BNDMR

Banque Nationale de Données
Maladies Rares

Number of cases per rare disease

registered in the French National Rare Disease Registry (BNDMR) as of 7th August 2025

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The French National Rare Disease Registry (Banque Nationale de Données Maladies Rares, BNDMR) is a national epidemiological and public health tool. It is funded within the framework of the 4th National Plan for Rare Diseases (PNMR4) by the French Ministry for Solidarity and Health. It is implemented by the AP-HP. According to the PNMR3, its main objective is to facilitate the « strategic and medical management of the Rare Disease Reference Centres (CRMR), to provide the indicators necessary for the plan follow-up and to allow the implementation of studies that can generate new knowledge on rare diseases, professional practices or clinical trials feasibility ».
The BNDMR gathers an administrative and medical data set (the rare disease minimum data set) for all patients treated in centres qualified for their expertise in rare diseases. The data is collected by these centres, through the BaMaRa application or directly in compatible electronic health records. For more information: https://www.bndmr.fr

This report is part of the French National Rare Disease Registry commitments and goals, at the service of the French rare disease community. It aims to share, for each rare disease, the number of patients registered in the BNDMR.

If you have any questions about this document, as well as any request for research on the BNDMR data, you can contact the BNDMR operational team at the following address: analyse.bndmr@aphp.fr.

BNDMR
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Maladies Rares

METHODOLOGY

Rare diseases coding in the BNDMR

To identify the patients' rare disease(s), the BNDMR uses the nomenclature produced by Orphanet (INSERM) [1], according to the European Commission recommendations.

This nomenclature is a rare diseases inventory. A disease is considered rare in Europe when it affects less than one person in 2000. Each clinical entity is assigned a unique identifier that is stable over time: the ORPHAcode.

ORPHAcodes are organised into three classification levels: groups of disorders, disorders and subtypes.

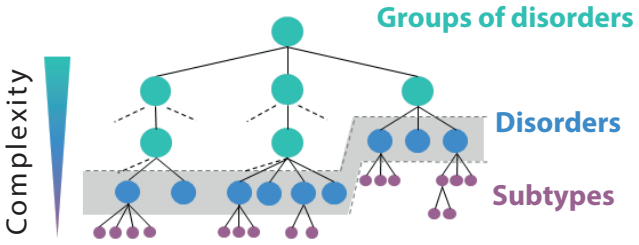

Figure 1 : Orphanet nomenclature classification levels representation

As diseases knowledge advances, the Orphanet nomenclature evolves and is updated. An annual version is published every summer. For the purpose of this report, we worked with the July 2025 version.

Some codes used in the BNDMR in the past are no longer part of this version. These are:

– **'Non rare in Europe' diseases** : the epidemiological data evolution showing that these diseases actually affect more than one person in 2000 in Europe, they can no longer be considered as rare.

– The ORPHAcodes presented in this report and the associated case counts vary from one version to another: new disease-level codes are introduced by the nomenclature package, some codes evolve (e.g., a disease code becomes a subtype and vice versa) and corrections are made by the centers.

Data source

The processed data were extracted from the BNDMR data warehouse (database containing all the national data collected within the framework of the project) of 07/08/2025, which does not include any name and surname (pseudonymised data) [2].

Only strictly useful and relevant data to the calculation and construction of this report results were used, in accordance with the data minimisation principle during processing.

The patients' number calculation (cases number) was based on the ORPHAcodes indicated by the expert centres to describe the patients' diagnosis. Living patients were calculated after matching the BNDMR data with the INSEE's death file based on the IdMR [3].

Methodology

All patients residing in France with at least one care activity in a rare disease expert centre were included. Patients without information on their place of residence were considered as resident in France by default.

Patients indicated as «not ill» (e.g. parents of a child

with a rare disease for whom a genetic sample has been taken) were excluded from the numbers, as were healthy carriers and foetuses. Other cases were excluded based on diagnostic criteria. Thus, were retired patients with a diagnosis:

– not filled in,

– classified as «non rare in Europe» according to the Orphanet nomenclature (*see previous page*),

When BaMaRa was opened, certain groups of diseases according to the Orphanet nomenclature (*see figure 1*) had been identified by the French Rare Diseases Health Networks (Filières de Santé Maladies Rares) as being of epidemiological interest. They had been made codable as a diagnosis in BaMaRa, and are still available today as an exception. They are included in this report and are identified with an **asterisk** *.

The ORPHAcodes rendered obsolete by Orphanet have been retained in this report and are the subject of a specific section at the document end.

Data preparation

In order to present only the patients number by disease (and not with a breakdown by disease subtype), the disease subtypes ORPHAcodes were reassigned at the disorder of which they are part. Thus, the cases number in this report aggregates patients coded in the BNDMR with either the ORPHAcode for the disorder or the ORPHAcode for a subtype of that disorder (*see Figure 1*).

The patients records with several identical ORPHA codes or with managements in several hospitals (several records) have been deduplicated so that the same disease is counted only once for the same patient.

When a disease concerns 10 or fewer patients, the precise number of patients is not published in compliance with the good practice rules. In this case, «≤10» is indicated.

Limitations and data quality

The patient unique identification (deduplication) was made on the basis of the national pseudonym used in the BNDMR, the IdMR [3], constructed from identity traits (surname, first name, date of birth, and sex). It is therefore sensitive to input errors and consequently does not eliminate the risk of undetected duplicates and death status (linkage between BNDMR and INSEE).

In the absence of information on the "healthy carrier" status, patients are counted as sick and therefore are part of the displayed figures. The Orphanet nomenclature evolves every year with assigning new codes or masking others (obsolet/ depreciated/non-rare). As a result, versions of this report may present fluctuations in listed diagnostic codes as well as the number of patients per disease. Rare disease networks are engaged in internal quality process which may have an impact on overall numbers per disease (deletion/de-duplication of records or recoding of the disease). The data entry for patients of the year N-1 continue over the first quarter of the following year. Therfore, this version of this report is more exaustive than the last version for patient medical files of the year N-1.

Moreover, deceased patients are included in the numbers (the patients vital status is not checked).

Presentation of the results

This report includes two lists:

1- Number of cases per rare disease registered in the BNDMR (list excluding ORPHAcodes that have become obsolete and depreciated) (p.6)

2- Number of cases per obsolete and depreciated rare disease registered in the BNDMR (p.98)

The results are presented in a table containing four columns:

– Rare disease: the disease main name according to the ORPHA nomenclature

– ORPHAcode: the rare disease unique identifier

– Cases in the BNDMR: number of distinct patients identified in the BNDMR at the report date.

– Living cases in the BNDMR: number of linving patients identified in the BNDMR at the report date.

They are listed in alphabetical order of the diseases.

References

[1] https://www.orpha.net/

[2] Anne-Sophie Jannot, Claude Messiaen, Ahlem Khatim, Thibaut Pichon, Arnaud Sandrin, the BNDMR infrastructure team, The ongoing French BaMaRa-BNDMR cohort: implementation and deployment of a nationwide information system on rare disease, Journal of the American Medical Informatics Association, Volume 29, Issue 3, November 2023, Pages 553–558, https://doi.org/10.1093/jamia/ocab237

[3] https://www.bndmr.fr/publications/identification-des-patients/

https://www.bndmr.fr

BNDMR

Banque Nationale de Données Maladies Rares

NUMBER OF CASES PER RARE DISEASE IDENTIFIED IN THE BNDMR

Excluding obsolete and depreciated ORPHAcodes

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
10q22.3q23.3 microdeletion syndrome	276413	18	18
10q22.3q23.3 microduplication syndrome	276422	10	10
11q22.2q22.3 microdeletion syndrome	444002	<10	<10
12q14 microdeletion syndrome	94063	13	13
13q12.3 microdeletion syndrome	412035	<10	<10
14q11.2 microdeletion syndrome	261120	<10	<10
14q11.2 microduplication syndrome	261229	<10	<10
14q22q23 microdeletion syndrome	264200	<10	<10
14q24.1q24.3 microdeletion syndrome	401935	<10	<10
14q32 duplication syndrome	488280	<10	<10
15q overgrowth syndrome	314585	146	142
15q11.2 microdeletion syndrome	261183	225	224
15q11q13 microduplication syndrome	238446	135	130
15q13.3 microdeletion syndrome	199318	151	150
16p11.2p12.2 microdeletion syndrome	261211	54	54
16p11.2p12.2 microduplication syndrome	261204	54	54
16p12.1p12.3 triplication syndrome	485405	<10	<10
16p13.11 microdeletion syndrome	261236	67	66
16p13.11 microduplication syndrome	261243	63	63

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
16p13.3 microduplication syndrome	96078	24	24
16q24.1 microdeletion syndrome	352629	<10	<10
16q24.3 microdeletion syndrome	261250	<10	<10
17p11.2 microduplication syndrome	1713	111	110
17p13.3 microduplication syndrome	217385	36	35
17q11.2 microduplication syndrome	139474	19	19
17q12 microdeletion syndrome	261265	77	75
17q12 microduplication syndrome	261272	81	81
17q21.31 microduplication syndrome	217340	<10	<10
17q23.1q23.2 microdeletion syndrome	261279	<10	<10
17q24.2 microdeletion syndrome	529962	<10	<10
19p13.12 microdeletion syndrome	254346	<10	<10
19p13.13 microdeletion syndrome	357001	<10	<10
19p13.3 microduplication syndrome	447980	12	12
19q13.11 microdeletion syndrome	217346	<10	<10
1p21.3 microdeletion syndrome	293948	<10	<10
1p31p32 microdeletion syndrome	401986	<10	<10
1p35.2 microdeletion syndrome	456298	<10	<10
1p36 deletion syndrome	1606	247	227
1q21.1 microdeletion syndrome	250989	134	134
1q21.1 microduplication syndrome	250994	85	85
1q41q42 microdeletion syndrome	250999	12	12

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
1q44 microdeletion syndrome	238769	**26**	**26**
20p12.3 microdeletion syndrome	261295	**<10**	**<10**
20p13 microdeletion syndrome	313781	**<10**	**<10**
20q11.2 microdeletion syndrome	444051	**<10**	**<10**
20q11.2 microduplication syndrome	363659	**<10**	**<10**
20q13.33 microdeletion syndrome	261311	**<10**	**<10**
21q deletion syndrome	574	**33**	**33**
21q22.11q22.12 microdeletion syndrome	261323	**23**	**23**
22q11.2 deletion syndrome	567	**3460**	**3333**
22q11.2 duplication syndrome	1727	**405**	**401**
2-aminoadipic 2-oxoadipic aciduria	79154	**<10**	**<10**
2-hydroxyglutaric aciduria*	19	**14**	**13**
2-methylbutyryl-CoA dehydrogenase deficiency	79157	**<10**	**<10**
2p13.2 microdeletion syndrome	363680	**<10**	**<10**
2p15p16.1 microdeletion syndrome	261349	**<10**	**<10**
2p21 microdeletion syndrome	163693	**<10**	**<10**
2q23.1 microdeletion syndrome	228402	**20**	**20**
2q23.1 microduplication syndrome	313947	**<10**	**<10**
2q31.1 microdeletion syndrome	251014	**11**	**11**
2q32q33 deletion syndrome	251019	**<10**	**<10**
2q37 microdeletion syndrome	1001	**141**	**135**
3C syndrome	7	**11**	**<10**
3-hydroxy-3-methylglutaric aciduria	20	**34**	**32**
3-hydroxy-3-methylglutaryl-CoA synthase deficiency	35701	**<10**	**<10**
3-hydroxyacyl-CoA dehydrogenase deficiency*	309127	**<10**	**<10**
3-hydroxyisobutyric aciduria	939	**<10**	**<10**
3M syndrome	2616	**94**	**90**
3MC syndrome	293843	**12**	**11**
3-methylcrotonyl-CoA carboxylase deficiency	6	**16**	**15**
3-methylglutaconic aciduria type 1	67046	**<10**	**<10**
3-methylglutaconic aciduria type 3	67047	**14**	**13**
3-methylglutaconic aciduria type 4	67048	**<10**	**<10**
3-methylglutaconic aciduria*	289902	**<10**	**<10**
3-methylglutaconic aciduria-neonatal cataract-neurologic involvement-congenital neutropenia syndrome	445038	**<10**	**<10**
3p25.3 microdeletion syndrome	435638	**14**	**14**
3q13 microdeletion syndrome	1621	**12**	**12**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
3q26 microduplication syndrome	96095	**21**	**20**
3q29 microdeletion syndrome	65286	**51**	**51**
3q29 microduplication syndrome	251038	**23**	**22**
45,X/46,XY mixed gonadal dysgenesis	1772	**304**	**303**
46,XX difference of sex development induced by endogenous maternal-derived androgen*	325093	**<10**	**<10**
46,XX difference of sex development induced by exogenous maternal-derived androgen*	325099	**<10**	**<10**
46,XX difference of sex development-anorectal anomalies syndrome	2973	**<10**	**<10**
46,XX gonadal dysgenesis	243	**94**	**92**
46,XX ovotesticular difference of sex development	2138	**83**	**83**
46,XX testicular difference of sex development	393	**65**	**65**
46,XY complete gonadal dysgenesis	242	**421**	**414**
46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	752	**34**	**33**
46,XY difference of sex development due to 5-alpha-reductase 2 deficiency	753	**60**	**60**
46,XY difference of sex development due to isolated 17,20-lyase deficiency	90796	**<10**	**<10**
46,XY difference of sex development due to testicular 17,20-desmolase deficiency	443087	**<10**	**<10**
46,XY difference of sex development-adrenal insufficiency due to CYP11A1 deficiency	168558	**<10**	**<10**
46,XY ovotesticular difference of sex development	325345	**56**	**56**
46,XY partial gonadal dysgenesis	251510	**336**	**334**
47,XYY syndrome	8	**303**	**298**
48,XXXY syndrome	96263	**33**	**33**
48,XXYY syndrome	10	**72**	**72**
49,XXXXY syndrome	96264	**37**	**37**
49,XXXYY syndrome	261534	**<10**	**<10**
49,XYYYY syndrome	99330	**<10**	**<10**
4H leukodystrophy	289494	**85**	**77**
4p16.3 microduplication syndrome	96072	**<10**	**<10**
4q21 microdeletion syndrome	238750	**17**	**16**
4q25 proximal deletion syndrome	502437	**<10**	**<10**
5p13 microduplication syndrome	329802	**<10**	**<10**
5q22 microdeletion syndrome	261584	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
5q35 microduplication syndrome	228415	<10	<10
6p22 microdeletion syndrome	251046	<10	<10
6-phosphogluconate dehydrogenase deficiency	99135	114	110
6q terminal deletion syndrome	75857	39	39
6q16 microdeletion syndrome	171829	24	24
6q25.2q25.3 microdeletion syndrome	251056	15	15
7p22.1 microduplication syndrome	314034	<10	<10
7q11.23 microduplication syndrome	96121	108	108
7q31 microdeletion syndrome	251061	13	13
8p inverted duplication/deletion syndrome	96092	15	15
8p11.2 deletion syndrome	251066	<10	<10
8p23.1 duplication syndrome	251076	28	27
8p23.1 microdeletion syndrome	251071	74	72
8q12 microduplication syndrome	228399	<10	<10
8q21.11 microdeletion syndrome	284160	<10	<10
8q22.1 microdeletion syndrome	178303	<10	<10
8q24.3 microdeletion syndrome	508488	31	31
9p13 microdeletion syndrome	324313	<10	<10
9q21.13 microdeletion syndrome	531151	<10	<10
9q31.1q31.3 microdeletion syndrome	401923	<10	<10
9q33.3q34.11 microdeletion syndrome	495818	<10	<10
AA amyloidosis	85445	369	292
AApoAIV amyloidosis	439232	<10	<10
Aarskog-Scott syndrome	915	172	171
Aase-Smith syndrome	916	<10	<10
ABeta2M amyloidosis*	439246	<10	<10
Abetalipoproteinemia	14	47	47
Ablepharon macrostomia syndrome	920	<10	<10
Abnormal number of coronary ostia	99089	<10	<10
Abnormal origin of right or left pulmonary artery from the aorta	99050	17	17
Abnormal origin of the pulmonary artery*	1138	<10	<10
Abruzzo-Erickson syndrome	921	<10	<10
Absence deformity of leg-cataract syndrome	2310	<10	<10
Absence of fingerprints-congenital milia syndrome	1658	<10	<10
Absence of the pulmonary artery	980	16	15
Absence of uterine body	180142	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Absent thumb-short stature-immunodeficiency syndrome	2951	<10	<10
Absent tibia-polydactyly-arachnoid cyst syndrome	3328	<10	<10
Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome	90301	<10	<10
Aceruloplasminemia	48818	45	41
Acetazolamide-responsive myotonia	99736	<10	<10
Achalasia-microcephaly syndrome	929	13	13
Achondrogenesis	932	10	<10
Achondroplasia	15	805	797
Achromatopsia	49382	371	370
Acinar cell carcinoma of pancreas	424046	<10	<10
Acquired aneurysmal subarachnoid hemorrhage	90065	11	10
Acquired angioedema with C1Inh deficiency	528663	126	119
Acquired angioedema*	91385	118	114
Acquired Creutzfeldt-Jakob disease*	454700	<10	<10
Acquired cutis laxa	228285	<10	<10
Acquired factor V deficiency	599490	<10	<10
Acquired factor VII deficiency	599495	56	53
Acquired factor X deficiency	599501	<10	<10
Acquired factor XI deficiency	599507	58	56
Acquired factor XIII deficiency	599513	<10	<10
Acquired generalized lipodystrophy	79086	73	63
Acquired hemophilia A	599480	250	203
Acquired hemophilia B	599485	<10	<10
Acquired ichthyosis	454	100	100
Acquired idiopathic sideroblastic anemia	75564	16	11
Acquired kinky hair syndrome	37559	<10	<10
Acquired methemoglobinemia	464453	<10	<10
Acquired monoclonal Ig light chain-associated Fanconi syndrome	91136	34	29
Acquired partial lipodystrophy	79087	67	62
Acquired prothrombin deficiency	26348	12	12
Acquired pseudoxanthoma elasticum	228247	<10	<10
Acquired purpura fulminans	49566	37	36
Acquired von Willebrand syndrome	99147	465	415
Acral peeling skin syndrome	263534	24	24
Acrocallosal syndrome	36	18	18

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Acrocapitofemoral dysplasia	63446	<10	<10
Acrocardiofacial syndrome	2008	<10	<10
Acrocraniofacial dysostosis	949	<10	<10
Acrodermatitis continua of Hallopeau	163931	<10	<10
Acrodermatitis enteropathica	37	28	26
Acrodysostosis	950	79	77
Acrodysplasia scoliosis	2956	<10	<10
Acrofacial dysostosis*	364574	<10	<10
Acrofacial dysostosis, Kennedy-Teebi type	64542	<10	<10
Acrofacial dysostosis, Weyers type	952	<10	<10
Acrofrontofacionasal dysostosis	1784	<10	<10
Acrogeria	2500	<10	<10
Acrokeratoelastoidosis of Costa	38	37	37
Acrokeratosis verruciformis of Hopf	79151	10	10
Acromegaly	963	3604	3422
Acromelic dysplasia*	93436	16	15
Acromelic frontonasal dysplasia	1827	<10	<10
Acromesomelic dysplasia*	93437	16	16
Acromesomelic dysplasia, Grebe type	2098	<10	<10
Acromesomelic dysplasia, Hunter-Thompson type	968	<10	<10
Acromesomelic dysplasia, Maroteaux type	40	18	18
Acromicric dysplasia	969	59	56
Acrootoocular syndrome	2980	<10	<10
Acropectoral syndrome	85203	<10	<10
Acropectorovertebral dysplasia	957	<10	<10
Acrorenal syndrome	971	<10	<10
Acro-renal-mandibular syndrome	958	<10	<10
Acro-renal-ocular syndrome	959	<10	<10
ACTH-dependent Cushing syndrome*	99892	82	77
Actinic lichen planus	254395	<10	<10
Actinic prurigo	330061	13	13
Actinomycosis	457095	11	10
Actinomyopathy-associated syndromic thrombocytopenia	674653	<10	<10
Action myoclonus-renal failure syndrome	163696	<10	<10
Activated PI3K-delta syndrome	397596	63	58
Acute ackee fruit intoxication	73423	<10	<10
Acute adrenal insufficiency	95409	188	176

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Acute annular outer retinopathy	284460	<10	<10
Acute bilirubin encephalopathy	529799	13	13
Acute disseminated encephalomyelitis	83597	514	507
Acute encephalopathy with biphasic seizures and late reduced diffusion	363549	<10	<10
Acute encephalopathy with inflammation-mediated status epilepticus*	363567	10	10
Acute erythroid leukemia	318	<10	<10
Acute fatty liver of pregnancy	243367	11	11
Acute flaccid myelitis	623801	<10	<10
Acute generalized exanthematous pustulosis	293173	355	303
Acute hepatic porphyria*	95157	10	<10
Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	217371	<10	<10
Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome	466794	<10	<10
Acute infantile liver failure-multisystemic involvement syndrome	370088	<10	<10
Acute inflammatory demyelinating polyradiculoneuropathy	98916	760	706
Acute intermittent porphyria	79276	734	548
Acute interstitial pneumonia	79126	415	356
Acute liver failure	90062	34	28
Acute lymphoblastic leukemia*	513	103	96
Acute macular neuroretinopathy	488239	13	13
Acute megakaryoblastic leukemia	518	<10	<10
Acute monoblastic/monocytic leukemia	514	<10	<10
Acute motor and sensory axonal neuropathy	98917	830	659
Acute motor axonal neuropathy	98918	347	307
Acute myeloblastic leukemia with maturation	98834	<10	<10
Acute myeloblastic leukemia without maturation	98833	10	<10
Acute myeloid leukaemia with myelodysplasia-related features	86845	<10	<10
Acute myeloid leukemia and myelodysplastic syndromes related to alkylating agent	102379	<10	<10
Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)	98829	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Acute myeloid leukemia with minimal differentiation	98832	<10	<10
Acute myeloid leukemia*	519	56	46
Acute myelomonocytic leukemia	517	23	19
Acute necrotizing encephalopathy of childhood	263524	10	<10
Acute pandysautonomia	231457	10	<10
Acute panmyelosis with myelofibrosis	86843	<10	<10
Acute peripheral arterial occlusion	90064	<10	<10
Acute poisoning by drugs with membrane-stabilizing effect	43119	<10	<10
Acute promyelocytic leukemia	520	<10	<10
Acute pure sensory neuropathy	231450	80	76
Acute sensory ataxic neuropathy	231466	91	80
Acute transverse myelitis	139417	424	417
Acute tricyclic antidepressant poisoning	43117	<10	<10
Acute undifferentiated leukemia	98835	<10	<10
Acute zonal occult outer retinopathy	284454	27	27
Acyl-CoA dehydrogenase 9 deficiency	99901	<10	<10
Acyl-CoA dehydrogenase deficiency*	309120	<10	<10
Adamantinoma	55881	<10	<10
Adams-Oliver syndrome	974	176	170
Addison disease	85138	1398	1313
Adducted thumbs-arthrogryposis syndrome, Christian type	2952	13	13
Adenine phosphoribosyltransferase deficiency	976	61	60
Adenocarcinoma of ovary	213504	29	19
Adenocarcinoma of the anal canal	424016	<10	<10
Adenocarcinoma of the cervix uteri	213772	<10	<10
Adenocarcinoma of the oesophagus and oesophagogastric junction	99976	<10	<10
Adenocarcinoma of the small intestine	104075	<10	<10
Adenohypophysitis	95512	566	463
Adenoma of pancreas	93292	<10	<10
Adenosarcoma of the corpus uteri	213600	<10	<10
Adenosine monophosphate deaminase deficiency	45	42	41
Adenovirus infection in immunocompromised patients	91127	<10	<10
Adenylosuccinate lyase deficiency	46	26	21
Adenylosuccinate synthetase-like 1-related distal myopathy	482601	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Adiposis dolorosa	36397	28	26
ADNP syndrome	404448	73	73
Adrenocortical carcinoma	1501	411	347
Adrenocortical carcinoma with pure aldosterone hypersecretion	231625	11	11
Adrenomyodystrophy	977	<10	<10
Adult acute respiratory distress syndrome	70578	35	25
Adult familial nephronophthisis-spastic quadriparesia syndrome	2666	<10	<10
Adult hepatocellular carcinoma	210159	<10	<10
Adult idiopathic neutropenia	2688	474	464
ADULT syndrome	978	30	29
Adult T-cell leukemia/lymphoma	86875	<10	<10
Adult-onset autosomal dominant leukodystrophy	99027	19	18
Adult-onset autosomal recessive cerebellar ataxia	284289	105	103
Adult-onset autosomal recessive sideroblastic anemia	255132	<10	<10
Adult-onset cervical dystonia, DYT23 type	420492	476	463
Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy	329336	76	70
Adult-onset distal myopathy due to VCP mutation	329478	61	52
Adult-onset dystonia-parkinsonism	199351	21	19
Adult-onset foveomacular vitelliform dystrophy	99000	128	126
Adult-onset immunodeficiency with anti-interferon-gamma autoantibodies	306431	178	165
Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	313808	28	25
Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency	329314	<10	<10
Adult-onset nemaline myopathy	171442	30	26
Adult-onset Still disease	829	884	869
African trypanosomiasis	3385	<10	<10
Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	83617	<10	<10
AGel amyloidosis	85448	23	22
Agenesis of the superior vena cava	99114	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Aggressive primary cutaneous B-cell lymphoma*	178554	**<10**	**<10**
Aggressive systemic mastocytosis	98850	**70**	**60**
Agnathia-holoprosencephaly-situs inversus syndrome	990	**<10**	**<10**
AH amyloidosis	442582	**<10**	**<10**
AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	412069	**45**	**45**
Aicardi syndrome	50	**68**	**65**
Aicardi-Goutières syndrome	51	**259**	**232**
AICA-ribosiduria	250977	**<10**	**<10**
Airway infantile hemangioma	137935	**53**	**52**
AL amyloidosis	85443	**2902**	**2043**
Alacrimia-choreoathetosis-liver dysfunction syndrome	404454	**10**	**<10**
Alagille syndrome	52	**549**	**508**
Åland Islands eye disease	178333	**<10**	**<10**
Alar cartilages hypoplasia-coloboma-telecanthus syndrome	2007	**<10**	**<10**
Alazami syndrome	319671	**23**	**23**
Albers-Schönberg osteopetrosis	53	**52**	**50**
Albinism-deafness syndrome	998	**<10**	**<10**
ALECT2 amyloidosis	439224	**<10**	**<10**
Alexander disease	58	**55**	**38**
ALG11-CDG	280071	**<10**	**<10**
ALG12-CDG	79324	**<10**	**<10**
ALG13-CDG	324422	**<10**	**<10**
ALG1-CDG	79327	**16**	**15**
ALG2-CDG	79326	**10**	**<10**
ALG3-CDG	79321	**<10**	**<10**
ALG6-CDG	79320	**<10**	**<10**
ALG8-CDG	79325	**<10**	**<10**
ALG9-CDG	79328	**<10**	**<10**
Alkaline ceramidase 3 deficiency	502444	**<10**	**<10**
Alkaptonuria	56	**113**	**108**
ALK-positive large B-cell lymphoma	364043	**<10**	**<10**
Allan-Herndon-Dudley syndrome	59	**59**	**48**
Allergic bronchopulmonary aspergillosis	1164	**500**	**467**
Alopecia antibody deficiency	1006	**<10**	**<10**
Alopecia totalis	700	**109**	**108**
Alopecia universalis	701	**123**	**123**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Alopecia-contractures-dwarfism-intellectual disability syndrome	1005	**<10**	**<10**
Alopecia-epilepsy-pyorrhea-intellectual disability syndrome	1008	**<10**	**<10**
Alopecia-intellectual disability syndrome	2850	**<10**	**<10**
Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome	1014	**<10**	**<10**
Alpers-Huttenlocher syndrome	726	**29**	**12**
Alpha delta granule deficiency	734	**156**	**149**
Alpha-1-antitrypsin deficiency	60	**1818**	**1744**
Alpha-B crystallin-related late-onset myopathy	399058	**<10**	**<10**
Alpha-crystallinopathy*	98910	**11**	**10**
Alpha-dystroglycan-related limb-girdle muscular dystrophy R16	280333	**13**	**13**
Alpha-mannosidosis	61	**69**	**69**
Alpha-N-acetylgalactosaminidase deficiency	3137	**<10**	**<10**
Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3	62	**156**	**142**
Alpha-thalassemia	846	**482**	**475**
Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	98791	**18**	**18**
Alpha-thalassemia-myelodysplastic syndrome	231401	**<10**	**<10**
Alport syndrome	63	**2635**	**2544**
Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	86818	**<10**	**<10**
Alström syndrome	64	**108**	**105**
Alternating hemiplegia of childhood	2131	**72**	**66**
Alternating hemiplegia*	209978	**80**	**77**
Alveolar echinococcosis	284	**16**	**15**
Alveolar soft tissue sarcoma	163699	**<10**	**<10**
Ameloblastoma	314419	**<10**	**<10**
Amelocerebrohypohidrotic syndrome	1946	**14**	**13**
Amelogenesis imperfecta	88661	**960**	**957**
Amelo-onycho-hypohidrotic syndrome	1028	**<10**	**<10**
Aminoacylase 1 deficiency	137754	**<10**	**<10**
Aminoacylase deficiency*	308448	**<10**	**<10**
Aminopterin/methotrexate embryofetopathy	1908	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Amish nemaline myopathy	98902	<10	<10
Amniotic band syndrome	295000	92	92
Amoebic keratitis	67043	24	24
Amyotrophic lateral sclerosis	803	15582	6408
Amyotrophic lateral sclerosis type 4	357043	26	20
Anal fistula	228113	<10	<10
Anaplastic astrocytoma	251589	14	13
Anaplastic ependymoma	251646	11	11
Anaplastic ganglioglioma	251957	<10	<10
Anaplastic large cell lymphoma	98841	<10	<10
Anaplastic oligoastrocytoma	251663	<10	<10
Anaplastic oligodendroglioma	251630	12	11
Anaplastic thyroid carcinoma	142	<10	<10
Anastomosing haemangioma	675359	<10	<10
Anauxetic dysplasia	93347	15	15
Andersen-Tawil syndrome	37553	114	112
Androgen insensitivity syndrome*	754	73	73
Aneurysm of sinus of Valsalva	1054	759	744
Aneurysmal bone cyst	480553	18	18
Aneurysm-osteoarthritis syndrome	284984	84	82
Angelman syndrome	72	832	819
Angel-shaped phalango-epiphyseal dysplasia	63442	14	14
Angiocentric glioma	251671	<10	<10
Angioimmunoblastic T-cell lymphoma	86886	<10	<10
Angioma serpiginosum	95429	22	22
Angiomatoid fibrous histiocytoma	569164	<10	<10
Angioosteohypertrophic syndrome*	2346	19	19
Angioosteohypotrophic syndrome	75508	<10	<10
Angiosarcoma	263413	12	<10
Angiostrongyliasis	74	<10	<10
Aniridia-cerebellar ataxia-intellectual disability syndrome	1065	26	26
Aniridia-intellectual disability syndrome	1068	<10	<10
Aniridia-ptosis-intellectual disability-familial obesity syndrome	1067	<10	<10
Aniridia-renal agenesis-psychomotor retardation syndrome	1064	<10	<10
ANK3-related intellectual disability-sleep disturbance syndrome	356996	<10	<10
Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	1071	72	71

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ankylosing vertebral hyperostosis with tylosis	2206	<10	<10
Annular pancreas	675	339	331
Anoctamin-5-related limb-girdle muscular dystrophy R12	206549	126	121
Anodontia	99797	<10	<10
Anomalous aortic origin of the left coronary artery	541443	48	48
Anomalous aortic origin of the right coronary artery	541454	66	66
Anomalous origin of coronary artery from the pulmonary artery	541507	116	113
Anomaly of the mitral subvalvular apparatus	101932	<10	<10
Anophthalmia plus syndrome	1104	<10	<10
Anophthalmia/microphthalmia-esophageal atresia syndrome	77298	<10	<10
Anotia	93976	39	39
Antecubital pterygium syndrome	2987	<10	<10
Anterior cutaneous nerve entrapment syndrome	51890	<10	<10
Anterior segment developmental anomaly*	88632	32	32
Anterior urethral valve	435372	22	22
Anti-glomerular basement membrane disease	375	160	147
Anti-neutrophil cytoplasmic antibody-associated vasculitis*	156152	1185	1026
Anti-p200 pemphigoid	454710	64	41
Antiphospholipid syndrome	80	5932	5794
Antisynthetase syndrome	81	1577	1448
Antley-Bixler syndrome	83	20	17
Aorta coarctation	1457	1478	1460
Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome	1110	<10	<10
Aortic arch interruption	2299	198	188
Aorto-ventricular tunnel	3400	12	12
Apert syndrome	87	123	112
Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome	1112	<10	<10
Aplasia cutis congenita	1114	84	83
Aplasia cutis congenita-intestinal lymphangiectasia syndrome	1116	<10	<10
Aplasia of lacrimal and salivary glands	86815	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Apnea of prematurity	99981	<10	<10
Apolipoprotein A-I deficiency	425	<10	<10
Apparent mineralocorticoid excess	320	<10	<10
Aquagenic palmoplantar keratoderma	498359	134	132
Arachnodactyly-abnormal ossification-intellectual disability syndrome	1129	<10	<10
Arachnodactyly-intellectual disability-dysmorphism syndrome	1130	<10	<10
Arachnoid cyst	2356	178	175
Arachnoiditis	137817	<10	<10
Arginine vasopressin deficiency	178029	1154	1099
Arginine vasopressin resistance	223	217	217
Arginine vasopressin resistance-intracranial calcification-short stature-facial dysmorphism syndrome	3145	<10	<10
Argininemia	90	39	34
Argininosuccinic aciduria	23	70	66
Arnold-Chiari malformation type I	268882	1628	1608
Aromatase deficiency	91	<10	<10
Aromatase excess syndrome	178345	11	11
Aromatic L-amino acid decarboxylase deficiency	35708	19	14
Arterial dissection-lentiginosis syndrome	1682	210	207
Arterial tortuosity syndrome	3342	48	48
Arthrochalasia Ehlers-Danlos syndrome	1899	16	16
Arthrogryposis multiplex congenita*	1037	149	137
Arthrogryposis multiplex congenita-whistling face syndrome	1150	<10	<10
Arthrogryposis syndrome*	109007	613	568
Arthrogryposis-hyperkeratosis syndrome, lethal form	1485	<10	<10
Arthrogryposis-like hand anomaly-sensorineural deafness syndrome	1144	<10	<10
Arthrogryposis-oculomotor limitation-electroretinal anomalies syndrome	1154	12	12
Arthrogryposis-renal dysfunction-cholestasis syndrome	2697	18	10
Arthrogryposis-severe scoliosis syndrome	65720	36	33
ARX-related encephalopathy-brain malformation spectrum*	423655	<10	<10
ARX-related epileptic encephalopathy*	182079	17	13

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Asbestos intoxication	2302	356	279
Ascher syndrome	1253	<10	<10
Aseptic abscess syndrome	54251	20	19
Asherman syndrome	137686	<10	<10
Aspartylglucosaminuria	93	12	12
Aspergillosis	1163	337	282
Astrocytoma*	94	76	70
Ataxia neuropathy spectrum*	254818	267	248
Ataxia with vitamin E deficiency	96	58	53
Ataxia-deafness-intellectual disability syndrome	1188	<10	<10
Ataxia-hypogonadism-choroidal dystrophy syndrome	1180	<10	<10
Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome	370022	13	13
Ataxia-oculomotor apraxia type 1	1168	46	44
Ataxia-oculomotor apraxia type 4	459033	<10	<10
Ataxia-pancytopenia syndrome	2585	12	12
Ataxia-tapetoretinal degeneration syndrome	1178	<10	<10
Ataxia-telangiectasia	100	250	209
Ataxia-telangiectasia variant	370109	<10	<10
Ataxia-telangiectasia-like disorder	251347	11	<10
Atelosteogenesis type I	1190	10	<10
Atelosteogenesis type III	56305	<10	<10
Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome	1192	<10	<10
Athyreosis	95713	651	649
Atopic keratoconjunctivitis	163934	54	54
Atresia of urethra	105	10	10
Atrial septal aneurysm	99107	<10	<10
Atrial septal defect-atrioventricular conduction defects syndrome	1479	13	12
Atrichia with papular lesions	86819	<10	<10
Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	1352	<10	<10
Atrioventricular septal defect*	98722	125	122
Atrophic lichen planus	254449	<10	<10
Atrophic papulosis	656071	<10	<10
Atrophoderma vermiculata	79100	<10	<10
Attenuated familial adenomatous polyposis	220460	93	90

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Atypical autism	199627	**2459**	**2456**
Atypical Fanconi syndrome-neonatal hyperinsulinism syndrome	544628	**<10**	**<10**
Atypical hemolytic uremic syndrome	2134	**829**	**735**
Atypical hypotonia-cystinuria syndrome	238523	**<10**	**<10**
Atypical juvenile parkinsonism	391411	**<10**	**<10**
Atypical lichen myxedematosus	86797	**<10**	**<10**
Atypical papilloma of choroid plexus	251902	**<10**	**<10**
Atypical Rett syndrome	3095	**271**	**258**
Atypical teratoid rhabdoid tumor	99966	**<10**	**<10**
Atypical Werner syndrome	79474	**<10**	**<10**
Audiogenic seizures	166415	**<10**	**<10**
Auditory neuropathy-optic atrophy syndrome	542585	**21**	**20**
Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome	77300	**36**	**35**
Auriculocondylar syndrome	137888	**22**	**22**
Autism spectrum disorder due to AUTS2 deficiency	352490	**70**	**70**
Autism spectrum disorder-epilepsy-arthrogryposis syndrome	370943	**10**	**10**
Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency	308410	**<10**	**<10**
Autoerythrocyte sensitization syndrome	324636	**14**	**14**
Autoimmune bullous skin disease*	79669	**39**	**33**
Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	420789	**10**	**<10**
Autoimmune hemolytic anemia*	98375	**1103**	**966**
Autoimmune hemolytic anemia, cold type*	228312	**59**	**51**
Autoimmune hemolytic anemia, warm type	90033	**595**	**529**
Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome due to TPP2 deficiency	444463	**32**	**31**
Autoimmune hepatitis	2137	**7528**	**7025**
Autoimmune hypoparathyroidism	36913	**40**	**40**
Autoimmune interstitial lung disease-arthritis syndrome	444092	**185**	**164**
Autoimmune limbic encephalitis	623615	**193**	**184**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autoimmune lymphoproliferative syndrome	3261	**130**	**128**
Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency	436159	**50**	**50**
Autoimmune lymphoproliferative syndrome-recurrent viral infections due to CASP8 deficiency	275517	**<10**	**<10**
Autoimmune pancreatitis type 2	280315	**138**	**133**
Autoimmune pancreatitis*	103919	**57**	**57**
Autoimmune polyendocrinopathy type 1	3453	**104**	**99**
Autoimmune polyendocrinopathy type 2	3143	**82**	**80**
Autoimmune polyendocrinopathy type 3	227982	**<10**	**<10**
Autoimmune polyendocrinopathy type 4	227990	**<10**	**<10**
Autoimmune polyendocrinopathy*	282196	**16**	**16**
Autoimmune pulmonary alveolar proteinosis	747	**127**	**113**
Autoimmune thrombocytopenia*	71203	**130**	**119**
Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation	324530	**<10**	**<10**
Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis	329173	**<10**	**<10**
Autosomal dominant adult-onset proximal spinal muscular atrophy	209335	**<10**	**<10**
Autosomal dominant aplasia and myelodysplasia	314399	**10**	**<10**
Autosomal dominant brachyolmia	93304	**<10**	**<10**
Autosomal dominant centronuclear myopathy	169189	**40**	**36**
Autosomal dominant cerebellar ataxia type I*	94145	**39**	**35**
Autosomal dominant cerebellar ataxia type II*	208508	**41**	**33**
Autosomal dominant cerebellar ataxia type III*	94148	**93**	**69**
Autosomal dominant cerebellar ataxia type IV*	94149	**<10**	**<10**
Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	314404	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation	324611	**22**	**21**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons	401964	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2*	64746	**1061**	**1010**
Autosomal dominant Charcot-Marie-Tooth disease type 2A1	99946	**120**	**114**
Autosomal dominant Charcot-Marie-Tooth disease type 2A2	99947	**114**	**112**
Autosomal dominant Charcot-Marie-Tooth disease type 2B	99936	**19**	**18**
Autosomal dominant Charcot-Marie-Tooth disease type 2C	99937	**35**	**33**
Autosomal dominant Charcot-Marie-Tooth disease type 2D	99938	**11**	**11**
Autosomal dominant Charcot-Marie-Tooth disease type 2DD	521414	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2E	99939	**27**	**26**
Autosomal dominant Charcot-Marie-Tooth disease type 2F	99940	**62**	**59**
Autosomal dominant Charcot-Marie-Tooth disease type 2I	99942	**38**	**37**
Autosomal dominant Charcot-Marie-Tooth disease type 2J	99943	**10**	**10**
Autosomal dominant Charcot-Marie-Tooth disease type 2K	99944	**37**	**36**
Autosomal dominant Charcot-Marie-Tooth disease type 2L	99945	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2M	228179	**12**	**12**
Autosomal dominant Charcot-Marie-Tooth disease type 2N	228174	**43**	**43**
Autosomal dominant Charcot-Marie-Tooth disease type 2O	284232	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2P	300319	**30**	**30**
Autosomal dominant Charcot-Marie-Tooth disease type 2Q	329258	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2U	397735	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2V	447964	**25**	**25**
Autosomal dominant Charcot-Marie-Tooth disease type 2Y	435387	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2Z	466768	**34**	**32**
Autosomal dominant childhood-onset proximal spinal muscular atrophy	363447	**50**	**49**
Autosomal dominant congenital benign spinal muscular atrophy	1216	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant cutis laxa	90348	**16**	**15**
Autosomal dominant deafness-onychodystrophy syndrome	79499	**<10**	**<10**
Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome	476093	**12**	**10**
Autosomal dominant dopa-responsive dystonia	98808	**57**	**56**
Autosomal dominant epidermolytic ichthyosis	312	**91**	**89**
Autosomal dominant focal dystonia, DYT25 type	329466	**53**	**53**
Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering	402003	**<10**	**<10**
Autosomal dominant generalized dystrophic epidermolysis bullosa	231568	**78**	**77**
Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form	79399	**<10**	**<10**
Autosomal dominant generalized epidermolysis bullosa simplex, severe form	79396	**45**	**44**
Autosomal dominant hereditary chronic pancreatitis	676	**377**	**368**
Autosomal dominant hyper-IgE syndrome due to STAT3 deficiency	2314	**133**	**129**
Autosomal dominant hyperinsulinism due to Kir6.2 deficiency	276580	**<10**	**<10**
Autosomal dominant hyperinsulinism due to SUR1 deficiency	276575	**10**	**10**
Autosomal dominant hypophosphatemic rickets	89937	**73**	**73**
Autosomal dominant intellectual disability-craniofacial dysmorphism-macrocephaly-hypotonia syndrome due to H1-4 mutation	642763	**<10**	**<10**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type A	100043	**90**	**88**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	100044	**11**	**11**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type C	100045	**<10**	**<10**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	100046	**<10**	**<10**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	93114	**14**	**13**
Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	324585	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant intermediate Charcot-Marie-Tooth disease*	90114	**34**	**32**
Autosomal dominant keratitis	2334	**<10**	**<10**
Autosomal dominant macrothrombocytopenia	140957	**215**	**213**
Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	319581	**<10**	**<10**
Autosomal dominant mitochondrial myopathy with exercise intolerance	457050	**15**	**15**
Autosomal dominant multiple pterygium syndrome	65743	**<10**	**<10**
Autosomal dominant myoglobinuria	99846	**<10**	**<10**
Autosomal dominant neovascular inflammatory vitreoretinopathy	329211	**<10**	**<10**
Autosomal dominant optic atrophy and cataract	67036	**13**	**12**
Autosomal dominant optic atrophy and peripheral neuropathy	250932	**<10**	**<10**
Autosomal dominant optic atrophy plus syndrome	1215	**49**	**49**
Autosomal dominant optic atrophy*	98672	**527**	**518**
Autosomal dominant optic atrophy, classic form	98673	**450**	**444**
Autosomal dominant osteopetrosis type 1	2783	**27**	**27**
Autosomal dominant otospondylomegaepiphyseal dysplasia	166100	**<10**	**<10**
Autosomal dominant palmoplantar keratoderma and congenital alopecia	1010	**<10**	**<10**
Autosomal dominant polycystic kidney disease	730	**8228**	**7855**
Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	88924	**69**	**69**
Autosomal dominant popliteal pterygium syndrome	1300	**11**	**11**
Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome	476119	**<10**	**<10**
Autosomal dominant primary hypomagnesemia with hypocalciuria	34528	**<10**	**<10**
Autosomal dominant prognathism	2964	**<10**	**<10**
Autosomal dominant progressive external ophthalmoplegia	254892	**39**	**38**
Autosomal dominant progressive nephropathy with hypertension	88659	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant proximal spinal muscular atrophy*	211037	**<10**	**<10**
Autosomal dominant rhegmatogenous retinal detachment	209867	**<10**	**<10**
Autosomal dominant secondary polycythemia	247511	**<10**	**<10**
Autosomal dominant severe congenital neutropenia	486	**46**	**45**
Autosomal dominant spastic ataxia type 1	251282	**<10**	**<10**
Autosomal dominant spastic paraplegia type 10	100991	**44**	**44**
Autosomal dominant spastic paraplegia type 12	100993	**<10**	**<10**
Autosomal dominant spastic paraplegia type 13	100994	**<10**	**<10**
Autosomal dominant spastic paraplegia type 17	100998	**<10**	**<10**
Autosomal dominant spastic paraplegia type 3	100984	**154**	**150**
Autosomal dominant spastic paraplegia type 31	101011	**45**	**45**
Autosomal dominant spastic paraplegia type 4	100985	**442**	**436**
Autosomal dominant spastic paraplegia type 41	320355	**<10**	**<10**
Autosomal dominant spastic paraplegia type 42	171863	**<10**	**<10**
Autosomal dominant spastic paraplegia type 6	100988	**<10**	**<10**
Autosomal dominant spastic paraplegia type 73	444099	**<10**	**<10**
Autosomal dominant spastic paraplegia type 8	100989	**22**	**22**
Autosomal dominant spastic paraplegia type 9A	447753	**<10**	**<10**
Autosomal dominant spastic paraplegia type 9B	447757	**<10**	**<10**
Autosomal dominant spondylocostal dysostosis	1797	**48**	**48**
Autosomal dominant thrombocytopenia with platelet secretion defect	466806	**18**	**18**
Autosomal dominant tubulointerstitial kidney disease	34149	**1239**	**1214**
Autosomal dominant vitreoretinochoroidopathy	3086	**16**	**16**
Autosomal erythropoietic protoporphyria	79278	**380**	**358**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal recessive anterior segment dysgenesis	519388	**18**	**18**
Autosomal recessive ataxia due to PEX10 deficiency	247815	**<10**	**<10**
Autosomal recessive ataxia due to PEX16 deficiency	642954	**<10**	**<10**
Autosomal recessive ataxia due to PEX2 deficiency	642965	**<10**	**<10**
Autosomal recessive ataxia due to ubiquinone deficiency	139485	**17**	**17**
Autosomal recessive ataxia, Beauce type	88644	**25**	**25**
Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect	521411	**<10**	**<10**
Autosomal recessive axonal hereditary motor and sensory neuropathy*	91024	**222**	**207**
Autosomal recessive axonal neuropathy with neuromyotonia	324442	**<10**	**<10**
Autosomal recessive bestrophinopathy	139455	**35**	**35**
Autosomal recessive brachyolmia	448242	**<10**	**<10**
Autosomal recessive centronuclear myopathy	169186	**28**	**26**
Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	453521	**<10**	**<10**
Autosomal recessive cerebellar ataxia due to STUB1 deficiency	412057	**13**	**12**
Autosomal recessive cerebellar ataxia with late-onset spasticity	352641	**<10**	**<10**
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency	404499	**<10**	**<10**
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency	404493	**<10**	**<10**
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency	284282	**17**	**13**
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome*	404481	**<10**	**<10**
Autosomal recessive cerebellar ataxia-movement disorder syndrome	95434	**<10**	**<10**
Autosomal recessive cerebellar ataxia-psychomotor delay syndrome	284271	**<10**	**<10**
Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome	363429	**<10**	**<10**
Autosomal recessive cerebelloparenchymal disorder type 3	1170	**26**	**25**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal recessive cerebral atrophy	363969	**<10**	**<10**
Autosomal recessive Charcot-Marie-Tooth disease type 2X	466775	**<10**	**<10**
Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	101097	**16**	**16**
Autosomal recessive chorioretinopathy-microcephaly syndrome	2518	**<10**	**<10**
Autosomal recessive congenital ichthyosis*	281097	**165**	**165**
Autosomal recessive cutis laxa type 1	90349	**<10**	**<10**
Autosomal recessive cutis laxa type 2A	357058	**10**	**10**
Autosomal recessive cutis laxa type 2B	357064	**<10**	**<10**
Autosomal recessive dopa-responsive dystonia	101150	**36**	**35**
Autosomal recessive epidermolytic ichthyosis	512103	**<10**	**<10**
Autosomal recessive faciodigitogenital syndrome	1974	**18**	**18**
Autosomal recessive frontotemporal pachygyria	329329	**<10**	**<10**
Autosomal recessive generalized dystrophic epidermolysis bullosa, intermediate form	89842	**35**	**33**
Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	79408	**26**	**19**
Autosomal recessive generalized epidermolysis bullosa simplex	89838	**10**	**<10**
Autosomal recessive hyperinsulinism due to Kir6.2 deficiency	79644	**<10**	**<10**
Autosomal recessive hyperinsulinism due to SUR1 deficiency	79643	**14**	**14**
Autosomal recessive hypophosphatemic rickets	289176	**21**	**21**
Autosomal recessive infantile hypercalcemia	300547	**196**	**195**
Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	217055	**21**	**21**
Autosomal recessive intermediate Charcot-Marie-Tooth disease type B	254334	**<10**	**<10**
Autosomal recessive intermediate Charcot-Marie-Tooth disease type C	369867	**<10**	**<10**
Autosomal recessive intermediate Charcot-Marie-Tooth disease*	268337	**223**	**217**
Autosomal recessive isolated optic atrophy	98676	**94**	**94**
Autosomal recessive lethal neonatal axonal sensorimotor polyneuropathy	538096	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome	314572	**<10**	**<10**
Autosomal recessive lower motor neuron disease with childhood onset	206580	**<10**	**<10**
Autosomal recessive malignant osteopetrosis	667	**53**	**40**
Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	319569	**<10**	**<10**
Autosomal recessive multiple pterygium syndrome	2990	**23**	**23**
Autosomal recessive myogenic arthrogryposis multiplex congenita	319332	**<10**	**<10**
Autosomal recessive optic atrophy, OPA7 type	227976	**10**	**10**
Autosomal recessive otospondylomegaepiphyseal dysplasia	1427	**<10**	**<10**
Autosomal recessive palmoplantar keratoderma and congenital alopecia	1366	**<10**	**<10**
Autosomal recessive polycystic kidney disease	731	**491**	**443**
Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity	437552	**<10**	**<10**
Autosomal recessive progressive external ophthalmoplegia	254886	**19**	**19**
Autosomal recessive secondary polycythemia not associated with VHL gene	247378	**<10**	**<10**
Autosomal recessive severe congenital neutropenia due to CSF3R deficiency	420702	**<10**	**<10**
Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency	420699	**<10**	**<10**
Autosomal recessive sideroblastic anemia	260305	**10**	**10**
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	98	**104**	**102**
Autosomal recessive spastic ataxia with leukoencephalopathy	314603	**<10**	**<10**
Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome	254343	**<10**	**<10**
Autosomal recessive spastic paraplegia type 11	2822	**81**	**79**
Autosomal recessive spastic paraplegia type 14	100995	**<10**	**<10**
Autosomal recessive spastic paraplegia type 15	100996	**11**	**11**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal recessive spastic paraplegia type 20	101000	**36**	**36**
Autosomal recessive spastic paraplegia type 21	101001	**<10**	**<10**
Autosomal recessive spastic paraplegia type 25	101005	**<10**	**<10**
Autosomal recessive spastic paraplegia type 26	101006	**<10**	**<10**
Autosomal recessive spastic paraplegia type 28	101008	**<10**	**<10**
Autosomal recessive spastic paraplegia type 35	171629	**12**	**11**
Autosomal recessive spastic paraplegia type 39	139480	**<10**	**<10**
Autosomal recessive spastic paraplegia type 43	320370	**<10**	**<10**
Autosomal recessive spastic paraplegia type 44	320401	**<10**	**<10**
Autosomal recessive spastic paraplegia type 46	320391	**<10**	**<10**
Autosomal recessive spastic paraplegia type 48	306511	**<10**	**<10**
Autosomal recessive spastic paraplegia type 54	320380	**10**	**10**
Autosomal recessive spastic paraplegia type 56	320411	**12**	**12**
Autosomal recessive spastic paraplegia type 5A	100986	**24**	**24**
Autosomal recessive spastic paraplegia type 61	401780	**<10**	**<10**
Autosomal recessive spastic paraplegia type 62	401785	**<10**	**<10**
Autosomal recessive spastic paraplegia type 69	401830	**<10**	**<10**
Autosomal recessive spastic paraplegia type 70	401835	**<10**	**<10**
Autosomal recessive spastic paraplegia type 75	459056	**<10**	**<10**
Autosomal recessive spastic paraplegia type 76	488594	**13**	**13**
Autosomal recessive spastic paraplegia type 77	466722	**<10**	**<10**
Autosomal recessive spastic paraplegia type 78	513436	**<10**	**<10**
Autosomal recessive spastic paraplegia type 82	631073	**<10**	**<10**
Autosomal recessive spondylocostal dysostosis	2311	**59**	**56**
Autosomal semi-dominant severe lipodystrophic laminopathy	280365	**26**	**24**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal spastic paraplegia type 18	209951	**10**	**10**
Autosomal spastic paraplegia type 30	101010	**48**	**48**
Autosomal spastic paraplegia type 58	397946	**<10**	**<10**
Autosomal spastic paraplegia type 72	401849	**<10**	**<10**
Autosomal systemic lupus erythematosus	300345	**63**	**61**
Axenfeld anomaly	98978	**48**	**46**
Axenfeld-Rieger syndrome	782	**191**	**189**
Axial mesodermal dysplasia spectrum	1834	**<10**	**<10**
Axial spondylometaphyseal dysplasia	168549	**<10**	**<10**
AXIN2-related polyposis	401911	**<10**	**<10**
Aymé-Gripp syndrome	1272	**<10**	**<10**
Azygos continuation of the inferior vena cava	99121	**10**	**<10**
B4GALT1-CDG	79332	**<10**	**<10**
Babesiosis	108	**<10**	**<10**
Bacterial myositis	206994	**17**	**17**
Bacterial toxic-shock syndrome	36234	**<10**	**<10**
Bainbridge-Ropers syndrome	352577	**47**	**47**
Balint syndrome	363746	**<10**	**<10**
Baller-Gerold syndrome	1225	**30**	**30**
Baló concentric sclerosis	228165	**<10**	**<10**
Bamforth-Lazarus syndrome	1226	**<10**	**<10**
Bangstad syndrome	1227	**<10**	**<10**
Banki syndrome	1228	**<10**	**<10**
BAP1-related tumor predisposition syndrome	289539	**31**	**30**
Baraitser-Winter cerebrofrontofacial syndrome	2995	**73**	**72**
Barber-Say syndrome	1231	**<10**	**<10**
Bardet-Biedl syndrome	110	**677**	**659**
Baroreflex failure	443084	**<10**	**<10**
Barth syndrome	111	**56**	**41**
Bartonella bacilliformis infection	64692	**<10**	**<10**
Bartsocas-Papas syndrome	1234	**<10**	**<10**
Bartter syndrome	112	**344**	**336**
Basel-Vanagaite-Smirin-Yosef syndrome	464738	**<10**	**<10**
Bathing suit ichthyosis	100976	**<10**	**<10**
Bazex-Dupré-Christol syndrome	113	**17**	**17**
B-cell chronic lymphocytic leukemia	67038	**28**	**22**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
B-cell immunodeficiency-limb anomaly-urogenital malformation syndrome	567502	**<10**	**<10**
Becker muscular dystrophy	98895	**1348**	**1217**
Becker nevus syndrome	64755	**45**	**45**
Beckwith-Wiedemann syndrome	116	**1427**	**1401**
Beemer-Ertbruggen syndrome	1237	**<10**	**<10**
Behavioral variant of frontotemporal dementia	275864	**1373**	**1108**
Behçet disease	117	**3806**	**3762**
Behr syndrome	1239	**<10**	**<10**
Benign cephalic histiocytosis	157997	**<10**	**<10**
Benign concentric annular macular dystrophy	251287	**<10**	**<10**
Benign focal seizures of adolescence	1544	**75**	**74**
Benign hereditary chorea	1429	**97**	**94**
Benign infantile focal epilepsy with midline spikes and waves during sleep	166308	**80**	**79**
Benign nocturnal alternating hemiplegia of childhood	209973	**<10**	**<10**
Benign paroxysmal tonic upgaze of childhood with ataxia	1179	**<10**	**<10**
Benign paroxysmal torticollis of infancy	71518	**22**	**22**
Benign partial infantile seizures*	166311	**28**	**28**
Benign recurrent intrahepatic cholestasis	65682	**119**	**116**
Benign Samaritan congenital myopathy	324581	**<10**	**<10**
Benign schwannoma	252164	**115**	**110**
Benign tumor of fallopian tubes	180237	**<10**	**<10**
BENTA disease	464336	**<10**	**<10**
Bernard-Soulier syndrome	274	**160**	**159**
Best vitelliform macular dystrophy	1243	**471**	**468**
Beta-ketothiolase deficiency	134	**64**	**64**
Beta-mannosidosis	118	**<10**	**<10**
Beta-propeller protein-associated neurodegeneration	329284	**50**	**49**
Beta-sarcoglycan-related limb-girdle muscular dystrophy R4	119	**45**	**43**
Beta-thalassemia	848	**1510**	**1483**
Beta-thalassemia-X-linked thrombocytopenia syndrome	231393	**<10**	**<10**
Bethlem muscular dystrophy	610	**250**	**236**
Bickerstaff brainstem encephalitis	79138	**40**	**38**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Bicornuate uterus*	180134	64	64
Bietti crystalline dystrophy	41751	28	28
Bifid nose	2695	<10	<10
Bifid uvula	99771	178	178
Bifunctional enzyme deficiency	300	13	<10
Bilateral microtia-deafness-cleft palate syndrome	140963	<10	<10
Bilateral polymicrogyria	268940	231	225
Bilateral striopallidodentate calcinosis	1980	225	214
Biliary atresia with splenic malformation syndrome	244283	32	30
Bilirubin encephalopathy*	415286	18	16
Biotinidase deficiency	79241	51	51
Biotin-thiamine-responsive basal ganglia disease	65284	15	14
Birdshot chorioretinopathy	179	1041	992
Birk-Barel syndrome	166108	12	11
Birt-Hogg-Dubé syndrome	122	650	640
Björnstad syndrome	123	<10	<10
Blake pouch cyst	98922	46	46
Blau syndrome	90340	85	83
Bleeding diathesis due to a collagen receptor defect	73271	16	15
Bleeding diathesis due to thromboxane synthesis deficiency	220443	22	22
Bleeding disorder due to CalDAG-GEFI deficiency	420566	<10	<10
Bleeding disorder due to P2Y12 defect	36355	48	48
Blepharo-cheilo-odontic syndrome	1997	27	25
Blepharophimosis-intellectual disability syndrome*	293642	<10	<10
Blepharophimosis-intellectual disability syndrome, MKB type	293707	<10	<10
Blepharophimosis-intellectual disability syndrome, Ohdo type	2728	42	40
Blepharophimosis-intellectual disability syndrome, SBBYS type	3047	21	21
Blepharophimosis-ptosis-epicanthus inversus syndrome	126	237	236
Blepharophimosis-ptosis-epicanthus inversus syndrome plus	572333	<10	<10
Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome	2057	<10	<10
Blepharoptosis-myopia-ectopia lentis syndrome	1259	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Blepharospasm-oromandibular dystonia syndrome	93964	207	204
Blomstrand lethal chondrodysplasia	50945	<10	<10
Bloom syndrome	125	17	14
Blount disease	2768	43	43
Blue cone monochromatism	16	118	118
Blue diaper syndrome	94086	<10	<10
Blue rubber bleb nevus	1059	108	108
Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	91135	<10	<10
Bohring-Opitz syndrome	97297	28	23
Bone sarcoma*	223727	<10	<10
BOR syndrome	107	537	533
Borjeson-Forssman-Lehmann syndrome	127	34	33
Bosley-Salih-Alorainy syndrome	69737	<10	<10
Bothnia retinal dystrophy	85128	<10	<10
Botulism	1267	12	12
Bowen-Conradi syndrome	1270	<10	<10
Brachydactyly type A1	93388	54	54
Brachydactyly type A2	93396	27	26
Brachydactyly type A4	93394	24	24
Brachydactyly type B	93383	52	51
Brachydactyly type C	93384	124	123
Brachydactyly type E	93387	65	65
Brachydactyly-arterial hypertension syndrome	1276	20	20
Brachydactyly-elbow wrist dysplasia syndrome	1275	<10	<10
Brachydactyly-mesomelia-intellectual disability-heart defects syndrome	1277	<10	<10
Brachydactyly-nystagmus-cerebellar ataxia syndrome	1246	<10	<10
Brachydactyly-preaxial hallux varus syndrome	1278	<10	<10
Brachydactyly-short stature-retinitis pigmentosa syndrome	166035	<10	<10
Brachydactyly-syndactyly, Zhao type	93409	<10	<10
Brachymorphism-onychodysplasia-dysphalangism syndrome	1292	<10	<10
Brachyolmia*	1293	40	40
Brachyolmia, Maroteaux type	93302	<10	<10
Brachyolmia-amelogenesis imperfecta syndrome	2899	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Brachytelephalangic chondrodysplasia punctata	79345	**36**	**35**
Brachytelephalangy-dysmorphism-Kallmann syndrome	1295	**<10**	**<10**
Braddock syndrome	52047	**<10**	**<10**
Bradyopsia	75374	**<10**	**<10**
Brain abnormalities-severe developmental delay-facial dysmorphism-intellectual disability syndrome	664410	**18**	**18**
Brain calcification, Rajab type	178506	**<10**	**<10**
Brain malformation-congenital heart disease-postaxial polydactyly syndrome	75389	**<10**	**<10**
Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	500150	**29**	**29**
Brain-lung-thyroid syndrome	209905	**42**	**40**
Branchiogenic deafness syndrome	50815	**20**	**20**
Branchio-oculo-facial syndrome	1297	**80**	**79**
Branchiootic syndrome	52429	**25**	**25**
Branchioskeletogenital syndrome	1299	**<10**	**<10**
BRESEK syndrome	85284	**<10**	**<10**
Brittle cornea syndrome	90354	**<10**	**<10**
Brody myopathy	53347	**23**	**23**
Bronchial neuroendocrine tumor	97287	**79**	**65**
Bronchiolitis obliterans*	1303	**770**	**731**
Bronchogenic cyst	2357	**106**	**106**
Bronchopulmonary dysplasia	70589	**2374**	**2342**
Brooke-Spiegler syndrome	79493	**112**	**111**
Brucellosis	1304	**<10**	**<10**
Bruck syndrome	2771	**30**	**29**
Brugada syndrome	130	**3927**	**3869**
Budd-Chiari syndrome	131	**803**	**719**
Buerger disease	36258	**537**	**515**
Bulbospinal muscular atrophy of adult*	206707	**72**	**60**
Bulbospinal muscular atrophy of childhood*	206704	**<10**	**<10**
Bullous impetigo	36237	**<10**	**<10**
Bullous lichen planus	33408	**11**	**10**
Bullous pemphigoid	703	**4187**	**2661**
Burkitt lymphoma	543	**22**	**18**
Burning mouth syndrome	353253	**99**	**98**
Burn-McKeown syndrome	1200	**<10**	**<10**
Butterfly-shaped pigment dystrophy	99001	**22**	**22**
BVES-related limb-girdle muscular dystrophy	476084	**<10**	**<10**
C syndrome	1308	**13**	**13**
C11ORF73-related autosomal recessive hypomyelinating leukodystrophy	495844	**<10**	**<10**
CACH syndrome	135	**92**	**74**
CAD-CDG	448010	**<10**	**<10**
CADDS	369942	**<10**	**<10**
Caffey disease	1310	**34**	**33**
Calciphylaxis	280062	**<10**	**<10**
Calpain-3-related limb-girdle muscular dystrophy D4	565909	**<10**	**<10**
Calpain-3-related limb-girdle muscular dystrophy R1	267	**524**	**495**
Calvarial doughnut lesions-bone fragility syndrome	85192	**<10**	**<10**
CAMOS syndrome	83472	**<10**	**<10**
Campomelia, Cumming type	1318	**<10**	**<10**
Campomelic dysplasia	140	**29**	**24**
Campomelic dysplasia and related disorders*	93439	**10**	**10**
Camptobrachydactyly	1319	**18**	**18**
Camptodactyly of fingers	295016	**82**	**82**
Camptodactyly syndrome, Guadalajara type 1	1327	**<10**	**<10**
Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	2848	**18**	**18**
Camptodactyly-fibrous tissue hyperplasia-skeletal anomalies syndrome	1321	**<10**	**<10**
Camptodactyly-tall stature-scoliosis-hearing loss syndrome	85164	**<10**	**<10**
Camptodactyly-taurinuria syndrome	1325	**<10**	**<10**
Camurati-Engelmann disease	1328	**51**	**51**
Canavan disease	141	**27**	**19**
Cancer-associated retinopathy	71505	**35**	**34**
CANOMAD syndrome	71279	**119**	**104**
Cantú syndrome	1517	**26**	**25**
Cap myopathy	171881	**18**	**16**
Capillary malformation-arteriovenous malformation	137667	**1149**	**1144**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Capillary-lymphatic-venous malformation with segmental distribution	90308	**482**	**480**
Carbamoyl-phosphate synthetase 1 deficiency	147	**58**	**39**
Carcinoid syndrome	100093	**18**	**16**
Cardiac anomalies-heterotaxy syndrome	137628	**88**	**79**
Cardiac anomalies-short stature-joint hypermobility-facial dysmorphism syndrome	228410	**23**	**22**
Cardiac diverticulum	1686	**<10**	**<10**
Cardiac-urogenital syndrome	647811	**<10**	**<10**
Cardiac-valvular Ehlers-Danlos syndrome	230851	**<10**	**<10**
Cardiocranial syndrome, Pfeiffer type	2872	**16**	**16**
Cardiofaciocutaneous syndrome	1340	**273**	**248**
Cardiogenic shock	97292	**14**	**11**
Cardiomyopathy-cataract-hip spine disease syndrome	1345	**<10**	**<10**
Cardiomyopathy-hypotonia-lactic acidosis syndrome	91130	**10**	**<10**
Cardiospondylocarpofacial syndrome	3238	**15**	**15**
Carey-Fineman-Ziter syndrome	1358	**17**	**16**
Caribbean parkinsonism	97355	**38**	**37**
Carney complex	1359	**172**	**168**
Carney complex-trismus-pseudocamptodactyly syndrome	319340	**<10**	**<10**
Carney triad	139411	**<10**	**<10**
Carney-Stratakis syndrome	97286	**<10**	**<10**
Carnitine palmitoyl transferase 1A deficiency	156	**20**	**20**
Carnitine palmitoyltransferase II deficiency	157	**163**	**157**
Carnitine-acylcarnitine translocase deficiency	159	**18**	**10**
Carnosinase deficiency	1361	**<10**	**<10**
Caroli disease	53035	**176**	**154**
Caroli syndrome	480520	**32**	**31**
Carpenter syndrome	65759	**10**	**<10**
Carpotarsal osteochondromatosis	2767	**<10**	**<10**
Cartilage-hair hypoplasia	175	**63**	**58**
Carvajal syndrome	65282	**<10**	**<10**
Castleman disease	160	**449**	**427**
Cataract-aberrant oral frenula-growth delay syndrome	1373	**<10**	**<10**
Cataract-congenital heart disease-neural tube defect syndrome	314993	**<10**	**<10**
Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	436174	**<10**	**<10**
Cataract-intellectual disability-hypogonadism syndrome	1387	**<10**	**<10**
Cataract-microcornea syndrome	1377	**27**	**27**
Cataract-nephropathy-encephalopathy syndrome	1380	**<10**	**<10**
Catastrophic antiphospholipid syndrome	464343	**189**	**176**
Catecholaminergic polymorphic ventricular tachycardia	3286	**461**	**446**
Catel-Manzke syndrome	1388	**<10**	**<10**
Cat-eye syndrome	195	**91**	**90**
Cathepsin A-related arteriopathy-strokes-leukoencephalopathy	575553	**<10**	**<10**
Cat-scratch disease	50839	**10**	**10**
Caudal appendage-deafness syndrome	1123	**<10**	**<10**
Caudal duplication	1756	**<10**	**<10**
Caudal regression syndrome	3027	**126**	**124**
Caudal regression-sirenomelia spectrum*	444941	**<10**	**<10**
CCDC115-CDG	468684	**<10**	**<10**
CDK13-related developmental delay-intellectual disability-facial dysmorphism-congenital heart defects syndrome	646278	**12**	**12**
CDKL5-deficiency disorder	505652	**80**	**77**
CEBPE-associated autoinflammation-immunodeficiency-neutrophil dysfunction syndrome	566067	**<10**	**<10**
Celiac artery compression syndrome	293208	**<10**	**<10**
Celiac disease-epilepsy-cerebral calcification syndrome	1459	**<10**	**<10**
CELSR1-related late-onset primary lymphedema	569816	**60**	**60**
Cenani-Lenz syndrome	3258	**<10**	**<10**
Central areolar choroidal dystrophy	75377	**45**	**43**
Central cloudy dystrophy of François	98972	**12**	**12**
Central congenital hypothyroidism*	226298	**122**	**121**
Central core disease	597	**413**	**395**
Central nervous system embryonal tumor*	251870	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Central nervous system tuberculosis	641396	**<10**	**<10**
Central neurocytoma	73256	**<10**	**<10**
Central polydactyly	295004	**12**	**12**
Central retinal artery occlusion	648684	**<10**	**<10**
Central retinal vein occlusion	411527	**151**	**146**
Central serous chorioretinopathy	443079	**40**	**39**
Centronuclear myopathy*	595	**142**	**129**
Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome	504476	**774**	**727**
Cerebellar ataxia, Cayman type	94122	**<10**	**<10**
Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome	1171	**31**	**30**
Cerebellar ataxia-ectodermal dysplasia syndrome	1174	**<10**	**<10**
Cerebellar ataxia-hypogonadism syndrome	1173	**<10**	**<10**
Cerebellar hypoplasia-intellectual disability-congenital microcephaly-dystonia-anemia-growth retardation syndrome	603448	**<10**	**<10**
Cerebellar hypoplasia-tapetoretinal degeneration syndrome	2246	**<10**	**<10**
Cerebellar-facial-dental syndrome	444072	**<10**	**<10**
Cerebral Amyloid Angiopathy	85458	**21**	**19**
Cerebral arteriovenous malformation	46724	**1619**	**1579**
Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	136	**779**	**722**
Cerebral autosomal recessive arteriopathy-subcortical infarcts-leukoencephalopathy	199354	**<10**	**<10**
Cerebral cortical dysplasia*	268950	**380**	**375**
Cerebral sinovenous thrombosis	329217	**787**	**761**
Cerebral visual impairment	447788	**24**	**23**
Cerebrocostomandibular syndrome	1393	**17**	**15**
Cerebrofacial arteriovenous metameric syndrome	141189	**<10**	**<10**
Cerebrofacioarticular syndrome	314679	**<10**	**<10**
Cerebrofaciothoracic dysplasia	1394	**<10**	**<10**
Cerebrooculonasal syndrome	66625	**<10**	**<10**
Cerebrotendinous xanthomatosis	909	**75**	**67**
Cernunnos-XLF deficiency	169079	**<10**	**<10**
Cervical aortic arch	99079	**<10**	**<10**
Cervical dermoid cyst	141046	**189**	**189**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Cervical hypertrichosis-peripheral neuropathy syndrome	2218	**<10**	**<10**
Cervicofacial fibrochondroma	141067	**527**	**527**
CHAND syndrome	1401	**<10**	**<10**
Chaotic conus spinal cord lipoma	645285	**<10**	**<10**
Char syndrome	46627	**24**	**24**
Charcot-Marie-Tooth disease type 1*	65753	**1202**	**1176**
Charcot-Marie-Tooth disease type 1A	101081	**3541**	**3461**
Charcot-Marie-Tooth disease type 1B	101082	**267**	**252**
Charcot-Marie-Tooth disease type 1C	101083	**70**	**70**
Charcot-Marie-Tooth disease type 1D	101084	**<10**	**<10**
Charcot-Marie-Tooth disease type 1E	90658	**16**	**15**
Charcot-Marie-Tooth disease type 1F	101085	**16**	**16**
Charcot-Marie-Tooth disease type 2B1	98856	**34**	**32**
Charcot-Marie-Tooth disease type 2B2	101101	**23**	**22**
Charcot-Marie-Tooth disease type 2B5	228374	**11**	**10**
Charcot-Marie-Tooth disease type 2H	101102	**26**	**23**
Charcot-Marie-Tooth disease type 2R	397968	**<10**	**<10**
Charcot-Marie-Tooth disease type 2S	443073	**20**	**20**
Charcot-Marie-Tooth disease type 2T	495274	**11**	**11**
Charcot-Marie-Tooth disease type 4*	64749	**107**	**102**
Charcot-Marie-Tooth disease type 4A	99948	**33**	**32**
Charcot-Marie-Tooth disease type 4B1	99955	**12**	**12**
Charcot-Marie-Tooth disease type 4B2	99956	**<10**	**<10**
Charcot-Marie-Tooth disease type 4C	99949	**112**	**106**
Charcot-Marie-Tooth disease type 4D	99950	**26**	**26**
Charcot-Marie-Tooth disease type 4F	99952	**54**	**53**
Charcot-Marie-Tooth disease type 4G	99953	**27**	**25**
Charcot-Marie-Tooth disease type 4H	99954	**<10**	**<10**
Charcot-Marie-Tooth disease type 4J	139515	**21**	**21**
Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome	90103	**<10**	**<10**
CHARGE syndrome	138	**741**	**649**
CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	599082	**37**	**37**
CHD4-related neurodevelopmental disorder	653712	**<10**	**<10**
CHD8 overgrowth syndrome	642675	**14**	**14**
Chédiak-Higashi syndrome	167	**26**	**24**
Cheilitis glandularis	1221	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Cherubism	184	**44**	**43**
Chilblain lupus	90280	**26**	**26**
CHILD syndrome	139	**15**	**15**
Childhood absence epilepsy	64280	**922**	**919**
Childhood-onset autosomal recessive myopathy with external ophthalmoplegia	363677	**<10**	**<10**
Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia	284324	**<10**	**<10**
Childhood-onset benign chorea with striatal involvement	494541	**<10**	**<10**
Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	500180	**<10**	**<10**
Childhood-onset nemaline myopathy	171439	**23**	**22**
Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome	466921	**<10**	**<10**
Childhood-onset schizophrenia	641496	**81**	**81**
Childhood-onset spasticity with hyperglycinemia	401866	**<10**	**<10**
CHIME syndrome	3474	**<10**	**<10**
Choanal atresia	137914	**306**	**306**
Choanal atresia-athelia-hypothyroidism-delayed puberty-short stature syndrome	589856	**<10**	**<10**
Cholangiocarcinoma	70567	**17**	**<10**
Choledochal cyst	480501	**243**	**240**
Cholestasis-lymphedema syndrome	1414	**<10**	**<10**
Chondrodysplasia punctata*	93442	**86**	**84**
Chondrodysplasia with joint dislocations, gPAPP type	280586	**<10**	**<10**
Chondrodysplasia-difference of sex development syndrome	1422	**<10**	**<10**
Chondromyxoid fibroma	404507	**<10**	**<10**
Chondrosarcoma	55880	**13**	**13**
Chordoid glioma	251674	**<10**	**<10**
Chordoma	178	**29**	**24**
Choreoacanthocytosis	2388	**15**	**13**
Choriocarcinoma of the central nervous system	252015	**<10**	**<10**
Choroid plexus carcinoma	251899	**<10**	**<10**
Choroid plexus tumor*	251896	**<10**	**<10**
Choroidal osteoma	674965	**<10**	**<10**
Choroideremia	180	**239**	**236**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Christianson syndrome	85278	**36**	**36**
Chromophobe renal cell carcinoma	319303	**38**	**38**
Chromosome Y microdeletion syndrome	1646	**73**	**71**
Chronic acquired demyelinating polyneuropathy*	208974	**440**	**394**
Chronic actinic dermatitis	330064	**<10**	**<10**
Chronic beryllium disease	133	**17**	**14**
Chronic bilirubin encephalopathy	529808	**<10**	**<10**
Chronic cutaneous lupus erythematosus*	163531	**276**	**267**
Chronic diarrhea due to glucoamylase deficiency	103907	**<10**	**<10**
Chronic diarrhea with villous atrophy	1670	**<10**	**<10**
Chronic enteropathy associated with SLCO2A1 gene	468641	**<10**	**<10**
Chronic eosinophilic leukemia	168940	**<10**	**<10**
Chronic Epstein-Barr virus infection syndrome	2566	**54**	**48**
Chronic granulomatous disease	379	**346**	**321**
Chronic inflammatory demyelinating polyneuropathy	2932	**4133**	**3574**
Chronic intervillositis of unknown etiology	615970	**<10**	**<10**
Chronic intestinal failure	294422	**243**	**217**
Chronic intestinal pseudoobstruction syndrome	2978	**504**	**446**
Chronic lymphoproliferative disorder of natural killer cells	512017	**13**	**12**
Chronic mucocutaneous candidiasis	1334	**31**	**30**
Chronic myeloid leukemia	521	**60**	**57**
Chronic myelomonocytic leukemia	98823	**19**	**13**
Chronic neurovisceral acid sphingomyelinase deficiency	618891	**<10**	**<10**
Chronic nonbacterial osteomyelitis/ Chronic recurrent multifocal osteomyelitis	324964	**901**	**901**
Chronic pneumonitis of infancy	91359	**132**	**129**
Chronic polyradiculoneuropathy*	208978	**1422**	**1272**
Chronic respiratory distress with surfactant metabolism deficiency	217566	**<10**	**<10**
Chronic thromboembolic pulmonary hypertension	70591	**3469**	**3053**
Chronic visceral acid sphingomyelinase deficiency	77293	**163**	**152**
CHST3-related skeletal dysplasia	263463	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Chudley-McCullough syndrome	314597	<10	<10
Chuvash erythrocytosis	238557	<10	<10
Chylomicron retention disease	71	27	26
Chylous ascites	1160	18	18
CIDEC-related familial partial lipodystrophy	435651	<10	<10
CINCA syndrome	1451	47	47
Citrullinemia type I	247525	83	71
Citrullinemia type II	247585	<10	<10
CK syndrome	251383	<10	<10
CLAPO syndrome	168984	10	10
Clark-Baraitser syndrome	600731	<10	<10
Class I glucose-6-phosphate dehydrogenase deficiency	466026	594	594
Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	90794	2172	2143
Classic eosinophilic pustular folliculitis	617408	<10	<10
Classic galactosemia	79239	296	294
Classic glucose transporter type 1 deficiency syndrome	71277	249	247
Classic hairy cell leukemia	58017	<10	<10
Classic heparin-induced thrombocytopenia	3325	19	16
Classic Hodgkin lymphoma	391	51	47
Classic lissencephaly*	102009	82	71
Classic mycosis fungoides	2584	23	20
Classical Ehlers-Danlos syndrome	287	538	535
Classical-like Ehlers-Danlos syndrome type 1	230839	44	44
CLCN4-related X-linked intellectual disability syndrome	485350	65	64
Clear cell renal carcinoma	319276	209	194
Clear cell sarcoma of kidney	457246	10	10
Cleft hard palate	101023	590	587
Cleft lip and alveolus	141291	1067	1065
Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-hearing loss syndrome	508476	<10	<10
Cleft lip with or without cleft palate*	1991	2522	2508
Cleft lip/palate	199306	3007	3003
Cleft lip/palate-deafness-sacral lipoma syndrome	2003	12	12
Cleft lip/palate-intestinal malrotation-cardiopathy syndrome	2001	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Cleft lip-retinopathy syndrome	1995	<10	<10
Cleft mitral valve	95465	64	64
Cleft palate*	2014	4311	4288
Cleft palate-congenital heart defect-intellectual disability syndrome	652519	12	12
Cleft palate-large ears-small head syndrome	2013	<10	<10
Cleft palate-lateral synechia syndrome	2016	<10	<10
Cleft palate-stapes fixation-oligodontia syndrome	2010	<10	<10
Cleft velum	99772	1412	1409
Cleidocranial dysplasia	1452	350	348
Climatic droplet keratopathy	98958	<10	<10
CLIPPERS	284448	10	10
CLN1 disease	228329	11	<10
CLN11 disease	314629	<10	<10
CLN2 disease	228349	19	15
CLN3 disease	228346	12	12
CLN5 disease	228360	<10	<10
CLN6 disease	228363	<10	<10
CLN7 disease	228366	<10	<10
CLN8 disease	228354	<10	<10
CLOVES syndrome	140944	519	519
CNTNAP2-related developmental and epileptic encephalopathy	163681	1942	1923
COASY protein-associated neurodegeneration	397725	<10	<10
Coats disease	190	332	329
Coats plus syndrome	313838	17	16
Cobblestone lissencephaly without muscular or ocular involvement	352682	<10	<10
Cobblestone lissencephaly*	51577	<10	<10
Cocaine embryofetopathy	1911	<10	<10
Cocaine intoxication	90068	<10	<10
Cochlear nerve deficiency	502318	22	22
Cochleovestibular malformation	502305	40	40
Cockayne syndrome	191	81	47
CODAS syndrome	1458	<10	<10
Coenzyme Q10 deficiency*	35656	27	23
Coffin-Lowry syndrome	192	156	153
Coffin-Siris syndrome	1465	423	418
COG1-CDG	263508	<10	<10
COG2-CDG	435934	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
COG4-CDG	263501	**<10**	**<10**
COG5-CDG	263487	**<10**	**<10**
COG6-CGD	464443	**<10**	**<10**
COG7-CDG	79333	**<10**	**<10**
COG8-CDG	95428	**<10**	**<10**
Cogan syndrome	1467	**181**	**178**
Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	444077	**<10**	**<10**
Cohen syndrome	193	**101**	**99**
Cohen-Gibson syndrome	659396	**<10**	**<10**
COL4A1/2-related familial vascular leukoencephalopathy	36383	**202**	**195**
Colchicine poisoning	31824	**<10**	**<10**
Cold agglutinin disease	56425	**348**	**311**
Cole-Carpenter syndrome	2050	**<10**	**<10**
Coloboma of choroid and retina	98942	**352**	**348**
Coloboma of eye lens	98943	**10**	**10**
Coloboma of eyelid	98946	**19**	**19**
Coloboma of inferior eyelid	155889	**<10**	**<10**
Coloboma of iris	98944	**254**	**251**
Coloboma of macula	98945	**12**	**12**
Coloboma of macula-brachydactyly type B syndrome	1471	**<10**	**<10**
Coloboma of optic disc	98947	**89**	**88**
Coloboma of superior eyelid	155884	**12**	**12**
Colobomatous microphthalmia	98938	**315**	**314**
Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	435930	**<10**	**<10**
Colonic atresia	1198	**20**	**18**
Combined deficiency of factor V and factor VIII	35909	**37**	**36**
Combined deficiency of factor VII and factor X	600691	**15**	**15**
Combined hamartoma of the retina and retinal pigment epithelium	440727	**14**	**14**
Combined hepatocellular carcinoma and cholangiocarcinoma	529852	**<10**	**<10**
Combined immunodeficiency due to CARD11 deficiency	357237	**<10**	**<10**
Combined immunodeficiency due to CARMIL2 deficiency	542301	**10**	**10**
Combined immunodeficiency due to CD27 deficiency	238505	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Combined immunodeficiency due to CD3gamma deficiency	169082	**<10**	**<10**
Combined immunodeficiency due to CD70 deficiency	538958	**<10**	**<10**
Combined immunodeficiency due to CRAC channel dysfunction	169090	**<10**	**<10**
Combined immunodeficiency due to DOCK2 deficiency	447737	**<10**	**<10**
Combined immunodeficiency due to DOCK8 deficiency	217390	**19**	**18**
Combined immunodeficiency due to GINS1 deficiency	505227	**<10**	**<10**
Combined immunodeficiency due to IL21R deficiency	357329	**<10**	**<10**
Combined immunodeficiency due to ITK deficiency	538963	**<10**	**<10**
Combined immunodeficiency due to LRBA deficiency	445018	**14**	**14**
Combined immunodeficiency due to MALT1 deficiency	397964	**<10**	**<10**
Combined immunodeficiency due to Moesin deficiency	504530	**<10**	**<10**
Combined immunodeficiency due to partial RAG1 deficiency	231154	**10**	**10**
Combined immunodeficiency due to RELA haploinsufficiency	596759	**<10**	**<10**
Combined immunodeficiency due to STK4 deficiency	314689	**<10**	**<10**
Combined immunodeficiency due to ZAP70 deficiency	911	**<10**	**<10**
Combined immunodeficiency with facio-oculo-skeletal anomalies	221139	**<10**	**<10**
Combined immunodeficiency with granulomatosis	157949	**205**	**193**
Combined immunodeficiency-megaloblastic anemia due to methylenetetrahydrofolate dehydrogenase 1 deficiency	658813	**<10**	**<10**
Combined immunodeficiency-multiple intestinal atresia	436252	**35**	**30**
Combined malonic and methylmalonic acidemia	289504	**<10**	**<10**
Combined oxidative phosphorylation defect type 11	324535	**<10**	**<10**
Combined oxidative phosphorylation defect type 13	319514	**<10**	**<10**
Combined oxidative phosphorylation defect type 14	319519	**<10**	**<10**
Combined oxidative phosphorylation defect type 15	319524	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Combined oxidative phosphorylation defect type 20	420728	**<10**	**<10**
Combined oxidative phosphorylation defect type 23	444013	**<10**	**<10**
Combined oxidative phosphorylation defect type 24	444458	**<10**	**<10**
Combined oxidative phosphorylation defect type 7	254930	**<10**	**<10**
Combined oxidative phosphorylation defect type 9	319509	**<10**	**<10**
Combined pancreatic lipase-colipase deficiency	309111	**10**	**10**
Combined pituitary hormone deficiencies, genetic forms	95494	**128**	**124**
Combined pulmonary fibrosis-emphysema syndrome	300564	**744**	**515**
Combined T and B cell immunodeficiency*	101972	**15**	**15**
Commissural lip fistula	141061	**<10**	**<10**
Common arterial trunk	3384	**355**	**325**
Common cystic lymphatic malformation*	458833	**330**	**328**
Common variable immunodeficiency phenotype due to IKAROS functional haploinsufficiency	317473	**10**	**10**
Common variable immunodeficiency without known genetic defect	231205	**<10**	**<10**
Complement component 3 deficiency	280133	**14**	**14**
Complement hyperactivation-angiopathic thrombosis-protein-losing enteropathy syndrome	566175	**<10**	**<10**
Complete androgen insensitivity syndrome	99429	**265**	**265**
Complete atrioventricular septal defect	1329	**1166**	**1092**
Complete septate uterus	180126	**24**	**24**
Complex hereditary spastic paraplegia*	102013	**163**	**156**
Complex lethal osteochondrodysplasia	457378	**<10**	**<10**
Complex regional pain syndrome	83452	**442**	**440**
Complication after organ transplantation	306644	**185**	**177**
Complication in hemodialysis	268316	**<10**	**<10**
Complications after hematopoietic stem cell transplantation	90053	**19**	**18**
Composite hemangioendothelioma	458758	**<10**	**<10**
Composite lymphoma	168966	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Conductive deafness-malformed external ear syndrome	3216	**65**	**65**
Conductive deafness-ptosis-skeletal anomalies syndrome	3236	**<10**	**<10**
Cone dystrophy with supernormal rod response	209932	**20**	**20**
Cone rod dystrophy	1872	**1180**	**1164**
Congenital abducens nerve palsy	440233	**<10**	**<10**
Congenital achiasma	324353	**<10**	**<10**
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	90795	**125**	**123**
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	90793	**40**	**39**
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	90791	**45**	**44**
Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	95699	**22**	**22**
Congenital adrenal hyperplasia*	418	**607**	**605**
Congenital agenesis of the scrotum	495879	**<10**	**<10**
Congenital alpha2-antiplasmin deficiency	79	**<10**	**<10**
Congenital alveolar capillary dysplasia	210122	**20**	**<10**
Congenital amegakaryocytic thrombocytopenia	3319	**22**	**21**
Congenital amyoplasia	488586	**25**	**25**
Congenital analbuminemia	86816	**<10**	**<10**
Congenital aortic valve stenosis	3093	**350**	**338**
Congenital aortopulmonary window	2037	**41**	**40**
Congenital atransferrinemia	1195	**13**	**13**
Congenital axonal neuropathy with encephalopathy	538101	**<10**	**<10**
Congenital bilateral absence of vas deferens	48	**182**	**182**
Congenital bile acid synthesis defect type 1	79301	**15**	**15**
Congenital bile acid synthesis defect type 2	79303	**11**	**11**
Congenital bile acid synthesis defect type 3	79302	**<10**	**<10**
Congenital bile acid synthesis defect type 4	79095	**<10**	**<10**
Congenital brain dysgenesis due to glutamine synthetase deficiency	71278	**<10**	**<10**
Congenital cataract microcornea with corneal opacity	289499	**10**	**10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital cataract-anterior segment dysgenesis syndrome	162	**51**	**51**
Congenital cataract-hearing loss-severe developmental delay syndrome	300313	**<10**	**<10**
Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome	1369	**11**	**<10**
Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome	330054	**<10**	**<10**
Congenital cataracts-facial dysmorphism-neuropathy syndrome	48431	**22**	**19**
Congenital central hypoventilation syndrome	661	**204**	**188**
Congenital cervical spinal stenosis	831	**17**	**16**
Congenital chloride diarrhea	53689	**21**	**19**
Congenital chronic diarrhea with protein-losing enteropathy	329242	**39**	**37**
Congenital chylothorax	264688	**76**	**69**
Congenital contractural arachnodactyly	115	**100**	**100**
Congenital cornea plana	53691	**<10**	**<10**
Congenital coronary artery aneurysm	95491	**16**	**16**
Congenital cystic eye	519384	**13**	**13**
Congenital diaphragmatic hernia	2140	**2188**	**1944**
Congenital dyserythropoietic anemia type I	98869	**27**	**26**
Congenital dyserythropoietic anemia type II	98873	**37**	**36**
Congenital dyserythropoietic anemia type III	98870	**<10**	**<10**
Congenital dyserythropoietic anemia type IV	293825	**<10**	**<10**
Congenital dyserythropoietic anemia*	85	**17**	**16**
Congenital ectropion uveae	91491	**<10**	**<10**
Congenital enteropathy due to enteropeptidase deficiency	168601	**<10**	**<10**
Congenital enterovirus infection	292	**<10**	**<10**
Congenital Epstein-Barr virus infection	70596	**<10**	**<10**
Congenital epulis	157826	**<10**	**<10**
Congenital erosive and vesicular dermatosis	231573	**<10**	**<10**
Congenital erythropoietic porphyria	79277	**45**	**34**
Congenital esophageal diverticulum	91358	**<10**	**<10**
Congenital esophageal stenosis	645749	**16**	**16**
Congenital eyelid retraction	99176	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital factor II deficiency	325	**63**	**63**
Congenital factor V deficiency	326	**700**	**691**
Congenital factor VII deficiency	327	**2802**	**2769**
Congenital factor X deficiency	328	**350**	**345**
Congenital factor XI deficiency	329	**1958**	**1921**
Congenital factor XII deficiency	330	**528**	**517**
Congenital factor XIII deficiency	331	**76**	**76**
Congenital fiber-type disproportion myopathy	2020	**188**	**178**
Congenital fibrinogen deficiency	335	**785**	**774**
Congenital fibrosis of extraocular muscles	45358	**63**	**63**
Congenital generalized hypercontractile muscle stiffness syndrome	476406	**<10**	**<10**
Congenital generalized lipodystrophy	528	**94**	**89**
Congenital Gerbode defect	99095	**<10**	**<10**
Congenital glaucoma	98976	**706**	**700**
Congenital glucokinase-related hyperinsulinism	79299	**11**	**11**
Congenital heart block	60041	**498**	**495**
Congenital heart defect-round face-developmental delay syndrome	1355	**<10**	**<10**
Congenital hemangioma*	458775	**366**	**365**
Congenital hereditary endothelial dystrophy type II	293603	**16**	**16**
Congenital hereditary facial paralysis-variable hearing loss syndrome	306530	**<10**	**<10**
Congenital herpes simplex virus infection	293	**<10**	**<10**
Congenital high-molecular-weight kininogen deficiency	483	**<10**	**<10**
Congenital Horner syndrome	91413	**35**	**35**
Congenital hydrocephalus	2185	**205**	**197**
Congenital hyperinsulinism due to HNF4A deficiency	263455	**11**	**11**
Congenital hypothyroidism due to maternal intake of antithyroid drugs	226313	**<10**	**<10**
Congenital hypothyroidism due to transplacental passage of TSH-binding inhibitory antibodies	95715	**33**	**33**
Congenital ichthyosiform erythroderma	79394	**156**	**154**
Congenital ichthyosis-microcephalus-tetraplegia syndrome	2271	**<10**	**<10**
Congenital infiltrating lipomatosis of the face	583097	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital insensitivity to pain with severe intellectual disability	453510	<10	<10
Congenital insensitivity to pain-anosmia-neuropathic arthropathy	88642	31	31
Congenital insensitivity to pain-hyperhidrosis-absence of cutaneous sensory innervation	217399	<10	<10
Congenital intrauterine infection-like syndrome	1229	<10	<10
Congenital intrinsic factor deficiency	332	26	26
Congenital isolated ACTH deficiency	199296	154	149
Congenital isolated hyperinsulinism*	657	629	621
Congenital knee dislocation	295034	<10	<10
Congenital lactase deficiency	53690	<10	<10
Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type	70472	<10	<10
Congenital laryngeal cyst	141124	49	48
Congenital laryngeal palsy	137932	147	146
Congenital laryngomalacia	2373	1407	1392
Congenital left ventricular aneurysm	1055	<10	<10
Congenital limbs-face contractures-hypotonia-developmental delay syndrome	562528	<10	<10
Congenital lipoid adrenal hyperplasia due to STAR deficency	90790	31	31
Congenital lobar emphysema	1928	150	148
Congenital long QT syndrome*	768	2325	2281
Congenital macroglossia	2430	149	149
Congenital megacalycosis	93109	16	16
Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	69063	80	75
Congenital mesoblastic nephroma	2665	18	18
Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome	391376	13	10
Congenital microcoria	566	19	17
Congenital microgastria	199293	<10	<10
Congenital mitral stenosis	99057	50	50
Congenital muscular alpha-dystroglycanopathy with brain and eye anomalies*	352687	23	21
Congenital muscular dystrophy due to LMNA mutation	157973	82	72
Congenital muscular dystrophy type 1B	98893	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital muscular dystrophy with cerebellar involvement	370959	<10	<10
Congenital muscular dystrophy with hyperlaxity	371007	28	28
Congenital muscular dystrophy with intellectual disability	370968	32	28
Congenital muscular dystrophy with intellectual disability and severe epilepsy	329178	<10	<10
Congenital muscular dystrophy without intellectual disability	370980	51	43
Congenital muscular dystrophy, Fukuyama type	272	<10	<10
Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome	1875	<10	<10
Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome	486815	<10	<10
Congenital myasthenic syndrome	590	601	572
Congenital myopathy with cores*	172976	126	119
Congenital myopathy with excess of thin filaments	98904	10	10
Congenital myopathy with internal nuclei and atypical cores	319160	12	12
Congenital myopathy with myasthenic-like onset	424107	20	18
Congenital myopathy with reduced type 2 muscle fibers	544602	14	13
Congenital myopathy, Paradas type	199329	<10	<10
Congenital myotonia*	206973	200	196
Congenital nemaline myopathy*	457074	20	20
Congenital nephrotic syndrome, Finnish type	839	92	85
Congenital neutropenia-combined immunodeficiency due to MKL1 deficiency	619941	<10	<10
Congenital neutropenia-myelofibrosis-nephromegaly syndrome	369852	<10	<10
Congenital oculomotor nerve palsy	440221	36	36
Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome	2772	<10	<10
Congenital pancreatic cyst	313906	112	111
Congenital panfollicular nevus	139414	14	14
Congenital partial pulmonary venous return anomaly	99124	359	354
Congenital patent ductus arteriosus aneurysm	99072	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital plasminogen activator inhibitor type 1 deficiency	465	**15**	**15**
Congenital portosystemic shunt	480531	**260**	**252**
Congenital prekallikrein deficiency	749	**<10**	**<10**
Congenital primary aphakia	83461	**<10**	**<10**
Congenital primary lymphedema of Gordon	569821	**17**	**17**
Congenital primary megaureter	617	**1346**	**1335**
Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome	508542	**<10**	**<10**
Congenital pseudoarthrosis of the clavicle	66630	**<10**	**<10**
Congenital ptosis	91411	**332**	**331**
Congenital pulmonary airway malformation	2444	**826**	**823**
Congenital pulmonary lymphangiectasia	2414	**44**	**41**
Congenital pulmonary sequestration	3161	**270**	**270**
Congenital pulmonary valvar stenosis	3189	**779**	**759**
Congenital pulmonary vein atresia	99126	**<10**	**<10**
Congenital pulmonary veins atresia or stenosis*	3188	**58**	**48**
Congenital pulmonary venous return anomaly*	3090	**177**	**173**
Congenital renal artery stenosis	97598	**120**	**120**
Congenital respiratory-biliary fistula	2040	**<10**	**<10**
Congenital reticular ichthyosiform erythroderma	281190	**<10**	**<10**
Congenital retinal arteriovenous communication	353334	**<10**	**<10**
Congenital rubella syndrome	290	**37**	**37**
Congenital short bowel syndrome	2301	**102**	**99**
Congenital short QT syndrome	51083	**67**	**67**
Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome	369861	**<10**	**<10**
Congenital smooth muscle hamartoma	263435	**35**	**35**
Congenital sodium diarrhea	103908	**<10**	**<10**
Congenital stationary night blindness	215	**307**	**307**
Congenital stromal corneal dystrophy	101068	**12**	**12**
Congenital subglottic stenosis	141121	**112**	**107**
Congenital sucrase-isomaltase deficiency	35122	**82**	**82**
Congenital supravalvular mitral ring	99059	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital syphilis	499009	**<10**	**<10**
Congenital systemic arteriovenous fistula	2039	**28**	**27**
Congenital temporomandibular joint ankylosis	210576	**12**	**11**
Congenital total pulmonary venous return anomaly	99125	**307**	**292**
Congenital toxoplasmosis	858	**58**	**55**
Congenital tracheal stenosis	141127	**142**	**133**
Congenital tracheomalacia	95430	**373**	**371**
Congenital tricuspid stenosis	95459	**<10**	**<10**
Congenital tricuspid valve dysplasia	555874	**48**	**46**
Congenital trigeminal anesthesia	231013	**<10**	**<10**
Congenital trochlear nerve palsy	98686	**46**	**46**
Congenital tufting enteropathy	92050	**58**	**53**
Congenital unguarded mitral orifice	99060	**22**	**21**
Congenital unilateral hypoplasia of depressor anguli oris	1166	**135**	**135**
Congenital varicella syndrome	291	**<10**	**<10**
Congenital velopharyngeal incompetence	2291	**217**	**217**
Congenital vertebral-cardiac-renal anomalies syndrome	521438	**<10**	**<10**
Congenital vertical talus	178382	**11**	**11**
Congenitally corrected transposition of the great arteries	216694	**1031**	**1004**
Congenitally uncorrected transposition of the great arteries	860	**2172**	**2126**
Conjunctival malignant melanoma	617910	**<10**	**<10**
Connective tissue disorder due to lysyl hydroxylase-3 deficiency	300284	**<10**	**<10**
Constitutional megaloblastic anemia with severe neurologic disease	319651	**<10**	**<10**
Constitutional mismatch repair deficiency syndrome	252202	**<10**	**<10**
Contractures-developmental delay-Pierre Robin syndrome	436003	**16**	**15**
Contractures-ectodermal dysplasia-cleft lip/palate syndrome	1484	**<10**	**<10**
Cooks syndrome	1487	**<10**	**<10**
Cooper-Jabs syndrome	1488	**<10**	**<10**
Cor triatriatum dexter	99098	**<10**	**<10**
Cor triatriatum sinister	99099	**14**	**14**
Corneal dystrophy-perceptive deafness syndrome	1490	**<10**	**<10**
Corneal endotheliitis	137602	**92**	**92**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome	352662	**<10**	**<10**
Cornelia de Lange syndrome	199	**461**	**438**
Corneodermatoosseous syndrome	3194	**<10**	**<10**
Coronary arterial fistula	2041	**57**	**56**
Coronary ostial stenosis or atresia	99087	**<10**	**<10**
Coronary sinus atresia	99118	**<10**	**<10**
Coronary sinus stenosis	99117	**<10**	**<10**
Corpus callosum agenesis-abnormal genitalia syndrome	2508	**11**	**11**
Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome	52055	**<10**	**<10**
Corpus callosum agenesis-macrocephaly-hypertelorism syndrome	459074	**11**	**11**
Corpus callosum agenesis-neuronopathy syndrome	1496	**39**	**38**
Cortical blindness-intellectual disability-polydactyly syndrome	1389	**<10**	**<10**
Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation	300570	**29**	**29**
Corticobasal syndrome	454887	**396**	**315**
Costello syndrome	3071	**88**	**78**
Coxopodopatellar syndrome	1509	**48**	**48**
Cramp-fasciculation syndrome	581271	**197**	**196**
Crane-Heise syndrome	1512	**<10**	**<10**
Cranial meningocele	268820	**26**	**26**
Cranial neuralgia*	221109	**<10**	**<10**
Cranio-cervical dystonia with laryngeal and upper-limb involvement	420485	**49**	**49**
Craniodiaphyseal dysplasia	1513	**<10**	**<10**
Craniodigital-intellectual disability syndrome	1514	**<10**	**<10**
Cranioectodermal dysplasia	1515	**31**	**29**
Craniofacial conodysplasia	85168	**<10**	**<10**
Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome	459061	**<10**	**<10**
Craniofacial-deafness-hand syndrome	1529	**<10**	**<10**
Craniofrontonasal dysplasia	1520	**59**	**59**
Craniofrontonasal dysplasia-Poland anomaly syndrome	1521	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Craniometadiaphyseal dysplasia, wormian bone type	85184	**<10**	**<10**
Craniometaphyseal dysplasia	1522	**29**	**28**
Cranio-osteoarthropathy	1525	**13**	**13**
Craniopharyngioma	54595	**2159**	**2022**
Craniosynostosis, Boston type	1541	**<10**	**<10**
Craniosynostosis-anal anomalies-porokeratosis syndrome	85199	**<10**	**<10**
Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome	1538	**<10**	**<10**
Craniosynostosis-dental anomalies	284149	**<10**	**<10**
Craniosynostosis-facial dysmorphism-Chiari-1 malformation-developmental and language delay syndrome	647681	**<10**	**<10**
Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome	171839	**<10**	**<10**
Craniosynostosis-intracranial calcifications syndrome	52054	**<10**	**<10**
Creatine deficiency syndrome*	79172	**20**	**20**
Crigler-Najjar syndrome	205	**50**	**48**
Crimean-Congo hemorrhagic fever	99827	**<10**	**<10**
Crisponi syndrome	1545	**24**	**18**
Criss-cross heart	1461	**10**	**<10**
Cronkhite-Canada syndrome	2930	**<10**	**<10**
Crossed polysyndactyly	2935	**<10**	**<10**
Crouzon syndrome	207	**402**	**395**
Crouzon syndrome-acanthosis nigricans syndrome	93262	**23**	**23**
Cryoglobulinemic vasculitis	91138	**895**	**810**
Cryptococcosis	1546	**<10**	**<10**
Cryptogenic late-onset epileptic spasms	163708	**24**	**24**
Cryptogenic organizing pneumonia	1302	**739**	**698**
Cryptomicrotia-brachydactyly-excess fingertip arch syndrome	1547	**<10**	**<10**
Cryptorchidism-arachnodactyly-intellectual disability syndrome	1548	**<10**	**<10**
CTCF-related neurodevelopmental disorder	363611	**15**	**15**
Currarino syndrome	1552	**272**	**264**
Curry-Jones syndrome	1553	**<10**	**<10**
Cushing disease	96253	**3075**	**2959**
Cushing syndrome due to bilateral macronodular adrenocortical disease	189427	**831**	**784**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Cushing syndrome due to cortisol-producing adrenocortical adenoma	642788	**186**	**184**
Cushing syndrome due to ectopic ACTH secretion	99889	**184**	**127**
Cutaneous collagenous vasculopathy	280779	**<10**	**<10**
Cutaneous mastocytoma	79455	**396**	**396**
Cutaneous mastocytosis*	66646	**838**	**823**
Cutaneous mastocytosis-deafness-microtia syndrome	2135	**<10**	**<10**
Cutaneous neuroendocrine carcinoma	79140	**27**	**21**
Cutaneous pseudolymphoma	451607	**<10**	**<10**
Cutaneous small vessel vasculitis	889	**170**	**161**
Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome	1555	**<10**	**<10**
Cutis laxa*	209	**82**	**76**
Cutis laxa-Marfanoid syndrome	171719	**<10**	**<10**
Cutis marmorata telangiectatica congenita	1556	**191**	**191**
Cyclic neutropenia	2686	**141**	**141**
Cylindrical spirals myopathy	171886	**<10**	**<10**
Cystadenoma of childhood	206470	**20**	**20**
Cystic echinococcosis	400	**<10**	**<10**
Cystic fibrosis	586	**9768**	**9467**
Cystic fibrosis-gastritis-megaloblastic anemia syndrome	2575	**<10**	**<10**
Cystic hamartoma of lung and kidney	2111	**<10**	**<10**
Cystic leukoencephalopathy without megalencephaly	85136	**<10**	**<10**
Cysticercosis	1560	**<10**	**<10**
Cystinosis	213	**170**	**153**
Cystinuria	214	**737**	**731**
Cystoid macular dystrophy	75381	**27**	**27**
Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	137698	**<10**	**<10**
Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder	477787	**<10**	**<10**
Czeizel-Losonci syndrome	2437	**<10**	**<10**
D,L-2-hydroxyglutaric aciduria	356978	**<10**	**<10**
D-2-hydroxyglutaric aciduria	79315	**<10**	**<10**
Dandy-Walker malformation-postaxial polydactyly syndrome	1566	**<10**	**<10**
Danon disease	34587	**46**	**42**
Darier disease	218	**266**	**260**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DDX41-related hematologic malignancy predisposition syndrome	488647	**<10**	**<10**
De Barsy syndrome	2962	**17**	**15**
De novo thrombotic microangiopathy after kidney transplantation	244275	**39**	**32**
Deaf blind hypopigmentation syndrome, Yemenite type	3214	**<10**	**<10**
Deafness with labyrinthine aplasia, microtia, and microdontia	90024	**<10**	**<10**
Deafness-craniofacial syndrome	3241	**<10**	**<10**
Deafness-ear malformation-facial palsy syndrome	3232	**16**	**15**
Deafness-enamel hypoplasia-nail defects syndrome	3220	**20**	**20**
Deafness-encephaloneuropathy-obesity-valvulopathy syndrome	254898	**<10**	**<10**
Deafness-epiphyseal dysplasia-short stature syndrome	3218	**<10**	**<10**
Deafness-hypogonadism syndrome	90646	**<10**	**<10**
Deafness-infertility syndrome	94064	**10**	**10**
Deafness-intellectual disability syndrome, Martin-Probst type	85321	**<10**	**<10**
Deafness-onychodystrophy syndrome*	3231	**10**	**<10**
Deep dermatophytosis	397587	**<10**	**<10**
Deficiency in anterior pituitary function-variable immunodeficiency syndrome	293978	**39**	**39**
Deficiency of adenosine deaminase 2	404553	**38**	**37**
Dehydrated hereditary stomatocytosis	3202	**93**	**92**
Dejerine-Sottas syndrome	64748	**19**	**15**
Delayed membranous cranial ossification	3034	**<10**	**<10**
Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome	3038	**18**	**16**
Deletion 5q35 syndrome	1627	**21**	**19**
Delta-beta-thalassemia	231237	**13**	**13**
Delta-sarcoglycan-related limb-girdle muscular dystrophy R6	219	**<10**	**<10**
Dementia pugilistica	97353	**<10**	**<10**
Demodicidosis	283	**<10**	**<10**
DEND syndrome	79134	**<10**	**<10**
Dengue fever	99828	**<10**	**<10**
Dent disease	1652	**357**	**354**
Dental ankylosis	1077	**<10**	**<10**
Dentatorubral pallidoluysian atrophy	101	**16**	**10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Dentin dysplasia	1653	30	30
Dentinogenesis imperfecta	49042	486	486
Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome	71267	<10	<10
Denys-Drash syndrome	220	88	82
Dermatitis herpetiformis	1656	89	88
Dermatofibrosarcoma protuberans	31112	16	16
Dermatoleukodystrophy	1659	<10	<10
Dermatomyositis	221	2311	2105
Dermatosparaxis Ehlers-Danlos syndrome	1901	<10	<10
Dermochondrocorneal dystrophy	79149	<10	<10
Dermoid or epidermoid cyst of the central nervous system	530033	22	22
Dermoodontodysplasia	1660	<10	<10
Desbuquois syndrome	1425	14	12
Desminopathy	98909	94	79
Desmin-related myopathy with Mallory body-like inclusions	84132	<10	<10
Desmoid tumor	873	26	26
Desmoplastic infantile astrocytoma/ganglioglioma	251940	<10	<10
Desmoplastic small round cell tumor	83469	<10	<10
Desquamative interstitial pneumonia	98852	389	352
Developmental and epileptic encephalopathy with spike-wave activation in sleep	725	636	631
Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	289307	<10	<10
Developmental delay with autism spectrum disorder and gait instability	329195	213	212
Developmental delay-ataxia-hypotonia-facial dysmorphism syndrome	658843	<10	<10
Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	369891	72	71
Developmental delay-language impairment-dopa responsive dystonia-parkinsonism syndrome	660017	33	30
Developmental malformations-deafness-dystonia syndrome	79107	<10	<10
Dextrocardia	1666	30	30
D-glyceric aciduria	941	<10	<10
Diabetic embryopathy	1926	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Diamond-Blackfan anemia	124	286	279
DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	494444	<10	<10
Diaphanospondylodysostosis	66637	18	18
Diaphragmatic defect-limb deficiency-skull defect syndrome	2141	<10	<10
Diaphragmatic hernia-short bowel-asplenia syndrome	527468	<10	<10
Diaphyseal medullary stenosis-bone malignancy syndrome	85182	<10	<10
Diastrophic dysplasia	628	65	65
Diazoxide-resistant diffuse hyperinsulinism*	165988	<10	<10
Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	276598	<10	<10
Diazoxide-resistant focal hyperinsulinism*	79298	<10	<10
Diazoxide-resistant hyperinsulinism*	276585	13	13
Diazoxide-sensitive diffuse hyperinsulinism*	165985	33	33
DICER1 tumor-predisposition syndrome	284343	26	26
Didelphys uterus	180086	97	96
Didymosis aplasticosebacea	370046	<10	<10
Diencephalic-mesencephalic junction dysplasia	319192	<10	<10
Dietary iron overload disease	139507	12	10
Difference of sex development-intellectual disability syndrome	2983	<10	<10
Differentiated thyroid carcinoma	146	171	168
Diffuse alveolar hemorrhage	90060	70	61
Diffuse astrocytoma	251595	<10	<10
Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	404437	<10	<10
Diffuse cutaneous mastocytosis	79456	125	121
Diffuse idiopathic pulmonary neuroendocrine cell hyperplasia	617916	24	22
Diffuse intrinsic pontine glioma	497188	10	<10
Diffuse large B-cell lymphoma of the central nervous system	300849	<10	<10
Diffuse large B-cell lymphoma with chronic inflammation	300888	<10	<10
Diffuse large B-cell lymphoma*	544	86	59
Diffuse leptomeningeal melanocytosis	252031	<10	<10
Diffuse lymphatic malformation	141209	162	157

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Diffuse palmoplantar keratoderma with painful fissures	369999	**<10**	**<10**
Diffuse palmoplantar keratoderma, Bothnian type	2337	**17**	**17**
Diffuse palmoplantar keratoderma-acrocyanosis syndrome	86918	**<10**	**<10**
Diffuse panbronchiolitis	171700	**24**	**22**
Diffuse unilateral subacute neuroretinitis	674947	**<10**	**<10**
Digenic hemochromatosis	648581	**<10**	**<10**
Digital extensor muscle aplasia-polyneuropathy	2926	**<10**	**<10**
Dihydropyrimidine dehydrogenase deficiency	1675	**<10**	**<10**
Dihydropyrimidinuria	38874	**<10**	**<10**
Dilated cardiomyopathy with ataxia	66634	**21**	**20**
Dimethylglycine dehydrogenase deficiency	243343	**<10**	**<10**
Diphallia	227	**<10**	**<10**
Diphtheria	1679	**<10**	**<10**
Discoid lupus erythematosus	90281	**207**	**204**
Dislocation of the hip-dysmorphism syndrome	2412	**18**	**18**
Dissecting cellulitis of the scalp	345	**<10**	**<10**
Disseminated peritoneal leiomyomatosis	71274	**<10**	**<10**
Disseminated superficial actinic porokeratosis	79152	**33**	**32**
Distal 16p11.2 microdeletion syndrome	261222	**176**	**176**
Distal 17p13.1 microdeletion syndrome	319171	**<10**	**<10**
Distal 17p13.3 microdeletion syndrome	261257	**15**	**14**
Distal 22q11.2 microdeletion syndrome	261330	**133**	**131**
Distal 22q11.2 microduplication syndrome	261337	**17**	**17**
Distal 7q11.23 microdeletion syndrome	254351	**13**	**12**
Distal 7q11.23 microduplication syndrome	261102	**<10**	**<10**
Distal anoctaminopathy	399096	**56**	**54**
Distal arthrogryposis type 1	1146	**32**	**32**
Distal arthrogryposis type 10	251515	**<10**	**<10**
Distal arthrogryposis type 5D	329457	**<10**	**<10**
Distal arthrogryposis*	97120	**188**	**184**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Distal deletion 10p syndrome	1580	**23**	**23**
Distal deletion 10q syndrome	96148	**45**	**45**
Distal deletion 12p syndrome	280325	**20**	**20**
Distal deletion 12q syndrome	96149	**<10**	**<10**
Distal deletion 13q syndrome	1590	**40**	**39**
Distal deletion 14q syndrome	96150	**20**	**20**
Distal deletion 15q syndrome	1596	**61**	**59**
Distal deletion 17q syndrome	1597	**12**	**11**
Distal deletion 19p syndrome	96129	**<10**	**<10**
Distal deletion 1q syndrome	36367	**24**	**24**
Distal deletion 3p syndrome	1620	**33**	**30**
Distal deletion 4q syndrome	96145	**57**	**52**
Distal deletion 6p syndrome	96125	**32**	**31**
Distal deletion 7p syndrome	96126	**12**	**12**
Distal deletion 9p syndrome	1642	**48**	**48**
Distal duplication 10q syndrome	96102	**11**	**11**
Distal duplication 11q syndrome	96103	**13**	**13**
Distal duplication 13q syndrome	96105	**12**	**12**
Distal duplication 14q syndrome	1705	**17**	**17**
Distal duplication 16q syndrome	96106	**17**	**17**
Distal duplication 17q syndrome	3379	**16**	**16**
Distal duplication 18q syndrome	1716	**19**	**18**
Distal duplication 19q syndrome	1717	**16**	**15**
Distal duplication 1p36 syndrome	96069	**<10**	**<10**
Distal duplication 20q syndrome	96107	**<10**	**<10**
Distal duplication 22q syndrome	96109	**19**	**19**
Distal duplication 2p syndrome	96070	**<10**	**<10**
Distal duplication 2q syndrome	96094	**10**	**10**
Distal duplication 3p syndrome	96071	**<10**	**<10**
Distal duplication 4q syndrome	96096	**12**	**11**
Distal duplication 5q syndrome	96097	**<10**	**<10**
Distal duplication 6p syndrome	1745	**13**	**13**
Distal duplication 6q syndrome	96098	**<10**	**<10**
Distal duplication 7p syndrome	96074	**<10**	**<10**
Distal duplication 8q syndrome	96100	**<10**	**<10**
Distal duplication 9q syndrome	96101	**12**	**11**
Distal hereditary motor neuropathy type 1	139518	**60**	**58**
Distal hereditary motor neuropathy type 2	139525	**14**	**12**
Distal hereditary motor neuropathy type 5	139536	**13**	**12**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Distal hereditary motor neuropathy type 7	139589	**<10**	**<10**
Distal hereditary motor neuropathy*	53739	**396**	**376**
Distal hereditary motor neuropathy, Jerash type	139552	**<10**	**<10**
Distal limb deficiencies-micrognathia syndrome	1307	**<10**	**<10**
Distal monosomy 7q36 syndrome	1636	**23**	**23**
Distal myopathy with anterior tibial onset	178400	**28**	**26**
Distal myopathy with posterior leg and anterior hand involvement	63273	**24**	**20**
Distal myopathy, Tateyama type	488650	**<10**	**<10**
Distal myopathy, Welander type	603	**<10**	**<10**
Distal myotilinopathy	98911	**39**	**37**
Distal nebulin myopathy	399103	**22**	**21**
Distal renal tubular acidosis	18	**364**	**363**
Distal spinal muscular atrophy type 3	139547	**76**	**75**
Distal Xq28 microduplication syndrome	293939	**23**	**22**
DITRA	404546	**<10**	**<10**
DK1-CDG	91131	**<10**	**<10**
DNA2-related mitochondrial DNA deletion syndrome	352470	**17**	**16**
DNAJB2-related Charcot-Marie-Tooth disease type 2	443950	**<10**	**<10**
DNAJB6-related limb-girdle muscular dystrophy D1	34516	**24**	**21**
DNMT3A-related microcephalic dwarfism	658595	**<10**	**<10**
Dominant hypophosphatemia with nephrolithiasis or osteoporosis	244305	**386**	**386**
Donnai-Barrow syndrome	2143	**13**	**12**
Donohue syndrome	508	**<10**	**<10**
DONSON-related microcephaly-short stature-limb abnormalities spectrum	572761	**<10**	**<10**
DOORS syndrome	79500	**<10**	**<10**
Dopamine beta-hydroxylase deficiency	230	**<10**	**<10**
Dopa-responsive dystonia due to sepiapterin reductase deficiency	70594	**11**	**11**
Dopa-responsive dystonia*	255	**45**	**45**
Dorsal spinal cord lipoma	645362	**19**	**19**
Double outlet left ventricle	3427	**55**	**51**
Double outlet right ventricle	3426	**703**	**663**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Double uterus-hemivagina-renal agenesis syndrome	3411	**120**	**120**
Dowling-Degos disease	79145	**14**	**14**
Down syndrome	870	**6364**	**6131**
DPAGT1-CDG	86309	**<10**	**<10**
DPM1-CDG	79322	**<10**	**<10**
DPM3-CDG	263494	**<10**	**<10**
Dravet syndrome	33069	**995**	**934**
Drug or radiation exposure-related interstitial lung disease	264978	**684**	**544**
Drug- or toxin-induced pulmonary arterial hypertension*	275786	**215**	**166**
Drug reaction with eosinophilia and systemic symptoms	139402	**1441**	**1230**
Drug-induced autoimmune hemolytic anemia	90037	**<10**	**<10**
Drug-induced localized lipodystrophy	90157	**<10**	**<10**
Drug-induced lupus erythematosus	231111	**53**	**50**
Drug-induced vasculitis	251325	**20**	**17**
Duane retraction syndrome	233	**200**	**198**
Duane retraction syndrome with congenital deafness	529574	**<10**	**<10**
Dubin-Johnson syndrome	234	**36**	**36**
Dubowitz syndrome	235	**12**	**12**
Duchenne and Becker muscular dystrophy*	262	**390**	**328**
Duchenne muscular dystrophy	98896	**2295**	**1834**
Duodenal atresia	1203	**161**	**157**
Duplication of the esophagus*	91357	**<10**	**<10**
Duplication of the pituitary gland	314621	**<10**	**<10**
Duplication of urethra	237	**66**	**66**
Dural sinus malformation	97339	**101**	**99**
Dyggve-Melchior-Clausen disease	239	**21**	**21**
DYRK1A-related intellectual disability syndrome	464306	**47**	**46**
Dysbetalipoproteinemia	412	**35**	**35**
Dyschromatosis symmetrica hereditaria	41	**<10**	**<10**
Dyschromatosis universalis hereditaria	241	**12**	**12**
Dysembryoplastic neuroepithelial tumor	251946	**351**	**347**
Dysequilibrium syndrome	1766	**105**	**103**
Dysferlin-related limb-girdle muscular dystrophy R2	268	**252**	**232**
Dyskeratosis congenita	1775	**257**	**215**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Dysmorphism-cleft palate-loose skin syndrome	1779	**<10**	**<10**
Dysmorphism-conductive hearing loss-heart defect syndrome	289553	**<10**	**<10**
Dysmorphism-pectus carinatum-joint laxity syndrome	2104	**<10**	**<10**
Dysmorphism-short stature-deafness-difference of sex development syndrome	2282	**<10**	**<10**
Dysosteosclerosis	1782	**<10**	**<10**
Dysphagia lusoria	99082	**<10**	**<10**
Dysplasia epiphysealis hemimelica	1822	**29**	**29**
Dysplasia of head of femur, Meyer type	168621	**<10**	**<10**
Dysplastic cortical hyperostosis	646139	**<10**	**<10**
Dysraphism-cleft lip/palate-limb reduction defects syndrome	2476	**11**	**11**
Dyssegmental dysplasia, Rolland-Desbuquois type	156731	**<10**	**<10**
Dyssegmental dysplasia, Silverman-Handmaker type	1865	**<10**	**<10**
Dysspondyloenchondromatosis	85198	**12**	**12**
Dystonia 16	210571	**12**	**10**
Dystonia 28	589618	**18**	**18**
Dystonia-aphonia syndrome	412217	**<10**	**<10**
Dystrophic epidermolysis bullosa pruriginosa	89843	**17**	**17**
Dystrophic epidermolysis bullosa*	303	**428**	**389**
Eales disease	40923	**70**	**69**
Early infantile developmental and epileptic encephalopathy	1934	**1384**	**1197**
Early onset non-syndromic cataract	91492	**2049**	**2031**
Early-onset autoimmunity-autoinflammation-immunodeficiency syndrome due to SOCS1 haploinsufficiency	619948	**<10**	**<10**
Early-onset autoinflammatory syndrome due to A20 haploinsufficiency	674762	**15**	**15**
Early-onset autosomal dominant Alzheimer disease	1020	**123**	**96**
Early-onset cerebellar ataxia with retained tendon reflexes	1177	**52**	**52**
Early-onset epilepsy-intellectual disability-brain anomalies syndrome	488635	**17**	**17**
Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation	289266	**51**	**50**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	411986	**<10**	**<10**
Early-onset generalized limb-onset dystonia	256	**333**	**319**
Early-onset immune dysregulation due to DOCK11 complete deficiency	658951	**<10**	**<10**
Early-onset immune dysregulation with autoimmunity due to DOCK11 partial deficiency	658946	**<10**	**<10**
Early-onset myopathy with fatal cardiomyopathy	289377	**<10**	**<10**
Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome	439212	**<10**	**<10**
Early-onset obesity-hyperphagia-severe developmental delay syndrome	99704	**<10**	**<10**
Early-onset parkinsonism-intellectual disability syndrome	2379	**<10**	**<10**
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	496641	**<10**	**<10**
Early-onset progressive encephalopathy with migrant continuous myoclonus	1943	**19**	**15**
Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	500144	**<10**	**<10**
Early-onset progressive leukoencephalopathy-central nervous system calcification-deafness-visual impairment syndrome	3240	**<10**	**<10**
Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	352654	**<10**	**<10**
Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	313772	**<10**	**<10**
Early-onset X-linked optic atrophy	98890	**52**	**51**
Ear-patella-short stature syndrome	2554	**28**	**27**
EAST syndrome	199343	**15**	**15**
Ebstein malformation of the tricuspid valve	1880	**486**	**467**
Eccrine angiomatous hamartoma	673568	**<10**	**<10**
Ectodermal dysplasia with natal teeth, Turnpenny type	69083	**<10**	**<10**
Ectodermal dysplasia, trichoodontoonychial type	1818	**10**	**10**
Ectodermal dysplasia-blindness syndrome	1806	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ectodermal dysplasia-hyperhidrosis-cutaneous syndactyly syndrome	247827	**<10**	**<10**
Ectodermal dysplasia-pili torti-cutaneous syndactyly syndrome	247820	**<10**	**<10**
Ectodermal dysplasia-sensorineural deafness syndrome	1883	**<10**	**<10**
Ectodermal dysplasia-skin fragility syndrome	158668	**<10**	**<10**
Ectopia lentis-chorioretinal dystrophy-myopia syndrome	1884	**12**	**12**
Ectrodactyly-polydactyly syndrome	1892	**<10**	**<10**
EDICT syndrome	293936	**162**	**162**
Edinburgh malformation syndrome	1895	**<10**	**<10**
EEC syndrome	1896	**148**	**146**
EEM syndrome	1897	**<10**	**<10**
Ehlers-Danlos syndrome*	98249	**2013**	**1992**
Ehlers-Danlos/osteogenesis imperfecta syndrome	230857	**31**	**30**
Eiken syndrome	79106	**<10**	**<10**
Eisenmenger syndrome	97214	**409**	**345**
Elastofibroma dorsi	228243	**<10**	**<10**
Ellis Van Creveld syndrome	289	**70**	**61**
Emanuel syndrome	96170	**21**	**20**
Embryonal carcinoma	180226	**<10**	**<10**
Emery-Dreifuss muscular dystrophy	261	**219**	**188**
Emery-Nelson syndrome	1927	**<10**	**<10**
EMILIN-1-related connective tissue disease	485418	**<10**	**<10**
Enamel-renal syndrome	1031	**56**	**56**
Encephalitis lethargica	83600	**<10**	**<10**
Encephaloclastic disorder*	269190	**<10**	**<10**
Encephalocraniocutaneous lipomatosis	2396	**10**	**10**
Encephalopathy due to mitochondrial and peroxisomal fission defect	527276	**11**	**<10**
Encephalopathy due to prosaposin deficiency	139406	**<10**	**<10**
Encephalopathy due to sulfite oxidase deficiency	833	**34**	**18**
Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	319678	**<10**	**<10**
Encircling double aortic arch	99075	**58**	**57**
Endocardial fibroelastosis	2022	**<10**	**<10**
Endometrial stromal sarcoma	213711	**<10**	**<10**
Endometrioid carcinoma of ovary	454723	**<10**	**<10**
Endophthalmitis	199323	**53**	**51**
Endosteal hyperostosis, Worth type	2790	**<10**	**<10**
Enlarged parietal foramina	60015	**21**	**21**
Enteric anendocrinosis	83620	**<10**	**<10**
Enteropathy-associated T-cell lymphoma	86880	**<10**	**<10**
Enthesitis-related juvenile idiopathic arthritis	85438	**2889**	**2881**
Eosinophilic angiocentric fibrosis	449566	**<10**	**<10**
Eosinophilic colitis	402035	**19**	**19**
Eosinophilic fasciitis	3165	**290**	**283**
Eosinophilic gastroenteritis	2070	**30**	**29**
Eosinophilic granulomatosis with polyangiitis	183	**1480**	**1402**
Ependymal tumor*	301	**<10**	**<10**
Ependymoma	251636	**65**	**59**
EPHB4-related lymphatic-related hydrops fetalis	568065	**<10**	**<10**
Epiblepharon	99169	**51**	**50**
Epidermal nevus syndrome	35125	**252**	**252**
Epidermolysis bullosa acquisita	46487	**241**	**227**
Epidermolysis bullosa simplex with mottled pigmentation	79397	**<10**	**<10**
Epidermolysis bullosa simplex with muscular dystrophy	257	**<10**	**<10**
Epidermolysis bullosa simplex*	304	**410**	**406**
Epidermolytic nevus	497737	**23**	**23**
Epidermolytic palmoplantar keratoderma	2199	**70**	**70**
Epilepsy of infancy with migrating focal seizures	293181	**72**	**66**
Epilepsy with auditory features	101046	**14**	**14**
Epilepsy with eyelid myoclonia	139431	**229**	**228**
Epilepsy with myoclonic absences	86911	**355**	**351**
Epilepsy with myoclonic-atonic seizures	1942	**574**	**571**
Epilepsy with reading-induced seizures	166433	**<10**	**<10**
Epilepsy-microcephaly-skeletal dysplasia syndrome	1948	**<10**	**<10**
Epilepsy-telangiectasia syndrome	1951	**<10**	**<10**
Epileptic encephalopathy with global cerebral demyelination	353217	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Epiphyseal dysplasia-hearing loss-dysmorphism syndrome	1825	**<10**	**<10**
Epiphyseal stippling-osteoclastic hyperplasia syndrome	1952	**<10**	**<10**
Epiphysiolysis of the hip	399329	**10**	**10**
Episodic ataxia type 1	37612	**52**	**52**
Episodic ataxia type 3	79135	**<10**	**<10**
Episodic ataxia type 4	79136	**<10**	**<10**
Episodic ataxia type 5	211067	**<10**	**<10**
Episodic ataxia type 6	209967	**<10**	**<10**
Episodic ataxia with slurred speech	401953	**<10**	**<10**
Episodic memory defect leukoencephalopathy	662229	**<10**	**<10**
Epithelial basement membrane dystrophy	98956	**142**	**140**
Epithelial recurrent erosion dystrophy	293381	**26**	**26**
Epithelioid hemangioendothelioma	157791	**43**	**38**
Epithelioid sarcoma	293202	**<10**	**<10**
Epstein-Barr virus-positive diffuse large B-cell lymphoma	289661	**<10**	**<10**
Erdheim-Chester disease	35687	**400**	**350**
Ermine phenotype	999	**<10**	**<10**
Erosive pustular dermatosis of the scalp	222	**<10**	**<10**
Erythema elevatum diutinum	90000	**<10**	**<10**
Erythema multiforme major	502499	**205**	**203**
Erythroderma desquamativum	314	**<10**	**<10**
Erythrokeratoderma ''en cocardes''	315	**<10**	**<10**
Erythrokeratoderma variabilis progressiva*	308166	**<10**	**<10**
Erythrokeratodermia variabilis	317	**26**	**26**
Esophageal atresia	1199	**2791**	**2699**
Essential fructosuria	2056	**<10**	**<10**
Essential thrombocythemia	3318	**145**	**138**
Estrogen resistance syndrome	785	**<10**	**<10**
Ethylene glycol poisoning	31826	**<10**	**<10**
Ethylmalonic encephalopathy	51188	**10**	**<10**
Euryblepharon	99172	**<10**	**<10**
Euthyroid dysprealbuminemic hyperthyroxinemia	597939	**<10**	**<10**
Euthyroid Graves orbitopathy	466682	**32**	**32**
Evans syndrome	1959	**629**	**578**
EVEN-plus syndrome	496751	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Exercise intolerance with lactic acidosis*	254843	**20**	**20**
Exercise-induced malignant hyperthermia	466650	**45**	**45**
Exfoliative ichthyosis	289586	**18**	**17**
Exostoses-anetodermia-brachydactyly type E syndrome	1962	**<10**	**<10**
Exstrophy-epispadias complex	322	**696**	**692**
Extensive peripapillary myelinated nerve fibers	440724	**17**	**17**
External auditory canal aplasia/ hypoplasia	141074	**1580**	**1570**
External auditory canal atresia-vertical talus-hypertelorism syndrome	3023	**<10**	**<10**
Extracranial carotid artery aneurysm	494424	**15**	**14**
Extracutaneous mastocytoma	66662	**<10**	**<10**
Extragonadal germinoma	182127	**163**	**159**
Extragonadal teratoma	883	**118**	**116**
Extramammary Paget disease	2800	**20**	**18**
Extramedullary conus spinal cord lipoma	645297	**52**	**52**
Extraneural perineurioma	100002	**<10**	**<10**
Extranodal nasal NK/T cell lymphoma	86879	**<10**	**<10**
Extrapelvic endometriosis	137820	**78**	**77**
Extraskeletal Ewing sarcoma	370334	**<10**	**<10**
Fabry disease	324	**1807**	**1705**
Facial arteriovenous malformation*	156230	**645**	**642**
Facial dermoid cyst	141051	**493**	**493**
Facial diplegia with paresthesias	480701	**<10**	**<10**
Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome	1969	**<10**	**<10**
Facial dysmorphism-global developmental delay-hypotonia-polymicrogyria syndrome	659609	**<10**	**<10**
Facial dysmorphism-immunodeficiency-livedo-short stature syndrome	352712	**15**	**15**
Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome	412022	**<10**	**<10**
Facial dysmorphism-shawl scrotum-joint laxity syndrome	1778	**<10**	**<10**
Facial onset sensory and motor neuronopathy	85162	**35**	**20**
Faciocardiorenal syndrome	1973	**<10**	**<10**
Facioscapulohumeral dystrophy	269	**3682**	**3381**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Fallot complex-intellectual disability-growth delay syndrome	3304	**<10**	**<10**
Familial abdominal aortic aneurysm	86	**67**	**63**
Familial acute necrotizing encephalopathy	88619	**<10**	**<10**
Familial adenomatous polyposis	733	**675**	**652**
Familial adrenal hypoplasia with absent pituitary luteinizing hormone	95700	**<10**	**<10**
Familial adult myoclonic epilepsy	86814	**24**	**24**
Familial Alzheimer-like prion disease	280397	**<10**	**<10**
Familial anetoderma	228277	**55**	**54**
Familial angiolipomatosis	199279	**<10**	**<10**
Familial aortic dissection	229	**333**	**313**
Familial articular hypermobility syndrome	2295	**1207**	**1204**
Familial atrial myxoma	615	**<10**	**<10**
Familial atypical multiple mole melanoma syndrome	404560	**135**	**132**
Familial avascular necrosis of femoral head	86820	**<10**	**<10**
Familial benign copper deficiency	1551	**10**	**10**
Familial benign flecked retina	363989	**<10**	**<10**
Familial bicuspid aortic valve	402075	**363**	**357**
Familial calcium pyrophosphate deposition	1416	**31**	**27**
Familial caudal dysgenesis	1768	**<10**	**<10**
Familial cavitary optic disc anomaly	464760	**<10**	**<10**
Familial cerebral cavernous malformation	221061	**673**	**655**
Familial cerebral saccular aneurysm	231160	**89**	**87**
Familial Chilblain lupus	481662	**<10**	**<10**
Familial chylomicronemia syndrome	444490	**83**	**83**
Familial clubfoot with or without associated lower limb anomalies	199315	**414**	**414**
Familial cold urticaria	47045	**42**	**42**
Familial congenital mirror movements	238722	**35**	**35**
Familial congenital nasolacrimal duct obstruction	451612	**<10**	**<10**
Familial congenital palsy of trochlear nerve	91498	**<10**	**<10**
Familial cortical myoclonus	319189	**22**	**22**
Familial cutaneous collagenoma	53296	**<10**	**<10**
Familial developmental dysphasia	1799	**52**	**52**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	300751	**589**	**565**
Familial drusen	75376	**104**	**103**
Familial dysautonomia	1764	**24**	**21**
Familial dyskinesia and facial myokymia	324588	**12**	**12**
Familial encephalopathy with neuroserpin inclusion bodies	85110	**<10**	**<10**
Familial episodic pain syndrome	391384	**11**	**11**
Familial expansile osteolysis	85195	**<10**	**<10**
Familial exudative vitreoretinopathy	891	**203**	**203**
Familial focal epilepsy with variable foci	98820	**89**	**89**
Familial generalized lentiginosis	231040	**10**	**10**
Familial gestational hyperthyroidism	99819	**16**	**16**
Familial glucocorticoid deficiency	361	**30**	**29**
Familial hemophagocytic lymphohistiocytosis	540	**120**	**108**
Familial hyperaldosteronism type I	403	**109**	**107**
Familial hyperaldosteronism type II	404	**25**	**25**
Familial hyperaldosteronism type III	251274	**<10**	**<10**
Familial hyperaldosteronism type IV	642671	**<10**	**<10**
Familial hyperaldosteronism*	235936	**47**	**47**
Familial hypercholanemia	238475	**17**	**17**
Familial hyperinflammatory lymphoproliferative immunodeficiency	619953	**<10**	**<10**
Familial hyperprolactinemia	397685	**20**	**19**
Familial hyperthyroidism due to mutations in TSH receptor	424	**138**	**134**
Familial hypoaldosteronism	427	**32**	**31**
Familial hypocalciuric hypercalcemia	405	**820**	**796**
Familial idiopathic dilatation of the right atrium	1677	**<10**	**<10**
Familial infantile bilateral striatal necrosis	225154	**<10**	**<10**
Familial infantile myoclonic epilepsy	352582	**26**	**26**
Familial intestinal malrotation	508410	**<10**	**<10**
Familial isolated café-au-lait macules	2678	**256**	**256**
Familial isolated clinodactyly of fingers	295014	**31**	**31**
Familial isolated congenital asplenia	101351	**37**	**37**
Familial isolated dilated cardiomyopathy	154	**12121**	**11452**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Familial isolated hyperparathyroidism	99879	**275**	**262**
Familial isolated hypoparathyroidism	2238	**372**	**359**
Familial isolated pituitary adenoma	314777	**<10**	**<10**
Familial isolated restrictive cardiomyopathy	75249	**265**	**231**
Familial isolated retinal arteriolar tortuosity	75326	**13**	**13**
Familial isolated trichomegaly	411788	**<10**	**<10**
Familial juvenile hypertrophy of the breast	180176	**49**	**49**
Familial keratoacanthoma	493	**<10**	**<10**
Familial median cleft of the upper and lower lips	401942	**<10**	**<10**
Familial Mediterranean fever	342	**2491**	**2454**
Familial melanoma	618	**410**	**382**
Familial mesial temporal lobe epilepsy	163717	**10**	**<10**
Familial mitral valve prolapse	741	**110**	**110**
Familial monosomy 7 syndrome	495930	**<10**	**<10**
Familial multinodular goiter	276399	**18**	**18**
Familial multiple lipomatosis	199276	**33**	**32**
Familial multiple meningioma	263662	**19**	**19**
Familial multiple nevi flammei	624	**10**	**10**
Familial or sporadic hemiplegic migraine	569	**282**	**277**
Familial ossifying fibroma	435329	**<10**	**<10**
Familial osteochondritis dissecans	251262	**<10**	**<10**
Familial pancreatic carcinoma	1333	**175**	**161**
Familial papillary or follicular thyroid carcinoma	319487	**13**	**13**
Familial paroxysmal ataxia	97	**178**	**173**
Familial partial epilepsy*	309	**107**	**107**
Familial partial lipodystrophy*	98306	**24**	**23**
Familial partial lipodystrophy, Dunnigan type	2348	**391**	**376**
Familial partial lipodystrophy, Köbberling type	79084	**42**	**42**
Familial patent arterial duct	466729	**24**	**24**
Familial peripheral male-limited precocious puberty	3000	**33**	**32**
Familial platelet disorder with associated myeloid malignancy	71290	**155**	**154**
Familial primary hyperparathyroidism*	2207	**704**	**639**
Familial primary localized cutaneous amyloidosis	353220	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Familial progressive hyper- and hypopigmentation	280628	**42**	**42**
Familial progressive hyperpigmentation	79146	**25**	**25**
Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome	488197	**<10**	**<10**
Familial prostate cancer	1331	**23**	**20**
Familial pseudohyperkalemia	90044	**<10**	**<10**
Familial reactive perforating collagenosis	79147	**<10**	**<10**
Familial recurrent peripheral facial palsy	2809	**26**	**26**
Familial renal glucosuria	69076	**46**	**46**
Familial retinal arterial macroaneurysm	284247	**<10**	**<10**
Familial scaphocephaly syndrome, McGillivray type	168624	**17**	**17**
Familial spontaneous pneumothorax	2903	**46**	**45**
Familial steroid-resistant nephrotic syndrome with adrenal insufficiency	506334	**<10**	**<10**
Familial steroid-resistant nephrotic syndrome with sensorineural deafness	280406	**<10**	**<10**
Familial supernumerary nipples	2456	**<10**	**<10**
Familial temporal lobe epilepsy	98819	**46**	**46**
Familial thoracic aortic aneurysm and aortic dissection	91387	**1088**	**1070**
Familial thrombocytosis	71493	**23**	**22**
Familial thrombomodulin anomalies	3324	**<10**	**<10**
Familial thyroglossal duct cyst	93953	**119**	**119**
Familial thyroid dyshormonogenesis	95716	**452**	**449**
Familial tumoral calcinosis	53715	**45**	**44**
Familial vesicoureteral reflux	289365	**209**	**208**
Familial visceral myopathy	2604	**<10**	**<10**
Fanconi anemia	84	**357**	**314**
Fanconi-Bickel syndrome	2088	**24**	**23**
Farber disease	333	**<10**	**<10**
FASTKD2-related infantile mitochondrial encephalomyopathy	166105	**<10**	**<10**
Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease	439854	**<10**	**<10**
Fatal familial insomnia	466	**<10**	**<10**
Fatal infantile cytochrome C oxidase deficiency	1561	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Fatal infantile hypertonic myofibrillar myopathy	280553	<10	<10
Fatal infantile lactic acidosis with methylmalonic aciduria	17	25	17
Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	168566	<10	<10
Fatal post-viral neurodegenerative disorder	391343	<10	<10
FATCO syndrome	2492	11	10
Fatty acid hydroxylase-associated neurodegeneration	329308	14	13
Fatty acyl-CoA reductase 1 deficiency	438178	<10	<10
Febrile infection-related epilepsy syndrome	163703	115	108
Feingold syndrome	1305	117	116
Felty syndrome	47612	11	10
Female infertility due to oocyte meiotic arrest	488191	11	11
Female infertility due to zona pellucida defect	404466	<10	<10
Female restricted epilepsy with intellectual disability	101039	123	122
Femoral-facial syndrome	1988	<10	<10
Femur-fibula-ulna complex	2019	14	14
Ferroportin disease	648562	14	13
Fetal akinesia deformation sequence	994	13	<10
Fetal akinesia-cerebral and retinal hemorrhage syndrome	363409	<10	<10
Fetal alcohol syndrome	1915	943	937
Fetal and neonatal alloimmune thrombocytopenia	853	35	35
Fetal anticonvulsant syndrome*	370068	<10	<10
Fetal cytomegalovirus syndrome	294	285	277
Fetal hydantoin syndrome	1912	<10	<10
Fetal iodine syndrome	1910	16	16
Fetal lower urinary tract obstruction*	435365	30	30
Fetal parvovirus syndrome	295	<10	<10
Fetal valproate spectrum disorder	1906	591	584
Fever-associated acute infantile liver failure syndrome	464724	<10	<10
FG syndrome type 1	93932	12	12
FGFR2-related bent bone dysplasia	313855	<10	<10
Fibroblastic rheumatism	477650	<10	<10
Fibrochondrogenesis	2021	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Fibrodysplasia ossificans progressiva	337	181	174
Fibronectin glomerulopathy	84090	<10	<10
Fibrosarcoma	2030	<10	<10
Fibrosis-neurodegeneration-cerebral angiomatosis syndrome	621758	<10	<10
Fibrous dysplasia of bone	249	2169	2133
Fibular aplasia-complex brachydactyly syndrome	2639	11	11
Fibular aplasia-ectrodactyly syndrome	1118	27	27
Fibular dimelia-diplopodia syndrome	1757	<10	<10
Filippi syndrome	3255	<10	<10
Fingerprint body myopathy	97232	<10	<10
First branchial cleft anomaly	141013	233	231
Fixed drug eruption	293812	159	136
Fixed subaortic stenosis	3092	146	144
FKRP-related limb-girdle muscular dystrophy R9	34515	133	115
Flat face-microstomia-ear anomaly syndrome	1968	<10	<10
Fleck corneal dystrophy	98970	<10	<10
FLNA-related X-linked myxomatous valvular dysplasia	555877	10	10
Floating-Harbor syndrome	2044	94	94
Florid cemento-osseous dysplasia	83451	<10	<10
Focal acral hyperkeratosis	308013	<10	<10
Focal dermal hypoplasia	2092	79	78
Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	352587	31	31
Focal facial dermal dysplasia	398166	31	31
Focal myositis	48918	211	200
Focal palmoplantar and gingival keratoderma	2200	<10	<10
Focal palmoplantar keratoderma with joint keratoses	370002	<10	<10
Foix-Alajouanine syndrome	79093	<10	<10
Foix-Chavany-Marie syndrome	2048	<10	<10
Follicular lymphoma	545	29	22
Folliculotropic mycosis fungoides	178512	<10	<10
Fontaine progeroid syndrome	697101	<10	<10
Formiminoglutamic aciduria	51208	<10	<10
Fountain syndrome	3219	<10	<10
Fourth branchial cleft anomaly	141037	115	114

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome	397618	**11**	**11**
Foveal hypoplasia-presenile cataract syndrome	2253	**25**	**25**
Fowler urethral sphincter dysfunction syndrome	2795	**<10**	**<10**
Fowler vasculopathy	221126	**<10**	**<10**
FOXG1 syndrome	561854	**61**	**59**
FOXP1 Syndrome	391372	**76**	**76**
Fragile X syndrome	908	**2168**	**2144**
Fragile X-associated primary ovarian insufficiency	642691	**50**	**50**
Fragile X-associated tremor/ataxia syndrome	93256	**116**	**110**
Fraser syndrome	2052	**27**	**22**
Frasier syndrome	347	**30**	**29**
FRAXE intellectual disability	100973	**55**	**53**
FRAXF syndrome	100974	**<10**	**<10**
Free sialic acid storage disease	834	**<10**	**<10**
Freeman-Sheldon syndrome	2053	**53**	**49**
Frey syndrome	662240	**<10**	**<10**
Fried syndrome	85335	**<10**	**<10**
Friedreich ataxia	95	**1115**	**997**
Frontal fibrosing alopecia	254492	**<10**	**<10**
Frontofacionasal dysplasia	1791	**16**	**16**
Frontometaphyseal dysplasia	1826	**23**	**23**
Frontonasal arteriovenous malformation	141168	**56**	**56**
Frontonasal dysplasia*	250	**63**	**63**
Frontonasal dysplasia-alopecia-genital anomalies syndrome	228390	**<10**	**<10**
Frontorhiny	391474	**<10**	**<10**
Frontotemporal dementia with motor neuron disease	275872	**580**	**206**
Frontotemporal dementia*	282	**794**	**663**
Fructose-1,6-bisphosphatase deficiency	348	**36**	**36**
Fryns syndrome	2059	**15**	**12**
FTH1-related iron overload	247790	**11**	**11**
Fuchs endothelial corneal dystrophy	98974	**1573**	**1556**
Fuchs heterochromic iridocyclitis	263479	**81**	**81**
Fucosidosis	349	**<10**	**<10**
Fuhrmann syndrome	2854	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Fukutin-related limb-girdle muscular dystrophy R13	206554	**10**	**10**
Full NF2-related schwannomatosis	637	**788**	**723**
Full schwannomatosis	93921	**539**	**532**
Fulminant viral hepatitis	35063	**18**	**18**
Fumaric aciduria	24	**13**	**10**
Functioning gonadotropic adenoma	91348	**1140**	**1034**
Functioning pituitary adenoma*	314753	**18**	**17**
Fundus albipunctatus	227796	**28**	**28**
Fungal keratitis	519930	**25**	**24**
Fungal myositis	207000	**<10**	**<10**
Furuncular myiasis	591	**<10**	**<10**
Gabriele-de Vries syndrome	506358	**16**	**16**
Galactokinase deficiency	79237	**<10**	**<10**
Galactose epimerase deficiency	79238	**<10**	**<10**
Galactosialidosis	351	**<10**	**<10**
Gallbladder neuroendocrine tumor	100086	**<10**	**<10**
Galloway-Mowat syndrome	2065	**46**	**29**
Gamma-aminobutyric acid transaminase deficiency	2066	**<10**	**<10**
Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5	353	**241**	**229**
Gangliocytoma	251937	**<10**	**<10**
Ganglioglioma	251949	**155**	**152**
Ganglioneuroblastoma	251877	**<10**	**<10**
Ganglioneuroma	251992	**18**	**18**
GAPO syndrome	2067	**<10**	**<10**
Gastrocutaneous syndrome	2069	**<10**	**<10**
Gastrointestinal stromal tumor	44890	**<10**	**<10**
Gastroschisis	2368	**315**	**303**
GATA2 deficiency spectrum	228423	**48**	**43**
Gaucher disease	355	**485**	**437**
Gelastic seizures with hypothalamic hamartoma	86906	**87**	**86**
Gelatinous drop-like corneal dystrophy	98957	**<10**	**<10**
Geleophysic dysplasia	2623	**22**	**18**
Gemignani syndrome	2074	**<10**	**<10**
Generalized arterial calcification of infancy	51608	**28**	**26**
Generalized basaloid follicular hamartoma syndrome	168632	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Generalized bulbospinal muscular atrophy*	206710	**60**	**52**
Generalized epilepsy-paroxysmal dyskinesia syndrome	79137	**13**	**13**
Generalized eruptive histiocytosis	157991	**<10**	**<10**
Generalized essential telangiectasia	280774	**13**	**13**
Generalized glucocorticoid resistance syndrome	786	**11**	**10**
Generalized peeling skin syndrome	263543	**<10**	**<10**
Generalized pustular psoriasis	247353	**34**	**33**
Genetic central precocious puberty in female	650077	**127**	**127**
Genetic epilepsy with febrile seizure plus	36387	**723**	**716**
Genetic hyperferritinemia without iron overload	254704	**212**	**210**
Genetic non-syndromic obesity	98267	**851**	**845**
Genetic recurrent myoglobinuria	99845	**16**	**15**
Genetic transient congenital hypothyroidism	226316	**14**	**14**
Genitopalatocardiac syndrome	2075	**<10**	**<10**
Genitopatellar syndrome	85201	**<10**	**<10**
Genochondromatosis type 1	85197	**<10**	**<10**
Genochondromatosis type 2	93398	**<10**	**<10**
Geroderma osteodysplastica	2078	**<10**	**<10**
Gerstmann-Straussler-Scheinker syndrome	356	**10**	**<10**
Ghosal hematodiaphyseal dysplasia	1802	**12**	**12**
Giant adenofibroma of the breast	180267	**87**	**87**
Giant axonal neuropathy	643	**13**	**12**
Giant cell arteritis	397	**6497**	**6064**
Giant cell tumor of bone	363976	**<10**	**<10**
Gingival fibromatosis-facial dysmorphism syndrome	2025	**<10**	**<10**
Gingival fibromatosis-hypertrichosis syndrome	2026	**<10**	**<10**
Gingival fibromatosis-progressive deafness syndrome	2027	**<10**	**<10**
Gitelman syndrome	358	**814**	**802**
Gitelman-like kidney tubulopathy due to mitochondrial DNA mutation	620371	**<10**	**<10**
GJC2-related late-onset primary lymphedema	568051	**70**	**70**
Glanzmann thrombasthenia	849	**394**	**381**
Glaucoma secondary to spherophakia/ectopia lentis and megalocornea	238763	**14**	**14**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Glaucoma-sleep apnea syndrome	2085	**<10**	**<10**
Glaucomatocyclitic crisis disease	636950	**20**	**20**
Glial tumor of neuroepithelial tissue with unknown origin*	251668	**<10**	**<10**
Glial tumor*	182067	**49**	**41**
Glioblastoma	360	**45**	**12**
Glioependymal/ependymal cyst	269197	**<10**	**<10**
Gliomatosis cerebri	251582	**<10**	**<10**
Global developmental delay-alopecia-macrocephaly-facial dysmorphism-structural brain anomalies syndrome	544488	**<10**	**<10**
Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome	404476	**<10**	**<10**
Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome	488613	**13**	**13**
Global developmental delay-osteopenia-ectodermal defect syndrome	73223	**<10**	**<10**
Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	480898	**14**	**13**
Glomus tumor	391651	**<10**	**<10**
Glomuvenous malformation	83454	**138**	**138**
Glossopalatine ankylosis	141163	**<10**	**<10**
Glucagonoma	97280	**<10**	**<10**
Glucose-galactose malabsorption	35710	**11**	**11**
Glutaric acidemia type 3	35706	**<10**	**<10**
Glutaryl-CoA dehydrogenase deficiency	25	**174**	**161**
Glutathione synthetase deficiency	32	**18**	**17**
Glycerol kinase deficiency*	308993	**<10**	**<10**
Glycine encephalopathy	407	**103**	**64**
Glycogen storage disease due to acid maltase deficiency	365	**488**	**408**
Glycogen storage disease due to aldolase A deficiency	57	**<10**	**<10**
Glycogen storage disease due to glucose-6-phosphatase deficiency	364	**280**	**273**
Glycogen storage disease due to glycogen branching enzyme deficiency	367	**40**	**36**
Glycogen storage disease due to glycogen debranching enzyme deficiency	366	**218**	**206**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Glycogen storage disease due to glycogen synthase deficiency*	308520	**<10**	**<10**
Glycogen storage disease due to hepatic glycogen synthase deficiency	2089	**21**	**20**
Glycogen storage disease due to lactate dehydrogenase deficiency	2364	**<10**	**<10**
Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency	79240	**10**	**10**
Glycogen storage disease due to liver glycogen phosphorylase deficiency	369	**42**	**42**
Glycogen storage disease due to liver phosphorylase kinase deficiency	264580	**23**	**22**
Glycogen storage disease due to muscle and heart glycogen synthase deficiency	137625	**<10**	**<10**
Glycogen storage disease due to muscle glycogen phosphorylase deficiency	368	**377**	**360**
Glycogen storage disease due to muscle phosphofructokinase deficiency	371	**12**	**12**
Glycogen storage disease due to muscle phosphorylase kinase deficiency	715	**10**	**10**
Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	713	**<10**	**<10**
Glycogen storage disease due to phosphoglycerate mutase deficiency	97234	**<10**	**<10**
Glycogen storage disease due to phosphorylase kinase deficiency*	370	**127**	**122**
Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	263297	**<10**	**<10**
GM1 gangliosidosis	354	**102**	**60**
GM2 gangliosidosis*	309152	**14**	**<10**
GM2 gangliosidosis, AB variant	309246	**<10**	**<10**
GM3 synthase deficiency	370933	**<10**	**<10**
GMPPB-related limb-girdle muscular dystrophy R19	363623	**<10**	**<10**
GMS syndrome	2090	**<10**	**<10**
GNAO1-related developmental delay-seizures-movement disorder spectrum	592564	**30**	**27**
Gnathodiaphyseal dysplasia	53697	**<10**	**<10**
GNB5-related intellectual disability-cardiac arrhythmia syndrome	542306	**<10**	**<10**
GNE myopathy	602	**97**	**89**
Goldberg-Shprintzen megacolon syndrome	66629	**10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Goldmann-Favre syndrome	53540	**59**	**59**
Gollop-Wolfgang complex	1986	**<10**	**<10**
Gómez-López-Hernández syndrome	1532	**<10**	**<10**
Gonadoblastoma	206484	**<10**	**<10**
Gonococcal conjunctivitis	1482	**<10**	**<10**
Good syndrome	169105	**84**	**72**
Gordon syndrome	376	**28**	**28**
Gorham-Stout disease	73	**55**	**52**
Gorlin syndrome	377	**513**	**501**
GRACILE syndrome	53693	**<10**	**<10**
Graft versus host disease	39812	**141**	**121**
Graham Little-Piccardi-Lassueur syndrome	505	**<10**	**<10**
Grange syndrome	79094	**<10**	**<10**
Granular corneal dystrophy type I	98962	**43**	**43**
Granular corneal dystrophy type II	98963	**17**	**17**
Granuloma faciale	615943	**<10**	**<10**
Granulomatosis with polyangiitis	900	**2449**	**2270**
Granulomatous mastitis	64722	**73**	**72**
Granulomatous slack skin	33111	**<10**	**<10**
Gray platelet syndrome	721	**38**	**38**
Greenberg dysplasia	1426	**<10**	**<10**
Greig cephalopolysyndactyly syndrome	380	**236**	**235**
GRFoma	97261	**<10**	**<10**
GRIN2B-related developmental delay, intellectual disability and autism spectrum disorder	589547	**59**	**59**
Griscelli syndrome	381	**26**	**24**
Growing teratoma syndrome	314613	**<10**	**<10**
Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	391348	**<10**	**<10**
Growth delay due to insulin-like growth factor I resistance	73273	**42**	**42**
Growth delay due to insulin-like growth factor type 1 deficiency	73272	**44**	**44**
Growth delay-hydrocephaly-lung hypoplasia syndrome	3035	**<10**	**<10**
Grubben-de Cock-Borghgraef syndrome	2101	**12**	**12**
Guanidinoacetate methyltransferase deficiency	382	**36**	**35**
Guillain-Barré syndrome*	2103	**2035**	**1868**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Guttmacher syndrome	2957	**<10**	**<10**
Gyrate atrophy of choroid and retina	414	**44**	**44**
H syndrome	168569	**26**	**26**
Haddad syndrome	99803	**23**	**15**
Hailey-Hailey disease	2841	**247**	**240**
Hajdu-Cheney syndrome	955	**50**	**46**
Hallermann-Streiff syndrome	2108	**25**	**23**
Hallermann-Streiff-like syndrome	2109	**<10**	**<10**
Hallux varus-preaxial polysyndactyly syndrome	2110	**<10**	**<10**
HANAC syndrome	73229	**33**	**29**
Hand-foot-genital syndrome	2438	**<10**	**<10**
Hao-Fountain syndrome	643549	**15**	**15**
Hardikar syndrome	1415	**<10**	**<10**
Harlequin ichthyosis	457	**26**	**23**
Harlequin syndrome	199282	**11**	**10**
Harrod syndrome	2115	**<10**	**<10**
Hartnup disease	2116	**<10**	**<10**
Hartsfield syndrome	2117	**<10**	**<10**
Hawkinsinuria	2118	**<10**	**<10**
Heart defects-limb shortening syndrome	1354	**10**	**<10**
Heart defect-tongue hamartoma-polysyndactyly syndrome	1338	**<10**	**<10**
Heart-hand syndrome type 2	1350	**<10**	**<10**
Heart-hand syndrome, Slovenian type	168796	**12**	**11**
Heavy chain disease	86864	**15**	**14**
Heiner syndrome	99932	**<10**	**<10**
Helicoid peripapillary chorioretinal degeneration	86813	**<10**	**<10**
HELLP syndrome	244242	**86**	**86**
Hemangioblastoma	252054	**12**	**11**
Hemiconvulsion-hemiplegia-epilepsy syndrome	86908	**82**	**82**
Hemidystonia-hemiatrophy syndrome	306741	**<10**	**<10**
Hemifacial hyperplasia	141145	**28**	**28**
Hemifacial spasm	221083	**342**	**331**
Hemihyperplasia-multiple lipomatosis syndrome	276280	**<10**	**<10**
Hemimegalencephaly	99802	**109**	**107**
Hemoglobin C disease	2132	**264**	**263**
Hemoglobin C-beta-thalassemia syndrome	231242	**53**	**53**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hemoglobin D disease	90039	**<10**	**<10**
Hemoglobin E disease	2133	**71**	**71**
Hemoglobin E-beta-thalassemia syndrome	231249	**61**	**59**
Hemoglobin Lepore-beta-thalassemia syndrome	330032	**<10**	**<10**
Hemoglobin M disease	330041	**12**	**12**
Hemoglobinopathy Toms River	280615	**<10**	**<10**
Hemolytic anemia due to glucophosphate isomerase deficiency	712	**<10**	**<10**
Hemolytic anemia due to glutathione reductase deficiency	90030	**<10**	**<10**
Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency	35120	**<10**	**<10**
Hemolytic anemia due to red cell pyruvate kinase deficiency	766	**151**	**144**
Hemolytic disease of the newborn with Kell alloimmunization	275944	**<10**	**<10**
Hemolytic uremic syndrome with DGKE deficiency	357008	**<10**	**<10**
Hemophagocytic syndrome associated with an infection	158048	**<10**	**<10**
Hemophilia A	98878	**9945**	**9715**
Hemophilia B	98879	**2427**	**2371**
Hemophilia*	448	**41**	**40**
Hemorrhagic fever-renal syndrome	340	**<10**	**<10**
Hennekam syndrome	2136	**32**	**31**
Hepatic cystic hamartoma	386	**65**	**63**
Hepatic fibrosis-renal cysts-intellectual disability syndrome	2031	**<10**	**<10**
Hepatic veno-occlusive disease	890	**369**	**303**
Hepatic veno-occlusive disease-immunodeficiency syndrome	79124	**<10**	**<10**
Hepatitis delta	402823	**<10**	**<10**
Hepatoblastoma	449	**24**	**23**
Hepatocellular adenoma	54272	**89**	**85**
Hepatocellular carcinoma*	88673	**16**	**11**
Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	137681	**<10**	**<10**
Hepatoerythropoietic porphyria	95159	**<10**	**<10**
Hepatosplenic T-cell lymphoma	86882	**<10**	**<10**
Hereditary acrokeratotic poikiloderma	2907	**<10**	**<10**
Hereditary amyloidosis with primary renal involvement	85450	**175**	**154**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hereditary angioedema with C1Inh deficiency	528623	**1103**	**1091**
Hereditary angioedema with normal C1Inh	528647	**447**	**442**
Hereditary angioedema*	91378	**69**	**67**
Hereditary arterial and articular multiple calcification syndrome	289601	**<10**	**<10**
Hereditary atrial fibrillation	334	**178**	**175**
Hereditary atrial tachyarrhythmia-infra-Hisian cardiac conduction disease	436242	**<10**	**<10**
Hereditary ATTR amyloidosis	271861	**2977**	**2541**
Hereditary benign intraepithelial dyskeratosis	352657	**10**	**10**
Hereditary breast and/or ovarian cancer syndrome	145	**1105**	**985**
Hereditary breast cancer	227535	**58**	**52**
Hereditary butyrylcholinesterase deficiency	132	**29**	**29**
Hereditary combined deficiency of vitamin K-dependent clotting factors	98434	**24**	**24**
Hereditary continuous muscle fiber activity	972	**12**	**10**
Hereditary coproporphyria	79273	**86**	**68**
Hereditary diffuse gastric cancer	26106	**<10**	**<10**
Hereditary elliptocytosis	288	**174**	**174**
Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome	221043	**<10**	**<10**
Hereditary folate malabsorption	90045	**12**	**11**
Hereditary fructose intolerance	469	**206**	**202**
Hereditary geniospasm	53372	**<10**	**<10**
Hereditary gingival fibromatosis	2024	**13**	**13**
Hereditary hemorrhagic telangiectasia	774	**4227**	**4022**
Hereditary hypercarotenemia and vitamin A deficiency	199285	**<10**	**<10**
Hereditary hyperekplexia	3197	**75**	**69**
Hereditary hyperferritinemia-cataract syndrome	163	**191**	**189**
Hereditary hypophosphatemic rickets with hypercalciuria	157215	**86**	**86**
Hereditary inclusion body myopathy type 4	324381	**<10**	**<10**
Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome	79091	**<10**	**<10**
Hereditary isolated aplastic anemia	397692	**43**	**41**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hereditary late-onset Parkinson disease	411602	**51**	**43**
Hereditary leiomyomatosis and renal cell cancer	523	**312**	**306**
Hereditary methemoglobinemia	621	**23**	**23**
Hereditary mixed polyposis syndrome	157794	**<10**	**<10**
Hereditary motor and sensory neuropathy type 5	64751	**<10**	**<10**
Hereditary motor and sensory neuropathy type 6	90120	**43**	**42**
Hereditary motor and sensory neuropathy with acrodystrophy	90119	**<10**	**<10**
Hereditary motor and sensory neuropathy, Okinawa type	90117	**<10**	**<10**
Hereditary mucoepithelial dysplasia	1839	**12**	**12**
Hereditary myopathy with early respiratory failure	178464	**110**	**106**
Hereditary myopathy with lactic acidosis due to ISCU deficiency	43115	**<10**	**<10**
Hereditary neurocutaneous malformation	1062	**<10**	**<10**
Hereditary neuroendocrine tumor of small intestine	456333	**<10**	**<10**
Hereditary neuropathy with liability to pressure palsies	640	**1479**	**1451**
Hereditary neutrophilia	279943	**17**	**17**
Hereditary nonpolyposis colon cancer*	443909	**55**	**41**
Hereditary orotic aciduria	30	**<10**	**<10**
Hereditary painful callosities	79141	**14**	**14**
Hereditary papillary renal cell carcinoma	47044	**12**	**10**
Hereditary persistence of alpha-fetoprotein	168615	**<10**	**<10**
Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome	46532	**<10**	**<10**
Hereditary persistence of fetal hemoglobin-intellectual disability syndrome	619233	**14**	**14**
Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	251380	**66**	**65**
Hereditary pheochromocytoma-paraganglioma	29072	**1060**	**1031**
Hereditary progressive cardiac conduction defect	871	**257**	**249**
Hereditary progressive mucinous histiocytosis	158025	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hereditary pulmonary alveolar proteinosis	264675	**26**	**24**
Hereditary renal hypouricemia	94088	**10**	**10**
Hereditary sensorimotor neuropathy with hyperelastic skin	280598	**<10**	**<10**
Hereditary sensory and autonomic neuropathy due to TECPR2 mutation	320385	**<10**	**<10**
Hereditary sensory and autonomic neuropathy type 1	36386	**31**	**29**
Hereditary sensory and autonomic neuropathy type 1B	139564	**15**	**15**
Hereditary sensory and autonomic neuropathy type 2	970	**22**	**21**
Hereditary sensory and autonomic neuropathy type 4	642	**21**	**18**
Hereditary sensory and autonomic neuropathy type 5	64752	**21**	**19**
Hereditary sensory and autonomic neuropathy type 6	314381	**<10**	**<10**
Hereditary sensory and autonomic neuropathy with deafness and global delay	139573	**<10**	**<10**
Hereditary sensory and autonomic neuropathy*	140471	**105**	**99**
Hereditary sensory neuropathy-deafness-dementia syndrome	456318	**<10**	**<10**
Hereditary sick sinus syndrome	166282	**18**	**15**
Hereditary sodium channelopathy-related small fibers neuropathy	306577	**504**	**491**
Hereditary spherocytosis	822	**1775**	**1765**
Hereditary steroid-resistant nephrotic syndrome	656	**279**	**272**
Hereditary stomatocytosis*	98365	**25**	**25**
Hereditary thermosensitive neuropathy	84093	**<10**	**<10**
Hereditary thrombocytopenia with early-onset myelofibrosis	480851	**<10**	**<10**
Hereditary thrombocytopenia with normal platelets	268322	**97**	**95**
Hereditary thrombophilia due to congenital antithrombin deficiency	82	**59**	**59**
Hereditary xanthinuria	3467	**13**	**13**
Hermansky-Pudlak syndrome	79430	**95**	**89**
Herpes simplex virus encephalitis	1930	**116**	**108**
Herpes simplex virus stromal keratitis	137599	**293**	**289**
Herpetiform pemphigus	208524	**25**	**23**
Hidrotic ectodermal dysplasia	189	**124**	**123**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
High bone mass osteogenesis imperfecta	314029	**<10**	**<10**
High myopia-sensorineural deafness syndrome	363396	**<10**	**<10**
High-grade astrocytoma*	251561	**<10**	**<10**
Hinman syndrome	84085	**193**	**188**
Hip dysplasia, Beukes type	2114	**11**	**11**
Hirschsprung disease	388	**2005**	**1973**
Hirschsprung disease-ganglioneuroblastoma syndrome	2151	**<10**	**<10**
Hirschsprung disease-nail hypoplasia-dysmorphism syndrome	2153	**<10**	**<10**
His bundle tachycardia	3283	**33**	**33**
Histidinemia	2157	**10**	**10**
Histiocytic sarcoma	86896	**27**	**15**
Histiocytoid cardiomyopathy	137675	**<10**	**<10**
Histoplasmosis	390	**<10**	**<10**
HJV or HAMP-related hemochromatosis	79230	**64**	**62**
HNRNPA1-related adult-onset distal myopathy	399086	**<10**	**<10**
HNRNPDL-related limb-girdle muscular dystrophy D3	55596	**<10**	**<10**
Hodgkin lymphoma*	98293	**54**	**54**
Holmes-Adie syndrome	454718	**<10**	**<10**
Holocarboxylase synthetase deficiency	79242	**11**	**11**
Holoprosencephaly	2162	**138**	**116**
Holoprosencephaly-craniosynostosis syndrome	2163	**<10**	**<10**
Holoprosencephaly-postaxial polydactyly syndrome	2166	**<10**	**<10**
Holoprosencephaly-radial heart renal anomalies syndrome	3186	**<10**	**<10**
Holt-Oram syndrome	392	**332**	**321**
Holzgreve syndrome	2167	**<10**	**<10**
Homocystinuria due to cystathionine beta-synthase deficiency	394	**244**	**233**
Homocystinuria due to methylene tetrahydrofolate reductase deficiency	395	**162**	**160**
Homocystinuria without methylmalonic aciduria	622	**34**	**31**
Homozygous 2p21 microdeletion syndrome*	369886	**<10**	**<10**
Homozygous familial hypercholesterolemia	391665	**190**	**189**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Horizontal gaze palsy with progressive scoliosis	2744	**16**	**16**
Hot water reflex epilepsy	166412	**<10**	**<10**
Hoyeraal-Hreidarsson syndrome	3322	**<10**	**<10**
HSD10 disease	391417	**<10**	**<10**
HTRA1-related autosomal dominant cerebral small vessel disease	482077	**40**	**39**
Huntington disease	399	**3647**	**3041**
Huntington disease-like 1	157941	**<10**	**<10**
Huntington disease-like 2	98934	**46**	**41**
Huntington disease-like 3	157946	**<10**	**<10**
Huntington disease-like syndrome due to C9ORF72 expansions	401901	**<10**	**<10**
Huntington disease-like syndrome*	158266	**<10**	**<10**
Huriez syndrome	384	**<10**	**<10**
Hutchinson-Gilford progeria syndrome	740	**20**	**19**
Hyaline fibromatosis syndrome	498474	**17**	**11**
Hydatidiform mole	99927	**10**	**10**
Hydranencephaly	2177	**<10**	**<10**
Hydroa vacciniforme	330058	**<10**	**<10**
Hydroa vacciniforme-like lymphoma	364039	**<10**	**<10**
Hydrocephalus-blue sclerae-nephropathy syndrome	2186	**<10**	**<10**
Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome	2180	**<10**	**<10**
Hydrocephalus-obesity-hypogonadism syndrome	2183	**<10**	**<10**
Hydrocephaly-cerebellar agenesis syndrome	1397	**<10**	**<10**
Hydrocephaly-tall stature-joint laxity syndrome	2181	**<10**	**<10**
Hydrops fetalis	1041	**36**	**23**
Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome	528091	**<10**	**<10**
Hyperammonemia due to N-acetylglutamate synthase deficiency	927	**31**	**26**
Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	401948	**<10**	**<10**
Hyperandrogenism due to cortisone reductase deficiency	168588	**<10**	**<10**
Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	209902	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hyperekplexia*	306773	**12**	**12**
Hyperekplexia-epilepsy syndrome	163985	**<10**	**<10**
Hypereosinophilic syndrome*	168956	**320**	**308**
Hypergonadotropic hypogonadism-cataract syndrome	2410	**<10**	**<10**
Hyper-IgE syndrome*	331223	**21**	**20**
Hyper-IgM syndrome with susceptibility to opportunistic infections	183663	**41**	**39**
Hyper-IgM syndrome without susceptibility to opportunistic infections	183666	**12**	**12**
Hyperinsulinism due to HNF1A deficiency	324575	**<10**	**<10**
Hyperinsulinism due to INSR deficiency	263458	**<10**	**<10**
Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	71212	**21**	**19**
Hyperinsulinism-hyperammonemia syndrome	35878	**47**	**47**
Hyperkalemic periodic paralysis	682	**116**	**112**
Hyperkeratosis lenticularis perstans	409	**<10**	**<10**
Hyperkeratosis-hyperpigmentation syndrome	1336	**<10**	**<10**
Hyperlysinemia	2203	**<10**	**<10**
Hypermethioninemia due to glycine N-methyltransferase deficiency	289891	**<10**	**<10**
Hypermethioninemia encephalopathy due to adenosine kinase deficiency	289290	**<10**	**<10**
Hypermobile Ehlers-Danlos syndrome	285	**2988**	**2975**
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	415	**26**	**25**
Hyperostosis corticalis generalisata	3416	**<10**	**<10**
Hyperostosis cranialis interna	443098	**<10**	**<10**
Hyperparathyroidism-jaw tumor syndrome	99880	**32**	**32**
Hyperphenylalaninemia due to DNAJC12 deficiency	508523	**<10**	**<10**
Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	238583	**129**	**127**
Hyperphosphatasia-intellectual disability syndrome	247262	**15**	**14**
Hyperprolinemia type 1	419	**23**	**22**
Hyperprolinemia type 2	79101	**<10**	**<10**
Hypersensitivity pneumonitis*	31740	**924**	**790**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hypertelorism-hypospadias-polysyndactyly syndrome	2211	**<10**	**<10**
Hypertelorism-microtia-facial clefting syndrome	2213	**<10**	**<10**
Hypertelorism-preauricular sinus-punctual pits-deafness syndrome	293958	**<10**	**<10**
Hypertension due to gain-of-function mutations in the mineralocorticoid receptor	88660	**<10**	**<10**
Hypertrichosis lanuginosa congenita	2222	**18**	**18**
Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation	324525	**<10**	**<10**
Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome	363694	**<10**	**<10**
Hyperzincemia and hypercalprotectinemia	251523	**<10**	**<10**
Hypnic headache	276429	**<10**	**<10**
Hypo- and hypermelanotic cutaneous macules-retarded growth-intellectual disability syndrome	2435	**<10**	**<10**
Hypobetalipoproteinemia*	31154	**31**	**31**
Hypocalcemic rickets*	289103	**66**	**65**
Hypocalcemic vitamin D-dependent rickets	289157	**201**	**196**
Hypocalcemic vitamin D-resistant rickets	93160	**52**	**52**
Hypochondroplasia	429	**450**	**448**
Hypocomplementemic urticarial vasculitis	36412	**155**	**152**
Hypodontia-dysplasia of nails syndrome	2228	**10**	**10**
Hypoglossia-hypodactyly syndrome	989	**28**	**27**
Hypogonadism-mitral valve prolapse-intellectual disability syndrome	2233	**<10**	**<10**
Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome	2235	**<10**	**<10**
Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome	293967	**<10**	**<10**
Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome	528105	**11**	**11**
Hypohidrotic ectodermal dysplasia	238468	**964**	**960**
Hypohidrotic ectodermal dysplasia with immunodeficiency	98813	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome	1882	**32**	**32**
Hypohidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome	69088	**10**	**<10**
Hypoinsulinemic hypoglycemia and body hemihypertrophy	293964	**<10**	**<10**
Hypokalemic periodic paralysis	681	**268**	**264**
Hypomandibular faciocranial dysostosis	1790	**<10**	**<10**
Hypomyelination neuropathy-arthrogryposis syndrome	2680	**<10**	**<10**
Hypomyelination of early myelinating structures	599376	**<10**	**<10**
Hypomyelination with atrophy of basal ganglia and cerebellum	139441	**11**	**11**
Hypomyelination with brain stem and spinal cord involvement and leg spasticity	363412	**<10**	**<10**
Hypomyelination-congenital cataract syndrome	85163	**10**	**10**
Hypoparathyroidism-sensorineural deafness-renal disease syndrome	2237	**132**	**129**
Hypophosphatasia	436	**542**	**532**
Hypophosphatemic rickets*	437	**456**	**454**
Hypopigmentation-punctate palmoplantar keratoderma syndrome	324561	**<10**	**<10**
Hypoplasia of the mitral valve annulus	99058	**<10**	**<10**
Hypoplasminogenemia	722	**11**	**11**
Hypoplastic left heart syndrome	2248	**264**	**199**
Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	293864	**<10**	**<10**
Hypoplastic right heart syndrome*	98723	**<10**	**<10**
Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome	2250	**<10**	**<10**
Hypothalamic adipsic hypernatraemia syndrome	443101	**<10**	**<10**
Hypothyroidism due to deficient transcription factors involved in pituitary development or function	226307	**<10**	**<10**
Hypothyroidism due to TSH receptor mutations	90673	**100**	**98**
Hypotonia-cystinuria syndrome	163690	**<10**	**<10**
Hypotonia-cystinuria type 1 syndrome*	238517	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hypotonia-failure to thrive-microcephaly syndrome	79507	**62**	**57**
Hypotonia-speech impairment-severe cognitive delay syndrome	371364	**12**	**12**
Hypotrichosis simplex	55654	**43**	**43**
Hypotrichosis simplex of the scalp	90368	**<10**	**<10**
Hypotrichosis with juvenile macular degeneration	1573	**<10**	**<10**
Hypotrichosis-deafness syndrome	330029	**<10**	**<10**
Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	69735	**<10**	**<10**
Hypoxanthine guanine phosphoribosyltransferase partial deficiency	79233	**10**	**10**
ICF syndrome	2268	**20**	**17**
Ichthyosis follicularis-alopecia-photophobia syndrome	2273	**17**	**17**
Ichthyosis hystrix of Curth-Macklin	79503	**10**	**10**
Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome	2269	**<10**	**<10**
Ichthyosis-hypotrichosis syndrome	91132	**<10**	**<10**
Ichthyosis-prematurity syndrome	88621	**21**	**20**
Ichthyosis-short stature-brachydactyly-microspherophakia syndrome	363992	**10**	**10**
Idiopathic achalasia	930	**138**	**136**
Idiopathic acute eosinophilic pneumonia	724	**15**	**12**
Idiopathic aplastic anemia	88	**1125**	**1041**
Idiopathic bilateral vestibulopathy	171684	**119**	**119**
Idiopathic bronchiectasis	60033	**2242**	**2174**
Idiopathic camptocormia	1320	**244**	**208**
Idiopathic catatonia	648919	**32**	**30**
Idiopathic CD4 lymphocytopenia	228000	**73**	**71**
Idiopathic chronic eosinophilic pneumonia	2902	**168**	**162**
Idiopathic congenital hypothyroidism	95717	**786**	**784**
Idiopathic copper-associated cirrhosis	209919	**<10**	**<10**
Idiopathic dropped head syndrome	447881	**25**	**22**
Idiopathic ductopenia	480512	**29**	**27**
Idiopathic eosinophilic myositis	247724	**<10**	**<10**
Idiopathic eosinophilic pneumonia*	182101	**26**	**25**
Idiopathic gastroparesis	558411	**187**	**184**
Idiopathic giant cell myocarditis	329874	**80**	**75**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Idiopathic hypercalciuria	2197	**1312**	**1307**
Idiopathic hypereosinophilic syndrome	3260	**716**	**682**
Idiopathic hypersomnia	33208	**4206**	**4187**
Idiopathic interstitial pneumonia*	98300	**718**	**635**
Idiopathic intracranial hypertension	238624	**110**	**106**
Idiopathic isolated micropenis	95707	**558**	**554**
Idiopathic juvenile osteoporosis	85193	**811**	**774**
Idiopathic linear interstitial keratitis	314017	**<10**	**<10**
Idiopathic localized lipodystrophy	90158	**<10**	**<10**
Idiopathic macular telangiectasia type 1	353344	**13**	**13**
Idiopathic macular telangiectasia type 3	353351	**<10**	**<10**
Idiopathic malabsorption due to bile acid synthesis defects	84065	**<10**	**<10**
Idiopathic neonatal atrial flutter	45452	**<10**	**<10**
Idiopathic nephrotic syndrome*	357502	**1085**	**1052**
Idiopathic non-lupus full-house nephropathy	567544	**11**	**11**
Idiopathic optic perineuritis	499107	**<10**	**<10**
Idiopathic panuveitis	280921	**1368**	**1363**
Idiopathic peliosis hepatis	480524	**172**	**168**
Idiopathic phalangeal acro-osteolysis	444316	**<10**	**<10**
Idiopathic pleuroparenchymal fibroelastosis	494428	**328**	**271**
Idiopathic posterior uveitis	280917	**672**	**664**
Idiopathic pregnancy-associated osteoporosis	647823	**<10**	**<10**
Idiopathic pulmonary artery dilatation	1676	**10**	**10**
Idiopathic pulmonary fibrosis	2032	**6278**	**4574**
Idiopathic pulmonary hemosiderosis	99931	**68**	**67**
Idiopathic recurrent pericarditis	251307	**575**	**569**
Idiopathic scleritis	648675	**129**	**128**
Idiopathic small fibers neuropathy	658549	**105**	**105**
Idiopathic spontaneous coronary artery dissection	458718	**96**	**96**
Idiopathic steroid-resistant nephrotic syndrome	567548	**380**	**373**
Idiopathic steroid-sensitive nephrotic syndrome	69061	**4252**	**4197**
Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance	567546	**50**	**50**
Idiopathic subglottic stenosis	652681	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Idiopathic trachyonychia	79153	**<10**	**<10**
Idiopathic uveal effusion syndrome	209956	**<10**	**<10**
Idiopathic ventricular fibrillation	228140	**999**	**757**
Idiopathic/heritable pulmonary arterial hypertension	422	**4040**	**3219**
IgA pemphigus	555905	**20**	**20**
IgG4-related disease*	284264	**351**	**318**
IgG4-related systemic disease	596448	**1280**	**1202**
Ileal neuroendocrine tumor	100078	**<10**	**<10**
IMAGe syndrome	85173	**16**	**16**
Imerslund-Gräsbeck syndrome	35858	**58**	**58**
Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome	238569	**689**	**689**
Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections-lymphopenia syndrome	529977	**12**	**12**
Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	37042	**68**	**57**
Immune thrombocytopenia	3002	**9799**	**9258**
Immune-mediated necrotizing myopathy	206569	**894**	**812**
Immune-mediated scleritis	648681	**22**	**22**
Immunodeficiency by defective expression of MHC class I	34592	**<10**	**<10**
Immunodeficiency by defective expression of MHC class II	572	**36**	**31**
Immunodeficiency due to a classical component pathway complement deficiency	169147	**60**	**60**
Immunodeficiency due to a late component of complement deficiency	169150	**52**	**52**
Immunodeficiency due to CD25 deficiency	169100	**<10**	**<10**
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	70593	**93**	**92**
Immunodeficiency with factor H anomaly	200421	**<10**	**<10**
Immunodeficiency with factor I anomaly	200418	**<10**	**<10**
Immunoglobulin A nephropathy	34145	**2018**	**1914**
Immunoglobulin A vasculitis	761	**3419**	**3342**
Immunoglobulin heavy chain deficiency	169110	**<10**	**<10**
Immunotactoid glomerulopathy	97567	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Immunotactoid or fibrillary glomerulopathy*	91137	**18**	**18**
Immunotherapy induced hypophysitis	641350	**77**	**74**
Incessant infant ventricular tachycardia	45453	**<10**	**<10**
Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	52430	**29**	**16**
Inclusion body myositis	611	**1457**	**1137**
Incontinentia pigmenti	464	**707**	**701**
Indeterminate cell histiocytosis	158019	**14**	**13**
Indolent primary cutaneous T-cell lymphoma*	178548	**<10**	**<10**
Indolent systemic mastocytosis	98848	**2212**	**2184**
Infantile apnea	70590	**2639**	**2589**
Infantile bilateral striatal necrosis*	1576	**<10**	**<10**
Infantile cerebellar-retinal degeneration	313850	**<10**	**<10**
Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	402364	**<10**	**<10**
Infantile choroidocerebral calcification syndrome	1313	**<10**	**<10**
Infantile convulsions and choreoathetosis	31709	**14**	**13**
Infantile digital fibromatosis	199267	**<10**	**<10**
Infantile dystonia-parkinsonism	238455	**12**	**11**
Infantile epileptic-dyskinetic encephalopathy	364063	**33**	**30**
Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	352563	**<10**	**<10**
Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome	522077	**<10**	**<10**
Infantile LAD-like disease due to RAC2 deficiency	183707	**10**	**10**
Infantile mercury poisoning	247165	**<10**	**<10**
Infantile multisystem neurologic-endocrine-pancreatic disease	456312	**<10**	**<10**
Infantile myofibromatosis	2591	**25**	**25**
Infantile neuroaxonal dystrophy	35069	**71**	**34**
Infantile neurodegeneration-progressive spasticity-intellectual disability-white matter lesions syndrome	641353	**<10**	**<10**
Infantile neurovisceral acid sphingomyelinase deficiency	77292	**31**	**15**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Infantile onset panniculitis with uveitis and systemic granulomatosis	251304	**14**	**13**
Infantile osteopetrosis with neuroaxonal dysplasia	85179	**<10**	**<10**
Infantile Refsum disease	772	**14**	**13**
Infantile spasms-broad thumbs syndrome	3173	**<10**	**<10**
Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome	263410	**<10**	**<10**
Infantile-onset ascending hereditary spastic paralysis	293168	**<10**	**<10**
Infantile-onset autosomal recessive nonprogressive cerebellar ataxia	284332	**14**	**14**
Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome	457205	**<10**	**<10**
Infantile-onset generalized dyskinesia with orofacial involvement	494526	**<10**	**<10**
Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression	391316	**<10**	**<10**
Infantile-onset periodic fever-panniculitis-dermatosis syndrome	500062	**<10**	**<10**
Infantile-onset pulmonary alveolar proteinosis-hypogammaglobulinemia	572428	**<10**	**<10**
Infantile-onset spinocerebellar ataxia	1186	**77**	**75**
Infantile-onset X-linked spinal muscular atrophy	1145	**11**	**<10**
Infection-related hemolytic uremic syndrome	544482	**2052**	**2007**
Infectious anterior uveitis	279922	**347**	**343**
Infectious epithelial keratitis	137593	**81**	**79**
Infectious panuveitis	279925	**107**	**102**
Infectious posterior uveitis	279919	**248**	**238**
Infectious scleritis	648665	**<10**	**<10**
Infective endocarditis	570762	**25**	**24**
Inferior vena cava interruption without azygos continuation	99123	**<10**	**<10**
Inflammatory bowel disease-recurrent sinopulmonary infections syndrome	529980	**<10**	**<10**
Inflammatory myofibroblastic tumor	178342	**19**	**19**
Inflammatory myopathy with abundant macrophages	247718	**11**	**11**
Inflammatory pseudotumor of the liver	90003	**11**	**<10**
Infundibulo-neurohypophysitis	238305	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Inhalational anthrax	247257	**<10**	**<10**
Inherited acute myeloid leukemia	319465	**10**	**<10**
Inherited arrhythmogenic cardiomyopathy*	247	**1047**	**998**
Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations	319462	**16**	**16**
Inherited congenital spastic tetraplegia	210141	**26**	**23**
Inherited Creutzfeldt-Jakob disease	282166	**<10**	**<10**
Inherited epidermodysplasia verruciformis	302	**34**	**32**
Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency	289548	**<10**	**<10**
Inherited isolated arrhythmogenic cardiomyopathy	217656	**2469**	**2413**
Insulin autoimmune syndrome	411593	**<10**	**<10**
Insulinoma	97279	**37**	**35**
Insulin-resistance syndrome type A	2297	**32**	**32**
Insulin-resistance syndrome type B	2298	**<10**	**<10**
Intellectual disability, Wolff type	3080	**<10**	**<10**
Intellectual disability-alacrima-achalasia syndrome	289483	**<10**	**<10**
Intellectual disability-autism-speech apraxia-craniofacial dysmorphism syndrome	529965	**<10**	**<10**
Intellectual disability-brachydactyly-Pierre Robin syndrome	364577	**<10**	**<10**
Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome	508498	**<10**	**<10**
Intellectual disability-cataracts-calcified pinnae-myopathy syndrome	3042	**36**	**36**
Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	397709	**<10**	**<10**
Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome	3044	**<10**	**<10**
Intellectual disability-early-onset cataract-microcephaly syndrome	633035	**<10**	**<10**
Intellectual disability-epilepsy-extrapyramidal syndrome	468620	**10**	**10**
Intellectual disability-expressive aphasia-facial dysmorphism syndrome	436151	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Intellectual disability-facial dysmorphism-hand anomalies syndrome	370010	**<10**	**<10**
Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome	1495	**20**	**19**
Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome	457279	**17**	**17**
Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome	457365	**<10**	**<10**
Intellectual disability-myopathy-short stature-endocrine defect syndrome	3068	**<10**	**<10**
Intellectual disability-nasal speech-craniofacial dysmorphism syndrome	697760	**<10**	**<10**
Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome	352530	**11**	**11**
Intellectual disability-peripheral spasticity-exudative vitreoretinopathy syndrome	404473	**53**	**52**
Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome	513456	**22**	**22**
Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	369837	**<10**	**<10**
Intellectual disability-seizures-macrocephaly-obesity syndrome	369950	**<10**	**<10**
Intellectual disability-short stature-hypertelorism syndrome	3074	**<10**	**<10**
Intellectual disability-spasticity-ectrodactyly syndrome	1891	**<10**	**<10**
Intellectual disability-speech delay-dysmorphic features-T cell abnormalities syndrome	662829	**<10**	**<10**
Intellectual disability-strabismus syndrome	363528	**28**	**28**
Interatrial communication	1478	**1558**	**1529**
Intermediate atrioventricular septal defect	576242	**165**	**162**
Intermediate collagen VI-related muscular dystrophy	646113	**12**	**12**
Intermediate generalized junctional epidermolysis bullosa	79402	**12**	**12**
Intermediate nemaline myopathy	171433	**11**	**11**
Intermediate osteopetrosis	210110	**<10**	**<10**
Intermediate uveitis	279914	**827**	**822**
Internal carotid absence	981	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Interstitial cystitis	37202	**<10**	**<10**
Interstitial granulomatous dermatitis with arthritis	79099	**<10**	**<10**
Interstitial lung disease due to ABCA3 deficiency	440402	**16**	**13**
Interstitial lung disease due to SP-C deficiency	440392	**57**	**52**
Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome	306504	**<10**	**<10**
Interventricular septum aneurysm	99092	**<10**	**<10**
Intestinal lymphangiectasia*	36204	**30**	**30**
Intestinal polyposis syndrome*	104010	**60**	**53**
Intraductal papillary mucinous carcinoma of pancreas	424058	**73**	**69**
Intrahepatic cholestasis of pregnancy	69665	**146**	**145**
Intramedullary non-dysraphic spinal cord lipoma	645359	**<10**	**<10**
Intraneural perineurioma	100003	**<10**	**<10**
Intrauterine growth restriction-congenital multiple café-au-lait macules-increased sister chromatid exchange syndrome	508512	**15**	**15**
Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	436144	**<10**	**<10**
Intrauterine growth retardation-micrognathia-short stature-facial dysmorphism-rhizomelic shortening syndrome	659702	**<10**	**<10**
Intravascular large B-cell lymphoma	98839	**<10**	**<10**
Intravascular papillary endothelial hyperplasia	673525	**<10**	**<10**
Invasive candidiasis	636945	**<10**	**<10**
Invasive mole	99925	**<10**	**<10**
Invasive non-typhoidal salmonellosis	324648	**<10**	**<10**
Inverted duplicated chromosome 15 syndrome	3306	**122**	**116**
IRF2BPL-related regressive neurodevelopmental disorder-dystonia-seizures syndrome	597623	**<10**	**<10**
IRIDA syndrome	209981	**23**	**22**
Iridocorneal endothelial syndrome	64734	**14**	**14**
IRVAN syndrome	209943	**11**	**11**
Isaacs syndrome	84142	**132**	**121**
Isobutyryl-CoA dehydrogenase deficiency	79159	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isochromosomy Yp syndrome	98797	<10	<10
Isochromosomy Yq syndrome	98798	<10	<10
Isolated absence of both forearm and hand	294979	214	213
Isolated absence of both lower leg and foot	294981	<10	<10
Isolated absence of upper arm and forearm with hand present	294975	<10	<10
Isolated absence/hypoplasia of fingers excluding thumb, unilateral	973	99	99
Isolated acheiria	294983	<10	<10
Isolated acheiropodia	931	<10	<10
Isolated agenesis of gallbladder	440987	10	<10
Isolated amelia of lower limb	294969	<10	<10
Isolated amelia of upper limb	294967	<10	<10
Isolated amyelia	268868	<10	<10
Isolated anencephaly/exencephaly	1048	<10	<10
Isolated angioid streaks	674943	<10	<10
Isolated aniridia	250923	552	546
Isolated ankyloblepharon filiforme adnatum	91397	<10	<10
Isolated anterior cervical hypertrichosis	3387	<10	<10
Isolated apodia	294986	<10	<10
Isolated arhinencephaly	268936	<10	<10
Isolated arrhinia	1134	<10	<10
Isolated asymptomatic elevation of creatine phosphokinase	206599	642	617
Isolated ATP synthase deficiency	254913	10	10
Isolated atrial standstill	1344	41	36
Isolated autosomal dominant hypomagnesemia, Glaudemans type	199326	<10	<10
Isolated bilateral hemispheric cerebellar hypoplasia	269221	<10	<10
Isolated biliary atresia	30391	1633	1540
Isolated blepharochalasis	519390	<10	<10
Isolated bone marrow mastocytosis	158778	14	14
Isolated cerebellar agenesis	1398	254	243
Isolated cerebellar vermis agenesis	269203	11	10
Isolated cerebellar vermis hypoplasia	199630	88	87
Isolated childhood apraxia of speech	209908	80	80
Isolated cleft lip	199302	719	718
Isolated complex I deficiency	2609	83	59
Isolated complex III deficiency	1460	37	32

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isolated congenital alacrima	91416	23	23
Isolated congenital anonychia	79143	<10	<10
Isolated congenital anosmia	88620	12	12
Isolated congenital auditory ossicle malformation	162526	114	114
Isolated congenital breast hypoplasia/ aplasia	180188	28	28
Isolated congenital ectropion	99171	<10	<10
Isolated congenital entropion	519386	<10	<10
Isolated congenital hepatic fibrosis	485426	235	222
Isolated congenital hypoglossia/ aglossia	141152	<10	<10
Isolated congenital hypogonadotropic hypogonadism	238666	2448	2425
Isolated congenital laryngeal web	2374	24	23
Isolated congenital megalocornea	91489	49	48
Isolated congenital microcephaly	199642	541	517
Isolated congenital nasal pyriform aperture stenosis	162516	173	170
Isolated congenital onychodysplasia	79144	<10	<10
Isolated congenital radial head dislocation	295032	<10	<10
Isolated congenital sclerocornea	91490	38	38
Isolated congenital syngnathia	141214	<10	<10
Isolated corpus callosum agenesis	200	760	754
Isolated cryptophthalmia	91396	<10	<10
Isolated cytochrome C oxidase deficiency	254905	31	25
Isolated Dandy-Walker malformation	217	99	92
Isolated delta-storage pool disease	248340	165	161
Isolated diffuse palmoplantar keratoderma*	307148	107	106
Isolated digestive duplication cyst of the tongue	141071	<10	<10
Isolated distal symphalangism	3248	10	10
Isolated distichiasis	99177	<10	<10
Isolated ectopia lentis	1885	171	169
Isolated encephalocele	199647	31	30
Isolated esophageal duplication cyst	100047	12	12
Isolated familial medullary thyroid carcinoma	99361	<10	<10
Isolated female hypospadias	603515	<10	<10
Isolated femoral agenesis/hypoplasia	1987	202	198
Isolated fibular hemimelia	93323	123	123

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isolated filum lipoma	645325	**133**	**133**
Isolated focal cortical dysplasia	65683	**1870**	**1849**
Isolated focal non-epidermolytic palmoplantar keratoderma	448264	**<10**	**<10**
Isolated focal palmoplantar keratoderma*	307846	**46**	**45**
Isolated follicle stimulating hormone deficiency	52901	**<10**	**<10**
Isolated foveal hypoplasia	519398	**78**	**78**
Isolated gastric duplication	662376	**<10**	**<10**
Isolated generalized anhidrosis with normal sweat glands	468666	**<10**	**<10**
Isolated glycerol kinase deficiency	408	**<10**	**<10**
Isolated hemihyperplasia	2128	**283**	**280**
Isolated hereditary congenital facial paralysis	306527	**28**	**28**
Isolated humeral agenesis/hypoplasia	294973	**11**	**11**
Isolated humero-radial synostosis	3265	**23**	**23**
Isolated humero-radio-ulnar synostosis	3266	**10**	**10**
Isolated humero-ulnar synostosis	94056	**<10**	**<10**
Isolated hyperchlorhidrosis	542657	**<10**	**<10**
Isolated hyperphalangy	295002	**<10**	**<10**
Isolated hypoplasia of thumb	294988	**69**	**69**
Isolated idiopathic anterior uveitis	280914	**1550**	**1531**
Isolated Joubert syndrome	475	**485**	**473**
Isolated Klippel-Feil syndrome	2345	**161**	**159**
Isolated left bronchial isomerism	649029	**<10**	**<10**
Isolated lissencephaly type 1 without known genetic defects	1084	**24**	**23**
Isolated lower lip fistula	141064	**12**	**12**
Isolated megalencephaly	2477	**34**	**33**
Isolated megalopapilla	519402	**<10**	**<10**
Isolated mesenteric vein thrombosis	583861	**33**	**25**
Isolated micronodular adrenocortical disease	647782	**<10**	**<10**
Isolated microphthalmia-anophthalmia-coloboma*	2542	**189**	**187**
Isolated microspherophakia	519396	**10**	**10**
Isolated multiple intestinal atresia	2300	**17**	**17**
Isolated nail clubbing	217059	**<10**	**<10**
Isolated neonatal sclerosing cholangitis	480556	**31**	**31**
Isolated optic nerve aplasia	637064	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isolated optic nerve hypoplasia	637061	**20**	**20**
Isolated optic neuritis	499096	**324**	**321**
Isolated osteopoikilosis	166119	**10**	**10**
Isolated partial vaginal agenesis	96269	**17**	**17**
Isolated patella aplasia/hypoplasia	86789	**<10**	**<10**
Isolated permanent neonatal diabetes mellitus	99885	**77**	**74**
Isolated persistent urogenital sinus	647794	**10**	**10**
Isolated Pierre Robin sequence	718	**2265**	**2234**
Isolated polycystic liver disease	2924	**619**	**585**
Isolated posterior meningocele	268810	**46**	**46**
Isolated primary pigmented nodular adrenocortical disease	647772	**<10**	**<10**
Isolated proximal femoral focal deficiency	633228	**<10**	**<10**
Isolated pseudoarthrosis of the limbs	157808	**17**	**17**
Isolated pulmonary artery sling	658574	**<10**	**<10**
Isolated pulmonary capillaritis	264691	**<10**	**<10**
Isolated punctate palmoplantar keratoderma*	2338	**28**	**28**
Isolated radial hemimelia	93321	**98**	**97**
Isolated radio-ulnar synostosis	3269	**42**	**42**
Isolated rectal duplication	171220	**<10**	**<10**
Isolated right ventricular hypoplasia	439	**30**	**27**
Isolated segmental infantile hemangioma	675380	**55**	**55**
Isolated small intestine duplication	662456	**<10**	**<10**
Isolated splenic vein thrombosis	583856	**<10**	**<10**
Isolated split hand-split foot malformation	2440	**433**	**430**
Isolated succinate-CoQ reductase deficiency	3208	**16**	**12**
Isolated thyroid-stimulating hormone deficiency	90674	**149**	**149**
Isolated thyrotropin-releasing hormone deficiency	238670	**<10**	**<10**
Isolated tibial hemimelia	93322	**41**	**41**
Isolated tracheoesophageal fistula	454750	**78**	**74**
Isolated transitional filum lipoma	645322	**<10**	**<10**
Isolated ulnar hemimelia	93320	**43**	**43**
Isolated unilateral hemispheric cerebellar hypoplasia	269218	**17**	**17**
Isotretinoin syndrome	2305	**<10**	**<10**
Isotretinoin-like syndrome	2306	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isovaleric acidemia	33	**113**	**105**
ISPD-related limb-girdle muscular dystrophy R20	352479	**<10**	**<10**
ITM2B amyloidosis	439254	**12**	**<10**
ITPA-related lethal infantile neurological disorder with cataract and cardiac involvement	457375	**<10**	**<10**
Jackson-Weiss syndrome	1540	**<10**	**<10**
Jacobsen syndrome	2308	**101**	**95**
Jalili syndrome	1873	**<10**	**<10**
Jansen-de Vries syndrome	653767	**<10**	**<10**
Jervell and Lange-Nielsen syndrome	90647	**26**	**26**
Jessner lymphocytic infiltration of the skin	33314	**<10**	**<10**
Jeune syndrome	474	**111**	**100**
Johanson-Blizzard syndrome	2315	**12**	**10**
Johnson neuroectodermal syndrome	2316	**<10**	**<10**
Joubert syndrome with hepatic defect	1454	**<10**	**<10**
Joubert syndrome with Jeune asphyxiating thoracic dystrophy	397715	**<10**	**<10**
Joubert syndrome with ocular defect	220493	**23**	**23**
Joubert syndrome with oculorenal defect	2318	**18**	**18**
Joubert syndrome with renal defect	220497	**15**	**15**
Junctional epidermolysis bullosa inversa	79405	**<10**	**<10**
Junctional epidermolysis bullosa with pyloric atresia	79403	**<10**	**<10**
Junctional epidermolysis bullosa*	305	**108**	**78**
Juvenile absence epilepsy	1941	**823**	**820**
Juvenile amyotrophic lateral sclerosis	300605	**21**	**16**
Juvenile dermatomyositis	93672	**406**	**404**
Juvenile glaucoma	98977	**96**	**95**
Juvenile Huntington disease	248111	**33**	**23**
Juvenile idiopathic arthritis*	92	**986**	**982**
Juvenile myelomonocytic leukemia	86834	**18**	**14**
Juvenile myoclonic epilepsy	307	**1547**	**1536**
Juvenile nasopharyngeal angiofibroma	289596	**23**	**23**
Juvenile overlap myositis	329894	**13**	**13**
Juvenile Paget disease	2801	**21**	**20**
Juvenile polymyositis	93568	**14**	**14**
Juvenile polyposis syndrome	2929	**94**	**93**
Juvenile primary lateral sclerosis	247604	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Juvenile temporal arteritis	26137	**<10**	**<10**
Juvenile xanthogranuloma	158000	**71**	**71**
Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	445062	**<10**	**<10**
Kabuki syndrome	2322	**582**	**562**
Kagami-Ogata syndrome	254519	**<10**	**<10**
Kallmann syndrome-heart disease syndrome	2326	**14**	**13**
Kaposi sarcoma	33276	**31**	**26**
Kaposiform hemangioendothelioma	2122	**41**	**40**
Kaposiform lymphangiomatosis	464329	**<10**	**<10**
Karyomegalic interstitial nephritis	401996	**13**	**10**
Kasabach-Merritt phenomenon	2330	**36**	**35**
KAT6-related intellectual disability-craniofacial anomalies-cardiac defects syndrome	457193	**28**	**28**
Kawasaki disease	2331	**913**	**909**
KBG syndrome	2332	**548**	**548**
KCNQ2-related developmental and epileptic encephalopathy	439218	**149**	**146**
KDM3B-related intellectual disability-facial dysmorphism-short stature syndrome	633004	**<10**	**<10**
KDM5C-related syndromic X-linked intellectual disability	85279	**36**	**36**
Kearns-Sayre syndrome	480	**217**	**187**
Keipert syndrome	2662	**<10**	**<10**
Kennedy disease	481	**311**	**238**
Kenny-Caffey syndrome	2333	**14**	**13**
Keratinopathic ichthyosis*	281103	**49**	**49**
Keratocystic odontogenic tumor	447777	**11**	**11**
Keratoderma hereditarium mutilans	494	**<10**	**<10**
Keratoderma hereditarium mutilans with ichthyosis	79395	**<10**	**<10**
Keratosis follicularis spinulosa decalvans	2340	**11**	**11**
Keratosis follicularis-dwarfism-cerebral atrophy syndrome	2339	**<10**	**<10**
Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome	281201	**<10**	**<10**
Keratosis palmaris et plantaris-clinodactyly syndrome	86919	**<10**	**<10**
Keratosis pilaris atrophicans*	498	**15**	**15**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ketoacidosis due to monocarboxylate transporter-1 deficiency	438075	<10	<10
Keutel syndrome	85202	<10	<10
KID syndrome	477	28	24
Kidney tubulopathy-dilated cardiomyopathy syndrome	73224	<10	<10
Kienbock disease	97332	<10	<10
Kikuchi-Fujimoto disease	50918	91	91
Kimura disease	482	18	18
Kindler epidermolysis bullosa	2908	30	29
King-Denborough syndrome	99741	<10	<10
Kleefstra syndrome	261494	213	211
Kleine-Levin syndrome	33543	381	380
KLHL7-related Bohring-Opitz-like and Crisponi/Cold-induced sweating-like overlap syndrome	603684	<10	<10
Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome	447974	<10	<10
Klüver-Bucy syndrome	157823	<10	<10
Kniest dysplasia	485	37	37
Knobloch syndrome	1571	35	35
Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome	2698	<10	<10
Kommerell diverticulum	99077	17	17
Koolen-De Vries syndrome	96169	125	124
Kosaki overgrowth syndrome	477831	<10	<10
Kostmann syndrome	99749	18	18
Krabbe disease	487	101	59
KRT1-related diffuse nonepidermolytic keratoderma	530838	<10	<10
Kufor-Rakeb syndrome	306674	<10	<10
Kuskokwim syndrome	1149	<10	<10
Kyphoscoliotic Ehlers-Danlos syndrome	536545	64	62
L1 syndrome	275543	119	115
L-2-hydroxyglutaric aciduria	79314	27	26
Lacrimoauriculodentodigital syndrome	2363	46	46
Lafora disease	501	16	<10
Laing distal myopathy	59135	69	68
LAMA5-related multisystemic syndrome	521450	<10	<10
Lambert syndrome	1296	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Lambert-Eaton myasthenic syndrome	43393	208	158
Lamb-Shaffer syndrome	530983	68	68
Lamellar ichthyosis	313	355	349
Laminin subunit alpha 2-related congenital muscular dystrophy	258	223	198
Landau-Kleffner syndrome	98818	54	54
Langer mesomelic dysplasia	2632	12	12
Langerhans cell histiocytosis	389	2013	1936
Langerhans cell sarcoma	86897	<10	<10
Large/giant congenital melanocytic nevus	626	1470	1460
Laron syndrome	633	74	74
Laron syndrome with immunodeficiency	220465	<10	<10
Larsen syndrome	503	82	79
Larsen-like osseous dysplasia-short stature syndrome	2370	<10	<10
Larsen-like syndrome, B3GAT3 type	284139	<10	<10
Laryngeal abductor paralysis	2808	21	20
Laryngeal abductor paralysis-intellectual disability syndrome	2375	<10	<10
Laryngeal neuroendocrine tumor	100083	<10	<10
Laryngocele	2372	<10	<10
Laryngo-onycho-cutaneous syndrome	2407	<10	<10
Laryngotracheoesophageal cleft	2004	114	110
Larynx atresia	1202	49	47
Late-onset distal myopathy, Markesbery-Griggs type	98912	28	26
Late-onset focal dermal elastosis	228227	<10	<10
Late-onset isolated ACTH deficiency	199299	295	282
Late-onset junctional epidermolysis bullosa	79406	<10	<10
Late-onset retinal degeneration	67042	13	13
Lateral facial cleft*	141269	16	16
Lateral meningocele syndrome	2789	<10	<10
Lattice corneal dystrophy type I	98964	43	39
Laubry-Pezzi syndrome	99094	206	205
Laurence-Moon syndrome	2377	<10	<10
Laurin-Sandrow syndrome	2378	<10	<10
LCAT deficiency	650	<10	<10
Lead poisoning	330015	<10	<10
Leber congenital amaurosis	65	555	552
Leber hereditary optic neuropathy	104	1100	1050

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Leber plus disease	99718	**56**	**49**
Ledderhose disease	199251	**<10**	**<10**
Left sided atrial isomerism	566862	**<10**	**<10**
Left ventricular noncompaction	54260	**1156**	**1123**
Legg-Calvé-Perthes disease	2380	**32**	**32**
Legionnaires disease	549	**<10**	**<10**
Legius syndrome	137605	**146**	**145**
Leigh syndrome	506	**178**	**120**
Leiomyosarcoma	64720	**21**	**20**
Leiomyosarcoma of small intestine	104076	**<10**	**<10**
Leiomyosarcoma of the cervix uteri	213807	**<10**	**<10**
Leiomyosarcoma of the corpus uteri	213625	**<10**	**<10**
Leishmaniasis	507	**<10**	**<10**
Lemierre syndrome	137839	**<10**	**<10**
Lennox-Gastaut syndrome	2382	**1227**	**1185**
Lenz-Majewski hyperostotic dwarfism	2658	**<10**	**<10**
Leprosy	548	**42**	**42**
Leptospirosis	509	**<10**	**<10**
Léri-Weill dyschondrosteosis	240	**1015**	**1015**
Lesch-Nyhan syndrome	510	**68**	**52**
Lethal ataxia with deafness and optic atrophy	1187	**<10**	**<10**
Lethal congenital contracture syndrome type 1	1486	**<10**	**<10**
Lethal congenital contracture syndrome*	294965	**<10**	**<10**
Lethal hemolytic anemia-genital anomalies syndrome	1046	**<10**	**<10**
Lethal infantile mitochondrial myopathy	254857	**36**	**10**
Lethal intrauterine growth restriction-cortical malformation-congenital contractures syndrome	2570	**<10**	**<10**
Lethal Larsen-like syndrome	2371	**<10**	**<10**
Lethal neonatal spasticity-epileptic encephalopathy syndrome	435845	**<10**	**<10**
Lethal occipital encephalocele-skeletal dysplasia syndrome	293925	**<10**	**<10**
Lethal polymalformative syndrome, Boissel type	210144	**38**	**35**
Lethal pontocerebellar hypoplasia-hypotonia-respiratory insufficiency syndrome	615954	**<10**	**<10**
Lethal recessive chondrodysplasia	1423	**<10**	**<10**
Leukocyte adhesion deficiency	2968	**14**	**10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Leukoencephalopathy with bilateral anterior temporal lobe cysts	139444	**<10**	**<10**
Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	137898	**10**	**<10**
Leukoencephalopathy with calcifications and cysts	542310	**14**	**13**
Leukoencephalopathy with mild cerebellar ataxia and white matter edema	363540	**<10**	**<10**
Leukoencephalopathy-palmoplantar keratoderma syndrome	2386	**<10**	**<10**
Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome	314051	**<10**	**<10**
Leukonychia totalis	2387	**<10**	**<10**
Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome	210133	**<10**	**<10**
Levocardia	95854	**<10**	**<10**
Leydig cell hypoplasia	755	**28**	**28**
L-ferritin deficiency	440731	**<10**	**<10**
Lichen amyloidosis	49804	**<10**	**<10**
Lichen myxedematosus*	402007	**<10**	**<10**
Lichen planopilaris	525	**<10**	**<10**
Lichen planus pemphigoides	254478	**124**	**119**
Lichen planus pigmentosus	254463	**<10**	**<10**
Lichtenstein syndrome	2390	**<10**	**<10**
Liddle syndrome	526	**33**	**33**
Li-Fraumeni syndrome	524	**65**	**56**
LIG4 syndrome	99812	**10**	**<10**
Limbal stem cell deficiency	171673	**20**	**20**
Limb-girdle muscular dystrophy due to POMK deficiency	445110	**<10**	**<10**
Limb-girdle muscular dystrophy*	263	**755**	**677**
Limb-mammary syndrome	69085	**<10**	**<10**
Linear and whorled nevoid hypermelanosis	79150	**22**	**22**
Linear atrophoderma of Moulin	140933	**<10**	**<10**
Linear hypopigmentation and craniofacial asymmetry with acral, ocular and brain anomalies	589608	**13**	**13**
Linear IgA dermatosis	46488	**222**	**199**
Linear lichen planus	254379	**21**	**21**
Linear nevus sebaceus syndrome	2612	**218**	**217**
Linear verrucous nevus syndrome	2611	**234**	**234**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
LIPE-related familial partial lipodystrophy	435660	<10	<10
Lipoblastoma	247762	<10	<10
Lipodystrophy due to peptidic growth factors deficiency	1979	<10	<10
Lipodystrophy-intellectual disability-deafness syndrome	50811	<10	<10
Lipoic acid synthetase deficiency	401859	<10	<10
Lipoid proteinosis	530	16	15
Lipoprotein glomerulopathy	329481	<10	<10
Liposarcoma	69078	32	30
Lipoyl transferase 1 deficiency	401862	<10	<10
Lisch epithelial corneal dystrophy	98955	<10	<10
Lissencephaly due to LIS1 mutation	95232	49	47
Lissencephaly due to TUBA1A mutation	171680	16	16
Lissencephaly type 1 due to doublecortin gene mutation	2148	59	57
Lissencephaly type 3*	102011	<10	<10
Lissencephaly type 3-metacarpal bone dysplasia syndrome	86822	<10	<10
Lissencephaly with cerebellar hypoplasia type A	100011	<10	<10
Lissencephaly with cerebellar hypoplasia*	86823	<10	<10
Listeriosis	533	<10	<10
Livedoid vasculopathy	542643	28	27
Liver adenomatosis	566841	<10	<10
LMNA-related cardiocutaneous progeria syndrome	363618	<10	<10
Localized dystrophic epidermolysis bullosa	595356	21	21
Localized epidermolysis bullosa simplex	79400	49	49
Localized junctional epidermolysis bullosa	251393	<10	<10
Localized lichen myxedematosus*	86795	<10	<10
Localized lipodystrophy*	79088	11	<10
Localized scleroderma	90289	319	317
Locked-in syndrome	2406	<10	<10
Loeffler endocarditis	75566	<10	<10
Loeys-Dietz syndrome	60030	663	637
Logopenic progressive aphasia	250831	111	101
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	5	91	81

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Loose anagen syndrome	168	<10	<10
Low phospholipid-associated cholelithiasis	69663	1593	1571
Lowe-Kohn-Cohen syndrome	2408	<10	<10
Lower limb hypertrophy	295051	32	32
Lower limb malformation-hypospadias syndrome	2487	<10	<10
Lower motor neuron syndrome with late-adult onset	276435	44	41
Low-grade astrocytoma*	251592	<10	<10
Lowry-Wood syndrome	1824	12	11
LRP5-related primary osteoporosis	498481	27	27
Lujan-Fryns syndrome	776	73	70
LUMBAR syndrome	83628	58	58
Lung agenesis-heart defect-thumb anomalies syndrome	1120	<10	<10
Lupus erythematosus panniculitis	90285	38	35
Lupus erythematosus tumidus	90283	172	170
Luscan-Lumish syndrome	597738	11	11
Lyme disease	91546	194	182
Lymphangioleiomyomatosis	538	612	594
Lymphatic filariasis	2035	<10	<10
Lymphedema with yellow nails	662	69	67
Lymphedema-cerebral arteriovenous anomaly-primary pulmonary hypertension syndrome	86914	<10	<10
Lymphedema-distichiasis syndrome	33001	71	70
Lymphoid interstitial pneumonia	79128	229	199
Lymphomatoid granulomatosis	86869	14	13
Lymphomatoid papulosis	98842	10	10
Lynch syndrome	144	281	244
Lysinuric protein intolerance	470	53	49
Lysosomal acid lipase deficiency	275761	42	39
Macrocephaly-developmental delay syndrome	397612	42	42
Macrocephaly-intellectual disability-autism syndrome	210548	38	38
Macrocephaly-intellectual disability-left ventricular non compaction syndrome	466791	<10	<10
Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	457485	29	29
Macrocephaly-short stature-paraplegia syndrome	2427	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Macrocephaly-spastic paraplegia-dysmorphism syndrome	2429	**16**	**16**
Macrocystic lymphatic malformation	79489	**663**	**661**
Macrodactyly of fingers	295044	**19**	**19**
Macrodactyly of toes	295047	**25**	**25**
Macrophage activation syndrome	158061	**130**	**99**
Macrophagic myofasciitis	592	**556**	**534**
Macrosomia-microphthalmia-cleft palate syndrome	2432	**<10**	**<10**
Macrostomia-preauricular tags-external ophthalmoplegia syndrome	83619	**<10**	**<10**
Macrothrombocytopenia with mitral valve insufficiency	220448	**<10**	**<10**
Macular coloboma-cleft palate-hallux valgus syndrome	91494	**<10**	**<10**
Macular corneal dystrophy	98969	**31**	**31**
Maculopapular cutaneous mastocytosis	79457	**706**	**705**
Madras motor neuron disease	137867	**<10**	**<10**
Maffucci syndrome	163634	**28**	**27**
MAGIC syndrome	324972	**26**	**25**
Majeed syndrome	77297	**<10**	**<10**
Mal de débarquement	210272	**13**	**13**
Mal de Meleda	87503	**25**	**25**
Malakoplakia	556	**<10**	**<10**
Malan overgrowth syndrome	420179	**51**	**51**
Malaria	673	**19**	**14**
Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome	2234	**<10**	**<10**
Male infertility with azoospermia or oligozoospermia due to single gene mutation	399805	**80**	**80**
Male infertility with teratozoospermia due to single gene mutation	399808	**20**	**20**
Malignant epithelial tumor of salivary glands	276145	**<10**	**<10**
Malignant germ cell tumor of the cervix uteri	213837	**<10**	**<10**
Malignant germ cell tumor of the vagina	206489	**<10**	**<10**
Malignant granulosa cell tumor of the ovary	99915	**24**	**24**
Malignant hyperthermia of anesthesia	423	**336**	**333**
Malignant melanoma of the mucosa	168999	**18**	**13**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Malignant non-dysgerminomatous germ cell tumor of ovary	206538	**<10**	**<10**
Malignant peripheral nerve sheath tumor	3148	**16**	**15**
Malignant peritoneal mesothelioma	168811	**<10**	**<10**
Malignant Sertoli-Leydig cell tumor of the ovary	99916	**21**	**20**
Malignant teratoma of ovary	398987	**35**	**35**
Malonic aciduria	943	**11**	**11**
Malposition of a coronary ostium	99090	**<10**	**<10**
MALT lymphoma	52417	**40**	**38**
MAN1B1-CDG	397941	**11**	**11**
Mandibular arteriovenous malformation	141174	**50**	**50**
Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome	363649	**<10**	**<10**
Mandibuloacral dysplasia	2457	**<10**	**<10**
Mandibulofacial dysostosis*	155899	**76**	**73**
Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome	357158	**<10**	**<10**
Mandibulofacial dysostosis-microcephaly syndrome	79113	**68**	**66**
Mantle cell lymphoma	52416	**19**	**14**
Maple syrup urine disease	511	**216**	**207**
Marburg acute multiple sclerosis	228157	**30**	**29**
Marcus-Gunn syndrome	91412	**46**	**45**
Marden-Walker syndrome	2461	**<10**	**<10**
Marfan syndrome	558	**8450**	**8254**
Marfanoid habitus-autosomal recessive intellectual disability syndrome	2463	**30**	**30**
Marfanoid habitus-facial dysmorphism-skeletal abnormality-heart defect syndrome	643503	**<10**	**<10**
Marfanoid syndrome, De Silva type	2464	**<10**	**<10**
Marginal zone lymphoma*	300912	**18**	**13**
Marie Unna hereditary hypotrichosis	444	**<10**	**<10**
Marinesco-Sjögren syndrome	559	**21**	**19**
Marshall syndrome	560	**68**	**68**
Marshall-Smith syndrome	561	**17**	**17**
Martinique crinkled retinal pigment epitheliopathy	466718	**<10**	**<10**
Mast cell leukemia	98851	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mast cell sarcoma	66661	<10	<10
Maternal phenylketonuria syndrome	2209	42	42
Maternal uniparental disomy of chromosome 1 syndrome	251009	<10	<10
Maternal uniparental disomy of chromosome 16 syndrome	96185	<10	<10
Maternal uniparental disomy of chromosome 20 syndrome	96186	<10	<10
Maternal uniparental disomy of chromosome 21 syndrome	96187	<10	<10
Maternal uniparental disomy of chromosome 6 syndrome	96181	<10	<10
Maternal uniparental disomy of chromosome X syndrome	261519	<10	<10
Matthew-Wood syndrome	2470	<10	<10
Maxillary arteriovenous malformation	141171	57	56
Maxillonasal dysplasia	1248	208	207
Mayer-Rokitansky-Küster-Hauser syndrome	3109	919	918
May-Thurner syndrome	675404	<10	<10
Mazabraud syndrome	57782	27	27
McCune-Albright syndrome	562	540	527
McDonough syndrome	2471	<10	<10
McKusick-Kaufman syndrome	2473	<10	<10
McLeod neuroacanthocytosis syndrome	59306	18	17
Meacham syndrome	3097	<10	<10
Meckel syndrome	564	12	<10
Meconium aspiration syndrome	70588	33	28
MECP2-related severe neonatal encephalopathy	209370	75	65
Medial condensing osteitis of the clavicle	57196	<10	<10
Median cleft lip/mandible	2006	<10	<10
Median cleft of the upper lip and maxilla	141239	49	48
Median facial cleft*	141234	12	12
Median nodule of the upper lip	2699	<10	<10
Medium chain acyl-CoA dehydrogenase deficiency	42	335	325
Medullary sponge kidney	1309	269	259
Medullary thyroid carcinoma	1332	120	114
Medulloblastoma	616	275	254
Meesmann corneal dystrophy	98954	13	13
Mega-cisterna magna	97252	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Megaconial congenital muscular dystrophy	280671	12	11
Megacystis-megaureter syndrome	238637	62	62
Megacystis-microcolon-intestinal hypoperistalsis syndrome	2241	13	12
Megalencephalic leukoencephalopathy with subcortical cysts	2478	44	43
Megalencephaly-capillary malformation-polymicrogyria syndrome	60040	159	157
Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	83473	26	25
Megalencephaly-severe kyphoscoliosis-overgrowth syndrome	457359	<10	<10
Megalocornea-intellectual disability syndrome	2479	<10	<10
MEGDEL syndrome	352328	<10	<10
MEHMO syndrome	85282	<10	<10
Meige disease	90186	2557	2507
Meigs syndrome	314451	<10	<10
Melanoma and neural system tumor syndrome	252206	<10	<10
Melanoma of soft tissue	97338	15	12
MELAS	550	1013	852
Melkersson-Rosenthal syndrome	2483	28	27
Melnick-Needles syndrome	2484	20	19
Melorheostosis	2485	70	70
Melorheostosis with osteopoikilosis	1879	<10	<10
MEND syndrome	401973	<10	<10
Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency	99898	<10	<10
Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency	319558	<10	<10
Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	319552	<10	<10
Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency	319600	<10	<10
Mendelian susceptibility to mycobacterial diseases due to partial JAK1 deficiency	574957	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency	319595	<10	<10
Mendelian susceptibility to mycobacterial diseases*	748	16	15
Ménétrier disease	2494	<10	<10
Meningeal melanocytoma	252046	<10	<10
Meningioma	2495	460	443
Meningococcal meningitis	33475	58	57
Menke-Hennekam syndrome	592574	21	21
Menkes disease	565	68	31
Menstrual cycle-dependent periodic fever	498251	<10	<10
MEPAN syndrome	508093	<10	<10
MERRF	551	109	92
Mesial temporal lobe epilepsy with hippocampal sclerosis	99701	988	973
Mesoaxial synostotic syndactyly with phalangeal reduction	157801	<10	<10
Mesocardia	95443	<10	<10
Mesomelia-synostoses syndrome	2496	<10	<10
Mesomelic and rhizo-mesomelic dysplasia*	93438	11	11
Mesomelic dysplasia, Savarirayan type	85170	<10	<10
Mesomelic dysplasia-digital anomalies-intellectual disability syndrome	632603	<10	<10
Metabolic myopathy due to lactate transporter defect	171690	18	18
Metachondromatosis	2499	51	50
Metachromatic leukodystrophy	512	197	116
Metaphyseal anadysplasia	1040	10	10
Metaphyseal chondrodysplasia, Jansen type	33067	<10	<10
Metaphyseal chondrodysplasia, Rosenberg type	1837	<10	<10
Metaphyseal chondrodysplasia, Schmid type	174	72	71
Metaphyseal chondrodysplasia, Spahr type	2501	<10	<10
Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria	99646	<10	<10
Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome	2504	<10	<10
Metatropic dysplasia	2635	24	23
Methanol poisoning	31825	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Methimazole embryofetopathy	1923	<10	<10
Methionine adenosyltransferase I/III deficiency	168598	<10	<10
Methotrexate toxicity	565782	<10	<10
Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	308425	<10	<10
Methylmalonic acidemia with homocystinuria	26	193	175
Methylmalonic acidemia without homocystinuria*	293355	63	51
Methylmalonic aciduria due to transcobalamin receptor defect	280183	<10	<10
Mevalonate kinase deficiency	309025	201	196
MGAT2-CDG	79329	<10	<10
Micro syndrome	2510	32	25
Microbrachycephaly-ptosis-cleft lip syndrome	2511	30	28
Microcephalic cortical malformations-short stature due to RTTN deficiency	468631	<10	<10
Microcephalic osteodysplastic dysplasia, Saul-Wilson type	85172	<10	<10
Microcephalic osteodysplastic primordial dwarfism type II	2637	21	20
Microcephalic osteodysplastic primordial dwarfism types I and III	2636	<10	<10
Microcephalic primordial dwarfism due to ZNF335 deficiency	329228	<10	<10
Microcephalic primordial dwarfism*	324761	21	20
Microcephalic primordial dwarfism, Toriello type	2643	<10	<10
Microcephalic primordial dwarfism-insulin resistance syndrome	436182	<10	<10
Microcephaly-albinism-digital anomalies syndrome	2513	<10	<10
Microcephaly-brachydactyly-kyphoscoliosis syndrome	3433	<10	<10
Microcephaly-brain defect-spasticity-hypernatremia syndrome	2523	<10	<10
Microcephaly-capillary malformation syndrome	294016	<10	<10
Microcephaly-cardiac defect-lung malsegmentation syndrome	2516	<10	<10
Microcephaly-cardiomyopathy syndrome	2515	<10	<10
Microcephaly-cervical spine fusion anomalies syndrome	2522	<10	<10
Microcephaly-cleft palate-abnormal retinal pigmentation syndrome	2521	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Microcephaly-complex motor and sensory axonal neuropathy syndrome	423894	<10	<10
Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrom	500159	<10	<10
Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome	457284	<10	<10
Microcephaly-deafness-intellectual disability syndrome	2533	14	14
Microcephaly-facial dysmorphism-ocular anomalies-multiple congenital anomalies syndrome	521445	<10	<10
Microcephaly-hearing loss-facial dysmorphism-intellectual disability syndrome	662179	<10	<10
Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome	457351	<10	<10
Microcephaly-lymphedema-chorioretinopathy syndrome	2526	33	32
Microcephaly-microcornea syndrome, Seemanova type	2528	<10	<10
Microcephaly-polymicrogyria-corpus callosum agenesis syndrome	171703	<10	<10
Microcephaly-seizures-intellectual disability-heart disease syndrome	2519	<10	<10
Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome	423306	<10	<10
Microcephaly-thin corpus callosum-intellectual disability syndrome	397951	<10	<10
Microcornea-posterior megalolenticonus-persistent fetal vasculature-coloboma syndrome	231736	<10	<10
Microcystic lymphatic malformation	79490	680	678
Microcytic anemia with liver iron overload	83642	25	25
Microduplication Xp11.22p11.23 syndrome	217377	22	21
Microform holoprosencephaly	280200	23	23
Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	476126	17	17
Microlissencephaly	1083	21	17
Microlissencephaly-micromelia syndrome	50810	<10	<10
Microphthalmia with brain and digit anomalies	139471	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Microphthalmia with limb anomalies	1106	<10	<10
Microphthalmia with linear skin defects syndrome	2556	15	14
Microphthalmia, Lenz type	568	15	15
Microphthalmia-brain atrophy syndrome	77299	<10	<10
Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	251279	10	10
Microscopic polyangiitis	727	880	773
Microspherophakia-metaphyseal dysplasia syndrome	2551	<10	<10
Microtia	83463	1013	1012
Microvenular haemangioma	675369	<10	<10
Microvillus inclusion disease	2290	43	36
Mid-dermal elastolysis	228299	<10	<10
Middle aortic syndrome	1456	151	146
Middle ear neuroendocrine tumor	100084	<10	<10
Middle East respiratory syndrome	576074	<10	<10
Midline cervical cleft	141288	27	27
Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis	93279	35	35
Miller Fisher syndrome	98919	231	213
Miller-Dieker syndrome	531	103	86
Mills syndrome	94091	19	16
Milroy disease	79452	487	481
MIRAGE syndrome	494433	16	10
Mirror polydactyly-vertebral segmentation-limbs defects syndrome	3004	<10	<10
Mirror-image polydactyly	498494	<10	<10
MiT family translocation renal cell carcinoma	319308	<10	<10
Mitchell Syndrome	631248	<10	<10
MITF-related melanoma and renal cell carcinoma predisposition syndrome	293822	31	31
Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	1933	<10	<10
Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy	255235	<10	<10
Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	369897	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mitochondrial DNA depletion syndrome, encephalomyopathic form*	254803	**<10**	**<10**
Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency	279934	**<10**	**<10**
Mitochondrial DNA depletion syndrome, hepatocerebral form*	254871	**<10**	**<10**
Mitochondrial DNA depletion syndrome, hepatocerebrorenal form	363534	**<10**	**<10**
Mitochondrial DNA depletion syndrome, myopathic form	254875	**23**	**20**
Mitochondrial DNA-associated Leigh syndrome	255210	**27**	**24**
Mitochondrial DNA-related cardiomyopathy and hearing loss	1349	**22**	**20**
Mitochondrial DNA-related dystonia	254851	**11**	**<10**
Mitochondrial DNA-related mitochondrial myopathy*	254788	**87**	**81**
Mitochondrial DNA-related progressive external ophthalmoplegia	663	**115**	**112**
Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	314637	**<10**	**<10**
Mitochondrial membrane protein-associated neurodegeneration	289560	**11**	**10**
Mitochondrial myopathy and sideroblastic anemia	2598	**<10**	**<10**
Mitochondrial myopathy with reversible cytochrome C oxidase deficiency	254864	**<10**	**<10**
Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	502423	**10**	**<10**
Mitochondrial myopathy-lactic acidosis-deafness syndrome	2597	**38**	**32**
Mitochondrial neurogastrointestinal encephalomyopathy	298	**47**	**33**
Mitochondrial pyruvate carrier deficiency	447784	**<10**	**<10**
Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	653880	**<10**	**<10**
Mitochondrial trifunctional protein deficiency	746	**23**	**18**
Mitral atresia	1205	**49**	**39**
Mitral valve agenesis	99062	**<10**	**<10**
Mixed connective tissue disease	809	**2476**	**2380**
Mixed cystic lymphatic malformation	458792	**513**	**512**
Mixed germ cell tumor	180234	**15**	**15**
Mixed neuronal-glial tumor	251934	**<10**	**<10**
Mixed phenotype acute leukemia	530995	**<10**	**<10**
Mixed-type autoimmune hemolytic anemia	90036	**67**	**60**
Miyoshi myopathy	45448	**50**	**50**
MMEP syndrome	3434	**<10**	**<10**
MME-related autosomal dominant Charcot Marie Tooth disease type 2	497757	**10**	**10**
Moderate and severe traumatic brain injury	90056	**33**	**32**
MODY	552	**1092**	**1080**
Moebius syndrome	570	**278**	**266**
Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome	2560	**<10**	**<10**
Mohr-Tranebjaerg syndrome	52368	**10**	**10**
MOMO syndrome	2563	**<10**	**<10**
Monilethrix	573	**30**	**30**
Monoamine oxidase A deficiency	3057	**<10**	**<10**
Monoclonal mast cell activation syndrome	529468	**2112**	**2104**
Monomelic amyotrophy	65684	**216**	**203**
Monosomy 13q14 syndrome	1587	**13**	**13**
Monosomy 13q34 syndrome	96168	**26**	**26**
Monosomy 18p syndrome	1598	**79**	**78**
Monosomy 18q syndrome	1600	**189**	**182**
Monosomy 22 syndrome	96123	**21**	**21**
Monosomy 5p syndrome	281	**223**	**216**
Monosomy 9p syndrome	261112	**37**	**37**
Monosomy 9q22.3 syndrome	77301	**10**	**10**
Mooren ulcer	519408	**25**	**21**
Morgagni-Stewart-Morel syndrome	77296	**<10**	**<10**
Morning glory disc anomaly	35737	**88**	**87**
Morvan syndrome	83467	**14**	**12**
Mosaic genome-wide paternal uniparental disomy syndrome	329813	**<10**	**<10**
Mosaic Legius syndrome	634511	**<10**	**<10**
Mosaic neurofibromatosis type 1	634461	**169**	**169**
Mosaic NF2-related schwannomatosis	634475	**15**	**15**
Mosaic schwannomatosis	634492	**18**	**18**
Mosaic trisomy 1 syndrome	1692	**<10**	**<10**
Mosaic trisomy 12 syndrome	1698	**18**	**16**
Mosaic trisomy 14 syndrome	1703	**22**	**22**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mosaic trisomy 15 syndrome	1706	**13**	**11**
Mosaic trisomy 16 syndrome	1708	**25**	**24**
Mosaic trisomy 17 syndrome	1711	**<10**	**<10**
Mosaic trisomy 2 syndrome	1723	**<10**	**<10**
Mosaic trisomy 20 syndrome	1724	**18**	**17**
Mosaic trisomy 22 syndrome	96068	**25**	**23**
Mosaic trisomy 3 syndrome	100071	**<10**	**<10**
Mosaic trisomy 5 syndrome	96060	**<10**	**<10**
Mosaic trisomy 7 syndrome	1747	**<10**	**<10**
Mosaic trisomy 8 syndrome	96061	**46**	**42**
Mosaic trisomy 9 syndrome	99776	**43**	**38**
Mosaic variegated aneuploidy syndrome	1052	**35**	**34**
Motor delay-microcephaly-speech impairment-ocular abnormalities syndrome	662762	**<10**	**<10**
Mounier-Kühn syndrome	3347	**26**	**25**
Mowat-Wilson syndrome	2152	**159**	**149**
Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	280679	**12**	**12**
Moyamoya disease	2573	**528**	**509**
Moyamoya disease with early-onset achalasia	401945	**<10**	**<10**
Moynahan syndrome	2574	**<10**	**<10**
MPI-CDG	79319	**15**	**12**
MSH3-related polyposis	480536	**<10**	**<10**
MT-ATP6-related mitochondrial spastic paraplegia	320360	**12**	**12**
Mucinous adenocarcinoma of ovary	398961	**<10**	**<10**
Mucinous cystadenocarcinoma of the pancreas	424053	**36**	**33**
Muckle-Wells syndrome	575	**115**	**111**
Mucocutaneous venous malformations	2451	**3287**	**3282**
Mucolipidosis type II	576	**43**	**21**
Mucolipidosis type III	577	**41**	**38**
Mucolipidosis type IV	578	**11**	**11**
Mucopolysaccharidosis type 1	579	**298**	**255**
Mucopolysaccharidosis type 2	580	**211**	**150**
Mucopolysaccharidosis type 3	581	**206**	**151**
Mucopolysaccharidosis type 4	582	**195**	**183**
Mucopolysaccharidosis type 6	583	**64**	**57**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mucopolysaccharidosis type 7	584	**15**	**14**
Mucous membrane pemphigoid	46486	**1748**	**1523**
Mueller-Weiss syndrome	566943	**<10**	**<10**
Muenke syndrome	53271	**145**	**143**
Mulibrey nanism	2576	**14**	**12**
Müllerian aplasia and hyperandrogenism	247768	**<10**	**<10**
Müllerian derivatives-lymphangiectasia-polydactyly syndrome	1655	**<10**	**<10**
Müllerian duct anomalies-limb anomalies syndrome	2491	**<10**	**<10**
Müllérienne aplasia*	73217	**<10**	**<10**
Multicentric carpo-tarsal osteolysis with or without nephropathy	2774	**11**	**<10**
Multicentric osteolysis-nodulosis-arthropathy spectrum	371428	**<10**	**<10**
Multicentric reticulohistiocytosis	139436	**14**	**14**
Multicystic dysplastic kidney	1851	**3291**	**3269**
Multifocal atrial tachycardia	3282	**20**	**19**
Multifocal infantile hemangioma with extracutenous involvement	2123	**106**	**105**
Multifocal lymphangioendotheliomatosis-thrombocytopenia syndrome	464321	**<10**	**<10**
Multifocal motor neuropathy	641	**935**	**872**
Multifocal pattern dystrophy simulating fundus flavimaculatus	99003	**27**	**26**
Multifocal tuberculosis	645854	**<10**	**<10**
Multiloculated renal cyst	97366	**45**	**45**
Multiminicore myopathy	598	**153**	**145**
Multinodular goiter-cystic kidney-polydactyly syndrome	2091	**14**	**14**
Multiple acyl-CoA dehydrogenase deficiency	26791	**134**	**122**
Multiple benign circumferential skin creases on limbs	2505	**<10**	**<10**
Multiple carboxylase deficiency*	148	**<10**	**<10**
Multiple congenital anomalies-hypotonia-seizures syndrome	280633	**36**	**27**
Multiple congenital anomalies-hypotonia-seizures syndrome type 2	300496	**<10**	**<10**
Multiple congenital anomalies-neurodevelopmental delay-ocular abnormalities syndrome	659904	**<10**	**<10**
Multiple endocrine neoplasia type 1	652	**574**	**554**
Multiple endocrine neoplasia type 2	653	**331**	**326**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Multiple endocrine neoplasia type 4	276152	<10	<10
Multiple endocrine neoplasia*	276161	39	33
Multiple epiphyseal dysplasia due to collagen 9 anomaly	166002	<10	<10
Multiple epiphyseal dysplasia type 1	93308	11	11
Multiple epiphyseal dysplasia type 4	93307	<10	<10
Multiple epiphyseal dysplasia type 5	93311	<10	<10
Multiple epiphyseal dysplasia*	251	467	465
Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome	166024	<10	<10
Multiple epiphyseal dysplasia-severe proximal femoral dysplasia syndrome	166029	<10	<10
Multiple evanescent white dot syndrome	674953	<10	<10
Multiple metaphyseal dysplasia*	93430	131	130
Multiple mitochondrial dysfunctions syndrome type 1	401869	<10	<10
Multiple myeloma	29073	516	322
Multiple osteochondromas	321	1297	1292
Multiple paragangliomas associated with polycythemia	324299	<10	<10
Multiple pterygium syndrome*	294060	<10	<10
Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome	3151	<10	<10
Multiple self-healing squamous epithelioma	65748	<10	<10
Multiple sulfatase deficiency	585	21	13
Multiple symmetric lipomatosis	2398	114	109
Multiple synostoses syndrome	3237	70	69
Multiple system atrophy	102	1526	935
Multisystem inflammatory syndrome in children and adults	598363	311	311
Multisystemic smooth muscle dysfunction syndrome	404463	<10	<10
Muscle filaminopathy	171445	80	68
Muscle-eye-brain disease	588	17	13
Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome	2579	<10	<10
Muscular dystrophy, Selcen type	199340	<10	<10
Muscular glycogenosis*	206959	101	94
Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome	324416	<10	<10
Musculocontractural Ehlers-Danlos syndrome	2953	18	18

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mutilating hereditary sensory neuropathy with spastic paraplegia	139578	34	32
Mutilating palmoplantar keratoderma with periorificial keratotic plaques	659	12	12
MUTYH-related polyposis	247798	48	45
Myalgia-eosinophilia syndrome associated with tryptophan	2582	<10	<10
Myasthenia gravis	589	8588	7669
MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome	498693	<10	<10
Mycophenolate mofetil embryopathy	268249	<10	<10
Mycoplasma encephalitis	83482	<10	<10
Mycosis fungoides and variants*	178566	<10	<10
Myelic limited dorsal malformation	645378	11	11
Myelocystocele*	268813	16	16
Myelodysplastic neoplasm with increased blasts	86839	<10	<10
Myelodysplastic neoplasm with low blasts	98826	<10	<10
Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality	86841	<10	<10
Myelodysplastic syndrome*	52688	87	70
Myelodysplastic/myeloproliferative disease*	98275	<10	<10
Myeloid/lymphoid neoplasm associated with FGFR1 rearrangement	168953	<10	<10
Myeloid/lymphoid neoplasm associated with JAK2 rearrangement	589542	<10	<10
Myeloperoxidase deficiency	2587	<10	<10
Myeloproliferative neoplasm*	98274	16	16
Myeloschisis	645398	<10	<10
MYH9-related syndromic thrombocytopenia	182050	276	269
Myhre syndrome	2588	65	58
Myoclonic epilepsy in non-progressive encephalopathies	86913	35	35
Myoclonic epilepsy of infancy	86909	231	228
Myoclonus-cerebellar ataxia-deafness syndrome	2589	<10	<10
Myoclonus-dystonia syndrome	36899	294	290
Myopathic Ehlers-Danlos syndrome	536516	<10	<10
Myopathy and diabetes mellitus	2596	18	18
Myosclerosis	289380	<10	<10
Myosin storage myopathy	53698	10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Myotonia fluctuans	99734	36	35
Myotonia permanens	99735	17	17
Myotonic dystrophy*	206647	114	96
MYT1L-related developmental delay-intellectual disability-obesity syndrome	647799	22	22
Myxopapillary ependymoma	251643	<10	<10
NAD(P)HX epimerase deficiency	555407	<10	<10
Naegeli-Franceschetti-Jadassohn syndrome	69087	<10	<10
Nager syndrome	245	49	45
Nail-patella syndrome	2614	462	455
Nail-patella-like renal disease	2613	<10	<10
Nance-Horan syndrome	627	40	40
Nanophthalmos	35612	40	39
Narcolepsy type 1	2073	3061	3039
Narcolepsy type 2	83465	1139	1130
NARP syndrome	644	81	73
Nasal dermoid cyst	141103	56	56
Nasal dorsum fistula	141219	189	189
Nasal ganglioglioma	141115	<10	<10
Nasal glial heterotopia	141112	<10	<10
Nasolacrimal duct cyst	141083	40	40
Nasopalpebral lipoma-coloboma syndrome	2399	<10	<10
Nasopharyngeal carcinoma	150	10	10
Nasu-Hakola disease	2770	<10	<10
Native American myopathy	168572	44	39
Navajo neurohepatopathy	255229	<10	<10
Naxos disease	34217	<10	<10
Necrobiosis lipoidica	542592	<10	<10
Necrobiotic xanthogranuloma	158011	17	17
Necrotizing enterocolitis	391673	104	102
Necrotizing soft tissue infection	440368	<10	<10
Nelson syndrome	199244	17	16
Nemaline myopathy*	607	159	144
Neonatal acute respiratory distress syndrome	217563	<10	<10
Neonatal adrenoleukodystrophy	44	14	10
Neonatal alloimmune neutropenia	464370	67	67
Neonatal antiphospholipid syndrome	398097	95	93
Neonatal autoimmune hemolytic anemia	398109	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Neonatal brainstem dysfunction	137929	257	248
Neonatal compartment syndrome	641829	<10	<10
Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome	79118	<10	<10
Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome	457185	<10	<10
Neonatal epileptic encephalopathy due to glutaminase deficiency	557064	<10	<10
Neonatal hemochromatosis	446	26	18
Neonatal hypoxic and ischemic brain injury	137577	440	412
Neonatal ichthyosis-sclerosing cholangitis syndrome	59303	11	10
Neonatal inflammatory skin and bowel disease	294023	<10	<10
Neonatal intrahepatic cholestasis due to citrin deficiency	247598	<10	<10
Neonatal lupus erythematosus	398124	30	30
Neonatal Marfan syndrome	284979	27	21
Neonatal osteosclerotic dysplasia*	93443	<10	<10
Neonatal renal venous thrombosis	664912	11	11
Neonatal scleroderma	398127	<10	<10
Neonatal severe primary hyperparathyroidism	417	19	16
Neovascular glaucoma	94058	<10	<10
Nephroblastoma	654	405	401
Nephrogenic syndrome of inappropriate antidiuresis	93606	27	27
Nephrogenic systemic fibrosis	137617	<10	<10
Nephronophthisis	655	488	477
Nephropathy-deafness-hyperparathyroidism syndrome	2668	<10	<10
Nephrosis-deafness-urinary tract-digital malformations syndrome	2669	<10	<10
NESCAV syndrome	662367	<10	<10
Netherton syndrome	634	126	115
Neuhauser anomaly	99078	38	38
Neuhauser-Eichner-Opitz syndrome	2672	14	13
Neu-Laxova syndrome	2671	<10	<10
Neuralgic amyotrophy	2901	850	823
Neurenteric cyst	268865	<10	<10
Neuroacanthocytosis*	263440	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Neuroblastoma	635	**199**	**183**
Neurocutaneous melanocytosis	2481	**21**	**19**
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	88639	**<10**	**<10**
Neurodegeneration with brain iron accumulation*	385	**63**	**54**
Neurodegenerative syndrome due to cerebral folate transport deficiency	217382	**20**	**19**
Neurodevelopmental delay-brain malformations-skeletal defects-intellectual disability syndrome	662207	**<10**	**<10**
Neurodevelopmental delay-congenital heart defects-intellectual disability syndrome	662234	**<10**	**<10**
Neurodevelopmental delay-intellectual disability-ataxia-feeding difficulty syndrome	647788	**<10**	**<10**
Neurodevelopmental delay-intellectual disability-skeletal defects syndrome	662198	**<10**	**<10**
Neurodevelopmental delay-seizures-ophthalmic anomalies-osteopenia-cerebellar atrophy syndrome	529665	**<10**	**<10**
Neurodevelopmental disorder-brain malformation-facial dysmorphism-brachydactyly syndrome	662189	**<10**	**<10**
Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome	453499	**35**	**34**
Neuroendocrine carcinoma of pancreas	506098	**<10**	**<10**
Neuroendocrine cell hyperplasia of infancy	217560	**100**	**99**
Neuroendocrine neoplasm of appendix	100079	**<10**	**<10**
Neuroendocrine tumor of stomach	100075	**<10**	**<10**
Neuroendocrine tumor of the colon	100080	**<10**	**<10**
Neurofaciodigitorenal syndrome	2673	**<10**	**<10**
Neuroferritinopathy	157846	**16**	**13**
Neurofibroma	252183	**156**	**155**
Neurofibromatosis type 1	636	**13806**	**13470**
Neurofibromatosis-Noonan syndrome	638	**109**	**104**
Neurogenic arthrogryposis multiplex congenita	1143	**70**	**65**
Neuroleptic malignant syndrome	94093	**<10**	**<10**
Neurolymphomatosis	206586	**42**	**28**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Neuromyelitis optica spectrum disorder	71211	**1516**	**1482**
Neuronal ceroid lipofuscinosis*	216	**37**	**21**
Neuronal tumor*	251924	**<10**	**<10**
Neuropathy with hearing impairment	139512	**11**	**11**
Neurotrophic keratopathy	137596	**61**	**57**
Neutral lipid storage disease with ichthyosis	98907	**28**	**28**
Neutral lipid storage disease with myopathy	98908	**15**	**15**
Neutral lipid storage disease*	165	**19**	**19**
NEVADA syndrome	370059	**<10**	**<10**
Nevus comedonicus syndrome	64754	**12**	**12**
Nevus of Ito	263432	**12**	**12**
Nevus of Ota	263425	**43**	**43**
New-onset refractory status epilepticus	363558	**46**	**36**
Nicolaides-Baraitser syndrome	3051	**68**	**66**
Nicolau syndrome	664787	**<10**	**<10**
Niemann-Pick disease type C	646	**187**	**134**
Night blindness-skeletal anomalies-dysmorphism syndrome	1390	**<10**	**<10**
Nijmegen breakage syndrome	647	**<10**	**<10**
NK-cell enteropathy	263665	**<10**	**<10**
NKX6-2-related autosomal recessive hypomyelinating leukodystrophy	527497	**<10**	**<10**
NLRC4-related familial cold autoinflammatory syndrome	576349	**<10**	**<10**
NLRP12-associated hereditary periodic fever syndrome	247868	**15**	**15**
NLRP3-associated autoinflammatory disease*	208650	**124**	**123**
NMDA receptor encephalitis	217253	**291**	**285**
Nocardiosis	31204	**11**	**10**
NOCARH syndrome	619363	**<10**	**<10**
Nodal marginal zone B-cell lymphoma	86867	**<10**	**<10**
Nodular cutaneous amyloidosis	137810	**<10**	**<10**
Nodular fasciitis	477742	**<10**	**<10**
Nodular lymphocyte predominant Hodgkin lymphoma	86893	**<10**	**<10**
Nodular neuronal heterotopia	2149	**550**	**547**
Nodular non-suppurative panniculitis	33577	**17**	**17**
Noma	2700	**<10**	**<10**
Non-24-hour sleep-wake syndrome	73267	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome	231720	**10**	**10**
Non-acquired isolated growth hormone deficiency	631	**6328**	**6294**
Non-acquired panhypopituitarism	90695	**1128**	**1063**
Non-amyloid fibrillary glomerulopathy	97566	**36**	**34**
Non-amyloid monoclonal immunoglobulin deposition disease	86861	**233**	**198**
Non-distal deletion 10q syndrome	1581	**44**	**44**
Non-distal deletion 12q syndrome	96160	**<10**	**<10**
Non-distal duplication 10q syndrome	1695	**11**	**10**
Non-distal duplication 13q syndrome	1702	**19**	**15**
Non-distal duplication 9q syndrome	96112	**<10**	**<10**
Non-functioning neuroendocrine tumor of pancreas	506075	**<10**	**<10**
Non-functioning paraganglioma	94080	**341**	**331**
Non-functioning pituitary adenoma	91349	**6015**	**5633**
Non-histaminic angioedema*	658	**587**	**580**
Non-insulinoma pancreatogenous hypoglycemia syndrome	276608	**20**	**20**
Non-involuting congenital hemangioma	141179	**348**	**348**
Non-Langerhans cell histiocytosis*	157987	**34**	**32**
Non-malignant and non-cirrhotic portal vein thrombosis	854	**4988**	**4310**
Non-paraneoplastic sensory ganglionopathy	208989	**128**	**121**
Non-progressive cerebellar ataxia with intellectual disability	314647	**38**	**38**
Non-recovering obstetric brachial plexus lesion	439202	**47**	**47**
Non-rhizomelic chondrodysplasia punctata*	176	**38**	**38**
Non-saccular limited dorsal myeloschisis	645343	**<10**	**<10**
Non-seminomatous germ cell tumor of testis	363494	**<10**	**<10**
Non-specific autoimmune brainstem encephalitis with characteristic antibodies	624199	**63**	**60**
Non-specific autoimmune brainstem encephalitis without characteristic antibodies	624216	**22**	**21**
Non-specific autoimmune cerebellar ataxia with characteristic antibodies	624259	**27**	**25**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Non-specific autoimmune cerebellar ataxia without characteristic antibodies	624268	**11**	**11**
Non-specific autoimmune supratentorial encephalitis with characteristic antibodies	624166	**237**	**214**
Non-specific autoimmune supratentorial encephalitis without characteristic antibodies	624178	**113**	**107**
Non-specific early-onset epileptic encephalopathy	442835	**453**	**424**
Non-specific interstitial pneumonia	91364	**1916**	**1608**
Non-specific syndromic intellectual disability	528084	**2057**	**2040**
Non-spherocytic hemolytic anemia due to hexokinase deficiency	90031	**<10**	**<10**
Non-syndromic agammaglobulinemia	229717	**487**	**474**
Non-syndromic amelia*	294925	**<10**	**<10**
Non-syndromic anal stenosis	601008	**25**	**24**
Non-syndromic anorectal malformation without fistula	601002	**51**	**51**
Non-syndromic anorectal malformation*	557	**533**	**529**
Non-syndromic bicoronal and metopic craniosynostosis	620198	**<10**	**<10**
Non-syndromic bicoronal and sagittal craniosynostosis	620205	**<10**	**<10**
Non-syndromic bicoronal craniosynostosis	35099	**228**	**228**
Non-syndromic bilambdoid and sagittal craniosynostosis	1516	**23**	**23**
Non-syndromic bilambdoid craniosynostosis	620178	**<10**	**<10**
Non-syndromic bridging bronchus	648992	**<10**	**<10**
Non-syndromic cloacal malformation	600998	**102**	**102**
Non-syndromic congenital bronchial atresia	649010	**19**	**19**
Non-syndromic craniosynostosis*	139390	**147**	**146**
Non-syndromic genetic deafness	87884	**6841**	**6808**
Non-syndromic hemimelia*	2130	**13**	**13**
Non-syndromic H-type fistula	601033	**<10**	**<10**
Non-syndromic intercalary limb defects*	294927	**10**	**10**
Non-syndromic male infertility due to sperm motility disorder	276234	**27**	**27**
Non-syndromic metopic craniosynostosis	3366	**1164**	**1160**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Non-syndromic non-specific multisutural craniosynostosis	620158	<10	<10
Non-syndromic perineal fistula	600952	218	218
Non-syndromic pontocerebellar hypoplasia*	98523	134	106
Non-syndromic posterior hypospadias	95706	1853	1838
Non-syndromic pouch colon	601013	<10	<10
Non-syndromic rectal atresia	601018	15	15
Non-syndromic rectal stenosis	601023	<10	<10
Non-syndromic rectourethral fistula	600961	101	100
Non-syndromic rectovaginal fistula	601028	29	29
Non-syndromic rectovesical fistula	600984	23	23
Non-syndromic sagittal craniosynostosis	35093	2210	2209
Non-syndromic supernumerary kidneys	652528	<10	<10
Non-syndromic unicoronal craniosynostosis	620102	32	32
Non-syndromic unifrontosphenoidal craniosynostosis	620139	<10	<10
Non-syndromic unilambdoid craniosynostosis	620113	<10	<10
Non-syndromic vestibular fistula	600993	72	72
Non-terminal myelocystocele	645340	<10	<10
Non-transplant-related bronchiolitis obliterans	658612	88	87
Noonan syndrome	648	2936	2830
Noonan syndrome with multiple lentigines	500	190	184
Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	363972	27	27
Noonan syndrome-like disorder with loose anagen hair	2701	80	79
Norrie disease	649	68	68
North Carolina macular dystrophy	75327	48	47
NPHP3-related Meckel-like syndrome	3032	<10	<10
NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	600663	13	13
NTHL1-related polyposis	454840	<10	<10
NUT midline carcinoma	443167	<10	<10
Oblique facial cleft*	141253	14	14
Occipital horn syndrome	198	14	12
Occipital pachygyria and polymicrogyria	280640	26	25

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Occult macular dystrophy	247834	127	125
Ocular albinism with late-onset sensorineural deafness	1000	<10	<10
Ocular albinism*	284804	252	252
Ocular anomalies-axonal neuropathy-developmental delay syndrome	496790	12	<10
Ocular cicatricial pemphigoid	99922	104	99
Ocular motor apraxia, Cogan type	1125	67	66
Oculoauricular syndrome, Schorderet type	157962	<10	<10
Oculoauriculofrontonasal syndrome	398156	<10	<10
Oculo-auriculo-vertebral spectrum	141132	190	189
Oculoauriculovertebral spectrum with radial defects	2549	41	41
Oculocerebral hypopigmentation syndrome, Cross type	2719	<10	<10
Oculocerebrocutaneous syndrome	1647	<10	<10
Oculocerebrofacial syndrome, Kaufman type	2707	<10	<10
Oculocerebrorenal syndrome of Lowe	534	111	100
Oculocutaneous albinism type 1	352731	244	244
Oculocutaneous albinism type 2	79432	180	179
Oculocutaneous albinism type 3	79433	15	15
Oculocutaneous albinism type 4	79435	35	34
Oculocutaneous albinism type 5	370091	<10	<10
Oculocutaneous albinism type 6	370097	<10	<10
Oculocutaneous albinism type 8	597733	<10	<10
Oculocutaneous albinism*	55	1228	1219
Oculodentodigital dysplasia	2710	67	64
Oculoectodermal syndrome	3339	<10	<10
Oculofaciocardiodental syndrome	2712	45	45
Oculomaxillofacial dysostosis	1794	<10	<10
Oculoosteocutaneous syndrome	2713	<10	<10
Oculootodental syndrome	99806	<10	<10
Oculo-palato-cerebral syndrome	2714	<10	<10
Oculopharyngeal muscular dystrophy	270	593	470
Oculopharyngodistal myopathy	98897	45	37
Odontochondrodysplasia	166272	<10	<10
Odontomatosis-aortae esophagus stenosis syndrome	2724	<10	<10
Odontomicronychial dysplasia	1811	14	14
Odonto-onycho-dermal dysplasia	2721	13	13
Ogden syndrome	276432	19	19

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Oguchi disease	75382	<10	<10
Okihiro syndrome	93293	169	168
Oligoarticular juvenile idiopathic arthritis	85410	4772	4764
Oligoastrocytoma	251656	<10	<10
Oligocone trichromacy	75378	<10	<10
Oligodendroglial tumor*	46484	<10	<10
Oligodendroglioma	251627	43	37
Oligodontia	99798	2973	2972
Oligomeganephronia	2260	245	242
Olivopontocerebellar atrophy-deafness syndrome	2732	<10	<10
Ollier disease	296	372	365
Omenn syndrome	39041	23	20
Omodysplasia	2733	<10	<10
Omphalocele	660	234	217
Omphalomesenteric cyst	490	<10	<10
Oncogenic osteomalacia	352540	40	38
Onychocytic matricoma	300504	<10	<10
Open spinal dysraphism with a myelomeningocele	93969	766	742
Open spinal dysraphism*	268369	54	49
Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome	2743	<10	<10
Opitz GBBB syndrome	2745	56	55
Opsismodysplasia	2746	<10	<10
Opsoclonus-myoclonus syndrome	1183	122	121
Optic atrophy-ataxia-peripheral neuropathy-global developmental delay syndrome	543470	<10	<10
Optic atrophy-intellectual disability syndrome	401777	57	56
Optic disc pit	519404	22	22
Optic pathway glioma	2086	349	331
Orbital leiomyoma	52994	<10	<10
Ornithine transcarbamylase deficiency	664	463	422
Orofaciodigital syndrome type 1	2750	69	68
Orofaciodigital syndrome type 2	2751	<10	<10
Orofaciodigital syndrome type 4	2753	<10	<10
Orofaciodigital syndrome type 5	2919	<10	<10
Orofaciodigital syndrome type 6	2754	<10	<10
Orofaciodigital syndrome type 9	141007	<10	<10
Orofaciodigital syndrome*	140997	40	40

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Oromandibular dystonia	93958	36	33
Osgood-Schlatter disease	97335	41	41
Ossification anomalies-psychomotor developmental delay syndrome	73230	<10	<10
Osteoblastoma	58040	<10	<10
Osteochondritis dissecans	2764	32	32
Osteochondrosis of the metatarsal bone	564003	<10	<10
Osteochondrosis of the tarsal bone	563991	<10	<10
Osteocraniostenosis	2763	360	357
Osteofibrous dysplasia	488265	27	27
Osteogenesis imperfecta	666	3639	3566
Osteoglosphonic dysplasia	2645	<10	<10
Osteomesopyknosis	2777	14	14
Osteonecrosis of the jaw	399293	44	41
Osteopathia striata-cranial sclerosis syndrome	2780	42	41
Osteopathia striata-pigmentary dermopathy-white forelock syndrome	2779	<10	<10
Osteopetrosis and related disorders*	2781	66	62
Osteopetrosis with renal tubular acidosis	2785	31	29
Osteopetrosis-hypogammaglobulinemia syndrome	178389	<10	<10
Osteoporosis-oculocutaneous hypopigmentation syndrome	2786	<10	<10
Osteoporosis-pseudoglioma syndrome	2788	19	18
Osteoradionecrosis of the mandible	521127	14	14
Osteosarcoma	668	80	76
Osteosclerosis-developmental delay-craniosynostosis syndrome	178377	<10	<10
Osteosclerosis-ichthyosis-premature ovarian failure syndrome	75325	<10	<10
Osteosclerotic bone dysplasia	1832	<10	<10
Osteosclerotic metaphyseal dysplasia	500548	<10	<10
O'Sullivan-McLeod syndrome	99965	12	12
Otodental syndrome	2791	13	13
Otofaciocervical syndrome	2792	<10	<10
Otopalatodigital syndrome spectrum disorder*	364541	<10	<10
Otopalatodigital syndrome type 1	90650	18	17
Otopalatodigital syndrome type 2	90652	11	10
Ovarian dysgerminoma	99912	13	13
Ovarian fibroma	314473	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ovarian fibrothecoma	314478	<10	<10
Ovarian hyperstimulation syndrome	64739	<10	<10
Overgrowth syndrome with 2q37 translocation	498488	<10	<10
Overgrowth-macrocephaly-facial dysmorphism syndrome	137634	17	16
Overhydrated hereditary stomatocytosis	3203	<10	<10
Overlap myositis	206572	774	716
Overlapping connective tissue disease*	251312	20	20
Pachydermoperiostosis	2796	49	48
Pachygyria-intellectual disability-epilepsy syndrome	2798	81	76
Pachyonychia congenita	2309	79	79
Paget disease of the nipple	180275	<10	<10
PAGOD syndrome	991	<10	<10
Pai syndrome	1993	26	26
Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	477993	<10	<10
Pallister-Hall syndrome	672	66	62
Pallister-Killian syndrome	884	91	82
Palmoplantar keratoderma, Nagashima type	140966	<10	<10
Palmoplantar keratoderma-deafness syndrome	2202	19	19
Palmoplantar keratoderma-spastic paralysis syndrome	2201	<10	<10
Pancreatic hypoplasia-diabetes-congenital heart disease syndrome	2255	<10	<10
Pancreatic solid pseudopapillary neoplasm	424065	33	33
Pancreatic triacylglycerol lipase deficiency	309031	<10	<10
Pancreatoblastoma	677	<10	<10
Pancytopenia-developmental delay syndrome	401764	<10	<10
PANDAS	66624	<10	<10
Panhypophysitis	95513	117	104
Panner disease	97336	<10	<10
Pantothenate kinase-associated neurodegeneration	157850	34	26
PAPA syndrome	69126	16	16
PAPASH syndrome	641380	<10	<10
Papillary glioneuronal tumor	251962	<10	<10
Papillary hemangioma	673543	<10	<10
Papillary renal cell carcinoma	319298	28	25
Papilloma of choroid plexus	2807	11	<10
Papillon-Lefèvre syndrome	678	38	36
Papular elastorrhexis	228264	<10	<10
Papular mucinosis of infancy	90395	<10	<10
Papular xanthoma	158008	<10	<10
Paracetamol poisoning	464458	<10	<10
Paramedian facial cleft*	155867	17	17
Paramedian nasal cleft	141242	22	22
Paramyotonia congenita of Von Eulenburg	684	308	295
Paraneoplastic cerebellar degeneration	623626	48	47
Paraneoplastic isolated brainstem encephalitis	624190	11	10
Paraneoplastic pemphigus	63455	52	34
Paraneoplastic sensory ganglionopathy	208999	62	55
Paraneoplastic uveitis	279928	<10	<10
Paraparetic variant of Guillain-Barré syndrome	231445	18	15
Paraplegia-intellectual disability-hyperkeratosis syndrome	2824	<10	<10
Paratesticular adenocarcinoma	363478	<10	<10
Parathyroid carcinoma	143	12	11
PARC syndrome	2825	<10	<10
Parenteral nutrition-associated cholestasis	567983	<10	<10
Parietal foramina with clavicular hypoplasia	251290	<10	<10
Paris-Trousseau thrombocytopenia	851	<10	<10
Parkes Weber syndrome	90307	103	101
Parkinson-dementia complex of Guam	90020	17	<10
Parkinsonian-pyramidal syndrome	171695	72	60
Paroxysmal cold hemoglobinuria	90035	<10	<10
Paroxysmal dyskinesia*	1431	69	68
Paroxysmal dystonia*	200037	40	39
Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	53583	17	17
Paroxysmal exertion-induced dyskinesia	98811	12	12
Paroxysmal extreme pain disorder	46348	51	49
Paroxysmal kinesigenic dyskinesia	98809	145	145

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Paroxysmal nocturnal hemoglobinuria	447	**396**	**362**
Paroxysmal non-kinesigenic dyskinesia	98810	**32**	**32**
Partial androgen insensitivity syndrome	90797	**160**	**159**
Partial atrioventricular septal defect	1330	**746**	**728**
Partial bilateral aplasia of the MÃ¼llerian ducts*	180068	**<10**	**<10**
Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome	401959	**<10**	**<10**
Partial deep dermal and full thickness burns	90076	**<10**	**<10**
Partial septate uterus	180129	**<10**	**<10**
Partially involuting congenital hemangioma	458785	**46**	**46**
Partington syndrome	94083	**27**	**25**
PASH syndrome	289478	**<10**	**<10**
PASS syndrome	641385	**<10**	**<10**
Paternal 20q13.2q13.3 microdeletion syndrome	261304	**<10**	**<10**
Paternal uniparental disomy of chromosome 6 syndrome	96191	**<10**	**<10**
Paternal uniparental disomy of chromosome 7 syndrome	96192	**<10**	**<10**
Patterson-Stevenson-Fontaine syndrome	2439	**10**	**10**
Pauci-immune glomerulonephritis	93126	**289**	**252**
PBX1-related congenital anomalies of kidney-urinary tract syndrome	656130	**<10**	**<10**
Pearson syndrome	699	**49**	**33**
Pectus excavatum-macrocephaly-dysplastic nails syndrome	2835	**<10**	**<10**
Pediatric arterial ischemic stroke	439175	**571**	**567**
Pediatric collagenous gastritis	487809	**<10**	**<10**
Pediatric multiple sclerosis	477738	**342**	**342**
Pediatric systemic lupus erythematosus	93552	**682**	**666**
Pediatric-onset Graves disease	525731	**991**	**987**
Peeling skin syndrome*	817	**29**	**29**
PEHO syndrome	2836	**<10**	**<10**
PEHO-like syndrome	99807	**<10**	**<10**
Pelizaeus-Merzbacher disease	702	**153**	**139**
Pelizaeus-Merzbacher-like disease	280270	**26**	**26**
Pellucid marginal degeneration	137672	**86**	**85**
Pelviscapular dysplasia	93333	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Pelvis-shoulder dysplasia	2839	**<10**	**<10**
Pemphigoid gestationis	63275	**112**	**111**
Pemphigus erythematosus	79480	**21**	**20**
Pemphigus foliaceus	79481	**22**	**20**
Pemphigus vegetans	79479	**13**	**10**
Pemphigus vulgaris	704	**1095**	**1034**
Pendred syndrome	705	**404**	**400**
Penile agenesis	49	**<10**	**<10**
Penoscrotal transposition	2842	**<10**	**<10**
PENS syndrome	313936	**<10**	**<10**
Pentalogy of Cantrell	1335	**<10**	**<10**
Pentasomy X syndrome	11	**<10**	**<10**
Pericardial and diaphragmatic defect	2847	**15**	**14**
Perineurioma*	85102	**12**	**12**
Periodic fever-infantile enterocolitis-autoinflammatory syndrome	436166	**16**	**15**
Periodic paralysis with later-onset distal motor neuropathy	397750	**<10**	**<10**
Periodic paralysis with transient compartment-like syndrome	397755	**<10**	**<10**
Periodic paralysis*	206976	**64**	**63**
Periodontal Ehlers-Danlos syndrome	75392	**24**	**23**
Perioral myoclonia with absences	139426	**<10**	**<10**
Peripapillary staphyloma	519400	**<10**	**<10**
Peripartum cardiomyopathy	563	**17**	**16**
Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	163746	**13**	**10**
Peripheral motor neuropathy-dysautonomia syndrome	2400	**63**	**58**
Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome	397744	**<10**	**<10**
Peripheral primitive neuroectodermal tumor	370348	**<10**	**<10**
Peripheral pulmonary stenosis	99084	**81**	**81**
Peritoneal inclusion cyst	168816	**<10**	**<10**
Perivascular epithelioid cell neoplasm	595133	**<10**	**<10**
Perlman syndrome	2849	**<10**	**<10**
Peroxisomal acyl-CoA oxidase deficiency	2971	**<10**	**<10**
Peroxisome biogenesis disorder*	79189	**25**	**17**
Perrault syndrome	2855	**60**	**59**
Perry syndrome	178509	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Persistent combined dystonia*	391711	**48**	**46**
Persistent fifth aortic arch	99076	**<10**	**<10**
Persistent hyperplastic primary vitreous	91495	**323**	**322**
Persistent idiopathic facial pain	398147	**<10**	**<10**
Persistent left superior vena cava connecting through coronary sinus to left-sided atrium	99109	**26**	**26**
Persistent left superior vena cava connecting to the roof of left-sided atrium	99111	**<10**	**<10**
Persistent Müllerian duct syndrome	2856	**44**	**42**
Persistent placoid maculopathy	97341	**<10**	**<10**
Persistent polyclonal B-cell lymphocytosis	300324	**<10**	**<10**
Peters anomaly	708	**230**	**226**
Peters plus syndrome	709	**60**	**59**
Peutz-Jeghers syndrome	2869	**200**	**193**
PFAPA syndrome	42642	**2477**	**2477**
Pfeiffer syndrome	710	**115**	**108**
PGM1-CDG	319646	**<10**	**<10**
PGM3-CDG	443811	**<10**	**<10**
PHACE syndrome	42775	**184**	**184**
Phacoanaphylactic uveitis	209959	**<10**	**<10**
Phakomatosis pigmentokeratotica	2874	**20**	**19**
Phakomatosis pigmentovascularis	2875	**104**	**102**
Phalangeal microgeodic syndrome	352636	**<10**	**<10**
Pharyngeal-cervical-brachial variant of Guillain-Barré syndrome	231426	**10**	**10**
Phelan-McDermid syndrome	48652	**370**	**363**
Phenobarbital embryopathy	1919	**<10**	**<10**
Phenylketonuria	716	**3024**	**3009**
PHIP-related behavioral problems-intellectual disability-obesity-dysmorphic features syndrome	589905	**27**	**27**
Phocomelia, Schinzel type	2879	**11**	**10**
Phosphoenolpyruvate carboxykinase deficiency	2880	**<10**	**<10**
Phosphoribosylpyrophosphate synthetase superactivity	3222	**<10**	**<10**
Photosensitive occipital lobe epilepsy	166409	**98**	**97**
Phyllodes tumor of the breast	180261	**16**	**16**
Piebald trait-neurologic defects syndrome	2885	**<10**	**<10**
Piebaldism	2884	**116**	**116**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Pierpont syndrome	487825	**17**	**17**
Pierre Robin syndrome-faciodigital anomaly syndrome	2888	**86**	**83**
Pierson syndrome	2670	**<10**	**<10**
PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis	568062	**<10**	**<10**
Pigmented paravenous retinochoroidal atrophy	251295	**13**	**13**
Pili bifurcati	720	**<10**	**<10**
Pilocytic astrocytoma	251612	**126**	**121**
Pilodental dysplasia-refractive errors syndrome	2892	**16**	**16**
Pilomatrixoma	91414	**85**	**85**
Pineal tumor of neuroepithelial tissue*	251905	**<10**	**<10**
Pineoblastoma	251909	**<10**	**<10**
Pineocytoma	251912	**<10**	**<10**
Pinnae fistula or cyst	155838	**190**	**189**
Pitt-Hopkins syndrome	2896	**217**	**214**
Pituicytoma	251623	**22**	**22**
Pituitary adenoma*	99408	**604**	**573**
Pituitary apoplexy	95613	**581**	**548**
Pituitary carcinoma	300385	**25**	**17**
Pituitary deficiency due to empty sella turcica syndrome	91354	**209**	**201**
Pituitary deficiency due to Rathke cleft cysts	91350	**788**	**767**
Pituitary dermoid and epidermoid cysts	91351	**217**	**216**
Pituitary gigantism	99725	**<10**	**<10**
Pituitary stalk interruption syndrome	95496	**1215**	**1207**
Pityriasis rubra pilaris	2897	**32**	**32**
PLA2G6-associated neurodegeneration*	329303	**10**	**10**
PLAA-associated neurodevelopmental disorder	521426	**<10**	**<10**
Placental insufficiency	439167	**<10**	**<10**
Placental site trophoblastic tumor	99928	**<10**	**<10**
Plague	707	**<10**	**<10**
Plasma cell leukemia	454714	**<10**	**<10**
Plasmacytoma	86855	**<10**	**<10**
Plastic bronchitis	439881	**24**	**23**
Platyspondylic dysplasia, Torrance type	85166	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
PLCG2-associated antibody deficiency and immune dysregulation	300359	<10	<10
PLEC-related intermediate epidermolysis bullosa simplex without extracutaneous involvement	79401	<10	<10
Plectin-related limb-girdle muscular dystrophy R17	254361	<10	<10
Pleomorphic xanthoastrocytoma	251607	<10	<10
Pleural empyema	449266	48	48
Pleural mesothelioma	50251	15	<10
Pleuro-pericardial cyst	99131	<10	<10
Pleuropulmonary blastoma	64742	<10	<10
PLIN1-related familial partial lipodystrophy	280356	24	24
Plummer-Vinson syndrome	54028	<10	<10
PMM2-CDG	79318	130	112
PMP22-RAI1 contiguous gene duplication syndrome	477817	<10	<10
PMP2-related Charcot-Marie-Tooth disease type 1	476394	23	23
Pneumococcal meningitis	55655	76	70
Pneumoconiosis*	182098	144	122
Pneumocystosis	723	28	26
Pneumonia caused by Pseudomonas aeruginosa infection	90066	40	38
POEMS syndrome	2905	195	166
POGLUT1-related limb-girdle muscular dystrophy R21	480682	<10	<10
Poikiloderma with neutropenia	221046	<10	<10
Poland syndrome	2911	525	524
Poliomyelitis	2912	96	86
Polyarteritis nodosa	767	757	715
Polyarticular juvenile idiopathic arthritis*	404580	192	191
Polycythemia vera	729	163	152
Polycythemia*	98427	103	102
Polydactyly of a biphalangeal thumb and/or hallux	93339	191	189
Polydactyly of a triphalangeal thumb	93336	22	22
Polydactyly of an index finger	93337	<10	<10
Polydactyly-myopia syndrome	2917	<10	<10
Polyendocrine-polyneuropathy syndrome	453533	<10	<10
Polyglucosan body myopathy type 1	397937	<10	<10
Polyglucosan body myopathy type 2	456369	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	500533	<10	<10
Polymerase proofreading-related polyposis	447877	<10	<10
Polymicrogyria due to TUBB2B mutation	300573	10	10
Polymicrogyria with optic nerve hypoplasia	250972	<10	<10
Polymicrogyria*	35981	303	283
Polymyositis	732	1139	966
Polyneuropathy associated with IgM monoclonal gammopathy	209004	191	142
Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	171848	11	10
Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome	2928	<10	<10
Polyrrhinia	141091	<10	<10
Polysyndactyly	93338	87	87
POMGNT1-related limb-girdle muscular dystrophy R15	206564	<10	<10
POMGNT2-related limb-girdle muscular dystrophy R24	565899	<10	<10
POMT1-related limb-girdle muscular dystrophy R11	86812	16	13
POMT2-related limb-girdle muscular dystrophy R14	206559	<10	<10
Pontine autosomal dominant microangiopathy with leukoencephalopathy	477749	14	11
Pontine tegmental cap dysplasia	269229	<10	<10
Pontocerebellar hypoplasia type 1	2254	35	25
Pontocerebellar hypoplasia type 10	411493	<10	<10
Pontocerebellar hypoplasia type 2	2524	50	38
Pontocerebellar hypoplasia type 3	97249	10	<10
Pontocerebellar hypoplasia type 4	166063	<10	<10
Pontocerebellar hypoplasia type 6	166073	15	11
Pontocerebellar hypoplasia type 7	284339	<10	<10
Pontocerebellar hypoplasia type 9	369920	<10	<10
Popliteal pterygium syndrome*	294963	<10	<10
Porencephaly	2940	95	90
Porencephaly-cerebellar hypoplasia-internal malformations syndrome	2941	<10	<10
Porencephaly-microcephaly-bilateral congenital cataract syndrome	306547	<10	<10
Porokeratosis of Mibelli	735	32	31

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Porokeratosis plantaris palmaris et disseminata	737	**17**	**16**
Porokeratotic eccrine ostial and dermal duct nevus	166286	**<10**	**<10**
Porphyria cutanea tarda	101330	**3488**	**2713**
Porphyria due to ALA dehydratase deficiency	100924	**<10**	**<10**
Porphyria*	738	**30**	**26**
Portosinusoidal vascular disease	596937	**2760**	**2424**
Postaxial acrofacial dysostosis	246	**15**	**14**
Postaxial polydactyly type A	93334	**91**	**91**
Postaxial polydactyly type B	93335	**181**	**181**
Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome	420584	**<10**	**<10**
Postaxial tetramelic oligodactyly	2730	**14**	**14**
Posterior amorphous corneal dystrophy	98971	**<10**	**<10**
Posterior column ataxia-retinitis pigmentosa syndrome	88628	**<10**	**<10**
Posterior cortical atrophy	54247	**84**	**73**
Posterior polymorphous corneal dystrophy	98973	**40**	**39**
Posterior urethral valve	93110	**2028**	**2008**
Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome	572013	**<10**	**<10**
Postinfectious cerebellitis	624244	**107**	**106**
Postinfectious vasculitis	48435	**61**	**61**
Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	477673	**<10**	**<10**
Postpartum psychosis	443173	**<10**	**<10**
Postpoliomyelitis syndrome	2942	**317**	**272**
Post-traumatic pituitary deficiency	95619	**2994**	**2745**
Postural orthostatic tachycardia syndrome due to NET deficiency	443236	**<10**	**<10**
Potassium-aggravated myotonia*	612	**<10**	**<10**
Potocki-Shaffer syndrome	52022	**23**	**23**
PPARG-related familial partial lipodystrophy	79083	**28**	**25**
Prader-Willi syndrome	739	**1790**	**1660**
Prader-Willi-like syndrome*	398073	**34**	**34**
Precursor B-cell acute lymphoblastic leukemia	99860	**56**	**51**
Precursor T-cell acute lymphoblastic leukemia	99861	**14**	**12**
Pre-Descemet corneal dystrophy	293462	**<10**	**<10**
Predisposition to invasive fungal disease due to CARD9 deficiency	457088	**<10**	**<10**
Predisposition to severe viral infection due to IRF7 deficiency	574918	**<10**	**<10**
Predominantly large-vessel vasculitis*	156140	**66**	**63**
Predominantly medium-vessel vasculitis*	156143	**38**	**36**
Predominantly small-vessel vasculitis*	156146	**66**	**55**
Preeclampsia	275555	**23**	**21**
Premature closure of the arterial duct	95486	**<10**	**<10**
Prenatal-onset spinal muscular atrophy with congenital bone fractures	486811	**<10**	**<10**
Prepubertal anorexia nervosa	525738	**777**	**775**
Pressure-induced localized lipoatrophy	90160	**<10**	**<10**
Primary acquired pure red cell aplasia	98872	**<10**	**<10**
Primary anetoderma	228272	**35**	**35**
Primary angiitis of the central nervous system	140989	**120**	**115**
Primary autoimmune enteropathy	522037	**10**	**<10**
Primary biliary cholangitis	186	**4529**	**4151**
Primary biliary cholangitis/ primary sclerosing cholangitis and autoimmune hepatitis overlap syndrome	562639	**125**	**120**
Primary bone and joint tuberculosis	645822	**<10**	**<10**
Primary bone dysplasia with multiple joint dislocations*	93441	**28**	**27**
Primary bone lymphoma	314684	**<10**	**<10**
Primary central nervous system lymphoma	46135	**<10**	**<10**
Primary central precocious puberty in male	650087	**34**	**34**
Primary ciliary dyskinesia	244	**1472**	**1457**
Primary ciliary dyskinesia-retinitis pigmentosa syndrome	247522	**<10**	**<10**
Primary congenital hypothyroidism*	226295	**215**	**214**
Primary cutaneous aggressive epidermotropic CD8+ T-cell lymphoma	178528	**<10**	**<10**
Primary cutaneous amyloidosis*	137807	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Primary cutaneous anaplastic large cell lymphoma	300865	**<10**	**<10**
Primary cutaneous CD4+ small/ medium-sized pleomorphic T-cell lymphoma	178522	**<10**	**<10**
Primary cutaneous follicle center lymphoma	178540	**<10**	**<10**
Primary cutaneous gamma/delta-positive T-cell lymphoma	178533	**<10**	**<10**
Primary cutaneous marginal zone B-cell lymphoma	178536	**<10**	**<10**
Primary cutaneous peripheral T-cell lymphoma not otherwise specified	86885	**<10**	**<10**
Primary cutaneous plasmacytosis	451602	**<10**	**<10**
Primary cutaneous tuberculosis	645849	**<10**	**<10**
Primary cutis verticis gyrata*	671	**<10**	**<10**
Primary dystonia, DYT13 type	98807	**25**	**25**
Primary dystonia, DYT17 type	370103	**<10**	**<10**
Primary dystonia, DYT2 type	99657	**<10**	**<10**
Primary dystonia, DYT21 type	306734	**19**	**19**
Primary dystonia, DYT27 type	464440	**138**	**136**
Primary dystonia, DYT4 type	98805	**<10**	**<10**
Primary dystonia, DYT6 type	98806	**32**	**32**
Primary early-onset glaucoma*	156005	**11**	**11**
Primary eosinophilic gastrointestinal disease*	402029	**<10**	**<10**
Primary erythromelalgia	90026	**207**	**204**
Primary essential cutis verticis gyrata	357220	**<10**	**<10**
Primary failure of tooth eruption	412206	**86**	**86**
Primary familial polycythemia	90042	**15**	**15**
Primary Fanconi renotubular syndrome	3337	**67**	**56**
Primary genito-urinary tuberculosis	645874	**<10**	**<10**
Primary hemophagocytic lymphohistiocytosis*	158038	**10**	**<10**
Primary hepatic neuroendocrine carcinoma	100085	**<10**	**<10**
Primary hyperaldosteronism-seizures-neurological abnormalities syndrome	369929	**<10**	**<10**
Primary hypereosinophilic syndrome	314950	**143**	**130**
Primary hypergonadotropic hypogonadism-partial alopecia syndrome	2232	**<10**	**<10**
Primary hyperoxaluria	416	**325**	**312**
Primary hypertrophic osteoarthropathy*	248095	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Primary hypomagnesemia with hypercalciuria and nephrocalcinosis	306516	**75**	**72**
Primary hypomagnesemia with secondary hypocalcemia	30924	**68**	**66**
Primary hypomagnesemia-refractory seizures-intellectual disability syndrome	564178	**<10**	**<10**
Primary hypophysitis*	95506	**54**	**46**
Primary immunodeficiency syndrome due to P14/LAMTOR2 deficiency	90023	**<10**	**<10**
Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency	75391	**<10**	**<10**
Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection	431166	**<10**	**<10**
Primary intestinal lymphangiectasia	90362	**112**	**105**
Primary intrahepatic lithiasis	480506	**42**	**39**
Primary intraocular lymphoma	279904	**12**	**10**
Primary intraosseous venous malformation	140436	**63**	**63**
Primary laryngeal lymphangioma	137926	**18**	**18**
Primary lateral sclerosis	35689	**596**	**470**
Primary mediastinal large B-cell lymphoma	98838	**<10**	**<10**
Primary melanoma of the central nervous system	252050	**<10**	**<10**
Primary membranoproliferative glomerulonephritis	54370	**879**	**809**
Primary membranous glomerulonephritis	97560	**1972**	**1806**
Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome	306558	**<10**	**<10**
Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	391408	**<10**	**<10**
Primary myelofibrosis	824	**27**	**21**
Primary non-essential cutis verticis gyrata	357225	**<10**	**<10**
Primary oculocerebral lymphoma	279897	**23**	**18**
Primary orthostatic hypotension*	182058	**<10**	**<10**
Primary orthostatic tremor	238606	**116**	**114**
Primary pediatric heart tumor	875	**22**	**20**
Primary peritoneal carcinoma	168829	**<10**	**<10**
Primary progressive aphasia*	95432	**78**	**69**
Primary progressive apraxia of speech	314566	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Primary progressive freezing gait	75567	<10	<10
Primary pulmonary hypoplasia	2257	71	66
Primary pulmonary lymphoma	2420	<10	<10
Primary pulmonary tuberculosis	645814	95	95
Primary pulmonary vein stenosis	642071	<10	<10
Primary renal tubular acidosis*	314822	17	17
Primary sclerosing cholangitis	171	3130	2926
Primary short bowel syndrome*	365563	11	11
Primary Sjögren disease	289390	11479	11174
Primary syringomyelia	99856	658	645
Primary tethered cord syndrome	268861	502	501
Primary tuberculosis of the digestive system	645859	<10	<10
Primary tuberculous lymphadenitis	645807	<10	<10
Primary unilateral adrenal hyperplasia	231580	1346	1338
Primitive neuroectodermal tumor of the cervix uteri	213812	<10	<10
Proboscis lateralis	141099	10	10
Progeria-short stature-pigmented nevi syndrome	2959	<10	<10
Progeroid and marfanoid aspect-lipodystrophy syndrome	300382	<10	<10
Progeroid features-hepatocellular carcinoma predisposition syndrome	435953	<10	<10
Progressive autosomal recessive ataxia-deafness syndrome	448251	<10	<10
Progressive bifocal chorioretinal atrophy	75373	<10	<10
Progressive cavitating leukoencephalopathy	139447	<10	<10
Progressive cerebello-cerebral atrophy	247198	24	21
Progressive cone dystrophy	1871	578	571
Progressive deafness with stapes fixation	3235	22	22
Progressive encephalopathy-severe neurodegeneration-lipodystrophy syndrome	363400	<10	<10
Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	457212	<10	<10
Progressive external ophthalmoplegia-myopathy-emaciation syndrome	352447	<10	<10
Progressive familial intrahepatic cholestasis	172	439	426
Progressive hemifacial atrophy	1214	73	73
Progressive hypotonia-intellectual disability-facial dysmorphism syndrome due to FYVE-defective RBSN	675782	<10	<10
Progressive multifocal leukoencephalopathy	217260	13	11
Progressive muscular atrophy	454706	61	49
Progressive myoclonic epilepsy type 1	308	111	109
Progressive myoclonic epilepsy type 3	263516	<10	<10
Progressive myoclonic epilepsy type 5	402082	<10	<10
Progressive myoclonic epilepsy type 7	435438	<10	<10
Progressive myoclonic epilepsy type 8	424027	<10	<10
Progressive myoclonic epilepsy type 9	457265	<10	<10
Progressive myoclonic epilepsy with dystonia	352596	<10	<10
Progressive myoclonic epilepsy*	98261	41	40
Progressive non-fluent aphasia	100070	415	347
Progressive non-infectious anterior vertebral fusion	2062	<10	<10
Progressive osseous heteroplasia	2762	22	22
Progressive pseudorheumatoid dysplasia	1159	26	26
Progressive scapulohumeroperoneal distal myopathy	447977	11	<10
Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	228012	<10	<10
Progressive supranuclear palsy	683	1445	1045
Progressive symmetric erythrokeratodermia	316	13	13
Prolactinoma	2965	7016	6870
Prolidase deficiency	742	<10	<10
Proliferating trichilemmal cyst	492	<10	<10
Properdin deficiency	2966	18	17
Propionic acidemia	35	156	123
Proteasome-associated autoinflammatory syndrome	324977	<10	<10
Protein S acquired deficiency	26349	14	14
Proteus syndrome	744	141	138
Proton-pump inhibitor-responsive esophageal eosinophilia	411696	12	11
Proximal 16p11.2 microdeletion syndrome	261197	232	231
Proximal 16p11.2 microduplication syndrome	370079	130	129
Proximal myopathy with extrapyramidal signs	401768	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Proximal myopathy with focal depletion of mitochondria	521305	**<10**	**<10**
Proximal myotonic myopathy	606	**826**	**717**
Proximal renal tubular acidosis	47159	**64**	**61**
Proximal spinal muscular atrophy	70	**2344**	**1905**
Proximal symphalangism	3250	**17**	**17**
Proximal Xq28 duplication syndrome	1762	**63**	**59**
PrP systemic amyloidosis	397606	**<10**	**<10**
Prune belly syndrome	2970	**92**	**88**
PRUNE1-related neurological syndrome	544469	**<10**	**<10**
Pruritic urticarial papules and plaques of pregnancy	64745	**<10**	**<10**
Pseudoachondroplasia	750	**89**	**89**
Pseudodiastrophic dysplasia	85174	**<10**	**<10**
Pseudohypoaldosteronism type 1	756	**117**	**112**
Pseudohypoaldosteronism type 2	757	**55**	**55**
Pseudohypoaldosteronism*	444916	**32**	**31**
Pseudohypoparathyroidism type 1A	79443	**322**	**319**
Pseudohypoparathyroidism type 1B	94089	**201**	**199**
Pseudohypoparathyroidism type 1C	79444	**<10**	**<10**
Pseudohypoparathyroidism type 2	94090	**12**	**12**
Pseudohypoparathyroidism with Albright hereditary osteodystrophy*	457059	**44**	**42**
Pseudohypoparathyroidism without Albright hereditary osteodystrophy*	457062	**11**	**11**
Pseudomyogenic hemangioendothelioma	673556	**<10**	**<10**
Pseudomyxoma peritonei	26790	**<10**	**<10**
Pseudoprogeria syndrome	2985	**<10**	**<10**
Pseudopseudohypoparathyroidism	79445	**128**	**126**
Pseudounicornuate uterus	180079	**<10**	**<10**
Pseudo-von Willebrand disease	52530	**31**	**31**
Pseudoxanthoma elasticum	758	**647**	**641**
Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa	436274	**<10**	**<10**
Psoriasis-related juvenile idiopathic arthritis	85436	**504**	**504**
Psychogenic movement disorders	71519	**199**	**193**
Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome	505242	**<10**	**<10**
PTEN hamartoma tumor syndrome*	306498	**727**	**713**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Pterygium colli-intellectual disability-digital anomalies syndrome	2988	**<10**	**<10**
Ptosis-strabismus-ectopic pupils syndrome	2999	**<10**	**<10**
Ptosis-vocal cord paralysis syndrome	2997	**<10**	**<10**
Pudendal nerve entrapment syndrome	60039	**12**	**12**
Pulmonary agenesis	984	**73**	**70**
Pulmonary alveolar microlithiasis	60025	**17**	**11**
Pulmonary arterial hypertension associated with chronic hemolytic anemia*	275828	**13**	**11**
Pulmonary arterial hypertension associated with congenital heart disease*	275803	**529**	**469**
Pulmonary arterial hypertension associated with connective tissue disease*	275798	**551**	**385**
Pulmonary arterial hypertension associated with HIV infection*	275808	**63**	**54**
Pulmonary arterial hypertension associated with portal hypertension*	275813	**321**	**223**
Pulmonary arterial hypertension associated with schistosomiasis*	275823	**<10**	**<10**
Pulmonary arteriovenous malformation	2038	**58**	**56**
Pulmonary artery coming from patent ductus arteriosus	99049	**<10**	**<10**
Pulmonary artery hypoplasia	99083	**30**	**30**
Pulmonary atresia with ventricular septal defect	1207	**728**	**684**
Pulmonary atresia-intact ventricular septum syndrome	1208	**431**	**414**
Pulmonary blastoma	64741	**<10**	**<10**
Pulmonary capillary hemangiomatosis	199241	**<10**	**<10**
Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome	210136	**34**	**27**
Pulmonary hypertension owing to lung disease and/or hypoxia*	275837	**1102**	**745**
Pulmonary hypertension with unclear multifactorial mechanism*	275844	**324**	**235**
Pulmonary interstitial glycogenosis	217557	**<10**	**<10**
Pulmonary nodular lymphoid hyperplasia	60026	**<10**	**<10**
Pulmonary non-tuberculous mycobacterial infection	411703	**299**	**276**
Pulmonary valve agenesis*	982	**64**	**64**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Pulmonary valve agenesis-intact ventricular septum-persistent ductus arteriosus syndrome	99048	**10**	**10**
Pulmonary valve agenesis-tetralogy of Fallot-absence of ductus arteriosus syndrome	101206	**29**	**28**
Pulmonary venoocclusive disease	31837	**328**	**192**
Punctate acrokeratoderma freckle-like pigmentation	99710	**<10**	**<10**
Punctate inner choroidopathy	580951	**27**	**27**
Punctate palmoplantar keratoderma type 1	79501	**15**	**15**
Punctate palmoplantar keratoderma type 2	79502	**10**	**10**
PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	438213	**45**	**45**
Pure autonomic failure	441	**54**	**49**
Pure hair and nail ectodermal dysplasia	69084	**43**	**43**
Pure hereditary spastic paraplegia*	102012	**417**	**406**
Pure mitochondrial myopathy	254854	**542**	**509**
Pure or complex hereditary spastic paraplegia*	320335	**76**	**73**
Purine nucleoside phosphorylase deficiency	760	**<10**	**<10**
Pustulosis palmaris et plantaris	163927	**15**	**15**
Pycnodysostosis	763	**87**	**87**
PYCR2-related microcephaly-progressive leukoencephalopathy	481152	**<10**	**<10**
Pyknoachondrogenesis	3003	**<10**	**<10**
Pyle disease	3005	**<10**	**<10**
Pyoderma gangrenosum	48104	**47**	**42**
Pyomyositis	764	**13**	**13**
Pyridoxamine-5-phosphate deficiency-developmental and epileptic encephalopathy	79096	**18**	**17**
Pyridoxine-dependent-developmental and epileptic encephalopathy	3006	**75**	**71**
Pyruvate carboxylase deficiency	3008	**<10**	**<10**
Pyruvate dehydrogenase deficiency	765	**150**	**131**
Q fever	781	**<10**	**<10**
Qazi-Markouizos syndrome	3010	**<10**	**<10**
QRICH1-related intellectual disability-chondrodysplasia syndrome	580940	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
QRSL1-related combined oxidative phosphorylation defect	570491	**<10**	**<10**
Quadricuspid aortic valve	542568	**<10**	**<10**
Quebec platelet disorder	220436	**57**	**56**
Quinquaud folliculitis decalvans	346	**<10**	**<10**
Rabson-Mendenhall syndrome	769	**<10**	**<10**
Radial deficiency-tibial hypoplasia syndrome	1121	**<10**	**<10**
Radial ray hypoplasia-choanal atresia syndrome	3026	**<10**	**<10**
Radiation myelitis	90021	**27**	**21**
Radiation-induced plexopathy	521123	**64**	**58**
Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome	71289	**<10**	**<10**
Radioulnar synostosis-microcephaly-scoliosis syndrome	3268	**<10**	**<10**
Ramsay Hunt syndrome	3020	**12**	**12**
RAPADILINO syndrome	3021	**<10**	**<10**
Rapidly involuting congenital hemangioma	141184	**264**	**264**
Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	293987	**47**	**43**
Rapid-onset dystonia-parkinsonism	71517	**19**	**17**
Rare adenocarcinoma of the breast	213528	**14**	**12**
Rare combined vascular malformation*	458837	**569**	**565**
Rare cutaneous lupus erythematosus*	535	**221**	**215**
Rare disorder without a determined diagnosis after full investigation	616874	**926**	**904**
Rare hereditary thrombophilia*	217454	**19**	**19**
Rare infantile hemangioma*	210589	**2543**	**2543**
Rare isolated myopia	98619	**516**	**512**
Rare non-syndromic intellectual disability	101685	**17775**	**17537**
RARS-related autosomal recessive hypomyelinating leukodystrophy	438114	**<10**	**<10**
RAS-associated autoimmune leukoproliferative disease	268114	**<10**	**<10**
Rasmussen subacute encephalitis	1929	**123**	**120**
Rat-bite fever	31205	**<10**	**<10**
Rauch-Steindl syndrome	659642	**<10**	**<10**
Ravine syndrome	99852	**63**	**29**
Reactive angioendotheliomatosis	673574	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Reactive arthritis	29207	**597**	**597**
Recessive dystrophic epidermolysis bullosa inversa	79409	**18**	**18**
Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome	280384	**<10**	**<10**
Recessive mitochondrial ataxia syndrome	94125	**14**	**12**
Recessive X-linked ichthyosis	461	**213**	**209**
Recombinant 8 syndrome	96167	**<10**	**<10**
Recurrent hepatitis C virus induced liver disease in liver transplant recipients	90052	**<10**	**<10**
Recurrent idiopathic neuroretinitis	499103	**<10**	**<10**
Recurrent infections associated with rare immunoglobulin isotypes deficiency	183675	**611**	**601**
Recurrent infections due to specific granule deficiency	169142	**<10**	**<10**
Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome	480864	**42**	**38**
Recurrent respiratory papillomatosis	60032	**122**	**119**
Reducing body myopathy	97239	**<10**	**<10**
Reflex epilepsy*	310	**27**	**27**
Refractory celiac disease	398063	**31**	**31**
Refsum disease	773	**31**	**27**
Regional odontodysplasia	83450	**<10**	**<10**
Regressive spondylometaphyseal dysplasia	448267	**<10**	**<10**
Reis-Bücklers corneal dystrophy	98961	**36**	**36**
RELA fusion-positive ependymoma	530792	**<10**	**<10**
Relapsing fever	91547	**190**	**190**
Relapsing polychondritis	728	**732**	**703**
Renal agenesis	411709	**3387**	**3338**
Renal caliceal diverticuli-deafness syndrome	2838	**<10**	**<10**
Renal cell carcinoma*	217071	**32**	**29**
Renal coloboma syndrome	1475	**198**	**194**
Renal dysplasia	93108	**2797**	**2764**
Renal hypoplasia	93101	**3454**	**3411**
Renal medullary carcinoma	319319	**<10**	**<10**
Renal nutcracker syndrome	71273	**16**	**16**
Renal tubular dysgenesis	3033	**46**	**37**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Renal tubulopathy-encephalopathy-liver failure syndrome	254902	**<10**	**<10**
Renal-hepatic-pancreatic dysplasia	294415	**<10**	**<10**
Renin-angiotensin-aldosterone system-blocker-induced angioedema	100057	**1145**	**1090**
Renpenning syndrome	3242	**39**	**39**
RERE-related neurodevelopmental syndrome	494344	**10**	**10**
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha	566231	**20**	**20**
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta	566243	**111**	**111**
Resistance to thyrotropin-releasing hormone syndrome	99832	**13**	**13**
Respiratory bronchiolitis-interstitial lung disease syndrome	79127	**233**	**213**
Restrictive dermopathy	1662	**<10**	**<10**
Reticular dysgenesis	33355	**<10**	**<10**
Reticular dystrophy of the retinal pigment epithelium	99002	**198**	**193**
Reticulate acropigmentation of Kitamura	178307	**<10**	**<10**
Retinal capillary malformation	71213	**25**	**25**
Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies	397758	**<10**	**<10**
Retinal dystrophy-optic nerve edema-splenomegaly-anhidrosis-migraine headache syndrome	313800	**17**	**17**
Retinal macular dystrophy type 2	319640	**52**	**52**
Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	247691	**12**	**10**
Retinitis pigmentosa	791	**5640**	**5599**
Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome	3085	**<10**	**<10**
Retinitis punctata albescens	52427	**37**	**37**
Retinoblastoma	790	**239**	**235**
Retinopathy of prematurity	90050	**575**	**574**
Rett syndrome	778	**823**	**740**
Reversible cerebral vasoconstriction syndrome	284388	**<10**	**<10**
Revesz syndrome	3088	**<10**	**<10**
Reye syndrome	3096	**21**	**15**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Reynolds syndrome	779	**55**	**50**
RFT1-CDG	244310	**<10**	**<10**
Rhabdoid tumor	69077	**22**	**15**
Rhabdomyosarcoma	780	**137**	**131**
Rhabdomyosarcoma of the cervix uteri	213802	**<10**	**<10**
Rheumatic fever	3099	**82**	**79**
Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	85408	**1244**	**1237**
Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	85435	**587**	**570**
Rhizomelic chondrodysplasia punctata	177	**19**	**16**
Rhombencephalosynapsis	59315	**37**	**35**
RHYNS syndrome	140976	**<10**	**<10**
Riboflavin transporter deficiency	97229	**40**	**40**
Richieri Costa-Pereira syndrome	3102	**<10**	**<10**
Rieger anomaly	91483	**41**	**39**
Right aortic arch	99081	**118**	**117**
Right inferior vena cava connecting to left-sided atrium	99119	**<10**	**<10**
Right sided atrial isomerism	97548	**48**	**39**
Right superior vena cava connecting to left-sided atrium	99110	**<10**	**<10**
Rigid spine syndrome	97244	**56**	**53**
Ring chromosome 1 syndrome	1437	**<10**	**<10**
Ring chromosome 10 syndrome	1438	**<10**	**<10**
Ring chromosome 11 syndrome	96175	**<10**	**<10**
Ring chromosome 12 syndrome	1439	**<10**	**<10**
Ring chromosome 13 syndrome	96176	**10**	**10**
Ring chromosome 14 syndrome	1440	**15**	**14**
Ring chromosome 15 syndrome	96177	**<10**	**<10**
Ring chromosome 16 syndrome	96178	**<10**	**<10**
Ring chromosome 17 syndrome	1441	**<10**	**<10**
Ring chromosome 18 syndrome	1442	**30**	**28**
Ring chromosome 19 syndrome	1443	**<10**	**<10**
Ring chromosome 2 syndrome	96171	**<10**	**<10**
Ring chromosome 20 syndrome	1444	**52**	**51**
Ring chromosome 21 syndrome	1445	**20**	**20**
Ring chromosome 22 syndrome	1446	**28**	**26**
Ring chromosome 3 syndrome	96172	**<10**	**<10**
Ring chromosome 4 syndrome	1447	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ring chromosome 5 syndrome	251043	**<10**	**<10**
Ring chromosome 7 syndrome	1449	**<10**	**<10**
Ring chromosome 8 syndrome	1450	**12**	**12**
Ring chromosome 9 syndrome	96173	**<10**	**<10**
Ring chromosome Y syndrome	261529	**<10**	**<10**
Ring dermoid of cornea	91481	**33**	**33**
Rippling muscle disease	97238	**12**	**12**
Rippling muscle disease with myasthenia gravis	206575	**<10**	**<10**
Roberts syndrome	3103	**<10**	**<10**
Robin sequence-oligodactyly syndrome	3104	**15**	**15**
Robinow syndrome	97360	**74**	**72**
Roch-Leri mesosomatous lipomatosis	529	**19**	**19**
Roifman syndrome	353298	**17**	**17**
Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	163727	**<10**	**<10**
Rolandic epilepsy-speech dyspraxia syndrome	163721	**12**	**12**
Romano-Ward syndrome	101016	**402**	**392**
Rosaï-Dorfman disease	158014	**153**	**149**
Rosette-forming glioneuronal tumor	251975	**<10**	**<10**
Rothmund-Thomson syndrome	2909	**42**	**36**
Rotor syndrome	3111	**<10**	**<10**
Roussy-Lévy syndrome	3115	**<10**	**<10**
Rubella panencephalitis	83616	**<10**	**<10**
Rubinstein-Taybi syndrome	783	**468**	**454**
Saccular limited dorsal myeloschisis	645354	**<10**	**<10**
Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome	397927	**17**	**17**
S-adenosylhomocysteine hydrolase deficiency	88618	**11**	**10**
Saethre-Chotzen syndrome	794	**185**	**184**
Saldino-Mainzer syndrome	140969	**<10**	**<10**
Salivary gland type cancer of the breast	213557	**<10**	**<10**
SAMD9L-associated autoinflammatory syndrome	619367	**<10**	**<10**
Sandhoff disease	796	**43**	**15**
Sandifer syndrome	71272	**<10**	**<10**
Sanjad-Sakati syndrome	2323	**<10**	**<10**
SAPHO syndrome	793	**357**	**356**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Sarcocystosis	54368	**<10**	**<10**
Sarcoidosis	797	**13522**	**13096**
Sarcoma of the corpus uteri*	213620	**<10**	**<10**
SATB2-associated syndrome	576278	**68**	**68**
Satoyoshi syndrome	3130	**<10**	**<10**
SBDS-related severe neonatal spondylometaphyseal dysplasia	622934	**<10**	**<10**
SCALP syndrome	370052	**<10**	**<10**
Scalp-ear-nipple syndrome	2036	**<10**	**<10**
Scapuloperoneal spinal muscular atrophy	431255	**18**	**18**
Schaaf-Yang syndrome	398069	**31**	**29**
Schilbach-Rott syndrome	2353	**<10**	**<10**
Schimke immuno-osseous dysplasia	1830	**28**	**17**
Schinzel-Giedion syndrome	798	**27**	**15**
Schistosomiasis	1247	**16**	**15**
Schizencephaly	799	**115**	**112**
Schneckenbecken dysplasia	3144	**<10**	**<10**
Schnitzler syndrome	37748	**92**	**88**
Schnyder corneal dystrophy	98967	**11**	**11**
Schöpf-Schulz-Passarge syndrome	50944	**<10**	**<10**
Schuurs-Hoeijmakers syndrome	329224	**27**	**27**
Schwartz-Jampel syndrome	800	**21**	**21**
Scimitar syndrome	185	**190**	**187**
Scleredema	352763	**<10**	**<10**
Scleroderma*	801	**1538**	**1368**
Scleromyxedema	167635	**22**	**21**
Sclerosing cholangitis*	447771	**231**	**220**
Sclerosteosis	3152	**<10**	**<10**
Scott syndrome	806	**41**	**39**
Sebocystomatosis	841	**22**	**22**
Seborrhea-like dermatitis with psoriasiform elements	168606	**<10**	**<10**
Seckel syndrome	808	**40**	**36**
Second branchial cleft anomaly	141022	**380**	**380**
Secondary central precocious puberty in female	650082	**17**	**17**
Secondary central precocious puberty in male	650092	**<10**	**<10**
Secondary erythromelalgia	529864	**33**	**33**
Secondary hypereosinophilic syndrome	314962	**186**	**175**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Secondary hypoparathyroidism due to impaired parathormon secretion	140286	**430**	**418**
Secondary intestinal lymphangiectasia	90363	**<10**	**<10**
Secondary non-traumatic avascular necrosis	399180	**<10**	**<10**
Secondary pulmonary alveolar proteinosis	420259	**17**	**14**
Secondary pulmonary hemosiderosis	99930	**<10**	**<10**
Secondary sclerosing cholangitis	447774	**202**	**175**
Secondary short bowel syndrome	95427	**1722**	**1442**
Secondary syringomyelia	99857	**368**	**331**
Segmental odontomaxillary dysplasia	67039	**<10**	**<10**
Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	314662	**33**	**33**
Segmental spinal dysgenesis	656126	**<10**	**<10**
Seizures-scoliosis-macrocephaly syndrome	466926	**<10**	**<10**
Selective IgM deficiency	331235	**44**	**42**
Selective intrauterine growth restriction	617301	**235**	**227**
Self-healing papular mucinosis	90397	**<10**	**<10**
Self-improving collodion baby	281122	**13**	**13**
Self-limited childhood occipital epilepsy	25968	**253**	**251**
Self-limited epilepsy with centrotemporal spikes	1945	**865**	**863**
Self-limited infantile epilepsy	306	**114**	**114**
Self-limited neonatal epilepsy	1949	**134**	**134**
Self-limited neonatal-infantile epilepsy	140927	**64**	**63**
Semantic dementia	100069	**385**	**344**
Semicircular canal dehiscence syndrome	420402	**49**	**49**
Senior-Boichis syndrome	84081	**16**	**10**
Senior-Loken syndrome	3156	**93**	**85**
Sensorineural deafness with dilated cardiomyopathy	217622	**<10**	**<10**
Sensorineural hearing loss-spastic quadriplegia-intellectual disability syndrome	659975	**<10**	**<10**
Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	70595	**37**	**28**
Sepsis in premature infants	90051	**<10**	**<10**
Septate uterus*	180122	**50**	**50**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Septate vagina	180154	**51**	**51**
Septo-optic dysplasia spectrum	3157	**456**	**451**
Serine biosynthesis pathway deficiency, infantile/juvenile form	583595	**<10**	**<10**
SERKAL syndrome	139466	**<10**	**<10**
Serous cystadenocarcinoma of pancreas	424073	**174**	**170**
Serpiginous choroiditis	35686	**81**	**81**
Serrated polyposis syndrome	157798	**66**	**66**
SETD2-related microcephaly-severe intellectual disability-multiple congenital anomalies syndrome	597743	**<10**	**<10**
Severe achondroplasia-developmental delay-acanthosis nigricans syndrome	85165	**<10**	**<10**
Severe acute respiratory syndrome	140896	**26**	**23**
Severe autosomal recessive macrothrombocytopenia	438207	**<10**	**<10**
Severe combined immunodeficiency due to adenosine deaminase deficiency	277	**18**	**16**
Severe combined immunodeficiency due to complete RAG1/2 deficiency	331206	**17**	**16**
Severe combined immunodeficiency due to CORO1A deficiency	228003	**<10**	**<10**
Severe combined immunodeficiency due to DCLRE1C deficiency	275	**40**	**35**
Severe combined immunodeficiency due to DNA-PKcs deficiency	317425	**<10**	**<10**
Severe combined immunodeficiency due to FOXN1 deficiency	169095	**<10**	**<10**
Severe combined immunodeficiency due to LCK deficiency	280142	**<10**	**<10**
Severe combined immunodeficiency*	183660	**91**	**83**
Severe congenital nemaline myopathy	171430	**41**	**31**
Severe congenital neutropenia due to G6PC3 deficiency	331176	**<10**	**<10**
Severe congenital neutropenia due to JAGN1 deficiency	423384	**<10**	**<10**
Severe congenital neutropenia*	42738	**809**	**806**
Severe congenital neutropenia-developmental delay syndrome due to SRP54 deficiency	675767	**<10**	**<10**
Severe dermatitis-multiple allergies-metabolic wasting syndrome	369992	**10**	**10**
Severe disseminated cytomegalovirus infection in immunocompetent patients	35062	**<10**	**<10**
Severe early-childhood-onset retinal dystrophy	364055	**129**	**128**
Severe early-onset axonal neuropathy due to MFN2 deficiency	90118	**20**	**19**
Severe early-onset pulmonary alveolar proteinosis due to MARS deficiency	440427	**36**	**35**
Severe generalized junctional epidermolysis bullosa	79404	**17**	**<10**
Severe hereditary thrombophilia due to congenital protein C deficiency	745	**118**	**115**
Severe hereditary thrombophilia due to congenital protein S deficiency	743	**77**	**77**
Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome	467176	**<10**	**<10**
Severe intellectual disability and progressive spastic paraplegia	280763	**35**	**35**
Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome	466688	**<10**	**<10**
Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia	94066	**<10**	**<10**
Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	363686	**<10**	**<10**
Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	397933	**23**	**23**
Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome	391307	**<10**	**<10**
Severe lateral tibial bowing-short stature-mild winged scapula-mild facial dysmorphism syndrome	324307	**<10**	**<10**
Severe microbrachycephaly-intellectual disability-athetoid cerebral palsy syndrome	1236	**<10**	**<10**
Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome	369939	**<10**	**<10**
Severe myopia-generalized joint laxity-short stature syndrome	527450	**<10**	**<10**
Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	397593	**<10**	**<10**
Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract	500545	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Severe oculo-renal-cerebellar syndrome	2715	<10	<10
Severe primary trimethylaminuria	468726	<10	<10
Severe X-linked intellectual disability, Gustavson type	3078	<10	<10
Severe X-linked mitochondrial encephalomyopathy	238329	<10	<10
Sex cord-stromal tumor of testis	363489	<10	<10
Sézary syndrome	3162	<10	<10
Sheehan syndrome	91355	143	138
Sheldon-Hall syndrome	1147	21	21
Shigellosis	810	<10	<10
Shone complex	99063	211	201
Short bowel syndrome*	104008	919	826
Short chain acyl-CoA dehydrogenase deficiency	26792	154	148
Short rib-polydactyly syndrome*	1505	11	<10
Short rib-polydactyly syndrome, Beemer-Langer type	93268	<10	<10
Short rib-polydactyly syndrome, Majewski type	93269	<10	<10
Short rib-polydactyly syndrome, Saldino-Noonan type	93270	<10	<10
Short rib-polydactyly syndrome, Verma-Naumoff type	93271	<10	<10
Short stature due to GHSR deficiency	314811	<10	<10
Short stature due to partial GHR deficiency	314802	45	45
Short stature, Brussels type	2867	<10	<10
Short stature-advanced bone age-early-onset osteoarthritis syndrome	435804	42	42
Short stature-brachydactyly-obesity-global developmental delay syndrome	464288	<10	<10
Short stature-delayed bone age due to thyroid hormone metabolism deficiency	171706	<10	<10
Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome	314394	<10	<10
Short stature-optic atrophy-Pelger-Huët anomaly syndrome	391677	<10	<10
Short stature-pituitary and cerebellar defects-small sella turcica syndrome	85442	<10	<10
Short stature-skeletal dysplasia-retinal degeneration-intellectual disability-sensorineural hearing loss syndrome	589442	<10	<10
Short stature-valvular heart disease-characteristic facies syndrome	2868	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Short stature-webbed neck-heart disease syndrome	2865	<10	<10
SHORT syndrome	3163	25	25
Short-limb skeletal dysplasia with severe combined immunodeficiency	935	<10	<10
SHOX-related short stature	314795	234	233
Shprintzen-Goldberg syndrome	2462	50	47
Shwachman-Diamond syndrome	811	138	131
Sialidosis type 1	812	<10	<10
Sialidosis type 2	87876	<10	<10
Sialidosis*	309294	<10	<10
Sialuria	3166	<10	<10
Sickle cell anemia	232	14866	14576
Sickle cell disease due to hemoglobin S and a non-S/non-C hemoglobin variant	700085	69	69
Sickle cell S-C disease	251365	3706	3651
Sickle cell-beta-thalassemia disease syndrome	251359	1740	1711
Silent sinus syndrome	71276	<10	<10
Sillence syndrome	3168	<10	<10
Silver-Russell syndrome	813	766	760
SIM1-related Prader-Willi-like syndrome	398079	<10	<10
Simple cryoglobulinemia	91139	195	178
Simpson-Golabi-Behmel syndrome	373	82	77
Sinding-Larsen-Johansson disease	97337	10	10
Singleton-Merten dysplasia	85191	<10	<10
Sinoatrial node dysfunction and deafness	324321	<10	<10
Sitosterolemia	2882	24	23
Situs ambiguus	157769	18	17
Situs inversus totalis	101063	82	77
Sjögren-Larsson syndrome	816	57	53
Skeletal dysplasia-epilepsy-short stature syndrome	1858	<10	<10
Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome	508533	<10	<10
Skeletal Ewing sarcoma	319	89	86
Skin fragility-woolly hair-palmoplantar keratoderma syndrome	293165	<10	<10
SLC35A1-CDG	238459	<10	<10
SLC35A2-CDG	356961	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
SLC39A8-CDG	468699	**<10**	**<10**
SLC40A1-related hemochromatosis	647834	**<10**	**<10**
Sleep-related hypermotor epilepsy	98784	**164**	**159**
Slender bone dysplasia*	93440	**10**	**10**
Small bowel atresia	1201	**243**	**239**
Small cell carcinoma of the bladder	284400	**<10**	**<10**
Small cell carcinoma of the ovary	370396	**<10**	**<10**
Small cell lung cancer	70573	**77**	**45**
SMARCA2-related blepharophimosis-intellectual disability syndrome	637013	**<10**	**<10**
Smith-Lemli-Opitz syndrome	818	**99**	**86**
Smith-Magenis syndrome	819	**408**	**401**
Smith-McCort dysplasia	178355	**<10**	**<10**
Smoldering systemic mastocytosis	158775	**37**	**35**
Sneddon syndrome	820	**91**	**85**
Snowflake vitreoretinal degeneration	91496	**<10**	**<10**
Sodium-dependent multivitamin transporter deficiency	521268	**<10**	**<10**
Soft and hard cleft palate	664372	**132**	**132**
Soft tissue sarcoma*	3394	**12**	**11**
Solitary bone cyst	83468	**<10**	**<10**
Solitary fibrous tumor	2126	**<10**	**<10**
Solitary rectal ulcer syndrome	209964	**18**	**18**
Somatomammotropinoma	314769	**379**	**368**
Somatotropic adenoma*	96256	**54**	**52**
Sorsby pseudoinflammatory fundus dystrophy	59181	**<10**	**<10**
Sotos syndrome	821	**656**	**644**
Southeast Asian ovalocytosis	98868	**<10**	**<10**
Spasmus nutans	279882	**<10**	**<10**
Spastic ataxia with congenital miosis	1182	**<10**	**<10**
Spastic ataxia*	316226	**123**	**120**
Spastic ataxia-corneal dystrophy syndrome	2572	**<10**	**<10**
Spastic ataxia-dysarthria due to glutaminase deficiency	557056	**<10**	**<10**
Spastic paraparesis-cataracts-speech delay syndrome	615938	**<10**	**<10**
Spastic paraparesis-deafness syndrome	2815	**30**	**25**
Spastic paraplegia type 2	99015	**92**	**89**
Spastic paraplegia type 7	99013	**223**	**210**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Spastic paraplegia-facial-cutaneous lesions syndrome	2819	**<10**	**<10**
Spastic paraplegia-glaucoma-intellectual disability syndrome	2818	**<10**	**<10**
Spastic paraplegia-intellectual disability-nystagmus-obesity syndrome	521390	**<10**	**<10**
Spastic paraplegia-nephritis-deafness syndrome	2820	**<10**	**<10**
Spastic paraplegia-neuropathy-poikiloderma syndrome	2821	**<10**	**<10**
Spastic paraplegia-optic atrophy-neuropathy and spastic paraplegia-optic atrophy-neuropathy-related disorder*	431320	**<10**	**<10**
Spastic paraplegia-optic atrophy-neuropathy syndrome	320406	**<10**	**<10**
Spastic paraplegia-Paget disease of bone syndrome	329475	**<10**	**<10**
Spastic paraplegia-precocious puberty syndrome	2826	**<10**	**<10**
Spastic paraplegia-severe developmental delay-epilepsy syndrome	464282	**<10**	**<10**
Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome	3011	**<10**	**<10**
Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	447997	**<10**	**<10**
SPECC1L-related hypertelorism syndrome	1519	**33**	**30**
Specific language disorder*	211053	**72**	**72**
Specific learning disability*	211047	**2204**	**2196**
Spectrin-associated autosomal recessive cerebellar ataxia	352403	**<10**	**<10**
Spigelian hernia-cryptorchidism syndrome	314432	**<10**	**<10**
Spina bifida and other spinal dysraphisms*	823	**714**	**693**
Spina bifida-hypospadias syndrome	3176	**<10**	**<10**
Spinal arteriovenous metameric syndrome	53721	**31**	**29**
Spinal atrophy-ophthalmoplegia-pyramidal syndrome	1217	**<10**	**<10**
Spinal cord injury	90058	**54**	**49**
Spinal dermal sinus	645188	**26**	**26**
Spinal dysraphism with a posterior meningocele*	268744	**22**	**22**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Spinal muscular atrophy associated with central nervous system anomaly*	207012	<10	<10
Spinal muscular atrophy with respiratory distress type 1	98920	79	35
Spinal muscular atrophy with respiratory distress type 2	404521	43	40
Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	2590	<10	<10
Spindle cell hemangioma	210584	<10	<10
Spinocerebellar ataxia type 1	98755	118	105
Spinocerebellar ataxia type 10	98761	<10	<10
Spinocerebellar ataxia type 11	98767	<10	<10
Spinocerebellar ataxia type 13	98768	10	10
Spinocerebellar ataxia type 14	98763	51	51
Spinocerebellar ataxia type 15/16	98769	20	18
Spinocerebellar ataxia type 17	98759	16	14
Spinocerebellar ataxia type 19/22	98772	38	38
Spinocerebellar ataxia type 2	98756	171	152
Spinocerebellar ataxia type 20	101110	<10	<10
Spinocerebellar ataxia type 21	98773	21	21
Spinocerebellar ataxia type 23	101108	<10	<10
Spinocerebellar ataxia type 25	101111	<10	<10
Spinocerebellar ataxia type 26	101112	<10	<10
Spinocerebellar ataxia type 27A	98764	119	118
Spinocerebellar ataxia type 27B	675216	55	55
Spinocerebellar ataxia type 28	101109	37	35
Spinocerebellar ataxia type 29	208513	47	46
Spinocerebellar ataxia type 3	98757	298	282
Spinocerebellar ataxia type 30	211017	<10	<10
Spinocerebellar ataxia type 31	217012	<10	<10
Spinocerebellar ataxia type 32	276183	<10	<10
Spinocerebellar ataxia type 34	1955	<10	<10
Spinocerebellar ataxia type 36	276198	26	25
Spinocerebellar ataxia type 4	98765	<10	<10
Spinocerebellar ataxia type 41	458798	<10	<10
Spinocerebellar ataxia type 42	458803	14	12
Spinocerebellar ataxia type 43	497764	<10	<10
Spinocerebellar ataxia type 45	589527	<10	<10
Spinocerebellar ataxia type 48	631103	<10	<10
Spinocerebellar ataxia type 49	631106	<10	<10
Spinocerebellar ataxia type 5	98766	27	27
Spinocerebellar ataxia type 6	98758	53	52
Spinocerebellar ataxia type 7	94147	96	83
Spinocerebellar ataxia type 8	98760	<10	<10
Spinocerebellar ataxia with axonal neuropathy type 1	94124	16	13
Spinocerebellar ataxia with axonal neuropathy type 2	64753	72	71
Spinocerebellar ataxia with epilepsy	254881	21	20
Spinocerebellar ataxia-dysmorphism syndrome	1185	<10	<10
Spinocerebellar degeneration-corneal dystrophy syndrome	3177	<10	<10
Splenic marginal zone lymphoma	86854	<10	<10
Split cord malformation type I	1671	83	83
Split cord malformation type II	573253	11	11
Split cord malformation*	573278	<10	<10
Split cord malformation, composite type	633076	<10	<10
Split hand-split foot-deafness syndrome	71271	<10	<10
SPONASTRIME dysplasia	93357	12	11
Spondylocamptodactyly syndrome	3180	<10	<10
Spondylocarpotarsal synostosis	3275	28	28
Spondylodysplastic dysplasia*	93434	14	14
Spondylodysplastic Ehlers-Danlos syndrome	536471	19	19
Spondyloenchondrodysplasia	1855	<10	<10
Spondyloepimetaphyseal dysplasia congenita, Strudwick type	93346	37	37
Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type	93360	<10	<10
Spondyloepimetaphyseal dysplasia, aggrecan type	171866	11	11
Spondyloepimetaphyseal dysplasia, Geneviève type	168454	<10	<10
Spondyloepimetaphyseal dysplasia, Isidor-Toutain type	370015	<10	<10
Spondyloepimetaphyseal dysplasia, Maroteaux type	263482	<10	<10
Spondyloepimetaphyseal dysplasia, matrilin-3 type	156728	<10	<10
Spondyloepimetaphyseal dysplasia, Missouri type	93356	<10	<10
Spondyloepimetaphyseal dysplasia, PAPSS2 type	93282	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome	93358	**<10**	**<10**
Spondyloepiphyseal dysplasia and spondyloepimetaphyseal dysplasia*	253	**109**	**109**
Spondyloepiphyseal dysplasia congenita	94068	**153**	**150**
Spondyloepiphyseal dysplasia tarda	93284	**89**	**86**
Spondyloepiphyseal dysplasia with metatarsal shortening	137678	**<10**	**<10**
Spondyloepiphyseal dysplasia, Reardon type	163662	**<10**	**<10**
Spondyloepiphyseal dysplasia, Stanescu type	459051	**<10**	**<10**
Spondylo-megaepiphyseal-metaphyseal dysplasia	228387	**<10**	**<10**
Spondylometaphyseal dysplasia*	254	**48**	**48**
Spondylometaphyseal dysplasia, 'corner fracture' type	93315	**<10**	**<10**
Spondylometaphyseal dysplasia, Golden type	168544	**<10**	**<10**
Spondylometaphyseal dysplasia, Kozlowski type	93314	**24**	**23**
Spondylometaphyseal dysplasia, Schmidt type	93316	**31**	**31**
Spondylometaphyseal dysplasia, Sedaghatian type	93317	**<10**	**<10**
Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	85167	**<10**	**<10**
Spondylo-ocular syndrome	85194	**22**	**22**
Spondyloperipheral dysplasia-short ulna syndrome	1856	**<10**	**<10**
Spontaneous intestinal perforation	645793	**<10**	**<10**
Spontaneous intracranial hypotension	443180	**<10**	**<10**
Spontaneous periodic hypothermia	29822	**16**	**12**
Sporadic adult-onset ataxia of unknown etiology	247234	**191**	**179**
Sporadic Creutzfeldt-Jakob disease	204	**18**	**<10**
Sporadic fetal brain disruption sequence	1665	**<10**	**<10**
Sporadic pheochromocytoma/secreting paraganglioma	276621	**1592**	**1520**
Sprengel deformity	3181	**35**	**35**
Squamous cell carcinoma of head and neck	67037	**<10**	**<10**
Squamous cell carcinoma of the cervix uteri	213767	**<10**	**<10**
Squamous cell carcinoma of the colon	423994	**<10**	**<10**
Squamous cell carcinoma of the esophagus	99977	**<10**	**<10**
Squamous cell carcinoma of the lip	502366	**<10**	**<10**
Squamous cell carcinoma of the nasal cavity and paranasal sinuses	500464	**<10**	**<10**
Squamous cell carcinoma of the oral cavity	502363	**11**	**10**
Squamous cell carcinoma of the oropharynx	500478	**<10**	**<10**
Squamous cell carcinoma of the penis	398058	**<10**	**<10**
Squamous cell carcinoma of the rectum	424002	**<10**	**<10**
SRD5A3-CDG	324737	**<10**	**<10**
SSR4-CDG	370927	**<10**	**<10**
STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome	502434	**<10**	**<10**
Stapes ankylosis with broad thumbs and toes	140917	**<10**	**<10**
Staphylococcal necrotizing pneumonia	36238	**17**	**17**
Staphylococcal scalded skin syndrome	36236	**13**	**13**
Stargardt disease	827	**1779**	**1761**
Startle epilepsy	166427	**<10**	**<10**
STAT1-related autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome	391487	**<10**	**<10**
STAT3-related early-onset multisystem autoimmune disease	438159	**19**	**19**
Steatocystoma multiplex-natal teeth syndrome	3184	**<10**	**<10**
Steel syndrome	438117	**<10**	**<10**
Steinert myotonic dystrophy	273	**7117**	**5817**
Sterile multifocal osteomyelitis with periostitis and pustulosis	210115	**107**	**107**
Sternal cleft	2017	**<10**	**<10**
Steroid-responsive encephalopathy associated with autoimmune thyroiditis	83601	**22**	**21**
Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	95455	**650**	**547**
Stickler syndrome	828	**978**	**969**
Stiff person spectrum disorder	3198	**129**	**119**
Stiff skin syndrome	2833	**<10**	**<10**
STING-associated vasculopathy with onset in infancy	425120	**18**	**13**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Stormorken-Sjaastad-Langslet syndrome	3204	**16**	**15**
Straddling or overriding tricuspid valve	95461	**11**	**11**
Striate palmoplantar keratoderma	50942	**<10**	**<10**
Stromme syndrome	506307	**<10**	**<10**
STT3B-CDG	370924	**<10**	**<10**
Sturge-Weber syndrome	3205	**600**	**594**
Stüve-Wiedemann syndrome	3206	**23**	**21**
STXBP1-related encephalopathy	599373	**73**	**73**
Subacute cutaneous lupus erythematosus	163525	**285**	**273**
Subacute inflammatory demyelinating polyneuropathy	206594	**218**	**179**
Subacute sclerosing leukoencephalitis	2806	**11**	**10**
Subaortic stenosis-short stature syndrome	3191	**<10**	**<10**
Subcorneal pustular dermatosis	48377	**16**	**16**
Subcortical band heterotopia	99796	**102**	**101**
Subcutaneous panniculitis-like T-cell lymphoma	86884	**<10**	**<10**
Subependymal giant cell astrocytoma	251618	**<10**	**<10**
Submucosal cleft palate	155878	**144**	**143**
Succinic semialdehyde dehydrogenase deficiency	22	**57**	**55**
Succinyl-CoA:3-oxoacid CoA transferase deficiency	832	**16**	**16**
Sudden infant death-dysgenesis of the testes syndrome	168593	**<10**	**<10**
Sudden sensorineural hearing loss	90059	**<10**	**<10**
SUNCT syndrome	57145	**<10**	**<10**
Superficial epidermolytic ichthyosis	455	**16**	**15**
Superficial fibromatosis*	199257	**<10**	**<10**
Superficial pemphigus*	46485	**306**	**288**
Superficial siderosis	247245	**<10**	**<10**
Superior limbic keratoconjunctivitis	88633	**<10**	**<10**
Supernumerary breasts	180182	**<10**	**<10**
Supratip dysplasia	466695	**<10**	**<10**
Supravalvular aortic stenosis	3193	**146**	**141**
SURF1-related Charcot-Marie-Tooth disease type 4	391351	**<10**	**<10**
Susac syndrome	838	**122**	**121**
Susceptibility to respiratory infections associated with CD8alpha chain mutation	169085	**<10**	**<10**
Susceptibility to viral and mycobacterial infections due to STAT1 deficiency	391311	**46**	**43**
Sweet syndrome	3243	**60**	**57**
Sydenham chorea	306731	**17**	**17**
Symbrachydactyly of hands and feet	1570	**35**	**35**
Symmetrical thalamic calcifications	1314	**<10**	**<10**
Sympathetic ophthalmia	79098	**44**	**42**
Symphalangism with multiple anomalies of hands and feet	3246	**<10**	**<10**
Symptomatic form of Coffin-Lowry syndrome in female carriers	276630	**<10**	**<10**
Symptomatic form of HFE-related hemochromatosis	465508	**387**	**373**
Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers	206546	**271**	**260**
Symptomatic form of X-linked centronuclear myopathy in female carriers	604680	**<10**	**<10**
Syndactyly type 1	93402	**153**	**153**
Syndactyly type 2	93403	**63**	**61**
Syndactyly type 3	93404	**24**	**24**
Syndactyly type 4	93405	**19**	**19**
Syndactyly type 5	93406	**13**	**13**
Syndactyly type 6	295012	**10**	**10**
Syndactyly type 8	2498	**<10**	**<10**
Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome	357332	**<10**	**<10**
Syndactyly-nystagmus syndrome due to 2q31.1 microduplication	294026	**<10**	**<10**
Syndactyly-telecanthus-anogenital and renal malformations syndrome	140952	**<10**	**<10**
Syndromic diarrhea	84064	**56**	**49**
Syndromic microphthalmia type 5	178364	**<10**	**<10**
Syndromic orbital border hypoplasia	98606	**<10**	**<10**
Syndromic recessive X-linked ichthyosis	281090	**37**	**37**
Syndromic X-linked intellectual disability 7	85274	**<10**	**<10**
SYNGAP1-related developmental and epileptic encephalopathy	544254	**101**	**101**
Synovial sarcoma	3273	**<10**	**<10**
Syringocystadenoma papilliferum	840	**<10**	**<10**
Syringomyelia*	3280	**475**	**458**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Systemic capillary leak syndrome	188	**45**	**40**
Systemic diseases with anterior uveitis*	280926	**244**	**241**
Systemic diseases with panuveitis*	280933	**149**	**146**
Systemic Epstein-Barr virus-positive T-cell lymphoproliferative disease of childhood	364033	**<10**	**<10**
Systemic lupus erythematosus	536	**15145**	**14797**
Systemic mastocytosis with associated hematologic neoplasm	98849	**66**	**55**
Systemic mastocytosis*	2467	**949**	**919**
Systemic monochloroacetate poisoning	90069	**<10**	**<10**
Systemic primary carnitine deficiency	158	**112**	**107**
Systemic sclerosis	90291	**9699**	**8775**
Systemic-onset juvenile idiopathic arthritis	85414	**1540**	**1521**
T+ B+ severe combined immunodeficiency*	397802	**<10**	**<10**
TAFRO syndrome	457077	**13**	**13**
Takayasu arteritis	3287	**914**	**883**
Takenouchi-Kosaki syndrome	487796	**<10**	**<10**
Tako-Tsubo cardiomyopathy	66529	**237**	**210**
Tall stature-intellectual disability-renal anomalies syndrome	500095	**<10**	**<10**
Tall stature-long halluces-multiple extra-epiphyses syndrome	329191	**<10**	**<10**
Tangier disease	31150	**<10**	**<10**
TARP syndrome	2886	**15**	**15**
Tarsal-carpal coalition syndrome	1412	**<10**	**<10**
Tatton-Brown-Rahman syndrome	404443	**79**	**77**
Tay-Sachs disease	845	**78**	**36**
T-B- severe combined immunodeficiency*	317419	**<10**	**<10**
T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta	169160	**<10**	**<10**
T-B+ severe combined immunodeficiency due to gamma chain deficiency	276	**40**	**37**
T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	169154	**24**	**23**
T-B+ severe combined immunodeficiency due to JAK3 deficiency	35078	**31**	**30**
T-B+ severe combined immunodeficiency*	317416	**<10**	**<10**
TBCK-related encephalopathy-severe hypotonia-craniofacial dysmorphism syndrome	488632	**<10**	**<10**
T-cell immunodeficiency with epidermodysplasia verruciformis	324294	**<10**	**<10**
T-cell immunodeficiency with thymic aplasia	83471	**<10**	**<10**
T-cell large granular lymphocyte leukemia	86872	**52**	**47**
T-cell/histiocyte rich large B cell lymphoma	300857	**<10**	**<10**
Telecanthus-hypertelorism-strabismus-pes cavus syndrome	3293	**<10**	**<10**
Telethonin-related limb-girdle muscular dystrophy R7	34514	**<10**	**<10**
TELO2-related intellectual disability-neurodevelopmental disorder	488642	**<10**	**<10**
TEMPI syndrome	284227	**<10**	**<10**
Temple syndrome	254516	**63**	**63**
Temple-Baraitser syndrome	420561	**<10**	**<10**
Temtamy preaxial brachydactyly syndrome	363417	**<10**	**<10**
Temtamy syndrome	1777	**11**	**11**
Tenosynovial giant cell tumor	66627	**22**	**22**
Terminal myelocystocele	645337	**<10**	**<10**
Terminal osseous dysplasia-pigmentary defects syndrome	88630	**<10**	**<10**
Terrien marginal degeneration	519410	**11**	**11**
Tessier number 4 facial cleft	141258	**11**	**11**
Tessier number 5 facial cleft	141261	**<10**	**<10**
Tessier number 6 facial cleft	141265	**<10**	**<10**
Tessier number 7 facial cleft	141276	**80**	**80**
Testicular agenesis	325124	**203**	**203**
Testicular regression syndrome	983	**199**	**199**
Testicular seminomatous germ cell tumor	842	**<10**	**<10**
Tetraamelia-multiple malformations syndrome	3301	**<10**	**<10**
Tetragametic chimerism syndrome	199310	**<10**	**<10**
Tetralogy of Fallot	3303	**3665**	**3585**
Tetramelic monodactyly	2564	**<10**	**<10**
Tetrasomy 18p syndrome	3307	**53**	**53**
Tetrasomy 21 syndrome	96055	**<10**	**<10**
Tetrasomy 5p syndrome	3309	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Tetrasomy 9p syndrome	3310	25	25
Tetrasomy X syndrome	9	19	19
TFR2-related hemochromatosis	225123	35	34
Thakker-Donnai syndrome	1780	44	40
Thalidomide embryopathy	3312	<10	<10
Thanatophoric dysplasia	2655	31	15
Theca steroid-producing cell malignant tumor of ovary, not further specified	99917	<10	<10
Thiamine-responsive encephalopathy	199348	11	10
Thiamine-responsive megaloblastic anemia syndrome	49827	<10	<10
Thickened earlobes-conductive deafness syndrome	2405	<10	<10
Thiel-Behnke corneal dystrophy	98960	18	18
Thiemann disease, familial form	3314	<10	<10
Thinking seizures	166424	<10	<10
Third branchial cleft anomaly	141030	21	21
THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	363444	<10	<10
Thomas syndrome	3316	<10	<10
Thomsen and Becker disease	614	500	488
Thoracic dysplasia-hydrocephalus syndrome	1861	<10	<10
Thoracic outlet syndrome	97330	36	35
Thoraco-abdominal enteric duplication	1759	<10	<10
Thoracolaryngopelvic dysplasia	3317	<10	<10
Thoracomelic dysplasia	1803	<10	<10
Thrombocytopenia with congenital dyserythropoietic anemia	67044	<10	<10
Thrombocytopenia-absent radius syndrome	3320	60	57
Thrombomodulin-related bleeding disorder	436169	18	18
Thrombotic microangiopathy*	93573	1122	873
Thrombotic thrombocytopenic purpura	54057	1113	1055
Thumb deformity-alopecia-pigmentation anomaly syndrome	2251	<10	<10
Thygeson superficial punctate keratitis	519406	46	46
Thymic carcinoma	99868	14	10
Thymic neuroendocrine carcinoma	99869	<10	<10
Thymic neuroendocrine tumor	97289	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Thymic-renal-anal-lung dysplasia	3326	<10	<10
Thymoma	99867	105	91
Thyroid ectopia	95712	1253	1251
Thyroid hemiagenesis	95719	45	45
Thyroid hypoplasia	95720	76	76
Thyroid lymphoma	97285	<10	<10
Thyrotoxic periodic paralysis	79102	15	15
Tibial aplasia-ectrodactyly syndrome	3329	32	32
Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome	988	15	15
Tibial muscular dystrophy	609	89	82
Tick-borne encephalitis	297	<10	<10
Tietz syndrome	42665	<10	<10
Timothy syndrome	65283	19	18
Titin-related limb-girdle muscular dystrophy R10	140922	81	77
TMEM165-CDG	314667	<10	<10
TMEM199-CDG	466703	<10	<10
TMEM70-related mitochondrial encephalo-cardio-myopathy	1194	18	17
TMEM94-associated congenital heart defect-facial dysmorphism-developmental delay syndrome	562569	<10	<10
TNP03-related limb-girdle muscular dystrophy D2	55595	<10	<10
Tolosa-Hunt syndrome	64686	24	22
TOR1AIP1-related limb-girdle muscular dystrophy	424261	<10	<10
Toriello-Carey syndrome	3338	<10	<10
Torpedo Maculopathy	674935	<10	<10
Torsade-de-pointes syndrome with short coupling interval	51084	37	36
Torticollis-keloids-cryptorchidism-renal dysplasia syndrome	3341	<10	<10
Townes-Brocks syndrome	857	197	193
Toxic maculopathy due to antimalarial drugs	279894	54	54
Toxocariasis	3343	18	18
Tracheal agenesis	3346	14	10
Tracheobronchopathia osteochondroplastica	3348	17	15

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
TRAF7-associated heart defect-digital anomalies-facial dysmorphism-motor and speech delay syndrome	592570	**<10**	**<10**
Transaldolase deficiency	101028	**14**	**13**
Transcobalamin deficiency	859	**11**	**11**
Transgrediens et progrediens palmoplantar keratoderma	495	**15**	**15**
Transient congenital hypothyroidism*	178045	**63**	**63**
Transient erythroblastopenia of childhood	98871	**16**	**12**
Transient familial neonatal hyperbilirubinemia	2312	**22**	**22**
Transient hyperammonemia of the newborn	289877	**<10**	**<10**
Transient hypogammaglobulinemia of infancy	169139	**14**	**14**
Transient infantile hypertriglyceridemia and hepatosteatosis	300293	**<10**	**<10**
Transient myeloproliferative syndrome	420611	**110**	**103**
Transient neonatal diabetes mellitus	99886	**38**	**37**
Transient predisposition to invasive pyogenic bacterial infection	70592	**12**	**10**
Transient pseudohypoaldosteronism	93164	**11**	**11**
Transketolase deficiency	488618	**<10**	**<10**
Transplant-related bronchiolitis obliterans	658602	**39**	**39**
TRAPPC11-related limb-girdle muscular dystrophy R18	369840	**<10**	**<10**
Traumatic avascular necrosis	399175	**<10**	**<10**
Treacher-Collins syndrome	861	**377**	**369**
Trehalase deficiency	103909	**<10**	**<10**
Tremor-nystagmus-duodenal ulcer syndrome	3350	**<10**	**<10**
Triatrial heart*	1463	**42**	**42**
Trichinellosis	863	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Trichodental syndrome	3351	**<10**	**<10**
Tricho-dento-osseous syndrome	3352	**12**	**12**
Trichofolliculoma	864	**<10**	**<10**
Trichomegaly-retina pigmentary degeneration-dwarfism syndrome	3363	**<10**	**<10**
Trichoodontoonychial dysplasia	3355	**<10**	**<10**
Tricho-retino-dento-digital syndrome	1264	**<10**	**<10**
Trichorhinophalangeal syndrome type 1	77258	**85**	**85**
Trichorhinophalangeal syndrome type 2	502	**31**	**31**
Trichorhinophalangeal syndrome*	324764	**75**	**75**
Trichothiodystrophy	33364	**61**	**53**
Tricuspid atresia	1209	**352**	**324**
Trigeminal autonomic cephalalgia*	157843	**<10**	**<10**
Trigeminal neuralgia	221091	**17**	**17**
Trigeminal trophic syndrome	664901	**<10**	**<10**
Trigonocephaly-broad thumbs syndrome	3365	**<10**	**<10**
Trigonocephaly-short stature-developmental delay syndrome	3369	**<10**	**<10**
TRIM32-related limb-girdle muscular dystrophy R8	1878	**15**	**14**
Triose phosphate-isomerase deficiency	868	**12**	**<10**
Triphalangeal thumbs-brachyectrodactyly syndrome	2947	**<10**	**<10**
Triple A syndrome	869	**107**	**103**
Triploidy syndrome	3376	**21**	**20**
Trismus-pseudocamptodactyly syndrome	3377	**<10**	**<10**
Trisomy 10p syndrome	171929	**<10**	**<10**
Trisomy 12p syndrome	1699	**35**	**34**
Trisomy 13 syndrome	3378	**147**	**73**
Trisomy 17p syndrome	261290	**<10**	**<10**
Trisomy 18 syndrome	3380	**223**	**113**
Trisomy 18p syndrome	1715	**18**	**16**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Trisomy 1q syndrome	261344	**29**	**28**
Trisomy 20p syndrome	261318	**13**	**13**
Trisomy 4p syndrome	1738	**24**	**22**
Trisomy 5p syndrome	1742	**20**	**20**
Trisomy 8p syndrome	264450	**24**	**24**
Trisomy 8q syndrome	1752	**18**	**18**
Trisomy 9p syndrome	236	**61**	**58**
Trisomy X syndrome	3375	**553**	**545**
Tritanopia	88629	**<10**	**<10**
Tropical endomyocardial fibrosis	75565	**<10**	**<10**
Tropical pancreatitis	103918	**183**	**181**
Tropical spastic paraparesis	289326	**66**	**61**
True unicornuate uterus	180074	**<10**	**<10**
TSH-secreting pituitary adenoma	91347	**196**	**189**
Tuberculosis*	3389	**325**	**325**
Tuberous sclerosis complex	805	**3240**	**3155**
Tubular aggregate myopathy	2593	**30**	**27**
Tubulinopathy-associated dysgyria	467166	**42**	**41**
Tubulointerstitial nephritis and uveitis syndrome	91500	**218**	**211**
Tufted angioma	1063	**57**	**57**
Tularemia	3392	**<10**	**<10**
Tumor necrosis factor receptor 1 associated periodic syndrome	32960	**150**	**147**
Turner syndrome	881	**4807**	**4750**
Twin anemia-polycythemia sequence	617294	**13**	**13**
Twin to twin transfusion syndrome	95431	**98**	**91**
Twin-reversed arterial perfusion sequence	617297	**<10**	**<10**
Typical nemaline myopathy	171436	**32**	**27**
Tyrosinemia type 1	882	**168**	**162**
Tyrosinemia type 2	28378	**12**	**12**
Tyrosinemia type 3	69723	**<10**	**<10**
Uhl anomaly	3403	**<10**	**<10**
Ulbright-Hodes syndrome	3404	**<10**	**<10**
Ulerythema ophryogenesis	3406	**<10**	**<10**
Ullrich congenital muscular dystrophy	75840	**222**	**204**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ulnar-mammary syndrome	3138	**52**	**51**
Unclassified myelodysplastic syndrome	98827	**36**	**28**
Unclassified myelodysplastic/ myeloproliferative disease	98825	**<10**	**<10**
Unclassified vasculitis	251328	**639**	**579**
Uncombable hair syndrome	1410	**18**	**18**
Undetermined colitis	103920	**3917**	**3903**
Undifferentiated carcinoma of the corpus uteri	213721	**<10**	**<10**
Undifferentiated connective tissue syndrome	90002	**1184**	**1154**
Undifferentiated embryonal sarcoma of the liver	178315	**<10**	**<10**
Undifferentiated pleomorphic sarcoma	2023	**<10**	**<10**
Unexplained long-lasting fever/ inflammatory syndrome	251332	**187**	**181**
Unicervical bicornuate uterus	180114	**<10**	**<10**
Unilateral aplasia of the MÃ¼llerian ducts*	180071	**<10**	**<10**
Unilateral polymicrogyria	268943	**154**	**153**
Univentricular heart	1464	**855**	**797**
Unspecified juvenile idiopathic arthritis	91140	**802**	**799**
Unspecified mitochondrial disorder*	254837	**1794**	**1556**
Unstable hemoglobin disease	99139	**46**	**46**
Upington disease	3408	**<10**	**<10**
Upper limb hypertrophy	295049	**13**	**13**
Upper limb mesomelic dysplasia, type Fryns	2497	**<10**	**<10**
Upper tract urothelial carcinoma	598216	**<10**	**<10**
Urachal cyst	488	**<10**	**<10**
Urachal diverticulum	431347	**<10**	**<10**
Urachal sinus	431344	**<10**	**<10**
Urban-Rogers-Meyer syndrome	3409	**<10**	**<10**
Uremic pruritus	94059	**<10**	**<10**
Urofacial syndrome	2704	**18**	**18**
Usher syndrome	886	**1518**	**1505**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Uterine cervical aplasia and agenesis	180145	**<10**	**<10**
Uterine hypoplasia	180139	**<10**	**<10**
Uveal coloboma-cleft lip and palate-intellectual disability	1473	**<10**	**<10**
Uveal melanoma	39044	**10**	**<10**
VACTERL with hydrocephalus	3412	**57**	**56**
VACTERL/VATER association	887	**945**	**905**
Vacuolar myopathy with sarcoplasmic reticulum protein aggregates	88635	**<10**	**<10**
Vaginal atresia	65681	**33**	**33**
Van den Ende-Gupta syndrome	2460	**<10**	**<10**
Van der Woude syndrome	888	**309**	**309**
Variant ABeta2M amyloidosis	314652	**<10**	**<10**
Variant Creutzfeldt-Jakob disease	576370	**<10**	**<10**
Variegate porphyria	79473	**379**	**310**
Vascular Ehlers-Danlos syndrome	286	**1130**	**1080**
Vascular Ehlers-Danlos-polymicrogyria syndrome	636941	**<10**	**<10**
Vasoproliferative tumor of the retina	353356	**41**	**39**
Vein of Galen aneurysmal malformation	1053	**131**	**112**
Velo-facial-skeletal syndrome	3424	**<10**	**<10**
Ventilator-induced diaphragmatic dysfunction	505395	**<10**	**<10**
Ventricular extrasystoles with syncopal episodes-perodactyly-Robin sequence syndrome	3201	**<10**	**<10**
Ventriculomegaly-cystic kidney disease	443988	**<10**	**<10**
Verloove Vanhorick-Brubakk syndrome	3429	**<10**	**<10**
Vernal keratoconjunctivitis	70476	**350**	**348**
Verrucous hemangioma	464318	**45**	**45**
Very long chain acyl-CoA dehydrogenase deficiency	26793	**98**	**88**
VEXAS syndrome	596753	**349**	**282**
Vibratory urticaria	493342	**<10**	**<10**
Vici syndrome	1493	**<10**	**<10**
VIPoma	97282	**<10**	**<10**
Viral myositis	206991	**123**	**121**
Visual snow syndrome	420556	**<10**	**<10**
Vitamin B12-responsive methylmalonic acidemia	28	**69**	**64**
Vitamin B12-unresponsive methylmalonic acidemia	27	**107**	**87**
Vitamin K antagonist embryofetopathy	1914	**<10**	**<10**
Vocal cord and pharyngeal distal myopathy	600	**<10**	**<10**
Vogt-Koyanagi-Harada disease	3437	**282**	**277**
Von Hippel-Lindau disease	892	**628**	**593**
Von Voss-Cherstvoy syndrome	3439	**<10**	**<10**
Von Willebrand disease	903	**11905**	**11760**
Vulvar carcinoma	494418	**<10**	**<10**
Vulvar intraepithelial neoplasia	137583	**<10**	**<10**
Vulvovaginal rhabdomyosarcoma	206492	**<10**	**<10**
Waardenburg syndrome	3440	**590**	**586**
Waardenburg-Shah syndrome	897	**58**	**56**
WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	466943	**47**	**47**
Wagner disease	898	**27**	**27**
WAGR syndrome	893	**68**	**60**
Waldenström macroglobulinemia	33226	**189**	**140**
Walker-Warburg syndrome	899	**28**	**17**
WARS2-related combined oxidative phosphorylation defect	572798	**<10**	**<10**
Warts-immunodeficiency-lymphedema-anogenital dysplasia syndrome	568056	**<10**	**<10**
Weaver syndrome	3447	**43**	**43**
Weill-Marchesani syndrome	3449	**30**	**30**
Weiss-Kruszka Syndrome	502430	**<10**	**<10**
Well-differentiated fetal adenocarcinoma of the lung	284395	**<10**	**<10**
Wells syndrome	901	**10**	**10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Werner syndrome	902	**25**	**20**
WHIM syndrome	51636	**15**	**14**
Whipple disease	3452	**121**	**117**
White forelock with malformations	2475	**<10**	**<10**
White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome	3207	**12**	**11**
White sponge nevus	171723	**<10**	**<10**
White-Sutton syndrome	468678	**80**	**79**
Whooping cough	1489	**<10**	**<10**
Wieacker-Wolff syndrome	3454	**25**	**23**
Wiedemann-Rautenstrauch syndrome	3455	**<10**	**<10**
Wiedemann-Steiner syndrome	319182	**193**	**192**
Wild type ABeta2M amyloidosis	85446	**<10**	**<10**
Wild type ATTR amyloidosis	330001	**4654**	**3587**
Wildervanck syndrome	3456	**10**	**10**
Williams syndrome	904	**1227**	**1196**
Williams-Campbell syndrome	411501	**<10**	**<10**
Wilson disease	905	**1328**	**1272**
Wilson-Turner syndrome	3459	**<10**	**<10**
Wiskott-Aldrich syndrome	906	**230**	**227**
Witteveen-Kolk syndrome	500163	**54**	**54**
Wolcott-Rallison syndrome	1667	**11**	**<10**
Wolf-Hirschhorn syndrome	280	**130**	**119**
Wolfram syndrome	3463	**317**	**289**
Wolfram-like syndrome	411590	**65**	**65**
Woodhouse-Sakati syndrome	3464	**37**	**37**
Woolly hair	170	**<10**	**<10**
Woolly hair nevus	79414	**<10**	**<10**
Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia	166277	**<10**	**<10**
Wound myiasis	165955	**<10**	**<10**
X small rings syndrome	96201	**<10**	**<10**
Xanthoma disseminatum	158003	**<10**	**<10**
Xeroderma pigmentosum	910	**142**	**129**
Xeroderma pigmentosum variant	90342	**10**	**<10**
Xeroderma pigmentosum-Cockayne syndrome complex	220295	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked acrogigantism	300373	**55**	**53**
X-linked adrenal hypoplasia congenita	95702	**53**	**51**
X-linked adrenoleukodystrophy	43	**569**	**509**
X-linked alpha-thalassemia-intellectual disability syndrome	847	**59**	**55**
X-linked calvarial hyperostosis	391327	**<10**	**<10**
X-linked central congenital hypothyroidism with late-onset testicular enlargement	329235	**<10**	**<10**
X-linked centronuclear myopathy	596	**79**	**47**
X-linked Charcot-Marie-Tooth disease type 1	101075	**202**	**200**
X-linked Charcot-Marie-Tooth disease type 2	101076	**36**	**32**
X-linked Charcot-Marie-Tooth disease type 3	101077	**<10**	**<10**
X-linked Charcot-Marie-Tooth disease type 4	101078	**11**	**11**
X-linked Charcot-Marie-Tooth disease type 5	99014	**<10**	**<10**
X-linked Charcot-Marie-Tooth disease*	64747	**356**	**346**
X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome	431140	**<10**	**<10**
X-linked cone dysfunction syndrome with myopia	90001	**17**	**17**
X-linked corneal dermoid	1661	**<10**	**<10**
X-linked creatine transporter deficiency	52503	**118**	**118**
X-linked distal spinal muscular atrophy type 3	139557	**<10**	**<10**
X-linked dominant chondrodysplasia punctata	35173	**63**	**60**
X-linked dominant chondrodysplasia, Chassaing-Lacombe type	163966	**<10**	**<10**
X-linked dyserythropoietic anemia with abnormal platelets and neutropenia	363727	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked Ehlers-Danlos syndrome	75497	**25**	**25**
X-linked epilepsy-learning disabilities-behavior disorders syndrome	85294	**23**	**23**
X-linked erythropoietic protoporphyria	443197	**17**	**15**
X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	480880	**<10**	**<10**
X-linked hereditary sensory and autonomic neuropathy with deafness	139583	**<10**	**<10**
X-linked hypophosphatemia	89936	**645**	**641**
X-linked ichthyosis syndrome*	281210	**<10**	**<10**
X-linked immunoneurologic disorder	2571	**<10**	**<10**
X-linked intellectual disability due to GRIA3 mutations	364028	**14**	**14**
X-linked intellectual disability, Cabezas type	85293	**11**	**11**
X-linked intellectual disability, Cantagrel type	85277	**<10**	**<10**
X-linked intellectual disability, Cilliers type	163971	**<10**	**<10**
X-linked intellectual disability, Hedera type	93952	**<10**	**<10**
X-linked intellectual disability, Najm type	163937	**23**	**22**
X-linked intellectual disability, Nascimento type	163956	**10**	**<10**
X-linked intellectual disability, Shashi type	85286	**<10**	**<10**
X-linked intellectual disability, Siderius type	85287	**<10**	**<10**
X-linked intellectual disability, Snyder type	3063	**16**	**15**
X-linked intellectual disability, Van Esch type	163976	**<10**	**<10**
X-linked intellectual disability-acromegaly-hyperactivity syndrome	85327	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked intellectual disability-ataxia-apraxia syndrome	85338	**<10**	**<10**
X-linked intellectual disability-cerebellar hypoplasia syndrome	137831	**32**	**32**
X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome	459070	**<10**	**<10**
X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	1568	**<10**	**<10**
X-linked intellectual disability-epilepsy syndrome*	2076	**36**	**35**
X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	480907	**<10**	**<10**
X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome	85317	**<10**	**<10**
X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome	3055	**<10**	**<10**
X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	85329	**<10**	**<10**
X-linked intellectual disability-hypotonia-movement disorder syndrome	457260	**25**	**25**
X-linked intellectual disability-macrocephaly-macroorchidism syndrome	85320	**<10**	**<10**
X-linked intellectual disability-plagiocephaly syndrome	2898	**15**	**15**
X-linked intellectual disability-psychosis-macroorchidism syndrome	3077	**<10**	**<10**
X-linked intellectual disability-retinitis pigmentosa syndrome	85332	**<10**	**<10**
X-linked intellectual disability-short stature-overweight syndrome	457240	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked lissencephaly with abnormal genitalia	452	**<10**	**<10**
X-linked lymphoproliferative disease due to SAP deficiency	538931	**20**	**20**
X-linked lymphoproliferative disease due to XIAP deficiency	538934	**39**	**39**
X-linked lymphoproliferative disease*	2442	**22**	**21**
X-linked mandibulofacial dysostosis	1131	**<10**	**<10**
X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	435938	**<10**	**<10**
X-linked myopathy with excessive autophagy	25980	**55**	**48**
X-linked myopathy with postural muscle atrophy	178461	**<10**	**<10**
X-linked myotubular myopathy-abnormal genitalia syndrome	456328	**<10**	**<10**
X-linked non progressive cerebellar ataxia	314978	**<10**	**<10**
X-linked osteoporosis with fractures	391330	**15**	**14**
X-linked progressive cerebellar ataxia	1175	**<10**	**<10**
X-linked recessive ocular albinism	54	**165**	**162**
X-linked reticulate pigmentary disorder	85453	**<10**	**<10**
X-linked retinal dysplasia	1852	**11**	**11**
X-linked retinoschisis	792	**493**	**490**
X-linked scapuloperoneal muscular dystrophy	431272	**<10**	**<10**
X-linked severe congenital neutropenia	86788	**<10**	**<10**
X-linked severe syndromic thoracic aortic aneurysm and dissection	622925	**<10**	**<10**
X-linked sideroblastic anemia	75563	**13**	**13**
X-linked sideroblastic anemia and spinocerebellar ataxia	2802	**<10**	**<10**
X-linked skeletal dysplasia-intellectual disability syndrome	1436	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked spasticity-intellectual disability-epilepsy syndrome	3175	**<10**	**<10**
X-linked spinocerebellar ataxia type 3	85297	**<10**	**<10**
X-linked spinocerebellar ataxia type 4	85292	**<10**	**<10**
X-linked spondyloepimetaphyseal dysplasia	93349	**<10**	**<10**
XMEN	317476	**10**	**10**
Xp21 deletion syndrome	261476	**15**	**15**
Xp22.13p22.2 duplication syndrome	284180	**21**	**21**
Xp22.3 microdeletion syndrome	1643	**47**	**46**
Xq12-q13.3 duplication syndrome	314389	**<10**	**<10**
Xq21 microdeletion syndrome	1435	**<10**	**<10**
Xq25 microduplication syndrome	521258	**16**	**16**
Xq27.3q28 duplication syndrome	261483	**18**	**17**
XY type gonadal dysgenesis-associated anomalies syndrome	1770	**28**	**28**
Yolk sac tumor	876	**<10**	**<10**
Young adult-onset distal hereditary motor neuropathy	314485	**18**	**17**
Young syndrome	3471	**<10**	**<10**
Young-onset Parkinson disease	2828	**224**	**198**
Yunis-Varon syndrome	3472	**<10**	**<10**
Zellweger syndrome	912	**57**	**18**
Zellweger-like syndrome without peroxisomal anomalies	50812	**<10**	**<10**
Zika virus disease	448237	**<10**	**<10**
Zimmermann-Laband syndrome	3473	**11**	**<10**
Zollinger-Ellison syndrome	913	**<10**	**<10**
Zygomycosis	73263	**<10**	**<10**

BNDMR
Banque Nationale de Données
Maladies Rares

NUMBER OF CASES PER OBSOLETE AND DEPRECIATED RARE DISEASE REGISTERED IN THE BNDMR

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : 3q26q27 microdeletion syndrome	356947	**<10**	**<10**
DEPRECATED : 3q27.3 microdeletion syndrome	397695	**<10**	**<10**
DEPRECATED : ABCD syndrome	918	**<10**	**<10**
DEPRECATED : Achalasia-alacrimia syndrome	99777	**<10**	**<10**
DEPRECATED : Acromegaloid facial appearance syndrome	965	**<10**	**<10**
DEPRECATED : Acromegaly-cutis verticis gyrata-corneal leukoma syndrome	964	**<10**	**<10**
DEPRECATED : Acute biphenotypic leukemia	98837	**<10**	**<10**
DEPRECATED : Albright hereditary osteodystrophy	665	**13**	**13**
DEPRECATED : Alopecia-hypogonadism-extrapyramidal syndrome	1011	**<10**	**<10**
DEPRECATED : Amelogenesis imperfecta-gingival hyperplasia syndrome	171836	**<10**	**<10**
DEPRECATED : Amish infantile epilepsy syndrome	171714	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : Anophthalmia-heart and pulmonary anomalies-intellectual disability syndrome	91129	**<10**	**<10**
DEPRECATED : Anophthalmia-hypothalamo-pituitary insufficiency syndrome	1102	**<10**	**<10**
DEPRECATED : Aortic dilatation-joint hypermobility-arterial tortuosity syndrome	88636	**<10**	**<10**
DEPRECATED : Arrhinia-choanal atresia-microphthalmia syndrome	1135	**<10**	**<10**
DEPRECATED : Aurocephalosyndactyly	1219	**<10**	**<10**
DEPRECATED : Autism-facial port-wine stain syndrome	137911	**<10**	**<10**
DEPRECATED : Autosomal dominant Charcot-Marie-Tooth disease type 2G	99941	**<10**	**<10**
DEPRECATED : Autosomal dominant chorioretinopathy-microcephaly syndrome	1432	**<10**	**<10**
DEPRECATED : Autosomal dominant limb-girdle muscular dystrophy type 1A	266	**50**	**45**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : Autosomal dominant limb-girdle muscular dystrophy type 1B	264	**100**	**82**
DEPRECATED : Autosomal dominant limb-girdle muscular dystrophy type 1C	265	**54**	**53**
DEPRECATED : Autosomal dominant limb-girdle muscular dystrophy type 1E	34517	**<10**	**<10**
DEPRECATED : Autosomal dominant optic atrophy and congenital deafness	3212	**<10**	**<10**
DEPRECATED : Autosomal recessive amelia	1027	**<10**	**<10**
DEPRECATED : Autosomal recessive carpotarsal osteolysis	2775	**<10**	**<10**
DEPRECATED : Autosomal recessive limb-girdle muscular dystrophy type 2R	363543	**<10**	**<10**
DEPRECATED : Ballard syndrome	93395	**<10**	**<10**
DEPRECATED : Benign idiopathic neonatal seizures	64545	**42**	**41**
DEPRECATED : Benign non-familial infantile seizures	166295	**10**	**10**
DEPRECATED : Benign partial epilepsy of infancy with complex partial seizures	166299	**58**	**58**
DEPRECATED : Benign partial epilepsy with secondarily generalized seizures in infancy	166302	**30**	**30**
DEPRECATED : Biliary tract malformation-renal failure syndrome	3438	**<10**	**<10**
DEPRECATED : Blepharofacioskeletal syndrome	1251	**<10**	**<10**
DEPRECATED : Blepharophimosis-intellectual disability syndrome due to UBE3B deficiency	329255	**<10**	**<10**
DEPRECATED : Bowen syndrome	1271	**<10**	**<10**
DEPRECATED : Braddock-Carey syndrome	3323	**11**	**11**
DEPRECATED : Bronchiectasis-oligospermia syndrome	1301	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : C3 deposition glomerulonephritis without proliferation	93559	**18**	**17**
DEPRECATED : CAMFAK syndrome	1317	**<10**	**<10**
DEPRECATED : Cavernous hemangiomas of face-supraumbilical midline raphe syndrome	2124	**43**	**43**
DEPRECATED : Central bilateral macrogyria	2431	**<10**	**<10**
DEPRECATED : Cerebral gigantism-jaw cysts syndrome	2081	**<10**	**<10**
DEPRECATED : Cerebroretinal vasculopathy	3421	**11**	**10**
DEPRECATED : Cheirospondyloenchondromatosis	99647	**<10**	**<10**
DEPRECATED : Chudley-Lowry-Hoar syndrome	93971	**<10**	**<10**
DEPRECATED : Combined cervical dystonia	370114	**109**	**105**
DEPRECATED : Common hereditary elliptocytosis	98864	**<10**	**<10**
DEPRECATED : Congenital and infantile nephrotic syndrome	97556	**53**	**46**
DEPRECATED : Congenital hereditary endothelial dystrophy type I	98975	**13**	**13**
DEPRECATED : Congenital hypothalamic hamartoma syndrome	2113	**13**	**13**
DEPRECATED : Congenital muscular dystrophy type 1C	52428	**<10**	**<10**
DEPRECATED : Connective tissue dysplasia, Spellacy type	3333	**<10**	**<10**
DEPRECATED : Corpus callosum dysgenesis-hypopituitarism syndrome	93943	**<10**	**<10**
DEPRECATED : Craniosynostosis-dysmorphism-brachydactyly syndrome	1535	**10**	**10**
DEPRECATED : Cutis verticis gyrata-intellectual disability syndrome	1557	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : Cystic hygroma	79486	**15**	**15**
DEPRECATED : Deafness-lymphedema-leukemia syndrome	3226	**<10**	**<10**
DEPRECATED : Deafness-opticoacoustic nerve atrophy-dementia syndrome	3213	**<10**	**<10**
DEPRECATED : Dincsoy-Salih-Patel syndrome	1678	**<10**	**<10**
DEPRECATED : Distal myopathy with early respiratory muscle involvement	34521	**<10**	**<10**
DEPRECATED : Dyssegmental dysplasia-glaucoma syndrome	1804	**<10**	**<10**
DEPRECATED : Dystonia 14	101151	**11**	**11**
DEPRECATED : Early myoclonic encephalopathy	1935	**63**	**51**
DEPRECATED : Early-onset familial noncirrhotic portal hypertension	494348	**<10**	**<10**
DEPRECATED : Early-onset sarcoidosis	90341	**22**	**22**
DEPRECATED : Ectrodactyly-cleft palate syndrome	1889	**<10**	**<10**
DEPRECATED : Ectrodactyly-ectodermal dysplasia without clefting syndrome	1888	**<10**	**<10**
DEPRECATED : Ehlers-Danlos syndrome with periventricular heterotopia	82004	**37**	**37**
DEPRECATED : Ependymoblastoma	251880	**<10**	**<10**
DEPRECATED : Familial mesial temporal lobe epilepsy with febrile seizures	165805	**<10**	**<10**
DEPRECATED : Familial multiple fibrofolliculoma	338	**<10**	**<10**
DEPRECATED : Familial progressive vestibulocochlear dysfunction	1767	**<10**	**<10**
DEPRECATED : Fanconi syndrome-ichthyosis-dysmorphism syndrome	1981	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : Frontotemporal dementia, right temporal atrophy variant	293848	**14**	**12**
DEPRECATED : Furlong syndrome	97295	**<10**	**<10**
DEPRECATED : Generalized resistance to thyroid hormone	3221	**416**	**393**
DEPRECATED : Goldenhar syndrome	374	**745**	**734**
DEPRECATED : Granulomatous arthritis of childhood	3274	**27**	**26**
DEPRECATED : Growth deficiency-brachydactyly-dysmorphism syndrome	2055	**<10**	**<10**
DEPRECATED : Hair defect-photosensitivity-intellectual disability syndrome	1408	**<10**	**<10**
DEPRECATED : Hereditary palmoplantar keratoderma, Gamborg-Nielsen type	86923	**<10**	**<10**
DEPRECATED : Hereditary pyropoikilocytosis	98867	**<10**	**<10**
DEPRECATED : Hereditary site-specific ovarian cancer syndrome	213524	**28**	**17**
DEPRECATED : Hereditary vascular retinopathy	71291	**<10**	**<10**
DEPRECATED : Homozygous hereditary elliptocytosis	98865	**<10**	**<10**
DEPRECATED : Hyperlipoproteinemia type 1	411	**11**	**11**
DEPRECATED : Hypopituitarism-micropenis-cleft lip/palate syndrome	2243	**<10**	**<10**
DEPRECATED : Hypoplastic tibiae-postaxial polydactyly syndrome	3332	**<10**	**<10**
DEPRECATED : Ichthyosis hystrix gravior	79504	**<10**	**<10**
DEPRECATED : Infant acute respiratory distress syndrome	70587	**55**	**48**
DEPRECATED : Intermittent neutropenia	2689	**78**	**78**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : Inverse Klippel-Trenaunay syndrome	329324	**20**	**20**
DEPRECATED : Ischio-vertebral syndrome	85200	**<10**	**<10**
DEPRECATED : Juberg-Marsidi syndrome	93972	**<10**	**<10**
DEPRECATED : LAMB2-related infantile-onset nephrotic syndrome	306507	**<10**	**<10**
DEPRECATED : Leigh syndrome with cardiomyopathy	70474	**<10**	**<10**
DEPRECATED : Leigh syndrome with leukodystrophy	255241	**17**	**13**
DEPRECATED : Ligneous conjunctivitis	97231	**<10**	**<10**
DEPRECATED : Lipoatrophy with diabetes, leukomelanodermic papules, liver steatosis, and hypertrophic cardiomyopathy	156156	**<10**	**<10**
DEPRECATED : Lipomyelomeningocele	268835	**151**	**149**
DEPRECATED : Long chain acyl-CoA dehydrogenase deficiency	99900	**<10**	**<10**
DEPRECATED : Lymphedema-ptosis syndrome	2419	**<10**	**<10**
DEPRECATED : Male infertility with normal virilization due to meiosis defect	217034	**13**	**13**
DEPRECATED : Malpuech syndrome	2453	**<10**	**<10**
DEPRECATED : Mediterranean macrothrombocytopenia	101022	**12**	**12**
DEPRECATED : Medulloepithelioma of the central nervous system	251883	**<10**	**<10**
DEPRECATED : Meningocele	93968	**91**	**90**
DEPRECATED : Metaphyseal acroscyphodysplasia	1240	**<10**	**<10**
DEPRECATED : Microcephaly-digital anomalies-intellectual disability syndrome	137653	**<10**	**<10**
DEPRECATED : Microcephaly-intellectual disability-phalangeal and neurological anomalies syndrome	137658	**<10**	**<10**
DEPRECATED : Mirhosseini-Holmes-Walton syndrome	3084	**<10**	**<10**
DEPRECATED : Muir-Torre syndrome	587	**129**	**120**
DEPRECATED : Multiple epiphyseal dysplasia, Beighton type	166011	**<10**	**<10**
DEPRECATED : Multiple non-ossifying fibromatosis	2029	**<10**	**<10**
DEPRECATED : Myoclonic dystonia 15	210566	**<10**	**<10**
DEPRECATED : NAME syndrome	623	**<10**	**<10**
DEPRECATED : Neonatal neutropenia	37629	**19**	**19**
DEPRECATED : Neutropenia-hyperlymphocytosis with large granular lymphocytes syndrome	2687	**<10**	**<10**
DEPRECATED : Non-hereditary congenital primary lymphedema	79450	**188**	**180**
DEPRECATED : Non-hereditary late-onset primary lymphedema	90185	**1580**	**1531**
DEPRECATED : Normokalemic periodic paralysis	680	**<10**	**<10**
DEPRECATED : Ocular albinism with congenital sensorineural deafness	352740	**<10**	**<10**
DEPRECATED : Onycho-tricho-dysplasia-neutropenia syndrome	2739	**<10**	**<10**
DEPRECATED : Otomandibular syndrome	141136	**553**	**549**
DEPRECATED : Palmoplantar porokeratosis of Mantoux	736	**<10**	**<10**
DEPRECATED : Paroxysmal hypnogenic dyskinesia	98812	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : Patella aplasia-coxa vara-tarsal synostosis syndrome	3112	**<10**	**<10**
DEPRECATED : PIBIDS syndrome	670	**<10**	**<10**
DEPRECATED : Pinsky-Di George-Harley syndrome	2895	**13**	**12**
DEPRECATED : Pipecolic acidemia	34	**<10**	**<10**
DEPRECATED : Pitt-Rogers-Danks syndrome	98788	**<10**	**<10**
DEPRECATED : Pituitary resistance to thyroid hormone	165994	**16**	**16**
DEPRECATED : Polyneuropathy associated with IgM monoclonal gammopathy with anti-MAG	639	**1415**	**1223**
DEPRECATED : Pontocerebellar hypoplasia type 5	166068	**<10**	**<10**
DEPRECATED : Premature chromosome condensation with microcephaly and intellectual disability	52183	**<10**	**0**
DEPRECATED : Proximal tubulopathy-diabetes mellitus-cerebellar ataxia syndrome	3390	**<10**	**<10**
DEPRECATED : Pseudo-Zellweger syndrome	2981	**<10**	**0**
DEPRECATED : Rapp-Hodgkin syndrome	3022	**<10**	**<10**
DEPRECATED : Renal-genital-middle ear anomalies	1092	**<10**	**<10**
DEPRECATED : Reunion Island Larsen-like syndrome	294049	**11**	**<10**
DEPRECATED : Robinow-like syndrome	3105	**<10**	**<10**
DEPRECATED : Sacral hemangiomas-multiple congenital abnormalities syndrome	2125	**<10**	**<10**
DEPRECATED : Sacrococcygeal dysgenesis association	1773	**<10**	**<10**
DEPRECATED : Salt-and-pepper syndrome	370938	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : Serpentine fibula-polycystic kidneys syndrome	2853	**<10**	**<10**
DEPRECATED : Severe dilated cardiomyopathy due to lamin A/C mutation	83618	**41**	**38**
DEPRECATED : Short stature-intellectual disability-eye anomalies-cleft lip/palate syndrome	2649	**<10**	**<10**
DEPRECATED : Shoulder and thorax deformity-congenital heart disease syndrome	1940	**<10**	**<10**
DEPRECATED : Simpson-Golabi-Behmel syndrome type 2	79022	**<10**	**<10**
DEPRECATED : Spastic paraplegia-epilepsy-intellectual disability syndrome	2816	**<10**	**<10**
DEPRECATED : Spinocerebellar ataxia type 16	98770	**<10**	**<10**
DEPRECATED : Spinocerebellar ataxia type 22	101107	**<10**	**<10**
DEPRECATED : Sporadic idiopathic steroid-resistant nephrotic syndrome	84271	**529**	**500**
DEPRECATED : Squamous cell carcinoma of the oral tongue	457252	**<10**	**<10**
DEPRECATED : Taurodontia-absent teeth-sparse hair syndrome	2731	**<10**	**<10**
DEPRECATED : Thost-Unna palmoplantar keratoderma	496	**<10**	**<10**
DEPRECATED : Triphalangeal thumb-polysyndactyly syndrome	2950	**<10**	**<10**
DEPRECATED : Vascular-like classical Ehlers-Danlos syndrome	230845	**<10**	**<10**
DEPRECATED : Weissenbacher-Zweymuller syndrome	3450	**48**	**48**
DEPRECATED : West syndrome	3451	**2162**	**2064**
DEPRECATED : Woolly hair-hypotrichosis-everted lower lip-outstanding ears syndrome	1409	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DEPRECATED : X-linked intellectual disability, Brooks type	3056	**<10**	**<10**
DEPRECATED : X-linked intellectual disability, Raymond type	163953	**<10**	**<10**
DEPRECATED : X-linked intellectual disability, Turner type	85328	**13**	**13**
DEPRECATED : X-linked intellectual disability-hypotonic face syndrome	73220	**<10**	**<10**
OBSOLETE: 3-Phosphoglycerate dehydrogenase deficiency	422519	**<10**	**<10**
OBSOLETE: Abnormal origin or aberrant course of coronary artery	95493	**69**	**68**
OBSOLETE: Acheiria, bilateral	295103	**<10**	**<10**
OBSOLETE: Acheiria, unilateral	295101	**40**	**40**
OBSOLETE: Acquired alimentary behavior disorder of infancy	138118	**762**	**750**
OBSOLETE: Acquired hemophilia	73274	**319**	**214**
OBSOLETE: Acquired metabolic neuropathy	206616	**306**	**253**
OBSOLETE: Acrocephalosyndactyly	946	**<10**	**<10**
OBSOLETE: Acrodysostosis with multiple hormone resistance	280651	**<10**	**<10**
OBSOLETE: ACTH-independent Cushing syndrome	99893	**135**	**125**
OBSOLETE: Acute cutaneous lupus erythematosus	163528	**62**	**62**
OBSOLETE: Adactyly of foot	435623	**17**	**16**
OBSOLETE: Adactyly of foot, bilateral	295118	**<10**	**<10**
OBSOLETE: Adactyly of foot, unilateral	295116	**<10**	**<10**
OBSOLETE: Adactyly of hand	294931	**58**	**58**
OBSOLETE: Adult neuronal ceroid lipofuscinosis	79262	**<10**	**<10**
OBSOLETE: Adult pulmonary Langerhans cell histiocytosis	99874	**36**	**33**
OBSOLETE: Alpha-1-antichymotrypsin deficiency	93594	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Amelia of upper limb, bilateral	295055	**<10**	**<10**
OBSOLETE: Amelia of upper limb, unilateral	295053	**<10**	**<10**
OBSOLETE: Amniotic bands	1034	**108**	**107**
OBSOLETE: Aneurysm or dilatation of ascending aorta	95484	**131**	**128**
OBSOLETE: Antenatal Bartter syndrome	93604	**32**	**32**
OBSOLETE: Anti-HLA hyperimmunization	2194	**<10**	**<10**
OBSOLETE: Aortopulmonary coronary arterial course	99086	**<10**	**<10**
OBSOLETE: APC-related attenuated familial adenomatous polyposis	247806	**161**	**161**
OBSOLETE: Apodia, unilateral	295105	**<10**	**<10**
OBSOLETE: Aregenerative anemia	101096	**37**	**36**
OBSOLETE: Argyrophilic grain disease	97342	**<10**	**<10**
OBSOLETE: Arnold-Chiari malformation type II	1136	**134**	**130**
OBSOLETE: Arterial hypertension due to renal artery stenosis secondary to vasculitis	97599	**<10**	**<10**
OBSOLETE: Arthrogryposis due to muscular dystrophy	1155	**<10**	**<10**
OBSOLETE: Aseptic osteitis	57194	**<10**	**<10**
OBSOLETE: Atrichia-intellectual disability and growth delay syndrome	1211	**<10**	**<10**
OBSOLETE: ATR-X-related syndrome	263355	**<10**	**<10**
OBSOLETE: Atypical hemolytic uremic syndrome with B factor anomaly	93578	**<10**	**<10**
OBSOLETE: Atypical hemolytic uremic syndrome with C3 anomaly	93575	**<10**	**<10**
OBSOLETE: Atypical hemolytic uremic syndrome with H factor anomaly	93579	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Atypical hemolytic uremic syndrome with I factor anomaly	93580	**<10**	**<10**
OBSOLETE: Atypical hemolytic uremic syndrome with MCP/CD46 anomaly	93576	**<10**	**<10**
OBSOLETE: Auriculoocular anomalies-cleft lip syndrome	71270	**<10**	**<10**
OBSOLETE: Autoimmune enteropathy type 2	103916	**<10**	**<10**
OBSOLETE: Autosomal dominant childhood-onset cortical cataract	306561	**<10**	**<10**
OBSOLETE: Autosomal dominant coarctation of aorta	1455	**<10**	**<10**
OBSOLETE: Autosomal dominant focal dystonia, DYT7 type	93963	**<10**	**<10**
OBSOLETE: Autosomal dominant Opitz G/BBB syndrome	306588	**<10**	**<10**
OBSOLETE: Autosomal dominant optic atrophy and late-onset deafness	255117	**<10**	**<10**
OBSOLETE: Autosomal dominant spastic paraplegia type 9	100990	**<10**	**<10**
OBSOLETE: Autosomal recessive optic atrophy, OPA6 type	99012	**13**	**13**
OBSOLETE: AymÃ -Gripp syndrome	477668	**<10**	**<10**
OBSOLETE: Bacterial susceptibility due to TLR signaling pathway deficiency	183713	**10**	**<10**
OBSOLETE: Basal epidermolysis bullosa simplex	158665	**32**	**31**
OBSOLETE: Basement membrane disease	93550	**<10**	**<10**
OBSOLETE: Benign essential blepharospasm	93955	**112**	**109**
OBSOLETE: Benign exophthalmos syndrome	71269	**<10**	**<10**
OBSOLETE: Benign infantile seizures associated with mild gastroenteritis	166305	**<10**	**<10**
OBSOLETE: Blaichman syndrome	1250	**<10**	**<10**
OBSOLETE: Blepharophimosis-epicanthus inversus-ptosis due to a point mutation syndrome	261572	**<10**	**<10**
OBSOLETE: Blepharophimosis-radioulnar synostosis syndrome	1256	**<10**	**<10**
OBSOLETE: Borderline epithelial tumor of ovary	206473	**<10**	**<10**
OBSOLETE: Bowed tibiae-radial anomalies-osteopenia-fractures syndrome	3331	**<10**	**<10**
OBSOLETE: Brachydactyly	294937	**113**	**112**
OBSOLETE: Brachydactyly of fingers	294996	**<10**	**<10**
OBSOLETE: Brachydactyly of fingers, bilateral	295130	**<10**	**<10**
OBSOLETE: Brachydactyly of fingers, unilateral	295128	**<10**	**<10**
OBSOLETE: Brachydactyly of toes	294998	**<10**	**<10**
OBSOLETE: Brachydactyly of toes, unilateral	295132	**<10**	**<10**
OBSOLETE: Bullous systemic lupus erythematosus	46489	**13**	**12**
OBSOLETE: Central polydactyly of toes	295010	**<10**	**<10**
OBSOLETE: Centripetalis recessive dystrophic epidermolysis bullosa	89841	**57**	**41**
OBSOLETE: Cerebrorenodigital syndrome	1396	**<10**	**<10**
OBSOLETE: Cervical dystonia	93962	**105**	**99**
OBSOLETE: Cervical spina bifida aperta	268392	**<10**	**<10**
OBSOLETE: Cervical spina bifida cystica	268762	**<10**	**<10**
OBSOLETE: Cervicofacial lymphatic malformation	137923	**<10**	**<10**
OBSOLETE: Cervicothoracic spina bifida aperta	268397	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Cholesterol-ester transfer protein deficiency	79506	**<10**	**<10**
OBSOLETE: Choristoma	91353	**<10**	**<10**
OBSOLETE: Chronic pain requiring intraspinal analgesia	95426	**<10**	**<10**
OBSOLETE: CINCA syndrome with NLRP3 mutations	93365	**<10**	**<10**
OBSOLETE: Circumscribed lymphatic malformation	217410	**13**	**13**
OBSOLETE: Classic mast cell leukemia	158796	**<10**	**<10**
OBSOLETE: Classic paraneoplastic limbic encephalitis	163898	**37**	**29**
OBSOLETE: Combined hyperlipidemia	79211	**<10**	**<10**
OBSOLETE: Common variable immunodeficiency	1572	**3533**	**3414**
OBSOLETE: Common variable immunodeficiency due to an intrinsic T cell defect	99831	**<10**	**<10**
OBSOLETE: Common variable immunodeficiency due to TNFR deficiency	183672	**<10**	**<10**
OBSOLETE: Complete atrioventricular canal-left heart obstruction syndrome	99066	**<10**	**<10**
OBSOLETE: Congenital absence of both forearm and hand, unilateral	295093	**<10**	**<10**
OBSOLETE: Congenital absence of both lower leg and foot, bilateral	295099	**<10**	**<10**
OBSOLETE: Congenital absence of both lower leg and foot, unilateral	295097	**<10**	**<10**
OBSOLETE: Congenital absence of thigh and lower leg with foot present, unilateral	295089	**<10**	**<10**
OBSOLETE: Congenital absence of upper arm and forearm with hand present, unilateral	295085	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Congenital absence/ hypoplasia of fingers excluding thumb	294990	**11**	**11**
OBSOLETE: Congenital absence/ hypoplasia of fingers excluding thumb, bilateral	295114	**<10**	**<10**
OBSOLETE: Congenital absence/ hypoplasia of thumb, bilateral	295112	**<10**	**<10**
OBSOLETE: Congenital absence/ hypoplasia of thumb, unilateral	295110	**<10**	**<10**
OBSOLETE: Congenital adrenal hypoplasia of maternal cause	95701	**<10**	**<10**
OBSOLETE: Congenital aortic valve insufficiency	95449	**25**	**25**
OBSOLETE: Congenital bowing of long bones-short stature-dolichomacrocephaly-ocular hypertelorism syndrome	2292	**12**	**12**
OBSOLETE: Congenital cataract, Volkmann type	98983	**<10**	**<10**
OBSOLETE: Congenital cataract-ichthyosis syndrome	1376	**<10**	**<10**
OBSOLETE: Congenital hydronephrosis	2190	**1536**	**1525**
OBSOLETE: Congenital liver hemangioma	238691	**31**	**31**
OBSOLETE: Congenital muscular dystrophy-muscle hypertrophy-severe intellectual disability syndrome	329206	**<10**	**<10**
OBSOLETE: Congenital nasal pyriform aperture stenosis with holoprosencephaly	162521	**30**	**30**
OBSOLETE: Congenital neuronal ceroid lipofuscinosis	168486	**<10**	**<10**
OBSOLETE: Congenital sucrase-isomaltase deficiency with minimal starch tolerance	306446	**<10**	**<10**
OBSOLETE: Congenital sucrase-isomaltase deficiency with starch and lactose intolerance	306474	**<10**	**<10**
OBSOLETE: Congenital sucrase-isomaltase deficiency with starch intolerance	306436	**25**	**25**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Congenital sucrase-isomaltase deficiency without starch intolerance	306462	**<10**	**<10**
OBSOLETE: Congenital sucrase-isomaltase deficiency without sucrose intolerance	306486	**<10**	**<10**
OBSOLETE: Congenital unilateral pulmonary hypoplasia	2258	**11**	**11**
OBSOLETE: Congenital valvular dysplasia	1864	**17**	**17**
OBSOLETE: Coronary artery intramyocardial course	99085	**<10**	**<10**
OBSOLETE: Corticobasal degeneration	278	**66**	**40**
OBSOLETE: Craniosynostosis-cataract syndrome	1530	**<10**	**<10**
OBSOLETE: Craniosynostosis-fibular aplasia syndrome	1533	**<10**	**<10**
OBSOLETE: Cushing syndrome	553	**629**	**580**
OBSOLETE: Dacryocystitis-osteopoikilosis syndrome	1562	**11**	**11**
OBSOLETE: Deletion 20p	1611	**<10**	**<10**
OBSOLETE: Deletion 4q	1625	**10**	**<10**
OBSOLETE: Developmental delay-deafness syndrome, Hildebrand type	163988	**<10**	**<10**
OBSOLETE: Digestive duplication	238	**20**	**20**
OBSOLETE: Distal monosomy 20q	96152	**<10**	**<10**
OBSOLETE: Duplication 4q	1739	**<10**	**<10**
OBSOLETE: Dyschondrosteosis-nephritis syndrome	1765	**<10**	**<10**
OBSOLETE: Early infantile epileptic encephalopathy without suppression burst	369894	**12**	**11**
OBSOLETE: Early-onset schizophrenia	96369	**161**	**158**
OBSOLETE: Ehlers-Danlos syndrome type 1	90309	**60**	**60**
OBSOLETE: Ehlers-Danlos syndrome type 2	90318	**33**	**33**
OBSOLETE: Ehlers-Danlos syndrome type 7A	99875	**<10**	**<10**
OBSOLETE: Ehlers-Danlos syndrome type 7B	99876	**<10**	**<10**
OBSOLETE: Ehlers-Danlos syndrome, fibronectinemic type	75501	**<10**	**<10**
OBSOLETE: Embryonary disorganization syndrome	1664	**<10**	**<10**
OBSOLETE: Eosinophilic granuloma	99871	**<10**	**<10**
OBSOLETE: Epidermal nevus-vitamin D-resistant rickets syndrome	2694	**<10**	**<10**
OBSOLETE: Epidermolysis bullosa simplex superficialis	89839	**<10**	**<10**
OBSOLETE: Epimetaphyseal skeletal dysplasia	1819	**<10**	**<10**
OBSOLETE: Epithelio-exfoliative colitis-deafness syndrome	103912	**<10**	**<10**
OBSOLETE: Erythromelalgia	1956	**44**	**44**
OBSOLETE: Facial asymmetry-temporal seizures syndrome	1167	**<10**	**<10**
OBSOLETE: Familial articular chondrocalcinosis type 1	99781	**<10**	**<10**
OBSOLETE: Familial capillary hemangioma	91415	**26**	**24**
OBSOLETE: Familial cervical artery dissection	36382	**217**	**211**
OBSOLETE: Familial chondromalacia patellae	1428	**<10**	**<10**
OBSOLETE: Familial esophageal achalasia	99723	**<10**	**<10**
OBSOLETE: Familial hyperreninemic hypoaldosteronism type 2	99764	**<10**	**<10**
OBSOLETE: Familial hypospadias	440	**275**	**274**
OBSOLETE: Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	93217	**<10**	**<10**
OBSOLETE: Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	93213	**49**	**48**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Familial idiopathic steroid-resistant nephrotic syndrome with minimal changes	93216	**11**	**11**
OBSOLETE: Familial juvenile hyperuricemic nephropathy type 1	209886	**49**	**45**
OBSOLETE: Familial lambdoid synostosis	3267	**<10**	**<10**
OBSOLETE: Familial ovarian cancer	213517	**<10**	**<10**
OBSOLETE: Familial parathyroid adenoma	99877	**<10**	**<10**
OBSOLETE: Familial primary hypomagnesemia with normocalciuria and normocalcemia	34527	**<10**	**<10**
OBSOLETE: Familial segmental neurofibromatosis	79428	**89**	**88**
OBSOLETE: Familial spinal neurofibromatosis	79429	**<10**	**<10**
OBSOLETE: Farmer's lung disease	99906	**54**	**49**
OBSOLETE: Femoral agenesis/ hypoplasia, bilateral	295067	**<10**	**<10**
OBSOLETE: Femoral agenesis/ hypoplasia, unilateral	295065	**<10**	**<10**
OBSOLETE: Fibular hemimelia, bilateral	295083	**<10**	**<10**
OBSOLETE: Fibular hemimelia, unilateral	295081	**<10**	**<10**
OBSOLETE: Follicular atrophoderma-basal cell carcinoma	79459	**<10**	**<10**
OBSOLETE: Generalized epilepsy and praxis-induced seizures	99649	**<10**	**<10**
OBSOLETE: Genetic primary hypomagnesemia with hypocalciuria	306519	**<10**	**<10**
OBSOLETE: Genetic primary hypomagnesemia with normocalciuria	306522	**<10**	**<10**
OBSOLETE: Giant infantile hemangioma	210592	**47**	**47**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Glycogen storage disease due to acid maltase deficiency, adult onset	308604	**<10**	**<10**
OBSOLETE: Glycogen storage disease due to acid maltase deficiency, juvenile onset	308573	**<10**	**<10**
OBSOLETE: Graft rejection after lung transplantation	91128	**<10**	**<10**
OBSOLETE: Hand-Schuller-Christian disease	99873	**<10**	**<10**
OBSOLETE: Hashimoto-Pritzker syndrome	99872	**<10**	**<10**
OBSOLETE: Hemochromatosis type 4	139491	**318**	**307**
OBSOLETE: Hemochromatosis type 5	447792	**<10**	**<10**
OBSOLETE: Hemolytic anemia due to glyceraldehyde-3-phosphate dehydrogenase deficiency	248305	**<10**	**<10**
OBSOLETE: Hereditary motor and sensory neuropathy	140450	**412**	**380**
OBSOLETE: Hereditary pediatric BehÃ§et-like disease	476102	**103**	**102**
OBSOLETE: High isolated anorectal malformation	171201	**513**	**507**
OBSOLETE: Hirsutism-skeletal dysplasia-intellectual disability syndrome	2156	**<10**	**<10**
OBSOLETE: House allergic alveolitis	99907	**553**	**454**
OBSOLETE: Humeral agenesis/ hypoplasia, unilateral	295061	**<10**	**<10**
OBSOLETE: Hyperlipoproteinemia type 5	70470	**<10**	**<10**
OBSOLETE: Hyperphalangy, bilateral	295142	**<10**	**<10**
OBSOLETE: Hypertrichotic osteochondrodysplasia	2765	**<10**	**<10**
OBSOLETE: Hypertrophic cardiomyopathy due to intensive athletic training	217601	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Hypopituitarism-short stature-skeletal anomalies syndrome	2626	**<10**	**<10**
OBSOLETE: Idiopathic hypersomnia with long sleep time	228315	**152**	**151**
OBSOLETE: Idiopathic hypersomnia without long sleep time	228318	**100**	**99**
OBSOLETE: Idiopathic recurrent and disabling cutaneous herpes	35061	**<10**	**<10**
OBSOLETE: Idiopathic steroid-sensitive nephrotic syndrome with diffuse mesangial proliferation	93209	**<10**	**<10**
OBSOLETE: Idiopathic steroid-sensitive nephrotic syndrome with focal segmental hyalinosis	93206	**187**	**170**
OBSOLETE: Idiopathic steroid-sensitive nephrotic syndrome with minimal change	93207	**359**	**352**
OBSOLETE: Infantile axonal neuropathy	2679	**37**	**35**
OBSOLETE: Infantile neuronal ceroid lipofuscinosis	79263	**20**	**13**
OBSOLETE: Infundibulopelvic stenosis-multicystic kidney syndrome	1849	**<10**	**<10**
OBSOLETE: Inherited predisposition to essential thrombocythemia	225968	**<10**	**<10**
OBSOLETE: Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	404440	**72**	**72**
OBSOLETE: Intellectual disability-hypotonia-skin hyperpigmentation syndrome	3050	**<10**	**<10**
OBSOLETE: Intellectual disability-microcephaly-phalangeal-facial abnormalities syndrome	3067	**<10**	**<10**
OBSOLETE: Intellectual disability-microcephaly-unusual facies syndrome	3313	**<10**	**<10**
OBSOLETE: Intellectual disability-unusual facies syndrome	3043	**<10**	**<10**
OBSOLETE: Intermediate isolated anorectal malformation	171208	**132**	**129**
OBSOLETE: Intracranial aneurysms-multiple congenital anomalies syndrome	1057	**<10**	**<10**
OBSOLETE: Isolated cloverleaf skull syndrome	2343	**<10**	**<10**
OBSOLETE: Isolated facial myokymia	221106	**<10**	**<10**
OBSOLETE: Isolated megalencephaly	268920	**<10**	**<10**
OBSOLETE: Isolated optic nerve hypoplasia/aplasia	137902	**33**	**33**
OBSOLETE: Isolated optic neuritis with anti-MOG antibodies	592888	**89**	**88**
OBSOLETE: Isolated optic neuritis without anti-MOG antibodies	592885	**41**	**40**
OBSOLETE: Isolated oxycephaly	63440	**63**	**62**
OBSOLETE: Isolated plagiocephaly	35098	**688**	**686**
OBSOLETE: Ito hypomelanosis	435	**297**	**293**
OBSOLETE: Junctional epidermolysis bullosa, non-Herlitz type	89840	**<10**	**<10**
OBSOLETE: Juvenile chronic recurrent multifocal osteomyelitis	2778	**105**	**105**
OBSOLETE: Juvenile neuronal ceroid lipofuscinosis	79264	**35**	**24**
OBSOLETE: Juvenile-onset SAPHO syndrome	324989	**<10**	**<10**
OBSOLETE: Laminopathy type Decaudain-Vigouroux	137871	**11**	**10**
OBSOLETE: Laryngeal dyskinesia	93961	**15**	**14**
OBSOLETE: Late infantile neuronal ceroid lipofuscinosis	168491	**23**	**10**
OBSOLETE: Leptomyelolipoma	268838	**58**	**58**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Letterer-Siwe disease	99870	**<10**	**<10**
OBSOLETE: Limb dystonia	93957	**41**	**40**
OBSOLETE: Limbic encephalitis	163892	**171**	**154**
OBSOLETE: Limbic encephalitis with caspr2 antibodies	276402	**<10**	**<10**
OBSOLETE: Limbic encephalitis with LGI1 antibodies	163908	**74**	**65**
OBSOLETE: Lipoma associated with neurospinal dysraphism	268832	**181**	**181**
OBSOLETE: Lissencephaly-demyelinating axonal neuropathy syndrome	101356	**<10**	**<10**
OBSOLETE: Localized epiphyseal dysplasia	1823	**<10**	**<10**
OBSOLETE: Low isolated anorectal malformation	171215	**1137**	**1116**
OBSOLETE: Low-grade ependymoma	251633	**<10**	**<10**
OBSOLETE: Lown-Ganong-Levine syndrome	844	**<10**	**<10**
OBSOLETE: Lumbosacral spina bifida aperta	268388	**23**	**22**
OBSOLETE: Lumbosacral spina bifida cystica	268758	**18**	**18**
OBSOLETE: Lymphedema praecox	77241	**10**	**10**
OBSOLETE: Lymphedema tarda	77242	**10**	**10**
OBSOLETE: Lymphocytic colitis	65279	**<10**	**<10**
OBSOLETE: Lymphomatous meningitis	329998	**<10**	**<10**
OBSOLETE: Lymphoproliferative syndrome	238510	**13**	**12**
OBSOLETE: Maculopapular lupus rash	90287	**<10**	**<10**
OBSOLETE: Madelung deformity	35688	**26**	**26**
OBSOLETE: Madelung deformity, bilateral	295223	**13**	**13**
OBSOLETE: Maternally-inherited mitochondrial hypertrophic cardiomyopathy	255225	**<10**	**<10**
OBSOLETE: MECP2 duplication syndrome	85281	**12**	**10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Metastatic spermatocytic seminoma	99866	**<10**	**<10**
OBSOLETE: Microcephaly-seizures-developmental delay syndrome	228418	**<10**	**<10**
OBSOLETE: Microcornea-corectopia-macular hypoplasia syndrome	2535	**<10**	**<10**
OBSOLETE: Microphthalmia-cataract syndrome	2543	**20**	**20**
OBSOLETE: Multicentric Castleman disease	93686	**13**	**11**
OBSOLETE: Multifocal muscular fibrosis-obstructed vessels syndrome	2033	**<10**	**<10**
OBSOLETE: Multiple fibroadenoma of the breast	50920	**320**	**317**
OBSOLETE: Multiple ventricular septal defects	99096	**12**	**12**
OBSOLETE: Mycobacterium xenopi infection	314946	**<10**	**<10**
OBSOLETE: MYH7-related late-onset scapuloperoneal muscular dystrophy	437572	**34**	**34**
OBSOLETE: Myopathy-growth delay-intellectual disability-hypospadias syndrome	2601	**<10**	**<10**
OBSOLETE: Myostatin-related muscle hypertrophy	275534	**<10**	**<10**
OBSOLETE: Natal teeth-intestinal pseudoobstruction-patent ductus syndrome	1654	**<10**	**<10**
OBSOLETE: Neuroepithelioma	2677	**<10**	**<10**
OBSOLETE: Neurogenic palpebral tumor	98593	**<10**	**<10**
OBSOLETE: Non-distal monosomy 7p	96136	**<10**	**<10**
OBSOLETE: Non-dystrophic myopathy with collagen 6 anomaly	206659	**72**	**70**
OBSOLETE: Non-herpetic acute limbic encephalitis	163924	**40**	**38**
OBSOLETE: Non-secreting chemodectoma	101106	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Not NOTCH3-related small vessel disease of the brain	77304	**<10**	**<10**
OBSOLETE: Ocular coloboma	194	**32**	**32**
OBSOLETE: Oculocerebral dysplasia	2705	**<10**	**<10**
OBSOLETE: Oligoarticular juvenile idiopathic arthritis with anti-nuclear antibodies	247839	**311**	**309**
OBSOLETE: Oligoarticular juvenile idiopathic arthritis without anti-nuclear antibodies	247846	**82**	**82**
OBSOLETE: Ophthalmoplegia-myalgia-tubular aggregates syndrome	2742	**<10**	**<10**
OBSOLETE: Orofaciodigital syndrome type 10	2756	**<10**	**<10**
OBSOLETE: Osteochondritis of tarsal/metatarsal bone	2054	**<10**	**<10**
OBSOLETE: Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome	2653	**<10**	**<10**
OBSOLETE: Osteoporosis-macrocephaly-blindness-joint hyperlaxity syndrome	2787	**<10**	**<10**
OBSOLETE: Otopalatodigital syndrome	669	**11**	**10**
OBSOLETE: Papillary fibroelastoma of the heart	208600	**<10**	**<10**
OBSOLETE: Paraneoplastic limbic encephalitis	163895	**14**	**11**
OBSOLETE: Patella aplasia/hypoplasia, unilateral	295038	**<10**	**<10**
OBSOLETE: Pediatric Castleman disease	93682	**<10**	**<10**
OBSOLETE: Pediatric polyarteritis nodosa	93564	**<10**	**<10**
OBSOLETE: Pediatric SjÃ¶gren syndrome	93566	**<10**	**<10**
OBSOLETE: Pediatric systemic sclerosis	93567	**<10**	**<10**
OBSOLETE: Peripheral dysostosis	1795	**<10**	**<10**
OBSOLETE: Peripheral hypothyroidism	226310	**23**	**22**
OBSOLETE: Peripheral resistance to thyroid hormones	97927	**<10**	**<10**
OBSOLETE: Peters anomaly-cataract syndrome	101033	**<10**	**<10**
OBSOLETE: Phosphoenolpyruvate carboxykinase 2 deficiency	79317	**<10**	**<10**
OBSOLETE: Pigeon-breeder lung disease	99908	**129**	**105**
OBSOLETE: Pitt-Hopkins-like syndrome	221150	**<10**	**<10**
OBSOLETE: Platelet function disease associated with renal insufficiency	99146	**<10**	**<10**
OBSOLETE: Pleomorphic salivary gland adenoma	454821	**<10**	**<10**
OBSOLETE: Polydactyly of a biphalangeal thumb, unilateral	295144	**<10**	**<10**
OBSOLETE: Polydactyly of a triphalangeal thumb, bilateral	295150	**<10**	**<10**
OBSOLETE: Polymicrogyria-turricephaly-hypogenitalism syndrome	2925	**<10**	**<10**
OBSOLETE: Polyradiculoneuropathy associated with IgG/IgA/IgM monoclonal gammopathy without known antibodies	208981	**138**	**99**
OBSOLETE: Polysyndactyly, bilateral	295161	**<10**	**<10**
OBSOLETE: Polysyndactyly, unilateral	295159	**<10**	**<10**
OBSOLETE: Postaxial polydactyly of fingers	294942	**26**	**26**
OBSOLETE: Postaxial polydactyly of toes	295008	**<10**	**<10**
OBSOLETE: Postaxial polydactyly of toes, unilateral	295179	**<10**	**<10**
OBSOLETE: Postaxial polydactyly type B, bilateral	295169	**<10**	**<10**
OBSOLETE: Postsurgical hypopituitarism	95621	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Posttraumatic diabetes insipidus	95625	**<10**	**<10**
OBSOLETE: Posttraumatic hypopituitarism	95623	**<10**	**<10**
OBSOLETE: Preaxial polydactyly of fingers	294939	**36**	**35**
OBSOLETE: Preaxial polydactyly of toes	295006	**<10**	**<10**
OBSOLETE: Preaxial polydactyly of toes, bilateral	295177	**<10**	**<10**
OBSOLETE: Primary parathyroid hyperplasia	99878	**68**	**66**
OBSOLETE: Primary pigmented nodular adrenocortical disease	189439	**17**	**17**
OBSOLETE: Radial hemimelia, bilateral	295071	**<10**	**<10**
OBSOLETE: Radial hemimelia, unilateral	295069	**<10**	**<10**
OBSOLETE: Radiation-induced hypopituitarism	95622	**<10**	**<10**
OBSOLETE: Radio-ulnar synostosis, bilateral	295219	**15**	**15**
OBSOLETE: Radio-ulnar synostosis, unilateral	295217	**12**	**12**
OBSOLETE: Rapidly progressive glomerulonephritis	280569	**51**	**50**
OBSOLETE: Rare idiopathic male infertility	98345	**29**	**29**
OBSOLETE: Rare major hypertriglyceridemia	181425	**11**	**11**
OBSOLETE: Rare variants of adenocarcinoma of the corpus uteri	213574	**<10**	**<10**
OBSOLETE: Recessive aplasia cutis congenita of limbs	1115	**<10**	**<10**
OBSOLETE: Recessive hereditary methemoglobinemia type 2	139380	**<10**	**<10**
OBSOLETE: Renier-Gabreels-Jasper syndrome	93975	**15**	**12**
OBSOLETE: Retrocerebellar cyst	269200	**<10**	**<10**
OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis with anti-nuclear antibodies	247854	**68**	**68**
OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis without anti-nuclear antibodies	247861	**35**	**35**
OBSOLETE: Sakati-Nyhan syndrome	3128	**<10**	**<10**
OBSOLETE: Secondary acute transverse myelitis	139420	**12**	**11**
OBSOLETE: Secondary ciliary dyskinesia	91365	**<10**	**<10**
OBSOLETE: Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency	35123	**<10**	**<10**
OBSOLETE: Short stature-heart defect-craniofacial anomalies syndrome	1088	**<10**	**<10**
OBSOLETE: Shoulder and girdle defects-familial intellectual disability syndrome	2580	**<10**	**<10**
OBSOLETE: Shy-Drager syndrome	98932	**<10**	**<10**
OBSOLETE: Single ventricular septal defect	99097	**12**	**11**
OBSOLETE: Sino-auricular heart block	1260	**<10**	**<10**
OBSOLETE: Solitary median maxillary central incisor syndrome	2286	**23**	**22**
OBSOLETE: Spastic diplegia, infantile type	1680	**62**	**61**
OBSOLETE: Split foot	294994	**<10**	**<10**
OBSOLETE: Split foot, bilateral	295126	**<10**	**<10**
OBSOLETE: Split hand	294992	**<10**	**<10**
OBSOLETE: Split hand or/and split foot malformation	294935	**11**	**11**
OBSOLETE: Split hand, bilateral	295122	**<10**	**<10**
OBSOLETE: Split hand, unilateral	295120	**<10**	**<10**
OBSOLETE: Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome	94095	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Spondylocostal dysostosis-hypospadias-intellectual disability syndrome	329252	**<10**	**<10**
OBSOLETE: Spondyloepimetaphyseal dysplasia with joint laxity	93359	**<10**	**<10**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation	93222	**<10**	**<10**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	93220	**<10**	**<10**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	93218	**113**	**107**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with minimal changes	93221	**90**	**88**
OBSOLETE: Sporadic Leigh syndrome	255199	**<10**	**<10**
OBSOLETE: Sporadic pheochromocytoma	276624	**141**	**134**
OBSOLETE: Sporadic secreting paraganglioma	276627	**23**	**22**
OBSOLETE: Squamous cell carcinoma of salivary glands	500481	**<10**	**<10**
OBSOLETE: Superior celosomia	93942	**<10**	**<10**
OBSOLETE: Suprabasal epidermolysis bullosa simplex	158661	**<10**	**<10**
OBSOLETE: Symbrachydactyly of hand and foot, unilateral	295136	**<10**	**<10**
OBSOLETE: Symptomatic form of fragile X syndrome in female carriers	449291	**36**	**36**
OBSOLETE: TACI-related selective deficiency of IgA	99974	**<10**	**<10**
OBSOLETE: Tay-Sachs disease, B1 variant	309239	**<10**	**<10**
OBSOLETE: Terminal limb defects	294929	**10**	**10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Terminal transverse defects of arm	93937	**<10**	**<10**
OBSOLETE: Thoracolumbosacral spina bifida aperta	268384	**<10**	**<10**
OBSOLETE: Tibial hemimelia, bilateral	295079	**<10**	**<10**
OBSOLETE: Tibial hemimelia, unilateral	295077	**<10**	**<10**
OBSOLETE: Total spina bifida aperta	268377	**16**	**16**
OBSOLETE: Total spina bifida cystica	268748	**<10**	**<10**
OBSOLETE: Toxic or/and iatrogenic neuropathy	206619	**203**	**161**
OBSOLETE: Tracheo-esophageal fistula-hypospadias syndrome	2042	**<10**	**<10**
OBSOLETE: Tricho-oculo-dermo-vertebral syndrome	3354	**<10**	**<10**
OBSOLETE: Tricuspid valve prolapse	95458	**<10**	**<10**
OBSOLETE: Trochlear nerve palsy	99664	**<10**	**<10**
OBSOLETE: Truncal dystonia	93956	**<10**	**<10**
OBSOLETE: Tuberculous meningitis	499004	**15**	**14**
OBSOLETE: Ulnar hemimelia, bilateral	295073	**<10**	**<10**
OBSOLETE: Ulnar hemimelia, unilateral	295075	**<10**	**<10**
OBSOLETE: Unclassified metaphyseal chondrodysplasia	90345	**<10**	**<10**
OBSOLETE: Unclassified overlapping connective tissue disease	251316	**<10**	**<10**
OBSOLETE: Univentricular heart with single atrio-ventricular valve	99069	**17**	**15**
OBSOLETE: Unknown leukodystrophy	84096	**111**	**99**
OBSOLETE: Upper thoracic spina bifida aperta	268740	**<10**	**<10**
OBSOLETE: Upper thoracic spina bifida cystica	268770	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
OBSOLETE: Vascular disruption sequence	3160	**<10**	**<10**
OBSOLETE: Vibratory angioedema	493348	**<10**	**<10**
OBSOLETE: Vitiligo-associated autoimmune disease	247871	**14**	**14**
OBSOLETE: Von Hippel anomaly	98941	**20**	**19**
OBSOLETE: Xeroderma pigmentosum complementation group A	276249	**<10**	**<10**
OBSOLETE: Xeroderma pigmentosum complementation group B	276252	**<10**	**<10**
OBSOLETE: Xeroderma pigmentosum complementation group C	276255	**27**	**25**
OBSOLETE: Xeroderma pigmentosum complementation group D	276258	**<10**	**<10**
OBSOLETE: Xeroderma pigmentosum complementation group E	276261	**<10**	**<10**
OBSOLETE: X-linked acrogigantism due to Xq26 microduplication	448372	**<10**	**<10**
OBSOLETE: X-linked dominant intellectual disability-epilepsy syndrome	93951	**<10**	**<10**
OBSOLETE: X-linked intellectual disability, Raynaud type	3061	**<10**	**<10**
OBSOLETE: X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency	319612	**14**	**13**
OBSOLETE: X-linked Opitz G/BBB syndrome	306597	**<10**	**<10**
OBSOLETE: X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome	83648	**<10**	**<10**

BNDMR

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