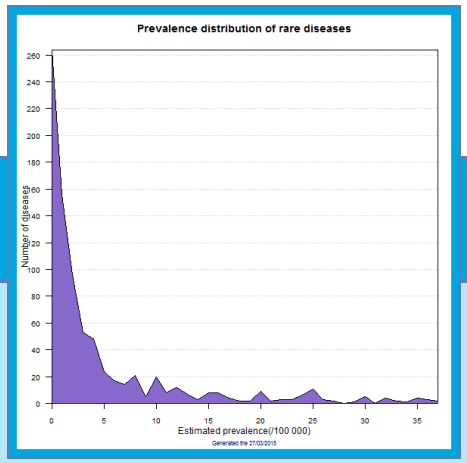

Orphanet Report Series
Rare Diseases collection

Number 1 | June 2018

Prevalence and incidence of rare diseases: Bibliographic data

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

www.orpha.net www.orphadata.org

Inserm
La science pour la santé
From science to health

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Methodology

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

This data is presented in the following reports published biannually:

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

Data collection

A number of different sources are used :

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

Data characteristics

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

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

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

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

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

Limitations of the study

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

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

Data presentation

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

Please note that this is just a selection of Orphanet's rare disease epidemiological data. Currently 5880 rare diseases are annotated with prevalence or incidence

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

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

ORPHA Number	Disease or Group of diseases	Estimated prevalence/incidence (/100,000)	Number of published cases or families
79154	2-aminoadipic 2-oxoadipic aciduria		20 Cases
79157	2-methylbutyryl-CoA dehydrogenase deficiency		30 Cases
35701	3-hydroxy-3-methylglutaryl-CoA synthase deficiency		9 Cases
939	3-hydroxyisobutyric aciduria		13 Cases
6	3-methylcrotonyl-CoA carboxylase deficiency	2.65 *BP* *	
67046	3-methylglutaconic aciduria type 1		20 Cases
445038	3-methylglutaconic aciduria type 7		22 Cases
505208	3-methylglutaconic aciduria type 8		9 Cases
505216	3-methylglutaconic aciduria type 9		4 Cases
79351	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form		15 Cases
79350	3-phosphoserine phosphatase deficiency		8 Cases
7	3C syndrome		25 Cases
2616	3M syndrome		200 Cases
293843	3MC syndrome		32 Cases
217064	5-fluorouracil poisoning	2.0 *P* *	
33572	5-oxoprolinase deficiency		8 Cases
2975	46,XX disorder of sex development-skeletal anomalies syndrome		2 Cases
444048	46,XX ovarian dysgenesis-short stature syndrome		3 Cases
2138	46,XX ovotesticular disorder of sex development	2.5 *BP*	
393	46,XX testicular disorder of sex development	2.5 *P*	
753	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency		50 Families
90796	46,XY disorder of sex development due to isolated 17,20-lyase deficiency		15 Cases
443087	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency		2 Families
168558	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency		9 Cases
168563	46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome		5 Cases
8	47,XYY syndrome	50.0 *BP* *	
96263	48,XXXY syndrome	1.0 *BP* *	
10	48,XXYY syndrome	1.9 *BP* *	
99329	48,XYYY syndrome		10 Cases
96264	49,XXXXY syndrome	0.55 *BP* *	
261534	49,XXXYY syndrome		2 Cases
99330	49,XYYYY syndrome		8 Cases
293948	1p21.3 microdeletion syndrome		9 Cases
401986	1p31p32 microdeletion syndrome		5 Cases
456298	1p35.2 microdeletion syndrome		2 Cases
250994	1q21.1 microduplication syndrome		46 Cases
238769	1q44 microdeletion syndrome		100 Cases
363680	2p13.2 microdeletion syndrome		2 Cases
261349	2p15p16.1 microdeletion syndrome		11 Cases
163693	2p21 microdeletion syndrome		7 Cases
369881	2p21 microdeletion syndrome without cystinuria		2 Cases
228402	2q23.1 microdeletion syndrome		18 Cases
313947	2q23.1 microduplication syndrome		2 Cases
1617	2q24 microdeletion syndrome		23 Cases
294026	2q31.1 microduplication syndrome		2 Cases
251019	2q32q33 microdeletion syndrome		25 Cases
251028	2q33.1 microdeletion syndrome		20 Cases
1001	2q37 microdeletion syndrome		115 Cases
435638	3p25.3 microdeletion syndrome		8 Cases
1621	3q13 microdeletion syndrome		42 Cases
96095	3q26 microduplication syndrome		100 Cases
356947	3q26q27 microdeletion syndrome		4 Cases
397695	3q27.3 microdeletion syndrome		7 Cases
238750	4q21 microdeletion syndrome		14 Cases
502437	4q25 proximal deletion syndrome		3 Cases
329802	5p13 microduplication syndrome		7 Cases
228384	5q14.3 microdeletion syndrome		40 Cases
228415	5q35 microduplication syndrome		30 Cases
251046	6p22 microdeletion syndrome		19 Cases
75857	6q terminal deletion syndrome		19 Cases
171829	6q16 deletion syndrome		12 Cases
251056	6q25 microdeletion syndrome		4 Cases

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ORPHA Number	Disease or Group of diseases	Estimated prevalence/incidence (/100,000)	Number of published cases or families
314034	7p22.1 microduplication syndrome		5 Cases
96121	7q11.23 microduplication syndrome		163 Cases
251061	7q31 microdeletion syndrome		20 Cases
96092	8p inverted duplication/deletion syndrome	3.9 *BP* *	
251066	8p11.2 deletion syndrome		3 Cases
251076	8p23.1 duplication syndrome	1.72 *P*	
228399	8q12 microduplication syndrome		4 Cases
284160	8q21.11 microdeletion syndrome		13 Cases
178303	8q22.1 microdeletion syndrome		6 Cases
324313	9p13 microdeletion syndrome		4 Cases
401923	9q31.1q31.3 microdeletion syndrome		2 Cases
495818	9q33.3q34.11 microdeletion syndrome		4 Cases
300305	11p15.4 microduplication syndrome		1 Family
444002	11q22.2q22.3 microdeletion syndrome		5 Cases
313884	12p12.1 microdeletion syndrome		6 Cases
94063	12q14 microdeletion syndrome		19 Cases
289513	12q15q21.1 microdeletion syndrome		6 Cases
412035	13q12.3 microdeletion syndrome		3 Cases
261120	14q11.2 microdeletion syndrome		3 Cases
261229	14q11.2 microduplication syndrome		7 Cases
261144	14q12 microdeletion syndrome		3 Cases
264200	14q22q23 microdeletion syndrome		5 Cases
401935	14q24.1q24.3 microdeletion syndrome		3 Cases
488280	14q32 duplication syndrome		33 Cases
314585	15q overgrowth syndrome		12 Cases
261183	15q11.2 microdeletion syndrome		200 Cases
238446	15q11q13 microduplication syndrome		30 Cases
199318	15q13.3 microdeletion syndrome		246 Cases
261190	15q14 microdeletion syndrome		9 Cases
94065	15q24 microdeletion syndrome		30 Cases
261211	16p11.2p12.2 microdeletion syndrome		8 Cases
261204	16p11.2p12.2 microduplication syndrome		7 Cases
485405	16p12.1p12.3 triplication syndrome		3 Cases
261236	16p13.11 microdeletion syndrome	7.0 *BP*	
261243	16p13.11 microduplication syndrome		162 Cases
500055	16p13.2 microdeletion syndrome		6 Cases
96078	16p13.3 microduplication syndrome		27 Cases
352629	16q24.1 microdeletion syndrome		42 Cases
261250	16q24.3 microdeletion syndrome		27 Cases
217385	17p13.3 microduplication syndrome		50 Cases
97685	17q11 microdeletion syndrome		170 Cases
139474	17q11.2 microduplication syndrome		7 Cases
261265	17q12 microdeletion syndrome		103 Cases
261272	17q12 microduplication syndrome		118 Cases
363958	17q21.31 microdeletion syndrome	6.25 *P* *	
261279	17q23.1q23.2 microdeletion syndrome		7 Cases
254346	19p13.12 microdeletion syndrome		6 Cases
357001	19p13.13 microdeletion syndrome		7 Cases
447980	19p13.3 microduplication syndrome		6 Cases
217346	19q13.11 microdeletion syndrome		12 Cases
261295	20p12.3 microdeletion syndrome		3 Cases
313781	20p13 microdeletion syndrome		4 Cases
444051	20q11.2 microdeletion syndrome		11 Cases
261323	21q22.11q22.12 microdeletion syndrome		12 Cases
567	22q11.2 deletion syndrome	37.5 *BP*	
439232	AApoAIV amyloidosis		2 Cases
915	Aarskog-Scott syndrome	0.5 *BP* *	
916	Aase-Smith syndrome		10 Cases
324723	ABeta amyloidosis, Arctic type		1 Family
100006	ABeta amyloidosis, Dutch type		250 Cases
324708	ABeta amyloidosis, Iowa type		2 Families
324713	ABeta amyloidosis, Italian type		7 Families
324718	ABetaA21G amyloidosis		2 Families
324703	ABetaL34V amyloidosis		1 Family
920	Ablepharon macrostomia syndrome		16 Cases
921	Abruzzo-Erickson syndrome		4 Cases
2310	Absence deformity of leg-cataract syndrome		2 Cases
1658	Absence of fingerprints-congenital milia syndrome		10 Families
980	Absence of the pulmonary artery	0.5 *I* *	

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ORPHA Number	Disease or Group of diseases	Estimated prevalence/incidence (/100,000)	Number of published cases or families
3016	Absent radius-anogenital anomalies syndrome		2 Cases
2951	Absent thumb-short stature-immunodeficiency syndrome		3 Cases
3328	Absent tibia-polydactyly-arachnoid cyst syndrome		3 Cases
67043	Acanthamoeba keratitis	1.0 *P* *	
90301	Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome		5 Cases
926	Acatalasemia	3.2 *P* *	
48818	Aceruloplasminemia	0.09 *P*	
929	Achalasia-microcephaly syndrome		7 Cases
15	Achondroplasia	4.0 *BP*	
49382	Achromatopsia	2.7 *P*	
424046	Acinar cell carcinoma of pancreas	0.02 *I* *	
40366	Acitretin/etretinate embryopathy		26 Cases
90065	Acquired aneurysmal subarachnoid hemorrhage	10.0 *P* *	
91385	Acquired angioedema		200 Cases
46487	Acquired epidermolysis bullosa	0.03 *I* *	
79086	Acquired generalized lipodystrophy	1.0 *P* *	
73274	Acquired hemophilia	0.1 *P* *	
73274	Acquired hemophilia	0.08 *I*	
2221	Acquired hypertrichosis lanuginosa		60 Cases
75564	Acquired idiopathic sideroblastic anemia	0.09 *I* *	
464453	Acquired methemoglobinemia		242 Cases
91136	Acquired monoclonal Ig light chain-associated Fanconi syndrome		100 Cases
79087	Acquired partial lipodystrophy	1.0 *P* *	
228247	Acquired pseudoxanthoma elasticum		20 Cases
99147	Acquired von Willebrand syndrome		300 Cases
158673	Acral dystrophic epidermolysis bullosa		10 Families
263534	Acral peeling skin syndrome		40 Cases
281127	Acral self-healing collodion baby		2 Cases
958	Acro-renal-mandibular syndrome		10 Cases
959	Acro-renal-ocular syndrome		20 Families
36	Acrocallosal syndrome		38 Cases
2008	Acrocardiofacial syndrome		9 Cases
221054	Acrocephalopolydactyly		8 Cases
949	Acrocraniofacial dysostosis		2 Cases
950	Acrodysostosis		80 Cases
280651	Acrodysostosis with multiple hormone resistance		40 Cases
1786	Acrofacial dysostosis, Catania type		2 Families
64542	Acrofacial dysostosis, Kennedy-Teebi type		2 Cases
1787	Acrofacial dysostosis, Palagonia type		4 Cases
1788	Acrofacial dysostosis, Rodríguez type		13 Cases
1784	Acrofrontofacionasal dysostosis		12 Cases
965	Acromegaloid facial appearance syndrome		23 Cases
963	Acromegaly	5.5 *P*	
963	Acromegaly	0.35 *I*	
39	Acromelanosis		10 Cases
1827	Acromelic frontonasal dysplasia		18 Cases
968	Acromesomelic dysplasia, Hunter-Thompson type		10 Cases
40	Acromesomelic dysplasia, Maroteaux type		50 Cases
969	Acromicric dysplasia		60 Cases
955	Acroosteolysis dominant type		100 Cases
363665	Acroosteolysis-keloid-like lesions-premature aging syndrome		5 Cases
85203	Acropectoral syndrome		25 Cases
957	Acropectorovertebral dysplasia		30 Cases
971	Acrorenal syndrome		20 Cases
99892	ACTH-dependent Cushing syndrome	0.55 *I*	
163696	Action myoclonus-renal failure syndrome		38 Cases
397596	Activated PI3K-delta syndrome		18 Cases
284460	Acute annular outer retinopathy		12 Cases
83597	Acute disseminated encephalomyelitis	0.6 *I* *	
363549	Acute encephalopathy with biphasic seizures and late reduced diffusion		283 Cases
293173	Acute generalized exanthematous pustulosis	0.3 *I*	
217371	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins		19 Cases
466794	Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome		3 Cases
370088	Acute infantile liver failure-multisystemic involvement syndrome		6 Cases
98916	Acute inflammatory demyelinating polyradiculoneuropathy	3.1 *P* *	

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ORPHA Number	Disease or Group of diseases	Estimated prevalence/incidence (/100,000)	Number of published cases or families
79276	Acute intermittent porphyria	0.54 *P* *	
79276	Acute intermittent porphyria	0.013 *I* *	
79126	Acute interstitial pneumonia	3.8 *P* *	
90062	Acute liver failure	20.0 *P* *	
178320	Acute lung injury	25.0 *I* *	
513	Acute lymphoblastic leukemia	11.0 *P* *	
513	Acute lymphoblastic leukemia	2.75 *I* *	
488239	Acute macular neuroretinopathy		101 Cases
518	Acute megakaryoblastic leukemia	0.02 *I* *	
514	Acute monoblastic leukemia	0.13 *I* *	
98834	Acute myeloblastic leukemia with maturation	0.02 *I* *	
98833	Acute myeloblastic leukemia without maturation	0.01 *I* *	
519	Acute myeloid leukemia	2.5 *I*	
98832	Acute myeloid leukemia with minimal differentiation	0.01 *I* *	
98277	Acute myeloid leukemia with recurrent genetic anomaly	0.11 *I* *	
517	Acute myelomonocytic leukemia	0.17 *I* *	
86843	Acute panmyelosis with myelofibrosis	0.06 *I* *	
90064	Acute peripheral arterial occlusion	16.0 *P* *	
520	Acute promyelocytic leukemia	0.11 *I* *	
90059	Acute sensorineural hearing loss by acute acoustic trauma or sudden deafness or surgery induced acoustic trauma	37.0 *P* *	
139417	Acute transverse myelitis	1.6 *I*	
284454	Acute zonal occult outer retinopathy		150 Cases
99901	Acyl-CoA dehydrogenase 9 deficiency		23 Cases
100008	ACys amyloidosis		9 Families
55881	Adamantinoma	0.01 *I* *	
974	Adams-Oliver syndrome		398 Cases
85138	Addison disease	12.5 *P* *	
2952	Adducted thumbs-arthrogryposis syndrome, Christian type		9 Cases
213504	Adenocarcinoma of ovary	5.97 *I* *	
424016	Adenocarcinoma of the anal canal	0.26 *I* *	
213772	Adenocarcinoma of the cervix uteri	1.01 *I* *	
99976	Adenocarcinoma of the esophagus	0.7 *I*	
424991	Adenocarcinoma of the gallbladder and extrahepatic biliary tract	2.62 *I* *	
424943	Adenocarcinoma of the liver and intrahepatic biliary tract	0.21 *I* *	
104075	Adenocarcinoma of the small instestine	0.57 *I* *	
45	Adenosine monophosphate deaminase deficiency		100 Cases
91127	Adenovirus infection in immunocompromised patients	18.0 *P* *	
46	Adenylosuccinate lyase deficiency		56 Cases
482601	Adenylosuccinate synthetase-like 1-related distal myopathy		19 Cases
404448	ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder		10 Cases
1501	Adrenocortical carcinoma	0.75 *P* *	
1501	Adrenocortical carcinoma	0.03 *I* *	
977	Adrenomyodystrophy		2 Cases
2666	Adult familial nephronophthisis-spastic quadriparesia syndrome		2 Cases
178487	Adult intestinal botulism		19 Cases
206583	Adult polyglucosan body disease		50 Cases
978	ADULT syndrome		50 Cases
86875	Adult T-cell leukemia/lymphoma	3.0 *P* *	
99027	Adult-onset autosomal dominant leukodystrophy		20 Families
284289	Adult-onset autosomal recessive cerebellar ataxia		14 Cases
420492	Adult-onset cervical dystonia, DYT23 type		2 Families
329478	Adult-onset distal myopathy due to VCP mutation		9 Cases
199351	Adult-onset dystonia-parkinsonism		14 Cases
183669	Agammaglobulinemia	0.13 *P* *	
83617	Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome		3 Cases
85448	AGel amyloidosis		475 Cases
98850	Aggressive systemic mastocytosis	0.33 *P* *	
442582	AH amyloidosis		12 Cases
412069	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome		4 Cases
250977	AICA-ribosiduria		1 Case
51	Aicardi-Goutières syndrome		120 Cases
90081	AIDS wasting syndrome	20.0 *P* *	
79085	AKT2-related familial partial lipodystrophy		1 Family
85443	AL amyloidosis	11.0 *P* *	
404454	Alacrimia-choreoathetosis-liver dysfunction syndrome		8 Cases
52	Alagille syndrome	0.8 *BP* *	
178333	Åland Islands eye disease		5 Families

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2007	Alar cartilages hypoplasia-coloboma-telecanthus syndrome		2 Cases
53	Albers-Schönberg osteopetrosis	1.0 *P*	
998	Albinism-deafness syndrome		1 Family
35664	ALDH18A1-related De Barsy syndrome		32 Cases
369929	Aldosterone-producing adenoma with seizures and neurological abnormalities		2 Cases
79327	ALG1-CDG		57 Cases
79326	ALG2-CDG		1 Case
79321	ALG3-CDG		15 Cases
79320	ALG6-CDG		54 Cases
79325	ALG8-CDG		15 Cases
79328	ALG9-CDG		12 Cases
280071	ALG11-CDG		8 Cases
79324	ALG12-CDG		11 Cases
324422	ALG13-CDG		1 Case
502444	Alkaline ceramidase 3 deficiency		2 Cases
59	Allan-Herndon-Dudley syndrome		320 Cases
1006	Alopecia antibody deficiency		3 Cases
700	Alopecia totalis	10.5 *P* *	
701	Alopecia universalis	25.0 *P* *	
1005	Alopecia-contractures-dwarfism-intellectual disability syndrome		5 Cases
1008	Alopecia-epilepsy-pyorrhea-intellectual disability syndrome		12 Cases
1014	Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome		2 Cases
726	Alpers-Huttenlocher syndrome	0.07 *P* *	
726	Alpers-Huttenlocher syndrome	0.7 *BP* *	
60	Alpha-1-antitrypsin deficiency	20.0 *P* *	
399058	Alpha-B crystallin-related late-onset distal myopathy		17 Cases
100025	Alpha-heavy chain disease		400 Cases
61	Alpha-mannosidosis	0.1 *P* *	
3137	Alpha-N-acetylgalactosaminidase deficiency		20 Cases
79279	Alpha-N-acetylgalactosaminidase deficiency type 1		10 Cases
79280	Alpha-N-acetylgalactosaminidase deficiency type 2		10 Cases
79281	Alpha-N-acetylgalactosaminidase deficiency type 3		10 Cases
98791	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16		20 Cases
231401	Alpha-thalassemia-myelodysplastic syndrome		80 Cases
847	Alpha-thalassemia-X-linked intellectual disability syndrome		200 Cases
63	Alport syndrome	2.0 *P* *	
86818	Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome		2 Families
64	Alström syndrome		950 Cases
284	Alveolar echinococcosis	0.16 *I* *	
169095	Alymphoid cystic thymic dysgenesis		9 Cases
93561	ALys amyloidosis		7 Families
1021	Amaurosis-hypertrichosis syndrome		2 Cases
314422	Ameloblastic carcinoma		40 Cases
171836	Amelogenesis imperfecta-gingival hyperplasia syndrome		8 Cases
1031	Amelogenesis imperfecta-nephrocalcinosis syndrome		11 Cases
1028	Ameloonychohypohidrotic syndrome		2 Cases
1908	Aminopterin/methotrexate embryofetopathy		17 Cases
319635	Amyloidosis cutis dyschromia		27 Cases
803	Amyotrophic lateral sclerosis	3.85 *P*	
803	Amyotrophic lateral sclerosis	1.35 *I*	
357043	Amyotrophic lateral sclerosis type 4		70 Cases
228113	Anal fistula	23.0 *P* *	
98841	Anaplastic large cell lymphoma	2.0 *P* *	
251630	Anaplastic oligodendroglioma	0.09 *I* *	
142	Anaplastic thyroid carcinoma	0.1 *P* *	
142	Anaplastic thyroid carcinoma	0.17 *I* *	
93347	Anauxetic dysplasia		10 Cases
157954	ANE syndrome		5 Cases
284984	Aneurysm-osteoarthritis syndrome		45 Cases
63442	Angel-shaped phalango-epiphyseal dysplasia		20 Cases
72	Angelman syndrome	7.5 *P*	
72	Angelman syndrome	1.3 *BP* *	
251671	Angiocentric glioma		52 Cases
2346	Angioosteohypertrophic syndrome	0.8 *BP* *	
370039	Angora hair nevus		2 Cases
69088	Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome		2 Cases
77	Aniridia	1.75 *P*	
77	Aniridia	1.3 *I* *	
1069	Aniridia-absent patella syndrome		3 Cases

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1065	Aniridia-cerebellar ataxia-intellectual disability syndrome		22 Families
1068	Aniridia-intellectual disability syndrome		2 Cases
1067	Aniridia-ptosis-intellectual disability-familial obesity syndrome		3 Cases
1064	Aniridia-renal agenesis-psychomotor retardation syndrome		2 Cases
1070	Anisakiasis	0.32 *I*	
1074	Ankyloblepharon filiforme adnatum-imperforate anus syndrome		3 Families
2206	Ankylosing vertebral hyperostosis with tylosis		8 Cases
254411	Annular atrophic lichen planus		10 Cases
281139	Annular epidermolytic ichthyosis		7 Families
675	Annular pancreas	1.8 *BP* *	
69125	Anonychia with flexural pigmentation		3 Cases
1094	Anonychia-microcephaly syndrome		4 Cases
90390	Anonychia-onychodystrophy syndrome		14 Cases
1104	Anophthalmia plus syndrome		17 Cases
1101	Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome		3 Cases
98555	Anophthalmia-microphthalmia syndrome	8.3 *BP* *	
77298	Anophthalmia/microphthalmia-esophageal atresia syndrome		30 Cases
93976	Anotia	0.028 *BP* *	
2987	Antecubital pterygium syndrome		11 Cases
90079	Anthracycline extravasation	0.3 *P* *	
375	Anti-glomerular basement membrane disease	0.08 *I* *	
454710	Anti-p200 pemphigoid		50 Cases
81	Antisynthetase syndrome	3.5 *P*	
83	Antley-Bixler syndrome		34 Cases
1457	Aorta coarctation	35.6 *BP* *	
1110	Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome		4 Cases
2299	Aortic arch interruption	0.3 *BP* *	
3400	Aorto-ventricular tunnel		130 Cases
1112	Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome		3 Cases
1113	Aphalangy-syndactyly-microcephaly syndrome		5 Cases
324540	Aphonia-deafness-retinal dystrophy-bifid halluces-intellectual disability syndrome		2 Cases
1114	Aplasia cutis congenita	10.0 *BP*	
1116	Aplasia cutis congenita-intestinal lymphangiectasia syndrome		3 Cases
1117	Aplasia cutis-myopia syndrome		4 Cases
99981	Apnea of prematurity	8.5 *P* *	
425	Apolipoprotein A-I deficiency		30 Families
1126	Aprosencephaly cerebellar dysgenesis		2 Cases
1129	Arachnodactyly-abnormal ossification-intellectual disability syndrome		5 Cases
1130	Arachnodactyly-intellectual disability-dysmorphism syndrome		3 Cases
1133	AREDYLD syndrome		3 Cases
23	Argininosuccinic aciduria	1.0 *P* *	
23	Argininosuccinic aciduria	0.46 *BP*	
91	Aromatase deficiency		35 Cases
178345	Aromatase excess syndrome		30 Cases
35708	Aromatic L-amino acid decarboxylase deficiency		100 Cases
1135	Arrhinia-choanal atresia-microphthalmia syndrome		4 Cases
247	Arrhythmogenic right ventricular cardiomyopathy	20.0 *P*	
1682	Arterial dissection-lentiginosis syndrome		4 Cases
3342	Arterial tortuosity syndrome		80 Cases
1037	Arthrogryposis multiplex congenita	5.7 *BP* *	
1150	Arthrogryposis multiplex congenita-whistling face syndrome		10 Cases
3200	Arthrogryposis-ectodermal dysplasia-other anomalies syndrome		2 Cases
1485	Arthrogryposis-hyperkeratosis syndrome, lethal form		2 Cases
1144	Arthrogryposis-like hand anomaly-sensorineural deafness syndrome		1 Family
1149	Arthrogryposis-like syndrome		8 Families
2697	Arthrogryposis-renal dysfunction-cholestasis syndrome		100 Cases
65720	Arthrogryposis-severe scoliosis syndrome		2 Families
1253	Ascher syndrome		50 Cases
137686	Asherman syndrome	44.0 *P* *	
85175	Astley-Kendall dysplasia		5 Cases
251679	Astroblastoma	0.02 *I* *	

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ORPHA Number	Disease or Group of diseases	Estimated prevalence/incidence (/100,000)	Number of published cases or families
94	Astrocytoma	2.5 *P* *	
94	Astrocytoma	4.8 *I* *	
96	Ataxia with vitamin E deficiency	0.33 *P* *	
1188	Ataxia-deafness-intellectual disability syndrome		8 Cases
370022	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome		7 Cases
459033	Ataxia-oculomotor apraxia type 4		12 Cases
1184	Ataxia-photosensitivity-short stature syndrome		2 Cases
100	Ataxia-telangiectasia	0.49 *P* *	
1190	Atelosteogenesis type I		12 Cases
56304	Atelosteogenesis type II		25 Cases
56305	Atelosteogenesis type III		25 Cases
69739	Athabaskan brainstem dysgenesis syndrome		13 Cases
1192	Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome		2 Cases
95713	Athyreosis	3.5 *P* *	
1193	Atkin-Flaitz syndrome		14 Cases
163934	Atopic keratoconjunctivitis	15.0 *P* *	
1201	Atresia of small intestine	16.0 *BP* *	
1479	Atrial septal defect-atrioventricular conduction defects syndrome		11 Cases
1352	Atrioventricular defect-blepharophimosis-radial and anal defect syndrome		2 Cases
352723	Attenuated Chédiak-Higashi syndrome		100 Cases
1456	Atypical coarctation of aorta	0.17 *BP* *	
314721	Atypical dentin dysplasia due to SMOC2 deficiency		4 Cases
289863	Atypical glycine encephalopathy		20 Cases
2134	Atypical hemolytic-uremic syndrome	1.0 *P* *	
357008	Atypical hemolytic-uremic syndrome with DGKE deficiency		13 Cases
238523	Atypical hypotonia-cystinuria syndrome		2 Cases
391411	Atypical juvenile parkinsonism		6 Families
86797	Atypical lichen myxedematosus		20 Cases
314466	Atypical Meigs syndrome		9 Cases
77300	Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome		2 Cases
137888	Auriculocondylar syndrome		50 Cases
114	Auriculoosteodysplasia		2 Families
352490	Autism spectrum disorder due to AUTS2 deficiency		60 Cases
370943	Autism spectrum disorder-epilepsy-arthrogryposis syndrome		8 Cases
308410	Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency		5 Families
137911	Autism-facial port-wine stain syndrome		4 Cases
324636	Autoerythrocyte sensitization syndrome		170 Cases
420789	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea		10 Cases
391487	Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome		5 Cases
98375	Autoimmune hemolytic anemia	2.02 *I* *	
444463	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome		6 Cases
2137	Autoimmune hepatitis	23.5 *P*	
2137	Autoimmune hepatitis	1.2 *I*	
444092	Autoimmune interstitial lung disease-arthritis syndrome		5 Families
3261	Autoimmune lymphoproliferative syndrome		500 Cases
436159	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency		17 Cases
275517	Autoimmune lymphoproliferative syndrome with recurrent viral infections		1 Family
747	Autoimmune pulmonary alveolar proteinosis	0.5 *P*	
747	Autoimmune pulmonary alveolar proteinosis	0.04 *I*	
324530	Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation		2 Cases
329173	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis		3 Cases
33110	Autosomal agammaglobulinemia		100 Cases
209335	Autosomal dominant adult-onset proximal spinal muscular atrophy	0.1 *P* *	
314399	Autosomal dominant aplasia and myelodysplasia		6 Cases
99	Autosomal dominant cerebellar ataxia	2.7 *P*	
314404	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome		24 Cases
487814	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation		2 Cases

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435819	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation		2 Cases
401964	Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons		2 Families
99946	Autosomal dominant Charcot-Marie-Tooth disease type 2A1		1 Family
99938	Autosomal dominant Charcot-Marie-Tooth disease type 2D		44 Cases
99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F		5 Families
99941	Autosomal dominant Charcot-Marie-Tooth disease type 2G		1 Family
99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K		30 Cases
99945	Autosomal dominant Charcot-Marie-Tooth disease type 2L		1 Family
228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M		20 Cases
228174	Autosomal dominant Charcot-Marie-Tooth disease type 2N		28 Cases
329258	Autosomal dominant Charcot-Marie-Tooth disease type 2Q		8 Cases
397735	Autosomal dominant Charcot-Marie-Tooth disease type 2U		2 Cases
447964	Autosomal dominant Charcot-Marie-Tooth disease type 2V		21 Cases
488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W		24 Cases
435387	Autosomal dominant Charcot-Marie-Tooth disease type 2Y		7 Cases
466768	Autosomal dominant Charcot-Marie-Tooth disease type 2Z		21 Cases
363454	Autosomal dominant childhood-onset proximal spinal muscular atrophy with contractures		25 Cases
447753	Autosomal dominant complex spastic paraplegia type 9A		2 Families
447757	Autosomal dominant complex spastic paraplegia type 9B		3 Families
90348	Autosomal dominant cutis laxa		50 Cases
79499	Autosomal dominant deafness-onychodystrophy syndrome		22 Cases
476093	Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome		8 Cases
329466	Autosomal dominant focal dystonia, DYT25 type		28 Cases
402003	Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering		21 Cases
2314	Autosomal dominant hyper-IgE syndrome	0.1 *I* *	
1810	Autosomal dominant hypohidrotic ectodermal dysplasia		40 Cases
89937	Autosomal dominant hypophosphatemic rickets		100 Cases
457193	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome		16 Cases
100043	Autosomal dominant intermediate Charcot-Marie-Tooth disease type A		20 Cases
100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B		37 Cases
100045	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C		35 Cases
100046	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D		12 Cases
93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E		21 Cases
352670	Autosomal dominant intermediate Charcot-Marie-Tooth disease type F		8 Cases
324585	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain		9 Cases
503	Autosomal dominant Larsen syndrome	0.4 *BP* *	
266	Autosomal dominant limb-girdle muscular dystrophy type 1A		4 Families
34516	Autosomal dominant limb-girdle muscular dystrophy type 1D		6 Families
34517	Autosomal dominant limb-girdle muscular dystrophy type 1E		20 Families
55595	Autosomal dominant limb-girdle muscular dystrophy type 1F		64 Cases
55596	Autosomal dominant limb-girdle muscular dystrophy type 1G		2 Families
238755	Autosomal dominant limb-girdle muscular dystrophy type 1H		11 Cases
140957	Autosomal dominant macrothrombocytopenia		100 Cases
319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency		68 Cases
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency		2 Cases
457050	Autosomal dominant mitochondrial myopathy with exercise intolerance		15 Cases
65743	Autosomal dominant multiple pterygium syndrome		4 Cases
99846	Autosomal dominant myoglobinuria		2 Families
440354	Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome		1 Family

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329211	Autosomal dominant neovascular inflammatory vitreoretinopathy		99 Cases
98784	Autosomal dominant nocturnal frontal lobe epilepsy		100 Families
98672	Autosomal dominant optic atrophy	3.3 *P*	
67036	Autosomal dominant optic atrophy and cataract		3 Families
1215	Autosomal dominant optic atrophy plus syndrome	0.4 *P* *	
2783	Autosomal dominant osteopetrosis type 1		33 Cases
1010	Autosomal dominant palmoplantar keratoderma and congenital alopecia		10 Cases
730	Autosomal dominant polycystic kidney disease	3.96 *P* *	
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis		30 Cases
1300	Autosomal dominant popliteal pterygium syndrome	0.3 *P*	
476119	Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome		1 Family
34528	Autosomal dominant primary hypomagnesemia with hypocalciuria		28 Cases
88659	Autosomal dominant progressive nephropathy with hypertension		14 Cases
314889	Autosomal dominant proximal renal tubular acidosis		1 Family
209867	Autosomal dominant rhegmatogenous retinal detachment		38 Cases
3107	Autosomal dominant Robinow syndrome		100 Cases
140481	Autosomal dominant slowed nerve conduction velocity		1 Family
251282	Autosomal dominant spastic ataxia type 1		53 Cases
100988	Autosomal dominant spastic paraplegia type 6		10 Families
100989	Autosomal dominant spastic paraplegia type 8		10 Families
100990	Autosomal dominant spastic paraplegia type 9		7 Families
100991	Autosomal dominant spastic paraplegia type 10		10 Families
100993	Autosomal dominant spastic paraplegia type 12		27 Cases
100994	Autosomal dominant spastic paraplegia type 13		10 Cases
100998	Autosomal dominant spastic paraplegia type 17		20 Families
100999	Autosomal dominant spastic paraplegia type 19		1 Family
101009	Autosomal dominant spastic paraplegia type 29		1 Family
320365	Autosomal dominant spastic paraplegia type 36		1 Family
171612	Autosomal dominant spastic paraplegia type 37		13 Cases
171617	Autosomal dominant spastic paraplegia type 38		1 Family
320355	Autosomal dominant spastic paraplegia type 41		7 Cases
171863	Autosomal dominant spastic paraplegia type 42		1 Family
444099	Autosomal dominant spastic paraplegia type 73		1 Family
228169	Autosomal dominant striatal neurodegeneration		11 Cases
466806	Autosomal dominant thrombocytopenia with platelet secretion defect		4 Families
3357	Autosomal dominant trichoodontoonychodysplasia-syndactyly		6 Cases
34149	Autosomal dominant tubulointerstitial kidney disease	0.11 *P* *	
3086	Autosomal dominant vitreoretinochoroidopathy		3 Cases
79278	Autosomal erythropoietic protoporphyria	0.92 *P* *	
79278	Autosomal erythropoietic protoporphyria	0.012 *I* *	
1027	Autosomal recessive amelia		3 Cases
247815	Autosomal recessive ataxia due to PEX10 deficiency		6 Cases
139485	Autosomal recessive ataxia due to ubiquinone deficiency		31 Cases
88644	Autosomal recessive ataxia, Beauce type		57 Cases
324442	Autosomal recessive axonal neuropathy with neuromyotonia		33 Families
139455	Autosomal recessive bestrophinopathy		20 Cases
448242	Autosomal recessive brachyolmia		20 Cases
1172	Autosomal recessive cerebellar ataxia	3.3 *P*	
453521	Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency		2 Cases
412057	Autosomal recessive cerebellar ataxia due to STUB1 deficiency		10 Families
352641	Autosomal recessive cerebellar ataxia with late-onset spasticity		10 Cases
404499	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency		2 Cases
404493	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency		3 Cases
363429	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome		17 Cases

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95434	Autosomal recessive cerebellar ataxia-saccadic intrusion syndrome		1 Family
363969	Autosomal recessive cerebral atrophy		4 Cases
466775	Autosomal recessive Charcot-Marie-Tooth disease type 2X		29 Cases
506353	Autosomal recessive complex spastic paraplegia due to Kennedy pathway dysfunction		4 Cases
447760	Autosomal recessive complex spastic paraplegia type 9B		2 Families
363432	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency		7 Cases
324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency		10 Cases
281097	Autosomal recessive congenital ichthyosis	0.5 *P* *	
90349	Autosomal recessive cutis laxa type 1		60 Cases
90350	Autosomal recessive cutis laxa type 2		40 Cases
101150	Autosomal recessive dopa-responsive dystonia		50 Cases
1974	Autosomal recessive faciodigitogenital syndrome		26 Cases
329329	Autosomal recessive frontotemporal pachygyria		7 Cases
300547	Autosomal recessive infantile hypercalcemia		12 Cases
217055	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A		8 Families
254334	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B		1 Case
369867	Autosomal recessive intermediate Charcot-Marie-Tooth disease type C		3 Cases
435998	Autosomal recessive intermediate Charcot-Marie-Tooth disease type D		4 Cases
98676	Autosomal recessive isolated optic atrophy		5 Cases
314572	Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome		3 Cases
267	Autosomal recessive limb-girdle muscular dystrophy type 2A	1.0 *P* *	
353	Autosomal recessive limb-girdle muscular dystrophy type 2C	0.2 *P* *	
119	Autosomal recessive limb-girdle muscular dystrophy type 2E	0.1 *P* *	
219	Autosomal recessive limb-girdle muscular dystrophy type 2F	0.3 *P* *	
34514	Autosomal recessive limb-girdle muscular dystrophy type 2G		16 Cases
34515	Autosomal recessive limb-girdle muscular dystrophy type 2I	1.0 *P* *	
140922	Autosomal recessive limb-girdle muscular dystrophy type 2J		1 Family
206554	Autosomal recessive limb-girdle muscular dystrophy type 2M		5 Families
206559	Autosomal recessive limb-girdle muscular dystrophy type 2N		1 Case
206564	Autosomal recessive limb-girdle muscular dystrophy type 2O		2 Cases
280333	Autosomal recessive limb-girdle muscular dystrophy type 2P		1 Case
254361	Autosomal recessive limb-girdle muscular dystrophy type 2Q		6 Cases
363543	Autosomal recessive limb-girdle muscular dystrophy type 2R		2 Cases
369840	Autosomal recessive limb-girdle muscular dystrophy type 2S		3 Cases
363623	Autosomal recessive limb-girdle muscular dystrophy type 2T		2 Cases
352479	Autosomal recessive limb-girdle muscular dystrophy type 2U		8 Cases
466801	Autosomal recessive limb-girdle muscular dystrophy type 2W		2 Cases
476084	Autosomal recessive limb-girdle muscular dystrophy type 2X		3 Cases
424261	Autosomal recessive limb-girdle muscular dystrophy type 2Y		3 Cases
480682	Autosomal recessive limb-girdle muscular dystrophy type 2Z		4 Cases
206580	Autosomal recessive lower motor neuron disease with childhood onset		5 Cases
238505	Autosomal recessive lymphoproliferative disease		17 Cases
667	Autosomal recessive malignant osteopetrosis	0.75 *BP* *	
477857	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency		7 Cases
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency		18 Cases
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency		6 Cases
319332	Autosomal recessive myogenic arthrogryposis multiplex congenita		1 Family
280654	Autosomal recessive nail dysplasia		4 Cases
93329	Autosomal recessive omodysplasia		23 Cases
227976	Autosomal recessive optic atrophy, OPA7 type		17 Cases

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1366	Autosomal recessive palmoplantar keratoderma and congenital alopecia		8 Cases
731	Autosomal recessive polycystic kidney disease	1.17 *P* *	
437552	Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity		3 Cases
1507	Autosomal recessive Robinow syndrome		100 Cases
420702	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency		4 Cases
420699	Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency		2 Cases
331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency		57 Cases
423384	Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency		14 Cases
314603	Autosomal recessive spastic ataxia with leukoencephalopathy		54 Cases
254343	Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome		6 Cases
100995	Autosomal recessive spastic paraplegia type 14		1 Family
100996	Autosomal recessive spastic paraplegia type 15		10 Families
209951	Autosomal recessive spastic paraplegia type 18		9 Cases
101000	Autosomal recessive spastic paraplegia type 20		29 Cases
101001	Autosomal recessive spastic paraplegia type 21		35 Cases
101003	Autosomal recessive spastic paraplegia type 23		5 Families
101004	Autosomal recessive spastic paraplegia type 24		1 Family
101005	Autosomal recessive spastic paraplegia type 25		1 Family
101006	Autosomal recessive spastic paraplegia type 26		10 Families
101007	Autosomal recessive spastic paraplegia type 27		10 Cases
101008	Autosomal recessive spastic paraplegia type 28		6 Cases
171622	Autosomal recessive spastic paraplegia type 32		1 Family
171629	Autosomal recessive spastic paraplegia type 35		38 Cases
139480	Autosomal recessive spastic paraplegia type 39		2 Families
320370	Autosomal recessive spastic paraplegia type 43		2 Cases
320401	Autosomal recessive spastic paraplegia type 44		3 Cases
320396	Autosomal recessive spastic paraplegia type 45		7 Families
320391	Autosomal recessive spastic paraplegia type 46		5 Cases
306511	Autosomal recessive spastic paraplegia type 48		2 Cases
320385	Autosomal recessive spastic paraplegia type 49		5 Cases
319199	Autosomal recessive spastic paraplegia type 53		9 Cases
320380	Autosomal recessive spastic paraplegia type 54		6 Families
320375	Autosomal recessive spastic paraplegia type 55		14 Cases
320411	Autosomal recessive spastic paraplegia type 56		5 Families
431329	Autosomal recessive spastic paraplegia type 57		2 Cases
397946	Autosomal recessive spastic paraplegia type 58		19 Cases
401795	Autosomal recessive spastic paraplegia type 59		3 Cases
401800	Autosomal recessive spastic paraplegia type 60		1 Case
401780	Autosomal recessive spastic paraplegia type 61		4 Cases
401785	Autosomal recessive spastic paraplegia type 62		7 Cases
401805	Autosomal recessive spastic paraplegia type 63		2 Cases
401810	Autosomal recessive spastic paraplegia type 64		4 Cases
401815	Autosomal recessive spastic paraplegia type 66		2 Cases
401820	Autosomal recessive spastic paraplegia type 67		2 Cases
401825	Autosomal recessive spastic paraplegia type 68		1 Case
401830	Autosomal recessive spastic paraplegia type 69		2 Cases
401835	Autosomal recessive spastic paraplegia type 70		4 Cases
401840	Autosomal recessive spastic paraplegia type 71		1 Case
468661	Autosomal recessive spastic paraplegia type 74		11 Cases
459056	Autosomal recessive spastic paraplegia type 75		5 Cases
488594	Autosomal recessive spastic paraplegia type 76		7 Families
466722	Autosomal recessive spastic paraplegia type 77		4 Cases
95433	Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome		3 Families
401979	Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type		4 Cases

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250984	Autosomal recessive Stickler syndrome		15 Cases
280365	Autosomal semi-dominant severe lipodystrophic laminopathy		7 Cases
101010	Autosomal spastic paraplegia type 30		3 Families
401849	Autosomal spastic paraplegia type 72		14 Cases
300345	Autosomal systemic lupus erythematosus		7 Families
454836	Avian influenza		826 Cases
782	Axenfeld-Rieger syndrome	0.5 *P* *	
168549	Axial spondylometaphyseal dysplasia		13 Cases
401911	AXIN2-related attenuated familial adenomatous polyposis		4 Families
1272	Aymé-Gripp syndrome		18 Cases
79332	B4GALT1-CDG		1 Case
67038	B-cell chronic lymphocytic leukemia	48.0 *P* *	
171915	B-cell non-Hodgkin lymphoma	17.45 *I* *	
86852	B-cell prolymphocytic leukemia	0.05 *I* *	
36234	Bacterial toxic-shock syndrome	3.0 *P*	
93395	Ballard syndrome		12 Cases
1225	Baller-Gerold syndrome		30 Cases
1226	Bamforth-Lazarus syndrome		8 Cases
1227	Bangstad syndrome		2 Cases
1228	Banki syndrome		1 Family
2995	Baraitser-Winter cerebrofrontofacial syndrome		60 Cases
1231	Barber-Say syndrome		16 Cases
110	Bardet-Biedl syndrome	0.7 *P* *	
110	Bardet-Biedl syndrome	0.5 *BP* *	
111	Barth syndrome	0.22 *P* *	
1234	Bartsocas-Papas syndrome		24 Cases
112	Bartter syndrome	0.1 *I* *	
100976	Bathing suit ichthyosis		20 Cases
166113	Bazex syndrome		145 Cases
113	Bazex-Dupré-Christol syndrome		143 Cases
98895	Becker muscular dystrophy	1.53 *P*	
98895	Becker muscular dystrophy	2.2 *BP* *	
116	Beckwith-Wiedemann syndrome	3.5 *BP* *	
1237	Beemer-Ertbruggen syndrome		2 Cases
1241	Bencze syndrome		2 Families
251287	Benign concentric annular macular dystrophy		27 Cases
1949	Benign familial neonatal epilepsy		100 Families
140927	Benign familial neonatal-infantile seizures		10 Families
166308	Benign infantile focal epilepsy with midline spikes and waves during sleep		36 Cases
166305	Benign infantile seizures associated with mild gastroenteritis		100 Cases
209973	Benign nocturnal alternating hemiplegia of childhood		12 Cases
1179	Benign paroxysmal tonic upgaze of childhood with ataxia		12 Cases
71518	Benign paroxysmal torticollis of infancy		50 Cases
324581	Benign Samaritan congenital myopathy		4 Cases
252164	Benign schwannoma	6.0 *P* *	
464336	BENTA disease		8 Cases
528	Berardinelli-Seip congenital lipodystrophy	0.5 *P* *	
274	Bernard-Soulier syndrome		100 Cases
118	Beta-mannosidosis	0.14 *BP* *	
1035	Beta-mercaptolactate cysteine disulfiduria		1 Case
329284	Beta-propeller protein-associated neurodegeneration		54 Cases
848	Beta-thalassemia	1.0 *I*	
65287	Beta-ureidopropionase deficiency		5 Cases
69736	Bilateral acute depigmentation of the iris		62 Cases
140963	Bilateral microtia-deafness-cleft palate syndrome		3 Families
1980	Bilateral striopallidodentate calcinosis		200 Cases
79241	Biotinidase deficiency	1.6 *P* *	
79241	Biotinidase deficiency	1.6 *BP*	
364198	Bipartite talus		23 Cases
179	Birdshot chorioretinopathy	0.35 *P*	
122	Birt-Hogg-Dubé syndrome	0.5 *P* *	
123	Björnstad syndrome		33 Cases
124	Blackfan-Diamond anemia	0.67 *BP* *	
93930	Bladder exstrophy	3.05 *BP*	
73271	Bleeding diathesis due to a collagen receptor defect		20 Cases
420566	Bleeding disorder due to CalDAG-GEFI deficiency		3 Cases
1997	Blepharo-cheilo-odontic syndrome		50 Cases
1252	Blepharonasofacial malformation syndrome		3 Families
126	Blepharophimosis-epicanthus inversus-ptosis syndrome	2.0 *P*	

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293642	Blepharophimosis-intellectual disability syndrome		58 Cases
2728	Blepharophimosis-intellectual disability syndrome, Ohdo type		30 Cases
3047	Blepharophimosis-intellectual disability syndrome, SBBYS type		20 Cases
2057	Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome		6 Cases
1259	Blepharoptosis-myopia-ectopia lentis syndrome		3 Cases
171844	Blindness-scoliosis-arachnodactyly syndrome		4 Cases
50945	Blomstrand lethal chondrodysplasia		13 Cases
125	Bloom syndrome		400 Cases
16	Blue cone monochromatism	1.0 *P*	
16	Blue cone monochromatism	1.0 *BP*	
1059	Blue rubber bleb nevus		200 Cases
217266	BNAR syndrome		9 Families
217008	Bockenheimer syndrome		40 Cases
91135	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency		11 Cases
97297	Bohring-Opitz syndrome		30 Cases
1842	Bone dysplasia, lethal Holmgren type		7 Cases
223727	Bone sarcoma	9.29 *P* *	
223727	Bone sarcoma	0.8 *I* *	
1261	Bonnemann-Meinecke-Reich syndrome		4 Cases
1262	Böök syndrome		26 Cases
1263	Boomerang dysplasia		10 Cases
127	Borjeson-Forssman-Lehmann syndrome		50 Cases
69737	Bosley-Salih-Alorainy syndrome		16 Cases
1267	Botulism	0.02 *I* *	
1270	Bowen-Conradi syndrome		60 Cases
93389	Brachydactyly type A5		2 Families
93382	Brachydactyly type A6		7 Cases
93397	Brachydactyly type A7		1 Family
1276	Brachydactyly-arterial hypertension syndrome		10 Families
1275	Brachydactyly-elbow wrist dysplasia syndrome		4 Families
2946	Brachydactyly-long thumb syndrome		4 Cases
1277	Brachydactyly-mesomelia-intellectual disability-heart defects syndrome		2 Cases
1246	Brachydactyly-nystagmus-cerebellar ataxia syndrome		1 Family
1278	Brachydactyly-preaxial hallux varus syndrome		8 Cases
166035	Brachydactyly-short stature-retinitis pigmentosa syndrome		12 Cases
93409	Brachydactyly-syndactyly, Zhao type		2 Families
1292	Brachymorphism-onychodysplasia-dysphalangism syndrome		9 Cases
1293	Brachyolmia		100 Cases
93302	Brachyolmia, Maroteaux type		4 Families
1295	Brachytelephalangy-dysmorphism-Kallmann syndrome		2 Cases
52047	Braddock syndrome		2 Cases
75374	Bradyopsia		5 Cases
178506	Brain calcification, Rajab type		8 Cases
168598	Brain demyelination due to methionine adenosyltransferase deficiency		2 Cases
352649	Brain dopamine-serotonin vesicular transport disease		8 Cases
75389	Brain malformation-congenital heart disease-postaxial polydactyly syndrome		2 Cases
500150	Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome		29 Cases
209905	Brain-lung-thyroid syndrome		100 Cases
1297	Branchio-oculo-facial syndrome		50 Cases
50815	Branchiogenic deafness syndrome		5 Cases
1299	Branchioskeletogenital syndrome		7 Cases
85284	BRESEK syndrome		5 Cases
90354	Brittle cornea syndrome		65 Cases
70589	Bronchopulmonary dysplasia	13.0 *P* *	
79493	Brooke-Spiegler syndrome		100 Cases
1304	Brucellosis	0.07 *I* *	
2771	Bruck syndrome		60 Cases
130	Brugada syndrome	20.0 *P* *	
131	Budd-Chiari syndrome	1.5 *P* *	
36258	Buerger disease	16.0 *P*	
280785	Bullous diffuse cutaneous mastocytosis		40 Cases
1867	Bullous dystrophy, macular type		2 Families
703	Bullous pemphigoid	26.0 *P* *	
543	Burkitt lymphoma	0.17 *I* *	
1306	Buschke-Ollendorff syndrome	5.0 *I*	
1308	C syndrome	0.11 *P* *	

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495844	C11ORF73-related autosomal recessive hypomyelinating leukodystrophy		6 Cases
497623	C12ORF65-related combined oxidative phosphorylation defect		30 Cases
135	CACH syndrome		148 Cases
448010	CAD-CDG		1 Case
136	CADASIL	3.0 *P* *	
369942	CADDS		4 Cases
1310	Caffey disease		100 Cases
280062	Calciphylaxis	5.0 *P* *	
85192	Calvarial doughnut lesions-bone fragility syndrome		20 Cases
83472	CAMOS syndrome		5 Cases
1318	Campomelia, Cumming type		8 Cases
140	Campomelic dysplasia	0.33 *BP* *	
1319	Camptobrachydactyly		1 Family
1327	Camptodactyly syndrome, Guadalajara type 1		8 Cases
1326	Camptodactyly syndrome, Guadalajara type 2		2 Cases
488434	Camptodactyly syndrome, Guadalajara type 3		5 Cases
2848	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome		30 Families
1321	Camptodactyly-fibrous tissue hyperplasia-skeletal dysplasia syndrome		3 Cases
1323	Camptodactyly-joint contractures-facial skeletal defects syndrome		4 Cases
85164	Camptodactyly-tall stature-scoliosis-hearing loss syndrome		30 Cases
1325	Camptodactyly-taurinuria syndrome		17 Cases
1328	Camurati-Engelmann disease		300 Cases
141	Canavan disease	1.0 *BP*	
325004	CANDLE syndrome		30 Cases
171881	Cap myopathy		21 Cases
160148	Cap polyposis		67 Cases
137667	Capillary malformation-arteriovenous malformation		261 Cases
147	Carbamoyl-phosphate synthetase 1 deficiency	0.31 *P*	
70482	Carcinoma of esophagus	9.8 *P*	
70482	Carcinoma of esophagus	7.0 *I*	
418945	Carcinoma of esophagus, salivary gland type	0.01 *I* *	
56044	Carcinoma of gallbladder and extrahepatic biliary tract	12.0 *I*	
423781	Carcinoma of stomach, salivary gland type	0.01 *I* *	
369891	Cardiac anomalies-developmental delay-facial dysmorphism syndrome		4 Cases
137628	Cardiac anomalies-heterotaxy syndrome		9 Cases
2872	Cardiocranial syndrome, Pfeiffer type		7 Cases
37553	Cardiodysrhythmic potassium-sensitive periodic paralysis	0.1 *I* *	
1340	Cardiofaciocutaneous syndrome		300 Cases
97292	Cardiogenic shock	40.0 *P* *	
1345	Cardiomyopathy-cataract-hip spine disease syndrome		9 Cases
91130	Cardiomyopathy-hypotonia-lactic acidosis syndrome		2 Cases
90022	Cardiomyopathy-renal anomalies syndrome		2 Cases
3238	Cardiospondylocarpofacial syndrome		5 Cases
1358	Carey-Fineman-Ziter syndrome		20 Cases
1359	Carney complex		160 Cases
319340	Carney complex-trismus-pseudocamptodactyly syndrome		3 Families
139411	Carney triad		150 Cases
97286	Carney-Stratakis syndrome		20 Families
156	Carnitine palmitoyl transferase 1A deficiency		60 Cases
228302	Carnitine palmitoyl transferase II deficiency, myopathic form		300 Cases
228308	Carnitine palmitoyl transferase II deficiency, neonatal form		20 Families
228305	Carnitine palmitoyl transferase II deficiency, severe infantile form		30 Families
157	Carnitine palmitoyltransferase II deficiency	1.0 *P* *	
159	Carnitine-acylcarnitine translocase deficiency		60 Cases
1361	Carnosinemia	0.2 *BP*	
53035	Caroli disease	0.1 *I*	
65759	Carpenter syndrome		70 Cases
93973	Carpenter-Waziri syndrome		6 Cases
65282	Carvajal syndrome		7 Cases
195	Cat-eye syndrome	1.35 *BP* *	
50839	Cat-scratch disease	6.6 *P* *	
1373	Cataract-aberrant oral frenula-growth delay syndrome		3 Cases
1368	Cataract-ataxia-deafness syndrome		2 Cases
314993	Cataract-congenital heart disease-neural tube defect syndrome		2 Cases
1383	Cataract-deafness-hypogonadism syndrome		3 Cases
162	Cataract-glaucoma syndrome		3 Families

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436174	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome		3 Cases
1381	Cataract-intellectual disability-anal atresia-urinary defects syndrome		3 Cases
1387	Cataract-intellectual disability-hypogonadism syndrome		20 Cases
1377	Cataract-microcornea syndrome		8 Families
1380	Cataract-nephropathy-encephalopathy syndrome		2 Cases
3286	Catecholaminergic polymorphic ventricular tachycardia	10.0 *P* *	
1388	Catel-Manzke syndrome		33 Cases
1123	Caudal appendage-deafness syndrome		2 Cases
468684	CCDC115-CDG		8 Cases
86870	CD4+/CD56+ hematodermic neoplasm	12.0 *P* *	
66631	CEDNIK syndrome		13 Cases
1459	Celiac disease-epilepsy-cerebral calcification syndrome		170 Cases
3258	Cenani-Lenz syndrome		30 Cases
2431	Central bilateral macrogyria		4 Cases
98972	Central cloudy dystrophy of François		24 Cases
178029	Central diabetes insipidus	4.0 *P* *	
3240	Central nervous system calcification-deafness-tubular acidosis-anemia syndrome		2 Cases
251870	Central nervous system primitive neuroectodermal tumor	0.07 *I* *	
73256	Central neurocytoma		500 Cases
411527	Central retinal vein occlusion	28.0 *P* *	
89841	Centripetalis recessive dystrophic epidermolysis bullosa		10 Cases
1171	Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome		10 Cases
2246	Cerebellar hypoplasia-tapetoretinal degeneration syndrome		3 Cases
444072	Cerebellar-facial-dental syndrome		3 Families
46724	Cerebral arteriovenous malformation	6.0 *P* *	
2081	Cerebral gigantism-jaw cysts syndrome		9 Cases
329217	Cerebral sinovenous thrombosis	0.35 *I* *	
1393	Cerebrocostomandibular syndrome		75 Cases
314679	Cerebrofacioarticular syndrome		9 Cases
1394	Cerebrofaciothoracic dysplasia		20 Cases
66625	Cerebrooculonasal syndrome		21 Cases
169079	Cernunnos-XLF deficiency		5 Cases
2218	Cervical hypertrichosis-peripheral neuropathy syndrome		4 Cases
88642	Channelopathy-associated congenital insensitivity to pain		20 Cases
46627	Char syndrome		10 Cases
65753	Charcot-Marie-Tooth disease type 1	17.5 *P*	
101101	Charcot-Marie-Tooth disease type 2B2		1 Family
228374	Charcot-Marie-Tooth disease type 2B5		4 Cases
101102	Charcot-Marie-Tooth disease type 2H		13 Cases
300319	Charcot-Marie-Tooth disease type 2P		18 Cases
397968	Charcot-Marie-Tooth disease type 2R		1 Case
443073	Charcot-Marie-Tooth disease type 2S		35 Cases
495274	Charcot-Marie-Tooth disease type 2T		10 Cases
99955	Charcot-Marie-Tooth disease type 4B1		11 Families
363981	Charcot-Marie-Tooth disease type 4B3		3 Cases
99954	Charcot-Marie-Tooth disease type 4H		15 Cases
139515	Charcot-Marie-Tooth disease type 4J		18 Cases
90103	Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome		7 Cases
166	Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy	25.0 *P* *	
138	CHARGE syndrome	6.5 *BP*	
1406	Charlie M syndrome		4 Cases
167	Chédiak-Higashi syndrome		500 Cases
1221	Cheilitis glandularis		100 Cases
184	Cherubism		300 Cases
324625	Chikungunya	0.01 *I* *	
139	CHILD syndrome		60 Cases
209908	Childhood apraxia of speech		22 Cases
168782	Childhood disintegrative disorder	2.0 *P* *	
293955	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency		5 Cases
363677	Childhood-onset autosomal recessive myopathy with external ophthalmoplegia		22 Cases
497906	Childhood-onset basal ganglia degeneration syndrome		4 Cases

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494541	Childhood-onset benign chorea with striatal involvement		3 Cases
500180	Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder		7 Cases
466921	Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome		3 Families
401866	Childhood-onset spasticity with hyperglycinemia		3 Cases
3474	CHIME syndrome		8 Cases
137914	Choanal atresia	8.6 *BP* *	
1200	Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome		11 Families
70567	Cholangiocarcinoma	2.1 *P*	
70567	Cholangiocarcinoma	4.2 *I*	
173	Cholera	0.01 *I* *	
1414	Cholestasis-lymphedema syndrome		47 Cases
1415	Cholestasis-pigmentary retinopathy-cleft palate syndrome		5 Cases
79347	Chondrodysplasia punctata, Toriello type		3 Cases
280586	Chondrodysplasia with joint dislocations, gPAPP type		4 Cases
1422	Chondrodysplasia-disorder of sex development syndrome		2 Cases
319195	Chondroectodermal dysplasia with night blindness		4 Cases
404507	Chondromyxoid fibroma		50 Cases
55880	Chondrosarcoma	0.24 *I* *	
251899	Choroid plexus carcinoma	0.01 *I* *	
1433	Choroidal atrophy-alopecia syndrome		2 Cases
180	Choroideremia	2.0 *P* *	
1435	Choroideremia-deafness-obesity syndrome		4 Cases
85278	Christianson syndrome		30 Cases
319303	Chromophobe renal cell carcinoma	0.01 *I* *	
435988	Chronic atrial and intestinal dysrhythmia syndrome		17 Cases
314373	Chronic diarrhea due to guanylate cyclase 2C overactivity		32 Cases
1670	Chronic diarrhea with villous atrophy		2 Cases
468641	Chronic enteropathy associated with SLCO2A1 gene		18 Cases
379	Chronic granulomatous disease	0.46 *BP*	
396	Chronic hiccup	1.0 *P* *	
2932	Chronic inflammatory demyelinating polyneuropathy	3.7 *P* *	
521	Chronic myeloid leukemia	6.0 *P* *	
521	Chronic myeloid leukemia	1.25 *I* *	
98823	Chronic myelomonocytic leukemia	0.29 *I* *	
86830	Chronic myeloproliferative disease, unclassifiable	0.53 *I* *	
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	0.3 *P*	
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	2.5 *I*	
95426	Chronic pain requiring intraspinal analgesia	12.0 *P* *	
101959	Chronic primary adrenal insufficiency	14.0 *P* *	
101959	Chronic primary adrenal insufficiency	0.4 *I* *	
70591	Chronic thromboembolic pulmonary hypertension	3.0 *P* *	
263463	CHST3-related skeletal dysplasia		2 Families
93971	Chudley-Lowry-Hoar syndrome		3 Cases
314597	Chudley-McCullough syndrome		25 Cases
71	Chylomicron retention disease		55 Cases
435651	CIDEC-related familial partial lipodystrophy		1 Case
1451	CINCA syndrome		200 Cases
69744	Circumscribed palmoplantar hypokeratosis		17 Cases
309854	Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome		20 Cases
247525	Citrullinemia type I	2.4 *P* *	
251383	CK syndrome		24 Cases
168984	CLAPO syndrome		6 Cases
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	7.0 *P* *	
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	7.0 *BP*	
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	7.5 *P* *	
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	7.5 *BP* *	
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	2.5 *P* *	
79239	Classic galactosemia	2.1 *I* *	
58017	Classic hairy cell leukemia	0.29 *I* *	
391	Classic Hodgkin lymphoma	2.38 *I* *	

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98846	Classic Hodgkin lymphoma, lymphocyte-depleted type	0.04 *I* *	
98845	Classic Hodgkin lymphoma, lymphocyte-rich type	0.1 *I* *	
98844	Classic Hodgkin lymphoma, mixed cellularity type	0.42 *I* *	
98843	Classic Hodgkin lymphoma, nodular sclerosis type	1.28 *I* *	
394	Classic homocystinuria	1.65 *P* *	
394	Classic homocystinuria	0.3 *BP*	
2584	Classic mycosis fungoides	0.5 *I* *	
329977	Classic neuroendocrine tumor of appendix	0.25 *I*	
485350	CLCN4-related X-linked intellectual disability syndrome		38 Cases
398971	Clear cell adenocarcinoma of the ovary	0.32 *I* *	
319276	Clear cell renal carcinoma	1.99 *I* *	
1995	Cleft lip-retinopathy syndrome		2 Cases
199306	Cleft lip/palate	80.0 *BP*	
2003	Cleft lip/palate-deafness-sacral lipoma syndrome		2 Cases
2001	Cleft lip/palate-intestinal malrotation-cardiopathy syndrome		5 Cases
2014	Cleft palate	53.6 *BP* *	
2013	Cleft palate-large ears-small head syndrome		8 Cases
2016	Cleft palate-lateral synechia syndrome		11 Cases
2015	Cleft palate-short stature-vertebral anomalies syndrome		2 Cases
2010	Cleft palate-stapes fixation-oligodontia syndrome		2 Cases
1452	Cleidocranial dysplasia	0.1 *P*	
1452	Cleidocranial dysplasia	0.4 *BP* *	
1453	Cleidorhizomelic syndrome		2 Cases
284448	CLIPPERS		50 Cases
228357	CLN9 disease		2 Cases
314629	CLN11 disease		2 Cases
352709	CLN13 disease		4 Cases
93929	Cloacal exstrophy	0.54 *BP*	
93267	Cloverleaf skull-multiple congenital anomalies syndrome		3 Cases
140944	CLOVES syndrome		40 Cases
397725	COASY protein-associated neurodegeneration		2 Cases
53721	Cobb syndrome		45 Cases
51577	Cobblestone lissencephaly	1.0 *BP* *	
352682	Cobblestone lissencephaly without muscular or ocular involvement		4 Cases
90068	Cocaine intoxication	1.0 *P* *	
3233	Cochleosaccular degeneration-cataract syndrome		2 Families
191	Cockayne syndrome	0.2 *BP* *	
191	Cockayne syndrome	0.5 *I* *	
1458	CODAS syndrome		12 Cases
192	Coffin-Lowry syndrome	1.5 *P*	
1465	Coffin-Siris syndrome		190 Cases
1466	COFS syndrome		20 Cases
263508	COG1-CDG		3 Cases
435934	COG2-CDG		1 Case
263501	COG4-CDG		2 Cases
263487	COG5-CDG		9 Cases
464443	COG6-CGD		10 Cases
79333	COG7-CDG		8 Cases
95428	COG8-CDG		2 Cases
1467	Cogan syndrome		300 Cases
444077	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome		3 Cases
193	Cohen syndrome		200 Cases
31824	Colchicine poisoning	0.1 *P* *	
157820	Cold-induced sweating syndrome		6 Cases
2050	Cole-Carpenter syndrome		3 Cases
1471	Coloboma of macula-brachydactyly type B syndrome		10 Cases
424099	Colobomatous microphthalmia-rhizomelic dysplasia syndrome		5 Families
435930	Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome		3 Cases
35909	Combined deficiency of factor V and factor VIII	0.5 *P* *	
440727	Combined hamartoma of the retina and retinal pigment epithelium		120 Cases
169090	Combined immunodeficiency due to CRAC channel dysfunction		10 Cases
217390	Combined immunodeficiency due to DOCK8 deficiency		11 Cases
505227	Combined immunodeficiency due to GINS1 deficiency		5 Cases
445018	Combined immunodeficiency due to LRBA deficiency		23 Cases
397964	Combined immunodeficiency due to MALT1 deficiency		3 Cases

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504530	Combined immunodeficiency due to Moesin deficiency		7 Cases
317428	Combined immunodeficiency due to ORAI1 deficiency		6 Cases
431149	Combined immunodeficiency due to OX40 deficiency		1 Case
231154	Combined immunodeficiency due to partial RAG1 deficiency		9 Cases
314689	Combined immunodeficiency due to STK4 deficiency		7 Cases
476113	Combined immunodeficiency due to TFRC deficiency		2 Families
221139	Combined immunodeficiency with facio oculoskeletal anomalies		2 Cases
254920	Combined oxidative phosphorylation defect type 2		1 Case
254925	Combined oxidative phosphorylation defect type 4		2 Cases
254930	Combined oxidative phosphorylation defect type 7		7 Cases
319504	Combined oxidative phosphorylation defect type 8		7 Cases
319509	Combined oxidative phosphorylation defect type 9		4 Cases
324535	Combined oxidative phosphorylation defect type 11		32 Cases
319514	Combined oxidative phosphorylation defect type 13		2 Cases
319519	Combined oxidative phosphorylation defect type 14		5 Cases
319524	Combined oxidative phosphorylation defect type 15		16 Cases
369913	Combined oxidative phosphorylation defect type 17		20 Families
420728	Combined oxidative phosphorylation defect type 20		2 Cases
420733	Combined oxidative phosphorylation defect type 21		2 Cases
444013	Combined oxidative phosphorylation defect type 23		11 Cases
444458	Combined oxidative phosphorylation defect type 24		3 Cases
447954	Combined oxidative phosphorylation defect type 25		2 Cases
477684	Combined oxidative phosphorylation defect type 26		2 Cases
477774	Combined oxidative phosphorylation defect type 27		3 Cases
478029	Combined oxidative phosphorylation defect type 29		1 Case
478042	Combined oxidative phosphorylation defect type 30		2 Cases
309111	Combined pancreatic lipase-colipase deficiency		3 Cases
280133	Complement component 3 deficiency		27 Cases
99429	Complete androgen insensitivity syndrome	0.83 *P*	
99429	Complete androgen insensitivity syndrome	3.0 *I* *	
1329	Complete atrioventricular canal	20.0 *BP* *	
98949	Complete cryptophthalmia		15 Cases
457378	Complex lethal osteochondrodysplasia		6 Cases
306644	Complication after organ transplantation	9.0 *P* *	
268316	Complication in hemodialysis	13.0 *I* *	
458758	Composite hemangioendothelioma		39 Cases
168966	Composite lymphoma	0.01 *I* *	
3216	Conductive deafness-malformed external ear syndrome		8 Cases
3236	Conductive deafness-ptosis-skeletal anomalies syndrome		3 Cases
209932	Cone dystrophy with supernormal rod response		45 Cases
1872	Cone rod dystrophy	2.5 *P* *	
221142	Confetti-like macular atrophy		2 Cases
294975	Congenital absence of upper arm and forearm with hand present	0.62 *BP*	
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral		2 Families
418	Congenital adrenal hyperplasia	6.0 *P* *	
418	Congenital adrenal hyperplasia	6.7 *BP* *	
418	Congenital adrenal hyperplasia	13.35 *I* *	
90791	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency		68 Cases
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	0.47 *P* *	
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	0.75 *BP* *	
90793	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	0.1 *P* *	
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	0.75 *BP* *	
495879	Congenital agenesis of the scrotum		6 Cases
79	Congenital alpha2-antiplasmin deficiency		40 Cases
210122	Congenital alveolar capillary dysplasia		40 Cases
3319	Congenital amegakaryocytic thrombocytopenia		100 Cases
86816	Congenital analbuminemia		50 Cases
1195	Congenital atransferrinemia		16 Cases
48	Congenital bilateral absence of vas deferens	50.0 *P* *	

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79302	Congenital bile acid synthesis defect type 3		2 Cases
79095	Congenital bile acid synthesis defect type 4		5 Cases
71278	Congenital brain dysgenesis due to glutamine synthetase deficiency		3 Cases
2040	Congenital bronchobiliary fistula		35 Cases
300313	Congenital cataract-hearing loss-severe developmental delay syndrome		5 Cases
1369	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome		40 Cases
464738	Congenital cataract-microcephaly-nevus flammeus simplex-severe intellectual disability syndrome		7 Cases
330054	Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome		3 Cases
48431	Congenital cataracts-facial dysmorphism-neuropathy syndrome		170 Cases
329242	Congenital chronic diarrhea with protein-losing enteropathy		2 Cases
168612	Congenital deficiency in alpha-fetoprotein		22 Cases
2140	Congenital diaphragmatic hernia	30.0 *BP*	
137	Congenital disorder of glycosylation	1.5 *BP* *	
85	Congenital dyserythropoietic anemia	0.16 *BP* *	
98870	Congenital dyserythropoietic anemia type III		60 Cases
293825	Congenital dyserythropoietic anemia type IV		4 Cases
103910	Congenital enterocyte heparan sulfate deficiency		3 Cases
231573	Congenital erosive and vesicular dermatosis		31 Cases
79277	Congenital erythropoietic porphyria	0.065 *I* *	
325	Congenital factor II deficiency	0.05 *P* *	
326	Congenital factor V deficiency	0.1 *P* *	
327	Congenital factor VII deficiency	0.33 *P* *	
329	Congenital factor XI deficiency	0.1 *P* *	
331	Congenital factor XIII deficiency	0.05 *P* *	
331	Congenital factor XIII deficiency	0.04 *I* *	
335	Congenital fibrinogen deficiency	0.15 *P* *	
476406	Congenital generalized hypercontractile muscle stiffness syndrome		2 Cases
1023	Congenital generalized hypertrichosis, Ambras type		40 Cases
98976	Congenital glaucoma	3.6 *BP* *	
60041	Congenital heart block	4.54 *BP*	
1355	Congenital heart defect-round face-developmental delay syndrome		3 Cases
98975	Congenital hereditary endothelial dystrophy type I		68 Cases
306530	Congenital hereditary facial paralysis-variable hearing loss syndrome		13 Cases
2185	Congenital hydrocephalus	46.5 *BP* *	
442	Congenital hypothyroidism	38.0 *BP* *	
95711	Congenital hypothyroidism due to developmental anomaly	21.3 *P* *	
95715	Congenital hypothyroidism due to transplacental passage of maternal TSH-binding inhibitory antibodies	1.0 *P* *	
352333	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome		2 Cases
2271	Congenital ichthyosis-microcephalus-tetraplegia syndrome		2 Cases
217399	Congenital insensitivity to pain with hyperhidrosis		2 Cases
453510	Congenital insensitivity to pain with severe intellectual disability		3 Cases
1229	Congenital intrauterine infection-like syndrome		30 Cases
332	Congenital intrinsic factor deficiency		100 Cases
657	Congenital isolated hyperinsulinism	2.0 *BP*	
495875	Congenital labioscrotal agenesis-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome		3 Cases
1954	Congenital lethal erythroderma		17 Cases
210163	Congenital lethal myopathy, Compton-North type		4 Cases
1928	Congenital lobar emphysema	4.0 *BP*	
93109	Congenital megacalycosis		25 Cases
69063	Congenital membranous nephropathy due to maternal anti-neutral endopeptidase alloimmunization		15 Cases
391376	Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome		20 Cases
157973	Congenital muscular dystrophy due to LMNA mutation		23 Cases
258	Congenital muscular dystrophy type 1A	0.3 *P* *	
98893	Congenital muscular dystrophy type 1B		6 Cases

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371007	Congenital muscular dystrophy with hyperlaxity		14 Cases
34520	Congenital muscular dystrophy with integrin alpha-7 deficiency	0.03 *P* *	
329178	Congenital muscular dystrophy with intellectual disability and severe epilepsy		3 Cases
1875	Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome		7 Cases
486815	Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome		4 Cases
590	Congenital myasthenic syndrome	0.3 *P* *	
319160	Congenital myopathy with internal nuclei and atypical cores		5 Cases
424107	Congenital myopathy with myasthenic-like onset		2 Cases
199329	Congenital myopathy, Paradas type		2 Cases
168486	Congenital neuronal ceroid lipofuscinosis		10 Cases
369852	Congenital neutropenia-myelofibrosis-nephromegaly syndrome		16 Cases
79394	Congenital non-bullous ichthyosiform erythroderma	0.3 *P* *	
2772	Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome		3 Cases
313906	Congenital pancreatic cyst		10 Cases
139414	Congenital panfollicular nevus		3 Cases
66630	Congenital pseudoarthrosis of the clavicle		200 Cases
2444	Congenital pulmonary airway malformation	8.2 *BP* *	
2414	Congenital pulmonary lymphangiectasia		100 Cases
3189	Congenital pulmonary valve stenosis	39.3 *BP* *	
3269	Congenital radioulnar synostosis		350 Cases
281190	Congenital reticular ichthyosiform erythroderma		40 Cases
290	Congenital rubella syndrome	0.35 *BP* *	
290	Congenital rubella syndrome	0.03 *I* *	
2301	Congenital short bowel syndrome		41 Cases
369861	Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome		16 Cases
103908	Congenital sodium diarrhea		15 Cases
101068	Congenital stromal corneal dystrophy		6 Families
35122	Congenital sucrase-isomaltase deficiency	20.0 *P* *	
99125	Congenital total pulmonary venous return anomaly	9.0 *BP*	
858	Congenital toxoplasmosis	33.0 *BP* *	
92050	Congenital tufting enteropathy	0.5 *BP* *	
291	Congenital varicella syndrome		130 Cases
216694	Congenitally corrected transposition of the great arteries	3.0 *BP*	
2391	Congenitally short costocoracoid ligament		1 Family
860	Congenitally uncorrected transposition of the great arteries	24.25 *BP* *	
300284	Connective tissue disorder due to lysyl hydroxylase-3 deficiency		2 Cases
420794	Cono-spondylar dysplasia		3 Cases
319651	Constitutional megaloblastic anemia with severe neurologic disease		6 Cases
436003	Contractures-developmental delay-Pierre Robin syndrome		6 Cases
1484	Contractures-ectodermal dysplasia-cleft lip/palate syndrome		2 Cases
314002	Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome		2 Cases
1487	Cooks syndrome		12 Cases
1488	Cooper-Jabs syndrome		2 Cases
1490	Corneal dystrophy-perceptive deafness syndrome		24 Cases
352662	Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome		2 Cases
3177	Corneal-cerebellar syndrome		2 Cases
199	Cornelia de Lange syndrome	1.9 *P* *	
199	Cornelia de Lange syndrome	1.3 *BP* *	
3194	Corneodermatoosseous syndrome		7 Cases
52055	Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome		2 Cases
459074	Corpus callosum agenesis-macrocephaly-hypertelorism syndrome		4 Cases
1389	Cortical blindness-intellectual disability-polydactyly syndrome		3 Cases
300570	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation		12 Cases
54251	Corticosteroid-sensitive aseptic abscess syndrome		49 Cases
3071	Costello syndrome		300 Cases
201	Cowden syndrome	0.5 *P* *	
1508	Coxoauricular syndrome		4 Cases

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1509	Coxopodopatellar syndrome		47 Cases
1512	Crane-Heise syndrome		9 Cases
1525	Cranio-osteoarthropathy		30 Cases
1513	Craniodiaphyseal dysplasia		20 Cases
1514	Craniodigital-intellectual disability syndrome		5 Cases
1515	Cranioectodermal dysplasia		39 Cases
85168	Craniofacial conodysplasia		1 Family
314555	Craniofacial dysplasia-osteopenia syndrome		5 Cases
459061	Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome		8 Cases
1516	Craniofacial dyssynostosis		14 Cases
1529	Craniofacial-deafness-hand syndrome		3 Cases
363705	Craniofaciofrontodigital syndrome		4 Cases
1521	Craniofrontonasal dysplasia-Poland anomaly syndrome		2 Cases
50814	Craniolenticulosutural dysplasia		28 Cases
85184	Craniometadiaphyseal dysplasia, wormian bone type		4 Cases
1522	Craniometaphyseal dysplasia		160 Cases
54595	Craniopharyngioma	2.0 *P* *	
54595	Craniopharyngioma	1.0 *I*	
157832	Craniorhiny		6 Cases
1531	Craniosynostosis	24.3 *BP* *	
1541	Craniosynostosis, Boston type		3 Families
2145	Craniosynostosis, Herrmann-Opitz type		2 Cases
1527	Craniosynostosis, Philadelphia type		1 Family
85199	Craniosynostosis-anal anomalies-porokeratosis syndrome		9 Cases
1538	Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome		4 Cases
1533	Craniosynostosis-fibular aplasia syndrome		2 Cases
171839	Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome		2 Cases
52054	Craniosynostosis-intracranial calcifications syndrome		3 Cases
1528	Craniotelencephalic dysplasia		4 Cases
90290	CREST syndrome	8.0 *P* *	
205	Crigler-Najjar syndrome	1.0 *P* *	
205	Crigler-Najjar syndrome	0.1 *BP* *	
1545	Crisponi syndrome		30 Cases
1461	Criss-cross heart	0.8 *BP* *	
2930	Cronkhite-Canada syndrome		500 Cases
2935	Crossed polysyndactyly		12 Cases
207	Crouzon disease	0.9 *BP* *	
93262	Crouzon syndrome-acanthosis nigricans syndrome	0.1 *BP*	
1546	Cryptococcosis	11.0 *I* *	
468635	Cryptogenic multifocal ulcerous stenosing enteritis		60 Cases
1547	Cryptomicrotia-brachydactyly-excess fingertip arch syndrome		2 Cases
1548	Cryptorchidism-arachnodactyly-intellectual disability syndrome		3 Cases
1549	Cryptosporidiosis	1.96 *I* *	
357329	Cryptosporidiosis-chronic cholangitis-liver disease syndrome		5 Cases
307766	Curly hair-acral keratoderma-caries syndrome		14 Cases
1552	Currarino syndrome	1.0 *P* *	
1553	Curry-Jones syndrome		9 Cases
96253	Cushing disease	4.0 *P* *	
96253	Cushing disease	0.2 *I* *	
553	Cushing syndrome	5.9 *P*	
553	Cushing syndrome	0.15 *I* *	
189427	Cushing syndrome due to macronodular adrenal hyperplasia	0.08 *P* *	
280779	Cutaneous collagenous vasculopathy		20 Cases
79140	Cutaneous neuroendocrine carcinoma	4.0 *P* *	
79140	Cutaneous neuroendocrine carcinoma	0.27 *I*	
2881	Cutaneous photosensitivity-lethal colitis syndrome		3 Cases
451607	Cutaneous pseudolymphoma		60 Cases
1555	Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome		12 Cases
209	Cutis laxa	0.1 *BP* *	
221145	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies		21 Cases
171719	Cutis laxa-Marfanoid syndrome		18 Cases
1556	Cutis marmorata telangiectatica congenita		300 Cases
2686	Cyclic neutropenia	0.1 *P* *	
2674	Cyprus facial-neuromusculoskeletal syndrome		1 Family
400	Cystic echinococcosis	1.0 *I* *	
586	Cystic fibrosis	7.4 *P* *	
2575	Cystic fibrosis-gastritis-megaloblastic anemia syndrome		2 Cases
2111	Cystic hamartoma of lung and kidney		3 Cases

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85136	Cystic leukoencephalopathy without megalencephaly		50 Cases
213	Cystinosis	1.5 *P* *	
213	Cystinosis	0.75 *BP*	
214	Cystinuria	14.0 *P*	
75381	Cystoid macular dystrophy		97 Cases
95702	Cytomegalic congenital adrenal hypoplasia	8.0 *BP*	
137698	Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	25.5 *P* *	
94087	Cytophagic histiocytic panniculitis		100 Cases
477787	Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder		2 Cases
137678	Czech dysplasia, metatarsal type		20 Cases
2437	Czeizel-Losonci syndrome		3 Cases
356978	D,L-2-hydroxyglutaric aciduria		13 Cases
79315	D-2-hydroxyglutaric aciduria		80 Cases
1562	Dacryocystitis-osteopoikilosis syndrome		5 Cases
1563	Dahlberg-Borer-Newcomer syndrome		2 Cases
1566	Dandy-Walker malformation-postaxial polydactyly syndrome		5 Cases
218	Darier disease	3.4 *P* *	
300536	DDOST-CDG		1 Case
2962	De Barsy syndrome		40 Cases
3214	Deaf blind hypopigmentation syndrome, Yemenite type		2 Cases
90024	Deafness with labyrinthine aplasia, microtia, and microdontia		56 Cases
3241	Deafness-craniofacial syndrome		2 Cases
3232	Deafness-ear malformation-facial palsy syndrome		4 Cases
3220	Deafness-enamel hypoplasia-nail defects syndrome		15 Families
254898	Deafness-encephaloneuropathy-obesity-valvulopathy syndrome		2 Cases
3218	Deafness-epiphyseal dysplasia-short stature syndrome		2 Cases
3224	Deafness-genital anomalies-metacarpal and metatarsal synostosis syndrome		2 Cases
90646	Deafness-hypogonadism syndrome		5 Cases
94064	Deafness-infertility syndrome		3 Families
85321	Deafness-intellectual disability syndrome, Martin-Probst type		3 Cases
3226	Deafness-lymphedema-leukemia syndrome		20 Cases
3230	Deafness-oligodontia syndrome		5 Cases
3231	Deafness-onychodystrophy syndrome		50 Cases
3217	Deafness-small bowel diverticulosis-neuropathy syndrome		5 Cases
3239	Deafness-vitiligo-achalasia syndrome		2 Cases
99970	Dedifferentiated liposarcoma	0.27 *I* *	
293978	Deficiency in anterior pituitary function-variable immunodeficiency syndrome		7 Cases
3202	Dehydrated hereditary stomatocytosis		20 Families
3034	Delayed membranous cranial ossification		2 Families
3038	Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome		6 Cases
1627	Deletion 5q35		10 Cases
79134	DEND syndrome		40 Cases
99828	Dengue fever	714.0 *I*	
93571	Dense deposit disease	0.25 *P*	
1652	Dent disease		250 Families
99789	Dentin dysplasia type I	1.0 *P* *	
99791	Dentin dysplasia type II		19 Families
99792	Dentin dysplasia-sclerotic bones syndrome		1 Family
49042	Dentinogenesis imperfecta	14.5 *P* *	
166260	Dentinogenesis imperfecta type 2	14.6 *P* *	
71267	Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome		2 Cases
220	Denys-Drash syndrome		200 Cases
1656	Dermatitis herpetiformis	27.0 *P* *	
31112	Dermatofibrosarcoma protuberans	10.0 *P* *	
1659	Dermatoleukodystrophy		2 Cases
221	Dermatomyositis	6.0 *P* *	
221	Dermatomyositis	0.55 *I* *	
1657	Dermatoosteolysis, Kirghizian type		5 Cases
86920	Dermatopathia pigmentosa reticularis		20 Cases
79149	Dermochondrocorneal dystrophy		15 Cases
1660	Dermoodontodysplasia		14 Cases
1425	Desbuquois syndrome		50 Cases
84132	Desmin-related myopathy with Mallory body-like inclusions		5 Cases
873	Desmoid tumor	0.3 *I* *	

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83469	Desmoplastic small round cell tumor		300 Cases
251863	Desmoplastic/nodular medulloblastoma	0.01 *I* *	
35107	Desmosterolosis		9 Cases
313892	Developmental and speech delay due to SOX5 deficiency		12 Cases
329195	Developmental delay with autism spectrum disorder and gait instability		22 Cases
163988	Developmental delay-deafness syndrome, Hildebrand type		1 Family
79107	Developmental malformations-deafness-dystonia syndrome		2 Cases
275523	Dianzani autoimmune lymphoproliferative disease		30 Cases
494444	DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome		8 Cases
66637	Diaphanospondylodysostosis		18 Cases
2141	Diaphragmatic defect-limb deficiency-skull defect syndrome		7 Cases
628	Diastrophic dwarfism	1.2 *P* *	
628	Diastrophic dwarfism	0.3 *BP* *	
370046	Didymosis aplasticosebacea		18 Cases
146	Differentiated thyroid carcinoma	5.25 *I*	
90060	Diffuse alveolar hemorrhage	1.0 *P* *	
404437	Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome		4 Cases
79456	Diffuse cutaneous mastocytosis		30 Cases
544	Diffuse large B-cell lymphoma	16.0 *P* *	
544	Diffuse large B-cell lymphoma	2.79 *I* *	
2123	Diffuse neonatal hemangiomatosis		70 Cases
86918	Diffuse palmoplantar keratoderma-acrocyanosis syndrome		10 Cases
352487	Digital anomalies-intellectual disability-short stature syndrome		8 Cases
2926	Digital extensor muscle aplasia-polyneuropathy		3 Cases
1146	Digitotalar dysmorphism	10.0 *P*	
226	Dihydropteridine reductase deficiency		150 Cases
2229	Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome		20 Families
243343	Dimethylglycine dehydrogenase deficiency		1 Case
227	Diphallia	0.02 *BP*	
1681	Diprosopus		33 Cases
2412	Dislocation of the hip-dysmorphism syndrome		4 Cases
79168	Disorder of bile acid synthesis	0.6 *P* *	
2983	Disorder of sex development-intellectual disability syndrome		3 Cases
71274	Disseminated peritoneal leiomyomatosis		150 Cases
254351	Distal 7q11.23 microdeletion syndrome		41 Cases
261102	Distal 7q11.23 microduplication syndrome		5 Cases
319171	Distal 17p13.1 microdeletion syndrome		16 Cases
261257	Distal 17p13.3 microdeletion syndrome		16 Cases
399096	Distal anoctaminopathy		8 Cases
329457	Distal arthrogryposis type 5D		37 Cases
251515	Distal arthrogryposis type 10		53 Cases
139525	Distal hereditary motor neuropathy type 2		4 Families
139552	Distal hereditary motor neuropathy, Jerash type		30 Cases
1307	Distal limb deficiencies-micrognathia syndrome		6 Cases
1620	Distal monosomy 3p		34 Cases
96125	Distal monosomy 6p		35 Cases
1642	Distal monosomy 9p		89 Cases
96148	Distal monosomy 10q		40 Cases
280325	Distal monosomy 12p		8 Cases
1590	Distal monosomy 13q		150 Cases
1596	Distal monosomy 15q		30 Cases
178400	Distal myopathy with anterior tibial onset		4 Cases
34521	Distal myopathy with early respiratory muscle involvement		24 Cases
63273	Distal myopathy with posterior leg and anterior hand involvement		16 Cases
488650	Distal myopathy, Tateyama type		7 Cases
399103	Distal nebulin myopathy		4 Families
139547	Distal spinal muscular atrophy type 3		28 Cases
3248	Distal symphalangism		8 Families
314588	Distal tetrasomy 15q		23 Cases
1745	Distal trisomy 6p		40 Cases
96102	Distal trisomy 10q		40 Cases
293939	Distal Xq28 microduplication syndrome		9 Cases
404546	DITRA		70 Cases
91131	DK1-CDG		17 Cases
443950	DNAJB2-related Charcot-Marie-Tooth disease type 2		2 Cases

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3262	Dobrow syndrome		2 Cases
447737	DOCK2 deficiency		5 Cases
158676	Dominant dystrophic epidermolysis bullosa, nails only		10 Families
244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis		6 Cases
2143	Donnai-Barrow syndrome		50 Cases
79500	DOORS syndrome		50 Cases
255	Dopa-responsive dystonia	0.5 *P*	
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency		43 Cases
230	Dopamine beta-hydroxylase deficiency		21 Cases
3427	Double outlet left ventricle	0.5 *BP*	
3411	Double uterus-hemivagina-renal agenesis syndrome		60 Cases
870	Down syndrome	95.0 *BP*	
86309	DPAGT1-CDG		18 Cases
79322	DPM1-CDG		9 Cases
263494	DPM3-CDG		1 Case
33069	Dravet syndrome	2.5 *BP*	
50817	Duane anomaly-myopathy-scoliosis syndrome		2 Cases
233	Duane retraction syndrome	10.0 *P* *	
235	Dubowitz syndrome	0.2 *BP* *	
98896	Duchenne muscular dystrophy	4.78 *P*	
98896	Duchenne muscular dystrophy	15.1 *BP* *	
1203	Duodenal atresia	9.0 *P* *	
1203	Duodenal atresia	9.0 *BP* *	
314621	Duplication of the pituitary gland		38 Cases
237	Duplication of urethra		300 Cases
239	Dyggve-Melchior-Clausen disease		60 Cases
464306	DYRK1A-related intellectual disability syndrome		54 Cases
268261	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion		19 Cases
1765	Dyschondrosteosis-nephritis syndrome		1 Family
41	Dyschromatosis symmetrica hereditaria		300 Cases
1766	Dysequilibrium syndrome		51 Cases
1775	Dyskeratosis congenita	0.1 *P* *	
2104	Dysmorphism-pectus carinatum-joint laxity syndrome		2 Cases
2282	Dysmorphism-short stature-deafness-disorder of sex development syndrome		2 Cases
1822	Dysplasia epiphysealis hemimelica	0.1 *I*	
2204	Dysplastic cortical hyperostosis		2 Cases
2476	Dysraphism-cleft lip/palate-limb reduction defects syndrome		3 Cases
1804	Dyssegmental dysplasia-glaucoma syndrome		2 Cases
85198	Dysspondyloenchondromatosis		16 Cases
210571	Dystonia 16		12 Cases
412217	Dystonia-aphonia syndrome		2 Cases
303	Dystrophic epidermolysis bullosa	0.7 *P*	
89843	Dystrophic epidermolysis bullosa pruriginosa		100 Families
2554	Ear-patella-short stature syndrome		67 Cases
1935	Early myoclonic encephalopathy		80 Cases
488635	Early-onset epilepsy-intellectual disability-brain anomalies syndrome		5 Cases
411986	Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome		3 Cases
494348	Early-onset familial noncirrhotic portal hypertension		3 Cases
256	Early-onset generalized limb-onset dystonia	0.4 *P* *	
324290	Early-onset Lafora body disease		3 Cases
439212	Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome		15 Cases
2379	Early-onset parkinsonism-intellectual disability syndrome		2 Families
496641	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome		22 Cases
500144	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome		3 Cases
496756	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome		6 Cases
352654	Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome		6 Cases
505237	Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome		12 Cases
313772	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome		2 Cases
98890	Early-onset X-linked optic atrophy		4 Families

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199343	EAST syndrome		26 Cases
391320	East Texas bleeding disorder		19 Cases
319218	Ebola hemorrhagic fever		28220 Cases
1880	Ebstein malformation	1.25 *P* *	
1880	Ebstein malformation	3.5 *BP* *	
1818	Ectodermal dysplasia, trichoodontoonychial type		7 Cases
1806	Ectodermal dysplasia-blindness syndrome		2 Cases
247827	Ectodermal dysplasia-cutaneous syndactyly syndrome		4 Cases
1883	Ectodermal dysplasia-sensorineural deafness syndrome		2 Cases
247820	Ectodermal dysplasia-syndactyly syndrome		6 Cases
448270	Ectopia cordis	0.67 *BP*	
1884	Ectopia lentis-chorioretinal dystrophy-myopia syndrome		4 Cases
1892	Ectrodactyly-polydactyly syndrome		1 Family
1894	Ectrodactyly-spina bifida-cardiopathy syndrome		1 Case
293936	EDICT syndrome		4 Families
1895	Edinburgh malformation syndrome		2 Families
1896	EEC syndrome	1.11 *BP* *	
1897	EEM syndrome		7 Families
98249	Ehlers-Danlos syndrome	0.9 *BP* *	
230839	Ehlers-Danlos syndrome due to tenascin-X deficiency		17 Cases
90309	Ehlers-Danlos syndrome type 1	5.0 *P* *	
230851	Ehlers-Danlos syndrome, cardiac valvular type		6 Cases
287	Ehlers-Danlos syndrome, classic type	5.0 *P*	
1901	Ehlers-Danlos syndrome, dermatosparaxis type		15 Cases
75501	Ehlers-Danlos syndrome, fibronectinemic type		1 Family
285	Ehlers-Danlos syndrome, hypermobility type	12.5 *P* *	
300179	Ehlers-Danlos syndrome, kyphoscoliotic and deafness type		9 Cases
1900	Ehlers-Danlos syndrome, kyphoscoliotic type	1.0 *BP*	
2953	Ehlers-Danlos syndrome, musculocontractural type		22 Cases
75392	Ehlers-Danlos syndrome, periodontitis type		62 Cases
157965	Ehlers-Danlos syndrome, spondylocheirodysplastic type		6 Cases
286	Ehlers-Danlos syndrome, vascular type	1.0 *P* *	
230845	Ehlers-Danlos syndrome, vascular-like type		3 Cases
1902	Ehrlichiosis		50 Cases
79106	Eiken syndrome		6 Cases
228240	Elastoderma		5 Cases
289	Ellis Van Creveld syndrome	1.1 *BP*	
96170	Emanuel syndrome		350 Cases
180226	Embryonal carcinoma	0.01 *I* *	
251852	Embryonal tumor of neuroepithelial tissue	0.22 *I* *	
261	Emery-Dreifuss muscular dystrophy	0.3 *P* *	
1927	Emery-Nelson syndrome		2 Cases
485418	EMILIN-1-related connective tissue disease		3 Cases
2396	Encephalocraniocutaneous lipomatosis		77 Cases
79155	Encephalopathy due to hydroxykynureninuria		30 Cases
139406	Encephalopathy due to prosaposin deficiency		10 Cases
833	Encephalopathy due to sulfite oxidase deficiency		100 Cases
319678	Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome		1 Case
296	Enchondromatosis	1.0 *P* *	
199332	Endocrine-cerebro-osteodysplasia syndrome		7 Cases
454723	Endometrioid carcinoma of ovary	0.81 *I* *	
2790	Endosteal hyperostosis, Worth type		6 Families
85186	Endosteal sclerosis-cerebellar hypoplasia syndrome		4 Cases
1937	Eng-Strom syndrome		2 Cases
60015	Enlarged parietal foramina	3.7 *P*	
83620	Enteric anendocrinosis		7 Cases
85438	Enthesitis-related juvenile idiopathic arthritis	5.7 *P* *	
449566	Eosinophilic angiocentric fibrosis		52 Cases
402035	Eosinophilic colitis		196 Cases
73247	Eosinophilic esophagitis	40.08 *P*	
3165	Eosinophilic fasciitis		200 Cases
2070	Eosinophilic gastroenteritis		280 Cases
183	Eosinophilic granulomatosis with polyangiitis	1.5 *P*	
183	Eosinophilic granulomatosis with polyangiitis	0.18 *I* *	
301	Ependymal tumor	0.2 *I* *	
251636	Ependymoma	0.16 *I* *	

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231742	Epibulbar lipodermoid-preauricular appendage-polythelia syndrome		1 Family
35125	Epidermal nevus syndrome		400 Cases
302	Epidermodysplasia verruciformis		200 Cases
304	Epidermolysis bullosa simplex	1.8 *P*	
304	Epidermolysis bullosa simplex	2.2 *BP* *	
412181	Epidermolysis bullosa simplex due to BP230 deficiency		2 Cases
412189	Epidermolysis bullosa simplex due to exophilin 5 deficiency		3 Cases
257	Epidermolysis bullosa simplex with muscular dystrophy		40 Cases
89838	Epidermolysis bullosa simplex, autosomal recessive K14		19 Cases
79401	Epidermolysis bullosa simplex, Ogna type		6 Families
141077	Epignathus	1.68 *BP*	
1948	Epilepsy-microcephaly-skeletal dysplasia syndrome		2 Cases
1951	Epilepsy-telangiectasia syndrome		6 Cases
1819	Epimetaphyseal skeletal dysplasia		4 Cases
1825	Epiphyseal dysplasia-hearing loss-dysmorphism syndrome		2 Cases
79135	Episodic ataxia type 3		1 Family
79136	Episodic ataxia type 4		2 Families
211067	Episodic ataxia type 5		7 Cases
209967	Episodic ataxia type 6		4 Cases
209970	Episodic ataxia type 7		7 Cases
401953	Episodic ataxia with slurred speech		13 Cases
93928	Epispadias	2.4 *BP* *	
293381	Epithelial recurrent erosion dystrophy		186 Cases
313920	Epstein-Barr virus-associated gastric carcinoma	1.2 *I*	
35687	Erdheim-Chester disease		500 Cases
999	Ermine phenotype		6 Cases
317	Erythrokeratodermia variabilis		200 Cases
476096	Erythrokeratodermia-cardiomyopathy syndrome		3 Cases
1199	Esophageal atresia	24.3 *BP* *	
3318	Essential thrombocythemia	0.48 *I* *	
1957	Esthesioneuroblastoma	0.02 *I* *	
785	Estrogen resistance syndrome		2 Cases
51188	Ethylmalonic encephalopathy		40 Cases
1959	Evans syndrome	0.1 *P* *	
496751	EVEN-plus syndrome		3 Cases
319	Ewing sarcoma	0.13 *I* *	
1962	Exostoses-anetodermia-brachydactyly type E syndrome		1 Family
3294	Extensor tendons of finger anomalies		2 Cases
3023	External auditory canal atresia-vertical talus-hypertelorism syndrome		10 Cases
363579	Extragonadal germ cell tumor	0.13 *I* *	
209916	Extraskeletal myxoid chondrosarcoma	0.2 *P* *	
1964	Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome		2 Cases
2725	Eye defects-arachnodactyly-cardiopathy syndrome		6 Cases
3172	Eyebrow duplication-syndactyly syndrome		3 Cases
324	Fabry disease	0.22 *BP* *	
1969	Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome		3 Cases
284169	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion		12 Cases
466950	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation		10 Cases
352712	Facial dysmorphism-immunodeficiency-livedo-short stature syndrome		11 Cases
412022	Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome		4 Families
1970	Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome		3 Cases
1778	Facial dysmorphism-shawl scrotum-joint laxity syndrome		2 Cases
85162	Facial onset sensory and motor neuronopathy		38 Cases
1973	Faciocardiorenal syndrome		4 Cases
269	Facioscapulohumeral dystrophy	4.5 *P* *	
306550	FADD-related immunodeficiency		4 Cases
3304	Fallot complex-intellectual disability-growth delay syndrome		5 Cases
88619	Familial acute necrotizing encephalopathy		11 Cases
733	Familial adenomatous polyposis	6.0 *P* *	
95700	Familial adrenal hypoplasia with absent pituitary luteinizing hormone		3 Cases
280397	Familial Alzheimer-like prion disease		2 Cases
228277	Familial anetoderma		12 Families

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615	Familial atrial myxoma		17 Families
436242	Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease		7 Cases
1551	Familial benign copper deficiency		1 Family
1416	Familial calcium pyrophosphate deposition		100 Families
1768	Familial caudal dysgenesis		4 Cases
464760	Familial cavitary optic disc anomaly		17 Cases
221061	Familial cerebral cavernous malformation	15.0 *P*	
481662	Familial Chilblain lupus		10 Families
1428	Familial chondromalacia patellae		5 Families
444490	Familial chylomicronemia syndrome	1.0 *P* *	
238578	Familial clubfoot due to 17q23.1q23.2 microduplication		4 Families
238722	Familial congenital mirror movements		75 Cases
451612	Familial congenital nasolacrimal duct obstruction		4 Cases
91498	Familial congenital palsy of trochlear nerve		6 Cases
319189	Familial cortical myoclonus		11 Cases
53296	Familial cutaneous collagenoma		16 Cases
313846	Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome		24 Cases
1799	Familial developmental dysphasia		6 Families
324588	Familial dyskinesia and facial myokymia		18 Cases
85110	Familial encephalopathy with neuroserpin inclusion bodies		6 Families
391392	Familial episodic pain syndrome with predominantly lower limb involvement		28 Cases
391389	Familial episodic pain syndrome with predominantly upper body involvement		21 Cases
464756	Familial gastric type 1 neuroendocrine tumor		5 Cases
361	Familial glucocorticoid deficiency		50 Cases
251274	Familial hyperaldosteronism type III		7 Families
238475	Familial hypercholanemia		23 Cases
99764	Familial hyperreninemic hypoaldosteronism type 2		5 Cases
424	Familial hyperthyroidism due to mutations in TSH receptor		28 Families
93372	Familial hypocalciuric hypercalcemia type 1	5.5 *P*	
300373	Familial infantile gigantism		3 Cases
352582	Familial infantile myoclonic epilepsy		7 Cases
154	Familial isolated dilated cardiomyopathy	17.5 *P* *	
154	Familial isolated dilated cardiomyopathy	2.91 *I* *	
99879	Familial isolated hyperparathyroidism		100 Families
2238	Familial isolated hypoparathyroidism		10 Families
2239	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland		2 Families
314777	Familial isolated pituitary adenoma		150 Cases
75249	Familial isolated restrictive cardiomyopathy	2.5 *P* *	
411788	Familial isolated trichomegaly		2 Families
79293	Familial LCAT deficiency		70 Cases
768	Familial long QT syndrome	40.0 *BP* *	
401942	Familial median cleft of the upper and lower lips		8 Cases
618	Familial melanoma	1.5 *I* *	
165805	Familial mesial temporal lobe epilepsy with febrile seizures		4 Cases
495930	Familial monosomy 7 syndrome		14 Families
338	Familial multiple fibrofolliculoma		7 Cases
922	Familial nasal acilia		8 Cases
280403	Familial omphalocele syndrome with facial dysmorphism		5 Cases
569	Familial or sporadic hemiplegic migraine	10.0 *P* *	
2769	Familial osteodysplasia, Anderson type		4 Cases
97290	Familial papillary thyroid carcinoma with renal papillary neoplasia		2 Cases
98306	Familial partial lipodystrophy	1.0 *P* *	
79084	Familial partial lipodystrophy, Köbberling type		20 Cases
34526	Familial primary hypomagnesemia		500 Cases
306516	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis		200 Cases
2196	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement		72 Cases
31043	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement		110 Cases

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34527	Familial primary hypomagnesemia with normocalciuria and normocalcemia		5 Families
871	Familial progressive cardiac conduction defect		50 Cases
280628	Familial progressive hyper- and hypopigmentation		3 Families
488197	Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome		9 Cases
1767	Familial progressive vestibulocochlear dysfunction		7 Cases
79147	Familial reactive perforating collagenosis		50 Cases
231108	Familial rhabdoid tumor		5 Families
168624	Familial scaphocephaly syndrome, McGillivray type		11 Cases
166282	Familial sick sinus syndrome		11 Cases
280406	Familial steroid-resistant nephrotic syndrome with sensorineural deafness		13 Cases
91387	Familial thoracic aortic aneurysm and aortic dissection		22 Cases
93953	Familial thyroglossal duct cyst		22 Cases
95716	Familial thyroid dyshormonogenesis	4.0 *P* *	
95716	Familial thyroid dyshormonogenesis	2.67 *I*	
84	Fanconi anemia	0.3 *P*	
84	Fanconi anemia	0.62 *BP* *	
333	Farber disease		96 Cases
166105	FASTKD2-related infantile mitochondrial encephalomyopathy		2 Cases
439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease		10 Cases
466	Fatal familial insomnia		27 Cases
280553	Fatal infantile hypertonic myofibrillar myopathy		11 Cases
168566	Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3		7 Cases
391343	Fatal post-viral neurodegenerative disorder		2 Cases
2492	FATCO syndrome		22 Cases
404451	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome		3 Cases
163703	Febrile infection-related epilepsy syndrome	1.0 *P* *	
1305	Feingold syndrome		123 Cases
391641	Feingold syndrome type 1		120 Cases
391646	Feingold syndrome type 2		7 Cases
488191	Female infertility due to oocyte meiotic arrest		16 Cases
404466	Female infertility due to zona pellucida defect		4 Cases
101039	Female restricted epilepsy with intellectual disability		5 Families
1988	Femoral-facial syndrome		62 Cases
2019	Femur-fibula-ulna complex	1.5 *BP* *	
397922	Ferro-cerebro-cutaneous syndrome		3 Cases
994	Fetal akinesia deformation sequence	0.6 *BP* *	
363409	Fetal akinesia-cerebral and retinal hemorrhage syndrome		3 Cases
1915	Fetal alcohol syndrome	1.6 *BP* *	
294	Fetal cytomegalovirus syndrome	40.0 *P* *	
465824	Fetal encasement syndrome		2 Cases
85212	Fetal Gaucher disease		50 Cases
1917	Fetal methylmercury syndrome		800 Cases
464724	Fever-associated acute infantile liver failure syndrome		11 Cases
313855	FGFR2-related bent bone dysplasia		11 Cases
477650	Fibroblastic rheumatism		30 Cases
2021	Fibrochondrogenesis		20 Cases
337	Fibrodysplasia ossificans progressiva	0.05 *P*	
84090	Fibronectin glomerulopathy		16 Families
2030	Fibrosarcoma	0.01 *I* *	
1118	Fibular aplasia-ectrodactyly syndrome		50 Cases
1757	Fibular dimelia-diplopodia syndrome		11 Cases
93323	Fibular hemimelia	2.0 *P* *	
2256	Fibulo-ulnar hypoplasia-renal anomalies syndrome		2 Cases
3255	Filippi syndrome		29 Cases
369979	Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome		2 Cases
97232	Fingerprint body myopathy		20 Cases
399086	Finnish upper limb-onset distal myopathy		7 Cases
79292	Fish-eye disease		30 Cases
1968	Flat face-microstomia-ear anomaly syndrome		2 Cases
98970	Fleck corneal dystrophy		30 Cases
2044	Floating-Harbor syndrome		87 Cases
2045	FLOTCH syndrome		6 Families
2047	Flynn-Aird syndrome		10 Cases

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2092	Focal dermal hypoplasia		300 Cases
352587	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation		7 Cases
398166	Focal facial dermal dysplasia		147 Cases
79133	Focal facial dermal dysplasia type I		81 Cases
398173	Focal facial dermal dysplasia type II		22 Cases
1807	Focal facial dermal dysplasia type III		20 Cases
398189	Focal facial dermal dysplasia type IV		21 Cases
48918	Focal myositis		115 Cases
1866	Focal, segmental or multifocal dystonia	11.7 *P* *	
1866	Focal, segmental or multifocal dystonia	2.0 *I* *	
2048	Foix-Chavany-Marie syndrome		150 Cases
300552	Follicular cholangitis and pancreatitis		5 Cases
545	Follicular lymphoma	28.0 *P* *	
545	Follicular lymphoma	2.12 *I* *	
228371	Foodborne botulism	0.1 *I* *	
3219	Fountain syndrome		8 Cases
397618	Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome		7 Families
2253	Foveal hypoplasia-presenile cataract syndrome		11 Cases
221126	Fowler syndrome		44 Cases
908	Fragile X syndrome	32.5 *P*	
908	Fragile X syndrome	2.4 *BP* *	
137834	Frank-Ter Haar syndrome		30 Cases
2052	Fraser syndrome	0.2 *BP* *	
347	Frasier syndrome		88 Cases
834	Free sialic acid storage disease		130 Cases
2053	Freeman-Sheldon syndrome		100 Cases
85335	Fried syndrome		1 Family
95	Friedreich ataxia	2.0 *P* *	
99672	Fried's tooth and nail syndrome		12 Cases
1791	Frontofacionasal dysplasia		14 Cases
1826	Frontometaphyseal dysplasia		100 Cases
250	Frontonasal dysplasia	0.7 *BP* *	
228390	Frontonasal dysplasia with alopecia and genital anomaly		5 Cases
306542	Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome		3 Cases

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282	Frontotemporal dementia	3.0 *P* *	
293848	Frontotemporal dementia, right temporal atrophy variant		200 Cases
2059	Fryns syndrome	7.0 *BP* *	
2058	Fryns-Smeets-Thiry syndrome		2 Cases
247790	FTH1-related iron overload		4 Cases
349	Fucosidosis		100 Cases
2854	Fuhrmann syndrome		11 Cases
24	Fumaric aciduria		40 Cases
506358	Gabriele de Vries syndrome		10 Cases
352	Galactosemia	2.0 *BP* *	
352	Galactosemia	2.1 *I* *	
351	Galactosialidosis		100 Cases
2065	Galloway-Mowat syndrome		60 Cases
2066	Gamma-aminobutyric acid transaminase deficiency		3 Families
33573	Gamma-glutamyl transpeptidase deficiency		7 Cases
100026	Gamma-heavy chain disease		120 Cases
2067	GAPO syndrome		38 Cases
79665	Gardner syndrome	9.1 *BP*	
314022	Gastric adenocarcinoma and proximal polyposis of the stomach		28 Cases
2069	Gastrocutaneous syndrome		24 Cases
44890	Gastrointestinal stromal tumor	13.0 *P* *	
44890	Gastrointestinal stromal tumor	1.0 *I*	
2368	Gastroschisis	23.7 *BP* *	
355	Gaucher disease	1.0 *P* *	
355	Gaucher disease	1.3 *BP*	
355	Gaucher disease	1.7 *I* *	
77259	Gaucher disease type 1	1.0 *P* *	
77260	Gaucher disease type 2	0.01 *P* *	
77261	Gaucher disease type 3	0.05 *P* *	
2072	Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome		10 Cases
438274	GCGR-related hyperglucagonemia		8 Cases
2623	Geleophysic dysplasia		27 Cases
2074	Gemignani syndrome		2 Cases
228429	Generalized congenital lipodystrophy with myopathy		22 Cases
411777	Generalized eruptive keratoacanthoma		40 Cases
98497	Genetic peripheral neuropathy	40.0 *P*	
2075	Genitopalatocardiac syndrome		15 Cases
85201	Genitopatellar syndrome		22 Cases

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93398	Genochondromatosis type 2		10 Cases
2077	German syndrome		5 Cases
2078	Geroderma osteodysplastica		50 Cases
356	Gerstmann-Straussler-Scheinker syndrome	0.0055 *I*	
643	Giant axonal neuropathy		50 Families
251579	Giant cell glioblastoma	0.02 *I* *	
2025	Gingival fibromatosis-facial dysmorphism syndrome		2 Cases
2027	Gingival fibromatosis-progressive deafness syndrome		2 Families
358	Gitelman syndrome	2.5 *P* *	
238763	Glaucoma secondary to spherophakia/ectopia lentis and megalocornea		12 Cases
2084	Glaucoma-ectopia-microspherophakia-stiff joints-short stature syndrome		3 Cases
2085	Glaucoma-sleep apnea syndrome		5 Cases
182067	Glial tumor	10.0 *P* *	
182067	Glial tumor	5.35 *I* *	
360	Glioblastoma	1.0 *P*	
360	Glioblastoma	3.0 *I*	
251582	Gliomatosis cerebri	0.01 *I* *	
251576	Gliosarcoma	0.03 *I* *	
404476	Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome		2 Cases
488613	Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome		26 Cases
73223	Global developmental delay-osteopenia-ectodermal defect syndrome		3 Cases
480898	Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome		6 Cases
141163	Glossopalatine ankylosis		30 Cases
97280	Glucagonoma	0.005 *I* *	
33574	Glutamate-cysteine ligase deficiency		10 Cases
25	Glutaryl-CoA dehydrogenase deficiency	1.0 *BP*	
32	Glutathione synthetase deficiency		70 Cases
407	Glycine encephalopathy	0.17 *P* *	
365	Glycogen storage disease due to acid maltase deficiency	0.8 *BP* *	
420429	Glycogen storage disease due to acid maltase deficiency, late-onset	1.75 *BP*	
364	Glycogen storage disease due to glucose-6-phosphatase deficiency	1.0 *BP*	
79258	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia	1.0 *BP* *	
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib		150 Cases
2088	Glycogen storage disease due to GLUT2 deficiency		200 Cases
367	Glycogen storage disease due to glycogen branching enzyme deficiency	0.1 *BP*	
2089	Glycogen storage disease due to hepatic glycogen synthase deficiency		16 Cases
34587	Glycogen storage disease due to LAMP-2 deficiency		84 Cases
264580	Glycogen storage disease due to liver phosphorylase kinase deficiency	1.0 *BP* *	
137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency		4 Cases
99849	Glycogen storage disease due to muscle beta-enolase deficiency		1 Case
371	Glycogen storage disease due to muscle phosphofructokinase deficiency		100 Cases
715	Glycogen storage disease due to muscle phosphorylase kinase deficiency		30 Cases
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency		30 Families
97234	Glycogen storage disease due to phosphoglycerate mutase deficiency		50 Cases
370	Glycogen storage disease due to phosphorylase kinase deficiency	1.0 *BP* *	
263297	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency		1 Case
354	GM1 gangliosidosis	0.75 *BP* *	
79255	GM1 gangliosidosis type 1		200 Cases
79256	GM1 gangliosidosis type 2		50 Cases
79257	GM1 gangliosidosis type 3		70 Cases
309152	GM2 gangliosidosis	5.0 *P* *	
309246	GM2 gangliosidosis, AB variant		10 Cases
2090	GMS syndrome		1 Family
602	GNE myopathy	1.0 *P*	
329984	Goblet cell carcinoma	0.025 *I*	
66629	Goldberg-Shprintzen megacolon syndrome		8 Families
374	Goldenhar syndrome	2.9 *BP* *	

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53540	Goldmann-Favre syndrome		50 Cases
1986	Gollop-Wolfgang complex		200 Cases
1532	Gómez-López-Hernández syndrome		36 Cases
169105	Good syndrome		241 Cases
65798	Goodman syndrome		3 Cases
73	Gorham-Stout disease		300 Cases
377	Gorlin syndrome	1.1 *P*	
2095	Gorlin-Chaudhry-Moss syndrome		7 Cases
39812	Graft versus host disease	3.0 *P* *	
79094	Grange syndrome		7 Cases
2097	Grant syndrome		1 Family
900	Granulomatosis with polyangiitis	9.0 *P* *	
900	Granulomatosis with polyangiitis	0.85 *I* *	
33111	Granulomatous slack skin		50 Cases
721	Gray platelet syndrome		60 Cases
293375	Grayson-Wilbrandt corneal dystrophy		1 Family
1426	Greenberg dysplasia		10 Cases
381	Griscelli disease		60 Cases
79476	Griscelli disease type 1		20 Cases
79477	Griscelli disease type 2		102 Cases
79478	Griscelli disease type 3		13 Cases
391348	Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome		2 Cases
73272	Growth delay due to insulin-like growth factor type 1 deficiency		5 Cases
3035	Growth delay-hydrocephaly-lung hypoplasia syndrome		4 Cases
391366	Growth retardation-mild developmental delay-chronic hepatitis syndrome		2 Cases
2101	Grubben-de Cock-Borghgraef syndrome		3 Cases
2102	GTP cyclohydrolase I deficiency		16 Cases
382	Guanidinoacetate methyltransferase deficiency		80 Cases
2103	Guillain-Barré syndrome	3.5 *P* *	
2103	Guillain-Barré syndrome	1.45 *I*	
2957	Guttmacher syndrome		3 Cases
414	Gyrate atrophy of choroid and retina		200 Cases
168569	H syndrome		100 Cases
99803	Haddad syndrome		60 Cases
2342	Haim-Munk syndrome		100 Cases
1408	Hair defect-photosensitivity-intellectual disability syndrome		3 Cases
2107	Hall-Riggs syndrome		8 Cases
2108	Hallermann-Streiff syndrome		150 Cases
2109	Hallermann-Streiff-like syndrome		2 Cases
2110	Hallux varus-preaxial polysyndactyly syndrome		2 Cases
93946	Hamel cerebro-palato-cardiac syndrome		4 Cases
73229	HANAC syndrome		6 Families
457	Harlequin ichthyosis		200 Cases
199282	Harlequin syndrome		100 Cases
2115	Harrod syndrome		3 Cases
2116	Hartnup disease	4.2 *P*	
2117	Hartsfield syndrome		17 Cases
99872	Hashimoto-Pritzker syndrome		50 Cases
2118	Hawkinsinuria		5 Families
3225	Hearing loss-familial salivary gland insensitivity to aldosterone syndrome		2 Cases
1338	Heart defect-tongue hamartoma-polysyndactyly syndrome		4 Cases
1354	Heart defects-limb shortening syndrome		2 Cases
1350	Heart-hand syndrome type 2		2 Families
1342	Heart-hand syndrome type 3		3 Cases
168796	Heart-hand syndrome, Slovenian type		14 Cases
2119	HEC syndrome		2 Cases
178330	Heinz body anemia		10 Cases
86813	Helicoid peripapillary chorioretinal degeneration		100 Cases
90053	Hematopoietic stem cell transplantation	0.65 *P* *	
306741	Hemidystonia-hemiatrophy syndrome		100 Cases
141148	Hemifacial myohyperplasia		12 Cases
276280	Hemihyperplasia-multiple lipomatosis syndrome		10 Cases
2130	Hemimelia	4.15 *P* *	
306669	Hemiparkinsonism-hemiatrophy syndrome		68 Cases
79230	Hemochromatosis type 2		74 Cases
225123	Hemochromatosis type 3		33 Cases
139491	Hemochromatosis type 4		200 Cases
280615	Hemoglobinopathy Toms River		10 Cases
86817	Hemolytic anemia due to adenylate kinase deficiency		7 Families

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712	Hemolytic anemia due to glucophosphate isomerase deficiency		50 Cases
90030	Hemolytic anemia due to glutathione reductase deficiency		3 Cases
766	Hemolytic anemia due to red cell pyruvate kinase deficiency	5.0 *P* *	
448	Hemophilia	7.7 *P* *	
448	Hemophilia	6.25 *I* *	
98878	Hemophilia A	4.85 *P*	
98878	Hemophilia A	11.25 *BP*	
98879	Hemophilia B	1.7 *P* *	
178396	Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation		4 Cases
340	Hemorrhagic fever-renal syndrome	37.0 *P* *	
340	Hemorrhagic fever-renal syndrome	0.65 *I* *	
324632	Hendra virus infection		7 Cases
2136	Hennekam syndrome		50 Cases
2135	Hennekam-Beemer syndrome		3 Cases
2031	Hepatic fibrosis-renal cysts-intellectual disability syndrome		4 Cases
890	Hepatic veno-occlusive disease	11.0 *P* *	
79124	Hepatic veno-occlusive disease-immunodeficiency syndrome		28 Cases
90073	Hepatitis B reinfection following liver transplantation	2.0 *P* *	
402823	Hepatitis delta	40.0 *P* *	
449	Hepatoblastoma	0.02 *I* *	
88673	Hepatocellular carcinoma	15.0 *P* *	
88673	Hepatocellular carcinoma	3.09 *I* *	
137681	Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1		2 Cases
95159	Hepatoerythropoietic porphyria		40 Cases
91378	Hereditary angioedema	1.5 *P* *	
289601	Hereditary arterial and articular multiple calcification syndrome		16 Cases
145	Hereditary breast and ovarian cancer syndrome	25.0 *P* *	
85458	Hereditary cerebral hemorrhage with amyloidosis		350 Cases
676	Hereditary chronic pancreatitis	0.43 *P* *	
98434	Hereditary combined deficiency of vitamin K-dependent clotting factors		30 Families
398088	Hereditary cryohydrocytosis with normal stomatin		53 Cases
168577	Hereditary cryohydrocytosis with reduced stomatin		3 Cases
26106	Hereditary diffuse gastric cancer	1.5 *I* *	
313808	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia		27 Cases
221043	Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome		15 Cases
90045	Hereditary folate malabsorption		30 Cases
469	Hereditary fructose intolerance	5.0 *P* *	
774	Hereditary hemorrhagic telangiectasia	20.0 *P* *	
3197	Hereditary hyperekplexia		150 Cases
163	Hereditary hyperferritinemia-cataract syndrome		64 Cases
217407	Hereditary hypotrichosis with recurrent skin vesicles		4 Cases
324381	Hereditary inclusion body myopathy type 4		17 Cases
79091	Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome		21 Cases
523	Hereditary leiomyomatosis and renal cell cancer		200 Cases
90117	Hereditary motor and sensory neuropathy, Okinawa type		120 Cases
43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency		19 Cases
1062	Hereditary neurocutaneous malformation		9 Families
640	Hereditary neuropathy with liability to pressure palsies	3.5 *P* *	
279943	Hereditary neutrophilia		16 Cases
168583	Hereditary North American Indian childhood cirrhosis		36 Cases
30	Hereditary orotic aciduria		20 Cases
79141	Hereditary painful callosities		2 Families
476102	Hereditary pediatric Behçet-like disease		13 Cases
168615	Hereditary persistence of alpha-fetoprotein		19 Families
29072	Hereditary pheochromocytoma-paraganglioma	0.3 *I*	
158025	Hereditary progressive mucinous histiocytosis		18 Cases
178464	Hereditary proximal myopathy with early respiratory failure		10 Families
221039	Hereditary sclerosing poikiloderma, Weary type		9 Cases
280598	Hereditary sensorimotor neuropathy with hyperelastic skin		4 Cases
139564	Hereditary sensory and autonomic neuropathy type 1B		2 Families
970	Hereditary sensory and autonomic neuropathy type 2		35 Cases

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314381	Hereditary sensory and autonomic neuropathy type 6		4 Cases
391397	Hereditary sensory and autonomic neuropathy type 7		3 Cases
478664	Hereditary sensory and autonomic neuropathy type 8		11 Families
139573	Hereditary sensory and autonomic neuropathy with deafness and global delay		4 Cases
139578	Hereditary sensory and autonomic neuropathy with spastic paraplegia		14 Cases
456318	Hereditary sensory neuropathy-deafness-dementia syndrome		6 Families
685	Hereditary spastic paraplegia	5.2 *P*	
84093	Hereditary thermosensitive neuropathy		1 Family
480851	Hereditary thrombocytopenia with early-onset myelofibrosis		9 Cases
3467	Hereditary xanthinuria	9.05 *I* *	
275777	Heritable pulmonary arterial hypertension	0.08 *P* *	
79430	Hermansky-Pudlak syndrome	0.15 *P*	
231531	Hermansky-Pudlak syndrome type 7		2 Cases
231537	Hermansky-Pudlak syndrome type 8		6 Cases
280663	Hermansky-Pudlak syndrome type 9		2 Cases
183678	Hermansky-Pudlak syndrome with neutropenia		40 Cases
1930	Herpes simplex virus encephalitis	0.3 *I*	
189	Hidrotic ectodermal dysplasia	1.0 *P* *	
1808	Hidrotic ectodermal dysplasia, Christianson-Fourie type		6 Cases
1809	Hidrotic ectodermal dysplasia, Halal type		4 Cases
314029	High bone mass osteogenesis imperfecta		2 Cases
363396	High myopia-sensorineural deafness syndrome		7 Cases
231080	High-grade dysplasia in patients with Barrett esophagus	36.0 *P* *	
388	Hirschsprung disease	10.9 *BP* *	
2155	Hirschsprung disease-deafness-polydactyly syndrome		2 Cases
2153	Hirschsprung disease-nail hypoplasia-dysmorphism syndrome		3 Cases
2150	Hirschsprung disease-type D brachydactyly syndrome		4 Cases
2158	Histidinuria-renal tubular defect syndrome		5 Cases
98287	Histiocytic and dendritic cell tumor	0.05 *I* *	
98293	Hodgkin lymphoma	2.4 *I* *	
93970	Holmes-Gang syndrome		3 Cases
79242	Holocarboxylase synthetase deficiency	0.5 *BP* *	
2162	Holoprosencephaly	13.4 *BP* *	
2163	Holoprosencephaly-craniosynostosis syndrome		11 Cases
2570	Holoprosencephaly-hypokinesia-congenital contractures syndrome		4 Cases
3186	Holoprosencephaly-radial heart renal anomalies syndrome		4 Cases
392	Holt-Oram syndrome	0.7 *BP* *	
2167	Holzgreve syndrome		3 Cases
2168	Homocarnosinosis		4 Cases
622	Homocystinuria without methylmalonic aciduria		73 Cases
391665	Homozygous familial hypercholesterolemia	0.1 *P*	
3322	Hoyeraal-Hreidarsson syndrome		33 Cases
391417	HSD10 disease		37 Cases
85295	HSD10 disease, atypical type		5 Cases
391457	HSD10 disease, neonatal type		3 Cases
482077	HTRA1-related autosomal dominant cerebral small vessel disease		21 Cases
228116	Hughes-Stovin syndrome		30 Cases
56970	Human prion disease	0.3 *P* *	
56970	Human prion disease	0.15 *I* *	
3265	Humero-radial synostosis		150 Cases
3266	Humero-radio-ulnar synostosis		30 Cases
94056	Humero-ulnar synostosis		5 Cases
3383	Humerus trochlea aplasia		5 Cases
97340	Hunter-McAlpine craniosynostosis		10 Cases
399	Huntington disease	2.7 *P*	
399	Huntington disease	0.38 *I*	
98934	Huntington disease-like 2		50 Families
401901	Huntington disease-like syndrome due to C9ORF72 expansions		10 Cases
93473	Hurler syndrome	0.5 *P* *	
93473	Hurler syndrome	0.7 *BP* *	
740	Hutchinson-Gilford progeria syndrome	0.005 *P*	
740	Hutchinson-Gilford progeria syndrome	0.025 *BP*	
498474	Hyaline fibromatosis syndrome		150 Cases
2177	Hydranencephaly	10.0 *BP*	
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	1.7 *P*	

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2182	Hydrocephalus with stenosis of the aqueduct of Sylvius	1.7 *BP*	
2186	Hydrocephalus-blue sclerae-nephropathy syndrome		1 Family
2180	Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome		8 Cases
2183	Hydrocephalus-obesity-hypogonadism syndrome		2 Cases
2184	Hydrocephaly-low insertion umbilicus syndrome		2 Cases
2181	Hydrocephaly-tall stature-joint laxity syndrome		2 Cases
401948	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency		4 Cases
168588	Hyperandrogenism due to cortisone reductase deficiency		11 Cases
276405	Hyperbiliverdinemia		2 Cases
209902	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency		24 Cases
83639	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency		2 Families
1032	Hyperdibasic aminoaciduria type 1		26 Cases
163985	Hyperekplexia-epilepsy syndrome		2 Cases
168956	Hypereosinophilic syndrome	1.5 *P* *	
2410	Hypergonadotropic hypogonadism-cataract syndrome		3 Cases
343	Hyperimmunoglobulinemia D with periodic fever		200 Cases
324575	Hyperinsulinism due to HNF1A deficiency		2 Cases
263458	Hyperinsulinism due to INSR deficiency		10 Cases
71212	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency		10 Cases
276556	Hyperinsulinism due to UCP2 deficiency		2 Cases
682	Hyperkalemic periodic paralysis	0.5 *P* *	
1336	Hyperkeratosis-hyperpigmentation syndrome		10 Cases
412	Hyperlipoproteinemia type 3	10.0 *P*	
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	12.0 *P* *	
3416	Hyperostosis corticalis generalisata		20 Cases
443098	Hyperostosis cranialis interna		13 Cases
99880	Hyperparathyroidism-jaw tumor syndrome		100 Cases
238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	0.2 *P*	
247262	Hyperphosphatasia-intellectual disability syndrome		24 Cases
157798	Hyperplastic polyposis syndrome	1.0 *I*	
1519	Hypertelorism, Teebi type		25 Cases
2211	Hypertelorism-hypospadias-polysyndactyly syndrome		3 Families
2213	Hypertelorism-microtia-facial clefting syndrome		9 Cases
293958	Hypertelorism-preauricular sinus-punctual pits-deafness syndrome		13 Cases
2220	Hypertrichosis cubiti		28 Cases
2222	Hypertrichosis lanuginosa congenita		100 Cases
966	Hypertrichosis-acromegaloid facial appearance syndrome		27 Cases
1517	Hypertrichotic osteochondrodysplasia, Cantu type		50 Cases
324525	Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation		3 Cases
2224	Hypertryptophanemia		12 Cases
363694	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome		4 Families
251523	Hyperzincemia and hypercalprotectinemia		5 Cases
2435	Hypo- and hypermelanotic cutaneous macules-retarded growth-intellectual disability syndrome		14 Cases
429	Hypochondroplasia	3.3 *P* *	
36412	Hypocomplementemic urticarial vasculitis		200 Cases
989	Hypoglossia-hypodactyly syndrome		47 Cases
2233	Hypogonadism-mitral valve prolapse-intellectual disability syndrome		2 Cases
2230	Hypogonadotropic hypogonadism-frontoparietal alopecia syndrome		6 Cases
2235	Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome		2 Cases
293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome		4 Cases
363523	Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome		12 Cases
238468	Hypohidrotic ectodermal dysplasia	6.7 *P* *	
98813	Hypohidrotic ectodermal dysplasia with immunodeficiency	0.2 *BP* *	

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1882	Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome		3 Cases
293964	Hypoinsulinemic hypoglycemia and body hemihypertrophy		5 Cases
681	Hypokalemic periodic paralysis	1.0 *P* *	
1790	Hypomandibular faciocranial dysostosis		3 Cases
137639	Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome		8 Cases
2680	Hypomyelination neuropathy-arthrogryposis syndrome		9 Cases
139441	Hypomyelination with atrophy of basal ganglia and cerebellum		19 Cases
363412	Hypomyelination with brain stem and spinal cord involvement and leg spasticity		13 Cases
447893	Hypomyelination-cerebellar atrophy-hypoplasia of the corpus callosum syndrome		4 Cases
85163	Hypomyelination-congenital cataract syndrome		10 Cases
88637	Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome		105 Cases
436	Hypophosphatasia	0.21 *BP* *	
324561	Hypopigmentation-punctate palmoplantar keratoderma syndrome		6 Families
722	Hypoplasminogenemia	0.2 *P* *	
2248	Hypoplastic left heart syndrome	24.0 *BP*	
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome		16 Cases
98723	Hypoplastic right heart syndrome	3.3 *BP* *	
2250	Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome		2 Cases
2261	Hypospadias-intellectual disability, Goldblatt type syndrome		3 Cases
137908	Hypotonia with lactic acidemia and hyperammonemia		4 Cases
163690	Hypotonia-cystinuria syndrome		22 Cases
79507	Hypotonia-failure to thrive-microcephaly syndrome		2 Cases
55654	Hypotrichosis simplex		38 Cases
1573	Hypotrichosis with juvenile macular degeneration		50 Cases
330029	Hypotrichosis-deafness syndrome		1 Case
2266	Hypotrichosis-intellectual disability, Lopes type		2 Cases
69735	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome		4 Cases
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar keratoderma syndrome		2 Cases
254509	Iatrogenic botulism		180 Cases
2268	ICF syndrome		66 Cases
2273	Ichthyosis follicularis-alopecia-photophobia syndrome		40 Cases
79504	Ichthyosis hystrix gravior		11 Cases
79503	Ichthyosis hystrix of Curth-Macklin		10 Cases
2269	Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome		4 Cases
2274	Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome		2 Cases
91132	Ichthyosis-hypotrichosis syndrome		11 Cases
2278	Ichthyosis-intellectual disability-dwarfism-renal impairment syndrome		4 Cases
2272	Ichthyosis-oral and digital anomalies syndrome		2 Cases
88621	Ichthyosis-prematurity syndrome		16 Families
363992	Ichthyosis-short stature-brachydactyly-microspherophakia syndrome		4 Cases
930	Idiopathic achalasia	8.0 *P*	
930	Idiopathic achalasia	0.77 *I*	
724	Idiopathic acute eosinophilic pneumonia		100 Cases
139423	Idiopathic acute transverse myelitis	0.25 *I* *	
422	Idiopathic and/or familial pulmonary arterial hypertension	1.0 *P* *	
88	Idiopathic aplastic anemia	0.4 *P* *	
33208	Idiopathic hypersomnia	30.0 *P* *	
238624	Idiopathic intracranial hypertension	14.0 *P* *	
45452	Idiopathic neonatal atrial flutter	1.5 *BP* *	
494428	Idiopathic pleuroparenchymal fibroelastosis		37 Cases
275766	Idiopathic pulmonary arterial hypertension	1.1 *P* *	
2032	Idiopathic pulmonary fibrosis	11.5 *P* *	
2032	Idiopathic pulmonary fibrosis	3.81 *I* *	
99931	Idiopathic pulmonary hemosiderosis	0.0425 *I* *	
90003	IgG4-related hepatopathy		140 Cases
49041	IgG4-related retroperitoneal fibrosis	0.35 *I* *	
477661	IL21-related infantile inflammatory bowel disease		3 Cases
238621	Ileal pouch anal anastomosis related faecal incontinence	3.0 *P* *	

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85173	IMAGe syndrome		25 Cases
42062	Iminoglycinuria	6.68 *P* *	
42062	Iminoglycinuria	6.67 *BP* *	
238569	Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome		80 Cases
37042	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome		150 Cases
3002	Immune thrombocytopenic purpura	25.0 *P* *	
3002	Immune thrombocytopenic purpura	6.75 *I* *	
206569	Immune-mediated necrotizing myopathy		300 Cases
572	Immunodeficiency by defective expression of HLA class 2		179 Cases
169100	Immunodeficiency due to CD25 deficiency		2 Cases
331190	Immunodeficiency due to ficolin3 deficiency		1 Case
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency		49 Cases
331187	Immunodeficiency due to MASP-2 deficiency		1 Case
70593	Immunodeficiency due to selective anti-polysaccharide antibody deficiency		100 Cases
200418	Immunodeficiency with factor I anomaly		35 Families
2759	Imperforate oropharynx-costovertebral anomalies syndrome		2 Cases
45453	Incessant infant ventricular tachycardia	1.5 *BP* *	
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia		26 Families
611	Inclusion body myositis	0.5 *P* *	
464	Incontinentia pigmenti	1.2 *BP* *	
98848	Indolent systemic mastocytosis	3.8 *P* *	
70587	Infant acute respiratory distress syndrome	20.0 *P* *	
70587	Infant acute respiratory distress syndrome	3.5 *I* *	
178478	Infant botulism	0.2 *BP* *	
178478	Infant botulism	0.3 *I* *	
1943	Infant epilepsy with migrant focal crisis		29 Cases
313850	Infantile cerebellar-retinal degeneration		11 Cases
402364	Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly		5 Cases
1313	Infantile choroidocerebral calcification syndrome		10 Cases
199267	Infantile digital fibromatosis		200 Cases
238455	Infantile dystonia-parkinsonism		16 Cases
352563	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency		2 Cases
456312	Infantile multisystem neurologic-endocrine-pancreatic disease		2 Cases
2591	Infantile myofibromatosis	0.67 *BP* *	
35069	Infantile neuroaxonal dystrophy		150 Cases
251304	Infantile onset panniculitis with uveitis and systemic granulomatosis		4 Cases
1186	Infantile onset spinocerebellar ataxia		25 Cases
263410	Infantile spams-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome		4 Cases
3173	Infantile spasms-broad thumbs syndrome		2 Cases
293168	Infantile-onset ascending hereditary spastic paralysis		17 Families
457205	Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome		2 Cases
494526	Infantile-onset generalized dyskinesia with orofacial involvement		8 Cases
391316	Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression		3 Cases
500062	Infantile-onset periodic fever-panniculitis-dermatosis syndrome		5 Cases
1145	Infantile-onset X-linked spinal muscular atrophy		14 Families
1849	Infundibulopelvic stenosis-multicystic kidney syndrome		3 Cases
247257	Inhalational anthrax	0.1 *P* *	
254504	Inhalational botulism		10 Cases
210141	Inherited congenital spastic tetraplegia		17 Cases
79361	Inherited epidermolysis bullosa	0.8 *P* *	
79361	Inherited epidermolysis bullosa	1.9 *BP* *	
63259	Iniencephaly	50.0 *P* *	
411593	Insulin autoimmune syndrome		404 Cases
97279	Insulinoma	0.25 *I*	
464311	Intellectual disability syndrome due to a DYRK1A point mutation		35 Cases
166108	Intellectual disability, Birk-Barel type		1 Family
3079	Intellectual disability, Buenos-Aires type		5 Cases

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3080	Intellectual disability, Wolff type		2 Cases
3041	Intellectual disability-balding-patella luxation-acromicria syndrome		3 Cases
364577	Intellectual disability-brachydactyly-Pierre Robin syndrome		4 Cases
3042	Intellectual disability-cataracts-calcified pinnae-myopathy syndrome		13 Cases
171860	Intellectual disability-cataracts-kyphosis syndrome		3 Cases
397709	Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome		30 Cases
329224	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome		2 Cases
3454	Intellectual disability-developmental delay-contractures syndrome		5 Families
3044	Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome		4 Cases
468620	Intellectual disability-epilepsy-extrapyramidal syndrome		3 Cases
436151	Intellectual disability-expressive aphasia-facial dysmorphism syndrome		13 Cases
404440	Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency		7 Cases
370010	Intellectual disability-facial dysmorphism-hand anomalies syndrome		3 Cases
363611	Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome		5 Cases
369847	Intellectual disability-hyperkinetic movement-truncal ataxia syndrome		5 Cases
1495	Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome		3 Cases
314575	Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome		2 Cases
356996	Intellectual disability-hypotonia-spasticity-sleep disorder syndrome		3 Cases
457279	Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome		16 Cases
457365	Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome		3 Cases
3068	Intellectual disability-myopathy-short stature-endocrine defect syndrome		2 Cases
352530	Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome		2 Cases
397973	Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome		2 Cases
3082	Intellectual disability-polydactyly-uncombable hair syndrome		2 Cases
369837	Intellectual disability-seizures-hypotonia-ophthalmologic-skeletal anomalies syndrome		4 Cases
369950	Intellectual disability-seizures-macrocephaly-obesity syndrome		7 Cases
391372	Intellectual disability-severe speech delay-mild dysmorphism syndrome		48 Cases
3074	Intellectual disability-short stature-hypertelorism syndrome		6 Cases
3051	Intellectual disability-sparse hair-brachydactyly syndrome		61 Cases
1891	Intellectual disability-spasticity-ectrodactyly syndrome		3 Cases
363528	Intellectual disability-strabismus syndrome		34 Cases
981	Internal carotid agenesis		100 Cases
79099	Interstitial granulomatous dermatitis with arthritis		53 Cases
182095	Interstitial lung disease	5.4 *I* *	
314376	Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency		16 Cases
137622	Intractable diarrhea-choanal atresia-eye anomalies syndrome		3 Cases
436144	Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome		15 Cases
329324	Inverse Klippel-Trénaunay syndrome		15 Cases
209981	IRIDA syndrome		74 Cases
209943	IRVAN syndrome		30 Cases
84142	Isaac syndrome		150 Cases
229717	Isolated agammaglobulinemia	0.3 *P*	
1048	Isolated anencephaly/exencephaly	35.0 *BP* *	
250923	Isolated aniridia	1.31 *I* *	
2542	Isolated anophthalmia-microphthalmia syndrome	5.3 *BP* *	
557	Isolated anorectal malformation	20.0 *BP*	
3387	Isolated anterior cervical hypertrichosis		20 Cases
1134	Isolated arrhinia		20 Cases
199326	Isolated autosomal dominant hypomagnesemia, Glaudemans type		21 Cases
30391	Isolated biliary atresia	18.5 *BP*	

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2343	Isolated cloverleaf skull syndrome		120 Cases
79143	Isolated congenital anonychia		50 Cases
88620	Isolated congenital anosmia		15 Cases
91396	Isolated cryptophthalmia		30 Cases
217	Isolated Dandy-Walker malformation	2.1 *P* *	
217	Isolated Dandy-Walker malformation	1.0 *BP* *	
1885	Isolated ectopia lentis		90 Cases
448264	Isolated focal non-epidermolytic palmoplantar keratoderma		2 Cases
468666	Isolated generalized anhidrosis with normal sweat glands		7 Cases
306527	Isolated hereditary congenital facial paralysis		8 Families
2345	Isolated Klippel-Feil syndrome	2.0 *P* *	
2345	Isolated Klippel-Feil syndrome	0.6 *BP* *	
480556	Isolated neonatal sclerosing cholangitis		4 Cases
718	Isolated Pierre Robin syndrome	5.0 *BP* *	
35098	Isolated plagiocephaly	3.0 *BP*	
2924	Isolated polycystic liver disease	1.0 *P* *	
440713	Isolated sedoheptulokinase deficiency		2 Cases
823	Isolated spina bifida	18.6 *BP* *	
457083	Isolated splenogonadal fusion		145 Cases
2440	Isolated split hand-split foot malformation	5.4 *BP* *	
3208	Isolated succinate-CoQ reductase deficiency		37 Cases
454750	Isolated tracheoesophageal fistula	2.2 *BP*	
3366	Isolated trigonocephaly	6.7 *BP* *	
2306	Isotretinoin-like syndrome		6 Cases
33	Isovaleric acidemia	1.0 *P* *	
439254	ITM2B amyloidosis		2 Families
435	Ito hypomelanosis	10.85 *I* *	
457375	ITPA-related encephalopathy		7 Cases
2307	IVIC syndrome		4 Families
1540	Jackson-Weiss syndrome		200 Cases
2308	Jacobsen syndrome	1.0 *BP* *	
1873	Jalili syndrome		49 Cases
79139	Japanese encephalitis	0.65 *I* *	
313795	Jawad syndrome		4 Cases
90647	Jervell and Lange-Nielsen syndrome	0.3 *P*	
474	Jeune syndrome	1.4 *BP* *	
324999	JMP syndrome		3 Cases
2315	Johanson-Blizzard syndrome	0.4 *BP* *	
475	Joubert syndrome	1.125 *BP*	
140874	Joubert syndrome and related disorders	1.1 *BP*	
1454	Joubert syndrome with hepatic defect		8 Cases
397715	Joubert syndrome with Jeune asphyxiating thoracic dystrophy		8 Cases
2318	Joubert syndrome with oculorenal defect		17 Cases
2319	Juberg-Hayward syndrome		13 Cases
93972	Juberg-Marsidi syndrome		16 Cases
79405	Junctional epidermolysis bullosa inversa		9 Cases
306504	Junctional epidermolysis bullosa with respiratory and renal involvement		3 Cases
79404	Junctional epidermolysis bullosa, generalized severe	0.17 *BP*	
79403	Junctional epidermolysis bullosa-pyloric atresia syndrome		100 Cases
2321	Jung-Wolff-Back-Stahl syndrome		2 Cases
1941	Juvenile absence epilepsy	7.5 *I* *	
247794	Juvenile cataract-microcornea-renal glucosuria syndrome		12 Cases
93672	Juvenile dermatomyositis	0.295 *I*	
248111	Juvenile Huntington disease	0.6 *P* *	
248111	Juvenile Huntington disease	0.04 *I* *	
2028	Juvenile hyaline fibromatosis		70 Cases
92	Juvenile idiopathic arthritis	1.5 *I* *	
86834	Juvenile myelomonocytic leukemia	0.1 *P* *	
2801	Juvenile Paget disease		50 Cases
79076	Juvenile polyposis of infancy		11 Cases
2929	Juvenile polyposis syndrome	3.85 *I* *	
247604	Juvenile primary lateral sclerosis		4 Cases
26137	Juvenile temporal arteritis		20 Cases
445062	Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome		5 Cases
2322	Kabuki syndrome	3.1 *P* *	
254519	Kagami-Ogata syndrome		84 Cases
478	Kallmann syndrome	3.75 *P* *	
2326	Kallmann syndrome-heart disease syndrome		8 Cases
33276	Kaposi sarcoma	0.34 *I* *	
2328	Kapur-Toriello syndrome		6 Cases
2329	Karsch-Neugebauer syndrome		11 Cases
401996	Karyomegalic interstitial nephritis		12 Families

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2330	Kasabach-Merritt syndrome		300 Cases
2332	KBG syndrome		59 Cases
439218	KCNQ2-related epileptic encephalopathy		11 Families
480	Kearns-Sayre syndrome	2.0 *P* *	
2662	Keipert syndrome		12 Cases
2333	Kenny-Caffey syndrome		65 Cases
435628	Keppen-Lubinsky syndrome		3 Cases
494	Keratoderma hereditarium mutilans		50 Cases
79395	Keratoderma hereditarium mutilans with ichthyosis		50 Cases
2339	Keratosis follicularis-dwarfism-cerebral atrophy syndrome		6 Cases
86919	Keratosis palmaris et plantaris-clinodactyly syndrome		20 Cases
293807	Ketamine-induced biliary dilatation		2 Cases
438075	Ketoacidosis due to monocarboxylate transporter-1 deficiency		9 Cases
85202	Keutel syndrome		30 Cases
477	KID syndrome		100 Cases
50918	Kikuchi-Fujimoto disease		1052 Cases
482	Kimura disease		300 Cases
2908	Kindler syndrome		250 Cases
99741	King-Denborough syndrome		18 Cases
261494	Kleefstra syndrome		114 Cases
96147	Kleefstra syndrome due to 9q34 microdeletion		86 Cases
261652	Kleefstra syndrome due to a point mutation		23 Cases
399081	KLHL9-related early-onset distal myopathy		10 Cases
447974	Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome		2 Cases
90308	Klippel-Trénaunay syndrome	0.007 *P* *	
96169	Koolen-De Vries syndrome	4.0 *P* *	
363965	Koolen-De Vries syndrome due to a point mutation		4 Cases
99749	Kostmann syndrome		45 Cases
2351	Kousseff syndrome		8 Cases
487	Krabbe disease	1.0 *P* *	
487	Krabbe disease	0.7 *BP*	
306674	Kufor-Rakeb syndrome		16 Cases
454745	Kuru		2700 Cases
496689	Kyphoscoliosis-lateral tongue atrophy-hereditary spastic paraplegia syndrome		12 Cases
496686	Kyphosis-lateral tongue atrophy-myofibrillar myopathy syndrome		3 Cases
79314	L-2-hydroxyglutaric aciduria		140 Cases
35704	L-Arginine:glycine amidinotransferase deficiency		9 Cases
440731	L-ferritin deficiency		2 Cases
2363	Lacrimoauriculodentodigital syndrome		100 Cases
501	Lafora disease		300 Cases
306507	LAMB2-related infantile-onset nephrotic syndrome		14 Cases
1296	Lambert syndrome		4 Cases
43393	Lambert-Eaton myasthenic syndrome	0.35 *P*	
313	Lamellar ichthyosis	0.55 *P* *	
2632	Langer mesomelic dysplasia		50 Cases
389	Langerhans cell histiocytosis	1.5 *P* *	
626	Large congenital melanocytic nevus	2.75 *P* *	
633	Laron syndrome	0.3 *P* *	
220465	Laron syndrome with immunodeficiency		10 Cases
2370	Larsen-like osseous dysplasia-short stature syndrome		3 Cases
284139	Larsen-like syndrome, B3GAT3 type		14 Cases
2808	Laryngeal abductor paralysis		9 Cases
2375	Laryngeal abductor paralysis-intellectual disability syndrome		20 Cases
2004	Laryngotracheoesophageal cleft	7.5 *BP* *	
93940	Laryngotracheoesophageal cleft type 3		30 Cases
93941	Laryngotracheoesophageal cleft type 4		20 Cases
98912	Late-onset distal myopathy, Markesbery-Griggs type		11 Cases
228227	Late-onset focal dermal elastosis		5 Cases
79406	Late-onset junctional epidermolysis bullosa		37 Cases
231556	Late-onset localized junctional epidermolysis bullosa-intellectual disability syndrome		2 Cases
2789	Lateral meningocele syndrome		14 Cases
46059	Lathosterolosis		4 Cases
2378	Laurin-Sandrow syndrome		14 Cases
650	LCAT deficiency		125 Cases
330015	Lead poisoning	2.3 *P* *	
65	Leber congenital amaurosis	2.5 *P*	
65	Leber congenital amaurosis	2.5 *BP*	
104	Leber hereditary optic neuropathy	4.3 *P*	

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99718	Leber plus disease	0.04 *P* *	
549	Legionellosis	0.97 *I* *	
137605	Legius syndrome	2.2 *BP*	
506	Leigh syndrome	2.0 *P* *	
506	Leigh syndrome	2.8 *BP* *	
485421	Leigh-like basal ganglia disease-optic atrophy-peripheral neuropathy syndrome		4 Cases
507	Leishmaniasis	0.1 *P* *	
507	Leishmaniasis	25.0 *I*	
140936	Lelis syndrome		9 Cases
137839	Lemierre syndrome	10.0 *I* *	
2382	Lennox-Gastaut syndrome	15.0 *P* *	
2382	Lennox-Gastaut syndrome	0.1 *I* *	
2658	Lenz-Majewski hyperostotic dwarfism		10 Cases
548	Leprosy	3.7 *I*	
509	Leptospirosis	0.11 *I* *	
510	Lesch-Nyhan syndrome	0.34 *BP* *	
158687	Lethal acantholytic epidermolysis bullosa		4 Cases
314718	Lethal arteriopathy syndrome due to fibulin-4 deficiency		22 Cases
53696	Lethal arthrogryposis-anterior horn cell disease syndrome		15 Cases
1187	Lethal ataxia with deafness and optic atrophy		4 Families
137776	Lethal congenital contracture syndrome type 2		1 Family
137783	Lethal congenital contracture syndrome type 3		14 Cases
330050	Lethal encephalopathy due to mitochondrial and peroxisomal fission defect		1 Case
1972	Lethal faciocardiomelic dysplasia		3 Cases
444069	Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome		4 Cases
439897	Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome		2 Cases
1046	Lethal hemolytic anemia-genital anomalies syndrome		2 Cases
480528	Lethal hydranencephaly-diaphragmatic hernia syndrome		2 Cases
2347	Lethal Kniest-like dysplasia		2 Cases
2371	Lethal Larsen-like syndrome		8 Cases
478049	Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome		4 Cases
33108	Lethal multiple pterygium syndrome		28 Families
435845	Lethal neonatal spasticity-epileptic encephalopathy syndrome		8 Cases
293925	Lethal occipital encephalocele-skeletal dysplasia syndrome		5 Cases
2736	Lethal omphalocele-cleft palate syndrome		3 Cases
1832	Lethal osteosclerotic bone dysplasia		40 Cases
210144	Lethal polymalformative syndrome, Boissel type		10 Cases
1423	Lethal recessive chondrodysplasia		4 Cases
99870	Letterer-Siwe disease	0.2 *P* *	
2968	Leukocyte adhesion deficiency		350 Cases
99842	Leukocyte adhesion deficiency type I	0.1 *P* *	
99843	Leukocyte adhesion deficiency type II		7 Cases
99844	Leukocyte adhesion deficiency type III		40 Cases
139444	Leukoencephalopathy with bilateral anterior temporal lobe cysts		29 Cases
137898	Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome		127 Cases
363540	Leukoencephalopathy with mild cerebellar ataxia and white matter edema		6 Cases
163684	Leukoencephalopathy-dystonia-motor neuropathy syndrome		2 Cases
83629	Leukoencephalopathy-metaphyseal chondrodysplasia syndrome		4 Cases
2386	Leukoencephalopathy-palmoplantar keratoderma syndrome		4 Cases
314051	Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome		14 Cases
1816	Leukomelanoderma-infantilism-intellectual disability-hypodontia-hypotrichosis syndrome		4 Cases
210133	Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome		11 Cases
48162	Lewis-Sumner syndrome	0.9 *P* *	
65285	Lhermitte-Duclos disease		220 Cases
524	Li-Fraumeni syndrome	6.0 *P*	
525	Lichen planopilaris		300 Cases
254478	Lichen planus pemphigoides		100 Cases
2390	Lichtenstein syndrome		2 Cases
526	Liddle syndrome		80 Cases

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99812	LIG4 syndrome		27 Cases
97231	Ligneous conjunctivitis		200 Cases
263	Limb-girdle muscular dystrophy	2.32 *P*	
445110	Limb-girdle muscular dystrophy due to POMK deficiency		2 Cases
69085	Limb-mammary syndrome		38 Cases
171673	Limbal stem cell deficiency	3.0 *P* *	
329341	Limbic encephalitis with DPP6 antibodies		4 Cases
498700	Limbic encephalitis with neurexin-3 antibodies		5 Cases
220407	Limited systemic sclerosis		200 Cases
140933	Linear atrophoderma of Moulin		30 Cases
228236	Linear focal elastosis		30 Cases
2612	Linear nevus sebaceus syndrome	10.0 *BP* *	
435660	LIPE-related familial partial lipodystrophy		2 Cases
156156	Lipoatrophy with diabetes, leukomelanodermic papules, liver steatosis, and hypertrophic cardiomyopathy		1 Case
1979	Lipodystrophy due to peptidic growth factors deficiency		1 Family
50811	Lipodystrophy-intellectual disability-deafness syndrome		3 Cases
401859	Lipoic acid synthetase deficiency		3 Cases
530	Lipoid proteinosis		300 Cases
69078	Liposarcoma	1.0 *I* *	
401862	Lipoyl transferase 1 deficiency		4 Cases
98955	Lisch epithelial corneal dystrophy		36 Cases
171680	Lissencephaly due to TUBA1A mutation		15 Cases
86821	Lissencephaly type 3-familial fetal akinesia sequence syndrome		5 Cases
86822	Lissencephaly type 3-metacarpal bone dysplasia syndrome		2 Cases
100012	Lissencephaly with cerebellar hypoplasia type B		50 Cases
100013	Lissencephaly with cerebellar hypoplasia type C		2 Cases
533	Listeriosis	0.337 *I*	
363618	LMNA-related cardiocutaneous progeria syndrome		5 Cases
2407	LOC syndrome		50 Cases
93685	Localized Castleman disease	1.0 *P*	
251393	Localized junctional epidermolysis bullosa, non-Herlitz type		20 Cases
90398	Localized lichen myxedematosus with mixed features of different subtypes		10 Cases
90399	Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms		5 Cases
2406	Locked-in syndrome		33 Cases
60030	Loeys-Dietz syndrome		52 Families
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	8.0 *P* *	
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	1.0 *BP* *	
2408	Lowe-Kohn-Cohen syndrome		1 Family
2487	Lower limb malformation-hypospadias syndrome		2 Cases
276435	Lower motor neuron syndrome with late-adult onset		31 Cases
844	Lown-Ganong-Levine syndrome		12 Cases
2409	Lowry-MacLean syndrome		3 Cases
1824	Lowry-Wood syndrome		8 Cases
83628	LUMBAR syndrome		54 Cases
1120	Lung agenesis-heart defect-thumb anomalies syndrome		9 Cases
137631	Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome		2 Cases
90283	Lupus erythematosus tumidus		250 Cases
91546	Lyme disease	21.9 *I*	
538	Lymphangioleiomyomatosis	0.15 *P*	
538	Lymphangioleiomyomatosis	0.0135 *I*	
86915	Lymphedema-atrial septal defects-facial changes syndrome		5 Cases
86914	Lymphedema-cerebral arteriovenous anomaly syndrome		5 Cases
99141	Lymphedema-posterior choanal atresia syndrome		6 Cases
275761	Lysosomal acid lipase deficiency	2.0 *P* *	
397612	Macrocephaly-developmental delay syndrome		9 Cases
210548	Macrocephaly-intellectual disability-autism syndrome		40 Cases
466791	Macrocephaly-intellectual disability-left ventricular non compaction syndrome		6 Cases
457485	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome		8 Cases
2427	Macrocephaly-short stature-paraplegia syndrome		2 Cases
2432	Macrosomia-microphthalmia-cleft palate syndrome		5 Cases
83619	Macrostomia-preauricular tags-external ophthalmoplegia syndrome		9 Cases
220448	Macrothrombocytopenia with mitral valve insufficiency		2 Cases

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487796	Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome		2 Cases
91494	Macular coloboma-cleft palate-hallux valgus syndrome		2 Cases
137867	Madras motor neuron disease		200 Cases
163634	Maffucci syndrome		250 Cases
324972	MAGIC syndrome		21 Cases
77297	Majeed syndrome		4 Families
87503	Mal de Meleda	1.0 *P*	
420179	Malan overgrowth syndrome		20 Cases
673	Malaria	3.0 *P* *	
673	Malaria	73.0 *I*	
2234	Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome		2 Cases
99915	Maligant granulosa cell tumor of the ovary	0.12 *I* *	
679	Malignant atrophic papulosis		200 Cases
99912	Malignant dysgerminomatous germ cell tumor of the ovary	0.04 *I* *	
398934	Malignant epithelial tumor of ovary	9.39 *I* *	
276145	Malignant epithelial tumor of salivary glands	0.73 *I* *	
35807	Malignant germ cell tumor of ovary	0.08 *I* *	
168999	Malignant melanoma of the mucosa	0.26 *I* *	
293181	Malignant migrating partial seizures of infancy		114 Cases
213512	Malignant mixed Müllerian tumor of the ovary	0.12 *I* *	
398940	Malignant non-epithelial tumor of ovary	0.43 *I* *	
3148	Malignant peripheral nerve sheath tumor	1.0 *I*	
168811	Malignant peritoneal mesothelioma	1.5 *P* *	
35808	Malignant sex cord stromal tumor of ovary	1.85 *P* *	
35808	Malignant sex cord stromal tumor of ovary	0.13 *I* *	
398987	Malignant teratoma of ovary	0.07 *I* *	
252212	Malignant triton tumor		170 Cases
180242	Malignant tumor of fallopian tubes	1.0 *P* *	
398043	Malignant tumor of penis	1.075 *I* *	
943	Malonic aciduria		34 Cases
52417	MALT lymphoma	4.0 *P* *	
52417	MALT lymphoma	0.3 *I* *	
238744	Mammary-digital-nail syndrome		11 Cases
397941	MAN1B1-CDG		25 Cases
363649	Mandibular hypoplasia-deafness-progeroid syndrome		21 Cases
2457	Mandibuloacral dysplasia		40 Cases
443995	Mandibulofacial dysostosis with alopecia		4 Cases
357158	Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome		2 Cases
79113	Mandibulofacial dysostosis-microcephaly syndrome		107 Cases
52416	Mantle cell lymphoma	3.5 *P* *	
511	Maple syrup urine disease	0.67 *BP*	
99826	Marburg hemorrhagic fever		500 Cases
221074	Marchiafava-Bignami disease		250 Cases
2461	Marden-Walker syndrome		50 Cases
558	Marfan syndrome	15.0 *P*	
558	Marfan syndrome	25.0 *I* *	
2463	Marfanoid habitus-autosomal recessive intellectual disability syndrome		4 Cases
314041	Marfanoid habitus-inguinal hernia-advanced bone age syndrome		2 Cases
2464	Marfanoid syndrome, De Silva type		6 Cases
300912	Marginal zone lymphoma	7.0 *P* *	
300912	Marginal zone lymphoma	0.3 *I* *	
559	Marinesco-Sjögren syndrome		200 Cases
560	Marshall syndrome		17 Cases
561	Marshall-Smith syndrome		33 Cases
466718	Martinique crinkled retinal pigment epitheliopathy		14 Cases
98292	Mastocytosis	9.0 *P* *	
254534	Maternal 14q32.2 hypermethylation syndrome		7 Cases
254528	Maternal 14q32.2 microdeletion syndrome		8 Cases
2209	Maternal phenylketonuria	10.0 *I* *	
411712	Maternal riboflavin deficiency		1 Case
96181	Maternal uniparental disomy of chromosome 6		15 Cases
97678	Maternal uniparental disomy of chromosome 13		3 Cases
96184	Maternal uniparental disomy of chromosome 14		64 Cases
96186	Maternal uniparental disomy of chromosome 20		12 Cases
96187	Maternal uniparental disomy of chromosome 21		2 Cases

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96188	Maternal uniparental disomy of chromosome 22		4 Cases
1349	Maternally-inherited cardiomyopathy and hearing loss		2 Families
225	Maternally-inherited diabetes and deafness	0.1 *P* *	
320360	Maternally-inherited spastic paraplegia		5 Cases
2470	Matthew-Wood syndrome		43 Cases
3109	Mayer-Rokitansky-Küster-Hauser syndrome	11.0 *BP*	
2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2	1.0 *BP* *	
57782	Mazabraud syndrome		54 Cases
562	McCune-Albright syndrome	0.55 *P* *	
2471	McDonough syndrome		2 Families
2473	McKusick-Kaufman syndrome		90 Cases
59306	McLeod neuroacanthocytosis syndrome		100 Cases
3097	Meacham syndrome		13 Cases
564	Meckel syndrome	4.0 *BP*	
70588	Meconium aspiration syndrome	2.44 *P* *	
2006	Median cleft lip/mandibule		70 Cases
2699	Median nodule of the upper lip		4 Families
370127	Medich giant platelet syndrome		3 Cases
42	Medium chain acyl-CoA dehydrogenase deficiency	6.85 *P*	
42	Medium chain acyl-CoA dehydrogenase deficiency	12.0 *BP* *	
171851	MEDNIK syndrome		5 Families
1332	Medullary thyroid carcinoma	7.0 *P* *	
1332	Medullary thyroid carcinoma	0.22 *I* *	
616	Medulloblastoma	1.0 *P* *	
616	Medulloblastoma	0.11 *I* *	
98954	Meesmann corneal dystrophy		250 Cases
280671	Megaconial congenital muscular dystrophy		19 Cases
2241	Megacystis-microcolon-intestinal hypoperistalsis syndrome		230 Cases
2478	Megalencephalic leukoencephalopathy with subcortical cysts		100 Cases
60040	Megalencephaly-capillary malformation-polymicrogyria syndrome		170 Cases
83473	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome		62 Cases
457359	Megalencephaly-severe kyphoscoliosis-overgrowth syndrome		2 Cases
352328	MEGDEL syndrome		20 Cases
85282	MEHMO syndrome		8 Cases
550	MELAS	0.6 *P* *	
2482	Melhem-Fahl syndrome		2 Cases
2484	Melnick-Needles syndrome		70 Cases
2485	Melorheostosis	0.09 *P* *	
1879	Melorheostosis with osteopoikilosis		5 Families
401973	MEND syndrome		19 Cases
99898	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency		31 Cases
319547	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency		13 Cases
319558	Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency		49 Cases
319552	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency		180 Cases
319563	Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency		6 Cases
319600	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency		2 Cases
319595	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency		17 Cases
2495	Meningioma	0.15 *I* *	
565	Menkes disease	0.33 *BP* *	
498251	Menstrual cycle-dependent periodic fever		5 Cases
157801	Mesoaxial synostotic syndactyly with phalangeal reduction		6 Families
2496	Mesomelia-synostoses syndrome		5 Cases
2631	Mesomelic dwarfism-cleft palate-camptodactyly syndrome		2 Cases
1836	Mesomelic dysplasia, Kantaputra type		5 Families
2499	Metachondromatosis		25 Cases
512	Metachromatic leukodystrophy	0.1 *P* *	
512	Metachromatic leukodystrophy	1.47 *BP* *	
1240	Metaphyseal acroscyphodysplasia		4 Cases
1040	Metaphyseal anadysplasia		27 Cases
33067	Metaphyseal chondrodysplasia, Jansen type		16 Cases
166038	Metaphyseal chondrodysplasia, Kaitila type		2 Cases
2501	Metaphyseal chondrodysplasia, Spahr type		18 Cases

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2502	Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome		3 Cases
2504	Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome		2 Families
213531	Metaplastic carcinoma of the breast	0.06 *I* *	
2635	Metatropic dysplasia	0.2 *BP* *	
1923	Methimazole embryofetopathy		40 Cases
413690	Methotrexate toxicity or dose selection	3.0 *P* *	
2169	Methylcobalamin deficiency type cblE		27 Cases
2170	Methylcobalamin deficiency type cblG		33 Cases
308425	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency		7 Cases
26	Methylmalonic acidemia with homocystinuria		500 Cases
79284	Methylmalonic acidemia with homocystinuria type cblF		15 Cases
79282	Methylmalonic acidemia with homocystinuria, type cblC		500 Cases
79283	Methylmalonic acidemia with homocystinuria, type cblD		17 Cases
369955	Methylmalonic acidemia with homocystinuria, type cblJ		2 Cases
369962	Methylmalonic acidemia with homocystinuria, type cblX		18 Cases
280183	Methylmalonic aciduria due to transcobalamin receptor defect		5 Cases
502430	Metopic ridging-ptosis-facial dysmorphism syndrome		8 Cases
309025	Mevalonate kinase deficiency		300 Cases
29	Mevalonic aciduria		30 Cases
79329	MGAT2-CDG		13 Cases
2510	Micro syndrome		203 Cases
2511	Microbrachycephaly-ptosis-cleft lip syndrome		2 Cases
85172	Microcephalic osteodysplastic dysplasia, Saul-Wilson type		4 Cases
2637	Microcephalic osteodysplastic primordial dwarfism type II		150 Cases
2636	Microcephalic osteodysplastic primordial dwarfism types I and III		30 Cases
468631	Microcephalic primordial dwarfism due to RTTN deficiency		12 Cases
329228	Microcephalic primordial dwarfism due to ZNF335 deficiency		7 Cases
319671	Microcephalic primordial dwarfism, Alazami type		10 Cases
319675	Microcephalic primordial dwarfism, Dauber type		2 Cases
2617	Microcephalic primordial dwarfism, Montreal type		3 Cases
2643	Microcephalic primordial dwarfism, Toriello type		2 Cases
436182	Microcephalic primordial dwarfism-insulin resistance syndrome		2 Cases
2513	Microcephaly-albinism-digital anomalies syndrome		2 Cases
3433	Microcephaly-brachydactyly-kyphoscoliosis syndrome		3 Cases
2523	Microcephaly-brain defect-spasticity-hypernatremia syndrome		3 Cases
294016	Microcephaly-capillary malformation syndrome		10 Cases
2516	Microcephaly-cardiac defect-lung malsegmentation syndrome		3 Cases
2515	Microcephaly-cardiomyopathy syndrome		3 Cases
329332	Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome		4 Cases
2522	Microcephaly-cervical spine fusion anomalies syndrome		2 Cases
2521	Microcephaly-cleft palate-abnormal retinal pigmentation syndrome		3 Cases
423894	Microcephaly-complex motor and sensory axonal neuropathy syndrome		3 Cases
488168	Microcephaly-congenital cataract-psoriasiform dermatitis syndrome		5 Cases
500159	Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrom		4 Cases
457284	Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome		5 Cases
2533	Microcephaly-deafness-intellectual disability syndrome		2 Cases
217026	Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type		5 Cases
2172	Microcephaly-glomerulonephritis-marfanoid habitus syndrome		2 Cases
457351	Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome		14 Cases
2526	Microcephaly-lymphedema-chorioretinopathy syndrome		50 Families
2528	Microcephaly-microcornea syndrome, Seemanova type		2 Cases
171703	Microcephaly-polymicrogyria-corpus callosum agenesis syndrome		4 Cases
2519	Microcephaly-seizures-intellectual disability-heart disease syndrome		2 Cases

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ORPHA Number	Disease or Group of diseases	Estimated prevalence/incidence (/100,000)	Number of published cases or families
423306	Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome		2 Cases
397951	Microcephaly-thin corpus callosum-intellectual disability syndrome		4 Cases
2535	Microcornea-corectopia-macular hypoplasia syndrome		3 Cases
2536	Microcornea-glaucoma-absent frontal sinuses syndrome		4 Cases
369970	Microcornea-myopic chorioretinal atrophy-telecanthus syndrome		14 Cases
231736	Microcornea-posterior megalolenticonus-persistent fetal vasculature-coloboma syndrome		8 Cases
83642	Microcytic anemia with liver iron overload		3 Cases
217377	Microduplication Xp11.22-p11.23 syndrome		12 Cases
2538	Microgastria-limb reduction defect syndrome		16 Cases
476126	Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome		4 Cases
50810	Microlissencephaly-micromelia syndrome		2 Cases
139471	Microphthalmia with brain and digit anomalies		2 Families
1106	Microphthalmia with limb anomalies		35 Families
2556	Microphthalmia with linear skin defects syndrome		55 Cases
77299	Microphthalmia-brain atrophy syndrome		3 Cases
2547	Microphthalmia-microtia-fetal akinesia syndrome		2 Cases
251279	Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome		9 Cases
727	Microscopic polyangiitis	1.0 *I* *	
83463	Microtia	15.5 *BP*	
139450	Microtia-eye coloboma-imperforation of the nasolacrimal duct syndrome		1 Family
289522	Microtriplication 11q24.1		2 Cases
2290	Microvillus inclusion disease		137 Cases
2557	Mietens syndrome		9 Cases
2558	Mikati-Najjar-Sahli syndrome		5 Cases
169799	Mild hemophilia B	0.6 *P* *	
93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis		4 Families
531	Miller-Dieker syndrome	1.0 *BP* *	
98919	Miller-Fisher syndrome	0.1 *I* *	
352734	Minimal pigment oculocutaneous albinism type 1		10 Cases
494433	MIRAGE syndrome		19 Cases
3004	Mirror polydactyly-vertebral segmentation-limbs defects syndrome	0.3 *P* *	
293822	MITF-related melanoma and renal cell carcinoma predisposition syndrome		30 Families
352470	Mitochondrial DNA deletion syndrome with progressive myopathy		4 Cases
1933	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria		2 Cases
255235	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy		5 Cases
369897	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies		20 Cases
363534	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form		3 Cases
254875	Mitochondrial DNA depletion syndrome, myopathic form		45 Cases
314637	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency		8 Cases
289560	Mitochondrial membrane protein-associated neurodegeneration	0.1 *P*	
2598	Mitochondrial myopathy and sideroblastic anemia		7 Cases
502423	Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome		9 Cases
2597	Mitochondrial myopathy-lactic acidosis-deafness syndrome		2 Cases
298	Mitochondrial neurogastrointestinal encephalomyopathy	0.1 *P* *	
2443	Mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies	9.0 *P* *	
447784	Mitochondrial pyruvate carrier deficiency		4 Cases
746	Mitochondrial trifunctional protein deficiency	1.0 *P* *	
180234	Mixed germ cell tumor	0.01 *I* *	
324364	Mixed sclerosing bone dystrophy with extra-skeletal manifestations		2 Cases
497757	MME-related autosomal dominant Charcot Marie Tooth disease type 2		19 Cases
90056	Moderate and severe traumatic brain injury	37.8 *P* *	
169796	Moderately severe hemophilia B	0.6 *P* *	

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570	Moebius syndrome		300 Cases
2560	Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome		7 Cases
79330	MOGS-CDG		3 Cases
52368	Mohr-Tranebjaerg syndrome		91 Cases
2563	MOMO syndrome		8 Cases
228423	Monocytopenia with susceptibility to infections		22 Cases
2565	Mononen-Karnes-Senac syndrome		1 Family
77301	Monosomy 9q22.3		42 Cases
1598	Monosomy 18p	2.0 *BP* *	
1600	Monosomy 18q	2.5 *BP*	
574	Monosomy 21		50 Cases
48652	Monosomy 22q13		200 Cases
83467	Morvan syndrome		60 Cases
329813	Mosaic genome-wide paternal uniparental disomy		13 Cases
1692	Mosaic trisomy 1		18 Cases
1723	Mosaic trisomy 2		22 Cases
100071	Mosaic trisomy 3		6 Cases
1747	Mosaic trisomy 7		31 Cases
96061	Mosaic trisomy 8	3.0 *I* *	
99776	Mosaic trisomy 9		50 Cases
1708	Mosaic trisomy 16		226 Cases
1711	Mosaic trisomy 17		31 Cases
1052	Mosaic variegated aneuploidy syndrome		41 Cases
254516	Motor developmental delay due to 14q32.2 paternally expressed gene defect		53 Cases
3347	Mounier-Kühn syndrome		300 Cases
2152	Mowat-Wilson syndrome	1.7 *BP* *	
280679	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome		9 Cases
2573	Moyamoya disease	0.035 *I* *	
401945	Moyamoya disease with early-onset achalasia		9 Cases
2574	Moynahan syndrome		26 Cases
79323	MPDU1-CDG		8 Cases
79319	MPI-CDG		25 Cases
263347	MRCS syndrome		7 Cases
480536	MSH3-related attenuated familial adenomatous polyposis		4 Cases
100024	Mu-heavy chain disease		35 Cases

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398961	Mucinous adenocarcinoma of ovary	0.85 *I* *	
424053	Mucinous cystadenocarcinoma of the pancreas	0.01 *I* *	
575	Muckle-Wells syndrome		200 Cases
576	Mucolipidosis type II	0.84 *BP* *	
577	Mucolipidosis type III	1.0 *BP* *	
423461	Mucolipidosis type III alpha/beta	13.0 *P*	
579	Mucopolysaccharidosis type 1	0.25 *P* *	
579	Mucopolysaccharidosis type 1	0.82 *BP*	
580	Mucopolysaccharidosis type 2	10.0 *P* *	
580	Mucopolysaccharidosis type 2	0.68 *BP*	
217085	Mucopolysaccharidosis type 2, severe form	0.4 *BP* *	
581	Mucopolysaccharidosis type 3	0.3 *P* *	
581	Mucopolysaccharidosis type 3	0.87 *BP* *	
309297	Mucopolysaccharidosis type 4A	15.0 *P* *	
583	Mucopolysaccharidosis type 6	0.16 *P* *	
583	Mucopolysaccharidosis type 6	0.16 *BP* *	
584	Mucopolysaccharidosis type 7	0.01 *P* *	
505248	Mucopolysaccharidosis-like syndrome with congenital heart defects and hematopoietic disorders		19 Cases
53271	Muenke syndrome	3.33 *BP*	
587	Muir-Torre syndrome		205 Cases
2576	MULIBREY nanism		140 Cases
1655	Müllerian derivatives-lymphangiectasia-polydactyly syndrome		8 Cases
2491	Müllerian duct anomalies-limb anomalies syndrome		5 Cases
93686	Multicentric Castleman disease		100 Cases
371428	Multicentric osteolysis-nodulosis-arthropathy spectrum		50 Cases
139436	Multicentric reticulohistiocytosis		200 Cases
1851	Multicystic dysplastic kidney	23.26 *BP*	
3282	Multifocal atrial tachycardia	0.67 *BP*	
641	Multifocal motor neuropathy	1.5 *P*	
2091	Multinodular goiter-cystic kidney-polydactyly syndrome		3 Cases
500135	Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome		3 Cases
280633	Multiple congenital anomalies-hypotonia-seizures syndrome		10 Cases
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2		15 Cases

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652	Multiple endocrine neoplasia type 1	3.3 *P* *	
653	Multiple endocrine neoplasia type 2	2.9 *P* *	
251	Multiple epiphyseal dysplasia	5.0 *P* *	
93311	Multiple epiphyseal dysplasia type 5		18 Families
166024	Multiple epiphyseal dysplasia, Al-Gazali type		4 Cases
166011	Multiple epiphyseal dysplasia, Beighton type		1 Family
166016	Multiple epiphyseal dysplasia, Lowry type		2 Cases
166032	Multiple epiphyseal dysplasia, with miniepiphyses		2 Cases
166029	Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia		3 Cases
401869	Multiple mitochondrial dysfunctions syndrome type 1		21 Cases
401874	Multiple mitochondrial dysfunctions syndrome type 2		6 Cases
363424	Multiple mitochondrial dysfunctions syndrome type 3		2 Cases
457406	Multiple mitochondrial dysfunctions syndrome type 4		8 Cases
29073	Multiple myeloma	11.9 *P* *	
29073	Multiple myeloma	6.0 *I*	
321	Multiple osteochondromas	1.0 *P* *	
324299	Multiple paragangliomas associated with polycythemia		2 Cases
2215	Multiple pterygium-malignant hyperthermia syndrome		4 Cases
3151	Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome		2 Cases
65748	Multiple self-healing squamous epithelioma		100 Cases
585	Multiple sulfatase deficiency		50 Cases
3237	Multiple synostoses syndrome		30 Families
102	Multiple system atrophy	3.5 *P*	
102	Multiple system atrophy	1.8 *I*	
98933	Multiple system atrophy, parkinsonian type	2.4 *P* *	
404463	Multisystemic smooth muscle dysfunction syndrome		7 Cases
370997	Muscle-eye-brain disease with bilateral multicystic leucodystrophy		2 Cases
2579	Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome		10 Cases
199340	Muscular dystrophy, Selcen type		12 Cases
324416	Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome		2 Cases
659	Mutilating palmoplantar keratoderma with periorificial keratotic plaques		73 Cases
589	Myasthenia gravis	7.77 *P*	
589	Myasthenia gravis	0.53 *I*	
498693	MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome		4 Cases
268249	Mycophenolate mofetil embryopathy		25 Cases
178566	Mycosis fungoides and variants	0.59 *I* *	
52688	Myelodysplastic syndrome	1.5 *I* *	
98275	Myelodysplastic/myeloproliferative disease	0.29 *I* *	
86850	Myeloid sarcoma	0.02 *I* *	
98274	Myeloproliferative neoplasm	3.07 *I* *	
437572	MYH7-related late-onset scapuloperoneal muscular dystrophy		12 Cases
182050	MYH9-related disease	0.3 *P* *	
2588	Myhre syndrome		55 Cases
480491	MYO5B-related progressive familial intrahepatic cholestasis		5 Cases
86909	Myoclonic epilepsy of infancy		106 Cases
2589	Myoclonus-cerebellar ataxia-deafness syndrome		4 Cases
88635	Myopathy due to calsequestrin and SERCA1 protein overload		4 Cases
2601	Myopathy-growth delay-intellectual disability-hypospadias syndrome		1 Case
206647	Myotonic dystrophy	6.7 *P*	
99967	Myxoid/round cell liposarcoma	0.1 *I* *	
2608	N syndrome		3 Cases
69087	Naegeli-Franceschetti-Jadassohn syndrome	0.035 *P* *	
245	Nager syndrome		100 Cases
423454	Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome		6 Cases
2614	Nail-patella syndrome	0.2 *BP* *	
2613	Nail-patella-like renal disease		3 Cases
2615	Nakajo-Nishimura syndrome		30 Cases
627	Nance-Horan syndrome		196 Cases
2073	Narcolepsy-cataplexy syndrome	25.0 *P* *	
2399	Nasopalpebral lipoma-coloboma syndrome		30 Cases
150	Nasopharyngeal carcinoma	2.0 *P* *	
150	Nasopharyngeal carcinoma	0.36 *I* *	

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1654	Natal teeth-intestinal pseudoobstruction-patent ductus syndrome		2 Cases
2663	Nathalie syndrome		1 Family
255229	Navajo neurohepatopathy		49 Cases
443162	NDE1-related microhydranencephaly		1 Family
391673	Necrotizing enterocolitis	45.0 *P*	
464366	NEK9-related lethal skeletal dysplasia		5 Cases
607	Nemaline myopathy	2.0 *BP* *	
217563	Neonatal acute respiratory distress due to SP-B deficiency	0.067 *BP*	
398097	Neonatal antiphospholipid syndrome		34 Cases
398109	Neonatal autoimmune hemolytic anemia		2 Cases
398117	Neonatal dermatomyositis		3 Cases
224	Neonatal diabetes mellitus	1.1 *BP* *	
79118	Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome		3 Cases
457185	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome		11 Cases
446	Neonatal hemochromatosis		35 Cases
59303	Neonatal ichthyosis-sclerosing cholangitis syndrome		12 Cases
294023	Neonatal inflammatory skin and bowel disease		3 Cases
398127	Neonatal scleroderma		6 Cases
466784	Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect		3 Cases
94058	Neovascular glaucoma	24.4 *P* *	
654	Nephroblastoma	10.0 *BP* *	
654	Nephroblastoma	0.14 *I* *	
223	Nephrogenic diabetes insipidus	0.15 *P* *	
3145	Nephrogenic diabetes insipidus-intracranial calcification syndrome		2 Cases
93606	Nephrogenic syndrome of inappropriate antidiuresis		21 Cases
2668	Nephropathy-deafness-hyperparathyroidism syndrome		5 Cases
2669	Nephrosis-deafness-urinary tract-digital malformations syndrome		5 Cases
300333	Nephrotic syndrome-deafness-pretibial epidermolysis bullosa syndrome		3 Cases
280576	Nestor-Guillermo progeria syndrome		2 Cases
634	Netherton syndrome	0.5 *P* *	
634	Netherton syndrome	0.5 *BP* *	
2671	Neu-Laxova syndrome		91 Cases
2672	Neuhauser-Eichner-Opitz syndrome		5 Cases
3388	Neural tube defect	91.05 *BP* *	
635	Neuroblastoma	11.0 *P* *	
635	Neuroblastoma	5.8 *BP* *	
635	Neuroblastoma	1.26 *I*	
2481	Neurocutaneous melanocytosis	1.25 *P* *	
88639	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency		20 Cases
385	Neurodegeneration with brain iron accumulation	0.2 *P* *	
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency		3 Cases
453499	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome		15 Cases
352665	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to 9q21 microdeletion		13 Cases
453504	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome due to a point mutation		2 Cases
33445	Neuroectodermal melanolysosomal disease		20 Cases
2676	Neuroectodermal-endocrine syndrome		4 Cases
877	Neuroendocrine neoplasm	2.53 *I* *	
97253	Neuroendocrine tumor of pancreas	0.21 *I* *	
100075	Neuroendocrine tumor of stomach	3.2 *P* *	
2673	Neurofaciodigitorenal syndrome		3 Cases
157846	Neuroferritinopathy		50 Cases
636	Neurofibromatosis type 1	21.3 *P* *	
636	Neurofibromatosis type 1	33.3 *BP*	
637	Neurofibromatosis type 2	1.7 *P* *	
1143	Neurogenic arthrogryposis multiplex congenita	4.3 *BP* *	
35705	Neurometabolic disorder due to serine deficiency		30 Cases
71211	Neuromyelitis optica	1.5 *P* *	
139512	Neuropathy with hearing impairment		1 Family
137596	Neurotrophic keratopathy	4.2 *P* *	
165	Neutral lipid storage disease		50 Cases
98908	Neutral lipid storage myopathy		36 Cases

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2690	Neutropenia-monocytopenia-deafness syndrome		3 Cases
183707	Neutrophil immunodeficiency syndrome		2 Cases
263432	Nevus of Ito	1.17 *P* *	
77292	Niemann-Pick disease type A	0.25 *BP* *	
77293	Niemann-Pick disease type B	0.4 *P* *	
646	Niemann-Pick disease type C	1.0 *P* *	
1390	Night blindness-skeletal anomalies-dysmorphism syndrome		2 Cases
647	Nijmegen breakage syndrome	1.0 *BP*	
240760	Nijmegen breakage syndrome-like disorder		1 Case
447731	NIK deficiency		2 Cases
99825	Nipah virus disease		556 Cases
263665	NK-cell enteropathy		8 Cases
247868	NLRP12-associated hereditary periodic fever syndrome		19 Cases
86893	Nodular lymphocyte predominant Hodgkin lymphoma	0.12 *I*	
467	Non-acquired combined pituitary hormone deficiency	29.0 *BP* *	
231720	Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome		13 Cases
631	Non-acquired isolated growth hormone deficiency	0.39 *P*	
2337	Non-epidermolytic palmoplantar keratoderma	2.5 *P* *	
2972	Non-eruption of teeth-maxillary hypoplasia-genu valgum syndrome		4 Cases
91349	Non-functioning pituitary adenoma	1.05 *I*	
357034	Non-hereditary retinoblastoma	0.038 *I* *	
547	Non-Hodgkin lymphoma	11.6 *I* *	
329883	Non-hypoproteinemic hypertrophic gastropathy		1 Family
363999	Non-immune hydrops fetalis	42.0 *BP*	
329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis	14.0 *P* *	
329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis	0.15 *I* *	
90061	Non-infectious posterior uveitis	18.0 *P* *	
209989	Non-papillary transitional cell carcinoma of the bladder	37.0 *P* *	
314647	Non-progressive cerebellar ataxia with intellectual disability		15 Cases
363494	Non-seminomatous germ cell tumor of testis	1.21 *I* *	
90031	Non-spherocytic hemolytic anemia due to hexokinase deficiency		17 Families
500	Noonan syndrome with multiple lentigines		296 Cases
2701	Noonan syndrome-like disorder with loose anagen hair		27 Cases
649	Norrie disease		400 Cases
75327	North Carolina macular dystrophy		2 Families
3032	NPHP3-related Meckel-like syndrome		10 Cases
397615	Obesity due to CEP19 deficiency		15 Cases
66628	Obesity due to congenital leptin deficiency		30 Cases
71526	Obesity due to pro-opiomelanocortin deficiency		7 Cases
71528	Obesity due to prohormone convertase I deficiency		16 Cases
88643	Obesity-colitis-hypothyroidism-cardiac hypertrophy-developmental delay syndrome		2 Cases
198	Occipital horn syndrome		20 Cases
280640	Occipital pachygyria and polymicrogyria		3 Cases
2704	Ochoa syndrome		100 Cases
352740	Ocular albinism with congenital sensorineural deafness		3 Families
1000	Ocular albinism with late-onset sensorineural deafness		9 Cases
194	Ocular coloboma	8.0 *BP* *	
1125	Ocular motor apraxia, Cogan type		50 Cases
2714	Oculo-palato-cerebral syndrome		5 Cases
157962	Oculoauricular syndrome, Schorderet type		5 Cases
398156	Oculoauriculofrontonasal syndrome		41 Cases
2705	Oculocerebral dysplasia		2 Cases
2719	Oculocerebral hypopigmentation syndrome, Cross type		14 Cases
2720	Oculocerebral hypopigmentation syndrome, Preus type		2 Cases
1647	Oculocerebrocutaneous syndrome		38 Cases
2707	Oculocerebrofacial syndrome, Kaufman type		14 Cases
534	Oculocerebrorenal syndrome of Lowe	0.2 *P*	
55	Oculocutaneous albinism	5.9 *P*	
352731	Oculocutaneous albinism type 1	2.5 *P*	
79431	Oculocutaneous albinism type 1A	1.3 *P*	
79434	Oculocutaneous albinism type 1B	1.3 *P*	
79432	Oculocutaneous albinism type 2	2.55 *P*	
79435	Oculocutaneous albinism type 4	1.0 *P*	

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370091	Oculocutaneous albinism type 5		1 Family
370097	Oculocutaneous albinism type 6		1 Case
352745	Oculocutaneous albinism type 7		9 Cases
2709	Oculodental syndrome, Rutherfurd type		1 Family
2710	Oculodentodigital dysplasia		243 Cases
1876	Oculogastrointestinal muscular dystrophy		1 Family
1794	Oculomaxillofacial dysostosis		4 Cases
2713	Oculoosteocutaneous syndrome		3 Cases
99806	Oculootodental syndrome		1 Family
2715	Oculorenocerebellar syndrome		5 Cases
2717	Oculotrichoanal syndrome		20 Cases
2718	Oculotrichodysplasia		2 Cases
2722	Odonto-onycho dysplasia-alopecia syndrome		2 Cases
2721	Odonto-onycho-dermal dysplasia		30 Cases
69082	Odonto-tricho-ungual-digito-palmar syndrome		21 Cases
166272	Odontochondrodysplasia		11 Cases
77295	Odontoleukodystrophy		4 Cases
2724	Odontomatosis-aortae esophagus stenosis syndrome		3 Cases
1811	Odontomicronychial dysplasia		5 Cases
2723	Odontotrichomelic syndrome		4 Cases
391655	Off-periods in Parkinson disease not responding to oral treatment	4.15 *P* *	
276432	Ogden syndrome		8 Cases
75382	Oguchi disease		50 Cases
2729	Okamoto syndrome		5 Cases
85410	Oligoarticular juvenile idiopathic arthritis	20.5 *P* *	
251651	Oligoastrocytic tumor	0.11 *I* *	
75378	Oligocone trichromacy		14 Cases
46484	Oligodendroglial tumor	0.35 *I* *	
251627	Oligodendroglioma	0.25 *I* *	
300576	Oligodontia-cancer predisposition syndrome		2 Families
2920	Oliver syndrome		7 Cases
39041	Omenn syndrome		25 Cases
2733	Omodysplasia		30 Cases
660	Omphalocele	11.7 *BP* *	
3164	Omphalocele syndrome, Shprintzen-Goldberg type		5 Cases
496693	Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome		7 Cases
352540	Oncogenic osteomalacia		400 Cases
300504	Onychocytic matricoma		5 Cases
300512	Onychomatricoma		50 Cases
2741	Ophthalmomandibulomelic dysplasia		3 Cases
2743	Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome		6 Cases
2745	Opitz G/BBB syndrome	3.0 *P* *	
2746	Opsismodysplasia		30 Cases
1183	Opsoclonus-myoclonus syndrome	0.02 *I* *	
401777	Optic atrophy-intellectual disability syndrome		6 Cases
496790	Optic atrophy-peripheral neuropathy-developmental delay syndrome		8 Cases
313800	Optic nerve edema-splenomegaly syndrome		3 Cases
2086	Optic pathway glioma	0.12 *I*	
52994	Orbital leiomyoma		26 Cases
664	Ornithine transcarbamylase deficiency	1.4 *P* *	
664	Ornithine transcarbamylase deficiency	1.77 *BP*	
2750	Orofaciodigital syndrome type 1	1.2 *BP* *	
2751	Orofaciodigital syndrome type 2		20 Cases
2752	Orofaciodigital syndrome type 3		5 Cases
2753	Orofaciodigital syndrome type 4		29 Cases
2919	Orofaciodigital syndrome type 5		12 Cases
2754	Orofaciodigital syndrome type 6		2 Families
2755	Orofaciodigital syndrome type 8		20 Cases
141007	Orofaciodigital syndrome type 9		10 Cases
141327	Orofaciodigital syndrome type 12		1 Case
141330	Orofaciodigital syndrome type 13		1 Case
434179	Orofaciodigital syndrome type 14		2 Families
443236	Orthostatic intolerance due to NET deficiency		2 Cases
2760	OSLAM syndrome		3 Cases
73230	Ossification anomalies-psychomotor developmental delay syndrome		2 Cases
2764	Osteochondritis dissecans	35.0 *P* *	
2653	Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome		2 Cases
2763	Osteocraniostenosis		30 Cases
666	Osteogenesis imperfecta	10.0 *P* *	
216804	Osteogenesis imperfecta type 2	0.4 *BP* *	

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216828	Osteogenesis imperfecta type 5		47 Cases
2773	Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome		2 Cases
2645	Osteoglosphonic dysplasia		7 Cases
2777	Osteomesopyknosis		35 Cases
2780	Osteopathia striata-cranial sclerosis syndrome		100 Cases
2779	Osteopathia striata-pigmentary dermopathy-white forelock syndrome		3 Cases
2324	Osteopenia-intellectual disability-sparse hair syndrome		2 Cases
91133	Osteopenia-myopia-hearing loss-intellectual disability-facial dysmorphism syndrome		2 Cases
2781	Osteopetrosis and related disorders	1.0 *I* *	
2785	Osteopetrosis with renal tubular acidosis		100 Cases
178389	Osteopetrosis-hypogammaglobulinemia syndrome		8 Cases
2786	Osteoporosis-oculocutaneous hypopigmentation syndrome		1 Case
2788	Osteoporosis-pseudoglioma syndrome	0.05 *P* *	
668	Osteosarcoma	0.23 *I* *	
178377	Osteosclerosis-developmental delay-craniosynostosis syndrome		13 Cases
75325	Osteosclerosis-ichthyosis-premature ovarian failure syndrome		3 Cases
500548	Osteosclerotic metaphyseal dysplasia		7 Cases
2791	Otodental syndrome		10 Families
2793	Otoonychoperoneal syndrome		6 Cases
90652	Otopalatodigital syndrome type 2		40 Cases
1427	Otospondylomegaepiphyseal dysplasia		30 Cases
213500	Ovarian cancer	30.0 *P* *	
99853	Ovarioleukodystrophy		17 Cases
498488	Overgrowth syndrome with 2q37 translocation		4 Cases
137634	Overgrowth-macrocephaly-facial dysmorphism syndrome		6 Families
498485	Overgrowth-metaphyseal undermodeling-spondylar dysplasia syndrome		4 Cases
3203	Overhydrated hereditary stomatocytosis		20 Families
36355	P2Y12 defect		14 Cases
2796	Pachydermoperiostosis		204 Cases
2798	Pachygyria-intellectual disability-epilepsy syndrome		5 Cases
2309	Pachyonychia congenita		1000 Cases
1952	Pacman dysplasia		4 Cases
180275	Paget disease of the nipple	0.51 *I* *	
991	PAGOD syndrome		6 Cases
1993	Pai syndrome		37 Cases
300501	Painful orbital and systemic neurofibromas-marfanoid habitus syndrome		4 Cases
477993	Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome		3 Cases
672	Pallister-Hall syndrome		100 Cases
140966	Palmoplantar keratoderma, Nagashima type		40 Cases
2202	Palmoplantar keratoderma-deafness syndrome		10 Families
2198	Palmoplantar keratoderma-esophageal carcinoma syndrome		10 Families
2201	Palmoplantar keratoderma-spastic paralysis syndrome		25 Cases
85112	Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome		5 Cases
2255	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome		10 Cases
199337	Pancreatic insufficiency-anemia-hyperostosis syndrome		5 Cases
677	Pancreatoblastoma		60 Cases
317473	Pancytopenia due to IKZF1 mutations		1 Case
401764	Pancytopenia-developmental delay syndrome		2 Cases
157850	Pantothenate kinase-associated neurodegeneration	0.15 *P* *	
319298	Papillary renal cell carcinoma	0.14 *I* *	
678	Papillon-Lefèvre syndrome	0.25 *P*	
2812	Parana hard skin syndrome		8 Cases
63455	Paraneoplastic pemphigus		60 Cases
2824	Paraplegia-intellectual disability-hyperkeratosis syndrome		6 Cases
143	Parathyroid carcinoma	0.02 *I* *	
2825	PARC syndrome		2 Cases
251290	Parietal foramina with clavicular hypoplasia		8 Cases
851	Paris-Trousseau thrombocytopenia		50 Cases
314632	Parkinsonism due to ATP13A2 deficiency		4 Cases
53583	Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity		20 Cases

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98811	Paroxysmal exertion-induced dyskinesia		50 Cases
46348	Paroxysmal extreme pain disorder		4 Families
157835	Paroxysmal hemicrania	2.0 *P* *	
98809	Paroxysmal kinesigenic dyskinesia	0.6 *P*	
447	Paroxysmal nocturnal hemoglobinuria	2.0 *P* *	
98810	Paroxysmal non-kinesigenic dyskinesia	0.1 *P*	
1330	Partial atrioventricular canal	30.0 *P* *	
1330	Partial atrioventricular canal	20.0 *BP* *	
1646	Partial chromosome Y deletion	20.8 *P*	
401959	Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome		2 Cases
90076	Partial deep dermal and full thickness burns	10.0 *P* *	
262941	Partial duplication of the long arm of chromosome 14		50 Cases
2805	Partial pancreatic agenesis		50 Cases
94083	Partington syndrome		2 Families
86789	Patella aplasia/hypoplasia		5 Families
228190	Patent ductus arteriosus-bicuspid aortic valve-hand anomalies syndrome		7 Cases
254531	Paternal 14q32.2 hypomethylation syndrome		12 Cases
254525	Paternal 14q32.2 microdeletion syndrome		9 Cases
261304	Paternal 20q13.2q13.3 microdeletion syndrome		2 Cases
96192	Paternal uniparental disomy of chromosome 7		4 Cases
96334	Paternal uniparental disomy of chromosome 14		37 Cases
2439	Patterson-Stevenson-Fontaine syndrome		7 Cases
438134	PCNA-related progressive neurodegenerative photosensitivity syndrome		4 Cases
439822	PDE4D haploinsufficiency syndrome		7 Cases
699	Pearson syndrome		95 Cases
2835	Pectus excavatum-macrocephaly-dysplastic nails syndrome		1 Family
93682	Pediatric Castleman disease		150 Cases
487809	Pediatric collagenous gastritis		24 Cases
33402	Pediatric hepatocellular carcinoma	0.15 *I* *	
263548	Peeling skin syndrome type A		40 Families
263553	Peeling skin syndrome type B		30 Families
444138	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome		4 Cases
99807	PEHO-like syndrome		10 Cases
702	Pelizaeus-Merzbacher disease	0.25 *P* *	
280219	Pelizaeus-Merzbacher disease, classic form	0.17 *P* *	
280210	Pelizaeus-Merzbacher disease, connatal form	0.03 *P* *	
280224	Pelizaeus-Merzbacher disease, transitional form	0.03 *P* *	
2840	Pelvic dysplasia-arthrogryposis of lower limbs syndrome		5 Cases
2839	Pelvis-shoulder dysplasia		10 Cases
93333	Pelviscapular dysplasia		4 Cases
704	Pemphigus vulgaris	18.0 *P* *	
705	Pendred syndrome	7.0 *P* *	
49	Penile agenesis		80 Cases
313936	PENS syndrome		13 Cases
1335	Pentalogy of Cantrell	0.67 *BP*	
2847	Pericardial and diaphragmatic defect		20 Cases
65250	Perineural cyst	50.0 *I* *	
436166	Periodic fever-infantile enterocolitis-autoinflammatory syndrome		4 Cases
397750	Periodic paralysis with later-onset distal motor neuropathy		9 Cases
397755	Periodic paralysis with transient compartment-like syndrome		4 Cases
139426	Perioral myoclonia with absences		10 Cases
563	Peripartum cardiomyopathy	30.0 *BP*	
163746	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease		40 Cases
2400	Peripheral motor neuropathy-dysautonomia syndrome		2 Cases
397744	Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome		15 Cases
97927	Peripheral resistance to thyroid hormones	2.5 *P* *	
168816	Peritoneal cystic mesothelioma		150 Cases
2849	Perlman syndrome		30 Cases
226292	Permanent congenital hypothyroidism	33.3 *BP* *	
99885	Permanent neonatal diabetes mellitus	0.38 *BP* *	
65288	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome		4 Cases

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2971	Peroxisomal acyl-CoA oxidase deficiency		40 Cases
2855	Perrault syndrome		61 Cases
178509	Perry syndrome		53 Cases
97341	Persistent placoid maculopathy		5 Cases
300324	Persistent polyclonal B-cell lymphocytosis		154 Cases
708	Peters anomaly		60 Cases
709	Peters plus syndrome		80 Cases
2869	Peutz-Jeghers syndrome	0.4 *P* *	
2869	Peutz-Jeghers syndrome	2.2 *BP*	
42642	PFAPA syndrome		500 Cases
710	Pfeiffer syndrome	1.0 *I* *	
2871	Pfeiffer-Palm-Teller syndrome		2 Cases
319646	PGM1-CDG		46 Cases
443811	PGM3-CDG		20 Cases
42775	PHACE syndrome		300 Cases
2874	Phakomatosis pigmentokeratotica		34 Cases
352636	Phalangeal microgeodic syndrome		50 Cases
2876	PHAVER syndrome		2 Cases
716	Phenylketonuria	10.0 *BP* *	
2878	Phocomelia-ectrodactyly-deafness-sinus arrhythmia syndrome		4 Cases
2880	Phosphoenolpyruvate carboxykinase deficiency		10 Cases
3222	Phosphoribosylpyrophosphate synthetase superactivity		30 Families
498228	Phyllodes tumor of the prostate		90 Cases
2885	Piebald trait-neurologic defects syndrome		2 Families
487825	Pierpont syndrome		7 Cases
2888	Pierre Robin syndrome-faciodigital anomaly syndrome		2 Cases
2670	Pierson syndrome		40 Families
447961	Pigmentation defects-palmoplantar keratoderma-skin carcinoma syndrome		2 Cases
251295	Pigmented paravenous retinochoroidal atrophy		100 Cases
66627	Pigmented villonodular synovitis	20.0 *P* *	
2891	Pili torti-developmental delay-neurological abnormalities syndrome		2 Cases
2890	Pili torti-onychodysplasia syndrome		1 Family
2892	Pilodental dysplasia-refractive errors syndrome		2 Cases
251909	Pineoblastoma	0.02 *I* *	
2896	Pitt-Hopkins syndrome		500 Cases
221150	Pitt-Hopkins-like syndrome		105 Cases
300385	Pituitary carcinoma	0.04 *I* *	
2897	Pityriasis rubra pilaris		48 Cases
439167	Placental insufficiency	33.0 *P*	
99928	Placental site trophoblastic tumor	0.02 *I* *	
707	Plague	2.2 *I* *	
454714	Plasma cell leukemia	0.04 *I* *	
300359	PLCG2-associated antibody deficiency and immune dysregulation		3 Families
99969	Pleomorphic liposarcoma	0.05 *I* *	
454821	Pleomorphic salivary gland adenoma	2.725 *I*	
251607	Pleomorphic xanthoastrocytoma	0.01 *I* *	
449266	Pleural empyema	13.0 *P* *	
50251	Pleural mesothelioma	3.1 *P* *	
50251	Pleural mesothelioma	1.9 *I* *	
64742	Pleuropulmonary blastoma	0.5 *BP* *	
284343	Pleuropulmonary blastoma familial tumor susceptibility syndrome	0.007 *I*	
280356	PLIN1-related familial partial lipodystrophy		3 Cases
54028	Plummer-Vinson syndrome		25 Cases
476394	PMP2-related Charcot-Marie-Tooth disease type 1		13 Cases
477817	PMP22-RAI1 contiguous gene duplication syndrome		23 Cases
90066	Pneumonia caused by Pseudomonas aeruginosa infection	50.0 *P* *	
221046	Poikiloderma with neutropenia		50 Cases
2911	Poland syndrome	1.5 *BP* *	
330009	Poliomyelitis in patients with immunodeficiencies deemed at risk	8.0E-4 *P* *	
767	Polyarteritis nodosa	3.0 *P* *	
2795	Polycystic ovaries-urethral sphincter dysfunction syndrome		33 Cases
729	Polycythemia vera	30.0 *P* *	
729	Polycythemia vera	1.9 *I* *	
2917	Polydactyly-myopia syndrome		1 Family
453533	Polyendocrine-polyneuropathy syndrome		3 Cases
397937	Polyglucosan body myopathy type 1		11 Cases
456369	Polyglucosan body myopathy type 2		15 Cases

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500533	Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome		17 Cases
183422	Polymalformative genetic syndrome with increased risk of developing cancer	10.0 *P* *	
300573	Polymicrogyria due to TUBB2B mutation		36 Cases
250972	Polymicrogyria with optic nerve hypoplasia		4 Cases
732	Polymyositis	7.1 *P* *	
732	Polymyositis	0.585 *I* *	
171848	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome		19 Cases
2928	Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome		3 Cases
2934	Polysyndactyly-cardiac malformation syndrome		8 Cases
228410	Polyvalvular heart disease syndrome		19 Cases
477749	Pontine autosomal dominant microangiopathy with leukoencephalopathy		11 Cases
269229	Pontine tegmental cap dysplasia		22 Cases
2254	Pontocerebellar hypoplasia type 1		40 Families
2524	Pontocerebellar hypoplasia type 2		81 Families
97249	Pontocerebellar hypoplasia type 3		3 Families
166063	Pontocerebellar hypoplasia type 4		10 Families
166068	Pontocerebellar hypoplasia type 5		3 Cases
166073	Pontocerebellar hypoplasia type 6		10 Cases
284339	Pontocerebellar hypoplasia type 7		4 Cases
324569	Pontocerebellar hypoplasia type 8		6 Cases
369920	Pontocerebellar hypoplasia type 9		14 Cases
411493	Pontocerebellar hypoplasia type 10		23 Cases
294963	Popliteal pterygium syndrome	0.3 *P* *	
2941	Porencephaly-cerebellar hypoplasia-internal malformations syndrome		2 Cases
306547	Porencephaly-microcephaly-bilateral congenital cataract syndrome		8 Cases
166286	Porokeratotic eccrine ostial and dermal duct nevus		45 Cases
738	Porphyria	5.25 *P*	
101330	Porphyria cutanea tarda	4.0 *P* *	
101330	Porphyria cutanea tarda	0.6 *I* *	
79473	Porphyria variegata	0.32 *P* *	
79473	Porphyria variegata	0.008 *I* *	
2703	Port-wine nevi-mega cisterna magna-hydrocephalus syndrome		5 Cases
70568	Post-transplant lymphoproliferative disease	26.2 *P* *	
246	Postaxial acrofacial dysostosis		30 Cases
420584	Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome		112 Cases
2916	Postaxial polydactyly-dental and vertebral anomalies syndrome		3 Cases
2730	Postaxial tetramelic oligodactyly		4 Cases
98971	Posterior amorphous corneal dystrophy		11 Families
88628	Posterior column ataxia-retinitis pigmentosa syndrome		20 Cases
2064	Posterior fusion of lumbosacral vertebrae-blepharoptosis syndrome		3 Cases
93110	Posterior urethral valve	2.0 *P* *	
93110	Posterior urethral valve	4.125 *BP* *	
477673	Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome		17 Cases
279947	Postorgasmic illness syndrome		45 Cases
52022	Potocki-Shaffer syndrome		40 Cases
217067	Pouchitis	22.0 *P* *	
79083	PPARG-related familial partial lipodystrophy		10 Cases
739	Prader-Willi syndrome	3.1 *BP* *	
398069	Prader-Willi syndrome due to a point mutation		28 Cases
398073	Prader-Willi-like syndrome		117 Cases
398079	Prader-Willi-like syndrome due to a point mutation		4 Cases
2956	Prata-Liberal-Goncalves syndrome		2 Cases
293462	Pre-Descemet corneal dystrophy		5 Cases
2921	Preaxial polydactyly-colobomata-intellectual disability syndrome		2 Cases
275555	Preeclampsia	45.0 *P* *	
79410	Pretibial dystrophic epidermolysis bullosa		40 Families
186	Primary biliary cholangitis	21.05 *P*	
186	Primary biliary cholangitis	3.0 *I*	
169464	Primary CD59 deficiency		6 Cases
244	Primary ciliary dyskinesia	5.0 *BP* *	
247522	Primary ciliary dyskinesia-retinitis pigmentosa syndrome		20 Cases
226295	Primary congenital hypothyroidism	37.5 *P* *	
541	Primary cutaneous CD30+ T-cell lymphoproliferative disease	0.18 *I* *	
542	Primary cutaneous lymphoma	0.75 *I* *	

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171901	Primary cutaneous T-cell lymphoma	24.0 *P* *	
171901	Primary cutaneous T-cell lymphoma	5.2 *I* *	
98805	Primary dystonia, DYT4 type		22 Cases
98806	Primary dystonia, DYT6 type		53 Cases
98807	Primary dystonia, DYT13 type		8 Cases
370103	Primary dystonia, DYT17 type		3 Cases
306734	Primary dystonia, DYT21 type		16 Cases
464440	Primary dystonia, DYT27 type		5 Cases
48686	Primary effusion lymphoma		200 Cases
90026	Primary erythermalgia		30 Families
100085	Primary hepatic neuroendocrine carcinoma	0.2 *I*	
2232	Primary hypergonadotropic hypogonadism-partial alopecia syndrome		7 Cases
93599	Primary hyperoxaluria type 2		10 Cases
93600	Primary hyperoxaluria type 3		50 Cases
30924	Primary hypomagnesemia with secondary hypocalcemia		100 Cases
90023	Primary immunodeficiency syndrome due to p14 deficiency		4 Cases
75391	Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency		4 Cases
431166	Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection		1 Case
458768	Primary intralymphatic angioendothelioma		30 Cases
35689	Primary lateral sclerosis	1.5 *P* *	
77240	Primary lymphedema	16.7 *P* *	
98838	Primary mediastinal large B-cell lymphoma	3.0 *P* *	
54370	Primary membranoproliferative glomerulonephritis	16.0 *P* *	
306558	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome		7 Cases
391408	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome		8 Cases
824	Primary myelofibrosis	3.0 *P* *	
824	Primary myelofibrosis	1.0 *I* *	
238606	Primary orthostatic tremor		390 Cases
189439	Primary pigmented nodular adrenocortical disease	0.04 *P* *	
95432	Primary progressive aphasia	7.0 *P*	
314566	Primary progressive apraxia of speech		16 Cases
171	Primary sclerosing cholangitis	8.1 *P*	
171	Primary sclerosing cholangitis	0.65 *I*	
289390	Primary Sjögren syndrome	48.99 *P* *	
314701	Primary systemic amyloidosis	30.0 *P* *	
412066	PRKAR1B-related neurodegenerative dementia with intermediate filaments		12 Cases
2959	Progeria-short stature-pigmented nevi syndrome		11 Cases
300382	Progeroid and marfanoid aspect-lipodystrophy syndrome		7 Cases
435953	Progeroid features-hepatocellular carcinoma predisposition syndrome		3 Cases
2963	Progeroid syndrome, Petty type		1 Case
448251	Progressive autosomal recessive ataxia-deafness syndrome		13 Cases
75373	Progressive bifocal chorioretinal atrophy		2 Families
139447	Progressive cavitating leukoencephalopathy		19 Cases
247198	Progressive cerebello-cerebral atrophy		7 Cases
431361	Progressive encephalopathy with leukodystrophy due to DECR deficiency		2 Cases
457212	Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome		5 Cases
352447	Progressive external ophthalmoplegia-myopathy-emaciation syndrome		6 Cases
480483	Progressive familial intrahepatic cholestasis type 4		14 Cases
480476	Progressive familial intrahepatic cholestasis type 5		4 Cases
477814	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome		9 Cases
263516	Progressive myoclonic epilepsy type 3		9 Families
402082	Progressive myoclonic epilepsy type 5		3 Cases
280620	Progressive myoclonic epilepsy type 6		12 Cases
435438	Progressive myoclonic epilepsy type 7		13 Cases
424027	Progressive myoclonic epilepsy type 8		4 Cases
457265	Progressive myoclonic epilepsy type 9		2 Cases
352596	Progressive myoclonic epilepsy with dystonia		5 Cases
100070	Progressive non-fluent aphasia	2.5 *P* *	
100070	Progressive non-fluent aphasia	0.7 *I* *	

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2062	Progressive non-infectious anterior vertebral fusion		67 Cases
217396	Progressive polyneuropathy with bilateral striatal necrosis		4 Cases
352718	Progressive retinal dystrophy due to retinol transport defect		5 Cases
447977	Progressive scapulohumeroperoneal distal myopathy		33 Cases
228012	Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome		4 Families
457395	Progressive spondyloepimetaphyseal dysplasia-short stature-short fourth metatarsals-intellectual disability syndrome		4 Cases
683	Progressive supranuclear palsy	6.0 *P*	
683	Progressive supranuclear palsy	0.65 *I*	
240103	Progressive supranuclear palsy-corticobasal syndrome	0.6 *P* *	
240112	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome		10 Cases
742	Prolidase deficiency		90 Cases
2083	Prominent glabella-microcephaly-hypogenitalism syndrome		2 Cases
35	Propionic acidemia	0.2 *P* *	
35	Propionic acidemia	1.5 *I*	
324977	Proteasome disability syndrome		40 Cases
251598	Protoplasmic astrocytoma	0.01 *I* *	
261197	Proximal 16p11.2 microdeletion syndrome	20.0 *P* *	
401768	Proximal myopathy with extrapyramidal signs		15 Cases
606	Proximal myotonic myopathy	1.0 *P* *	
70	Proximal spinal muscular atrophy	20.0 *BP* *	
70	Proximal spinal muscular atrophy	2.6 *I* *	
83330	Proximal spinal muscular atrophy type 1	0.26 *I* *	
83418	Proximal spinal muscular atrophy type 2	1.23 *I* *	
83419	Proximal spinal muscular atrophy type 3	1.1 *I* *	
83420	Proximal spinal muscular atrophy type 4	0.32 *I* *	
397606	PrP systemic amyloidosis		16 Cases
52530	Pseudo-von Willebrand disease		60 Cases
750	Pseudoachondroplasia	3.3 *P*	
221120	Pseudoaminopterin syndrome		11 Cases
85174	Pseudodiastrophic dysplasia		13 Cases
756	Pseudohypoaldosteronism type 1		70 Cases
757	Pseudohypoaldosteronism type 2		80 Families
300525	Pseudohypoaldosteronism type 2D		24 Cases
300530	Pseudohypoaldosteronism type 2E		17 Cases
2976	Pseudoleprechaunism syndrome, Patterson type		2 Cases
26790	Pseudomyxoma peritonei	0.1 *I*	
2985	Pseudoprogeria syndrome		2 Cases
758	Pseudoxanthoma elasticum	2.5 *P* *	
436274	Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa		13 Cases
280794	Pseudoxanthomatous diffuse cutaneous mastocytosis		10 Cases
85436	Psoriasis-related juvenile idiopathic arthritis	4.2 *P* *	
505242	Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome		6 Cases
88618	Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency		4 Cases
1578	Pterin-4 alpha-carbinolamine dehydratase deficiency		21 Cases
2988	Pterygium colli-intellectual disability-digital anomalies syndrome		2 Cases
2999	Ptosis-strabismus-ectopic pupils syndrome		1 Family
238766	Ptosis-syndactyly-learning difficulties syndrome		5 Cases
228396	Ptosis-upper ocular movement limitation-absence of lacrimal punctum syndrome		3 Cases
2997	Ptosis-vocal cord paralysis syndrome		2 Cases
182090	Pulmonary arterial hypertension	3.3 *P* *	
275803	Pulmonary arterial hypertension associated with congenital heart disease	0.57 *P* *	
275798	Pulmonary arterial hypertension associated with connective tissue disease	0.25 *P* *	
2038	Pulmonary arteriovenous malformation	2.5 *I*	
99050	Pulmonary artery coming from the aorta		200 Cases
64741	Pulmonary blastoma		350 Cases
199241	Pulmonary capillary hemangiomatosis		100 Cases
210136	Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome		4 Cases
217080	Pulmonary fungal infections in patients deemed at risk	22.0 *P* *	

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411703	Pulmonary non-tuberculous mycobacterial infection	6.0 *P* *	
31837	Pulmonary venoocclusive disease	0.015 *I* *	
79501	Punctate palmoplantar keratoderma type 1		35 Families
79502	Punctate palmoplantar keratoderma type 2		13 Cases
438213	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome		24 Cases
438216	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation		24 Cases
69084	Pure hair and nail ectodermal dysplasia		20 Cases
760	Purine nucleoside phosphorylase deficiency		70 Cases
763	Pycnodysostosis	0.13 *P*	
481152	PYCR2-related microcephaly-progressive leukoencephalopathy		18 Cases
3003	Pyknoachondrogenesis		5 Cases
3005	Pyle disease		30 Cases
48104	Pyoderma gangrenosum	0.74 *I*	
69126	Pyogenic arthritis-pyoderma gangrenosum-acne syndrome		34 Cases
183713	Pyogenic bacterial infections due to MyD88 deficiency		24 Cases
2561	Pyramidal molar-glaucoma-upper abnormal lip syndrome		8 Cases
79096	Pyridoxal phosphate-responsive seizures	0.2 *P* *	
3006	Pyridoxine-dependent epilepsy	0.2 *BP* *	
3008	Pyruvate carboxylase deficiency	0.4 *BP* *	
353320	Pyruvate carboxylase deficiency, benign type		5 Cases
2394	Pyruvate dehydrogenase E3 deficiency		20 Cases
781	Q fever	0.19 *I* *	
3010	Qazi-Markouizos syndrome		3 Cases
2252	Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome		8 Cases
3026	Radial ray hypoplasia-choanal atresia syndrome		3 Cases
70475	Radiation proctitis	35.0 *P* *	
3015	Radio-renal syndrome		4 Cases
71289	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome		20 Cases
3270	Radioulnar synostosis-developmental delay-hypotonia syndrome		4 Cases
3268	Radioulnar synostosis-microcephaly-scoliosis syndrome		13 Cases
3019	Ramon syndrome		8 Cases
1051	Ramos-Arroyo syndrome		6 Cases
3021	RAPADILINO syndrome		20 Cases
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome		48 Cases
71517	Rapid-onset dystonia-parkinsonism		100 Cases
213528	Rare adenocarcinoma of the breast	3.55 *I* *	
217074	Rare carcinoma of pancreas	3.5 *P*	
217074	Rare carcinoma of pancreas	3.9 *I*	
88991	Rare congenital non-syndromic heart malformation	7.8 *BP* *	
535	Rare cutaneous lupus erythematosus	50.0 *P* *	
63443	Rare epithelial tumor of stomach	18.6 *I* *	
2415	Rare lymphatic malformation	12.5 *P* *	
182114	Rare urogenital tumor	0.13 *I* *	
438114	RARS-related autosomal recessive hypomyelinating leukodystrophy		4 Cases
268114	RAS-associated autoimmune leukoproliferative disease		20 Cases
1929	Rasmussen subacute encephalitis		100 Cases
99852	Ravine syndrome		38 Cases
1115	Recessive aplasia cutis congenita of limbs		6 Cases
79409	Recessive dystrophic epidermolysis bullosa inversa		100 Cases
280384	Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome		12 Cases
461	Recessive X-linked ichthyosis	16.6 *P* *	
461	Recessive X-linked ichthyosis	15.0 *I* *	
64740	Recurrent acute pancreatitis	10.0 *P* *	
90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients	7.0 *P* *	
480864	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome		24 Cases
97239	Reducing body myopathy		4 Families
86839	Refractory anemia with excess blasts	0.15 *I* *	
168960	Refractory anemia with excess blasts in transformation	0.04 *I* *	
773	Refsum disease	0.1 *P* *	
83450	Regional odontodysplasia		140 Cases
448267	Regressive spondylometaphyseal dysplasia		2 Cases

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98961	Reis-Bücklers corneal dystrophy		81 Cases
728	Relapsing polychondritis	0.35 *I*	
217330	REN-related autosomal dominant tubulointerstitial kidney disease		21 Cases
1848	Renal agenesis, bilateral	1.7 *BP* *	
93100	Renal agenesis, unilateral	50.0 *BP*	
2838	Renal caliceal diverticuli-deafness syndrome		4 Cases
217071	Renal cell carcinoma	42.0 *P* *	
217071	Renal cell carcinoma	8.35 *I* *	
1475	Renal coloboma syndrome		180 Cases
93108	Renal dysplasia	43.5 *BP* *	
1092	Renal-genital-middle ear anomalies		2 Families
93975	Renier-Gabreels-Jasper syndrome		5 Cases
3242	Renpenning syndrome		64 Cases
494344	RERE-related neurodevelopmental syndrome		10 Cases
99832	Resistance to thyrotropin-releasing hormone syndrome		2 Cases
1662	Restrictive dermopathy		30 Cases
33355	Reticular dysgenesis	0.03 *I* *	
178307	Reticulate acropigmentation of Kitamura		130 Cases
458763	Retiform hemangioendothelioma		32 Cases
75326	Retinal arterial tortuosity		100 Cases
1574	Retinal degeneration-nanophthalmos-glaucoma syndrome		7 Cases
397758	Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies		14 Cases
3018	Retinal ischemic syndrome-digestive tract small vessel hyalinosis-diffuse cerebral calcifications syndrome		3 Cases
319640	Retinal macular dystrophy type 2		5 Families
791	Retinitis pigmentosa	26.7 *P*	
494439	Retinitis pigmentosa-hearing loss-premature aging-short stature-facial dysmorphism syndrome		3 Cases
3085	Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome		2 Families
436245	Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome		3 Cases
790	Retinoblastoma	6.0 *BP*	
790	Retinoblastoma	0.05 *I* *	
3087	Retinohepatoendocrinologic syndrome		7 Cases
778	Rett syndrome	10.0 *P* *	
778	Rett syndrome	5.0 *BP* *	
294049	Reunion Island Larsen syndrome		30 Cases
3088	Revesz syndrome		4 Cases
244310	RFT1-CDG		8 Cases
69077	Rhabdoid tumor		500 Cases
780	Rhabdomyosarcoma	0.59 *I* *	
3099	Rheumatic fever	5.0 *I* *	
85408	Rheumatoid factor-negative juvenile idiopathic arthritis	8.0 *P* *	
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	4.2 *P* *	
177	Rhizomelic chondrodysplasia punctata	1.0 *P* *	
468717	Rhizomelic chondrodysplasia punctata type 5		4 Cases
2831	Rhizomelic dysplasia, Patterson-Lowry type		5 Cases
3098	Rhizomelic syndrome, Urbach type		3 Cases
59315	Rhombencephalosynapsis		100 Cases
140976	RHYNS syndrome		4 Cases
97229	Riboflavin transporter deficiency		80 Cases
440706	Ribose-5-P isomerase deficiency		1 Case
3101	Richieri Costa-da Silva syndrome		4 Cases
3102	Richieri Costa-Pereira syndrome		33 Cases
83312	Rickettsialpox		800 Cases
420741	RIDDLE syndrome		2 Cases
217335	RIN2 syndrome		10 Cases
363203	Ring chromosome	2.0 *BP*	
1437	Ring chromosome 1 syndrome		35 Cases
96171	Ring chromosome 2 syndrome		18 Cases
96172	Ring chromosome 3 syndrome		11 Cases
1447	Ring chromosome 4 syndrome		20 Cases
1448	Ring chromosome 6 syndrome		25 Cases
1449	Ring chromosome 7 syndrome		18 Cases
1450	Ring chromosome 8 syndrome		8 Cases
96173	Ring chromosome 9 syndrome		31 Cases
1438	Ring chromosome 10 syndrome		16 Cases
96175	Ring chromosome 11 syndrome		20 Cases
1439	Ring chromosome 12 syndrome		10 Cases
1440	Ring chromosome 14 syndrome		80 Cases
96177	Ring chromosome 15 syndrome		50 Cases
96178	Ring chromosome 16 syndrome		10 Cases

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1441	Ring chromosome 17 syndrome		18 Cases
1442	Ring chromosome 18 syndrome		70 Cases
1443	Ring chromosome 19 syndrome		10 Cases
1444	Ring chromosome 20 syndrome		50 Cases
1446	Ring chromosome 22 syndrome		100 Cases
91481	Ring dermoid of cornea		30 Cases
3103	Roberts syndrome		150 Cases
3104	Robin sequence-oligodactyly syndrome		3 Cases
97360	Robinow syndrome		200 Cases
3105	Robinow-like syndrome		2 Cases
353298	Roifman syndrome		7 Cases
163727	Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome		1 Family
101016	Romano-Ward syndrome	40.0 *P* *	
158014	Rosaï-Dorfman disease		1000 Cases
2909	Rothmund-Thomson syndrome		300 Cases
221008	Rothmund-Thomson syndrome type 1		100 Cases
221016	Rothmund-Thomson syndrome type 2		200 Cases
3111	Rotor syndrome		50 Cases
83616	Rubella panencephalitis		20 Cases
783	Rubinstein-Taybi syndrome	0.7 *BP* *	
397927	Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome		4 Cases
794	Saethre-Chotzen syndrome	3.0 *BP* *	
300493	Sagliker syndrome		60 Cases
140969	Saldino-Mainzer syndrome		10 Cases
213557	Salivary gland type cancer of the breast	0.05 *I* *	
370938	Salt-and-pepper syndrome		3 Cases
796	Sandhoff disease	0.67 *BP* *	
79269	Sanfilippo syndrome type A	0.32 *P* *	
79269	Sanfilippo syndrome type A	1.4 *BP*	
79270	Sanfilippo syndrome type B	0.2 *P* *	
79271	Sanfilippo syndrome type C	5.0 *P* *	
797	Sarcoidosis	12.5 *P*	
3129	Sarcosinemia	2.0 *BP*	
3130	Satoyoshi syndrome		50 Cases
3132	Say-Barber-Miller syndrome		2 Cases
3133	Say-Field-Coldwell syndrome		4 Cases
1003	Scalp defects-postaxial polydactyly syndrome		2 Cases
370052	SCALP syndrome		4 Cases
2036	Scalp-ear-nipple syndrome		30 Cases
431255	Scapuloperoneal spinal muscular atrophy		31 Cases
3134	SCARF syndrome		2 Cases
90080	Scarring in glaucoma filtration surgical procedures	22.0 *P* *	
2353	Schilbach-Rott syndrome		18 Cases
1830	Schimke immuno-osseous dysplasia		71 Cases
798	Schinzel-Giedion syndrome		46 Cases
37748	Schnitzler syndrome		150 Cases
98967	Schnyder corneal dystrophy		115 Cases
50944	Schöpf-Schulz-Passarge syndrome		25 Cases
800	Schwartz-Jampel syndrome		129 Cases
185	Scimitar syndrome	2.0 *BP* *	
801	Scleroderma	42.0 *P*	
801	Scleroderma	1.41 *I*	
167635	Scleromyxedema		250 Cases
90400	Scleromyxedema without monoclonal gammopathy		15 Cases
3152	Sclerosteosis		80 Cases
806	Scott syndrome		4 Cases
158029	Sea-blue histiocytosis		60 Cases
168606	Seborrhea-like dermatitis with psoriasiform elements		44 Cases
808	Seckel syndrome	0.2 *BP* *	
67039	Segmental odontomaxillary dysplasia		32 Cases
314662	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia		10 Cases
79156	Seizures-intellectual disability due to hydroxylysinuria syndrome		3 Cases
466926	Seizures-scoliosis-macrocephaly syndrome		4 Cases
281122	Self-improving collodion baby		25 Cases
3156	Senior-Loken syndrome		150 Cases
217622	Sensorineural deafness with dilated cardiomyopathy		2 Families
66633	Sensorineural hearing loss-early graying-essential tremor syndrome		3 Cases
90051	Sepsis in premature infants	32.0 *P* *	
3157	Septo-optic dysplasia spectrum	10.0 *BP* *	
139466	SERKAL syndrome		3 Cases

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85165	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome		5 Cases
438207	Severe autosomal recessive macrothrombocytopenia		2 Cases
183660	Severe combined immunodeficiency	1.65 *BP* *	
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	0.2 *P* *	
277	Severe combined immunodeficiency due to adenosine deaminase deficiency	0.3 *BP* *	
357237	Severe combined immunodeficiency due to CARD11 deficiency		3 Cases
331206	Severe combined immunodeficiency due to complete RAG1/2 deficiency	1.0 *P* *	
228003	Severe combined immunodeficiency due to CORO1A deficiency		6 Cases
420573	Severe combined immunodeficiency due to CTPS1 deficiency		8 Cases
317425	Severe combined immunodeficiency due to DNA-PKcs deficiency		2 Cases
397787	Severe combined immunodeficiency due to IKK2 deficiency		9 Cases
504523	Severe combined immunodeficiency due to LAT deficiency		3 Cases
280142	Severe combined immunodeficiency due to LCK deficiency		4 Cases
300298	Severe congenital hypochromic anemia with ringed sideroblasts		3 Cases
42738	Severe congenital neutropenia	0.07 *P*	
42738	Severe congenital neutropenia	0.4 *BP* *	
369992	Severe dermatitis-multiple allergies-metabolic wasting syndrome		3 Cases
329249	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency		13 Cases
352577	Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome		28 Cases
488627	Severe growth deficiency-strabismus-extensive dermal melanocytosis-intellectual disability syndrome		3 Cases
169802	Severe hemophilia A	2.8 *P* *	
169793	Severe hemophilia B	0.8 *P* *	
745	Severe hereditary thrombophilia due to congenital protein C deficiency	0.16 *BP*	
467176	Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome		6 Cases
280763	Severe intellectual disability and progressive spastic paraplegia		15 Cases
466688	Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome		6 Cases
94066	Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia		2 Cases
438178	Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency		3 Cases
436141	Severe intellectual disability-hypotonia-strabismus-coarse face-planovalgus syndrome		6 Cases
363686	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome		4 Cases
397933	Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome		3 Cases
404473	Severe intellectual disability-progressive spastic diplegia syndrome		4 Cases
391307	Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome		3 Cases
324307	Severe lateral tibial bowing with short stature		2 Cases
1236	Severe microbrachycephaly-intellectual disability-athetoid cerebral palsy syndrome		2 Cases
369939	Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome		7 Cases
314655	Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion		7 Cases
397593	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency		5 Cases
209370	Severe neonatal-onset encephalopathy with microcephaly		30 Cases
363400	Severe neurodegenerative syndrome with lipodystrophy		10 Cases
500545	Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract		6 Cases

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3078	Severe X-linked intellectual disability, Gustavson type		7 Cases
238329	Severe X-linked mitochondrial encephalomyopathy		2 Cases
363489	Sex cord-stromal tumor of testis	0.02 *I* *	
810	Shigellosis	1.59 *I* *	
99063	Shone complex		100 Cases
104008	Short bowel syndrome	3.4 *P* *	
66518	Short fifth metacarpals-insulin resistance syndrome		6 Cases
498497	Short rib-polydactyly syndrome type 5		2 Cases
93269	Short rib-polydactyly syndrome, Majewski type		34 Cases
314811	Short stature due to GHSR deficiency		8 Cases
629	Short stature due to growth hormone qualitative anomaly		3 Cases
2867	Short stature, Brussels type		2 Cases
435804	Short stature-advanced bone age-early-onset osteoarthritis syndrome		3 Families
397623	Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome		4 Cases
464288	Short stature-brachydactyly-obesity-global developmental delay syndrome		6 Cases
2994	Short stature-craniofacial anomalies-genital hypoplasia syndrome		3 Families
2866	Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome		2 Cases
314394	Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome		14 Cases
391677	Short stature-optic atrophy-Pelger-Huët anomaly syndrome		34 Cases
85442	Short stature-pituitary and cerebellar defects-small sella turcica syndrome		5 Families
2868	Short stature-valvular heart disease-characteristic facies syndrome		3 Cases
2865	Short stature-webbed neck-heart disease syndrome		4 Cases
2863	Short stature-wormian bones-dextrocardia syndrome		3 Cases
3163	SHORT syndrome		32 Cases
2832	Short tarsus-absence of lower eyelashes syndrome		11 Cases
357175	Short ulna-dysmorphism-hypotonia-intellectual disability syndrome		4 Cases
935	Short-limb skeletal dysplasia with severe combined immunodeficiency		19 Cases
2462	Shprintzen-Goldberg syndrome		60 Cases
811	Shwachman-Diamond syndrome	0.28 *P*	
811	Shwachman-Diamond syndrome	0.5 *BP*	
309294	Sialidosis	0.05 *BP* *	
3166	Sialuria		5 Cases
232	Sickle cell anemia	22.0 *P* *	
3167	Siegler-Brewer-Carey syndrome		2 Cases
71276	Silent sinus syndrome		98 Cases
3168	Sillence syndrome		5 Cases
813	Silver-Russell syndrome	0.7 *BP* *	
813	Silver-Russell syndrome	15.5 *I* *	
397590	Silver-Russell syndrome due to a point mutation		8 Cases
373	Simpson-Golabi-Behmel syndrome		250 Cases
79022	Simpson-Golabi-Behmel syndrome type 2		9 Cases
500163	SIN3A-related intellectual disability syndrome		15 Cases
85191	Singleton-Merten dysplasia		22 Cases
324321	Sinoatrial node dysfunction and deafness		8 Cases
3169	Sirenomelia	0.01 *P*	
3169	Sirenomelia	0.98 *BP*	
2882	Sitosterolemia		100 Cases
488437	SIX2-related frontonasal dysplasia		1 Family
1858	Skeletal dysplasia-epilepsy-short stature syndrome		7 Cases
477831	Skeletal overgrowth-craniofacial dysmorphism-hyperelastic skin-white matter lesions syndrome		2 Cases
293165	Skin fragility-woolly hair-palmoplantar keratoderma syndrome		7 Cases
238459	SLC35A1-CDG		1 Case
356961	SLC35A2-CDG		4 Cases
468699	SLC39A8-CDG		10 Cases
70573	Small cell lung cancer	11.2 *P* *	
466962	SMARCA4-deficient sarcoma of thorax		19 Cases
93974	Smith-Fineman-Myers syndrome		11 Families
818	Smith-Lemli-Opitz syndrome	3.7 *BP* *	
819	Smith-Magenis syndrome	4.0 *P*	
178355	Smith-McCort dysplasia		12 Families
820	Sneddon syndrome	0.4 *I* *	

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91496	Snowflake vitreoretinal degeneration		50 Cases
306577	Sodium channelopathy-related small fiber neuropathy		8 Cases
3394	Soft tissue sarcoma	30.0 *P* *	
3394	Soft tissue sarcoma	4.74 *I* *	
97230	Solar urticaria	36.0 *P* *	
209964	Solitary rectal ulcer syndrome	1.0 *I* *	
97283	Somatostatinoma	0.0025 *I* *	
821	Sotos syndrome	7.1 *BP*	
79132	Sparse hair-short stature-skin anomalies syndrome		4 Cases
1182	Spastic ataxia with congenital miosis		3 Families
2572	Spastic ataxia-corneal dystrophy syndrome		1 Family
2815	Spastic paraparesis-deafness syndrome		6 Cases
99015	Spastic paraplegia type 2		100 Cases
99013	Spastic paraplegia type 7	4.0 *P* *	
2816	Spastic paraplegia-epilepsy-intellectual disability syndrome		5 Cases
2819	Spastic paraplegia-facial-cutaneous lesions syndrome		5 Cases
2818	Spastic paraplegia-glaucoma-intellectual disability syndrome		2 Families
2820	Spastic paraplegia-nephritis-deafness syndrome		4 Cases
2821	Spastic paraplegia-neuropathy-poikiloderma syndrome		1 Family
320406	Spastic paraplegia-optic atrophy-neuropathy syndrome		75 Cases
329475	Spastic paraplegia-Paget disease of bone syndrome		1 Family
2826	Spastic paraplegia-precocious puberty syndrome		2 Cases
464282	Spastic paraplegia-severe developmental delay-epilepsy syndrome		16 Cases
3011	Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome		2 Cases
447997	Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome		15 Cases
352403	Spectrin-associated autosomal recessive cerebellar ataxia		2 Families
99865	Spermatocytic seminoma	0.03 *I* *	
314432	Spigelian hernia-cryptorchidism syndrome		15 Cases
1217	Spinal atrophy-ophthalmoplegia-pyramidal syndrome		2 Cases
90058	Spinal cord injury	32.0 *P* *	
98920	Spinal muscular atrophy with respiratory distress type 1		128 Cases
404521	Spinal muscular atrophy with respiratory distress type 2		1 Case
73245	Spinal muscular atrophy-Dandy-Walker malformation-cataracts syndrome		2 Cases
2590	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome		10 Cases
98755	Spinocerebellar ataxia type 1	1.5 *P*	
98756	Spinocerebellar ataxia type 2	1.5 *P*	
98757	Spinocerebellar ataxia type 3	1.5 *P*	
98766	Spinocerebellar ataxia type 5		3 Families
98767	Spinocerebellar ataxia type 11		51 Cases
98762	Spinocerebellar ataxia type 12		40 Families
98768	Spinocerebellar ataxia type 13		20 Cases
98763	Spinocerebellar ataxia type 14		20 Families
98769	Spinocerebellar ataxia type 15/16		80 Cases
98759	Spinocerebellar ataxia type 17		100 Families
98771	Spinocerebellar ataxia type 18		26 Cases
98772	Spinocerebellar ataxia type 19/22		12 Cases
101110	Spinocerebellar ataxia type 20		20 Cases
98773	Spinocerebellar ataxia type 21		35 Cases
101108	Spinocerebellar ataxia type 23		4 Families
101111	Spinocerebellar ataxia type 25		10 Cases
101112	Spinocerebellar ataxia type 26		1 Family
98764	Spinocerebellar ataxia type 27		30 Cases
208513	Spinocerebellar ataxia type 29		50 Cases
211017	Spinocerebellar ataxia type 30		6 Cases
217012	Spinocerebellar ataxia type 31		30 Families
276183	Spinocerebellar ataxia type 32		1 Family
1955	Spinocerebellar ataxia type 34		27 Cases
276193	Spinocerebellar ataxia type 35		3 Families
276198	Spinocerebellar ataxia type 36		90 Cases
363710	Spinocerebellar ataxia type 37		9 Cases
423296	Spinocerebellar ataxia type 38		4 Families
423275	Spinocerebellar ataxia type 40		5 Cases
458798	Spinocerebellar ataxia type 41		1 Case
458803	Spinocerebellar ataxia type 42		25 Cases
497764	Spinocerebellar ataxia type 43		7 Cases
94124	Spinocerebellar ataxia with axonal neuropathy type 1		9 Cases
1185	Spinocerebellar ataxia-dysmorphism syndrome		3 Cases
2063	Splenogonadal fusion-limb defects-micrognathia syndrome		30 Cases

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71271	Split hand-split foot-deafness syndrome		22 Cases
488232	Split-foot malformation-mesoaxial polydactyly syndrome		5 Cases
93357	SPONASTRIME dysplasia		16 Cases
228387	Spondylo-megaepiphyseal-metaphyseal dysplasia		19 Cases
85194	Spondylo-ocular syndrome		7 Cases
3180	Spondylocamptodactyly syndrome		5 Cases
3275	Spondylocarpotarsal synostosis		24 Cases
94095	Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome		3 Cases
329252	Spondylocostal dysostosis-hypospadias-intellectual disability syndrome		2 Cases
1855	Spondyloenchondrodysplasia		36 Cases
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type		30 Cases
171866	Spondyloepimetaphyseal dysplasia, aggrecan type		3 Cases
168448	Spondyloepimetaphyseal dysplasia, Bieganski type		7 Cases
168454	Spondyloepimetaphyseal dysplasia, Geneviève type		6 Families
99642	Spondyloepimetaphyseal dysplasia, Handigodu type		234 Cases
370015	Spondyloepimetaphyseal dysplasia, Isidor type		2 Cases
156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type		1 Family
93356	Spondyloepimetaphyseal dysplasia, Missouri type		14 Cases
93282	Spondyloepimetaphyseal dysplasia, PAPSS2 type		17 Cases
93352	Spondyloepimetaphyseal dysplasia, Shohat type		4 Cases
168451	Spondyloepimetaphyseal dysplasia-abnormal dentition syndrome		2 Cases
168443	Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome		5 Cases
93358	Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome		27 Cases
94068	Spondyloepiphyseal dysplasia congenita	1.0 *BP* *	
163665	Spondyloepiphyseal dysplasia tarda, Kohn type		3 Cases
163654	Spondyloepiphyseal dysplasia, Cantu type		4 Cases
93283	Spondyloepiphyseal dysplasia, Kimberley type		1 Family
163668	Spondyloepiphyseal dysplasia, MacDermot type		4 Cases
263482	Spondyloepiphyseal dysplasia, Maroteaux type		10 Cases
163649	Spondyloepiphyseal dysplasia, Nishimura type		4 Cases
163662	Spondyloepiphyseal dysplasia, Reardon type		1 Family
459051	Spondyloepiphyseal dysplasia, Stanescu type		7 Cases
254	Spondylometaphyseal dysplasia	1.0 *BP* *	
168555	Spondylometaphyseal dysplasia, A4 type		3 Cases
93315	Spondylometaphyseal dysplasia, 'corner fracture' type		30 Cases
370019	Spondylometaphyseal dysplasia, Czarny-Ratajczak type		2 Cases
168544	Spondylometaphyseal dysplasia, Golden type		3 Cases
93316	Spondylometaphyseal dysplasia, Schmidt type		7 Cases
93317	Spondylometaphyseal dysplasia, Sedaghatian type		9 Cases
168552	Spondylometaphyseal dysplasia-bowed forearms-facial dysmorphism syndrome		2 Cases
85167	Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome		18 Cases
1856	Spondyloperipheral dysplasia-short ulna syndrome		10 Families
29822	Spontaneous periodic hypothermia		50 Cases
247234	Sporadic adult-onset ataxia of unknown etiology	7.6 *P* *	
204	Sporadic Creutzfeldt-Jakob disease	0.1 *P* *	
204	Sporadic Creutzfeldt-Jakob disease	0.15 *I*	
424996	Squamous cell carcinoma of gallbladder and extrahepatic biliary tract	0.04 *I* *	
424975	Squamous cell carcinoma of liver and intrahepatic biliary tract	0.01 *I* *	
424019	Squamous cell carcinoma of the anal canal	0.73 *I* *	
213767	Squamous cell carcinoma of the cervix uteri	4.28 *I* *	
423994	Squamous cell carcinoma of the colon	0.02 *I* *	
213716	Squamous cell carcinoma of the corpus uteri	0.12 *I* *	
99977	Squamous cell carcinoma of the esophagus	5.2 *I*	
494547	Squamous cell carcinoma of the hypopharynx	1.27 *I* *	
494550	Squamous cell carcinoma of the larynx	4.61 *I* *	
502366	Squamous cell carcinoma of the lip	1.02 *I*	
500464	Squamous cell carcinoma of the nasal cavity and paranasal sinuses	0.35 *I*	

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502363	Squamous cell carcinoma of the oral cavity	3.51 *I* *	
500478	Squamous cell carcinoma of the oropharynx	3.12 *I* *	
424039	Squamous cell carcinoma of the pancreas	0.03 *I* *	
398058	Squamous cell carcinoma of the penis	0.57 *I* *	
424002	Squamous cell carcinoma of the rectum	0.07 *I* *	
423968	Squamous cell carcinoma of the small intestine	0.01 *I* *	
418959	Squamous cell carcinoma of the stomach	0.13 *I* *	
324737	SRD5A3-CDG		7 Families
370927	SSR4-CDG		9 Cases
83484	St. Louis encephalitis	0.38 *I* *	
502434	STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome		17 Cases
140917	Stapes ankylosis with broad thumbs and toes		6 Families
827	Stargardt disease	10.0 *P* *	
438159	STAT3-related early-onset multisystem autoimmune disease		19 Cases
438117	Steel syndrome		40 Cases
273	Steinert myotonic dystrophy	12.5 *P*	
210115	Sterile multifocal osteomyelitis with periostitis and pustulosis		17 Cases
2017	Sternal cleft	2.0 *BP* *	
3196	Steroid dehydrogenase deficiency-dental anomalies syndrome		1 Family
36426	Stevens-Johnson syndrome	0.36 *I* *	
95455	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	0.19 *I*	
828	Stickler syndrome	12.2 *BP*	
2833	Stiff skin syndrome		54 Cases
3199	Stimmler syndrome		2 Cases
425120	STING-associated vasculopathy with onset in infancy		9 Cases
3204	Stormorken-Sjaastad-Langslet syndrome		17 Cases
137599	Stromal keratitis	16.0 *P* *	
370921	STT3A-CDG		2 Cases
370924	STT3B-CDG		1 Case
3205	Sturge-Weber syndrome	3.5 *BP* *	
3206	Stüve-Wiedemann syndrome		56 Cases
3191	Subaortic stenosis-short stature syndrome		1 Family
48377	Subcorneal pustular dermatosis		200 Cases
98959	Subepithelial mucinous corneal dystrophy		1 Family
22	Succinic semialdehyde dehydrogenase deficiency		450 Cases
832	Succinyl-CoA:3-ketoacid CoA transferase deficiency		33 Cases
168593	Sudden infant death-dysgenesis of the testes syndrome		21 Cases
498602	Sugarman brachydactyly		1 Family
3210	Summitt syndrome		3 Cases
57145	SUNCT syndrome	6.7 *P* *	
455	Superficial epidermolytic ichthyosis		20 Cases
46485	Superficial pemphigus	1.2 *P* *	
247245	Superficial siderosis		300 Cases
141096	Supernumerary nostril		32 Cases
466695	Supratip dysplasia		5 Cases
3193	Supravalvular aortic stenosis	13.3 *P* *	
3193	Supravalvular aortic stenosis	4.0 *BP* *	
391351	SURF1-related Charcot-Marie-Tooth disease type 4		3 Cases
838	Susac syndrome		304 Cases
331226	Susceptibility to infection due to TYK2 deficiency		8 Cases
3243	Sweet syndrome		100 Cases
1570	Symbrachydactyly of hands and feet		2 Cases
1314	Symmetrical thalamic calcifications		30 Cases
79098	Sympathetic ophthalmia	0.6 *P* *	
3246	Symphalangism with multiple anomalies of hands and feet		6 Cases
93402	Syndactyly type 1	25.0 *BP* *	
93405	Syndactyly type 4		4 Cases
93406	Syndactyly type 5		10 Cases
357332	Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome		26 Cases
3259	Syndactyly-polydactyly-ear lobe syndrome		10 Cases
140952	Syndactyly-telecanthus-anogenital and renal malformations syndrome		6 Cases
84064	Syndromic diarrhea		44 Cases
178364	Syndromic microphthalmia type 5		20 Cases
228426	Syndromic multisystem autoimmune disease due to Itch deficiency		10 Cases
98606	Syndromic orbital border hypoplasia		2 Families
281090	Syndromic recessive X-linked ichthyosis	1.3 *P* *	

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457223	Syndromic sensorineural deafness due to combined oxidative phosphorylation defect		2 Cases
85274	Syndromic X-linked intellectual disability 7		10 Cases
85279	Syndromic X-linked intellectual disability due to JARID1C mutation		10 Families
840	Syringocystadenoma papilliferum		300 Cases
3280	Syringomyelia	8.4 *P* *	
188	Systemic capillary leak syndrome		150 Cases
2467	Systemic mastocytosis	3.75 *P*	
2467	Systemic mastocytosis	0.9 *I* *	
158	Systemic primary carnitine deficiency	3.2 *BP* *	
90291	Systemic sclerosis	15.4 *P* *	
85414	Systemic-onset juvenile idiopathic arthritis	5.0 *P* *	
169157	T-B+ severe combined immunodeficiency due to CD45 deficiency		3 Cases
324294	T-cell immunodeficiency with epidermodysplasia verruciformis		2 Cases
86872	T-cell large granular lymphocyte leukemia	0.4 *I* *	
171918	T-cell non-Hodgkin lymphoma	0.99 *I* *	
457077	TAFRO syndrome		28 Cases
3287	Takayasu arteritis	1.34 *P* *	
3287	Takayasu arteritis	0.084 *I* *	
404443	Tall stature-intellectual disability-facial dysmorphism syndrome		13 Cases
500095	Tall stature-intellectual disability-renal anomalies syndrome		4 Cases
329191	Tall stature-scoliosis-macrodactyly of the great toes syndrome		2 Families
50809	Talo-patello-scaphoid osteolysis		2 Cases
31150	Tangier disease		100 Cases
2886	TARP syndrome		6 Families
1412	Tarsal-carpal coalition syndrome		10 Families
2731	Taurodontia-absent teeth-sparse hair syndrome		15 Cases
845	Tay-Sachs disease	0.28 *BP*	
488632	TBCK-related intellectual disability syndrome		25 Cases
397959	TCR-alpha-beta-positive T-cell deficiency		2 Cases
3291	Teebi-Shaltout syndrome		5 Cases
3293	Telecanthus-hypertelorism-strabismus-pes cavus syndrome		2 Cases
488642	TELO2-related intellectual disability-neurodevelopmental disorder		6 Cases
352737	Temperature-sensitive oculocutaneous albinism type 1		10 Cases
284227	TEMPI syndrome		10 Cases
420561	Temple-Baraitser syndrome		7 Cases
363417	Temtamy preaxial brachydactyly syndrome		18 Cases
1777	Temtamy syndrome		7 Families
88630	Terminal osseous dysplasia-pigmentary defects syndrome		23 Cases
141258	Tessier number 4 facial cleft		2 Cases
842	Testicular seminomatous germ cell tumor	1.71 *I* *	
3299	Tetanus	0.04 *I* *	
3303	Tetralogy of Fallot	34.0 *BP*	
3310	Tetrasomy 9p		70 Cases
884	Tetrasomy 12p	4.0 *BP* *	
96055	Tetrasomy 21		13 Cases
9	Tetrasomy X		50 Cases
1780	Thakker-Donnai syndrome		2 Cases
3312	Thalidomide embryopathy	0.77 *P*	
2655	Thanatophoric dysplasia	3.5 *BP* *	
199348	Thiamine-responsive encephalopathy		2 Cases
49827	Thiamine-responsive megaloblastic anemia syndrome		80 Cases
2405	Thickened earlobes-conductive deafness syndrome		2 Families
98960	Thiel-Behnke corneal dystrophy		173 Cases
1506	Thin ribs-tubular bones-dysmorphism syndrome		2 Cases
363444	THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome		4 Cases
3316	Thomas syndrome		6 Cases
614	Thomsen and Becker disease	1.0 *P*	
1861	Thoracic dysplasia-hydrocephalus syndrome		2 Cases
3317	Thoracolaryngopelvic dysplasia		10 Cases
1803	Thoracomelic dysplasia		2 Cases
329319	Thrombocythemia with distal limb defects		3 Families
67044	Thrombocytopenia with congenital dyserythropoietic anemia		3 Families
3320	Thrombocytopenia-absent radius syndrome	0.5 *BP* *	
3323	Thrombocytopenia-Robin sequence syndrome		2 Cases
436169	Thrombomodulin-related bleeding disorder		15 Cases

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54057	Thrombotic thrombocytopenic purpura	25.5 *P* *	
2251	Thumb deformity-alopecia-pigmentation anomaly syndrome		1 Family
1078	Thumb stiffness-brachydactyly-intellectual disability syndrome		6 Cases
3398	Thymic epithelial neoplasm	0.17 *I* *	
3326	Thymic-renal-anal-lung dysplasia		3 Cases
99867	Thymoma	0.14 *I* *	
3327	Thyrocerebrorenal syndrome		2 Cases
100088	Thyroid carcinoma	12.7 *P*	
100088	Thyroid carcinoma	3.1 *I*	
95712	Thyroid ectopia	14.3 *P* *	
95719	Thyroid hemiagenesis	25.0 *P*	
95720	Thyroid hypoplasia	3.5 *P*	
100087	Thyroid tumor	3.2 *I*	
3329	Tibial aplasia-ectrodactyly syndrome	0.1 *P* *	
93322	Tibial hemimelia	0.1 *BP* *	
609	Tibial muscular dystrophy	6.0 *P* *	
42665	Tietz syndrome		2 Families
65283	Timothy syndrome		20 Cases
314667	TMEM165-CDG		6 Cases
466703	TMEM199-CDG		7 Cases
3460	Torg-Winchester syndrome		12 Cases
3338	Toriello-Carey syndrome		59 Cases
3339	Toriello-Lacassie-Droste syndrome		19 Cases
3341	Torticollis-keloids-cryptorchidism-renal dysplasia syndrome		7 Cases
227972	Toxic oil syndrome		20000 Cases
3346	Tracheal agenesis	2.0 *BP* *	
3348	Tracheobronchopathia osteochondroplastica		400 Cases
101028	Transaldolase deficiency		23 Cases
859	Transcobalamin deficiency		40 Cases
79411	Transient bullous dermolysis of the newborn		30 Cases
300293	Transient infantile hypertriglyceridemia and hepatosteatosis		11 Cases
99886	Transient neonatal diabetes mellitus	0.3 *BP* *	
329942	Transient neonatal multiple acyl-CoA dehydrogenase deficiency		1 Case
488618	Transketolase deficiency		5 Cases
216675	Transposition of the great arteries	31.7 *BP* *	
861	Treacher-Collins syndrome	2.0 *BP* *	
447896	Tremor-ataxia-central hypomyelination syndrome		7 Cases
3350	Tremor-nystagmus-duodenal ulcer syndrome		17 Cases
863	Trichinellosis	0.06 *I* *	
3352	Tricho-dento-osseous syndrome		30 Cases
1264	Tricho-retino-dento-digital syndrome		9 Cases
3351	Trichodental syndrome		5 Families
3353	Trichodermodysplasia-dental alterations syndrome		3 Cases
79129	Trichodysplasia-amelogenesis imperfecta syndrome		1 Family
3361	Trichodysplasia-xeroderma syndrome		1 Family
3363	Trichomegaly-retina pigmentary degeneration-dwarfism syndrome		14 Cases
3355	Trichoodontoonychial dysplasia		4 Cases
77258	Trichorhinophalangeal syndrome type 1 and 3		100 Cases
502	Trichorhinophalangeal syndrome type 2		100 Cases
33364	Trichothiodystrophy	0.12 *BP* *	
1209	Tricuspid atresia	4.2 *BP* *	
3368	Trigonocephaly-bifid nose-acral anomalies syndrome		2 Cases
3365	Trigonocephaly-broad thumbs syndrome		2 Cases
3369	Trigonocephaly-short stature-developmental delay syndrome		3 Cases
868	Triose phosphate-isomerase deficiency		50 Cases
2950	Triphalangeal thumb-polysyndactyly syndrome		15 Families
2947	Triphalangeal thumbs-brachyectrodactyly syndrome		4 Families
869	Triple A syndrome		100 Cases
3376	Triploidy	12.6 *BP* *	
261344	Trisomy 1q		18 Cases
1738	Trisomy 4p		85 Cases
1742	Trisomy 5p		40 Cases
1752	Trisomy 8q		30 Cases
236	Trisomy 9p		150 Cases
171929	Trisomy 10p		50 Cases
1699	Trisomy 12p	2.0 *BP*	
3378	Trisomy 13	3.7 *BP* *	
3380	Trisomy 18	16.7 *BP*	
1715	Trisomy 18p		25 Cases
3375	Trisomy X	42.5 *P* *	
88629	Tritanopia	4.8 *P* *	

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3384	Truncus arteriosus	4.3 *BP*	
3389	Tuberculosis	20.0 *P* *	
3389	Tuberculosis	139.0 *I*	
805	Tuberous sclerosis complex	12.0 *P* *	
805	Tuberous sclerosis complex	10.0 *BP* *	
73224	Tubular renal disease-cardiomyopathy syndrome		2 Cases
467166	Tubulinopathy-associated dysgyria		7 Cases
1063	Tufted angioma		200 Cases
3392	Tularemia	2.0 *P* *	
3392	Tularemia	0.12 *I* *	
32960	Tumor necrosis factor receptor 1 associated periodic syndrome	0.1 *P* *	
182130	Tumor of endocrine glands	64.0 *P* *	
182130	Tumor of endocrine glands	3.75 *I* *	
363472	Tumor of testis and paratestis	3.15 *I* *	
881	Turner syndrome	5.5 *BP* *	
99745	Typhoid	3.0 *I* *	
882	Tyrosinemia type 1	0.9 *BP*	
28378	Tyrosinemia type 2		150 Cases
69723	Tyrosinemia type 3		20 Cases
3403	Uhl anomaly	1.0 *BP*	
3404	Ulbright-Hodes syndrome		3 Cases
2249	Ulna hypoplasia-intellectual disability syndrome		2 Cases
1837	Ulna metaphyseal dysplasia syndrome		3 Cases
1122	Ulnar hypoplasia-split foot syndrome		1 Family
3138	Ulnar-mammary syndrome		117 Cases
52056	Ulnar/fibula ray defect-brachydactyly syndrome		1 Family
3405	Umbilical cord ulceration-intestinal atresia syndrome		55 Cases
167714	Unclassified acute myeloid leukemia	0.49 *I* *	
418951	Undifferentiated carcinoma of esophagus	0.07 *I* *	
424970	Undifferentiated carcinoma of liver and intrahepatic biliary tract	0.02 *I* *	
423786	Undifferentiated carcinoma of stomach	0.17 *I* *	
2023	Undifferentiated pleomorphic sarcoma	0.9 *I* *	
97363	Unilateral multicystic dysplastic kidney	23.2 *BP*	
1464	Univentricular heart	7.5 *BP*	
99069	Univentricular heart with single atrio-ventricular valve		2 Cases
3408	Upington disease		1 Family
2489	Upper limb defect-eye and ear abnormalities syndrome		2 Cases
2497	Upper limb mesomelic dysplasia		4 Cases
3409	Urban-Rogers-Meyer syndrome		3 Cases
94059	Uremic pruritus	35.0 *P* *	
210128	Urocanic aciduria		4 Cases
481665	USP18 deficiency		5 Cases
178338	UV-sensitive syndrome		7 Cases
1473	Uveal coloboma-cleft lip and palate-intellectual disability		12 Cases
39044	Uveal melanoma	0.5 *I* *	
98715	Uveitis	38.0 *P* *	
98715	Uveitis	17.0 *I* *	
3412	VACTERL with hydrocephalus		10 Families
887	VACTERL/VATER association	6.25 *BP* *	
3417	Van den Bosch syndrome		1 Family
2460	Van den Ende-Gupta syndrome		29 Cases
3419	Van Regemorter-Pierquin-Vamos syndrome		3 Cases
314652	Variant ABeta2M amyloidosis		5 Cases
52759	Vasculitis	6.3 *P* *	
404553	Vasculitis due to ADA2 deficiency		48 Cases
3424	Velo-facial-skeletal syndrome		2 Cases
443988	Ventriculomegaly-cystic kidney disease		11 Cases
3429	Verloove Vanhorick-Brubakk syndrome		2 Cases
70476	Vernal keratoconjunctivitis	32.0 *P* *	
493342	Vibratory urticaria		37 Cases
1493	Vici syndrome		50 Cases
228379	Virus-associated trichodysplasia spinulosa		7 Cases
73246	Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome		2 Cases
28	Vitamin B12-responsive methylmalonic acidemia		192 Cases
79310	Vitamin B12-responsive methylmalonic acidemia type cblA		60 Cases
79312	Vitamin B12-unresponsive methylmalonic acidemia type mut-		450 Cases
600	Vocal cord and pharyngeal distal myopathy		78 Cases
3439	Von Voss-Cherstvoy syndrome		10 Cases
903	Von Willebrand disease	12.5 *P*	

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466934	VPS11-related autosomal recessive hypomyelinating leukodystrophy		13 Cases
137583	Vulvar intraepithelial neoplasia	20.0 *P* *	
83453	Vulvovaginal gingival syndrome		380 Cases
2804	W syndrome		6 Cases
3440	Waardenburg syndrome	0.37 *BP* *	
897	Waardenburg-Shah syndrome		100 Cases
466943	WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome		22 Cases
898	Wagner disease		100 Cases
893	WAGR syndrome	0.2 *BP*	
33226	Waldenström macroglobulinemia	0.81 *I* *	
899	Walker-Warburg syndrome	1.65 *BP* *	
280558	Warsaw breakage syndrome		4 Cases
3447	Weaver syndrome		48 Cases
3448	Weaver-Williams syndrome		2 Cases
3449	Weill-Marchesani syndrome	1.0 *P*	
3344	Weismann-Netter syndrome		100 Cases
99971	Well-differentiated liposarcoma	0.51 *I* *	
901	Wells syndrome		200 Cases
902	Werner syndrome	0.5 *P* *	
3451	West syndrome	6.0 *P* *	
3451	West syndrome	3.7 *BP*	
83476	West-Nile encephalitis	0.04 *I* *	
51636	WHIM syndrome		65 Cases
2475	White forelock with malformations		2 Cases
3207	White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome		4 Cases
370131	White platelet syndrome		1 Family
1489	Whooping cough	4.37 *I* *	
3455	Wiedemann-Rautenstrauch syndrome		54 Cases
319182	Wiedemann-Steiner syndrome		18 Cases
85446	Wild type ABeta2M amyloidosis	4.5 *P* *	
330001	Wild type ATTR amyloidosis	30.0 *P* *	
904	Williams syndrome	10.8 *BP*	
905	Wilson disease	3.3 *P*	
905	Wilson disease	2.2 *BP*	
3459	Wilson-Turner syndrome		28 Cases
906	Wiskott-Aldrich syndrome	0.1 *P* *	
1667	Wolcott-Rallison syndrome		60 Cases
280	Wolf-Hirschhorn syndrome	2.0 *BP* *	
3463	Wolfram syndrome	0.13 *P*	
3464	Woodhouse-Sakati syndrome		30 Cases
1409	Woolly hair-hypotrichosis-everted lower lip-outstanding ears syndrome		1 Family
420686	Woolly hair-palmoplantar keratoderma syndrome		8 Cases
166277	Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia		3 Cases
3465	Worster-Drought syndrome	3.7 *P* *	
178475	Wound botulism	0.1 *I* *	
2834	Wrinkly skin syndrome		30 Cases
3466	WT limb-blood syndrome		3 Families
53719	Wyburn-Mason syndrome		90 Cases
448372	X-linked acrogigantism due to Xq26 microduplication		22 Cases
43	X-linked adrenoleukodystrophy	5.0 *BP*	
47	X-linked agammaglobulinemia	0.22 *P*	
391327	X-linked calvarial hyperostosis		1 Family
329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement		27 Cases
596	X-linked centronuclear myopathy	0.2 *P* *	
163961	X-linked cerebral-cerebellar-coloboma syndrome		3 Cases
64747	X-linked Charcot-Marie-Tooth disease	1.6 *P* *	
101076	X-linked Charcot-Marie-Tooth disease type 2		5 Cases
101077	X-linked Charcot-Marie-Tooth disease type 3		4 Families
101078	X-linked Charcot-Marie-Tooth disease type 4		7 Cases
99014	X-linked Charcot-Marie-Tooth disease type 5		9 Cases
352675	X-linked Charcot-Marie-Tooth disease type 6		8 Cases
431140	X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome		1 Family
1497	X-linked complicated corpus callosum dysgenesis		11 Cases
90001	X-linked cone dysfunction syndrome with myopia		10 Families
1661	X-linked corneal dermoid		6 Cases
52503	X-linked creatine transporter deficiency		150 Cases
139557	X-linked distal spinal muscular atrophy type 3		2 Families
35173	X-linked dominant chondrodysplasia punctata	0.25 *BP* *	

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ORPHA Number	Disease or Group of diseases	Estimated prevalence/incidence (/100,000)	Number of published cases or families
163966	X-linked dominant chondrodysplasia, Chassaing-Lacombe type		10 Cases
363727	X-linked dyserythropoetic anemia with abnormal platelets and neutropenia		1 Family
75497	X-linked Ehlers-Danlos syndrome		2 Families
98863	X-linked Emery-Dreifuss muscular dystrophy	1.0 *P*	
98863	X-linked Emery-Dreifuss muscular dystrophy	1.0 *BP*	
293621	X-linked endothelial corneal dystrophy		35 Cases
443197	X-linked erythropoietic protoporphyria		50 Cases
500188	X-linked external auditory canal atresia-dilated internal auditory canal-facial dysmorphism syndrome		4 Cases
480880	X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability		17 Cases
139583	X-linked hereditary sensory and autonomic neuropathy with deafness		5 Families
181	X-linked hypohidrotic ectodermal dysplasia	0.75 *BP* *	
89936	X-linked hypophosphatemia	0.21 *P* *	
89936	X-linked hypophosphatemia	4.45 *BP*	
317476	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia		7 Cases
2571	X-linked immunoneurologic disorder		5 Cases
364028	X-linked intellectual disability due to GRIA3 anomalies		14 Cases
67045	X-linked intellectual disability with isolated growth hormone deficiency		2 Families
85273	X-linked intellectual disability, Abidi type		8 Cases
85276	X-linked intellectual disability, Armfield type		6 Cases
3056	X-linked intellectual disability, Brooks type		9 Cases
85293	X-linked intellectual disability, Cabezas type		24 Families
85277	X-linked intellectual disability, Cantagrel type		30 Cases
163971	X-linked intellectual disability, Cilliers type		4 Cases
93947	X-linked intellectual disability, Golabi-Ito-Hall type		3 Cases
93952	X-linked intellectual disability, Hedera type		9 Cases
85283	X-linked intellectual disability, Miles-Carpenter type		4 Cases
163937	X-linked intellectual disability, Najm type		35 Families
163956	X-linked intellectual disability, Nascimento type		8 Cases
85322	X-linked intellectual disability, Pai type		1 Family
85285	X-linked intellectual disability, Schimke type		4 Cases
85323	X-linked intellectual disability, Seemanova type		4 Cases
85286	X-linked intellectual disability, Shashi type		9 Cases
85324	X-linked intellectual disability, Shrimpton type		3 Cases
85287	X-linked intellectual disability, Siderius type		2 Families
3063	X-linked intellectual disability, Snyder type		21 Cases
85325	X-linked intellectual disability, Stevenson type		4 Cases
85288	X-linked intellectual disability, Stocco Dos Santos type		1 Family
85326	X-linked intellectual disability, Stoll type		4 Cases
163976	X-linked intellectual disability, Van Esch type		7 Cases
85290	X-linked intellectual disability, Wilson type		3 Cases
85327	X-linked intellectual disability-acromegaly-hyperactivity syndrome		2 Cases
85338	X-linked intellectual disability-ataxia-apraxia syndrome		9 Cases
324410	X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome		2 Cases
137831	X-linked intellectual disability-cerebellar hypoplasia syndrome		14 Families
459070	X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome		2 Cases
85330	X-linked intellectual disability-corpus callosum agenesis-spastic quadriparesis syndrome		4 Cases
163979	X-linked intellectual disability-craniofacioskeletal syndrome		9 Cases
85280	X-linked intellectual disability-cubitus valgus-dysmorphism syndrome		5 Cases
1568	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome		10 Cases
2958	X-linked intellectual disability-dysmorphism-cerebral atrophy syndrome		8 Cases
85319	X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome		2 Cases

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480907	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome		14 Cases
85317	X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome		3 Cases
3055	X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome		4 Cases
85329	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome		10 Cases
457260	X-linked intellectual disability-hypotonia-movement disorder syndrome		38 Cases
423479	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome		2 Cases
85320	X-linked intellectual disability-macrocephaly-macroorchidism syndrome		12 Cases
2898	X-linked intellectual disability-plagiocephaly syndrome		2 Cases
85318	X-linked intellectual disability-precocious puberty-obesity syndrome		3 Cases
3077	X-linked intellectual disability-psychosis-macroorchidism syndrome		6 Cases
3052	X-linked intellectual disability-seizures-psoriasis syndrome		4 Cases
457240	X-linked intellectual disability-short stature-overweight syndrome		20 Cases
163982	X-linked intellectual disability-spastic quadriparesis syndrome		9 Cases
482606	X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome		15 Cases
79447	X-linked lethal multiple pterygium syndrome		6 Families
452	X-linked lissencephaly with abnormal genitalia		30 Families
2442	X-linked lymphoproliferative disease	0.05 *P* *	
1131	X-linked mandibulofacial dysostosis		7 Cases
319605	X-linked mendelian susceptibility to mycobacterial diseases		13 Cases
319623	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency		7 Cases
319612	X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency		6 Cases
435938	X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome		3 Cases
25980	X-linked myopathy with excessive autophagy		18 Families
178461	X-linked myopathy with postural muscle atrophy		7 Families
456328	X-linked myotubular myopathy-abnormal genitalia syndrome		4 Cases
85334	X-linked neurodegenerative syndrome, Bertini type		7 Cases
85336	X-linked neurodegenerative syndrome, Hamel type		11 Cases
314978	X-linked non progressive cerebellar ataxia		3 Families
391330	X-linked osteoporosis with fractures		8 Families
363654	X-linked parkinsonism-spasticity syndrome		5 Cases
83648	X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome		1 Family
54	X-linked recessive ocular albinism	0.58 *BP* *	
85453	X-linked reticulate pigmentary disorder		6 Families
1852	X-linked retinal dysplasia		8 Cases
792	X-linked retinoschisis	5.0 *P*	
431272	X-linked scapuloperoneal muscular dystrophy		22 Cases
86788	X-linked severe congenital neutropenia		45 Cases
75563	X-linked sideroblastic anemia		200 Cases
2802	X-linked sideroblastic anemia and spinocerebellar ataxia		5 Families
1436	X-linked skeletal dysplasia-intellectual disability syndrome		4 Cases
100997	X-linked spastic paraplegia type 16		1 Family
171607	X-linked spastic paraplegia type 34		24 Cases
3175	X-linked spasticity-intellectual disability-epilepsy syndrome		6 Cases
85297	X-linked spinocerebellar ataxia type 3		5 Cases
85292	X-linked spinocerebellar ataxia type 4		1 Family
910	Xeroderma pigmentosum	0.23 *BP* *	
90342	Xeroderma pigmentosum variant		50 Cases
220295	Xeroderma pigmentosum-Cockayne syndrome complex		30 Cases
3469	XK aprosencephaly syndrome		10 Cases
261476	Xp21 microdeletion syndrome		100 Cases
314389	Xq12-q13.3 duplication syndrome		3 Cases
261483	Xq27.3q28 duplication syndrome		8 Cases
1770	XY type gonadal dysgenesis-associated anomalies syndrome		2 Cases
370930	XYLT1-CDG		2 Cases

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662	Yellow nail syndrome		400 Cases
314485	Young adult-onset distal hereditary motor neuropathy		3 Cases
2828	Young-onset Parkinson disease	15.0 *P* *	
3472	Yunis-Varon syndrome		25 Cases
97240	Zebra body myopathy		10 Cases
217017	Zechi-Ceide syndrome		3 Cases
50812	Zellweger-like syndrome without peroxisomal anomalies		2 Cases
3473	Zimmermann-Laband syndrome		52 Cases
3253	Zlotogora-Ogur syndrome		50 Cases
913	Zollinger-Ellison syndrome	0.125 *I*	

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To access the complete Orphanet epidemiological data sets visit Orphadata (www.orphadata.org).
For any questions or comments, please contact us: contact.orphanet@inserm.fr

Editor-in-chief : Ana Rath – Editor of the report : Stéphanie NGUENGANG WAKAP - Technical support : Samuel Demarest and Valérie Lanneau
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