2.0 *I*	
99885	Isolated permanent neonatal diabetes mellitus	Disorder	0.38 *BP**	
2924	Isolated polycystic liver disease	Disorder	1.0 *P**	
440713	Isolated sedoheptulokinase deficiency	Disorder		2 *Cases*
457083	Isolated splenogonadal fusion	Disorder		145 *Cases*
2440	Isolated split hand-split foot malformation	Disorder	5.4 *BP**	
3208	Isolated succinate-CoQ reductase deficiency	Disorder		37 *Cases*
99731	Isolated sulfite oxidase deficiency	Subtype of disorder		50 *Cases*
454750	Isolated tracheoesophageal fistula	Disorder	2.2 *BP*	
2306	Isotretinoin-like syndrome	Disorder		6 *Cases*
33	Isovaleric acidemia	Disorder	1.0 *P**	
1540	Jackson-Weiss syndrome	Disorder		200 *Cases*
1873	Jalili syndrome	Disorder		49 *Cases*
79139	Japanese encephalitis	Disorder	0.65 *I**	
313795	Jawad syndrome	Disorder		4 *Cases*
90647	Jervell and Lange-Nielsen syndrome	Disorder	0.3 *P*	
474	Jeune syndrome	Disorder	1.4 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
2315	Johanson-Blizzard syndrome	Disorder	0.4 *BP**	
475	Joubert syndrome	Disorder	1.6666 *BP*	
397715	Joubert syndrome with Jeune asphyxiating thoracic dystrophy	Disorder		8 *Cases*
1454	Joubert syndrome with hepatic defect	Disorder		8 *Cases*
2318	Joubert syndrome with oculorenal defect	Disorder		17 *Cases*
2319	Juberg-Hayward syndrome	Disorder		13 *Cases*
79405	Junctional epidermolysis bullosa inversa	Disorder		9 *Cases*
79403	Junctional epidermolysis bullosa with pyloric atresia	Disorder		100 *Cases*
2321	Jung syndrome	Disorder		2 *Cases*
248111	Juvenile Huntington disease	Disorder	0.04 *I**	
248111	Juvenile Huntington disease	Disorder	0.6 *P**	
2801	Juvenile Paget disease	Disorder		50 *Cases*
1941	Juvenile absence epilepsy	Disorder	7.5 *I**	
247794	Juvenile cataract-microcornea-renal glucosuria syndrome	Disorder		12 *Cases*
93672	Juvenile dermatomyositis	Disorder	0.295 *I*	
2028	Juvenile hyaline fibromatosis	Subtype of disorder		70 *Cases*
86834	Juvenile myelomonocytic leukemia	Disorder	0.1 *P**	
289596	Juvenile nasopharyngeal angiofibroma	Disorder	0.6666 *I*	
79076	Juvenile polyposis of infancy	Subtype of disorder		11 *Cases*
2929	Juvenile polyposis syndrome	Disorder	3.85 *I**	
247604	Juvenile primary lateral sclerosis	Disorder		4 *Cases*
26137	Juvenile temporal arteritis	Disorder		20 *Cases*
445062	Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	Disorder		5 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
2332	KBG syndrome	Disorder		164 *Cases*
439218	KCNQ2-related epileptic encephalopathy	Disorder		11 *Families*
85279	KDM5C-related syndromic X-linked intellectual disability	Disorder		10 *Families*
477	KID syndrome	Disorder		100 *Cases*
603684	KLHL7-related Bohring-Opitz-like and Crisponi/Cold-induced sweating-like overlap syndrome	Disorder		3 *Cases*
603689	KLHL7-related Bohring-Opitz-like syndrome	Disorder		12 *Cases*
399081	KLHL9-related early-onset distal myopathy	Disorder		10 *Cases*
2322	Kabuki syndrome	Disorder	3.1 *P**	
254519	Kagami-Ogata syndrome	Disorder		84 *Cases*
254534	Kagami-Ogata syndrome due to maternal 14q32.2 hypermethylation	Subtype of disorder		7 *Cases*
254528	Kagami-Ogata syndrome due to maternal 14q32.2 microdeletion	Subtype of disorder		8 *Cases*
96334	Kagami-Ogata syndrome due to paternal uniparental disomy of chromosome 14	Subtype of disorder		37 *Cases*
478	Kallmann syndrome	Subtype of disorder	3.75 *P**	
2326	Kallmann syndrome-heart disease syndrome	Disorder		8 *Cases*
33276	Kaposi sarcoma	Disorder	0.34 *I**	
33276	Kaposi sarcoma	Disorder	2.11	
2328	Kapur-Toriello syndrome	Disorder		6 *Cases*
2329	Karsch-Neugebauer syndrome	Disorder		11 *Cases*
401996	Karyomegalic interstitial nephritis	Disorder		12 *Families*
2330	Kasabach-Merritt phenomenon	Disorder		300 *Cases*
480	Kearns-Sayre syndrome	Disorder	2.0 *P**	
2662	Keipert syndrome	Disorder		12 *Cases*
2333	Kenny-Caffey syndrome	Disorder		65 *Cases*
435628	Keppen-Lubinsky syndrome	Disorder		3 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
494	Keratoderma hereditarium mutilans	Disorder		50 *Cases*
79395	Keratoderma hereditarium mutilans with ichthyosis	Disorder		50 *Cases*
2339	Keratosis follicularis-dwarfism-cerebral atrophy syndrome	Disorder		6 *Cases*
86919	Keratosis palmaris et plantaris-clinodactyly syndrome	Disorder		20 *Cases*
293807	Ketamine-induced biliary dilatation	Disorder		2 *Cases*
438075	Ketoacidosis due to monocarboxylate transporter-1 deficiency	Disorder		9 *Cases*
85202	Keutel syndrome	Disorder		30 *Cases*
73224	Kidney tubulopathy-dilated cardiomyopathy syndrome	Disorder		2 *Cases*
50918	Kikuchi-Fujimoto disease	Disorder		1052 *Cases*
482	Kimura disease	Disorder		300 *Cases*
2908	Kindler epidermolysis bullosa	Disorder		250 *Cases*
99741	King-Denborough syndrome	Disorder		18 *Cases*
261494	Kleefstra syndrome	Disorder		114 *Cases*
96147	Kleefstra syndrome due to 9q34 microdeletion	Subtype of disorder		86 *Cases*
261652	Kleefstra syndrome due to a point mutation	Subtype of disorder		23 *Cases*
447974	Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome	Disorder		2 *Cases*
90308	Klippel-Trénaunay syndrome	Disorder	0.007 *P**	
1571	Knobloch syndrome	Disorder		119 *Cases*
363965	Koolen-De Vries syndrome due to a point mutation	Subtype of disorder		4 *Cases*
477831	Kosaki overgrowth syndrome	Disorder		2 *Cases*
99749	Kostmann syndrome	Disorder		45 *Cases*
2351	Kousseff syndrome	Disorder		8 *Cases*
487	Krabbe disease	Disorder	1.0 *P**	
487	Krabbe disease	Disorder	1.0 *BP**	

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487	Krabbe disease	Disorder	0.7 *BP*	
306674	Kufor-Rakeb syndrome	Disorder		16 *Cases*
454745	Kuru	Disorder		2700 *Cases*
1149	Kuskokwim syndrome	Disorder		8 *Families*
496689	Kyphoscoliosis-lateral tongue atrophy-hereditary spastic paraplegia syndrome	Disorder		12 *Cases*
300179	Kyphoscoliotic Ehlers-Danlos syndrome due to FKBP22 deficiency	Subtype of disorder		9 *Cases*
1900	Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency	Subtype of disorder	1.0 *BP*	
496686	Kyphosis-lateral tongue atrophy-myofibrillar myopathy syndrome	Disorder		3 *Cases*
79314	L-2-hydroxyglutaric aciduria	Disorder		140 *Cases*
35704	L-Arginine:glycine amidinotransferase deficiency	Disorder		9 *Cases*
440731	L-ferritin deficiency	Disorder		2 *Cases*
521450	LAMA5-related multisystemic syndrome	Disorder		11 *Cases*
650	LCAT deficiency	Disorder		125 *Cases*
99812	LIG4 syndrome	Disorder		28 *Cases*
435660	LIPE-related familial partial lipodystrophy	Disorder		4 *Cases*
363618	LMNA-related cardiocutaneous progeria syndrome	Disorder		5 *Cases*
83628	LUMBAR syndrome	Disorder		54 *Cases*
2363	Lacrimoauriculodentodigital syndrome	Disorder		100 *Cases*
501	Lafora disease	Disorder		300 *Cases*
530983	Lamb-Shaffer syndrome	Disorder		70 *Cases*
1296	Lambert syndrome	Disorder		4 *Cases*
43393	Lambert-Eaton myasthenic syndrome	Disorder	1.0 *P**	
43393	Lambert-Eaton myasthenic syndrome	Disorder	0.35 *P*	
258	Laminin subunit alpha 2-related	Disorder	0.3 *P**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	congenital muscular dystrophy			
2632	Langer mesomelic dysplasia	Disorder		100 *Cases*
389	Langerhans cell histiocytosis	Disorder	1.5 *P**	
626	Large congenital melanocytic nevus	Disorder	2.75 *P**	
633	Laron syndrome	Disorder	0.3 *P**	
220465	Laron syndrome with immunodeficiency	Disorder		10 *Cases*
503	Larsen syndrome	Disorder	0.4 *BP**	
2370	Larsen-like osseous dysplasia-short stature syndrome	Disorder		3 *Cases*
284139	Larsen-like syndrome, B3GAT3 type	Disorder		14 *Cases*
2808	Laryngeal abductor paralysis	Disorder		9 *Cases*
2375	Laryngeal abductor paralysis-intellectual disability syndrome	Disorder		20 *Cases*
2407	Laryngo-onycho-cutaneous syndrome	Disorder		50 *Cases*
2004	Laryngotracheoesophageal cleft	Disorder	7.5 *BP**	
93940	Laryngotracheoesophageal cleft type 3	Subtype of disorder		30 *Cases*
93941	Laryngotracheoesophageal cleft type 4	Subtype of disorder		20 *Cases*
98912	Late-onset distal myopathy, Markesbery-Griggs type	Disorder		11 *Cases*
228227	Late-onset focal dermal elastosis	Disorder		5 *Cases*
79406	Late-onset junctional epidermolysis bullosa	Disorder		37 *Cases*
231556	Late-onset localized junctional epidermolysis bullosa-intellectual disability syndrome	Disorder		2 *Cases*
2789	Lateral meningocele syndrome	Disorder		14 *Cases*
46059	Lathosterolosis	Disorder		4 *Cases*
2378	Laurin-Sandrow syndrome	Disorder		14 *Cases*
330015	Lead poisoning	Disorder	2.3 *P**	
65	Leber congenital amaurosis	Disorder	2.5 *BP*	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
65	Leber congenital amaurosis	Disorder	2.5 *P*	
104	Leber hereditary optic neuropathy	Disorder	4.3 *P*	
104	Leber hereditary optic neuropathy	Disorder	2.3 *P**	
99718	Leber plus disease	Disorder	0.04 *P**	
549	Legionnaires disease	Disorder	1.4 *I**	
137605	Legius syndrome	Disorder	2.2 *BP*	
506	Leigh syndrome	Disorder	2.8 *BP**	
506	Leigh syndrome	Disorder	2.0 *P**	
507	Leishmaniasis	Disorder	0.1 *P**	
507	Leishmaniasis	Disorder	25.0 *I*	
140936	Lelis syndrome	Disorder		9 *Cases*
137839	Lemierre syndrome	Disorder	10.0 *I**	
2382	Lennox-Gastaut syndrome	Disorder	0.1 *I**	
2382	Lennox-Gastaut syndrome	Disorder	15.0 *P**	
2658	Lenz-Majewski hyperostotic dwarfism	Disorder		10 *Cases*
548	Leprosy	Disorder	3.7 *I*	
509	Leptospirosis	Disorder	0.12 *I**	
510	Lesch-Nyhan syndrome	Disorder	0.34 *BP**	
2347	Lethal Kniest-like dysplasia	Disorder		2 *Cases*
2371	Lethal Larsen-like syndrome	Disorder		8 *Cases*
158687	Lethal acantholytic erosive disorder	Disorder		4 *Cases*
314718	Lethal arteriopathy syndrome due to fibulin-4 deficiency	Disorder		22 *Cases*
1187	Lethal ataxia with deafness and optic atrophy	Disorder		4 *Families*
580933	Lethal brain and heart developmental defects	Disorder		4 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
137776	Lethal congenital contracture syndrome type 2	Disorder		1 *Family*
137783	Lethal congenital contracture syndrome type 3	Disorder		14 *Cases*
1972	Lethal faciocardiomelic dysplasia	Disorder		3 *Cases*
444069	Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome	Disorder		4 *Cases*
439897	Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome	Disorder		2 *Cases*
1046	Lethal hemolytic anemia-genital anomalies syndrome	Disorder		2 *Cases*
480528	Lethal hydranencephaly-diaphragmatic hernia syndrome	Disorder		2 *Cases*
2570	Lethal intrauterine growth restriction-cortical malformation-congenital contractures syndrome	Disorder		4 *Cases*
478049	Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome	Disorder		4 *Cases*
33108	Lethal multiple pterygium syndrome	Disorder		28 *Families*
435845	Lethal neonatal spasticity-epileptic encephalopathy syndrome	Disorder		8 *Cases*
293925	Lethal occipital encephalocele-skeletal dysplasia syndrome	Disorder		5 *Cases*
2736	Lethal omphalocele-cleft palate syndrome	Disorder		5 *Cases*
1832	Lethal osteosclerotic bone dysplasia	Disorder		40 *Cases*
210144	Lethal polymalformative syndrome, Boissel type	Disorder		10 *Cases*
1423	Lethal recessive chondrodysplasia	Disorder		4 *Cases*
2968	Leukocyte adhesion deficiency	Disorder		350 *Cases*
99842	Leukocyte adhesion deficiency type I	Subtype of disorder	0.1 *P**	
99843	Leukocyte adhesion deficiency type II	Subtype of disorder		7 *Cases*
99844	Leukocyte adhesion deficiency type III	Subtype of disorder		40 *Cases*
139444	Leukoencephalopathy with bilateral anterior temporal lobe cysts	Disorder		29 *Cases*
137898	Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	Disorder		127 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
542310	Leukoencephalopathy with calcifications and cysts	Disorder		50 *Cases*
363540	Leukoencephalopathy with mild cerebellar ataxia and white matter edema	Disorder		6 *Cases*
163684	Leukoencephalopathy-dystonia-motor neuropathy syndrome	Disorder		2 *Cases*
2386	Leukoencephalopathy-palmoplantar keratoderma syndrome	Disorder		4 *Cases*
83629	Leukoencephalopathy-spondyloepimetaphyseal dysplasia syndrome	Disorder		11 *Cases*
314051	Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome	Disorder		14 *Cases*
1816	Leukomelanoderma-infantilism-intellectual disability-hypodontia-hypotrichosis syndrome	Disorder		4 *Cases*
210133	Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome	Disorder		11 *Cases*
48162	Lewis-Sumner syndrome	Subtype of disorder	0.9 *P**	
65285	Lhermitte-Duclos disease	Disorder		220 *Cases*
525	Lichen planopilaris	Disorder		300 *Cases*
254478	Lichen planus pemphigoides	Disorder		100 *Cases*
2390	Lichtenstein syndrome	Disorder		2 *Cases*
526	Liddle syndrome	Disorder		72 *Families*
445110	Limb-girdle muscular dystrophy due to POMK deficiency	Disorder		2 *Cases*
69085	Limb-mammary syndrome	Disorder		38 *Cases*
171673	Limbal stem cell deficiency	Disorder	20.0 *P**	
220407	Limited systemic sclerosis	Subtype of disorder		200 *Cases*
140933	Linear atrophoderma of Moulin	Disorder		30 *Cases*
228236	Linear focal elastosis	Disorder		30 *Cases*
589608	Linear hypopigmentation and craniofacial asymmetry with acral, ocular and brain anomalies	Disorder		7 *Cases*
1979	Lipodystrophy due to peptidic growth	Disorder		1 *Family*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	factors deficiency			
50811	Lipodystrophy-intellectual disability-deafness syndrome	Disorder		3 *Cases*
401859	Lipoic acid synthetase deficiency	Disorder		3 *Cases*
530	Lipoid proteinosis	Disorder		500 *Cases*
329481	Lipoprotein glomerulopathy	Disorder		150 *Cases*
69078	Liposarcoma	Disorder	1.0 *I**	
401862	Lipoyl transferase 1 deficiency	Disorder		4 *Cases*
98955	Lisch epithelial corneal dystrophy	Disorder		36 *Cases*
171680	Lissencephaly due to TUBA1A mutation	Disorder		15 *Cases*
86821	Lissencephaly type 3-familial fetal akinesia sequence syndrome	Disorder		5 *Cases*
86822	Lissencephaly type 3-metacarpal bone dysplasia syndrome	Disorder		2 *Cases*
100012	Lissencephaly with cerebellar hypoplasia type B	Disorder		50 *Cases*
100013	Lissencephaly with cerebellar hypoplasia type C	Disorder		2 *Cases*
533	Listeriosis	Disorder	0.43 *I**	
533	Listeriosis	Disorder	0.337 *I*	
158673	Localized dystrophic epidermolysis bullosa, acral form	Subtype of disorder		10 *Families*
158676	Localized dystrophic epidermolysis bullosa, nails only	Subtype of disorder		10 *Families*
79410	Localized dystrophic epidermolysis bullosa, pretibial form	Subtype of disorder		40 *Families*
251393	Localized junctional epidermolysis bullosa	Disorder		20 *Cases*
90398	Localized lichen myxedematosus with mixed features of different subtypes	Subtype of disorder		10 *Cases*
90399	Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms	Subtype of disorder		5 *Cases*
2406	Locked-in syndrome	Disorder		33 *Cases*
60030	Loeys-Dietz syndrome	Disorder		52 *Families*
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Disorder	1.0 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Disorder	8.0 *P**	
2408	Lowe-Kohn-Cohen syndrome	Disorder		1 *Family*
2487	Lower limb malformation-hypospadias syndrome	Disorder		2 *Cases*
276435	Lower motor neuron syndrome with late-adult onset	Disorder		55 *Cases*
844	Lown-Ganong-Levine syndrome	Disorder		12 *Cases*
2409	Lowry-MacLean syndrome	Disorder		3 *Cases*
1824	Lowry-Wood syndrome	Disorder		8 *Cases*
1120	Lung agenesis-heart defect-thumb anomalies syndrome	Disorder		9 *Cases*
137631	Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome	Disorder		2 *Cases*
90283	Lupus erythematosus tumidus	Disorder		250 *Cases*
597738	Luscan-Lumish syndrome	Disorder		11 *Cases*
91546	Lyme disease	Disorder	177.5 *I**	
91546	Lyme disease	Disorder	21.9 *I*	
538	Lymphangioleiomyomatosis	Disorder	0.0135 *I*	
538	Lymphangioleiomyomatosis	Disorder	0.25 *P**	
538	Lymphangioleiomyomatosis	Disorder	0.15 *P*	
86915	Lymphedema-atrial septal defects-facial changes syndrome	Disorder		5 *Cases*
86914	Lymphedema-cerebral arteriovenous anomaly-primary pulmonary hypertension syndrome	Disorder		5 *Cases*
99141	Lymphedema-posterior choanal atresia syndrome	Disorder		6 *Cases*
275761	Lysosomal acid lipase deficiency	Disorder	2.0 *P**	
398069	MAGEL2-related Prader-Willi-like syndrome	Disorder		28 *Cases*
324972	MAGIC syndrome	Disorder		21 *Cases*
52417	MALT lymphoma	Disorder	0.3 *I**	
52417	MALT lymphoma	Disorder	4.0 *P**	

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397941	MAN1B1-CDG	Disorder		25 *Cases*
171851	MEDNIK syndrome	Disorder		5 *Families*
352328	MEGDEL syndrome	Disorder		67 *Cases*
85282	MEHMO syndrome	Disorder		22 *Cases*
550	MELAS	Disorder	0.6 *P**	
401973	MEND syndrome	Disorder		24 *Cases*
508093	MEPAN syndrome	Disorder		7 *Cases*
485421	MFF-related encephalopathy due to mitochondrial and peroxisomal fission defect	Subtype of disorder		4 *Cases*
79329	MGAT2-CDG	Disorder		13 *Cases*
494433	MIRAGE syndrome	Disorder		19 *Cases*
293822	MITF-related melanoma and renal cell carcinoma predisposition syndrome	Disorder		30 *Families*
497757	MME-related autosomal dominant Charcot Marie Tooth disease type 2	Disorder		19 *Cases*
79330	MOGS-CDG	Disorder		3 *Cases*
2563	MOMO syndrome	Disorder		8 *Cases*
79323	MPDU1-CDG	Disorder		8 *Cases*
79319	MPI-CDG	Disorder		25 *Cases*
263347	MRCS syndrome	Disorder		7 *Cases*
480536	MSH3-related attenuated familial adenomatous polyposis	Subtype of disorder		4 *Cases*
320360	MT-ATP6-related mitochondrial spastic paraplegia	Disorder		5 *Cases*
597874	MTHFS-related developmental delay-microcephaly-short stature-epilepsy syndrome	Disorder		3 *Cases*
498693	MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome	Disorder		4 *Cases*
182050	MYH9-related disease	Disorder	0.3 *P**	

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480491	MYO5B-related progressive familial intrahepatic cholestasis	Subtype of disorder		5 *Cases*
397612	Macrocephaly-developmental delay syndrome	Disorder		9 *Cases*
210548	Macrocephaly-intellectual disability-autism syndrome	Disorder		40 *Cases*
466791	Macrocephaly-intellectual disability-left ventricular non compaction syndrome	Disorder		6 *Cases*
457485	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	Disorder		8 *Cases*
2427	Macrocephaly-short stature-paraplegia syndrome	Disorder		2 *Cases*
2432	Macrosomia-microphthalmia-cleft palate syndrome	Disorder		5 *Cases*
83619	Macrostomia-preauricular tags-external ophthalmoplegia syndrome	Disorder		9 *Cases*
220448	Macrothrombocytopenia with mitral valve insufficiency	Disorder		2 *Cases*
487796	Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome	Disorder		2 *Cases*
91494	Macular coloboma-cleft palate-hallux valgus syndrome	Disorder		2 *Cases*
137867	Madras motor neuron disease	Disorder		200 *Cases*
163634	Maffucci syndrome	Disorder		250 *Cases*
77297	Majeed syndrome	Disorder		4 *Families*
87503	Mal de Meleda	Disorder	1.0 *P*	
420179	Malan overgrowth syndrome	Disorder		20 *Cases*
673	Malaria	Disorder	73.0 *I*	
673	Malaria	Disorder	1.2 *I**	
673	Malaria	Disorder	3.0 *P**	
2234	Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome	Disorder		2 *Cases*
679	Malignant atrophic papulosis	Subtype of disorder		200 *Cases*
276145	Malignant epithelial tumor of salivary glands	Disorder	0.73 *I**	
99915	Malignant granulosa cell tumor of the	Disorder	0.12 *I**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	ovary			
168999	Malignant melanoma of the mucosa	Disorder	0.26 *I**	
168999	Malignant melanoma of the mucosa	Disorder	1.5	
293181	Malignant migrating focal seizures of infancy	Disorder		114 *Cases*
213512	Malignant mixed Müllerian tumor of the ovary	Disorder	0.12 *I**	
3148	Malignant peripheral nerve sheath tumor	Disorder	1.0 *I*	
398987	Malignant teratoma of ovary	Disorder	0.07 *I**	
252212	Malignant triton tumor	Subtype of disorder		170 *Cases*
180242	Malignant tumor of fallopian tubes	Disorder	1.0 *P**	
943	Malonic aciduria	Disorder		34 *Cases*
238744	Mammary-digital-nail syndrome	Disorder		11 *Cases*
363649	Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome	Disorder		21 *Cases*
2457	Mandibuloacral dysplasia	Disorder		40 *Cases*
443995	Mandibulofacial dysostosis with alopecia	Disorder		4 *Cases*
357158	Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome	Disorder		2 *Cases*
79113	Mandibulofacial dysostosis-microcephaly syndrome	Disorder		107 *Cases*
52416	Mantle cell lymphoma	Disorder	3.5 *P**	
511	Maple syrup urine disease	Disorder	0.67 *BP*	
99826	Marburg hemorrhagic fever	Disorder		500 *Cases*
221074	Marchiafava-Bignami disease	Disorder		250 *Cases*
2461	Marden-Walker syndrome	Disorder		50 *Cases*
558	Marfan syndrome	Disorder	25.0 *I**	
558	Marfan syndrome	Disorder	20.0 *P**	
558	Marfan syndrome	Disorder	15.0 *P*	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
2463	Marfanoid habitus-autosomal recessive intellectual disability syndrome	Disorder		4 *Cases*
314041	Marfanoid habitus-inguinal hernia-advanced bone age syndrome	Disorder		2 *Cases*
2464	Marfanoid syndrome, De Silva type	Disorder		6 *Cases*
559	Marinesco-Sjögren syndrome	Disorder		200 *Cases*
560	Marshall syndrome	Disorder		17 *Cases*
561	Marshall-Smith syndrome	Disorder		74 *Cases*
466718	Martinique crinkled retinal pigment epitheliopathy	Disorder		14 *Cases*
2209	Maternal phenylketonuria	Disorder	10.0 *I**	
411712	Maternal riboflavin deficiency	Disorder		2 *Cases*
97678	Maternal uniparental disomy of chromosome 13	Disorder		3 *Cases*
96186	Maternal uniparental disomy of chromosome 20	Disorder		12 *Cases*
96187	Maternal uniparental disomy of chromosome 21	Disorder		2 *Cases*
96188	Maternal uniparental disomy of chromosome 22	Disorder		4 *Cases*
96181	Maternal uniparental disomy of chromosome 6	Disorder		15 *Cases*
2470	Matthew-Wood syndrome	Disorder		43 *Cases*
3109	Mayer-Rokitansky-Küster-Hauser syndrome	Disorder	11.0 *BP*	
2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2	Subtype of disorder	1.0 *BP**	
57782	Mazabraud syndrome	Disorder		54 *Cases*
562	McCune-Albright syndrome	Disorder	0.55 *P**	
2471	McDonough syndrome	Disorder		2 *Families*
2473	McKusick-Kaufman syndrome	Disorder		90 *Cases*
59306	McLeod neuroacanthocytosis syndrome	Disorder		100 *Cases*
3097	Meacham syndrome	Disorder		13 *Cases*
564	Meckel syndrome	Disorder	4.0 *BP*	

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564	Meckel syndrome	Disorder	2.6 *BP**	
70588	Meconium aspiration syndrome	Disorder	2.44 *P**	
57196	Medial condensing osteitis of the clavicle	Disorder		58 *Cases*
2006	Median cleft lip/mandible	Disorder		70 *Cases*
2699	Median nodule of the upper lip	Disorder		4 *Families*
370127	Medich giant platelet syndrome	Disorder		3 *Cases*
42	Medium chain acyl-CoA dehydrogenase deficiency	Disorder	12.0 *BP**	
42	Medium chain acyl-CoA dehydrogenase deficiency	Disorder	6.85 *P*	
1332	Medullary thyroid carcinoma	Disorder	5.0 *P**	
1332	Medullary thyroid carcinoma	Disorder	0.22 *I**	
616	Medulloblastoma	Disorder	0.11 *I**	
616	Medulloblastoma	Disorder	1.0 *P**	
98954	Meesmann corneal dystrophy	Disorder		250 *Cases*
280671	Megaconial congenital muscular dystrophy	Disorder		19 *Cases*
2241	Megacystis-microcolon-intestinal hypoperistalsis syndrome	Disorder		230 *Cases*
2478	Megalencephalic leukoencephalopathy with subcortical cysts	Disorder		100 *Cases*
60040	Megalencephaly-capillary malformation-polymicrogyria syndrome	Disorder		170 *Cases*
83473	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	Disorder		62 *Cases*
457359	Megalencephaly-severe kyphoscoliosis-overgrowth syndrome	Disorder		2 *Cases*
2482	Melhem-Fahl syndrome	Disorder		2 *Cases*
2484	Melnick-Needles syndrome	Disorder		70 *Cases*
2485	Melorheostosis	Disorder	0.09 *P**	
1879	Melorheostosis with osteopoikilosis	Disorder		5 *Families*
99898	Mendelian susceptibility to	Disorder		31 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	mycobacterial diseases due to complete IFNgammaR1 deficiency			
319547	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency	Disorder		13 *Cases*
319558	Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency	Disorder		49 *Cases*
319552	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	Disorder		180 *Cases*
319563	Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency	Disorder		6 *Cases*
319600	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency	Disorder		2 *Cases*
319595	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency	Disorder		17 *Cases*
592574	Menke-Hennekam syndrome	Disorder		27 *Cases*
565	Menkes disease	Disorder	0.33 *BP**	
498251	Menstrual cycle-dependent periodic fever	Disorder		5 *Cases*
157801	Mesoaxial synostotic syndactyly with phalangeal reduction	Disorder		6 *Families*
2496	Mesomelia-synostoses syndrome	Disorder		10 *Cases*
2631	Mesomelic dwarfism-cleft palate-camptodactyly syndrome	Disorder		2 *Cases*
1836	Mesomelic dysplasia, Kantaputra type	Disorder		5 *Families*
2499	Metachondromatosis	Disorder		25 *Cases*
512	Metachromatic leukodystrophy	Disorder	1.47 *BP**	
512	Metachromatic leukodystrophy	Disorder	0.1 *P**	
1040	Metaphyseal anadysplasia	Disorder		27 *Cases*
33067	Metaphyseal chondrodysplasia, Jansen type	Disorder		16 *Cases*
166038	Metaphyseal chondrodysplasia, Kaitila type	Disorder		2 *Cases*
2501	Metaphyseal chondrodysplasia, Spahr type	Disorder		18 *Cases*

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2502	Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome	Disorder		3 *Cases*
2504	Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome	Disorder		2 *Families*
213531	Metaplastic carcinoma of the breast	Disorder	0.06 *I**	
2635	Metatropic dysplasia	Disorder		81 *Cases*
2635	Metatropic dysplasia	Disorder	0.2 *BP**	
1923	Methimazole embryofetopathy	Disorder		40 *Cases*
168598	Methionine adenosyltransferase I/III deficiency	Disorder		2 *Cases*
565782	Methotrexate toxicity	Disorder	3.0 *P**	
2169	Methylcobalamin deficiency type cblE	Subtype of disorder		27 *Cases*
2170	Methylcobalamin deficiency type cblG	Subtype of disorder		33 *Cases*
308425	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	Disorder		7 *Cases*
26	Methylmalonic acidemia with homocystinuria	Disorder		500 *Cases*
79284	Methylmalonic acidemia with homocystinuria type cblF	Subtype of disorder		15 *Cases*
79282	Methylmalonic acidemia with homocystinuria, type cblC	Subtype of disorder		500 *Cases*
79283	Methylmalonic acidemia with homocystinuria, type cblD	Subtype of disorder		17 *Cases*
369955	Methylmalonic acidemia with homocystinuria, type cblJ	Subtype of disorder		2 *Cases*
369962	Methylmalonic acidemia with homocystinuria, type cblX	Subtype of disorder		18 *Cases*
280183	Methylmalonic aciduria due to transcobalamin receptor defect	Disorder		5 *Cases*
309025	Mevalonate kinase deficiency	Disorder		300 *Cases*
29	Mevalonic aciduria	Subtype of disorder		30 *Cases*
2510	Micro syndrome	Disorder		203 *Cases*
2511	Microbrachycephaly-ptosis-cleft lip syndrome	Disorder		2 *Cases*
468631	Microcephalic cortical malformations-short stature due to RTTN deficiency	Disorder		28 *Cases*
85172	Microcephalic osteodysplastic dysplasia, Saul-Wilson type	Disorder		4 *Cases*

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2637	Microcephalic osteodysplastic primordial dwarfism type II	Disorder		150 *Cases*
2636	Microcephalic osteodysplastic primordial dwarfism types I and III	Disorder		53 *Cases*
329228	Microcephalic primordial dwarfism due to ZNF335 deficiency	Disorder		10 *Cases*
319675	Microcephalic primordial dwarfism, Dauber type	Disorder		2 *Cases*
2617	Microcephalic primordial dwarfism, Montreal type	Disorder		3 *Cases*
2643	Microcephalic primordial dwarfism, Toriello type	Disorder		2 *Cases*
436182	Microcephalic primordial dwarfism-insulin resistance syndrome	Disorder		2 *Cases*
2513	Microcephaly-albinism-digital anomalies syndrome	Disorder		2 *Cases*
3433	Microcephaly-brachydactyly-kyphoscoliosis syndrome	Disorder		3 *Cases*
2523	Microcephaly-brain defect-spasticity-hypernatremia syndrome	Disorder		3 *Cases*
294016	Microcephaly-capillary malformation syndrome	Disorder		10 *Cases*
2516	Microcephaly-cardiac defect-lung malsegmentation syndrome	Disorder		3 *Cases*
2515	Microcephaly-cardiomyopathy syndrome	Disorder		3 *Cases*
329332	Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome	Disorder		4 *Cases*
2522	Microcephaly-cervical spine fusion anomalies syndrome	Disorder		2 *Cases*
2521	Microcephaly-cleft palate-abnormal retinal pigmentation syndrome	Disorder		3 *Cases*
423894	Microcephaly-complex motor and sensory axonal neuropathy syndrome	Disorder		3 *Cases*
488168	Microcephaly-congenital cataract-psoriasiform dermatitis syndrome	Disorder		5 *Cases*
500159	Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrom	Disorder		4 *Cases*
457284	Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome	Disorder		5 *Cases*
2533	Microcephaly-deafness-intellectual disability syndrome	Disorder		2 *Cases*
521445	Microcephaly-facial dysmorphism-ocular anomalies-multiple congenital anomalies syndrome	Disorder		10 *Cases*

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217026	Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type	Disorder		5 *Cases*
2172	Microcephaly-glomerulonephritis-marfanoid habitus syndrome	Disorder		2 *Cases*
457351	Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome	Disorder		14 *Cases*
2526	Microcephaly-lymphedema-chorioretinopathy syndrome	Disorder		50 *Families*
2528	Microcephaly-microcornea syndrome, Seemanova type	Disorder		2 *Cases*
572768	Microcephaly-micromelia syndrome	Subtype of disorder		32 *Cases*
171703	Microcephaly-polymicrogyria-corpus callosum agenesis syndrome	Disorder		4 *Cases*
2519	Microcephaly-seizures-intellectual disability-heart disease syndrome	Disorder		2 *Cases*
423306	Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome	Disorder		2 *Cases*
572773	Microcephaly-short stature-limb abnormalities syndrome	Subtype of disorder		29 *Cases*
397951	Microcephaly-thin corpus callosum-intellectual disability syndrome	Disorder		4 *Cases*
2536	Microcornea-glaucoma-absent frontal sinuses syndrome	Disorder		5 *Cases*
369970	Microcornea-myopic chorioretinal atrophy-telecanthus syndrome	Disorder		14 *Cases*
231736	Microcornea-posterior megalolenticonus-persistent fetal vasculature-coloboma syndrome	Disorder		8 *Cases*
83642	Microcytic anemia with liver iron overload	Disorder		3 *Cases*
217377	Microduplication Xp11.22p11.23 syndrome	Disorder		12 *Cases*
2538	Microgastria-limb reduction defect syndrome	Disorder		16 *Cases*
476126	Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	Disorder		4 *Cases*
50810	Microlissencephaly-micromelia syndrome	Disorder		2 *Cases*
139471	Microphthalmia with brain and digit anomalies	Disorder		2 *Families*
1106	Microphthalmia with limb anomalies	Disorder		35 *Families*
2556	Microphthalmia with linear skin defects syndrome	Disorder		55 *Cases*
77299	Microphthalmia-brain atrophy syndrome	Disorder		3 *Cases*

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2547	Microphthalmia-microtia-fetal akinesia syndrome	Disorder		2 *Cases*
251279	Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	Disorder		9 *Cases*
727	Microscopic polyangiitis	Disorder	1.0 *I**	
727	Microscopic polyangiitis	Disorder	4.2843 *P*	
83463	Microtia	Disorder	13.0 *BP**	
83463	Microtia	Disorder	15.5 *BP*	
139450	Microtia-eye coloboma-imperforation of the nasolacrimal duct syndrome	Disorder		1 *Family*
289522	Microtriplication 11q24.1	Disorder		2 *Cases*
2290	Microvillus inclusion disease	Disorder		137 *Cases*
2557	Mietens syndrome	Disorder		9 *Cases*
2558	Mikati-Najjar-Sahli syndrome	Disorder		5 *Cases*
169799	Mild hemophilia B	Subtype of disorder	0.6 *P**	
93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis	Disorder		4 *Families*
98919	Miller Fisher syndrome	Disorder	0.1 *I**	
531	Miller-Dieker syndrome	Disorder	1.0 *BP**	
352734	Minimal pigment oculocutaneous albinism type 1	Subtype of disorder		10 *Cases*
3004	Mirror polydactyly-vertebral segmentation-limbs defects syndrome	Disorder	0.3 *P**	
1933	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	Disorder		2 *Cases*
255235	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy	Disorder		5 *Cases*
369897	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	Disorder		20 *Cases*
279934	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency	Disorder		100 *Cases*

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363534	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form	Disorder		3 *Cases*
254875	Mitochondrial DNA depletion syndrome, myopathic form	Disorder		45 *Cases*
1349	Mitochondrial DNA-related cardiomyopathy and hearing loss	Disorder		2 *Families*
314637	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	Disorder		8 *Cases*
289560	Mitochondrial membrane protein-associated neurodegeneration	Disorder	0.1 *P*	
2598	Mitochondrial myopathy and sideroblastic anemia	Disorder		7 *Cases*
502423	Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	Disorder		9 *Cases*
2597	Mitochondrial myopathy-lactic acidosis-deafness syndrome	Disorder		2 *Cases*
298	Mitochondrial neurogastrointestinal encephalomyopathy	Disorder	0.1 *P**	
447784	Mitochondrial pyruvate carrier deficiency	Disorder		4 *Cases*
746	Mitochondrial trifunctional protein deficiency	Disorder		100 *Cases*
746	Mitochondrial trifunctional protein deficiency	Disorder	1.0 *P**	
180234	Mixed germ cell tumor	Disorder	0.01 *I**	
324364	Mixed sclerosing bone dystrophy with extra-skeletal manifestations	Disorder		2 *Cases*
90056	Moderate and severe traumatic brain injury	Disorder	37.8 *P**	
169796	Moderate hemophilia B	Subtype of disorder	0.6 *P**	
570	Moebius syndrome	Disorder		300 *Cases*
2560	Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome	Disorder		7 *Cases*
52368	Mohr-Tranebjaerg syndrome	Disorder		91 *Cases*
228423	Monocytopenia with susceptibility to infections	Disorder		22 *Cases*
2565	Mononen-Karnes-Senac syndrome	Disorder		1 *Family*
1598	Monosomy 18p	Disorder	2.0 *BP**	
1600	Monosomy 18q	Disorder	2.5 *BP*	

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48652	Monosomy 22q13.3	Disorder		200 *Cases*
77301	Monosomy 9q22.3	Disorder		42 *Cases*
83467	Morvan syndrome	Disorder		60 *Cases*
329813	Mosaic genome-wide paternal uniparental disomy	Disorder		13 *Cases*
1692	Mosaic trisomy 1	Disorder		1 *Case*
1708	Mosaic trisomy 16	Disorder		226 *Cases*
1711	Mosaic trisomy 17	Disorder		31 *Cases*
1723	Mosaic trisomy 2	Disorder		22 *Cases*
100071	Mosaic trisomy 3	Disorder		6 *Cases*
1747	Mosaic trisomy 7	Disorder		31 *Cases*
99776	Mosaic trisomy 9	Disorder		50 *Cases*
1052	Mosaic variegated aneuploidy syndrome	Disorder		41 *Cases*
3347	Mounier-Kühn syndrome	Disorder		300 *Cases*
2152	Mowat-Wilson syndrome	Disorder	1.7 *BP**	
280679	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	Disorder		9 *Cases*
2573	Moyamoya disease	Disorder	0.035 *I**	
401945	Moyamoya disease with early-onset achalasia	Disorder		9 *Cases*
2574	Moynahan syndrome	Disorder		26 *Cases*
100024	Mu-heavy chain disease	Subtype of disorder		35 *Cases*
398961	Mucinous adenocarcinoma of ovary	Disorder	0.85 *I**	
424053	Mucinous cystadenocarcinoma of the pancreas	Disorder	0.01 *I**	
575	Muckle-Wells syndrome	Disorder		200 *Cases*
576	Mucolipidosis type II	Disorder	0.34 *BP**	
577	Mucolipidosis type III	Disorder	0.985 *I**	

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577	Mucolipidosis type III	Disorder	29.55 *P**	
423461	Mucolipidosis type III alpha/beta	Subtype of disorder	13.0 *P*	
579	Mucopolysaccharidosis type 1	Disorder	1.0 *BP**	
579	Mucopolysaccharidosis type 1	Disorder	0.82 *BP*	
579	Mucopolysaccharidosis type 1	Disorder	0.5 *P**	
580	Mucopolysaccharidosis type 2	Disorder	0.7 *BP**	
580	Mucopolysaccharidosis type 2	Disorder	0.68 *BP*	
580	Mucopolysaccharidosis type 2	Disorder	0.2 *P**	
217085	Mucopolysaccharidosis type 2, severe form	Subtype of disorder	0.4 *BP**	
581	Mucopolysaccharidosis type 3	Disorder	0.87 *BP**	
581	Mucopolysaccharidosis type 3	Disorder	0.3 *P**	
582	Mucopolysaccharidosis type 4	Disorder	0.45 *BP**	
582	Mucopolysaccharidosis type 4	Disorder	0.07 *BP*	
309297	Mucopolysaccharidosis type 4A	Subtype of disorder	15.0 *P**	
583	Mucopolysaccharidosis type 6	Disorder	0.16 *BP**	
583	Mucopolysaccharidosis type 6	Disorder	0.16 *P**	
584	Mucopolysaccharidosis type 7	Disorder	0.01 *P**	
505248	Mucopolysaccharidosis-like syndrome with congenital heart defects and hematopoietic disorders	Disorder		19 *Cases*
566943	Mueller-Weiss syndrome	Disorder		277 *Cases*
53271	Muenke syndrome	Disorder	3.33 *BP*	
2576	Mulibrey nanism	Disorder		150 *Cases*
371428	Multicentric osteolysis-nodulosis-arthropathy spectrum	Disorder		50 *Cases*
139436	Multicentric reticulohistiocytosis	Disorder		200 *Cases*

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1851	Multicystic dysplastic kidney	Disorder	23.26 *BP*	
3282	Multifocal atrial tachycardia	Disorder	0.67 *BP*	
641	Multifocal motor neuropathy	Disorder	1.5 *P*	
2091	Multinodular goiter-cystic kidney-polydactyly syndrome	Disorder		3 *Cases*
500135	Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome	Disorder		3 *Cases*
280633	Multiple congenital anomalies-hypotonia-seizures syndrome	Disorder		15 *Cases*
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	Disorder		24 *Cases*
652	Multiple endocrine neoplasia type 1	Disorder	3.3 *P**	
653	Multiple endocrine neoplasia type 2	Disorder	2.9 *P**	
93311	Multiple epiphyseal dysplasia type 5	Disorder		18 *Families*
166024	Multiple epiphyseal dysplasia, Al-Gazali type	Disorder		4 *Cases*
166011	Multiple epiphyseal dysplasia, Beighton type	Disorder		1 *Family*
166016	Multiple epiphyseal dysplasia, Lowry type	Disorder		2 *Cases*
166032	Multiple epiphyseal dysplasia, with miniepiphyses	Disorder		2 *Cases*
166029	Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia	Disorder		3 *Cases*
2300	Multiple intestinal atresia	Disorder	4.05 *BP*	
401869	Multiple mitochondrial dysfunctions syndrome type 1	Disorder		21 *Cases*
401874	Multiple mitochondrial dysfunctions syndrome type 2	Disorder		6 *Cases*
363424	Multiple mitochondrial dysfunctions syndrome type 3	Disorder		2 *Cases*
457406	Multiple mitochondrial dysfunctions syndrome type 4	Disorder		8 *Cases*
569274	Multiple mitochondrial dysfunctions syndrome type 5	Disorder		6 *Cases*
569290	Multiple mitochondrial dysfunctions syndrome type 6	Disorder		5 *Cases*
29073	Multiple myeloma	Disorder	6.0 *I*	
29073	Multiple myeloma	Disorder	2.4 *I**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
29073	Multiple myeloma	Disorder	11.9 *P**	
321	Multiple osteochondromas	Disorder	3.0 *P**	
324299	Multiple paragangliomas associated with polycythemia	Disorder		2 *Cases*
2215	Multiple pterygium-malignant hyperthermia syndrome	Disorder		4 *Cases*
3151	Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome	Disorder		2 *Cases*
65748	Multiple self-healing squamous epithelioma	Disorder		100 *Cases*
585	Multiple sulfatase deficiency	Disorder		50 *Cases*
3237	Multiple synostoses syndrome	Disorder		30 *Families*
102	Multiple system atrophy	Disorder	3.7 *P**	
102	Multiple system atrophy	Disorder	1.8 *I*	
102	Multiple system atrophy	Disorder	3.5 *P*	
98933	Multiple system atrophy, parkinsonian type	Subtype of disorder	2.4 *P**	
404463	Multisystemic smooth muscle dysfunction syndrome	Disorder		7 *Cases*
370997	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	Disorder		2 *Cases*
2579	Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome	Disorder		12 *Cases*
199340	Muscular dystrophy, Selcen type	Disorder		12 *Cases*
324416	Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome	Disorder		2 *Cases*
2953	Musculocontractural Ehlers-Danlos syndrome	Disorder		34 *Cases*
139578	Mutilating hereditary sensory neuropathy with spastic paraplegia	Disorder		14 *Cases*
659	Mutilating palmoplantar keratoderma with periorificial keratotic plaques	Disorder		73 *Cases*
589	Myasthenia gravis	Disorder	1.7 *I**	
589	Myasthenia gravis	Disorder	7.77 *P*	
589	Myasthenia gravis	Disorder	20.0 *P**	
589	Myasthenia gravis	Disorder	0.53 *I*	

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268249	Mycophenolate mofetil embryopathy	Disorder		25 *Cases*
86850	Myeloid sarcoma	Disorder	0.02 *I**	
86909	Myoclonic epilepsy of infancy	Disorder		106 *Cases*
2589	Myoclonus-cerebellar ataxia-deafness syndrome	Disorder		4 *Cases*
536516	Myopathic Ehlers-Danlos syndrome	Disorder		8 *Cases*
99967	Myxoid/round cell liposarcoma	Subtype of disorder	0.1 *I**	
1655	Müllerian derivatives-lymphangiectasia-polydactyly syndrome	Disorder		8 *Cases*
2491	Müllerian duct anomalies-limb anomalies syndrome	Disorder		5 *Cases*
2608	N syndrome	Disorder		3 *Cases*
555402	NAD(P)HX dehydratase deficiency	Disorder		6 *Cases*
555407	NAD(P)HX epimerase deficiency	Disorder		11 *Cases*
443162	NDE1-related microhydranencephaly	Disorder		1 *Family*
464366	NEK9-related lethal skeletal dysplasia	Disorder		5 *Cases*
447731	NIK deficiency	Disorder		2 *Cases*
263665	NK-cell enteropathy	Disorder		8 *Cases*
527497	NKX6-2-related autosomal recessive hypomyelinating leukodystrophy	Disorder		25 *Cases*
247868	NLRP12-associated hereditary periodic fever syndrome	Disorder		19 *Cases*
3032	NPHP3-related Meckel-like syndrome	Disorder		10 *Cases*
600663	NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	Disorder		11 *Cases*
69087	Naegeli-Franceschetti-Jadassohn syndrome	Disorder	0.035 *P**	
245	Nager syndrome	Disorder		100 *Cases*
423454	Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome	Disorder		6 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
2614	Nail-patella syndrome	Disorder	0.2 *BP**	
2613	Nail-patella-like renal disease	Disorder		3 *Cases*
627	Nance-Horan syndrome	Disorder		196 *Cases*
2073	Narcolepsy type 1	Disorder	30.0 *P**	
2399	Nasopalpebral lipoma-coloboma syndrome	Disorder		19 *Cases*
150	Nasopharyngeal carcinoma	Disorder	2.0 *P**	
150	Nasopharyngeal carcinoma	Disorder	0.36 *I**	
2663	Nathalie syndrome	Disorder		1 *Family*
255229	Navajo neurohepatopathy	Disorder		49 *Cases*
391673	Necrotizing enterocolitis	Disorder	45.0 *P*	
217563	Neonatal acute respiratory distress due to SP-B deficiency	Disorder	0.067 *BP*	
398097	Neonatal antiphospholipid syndrome	Disorder		34 *Cases*
398109	Neonatal autoimmune hemolytic anemia	Disorder		2 *Cases*
398117	Neonatal dermatomyositis	Disorder		3 *Cases*
79118	Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome	Disorder		3 *Cases*
457185	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome	Disorder		11 *Cases*
557064	Neonatal epileptic encephalopathy due to glutaminase deficiency	Disorder		4 *Cases*
446	Neonatal hemochromatosis	Disorder		35 *Cases*
59303	Neonatal ichthyosis-sclerosing cholangitis syndrome	Disorder		12 *Cases*
294023	Neonatal inflammatory skin and bowel disease	Disorder		3 *Cases*
398127	Neonatal scleroderma	Disorder		6 *Cases*
466784	Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect	Disorder		3 *Cases*
94058	Neovascular glaucoma	Disorder	24.4 *P**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
654	Nephroblastoma	Disorder	0.14 *I**	
654	Nephroblastoma	Disorder	10.0 *BP**	
654	Nephroblastoma	Disorder	3.65	
223	Nephrogenic diabetes insipidus	Disorder	0.15 *P**	
3145	Nephrogenic diabetes insipidus-intracranial calcification-short stature-facial dysmorphism syndrome	Disorder		19 *Cases*
93606	Nephrogenic syndrome of inappropriate antidiuresis	Disorder		21 *Cases*
2668	Nephropathy-deafness-hyperparathyroidism syndrome	Disorder		5 *Cases*
2669	Nephrosis-deafness-urinary tract-digital malformations syndrome	Disorder		5 *Cases*
300333	Nephrotic syndrome-epidermolysis bullosa-sensorineural deafness syndrome	Disorder		3 *Cases*
280576	Nestor-Guillermo progeria syndrome	Disorder		2 *Cases*
634	Netherton syndrome	Disorder	0.5 *BP**	
634	Netherton syndrome	Disorder	0.5 *P**	
2671	Neu-Laxova syndrome	Disorder		91 *Cases*
2672	Neuhauser-Eichner-Opitz syndrome	Disorder		5 *Cases*
635	Neuroblastoma	Disorder	11.0 *P**	
635	Neuroblastoma	Disorder	1.26 *I*	
635	Neuroblastoma	Disorder	5.8 *BP**	
2481	Neurocutaneous melanocytosis	Disorder	1.25 *P**	
88639	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	Disorder		20 *Cases*
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency	Disorder		3 *Cases*
529665	Neurodevelopmental delay-seizures-ophthalmic anomalies-osteopenia-cerebellar atrophy syndrome	Disorder		10 *Cases*
453499	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-	Disorder		25 *Cases*

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	skeletal anomalies syndrome			
352665	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome due to 9q21.3 microdeletion	Subtype of disorder		2 *Cases*
453504	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome due to a point mutation	Subtype of disorder		10 *Cases*
33445	Neuroectodermal melanolysosomal disease	Disorder		20 *Cases*
100075	Neuroendocrine tumor of stomach	Disorder	3.2 *P**	
2673	Neurofaciodigitorenal syndrome	Disorder		3 *Cases*
157846	Neuroferritinopathy	Disorder		90 *Cases*
636	Neurofibromatosis type 1	Disorder	21.3 *P**	
636	Neurofibromatosis type 1	Disorder	33.3 *BP*	
1143	Neurogenic arthrogryposis multiplex congenita	Disorder	4.3 *BP**	
85146	Neurogenic scapuloperoneal syndrome, Kaeser type	Disorder		15 *Cases*
137754	Neurological conditions associated with aminoacylase 1 deficiency	Disorder		15 *Cases*
71211	Neuromyelitis optica spectrum disorder	Disorder	0.1877 *I*	
71211	Neuromyelitis optica spectrum disorder	Disorder	2.071 *P*	
139512	Neuropathy with hearing impairment	Disorder		1 *Family*
137596	Neurotrophic keratopathy	Disorder	4.2 *P**	
98908	Neutral lipid storage myopathy	Disorder		36 *Cases*
2690	Neutropenia-monocytopenia-deafness syndrome	Disorder		3 *Cases*
183707	Neutrophil immunodeficiency syndrome	Disorder		2 *Cases*
263432	Nevus of Ito	Disorder	1.17 *P**	
3051	Nicolaides-Baraitser syndrome	Disorder		61 *Cases*
646	Niemann-Pick disease type C	Disorder	1.0 *P**	
1390	Night blindness-skeletal anomalies-	Disorder		2 *Cases*

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	dysmorphism syndrome			
647	Nijmegen breakage syndrome	Disorder	1.0 *BP*	
240760	Nijmegen breakage syndrome-like disorder	Disorder		1 *Case*
99825	Nipah virus disease	Disorder		556 *Cases*
86867	Nodal marginal zone B-cell lymphoma	Disorder	1.0 *P**	
86893	Nodular lymphocyte predominant Hodgkin lymphoma	Disorder	0.12 *I*	
86893	Nodular lymphocyte predominant Hodgkin lymphoma	Disorder	0.095 *I**	
231720	Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome	Disorder		13 *Cases*
2972	Non-eruption of teeth-maxillary hypoplasia-genu valgum syndrome	Disorder		4 *Cases*
91349	Non-functioning pituitary adenoma	Disorder	1.05 *I*	
91349	Non-functioning pituitary adenoma	Disorder	2.55 *I**	
357034	Non-hereditary retinoblastoma	Subtype of disorder	0.038 *I**	
329883	Non-hypoproteinemic hypertrophic gastropathy	Disorder		1 *Family*
363999	Non-immune hydrops fetalis	Subtype of disorder	42.0 *BP*	
209989	Non-papillary transitional cell carcinoma of the bladder	Disorder	37.0 *P**	
314647	Non-progressive cerebellar ataxia with intellectual disability	Disorder		15 *Cases*
363494	Non-seminomatous germ cell tumor of testis	Disorder	1.21 *I**	
363494	Non-seminomatous germ cell tumor of testis	Disorder	33.53	
90031	Non-spherocytic hemolytic anemia due to hexokinase deficiency	Disorder		17 *Families*
1516	Non-syndromic bilambdoid and sagittal craniosynostosis	Disorder		14 *Cases*
3366	Non-syndromic metopic craniosynostosis	Disorder	6.7 *BP**	
3366	Non-syndromic metopic craniosynostosis	Disorder	10.2979 *P**	
95706	Non-syndromic posterior hypospadias	Disorder	19.25 *BP**	
500	Noonan syndrome with multiple	Disorder		296 *Cases*

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	lentigines			
2701	Noonan syndrome-like disorder with loose anagen hair	Disorder		70 *Cases*
649	Norrie disease	Disorder		400 *Cases*
75327	North Carolina macular dystrophy	Disorder		2 *Families*
2760	OSLAM syndrome	Disorder		3 *Cases*
397615	Obesity due to CEP19 deficiency	Subtype of disorder		15 *Cases*
66628	Obesity due to congenital leptin deficiency	Subtype of disorder		30 *Cases*
71526	Obesity due to pro-opiomelanocortin deficiency	Subtype of disorder		7 *Cases*
71528	Obesity due to prohormone convertase I deficiency	Subtype of disorder		16 *Cases*
88643	Obesity-colitis-hypothyroidism-cardiac hypertrophy-developmental delay syndrome	Disorder		2 *Cases*
198	Occipital horn syndrome	Disorder		35 *Cases*
280640	Occipital pachygyria and polymicrogyria	Disorder		3 *Cases*
2704	Ochoa syndrome	Disorder		100 *Cases*
1000	Ocular albinism with late-onset sensorineural deafness	Disorder		9 *Cases*
496790	Ocular anomalies-axonal neuropathy-developmental delay syndrome	Disorder		8 *Cases*
1125	Ocular motor apraxia, Cogan type	Disorder		50 *Cases*
2714	Oculo-palato-cerebral syndrome	Disorder		5 *Cases*
157962	Oculoauricular syndrome, Schorderet type	Disorder		5 *Cases*
398156	Oculoauriculofrontonasal syndrome	Disorder		41 *Cases*
2719	Oculocerebral hypopigmentation syndrome, Cross type	Disorder		14 *Cases*
2720	Oculocerebral hypopigmentation syndrome, Preus type	Disorder		2 *Cases*
1647	Oculocerebrocutaneous syndrome	Disorder		38 *Cases*
2707	Oculocerebrofacial syndrome, Kaufman type	Disorder		19 *Cases*
534	Oculocerebrorenal syndrome of Lowe	Disorder	0.2 *P*	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
534	Oculocerebrorenal syndrome of Lowe	Disorder	0.2 *P**	
352731	Oculocutaneous albinism type 1	Disorder	2.5 *P*	
79431	Oculocutaneous albinism type 1A	Subtype of disorder	1.3 *P*	
79434	Oculocutaneous albinism type 1B	Subtype of disorder	1.3 *P*	
79432	Oculocutaneous albinism type 2	Disorder	2.55 *P*	
79435	Oculocutaneous albinism type 4	Disorder	1.0 *P*	
370091	Oculocutaneous albinism type 5	Disorder		1 *Family*
370097	Oculocutaneous albinism type 6	Disorder		1 *Case*
352745	Oculocutaneous albinism type 7	Disorder		9 *Cases*
597733	Oculocutaneous albinism type 8	Disorder		2 *Cases*
2709	Oculodental syndrome, Rutherfurd type	Disorder		1 *Family*
2710	Oculodentodigital dysplasia	Disorder		243 *Cases*
1876	Oculogastrointestinal muscular dystrophy	Disorder		1 *Family*
1794	Oculomaxillofacial dysostosis	Disorder		4 *Cases*
2713	Oculoosteocutaneous syndrome	Disorder		3 *Cases*
99806	Oculootodental syndrome	Disorder		1 *Family*
557003	Oculoskeletodental syndrome	Disorder		5 *Cases*
2717	Oculotrichoanal syndrome	Disorder		20 *Cases*
2718	Oculotrichodysplasia	Disorder		2 *Cases*
2722	Odonto-onycho dysplasia-alopecia syndrome	Disorder		2 *Cases*
2721	Odonto-onycho-dermal dysplasia	Disorder		30 *Cases*
69082	Odonto-tricho-ungual-digito-palmar syndrome	Disorder		21 *Cases*
166272	Odontochondrodysplasia	Disorder		11 *Cases*
77295	Odontoleukodystrophy	Subtype of disorder		4 *Cases*

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2724	Odontomatosis-aortae esophagus stenosis syndrome	Disorder		3 *Cases*
1811	Odontomicronychial dysplasia	Disorder		5 *Cases*
2723	Odontotrichomelic syndrome	Disorder		4 *Cases*
391655	Off-periods in Parkinson disease not responding to oral treatment	Disorder	4.15 *P**	
276432	Ogden syndrome	Disorder		8 *Cases*
75382	Oguchi disease	Disorder		50 *Cases*
2729	Okamoto syndrome	Disorder		5 *Cases*
85410	Oligoarticular juvenile idiopathic arthritis	Disorder	20.5 *P**	
75378	Oligocone trichromacy	Disorder		14 *Cases*
251627	Oligodendroglioma	Disorder	0.25 *I**	
300576	Oligodontia-cancer predisposition syndrome	Disorder		2 *Families*
2920	Oliver syndrome	Disorder		7 *Cases*
296	Ollier disease	Disorder	1.0 *P**	
39041	Omenn syndrome	Disorder		25 *Cases*
2733	Omodysplasia	Disorder		30 *Cases*
660	Omphalocele	Disorder	11.7 *BP**	
3164	Omphalocele syndrome, Shprintzen-Goldberg type	Disorder		5 *Cases*
496693	Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome	Disorder		7 *Cases*
352540	Oncogenic osteomalacia	Disorder		400 *Cases*
300504	Onychocytic matricoma	Disorder		5 *Cases*
300512	Onychomatricoma	Disorder		50 *Cases*
2741	Ophthalmomandibulomelic dysplasia	Disorder		3 *Cases*
2743	Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome	Disorder		6 *Cases*
2746	Opsismodysplasia	Disorder		30 *Cases*

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1183	Opsoclonus-myoclonus syndrome	Disorder	0.02 *I**	
543470	Optic atrophy-ataxia-peripheral neuropathy-global developmental delay syndrome	Disorder		17 *Cases*
401777	Optic atrophy-intellectual disability syndrome	Disorder		6 *Cases*
2086	Optic pathway glioma	Disorder	0.12 *I*	
508501	Oral-facial-digital syndrome with short stature and brachymesophalangy	Disorder		3 *Cases*
52994	Orbital leiomyoma	Disorder		26 *Cases*
664	Ornithine transcarbamylase deficiency	Disorder	1.0 *P**	
664	Ornithine transcarbamylase deficiency	Disorder	1.77 *BP*	
2750	Orofaciodigital syndrome type 1	Disorder	1.2 *BP**	
141327	Orofaciodigital syndrome type 12	Disorder		1 *Case*
141330	Orofaciodigital syndrome type 13	Disorder		1 *Case*
434179	Orofaciodigital syndrome type 14	Disorder		2 *Families*
2751	Orofaciodigital syndrome type 2	Disorder		20 *Cases*
2752	Orofaciodigital syndrome type 3	Disorder		5 *Cases*
2753	Orofaciodigital syndrome type 4	Disorder		29 *Cases*
2919	Orofaciodigital syndrome type 5	Disorder		12 *Cases*
2754	Orofaciodigital syndrome type 6	Disorder		2 *Families*
2755	Orofaciodigital syndrome type 8	Disorder		20 *Cases*
141007	Orofaciodigital syndrome type 9	Disorder		10 *Cases*
73230	Ossification anomalies-psychomotor developmental delay syndrome	Disorder		2 *Cases*
2764	Osteochondritis dissecans	Disorder	35.0 *P**	
2653	Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome	Disorder		2 *Cases*
2763	Osteocraniostenosis	Disorder		30 *Cases*

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666	Osteogenesis imperfecta	Disorder	8.06 *P*	
216828	Osteogenesis imperfecta type 5	Subtype of disorder		47 *Cases*
2773	Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome	Disorder		2 *Cases*
2645	Osteoglosphonic dysplasia	Disorder		7 *Cases*
2777	Osteomesopyknosis	Disorder		35 *Cases*
2780	Osteopathia striata-cranial sclerosis syndrome	Disorder		100 *Cases*
2779	Osteopathia striata-pigmentary dermopathy-white forelock syndrome	Disorder		3 *Cases*
2324	Osteopenia-intellectual disability-sparse hair syndrome	Disorder		2 *Cases*
2785	Osteopetrosis with renal tubular acidosis	Disorder		100 *Cases*
178389	Osteopetrosis-hypogammaglobulinemia syndrome	Disorder		8 *Cases*
2786	Osteoporosis-oculocutaneous hypopigmentation syndrome	Disorder		1 *Case*
2788	Osteoporosis-pseudoglioma syndrome	Disorder	0.05 *P**	
668	Osteosarcoma	Disorder	0.23 *I**	
668	Osteosarcoma	Disorder	3.17	
178377	Osteosclerosis-developmental delay-craniosynostosis syndrome	Disorder		13 *Cases*
75325	Osteosclerosis-ichthyosis-premature ovarian failure syndrome	Disorder		3 *Cases*
500548	Osteosclerotic metaphyseal dysplasia	Disorder		7 *Cases*
2791	Otodental syndrome	Disorder		10 *Families*
2793	Otoonychoperoneal syndrome	Disorder		6 *Cases*
90652	Otopalatodigital syndrome type 2	Disorder		40 *Cases*
1427	Otospondylomegaepiphyseal dysplasia	Disorder		30 *Cases*
99912	Ovarian dysgerminoma	Disorder	0.04 *I**	
99853	Ovarioleukodystrophy	Subtype of disorder		17 *Cases*
498488	Overgrowth syndrome with 2q37 translocation	Disorder		4 *Cases*

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137634	Overgrowth-macrocephaly-facial dysmorphism syndrome	Disorder		6 *Families*
498485	Overgrowth-metaphyseal undermodeling-spondylar dysplasia syndrome	Disorder		4 *Cases*
3203	Overhydrated hereditary stomatocytosis	Disorder		20 *Families*
991	PAGOD syndrome	Disorder		6 *Cases*
69126	PAPA syndrome	Disorder		53 *Cases*
2825	PARC syndrome	Disorder		2 *Cases*
289478	PASH syndrome	Disorder		36 *Cases*
438134	PCNA-related progressive neurodegenerative photosensitivity syndrome	Disorder		4 *Cases*
439822	PDE4D haploinsufficiency syndrome	Disorder		7 *Cases*
99807	PEHO-like syndrome	Disorder		10 *Cases*
313936	PENS syndrome	Disorder		13 *Cases*
42642	PFAPA syndrome	Disorder		500 *Cases*
319646	PGM1-CDG	Disorder		46 *Cases*
443811	PGM3-CDG	Disorder		20 *Cases*
42775	PHACE syndrome	Disorder		300 *Cases*
2876	PHAVER syndrome	Disorder		2 *Cases*
589905	PHIP-related behavioral problems-intellectual disability-obesity-dysmorphic features syndrome	Disorder		35 *Cases*
568062	PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis	Disorder		10 *Cases*
521426	PLAA-associated neurodevelopmental disorder	Disorder		15 *Cases*
300359	PLCG2-associated antibody deficiency and immune dysregulation	Disorder		3 *Families*
79401	PLEC-related intermediate epidermolysis bullosa simplex without extracutaneous involvement	Disorder		6 *Families*
537072	PLG-related hereditary angioedema with	Subtype of		105 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	normal C1Inh	disorder		
280356	PLIN1-related familial partial lipodystrophy	Disorder		3 *Cases*
476394	PMP2-related Charcot-Marie-Tooth disease type 1	Disorder		13 *Cases*
477817	PMP22-RAI1 contiguous gene duplication syndrome	Disorder		23 *Cases*
480682	POGLUT1-related limb-girdle muscular dystrophy R21	Disorder		4 *Cases*
206564	POMGNT1-related limb-girdle muscular dystrophy R15	Disorder		2 *Cases*
565899	POMGNT2-related limb-girdle muscular dystrophy R24	Disorder		3 *Cases*
206559	POMT2-related limb-girdle muscular dystrophy R14	Disorder		1 *Case*
79083	PPARG-related familial partial lipodystrophy	Disorder		10 *Cases*
412066	PRKAR1B-related neurodegenerative dementia with intermediate filaments	Disorder		12 *Cases*
544469	PRUNE1-related neurological syndrome	Disorder		48 *Cases*
589515	PUM1-associated developmental disability-ataxia-seizure syndrome	Disorder		14 *Cases*
438213	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	Disorder		24 *Cases*
438216	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation	Subtype of disorder		24 *Cases*
481152	PYCR2-related microcephaly-progressive leukoencephalopathy	Disorder		18 *Cases*
2796	Pachydermoperiostosis	Disorder		204 *Cases*
2798	Pachygyria-intellectual disability-epilepsy syndrome	Disorder		5 *Cases*
2309	Pachyonychia congenita	Disorder		1000 *Cases*
180275	Paget disease of the nipple	Disorder	0.51 */**	
1993	Pai syndrome	Disorder		67 *Cases*
300501	Painful orbital and systemic neurofibromas-marfanoid habitus syndrome	Disorder		4 *Cases*
477993	Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	Disorder		3 *Cases*
672	Pallister-Hall syndrome	Disorder		100 *Cases*

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140966	Palmoplantar keratoderma, Nagashima type	Disorder		40 *Cases*
85112	Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome	Disorder		5 *Cases*
2202	Palmoplantar keratoderma-deafness syndrome	Disorder		10 *Families*
2198	Palmoplantar keratoderma-esophageal carcinoma syndrome	Disorder		10 *Families*
538574	Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome	Disorder		23 *Cases*
2201	Palmoplantar keratoderma-spastic paralysis syndrome	Disorder		1 *Family*
556955	Pancreatic agenesis-holoprosencephaly syndrome	Disorder		4 *Cases*
309108	Pancreatic colipase deficiency	Disorder		2 *Cases*
2255	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome	Disorder		10 *Cases*
199337	Pancreatic insufficiency-anemia-hyperostosis syndrome	Disorder		5 *Cases*
677	Pancreatoblastoma	Disorder		60 *Cases*
317473	Pancytopenia due to IKZF1 mutations	Disorder		39 *Cases*
401764	Pancytopenia-developmental delay syndrome	Disorder		3 *Cases*
157850	Pantothenate kinase-associated neurodegeneration	Disorder	0.15 *P**	
319298	Papillary renal cell carcinoma	Disorder	0.14 *I**	
678	Papillon-Lefèvre syndrome	Disorder	0.25 *P*	
2812	Parana hard skin syndrome	Disorder		8 *Cases*
2824	Paraplegia-intellectual disability-hyperkeratosis syndrome	Disorder		6 *Cases*
363478	Paratesticular adenocarcinoma	Disorder	0.01	
143	Parathyroid carcinoma	Disorder	0.02 *I**	
143	Parathyroid carcinoma	Disorder	0.28	
251290	Parietal foramina with clavicular hypoplasia	Disorder		8 *Cases*
851	Paris-Trousseau thrombocytopenia	Disorder		50 *Cases*
53583	Paroxysmal dystonic choreathetosis with	Disorder		20 *Cases*

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	episodic ataxia and spasticity			
98811	Paroxysmal exertion-induced dyskinesia	Disorder		50 *Cases*
46348	Paroxysmal extreme pain disorder	Disorder		4 *Families*
98809	Paroxysmal kinesigenic dyskinesia	Disorder	0.6 *P*	
447	Paroxysmal nocturnal hemoglobinuria	Disorder	2.0 *P**	
98810	Paroxysmal non-kinesigenic dyskinesia	Disorder	0.1 *P*	
1330	Partial atrioventricular septal defect	Disorder	20.0 *BP**	
1330	Partial atrioventricular septal defect	Disorder	30.0 *P**	
1646	Partial chromosome Y deletion	Disorder	20.8 *P*	
1646	Partial chromosome Y deletion	Disorder	20.0 *P**	
401959	Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome	Disorder		2 *Cases*
90076	Partial deep dermal and full thickness burns	Disorder	10.0 *P**	
2805	Partial pancreatic agenesis	Disorder		50 *Cases*
94083	Partington syndrome	Disorder		2 *Families*
86789	Patella aplasia/hypoplasia	Disorder		5 *Families*
228190	Patent ductus arteriosus-bicuspid aortic valve-hand anomalies syndrome	Disorder		7 *Cases*
261304	Paternal 20q13.2q13.3 microdeletion syndrome	Disorder		2 *Cases*
96192	Paternal uniparental disomy of chromosome 7	Disorder		4 *Cases*
2439	Patterson-Stevenson-Fontaine syndrome	Disorder		7 *Cases*
699	Pearson syndrome	Disorder		194 *Cases*
2835	Pectus excavatum-macrocephaly-dysplastic nails syndrome	Disorder		1 *Family*
487809	Pediatric collagenous gastritis	Disorder		24 *Cases*
33402	Pediatric hepatocellular carcinoma	Disorder	0.15 *I**	
263548	Peeling skin syndrome type A	Subtype of disorder		40 *Families*

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263553	Peeling skin syndrome type B	Subtype of disorder		30 *Families*
444138	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	Disorder		4 *Cases*
702	Pelizaeus-Merzbacher disease	Disorder	0.25 *P**	
280219	Pelizaeus-Merzbacher disease, classic form	Subtype of disorder	0.17 *P**	
280210	Pelizaeus-Merzbacher disease, connatal form	Subtype of disorder	0.03 *P**	
280224	Pelizaeus-Merzbacher disease, transitional form	Subtype of disorder	0.03 *P**	
2840	Pelvic dysplasia-arthrogryposis of lower limbs syndrome	Disorder		5 *Cases*
2839	Pelvis-shoulder dysplasia	Disorder		10 *Cases*
93333	Pelviscapular dysplasia	Disorder		4 *Cases*
704	Pemphigus vulgaris	Disorder	18.0 *P**	
705	Pendred syndrome	Disorder	7.0 *P**	
49	Penile agenesis	Disorder		80 *Cases*
1335	Pentalogy of Cantrell	Disorder	0.55 *BP**	
1335	Pentalogy of Cantrell	Disorder	0.67 *BP*	
2847	Pericardial and diaphragmatic defect	Disorder		20 *Cases*
436166	Periodic fever-infantile enterocolitis-autoinflammatory syndrome	Disorder		4 *Cases*
397750	Periodic paralysis with later-onset distal motor neuropathy	Disorder		9 *Cases*
397755	Periodic paralysis with transient compartment-like syndrome	Disorder		4 *Cases*
75392	Periodontal Ehlers-Danlos syndrome	Disorder		62 *Cases*
139426	Perioral myoclonia with absences	Disorder		10 *Cases*
563	Peripartum cardiomyopathy	Disorder	30.0 *BP*	
163746	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	Disorder		40 *Cases*
2400	Peripheral motor neuropathy-	Disorder		2 *Cases*

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	dysautonomia syndrome			
397744	Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome	Disorder		15 *Cases*
168816	Peritoneal cystic mesothelioma	Disorder		150 *Cases*
2849	Perlman syndrome	Disorder		30 *Cases*
65288	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome	Disorder		4 *Cases*
2971	Peroxisomal acyl-CoA oxidase deficiency	Disorder		40 *Cases*
2855	Perrault syndrome	Disorder		124 *Cases*
178509	Perry syndrome	Disorder		53 *Cases*
97341	Persistent placoid maculopathy	Disorder		5 *Cases*
300324	Persistent polyclonal B-cell lymphocytosis	Disorder		154 *Cases*
708	Peters anomaly	Disorder		60 *Cases*
709	Peters plus syndrome	Disorder		100 *Cases*
2869	Peutz-Jeghers syndrome	Disorder	2.2 *BP*	
2869	Peutz-Jeghers syndrome	Disorder	0.4 *P**	
710	Pfeiffer syndrome	Disorder	1.0 *BP**	
2871	Pfeiffer-Palm-Teller syndrome	Disorder		2 *Cases*
2874	Phakomatosis pigmentokeratotica	Disorder		34 *Cases*
352636	Phalangeal microgeodic syndrome	Disorder		50 *Cases*
716	Phenylketonuria	Disorder	11.4 *BP**	
716	Phenylketonuria	Disorder	4.1366 *P*	
716	Phenylketonuria	Disorder	11.5079 *P**	
716	Phenylketonuria	Disorder	6.4 *BP*	
2878	Phocomelia-ectrodactyly-deafness-sinus arrhythmia syndrome	Disorder		4 *Cases*
2880	Phosphoenolpyruvate carboxykinase deficiency	Disorder		10 *Cases*

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3222	Phosphoribosylpyrophosphate synthetase superactivity	Disorder		30 *Families*
498228	Phyllodes tumor of the prostate	Disorder		90 *Cases*
2885	Piebald trait-neurologic defects syndrome	Disorder		8 *Cases*
487825	Pierpont syndrome	Disorder		7 *Cases*
2888	Pierre Robin syndrome-faciodigital anomaly syndrome	Disorder		2 *Cases*
2670	Pierson syndrome	Disorder		98 *Cases*
447961	Pigmentation defects-palmoplantar keratoderma-skin carcinoma syndrome	Disorder		2 *Cases*
251295	Pigmented paravenous retinochoroidal atrophy	Disorder		100 *Cases*
2891	Pili torti-developmental delay-neurological abnormalities syndrome	Disorder		2 *Cases*
2890	Pili torti-onychodysplasia syndrome	Disorder		1 *Family*
2892	Pilodental dysplasia-refractive errors syndrome	Disorder		2 *Cases*
251909	Pineoblastoma	Disorder	0.02 *I**	
300385	Pituitary carcinoma	Disorder	0.04 *I**	
300385	Pituitary carcinoma	Disorder	0.87	
2897	Pityriasis rubra pilaris	Disorder		48 *Cases*
439167	Placental insufficiency	Disorder	33.0 *P*	
99928	Placental site trophoblastic tumor	Disorder	0.02 *I**	
99928	Placental site trophoblastic tumor	Disorder	0.86	
707	Plague	Disorder	2.2 *I**	
454714	Plasma cell leukemia	Disorder	0.04 *I**	
254361	Plectin-related limb-girdle muscular dystrophy R17	Disorder		6 *Cases*
99969	Pleomorphic liposarcoma	Subtype of disorder	0.05 *I**	
454821	Pleomorphic salivary gland adenoma	Subtype of disorder	2.725 *I*	
251607	Pleomorphic xanthoastrocytoma	Disorder	0.01 *I**	

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449266	Pleural empyema	Disorder	13.0 *P**	
50251	Pleural mesothelioma	Disorder	3.1 *P**	
50251	Pleural mesothelioma	Disorder	1.9 *I**	
64742	Pleuropulmonary blastoma	Disorder	0.5 *BP**	
54028	Plummer-Vinson syndrome	Disorder		25 *Cases*
90066	Pneumonia caused by Pseudomonas aeruginosa infection	Disorder	50.0 *P**	
221046	Poikiloderma with neutropenia	Disorder		50 *Cases*
2911	Poland syndrome	Disorder	1.5 *BP**	
767	Polyarteritis nodosa	Disorder	3.16 *P**	
729	Polycythemia vera	Disorder	1.9 *I**	
729	Polycythemia vera	Disorder	30.0 *P**	
2917	Polydactyly-myopia syndrome	Disorder		1 *Family*
453533	Polyendocrine-polyneuropathy syndrome	Disorder		3 *Cases*
397937	Polyglucosan body myopathy type 1	Disorder		11 *Cases*
456369	Polyglucosan body myopathy type 2	Disorder		15 *Cases*
500533	Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	Disorder		17 *Cases*
300573	Polymicrogyria due to TUBB2B mutation	Disorder		36 *Cases*
250972	Polymicrogyria with optic nerve hypoplasia	Disorder		4 *Cases*
732	Polymyositis	Disorder	0.585 *I**	
732	Polymyositis	Disorder	7.1 *P**	
171848	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	Disorder		19 *Cases*
2928	Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome	Disorder		3 *Cases*
2934	Polysyndactyly-cardiac malformation syndrome	Disorder		8 *Cases*
228410	Polyvalvular heart disease syndrome	Disorder		19 *Cases*

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477749	Pontine autosomal dominant microangiopathy with leukoencephalopathy	Disorder		11 *Cases*
269229	Pontine tegmental cap dysplasia	Disorder		22 *Cases*
2254	Pontocerebellar hypoplasia type 1	Disorder		40 *Families*
411493	Pontocerebellar hypoplasia type 10	Disorder		23 *Cases*
2524	Pontocerebellar hypoplasia type 2	Disorder		81 *Families*
97249	Pontocerebellar hypoplasia type 3	Disorder		3 *Families*
166063	Pontocerebellar hypoplasia type 4	Disorder		10 *Families*
166073	Pontocerebellar hypoplasia type 6	Disorder		10 *Cases*
284339	Pontocerebellar hypoplasia type 7	Disorder		4 *Cases*
324569	Pontocerebellar hypoplasia type 8	Disorder		6 *Cases*
369920	Pontocerebellar hypoplasia type 9	Disorder		14 *Cases*
2941	Porencephaly-cerebellar hypoplasia-internal malformations syndrome	Disorder		2 *Cases*
306547	Porencephaly-microcephaly-bilateral congenital cataract syndrome	Disorder		8 *Cases*
166286	Porokeratotic eccrine ostial and dermal duct nevus	Disorder		45 *Cases*
101330	Porphyria cutanea tarda	Disorder	0.6 *I**	
101330	Porphyria cutanea tarda	Disorder	4.0 *P**	
79473	Porphyria variegata	Disorder	0.008 *I**	
79473	Porphyria variegata	Disorder	0.32 *P**	
2703	Port-wine nevi-mega cisterna magna-hydrocephalus syndrome	Disorder		5 *Cases*
70568	Post-transplant lymphoproliferative disease	Disorder	26.2 *P**	
246	Postaxial acrofacial dysostosis	Disorder		30 *Cases*
420584	Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome	Disorder		112 *Cases*
2916	Postaxial polydactyly-dental and vertebral anomalies syndrome	Disorder		3 *Cases*

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2730	Postaxial tetramelic oligodactyly	Disorder		4 *Cases*
98971	Posterior amorphous corneal dystrophy	Disorder		11 *Families*
88628	Posterior column ataxia-retinitis pigmentosa syndrome	Disorder		20 *Cases*
2064	Posterior fusion of lumbosacral vertebrae-blepharoptosis syndrome	Disorder		3 *Cases*
93110	Posterior urethral valve	Disorder	2.0 *P**	
93110	Posterior urethral valve	Disorder	4.125 *BP**	
572013	Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome	Disorder		8 *Cases*
477673	Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	Disorder		17 *Cases*
279947	Postorgasmic illness syndrome	Disorder		45 *Cases*
443236	Postural orthostatic tachycardia syndrome due to NET deficiency	Disorder		2 *Cases*
52022	Potocki-Shaffer syndrome	Disorder		40 *Cases*
217067	Pouchitis	Disorder	22.0 *P**	
397606	PrP systemic amyloidosis	Disorder		16 *Cases*
739	Prader-Willi syndrome	Disorder	3.1 *BP**	
293462	Pre-Descemet corneal dystrophy	Disorder		5 *Cases*
2921	Preaxial polydactyly-colobomata-intellectual disability syndrome	Disorder		2 *Cases*
574918	Predisposition to severe viral infection due to IRF7 deficiency	Disorder		1 *Family*
275555	Preeclampsia	Disorder	45.0 *P**	
486811	Prenatal-onset spinal muscular atrophy with congenital bone fractures	Disorder		7 *Cases*
169464	Primary CD59 deficiency	Disorder		6 *Cases*
289390	Primary Sjögren syndrome	Disorder	48.99 *P**	
289390	Primary Sjögren syndrome	Disorder	6.92 *I*	
186	Primary biliary cholangitis	Disorder	2.57 *I**	

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186	Primary biliary cholangitis	Disorder	3.0 *I*	
186	Primary biliary cholangitis	Disorder	21.05 *P*	
186	Primary biliary cholangitis	Disorder	25.0 *P**	
244	Primary ciliary dyskinesia	Disorder	5.0 *BP**	
247522	Primary ciliary dyskinesia-retinitis pigmentosa syndrome	Disorder		20 *Cases*
98807	Primary dystonia, DYT13 type	Disorder		8 *Cases*
370103	Primary dystonia, DYT17 type	Disorder		3 *Cases*
306734	Primary dystonia, DYT21 type	Disorder		16 *Cases*
464440	Primary dystonia, DYT27 type	Disorder		5 *Cases*
98805	Primary dystonia, DYT4 type	Disorder		22 *Cases*
98806	Primary dystonia, DYT6 type	Disorder		53 *Cases*
48686	Primary effusion lymphoma	Disorder		200 *Cases*
100085	Primary hepatic neuroendocrine carcinoma	Disorder	0.2 *I*	
369929	Primary hyperaldosteronism-seizures-neurological abnormalities syndrome	Disorder		2 *Cases*
2232	Primary hypergonadotropic hypogonadism-partial alopecia syndrome	Disorder		7 *Cases*
93599	Primary hyperoxaluria type 2	Subtype of disorder		10 *Cases*
93600	Primary hyperoxaluria type 3	Subtype of disorder		50 *Cases*
306516	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis	Disorder		200 *Cases*
2196	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement	Subtype of disorder		72 *Cases*
31043	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement	Subtype of disorder		110 *Cases*
30924	Primary hypomagnesemia with secondary hypocalcemia	Disorder		100 *Cases*
564178	Primary hypomagnesemia-refractory seizures-intellectual disability syndrome	Disorder		3 *Cases*
90023	Primary immunodeficiency syndrome due to LAMTOR2 deficiency	Disorder		4 *Cases*

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75391	Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency	Disorder		4 *Cases*
431166	Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection	Disorder		1 *Case*
458768	Primary intralymphatic angioendothelioma	Disorder		30 *Cases*
35689	Primary lateral sclerosis	Disorder	1.5 *P**	
98838	Primary mediastinal large B-cell lymphoma	Disorder	5.0 *P**	
54370	Primary membranoproliferative glomerulonephritis	Disorder	16.0 *P**	
97560	Primary membranous glomerulonephritis	Disorder	0.8103 *I*	
97560	Primary membranous glomerulonephritis	Disorder	0.9194 *I**	
306558	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome	Disorder		8 *Cases*
391408	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	Disorder		8 *Cases*
824	Primary myelofibrosis	Disorder	1.0 *I**	
824	Primary myelofibrosis	Disorder	3.0 *P**	
238606	Primary orthostatic tremor	Disorder		390 *Cases*
314566	Primary progressive apraxia of speech	Disorder		16 *Cases*
171	Primary sclerosing cholangitis	Disorder	0.77 *I**	
171	Primary sclerosing cholangitis	Disorder	8.1 *P*	
171	Primary sclerosing cholangitis	Disorder	7.84 *P**	
171	Primary sclerosing cholangitis	Disorder	0.65 *I*	
314701	Primary systemic amyloidosis	Subtype of disorder	30.0 *P**	
565612	Primary triglyceride deposit cardiomyovasculopathy	Disorder		200 *Cases*
854	Primitive portal vein thrombosis	Disorder	1.72 *I**	
2959	Progeria-short stature-pigmented nevi syndrome	Disorder		11 *Cases*
300382	Progeroid and marfanoid aspect-	Disorder		7 *Cases*

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	lipodystrophy syndrome			
435953	Progeroid features-hepatocellular carcinoma predisposition syndrome	Disorder		3 *Cases*
2963	Progeroid syndrome, Petty type	Disorder		1 *Case*
448251	Progressive autosomal recessive ataxia-deafness syndrome	Disorder		13 *Cases*
75373	Progressive bifocal chorioretinal atrophy	Disorder		2 *Families*
139447	Progressive cavitating leukoencephalopathy	Disorder		19 *Cases*
247198	Progressive cerebello-cerebral atrophy	Disorder		7 *Cases*
431361	Progressive encephalopathy with leukodystrophy due to DECR deficiency	Disorder		2 *Cases*
457212	Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	Disorder		5 *Cases*
352447	Progressive external ophthalmoplegia-myopathy-emaciation syndrome	Disorder		6 *Cases*
480483	Progressive familial intrahepatic cholestasis type 4	Subtype of disorder		14 *Cases*
480476	Progressive familial intrahepatic cholestasis type 5	Subtype of disorder		4 *Cases*
477814	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome	Disorder		9 *Cases*
263516	Progressive myoclonic epilepsy type 3	Subtype of disorder		9 *Families*
402082	Progressive myoclonic epilepsy type 5	Disorder		3 *Cases*
280620	Progressive myoclonic epilepsy type 6	Disorder		12 *Cases*
435438	Progressive myoclonic epilepsy type 7	Disorder		13 *Cases*
424027	Progressive myoclonic epilepsy type 8	Disorder		4 *Cases*
457265	Progressive myoclonic epilepsy type 9	Disorder		2 *Cases*
352596	Progressive myoclonic epilepsy with dystonia	Disorder		5 *Cases*
100070	Progressive non-fluent aphasia	Disorder	2.5 *P**	
100070	Progressive non-fluent aphasia	Disorder	0.7 *I**	
2062	Progressive non-infectious anterior vertebral fusion	Disorder		67 *Cases*

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217396	Progressive polyneuropathy with bilateral striatal necrosis	Disorder		4 *Cases*
352718	Progressive retinal dystrophy due to retinol transport defect	Disorder		5 *Cases*
447977	Progressive scapulohumeroperoneal distal myopathy	Disorder		33 *Cases*
228012	Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	Disorder		4 *Families*
457395	Progressive spondyloepimetaphyseal dysplasia-short stature-short fourth metatarsals-intellectual disability syndrome	Disorder		4 *Cases*
683	Progressive supranuclear palsy	Disorder	0.65 *I*	
683	Progressive supranuclear palsy	Disorder	14.0 *P**	
683	Progressive supranuclear palsy	Disorder	5.26 *P*	
240103	Progressive supranuclear palsy-corticobasal syndrome	Subtype of disorder	0.6 *P**	
240112	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome	Subtype of disorder		10 *Cases*
742	Prolidase deficiency	Disorder		90 *Cases*
2083	Prominent glabella-microcephaly-hypogenitalism syndrome	Disorder		2 *Cases*
35	Propionic acidemia	Disorder	1.5 *I*	
35	Propionic acidemia	Disorder	0.2 *P**	
324977	Proteasome-associated autoinflammatory syndrome	Disorder		40 *Cases*
251598	Protoplasmic astrocytoma	Subtype of disorder	0.01 *I**	
261197	Proximal 16p11.2 microdeletion syndrome	Disorder	20.0 *P**	
401768	Proximal myopathy with extrapyramidal signs	Disorder		15 *Cases*
521305	Proximal myopathy with focal depletion of mitochondria	Disorder		4 *Cases*
606	Proximal myotonic myopathy	Disorder	1.0 *P**	
70	Proximal spinal muscular atrophy	Disorder	2.6 *I**	
70	Proximal spinal muscular atrophy	Disorder	20.0 *BP**	
83330	Proximal spinal muscular atrophy type 1	Subtype of disorder	0.26 *I**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
83418	Proximal spinal muscular atrophy type 2	Subtype of disorder	2.0 *BP**	
52530	Pseudo-von Willebrand disease	Disorder		60 *Cases*
750	Pseudoachondroplasia	Disorder	3.3 *P*	
221120	Pseudoaminopterin syndrome	Disorder		11 *Cases*
85174	Pseudodiastrophic dysplasia	Disorder		13 *Cases*
757	Pseudohypoaldosteronism type 2	Disorder		180 *Cases*
300525	Pseudohypoaldosteronism type 2D	Subtype of disorder		24 *Cases*
300530	Pseudohypoaldosteronism type 2E	Subtype of disorder		17 *Cases*
2976	Pseudoleprechaunism syndrome, Patterson type	Disorder		2 *Cases*
26790	Pseudomyxoma peritonei	Disorder	0.1 *I*	
26790	Pseudomyxoma peritonei	Disorder	2.0 *P**	
2985	Pseudoprogeria syndrome	Disorder		2 *Cases*
758	Pseudoxanthoma elasticum	Disorder	2.5 *P**	
436274	Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa	Disorder		13 *Cases*
280794	Pseudoxanthomatous diffuse cutaneous mastocytosis	Subtype of disorder		10 *Cases*
505242	Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome	Disorder		6 *Cases*
1578	Pterin-4 alpha-carbinolamine dehydratase deficiency	Subtype of disorder		21 *Cases*
2988	Pterygium colli-intellectual disability-digital anomalies syndrome	Disorder		2 *Cases*
2999	Ptosis-strabismus-ectopic pupils syndrome	Disorder		1 *Family*
228396	Ptosis-upper ocular movement limitation-absence of lacrimal punctum syndrome	Disorder		3 *Cases*
2997	Ptosis-vocal cord paralysis syndrome	Disorder		2 *Cases*
2038	Pulmonary arteriovenous malformation	Disorder	2.5 *I*	
64741	Pulmonary blastoma	Disorder		350 *Cases*

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199241	Pulmonary capillary hemangiomatosis	Disorder		100 *Cases*
210136	Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome	Disorder		4 *Cases*
217080	Pulmonary fungal infections in patients deemed at risk	Disorder	22.0 *P**	
411703	Pulmonary non-tuberculous mycobacterial infection	Disorder	6.0 *P**	
31837	Pulmonary venoocclusive disease	Disorder	0.015 *I**	
99710	Punctate acrokeratoderma freckle-like pigmentation	Disorder		7 *Cases*
79501	Punctate palmoplantar keratoderma type 1	Disorder		437 *Cases*
79502	Punctate palmoplantar keratoderma type 2	Disorder		13 *Cases*
69084	Pure hair and nail ectodermal dysplasia	Disorder		20 *Cases*
760	Purine nucleoside phosphorylase deficiency	Disorder		72 *Cases*
763	Pycnodysostosis	Disorder	0.13 *P*	
3003	Pyknoachondrogenesis	Disorder		5 *Cases*
3005	Pyle disease	Disorder		30 *Cases*
48104	Pyoderma gangrenosum	Disorder	0.74 *I*	
2561	Pyramidal molars-abnormal upper lip syndrome	Disorder		8 *Cases*
3006	Pyridoxine-dependent epilepsy	Disorder	0.2 *BP**	
3008	Pyruvate carboxylase deficiency	Disorder	0.4 *BP**	
353320	Pyruvate carboxylase deficiency, benign type	Subtype of disorder		5 *Cases*
2394	Pyruvate dehydrogenase E3 deficiency	Subtype of disorder		20 *Cases*
781	Q fever	Disorder	0.16 *I**	
3010	Qazi-Markouizos syndrome	Disorder		3 *Cases*
3021	RAPADILINO syndrome	Disorder		20 *Cases*
438114	RARS-related autosomal recessive hypomyelinating leukodystrophy	Disorder		4 *Cases*
268114	RAS-associated autoimmune leukoproliferative disease	Disorder		20 *Cases*

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217330	REN-related autosomal dominant tubulointerstitial kidney disease	Subtype of disorder		35 *Families*
494344	RERE-related neurodevelopmental syndrome	Disorder		10 *Cases*
244310	RFT1-CDG	Disorder		8 *Cases*
140976	RHYNS syndrome	Disorder		4 *Cases*
420741	RIDDLE syndrome	Disorder		2 *Cases*
217335	RIN2 syndrome	Disorder		10 *Cases*
544503	RNF13-related severe early-onset epileptic encephalopathy	Disorder		3 *Cases*
93321	Radial hemimelia	Disorder	2.5 *BP*	
2252	Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome	Disorder		8 *Cases*
3026	Radial ray hypoplasia-choanal atresia syndrome	Disorder		3 *Cases*
70475	Radiation proctitis	Disorder	35.0 *P**	
3015	Radio-renal syndrome	Disorder		4 *Cases*
71289	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome	Disorder		20 *Cases*
3270	Radioulnar synostosis-developmental delay-hypotonia syndrome	Disorder		4 *Cases*
3268	Radioulnar synostosis-microcephaly-scoliosis syndrome	Disorder		13 *Cases*
3019	Ramon syndrome	Disorder		8 *Cases*
1051	Ramos-Arroyo syndrome	Disorder		6 *Cases*
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	Disorder		96 *Cases*
71517	Rapid-onset dystonia-parkinsonism	Disorder		100 *Cases*
213528	Rare adenocarcinoma of the breast	Disorder	3.55 *I**	
1929	Rasmussen subacute encephalitis	Disorder		100 *Cases*
99852	Ravine syndrome	Disorder		38 *Cases*
461	Recessive X-linked ichthyosis	Disorder	15.0 *I**	

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461	Recessive X-linked ichthyosis	Disorder	16.6 *P**	
79409	Recessive dystrophic epidermolysis bullosa inversa	Disorder		100 *Cases*
280384	Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome	Disorder		12 *Cases*
90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients	Disorder	7.0 *P**	
480864	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome	Disorder		24 *Cases*
97239	Reducing body myopathy	Disorder		4 *Families*
86839	Refractory anemia with excess blasts	Disorder	0.15 *I**	
168960	Refractory anemia with excess blasts in transformation	Disorder	0.04 *I**	
773	Refsum disease	Disorder	0.1 *P**	
773	Refsum disease	Disorder		60 *Cases*
83450	Regional odontodysplasia	Disorder		140 *Cases*
448267	Regressive spondylometaphyseal dysplasia	Disorder		2 *Cases*
98961	Reis-Bücklers corneal dystrophy	Disorder		81 *Cases*
728	Relapsing polychondritis	Disorder	0.35 *I*	
1848	Renal agenesis, bilateral	Subtype of disorder	1.7 *BP**	
93100	Renal agenesis, unilateral	Subtype of disorder	50.0 *BP*	
2838	Renal caliceal diverticuli-deafness syndrome	Disorder		4 *Cases*
1475	Renal coloboma syndrome	Disorder		180 *Cases*
93108	Renal dysplasia	Disorder	43.5 *BP**	
3242	Renpenning syndrome	Disorder		64 *Cases*
566231	Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha	Disorder		35 *Cases*
99832	Resistance to thyrotropin-releasing	Disorder		2 *Cases*

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	hormone syndrome			
1662	Restrictive dermopathy	Disorder		30 *Cases*
33355	Reticular dysgenesis	Disorder	0.03 *I**	
178307	Reticulate acropigmentation of Kitamura	Disorder		130 *Cases*
458763	Retiform hemangioendothelioma	Disorder		32 *Cases*
75326	Retinal arterial tortuosity	Disorder		100 *Cases*
1574	Retinal degeneration-nanophthalmos-glaucoma syndrome	Disorder		7 *Cases*
397758	Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies	Disorder		14 *Cases*
313800	Retinal dystrophy-optic nerve edema-splenomegaly-anhidrosis-migraine headache syndrome	Disorder		3 *Cases*
3018	Retinal ischemic syndrome-digestive tract small vessel hyalinosis-diffuse cerebral calcifications syndrome	Disorder		3 *Cases*
319640	Retinal macular dystrophy type 2	Disorder		5 *Families*
791	Retinitis pigmentosa	Disorder	30.0 *P**	
791	Retinitis pigmentosa	Disorder	26.7 *P*	
494439	Retinitis pigmentosa-hearing loss-premature aging-short stature-facial dysmorphism syndrome	Disorder		3 *Cases*
3085	Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome	Disorder		2 *Families*
436245	Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome	Disorder		3 *Cases*
52427	Retinitis punctata albescens	Disorder	0.125 *P*	
52427	Retinitis punctata albescens	Disorder	0.175 *P**	
790	Retinoblastoma	Disorder	0.05 *I**	
790	Retinoblastoma	Disorder	6.0 *BP*	
790	Retinoblastoma	Disorder	1.05	
778	Rett syndrome	Disorder	5.0 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
778	Rett syndrome	Disorder	10.0 *P**	
3088	Revesz syndrome	Disorder		4 *Cases*
69077	Rhabdoid tumor	Disorder		500 *Cases*
231108	Rhabdoid tumor predisposition syndrome	Disorder		5 *Families*
780	Rhabdomyosarcoma	Disorder	0.59 *I**	
3099	Rheumatic fever	Disorder	5.0 *I**	
85408	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	Disorder	8.0 *P**	
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	Disorder	4.2 *P**	
177	Rhizomelic chondrodysplasia punctata	Disorder	0.7 *BP**	
468717	Rhizomelic chondrodysplasia punctata type 5	Subtype of disorder		4 *Cases*
2831	Rhizomelic dysplasia, Patterson-Lowry type	Disorder		5 *Cases*
3098	Rhizomelic syndrome, Urbach type	Disorder		3 *Cases*
59315	Rhombencephalosynapsis	Disorder		100 *Cases*
97229	Riboflavin transporter deficiency	Disorder		109 *Cases*
440706	Ribose-5-P isomerase deficiency	Disorder		1 *Case*
3102	Richieri Costa-Pereira syndrome	Disorder		33 *Cases*
3101	Richieri Costa-da Silva syndrome	Disorder		4 *Cases*
83312	Rickettsialpox	Disorder		800 *Cases*
1437	Ring chromosome 1 syndrome	Disorder		35 *Cases*
1438	Ring chromosome 10 syndrome	Disorder		16 *Cases*
96175	Ring chromosome 11 syndrome	Disorder		20 *Cases*
1439	Ring chromosome 12 syndrome	Disorder		10 *Cases*
1440	Ring chromosome 14 syndrome	Disorder		80 *Cases*
96177	Ring chromosome 15 syndrome	Disorder		50 *Cases*

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96178	Ring chromosome 16 syndrome	Disorder		10 *Cases*
1441	Ring chromosome 17 syndrome	Disorder		18 *Cases*
1442	Ring chromosome 18 syndrome	Disorder		70 *Cases*
1443	Ring chromosome 19 syndrome	Disorder		10 *Cases*
96171	Ring chromosome 2 syndrome	Disorder		18 *Cases*
1444	Ring chromosome 20 syndrome	Disorder		50 *Cases*
1446	Ring chromosome 22 syndrome	Disorder		100 *Cases*
96172	Ring chromosome 3 syndrome	Disorder		11 *Cases*
1447	Ring chromosome 4 syndrome	Disorder		20 *Cases*
1448	Ring chromosome 6 syndrome	Disorder		25 *Cases*
1449	Ring chromosome 7 syndrome	Disorder		18 *Cases*
1450	Ring chromosome 8 syndrome	Disorder		8 *Cases*
96173	Ring chromosome 9 syndrome	Disorder		31 *Cases*
91481	Ring dermoid of cornea	Disorder		30 *Cases*
3103	Roberts syndrome	Disorder		150 *Cases*
3104	Robin sequence-oligodactyly syndrome	Disorder		3 *Cases*
97360	Robinow syndrome	Disorder		200 *Cases*
353298	Roifman syndrome	Disorder		17 *Cases*
163727	Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	Disorder		1 *Family*
101016	Romano-Ward syndrome	Disorder	40.0 *P**	
158014	Rosaï-Dorfman disease	Disorder		1000 *Cases*
2909	Rothmund-Thomson syndrome	Disorder		400 *Cases*
221008	Rothmund-Thomson syndrome type 1	Subtype of disorder		100 *Cases*
221016	Rothmund-Thomson syndrome type 2	Subtype of		200 *Cases*

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		disorder		
3111	Rotor syndrome	Disorder		50 *Cases*
83616	Rubella panencephalitis	Disorder		20 *Cases*
783	Rubinstein-Taybi syndrome	Disorder	0.7 *BP**	
353284	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency	Subtype of disorder		34 *Cases*
88618	S-adenosylhomocysteine hydrolase deficiency	Disorder		15 *Cases*
576278	SATB2-associated syndrome	Disorder		171 *Cases*
251028	SATB2-associated syndrome due to a chromosomal rearrangement	Subtype of disorder		20 *Cases*
370052	SCALP syndrome	Disorder		4 *Cases*
3134	SCARF syndrome	Disorder		2 *Cases*
139466	SERKAL syndrome	Disorder		3 *Cases*
597743	SETD2-related microcephaly-severe intellectual disability-multiple congenital anomalies syndrome	Disorder		12 *Cases*
3163	SHORT syndrome	Disorder		32 *Cases*
398079	SIM1-related Prader-Willi-like syndrome	Disorder		4 *Cases*
488437	SIX2-related frontonasal dysplasia	Disorder		1 *Family*
238459	SLC35A1-CDG	Disorder		3 *Cases*
356961	SLC35A2-CDG	Disorder		4 *Cases*
157965	SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome	Subtype of disorder		8 *Cases*
468699	SLC39A8-CDG	Disorder		10 *Cases*
466962	SMARCA4-deficient sarcoma of thorax	Disorder		19 *Cases*
1519	SPECC1L-related hypertelorism syndrome	Disorder		25 *Cases*
93357	SPONASTRIME dysplasia	Disorder		16 *Cases*
324737	SRD5A3-CDG	Disorder		7 *Families*
370927	SSR4-CDG	Disorder		9 *Cases*

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502434	STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome	Disorder		17 *Cases*
438159	STAT3-related early-onset multisystem autoimmune disease	Disorder		19 *Cases*
425120	STING-associated vasculopathy with onset in infancy	Disorder		9 *Cases*
370921	STT3A-CDG	Disorder		2 *Cases*
370924	STT3B-CDG	Disorder		1 *Case*
599373	STXBP1-related encephalopathy	Disorder		282 *Cases*
57145	SUNCT syndrome	Disorder	6.7 *P**	
391351	SURF1-related Charcot-Marie-Tooth disease type 4	Disorder		3 *Cases*
544254	SYNGAP1-related developmental and epileptic encephalopathy	Disorder		57 *Cases*
397927	Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome	Disorder		4 *Cases*
794	Saethre-Chotzen syndrome	Disorder	3.0 *BP**	
300493	Sagliker syndrome	Disorder		60 *Cases*
140969	Saldino-Mainzer syndrome	Disorder		13 *Cases*
213557	Salivary gland type cancer of the breast	Disorder	0.05 *I**	
796	Sandhoff disease	Disorder	0.67 *BP**	
79269	Sanfilippo syndrome type A	Subtype of disorder	0.32 *P**	
79269	Sanfilippo syndrome type A	Subtype of disorder	1.4 *BP*	
79270	Sanfilippo syndrome type B	Subtype of disorder	0.2 *P**	
79271	Sanfilippo syndrome type C	Subtype of disorder	5.0 *P**	
797	Sarcoidosis	Disorder	20.0 *P**	
3129	Sarcosinemia	Disorder	2.0 *BP*	
3130	Satoyoshi syndrome	Disorder		50 *Cases*
3132	Say-Barber-Miller syndrome	Disorder		4 *Cases*

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1003	Scalp defects-postaxial polydactyly syndrome	Disorder		2 *Cases*
2036	Scalp-ear-nipple syndrome	Disorder		30 *Cases*
431255	Scapuloperoneal spinal muscular atrophy	Disorder		31 *Cases*
90080	Scarring in glaucoma filtration surgical procedures	Disorder	22.0 *P**	
2353	Schilbach-Rott syndrome	Disorder		18 *Cases*
1830	Schimke immuno-osseous dysplasia	Disorder		133 *Cases*
798	Schinzel-Giedion syndrome	Disorder		46 *Cases*
37748	Schnitzler syndrome	Disorder		150 *Cases*
98967	Schnyder corneal dystrophy	Disorder		115 *Cases*
800	Schwartz-Jampel syndrome	Disorder		129 *Cases*
50944	Schöpf-Schulz-Passarge syndrome	Disorder		25 *Cases*
185	Scimitar syndrome	Disorder	2.0 *BP**	
167635	Scleromyxedema	Disorder		250 *Cases*
90400	Scleromyxedema without monoclonal gammopathy	Subtype of disorder		15 *Cases*
3152	Sclerosteosis	Disorder		80 *Cases*
806	Scott syndrome	Disorder		4 *Cases*
158029	Sea-blue histiocytosis	Disorder		60 *Cases*
168606	Seborrhea-like dermatitis with psoriasiform elements	Disorder		44 *Cases*
808	Seckel syndrome	Disorder		50 *Cases*
808	Seckel syndrome	Disorder	0.2 *BP**	
140286	Secondary hypoparathyroidism due to impaired parathormon secretion	Disorder	24.75 *P**	
67039	Segmental odontomaxillary dysplasia	Disorder		32 *Cases*
314662	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	Disorder		10 *Cases*
79156	Seizures-intellectual disability due to hydroxylysinuria syndrome	Disorder		6 *Cases*

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466926	Seizures-scoliosis-macrocephaly syndrome	Disorder		10 *Cases*
281122	Self-improving collodion baby	Disorder		25 *Cases*
79411	Self-improving dystrophic epidermolysis bullosa	Disorder		52 *Cases*
217622	Sensorineural deafness with dilated cardiomyopathy	Disorder		2 *Families*
66633	Sensorineural hearing loss-early graying-essential tremor syndrome	Disorder		3 *Cases*
90051	Sepsis in premature infants	Disorder	32.0 *P**	
3157	Septo-optic dysplasia spectrum	Disorder	10.0 *BP**	
157798	Serrated polyposis syndrome	Disorder	1.0 *I*	
3078	Severe X-linked intellectual disability, Gustavson type	Disorder		7 *Cases*
238329	Severe X-linked mitochondrial encephalomyopathy	Disorder		2 *Cases*
85165	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome	Disorder		5 *Cases*
438207	Severe autosomal recessive macrothrombocytopenia	Disorder		2 *Cases*
228003	Severe combined immunodeficiency due to CORO1A deficiency	Disorder		9 *Cases*
420573	Severe combined immunodeficiency due to CTPS1 deficiency	Disorder		12 *Cases*
317425	Severe combined immunodeficiency due to DNA-PKcs deficiency	Disorder		2 *Cases*
169095	Severe combined immunodeficiency due to FOXN1 deficiency	Disorder		9 *Cases*
397787	Severe combined immunodeficiency due to IKK2 deficiency	Disorder		9 *Cases*
504523	Severe combined immunodeficiency due to LAT deficiency	Disorder		3 *Cases*
280142	Severe combined immunodeficiency due to LCK deficiency	Disorder		4 *Cases*
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	Disorder	0.3 *BP**	
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	Disorder	0.2 *P**	
331206	Severe combined immunodeficiency due to complete RAG1/2 deficiency	Disorder	1.0 *P**	
300298	Severe congenital hypochromic anemia with ringed sideroblasts	Disorder		3 *Cases*
369992	Severe dermatitis-multiple allergies-metabolic wasting syndrome	Disorder		3 *Cases*

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329249	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency	Subtype of disorder		13 *Cases*
79404	Severe generalized junctional epidermolysis bullosa	Disorder	0.17 *BP*	
488627	Severe growth deficiency-strabismus-extensive dermal melanocytosis-intellectual disability syndrome	Disorder		3 *Cases*
169802	Severe hemophilia A	Subtype of disorder	2.8 *P**	
169793	Severe hemophilia B	Subtype of disorder	0.8 *P**	
745	Severe hereditary thrombophilia due to congenital protein C deficiency	Disorder	0.16 *BP*	
467176	Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome	Disorder		6 *Cases*
280763	Severe intellectual disability and progressive spastic paraplegia	Disorder		15 *Cases*
466688	Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome	Disorder		6 *Cases*
94066	Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia	Disorder		2 *Cases*
436141	Severe intellectual disability-hypotonia-strabismus-coarse face-planovalgus syndrome	Disorder		6 *Cases*
363686	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	Disorder		4 *Cases*
397933	Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	Disorder		3 *Cases*
404473	Severe intellectual disability-progressive spastic diplegia syndrome	Disorder		4 *Cases*
391307	Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome	Disorder		3 *Cases*
324307	Severe lateral tibial bowing-short stature-mild winged scapula-mild facial dysmorphism syndrome	Disorder		2 *Cases*
1236	Severe microbrachycephaly-intellectual disability-athetoid cerebral palsy syndrome	Disorder		2 *Cases*
369939	Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome	Disorder		7 *Cases*
527450	Severe myopia-generalized joint laxity-	Disorder		5 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	short stature syndrome			
314655	Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion	Subtype of disorder		7 *Cases*
397593	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	Disorder		5 *Cases*
209370	Severe neonatal-onset encephalopathy with microcephaly	Disorder		30 *Cases*
363400	Severe neurodegenerative syndrome with lipodystrophy	Disorder		10 *Cases*
500545	Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract	Disorder		6 *Cases*
2715	Severe oculo-renal-cerebellar syndrome	Disorder		5 *Cases*
411543	Severe phosphoribosylpyrophosphate synthetase superactivity	Subtype of disorder		33 *Cases*
363489	Sex cord-stromal tumor of testis	Disorder	0.02 *I**	
363489	Sex cord-stromal tumor of testis	Disorder	0.44	
810	Shigellosis	Disorder	1.68 *I**	
99063	Shone complex	Disorder		100 *Cases*
66518	Short fifth metacarpals-insulin resistance syndrome	Disorder		6 *Cases*
498497	Short rib-polydactyly syndrome type 5	Disorder		2 *Cases*
93269	Short rib-polydactyly syndrome, Majewski type	Disorder		34 *Cases*
314811	Short stature due to GHSR deficiency	Disorder		8 *Cases*
629	Short stature due to growth hormone qualitative anomaly	Subtype of disorder		3 *Cases*
2867	Short stature, Brussels type	Disorder		2 *Cases*
435804	Short stature-advanced bone age-early-onset osteoarthritis syndrome	Disorder		3 *Families*
397623	Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome	Disorder		4 *Cases*
464288	Short stature-brachydactyly-obesity-global developmental delay syndrome	Disorder		6 *Cases*
2994	Short stature-craniofacial anomalies-genital hypoplasia syndrome	Disorder		3 *Families*
2866	Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome	Disorder		2 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
314394	Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome	Disorder		14 *Cases*
391677	Short stature-optic atrophy-Pelger-Huët anomaly syndrome	Disorder		39 *Cases*
85442	Short stature-pituitary and cerebellar defects-small sella turcica syndrome	Disorder		5 *Families*
589442	Short stature-skeletal dysplasia-retinal degeneration-intellectual disability-sensorineural hearing loss syndrome	Disorder		7 *Cases*
2868	Short stature-valvular heart disease-characteristic facies syndrome	Disorder		3 *Cases*
2865	Short stature-webbed neck-heart disease syndrome	Disorder		4 *Cases*
2863	Short stature-wormian bones-dextrocardia syndrome	Disorder		3 *Cases*
2832	Short tarsus-absence of lower eyelashes syndrome	Disorder		11 *Cases*
357175	Short ulna-dysmorphism-hypotonia-intellectual disability syndrome	Disorder		4 *Cases*
935	Short-limb skeletal dysplasia with severe combined immunodeficiency	Disorder		19 *Cases*
2462	Shprintzen-Goldberg syndrome	Disorder		60 *Cases*
811	Shwachman-Diamond syndrome	Disorder	0.5 *BP*	
811	Shwachman-Diamond syndrome	Disorder	0.28 *P*	
3166	Sialuria	Disorder		5 *Cases*
232	Sickle cell anemia	Disorder	10.0 *P**	
3167	Siegler-Brewer-Carey syndrome	Disorder		2 *Cases*
71276	Silent sinus syndrome	Disorder		558 *Cases*
3168	Sillence syndrome	Disorder		5 *Cases*
813	Silver-Russell syndrome	Disorder	15.5 *I**	
813	Silver-Russell syndrome	Disorder	0.7 *BP**	
397590	Silver-Russell syndrome due to a point mutation	Subtype of disorder		8 *Cases*
373	Simpson-Golabi-Behmel syndrome	Disorder		250 *Cases*
85191	Singleton-Merten dysplasia	Disorder		22 *Cases*
324321	Sinoatrial node dysfunction and deafness	Disorder		8 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
3169	Sirenomelia	Disorder	0.98 *BP*	
3169	Sirenomelia	Disorder	0.71 *BP**	
3169	Sirenomelia	Disorder	0.01 *P*	
3169	Sirenomelia	Disorder	0.009 *P**	
2882	Sitosterolemia	Disorder		100 *Cases*
319	Skeletal Ewing sarcoma	Disorder	0.13 *I**	
319	Skeletal Ewing sarcoma	Disorder	2.33	
508533	Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome	Disorder		12 *Cases*
1858	Skeletal dysplasia-epilepsy-short stature syndrome	Disorder		7 *Cases*
293165	Skin fragility-woolly hair-palmoplantar keratoderma syndrome	Disorder		7 *Cases*
1201	Small bowel atresia	Disorder	9.0 *BP**	
70573	Small cell lung cancer	Disorder	12.0 *P**	
818	Smith-Lemli-Opitz syndrome	Disorder	3.7 *BP**	
819	Smith-Magenis syndrome	Disorder	4.0 *P*	
819	Smith-Magenis syndrome	Disorder	5.35 *P**	
178355	Smith-McCort dysplasia	Disorder		16 *Cases*
820	Sneddon syndrome	Disorder	0.4 *I**	
91496	Snowflake vitreoretinal degeneration	Disorder		50 *Cases*
424065	Solid pseudopapillary carcinoma of pancreas	Disorder	0.003 *I**	
209964	Solitary rectal ulcer syndrome	Disorder	1.0 *I**	
97283	Somatostatinoma	Disorder	0.0025 *I**	
821	Sotos syndrome	Disorder	7.1 *BP*	
821	Sotos syndrome	Disorder	0.5 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
1182	Spastic ataxia with congenital miosis	Disorder		3 *Families*
2572	Spastic ataxia-corneal dystrophy syndrome	Disorder		1 *Family*
557056	Spastic ataxia-dysarthria due to glutaminase deficiency	Disorder		5 *Cases*
2815	Spastic paraparesis-deafness syndrome	Disorder		6 *Cases*
99015	Spastic paraplegia type 2	Disorder		100 *Cases*
329475	Spastic paraplegia-Paget disease of bone syndrome	Disorder		1 *Family*
2819	Spastic paraplegia-facial-cutaneous lesions syndrome	Disorder		5 *Cases*
2818	Spastic paraplegia-glaucoma-intellectual disability syndrome	Disorder		2 *Families*
521390	Spastic paraplegia-intellectual disability-nystagmus-obesity syndrome	Disorder		4 *Cases*
2820	Spastic paraplegia-nephritis-deafness syndrome	Disorder		4 *Cases*
2821	Spastic paraplegia-neuropathy-poikiloderma syndrome	Disorder		1 *Family*
320406	Spastic paraplegia-optic atrophy-neuropathy syndrome	Disorder		75 *Cases*
2826	Spastic paraplegia-precocious puberty syndrome	Disorder		2 *Cases*
464282	Spastic paraplegia-severe developmental delay-epilepsy syndrome	Disorder		16 *Cases*
3011	Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome	Disorder		2 *Cases*
447997	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	Disorder		15 *Cases*
352403	Spectrin-associated autosomal recessive cerebellar ataxia	Disorder		2 *Families*
99865	Spermatocytic seminoma	Disorder	0.03 *I**	
314432	Spigelian hernia-cryptorchidism syndrome	Disorder		15 *Cases*
53721	Spinal arteriovenous metameric syndrome	Disorder		45 *Cases*
1217	Spinal atrophy-ophthalmoplegia-pyramidal syndrome	Disorder		2 *Cases*
90058	Spinal cord injury	Disorder	32.0 *P**	
98920	Spinal muscular atrophy with respiratory distress type 1	Disorder		128 *Cases*
404521	Spinal muscular atrophy with respiratory	Disorder		1 *Case*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	distress type 2			
73245	Spinal muscular atrophy-Dandy-Walker malformation-cataracts syndrome	Disorder		2 *Cases*
2590	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	Disorder		10 *Cases*
98755	Spinocerebellar ataxia type 1	Disorder	1.5 *P*	
98767	Spinocerebellar ataxia type 11	Disorder		51 *Cases*
98762	Spinocerebellar ataxia type 12	Disorder		40 *Families*
98768	Spinocerebellar ataxia type 13	Disorder		20 *Cases*
98763	Spinocerebellar ataxia type 14	Disorder		20 *Families*
98769	Spinocerebellar ataxia type 15/16	Disorder		80 *Cases*
98759	Spinocerebellar ataxia type 17	Disorder		100 *Families*
98771	Spinocerebellar ataxia type 18	Disorder		26 *Cases*
98772	Spinocerebellar ataxia type 19/22	Disorder		12 *Cases*
98756	Spinocerebellar ataxia type 2	Disorder	1.5 *P*	
101110	Spinocerebellar ataxia type 20	Disorder		20 *Cases*
98773	Spinocerebellar ataxia type 21	Disorder		35 *Cases*
101108	Spinocerebellar ataxia type 23	Disorder		4 *Families*
101111	Spinocerebellar ataxia type 25	Disorder		10 *Cases*
101112	Spinocerebellar ataxia type 26	Disorder		1 *Family*
98764	Spinocerebellar ataxia type 27	Disorder		30 *Cases*
208513	Spinocerebellar ataxia type 29	Disorder		50 *Cases*
98757	Spinocerebellar ataxia type 3	Disorder	1.5 *P*	
211017	Spinocerebellar ataxia type 30	Disorder		6 *Cases*
217012	Spinocerebellar ataxia type 31	Disorder		30 *Families*
276183	Spinocerebellar ataxia type 32	Disorder		1 *Family*
1955	Spinocerebellar ataxia type 34	Disorder		45 *Cases*

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276193	Spinocerebellar ataxia type 35	Disorder		28 *Cases*
276198	Spinocerebellar ataxia type 36	Disorder		100 *Families*
363710	Spinocerebellar ataxia type 37	Disorder		9 *Cases*
423296	Spinocerebellar ataxia type 38	Disorder		4 *Families*
423275	Spinocerebellar ataxia type 40	Disorder		5 *Cases*
458798	Spinocerebellar ataxia type 41	Disorder		1 *Case*
458803	Spinocerebellar ataxia type 42	Disorder		25 *Cases*
497764	Spinocerebellar ataxia type 43	Disorder		7 *Cases*
589527	Spinocerebellar ataxia type 45	Disorder		7 *Cases*
589522	Spinocerebellar ataxia type 46	Disorder		1 *Family*
98766	Spinocerebellar ataxia type 5	Disorder		5 *Families*
94124	Spinocerebellar ataxia with axonal neuropathy type 1	Disorder		9 *Cases*
1185	Spinocerebellar ataxia-dysmorphism syndrome	Disorder		3 *Cases*
3177	Spinocerebellar degeneration-corneal dystrophy syndrome	Disorder		2 *Cases*
86854	Splenic marginal zone lymphoma	Disorder	0.5 *P**	
2063	Splenogonadal fusion-limb defects-micrognathia syndrome	Disorder		30 *Cases*
71271	Split hand-split foot-deafness syndrome	Disorder		22 *Cases*
488232	Split-foot malformation-mesoaxial polydactyly syndrome	Disorder		5 *Cases*
228387	Spondylo-megaepiphyseal-metaphyseal dysplasia	Disorder		19 *Cases*
85194	Spondylo-ocular syndrome	Disorder		7 *Cases*
3180	Spondylocamptodactyly syndrome	Disorder		5 *Cases*
3275	Spondylocarpotarsal synostosis	Disorder		35 *Cases*
536471	Spondylodysplastic Ehlers-Danlos syndrome	Disorder		24 *Families*
1855	Spondyloenchondrodysplasia	Disorder		36 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type	Disorder		30 *Cases*
168454	Spondyloepimetaphyseal dysplasia, Geneviève type	Disorder		6 *Families*
99642	Spondyloepimetaphyseal dysplasia, Handigodu type	Disorder		234 *Cases*
370015	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type	Disorder		2 *Cases*
93356	Spondyloepimetaphyseal dysplasia, Missouri type	Disorder		14 *Cases*
93282	Spondyloepimetaphyseal dysplasia, PAPSS2 type	Disorder		17 *Cases*
93352	Spondyloepimetaphyseal dysplasia, Shohat type	Disorder		5 *Cases*
171866	Spondyloepimetaphyseal dysplasia, aggrecan type	Disorder		3 *Cases*
156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type	Disorder		5 *Cases*
168451	Spondyloepimetaphyseal dysplasia-abnormal dentition syndrome	Disorder		2 *Cases*
168443	Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome	Disorder		5 *Cases*
93358	Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome	Disorder		27 *Cases*
94068	Spondyloepiphyseal dysplasia congenita	Disorder	1.0 *BP**	
163665	Spondyloepiphyseal dysplasia tarda, Kohn type	Disorder		3 *Cases*
137678	Spondyloepiphyseal dysplasia with metatarsal shortening	Disorder		13 *Families*
93283	Spondyloepiphyseal dysplasia, Kimberley type	Disorder		1 *Family*
163668	Spondyloepiphyseal dysplasia, MacDermot type	Disorder		4 *Cases*
263482	Spondyloepiphyseal dysplasia, Maroteaux type	Disorder		10 *Cases*
163662	Spondyloepiphyseal dysplasia, Reardon type	Disorder		1 *Family*
459051	Spondyloepiphyseal dysplasia, Stanescu type	Disorder		7 *Cases*
163654	Spondyloepiphyseal dysplasia-brachydactyly-speech disorder syndrome	Disorder		4 *Cases*
163649	Spondyloepiphyseal dysplasia-craniosynostosis-cleft palate-cataracts-intellectual disability syndrome	Disorder		4 *Cases*
93315	Spondylometaphyseal dysplasia, 'corner fracture' type	Disorder		30 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
168555	Spondylometaphyseal dysplasia, A4 type	Disorder		3 *Cases*
168544	Spondylometaphyseal dysplasia, Golden type	Disorder		3 *Cases*
93316	Spondylometaphyseal dysplasia, Schmidt type	Disorder		7 *Cases*
93317	Spondylometaphyseal dysplasia, Sedaghatian type	Disorder		9 *Cases*
168552	Spondylometaphyseal dysplasia-bowed forearms-facial dysmorphism syndrome	Disorder		2 *Cases*
85167	Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	Disorder		18 *Cases*
589435	Spondylometaphyseal dysplasia-corneal dystrophy syndrome	Disorder		2 *Cases*
1856	Spondyloperipheral dysplasia-short ulna syndrome	Disorder		10 *Families*
29822	Spontaneous periodic hypothermia	Disorder		50 *Cases*
204	Sporadic Creutzfeldt-Jakob disease	Disorder	0.088 *P*	
204	Sporadic Creutzfeldt-Jakob disease	Disorder	0.118 *I*	
247234	Sporadic adult-onset ataxia of unknown etiology	Disorder	7.6 *P**	
586130	Sporadic fatal insomnia	Disorder		27 *Cases*
424996	Squamous cell carcinoma of gallbladder and extrahepatic biliary tract	Disorder	0.032 *I**	
424975	Squamous cell carcinoma of liver and intrahepatic biliary tract	Disorder	0.009 *I**	
424039	Squamous cell carcinoma of pancreas	Disorder	0.023 *I**	
424019	Squamous cell carcinoma of the anal canal	Disorder	0.81 *I**	
213767	Squamous cell carcinoma of the cervix uteri	Disorder	4.28 *I**	
423994	Squamous cell carcinoma of the colon	Disorder	0.026 *I**	
213716	Squamous cell carcinoma of the corpus uteri	Disorder	0.12 *I**	
99977	Squamous cell carcinoma of the esophagus	Disorder	3.357 *I**	
99977	Squamous cell carcinoma of the esophagus	Disorder	5.2 *I*	
99977	Squamous cell carcinoma of the esophagus	Disorder	5.42	
494547	Squamous cell carcinoma of the hypopharynx	Disorder	1.27 *I**	

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494550	Squamous cell carcinoma of the larynx	Disorder	4.61 *I**	
502366	Squamous cell carcinoma of the lip	Disorder	1.02 *I*	
500464	Squamous cell carcinoma of the nasal cavity and paranasal sinuses	Disorder	0.35 *I*	
502363	Squamous cell carcinoma of the oral cavity	Disorder	3.51 *I**	
500478	Squamous cell carcinoma of the oropharynx	Disorder	3.12 *I**	
398058	Squamous cell carcinoma of the penis	Disorder	0.57 *I**	
424002	Squamous cell carcinoma of the rectum	Disorder	0.113 *I**	
423968	Squamous cell carcinoma of the small intestine	Disorder	0.008 *I**	
418959	Squamous cell carcinoma of the stomach	Disorder	0.115 *I**	
83484	St. Louis encephalitis	Disorder	0.38 *I**	
140917	Stapes ankylosis with broad thumbs and toes	Disorder		6 *Families*
827	Stargardt disease	Disorder	13.0 *P**	
438117	Steel syndrome	Disorder		40 *Cases*
273	Steinert myotonic dystrophy	Disorder	5.0 *P**	
273	Steinert myotonic dystrophy	Disorder	12.5 *P*	
210115	Sterile multifocal osteomyelitis with periostitis and pustulosis	Disorder		17 *Cases*
2017	Sternal cleft	Disorder	2.0 *BP**	
3196	Steroid dehydrogenase deficiency-dental anomalies syndrome	Disorder		1 *Family*
36426	Stevens-Johnson syndrome	Subtype of disorder	0.36 *I**	
95455	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	Disorder	0.19 *I*	
828	Stickler syndrome	Disorder	1.0 *BP**	
828	Stickler syndrome	Disorder	12.2 *BP*	
2833	Stiff skin syndrome	Disorder		54 *Cases*
3199	Stimmler syndrome	Disorder		2 *Cases*

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3204	Stormorken-Sjaastad-Langslet syndrome	Disorder		17 *Cases*
506307	Stromme syndrome	Disorder		11 *Cases*
3205	Sturge-Weber syndrome	Disorder	3.5 *BP**	
3206	Stüve-Wiedemann syndrome	Disorder		56 *Cases*
3191	Subaortic stenosis-short stature syndrome	Disorder		1 *Family*
48377	Subcorneal pustular dermatosis	Disorder		200 *Cases*
98959	Subepithelial mucinous corneal dystrophy	Disorder		1 *Family*
22	Succinic semialdehyde dehydrogenase deficiency	Disorder		450 *Cases*
832	Succinyl-CoA:3-oxoacid CoA transferase deficiency	Disorder		32 *Cases*
168593	Sudden infant death-dysgenesis of the testes syndrome	Disorder		21 *Cases*
90059	Sudden sensorineural hearing loss	Disorder	40.0 *P**	
498602	Sugarman brachydactyly	Disorder		1 *Family*
3210	Summitt syndrome	Disorder		3 *Cases*
455	Superficial epidermolytic ichthyosis	Disorder		20 *Cases*
247245	Superficial siderosis	Disorder		300 *Cases*
141096	Supernumerary nostril	Disorder		32 *Cases*
466695	Supratip dysplasia	Disorder		5 *Cases*
3193	Supravalvular aortic stenosis	Disorder	4.0 *BP**	
3193	Supravalvular aortic stenosis	Disorder	13.3 *P**	
838	Susac syndrome	Disorder		304 *Cases*
331226	Susceptibility to infection due to TYK2 deficiency	Disorder		8 *Cases*
1570	Symbrachydactyly of hands and feet	Disorder		2 *Cases*
1314	Symmetrical thalamic calcifications	Disorder		30 *Cases*
79098	Sympathetic ophthalmia	Disorder	0.6 *P**	

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3246	Symphalangism with multiple anomalies of hands and feet	Disorder		6 *Cases*
93402	Syndactyly type 1	Disorder	25.0 *BP**	
93405	Syndactyly type 4	Disorder		4 *Cases*
93406	Syndactyly type 5	Disorder		10 *Cases*
357332	Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome	Disorder		26 *Cases*
294026	Syndactyly-nystagmus syndrome due to 2q31.1 microduplication	Disorder		2 *Cases*
3259	Syndactyly-polydactyly-ear lobe syndrome	Disorder		10 *Cases*
140952	Syndactyly-telecanthus-anogenital and renal malformations syndrome	Disorder		7 *Cases*
85274	Syndromic X-linked intellectual disability 7	Disorder		10 *Cases*
84064	Syndromic diarrhea	Disorder		116 *Cases*
178364	Syndromic microphthalmia type 5	Disorder		20 *Cases*
228426	Syndromic multisystem autoimmune disease due to Itch deficiency	Disorder		10 *Cases*
98606	Syndromic orbital border hypoplasia	Disorder		2 *Families*
281090	Syndromic recessive X-linked ichthyosis	Disorder	1.3 *P**	
457223	Syndromic sensorineural deafness due to combined oxidative phosphorylation defect	Disorder		2 *Cases*
840	Syringocystadenoma papilliferum	Disorder		730 *Cases*
188	Systemic capillary leak syndrome	Disorder		150 *Cases*
536	Systemic lupus erythematosus	Disorder	5.14 *I*	
536	Systemic lupus erythematosus	Disorder	43.7 *P*	
158	Systemic primary carnitine deficiency	Disorder	3.2 *BP**	
90291	Systemic sclerosis	Disorder	15.4 *P**	
85414	Systemic-onset juvenile idiopathic arthritis	Disorder	3.1 *P*	
85414	Systemic-onset juvenile idiopathic arthritis	Disorder	0.6 *I*	

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169157	T-B+ severe combined immunodeficiency due to CD45 deficiency	Disorder		3 *Cases*
169154	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	Disorder	0.15 *BP*	
324294	T-cell immunodeficiency with epidermodysplasia verruciformis	Disorder		2 *Cases*
86872	T-cell large granular lymphocyte leukemia	Disorder	0.4 *I**	
457077	TAFRO syndrome	Disorder		28 *Cases*
2886	TARP syndrome	Disorder		6 *Families*
488632	TBCK-related intellectual disability syndrome	Disorder		25 *Cases*
397959	TCR-alpha-beta-positive T-cell deficiency	Disorder		2 *Cases*
488642	TELO2-related intellectual disability-neurodevelopmental disorder	Disorder		6 *Cases*
284227	TEMPI syndrome	Disorder		10 *Cases*
225123	TFR2-related hemochromatosis	Disorder		33 *Cases*
363444	THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	Disorder		4 *Cases*
314667	TMEM165-CDG	Disorder		6 *Cases*
466703	TMEM199-CDG	Disorder		7 *Cases*
562569	TMEM94-associated congenital heart defect-facial dysmorphism-developmental delay syndrome	Disorder		10 *Cases*
55595	TNP03-related limb-girdle muscular dystrophy D2	Disorder		64 *Cases*
424261	TOR1AIP1-related limb-girdle muscular dystrophy	Disorder		3 *Cases*
592570	TRAF7-associated heart defect-digital anomalies-facial dysmorphism-motor and speech delay syndrome	Disorder		55 *Cases*
369840	TRAPPC11-related limb-girdle muscular dystrophy R18	Disorder		3 *Cases*
597201	TRIM22-related inflammatory bowel disease	Disorder		8 *Cases*
3287	Takayasu arteritis	Disorder	0.084 *I**	
3287	Takayasu arteritis	Disorder	1.34 *P**	
500095	Tall stature-intellectual disability-renal	Disorder		4 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	anomalies syndrome			
329191	Tall stature-long halluces-multiple extra-epiphyses syndrome	Disorder		2 *Families*
50809	Talo-patello-scaphoid osteolysis	Disorder		2 *Cases*
31150	Tangier disease	Disorder		185 *Cases*
1412	Tarsal-carpal coalition syndrome	Disorder		10 *Families*
404443	Tatton-Brown-Rahman syndrome	Disorder		17 *Cases*
845	Tay-Sachs disease	Disorder	0.31 *BP**	
845	Tay-Sachs disease	Disorder	0.28 *BP*	
3291	Teebi-Shaltout syndrome	Disorder		5 *Cases*
3293	Telecanthus-hypertelorism-strabismus-pes cavus syndrome	Disorder		2 *Cases*
34514	Telethonin-related limb-girdle muscular dystrophy R7	Disorder		16 *Cases*
352737	Temperature-sensitive oculocutaneous albinism type 1	Subtype of disorder		10 *Cases*
254516	Temple syndrome	Disorder		53 *Cases*
96184	Temple syndrome due to maternal uniparental disomy of chromosome 14	Subtype of disorder		64 *Cases*
254531	Temple syndrome due to paternal 14q32.2 hypomethylation	Subtype of disorder		12 *Cases*
254525	Temple syndrome due to paternal 14q32.2 microdeletion	Subtype of disorder		9 *Cases*
420561	Temple-Baraitser syndrome	Disorder		9 *Cases*
363417	Temtamy preaxial brachydactyly syndrome	Disorder		18 *Cases*
1777	Temtamy syndrome	Disorder		56 *Cases*
66627	Tenosynovial giant cell tumor	Disorder	20.0 *P**	
141258	Tessier number 4 facial cleft	Disorder		2 *Cases*
842	Testicular seminomatous germ cell tumor	Disorder	1.71 *I**	
842	Testicular seminomatous germ cell tumor	Disorder	46.01	
363483	Testicular teratoma	Disorder	0.04	
3299	Tetanus	Disorder	0.024 *I**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
3301	Tetraamelia-multiple malformations syndrome	Disorder		5 *Families*
3303	Tetralogy of Fallot	Disorder	34.0 *BP*	
3303	Tetralogy of Fallot	Disorder	29.3 *BP**	
884	Tetrasomy 12p	Disorder	4.0 *BP**	
96055	Tetrasomy 21	Disorder		13 *Cases*
3310	Tetrasomy 9p	Disorder		70 *Cases*
9	Tetrasomy X	Disorder		50 *Cases*
1780	Thakker-Donnai syndrome	Disorder		2 *Cases*
3312	Thalidomide embryopathy	Disorder	0.77 *P*	
2655	Thanatophoric dysplasia	Disorder	3.5 *BP**	
199348	Thiamine-responsive encephalopathy	Disorder		2 *Cases*
49827	Thiamine-responsive megaloblastic anemia syndrome	Disorder		80 *Cases*
2405	Thickened earlobes-conductive deafness syndrome	Disorder		2 *Families*
98960	Thiel-Behnke corneal dystrophy	Disorder		173 *Cases*
3314	Thiemann disease, familial form	Disorder		33 *Cases*
1506	Thin ribs-tubular bones-dysmorphism syndrome	Disorder		2 *Cases*
3316	Thomas syndrome	Disorder		6 *Cases*
614	Thomsen and Becker disease	Disorder	1.0 *P*	
1861	Thoracic dysplasia-hydrocephalus syndrome	Disorder		2 *Cases*
3317	Thoracolaryngopelvic dysplasia	Disorder		10 *Cases*
1803	Thoracomelic dysplasia	Disorder		2 *Cases*
329319	Thrombocythemia with distal limb defects	Disorder		3 *Families*
67044	Thrombocytopenia with congenital dyserythropoietic anemia	Disorder		3 *Families*
3320	Thrombocytopenia-absent radius	Disorder	0.5 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	syndrome			
436169	Thrombomodulin-related bleeding disorder	Disorder		15 *Cases*
54057	Thrombotic thrombocytopenic purpura	Disorder	0.35 *I*	
2251	Thumb deformity-alopecia-pigmentation anomaly syndrome	Disorder		2 *Families*
1078	Thumb stiffness-brachydactyly-intellectual disability syndrome	Disorder		7 *Cases*
3326	Thymic-renal-anal-lung dysplasia	Disorder		3 *Cases*
99867	Thymoma	Disorder	0.14 *I**	
99867	Thymoma	Disorder	1.22	
3327	Thyrocerebrorenal syndrome	Disorder		2 *Cases*
95712	Thyroid ectopia	Disorder	14.3 *P**	
95719	Thyroid hemiagenesis	Disorder	25.0 *P*	
95720	Thyroid hypoplasia	Disorder	3.5 *P*	
3329	Tibial aplasia-ectrodactyly syndrome	Disorder	0.1 *P**	
93322	Tibial hemimelia	Disorder	0.1 *BP**	
93322	Tibial hemimelia	Disorder	0.1 *P**	
609	Tibial muscular dystrophy	Disorder	6.0 *P**	
42665	Tietz syndrome	Disorder		2 *Families*
65283	Timothy syndrome	Disorder		56 *Cases*
140922	Titin-related limb-girdle muscular dystrophy R10	Disorder		1 *Family*
3338	Toriello-Carey syndrome	Disorder		59 *Cases*
3339	Toriello-Lacassie-Droste syndrome	Disorder		19 *Cases*
3341	Torticollis-keloids-cryptorchidism-renal dysplasia syndrome	Disorder		7 *Cases*
227972	Toxic oil syndrome	Disorder		20000 *Cases*
3346	Tracheal agenesis	Disorder	2.0 *BP**	
3348	Tracheobronchopathia	Disorder		400 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	osteochondroplastica			
101028	Transaldolase deficiency	Disorder		23 *Cases*
859	Transcobalamin deficiency	Disorder		40 *Cases*
300293	Transient infantile hypertriglyceridemia and hepatosteatosis	Disorder		11 *Cases*
99886	Transient neonatal diabetes mellitus	Disorder	0.3 *BP**	
329942	Transient neonatal multiple acyl-CoA dehydrogenase deficiency	Disorder		1 *Case*
93164	Transient pseudohypoaldosteronism	Disorder		152 *Cases*
488618	Transketolase deficiency	Disorder		5 *Cases*
861	Treacher-Collins syndrome	Disorder	2.0 *BP**	
447896	Tremor-ataxia-central hypomyelination syndrome	Subtype of disorder		7 *Cases*
3350	Tremor-nystagmus-duodenal ulcer syndrome	Disorder		17 *Cases*
863	Trichinellosis	Disorder	0.06 *I**	
3352	Tricho-dento-osseous syndrome	Disorder		30 *Cases*
1264	Tricho-retino-dento-digital syndrome	Disorder		9 *Cases*
3351	Trichodental syndrome	Disorder		5 *Families*
3353	Trichodermodysplasia-dental alterations syndrome	Disorder		3 *Cases*
79129	Trichodysplasia-amelogenesis imperfecta syndrome	Disorder		1 *Family*
3361	Trichodysplasia-xeroderma syndrome	Disorder		1 *Family*
3363	Trichomegaly-retina pigmentary degeneration-dwarfism syndrome	Disorder		14 *Cases*
3355	Trichoodontoonychial dysplasia	Disorder		4 *Cases*
77258	Trichorhinophalangeal syndrome type 1	Disorder		250 *Cases*
502	Trichorhinophalangeal syndrome type 2	Disorder		100 *Cases*
33364	Trichothiodystrophy	Disorder		201 *Cases*
33364	Trichothiodystrophy	Disorder	0.12 *BP**	
1209	Tricuspid atresia	Disorder	5.5625 *BP**	

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
3368	Trigonocephaly-bifid nose-acral anomalies syndrome	Disorder		2 *Cases*
3365	Trigonocephaly-broad thumbs syndrome	Disorder		2 *Cases*
3369	Trigonocephaly-short stature-developmental delay syndrome	Disorder		3 *Cases*
868	Triose phosphate-isomerase deficiency	Disorder		50 *Cases*
2947	Triphalangeal thumbs-brachyectrodactyly syndrome	Disorder		4 *Families*
869	Triple A syndrome	Disorder		100 *Cases*
3376	Triploidy	Disorder	12.6 *BP**	
171929	Trisomy 10p	Disorder		50 *Cases*
1699	Trisomy 12p	Disorder		40 *Cases*
1699	Trisomy 12p	Disorder	2.0 *BP*	
3378	Trisomy 13	Disorder	3.7 *BP**	
3380	Trisomy 18	Disorder	16.7 *BP*	
3380	Trisomy 18	Disorder	10.4 *BP**	
1715	Trisomy 18p	Disorder		25 *Cases*
261344	Trisomy 1q	Disorder		18 *Cases*
1738	Trisomy 4p	Disorder		85 *Cases*
1742	Trisomy 5p	Disorder		40 *Cases*
1752	Trisomy 8q	Disorder		30 *Cases*
236	Trisomy 9p	Disorder		150 *Cases*
3375	Trisomy X	Disorder	42.5 *P**	
88629	Tritanopia	Disorder	4.8 *P**	
3384	Truncus arteriosus	Disorder	4.3 *BP*	
3384	Truncus arteriosus	Disorder	4.8 *BP**	
467166	Tubulinopathy-associated dysgyria	Disorder		7 *Cases*

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1063	Tufted angioma	Disorder		200 *Cases*
3392	Tularemia	Disorder	0.2 *I**	
3392	Tularemia	Disorder	2.0 *P**	
32960	Tumor necrosis factor receptor 1 associated periodic syndrome	Disorder	0.1 *P**	
881	Turner syndrome	Disorder	5.5 *BP**	
99745	Typhoid	Disorder	3.0 *I**	
882	Tyrosinemia type 1	Disorder	0.9 *BP*	
28378	Tyrosinemia type 2	Disorder		150 *Cases*
69723	Tyrosinemia type 3	Disorder		20 *Cases*
481665	USP18 deficiency	Disorder		5 *Cases*
178338	UV-sensitive syndrome	Disorder		7 *Cases*
3403	Uhl anomaly	Disorder		84 *Cases*
3403	Uhl anomaly	Disorder	1.0 *BP*	
3404	Ulbright-Hodes syndrome	Disorder		3 *Cases*
2249	Ulna hypoplasia-intellectual disability syndrome	Disorder		2 *Cases*
1837	Ulna metaphyseal dysplasia syndrome	Disorder		3 *Cases*
1122	Ulnar hypoplasia-split foot syndrome	Disorder		1 *Family*
3138	Ulnar-mammary syndrome	Disorder		128 *Cases*
52056	Ulnar/fibula ray defect-brachydactyly syndrome	Disorder		1 *Family*
3405	Umbilical cord ulceration-intestinal atresia syndrome	Disorder		66 *Cases*
418951	Undifferentiated carcinoma of esophagus	Disorder	0.044 *I**	
424970	Undifferentiated carcinoma of liver and intrahepatic biliary tract	Disorder	0.015 *I**	
423786	Undifferentiated carcinoma of stomach	Disorder	0.211 *I**	
424080	Undifferentiated carcinoma with	Disorder	0.001 *I**	

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	osteoclast-like giant cells of pancreas			
2023	Undifferentiated pleomorphic sarcoma	Disorder	0.9 *I**	
97363	Unilateral multicystic dysplastic kidney	Subtype of disorder	23.2 *BP*	
97363	Unilateral multicystic dysplastic kidney	Subtype of disorder	14.8 *BP**	
1464	Univentricular heart	Disorder	7.5 *BP*	
3408	Upington disease	Disorder		1 *Family*
2489	Upper limb defect-eye and ear abnormalities syndrome	Disorder		2 *Cases*
2497	Upper limb mesomelic dysplasia	Disorder		4 *Cases*
598216	Upper tract urothelial carcinoma	Disorder	1.5 *I*	
3409	Urban-Rogers-Meyer syndrome	Disorder		3 *Cases*
94059	Uremic pruritus	Disorder	35.0 *P**	
210128	Urocanic aciduria	Disorder		4 *Cases*
1473	Uveal coloboma-cleft lip and palate-intellectual disability	Disorder		12 *Cases*
39044	Uveal melanoma	Disorder	0.5 *I**	
39044	Uveal melanoma	Disorder	6.0	
3412	VACTERL with hydrocephalus	Disorder		10 *Families*
887	VACTERL/VATER association	Disorder	6.25 *BP**	
596753	VEXAS syndrome	Disorder		37 *Cases*
466934	VPS11-related autosomal recessive hypomyelinating leukodystrophy	Disorder		13 *Cases*
88635	Vacuolar myopathy with sarcoplasmic reticulum protein aggregates	Disorder		4 *Cases*
3417	Van den Bosch syndrome	Disorder		1 *Family*
2460	Van den Ende-Gupta syndrome	Disorder		29 *Cases*
314652	Variant ABeta2M amyloidosis	Disorder		5 *Cases*
286	Vascular Ehlers-Danlos syndrome	Disorder	1.0 *P*	
404553	Vasculitis due to ADA2 deficiency	Disorder		48 *Cases*

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3424	Velo-facial-skeletal syndrome	Disorder		2 *Cases*
443988	Ventriculomegaly-cystic kidney disease	Disorder		11 *Cases*
3429	Verloove Vanhorick-Brubakk syndrome	Disorder		2 *Cases*
70476	Vernal keratoconjunctivitis	Disorder	32.0 *P**	
493342	Vibratory urticaria	Disorder		37 *Cases*
1493	Vici syndrome	Disorder		50 *Cases*
228379	Virus-associated trichodysplasia spinulosa	Disorder		7 *Cases*
73246	Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome	Disorder		2 *Cases*
28	Vitamin B12-responsive methylmalonic acidemia	Disorder		192 *Cases*
79310	Vitamin B12-responsive methylmalonic acidemia type cblA	Subtype of disorder		60 *Cases*
79312	Vitamin B12-unresponsive methylmalonic acidemia type mut-	Subtype of disorder		450 *Cases*
3439	Von Voss-Cherstvoy syndrome	Disorder		10 *Cases*
903	Von Willebrand disease	Disorder	10.0 *P*	
166096	Von Willebrand disease type 3	Subtype of disorder	0.1865 *P*	
137583	Vulvar intraepithelial neoplasia	Disorder	20.0 *P**	
83453	Vulvovaginal gingival syndrome	Disorder		380 *Cases*
2804	W syndrome	Disorder		6 *Cases*
466943	WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	Disorder		22 *Cases*
893	WAGR syndrome	Disorder	0.2 *BP*	
572798	WARS2-related combined oxidative phosphorylation defect	Disorder		11 *Cases*
51636	WHIM syndrome	Disorder		65 *Cases*
3466	WT limb-blood syndrome	Disorder		3 *Families*
3440	Waardenburg syndrome	Disorder	0.37 *BP**	

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895	Waardenburg syndrome type 2	Subtype of disorder		3 *Families*
897	Waardenburg-Shah syndrome	Disorder		100 *Cases*
898	Wagner disease	Disorder		100 *Cases*
33226	Waldenström macroglobulinemia	Disorder	0.81 *I**	
899	Walker-Warburg syndrome	Disorder	1.65 *BP**	
280558	Warsaw breakage syndrome	Disorder		4 *Cases*
568056	Warts-immunodeficiency-lymphedema-anogenital dysplasia syndrome	Disorder		2 *Cases*
3447	Weaver syndrome	Disorder		48 *Cases*
3448	Weaver-Williams syndrome	Disorder		2 *Cases*
3449	Weill-Marchesani syndrome	Disorder	1.0 *P*	
3344	Weismann-Netter syndrome	Disorder		100 *Cases*
502430	Weiss-Kruszka Syndrome	Disorder		8 *Cases*
99971	Well-differentiated liposarcoma	Subtype of disorder	0.51 *I**	
901	Wells syndrome	Disorder		200 *Cases*
902	Werner syndrome	Disorder	0.5 *P**	
83476	West-Nile encephalitis	Disorder	0.036 *I**	
2475	White forelock with malformations	Disorder		2 *Cases*
3207	White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome	Disorder		4 *Cases*
370131	White platelet syndrome	Disorder		1 *Family*
1489	Whooping cough	Disorder	8.9 *I**	
3455	Wiedemann-Rautenstrauch syndrome	Disorder		37 *Cases*
319182	Wiedemann-Steiner syndrome	Disorder		84 *Cases*
85446	Wild type ABeta2M amyloidosis	Disorder	4.5 *P**	

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330001	Wild type ATTR amyloidosis	Disorder	1.72 *P*	
905	Wilson disease	Disorder	2.25 *BP*	
905	Wilson disease	Disorder	2.02 *P*	
905	Wilson disease	Disorder	6.0 *P**	
3459	Wilson-Turner syndrome	Disorder		28 *Cases*
906	Wiskott-Aldrich syndrome	Disorder	0.1 *P**	
500163	Witteveen-Kolk syndrome	Disorder		40 *Cases*
1667	Wolcott-Rallison syndrome	Disorder		60 *Cases*
280	Wolf-Hirschhorn syndrome	Disorder	2.0 *BP**	
3463	Wolfram syndrome	Disorder	0.13 *P*	
3463	Wolfram syndrome	Disorder	0.62 *P**	
3464	Woodhouse-Sakati syndrome	Disorder		25 *Families*
420686	Woolly hair-palmoplantar keratoderma syndrome	Disorder		8 *Cases*
166277	Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia	Disorder		3 *Cases*
3465	Worster-Drought syndrome	Disorder	3.7 *P**	
178475	Wound botulism	Subtype of disorder	0.1 *I**	
2834	Wrinkly skin syndrome	Subtype of disorder		30 *Cases*
53719	Wyburn-Mason syndrome	Disorder		90 *Cases*
101076	X-linked Charcot-Marie-Tooth disease type 2	Disorder		5 *Cases*
101077	X-linked Charcot-Marie-Tooth disease type 3	Disorder		4 *Families*
101078	X-linked Charcot-Marie-Tooth disease type 4	Disorder		7 *Cases*
99014	X-linked Charcot-Marie-Tooth disease type 5	Disorder		9 *Cases*
352675	X-linked Charcot-Marie-Tooth disease type 6	Disorder		8 *Cases*
75497	X-linked Ehlers-Danlos syndrome	Disorder		2 *Families*

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98863	X-linked Emery-Dreifuss muscular dystrophy	Subtype of disorder	1.0 *BP*	
98863	X-linked Emery-Dreifuss muscular dystrophy	Subtype of disorder	1.0 *P*	
300373	X-linked acrogigantism	Disorder		33 *Cases*
95702	X-linked adrenal hypoplasia congenita	Disorder	8.0 *BP*	
47	X-linked agammaglobulinemia	Subtype of disorder	0.1 *P**	
47	X-linked agammaglobulinemia	Subtype of disorder	0.22 *P*	
391327	X-linked calvarial hyperostosis	Disorder		1 *Family*
329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement	Disorder		27 *Cases*
596	X-linked centronuclear myopathy	Disorder	0.2 *P**	
163961	X-linked cerebral-cerebellar-coloboma syndrome	Disorder		3 *Cases*
431140	X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome	Disorder		1 *Family*
1497	X-linked complicated corpus callosum dysgenesis	Subtype of disorder		11 *Cases*
90001	X-linked cone dysfunction syndrome with myopia	Disorder		10 *Families*
1661	X-linked corneal dermoid	Disorder		6 *Cases*
52503	X-linked creatine transporter deficiency	Disorder		150 *Cases*
139557	X-linked distal spinal muscular atrophy type 3	Disorder		2 *Families*
35173	X-linked dominant chondrodysplasia punctata	Disorder	0.25 *BP**	
163966	X-linked dominant chondrodysplasia, Chassaing-Lacombe type	Disorder		10 *Cases*
363727	X-linked dyserythropoietic anemia with abnormal platelets and neutropenia	Disorder		1 *Family*
293621	X-linked endothelial corneal dystrophy	Disorder		35 *Cases*
443197	X-linked erythropoietic protoporphyria	Disorder		50 *Cases*
500188	X-linked external auditory canal atresia-dilated internal auditory canal-facial dysmorphism syndrome	Disorder		4 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
480880	X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	Disorder		17 *Cases*
139583	X-linked hereditary sensory and autonomic neuropathy with deafness	Disorder		5 *Families*
181	X-linked hypohidrotic ectodermal dysplasia	Subtype of disorder	0.75 *BP**	
89936	X-linked hypophosphatemia	Disorder	1.66 *P**	
89936	X-linked hypophosphatemia	Disorder	2.14 *P*	
89936	X-linked hypophosphatemia	Disorder	4.5 *I*	
317476	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia	Disorder		7 *Cases*
2571	X-linked immunoneurologic disorder	Disorder		5 *Cases*
364028	X-linked intellectual disability due to GRIA3 mutations	Disorder		14 *Cases*
67045	X-linked intellectual disability with isolated growth hormone deficiency	Subtype of disorder		2 *Families*
85273	X-linked intellectual disability, Abidi type	Disorder		8 *Cases*
85276	X-linked intellectual disability, Armfield type	Disorder		6 *Cases*
85293	X-linked intellectual disability, Cabezas type	Disorder		24 *Families*
85277	X-linked intellectual disability, Cantagrel type	Disorder		30 *Cases*
163971	X-linked intellectual disability, Cilliers type	Disorder		4 *Cases*
93947	X-linked intellectual disability, Golabi-Ito-Hall type	Subtype of disorder		3 *Cases*
93952	X-linked intellectual disability, Hedera type	Disorder		9 *Cases*
85283	X-linked intellectual disability, Miles-Carpenter type	Disorder		4 *Cases*
163937	X-linked intellectual disability, Najm type	Disorder		35 *Families*
163956	X-linked intellectual disability, Nascimento type	Disorder		8 *Cases*
85322	X-linked intellectual disability, Pai type	Disorder		1 *Family*
85285	X-linked intellectual disability, Schimke type	Disorder		4 *Cases*
85323	X-linked intellectual disability, Seemanova type	Disorder		4 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
85286	X-linked intellectual disability, Shashi type	Disorder		9 *Cases*
85324	X-linked intellectual disability, Shrimpton type	Disorder		3 *Cases*
85287	X-linked intellectual disability, Siderius type	Disorder		2 *Families*
3063	X-linked intellectual disability, Snyder type	Disorder		21 *Cases*
85325	X-linked intellectual disability, Stevenson type	Disorder		4 *Cases*
85288	X-linked intellectual disability, Stocco Dos Santos type	Disorder		1 *Family*
85326	X-linked intellectual disability, Stoll type	Disorder		4 *Cases*
163976	X-linked intellectual disability, Van Esch type	Disorder		7 *Cases*
85290	X-linked intellectual disability, Wilson type	Disorder		3 *Cases*
1568	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	Disorder		10 *Cases*
85327	X-linked intellectual disability-acromegaly-hyperactivity syndrome	Disorder		2 *Cases*
85338	X-linked intellectual disability-ataxia-apraxia syndrome	Disorder		9 *Cases*
324410	X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome	Disorder		2 *Cases*
137831	X-linked intellectual disability-cerebellar hypoplasia syndrome	Disorder		14 *Families*
459070	X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome	Disorder		2 *Cases*
163979	X-linked intellectual disability-craniofacioskeletal syndrome	Disorder		9 *Cases*
85280	X-linked intellectual disability-cubitus valgus-dysmorphism syndrome	Disorder		5 *Cases*
2958	X-linked intellectual disability-dysmorphism-cerebral atrophy syndrome	Disorder		8 *Cases*
85319	X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome	Disorder		2 *Cases*
480907	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	Disorder		14 *Cases*
85317	X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome	Disorder		3 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
3055	X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome	Disorder		4 *Cases*
85329	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	Disorder		10 *Cases*
457260	X-linked intellectual disability-hypotonia-movement disorder syndrome	Disorder		38 *Cases*
423479	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	Disorder		2 *Cases*
85320	X-linked intellectual disability-macrocephaly-macroorchidism syndrome	Disorder		12 *Cases*
2898	X-linked intellectual disability-plagiocephaly syndrome	Disorder		2 *Cases*
3077	X-linked intellectual disability-psychosis-macroorchidism syndrome	Disorder		6 *Cases*
3052	X-linked intellectual disability-seizures-psoriasis syndrome	Disorder		4 *Cases*
457240	X-linked intellectual disability-short stature-overweight syndrome	Disorder		20 *Cases*
482606	X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome	Disorder		15 *Cases*
79447	X-linked lethal multiple pterygium syndrome	Disorder		6 *Families*
452	X-linked lissencephaly with abnormal genitalia	Disorder		30 *Families*
538931	X-linked lymphoproliferative disease due to SH2D1A deficiency	Disorder		100 *Cases*
538934	X-linked lymphoproliferative disease due to XIAP deficiency	Disorder		100 *Cases*
1131	X-linked mandibulofacial dysostosis	Disorder		7 *Cases*
319605	X-linked mendelian susceptibility to mycobacterial diseases	Disorder		13 *Cases*
319623	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency	Subtype of disorder		7 *Cases*
319612	X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency	Subtype of disorder		6 *Cases*
435938	X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	Disorder		3 *Cases*
25980	X-linked myopathy with excessive autophagy	Disorder		18 *Families*
178461	X-linked myopathy with postural muscle	Disorder		7 *Families*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
	atrophy			
456328	X-linked myotubular myopathy-abnormal genitalia syndrome	Disorder		4 *Cases*
85334	X-linked neurodegenerative syndrome, Bertini type	Disorder		7 *Cases*
85336	X-linked neurodegenerative syndrome, Hamel type	Disorder		11 *Cases*
314978	X-linked non progressive cerebellar ataxia	Disorder		3 *Families*
391330	X-linked osteoporosis with fractures	Disorder		8 *Families*
363654	X-linked parkinsonism-spasticity syndrome	Disorder		5 *Cases*
54	X-linked recessive ocular albinism	Disorder	0.58 *BP**	
85453	X-linked reticulate pigmentary disorder	Disorder		6 *Families*
1852	X-linked retinal dysplasia	Disorder		8 *Cases*
792	X-linked retinoschisis	Disorder	5.0 *P*	
792	X-linked retinoschisis	Disorder	4.5 *P**	
431272	X-linked scapuloperoneal muscular dystrophy	Disorder		22 *Cases*
86788	X-linked severe congenital neutropenia	Disorder		45 *Cases*
75563	X-linked sideroblastic anemia	Disorder		200 *Cases*
2802	X-linked sideroblastic anemia and spinocerebellar ataxia	Disorder		13 *Cases*
1436	X-linked skeletal dysplasia-intellectual disability syndrome	Disorder		4 *Cases*
100997	X-linked spastic paraplegia type 16	Disorder		1 *Family*
171607	X-linked spastic paraplegia type 34	Disorder		24 *Cases*
3175	X-linked spasticity-intellectual disability-epilepsy syndrome	Disorder		6 *Cases*
85297	X-linked spinocerebellar ataxia type 3	Disorder		5 *Cases*
85292	X-linked spinocerebellar ataxia type 4	Disorder		1 *Family*
3469	XK aprosencephaly syndrome	Disorder		10 *Cases*
1770	XY type gonadal dysgenesis-associated anomalies syndrome	Disorder		2 *Cases*
370930	XYLT1-CDG	Disorder		2 *Cases*

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ORPHACode	Disease or Subtype of disease	Classification Level	Estimated prevalence/incidence (/100,000)	Number of published cases or families
910	Xeroderma pigmentosum	Disorder	0.23 *BP**	
90342	Xeroderma pigmentosum variant	Disorder		50 *Cases*
220295	Xeroderma pigmentosum-Cockayne syndrome complex	Disorder		30 *Cases*
261476	Xp21 deletion syndrome	Disorder		100 *Cases*
314389	Xq12-q13.3 duplication syndrome	Disorder		3 *Cases*
1435	Xq21 microdeletion syndrome	Disorder		13 *Cases*
521258	Xq25 microduplication syndrome	Disorder		28 *Cases*
261483	Xq27.3q28 duplication syndrome	Disorder		8 *Cases*
662	Yellow nail syndrome	Disorder		400 *Cases*
314485	Young adult-onset distal hereditary motor neuropathy	Disorder		3 *Cases*
2828	Young-onset Parkinson disease	Disorder	15.0 *P**	
3472	Yunis-Varon syndrome	Disorder		25 *Cases*
97240	Zebra body myopathy	Disorder		10 *Cases*
217017	Zechi-Ceide syndrome	Disorder		3 *Cases*
50812	Zellweger-like syndrome without peroxisomal anomalies	Disorder		2 *Cases*
3473	Zimmermann-Laband syndrome	Disorder		52 *Cases*
913	Zollinger-Ellison syndrome	Disorder	0.15 *I**	
913	Zollinger-Ellison syndrome	Disorder	0.125 *I*	
178333	Åland Islands eye disease	Disorder		5 *Families*

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