BNDMR

Banque Nationale de Données
Maladies Rares

Number of cases per rare disease

registered in the French National Rare Disease Registry (BNDMR) as of 1st May 2024

ASSISTANCE PUBLIQUE HÔPITAUX DE PARIS

MINISTÈRE DES SOLIDARITÉS ET DE LA SANTÉ
Liberté
Égalité
Fraternité

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 3rd National Plan for Rare Diseases (PNMR3) 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
Banque Nationale de Données
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.

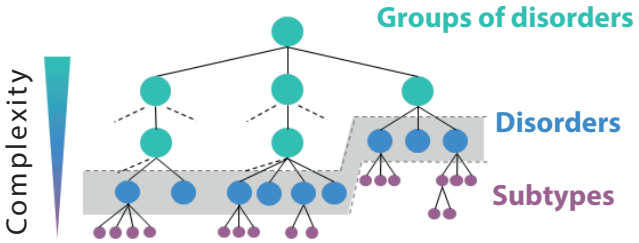

Figure 1 : Orphanet nomenclature classification levels representation

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 2023 version.

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

- **Codes that have become obsolete** : these codes have been removed from the Orphanet nomenclature because they are, for instance, codes assigned to a disease that was not well defined, or that already had one.
- **'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.

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 01/05/2024, 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 engagend 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, the May version of this report is more exaustive than the version of november 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) (p.6)

2- Number of cases per obsolete rare disease registered in the BNDMR (p.95)

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 ORPHAcodes

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
10q22.3q23.3 microdeletion syndrome	276413	**14**	**14**
10q22.3q23.3 microduplication syndrome	276422	**<10**	**<10**
11q22.2q22.3 microdeletion syndrome	444002	**<10**	**<10**
12q14 microdeletion syndrome	94063	**11**	**11**
12q15q21.1 microdeletion syndrome	289513	**<10**	**<10**
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	**136**	**133**
15q11.2 microdeletion syndrome	261183	**174**	**173**
15q11q13 microduplication syndrome	238446	**107**	**103**
15q13.3 microdeletion syndrome	199318	**119**	**119**
15q14 microdeletion syndrome	261190	**<10**	**<10**
16p11.2p12.2 microdeletion syndrome	261211	**47**	**47**
16p11.2p12.2 microduplication syndrome	261204	**44**	**44**
16p12.1p12.3 triplication syndrome	485405	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
16p13.11 microdeletion syndrome	261236	**49**	**49**
16p13.11 microduplication syndrome	261243	**55**	**55**
16p13.3 microduplication syndrome	96078	**17**	**17**
16q24.1 microdeletion syndrome	352629	**<10**	**<10**
16q24.3 microdeletion syndrome	261250	**<10**	**<10**
17p11.2 microduplication syndrome	1713	**100**	**99**
17p13.3 microduplication syndrome	217385	**31**	**30**
17q11.2 microduplication syndrome	139474	**17**	**17**
17q12 microdeletion syndrome	261265	**60**	**60**
17q12 microduplication syndrome	261272	**67**	**67**
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	**<10**	**<10**
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	**232**	**213**
1q21.1 microdeletion syndrome	250989	**111**	**111**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
1q21.1 microduplication syndrome	250994	**71**	**71**
1q41q42 microdeletion syndrome	250999	**12**	**12**
1q44 microdeletion syndrome	238769	**20**	**20**
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	**26**	**26**
21q22.11q22.12 microdeletion syndrome	261323	**21**	**21**
22q11.2 deletion syndrome	567	**3266**	**3154**
22q11.2 duplication syndrome	1727	**352**	**348**
2-aminoadipic 2-oxoadipic aciduria	79154	**<10**	**<10**
2-hydroxyglutaric aciduria*	19	**11**	**<10**
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	**14**	**14**
2q23.1 microduplication syndrome	313947	**<10**	**<10**
2q24 microdeletion syndrome	1617	**25**	**23**
2q31.1 microdeletion syndrome	251014	**<10**	**<10**
2q32q33 microdeletion syndrome	251019	**<10**	**<10**
2q37 microdeletion syndrome	1001	**127**	**123**
3C syndrome	7	**<10**	**<10**
3-hydroxy-3-methylglutaric aciduria	20	**32**	**31**
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	**89**	**85**
3MC syndrome	293843	**12**	**11**
3-methylcrotonyl-CoA carboxylase deficiency	6	**14**	**13**
3-methylglutaconic aciduria type 1	67046	**<10**	**<10**
3-methylglutaconic aciduria type 3	67047	**12**	**11**
3-methylglutaconic aciduria type 4	67048	**<10**	**<10**
3-methylglutaconic aciduria*	289902	**<10**	**<10**
3p25.3 microdeletion syndrome	435638	**12**	**12**
3q13 microdeletion syndrome	1621	**12**	**12**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
3q26 microduplication syndrome	96095	**20**	**19**
3q26q27 microdeletion syndrome	356947	**<10**	**<10**
3q27.3 microdeletion syndrome	397695	**<10**	**<10**
3q29 microdeletion syndrome	65286	**44**	**44**
3q29 microduplication syndrome	251038	**18**	**17**
45,X/46,XY mixed gonadal dysgenesis	1772	**282**	**281**
46,XX difference of sex development-anorectal anomalies syndrome	2973	**<10**	**<10**
46,XX disorder of sex development induced by endogenous maternal-derived androgen*	325093	**<10**	**<10**
46,XX disorder of sex development induced by exogenous maternal-derived androgen*	325099	**<10**	**<10**
46,XX gonadal dysgenesis	243	**83**	**82**
46,XX ovotesticular difference of sex development	2138	**76**	**76**
46,XX testicular difference of sex development	393	**62**	**62**
46,XY complete gonadal dysgenesis	242	**398**	**391**
46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	752	**29**	**29**
46,XY difference of sex development due to 5-alpha-reductase 2 deficiency	753	**56**	**56**
46,XY difference of sex development due to isolated 17,20-lyase deficiency	90796	**<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	**50**	**50**
46,XY partial gonadal dysgenesis	251510	**304**	**303**
47,XYY syndrome	8	**279**	**274**
48,XXXY syndrome	96263	**28**	**28**
48,XXYY syndrome	10	**65**	**65**
49,XXXXY syndrome	96264	**36**	**36**
49,XXXYY syndrome	261534	**<10**	**<10**
49,XYYYY syndrome	99330	**<10**	**<10**
4H leukodystrophy	289494	**78**	**74**
4p16.3 microduplication syndrome	96072	**<10**	**<10**
4q21 microdeletion syndrome	238750	**14**	**13**
4q25 proximal deletion syndrome	502437	**<10**	**<10**
5p13 microduplication syndrome	329802	**<10**	**<10**
5q14.3 microdeletion syndrome	228384	**14**	**14**
5q35 microduplication syndrome	228415	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
6p22 microdeletion syndrome	251046	<10	<10
6-phosphogluconate dehydrogenase deficiency	99135	125	124
6q terminal deletion syndrome	75857	37	36
6q16 microdeletion syndrome	171829	18	18
6q25 microdeletion syndrome	251056	15	15
7p22.1 microduplication syndrome	314034	<10	<10
7q11.23 microduplication syndrome	96121	93	93
7q31 microdeletion syndrome	251061	12	12
8p inverted duplication/deletion syndrome	96092	13	13
8p11.2 deletion syndrome	251066	<10	<10
8p23.1 duplication syndrome	251076	23	22
8p23.1 microdeletion syndrome	251071	64	63
8q12 microduplication syndrome	228399	<10	<10
8q21.11 microdeletion syndrome	284160	<10	<10
8q24.3 microdeletion syndrome	508488	25	25
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	317	264
AApoAIV amyloidosis	439232	<10	<10
Aarskog-Scott syndrome	915	155	155
Aase-Smith syndrome	916	<10	<10
ABeta2M amyloidosis*	439246	<10	<10
Abetalipoproteinemia	14	39	39
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	13	13
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	13	12
Absence of uterine body	180142	<10	<10
Absent thumb-short stature-immunodeficiency syndrome	2951	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Absent tibia-polydactyly-arachnoid cyst syndrome	3328	<10	<10
Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome	90301	<10	<10
Aceruloplasminemia	48818	43	40
Acetazolamide-responsive myotonia	99736	<10	<10
Achalasia-microcephaly syndrome	929	13	13
Acheiria	294983	<10	<10
Acheiropodia	931	<10	<10
Achondrogenesis	932	<10	<10
Achondroplasia	15	773	764
Achromatopsia	49382	326	325
Acinar cell carcinoma of pancreas	424046	<10	<10
Acquired aneurysmal subarachnoid hemorrhage	90065	<10	<10
Acquired angioedema with C1Inh deficiency	528663	86	81
Acquired angioedema*	91385	65	62
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	23	23
Acquired factor X deficiency	599501	<10	<10
Acquired factor XI deficiency	599507	25	25
Acquired factor XIII deficiency	599513	<10	<10
Acquired generalized lipodystrophy	79086	68	63
Acquired hemophilia A	599480	105	96
Acquired ichthyosis	454	82	82
Acquired idiopathic sideroblastic anemia	75564	11	<10
Acquired kinky hair syndrome	37559	<10	<10
Acquired methemoglobinemia	464453	<10	<10
Acquired monoclonal Ig light chain-associated Fanconi syndrome	91136	26	22
Acquired partial lipodystrophy	79087	60	56
Acquired prothrombin deficiency	26348	<10	<10
Acquired pseudoxanthoma elasticum	228247	<10	<10
Acquired purpura fulminans	49566	34	33
Acquired von Willebrand syndrome	99147	377	352
Acral peeling skin syndrome	263534	22	22
Acrocallosal syndrome	36	18	18
Acrocapitofemoral dysplasia	63446	<10	<10
Acrocardiofacial syndrome	2008	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Acrocraniofacial dysostosis	949	<10	<10
Acrodermatitis enteropathica	37	26	24
Acrodysostosis	950	77	75
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	3266	3142
Acromelic dysplasia*	93436	18	17
Acromesomelic dysplasia*	93437	14	14
Acromesomelic dysplasia, Grebe type	2098	<10	<10
Acromesomelic dysplasia, Hunter-Thompson type	968	<10	<10
Acromesomelic dysplasia, Maroteaux type	40	16	16
Acromicric dysplasia	969	46	43
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	73	70
Actinic lichen planus	254395	<10	<10
Actinic prurigo	330061	<10	<10
Actinomycosis	457095	<10	<10
Action myoclonus-renal failure syndrome	163696	<10	<10
Activated PI3K-delta syndrome	397596	53	52
Acute ackee fruit intoxication	73423	<10	<10
Acute adrenal insufficiency	95409	161	155
Acute annular outer retinopathy	284460	<10	<10
Acute bilirubin encephalopathy	529799	<10	<10
Acute disseminated encephalomyelitis	83597	429	426
Acute encephalopathy with biphasic seizures and late reduced diffusion	363549	<10	<10
Acute encephalopathy with inflammation-mediated status epilepticus*	363567	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Acute erythroid leukemia	318	<10	<10
Acute fatty liver of pregnancy	243367	<10	<10
Acute flaccid myelitis	623801	<10	<10
Acute generalized exanthematous pustulosis	293173	292	267
Acute hepatic porphyria*	95157	11	11
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	623	588
Acute intermittent porphyria	79276	709	534
Acute interstitial pneumonia	79126	260	235
Acute liver failure	90062	35	29
Acute lymphoblastic leukemia*	513	92	87
Acute macular neuroretinopathy	488239	<10	<10
Acute megakaryoblastic leukemia	518	<10	<10
Acute monoblastic/monocytic leukemia	514	<10	<10
Acute motor and sensory axonal neuropathy	98917	790	650
Acute motor axonal neuropathy	98918	305	278
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
Acute myeloid leukemia with minimal differentiation	98832	<10	<10
Acute myeloid leukemia*	519	41	33
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

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Acute poisoning by drugs with membrane-stabilizing effect	43119	<10	<10
Acute promyelocytic leukemia	520	<10	<10
Acute pure sensory neuropathy	231450	67	63
Acute sensory ataxic neuropathy	231466	79	72
Acute transverse myelitis	139417	345	340
Acute tricyclic antidepressant poisoning	43117	<10	<10
Acute undifferentiated leukemia	98835	<10	<10
Acute zonal occult outer retinopathy	284454	17	17
Acyl-CoA dehydrogenase 9 deficiency	99901	<10	<10
Acyl-CoA dehydrogenase deficiency*	309120	<10	<10
Adamantinoma	55881	<10	<10
Adams-Oliver syndrome	974	163	157
Addison disease	85138	1216	1168
Adducted thumbs-arthrogryposis syndrome, Christian type	2952	13	13
Adenine phosphoribosyltransferase deficiency	976	59	58
Adenocarcinoma of ovary	213504	28	20
Adenocarcinoma of the cervix uteri	213772	<10	<10
Adenocarcinoma of the esophagus	99976	<10	<10
Adenocarcinoma of the small intestine	104075	<10	<10
Adenohypophysitis	95512	511	452
Adenoma of pancreas	93292	<10	<10
Adenosarcoma of the corpus uteri	213600	<10	<10
Adenosine monophosphate deaminase deficiency	45	40	39
Adenylosuccinate lyase deficiency	46	24	19
Adenylosuccinate synthetase-like 1-related distal myopathy	482601	<10	<10
Adiposis dolorosa	36397	21	19
ADNP syndrome	404448	46	46
Adrenocortical carcinoma	1501	348	314
Adrenocortical carcinoma with pure aldosterone hypersecretion	231625	<10	<10
Adrenomyodystrophy	977	<10	<10
Adult acute respiratory distress syndrome	70578	28	20
Adult familial nephronophthisis-spastic quadriparesia syndrome	2666	<10	<10
Adult hepatocellular carcinoma	210159	<10	<10
Adult idiopathic neutropenia	2688	376	368
Adult neuronal ceroid lipofuscinosis	79262	11	<10
ADULT syndrome	978	27	27
Adult T-cell leukemia/lymphoma	86875	<10	<10
Adult-onset autosomal dominant leukodystrophy	99027	11	11
Adult-onset autosomal recessive cerebellar ataxia	284289	62	62
Adult-onset autosomal recessive sideroblastic anemia	255132	<10	<10
Adult-onset cervical dystonia, DYT23 type	420492	414	410
Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy	329336	67	63
Adult-onset distal myopathy due to VCP mutation	329478	55	51
Adult-onset dystonia-parkinsonism	199351	18	17
Adult-onset foveomacular vitelliform dystrophy	99000	104	103
Adult-onset immunodeficiency with anti-interferon-gamma autoantibodies	306431	170	166
Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency	329314	<10	<10
Adult-onset nemaline myopathy	171442	27	24
Adult-onset Still disease	829	712	702
African trypanosomiasis	3385	<10	<10
Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	83617	<10	<10
AGel amyloidosis	85448	18	17
Agenesis of the superior vena cava	99114	<10	<10
Aggressive primary cutaneous B-cell lymphoma*	178554	<10	<10
Aggressive systemic mastocytosis	98850	63	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	38	38
Aicardi syndrome	50	65	61
Aicardi-Goutières syndrome	51	224	206
AICA-ribosiduria	250977	<10	<10
AL amyloidosis	85443	2464	1832
Alacrimia-choreoathetosis-liver dysfunction syndrome	404454	<10	<10
Alagille syndrome	52	520	484

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Åland Islands eye disease	178333	<10	<10
Alar cartilages hypoplasia-coloboma-telecanthus syndrome	2007	<10	<10
Alazami syndrome	319671	15	15
Albers-Schönberg osteopetrosis	53	48	47
Albinism-deafness syndrome	998	<10	<10
Alexander disease	58	54	43
ALG11-CDG	280071	<10	<10
ALG12-CDG	79324	<10	<10
ALG13-CDG	324422	<10	<10
ALG1-CDG	79327	12	11
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
Alkaptonuria	56	113	109
ALK-positive large B-cell lymphoma	364043	<10	<10
Allan-Herndon-Dudley syndrome	59	54	48
Allergic bronchopulmonary aspergillosis	1164	379	365
Alopecia antibody deficiency	1006	<10	<10
Alopecia totalis	700	88	87
Alopecia universalis	701	114	114
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	26	12
Alpha delta granule deficiency	734	130	128
Alpha-1-antitrypsin deficiency	60	1463	1421
Alpha-B crystallin-related late-onset myopathy	399058	<10	<10
Alpha-crystallinopathy*	98910	<10	<10
Alpha-dystroglycan-related limb-girdle muscular dystrophy R16	280333	<10	<10
Alpha-mannosidosis	61	67	67
Alpha-N-acetylgalactosaminidase deficiency	3137	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3	62	147	136
Alpha-thalassemia	846	457	452
Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	98791	16	16
Alpha-thalassemia-myelodysplastic syndrome	231401	<10	<10
Alpha-thalassemia-X-linked intellectual disability syndrome	847	55	51
Alport syndrome	63	2478	2392
Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	86818	<10	<10
Alström syndrome	64	87	84
Alternating hemiplegia of childhood	2131	60	56
Alternating hemiplegia*	209978	73	71
Alveolar echinococcosis	284	16	16
Alveolar soft tissue sarcoma	163699	<10	<10
Amelia of lower limb	294969	<10	<10
Amelia of upper limb	294967	<10	<10
Amelia*	294925	<10	<10
Ameloblastoma	314419	<10	<10
Amelocerebrohypohidrotic syndrome	1946	12	11
Amelogenesis imperfecta	88661	816	815
Amelo-onycho-hypohidrotic syndrome	1028	<10	<10
Aminoacylase deficiency*	308448	<10	<10
Aminopterin/methotrexate embryofetopathy	1908	<10	<10
Amoebic keratitis	67043	20	20
Amyotrophic lateral sclerosis	803	13711	6798
Amyotrophic lateral sclerosis type 4	357043	20	17
Anal fistula	228113	<10	<10
Anaplastic astrocytoma	251589	<10	<10
Anaplastic ependymoma	251646	<10	<10
Anaplastic ganglioglioma	251957	<10	<10
Anaplastic large cell lymphoma	98841	<10	<10
Anaplastic oligoastrocytoma	251663	<10	<10
Anaplastic oligodendroglioma	251630	<10	<10
Anaplastic thyroid carcinoma	142	<10	<10
Anauxetic dysplasia	93347	14	14
Andersen-Tawil syndrome	37553	91	91
Androgen insensitivity syndrome*	754	68	68
Aneurysm of sinus of Valsalva	1054	624	619

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Aneurysmal bone cyst	480553	<10	<10
Aneurysm-osteoarthritis syndrome	284984	79	77
Angelman syndrome	72	799	786
Angel-shaped phalango-epiphyseal dysplasia	63442	<10	<10
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
Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome	69088	<10	<10
Aniridia-cerebellar ataxia-intellectual disability syndrome	1065	25	25
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	62	62
Ankylosing vertebral hyperostosis with tylosis	2206	<10	<10
Annular pancreas	675	286	283
Anoctamin-5-related limb-girdle muscular dystrophy R12	206549	110	107
Anodontia	99797	<10	<10
Anomalous aortic origin of the left coronary artery	541443	32	32
Anomalous aortic origin of the right coronary artery	541454	42	42
Anomalous origin of coronary artery from the pulmonary artery	541507	92	90
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	37	37
Antecubital pterygium syndrome	2987	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Anterior cutaneous nerve entrapment syndrome	51890	<10	<10
Anterior segment developmental anomaly*	88632	17	17
Anterior urethral valve	435372	18	18
Anti-glomerular basement membrane disease	375	126	119
Anti-HLA hyperimmunization	2194	<10	<10
Anti-neutrophil cytoplasmic antibody-associated vasculitis*	156152	896	808
Anti-p200 pemphigoid	454710	50	36
Antiphospholipid syndrome	80	4696	4642
Antisynthetase syndrome	81	1222	1156
Antley-Bixler syndrome	83	19	17
Aorta coarctation	1457	1299	1284
Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome	1110	<10	<10
Aortic arch interruption	2299	153	145
Aorto-ventricular tunnel	3400	<10	<10
Apert syndrome	87	115	108
Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome	1112	<10	<10
Aplasia cutis congenita	1114	77	76
Aplasia cutis congenita-intestinal lymphangiectasia syndrome	1116	<10	<10
Aplasia of lacrimal and salivary glands	86815	<10	<10
Apnea of prematurity	99981	<10	<10
Apodia	294986	<10	<10
Apolipoprotein A-I deficiency	425	<10	<10
Apparent mineralocorticoid excess	320	<10	<10
Aquagenic palmoplantar keratoderma	498359	106	105
Arachnodactyly-abnormal ossification-intellectual disability syndrome	1129	<10	<10
Arachnodactyly-intellectual disability-dysmorphism syndrome	1130	<10	<10
Arachnoid cyst	2356	119	119
Arachnoiditis	137817	<10	<10
Aregenerative anemia	101096	33	32
Argininemia	90	33	29
Argininosuccinic aciduria	23	69	64
Arnold-Chiari malformation type I	268882	1247	1235
Aromatase deficiency	91	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Aromatase excess syndrome	178345	<10	<10
Aromatic L-amino acid decarboxylase deficiency	35708	17	14
Arrhinia-choanal atresia-microphthalmia syndrome	1135	<10	<10
Arrhythmogenic right ventricular cardiomyopathy*	247	871	843
Arterial dissection-lentiginosis syndrome	1682	194	193
Arterial tortuosity syndrome	3342	47	47
Arthrochalasia Ehlers-Danlos syndrome	1899	16	16
Arthrogryposis multiplex congenita*	1037	143	128
Arthrogryposis syndrome*	109007	596	554
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	<10	<10
Arthrogryposis-renal dysfunction-cholestasis syndrome	2697	18	<10
Arthrogryposis-severe scoliosis syndrome	65720	35	33
ARX-related encephalopathy-brain malformation spectrum*	423655	<10	<10
ARX-related epileptic encephalopathy*	182079	15	12
Asbestos intoxication	2302	273	252
Ascher syndrome	1253	<10	<10
Asherman syndrome	137686	<10	<10
Aspartylglucosaminuria	93	<10	<10
Aspergillosis	1163	219	194
Astrocytoma*	94	64	61
Ataxia neuropathy spectrum*	254818	272	262
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	<10	<10
Ataxia-oculomotor apraxia type 1	1168	48	47
Ataxia-oculomotor apraxia type 4	459033	<10	<10
Ataxia-pancytopenia syndrome	2585	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ataxia-tapetoretinal degeneration syndrome	1178	<10	<10
Ataxia-telangiectasia	100	240	203
Ataxia-telangiectasia variant	370109	<10	<10
Ataxia-telangiectasia-like disorder	251347	<10	<10
Atelosteogenesis type I	1190	<10	<10
Atelosteogenesis type III	56305	<10	<10
Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome	1192	<10	<10
Athyreosis	95713	611	609
Atopic keratoconjunctivitis	163934	35	35
Atresia of urethra	105	<10	<10
Atrial septal aneurysm	99107	<10	<10
Atrial septal defect-atrioventricular conduction defects syndrome	1479	<10	<10
Atrial standstill	1344	38	37
Atrichia with papular lesions	86819	<10	<10
Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	1352	<10	<10
Atrioventricular septal defect*	98722	86	86
Atrophic lichen planus	254449	<10	<10
Atrophoderma vermiculata	79100	<10	<10
Attenuated familial adenomatous polyposis	220460	123	122
ATTRV122I amyloidosis	85451	351	319
ATTRV30M amyloidosis	85447	904	825
Atypical autism	199627	2070	2068
Atypical Fanconi syndrome-neonatal hyperinsulinism syndrome	544628	<10	<10
Atypical hemolytic uremic syndrome	2134	682	631
Atypical hypotonia-cystinuria syndrome	238523	<10	<10
Atypical juvenile parkinsonism	391411	<10	<10
Atypical Norrie disease due to Xp11.3 microdeletion	261501	<10	<10
Atypical papilloma of choroid plexus	251902	<10	<10
Atypical Rett syndrome	3095	262	257
Atypical teratoid rhabdoid tumor	99966	<10	<10
Atypical Werner syndrome	79474	<10	<10
Audiogenic seizures	166415	<10	<10
Auditory neuropathy-optic atrophy syndrome	542585	18	17

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome	77300	**35**	**34**
Auriculocondylar syndrome	137888	**17**	**17**
Autism spectrum disorder due to AUTS2 deficiency	352490	**36**	**36**
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	**11**	**11**
Autoimmune bullous skin disease*	79669	**39**	**34**
Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	420789	**<10**	**<10**
Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome	391487	**<10**	**<10**
Autoimmune hemolytic anemia*	98375	**934**	**849**
Autoimmune hemolytic anemia, cold type*	228312	**50**	**46**
Autoimmune hemolytic anemia, warm type	90033	**397**	**360**
Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome	444463	**29**	**28**
Autoimmune hepatitis	2137	**6777**	**6445**
Autoimmune hypoparathyroidism	36913	**37**	**37**
Autoimmune interstitial lung disease-arthritis syndrome	444092	**135**	**125**
Autoimmune limbic encephalitis	623615	**67**	**66**
Autoimmune lymphoproliferative syndrome	3261	**115**	**114**
Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency	436159	**37**	**37**
Autoimmune lymphoproliferative syndrome with recurrent viral infections	275517	**<10**	**<10**
Autoimmune pancreatitis type 2	280315	**124**	**123**
Autoimmune pancreatitis*	103919	**48**	**48**
Autoimmune polyendocrinopathy type 1	3453	**87**	**84**
Autoimmune polyendocrinopathy type 2	3143	**62**	**62**
Autoimmune polyendocrinopathy type 3	227982	**<10**	**<10**
Autoimmune polyendocrinopathy*	282196	**<10**	**<10**
Autoimmune pulmonary alveolar proteinosis	747	**108**	**95**
Autoimmune thrombocytopenia*	71203	**131**	**122**
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	**39**	**36**
Autosomal dominant cerebellar ataxia type I*	94145	**30**	**27**
Autosomal dominant cerebellar ataxia type II*	208508	**39**	**32**
Autosomal dominant cerebellar ataxia type III*	94148	**93**	**75**
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	**15**	**14**
Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons	401964	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2*	64746	**991**	**954**
Autosomal dominant Charcot-Marie-Tooth disease type 2A1	99946	**111**	**106**
Autosomal dominant Charcot-Marie-Tooth disease type 2A2	99947	**93**	**91**
Autosomal dominant Charcot-Marie-Tooth disease type 2B	99936	**18**	**17**
Autosomal dominant Charcot-Marie-Tooth disease type 2C	99937	**28**	**27**
Autosomal dominant Charcot-Marie-Tooth disease type 2D	99938	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2DD	521414	**<10**	**<10**
Autosomal dominant Charcot-Marie-Tooth disease type 2E	99939	**26**	**26**
Autosomal dominant Charcot-Marie-Tooth disease type 2F	99940	**52**	**50**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant Charcot-Marie-Tooth disease type 2G	99941	<10	<10
Autosomal dominant Charcot-Marie-Tooth disease type 2I	99942	32	31
Autosomal dominant Charcot-Marie-Tooth disease type 2J	99943	<10	<10
Autosomal dominant Charcot-Marie-Tooth disease type 2K	99944	33	33
Autosomal dominant Charcot-Marie-Tooth disease type 2L	99945	<10	<10
Autosomal dominant Charcot-Marie-Tooth disease type 2M	228179	<10	<10
Autosomal dominant Charcot-Marie-Tooth disease type 2N	228174	36	36
Autosomal dominant Charcot-Marie-Tooth disease type 2O	284232	<10	<10
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	17	17
Autosomal dominant Charcot-Marie-Tooth disease type 2Y	435387	<10	<10
Autosomal dominant Charcot-Marie-Tooth disease type 2Z	466768	25	23
Autosomal dominant childhood-onset proximal spinal muscular atrophy	363447	41	41
Autosomal dominant congenital benign spinal muscular atrophy	1216	<10	<10
Autosomal dominant cutis laxa	90348	14	13
Autosomal dominant deafness-onychodystrophy syndrome	79499	<10	<10
Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome	476093	11	<10
Autosomal dominant dopa-responsive dystonia	98808	50	49
Autosomal dominant epidermolytic ichthyosis	312	89	88
Autosomal dominant epilepsy with auditory features	101046	12	12
Autosomal dominant focal dystonia, DYT25 type	329466	32	32
Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering	402003	<10	<10
Autosomal dominant generalized dystrophic epidermolysis bullosa	231568	75	75

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form	79399	<10	<10
Autosomal dominant generalized epidermolysis bullosa simplex, severe form	79396	41	40
Autosomal dominant hyper-IgE syndrome	2314	115	111
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	31	31
Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	457193	20	20
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	86	85
Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	100044	12	12
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	11	<10
Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	324585	<10	<10
Autosomal dominant intermediate Charcot-Marie-Tooth disease*	90114	36	34
Autosomal dominant keratitis	2334	<10	<10
Autosomal dominant limb-girdle muscular dystrophy type 1A	266	42	38
Autosomal dominant macrothrombocytopenia	140957	183	182
Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	319581	<10	<10
Autosomal dominant mitochondrial myopathy with exercise intolerance	457050	14	14
Autosomal dominant multiple pterygium syndrome	65743	<10	<10
Autosomal dominant myoglobinuria	99846	<10	<10
Autosomal dominant neovascular inflammatory vitreoretinopathy	329211	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant nocturnal frontal lobe epilepsy	98784	**128**	**123**
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	**45**	**45**
Autosomal dominant optic atrophy*	98672	**448**	**441**
Autosomal dominant optic atrophy, classic form	98673	**401**	**396**
Autosomal dominant osteopetrosis type 1	2783	**25**	**25**
Autosomal dominant otospondylomegaepiphyseal dysplasia	166100	**<10**	**<10**
Autosomal dominant palmoplantar keratoderma and congenital alopecia	1010	**<10**	**<10**
Autosomal dominant polycystic kidney disease	730	**7060**	**6845**
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	**31**	**31**
Autosomal dominant progressive nephropathy with hypertension	88659	**<10**	**<10**
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	**28**	**27**
Autosomal dominant spastic ataxia type 1	251282	**<10**	**<10**
Autosomal dominant spastic paraplegia type 10	100991	**39**	**39**
Autosomal dominant spastic paraplegia type 13	100994	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal dominant spastic paraplegia type 17	100998	**<10**	**<10**
Autosomal dominant spastic paraplegia type 3	100984	**130**	**128**
Autosomal dominant spastic paraplegia type 31	101011	**34**	**34**
Autosomal dominant spastic paraplegia type 4	100985	**356**	**354**
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	**17**	**17**
Autosomal dominant spastic paraplegia type 9A	447753	**<10**	**<10**
Autosomal dominant spastic paraplegia type 9B	447757	**<10**	**<10**
Autosomal dominant spondylocostal dysostosis	1797	**44**	**43**
Autosomal dominant thrombocytopenia with platelet secretion defect	466806	**14**	**14**
Autosomal dominant tubulointerstitial kidney disease	34149	**1084**	**1070**
Autosomal dominant vitreoretinochoroidopathy	3086	**14**	**14**
Autosomal erythropoietic protoporphyria	79278	**373**	**350**
Autosomal recessive amelia	1027	**<10**	**<10**
Autosomal recessive anterior segment dysgenesis	519388	**17**	**17**
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 ubiquinone deficiency	139485	**17**	**17**
Autosomal recessive ataxia, Beauce type	88644	**22**	**22**
Autosomal recessive axonal hereditary motor and sensory neuropathy*	91024	**224**	**215**
Autosomal recessive axonal neuropathy with neuromyotonia	324442	**<10**	**<10**
Autosomal recessive bestrophinopathy	139455	**28**	**28**
Autosomal recessive brachyolmia	448242	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Autosomal recessive centronuclear myopathy	169186	**26**	**24**
Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	453521	**<10**	**<10**
Autosomal recessive cerebellar ataxia due to STUB1 deficiency	412057	**14**	**13**
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 WWOX deficiency	284282	**16**	**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	**23**	**22**
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	**12**	**12**
Autosomal recessive chorioretinopathy-microcephaly syndrome	2518	**<10**	**<10**
Autosomal recessive congenital ichthyosis*	281097	**135**	**135**
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	**35**	**35**
Autosomal recessive epidermolytic ichthyosis	512103	**<10**	**<10**
Autosomal recessive faciodigitogenital syndrome	1974	**19**	**19**
Autosomal recessive frontotemporal pachygyria	329329	**<10**	**<10**
Autosomal recessive generalized dystrophic epidermolysis bullosa, intermediate form	89842	**33**	**32**
Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	79408	**27**	**24**
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	**12**	**12**
Autosomal recessive hypophosphatemic rickets	289176	**18**	**18**
Autosomal recessive infantile hypercalcemia	300547	**155**	**154**
Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	217055	**11**	**11**
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	**170**	**168**
Autosomal recessive isolated optic atrophy	98676	**80**	**80**
Autosomal recessive lethal neonatal axonal sensorimotor polyneuropathy	538096	**<10**	**<10**
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	**50**	**37**
Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	319569	**<10**	**<10**
Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial JAK1 deficiency	574957	**<10**	**<10**
Autosomal recessive multiple pterygium syndrome	2990	**20**	**20**
Autosomal recessive myogenic arthrogryposis multiplex congenita	319332	**<10**	**<10**
Autosomal recessive optic atrophy, OPA7 type	227976	**<10**	**<10**
Autosomal recessive palmoplantar keratoderma and congenital alopecia	1366	**<10**	**<10**
Autosomal recessive polycystic kidney disease	731	**465**	**410**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
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 severe congenital neutropenia due to G6PC3 deficiency	331176	<10	<10
Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency	423384	<10	<10
Autosomal recessive sideroblastic anemia	260305	<10	<10
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	98	97	95
Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome	254343	<10	<10
Autosomal recessive spastic paraplegia type 11	2822	69	68
Autosomal recessive spastic paraplegia type 14	100995	<10	<10
Autosomal recessive spastic paraplegia type 15	100996	11	11
Autosomal recessive spastic paraplegia type 20	101000	31	31
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	11	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	<10	<10
Autosomal recessive spastic paraplegia type 5A	100986	21	21
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 76	488594	11	11
Autosomal recessive spastic paraplegia type 78	513436	<10	<10
Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome	95433	<10	<10
Autosomal recessive spondylocostal dysostosis	2311	56	49
Autosomal semi-dominant severe lipodystrophic laminopathy	280365	15	14
Autosomal spastic paraplegia type 18	209951	<10	<10
Autosomal spastic paraplegia type 30	101010	39	39
Autosomal spastic paraplegia type 58	397946	<10	<10
Autosomal spastic paraplegia type 72	401849	<10	<10
Autosomal systemic lupus erythematosus	300345	57	55
Axenfeld anomaly	98978	48	46
Axenfeld-Rieger syndrome	782	159	157
Axial mesodermal dysplasia spectrum	1834	<10	<10
Axial spondylometaphyseal dysplasia	168549	<10	<10
Axonal polyneuropathy associated with IgG/IgM/IgA monoclonal gammopathy	209004	178	146
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	15	15
Bacterial susceptibility due to TLR signaling pathway deficiency	183713	<10	<10
Bacterial toxic-shock syndrome	36234	<10	<10
Bainbridge-Ropers syndrome	352577	40	40

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Balantidiasis	1223	**<10**	**<10**
Balint syndrome	363746	**<10**	**<10**
Baller-Gerold syndrome	1225	**71**	**71**
Baló concentric sclerosis	228165	**<10**	**<10**
Bamforth-Lazarus syndrome	1226	**<10**	**<10**
Bangstad syndrome	1227	**<10**	**<10**
Bannayan-Riley-Ruvalcaba syndrome	109	**39**	**39**
BAP1-related tumor predisposition syndrome	289539	**24**	**23**
Baraitser-Winter cerebrofrontofacial syndrome	2995	**63**	**62**
Barber-Say syndrome	1231	**<10**	**<10**
Bardet-Biedl syndrome	110	**664**	**647**
Baroreflex failure	443084	**<10**	**<10**
Barth syndrome	111	**53**	**38**
Bartsocas-Papas syndrome	1234	**<10**	**<10**
Bartter syndrome	112	**322**	**317**
Basel-Vanagaite-Smirin-Yosef syndrome	464738	**<10**	**<10**
Bathing suit ichthyosis	100976	**<10**	**<10**
Bazex-Dupré-Christol syndrome	113	**14**	**14**
B-cell chronic lymphocytic leukemia	67038	**25**	**23**
B-cell immunodeficiency-limb anomaly-urogenital malformation syndrome	567502	**<10**	**<10**
Becker muscular dystrophy	98895	**1282**	**1174**
Becker nevus syndrome	64755	**36**	**36**
Beckwith-Wiedemann syndrome	116	**1437**	**1408**
Beemer-Ertbruggen syndrome	1237	**<10**	**<10**
Behavioral variant of frontotemporal dementia	275864	**1018**	**919**
Behçet disease	117	**3176**	**3147**
Benign adult familial myoclonic epilepsy	86814	**22**	**22**
Benign cephalic histiocytosis	157997	**<10**	**<10**
Benign epithelial tumor of salivary glands	276148	**<10**	**<10**
Benign familial infantile epilepsy	306	**103**	**103**
Benign familial mesial temporal lobe epilepsy	163717	**38**	**38**
Benign familial neonatal epilepsy	1949	**119**	**119**
Benign familial neonatal-infantile seizures	140927	**52**	**51**
Benign focal seizures of adolescence	1544	**51**	**51**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Benign hereditary chorea	1429	**81**	**78**
Benign idiopathic neonatal seizures	64545	**40**	**39**
Benign infantile focal epilepsy with midline spikes and waves during sleep	166308	**68**	**67**
Benign infantile seizures associated with mild gastroenteritis	166305	**<10**	**<10**
Benign nocturnal alternating hemiplegia of childhood	209973	**<10**	**<10**
Benign non-familial infantile seizures*	166295	**<10**	**<10**
Benign occipital epilepsy	25968	**208**	**207**
Benign paroxysmal tonic upgaze of childhood with ataxia	1179	**<10**	**<10**
Benign paroxysmal torticollis of infancy	71518	**20**	**20**
Benign partial epilepsy of infancy with complex partial seizures	166299	**52**	**52**
Benign partial epilepsy with secondarily generalized seizures in infancy	166302	**27**	**27**
Benign partial infantile seizures*	166311	**27**	**27**
Benign recurrent intrahepatic cholestasis	65682	**102**	**100**
Benign Samaritan congenital myopathy	324581	**<10**	**<10**
Benign schwannoma	252164	**94**	**91**
Benign tumor of fallopian tubes	180237	**<10**	**<10**
BENTA disease	464336	**<10**	**<10**
Bernard-Soulier syndrome	274	**129**	**129**
Best vitelliform macular dystrophy	1243	**420**	**418**
Beta-ketothiolase deficiency	134	**62**	**62**
Beta-mannosidosis	118	**<10**	**<10**
Beta-propeller protein-associated neurodegeneration	329284	**36**	**36**
Beta-sarcoglycan-related limb-girdle muscular dystrophy R4	119	**40**	**38**
Beta-thalassemia	848	**1353**	**1334**
Beta-thalassemia-X-linked thrombocytopenia syndrome	231393	**<10**	**<10**
Bethlem myopathy	610	**223**	**213**
Bickerstaff brainstem encephalitis	79138	**33**	**32**
Bicornuate uterus*	180134	**53**	**53**
Bietti crystalline dystrophy	41751	**24**	**24**
Bifid nose	2695	**<10**	**<10**
Bifid uvula	99771	**150**	**150**
Bifunctional enzyme deficiency	300	**11**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Bilateral microtia-deafness-cleft palate syndrome	140963	<10	<10
Bilateral polymicrogyria	268940	208	202
Bilateral striopallidodentate calcinosis	1980	193	188
Biliary atresia with splenic malformation syndrome	244283	32	30
Bilirubin encephalopathy*	415286	16	15
Biotinidase deficiency	79241	48	48
Biotin-thiamine-responsive basal ganglia disease	65284	14	13
Birdshot chorioretinopathy	179	920	886
Birt-Hogg-Dubé syndrome	122	539	531
Björnstad syndrome	123	<10	<10
Blake pouch cyst	98922	35	35
Blau syndrome	90340	76	75
Bleeding diathesis due to a collagen receptor defect	73271	16	16
Bleeding diathesis due to thromboxane synthesis deficiency	220443	18	18
Bleeding disorder due to CalDAG-GEFI deficiency	420566	<10	<10
Bleeding disorder due to P2Y12 defect	36355	43	43
Blepharo-cheilo-odontic syndrome	1997	25	24
Blepharophimosis-intellectual disability syndrome*	293642	<10	<10
Blepharophimosis-intellectual disability syndrome, MKB type	293707	<10	<10
Blepharophimosis-intellectual disability syndrome, Ohdo type	2728	39	37
Blepharophimosis-intellectual disability syndrome, SBBYS type	3047	19	19
Blepharophimosis-ptosis-epicanthus inversus syndrome	126	220	220
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
Blepharospasm-oromandibular dystonia syndrome	93964	173	172
Blomstrand lethal chondrodysplasia	50945	<10	<10
Bloom syndrome	125	16	13
Blount disease	2768	34	34
Blue cone monochromatism	16	97	97
Blue diaper syndrome	94086	<10	<10
Blue rubber bleb nevus	1059	99	99

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	91135	<10	<10
Bohring-Opitz syndrome	97297	25	23
Bone sarcoma*	223727	<10	<10
BOR syndrome	107	501	496
Borjeson-Forssman-Lehmann syndrome	127	28	28
Bosley-Salih-Alorainy syndrome	69737	<10	<10
Bothnia retinal dystrophy	85128	<10	<10
Botulism	1267	11	11
Bowen-Conradi syndrome	1270	<10	<10
Brachydactyly type A1	93388	44	44
Brachydactyly type A2	93396	25	24
Brachydactyly type A4	93394	17	17
Brachydactyly type B	93383	51	51
Brachydactyly type C	93384	94	93
Brachydactyly type E	93387	60	60
Brachydactyly-arterial hypertension syndrome	1276	18	18
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	38	38
Brachyolmia, Maroteaux type	93302	<10	<10
Brachyolmia-amelogenesis imperfecta syndrome	2899	<10	<10
Brachytelephalangic chondrodysplasia punctata	79345	27	26
Brachytelephalangy-dysmorphism-Kallmann syndrome	1295	<10	<10
Braddock syndrome	52047	<10	<10
Bradyopsia	75374	<10	<10
Brain calcification, Rajab type	178506	<10	<10
Brain malformation-congenital heart disease-postaxial polydactyly syndrome	75389	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	500150	**19**	**19**
Brain-lung-thyroid syndrome	209905	**35**	**34**
Branchiogenic deafness syndrome	50815	**22**	**22**
Branchio-oculo-facial syndrome	1297	**68**	**67**
Branchiootic syndrome	52429	**20**	**20**
Branchioskeletogenital syndrome	1299	**<10**	**<10**
BRESEK syndrome	85284	**<10**	**<10**
Brittle cornea syndrome	90354	**<10**	**<10**
Brody myopathy	53347	**19**	**19**
Bronchial neuroendocrine tumor	97287	**46**	**39**
Bronchiolitis obliterans with obstructive pulmonary disease	1303	**709**	**688**
Bronchogenic cyst	2357	**85**	**85**
Bronchopulmonary dysplasia	70589	**1833**	**1813**
Brooke-Spiegler syndrome	79493	**99**	**98**
Brucellosis	1304	**<10**	**<10**
Bruck syndrome	2771	**25**	**24**
Brugada syndrome	130	**3054**	**3023**
Budd-Chiari syndrome	131	**730**	**684**
Buerger disease	36258	**442**	**429**
Bulbospinal muscular atrophy of adult*	206707	**75**	**66**
Bulbospinal muscular atrophy of childhood*	206704	**<10**	**<10**
Bullous impetigo	36237	**<10**	**<10**
Bullous lichen planus	33408	**<10**	**<10**
Bullous pemphigoid	703	**3433**	**2387**
Burkitt lymphoma	543	**22**	**19**
Burning mouth syndrome	353253	**15**	**15**
Burn-McKeown syndrome	1200	**<10**	**<10**
Butterfly-shaped pigment dystrophy	99001	**11**	**11**
Butyrylcholinesterase deficiency	132	**24**	**24**
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	**97**	**86**
CAD-CDG	448010	**<10**	**<10**
CADDS	369942	**<10**	**<10**
Caffey disease	1310	**32**	**31**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Calciphylaxis	280062	**<10**	**<10**
Calpain-3-related limb-girdle muscular dystrophy D4	565909	**<10**	**<10**
Calpain-3-related limb-girdle muscular dystrophy R1	267	**485**	**463**
CAMOS syndrome	83472	**<10**	**<10**
Campomelia, Cumming type	1318	**<10**	**<10**
Campomelic dysplasia	140	**27**	**23**
Campomelic dysplasia and related disorders*	93439	**12**	**12**
Camptobrachydactyly	1319	**14**	**14**
Camptodactyly of fingers	295016	**70**	**70**
Camptodactyly syndrome, Guadalajara type 1	1327	**<10**	**<10**
Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	2848	**14**	**14**
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	**45**	**45**
Canavan disease	141	**29**	**23**
Cancer-associated retinopathy	71505	**26**	**25**
CANOMAD syndrome	71279	**108**	**97**
Cantú syndrome	1517	**22**	**21**
Cap myopathy	171881	**20**	**18**
Cap polyposis	160148	**<10**	**<10**
Capillary malformation-arteriovenous malformation	137667	**888**	**884**
Carbamoyl-phosphate synthetase 1 deficiency	147	**54**	**37**
Carcinoid syndrome	100093	**<10**	**<10**
Cardiac anomalies-heterotaxy syndrome	137628	**84**	**72**
Cardiac diverticulum	1686	**<10**	**<10**
Cardiac-valvular Ehlers-Danlos syndrome	230851	**<10**	**<10**
Cardiocranial syndrome, Pfeiffer type	2872	**15**	**15**
Cardiofaciocutaneous syndrome	1340	**259**	**238**
Cardiogenic shock	97292	**<10**	**<10**
Cardiomyopathy-cataract-hip spine disease syndrome	1345	**<10**	**<10**
Cardiomyopathy-hypotonia-lactic acidosis syndrome	91130	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Cardiospondylocarpofacial syndrome	3238	**11**	**11**
Carey-Fineman-Ziter syndrome	1358	**16**	**15**
Caribbean parkinsonism	97355	**<10**	**<10**
Carney complex	1359	**160**	**158**
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	**16**	**16**
Carnitine palmitoyltransferase II deficiency	157	**157**	**152**
Carnitine-acylcarnitine translocase deficiency	159	**19**	**12**
Carnosinase deficiency	1361	**<10**	**<10**
Caroli disease	53035	**155**	**142**
Caroli syndrome	480520	**27**	**27**
Carpenter syndrome	65759	**<10**	**<10**
Carpotarsal osteochondromatosis	2767	**<10**	**<10**
Cartilage-hair hypoplasia	175	**62**	**59**
Carvajal syndrome	65282	**<10**	**<10**
Castleman disease	160	**378**	**363**
Cataract-congenital heart disease-neural tube defect syndrome	314993	**<10**	**<10**
Cataract-glaucoma syndrome	162	**50**	**50**
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	**26**	**26**
Cataract-nephropathy-encephalopathy syndrome	1380	**<10**	**<10**
Catastrophic antiphospholipid syndrome	464343	**176**	**170**
Catecholaminergic polymorphic ventricular tachycardia	3286	**378**	**370**
Catel-Manzke syndrome	1388	**<10**	**<10**
Cat-eye syndrome	195	**87**	**86**
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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Caudal regression syndrome	3027	**112**	**110**
Caudal regression-sirenomelia spectrum*	444941	**<10**	**<10**
CCDC115-CDG	468684	**<10**	**<10**
CDK13-related congenital heart defects-intellectual disability-facial dysmorphism syndrome	646278	**<10**	**<10**
CDKL5-deficiency disorder	505652	**63**	**61**
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	**11**	**11**
Cenani-Lenz syndrome	3258	**<10**	**<10**
Central areolar choroidal dystrophy	75377	**33**	**32**
Central cloudy dystrophy of François	98972	**<10**	**<10**
Central congenital hypothyroidism*	226298	**88**	**88**
Central core disease	597	**383**	**369**
Central diabetes insipidus	178029	**1049**	**1012**
Central nervous system calcification-deafness-tubular acidosis-anemia syndrome	3240	**<10**	**<10**
Central nervous system embryonal tumor*	251870	**<10**	**<10**
Central neurocytoma	73256	**<10**	**<10**
Central polydactyly	295004	**<10**	**<10**
Central retinal vein occlusion	411527	**128**	**127**
Central serous chorioretinopathy	443079	**27**	**27**
Centronuclear myopathy*	595	**138**	**129**
Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome	504476	**609**	**591**
Cerebellar ataxia, Cayman type	94122	**<10**	**<10**
Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome	1171	**32**	**31**
Cerebellar ataxia-ectodermal dysplasia syndrome	1174	**<10**	**<10**
Cerebellar ataxia-hypogonadism syndrome	1173	**<10**	**<10**
Cerebellar hypoplasia-tapetoretinal degeneration syndrome	2246	**<10**	**<10**
Cerebellar-facial-dental syndrome	444072	**<10**	**<10**
Cerebral arteriovenous malformation	46724	**971**	**957**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	136	**668**	**631**
Cerebral autosomal recessive arteriopathy-subcortical infarcts-leukoencephalopathy	199354	**<10**	**<10**
Cerebral cortical dysplasia*	268950	**334**	**331**
Cerebral sinovenous thrombosis	329217	**553**	**541**
Cerebral visual impairment	447788	**<10**	**<10**
Cerebrocostomandibular syndrome	1393	**15**	**13**
Cerebrofacioarticular syndrome	314679	**<10**	**<10**
Cerebrofaciothoracic dysplasia	1394	**<10**	**<10**
Cerebrooculonasal syndrome	66625	**<10**	**<10**
Cerebrotendinous xanthomatosis	909	**68**	**64**
Cernunnos-XLF deficiency	169079	**<10**	**<10**
Cervical aortic arch	99079	**<10**	**<10**
Cervical dermoid cyst	141046	**150**	**150**
Cervical hypertrichosis-peripheral neuropathy syndrome	2218	**<10**	**<10**
Cervicofacial fibrochondroma	141067	**474**	**474**
Char syndrome	46627	**22**	**22**
Charcot-Marie-Tooth disease type 1*	65753	**1185**	**1168**
Charcot-Marie-Tooth disease type 1A	101081	**3158**	**3094**
Charcot-Marie-Tooth disease type 1B	101082	**247**	**237**
Charcot-Marie-Tooth disease type 1C	101083	**61**	**61**
Charcot-Marie-Tooth disease type 1D	101084	**<10**	**<10**
Charcot-Marie-Tooth disease type 1E	90658	**17**	**16**
Charcot-Marie-Tooth disease type 1F	101085	**15**	**15**
Charcot-Marie-Tooth disease type 2B1	98856	**29**	**28**
Charcot-Marie-Tooth disease type 2B2	101101	**21**	**20**
Charcot-Marie-Tooth disease type 2B5	228374	**<10**	**<10**
Charcot-Marie-Tooth disease type 2H	101102	**22**	**19**
Charcot-Marie-Tooth disease type 2P	300319	**16**	**16**
Charcot-Marie-Tooth disease type 2R	397968	**<10**	**<10**
Charcot-Marie-Tooth disease type 2S	443073	**17**	**17**
Charcot-Marie-Tooth disease type 2T	495274	**<10**	**<10**
Charcot-Marie-Tooth disease type 4*	64749	**101**	**99**
Charcot-Marie-Tooth disease type 4A	99948	**30**	**29**
Charcot-Marie-Tooth disease type 4B1	99955	**<10**	**<10**
Charcot-Marie-Tooth disease type 4B2	99956	**<10**	**<10**
Charcot-Marie-Tooth disease type 4C	99949	**97**	**91**
Charcot-Marie-Tooth disease type 4D	99950	**20**	**20**
Charcot-Marie-Tooth disease type 4F	99952	**50**	**49**
Charcot-Marie-Tooth disease type 4G	99953	**25**	**24**
Charcot-Marie-Tooth disease type 4H	99954	**<10**	**<10**
Charcot-Marie-Tooth disease type 4J	139515	**18**	**18**
Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome	90103	**<10**	**<10**
CHARGE syndrome	138	**712**	**625**
CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	599082	**26**	**26**
CHD8 overgrowth syndrome	642675	**<10**	**<10**
Chédiak-Higashi syndrome	167	**26**	**25**
Cheilitis glandularis	1221	**<10**	**<10**
Cheirospondyloenchondromatosis	99647	**<10**	**<10**
Cherubism	184	**41**	**40**
Chilblain lupus	90280	**24**	**24**
CHILD syndrome	139	**12**	**12**
Childhood absence epilepsy	64280	**723**	**721**
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	**<10**	**<10**
Childhood-onset spasticity with hyperglycinemia	401866	**<10**	**<10**
CHIME syndrome	3474	**<10**	**<10**
Choanal atresia	137914	**284**	**284**
Choanal atresia-athelia-hypothyroidism-delayed puberty-short stature syndrome	589856	**<10**	**<10**
Cholangiocarcinoma	70567	**13**	**<10**
Choledochal cyst	480501	**207**	**206**
Cholestasis-lymphedema syndrome	1414	**<10**	**<10**
Cholestasis-pigmentary retinopathy-cleft palate syndrome	1415	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Chondrodysplasia punctata*	93442	68	66
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	11	11
Chordoid glioma	251674	<10	<10
Chordoma	178	17	15
Choreoacanthocytosis	2388	13	12
Choriocarcinoma of the central nervous system	252015	<10	<10
Choroid plexus carcinoma	251899	<10	<10
Choroid plexus tumor*	251896	<10	<10
Choroideremia	180	212	211
Christianson syndrome	85278	30	30
Chromophobe renal cell carcinoma	319303	35	35
Chronic acquired demyelinating polyneuropathy*	208974	371	346
Chronic actinic dermatitis	330064	<10	<10
Chronic beryllium disease	133	15	14
Chronic bilirubin encephalopathy	529808	<10	<10
Chronic cutaneous lupus erythematosus*	163531	221	215
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	49	45
Chronic granulomatous disease	379	292	272
Chronic inflammatory demyelinating polyneuropathy	2932	3877	3479
Chronic intervillositis of unknown etiology	615970	<10	<10
Chronic intestinal failure	294422	194	186
Chronic intestinal pseudoobstruction	2978	434	400
Chronic lymphoproliferative disorder of natural killer cells	512017	11	11
Chronic mucocutaneous candidiasis	1334	29	28
Chronic myeloid leukemia	521	55	53
Chronic myelomonocytic leukemia	98823	11	<10
Chronic neurovisceral acid sphingomyelinase deficiency	618891	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Chronic nonbacterial osteomyelitis/ Chronic recurrent multifocal osteomyelitis	324964	766	766
Chronic pneumonitis of infancy	91359	118	117
Chronic polyradiculoneuropathy*	208978	1274	1180
Chronic relapsing inflammatory optic neuropathy	499085	54	54
Chronic respiratory distress with surfactant metabolism deficiency	217566	<10	<10
Chronic thromboembolic pulmonary hypertension	70591	2761	2556
Chronic visceral acid sphingomyelinase deficiency	77293	149	139
CHST3-related skeletal dysplasia	263463	<10	<10
Chudley-McCullough syndrome	314597	<10	<10
Chuvash erythrocytosis	238557	<10	<10
Chylomicron retention disease	71	24	24
Chylous ascites	1160	17	17
CIDEC-related familial partial lipodystrophy	435651	<10	<10
CINCA syndrome	1451	45	45
Cirrhotic cardiomyopathy	57777	<10	<10
Citrullinemia type I	247525	84	73
Citrullinemia type II	247585	<10	<10
CLAPO syndrome	168984	<10	<10
Clark-Baraitser syndrome	600731	<10	<10
Class I glucose-6-phosphate dehydrogenase deficiency	466026	480	480
Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	90794	2161	2140
Classic eosinophilic pustular folliculitis	617408	<10	<10
Classic galactosemia	79239	279	277
Classic glucose transporter type 1 deficiency syndrome	71277	225	223
Classic hairy cell leukemia	58017	<10	<10
Classic Hodgkin lymphoma	391	45	42
Classic homocystinuria	394	220	212
Classic lissencephaly*	102009	72	65
Classic mycosis fungoides	2584	18	15
Classical Ehlers-Danlos syndrome	287	483	480
Classical-like Ehlers-Danlos syndrome type 1	230839	33	33
CLCN4-related X-linked intellectual disability syndrome	485350	47	47
Clear cell renal carcinoma	319276	195	181
Clear cell sarcoma of kidney	457246	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Cleft hard palate	101023	**561**	**559**
Cleft lip and alveolus	141291	**920**	**918**
Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-hearing loss syndrome	508476	**<10**	**<10**
Cleft lip with or without cleft palate*	1991	**2468**	**2458**
Cleft lip/palate	199306	**2600**	**2600**
Cleft lip/palate-deafness-sacral lipoma syndrome	2003	**12**	**12**
Cleft lip/palate-ectodermal dysplasia syndrome	3253	**<10**	**<10**
Cleft lip/palate-intestinal malrotation-cardiopathy syndrome	2001	**<10**	**<10**
Cleft lip-retinopathy syndrome	1995	**<10**	**<10**
Cleft mitral valve	95465	**57**	**57**
Cleft palate*	2014	**4264**	**4246**
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	**1296**	**1294**
Cleidocranial dysplasia	1452	**324**	**323**
Climatic droplet keratopathy	98958	**<10**	**<10**
CLIPPERS	284448	**<10**	**<10**
CLOVES syndrome	140944	**377**	**377**
CNTNAP2-related developmental and epileptic encephalopathy	163681	**1720**	**1714**
COASY protein-associated neurodegeneration	397725	**<10**	**<10**
Coats disease	190	**284**	**282**
Coats plus syndrome	313838	**17**	**17**
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	**19**	**19**
Cochleovestibular malformation	502305	**13**	**13**
Cockayne syndrome	191	**76**	**49**
CODAS syndrome	1458	**<10**	**<10**
Coenzyme Q10 deficiency*	35656	**24**	**21**
Coffin-Lowry syndrome	192	**150**	**148**
Coffin-Siris syndrome	1465	**364**	**359**
COG1-CDG	263508	**<10**	**<10**
COG2-CDG	435934	**<10**	**<10**
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	**100**	**98**
Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	444077	**<10**	**<10**
Cohen syndrome	193	**98**	**96**
COL4A1-related familial vascular leukoencephalopathy	36383	**159**	**157**
Colchicine poisoning	31824	**<10**	**<10**
Cold agglutinin disease	56425	**253**	**236**
Cole-Carpenter syndrome	2050	**<10**	**<10**
Collagen type III glomerulopathy	84087	**<10**	**<10**
Coloboma of choroid and retina	98942	**300**	**297**
Coloboma of eye lens	98943	**<10**	**<10**
Coloboma of eyelid	98946	**15**	**15**
Coloboma of inferior eyelid	155889	**<10**	**<10**
Coloboma of iris	98944	**192**	**189**
Coloboma of macula	98945	**<10**	**<10**
Coloboma of macula-brachydactyly type B syndrome	1471	**<10**	**<10**
Coloboma of optic disc	98947	**81**	**81**
Coloboma of superior eyelid	155884	**12**	**12**
Colobomatous microphthalmia	98938	**276**	**275**
Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	435930	**<10**	**<10**
Colonic atresia	1198	**16**	**16**
Combined deficiency of factor V and factor VIII	35909	**36**	**35**
Combined deficiency of factor VII and factor X	600691	**<10**	**<10**
Combined hamartoma of the retina and retinal pigment epithelium	440727	**<10**	**<10**
Combined immunodeficiency due to CARMIL2 deficiency	542301	**<10**	**<10**
Combined immunodeficiency due to CD27 deficiency	238505	**<10**	**<10**
Combined immunodeficiency due to CD3gamma deficiency	169082	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Combined immunodeficiency due to CD70 deficiency	538958	**<10**	**<10**
Combined immunodeficiency due to CRAC channel dysfunction	169090	**<10**	**<10**
Combined immunodeficiency due to DOCK8 deficiency	217390	**16**	**15**
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	**11**	**11**
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 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	**179**	**174**
Combined immunodeficiency-enteropathy spectrum	436252	**32**	**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**
Combined oxidative phosphorylation defect type 20	420728	**<10**	**<10**
Combined oxidative phosphorylation defect type 23	444013	**<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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Combined pituitary hormone deficiencies, genetic forms	95494	**121**	**119**
Combined pulmonary fibrosis-emphysema syndrome	300564	**568**	**450**
Combined T and B cell immunodeficiency*	101972	**<10**	**<10**
Commissural lip fistula	141061	**<10**	**<10**
Common cystic lymphatic malformation*	458833	**285**	**284**
Common variable immunodeficiency	1572	**2912**	**2847**
Complement component 3 deficiency	280133	**12**	**12**
Complement hyperactivation-angiopathic thrombosis-protein-losing enteropathy syndrome	566175	**<10**	**<10**
Complete androgen insensitivity syndrome	99429	**244**	**244**
Complete atrioventricular septal defect	1329	**952**	**904**
Complete septate uterus	180126	**23**	**23**
Complex hereditary spastic paraplegia*	102013	**150**	**146**
Complex lethal osteochondrodysplasia	457378	**<10**	**<10**
Complex regional pain syndrome	83452	**407**	**407**
Complication after organ transplantation	306644	**161**	**158**
Complication in hemodialysis	268316	**<10**	**<10**
Complications after hematopoietic stem cell transplantation	90053	**11**	**11**
Composite hemangioendothelioma	458758	**<10**	**<10**
Composite lymphoma	168966	**<10**	**<10**
Conductive deafness-malformed external ear syndrome	3216	**61**	**61**
Conductive deafness-ptosis-skeletal anomalies syndrome	3236	**<10**	**<10**
Cone dystrophy with supernormal rod response	209932	**13**	**13**
Cone rod dystrophy	1872	**1018**	**1011**
Congenital abducens nerve palsy	440233	**<10**	**<10**
Congenital absence of both forearm and hand	294979	**87**	**86**
Congenital absence of both lower leg and foot	294981	**<10**	**<10**
Congenital absence of upper arm and forearm with hand present	294975	**<10**	**<10**
Congenital absence/hypoplasia of fingers excluding thumb, unilateral	973	**68**	**68**
Congenital achiasma	324353	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	90795	**116**	**114**
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	90793	**38**	**37**
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	90791	**45**	**44**
Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	95699	**19**	**19**
Congenital adrenal hyperplasia*	418	**574**	**571**
Congenital agenesis of the scrotum	495879	**<10**	**<10**
Congenital alpha2-antiplasmin deficiency	79	**<10**	**<10**
Congenital alveolar capillary dysplasia	210122	**12**	**<10**
Congenital amegakaryocytic thrombocytopenia	3319	**19**	**18**
Congenital amyoplasia	488586	**16**	**16**
Congenital aortic valve stenosis	3093	**299**	**287**
Congenital aortopulmonary window	2037	**27**	**27**
Congenital atransferrinemia	1195	**13**	**13**
Congenital axonal neuropathy with encephalopathy	538101	**<10**	**<10**
Congenital bilateral absence of vas deferens	48	**152**	**152**
Congenital bile acid synthesis defect type 1	79301	**<10**	**<10**
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 cataract microcornea with corneal opacity	289499	**<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	**21**	**18**
Congenital central hypoventilation syndrome	661	**191**	**172**
Congenital cervical spinal stenosis	831	**15**	**14**
Congenital chloride diarrhea	53689	**19**	**17**
Congenital chronic diarrhea with protein-losing enteropathy	329242	**31**	**30**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital chylothorax	264688	**60**	**57**
Congenital contractural arachnodactyly	115	**99**	**99**
Congenital cornea plana	53691	**<10**	**<10**
Congenital coronary artery aneurysm	95491	**13**	**13**
Congenital cystic eye	519384	**<10**	**<10**
Congenital diaphragmatic hernia	2140	**1953**	**1758**
Congenital dyserythropoietic anemia type I	98869	**22**	**21**
Congenital dyserythropoietic anemia type II	98873	**36**	**35**
Congenital dyserythropoietic anemia type III	98870	**<10**	**<10**
Congenital dyserythropoietic anemia type IV	293825	**<10**	**<10**
Congenital dyserythropoietic anemia*	85	**16**	**15**
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 erythropoietic porphyria	79277	**44**	**34**
Congenital esophageal diverticulum	91358	**<10**	**<10**
Congenital esophageal stenosis	645749	**<10**	**<10**
Congenital eyelid retraction	99176	**<10**	**<10**
Congenital factor II deficiency	325	**51**	**51**
Congenital factor V deficiency	326	**594**	**590**
Congenital factor VII deficiency	327	**2233**	**2212**
Congenital factor X deficiency	328	**311**	**309**
Congenital factor XI deficiency	329	**1590**	**1566**
Congenital factor XII deficiency	330	**431**	**424**
Congenital factor XIII deficiency	331	**68**	**68**
Congenital fiber-type disproportion myopathy	2020	**177**	**168**
Congenital fibrinogen deficiency	335	**640**	**635**
Congenital fibrosis of extraocular muscles	45358	**60**	**60**
Congenital generalized lipodystrophy	528	**90**	**87**
Congenital Gerbode defect	99095	**<10**	**<10**
Congenital glaucoma	98976	**609**	**605**
Congenital glucokinase-related hyperinsulinism	79299	**<10**	**<10**
Congenital heart block	60041	**368**	**367**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital heart defect-round face-developmental delay syndrome	1355	**<10**	**<10**
Congenital hemangioma*	458775	**309**	**309**
Congenital hereditary endothelial dystrophy type I	98975	**15**	**15**
Congenital hereditary endothelial dystrophy type II	293603	**<10**	**<10**
Congenital hereditary facial paralysis-variable hearing loss syndrome	306530	**<10**	**<10**
Congenital herpes simplex virus infection	293	**<10**	**<10**
Congenital Horner syndrome	91413	**34**	**34**
Congenital hydrocephalus	2185	**145**	**140**
Congenital hyperinsulinism due to HNF4A deficiency	263455	**<10**	**<10**
Congenital hypoplasia of thumb	294988	**60**	**60**
Congenital hypothyroidism due to maternal intake of antithyroid drugs	226313	**<10**	**<10**
Congenital hypothyroidism due to transplacental passage of TSH-binding inhibitory antibodies	95715	**30**	**30**
Congenital ichthyosiform erythroderma	79394	**153**	**152**
Congenital ichthyosis-microcephalus-tetraplegia syndrome	2271	**<10**	**<10**
Congenital infiltrating lipomatosis of the face	583097	**<10**	**<10**
Congenital insensitivity to pain with severe intellectual disability	453510	**<10**	**<10**
Congenital insensitivity to pain-anosmia-neuropathic arthropathy	88642	**32**	**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	**25**	**25**
Congenital isolated ACTH deficiency	199296	**147**	**143**
Congenital isolated hyperinsulinism*	657	**596**	**592**
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	**37**	**36**
Congenital laryngeal palsy	137932	**137**	**137**
Congenital laryngeal web	2374	**17**	**17**
Congenital laryngomalacia	2373	**1178**	**1175**
Congenital left ventricular aneurysm	1055	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
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	**122**	**121**
Congenital macroglossia	2430	**139**	**139**
Congenital megacalycosis	93109	**17**	**17**
Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	69063	**11**	**<10**
Congenital mesoblastic nephroma	2665	**18**	**18**
Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome	391376	**11**	**<10**
Congenital microcoria	566	**18**	**17**
Congenital microgastria	199293	**<10**	**<10**
Congenital mitral stenosis	99057	**41**	**41**
Congenital muscular alpha-dystroglycanopathy with brain and eye anomalies*	352687	**23**	**21**
Congenital muscular dystrophy due to LMNA mutation	157973	**67**	**59**
Congenital muscular dystrophy type 1B	98893	**<10**	**<10**
Congenital muscular dystrophy with cerebellar involvement	370959	**<10**	**<10**
Congenital muscular dystrophy with hyperlaxity	371007	**28**	**28**
Congenital muscular dystrophy with intellectual disability	370968	**28**	**26**
Congenital muscular dystrophy with intellectual disability and severe epilepsy	329178	**<10**	**<10**
Congenital muscular dystrophy without intellectual disability	370980	**47**	**44**
Congenital muscular dystrophy, Fukuyama type	272	**<10**	**<10**
Congenital muscular dystrophy, Ullrich type	75840	**212**	**198**
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	**633**	**605**
Congenital myopathy with cores*	172976	**125**	**118**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital myopathy with excess of thin filaments	98904	**<10**	**<10**
Congenital myopathy with internal nuclei and atypical cores	319160	**<10**	**<10**
Congenital myopathy with myasthenic-like onset	424107	**18**	**16**
Congenital myopathy with reduced type 2 muscle fibers	544602	**14**	**13**
Congenital myopathy, Paradas type	199329	**<10**	**<10**
Congenital myotonia*	206973	**181**	**178**
Congenital nemaline myopathy*	457074	**20**	**20**
Congenital nephrotic syndrome, Finnish type	839	**99**	**87**
Congenital neuronal ceroid lipofuscinosis	168486	**<10**	**<10**
Congenital neutropenia-myelofibrosis-nephromegaly syndrome	369852	**<10**	**<10**
Congenital oculomotor nerve palsy	440221	**25**	**25**
Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome	2772	**<10**	**<10**
Congenital pancreatic cyst	313906	**105**	**104**
Congenital panfollicular nevus	139414	**13**	**13**
Congenital partial pulmonary venous return anomaly	99124	**274**	**271**
Congenital patent ductus arteriosus aneurysm	99072	**<10**	**<10**
Congenital plasminogen activator inhibitor type 1 deficiency	465	**14**	**14**
Congenital portosystemic shunt	480531	**223**	**220**
Congenital prekallikrein deficiency	749	**<10**	**<10**
Congenital primary aphakia	83461	**<10**	**<10**
Congenital primary lymphedema of Gordon	569821	**<10**	**<10**
Congenital primary megaureter	617	**1157**	**1149**
Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome	508542	**<10**	**<10**
Congenital pseudoarthrosis of the clavicle	66630	**<10**	**<10**
Congenital pseudoarthrosis of the limbs	157808	**17**	**17**
Congenital ptosis	91411	**297**	**297**
Congenital pulmonary airway malformation	2444	**697**	**695**
Congenital pulmonary lymphangiectasia	2414	**36**	**33**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital pulmonary sequestration	3161	**223**	**223**
Congenital pulmonary valvar stenosis	3189	**620**	**605**
Congenital pulmonary vein atresia	99126	**<10**	**<10**
Congenital pulmonary veins atresia or stenosis*	3188	**57**	**47**
Congenital pulmonary venous return anomaly*	3090	**120**	**117**
Congenital radioulnar synostosis	3269	**61**	**61**
Congenital renal artery stenosis	97598	**114**	**114**
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	**29**	**29**
Congenital short bowel syndrome	2301	**95**	**92**
Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome	369861	**<10**	**<10**
Congenital smooth muscle hamartoma	263435	**33**	**33**
Congenital sodium diarrhea	103908	**<10**	**<10**
Congenital stationary night blindness	215	**253**	**253**
Congenital stromal corneal dystrophy	101068	**<10**	**<10**
Congenital subglottic stenosis	141121	**98**	**95**
Congenital sucrase-isomaltase deficiency	35122	**70**	**70**
Congenital supravalvular mitral ring	99059	**<10**	**<10**
Congenital syphilis	499009	**<10**	**<10**
Congenital systemic arteriovenous fistula	2039	**22**	**21**
Congenital temporomandibular joint ankylosis	210576	**<10**	**<10**
Congenital total pulmonary venous return anomaly	99125	**247**	**238**
Congenital toxoplasmosis	858	**40**	**38**
Congenital tracheal stenosis	141127	**120**	**116**
Congenital tracheomalacia	95430	**295**	**294**
Congenital tricuspid stenosis	95459	**<10**	**<10**
Congenital tricuspid valve dysplasia	555874	**35**	**34**
Congenital trigeminal anesthesia	231013	**<10**	**<10**
Congenital trochlear nerve palsy	98686	**35**	**35**
Congenital tufting enteropathy	92050	**56**	**51**
Congenital unguarded mitral orifice	99060	**21**	**20**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Congenital unilateral hypoplasia of depressor anguli oris	1166	**130**	**130**
Congenital varicella syndrome	291	**<10**	**<10**
Congenital velopharyngeal incompetence	2291	**205**	**205**
Congenital vertebral-cardiac-renal anomalies syndrome	521438	**<10**	**<10**
Congenital vertical talus	178382	**<10**	**<10**
Congenitally corrected transposition of the great arteries	216694	**887**	**868**
Congenitally uncorrected transposition of the great arteries	860	**1763**	**1731**
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**
Continuous spikes and waves during sleep	725	**572**	**569**
Contractures-developmental delay-Pierre Robin syndrome	436003	**15**	**14**
Contractures-ectodermal dysplasia-cleft lip/palate syndrome	1484	**<10**	**<10**
Cooks syndrome	1487	**<10**	**<10**
Cor triatriatum dexter	99098	**<10**	**<10**
Cor triatriatum sinister	99099	**11**	**11**
Corneal dystrophy-perceptive deafness syndrome	1490	**<10**	**<10**
Corneal endotheliitis	137602	**81**	**81**
Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome	352662	**<10**	**<10**
Cornelia de Lange syndrome	199	**439**	**417**
Corneodermatoosseous syndrome	3194	**<10**	**<10**
Coronary arterial fistula	2041	**39**	**38**
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	**<10**	**<10**
Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome	52055	**<10**	**<10**
Corpus callosum agenesis-macrocephaly-hypertelorism syndrome	459074	**<10**	**<10**
Corpus callosum agenesis-neuronopathy syndrome	1496	**38**	**37**
Cortical blindness-intellectual disability-polydactyly syndrome	1389	**<10**	**<10**
Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation	300570	**27**	**27**
Corticobasal syndrome	454887	**262**	**238**
Corticosteroid-sensitive aseptic abscess syndrome	54251	**18**	**18**
Costello syndrome	3071	**83**	**76**
Cowden syndrome	201	**539**	**531**
Coxopodopatellar syndrome	1509	**48**	**48**
Cramp-fasciculation syndrome	581271	**156**	**156**
Crane-Heise syndrome	1512	**<10**	**<10**
Cranial meningocele	268820	**15**	**15**
Cranial neuralgia*	221109	**<10**	**<10**
Cranio-cervical dystonia with laryngeal and upper-limb involvement	420485	**21**	**21**
Craniodiaphyseal dysplasia	1513	**<10**	**<10**
Craniodigital-intellectual disability syndrome	1514	**<10**	**<10**
Cranioectodermal dysplasia	1515	**29**	**26**
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	**56**	**56**
Craniofrontonasal dysplasia-Poland anomaly syndrome	1521	**<10**	**<10**
Craniometadiaphyseal dysplasia, wormian bone type	85184	**<10**	**<10**
Craniometaphyseal dysplasia	1522	**26**	**25**
Cranio-osteoarthropathy	1525	**<10**	**<10**
Craniopharyngioma	54595	**1959**	**1859**
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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome	171839	**<10**	**<10**
Craniosynostosis-intracranial calcifications syndrome	52054	**<10**	**<10**
Creatine deficiency syndrome*	79172	**18**	**18**
Crigler-Najjar syndrome	205	**52**	**48**
Crimean-Congo hemorrhagic fever	99827	**<10**	**<10**
Crisponi syndrome	1545	**25**	**18**
Criss-cross heart	1461	**<10**	**<10**
Crossed polysyndactyly	2935	**<10**	**<10**
Crouzon syndrome	207	**384**	**378**
Crouzon syndrome-acanthosis nigricans syndrome	93262	**21**	**21**
Cryoglobulinemic vasculitis	91138	**675**	**628**
Cryopyrin-associated periodic syndrome*	208650	**98**	**98**
Cryptococcosis	1546	**<10**	**<10**
Cryptogenic late-onset epileptic spasms	163708	**21**	**21**
Cryptogenic organizing pneumonia	1302	**555**	**540**
Cryptomicrotia-brachydactyly-excess fingertip arch syndrome	1547	**<10**	**<10**
Cryptorchidism-arachnodactyly-intellectual disability syndrome	1548	**<10**	**<10**
CTCF-related neurodevelopmental disorder	363611	**13**	**13**
Currarino syndrome	1552	**262**	**256**
Curry-Jones syndrome	1553	**<10**	**<10**
Cushing disease	96253	**2746**	**2672**
Cushing syndrome due to bilateral macronodular adrenocortical disease	189427	**701**	**681**
Cushing syndrome due to cortisol-producing adrenocortical adenoma	642788	**18**	**18**
Cushing syndrome due to ectopic ACTH secretion	99889	**147**	**116**
Cutaneous collagenous vasculopathy	280779	**<10**	**<10**
Cutaneous mastocytoma	79455	**330**	**330**
Cutaneous mastocytosis*	66646	**723**	**715**
Cutaneous neuroendocrine carcinoma	79140	**24**	**19**
Cutaneous pseudolymphoma	451607	**<10**	**<10**
Cutaneous small vessel vasculitis	889	**135**	**132**
Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome	1555	**<10**	**<10**
Cutis laxa*	209	**85**	**78**
Cutis laxa-Marfanoid syndrome	171719	**<10**	**<10**
Cutis marmorata telangiectatica congenita	1556	**169**	**169**
Cyclic neutropenia	2686	**111**	**111**
Cylindrical spirals myopathy	171886	**<10**	**<10**
Cystadenoma of childhood	206470	**17**	**17**
Cystic echinococcosis	400	**<10**	**<10**
Cystic fibrosis	586	**9586**	**9293**
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	**200**	**184**
Cystinuria	214	**675**	**671**
Cystoid macular dystrophy	75381	**21**	**21**
Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	137698	**<10**	**<10**
Czeizel-Losonci syndrome	2437	**<10**	**<10**
D-2-hydroxyglutaric aciduria	79315	**<10**	**<10**
Dacryocystitis-osteopoikilosis syndrome	1562	**11**	**11**
Dandy-Walker malformation-postaxial polydactyly syndrome	1566	**<10**	**<10**
Darier disease	218	**240**	**234**
DDX41-related hematologic malignancy predisposition syndrome	488647	**<10**	**<10**
De Barsy syndrome	2962	**19**	**17**
De novo thrombotic microangiopathy after kidney transplantation	244275	**27**	**25**
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	**14**	**13**
Deafness-enamel hypoplasia-nail defects syndrome	3220	**19**	**19**
Deafness-encephaloneuropathy-obesity-valvulopathy syndrome	254898	**<10**	**<10**
Deafness-hypogonadism syndrome	90646	**<10**	**<10**
Deafness-infertility syndrome	94064	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Deafness-intellectual disability syndrome, Martin-Probst type	85321	<10	<10
Deafness-lymphedema-leukemia syndrome	3226	<10	<10
Deafness-onychodystrophy syndrome*	3231	11	<10
Deep dermatophytosis	397587	<10	<10
Deficiency in anterior pituitary function-variable immunodeficiency syndrome	293978	35	35
Dehydrated hereditary stomatocytosis	3202	65	64
Dejerine-Sottas syndrome	64748	19	16
Delayed membranous cranial ossification	3034	<10	<10
Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome	3038	20	18
Deletion 5q35	1627	18	16
Delta-beta-thalassemia	231237	11	11
Delta-sarcoglycan-related limb-girdle muscular dystrophy R6	219	<10	<10
Demodicidosis	283	<10	<10
DEND syndrome	79134	<10	<10
Dengue fever	99828	<10	<10
Dent disease	1652	343	342
Dental ankylosis	1077	<10	<10
Dentatorubral pallidoluysian atrophy	101	13	<10
Dentin dysplasia	1653	27	27
Dentinogenesis imperfecta	49042	419	418
Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome	71267	<10	<10
Denys-Drash syndrome	220	85	82
Dermatitis herpetiformis	1656	83	82
Dermatofibrosarcoma protuberans	31112	14	14
Dermatoleukodystrophy	1659	<10	<10
Dermatomyositis	221	1888	1761
Dermatosparaxis Ehlers-Danlos syndrome	1901	<10	<10
Dermochondrocorneal dystrophy	79149	<10	<10
Dermoid or epidermoid cyst of the central nervous system	530033	17	17
Dermoodontodysplasia	1660	<10	<10
Desbuquois syndrome	1425	15	13
Desminopathy	98909	86	73
Desmin-related myopathy with Mallory body-like inclusions	84132	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Desmoid tumor	873	24	24
Desmoplastic infantile astrocytoma/ganglioglioma	251940	<10	<10
Desmoplastic small round cell tumor	83469	<10	<10
Desmosterolosis	35107	<10	<10
Desquamative interstitial pneumonia	98852	297	280
Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	289307	<10	<10
Developmental delay with autism spectrum disorder and gait instability	329195	196	195
Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	369891	59	58
Developmental malformations-deafness-dystonia syndrome	79107	<10	<10
Dextrocardia	1666	25	25
D-glyceric aciduria	941	<10	<10
Diabetic embryopathy	1926	<10	<10
Diamond-Blackfan anemia	124	246	244
DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	494444	<10	<10
Diaphanospondylodysostosis	66637	<10	<10
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	59	58
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	12	12
Diazoxide-sensitive diffuse hyperinsulinism*	165985	32	32
DICER1 tumor-predisposition syndrome	284343	22	22
Didelphys uterus	180086	89	88
Didymosis aplasticosebacea	370046	<10	<10
Diencephalic-mesencephalic junction dysplasia	319192	<10	<10
Dietary iron overload disease	139507	11	11

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Difference of sex development-intellectual disability syndrome	2983	<10	<10
Differentiated thyroid carcinoma	146	73	72
Diffuse alveolar hemorrhage	90060	59	54
Diffuse astrocytoma	251595	<10	<10
Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	404437	<10	<10
Diffuse cutaneous mastocytosis	79456	104	101
Diffuse idiopathic pulmonary neuroendocrine cell hyperplasia	617916	<10	<10
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	68	53
Diffuse leptomeningeal melanocytosis	252031	<10	<10
Diffuse lymphatic malformation	141209	147	143
Diffuse neonatal hemangiomatosis	2123	100	99
Diffuse palmoplantar keratoderma with painful fissures	369999	<10	<10
Diffuse palmoplantar keratoderma-acrocyanosis syndrome	86918	<10	<10
Diffuse panbronchiolitis	171700	18	16
Digestive duplication	238	20	20
Digestive duplication cyst of the tongue	141071	<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	19	18
Dimethylglycine dehydrogenase deficiency	243343	<10	<10
Diphallia	227	<10	<10
Diphtheria	1679	<10	<10
Discoid lupus erythematosus	90281	78	78
Dislocation of the hip-dysmorphism syndrome	2412	15	15
Dissecting cellulitis of the scalp	345	<10	<10
Disseminated peritoneal leiomyomatosis	71274	<10	<10
Disseminated superficial actinic porokeratosis	79152	28	27

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Distal 16p11.2 microdeletion syndrome	261222	149	149
Distal 17p13.1 microdeletion syndrome	319171	<10	<10
Distal 17p13.3 microdeletion syndrome	261257	15	14
Distal 22q11.2 microdeletion syndrome	261330	99	98
Distal 22q11.2 microduplication syndrome	261337	14	14
Distal 7q11.23 microdeletion syndrome	254351	13	13
Distal 7q11.23 microduplication syndrome	261102	<10	<10
Distal anoctaminopathy	399096	52	50
Distal arthrogryposis type 1	1146	32	32
Distal arthrogryposis type 10	251515	<10	<10
Distal arthrogryposis type 5D	329457	<10	<10
Distal arthrogryposis*	97120	180	175
Distal deletion 10p	1580	22	22
Distal deletion 10q	96148	45	45
Distal deletion 12p	280325	19	19
Distal deletion 12q	96149	<10	<10
Distal deletion 13q	1590	37	36
Distal deletion 14q	96150	18	18
Distal deletion 15q	1596	52	51
Distal deletion 17q	1597	14	13
Distal deletion 19p	96129	<10	<10
Distal deletion 1q	36367	22	22
Distal deletion 3p	1620	32	30
Distal deletion 4q	96145	52	47
Distal deletion 6p	96125	25	25
Distal deletion 7p	96126	<10	<10
Distal deletion 9p	1642	45	45
Distal duplication 10q	96102	<10	<10
Distal duplication 11q	96103	<10	<10
Distal duplication 13q	96105	<10	<10
Distal duplication 14q	1705	16	15
Distal duplication 16q	96106	17	17
Distal duplication 17q	3379	12	12
Distal duplication 18q	1716	19	18
Distal duplication 19q	1717	17	17
Distal duplication 1p36	96069	<10	<10
Distal duplication 20q	96107	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Distal duplication 22q	96109	**15**	**15**
Distal duplication 2p	96070	**<10**	**<10**
Distal duplication 2q	96094	**<10**	**<10**
Distal duplication 3p	96071	**<10**	**<10**
Distal duplication 4q	96096	**<10**	**<10**
Distal duplication 5q	96097	**<10**	**<10**
Distal duplication 6p	1745	**11**	**11**
Distal duplication 6q	96098	**<10**	**<10**
Distal duplication 7p	96074	**<10**	**<10**
Distal duplication 8q	96100	**<10**	**<10**
Distal duplication 9q	96101	**11**	**11**
Distal hereditary motor neuropathy type 1	139518	**54**	**52**
Distal hereditary motor neuropathy type 2	139525	**12**	**<10**
Distal hereditary motor neuropathy type 5	139536	**11**	**11**
Distal hereditary motor neuropathy type 7	139589	**<10**	**<10**
Distal hereditary motor neuropathy*	53739	**347**	**335**
Distal hereditary motor neuropathy, Jerash type	139552	**<10**	**<10**
Distal limb deficiencies-micrognathia syndrome	1307	**<10**	**<10**
Distal monosomy 7q36	1636	**24**	**24**
Distal myopathy with anterior tibial onset	178400	**23**	**22**
Distal myopathy with posterior leg and anterior hand involvement	63273	**26**	**23**
Distal myopathy, Tateyama type	488650	**<10**	**<10**
Distal myopathy, Welander type	603	**<10**	**<10**
Distal myotilinopathy	98911	**34**	**32**
Distal nebulin myopathy	399103	**17**	**16**
Distal renal tubular acidosis	18	**336**	**336**
Distal spinal muscular atrophy type 3	139547	**61**	**60**
Distal symphalangism	3248	**<10**	**<10**
Distal Xq28 microduplication syndrome	293939	**16**	**16**
DITRA	404546	**<10**	**<10**
DK1-CDG	91131	**<10**	**<10**
DNA2-related mitochondrial DNA deletion syndrome	352470	**15**	**14**
DNAJB2-related Charcot-Marie-Tooth disease type 2	443950	**<10**	**<10**
DNAJB6-related limb-girdle muscular dystrophy D1	34516	**23**	**20**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
DOCK2 deficiency	447737	**<10**	**<10**
Dominant hypophosphatemia with nephrolithiasis or osteoporosis	244305	**327**	**327**
Donnai-Barrow syndrome	2143	**12**	**11**
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	**<10**	**<10**
Dopa-responsive dystonia*	255	**36**	**36**
Dorsal spinal cord lipoma	645362	**<10**	**<10**
Double outlet left ventricle	3427	**40**	**38**
Double outlet right ventricle	3426	**581**	**550**
Double uterus-hemivagina-renal agenesis syndrome	3411	**103**	**103**
Dowling-Degos disease	79145	**13**	**13**
Down syndrome	870	**5884**	**5677**
DPAGT1-CDG	86309	**<10**	**<10**
DPM1-CDG	79322	**<10**	**<10**
DPM3-CDG	263494	**<10**	**<10**
Dravet syndrome	33069	**906**	**856**
Drug or radiation exposure-related interstitial lung disease	264978	**508**	**433**
Drug- or toxin-induced pulmonary arterial hypertension*	275786	**169**	**145**
Drug reaction with eosinophilia and systemic symptoms	139402	**1196**	**1070**
Drug-induced autoimmune hemolytic anemia	90037	**<10**	**<10**
Drug-induced localized lipodystrophy	90157	**<10**	**<10**
Drug-induced lupus erythematosus	231111	**41**	**38**
Drug-induced vasculitis	251325	**17**	**15**
Duane retraction syndrome	233	**161**	**159**
Dubin-Johnson syndrome	234	**36**	**36**
Dubowitz syndrome	235	**17**	**16**
Duchenne and Becker muscular dystrophy*	262	**376**	**328**
Duchenne muscular dystrophy	98896	**2222**	**1836**
Duodenal atresia	1203	**114**	**111**
Duplication of the esophagus*	91357	**<10**	**<10**
Duplication of the pituitary gland	314621	**<10**	**<10**
Duplication of urethra	237	**58**	**58**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Dural sinus malformation	97339	**61**	**60**
Dyggve-Melchior-Clausen disease	239	**20**	**20**
DYRK1A-related intellectual disability syndrome	464306	**43**	**43**
Dysbetalipoproteinemia	412	**14**	**14**
Dyschondrosteosis-nephritis syndrome	1765	**<10**	**<10**
Dyschromatosis symmetrica hereditaria	41	**<10**	**<10**
Dyschromatosis universalis hereditaria	241	**<10**	**<10**
Dysembryoplastic neuroepithelial tumor	251946	**290**	**289**
Dysequilibrium syndrome	1766	**109**	**107**
Dysferlin-related limb-girdle muscular dystrophy R2	268	**241**	**225**
Dyskeratosis congenita	1775	**216**	**182**
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	**26**	**26**
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, Silverman-Handmaker type	1865	**<10**	**<10**
Dysspondyloenchondromatosis	85198	**11**	**11**
Dystonia 16	210571	**12**	**12**
Dystonia 28	589618	**15**	**15**
Dystonia-aphonia syndrome	412217	**<10**	**<10**
Dystrophic epidermolysis bullosa pruriginosa	89843	**15**	**15**
Dystrophic epidermolysis bullosa*	303	**407**	**374**
Eales disease	40923	**55**	**54**
Early infantile epileptic encephalopathy	1934	**1171**	**1043**
Early myoclonic encephalopathy	1935	**59**	**48**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Early-onset autoimmunity-autoinflammation-immunodeficiency syndrome	619948	**<10**	**<10**
Early-onset autosomal dominant Alzheimer disease	1020	**114**	**100**
Early-onset cerebellar ataxia with retained tendon reflexes	1177	**49**	**49**
Early-onset epilepsy-intellectual disability-brain anomalies syndrome	488635	**14**	**14**
Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation	289266	**35**	**35**
Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	411986	**<10**	**<10**
Early-onset generalized limb-onset dystonia	256	**296**	**287**
Early-onset myopathy with fatal cardiomyopathy	289377	**<10**	**<10**
Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome	439212	**<10**	**<10**
Early-onset non-syndromic cataract	91492	**1189**	**1180**
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**	**17**
Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	500144	**<10**	**<10**
Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	313772	**<10**	**<10**
Early-onset X-linked optic atrophy	98890	**37**	**37**
Ear-patella-short stature syndrome	2554	**25**	**24**
EAST syndrome	199343	**15**	**15**
Ebstein malformation of the tricuspid valve	1880	**390**	**381**
Ectasia of the right atrial appendage	99101	**<10**	**<10**
Ectodermal dysplasia with natal teeth, Turnpenny type	69083	**<10**	**<10**
Ectodermal dysplasia, trichoodontoonychial type	1818	**<10**	**<10**
Ectodermal dysplasia-hyperhidrosis-cutaneous syndactyly syndrome	247827	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ectodermal dysplasia-intellectual disability-central nervous system malformation syndrome	1812	<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	130	130
Edinburgh malformation syndrome	1895	<10	<10
EEC syndrome	1896	127	126
EEM syndrome	1897	<10	<10
Ehlers-Danlos syndrome*	98249	1893	1876
Ehlers-Danlos/osteogenesis imperfecta syndrome	230857	28	28
Eisenmenger syndrome	97214	361	334
Elastofibroma dorsi	228243	<10	<10
Ellis Van Creveld syndrome	289	65	56
Emanuel syndrome	96170	19	18
Embryonal carcinoma	180226	<10	<10
Emery-Dreifuss muscular dystrophy	261	231	202
Emery-Nelson syndrome	1927	<10	<10
Enamel-renal syndrome	1031	49	49
Encephalitis lethargica	83600	<10	<10
Encephaloclastic disorder*	269190	<10	<10
Encephalocraniocutaneous lipomatosis	2396	<10	<10
Encephalopathy due to mitochondrial and peroxisomal fission defect	527276	<10	<10
Encephalopathy due to prosaposin deficiency	139406	<10	<10
Encephalopathy due to sulfite oxidase deficiency	833	35	15
Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	319678	<10	<10
Encircling double aortic arch	99075	42	42
Endocardial fibroelastosis	2022	<10	<10
Endometrial stromal sarcoma	213711	<10	<10
Endometrioid carcinoma of ovary	454723	<10	<10
Endophthalmitis	199323	43	43
Endosteal hyperostosis, Worth type	2790	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Enlarged parietal foramina	60015	21	21
Enthesitis-related juvenile idiopathic arthritis	85438	2726	2722
Eosinophilic angiocentric fibrosis	449566	<10	<10
Eosinophilic colitis	402035	14	14
Eosinophilic fasciitis	3165	196	195
Eosinophilic gastroenteritis	2070	14	14
Eosinophilic granulomatosis with polyangiitis	183	1230	1186
Ependymal tumor*	301	<10	<10
Ependymoblastoma	251880	<10	<10
Ependymoma	251636	54	48
Epiblepharon	99169	46	45
Epidermal nevus syndrome	35125	224	224
Epidermodysplasia verruciformis	302	31	29
Epidermolysis bullosa acquisita	46487	209	196
Epidermolysis bullosa simplex with mottled pigmentation	79397	<10	<10
Epidermolysis bullosa simplex with muscular dystrophy	257	<10	<10
Epidermolysis bullosa simplex*	304	357	355
Epidermolytic nevus	497737	21	21
Epidermolytic palmoplantar keratoderma	2199	64	64
Epilepsy with myoclonic absences	86911	305	302
Epilepsy-microcephaly-skeletal dysplasia syndrome	1948	<10	<10
Epilepsy-telangiectasia syndrome	1951	<10	<10
Epileptic encephalopathy with global cerebral demyelination	353217	<10	<10
Epiphyseal stippling-osteoclastic hyperplasia syndrome	1952	<10	<10
Epiphysiolysis of the hip	399329	<10	<10
Episodic ataxia type 1	37612	47	47
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
Epithelial basement membrane dystrophy	98956	53	53
Epithelial recurrent erosion dystrophy	293381	26	26
Epithelioid hemangioendothelioma	157791	40	37
Epstein-Barr virus-positive diffuse large B-cell lymphoma of the elderly	289661	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Erdheim-Chester disease	35687	**338**	**310**
Ermine phenotype	999	**<10**	**<10**
Erosive pustular dermatosis of the scalp	222	**<10**	**<10**
Erythema elevatum diutinum	90000	**<10**	**<10**
Erythema multiforme major	502499	**164**	**163**
Erythroderma desquamativum	314	**<10**	**<10**
Erythrokeratoderma ''en cocardes''	315	**<10**	**<10**
Erythrokeratoderma variabilis progressiva*	308166	**<10**	**<10**
Erythrokeratodermia variabilis	317	**25**	**25**
Esophageal atresia	1199	**2511**	**2434**
Esophageal duplication cyst	100047	**<10**	**<10**
Essential fructosuria	2056	**<10**	**<10**
Essential thrombocythemia	3318	**101**	**96**
Estrogen resistance syndrome	785	**<10**	**<10**
Ethylmalonic encephalopathy	51188	**<10**	**<10**
Euryblepharon	99172	**<10**	**<10**
Euthyroid dysprealbuminemic hyperthyroxinemia	597939	**<10**	**<10**
Euthyroid Graves orbitopathy	466682	**22**	**22**
Evans syndrome	1959	**545**	**516**
EVEN-plus syndrome	496751	**<10**	**<10**
Exercise intolerance with lactic acidosis*	254843	**21**	**21**
Exercise-induced malignant hyperthermia	466650	**22**	**22**
Exfoliative ichthyosis	289586	**13**	**12**
Exostoses-anetodermia-brachydactyly type E syndrome	1962	**<10**	**<10**
Exstrophy-epispadias complex	322	**615**	**612**
Extensive peripapillary myelinated nerve fibers	440724	**14**	**14**
External auditory canal aplasia/ hypoplasia	141074	**1411**	**1404**
External auditory canal atresia-vertical talus-hypertelorism syndrome	3023	**<10**	**<10**
Extracranial carotid artery aneurysm	494424	**12**	**12**
Extracutaneous mastocytoma	66662	**<10**	**<10**
Extragonadal germinoma	182127	**148**	**147**
Extragonadal teratoma	883	**115**	**112**
Extramammary Paget disease	2800	**14**	**12**
Extramedullary conus spinal cord lipoma	645297	**<10**	**<10**
Extraneural perineurioma	100002	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Extranodal nasal NK/T cell lymphoma	86879	**<10**	**<10**
Extrapelvic endometriosis	137820	**74**	**73**
Extraskeletal Ewing sarcoma	370334	**<10**	**<10**
Fabry disease	324	**1657**	**1590**
Facial arteriovenous malformation*	156230	**552**	**550**
Facial dermoid cyst	141051	**422**	**422**
Facial diplegia with paresthesias	480701	**<10**	**<10**
Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome	1969	**<10**	**<10**
Facial dysmorphism-immunodeficiency-livedo-short stature syndrome	352712	**12**	**12**
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	**31**	**21**
Faciocardiorenal syndrome	1973	**<10**	**<10**
Facioscapulohumeral dystrophy	269	**3499**	**3275**
Fallot complex-intellectual disability-growth delay syndrome	3304	**<10**	**<10**
Familial abdominal aortic aneurysm	86	**67**	**65**
Familial acute necrotizing encephalopathy	88619	**<10**	**<10**
Familial adenomatous polyposis	733	**477**	**459**
Familial adrenal hypoplasia with absent pituitary luteinizing hormone	95700	**<10**	**<10**
Familial Alzheimer-like prion disease	280397	**<10**	**<10**
Familial anetoderma	228277	**47**	**47**
Familial angiolipomatosis	199279	**<10**	**<10**
Familial aortic dissection	229	**313**	**300**
Familial articular hypermobility syndrome	2295	**842**	**841**
Familial atrial fibrillation	334	**139**	**137**
Familial atrial myxoma	615	**<10**	**<10**
Familial atypical multiple mole melanoma syndrome	404560	**127**	**124**
Familial avascular necrosis of femoral head	86820	**<10**	**<10**
Familial benign chronic pemphigus	2841	**224**	**220**
Familial benign copper deficiency	1551	**<10**	**<10**
Familial benign flecked retina	363989	**<10**	**<10**
Familial bicuspid aortic valve	402075	**319**	**316**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Familial calcium pyrophosphate deposition	1416	**33**	**27**
Familial caudal dysgenesis	1768	**<10**	**<10**
Familial cavitary optic disc anomaly	464760	**<10**	**<10**
Familial cerebral cavernous malformation	221061	**565**	**556**
Familial cerebral saccular aneurysm	231160	**81**	**79**
Familial cervical artery dissection	36382	**202**	**198**
Familial Chilblain lupus	481662	**<10**	**<10**
Familial chylomicronemia syndrome	444490	**44**	**44**
Familial clubfoot with or without associated lower limb anomalies	199315	**140**	**140**
Familial cold urticaria	47045	**37**	**37**
Familial congenital mirror movements	238722	**32**	**32**
Familial congenital nasolacrimal duct obstruction	451612	**<10**	**<10**
Familial congenital palsy of trochlear nerve	91498	**<10**	**<10**
Familial cortical myoclonus	319189	**14**	**14**
Familial cutaneous collagenoma	53296	**<10**	**<10**
Familial developmental dysphasia	1799	**52**	**52**
Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	300751	**452**	**438**
Familial drusen	75376	**87**	**86**
Familial dysautonomia	1764	**22**	**20**
Familial dyskinesia and facial myokymia	324588	**<10**	**<10**
Familial encephalopathy with neuroserpin inclusion bodies	85110	**<10**	**<10**
Familial episodic pain syndrome	391384	**<10**	**<10**
Familial expansile osteolysis	85195	**<10**	**<10**
Familial exudative vitreoretinopathy	891	**148**	**148**
Familial focal epilepsy with variable foci	98820	**68**	**68**
Familial generalized lentiginosis	231040	**<10**	**<10**
Familial gestational hyperthyroidism	99819	**15**	**15**
Familial glucocorticoid deficiency	361	**29**	**29**
Familial hemophagocytic lymphohistiocytosis	540	**109**	**95**
Familial hyperaldosteronism type I	403	**93**	**91**
Familial hyperaldosteronism type II	404	**18**	**18**
Familial hyperaldosteronism type III	251274	**<10**	**<10**
Familial hyperaldosteronism*	235936	**41**	**41**
Familial hypercholanemia	238475	**24**	**24**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Familial hyperinflammatory lymphoproliferative immunodeficiency	619953	**<10**	**<10**
Familial hyperprolactinemia	397685	**17**	**16**
Familial hyperthyroidism due to mutations in TSH receptor	424	**116**	**114**
Familial hypoaldosteronism	427	**24**	**23**
Familial hypocalciuric hypercalcemia	405	**660**	**647**
Familial idiopathic dilatation of the right atrium	1677	**<10**	**<10**
Familial infantile bilateral striatal necrosis	225154	**<10**	**<10**
Familial infantile myoclonic epilepsy	352582	**23**	**23**
Familial intestinal malrotation	508410	**<10**	**<10**
Familial isolated arrhythmogenic right ventricular dysplasia	217656	**1651**	**1635**
Familial isolated café-au-lait macules	2678	**239**	**239**
Familial isolated clinodactyly of fingers	295014	**24**	**24**
Familial isolated congenital asplenia	101351	**28**	**28**
Familial isolated dilated cardiomyopathy	154	**8112**	**7825**
Familial isolated hyperparathyroidism	99879	**227**	**223**
Familial isolated hypoparathyroidism	2238	**315**	**306**
Familial isolated pituitary adenoma	314777	**<10**	**<10**
Familial isolated restrictive cardiomyopathy	75249	**200**	**183**
Familial isolated trichomegaly	411788	**<10**	**<10**
Familial juvenile hypertrophy of the breast	180176	**48**	**48**
Familial keratoacanthoma	493	**<10**	**<10**
Familial long QT syndrome*	768	**2051**	**2023**
Familial median cleft of the upper and lower lips	401942	**<10**	**<10**
Familial Mediterranean fever	342	**2181**	**2155**
Familial medullary thyroid carcinoma	99361	**<10**	**<10**
Familial melanoma	618	**383**	**362**
Familial mesial temporal lobe epilepsy with febrile seizures	165805	**<10**	**<10**
Familial mitral valve prolapse	741	**77**	**77**
Familial monosomy 7 syndrome	495930	**<10**	**<10**
Familial multinodular goiter	276399	**<10**	**<10**
Familial multiple lipomatosis	199276	**28**	**27**
Familial multiple meningioma	263662	**17**	**17**
Familial multiple nevi flammei	624	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Familial or sporadic hemiplegic migraine	569	**240**	**238**
Familial ossifying fibroma	435329	**<10**	**<10**
Familial osteochondritis dissecans	251262	**<10**	**<10**
Familial pancreatic carcinoma	1333	**161**	**154**
Familial papillary or follicular thyroid carcinoma	319487	**11**	**11**
Familial paroxysmal ataxia	97	**147**	**143**
Familial partial epilepsy*	309	**102**	**102**
Familial partial lipodystrophy*	98306	**24**	**22**
Familial partial lipodystrophy, Dunnigan type	2348	**345**	**333**
Familial partial lipodystrophy, Köbberling type	79084	**26**	**26**
Familial patent arterial duct	466729	**17**	**17**
Familial peripheral male-limited precocious puberty	3000	**30**	**29**
Familial platelet disorder with associated myeloid malignancy	71290	**133**	**133**
Familial primary hyperparathyroidism*	2207	**638**	**600**
Familial primary localized cutaneous amyloidosis	353220	**<10**	**<10**
Familial progressive cardiac conduction defect	871	**225**	**222**
Familial progressive hyper- and hypopigmentation	280628	**11**	**11**
Familial progressive hyperpigmentation	79146	**23**	**23**
Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome	488197	**<10**	**<10**
Familial prostate cancer	1331	**21**	**20**
Familial pseudohyperkalemia	90044	**<10**	**<10**
Familial reactive perforating collagenosis	79147	**<10**	**<10**
Familial recurrent peripheral facial palsy	2809	**25**	**25**
Familial renal glucosuria	69076	**42**	**42**
Familial scaphocephaly syndrome, McGillivray type	168624	**15**	**15**
Familial short QT syndrome	51083	**54**	**54**
Familial sick sinus syndrome	166282	**16**	**15**
Familial spontaneous pneumothorax	2903	**42**	**42**
Familial steroid-resistant nephrotic syndrome with adrenal insufficiency	506334	**<10**	**<10**
Familial steroid-resistant nephrotic syndrome with sensorineural deafness	280406	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Familial supernumerary nipples	2456	**<10**	**<10**
Familial temporal lobe epilepsy	98819	**39**	**39**
Familial thoracic aortic aneurysm and aortic dissection	91387	**932**	**925**
Familial thrombocytosis	71493	**20**	**19**
Familial thrombomodulin anomalies	3324	**<10**	**<10**
Familial thyroglossal duct cyst	93953	**112**	**112**
Familial thyroid dyshormonogenesis	95716	**429**	**426**
Familial tumoral calcinosis	53715	**44**	**43**
Familial vesicoureteral reflux	289365	**157**	**157**
Familial visceral myopathy	2604	**<10**	**<10**
Fanconi anemia	84	**337**	**297**
Fanconi-Bickel syndrome	2088	**24**	**23**
Farber disease	333	**<10**	**<10**
Farmer's lung disease	99906	**40**	**38**
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**
Fatal infantile hypertonic myofibrillar myopathy	280553	**<10**	**<10**
Fatal infantile lactic acidosis with methylmalonic aciduria	17	**25**	**18**
Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	168566	**<10**	**<10**
FATCO syndrome	2492	**<10**	**<10**
Fatty acid hydroxylase-associated neurodegeneration	329308	**14**	**14**
Fatty acyl-CoA reductase 1 deficiency	438178	**<10**	**<10**
Febrile infection-related epilepsy syndrome	163703	**89**	**86**
Feingold syndrome	1305	**111**	**110**
Felty syndrome	47612	**<10**	**<10**
Female infertility due to oocyte meiotic arrest	488191	**<10**	**<10**
Female infertility due to zona pellucida defect	404466	**<10**	**<10**
Female restricted epilepsy with intellectual disability	101039	**115**	**115**
Femoral agenesis/hypoplasia	1987	**185**	**182**
Femoral-facial syndrome	1988	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Femur-fibula-ulna complex	2019	**14**	**14**
Ferroportin Disease	648562	**<10**	**<10**
Fetal akinesia deformation sequence	994	**17**	**<10**
Fetal akinesia-cerebral and retinal hemorrhage syndrome	363409	**<10**	**<10**
Fetal alcohol syndrome	1915	**814**	**809**
Fetal and neonatal alloimmune thrombocytopenia	853	**30**	**30**
Fetal anticonvulsant syndrome*	370068	**<10**	**<10**
Fetal cytomegalovirus syndrome	294	**243**	**238**
Fetal hydantoin syndrome	1912	**<10**	**<10**
Fetal iodine syndrome	1910	**12**	**12**
Fetal lower urinary tract obstruction*	435365	**30**	**30**
Fetal parvovirus syndrome	295	**<10**	**<10**
Fetal valproate spectrum disorder	1906	**589**	**584**
Fever-associated acute infantile liver failure syndrome	464724	**<10**	**<10**
FG syndrome type 1	93932	**<10**	**<10**
FGFR2-related bent bone dysplasia	313855	**<10**	**<10**
Fibroblastic rheumatism	477650	**<10**	**<10**
Fibrochondrogenesis	2021	**<10**	**<10**
Fibrodysplasia ossificans progressiva	337	**173**	**166**
Fibronectin glomerulopathy	84090	**<10**	**<10**
Fibrosarcoma	2030	**<10**	**<10**
Fibrosis-neurodegeneration-cerebral angiomatosis syndrome	621758	**<10**	**<10**
Fibrous dysplasia of bone	249	**2271**	**2239**
Fibular aplasia-complex brachydactyly syndrome	2639	**11**	**11**
Fibular aplasia-ectrodactyly syndrome	1118	**27**	**27**
Fibular dimelia-diplopodia syndrome	1757	**<10**	**<10**
Fibular hemimelia	93323	**102**	**102**
Filippi syndrome	3255	**<10**	**<10**
Fingerprint body myopathy	97232	**<10**	**<10**
Finnish upper limb-onset distal myopathy	399086	**<10**	**<10**
First branchial cleft anomaly	141013	**200**	**199**
Fixed drug eruption	293812	**138**	**125**
Fixed subaortic stenosis	3092	**90**	**89**
FKRP-related limb-girdle muscular dystrophy R9	34515	**129**	**116**
Flat face-microstomia-ear anomaly syndrome	1968	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Fleck corneal dystrophy	98970	**<10**	**<10**
FLNA-related X-linked myxomatous valvular dysplasia	555877	**<10**	**<10**
Floating-Harbor syndrome	2044	**88**	**88**
Florid cemento-osseous dysplasia	83451	**<10**	**<10**
Focal acral hyperkeratosis	308013	**<10**	**<10**
Focal dermal hypoplasia	2092	**75**	**75**
Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	352587	**27**	**27**
Focal facial dermal dysplasia	398166	**30**	**30**
Focal myositis	48918	**171**	**160**
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	**22**	**19**
Folliculotropic mycosis fungoides	178512	**<10**	**<10**
Formiminoglutamic aciduria	51208	**<10**	**<10**
Fountain syndrome	3219	**<10**	**<10**
Fourth branchial cleft anomaly	141037	**94**	**93**
Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome	397618	**<10**	**<10**
Foveal hypoplasia-presenile cataract syndrome	2253	**22**	**22**
Fowler urethral sphincter dysfunction syndrome	2795	**<10**	**<10**
Fowler vasculopathy	221126	**<10**	**<10**
FOXG1 syndrome	561854	**58**	**56**
Fragile X syndrome	908	**2147**	**2126**
Fragile X-associated primary ovarian insufficiency	642691	**<10**	**<10**
Fragile X-associated tremor/ataxia syndrome	93256	**101**	**98**
Fraser syndrome	2052	**24**	**19**
Frasier syndrome	347	**26**	**26**
FRAXE intellectual disability	100973	**52**	**50**
FRAXF syndrome	100974	**<10**	**<10**
Free sialic acid storage disease	834	**<10**	**<10**
Freeman-Sheldon syndrome	2053	**46**	**43**
Fried syndrome	85335	**<10**	**<10**
Friedreich ataxia	95	**1009**	**916**
Frontal fibrosing alopecia	254492	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Frontofacionasal dysplasia	1791	13	13
Frontometaphyseal dysplasia	1826	23	23
Frontonasal arteriovenous malformation	141168	42	42
Frontonasal dysplasia*	250	64	64
Frontorhiny	391474	<10	<10
Frontotemporal dementia with motor neuron disease	275872	449	197
Frontotemporal dementia*	282	616	557
Fructose-1,6-bisphosphatase deficiency	348	35	35
Fryns syndrome	2059	16	<10
FTH1-related iron overload	247790	11	11
Fuchs endothelial corneal dystrophy	98974	767	766
Fuchs heterochromic iridocyclitis	263479	62	62
Fucosidosis	349	<10	<10
Fuhrmann syndrome	2854	<10	<10
Fukutin-related limb-girdle muscular dystrophy R13	206554	<10	<10
Full NF2-related schwannomatosis	637	740	700
Full schwannomatosis	93921	481	477
Fulminant viral hepatitis	35063	18	18
Fumaric aciduria	24	13	11
Functioning gonadotropic adenoma	91348	1085	1019
Functioning pituitary adenoma*	314753	<10	<10
Fundus albipunctatus	227796	23	23
Fungal keratitis	519930	19	19
Fungal myositis	207000	<10	<10
Furuncular myiasis	591	<10	<10
Gabriele-de Vries syndrome	506358	14	14
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	44	28
Gamma-aminobutyric acid transaminase deficiency	2066	<10	<10
Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5	353	241	232
Gangliocytoma	251937	<10	<10
Ganglioglioma	251949	141	139
Ganglioneuroblastoma	251877	<10	<10
Ganglioneuroma	251992	14	14
GAPO syndrome	2067	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Gastrocutaneous syndrome	2069	<10	<10
Gastrointestinal stromal tumor	44890	<10	<10
Gastroschisis	2368	278	266
Gaucher disease	355	514	472
Geleophysic dysplasia	2623	22	19
Gemignani syndrome	2074	<10	<10
Generalized arterial calcification of infancy	51608	24	21
Generalized basaloid follicular hamartoma syndrome	168632	<10	<10
Generalized bulbospinal muscular atrophy*	206710	56	52
Generalized epilepsy with febrile seizures-plus	36387	603	598
Generalized epilepsy-paroxysmal dyskinesia syndrome	79137	<10	<10
Generalized essential telangiectasia	280774	11	11
Generalized glucocorticoid resistance syndrome	786	11	<10
Generalized peeling skin syndrome	263543	<10	<10
Generalized pustular psoriasis	247353	20	19
Genetic central precocious puberty in female	650077	24	24
Genetic hyperferritinemia without iron overload	254704	199	197
Genetic non-syndromic obesity	98267	615	612
Genetic recurrent myoglobinuria	99845	15	15
Genetic steroid-resistant nephrotic syndrome	656	223	219
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	11	11
Giant adenofibroma of the breast	180267	77	77
Giant axonal neuropathy	643	13	12
Giant cell arteritis	397	4770	4578
Giant cell tumor of bone	363976	<10	<10
Gingival fibromatosis-facial dysmorphism syndrome	2025	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Gingival fibromatosis-hypertrichosis syndrome	2026	**<10**	**<10**
Gingival fibromatosis-progressive deafness syndrome	2027	**<10**	**<10**
Gitelman syndrome	358	**729**	**724**
Gitelman-like kidney tubulopathy due to mitochondrial DNA mutation	620371	**<10**	**<10**
GJC2-related late-onset primary lymphedema	568051	**<10**	**<10**
Glanzmann thrombasthenia	849	**376**	**367**
Glaucoma secondary to spherophakia/ectopia lentis and megalocornea	238763	**13**	**13**
Glaucoma-sleep apnea syndrome	2085	**<10**	**<10**
Glaucomatocyclitic crisis disease	636950	**<10**	**<10**
Glial tumor of neuroepithelial tissue with unknown origin*	251668	**<10**	**<10**
Glial tumor*	182067	**36**	**32**
Glioblastoma	360	**42**	**15**
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-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome	488613	**<10**	**<10**
Global developmental delay-osteopenia-ectodermal defect syndrome	73223	**<10**	**<10**
Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	480898	**11**	**<10**
Glomus tumor	391651	**<10**	**<10**
Glomuvenous malformation	83454	**102**	**102**
Glossopalatine ankylosis	141163	**<10**	**<10**
Glucagonoma	97280	**<10**	**<10**
Glucose-galactose malabsorption	35710	**<10**	**<10**
Glutaric acidemia type 3	35706	**<10**	**<10**
Glutaryl-CoA dehydrogenase deficiency	25	**160**	**150**
Glutathione synthetase deficiency	32	**16**	**14**
Glycerol kinase deficiency*	308993	**<10**	**<10**
Glycine encephalopathy	407	**103**	**68**
Glycogen storage disease due to acid maltase deficiency	365	**472**	**397**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Glycogen storage disease due to aldolase A deficiency	57	**<10**	**<10**
Glycogen storage disease due to glucose-6-phosphatase deficiency	364	**314**	**308**
Glycogen storage disease due to glycogen branching enzyme deficiency	367	**36**	**33**
Glycogen storage disease due to glycogen debranching enzyme deficiency	366	**213**	**205**
Glycogen storage disease due to glycogen synthase deficiency*	308520	**<10**	**<10**
Glycogen storage disease due to hepatic glycogen synthase deficiency	2089	**20**	**19**
Glycogen storage disease due to lactate dehydrogenase deficiency	2364	**<10**	**<10**
Glycogen storage disease due to LAMP-2 deficiency	34587	**41**	**37**
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	**22**	**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	**348**	**335**
Glycogen storage disease due to muscle phosphofructokinase deficiency	371	**<10**	**<10**
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	**121**	**117**
Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	263297	**<10**	**<10**
GM1 gangliosidosis	354	**115**	**74**
GM2 gangliosidosis*	309152	**15**	**<10**
GM2 gangliosidosis, AB variant	309246	**<10**	**<10**
GM3 synthase deficiency	370933	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
GMPPB-related limb-girdle muscular dystrophy R19	363623	<10	<10
GMS syndrome	2090	<10	<10
GNAO1-related developmental delay-seizures-movement disorder spectrum	592564	22	22
Gnathodiaphyseal dysplasia	53697	<10	<10
GNB5-related intellectual disability-cardiac arrhythmia syndrome	542306	<10	<10
GNE myopathy	602	93	87
Goldberg-Shprintzen megacolon syndrome	66629	<10	<10
Goldmann-Favre syndrome	53540	48	48
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	73	64
Gordon syndrome	376	19	19
Gorham-Stout disease	73	51	48
Gorlin syndrome	377	472	465
Gorlin-Chaudhry-Moss syndrome	2095	<10	<10
GRACILE syndrome	53693	<10	<10
Graft versus host disease	39812	101	88
Graham Little-Piccardi-Lassueur syndrome	505	<10	<10
Grange syndrome	79094	<10	<10
Granular corneal dystrophy type I	98962	34	34
Granular corneal dystrophy type II	98963	13	13
Granuloma faciale	615943	<10	<10
Granulomatosis with polyangiitis	900	2064	1952
Granulomatous mastitis	64722	53	52
Granulomatous slack skin	33111	<10	<10
Gray platelet syndrome	721	29	29
Greenberg dysplasia	1426	<10	<10
Greig cephalopolysyndactyly syndrome	380	226	226
GRFoma	97261	<10	<10
GRIN2B-related developmental delay, intellectual disability and autism spectrum disorder	589547	36	36
Griscelli syndrome	381	27	25
Growing teratoma syndrome	314613	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	391348	<10	<10
Growth delay due to insulin-like growth factor I resistance	73273	32	32
Growth delay due to insulin-like growth factor type 1 deficiency	73272	42	42
Growth delay-hydrocephaly-lung hypoplasia syndrome	3035	<10	<10
Grubben-de Cock-Borghgraef syndrome	2101	<10	<10
Guanidinoacetate methyltransferase deficiency	382	32	31
Guillain-Barré syndrome*	2103	1787	1660
Guttmacher syndrome	2957	<10	<10
Gyrate atrophy of choroid and retina	414	38	37
H syndrome	168569	17	17
Haddad syndrome	99803	22	14
Hajdu-Cheney syndrome	955	49	45
Hallermann-Streiff syndrome	2108	25	23
Hallermann-Streiff-like syndrome	2109	<10	<10
Hallux varus-preaxial polysyndactyly syndrome	2110	<10	<10
HANAC syndrome	73229	32	28
Hand-foot-genital syndrome	2438	<10	<10
Hao-Fountain syndrome	643549	<10	<10
Harlequin ichthyosis	457	25	18
Harlequin syndrome	199282	<10	<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	11	<10
HELLP syndrome	244242	37	36
Hemangioblastoma	252054	11	11
Hemifacial hyperplasia	141145	29	29
Hemifacial spasm	221083	285	280
Hemihyperplasia-multiple lipomatosis syndrome	276280	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hemimegalencephaly	99802	**96**	**94**
Hemimelia*	2130	**<10**	**<10**
Hemoglobin C disease	2132	**217**	**216**
Hemoglobin C-beta-thalassemia syndrome	231242	**54**	**54**
Hemoglobin D disease	90039	**<10**	**<10**
Hemoglobin E disease	2133	**62**	**62**
Hemoglobin E-beta-thalassemia syndrome	231249	**55**	**55**
Hemoglobin Lepore-beta-thalassemia syndrome	330032	**<10**	**<10**
Hemoglobin M disease	330041	**<10**	**<10**
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	**135**	**130**
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	**9028**	**8884**
Hemophilia B	98879	**2143**	**2105**
Hemophilia*	448	**34**	**33**
Hemorrhagic fever-renal syndrome	340	**<10**	**<10**
Hennekam syndrome	2136	**30**	**29**
Hennekam-Beemer syndrome	2135	**<10**	**<10**
Heparin-induced thrombocytopenia	3325	**<10**	**<10**
Hepatic cystic hamartoma	386	**48**	**46**
Hepatic fibrosis-renal cysts-intellectual disability syndrome	2031	**<10**	**<10**
Hepatic veno-occlusive disease	890	**334**	**298**
Hepatitis delta	402823	**<10**	**<10**
Hepatoblastoma	449	**20**	**20**
Hepatocellular adenoma	54272	**87**	**84**
Hepatocellular carcinoma*	88673	**17**	**12**
Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	137681	**<10**	**<10**
Hepatoerythropoietic porphyria	95159	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hepatosplenic T-cell lymphoma	86882	**<10**	**<10**
Hereditary acrokeratotic poikiloderma	2907	**<10**	**<10**
Hereditary amyloidosis with primary renal involvement	85450	**180**	**175**
Hereditary angioedema with C1Inh deficiency	528623	**979**	**974**
Hereditary angioedema with normal C1Inh	528647	**372**	**370**
Hereditary angioedema*	91378	**68**	**66**
Hereditary arterial and articular multiple calcification syndrome	289601	**<10**	**<10**
Hereditary ATTR amyloidosis*	271861	**1357**	**1185**
Hereditary benign intraepithelial dyskeratosis	352657	**<10**	**<10**
Hereditary breast and/or ovarian cancer syndrome	145	**1054**	**956**
Hereditary breast cancer	227535	**47**	**43**
Hereditary cerebral hemorrhage with amyloidosis	85458	**<10**	**<10**
Hereditary chronic pancreatitis	676	**322**	**318**
Hereditary combined deficiency of vitamin K-dependent clotting factors	98434	**19**	**19**
Hereditary continuous muscle fiber activity	972	**12**	**<10**
Hereditary coproporphyria	79273	**82**	**66**
Hereditary diffuse gastric cancer	26106	**<10**	**<10**
Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia	313808	**19**	**16**
Hereditary elliptocytosis	288	**141**	**140**
Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome	221043	**<10**	**<10**
Hereditary folate malabsorption	90045	**<10**	**<10**
Hereditary fructose intolerance	469	**196**	**194**
Hereditary geniospasm	53372	**<10**	**<10**
Hereditary gingival fibromatosis	2024	**11**	**11**
Hereditary hemorrhagic telangiectasia	774	**3695**	**3561**
Hereditary hypercarotenemia and vitamin A deficiency	199285	**<10**	**<10**
Hereditary hyperekplexia	3197	**76**	**72**
Hereditary hyperferritinemia-cataract syndrome	163	**175**	**173**
Hereditary hypophosphatemic rickets with hypercalciuria	157215	**72**	**72**
Hereditary inclusion body myopathy type 4	324381	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome	79091	**<10**	**<10**
Hereditary isolated aplastic anemia	397692	**39**	**37**
Hereditary late-onset Parkinson disease	411602	**47**	**44**
Hereditary leiomyomatosis and renal cell cancer	523	**276**	**274**
Hereditary methemoglobinemia	621	**21**	**21**
Hereditary motor and sensory neuropathy type 5	64751	**<10**	**<10**
Hereditary motor and sensory neuropathy type 6	90120	**36**	**35**
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	**90**	**89**
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	**1333**	**1311**
Hereditary neutrophilia	279943	**13**	**13**
Hereditary nonpolyposis colon cancer*	443909	**55**	**44**
Hereditary orotic aciduria	30	**<10**	**<10**
Hereditary painful callosities	79141	**15**	**15**
Hereditary palmoplantar keratoderma, Gamborg-Nielsen type	86923	**<10**	**<10**
Hereditary papillary renal cell carcinoma	47044	**12**	**12**
Hereditary pediatric Behçet-like disease	476102	**98**	**98**
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	**<10**	**<10**
Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	251380	**62**	**62**
Hereditary pheochromocytoma-paraganglioma	29072	**895**	**877**
Hereditary progressive mucinous histiocytosis	158025	**<10**	**<10**
Hereditary pulmonary alveolar proteinosis	264675	**23**	**22**
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	**30**	**28**
Hereditary sensory and autonomic neuropathy type 1B	139564	**16**	**16**
Hereditary sensory and autonomic neuropathy type 2	970	**19**	**18**
Hereditary sensory and autonomic neuropathy type 4	642	**20**	**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	**97**	**95**
Hereditary sensory neuropathy-deafness-dementia syndrome	456318	**<10**	**<10**
Hereditary spherocytosis	822	**1487**	**1481**
Hereditary stomatocytosis*	98365	**21**	**21**
Hereditary thermosensitive neuropathy	84093	**<10**	**<10**
Hereditary thrombocytopenia with early-onset myelofibrosis	480851	**<10**	**<10**
Hereditary thrombocytopenia with normal platelets	268322	**85**	**84**
Hereditary thrombophilia due to congenital antithrombin deficiency	82	**34**	**34**
Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	217467	**<10**	**<10**
Hereditary xanthinuria	3467	**12**	**12**
Hermansky-Pudlak syndrome	79430	**86**	**81**
Herpes simplex virus encephalitis	1930	**93**	**88**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Herpes simplex virus stromal keratitis	137599	**194**	**193**
Herpetiform pemphigus	208524	**19**	**18**
Hidrotic ectodermal dysplasia	189	**119**	**118**
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	**174**	**171**
Hip dysplasia, Beukes type	2114	**12**	**12**
Hirschsprung disease	388	**1789**	**1765**
Hirschsprung disease-ganglioneuroblastoma syndrome	2151	**<10**	**<10**
Hirschsprung disease-nail hypoplasia-dysmorphism syndrome	2153	**<10**	**<10**
His bundle tachycardia	3283	**18**	**18**
Histidinemia	2157	**<10**	**<10**
Histiocytic sarcoma	86896	**24**	**15**
Histiocytoid cardiomyopathy	137675	**<10**	**<10**
Histoplasmosis	390	**<10**	**<10**
HJV or HAMP-related hemochromatosis	79230	**61**	**59**
HNRNPDL-related limb-girdle muscular dystrophy D3	55596	**<10**	**<10**
Hodgkin lymphoma*	98293	**36**	**36**
Holmes-Adie syndrome	454718	**<10**	**<10**
Holocarboxylase synthetase deficiency	79242	**11**	**11**
Holoprosencephaly	2162	**137**	**119**
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	**326**	**316**
Holzgreve syndrome	2167	**<10**	**<10**
Homocystinuria due to methylene tetrahydrofolate reductase deficiency	395	**139**	**138**
Homocystinuria without methylmalonic aciduria	622	**36**	**33**
Homozygous familial hypercholesterolemia	391665	**71**	**71**
Horizontal gaze palsy with progressive scoliosis	2744	**13**	**13**
Hot water reflex epilepsy	166412	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
House allergic alveolitis	99907	**442**	**394**
Hoyeraal-Hreidarsson syndrome	3322	**<10**	**<10**
HSD10 disease	391417	**<10**	**<10**
HTRA1-related autosomal dominant cerebral small vessel disease	482077	**32**	**32**
Humeral agenesis/hypoplasia	294973	**<10**	**<10**
Humero-radial synostosis	3265	**23**	**23**
Humero-radio-ulnar synostosis	3266	**<10**	**<10**
Humero-ulnar synostosis	94056	**<10**	**<10**
Huntington disease	399	**3221**	**2876**
Huntington disease-like 1	157941	**<10**	**<10**
Huntington disease-like 2	98934	**47**	**47**
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	**18**	**16**
Hyaline fibromatosis syndrome	498474	**17**	**13**
Hydatidiform mole	99927	**11**	**<10**
Hydranencephaly	2177	**<10**	**<10**
Hydroa vacciniforme	330058	**<10**	**<10**
Hydroa vacciniforme-like lymphoma	364039	**<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	**32**	**20**
Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome	528091	**<10**	**<10**
Hyperammonemia due to N-acetylglutamate synthase deficiency	927	**30**	**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	214	210
Hypergonadotropic hypogonadism-cataract syndrome	2410	<10	<10
Hyper-IgE syndrome*	331223	20	19
Hyper-IgM syndrome with susceptibility to opportunistic infections	183663	36	34
Hyper-IgM syndrome without susceptibility to opportunistic infections	183666	13	13
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	22	20
Hyperinsulinism-hyperammonemia syndrome	35878	44	44
Hyperkalemic periodic paralysis	682	111	109
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	2905	2900
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	415	26	25
Hyperostosis corticalis generalisata	3416	<10	<10
Hyperostosis cranialis interna	443098	<10	<10
Hyperparathyroidism-jaw tumor syndrome	99880	29	29
Hyperphalangy	295002	<10	<10
Hyperphenylalaninemia due to DNAJC12 deficiency	508523	<10	<10
Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	238583	128	127
Hyperphosphatasia-intellectual disability syndrome	247262	12	12
Hyperprolinemia type 1	419	23	22
Hyperprolinemia type 2	79101	<10	<10
Hypersensitivity pneumonitis*	31740	605	551

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
Hypobetalipoproteinemia*	31154	20	20
Hypocalcemic rickets*	289103	71	71
Hypocalcemic vitamin D-dependent rickets	289157	153	151
Hypocalcemic vitamin D-resistant rickets	93160	51	51
Hypochondroplasia	429	418	417
Hypocomplementemic urticarial vasculitis	36412	107	105
Hypodontia-dysplasia of nails syndrome	2228	<10	<10
Hypoglossia-hypodactyly syndrome	989	29	28
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	<10	<10
Hypohidrotic ectodermal dysplasia	238468	932	928
Hypohidrotic ectodermal dysplasia with immunodeficiency	98813	<10	<10
Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome	1882	32	32
Hypoinsulinemic hypoglycemia and body hemihypertrophy	293964	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hypokalemic periodic paralysis	681	**256**	**254**
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	**112**	**111**
Hypophosphatasia	436	**492**	**486**
Hypophosphatemic rickets*	437	**444**	**442**
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	**227**	**177**
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**
Hypospadias-intellectual disability, Goldblatt type syndrome	2261	**<10**	**<10**
Hypothalamic adipsic hypernatraemia syndrome	443101	**<10**	**<10**
Hypothalamic hamartomas with gelastic seizures	86906	**63**	**63**
Hypothyroidism due to deficient transcription factors involved in pituitary development or function	226307	**<10**	**<10**
Hypothyroidism due to TSH receptor mutations	90673	**92**	**91**
Hypotonia-cystinuria syndrome	163690	**<10**	**<10**
Hypotonia-cystinuria type 1 syndrome*	238517	**<10**	**<10**
Hypotonia-failure to thrive-microcephaly syndrome	79507	**61**	**58**
Hypotonia-speech impairment-severe cognitive delay syndrome	371364	**12**	**12**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Hypotrichosis simplex	55654	**35**	**35**
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	**19**	**16**
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	**17**	**17**
Ichthyosis-short stature-brachydactyly-microspherophakia syndrome	363992	**<10**	**<10**
Idiopathic achalasia	930	**104**	**103**
Idiopathic acute eosinophilic pneumonia	724	**13**	**12**
Idiopathic aplastic anemia	88	**951**	**894**
Idiopathic bilateral vestibulopathy	171684	**13**	**13**
Idiopathic bronchiectasis	60033	**1559**	**1541**
Idiopathic camptocormia	1320	**223**	**197**
Idiopathic CD4 lymphocytopenia	228000	**66**	**65**
Idiopathic chronic eosinophilic pneumonia	2902	**133**	**130**
Idiopathic congenital hypothyroidism	95717	**675**	**673**
Idiopathic dropped head syndrome	447881	**21**	**21**
Idiopathic ductopenia	480512	**22**	**20**
Idiopathic eosinophilic myositis	247724	**<10**	**<10**
Idiopathic eosinophilic pneumonia*	182101	**20**	**20**
Idiopathic gastroparesis	558411	**161**	**160**
Idiopathic giant cell myocarditis	329874	**57**	**57**
Idiopathic hemiconvulsion-hemiplegia syndrome	86908	**64**	**64**
Idiopathic hypercalciuria	2197	**1017**	**1013**
Idiopathic hypereosinophilic syndrome	3260	**585**	**567**
Idiopathic hypersomnia	33208	**3275**	**3269**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Idiopathic interstitial pneumonia*	98300	**439**	**401**
Idiopathic intracranial hypertension	238624	**58**	**58**
Idiopathic isolated micropenis	95707	**539**	**536**
Idiopathic juvenile osteoporosis	85193	**735**	**721**
Idiopathic linear interstitial keratitis	314017	**<10**	**<10**
Idiopathic localized lipodystrophy	90158	**<10**	**<10**
Idiopathic macular telangiectasia type 1	353344	**12**	**12**
Idiopathic macular telangiectasia type 3	353351	**<10**	**<10**
Idiopathic neonatal atrial flutter	45452	**<10**	**<10**
Idiopathic nephrotic syndrome*	357502	**906**	**886**
Idiopathic non-lupus full-house nephropathy	567544	**<10**	**<10**
Idiopathic optic perineuritis	499107	**<10**	**<10**
Idiopathic panuveitis	280921	**960**	**958**
Idiopathic peliosis hepatis	480524	**154**	**151**
Idiopathic phalangeal acro-osteolysis	444316	**<10**	**<10**
Idiopathic pleuroparenchymal fibroelastosis	494428	**223**	**192**
Idiopathic posterior uveitis	280917	**512**	**508**
Idiopathic pregnancy-associated osteoporosis	647823	**<10**	**<10**
Idiopathic pulmonary artery dilatation	1676	**<10**	**<10**
Idiopathic pulmonary fibrosis	2032	**4987**	**4117**
Idiopathic pulmonary hemosiderosis	99931	**55**	**54**
Idiopathic recurrent pericarditis	251307	**386**	**383**
Idiopathic scleritis	648675	**26**	**26**
Idiopathic spontaneous coronary artery dissection	458718	**87**	**87**
Idiopathic steroid-resistant nephrotic syndrome	567548	**290**	**290**
Idiopathic steroid-sensitive nephrotic syndrome	69061	**4212**	**4169**
Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance	567546	**45**	**45**
Idiopathic trachyonychia	79153	**<10**	**<10**
Idiopathic uveal effusion syndrome	209956	**<10**	**<10**
Idiopathic ventricular fibrillation, non Brugada type	228140	**733**	**608**
Idiopathic/heritable pulmonary arterial hypertension	422	**3339**	**2893**
IgA pemphigus	555905	**18**	**18**
IgG4-related disease*	284264	**297**	**282**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
IgG4-related systemic disease	596448	**958**	**918**
Ileal neuroendocrine tumor	100078	**<10**	**<10**
IMAGe syndrome	85173	**14**	**14**
Imerslund-Gräsbeck syndrome	35858	**49**	**49**
Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome	238569	**632**	**632**
Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections-lymphopenia syndrome	529977	**<10**	**<10**
Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	37042	**60**	**52**
Immune thrombocytopenia	3002	**7957**	**7675**
Immune-mediated necrotizing myopathy	206569	**682**	**627**
Immune-mediated scleritis	648681	**<10**	**<10**
Immunodeficiency by defective expression of MHC class I	34592	**<10**	**<10**
Immunodeficiency by defective expression of MHC class II	572	**33**	**30**
Immunodeficiency due to a classical component pathway complement deficiency	169147	**44**	**44**
Immunodeficiency due to a late component of complement deficiency	169150	**42**	**42**
Immunodeficiency due to CD25 deficiency	169100	**<10**	**<10**
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	70592	**11**	**<10**
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	70593	**82**	**81**
Immunodeficiency with factor H anomaly	200421	**<10**	**<10**
Immunodeficiency with factor I anomaly	200418	**<10**	**<10**
Immunoglobulin A vasculitis	761	**2842**	**2801**
Immunoglobulin heavy chain deficiency	169110	**<10**	**<10**
Immunotactoid glomerulopathy	97567	**<10**	**<10**
Immunotactoid or fibrillary glomerulopathy*	91137	**12**	**12**
Immunotherapy induced hypophysitis	641350	**<10**	**<10**
Incessant infant ventricular tachycardia	45453	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	52430	**29**	**19**
Inclusion body myositis	611	**1186**	**948**
Incontinentia pigmenti	464	**650**	**646**
Indeterminate cell histiocytosis	158019	**<10**	**<10**
Indolent primary cutaneous T-cell lymphoma	178548	**<10**	**<10**
Indolent systemic mastocytosis	98848	**1655**	**1645**
Infant acute respiratory distress syndrome	70587	**40**	**37**
Infantile apnea	70590	**2018**	**1992**
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	**12**	**12**
Infantile digital fibromatosis	199267	**<10**	**<10**
Infantile dystonia-parkinsonism	238455	**12**	**11**
Infantile epileptic-dyskinetic encephalopathy	364063	**34**	**34**
Infantile hemangioma of rare localization*	210589	**2154**	**2154**
Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	352563	**<10**	**<10**
Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome	522077	**<10**	**<10**
Infantile multisystem neurologic-endocrine-pancreatic disease	456312	**<10**	**<10**
Infantile myofibromatosis	2591	**24**	**23**
Infantile neuroaxonal dystrophy	35069	**66**	**40**
Infantile neuronal ceroid lipofuscinosis	79263	**27**	**19**
Infantile neurovisceral acid sphingomyelinase deficiency	77292	**31**	**14**
Infantile onset panniculitis with uveitis and systemic granulomatosis	251304	**<10**	**<10**
Infantile osteopetrosis with neuroaxonal dysplasia	85179	**<10**	**<10**
Infantile Refsum disease	772	**13**	**12**
Infantile spasms syndrome	3451	**1944**	**1876**
Infantile spasms-broad thumbs syndrome	3173	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Infantile-onset ascending hereditary spastic paralysis	293168	**<10**	**<10**
Infantile-onset autosomal recessive nonprogressive cerebellar ataxia	284332	**13**	**13**
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**	**76**
Infantile-onset X-linked spinal muscular atrophy	1145	**12**	**11**
Infection-related hemolytic uremic syndrome	544482	**1897**	**1867**
Infectious anterior uveitis	279922	**172**	**172**
Infectious epithelial keratitis	137593	**71**	**69**
Infectious panuveitis	279925	**79**	**79**
Infectious posterior uveitis	279919	**196**	**188**
Infective endocarditis	570762	**12**	**12**
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	**<10**	**<10**
Inflammatory pseudotumor of the liver	90003	**13**	**11**
Infundibulo-neurohypophysitis	238305	**<10**	**<10**
Inherited acute myeloid leukemia	319465	**<10**	**<10**
Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations	319462	**14**	**14**
Inherited congenital spastic tetraplegia	210141	**25**	**23**
Inherited Creutzfeldt-Jakob disease	282166	**<10**	**<10**
Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency	289548	**<10**	**<10**
Insulin autoimmune syndrome	411593	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Insulinoma	97279	**25**	**24**
Insulin-resistance syndrome type A	2297	**23**	**23**
Insulin-resistance syndrome type B	2298	**<10**	**<10**
Intellectual disability, Birk-Barel type	166108	**<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	**33**	**33**
Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	397709	**<10**	**<10**
Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	329224	**21**	**21**
Intellectual disability-developmental delay-contractures syndrome	3454	**18**	**18**
Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome	3044	**<10**	**<10**
Intellectual disability-epilepsy-extrapyramidal syndrome	468620	**<10**	**<10**
Intellectual disability-expressive aphasia-facial dysmorphism syndrome	436151	**<10**	**<10**
Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	404440	**61**	**61**
Intellectual disability-facial dysmorphism-hand anomalies syndrome	370010	**<10**	**<10**
Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome	1495	**18**	**18**
Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome	457279	**13**	**13**
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-obesity-brain malformations-facial dysmorphism syndrome	352530	**<10**	**<10**
Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome	513456	**21**	**21**
Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	369837	**<10**	**<10**
Intellectual disability-seizures-macrocephaly-obesity syndrome	369950	**<10**	**<10**
Intellectual disability-severe speech delay-mild dysmorphism syndrome	391372	**64**	**64**
Intellectual disability-short stature-hypertelorism syndrome	3074	**<10**	**<10**
Intellectual disability-spasticity-ectrodactyly syndrome	1891	**<10**	**<10**
Intellectual disability-strabismus syndrome	363528	**23**	**23**
Interatrial communication	1478	**1144**	**1128**
Intercalary limb defects*	294927	**<10**	**<10**
Intermediate atrioventricular septal defect	576242	**134**	**132**
Intermediate collagen VI-related muscular dystrophy	646113	**<10**	**<10**
Intermediate generalized junctional epidermolysis bullosa	79402	**11**	**11**
Intermediate nemaline myopathy	171433	**11**	**11**
Intermediate osteopetrosis	210110	**<10**	**<10**
Intermediate uveitis	279914	**584**	**584**
Internal carotid absence	981	**<10**	**<10**
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	**50**	**45**
Interventricular septum aneurysm	99092	**<10**	**<10**
Intestinal lymphangiectasia*	36204	**29**	**29**
Intestinal polyposis syndrome*	104010	**62**	**58**
Intraductal papillary mucinous carcinoma of pancreas	424058	**44**	**42**
Intrahepatic cholestasis of pregnancy	69665	**124**	**123**
Intraneural perineurioma	100003	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
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**
Intravascular large B-cell lymphoma	98839	**<10**	**<10**
Invasive Candidiasis	636945	**<10**	**<10**
Invasive mole	99925	**<10**	**<10**
Invasive non-typhoidal salmonellosis	324648	**<10**	**<10**
Inverse Klippel-Trénaunay syndrome	329324	**13**	**13**
Inverted duplicated chromosome 15 syndrome	3306	**116**	**113**
IRF2BPL-related regressive neurodevelopmental disorder-dystonia-seizures syndrome	597623	**<10**	**<10**
IRIDA syndrome	209981	**22**	**21**
Iridocorneal endothelial syndrome	64734	**12**	**12**
IRVAN syndrome	209943	**<10**	**<10**
Isaacs syndrome	84142	**129**	**121**
Ischiovertebral syndrome	85200	**<10**	**<10**
Isobutyryl-CoA dehydrogenase deficiency	79159	**<10**	**<10**
Isochromosomy Yp	98797	**<10**	**<10**
Isochromosomy Yq	98798	**<10**	**<10**
Isolated agammaglobulinemia	229717	**423**	**413**
Isolated agenesis of gallbladder	440987	**<10**	**<10**
Isolated amyelia	268868	**<10**	**<10**
Isolated anencephaly/exencephaly	1048	**<10**	**<10**
Isolated aniridia	250923	**516**	**511**
Isolated ankyloblepharon filiforme adnatum	91397	**<10**	**<10**
Isolated anterior cervical hypertrichosis	3387	**<10**	**<10**
Isolated arhinencephaly	268936	**<10**	**<10**
Isolated arrhinia	1134	**<10**	**<10**
Isolated asymptomatic elevation of creatine phosphokinase	206599	**612**	**593**
Isolated ATP synthase deficiency	254913	**<10**	**<10**
Isolated autosomal dominant hypomagnesemia, Glaudemans type	199326	**<10**	**<10**
Isolated bilateral hemispheric cerebellar hypoplasia	269221	**<10**	**<10**
Isolated biliary atresia	30391	**1557**	**1488**
Isolated blepharochalasis	519390	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isolated bone marrow mastocytosis	158778	**<10**	**<10**
Isolated cerebellar agenesis	1398	**254**	**248**
Isolated cerebellar vermis agenesis	269203	**<10**	**<10**
Isolated cerebellar vermis hypoplasia	199630	**81**	**80**
Isolated childhood apraxia of speech	209908	**71**	**71**
Isolated cleft lip	199302	**595**	**594**
Isolated complex I deficiency	2609	**82**	**59**
Isolated complex III deficiency	1460	**40**	**29**
Isolated congenital alacrima	91416	**14**	**14**
Isolated congenital anonychia	79143	**<10**	**<10**
Isolated congenital anosmia	88620	**11**	**11**
Isolated congenital auditory ossicle malformation	162526	**83**	**83**
Isolated congenital breast hypoplasia/aplasia	180188	**25**	**25**
Isolated congenital digital clubbing	217059	**<10**	**<10**
Isolated congenital ectropion	99171	**<10**	**<10**
Isolated congenital hepatic fibrosis	485426	**207**	**197**
Isolated congenital hypoglossia/aglossia	141152	**<10**	**<10**
Isolated congenital hypogonadotropic hypogonadism	238666	**2186**	**2170**
Isolated congenital megalocornea	91489	**39**	**38**
Isolated congenital microcephaly	199642	**482**	**466**
Isolated congenital nasal pyriform aperture stenosis	162516	**148**	**147**
Isolated congenital onychodysplasia	79144	**<10**	**<10**
Isolated congenital radial head dislocation	295032	**<10**	**<10**
Isolated congenital sclerocornea	91490	**40**	**40**
Isolated congenital syngnathia	141214	**<10**	**<10**
Isolated corpus callosum agenesis	200	**683**	**678**
Isolated cryptophthalmia	91396	**<10**	**<10**
Isolated cytochrome C oxidase deficiency	254905	**29**	**26**
Isolated Dandy-Walker malformation	217	**91**	**85**
Isolated delta-storage pool disease	248340	**125**	**125**
Isolated diffuse palmoplantar keratoderma*	307148	**84**	**84**
Isolated distichiasis	99177	**<10**	**<10**
Isolated ectopia lentis	1885	**153**	**152**
Isolated encephalocele	199647	**21**	**21**
Isolated female hypospadias	603515	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isolated filum lipoma	645325	**57**	**57**
Isolated focal cortical dysplasia	65683	**1366**	**1358**
Isolated focal non-epidermolytic palmoplantar keratoderma	448264	**<10**	**<10**
Isolated focal palmoplantar keratoderma*	307846	**34**	**34**
Isolated follicle stimulating hormone deficiency	52901	**<10**	**<10**
Isolated foveal hypoplasia	519398	**65**	**65**
Isolated generalized anhidrosis with normal sweat glands	468666	**<10**	**<10**
Isolated glycerol kinase deficiency	408	**<10**	**<10**
Isolated hemihyperplasia	2128	**279**	**277**
Isolated hereditary congenital facial paralysis	306527	**22**	**22**
Isolated hyperchlorhidrosis	542657	**<10**	**<10**
Isolated Klippel-Feil syndrome	2345	**156**	**154**
Isolated lissencephaly type 1 without known genetic defects	1084	**24**	**23**
Isolated megalopapilla	519402	**<10**	**<10**
Isolated mesenteric vein thrombosis	583861	**26**	**23**
Isolated microphthalmia-anophthalmia-coloboma*	2542	**140**	**139**
Isolated microspherophakia	519396	**<10**	**<10**
Isolated neonatal sclerosing cholangitis	480556	**29**	**29**
Isolated optic nerve hypoplasia	637061	**<10**	**<10**
Isolated optic neuritis	499096	**234**	**233**
Isolated osteopoikilosis	166119	**<10**	**<10**
Isolated partial vaginal agenesis	96269	**16**	**16**
Isolated permanent neonatal diabetes mellitus	99885	**69**	**65**
Isolated persistent urogenital sinus	647794	**<10**	**<10**
Isolated Pierre Robin syndrome	718	**2125**	**2101**
Isolated polycystic liver disease	2924	**527**	**506**
Isolated posterior meningocele	268810	**29**	**29**
Isolated pulmonary capillaritis	264691	**<10**	**<10**
Isolated punctate palmoplantar keratoderma*	2338	**26**	**26**
Isolated right ventricular hypoplasia	439	**27**	**26**
Isolated spina bifida*	823	**616**	**598**
Isolated splenic vein thrombosis	583856	**<10**	**<10**
Isolated split hand-split foot malformation	2440	**387**	**384**
Isolated succinate-CoQ reductase deficiency	3208	**18**	**12**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Isolated thyroid-stimulating hormone deficiency	90674	**128**	**128**
Isolated thyrotropin-releasing hormone deficiency	238670	**<10**	**<10**
Isolated tracheoesophageal fistula	454750	**71**	**69**
Isolated transitional filum lipoma	645322	**<10**	**<10**
Isolated unilateral hemispheric cerebellar hypoplasia	269218	**14**	**14**
Isotretinoin syndrome	2305	**<10**	**<10**
Isotretinoin-like syndrome	2306	**<10**	**<10**
Isovaleric acidemia	33	**105**	**98**
ISPD-related limb-girdle muscular dystrophy R20	352479	**<10**	**<10**
ITM2B amyloidosis	439254	**13**	**12**
ITPA-related lethal infantile neurological disorder with cataract and cardiac involvement	457375	**<10**	**<10**
Jackson-Weiss syndrome	1540	**<10**	**<10**
Jacobsen syndrome	2308	**81**	**76**
Jalili syndrome	1873	**<10**	**<10**
Jeavons syndrome	139431	**172**	**171**
Jervell and Lange-Nielsen syndrome	90647	**27**	**27**
Jessner lymphocytic infiltration of the skin	33314	**<10**	**<10**
Jeune syndrome	474	**108**	**97**
Johanson-Blizzard syndrome	2315	**11**	**<10**
Johnson neuroectodermal syndrome	2316	**<10**	**<10**
Joubert syndrome	475	**462**	**450**
Joubert syndrome with hepatic defect	1454	**<10**	**<10**
Joubert syndrome with Jeune asphyxiating thoracic dystrophy	397715	**<10**	**<10**
Joubert syndrome with ocular defect	220493	**20**	**19**
Joubert syndrome with oculorenal defect	2318	**15**	**15**
Joubert syndrome with renal defect	220497	**14**	**14**
Junctional epidermolysis bullosa inversa	79405	**<10**	**<10**
Junctional epidermolysis bullosa with pyloric atresia	79403	**<10**	**<10**
Junctional epidermolysis bullosa*	305	**105**	**79**
Juvenile absence epilepsy	1941	**653**	**651**
Juvenile amyotrophic lateral sclerosis	300605	**20**	**17**
Juvenile glaucoma	98977	**74**	**73**
Juvenile Huntington disease	248111	**29**	**24**
Juvenile idiopathic arthritis*	92	**863**	**861**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Juvenile myelomonocytic leukemia	86834	**16**	**12**
Juvenile myoclonic epilepsy	307	**1029**	**1023**
Juvenile nasopharyngeal angiofibroma	289596	**20**	**20**
Juvenile neuronal ceroid lipofuscinosis	79264	**66**	**53**
Juvenile overlap myositis	329894	**<10**	**<10**
Juvenile Paget disease	2801	**20**	**19**
Juvenile polymyositis	93568	**<10**	**<10**
Juvenile polyposis syndrome	2929	**62**	**62**
Juvenile primary lateral sclerosis	247604	**<10**	**<10**
Juvenile temporal arteritis	26137	**<10**	**<10**
Juvenile xanthogranuloma	158000	**63**	**63**
Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	445062	**<10**	**<10**
Kabuki syndrome	2322	**545**	**530**
Kagami-Ogata syndrome	254519	**<10**	**<10**
Kallmann syndrome-heart disease syndrome	2326	**14**	**13**
Kaposi sarcoma	33276	**25**	**21**
Kaposiform hemangioendothelioma	2122	**37**	**37**
Karyomegalic interstitial nephritis	401996	**12**	**<10**
Kasabach-Merritt syndrome	2330	**33**	**33**
Kawasaki disease	2331	**776**	**772**
KBG syndrome	2332	**468**	**468**
KCNQ2-related epileptic encephalopathy	439218	**126**	**124**
KDM3B-related intellectual disability-facial dysmorphism-short stature syndrome	633004	**<10**	**<10**
KDM5C-related syndromic X-linked intellectual disability	85279	**28**	**28**
Kearns-Sayre syndrome	480	**204**	**181**
Keipert syndrome	2662	**<10**	**<10**
Kennedy disease	481	**293**	**237**
Kenny-Caffey syndrome	2333	**15**	**14**
Keratinopathic ichthyosis*	281103	**41**	**41**
Keratocystic odontogenic tumor	447777	**<10**	**<10**
Keratoderma hereditarium mutilans	494	**<10**	**<10**
Keratoderma hereditarium mutilans with ichthyosis	79395	**<10**	**<10**
Keratosis follicularis spinulosa decalvans	2340	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
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**
Ketoacidosis due to monocarboxylate transporter-1 deficiency	438075	**<10**	**<10**
Keutel syndrome	85202	**<10**	**<10**
KID syndrome	477	**29**	**23**
Kidney tubulopathy-dilated cardiomyopathy syndrome	73224	**<10**	**<10**
Kienbock disease	97332	**<10**	**<10**
Kikuchi-Fujimoto disease	50918	**64**	**64**
Kimura disease	482	**15**	**15**
Kindler epidermolysis bullosa	2908	**27**	**27**
King-Denborough syndrome	99741	**<10**	**<10**
Kleefstra syndrome	261494	**194**	**192**
Kleine-Levin syndrome	33543	**322**	**322**
Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome	447974	**<10**	**<10**
Klippel-Trénaunay syndrome	90308	**375**	**374**
Kniest dysplasia	485	**31**	**31**
Knobloch syndrome	1571	**28**	**28**
Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome	2698	**<10**	**<10**
Kommerell diverticulum	99077	**13**	**13**
Koolen-De Vries syndrome	96169	**106**	**105**
Kosaki overgrowth syndrome	477831	**<10**	**<10**
Kostmann syndrome	99749	**12**	**12**
Krabbe disease	487	**107**	**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	**65**	**62**
L1 syndrome	275543	**115**	**109**
L-2-hydroxyglutaric aciduria	79314	**26**	**25**
Lacrimoauriculodentodigital syndrome	2363	**41**	**41**
Lafora disease	501	**15**	**<10**
Laing early-onset distal myopathy	59135	**61**	**60**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
LAMA5-related multisystemic syndrome	521450	<10	<10
Lambert syndrome	1296	<10	<10
Lambert-Eaton myasthenic syndrome	43393	194	151
Lamb-Shaffer syndrome	530983	51	51
Lamellar ichthyosis	313	368	365
Laminin subunit alpha 2-related congenital muscular dystrophy	258	214	191
Landau-Kleffner syndrome	98818	49	49
Langer mesomelic dysplasia	2632	11	11
Langerhans cell histiocytosis	389	1691	1652
Langerhans cell sarcoma	86897	<10	<10
Large congenital melanocytic nevus	626	1367	1358
Laron syndrome	633	49	49
Laron syndrome with immunodeficiency	220465	<10	<10
Larsen syndrome	503	79	77
Larsen-like osseous dysplasia-short stature syndrome	2370	<10	<10
Larsen-like syndrome, B3GAT3 type	284139	<10	<10
Laryngeal abductor paralysis	2808	17	16
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
Laryngotracheal angioma	137935	47	47
Laryngotracheoesophageal cleft	2004	96	92
Larynx atresia	1202	38	36
Late infantile neuronal ceroid lipofuscinosis	168491	30	21
Late-onset distal myopathy, Markesbery-Griggs type	98912	26	25
Late-onset focal dermal elastosis	228227	<10	<10
Late-onset isolated ACTH deficiency	199299	261	258
Late-onset junctional epidermolysis bullosa	79406	<10	<10
Late-onset retinal degeneration	67042	12	12
Lateral facial cleft*	141269	13	13
Lateral meningocele syndrome	2789	<10	<10
Lattice corneal dystrophy type I	98964	30	29
Laubry-Pezzi syndrome	99094	154	153
Laurence-Moon syndrome	2377	<10	<10
Laurin-Sandrow syndrome	2378	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
LCAT deficiency	650	<10	<10
Lead poisoning	330015	<10	<10
Leber congenital amaurosis	65	521	519
Leber hereditary optic neuropathy	104	988	954
Leber plus disease	99718	57	52
Ledderhose disease	199251	<10	<10
Left sided atrial isomerism	566862	<10	<10
Left ventricular noncompaction	54260	919	906
Legg-Calvé-Perthes disease	2380	30	30
Legionnaires disease	549	<10	<10
Legius syndrome	137605	115	114
Leigh syndrome	506	152	113
Leiomyosarcoma	64720	22	21
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	1062	1035
Lenz-Majewski hyperostotic dwarfism	2658	<10	<10
Leprechaunism	508	<10	<10
Leprosy	548	34	34
Leptospirosis	509	<10	<10
Léri-Weill dyschondrosteosis	240	979	979
Lesch-Nyhan syndrome	510	65	51
Lethal ataxia with deafness and optic atrophy	1187	<10	<10
Lethal congenital contracture syndrome	294965	<10	<10
Lethal congenital contracture syndrome type 1	1486	<10	<10
Lethal hemolytic anemia-genital anomalies syndrome	1046	<10	<10
Lethal infantile mitochondrial myopathy	254857	30	<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 osteosclerotic bone dysplasia	1832	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Lethal polymalformative syndrome, Boissel type	210144	**14**	**12**
Lethal pontocerebellar hypoplasia-hypotonia-respiratory insufficiency syndrome	615954	**<10**	**<10**
Lethal recessive chondrodysplasia	1423	**<10**	**<10**
Leukocyte adhesion deficiency	2968	**12**	**<10**
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	**11**	**11**
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**
Lhermitte-Duclos disease	65285	**<10**	**<10**
Lichen amyloidosis	49804	**<10**	**<10**
Lichen myxedematosus*	402007	**<10**	**<10**
Lichen planopilaris	525	**<10**	**<10**
Lichen planus pemphigoides	254478	**106**	**102**
Lichen planus pigmentosus	254463	**<10**	**<10**
Liddle syndrome	526	**33**	**33**
Li-Fraumeni syndrome	524	**57**	**48**
LIG4 syndrome	99812	**<10**	**<10**
Limbal stem cell deficiency	171673	**19**	**19**
Limb-girdle muscular dystrophy due to POMK deficiency	445110	**<10**	**<10**
Limb-girdle muscular dystrophy*	263	**698**	**643**
Limb-mammary syndrome	69085	**<10**	**<10**
Linear and whorled nevoid hypermelanosis	79150	**19**	**19**
Linear atrophoderma of Moulin	140933	**<10**	**<10**
Linear hypopigmentation and craniofacial asymmetry with acral, ocular and brain anomalies	589608	**<10**	**<10**
Linear IgA dermatosis	46488	**189**	**175**
Linear lichen planus	254379	**17**	**17**
Linear nevus sebaceus syndrome	2612	**198**	**198**
Linear verrucous nevus syndrome	2611	**221**	**221**
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	**15**	**14**
Lipoprotein glomerulopathy	329481	**<10**	**<10**
Liposarcoma	69078	**31**	**31**
Lipoyl transferase 1 deficiency	401862	**<10**	**<10**
Lisch epithelial corneal dystrophy	98955	**<10**	**<10**
Lissencephaly due to LIS1 mutation	95232	**47**	**45**
Lissencephaly due to TUBA1A mutation	171680	**14**	**14**
Lissencephaly type 1 due to doublecortin gene mutation	2148	**57**	**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	**20**	**20**
Liver adenomatosis	566841	**<10**	**<10**
LMNA-related cardiocutaneous progeria syndrome	363618	**<10**	**<10**
Localized dystrophic epidermolysis bullosa	595356	**20**	**20**
Localized epidermolysis bullosa simplex	79400	**44**	**44**
Localized junctional epidermolysis bullosa	251393	**<10**	**<10**
Localized lichen myxedematosus*	86795	**<10**	**<10**
Localized lipodystrophy*	79088	**11**	**<10**
Localized scleroderma	90289	**268**	**267**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Locked-in syndrome	2406	**<10**	**<10**
Loeys-Dietz syndrome	60030	**576**	**556**
Logopenic progressive aphasia	250831	**78**	**76**
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	5	**81**	**72**
Loose anagen syndrome	168	**<10**	**<10**
Low phospholipid-associated cholelithiasis	69663	**1380**	**1374**
Lowe-Kohn-Cohen syndrome	2408	**<10**	**<10**
Lower limb hypertrophy	295051	**27**	**27**
Lower limb malformation-hypospadias syndrome	2487	**<10**	**<10**
Lower lip fistula	141064	**12**	**12**
Lower motor neuron syndrome with late-adult onset	276435	**38**	**35**
Low-grade astrocytoma*	251592	**<10**	**<10**
Lown-Ganong-Levine syndrome	844	**<10**	**<10**
Lowry-MacLean syndrome	2409	**<10**	**<10**
Lowry-Wood syndrome	1824	**<10**	**<10**
LRP5-related primary osteoporosis	498481	**17**	**17**
Lujan-Fryns syndrome	776	**72**	**70**
LUMBAR syndrome	83628	**54**	**54**
Lung agenesis-heart defect-thumb anomalies syndrome	1120	**<10**	**<10**
Lupus erythematosus panniculitis	90285	**28**	**25**
Lupus erythematosus tumidus	90283	**88**	**88**
Luscan-Lumish syndrome	597738	**<10**	**<10**
Lyme disease	91546	**179**	**171**
Lymphangioleiomyomatosis	538	**513**	**507**
Lymphatic filariasis	2035	**<10**	**<10**
Lymphedema-distichiasis syndrome	33001	**65**	**65**
Lymphoid interstitial pneumonia	79128	**168**	**156**
Lymphomatoid granulomatosis	86869	**<10**	**<10**
Lymphomatoid papulosis	98842	**<10**	**<10**
Lymphoproliferative syndrome*	238510	**14**	**13**
Lynch syndrome	144	**262**	**233**
Lysinuric protein intolerance	470	**51**	**49**
Lysosomal acid lipase deficiency	275761	**46**	**44**
Macrocephaly-developmental delay syndrome	397612	**40**	**40**
Macrocephaly-intellectual disability-autism syndrome	210548	**35**	**35**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Macrocephaly-intellectual disability-left ventricular non compaction syndrome	466791	**<10**	**<10**
Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	457485	**27**	**27**
Macrocephaly-short stature-paraplegia syndrome	2427	**<10**	**<10**
Macrocephaly-spastic paraplegia-dysmorphism syndrome	2429	**13**	**13**
Macrocystic lymphatic malformation	79489	**501**	**501**
Macrodactyly of fingers	295044	**14**	**14**
Macrodactyly of toes	295047	**25**	**25**
Macrophage activation syndrome	158061	**100**	**79**
Macrophagic myofasciitis	592	**523**	**509**
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**
Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome	487796	**<10**	**<10**
Macular coloboma-cleft palate-hallux valgus syndrome	91494	**<10**	**<10**
Macular corneal dystrophy	98969	**23**	**23**
Maculopapular cutaneous mastocytosis	79457	**655**	**654**
Madras motor neuron disease	137867	**<10**	**<10**
Maffucci syndrome	163634	**22**	**21**
MAGEL2-related Prader-Willi-like syndrome	398069	**30**	**30**
MAGIC syndrome	324972	**22**	**21**
Majeed syndrome	77297	**<10**	**<10**
Mal de débarquement	210272	**<10**	**<10**
Mal de Meleda	87503	**23**	**23**
Malakoplakia	556	**<10**	**<10**
Malan overgrowth syndrome	420179	**40**	**40**
Malaria	673	**17**	**13**
Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome	2234	**<10**	**<10**
Male infertility with azoospermia or oligozoospermia due to single gene mutation	399805	**64**	**64**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Male infertility with teratozoospermia due to single gene mutation	399808	**14**	**14**
Maligant granulosa cell tumor of the ovary	99915	**19**	**19**
Malignant atrophic papulosis	679	**<10**	**<10**
Malignant dysgerminomatous germ cell tumor of the ovary	99912	**<10**	**<10**
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 hyperthermia of anesthesia	423	**308**	**306**
Malignant melanoma of the mucosa	168999	**17**	**13**
Malignant migrating focal seizures of infancy	293181	**52**	**50**
Malignant non-dysgerminomatous germ cell tumor of ovary	206538	**<10**	**<10**
Malignant peripheral nerve sheath tumor	3148	**15**	**15**
Malignant peritoneal mesothelioma	168811	**<10**	**<10**
Malignant Sertoli-Leydig cell tumor of the ovary	99916	**16**	**15**
Malignant teratoma of ovary	398987	**24**	**24**
Malonic aciduria	943	**11**	**11**
Malposition of a coronary ostium	99090	**<10**	**<10**
MALT lymphoma	52417	**37**	**36**
MAN1B1-CDG	397941	**<10**	**<10**
Mandibular arteriovenous malformation	141174	**48**	**48**
Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome	363649	**<10**	**<10**
Mandibuloacral dysplasia	2457	**<10**	**<10**
Mandibulofacial dysostosis*	155899	**73**	**70**
Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome	357158	**<10**	**<10**
Mandibulofacial dysostosis-microcephaly syndrome	79113	**68**	**67**
Mantle cell lymphoma	52416	**18**	**17**
Maple syrup urine disease	511	**223**	**217**
Marburg acute multiple sclerosis	228157	**31**	**30**
Marcus-Gunn syndrome	91412	**41**	**40**
Marden-Walker syndrome	2461	**<10**	**<10**
Marfan syndrome	558	**7979**	**7829**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Marfanoid habitus-autosomal recessive intellectual disability syndrome	2463	**30**	**30**
Marfanoid syndrome, De Silva type	2464	**<10**	**<10**
Marginal zone lymphoma*	300912	**11**	**<10**
Marie Unna hereditary hypotrichosis	444	**<10**	**<10**
Marinesco-Sjögren syndrome	559	**20**	**18**
Marshall syndrome	560	**70**	**70**
Marshall-Smith syndrome	561	**17**	**17**
Martinique crinkled retinal pigment epitheliopathy	466718	**<10**	**<10**
Mast cell leukemia	98851	**<10**	**<10**
Mast cell sarcoma	66661	**<10**	**<10**
Maternal phenylketonuria	2209	**37**	**37**
Maternal uniparental disomy of chromosome 1	251009	**<10**	**<10**
Maternal uniparental disomy of chromosome 16	96185	**<10**	**<10**
Maternal uniparental disomy of chromosome 20	96186	**<10**	**<10**
Maternal uniparental disomy of chromosome 21	96187	**<10**	**<10**
Maternal uniparental disomy of chromosome X	261519	**<10**	**<10**
Maternally-inherited diabetes and deafness	225	**328**	**313**
Matthew-Wood syndrome	2470	**<10**	**<10**
Maxillary arteriovenous malformation	141171	**45**	**45**
Maxillonasal dysplasia	1248	**187**	**186**
Mayer-Rokitansky-Küster-Hauser syndrome	3109	**889**	**889**
Mazabraud syndrome	57782	**19**	**19**
McCune-Albright syndrome	562	**499**	**488**
McDonough syndrome	2471	**<10**	**<10**
McKusick-Kaufman syndrome	2473	**<10**	**<10**
McLeod neuroacanthocytosis syndrome	59306	**13**	**13**
Meacham syndrome	3097	**<10**	**<10**
Meckel syndrome	564	**15**	**<10**
Meconium aspiration syndrome	70588	**32**	**29**
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	**47**	**47**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Median facial cleft*	141234	**<10**	**<10**
Median nodule of the upper lip	2699	**<10**	**<10**
Medium chain acyl-CoA dehydrogenase deficiency	42	**294**	**284**
Medullary sponge kidney	1309	**215**	**210**
Medullary thyroid carcinoma	1332	**77**	**75**
Medulloblastoma	616	**224**	**208**
Medulloepithelioma of the central nervous system	251883	**<10**	**<10**
Meesmann corneal dystrophy	98954	**<10**	**<10**
Mega-cisterna magna	97252	**<10**	**<10**
Megaconial congenital muscular dystrophy	280671	**14**	**14**
Megacystis-megaureter syndrome	238637	**54**	**54**
Megacystis-microcolon-intestinal hypoperistalsis syndrome	2241	**<10**	**<10**
Megalencephalic leukoencephalopathy with subcortical cysts	2478	**41**	**40**
Megalencephaly	2477	**36**	**35**
Megalencephaly-capillary malformation-polymicrogyria syndrome	60040	**153**	**153**
Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	83473	**24**	**23**
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	**1944**	**1917**
Meigs syndrome	314451	**<10**	**<10**
Melanoma and neural system tumor syndrome	252206	**<10**	**<10**
Melanoma of soft tissue	97338	**14**	**12**
MELAS	550	**885**	**756**
Melkersson-Rosenthal syndrome	2483	**24**	**23**
Melnick-Needles syndrome	2484	**20**	**20**
Melorheostosis	2485	**57**	**57**
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 STAT1 deficiency	319595	**<10**	**<10**
Mendelian susceptibility to mycobacterial diseases*	748	**<10**	**<10**
Ménétrier disease	2494	**<10**	**<10**
Meningeal melanocytoma	252046	**<10**	**<10**
Meningioma	2495	**346**	**338**
Meningococcal meningitis	33475	**51**	**50**
Menke-Hennekam syndrome	592574	**13**	**13**
Menkes disease	565	**66**	**34**
Menstrual cycle-dependent periodic fever	498251	**<10**	**<10**
MEPAN syndrome	508093	**<10**	**<10**
MERRF	551	**99**	**85**
Mesial temporal lobe epilepsy with hippocampal sclerosis	99701	**764**	**759**
Mesoaxial synostotic syndactyly with phalangeal reduction	157801	**<10**	**<10**
Mesomelia-synostoses syndrome	2496	**<10**	**<10**
Mesomelic and rhizo-mesomelic dysplasia*	93438	**11**	**11**
Mesomelic dysplasia, Savarirayan type	85170	**<10**	**<10**
Metabolic myopathy due to lactate transporter defect	171690	**17**	**17**
Metachondromatosis	2499	**48**	**47**
Metachromatic leukodystrophy	512	**203**	**137**
Metaphyseal acroscyphodysplasia	1240	**<10**	**<10**
Metaphyseal anadysplasia	1040	**<10**	**<10**
Metaphyseal chondrodysplasia, Schmid type	174	**70**	**69**
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	**22**	**21**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Methanol poisoning	31825	<10	<10
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	197	182
Methylmalonic acidemia without homocystinuria*	293355	55	48
Methylmalonic aciduria due to transcobalamin receptor defect	280183	<10	<10
Metopic ridging-ptosis-facial dysmorphism syndrome	502430	<10	<10
Mevalonate kinase deficiency	309025	206	203
MGAT2-CDG	79329	<10	<10
Micro syndrome	2510	31	28
Microbrachycephaly-ptosis-cleft lip syndrome	2511	22	22
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	19	18
Microcephalic osteodysplastic primordial dwarfism types I and III	2636	<10	<10
Microcephalic primordial dwarfism*	324761	18	16
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

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Microcephaly-cleft palate-abnormal retinal pigmentation syndrome	2521	<10	<10
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-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome	457351	<10	<10
Microcephaly-lymphedema-chorioretinopathy syndrome	2526	31	30
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
Microcystic lymphatic malformation	79490	533	532
Microcytic anemia with liver iron overload	83642	<10	<10
Microduplication Xp11.22p11.23 syndrome	217377	19	18
Microform holoprosencephaly	280200	20	19
Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	476126	12	12
Microlissencephaly	1083	18	16
Microlissencephaly-micromelia syndrome	50810	<10	<10
Microphthalmia with brain and digit anomalies	139471	<10	<10
Microphthalmia with limb anomalies	1106	<10	<10
Microphthalmia with linear skin defects syndrome	2556	15	15
Microphthalmia, Lenz type	568	<10	<10
Microphthalmia-brain atrophy syndrome	77299	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	251279	<10	<10
Microscopic polyangiitis	727	672	619
Microspherophakia-metaphyseal dysplasia syndrome	2551	<10	<10
Microtia	83463	882	881
Microvillus inclusion disease	2290	46	38
Mid-dermal elastolysis	228299	<10	<10
Middle ear neuroendocrine tumor	100084	<10	<10
Middle East respiratory syndrome	576074	<10	<10
Midline cervical cleft	141288	25	25
Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis	93279	35	35
Miller Fisher syndrome	98919	206	192
Miller-Dieker syndrome	531	103	91
Mills syndrome	94091	13	11
Milroy disease	79452	410	407
MIRAGE syndrome	494433	14	12
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
MITF-related melanoma and renal cell carcinoma predisposition syndrome	293822	28	28
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
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	22	21

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mitochondrial DNA-associated Leigh syndrome	255210	21	21
Mitochondrial DNA-related cardiomyopathy and hearing loss	1349	14	13
Mitochondrial DNA-related dystonia	254851	11	<10
Mitochondrial DNA-related mitochondrial myopathy*	254788	57	55
Mitochondrial DNA-related progressive external ophthalmoplegia	663	106	104
Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	314637	<10	<10
Mitochondrial membrane protein-associated neurodegeneration	289560	11	11
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	34
Mitochondrial neurogastrointestinal encephalomyopathy	298	41	30
Mitochondrial pyruvate carrier deficiency	447784	<10	<10
Mitochondrial trifunctional protein deficiency	746	23	18
Mitral atresia	1205	38	33
Mitral valve agenesis	99062	<10	<10
Mixed connective tissue disease	809	1952	1913
Mixed cystic lymphatic malformation	458792	325	325
Mixed germ cell tumor	180234	<10	<10
Mixed phenotype acute leukemia	530995	<10	<10
Mixed-type autoimmune hemolytic anemia	90036	60	55
Miyoshi myopathy	45448	48	48
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	27	27
MODY	552	941	936
Moebius syndrome	570	263	252

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome	2560	<10	<10
Mohr-Tranebjaerg syndrome	52368	11	<10
MOMO syndrome	2563	<10	<10
Monilethrix	573	26	26
Monoamine oxidase A deficiency	3057	<10	<10
Monoclonal mast cell activation syndrome	529468	1679	1676
Monocytopenia with susceptibility to infections	228423	39	34
Monomelic amyotrophy	65684	204	193
Monosomy 13q14	1587	12	12
Monosomy 13q34	96168	23	23
Monosomy 18p	1598	70	68
Monosomy 18q	1600	176	169
Monosomy 22	96123	14	14
Monosomy 22q13.3	48652	320	315
Monosomy 5p	281	215	210
Monosomy 9p	261112	36	36
Monosomy 9q22.3	77301	<10	<10
Mooren ulcer	519408	19	17
Morgagni-Stewart-Morel syndrome	77296	<10	<10
Morning glory disc anomaly	35737	80	79
Morvan syndrome	83467	<10	<10
Mosaic genome-wide paternal uniparental disomy	329813	<10	<10
Mosaic neurofibromatosis type 1	634461	26	26
Mosaic NF2-related schwannomatosis	634475	<10	<10
Mosaic schwannomatosis	634492	<10	<10
Mosaic trisomy 1	1692	<10	<10
Mosaic trisomy 12	1698	15	13
Mosaic trisomy 14	1703	20	20
Mosaic trisomy 15	1706	12	12
Mosaic trisomy 16	1708	20	19
Mosaic trisomy 2	1723	<10	<10
Mosaic trisomy 20	1724	20	18
Mosaic trisomy 22	96068	17	16
Mosaic trisomy 3	100071	<10	<10
Mosaic trisomy 5	96060	<10	<10
Mosaic trisomy 7	1747	<10	<10
Mosaic trisomy 8	96061	43	39

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Mosaic trisomy 9	99776	41	35
Mosaic variegated aneuploidy syndrome	1052	34	33
Mounier-Kühn syndrome	3347	22	22
Mowat-Wilson syndrome	2152	151	142
Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	280679	<10	<10
Moyamoya disease	2573	445	430
Moyamoya disease with early-onset achalasia	401945	<10	<10
Moynahan syndrome	2574	<10	<10
MPI-CDG	79319	15	13
MT-ATP6-related mitochondrial spastic paraplegia	320360	<10	<10
Mucinous adenocarcinoma of ovary	398961	<10	<10
Mucinous cystadenocarcinoma of the pancreas	424053	36	35
Muckle-Wells syndrome	575	103	100
Mucocutaneous venous malformations	2451	2606	2605
Mucolipidosis type II	576	42	21
Mucolipidosis type III	577	44	41
Mucolipidosis type IV	578	11	11
Mucopolysaccharidosis type 1	579	305	266
Mucopolysaccharidosis type 2	580	222	164
Mucopolysaccharidosis type 3	581	237	178
Mucopolysaccharidosis type 4	582	206	197
Mucopolysaccharidosis type 6	583	61	55
Mucopolysaccharidosis type 7	584	12	11
Mucous membrane pemphigoid	46486	1457	1318
Mueller-Weiss syndrome	566943	<10	<10
Muenke syndrome	53271	132	130
Muir-Torre syndrome	587	128	121
Mulibrey nanism	2576	14	14
Müllerian aplasia and hyperandrogenism	247768	<10	<10
Müllerian aplasia*	73217	<10	<10
Müllerian derivatives-lymphangiectasia-polydactyly syndrome	1655	<10	<10
Müllerian duct anomalies-limb anomalies syndrome	2491	<10	<10
Multicentric carpo-tarsal osteolysis with or without nephropathy	2774	11	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Multicentric osteolysis-nodulosis-arthropathy spectrum	371428	**<10**	**<10**
Multicentric reticulohistiocytosis	139436	**14**	**14**
Multicystic dysplastic kidney	1851	**3059**	**3039**
Multifocal atrial tachycardia	3282	**13**	**12**
Multifocal lymphangioendotheliomatosis-thrombocytopenia syndrome	464321	**<10**	**<10**
Multifocal motor neuropathy	641	**884**	**840**
Multifocal pattern dystrophy simulating fundus flavimaculatus	99003	**23**	**23**
Multiloculated renal cyst	97366	**34**	**34**
Multiminicore myopathy	598	**148**	**142**
Multinodular goiter-cystic kidney-polydactyly syndrome	2091	**14**	**14**
Multiple acyl-CoA dehydrogenase deficiency	26791	**116**	**105**
Multiple benign circumferential skin creases on limbs	2505	**<10**	**<10**
Multiple carboxylase deficiency*	148	**<10**	**<10**
Multiple congenital anomalies-hypotonia-seizures syndrome	280633	**35**	**28**
Multiple congenital anomalies-hypotonia-seizures syndrome type 2	300496	**<10**	**<10**
Multiple endocrine neoplasia type 1	652	**473**	**460**
Multiple endocrine neoplasia type 2	653	**261**	**257**
Multiple endocrine neoplasia type 4	276152	**<10**	**<10**
Multiple endocrine neoplasia*	276161	**48**	**47**
Multiple epiphyseal dysplasia due to collagen 9 anomaly	166002	**<10**	**<10**
Multiple epiphyseal dysplasia type 1	93308	**<10**	**<10**
Multiple epiphyseal dysplasia type 4	93307	**<10**	**<10**
Multiple epiphyseal dysplasia type 5	93311	**<10**	**<10**
Multiple epiphyseal dysplasia*	251	**440**	**438**
Multiple epiphyseal dysplasia, Beighton type	166011	**<10**	**<10**
Multiple epiphyseal dysplasia, Lowry type	166016	**<10**	**<10**
Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia	166029	**<10**	**<10**
Multiple intestinal atresia	2300	**15**	**15**
Multiple metaphyseal dysplasia*	93430	**128**	**127**
Multiple mitochondrial dysfunctions syndrome type 1	401869	**<10**	**<10**
Multiple myeloma	29073	**458**	**306**
Multiple osteochondromas	321	**1181**	**1176**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
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	**19**	**13**
Multiple symmetric lipomatosis	2398	**103**	**99**
Multiple synostoses syndrome	3237	**62**	**62**
Multiple system atrophy	102	**1223**	**832**
Multisystem inflammatory syndrome in children and adults	598363	**306**	**306**
Multisystemic smooth muscle dysfunction syndrome	404463	**<10**	**<10**
Muscle filaminopathy	171445	**77**	**68**
Muscle-eye-brain disease	588	**16**	**12**
Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome	2579	**<10**	**<10**
Muscular dystrophy, Selcen type	199340	**<10**	**<10**
Muscular glycogenosis*	206959	**104**	**96**
Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome	324416	**<10**	**<10**
Musculocontractural Ehlers-Danlos syndrome	2953	**14**	**14**
Mutilating hereditary sensory neuropathy with spastic paraplegia	139578	**36**	**34**
Mutilating palmoplantar keratoderma with periorificial keratotic plaques	659	**<10**	**<10**
Myalgia-eosinophilia syndrome associated with tryptophan	2582	**<10**	**<10**
Myasthenia gravis	589	**7573**	**6882**
MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome	498693	**<10**	**<10**
Mycophenolate mofetil embryopathy	268249	**<10**	**<10**
Mycoplasma encephalitis	83482	**<10**	**<10**
Myelocystocele*	268813	**12**	**12**
Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality	86841	**<10**	**<10**
Myelodysplastic syndrome*	52688	**80**	**69**
Myelodysplastic/myeloproliferative disease*	98275	**<10**	**<10**
Myeloid/lymphoid neoplasm associated with FGFR1 rearrangement	168953	**<10**	**<10**
Myeloperoxidase deficiency	2587	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Myeloproliferative neoplasm*	98274	<10	<10
MYH9-related disease	182050	243	241
Myhre syndrome	2588	60	55
Myoclonic epilepsy in non-progressive encephalopathies	86913	31	31
Myoclonic epilepsy of infancy	86909	166	165
Myoclonic-astatic epilepsy	1942	522	521
Myoclonus-cerebellar ataxia-deafness syndrome	2589	<10	<10
Myoclonus-dystonia syndrome	36899	259	256
Myopathic Ehlers-Danlos syndrome	536516	<10	<10
Myopathy and diabetes mellitus	2596	<10	<10
Myopic macular degeneration	178493	30	29
Myosclerosis	289380	<10	<10
Myosin storage myopathy	53698	<10	<10
Myotonia fluctuans	99734	33	32
Myotonia permanens	99735	11	11
Myotonic dystrophy*	206647	116	106
MYT1L-related developmental delay-intellectual disability-obesity syndrome	647799	<10	<10
Myxopapillary ependymoma	251643	<10	<10
NAD(P)HX epimerase deficiency	555407	<10	<10
Naegeli-Franceschetti-Jadassohn syndrome	69087	<10	<10
Nager syndrome	245	46	40
Nail-patella syndrome	2614	425	420
Nail-patella-like renal disease	2613	<10	<10
Nance-Horan syndrome	627	35	35
Nanophthalmos	35612	28	28
Narcolepsy type 1	2073	2687	2678
Narcolepsy type 2	83465	995	991
NARP syndrome	644	78	74
Nasal dermoid cyst	141103	43	43
Nasal dorsum fistula	141219	166	166
Nasal glial heterotopia	141112	<10	<10
Nasolacrimal duct cyst	141083	27	27
Nasopalpebral lipoma-coloboma syndrome	2399	<10	<10
Nasopharyngeal carcinoma	150	<10	<10
Nasu-Hakola disease	2770	<10	<10
Native American myopathy	168572	40	38
Navajo neurohepatopathy	255229	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Naxos disease	34217	<10	<10
Necrobiosis lipoidica	542592	<10	<10
Necrobiotic xanthogranuloma	158011	16	16
Necrotizing enterocolitis	391673	99	96
Necrotizing soft tissue infection	440368	<10	<10
Nelson syndrome	199244	16	15
Nemaline myopathy*	607	139	123
Neonatal acute respiratory distress due to SP-B deficiency	217563	<10	<10
Neonatal adrenoleukodystrophy	44	16	12
Neonatal alloimmune neutropenia	464370	29	29
Neonatal antiphospholipid syndrome	398097	85	85
Neonatal autoimmune hemolytic anemia	398109	<10	<10
Neonatal brainstem dysfunction	137929	241	235
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	27	18
Neonatal hypoxic and ischemic brain injury	137577	350	341
Neonatal ichthyosis-sclerosing cholangitis syndrome	59303	12	11
Neonatal inflammatory skin and bowel disease	294023	<10	<10
Neonatal intrahepatic cholestasis due to citrin deficiency	247598	<10	<10
Neonatal lupus erythematosus	398124	23	23
Neonatal Marfan syndrome	284979	27	22
Neonatal osteosclerotic dysplasia*	93443	<10	<10
Neonatal scleroderma	398127	<10	<10
Neonatal severe primary hyperparathyroidism	417	18	16
Neovascular glaucoma	94058	<10	<10
Nephroblastoma	654	367	365
Nephrogenic diabetes insipidus	223	204	203
Nephrogenic syndrome of inappropriate antidiuresis	93606	16	16
Nephrogenic systemic fibrosis	137617	<10	<10
Nephronophthisis	655	468	459

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Nephropathy-deafness-hyperparathyroidism syndrome	2668	<10	<10
Nephrosis-deafness-urinary tract-digital malformations syndrome	2669	<10	<10
Netherton syndrome	634	117	108
Neuhauser anomaly	99078	28	28
Neuhauser-Eichner-Opitz syndrome	2672	<10	<10
Neu-Laxova syndrome	2671	<10	<10
Neuralgic amyotrophy	2901	776	762
Neurenteric cyst	268865	<10	<10
Neuroacanthocytosis*	263440	<10	<10
Neuroblastoma	635	171	156
Neurocutaneous melanocytosis	2481	20	19
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	88639	<10	<10
Neurodegeneration with brain iron accumulation*	385	55	48
Neurodegenerative syndrome due to cerebral folate transport deficiency	217382	19	18
Neurodevelopmental delay-intellectual disability-ataxia-feeding difficulty syndrome	647788	<10	<10
Neurodevelopmental delay-seizures-ophthalmic anomalies-osteopenia-cerebellar atrophy syndrome	529665	<10	<10
Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome	453499	31	30
Neuroendocrine carcinoma of pancreas	506098	<10	<10
Neuroendocrine cell hyperplasia of infancy	217560	87	86
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	15	14
Neurofibroma	252183	147	146
Neurofibromatosis type 1	636	12874	12625
Neurofibromatosis-Noonan syndrome	638	103	99
Neurogenic arthrogryposis multiplex congenita	1143	63	58
Neuroleptic malignant syndrome	94093	<10	<10
Neurological conditions associated with aminoacylase 1 deficiency	137754	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Neurolymphomatosis	206586	34	25
Neuromyelitis optica spectrum disorder	71211	1262	1243
Neuronal ceroid lipofuscinosis*	216	35	22
Neuronal tumor*	251924	<10	<10
Neuropathy with hearing impairment	139512	<10	<10
Neurotrophic keratopathy	137596	53	51
Neutral lipid storage disease with ichthyosis	98907	24	24
Neutral lipid storage disease*	165	18	18
Neutral lipid storage myopathy	98908	14	14
Neutrophil immunodeficiency syndrome	183707	<10	<10
NEVADA syndrome	370059	<10	<10
Nevus comedonicus syndrome	64754	12	12
Nevus of Ito	263432	<10	<10
Nevus of Ota	263425	31	31
New-onset refractory status epilepticus	363558	34	28
Nicolaides-Baraitser syndrome	3051	62	61
Niemann-Pick disease type C	646	203	160
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	11	11
NMDA receptor encephalitis	217253	228	225
Nocardiosis	31204	<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	476	474
Nodular non-suppurative panniculitis	33577	13	13
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	**5598**	**5573**
Non-acquired panhypopituitarism	90695	**1073**	**1032**
Non-amyloid fibrillary glomerulopathy	97566	**32**	**31**
Non-amyloid monoclonal immunoglobulin deposition disease	86861	**197**	**179**
Non-distal deletion 10q	1581	**43**	**43**
Non-distal deletion 12q	96160	**<10**	**<10**
Non-distal duplication 10q	1695	**<10**	**<10**
Non-distal duplication 13q	1702	**17**	**14**
Non-epidermolytic palmoplantar keratoderma	2337	**19**	**19**
Non-functioning neuroendocrine tumor of pancreas	506075	**<10**	**<10**
Non-functioning paraganglioma	94080	**237**	**233**
Non-functioning pituitary adenoma	91349	**5128**	**4919**
Non-histaminic angioedema*	658	**432**	**431**
Non-insulinoma pancreatogenous hypoglycemia syndrome	276608	**14**	**14**
Non-involuting congenital hemangioma	141179	**338**	**338**
Non-Langerhans cell histiocytosis*	157987	**22**	**20**
Non-papillary transitional cell carcinoma of the bladder	209989	**<10**	**<10**
Non-paraneoplastic sensory ganglionopathy	208989	**69**	**67**
Non-progressive cerebellar ataxia with intellectual disability	314647	**37**	**37**
Non-recovering obstetric brachial plexus lesion	439202	**43**	**43**
Non-rhizomelic chondrodysplasia punctata*	176	**39**	**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	**16**	**16**
Non-specific autoimmune brainstem encephalitis without characteristic antibodies	624216	**11**	**11**
Non-specific autoimmune cerebellar ataxia with characteristic antibodies	624259	**<10**	**<10**
Non-specific autoimmune cerebellar ataxia without characteristic antibodies	624268	**<10**	**<10**
Non-specific autoimmune supratentorial encephalitis with characteristic antibodies	624166	**109**	**102**
Non-specific autoimmune supratentorial encephalitis without characteristic antibodies	624178	**30**	**29**
Non-specific early-onset epileptic encephalopathy	442835	**360**	**346**
Non-specific interstitial pneumonia	91364	**1516**	**1353**
Non-specific syndromic intellectual disability	528084	**1626**	**1617**
Non-spherocytic hemolytic anemia due to hexokinase deficiency	90031	**<10**	**<10**
Non-syndromic anorectal malformation with anal stenosis	601008	**22**	**22**
Non-syndromic anorectal malformation with perineal fistula	600952	**184**	**184**
Non-syndromic anorectal malformation with pouch colon	601013	**<10**	**<10**
Non-syndromic anorectal malformation with rectal atresia	601018	**13**	**13**
Non-syndromic anorectal malformation with rectal stenosis	601023	**<10**	**<10**
Non-syndromic anorectal malformation with rectourethral fistula	600961	**83**	**82**
Non-syndromic anorectal malformation with rectovaginal fistula	601028	**25**	**25**
Non-syndromic anorectal malformation with rectovesical fistula	600984	**17**	**17**
Non-syndromic anorectal malformation with vestibular fistula	600993	**58**	**58**
Non-syndromic anorectal malformation without fistula	601002	**37**	**37**
Non-syndromic anorectal malformation*	557	**364**	**361**
Non-syndromic bicoronal and sagittal craniosynostosis	620205	**<10**	**<10**
Non-syndromic bicoronal craniosynostosis	35099	**194**	**194**
Non-syndromic bilambdoid and sagittal craniosynostosis	1516	**21**	**21**
Non-syndromic bilambdoid craniosynostosis	620178	**<10**	**<10**
Non-syndromic cloacal malformation	600998	**69**	**69**
Non-syndromic congenital bronchial atresia	649010	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Non-syndromic craniosynostosis*	139390	**117**	**117**
Non-syndromic genetic deafness	87884	**5972**	**5948**
Non-syndromic male infertility due to sperm motility disorder	276234	**21**	**21**
Non-syndromic metopic craniosynostosis	3366	**1053**	**1050**
Non-syndromic pontocerebellar hypoplasia*	98523	**127**	**107**
Non-syndromic posterior hypospadias	95706	**1528**	**1517**
Non-syndromic sagittal craniosynostosis	35093	**2016**	**2016**
Non-syndromic unicoronal craniosynostosis	620102	**<10**	**<10**
Non-syndromic unifrontosphenoidal craniosynostosis	620139	**<10**	**<10**
Non-syndromic unilambdoid craniosynostosis	620113	**<10**	**<10**
Non-terminal myelocystocele	645340	**<10**	**<10**
Noonan syndrome	648	**2702**	**2600**
Noonan syndrome with multiple lentigines	500	**187**	**181**
Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	363972	**22**	**22**
Noonan syndrome-like disorder with loose anagen hair	2701	**77**	**76**
Norrie disease	649	**63**	**63**
North Carolina macular dystrophy	75327	**38**	**37**
NPHP3-related Meckel-like syndrome	3032	**<10**	**<10**
NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	600663	**<10**	**<10**
NUT midline carcinoma	443167	**<10**	**<10**
Oblique facial cleft*	141253	**11**	**11**
Occipital horn syndrome	198	**12**	**11**
Occipital pachygyria and polymicrogyria	280640	**22**	**21**
Occult macular dystrophy	247834	**100**	**100**
Ochoa syndrome	2704	**17**	**17**
Ocular albinism with late-onset sensorineural deafness	1000	**<10**	**<10**
Ocular albinism*	284804	**205**	**205**
Ocular anomalies-axonal neuropathy-developmental delay syndrome	496790	**<10**	**<10**
Ocular cicatricial pemphigoid	99922	**69**	**66**
Ocular motor apraxia, Cogan type	1125	**56**	**56**
Oculoauriculofrontonasal syndrome	398156	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Oculo-auriculo-vertebral spectrum	141132	**139**	**138**
Oculoauriculovertebral spectrum with radial defects	2549	**15**	**15**
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	**107**	**100**
Oculocutaneous albinism type 1	352731	**211**	**211**
Oculocutaneous albinism type 2	79432	**153**	**153**
Oculocutaneous albinism type 3	79433	**13**	**13**
Oculocutaneous albinism type 4	79435	**31**	**30**
Oculocutaneous albinism type 5	370091	**<10**	**<10**
Oculocutaneous albinism type 6	370097	**<10**	**<10**
Oculocutaneous albinism*	55	**1101**	**1095**
Oculodentodigital dysplasia	2710	**64**	**61**
Oculofaciocardiodental syndrome	2712	**40**	**40**
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	**552**	**453**
Oculopharyngodistal myopathy	98897	**42**	**36**
Odontochondrodysplasia	166272	**<10**	**<10**
Odontomatosis-aortae esophagus stenosis syndrome	2724	**<10**	**<10**
Odontomicronychial dysplasia	1811	**15**	**15**
Odonto-onycho-dermal dysplasia	2721	**13**	**13**
Ogden syndrome	276432	**15**	**15**
Oguchi disease	75382	**<10**	**<10**
Okihiro syndrome	93293	**172**	**171**
Oligoarticular juvenile idiopathic arthritis	85410	**4372**	**4366**
Oligoastrocytoma	251656	**<10**	**<10**
Oligocone trichromacy	75378	**<10**	**<10**
Oligodendroglial tumor*	46484	**<10**	**<10**
Oligodendroglioma	251627	**31**	**29**
Oligodontia	99798	**2480**	**2480**
Oligomeganephronia	2260	**216**	**214**
Olivopontocerebellar atrophy-deafness syndrome	2732	**<10**	**<10**
Ollier disease	296	**359**	**353**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Omenn syndrome	39041	**23**	**22**
Omodysplasia	2733	**<10**	**<10**
Omphalocele	660	**202**	**189**
Omphalomesenteric cyst	490	**<10**	**<10**
Oncogenic osteomalacia	352540	**29**	**29**
Open spinal dysraphism with a myelomeningocele	93969	**664**	**652**
Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome	2743	**<10**	**<10**
Opitz GBBB syndrome	2745	**56**	**55**
Opsismodysplasia	2746	**<10**	**<10**
Opsoclonus-myoclonus syndrome	1183	**108**	**107**
Optic atrophy-ataxia-peripheral neuropathy-global developmental delay syndrome	543470	**<10**	**<10**
Optic atrophy-intellectual disability syndrome	401777	**44**	**43**
Optic disc pit	519404	**11**	**11**
Optic pathway glioma	2086	**300**	**285**
Oral-facial-digital syndrome with short stature and brachymesophalangy	508501	**<10**	**<10**
Orbital leiomyoma	52994	**<10**	**<10**
Ornithine transcarbamylase deficiency	664	**447**	**401**
Orofaciodigital syndrome type 1	2750	**62**	**61**
Orofaciodigital syndrome type 10	2756	**<10**	**<10**
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	**38**	**38**
Oromandibular dystonia	93958	**30**	**28**
Osgood-Schlatter disease	97335	**39**	**39**
Ossification anomalies-psychomotor developmental delay syndrome	73230	**<10**	**<10**
Osteochondritis dissecans	2764	**33**	**33**
Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome	2653	**<10**	**<10**
Osteochondrosis of the metatarsal bone	564003	**<10**	**<10**
Osteochondrosis of the tarsal bone	563991	**<10**	**<10**
Osteocraniostenosis	2763	**277**	**274**
Osteofibrous dysplasia	488265	**23**	**23**
Osteogenesis imperfecta	666	**3575**	**3503**
Osteoglosphonic dysplasia	2645	**<10**	**<10**
Osteomesopyknosis	2777	**12**	**12**
Osteonecrosis of the jaw	399293	**<10**	**<10**
Osteopathia striata-cranial sclerosis syndrome	2780	**41**	**41**
Osteopathia striata-pigmentary dermopathy-white forelock syndrome	2779	**<10**	**<10**
Osteopetrosis and related disorders*	2781	**64**	**60**
Osteopetrosis with renal tubular acidosis	2785	**31**	**29**
Osteopetrosis-hypogammaglobulinemia syndrome	178389	**<10**	**<10**
Osteoporosis-macrocephaly-blindness-joint hyperlaxity syndrome	2787	**<10**	**<10**
Osteoporosis-oculocutaneous hypopigmentation syndrome	2786	**<10**	**<10**
Osteoporosis-pseudoglioma syndrome	2788	**18**	**18**
Osteosarcoma	668	**68**	**65**
Osteosclerosis-developmental delay-craniosynostosis syndrome	178377	**<10**	**<10**
Osteosclerosis-ichthyosis-premature ovarian failure syndrome	75325	**<10**	**<10**
Osteosclerotic metaphyseal dysplasia	500548	**<10**	**<10**
O'Sullivan-McLeod syndrome	99965	**13**	**13**
Otodental syndrome	2791	**14**	**14**
Otofaciocervical syndrome	2792	**<10**	**<10**
Otopalatodigital syndrome spectrum disorder*	364541	**<10**	**<10**
Otopalatodigital syndrome type 1	90650	**18**	**17**
Otopalatodigital syndrome type 2	90652	**<10**	**<10**
Otospondylomegaepiphyseal dysplasia	1427	**<10**	**<10**
Ovarian fibroma	314473	**<10**	**<10**
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	**16**	**16**
Overhydrated hereditary stomatocytosis	3203	**<10**	**<10**
Overlap myositis	206572	**568**	**543**
Overlapping connective tissue disease*	251312	**20**	**20**
Pachydermoperiostosis	2796	**43**	**42**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Pachygyria-intellectual disability-epilepsy syndrome	2798	64	61
Pachyonychia congenita	2309	74	74
Paget disease of the nipple	180275	<10	<10
PAGOD syndrome	991	<10	<10
Pai syndrome	1993	25	25
PAICS deficiency	633099	<10	<10
Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	477993	<10	<10
Pallister-Hall syndrome	672	65	62
Palmoplantar keratoderma, Nagashima type	140966	<10	<10
Palmoplantar keratoderma-deafness syndrome	2202	17	17
Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome	538574	<10	<10
Palmoplantar keratoderma-spastic paralysis syndrome	2201	<10	<10
Pancytopenia due to IKZF1 mutations	317473	<10	<10
Pancytopenia-developmental delay syndrome	401764	<10	<10
PANDAS	66624	<10	<10
Panhypophysitis	95513	101	94
Pantothenate kinase-associated neurodegeneration	157850	32	28
PAPA syndrome	69126	15	15
Papillary renal cell carcinoma	319298	27	26
Papilloma of choroid plexus	2807	11	<10
Papillon-Lefèvre syndrome	678	36	35
Papular elastorrhexis	228264	<10	<10
Papular mucinosis of infancy	90395	<10	<10
Paracetamol poisoning	464458	<10	<10
Paramedian facial cleft*	155867	15	15
Paramedian nasal cleft	141242	21	21
Paramyotonia congenita of Von Eulenburg	684	278	268
Paraneoplastic cerebellar degeneration	623626	17	17
Paraneoplastic isolated brainstem encephalitis	624190	<10	<10
Paraneoplastic pemphigus	63455	47	36
Paraneoplastic sensory ganglionopathy	208999	33	30
Paraneoplastic uveitis	279928	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Paraparetic variant of Guillain-Barré syndrome	231445	16	15
Paraplegia-intellectual disability-hyperkeratosis syndrome	2824	<10	<10
Paratesticular adenocarcinoma	363478	<10	<10
Parathyroid carcinoma	143	<10	<10
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	75	74
Parkinson-dementia complex of Guam	90020	15	<10
Parkinsonian-pyramidal syndrome	171695	68	64
Paroxysmal cold hemoglobinuria	90035	<10	<10
Paroxysmal dyskinesia*	1431	63	62
Paroxysmal dystonia*	200037	39	38
Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	53583	15	15
Paroxysmal exertion-induced dyskinesia	98811	<10	<10
Paroxysmal extreme pain disorder	46348	48	47
Paroxysmal kinesigenic dyskinesia	98809	124	124
Paroxysmal nocturnal hemoglobinuria	447	364	341
Paroxysmal non-kinesigenic dyskinesia	98810	31	31
Partial androgen insensitivity syndrome	90797	152	151
Partial atrioventricular septal defect	1330	614	606
Partial bilateral aplasia of the Müllerian ducts*	180068	<10	<10
Partial chromosome Y deletion	1646	68	66
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 pancreatic agenesis	2805	<10	<10
Partial septate uterus	180129	<10	<10
Partially involuting congenital hemangioma	458785	36	36
Partington syndrome	94083	23	21
PASH syndrome	289478	<10	<10
PASS syndrome	641385	<10	<10
Patella aplasia/hypoplasia	86789	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Paternal 20q13.2q13.3 microdeletion syndrome	261304	<10	<10
Paternal uniparental disomy of chromosome 6	96191	<10	<10
Paternal uniparental disomy of chromosome 7	96192	<10	<10
Patterson-Stevenson-Fontaine syndrome	2439	<10	<10
Pauci-immune glomerulonephritis	93126	245	222
Pearson syndrome	699	45	33
Pectus excavatum-macrocephaly-dysplastic nails syndrome	2835	<10	<10
Pediatric arterial ischemic stroke	439175	451	450
Pediatric collagenous gastritis	487809	<10	<10
Pediatric multiple sclerosis	477738	291	291
Pediatric systemic lupus erythematosus	93552	644	634
Pediatric-onset Graves disease	525731	867	863
Peeling skin syndrome*	817	28	28
PEHO syndrome	2836	<10	<10
PEHO-like syndrome	99807	<10	<10
Pelizaeus-Merzbacher disease	702	162	151
Pelizaeus-Merzbacher-like disease	280270	26	26
Pellucid marginal degeneration	137672	70	70
Pelviscapular dysplasia	93333	<10	<10
Pelvis-shoulder dysplasia	2839	<10	<10
Pemphigoid gestationis	63275	94	93
Pemphigus erythematosus	79480	17	16
Pemphigus foliaceus	79481	18	17
Pemphigus vegetans	79479	<10	<10
Pemphigus vulgaris	704	944	905
Pendred syndrome	705	382	378
Penile agenesis	49	<10	<10
Penoscrotal transposition	2842	<10	<10
PENS syndrome	313936	<10	<10
Pentalogy of Cantrell	1335	<10	<10
Pentasomy X	11	<10	<10
Pericardial and diaphragmatic defect	2847	13	12
Perineural cyst	65250	172	172
Perineurioma*	85102	<10	<10
Periodic fever-infantile enterocolitis-autoinflammatory syndrome	436166	15	15

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Periodic paralysis with later-onset distal motor neuropathy	397750	<10	<10
Periodic paralysis with transient compartment-like syndrome	397755	<10	<10
Periodic paralysis*	206976	65	65
Periodontal Ehlers-Danlos syndrome	75392	24	24
Perioral myoclonia with absences	139426	<10	<10
Peripapillary staphyloma	519400	<10	<10
Peripartum cardiomyopathy	563	11	<10
Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	163746	12	<10
Peripheral dysostosis	1795	<10	<10
Peripheral motor neuropathy-dysautonomia syndrome	2400	64	59
Peripheral primitive neuroectodermal tumor	370348	<10	<10
Peripheral pulmonary stenosis	99084	61	61
Peritoneal cystic mesothelioma	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	49	48
Persistent combined dystonia*	391711	36	35
Persistent fifth aortic arch	99076	<10	<10
Persistent hyperplastic primary vitreous	91495	292	292
Persistent idiopathic facial pain	398147	<10	<10
Persistent left superior vena cava connecting through coronary sinus to left-sided atrium	99109	18	18
Persistent left superior vena cava connecting to the roof of left-sided atrium	99111	<10	<10
Persistent Müllerian duct syndrome	2856	41	40
Persistent placoid maculopathy	97341	<10	<10
Persistent polyclonal B-cell lymphocytosis	300324	<10	<10
Peters anomaly	708	212	209
Peters plus syndrome	709	50	49
Peutz-Jeghers syndrome	2869	151	148
PFAPA syndrome	42642	2209	2209
Pfeiffer syndrome	710	121	113

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
PGM1-CDG	319646	**<10**	**<10**
PGM3-CDG	443811	**<10**	**<10**
PHACE syndrome	42775	**165**	**165**
Phakomatosis pigmentokeratotica	2874	**19**	**18**
Phakomatosis pigmentovascularis	2875	**89**	**87**
Phalangeal microgeodic syndrome	352636	**<10**	**<10**
Pharyngeal-cervical-brachial variant of Guillain-Barré syndrome	231426	**<10**	**<10**
Phenobarbital embryopathy	1919	**<10**	**<10**
Phenylketonuria	716	**3044**	**3028**
PHIP-related behavioral problems-intellectual disability-obesity-dysmorphic features syndrome	589905	**18**	**18**
Phocomelia, Schinzel type	2879	**<10**	**<10**
Phosphoenolpyruvate carboxykinase deficiency	2880	**<10**	**<10**
Phosphoribosylpyrophosphate synthetase superactivity	3222	**<10**	**<10**
Photosensitive epilepsy	166409	**67**	**67**
Phyllodes tumor of the breast	180261	**17**	**17**
Piebald trait-neurologic defects syndrome	2885	**<10**	**<10**
Piebaldism	2884	**105**	**105**
Pierpont syndrome	487825	**13**	**13**
Pierre Robin syndrome-faciodigital anomaly syndrome	2888	**76**	**73**
Pierson syndrome	2670	**<10**	**<10**
PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis	568062	**<10**	**<10**
Pigeon-breeder lung disease	99908	**101**	**92**
Pigmented paravenous retinochoroidal atrophy	251295	**<10**	**<10**
Pili bifurcati	720	**<10**	**<10**
Pilocytic astrocytoma	251612	**81**	**80**
Pilodental dysplasia-refractive errors syndrome	2892	**12**	**12**
Pilomatrixoma	91414	**77**	**77**
Pineal tumor of neuroepithelial tissue*	251905	**<10**	**<10**
Pineoblastoma	251909	**<10**	**<10**
Pinnae fistula or cyst	155838	**177**	**176**
Pitt-Hopkins syndrome	2896	**205**	**203**
Pituicytoma	251623	**18**	**18**
Pituitary adenoma*	99408	**525**	**505**
Pituitary apoplexy	95613	**510**	**491**
Pituitary carcinoma	300385	**20**	**17**
Pituitary deficiency due to empty sella turcica syndrome	91354	**168**	**165**
Pituitary deficiency due to Rathke cleft cysts	91350	**697**	**686**
Pituitary dermoid and epidermoid cysts	91351	**159**	**159**
Pituitary gigantism	99725	**<10**	**<10**
Pituitary stalk interruption syndrome	95496	**1135**	**1131**
Pityriasis rubra pilaris	2897	**30**	**30**
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	**16**	**16**
Platyspondylic dysplasia, Torrance type	85166	**<10**	**<10**
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	**35**	**35**
Pleural mesothelioma	50251	**<10**	**<10**
Pleuro-pericardial cyst	99131	**<10**	**<10**
Pleuropulmonary blastoma	64742	**<10**	**<10**
PLIN1-related familial partial lipodystrophy	280356	**21**	**21**
Plummer-Vinson syndrome	54028	**<10**	**<10**
PMM2-CDG	79318	**125**	**110**
PMP22-RAI1 contiguous gene duplication syndrome	477817	**<10**	**<10**
PMP2-related Charcot-Marie-Tooth disease type 1	476394	**14**	**14**
Pneumococcal meningitis	55655	**68**	**63**
Pneumoconiosis*	182098	**104**	**97**
Pneumocystosis	723	**20**	**18**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Pneumonia caused by Pseudomonas aeruginosa infection	90066	**12**	**12**
POEMS syndrome	2905	**184**	**170**
POGLUT1-related limb-girdle muscular dystrophy R21	480682	**<10**	**<10**
Poikiloderma with neutropenia	221046	**<10**	**<10**
Poland syndrome	2911	**491**	**490**
Poliomyelitis	2912	**94**	**88**
Polyarteritis nodosa	767	**609**	**583**
Polyarticular juvenile idiopathic arthritis*	404580	**117**	**116**
Polycythemia vera	729	**130**	**125**
Polycythemia*	98427	**74**	**74**
Polydactyly of a biphalangeal thumb and/or hallux	93339	**162**	**161**
Polydactyly of a triphalangeal thumb	93336	**13**	**13**
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**
Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	500533	**<10**	**<10**
Polymicrogyria due to TUBB2B mutation	300573	**<10**	**<10**
Polymicrogyria with optic nerve hypoplasia	250972	**<10**	**<10**
Polymicrogyria*	35981	**283**	**268**
Polymyalgia rheumatica	93569	**1615**	**1569**
Polymyositis	732	**1008**	**888**
Polyneuropathy associated with IgM monoclonal gammopathy with anti-MAG	639	**1264**	**1117**
Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	171848	**<10**	**<10**
Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome	2928	**<10**	**<10**
Polyrrhinia	141091	**<10**	**<10**
Polysyndactyly	93338	**75**	**74**
Polyvalvular heart disease syndrome	228410	**15**	**14**
POMGNT1-related limb-girdle muscular dystrophy R15	206564	**<10**	**<10**
POMT1-related limb-girdle muscular dystrophy R11	86812	**17**	**16**
POMT2-related limb-girdle muscular dystrophy R14	206559	**<10**	**<10**
Pontine autosomal dominant microangiopathy with leukoencephalopathy	477749	**11**	**<10**
Pontine tegmental cap dysplasia	269229	**<10**	**<10**
Pontocerebellar hypoplasia type 1	2254	**28**	**17**
Pontocerebellar hypoplasia type 10	411493	**<10**	**<10**
Pontocerebellar hypoplasia type 2	2524	**45**	**36**
Pontocerebellar hypoplasia type 3	97249	**<10**	**<10**
Pontocerebellar hypoplasia type 4	166063	**<10**	**<10**
Pontocerebellar hypoplasia type 6	166073	**<10**	**<10**
Pontocerebellar hypoplasia type 7	284339	**<10**	**<10**
Pontocerebellar hypoplasia type 9	369920	**<10**	**<10**
Popliteal pterygium syndrome*	294963	**<10**	**<10**
Porencephaly	2940	**85**	**82**
Porencephaly-cerebellar hypoplasia-internal malformations syndrome	2941	**<10**	**<10**
Porencephaly-microcephaly-bilateral congenital cataract syndrome	306547	**<10**	**<10**
Porokeratosis of Mibelli	735	**30**	**29**
Porokeratosis plantaris palmaris et disseminata	737	**14**	**13**
Porphyria cutanea tarda	101330	**3413**	**2774**
Porphyria due to ALA dehydratase deficiency	100924	**<10**	**<10**
Porphyria variegata	79473	**370**	**306**
Porphyria*	738	**26**	**22**
Portosinusoidal vascular disease	596937	**2494**	**2301**
Postaxial acrofacial dysostosis	246	**11**	**<10**
Postaxial polydactyly type A	93334	**84**	**84**
Postaxial polydactyly type B	93335	**118**	**118**
Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome	420584	**<10**	**<10**
Postaxial tetramelic oligodactyly	2730	**15**	**15**
Posterior amorphous corneal dystrophy	98971	**<10**	**<10**
Posterior column ataxia-retinitis pigmentosa syndrome	88628	**<10**	**<10**
Posterior cortical atrophy	54247	**71**	**66**
Posterior polymorphous corneal dystrophy	98973	**34**	**34**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Posterior urethral valve	93110	**1799**	**1782**
Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome	572013	**<10**	**<10**
Postinfectious cerebellitis	624244	**30**	**30**
Postinfectious vasculitis	48435	**53**	**53**
Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	477673	**<10**	**<10**
Postpartum psychosis	443173	**<10**	**<10**
Postpoliomyelitis syndrome	2942	**301**	**272**
Post-traumatic pituitary deficiency	95619	**2758**	**2597**
Postural orthostatic tachycardia syndrome due to NET deficiency	443236	**<10**	**<10**
Potassium-aggravated myotonia*	612	**<10**	**<10**
Potocki-Shaffer syndrome	52022	**20**	**20**
PPARG-related familial partial lipodystrophy	79083	**21**	**19**
Prader-Willi syndrome	739	**2180**	**2056**
Prader-Willi-like syndrome*	398073	**33**	**33**
Precursor B-cell acute lymphoblastic leukemia	99860	**43**	**42**
Precursor T-cell acute lymphoblastic leukemia	99861	**<10**	**<10**
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	**50**	**49**
Predominantly medium-vessel vasculitis*	156143	**33**	**32**
Predominantly small-vessel vasculitis*	156146	**58**	**54**
Preeclampsia	275555	**22**	**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	**541**	**540**
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	**70**	**69**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Primary autoimmune enteropathy	522037	**<10**	**<10**
Primary biliary cholangitis	186	**4130**	**3876**
Primary biliary cholangitis/primary sclerosing cholangitis and autoimmune hepatitis overlap syndrome	562639	**73**	**72**
Primary bone dysplasia with multiple joint dislocations*	93441	**27**	**26**
Primary bone lymphoma	314684	**<10**	**<10**
Primary central nervous system lymphoma	46135	**<10**	**<10**
Primary central precocious puberty in male	650087	**<10**	**<10**
Primary ciliary dyskinesia	244	**1302**	**1292**
Primary ciliary dyskinesia-retinitis pigmentosa syndrome	247522	**<10**	**<10**
Primary congenital hypothyroidism*	226295	**199**	**198**
Primary cutaneous aggressive epidermotropic CD8+ T-cell lymphoma	178528	**<10**	**<10**
Primary cutaneous amyloidosis*	137807	**<10**	**<10**
Primary cutaneous anaplastic large cell lymphoma	300865	**<10**	**<10**
Primary cutaneous follicle center lymphoma	178540	**<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 cutis verticis gyrata*	671	**<10**	**<10**
Primary dystonia, DYT13 type	98807	**20**	**20**
Primary dystonia, DYT2 type	99657	**<10**	**<10**
Primary dystonia, DYT21 type	306734	**19**	**19**
Primary dystonia, DYT27 type	464440	**132**	**131**
Primary dystonia, DYT4 type	98805	**<10**	**<10**
Primary dystonia, DYT6 type	98806	**26**	**26**
Primary early-onset glaucoma*	156005	**<10**	**<10**
Primary eosinophilic gastrointestinal disease*	402029	**<10**	**<10**
Primary erythromelalgia	90026	**170**	**170**
Primary essential cutis verticis gyrata	357220	**<10**	**<10**
Primary failure of tooth eruption	412206	**58**	**58**
Primary familial polycythemia	90042	**13**	**13**
Primary Fanconi renotubular syndrome	3337	**62**	**53**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
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	121	114
Primary hypergonadotropic hypogonadism-partial alopecia syndrome	2232	<10	<10
Primary hyperoxaluria	416	329	320
Primary hypertrophic osteoarthropathy*	248095	<10	<10
Primary hypomagnesemia with hypercalciuria and nephrocalcinosis	306516	71	71
Primary hypomagnesemia with secondary hypocalcemia	30924	48	48
Primary hypomagnesemia-refractory seizures-intellectual disability syndrome	564178	<10	<10
Primary hypophysitis*	95506	41	38
Primary immunodeficiency syndrome due to 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	83	80
Primary intrahepatic lithiasis	480506	36	33
Primary intraocular lymphoma	279904	<10	<10
Primary intraosseous venous malformation	140436	50	50
Primary laryngeal lymphangioma	137926	18	18
Primary lateral sclerosis	35689	504	425
Primary mediastinal large B-cell lymphoma	98838	<10	<10
Primary melanoma of the central nervous system	252050	<10	<10
Primary membranoproliferative glomerulonephritis	54370	790	742
Primary membranous glomerulonephritis	97560	1598	1489
Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	391408	<10	<10
Primary myelofibrosis	824	20	15

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Primary non-essential cutis verticis gyrata	357225	<10	<10
Primary oculocerebral lymphoma	279897	18	15
Primary orthostatic hypotension*	182058	<10	<10
Primary orthostatic tremor	238606	99	99
Primary pediatric heart tumor	875	20	18
Primary peritoneal carcinoma	168829	<10	<10
Primary progressive aphasia*	95432	56	51
Primary progressive apraxia of speech	314566	<10	<10
Primary progressive freezing gait	75567	<10	<10
Primary pulmonary hypoplasia	2257	57	55
Primary pulmonary lymphoma	2420	<10	<10
Primary pulmonary tuberculosis	645814	34	34
Primary renal tubular acidosis*	314822	17	17
Primary sclerosing cholangitis	171	2760	2620
Primary short bowel syndrome*	365563	<10	<10
Primary Sjögren syndrome	289390	9105	8956
Primary syringomyelia	99856	668	660
Primary tethered cord syndrome	268861	335	334
Primary tuberculous lymphadenitis	645807	<10	<10
Primary unilateral adrenal hyperplasia	231580	966	964
Primitive neuroectodermal tumor of the cervix uteri	213812	<10	<10
Primitive portal vein thrombosis	854	4369	3901
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
Progeroid syndrome, Petty type	2963	<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	22	19
Progressive cone dystrophy	1871	487	481
Progressive deafness with stapes fixation	3235	18	18

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	457212	**<10**	**<10**
Progressive familial intrahepatic cholestasis	172	**419**	**410**
Progressive hemifacial atrophy	1214	**68**	**68**
Progressive multifocal leukoencephalopathy	217260	**<10**	**<10**
Progressive muscular atrophy	454706	**53**	**47**
Progressive myoclonic epilepsy type 1	308	**108**	**106**
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	**34**	**33**
Progressive non-fluent aphasia	100070	**321**	**296**
Progressive non-infectious anterior vertebral fusion	2062	**<10**	**<10**
Progressive osseous heteroplasia	2762	**22**	**22**
Progressive pseudorheumatoid arthropathy of childhood	1159	**25**	**25**
Progressive scapulohumeroperoneal distal myopathy	447977	**<10**	**<10**
Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	228012	**<10**	**<10**
Progressive supranuclear palsy	683	**1036**	**880**
Progressive symmetric erythrokeratodermia	316	**13**	**13**
Prolactinoma	2965	**6304**	**6221**
Prolidase deficiency	742	**<10**	**<10**
Proliferating trichilemmal cyst	492	**<10**	**<10**
Properdin deficiency	2966	**11**	**<10**
Propionic acidemia	35	**154**	**130**
Proteasome-associated autoinflammatory syndrome	324977	**<10**	**<10**
Protein S acquired deficiency	26349	**11**	**11**
Proteus syndrome	744	**147**	**145**
Proteus-like syndrome	2969	**<10**	**<10**
Proton-pump inhibitor-responsive esophageal eosinophilia	411696	**<10**	**<10**
Proximal 16p11.2 microdeletion syndrome	261197	**190**	**190**
Proximal 16p11.2 microduplication syndrome	370079	**106**	**106**
Proximal femoral focal deficiency	633228	**<10**	**<10**
Proximal myopathy with focal depletion of mitochondria	521305	**<10**	**<10**
Proximal myotonic myopathy	606	**776**	**694**
Proximal renal tubular acidosis	47159	**57**	**54**
Proximal spinal muscular atrophy	70	**2361**	**1929**
Proximal symphalangism	3250	**15**	**15**
Proximal Xq28 duplication syndrome	1762	**49**	**48**
PrP systemic amyloidosis	397606	**<10**	**<10**
Prune belly syndrome	2970	**82**	**78**
PRUNE1-related neurological syndrome	544469	**<10**	**<10**
Pseudoachondroplasia	750	**85**	**85**
Pseudodiastrophic dysplasia	85174	**<10**	**<10**
Pseudohypoaldosteronism type 1	756	**107**	**103**
Pseudohypoaldosteronism type 2	757	**52**	**52**
Pseudohypoaldosteronism*	444916	**29**	**28**
Pseudohypoparathyroidism type 1A	79443	**290**	**289**
Pseudohypoparathyroidism type 1B	94089	**166**	**164**
Pseudohypoparathyroidism type 1C	79444	**<10**	**<10**
Pseudohypoparathyroidism type 2	94090	**<10**	**<10**
Pseudohypoparathyroidism with Albright hereditary osteodystrophy*	457059	**37**	**36**
Pseudohypoparathyroidism without Albright hereditary osteodystrophy*	457062	**<10**	**<10**
Pseudomyxoma peritonei	26790	**<10**	**<10**
Pseudoprogeria syndrome	2985	**<10**	**<10**
Pseudopseudohypoparathyroidism	79445	**107**	**106**
Pseudounicornuate uterus	180079	**<10**	**<10**
Pseudo-von Willebrand disease	52530	**27**	**27**
Pseudoxanthoma elasticum	758	**570**	**564**
Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa	436274	**<10**	**<10**
Psoriasis-related juvenile idiopathic arthritis	85436	**462**	**461**
Psychogenic movement disorders	71519	**194**	**190**
Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome	505242	**<10**	**<10**
PTEN hamartoma tumor syndrome*	306498	**57**	**57**

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 neuralgia	60039	<10	<10
Pulmonary agenesis	984	64	61
Pulmonary alveolar microlithiasis	60025	16	14
Pulmonary arterial hypertension associated with chronic hemolytic anemia*	275828	<10	<10
Pulmonary arterial hypertension associated with congenital heart disease*	275803	477	446
Pulmonary arterial hypertension associated with connective tissue disease*	275798	452	364
Pulmonary arterial hypertension associated with HIV infection*	275808	58	53
Pulmonary arterial hypertension associated with portal hypertension*	275813	258	212
Pulmonary arterial hypertension associated with schistosomiasis*	275823	<10	<10
Pulmonary arteriovenous malformation	2038	51	50
Pulmonary artery coming from patent ductus arteriosus	99049	<10	<10
Pulmonary artery hypoplasia	99083	26	26
Pulmonary atresia with ventricular septal defect	1207	607	582
Pulmonary atresia-intact ventricular septum syndrome	1208	354	343
Pulmonary blastoma	64741	<10	<10
Pulmonary capillary hemangiomatosis	199241	<10	<10
Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome	210136	29	25
Pulmonary hypertension owing to lung disease and/or hypoxia*	275837	765	594
Pulmonary hypertension with unclear multifactorial mechanism*	275844	236	191
Pulmonary interstitial glycogenosis	217557	<10	<10
Pulmonary nodular lymphoid hyperplasia	60026	<10	<10
Pulmonary non-tuberculous mycobacterial infection	411703	202	194
Pulmonary valve agenesis*	982	60	60
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	19	18
Pulmonary venoocclusive disease	31837	287	214
Punctate inner choroidopathy	580951	18	18
Punctate palmoplantar keratoderma type 1	79501	12	12
Punctate palmoplantar keratoderma type 2	79502	<10	<10
PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	438213	39	38
Pure autonomic failure	441	45	41
Pure hair and nail ectodermal dysplasia	69084	43	43
Pure hereditary spastic paraplegia*	102012	369	363
Pure mitochondrial myopathy	254854	527	509
Pure or complex hereditary spastic paraplegia*	320335	67	67
Purine nucleoside phosphorylase deficiency	760	<10	<10
Pustulosis palmaris et plantaris	163927	<10	<10
Pycnodysostosis	763	81	81
PYCR2-related microcephaly-progressive leukoencephalopathy	481152	<10	<10
Pyknoachondrogenesis	3003	<10	<10
Pyle disease	3005	<10	<10
Pyoderma gangrenosum	48104	29	25
Pyomyositis	764	<10	<10
Pyridoxal phosphate-responsive seizures	79096	19	18
Pyridoxine-dependent epilepsy	3006	67	63
Pyruvate carboxylase deficiency	3008	<10	<10
Pyruvate dehydrogenase deficiency	765	153	140
Q fever	781	<10	<10
Qazi-Markouizos syndrome	3010	<10	<10
QRICH1-related intellectual disability-chondrodysplasia syndrome	580940	<10	<10
QRSL1-related combined oxidative phosphorylation defect	570491	<10	<10
Quadricuspid aortic valve	542568	<10	<10
Quebec platelet disorder	220436	54	53
Quinquaud folliculitis decalvans	346	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Rabson-Mendenhall syndrome	769	**<10**	**<10**
Radial deficiency-tibial hypoplasia syndrome	1121	**<10**	**<10**
Radial hemimelia	93321	**89**	**88**
Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome	2252	**<10**	**<10**
Radial ray hypoplasia-choanal atresia syndrome	3026	**<10**	**<10**
Radiation myelitis	90021	**24**	**22**
Radiation-induced plexopathy	521123	**51**	**47**
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	**247**	**247**
Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	293987	**45**	**41**
Rapid-onset dystonia-parkinsonism	71517	**17**	**15**
Rare adenocarcinoma of the breast	213528	**12**	**<10**
Rare combined vascular malformation*	458837	**526**	**524**
Rare cutaneous lupus erythematosus*	535	**239**	**233**
Rare disorder without a determined diagnosis after full investigation	616874	**256**	**256**
Rare hereditary thrombophilia*	217454	**<10**	**<10**
Rare isolated myopia	98619	**315**	**312**
Rare non-syndromic intellectual disability	101685	**16482**	**16316**
RARS-related autosomal recessive hypomyelinating leukodystrophy	438114	**<10**	**<10**
RAS-associated autoimmune leukoproliferative disease	268114	**<10**	**<10**
Rasmussen subacute encephalitis	1929	**109**	**107**
Rat-bite fever	31205	**<10**	**<10**
Ravine syndrome	99852	**62**	**31**
Reactive arthritis	29207	**513**	**513**
Reading seizures	166433	**<10**	**<10**
Recessive dystrophic epidermolysis bullosa inversa	79409	**17**	**17**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome	280384	**<10**	**<10**
Recessive mitochondrial ataxia syndrome	94125	**18**	**16**
Recessive X-linked ichthyosis	461	**191**	**189**
Recombinant 8 syndrome	96167	**<10**	**<10**
Rectal duplication	171220	**<10**	**<10**
Recurrent hepatitis C virus induced liver disease in liver transplant recipients	90052	**<10**	**<10**
Recurrent idiopathic neuroretinitis	499103	**<10**	**<10**
Recurrent infection due to specific granule deficiency	169142	**<10**	**<10**
Recurrent infections associated with rare immunoglobulin isotypes deficiency	183675	**498**	**496**
Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome	480864	**33**	**29**
Recurrent respiratory papillomatosis	60032	**105**	**105**
Reducing body myopathy	97239	**<10**	**<10**
Reflex epilepsy*	310	**25**	**25**
Refractory anemia	98826	**<10**	**<10**
Refractory anemia with excess blasts	86839	**<10**	**<10**
Refractory celiac disease	398063	**28**	**28**
Refsum disease	773	**31**	**27**
Regional odontodysplasia	83450	**<10**	**<10**
Regressive spondylometaphyseal dysplasia	448267	**<10**	**<10**
Reis-Bücklers corneal dystrophy	98961	**29**	**29**
Relapsing fever	91547	**209**	**209**
Relapsing polychondritis	728	**595**	**580**
Renal agenesis	411709	**3029**	**2995**
Renal caliceal diverticuli-deafness syndrome	2838	**<10**	**<10**
Renal cell carcinoma*	217071	**29**	**27**
Renal coloboma syndrome	1475	**179**	**177**
Renal dysplasia	93108	**2502**	**2479**
Renal hypoplasia	93101	**3193**	**3163**
Renal medullary carcinoma	319319	**<10**	**<10**
Renal nutcracker syndrome	71273	**15**	**15**
Renal tubular dysgenesis	3033	**45**	**36**

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	**787**	**766**
Renpenning syndrome	3242	**37**	**37**
RERE-related neurodevelopmental syndrome	494344	**<10**	**<10**
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha	566231	**<10**	**<10**
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta	566243	**66**	**66**
Resistance to thyrotropin-releasing hormone syndrome	99832	**<10**	**<10**
Respiratory bronchiolitis-interstitial lung disease syndrome	79127	**163**	**159**
Restrictive dermopathy	1662	**<10**	**<10**
Reticular dysgenesis	33355	**<10**	**<10**
Reticular dystrophy of the retinal pigment epithelium	99002	**167**	**163**
Reticulate acropigmentation of Kitamura	178307	**<10**	**<10**
Retinal arterial tortuosity	75326	**13**	**13**
Retinal capillary malformation	71213	**12**	**12**
Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies	397758	**<10**	**<10**
Retinal dystrophy-optic nerve edema-splenomegaly-anhidrosis-migraine headache syndrome	313800	**<10**	**<10**
Retinal macular dystrophy type 2	319640	**27**	**27**
Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	247691	**<10**	**<10**
Retinitis pigmentosa	791	**4715**	**4695**
Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome	3085	**<10**	**<10**
Retinitis punctata albescens	52427	**30**	**30**
Retinoblastoma	790	**224**	**223**
Retinopathy of prematurity	90050	**363**	**363**
Rett syndrome	778	**786**	**720**
Reversible cerebral vasoconstriction syndrome	284388	**<10**	**<10**
Revesz syndrome	3088	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Reye syndrome	3096	**15**	**13**
Reynolds syndrome	779	**43**	**39**
RFT1-CDG	244310	**<10**	**<10**
Rhabdoid tumor	69077	**21**	**16**
Rhabdomyosarcoma	780	**114**	**110**
Rhabdomyosarcoma of the cervix uteri	213802	**<10**	**<10**
Rheumatic fever	3099	**72**	**71**
Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	85408	**1175**	**1169**
Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	85435	**536**	**522**
Rhizomelic chondrodysplasia punctata	177	**18**	**15**
Rhombencephalosynapsis	59315	**35**	**33**
RHYNS syndrome	140976	**<10**	**<10**
Riboflavin transporter deficiency	97229	**34**	**34**
Richieri Costa-Pereira syndrome	3102	**<10**	**<10**
Rieger anomaly	91483	**36**	**35**
Right aortic arch	99081	**93**	**93**
Right inferior vena cava connecting to left-sided atrium	99119	**<10**	**<10**
Right sided atrial isomerism	97548	**50**	**42**
Right superior vena cava connecting to left-sided atrium	99110	**<10**	**<10**
Rigid spine syndrome	97244	**51**	**49**
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	**12**	**11**
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	**28**	**27**
Ring chromosome 19 syndrome	1443	**<10**	**<10**
Ring chromosome 2 syndrome	96171	**<10**	**<10**
Ring chromosome 20 syndrome	1444	**43**	**43**
Ring chromosome 21 syndrome	1445	**18**	**18**
Ring chromosome 22 syndrome	1446	**25**	**23**
Ring chromosome 3 syndrome	96172	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Ring chromosome 4 syndrome	1447	<10	<10
Ring chromosome 7 syndrome	1449	<10	<10
Ring chromosome 8 syndrome	1450	11	11
Ring chromosome 9 syndrome	96173	<10	<10
Ring chromosome Y syndrome	261529	<10	<10
Ring dermoid of cornea	91481	32	32
Rippling muscle disease	97238	<10	<10
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	18	18
Roifman syndrome	353298	11	11
Rolandic epilepsy	1945	712	712
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	292	287
Rosaï-Dorfman disease	158014	127	125
Rosette-forming glioneuronal tumor	251975	<10	<10
Rothmund-Thomson syndrome	2909	41	37
Rotor syndrome	3111	<10	<10
Roussy-Lévy syndrome	3115	<10	<10
Rubella panencephalitis	83616	<10	<10
Rubinstein-Taybi syndrome	783	467	457
Saccular limited dorsal myeloschisis	645354	<10	<10
Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome	397927	16	16
S-adenosylhomocysteine hydrolase deficiency	88618	<10	<10
Saethre-Chotzen syndrome	794	176	175
Saldino-Mainzer syndrome	140969	<10	<10
Sandhoff disease	796	46	25
Sandifer syndrome	71272	<10	<10
Sanjad-Sakati syndrome	2323	<10	<10
SAPHO syndrome	793	295	295
Sarcoidosis	797	10384	10174
Sarcoma of the corpus uteri*	213620	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
SATB2-associated syndrome	576278	60	60
SCALP syndrome	370052	<10	<10
Scalp-ear-nipple syndrome	2036	<10	<10
Scapuloperoneal spinal muscular atrophy	431255	18	18
Schilbach-Rott syndrome	2353	<10	<10
Schimke immuno-osseous dysplasia	1830	29	18
Schinzel-Giedion syndrome	798	23	12
Schistosomiasis	1247	14	13
Schizencephaly	799	97	96
Schneckenbecken dysplasia	3144	<10	<10
Schnitzler syndrome	37748	68	67
Schnyder corneal dystrophy	98967	<10	<10
Schöpf-Schulz-Passarge syndrome	50944	<10	<10
Schwartz-Jampel syndrome	800	18	18
Scimitar syndrome	185	152	150
Scleredema	352763	<10	<10
Scleroderma*	801	1267	1157
Scleromyxedema	167635	12	12
Sclerosing cholangitis*	447771	229	219
Sclerosteosis	3152	<10	<10
Scott syndrome	806	39	38
Sebocystomatosis	841	21	21
Seborrhea-like dermatitis with psoriasiform elements	168606	<10	<10
Seckel syndrome	808	42	38
Second branchial cleft anomaly	141022	337	337
Secondary central precocious puberty in female	650082	<10	<10
Secondary erythromelalgia	529864	37	37
Secondary hypereosinophilic syndrome	314962	149	145
Secondary hypoparathyroidism due to impaired parathormon secretion	140286	323	318
Secondary intestinal lymphangiectasia	90363	<10	<10
Secondary non-traumatic avascular necrosis	399180	<10	<10
Secondary pulmonary alveolar proteinosis	420259	11	<10
Secondary pulmonary hemosiderosis	99930	<10	<10
Secondary sclerosing cholangitis	447774	175	156
Secondary short bowel syndrome	95427	1515	1357

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Secondary syringomyelia	99857	**361**	**338**
Segmental odontomaxillary dysplasia	67039	**<10**	**<10**
Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome	137608	**<10**	**<10**
Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	314662	**30**	**30**
Seizures-scoliosis-macrocephaly syndrome	466926	**<10**	**<10**
Selective IgM deficiency	331235	**39**	**38**
Selective intrauterine growth restriction	617301	**31**	**31**
Self-healing papular mucinosis	90397	**<10**	**<10**
Self-improving collodion baby	281122	**<10**	**<10**
Semantic dementia	100069	**268**	**255**
Semicircular canal dehiscence syndrome	420402	**<10**	**<10**
Senior-Boichis syndrome	84081	**16**	**11**
Senior-Loken syndrome	3156	**85**	**81**
Sensorineural deafness with dilated cardiomyopathy	217622	**<10**	**<10**
Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	70595	**36**	**29**
Sepsis in premature infants	90051	**<10**	**<10**
Septate uterus*	180122	**32**	**32**
Septate vagina	180154	**40**	**40**
Septo-optic dysplasia spectrum	3157	**426**	**424**
Serine biosynthesis pathway deficiency, infantile/juvenile form	583595	**<10**	**<10**
SERKAL syndrome	139466	**<10**	**<10**
Serous cystadenocarcinoma of pancreas	424073	**159**	**158**
Serpiginous choroiditis	35686	**61**	**61**
Serrated polyposis syndrome	157798	**19**	**19**
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 CARD11 deficiency	357237	**<10**	**<10**
Severe combined immunodeficiency due to complete RAG1/2 deficiency	331206	**16**	**15**
Severe combined immunodeficiency due to CORO1A deficiency	228003	**<10**	**<10**
Severe combined immunodeficiency due to DCLRE1C deficiency	275	**38**	**34**
Severe combined immunodeficiency due to FOXN1 deficiency	169095	**<10**	**<10**
Severe combined immunodeficiency due to LCK deficiency	280142	**<10**	**<10**
Severe combined immunodeficiency*	183660	**68**	**65**
Severe congenital nemaline myopathy	171430	**38**	**30**
Severe congenital neutropenia*	42738	**716**	**714**
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	**93**	**92**
Severe early-onset axonal neuropathy due to MFN2 deficiency	90118	**21**	**21**
Severe early-onset pulmonary alveolar proteinosis due to MARS deficiency	440427	**34**	**33**
Severe generalized junctional epidermolysis bullosa	79404	**16**	**<10**
Severe hereditary thrombophilia due to congenital protein C deficiency	745	**65**	**64**
Severe hereditary thrombophilia due to congenital protein S deficiency	743	**41**	**40**
Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome	467176	**<10**	**<10**
Severe intellectual disability and progressive spastic paraplegia	280763	**32**	**32**
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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	397933	**21**	**21**
Severe intellectual disability-progressive spastic diplegia syndrome	404473	**44**	**44**
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 neonatal-onset encephalopathy with microcephaly	209370	**77**	**70**
Severe neurodegenerative syndrome with lipodystrophy	363400	**<10**	**<10**
Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract	500545	**<10**	**<10**
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	**113**	**109**
Sheldon-Hall syndrome	1147	**19**	**19**
Shigellosis	810	**<10**	**<10**
Shone complex	99063	**182**	**177**
Short bowel syndrome*	104008	**758**	**709**
Short chain acyl-CoA dehydrogenase deficiency	26792	**151**	**145**
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	**31**	**31**
Short stature, Brussels type	2867	**<10**	**<10**
Short stature-advanced bone age-early-onset osteoarthritis syndrome	435804	**29**	**29**
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**
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	**183**	**182**
Shprintzen-Goldberg syndrome	2462	**47**	**44**
Shwachman-Diamond syndrome	811	**127**	**120**
Sialidosis type 1	812	**<10**	**<10**
Sialidosis type 2	87876	**<10**	**<10**
Sialidosis*	309294	**<10**	**<10**
Sialuria	3166	**<10**	**<10**
Sickle cell anemia	232	**13296**	**13129**
Sickle cell-beta-thalassemia disease syndrome	251359	**1520**	**1505**
Sickle cell-hemoglobin C disease syndrome	251365	**3262**	**3234**
Sickle cell-hemoglobin D disease syndrome	251370	**20**	**20**
Sickle cell-hemoglobin E disease syndrome	251375	**38**	**38**
Silent sinus syndrome	71276	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Sillence syndrome	3168	<10	<10
Silver-Russell syndrome	813	821	816
SIM1-related Prader-Willi-like syndrome	398079	<10	<10
Simple cryoglobulinemia	91139	142	131
Simpson-Golabi-Behmel syndrome	373	75	68
Sinding-Larsen-Johansson disease	97337	<10	<10
Singleton-Merten dysplasia	85191	<10	<10
Sinoatrial node dysfunction and deafness	324321	<10	<10
Sitosterolemia	2882	16	15
Situs ambiguus	157769	15	14
Situs inversus totalis	101063	69	66
Sjögren-Larsson syndrome	816	54	51
Skeletal dysplasia-epilepsy-short stature syndrome	1858	<10	<10
Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome	508533	<10	<10
Skeletal Ewing sarcoma	319	71	68
Skin fragility-woolly hair-palmoplantar keratoderma syndrome	293165	<10	<10
SLC35A1-CDG	238459	<10	<10
SLC35A2-CDG	356961	<10	<10
SLC39A8-CDG	468699	<10	<10
SLC40A1-related hemochromatosis	647834	<10	<10
Slender bone dysplasia*	93440	<10	<10
Small bowel atresia	1201	218	215
Small cell carcinoma of the bladder	284400	<10	<10
Small cell carcinoma of the ovary	370396	<10	<10
Small cell lung cancer	70573	28	18
Smith-Lemli-Opitz syndrome	818	93	82
Smith-Magenis syndrome	819	394	387
Smith-McCort dysplasia	178355	<10	<10
Smoldering systemic mastocytosis	158775	28	28
Sneddon syndrome	820	61	56
Sodium channelopathy-related small fiber neuropathy	306577	498	492
Soft tissue sarcoma*	3394	11	<10
Solid pseudopapillary carcinoma of pancreas	424065	26	26
Solitary bone cyst	83468	<10	<10
Solitary fibrous tumor	2126	<10	<10
Solitary rectal ulcer syndrome	209964	<10	<10
Somatomammotropinoma	314769	347	342
Somatotropic adenoma*	96256	52	50
Sorsby pseudoinflammatory fundus dystrophy	59181	<10	<10
Sotos syndrome	821	622	615
Southeast Asian ovalocytosis	98868	<10	<10
Spasmus nutans	279882	<10	<10
Spastic ataxia with congenital miosis	1182	<10	<10
Spastic ataxia*	316226	125	124
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	29	25
Spastic paraplegia type 2	99015	83	82
Spastic paraplegia type 7	99013	183	176
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

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	447997	<10	<10
SPECC1L-related hypertelorism syndrome	1519	28	26
Specific language disorder*	211053	62	62
Specific learning disability*	211047	1949	1941
Spectrin-associated autosomal recessive cerebellar ataxia	352403	<10	<10
Spigelian hernia-cryptorchidism syndrome	314432	<10	<10
Spina bifida aperta*	268369	54	50
Spina bifida cystica*	268744	11	11
Spina bifida-hypospadias syndrome	3176	<10	<10
Spinal arteriovenous metameric syndrome	53721	27	27
Spinal atrophy-ophthalmoplegia-pyramidal syndrome	1217	<10	<10
Spinal cord injury	90058	45	42
Spinal dermal sinus	645188	<10	<10
Spinal muscular atrophy associated with central nervous system anomaly*	207012	<10	<10
Spinal muscular atrophy with respiratory distress type 1	98920	73	33
Spinal muscular atrophy with respiratory distress type 2	404521	33	30
Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	2590	<10	<10
Spindle cell hemangioma	210584	<10	<10
Spinocerebellar ataxia type 1	98755	108	102
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	44	44
Spinocerebellar ataxia type 15/16	98769	19	19
Spinocerebellar ataxia type 17	98759	14	13
Spinocerebellar ataxia type 19/22	98772	33	33
Spinocerebellar ataxia type 2	98756	158	146
Spinocerebellar ataxia type 20	101110	<10	<10
Spinocerebellar ataxia type 21	98773	22	22
Spinocerebellar ataxia type 23	101108	<10	<10
Spinocerebellar ataxia type 25	101111	<10	<10
Spinocerebellar ataxia type 26	101112	<10	<10
Spinocerebellar ataxia type 27	98764	66	66
Spinocerebellar ataxia type 28	101109	28	27
Spinocerebellar ataxia type 29	208513	44	43
Spinocerebellar ataxia type 3	98757	258	249
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	21	21
Spinocerebellar ataxia type 37	363710	<10	<10
Spinocerebellar ataxia type 4	98765	<10	<10
Spinocerebellar ataxia type 41	458798	<10	<10
Spinocerebellar ataxia type 42	458803	13	13
Spinocerebellar ataxia type 43	497764	<10	<10
Spinocerebellar ataxia type 45	589527	<10	<10
Spinocerebellar ataxia type 48	631103	<10	<10
Spinocerebellar ataxia type 5	98766	23	23
Spinocerebellar ataxia type 6	98758	50	50
Spinocerebellar ataxia type 7	94147	81	71
Spinocerebellar ataxia type 8	98760	<10	<10
Spinocerebellar ataxia with axonal neuropathy type 1	94124	17	13
Spinocerebellar ataxia with axonal neuropathy type 2	64753	56	55
Spinocerebellar ataxia with epilepsy	254881	17	16
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	573278	<10	<10
Split cord malformation type I	1671	59	59
Split cord malformation type II	573253	<10	<10
Split hand-split foot-deafness syndrome	71271	<10	<10
SPONASTRIME dysplasia	93357	11	<10
Spondylocamptodactyly syndrome	3180	<10	<10
Spondylocarpotarsal synostosis	3275	27	27
Spondylodysplastic dysplasia*	93434	12	12
Spondylodysplastic Ehlers-Danlos syndrome	536471	14	14
Spondyloenchondrodysplasia	1855	<10	<10
Spondyloepimetaphyseal dysplasia congenita, Strudwick type	93346	37	37

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type	93360	<10	<10
Spondyloepimetaphyseal dysplasia, aggrecan type	171866	11	11
Spondyloepimetaphyseal dysplasia, Isidor-Toutain type	370015	<10	<10
Spondyloepimetaphyseal dysplasia, matrilin-3 type	156728	<10	<10
Spondyloepimetaphyseal dysplasia, Missouri type	93356	<10	<10
Spondyloepimetaphyseal dysplasia, PAPSS2 type	93282	<10	<10
Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome	93358	<10	<10
Spondyloepiphyseal dysplasia and spondyloepimetaphyseal dysplasia*	253	112	110
Spondyloepiphyseal dysplasia congenita	94068	132	130
Spondyloepiphyseal dysplasia tarda	93284	79	76
Spondyloepiphyseal dysplasia with metatarsal shortening	137678	<10	<10
Spondyloepiphyseal dysplasia, Maroteaux type	263482	<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	44	44
Spondylometaphyseal dysplasia, 'corner fracture' type	93315	<10	<10
Spondylometaphyseal dysplasia, Golden type	168544	<10	<10
Spondylometaphyseal dysplasia, Kozlowski type	93314	23	22
Spondylometaphyseal dysplasia, Schmidt type	93316	29	29
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	13	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Sporadic adult-onset ataxia of unknown etiology	247234	131	126
Sporadic Creutzfeldt-Jakob disease	204	15	<10
Sporadic fetal brain disruption sequence	1665	<10	<10
Sporadic pheochromocytoma/secreting paraganglioma	276621	1289	1255
Sprengel deformity	3181	32	32
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	<10	<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	<10	<10
Staphylococcal scalded skin syndrome	36236	13	13
Stargardt disease	827	1513	1504
Startle epilepsy	166427	<10	<10
STAT3-related early-onset multisystem autoimmune disease	438159	15	15
Steatocystoma multiplex-natal teeth syndrome	3184	<10	<10
Steel syndrome	438117	<10	<10
Steinert myotonic dystrophy	273	6827	5778
Sterile multifocal osteomyelitis with periostitis and pustulosis	210115	91	91
Sternal cleft	2017	<10	<10
Steroid-responsive encephalopathy associated with autoimmune thyroiditis	83601	20	20

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	95455	546	481
Stickler syndrome	828	873	869
Stiff person spectrum disorder	3198	103	96
Stiff skin syndrome	2833	<10	<10
STING-associated vasculopathy with onset in infancy	425120	15	13
Stormorken-Sjaastad-Langslet syndrome	3204	13	12
Straddling or overriding tricuspid valve	95461	<10	<10
Striate palmoplantar keratoderma	50942	<10	<10
Stromme syndrome	506307	<10	<10
STT3B-CDG	370924	<10	<10
Sturge-Weber syndrome	3205	519	516
Stüve-Wiedemann syndrome	3206	22	21
STXBP1-related encephalopathy	599373	45	45
Subacute cutaneous lupus erythematosus	163525	179	176
Subacute inflammatory demyelinating polyneuropathy	206594	202	168
Subacute sclerosing leukoencephalitis	2806	<10	<10
Subaortic stenosis-short stature syndrome	3191	<10	<10
Subcorneal pustular dermatosis	48377	12	12
Subcortical band heterotopia	99796	86	86
Subcutaneous panniculitis-like T-cell lymphoma	86884	<10	<10
Subependymal giant cell astrocytoma	251618	<10	<10
Submucosal cleft palate	155878	128	127
Succinic semialdehyde dehydrogenase deficiency	22	53	52
Succinyl-CoA:3-oxoacid CoA transferase deficiency	832	15	15
Sudden infant death-dysgenesis of the testes syndrome	168593	<10	<10
Sudden sensorineural hearing loss	90059	<10	<10
Suggest an update	93672	347	347
Suggest an update*	178566	<10	<10
SUNCT syndrome	57145	<10	<10
Superficial epidermolytic ichthyosis	455	15	15
Superficial fibromatosis*	199257	<10	<10
Superficial pemphigus*	46485	256	246
Superficial siderosis	247245	<10	<10
Superior limbic keratoconjunctivitis	88633	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Supernumerary breasts	180182	<10	<10
Supratip dysplasia	466695	<10	<10
Supravalvular aortic stenosis	3193	116	112
SURF1-related Charcot-Marie-Tooth disease type 4	391351	<10	<10
Susac syndrome	838	88	88
Susceptibility to respiratory infections associated with CD8alpha chain mutation	169085	<10	<10
Susceptibility to viral and mycobacterial infections due to STAT1 deficiency	391311	40	37
Sweet syndrome	3243	48	47
Sydenham chorea	306731	11	11
Symbrachydactyly of hands and feet	1570	25	25
Symmetrical thalamic calcifications	1314	<10	<10
Sympathetic ophthalmia	79098	37	35
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 fragile X syndrome in female carriers	449291	30	30
Symptomatic form of HFE-related hemochromatosis	465508	254	245
Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers	206546	231	222
Symptomatic form of X-linked centronuclear myopathy in female carriers	604680	<10	<10
Syndactyly type 1	93402	118	118
Syndactyly type 2	93403	54	53
Syndactyly type 3	93404	22	22
Syndactyly type 4	93405	17	17
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	51	45

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Syndromic microphthalmia type 5	178364	**<10**	**<10**
Syndromic orbital border hypoplasia	98606	**<10**	**<10**
Syndromic recessive X-linked ichthyosis	281090	**32**	**32**
Syndromic X-linked intellectual disability 7	85274	**<10**	**<10**
SYNGAP1-related developmental and epileptic encephalopathy	544254	**77**	**77**
Synovial sarcoma	3273	**<10**	**<10**
Syringocystadenoma papilliferum	840	**<10**	**<10**
Syringomyelia*	3280	**383**	**374**
Systemic capillary leak syndrome	188	**32**	**30**
Systemic diseases with anterior uveitis*	280926	**161**	**160**
Systemic diseases with panuveitis*	280933	**109**	**109**
Systemic Epstein-Barr virus-positive T-cell lymphoproliferative disease of childhood	364033	**<10**	**<10**
Systemic lupus erythematosus	536	**12337**	**12145**
Systemic mastocytosis with associated hematologic neoplasm	98849	**56**	**52**
Systemic mastocytosis*	2467	**914**	**901**
Systemic primary carnitine deficiency	158	**99**	**95**
Systemic sclerosis	90291	**7895**	**7411**
Systemic-onset juvenile idiopathic arthritis	85414	**1383**	**1369**
T+ B+ severe combined immunodeficiency*	397802	**<10**	**<10**
TAFRO syndrome	457077	**12**	**12**
Takayasu arteritis	3287	**785**	**768**
Tako-Tsubo cardiomyopathy	66529	**169**	**158**
Tall stature-intellectual disability-renal anomalies syndrome	500095	**<10**	**<10**
Tangier disease	31150	**<10**	**<10**
TARP syndrome	2886	**12**	**12**
Tarsal-carpal coalition syndrome	1412	**<10**	**<10**
Tatton-Brown-Rahman syndrome	404443	**63**	**63**
Tay-Sachs disease	845	**77**	**44**
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	**39**	**35**
T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	169154	**18**	**17**
T-B+ severe combined immunodeficiency due to JAK3 deficiency	35078	**29**	**29**
T-B+ severe combined immunodeficiency*	317416	**<10**	**<10**
TBCK-related intellectual disability 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	**41**	**40**
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	**53**	**53**
Temple-Baraitser syndrome	420561	**<10**	**<10**
Temtamy preaxial brachydactyly syndrome	363417	**<10**	**<10**
Temtamy syndrome	1777	**11**	**11**
Tenosynovial giant cell tumor	66627	**16**	**16**
Terminal myelocystocele	645337	**<10**	**<10**
Terminal osseous dysplasia-pigmentary defects syndrome	88630	**<10**	**<10**
Terrien marginal degeneration	519410	**<10**	**<10**
Tessier number 4 facial cleft	141258	**<10**	**<10**
Tessier number 5 facial cleft	141261	**<10**	**<10**
Tessier number 6 facial cleft	141265	**<10**	**<10**
Tessier number 7 facial cleft	141276	**75**	**75**
Testicular agenesis	325124	**194**	**194**
Testicular regression syndrome	983	**175**	**175**
Testicular seminomatous germ cell tumor	842	**<10**	**<10**
Tetraamelia-multiple malformations syndrome	3301	**<10**	**<10**
Tetragametic chimerism	199310	**<10**	**<10**
Tetralogy of Fallot	3303	**3029**	**2990**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Tetramelic monodactyly	2564	<10	<10
Tetrasomy 12p	884	89	81
Tetrasomy 18p	3307	51	51
Tetrasomy 21	96055	<10	<10
Tetrasomy 9p	3310	23	22
Tetrasomy X	9	17	17
TFR2-related hemochromatosis	225123	34	33
Thakker-Donnai syndrome	1780	45	41
Thalidomide embryopathy	3312	<10	<10
Thanatophoric dysplasia	2655	32	14
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	17	17
Thiemann disease, familial form	3314	<10	<10
Thinking seizures	166424	<10	<10
Third branchial cleft anomaly	141030	20	20
THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	363444	<10	<10
Thomas syndrome	3316	<10	<10
Thomsen and Becker disease	614	468	457
Thoracic dysplasia-hydrocephalus syndrome	1861	<10	<10
Thoracic outlet syndrome	97330	31	31
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	53	51
Thrombomodulin-related bleeding disorder	436169	12	12
Thrombotic microangiopathy*	93573	618	506
Thrombotic thrombocytopenic purpura	54057	899	861
Thumb deformity-alopecia-pigmentation anomaly syndrome	2251	<10	<10

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Thygeson superficial punctate keratitis	519406	15	15
Thymic carcinoma	99868	<10	<10
Thymic neuroendocrine carcinoma	99869	<10	<10
Thymic neuroendocrine tumor	97289	<10	<10
Thymic-renal-anal-lung dysplasia	3326	<10	<10
Thymoma	99867	68	55
Thyroid ectopia	95712	1182	1180
Thyroid hemiagenesis	95719	46	46
Thyroid hypoplasia	95720	59	59
Thyroid lymphoma	97285	<10	<10
Thyrotoxic periodic paralysis	79102	13	13
Tibial aplasia-ectrodactyly syndrome	3329	27	27
Tibial hemimelia	93322	35	35
Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome	988	15	15
Tibial muscular dystrophy	609	75	70
Tick-borne encephalitis	297	<10	<10
Tietz syndrome	42665	<10	<10
Timothy syndrome	65283	15	14
Titin-related limb-girdle muscular dystrophy R10	140922	68	68
TMEM165-CDG	314667	<10	<10
TMEM199-CDG	466703	<10	<10
TMEM70-related mitochondrial encephalo-cardio-myopathy	1194	18	17
TNP03-related limb-girdle muscular dystrophy D2	55595	<10	<10
Tolosa-Hunt syndrome	64686	22	21
TOR1AIP1-related limb-girdle muscular dystrophy	424261	<10	<10
Toriello-Carey syndrome	3338	<10	<10
Toriello-Lacassie-Droste syndrome	3339	<10	<10
Torsade-de-pointes syndrome with short coupling interval	51084	30	30
Torticollis-keloids-cryptorchidism-renal dysplasia syndrome	3341	<10	<10
Townes-Brocks syndrome	857	179	177
Toxic maculopathy due to antimalarial drugs	279894	40	40
Toxocariasis	3343	16	16
Tracheal agenesis	3346	14	<10
Tracheobronchopathia osteochondroplastica	3348	15	14

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	14
Transcobalamin deficiency	859	<10	<10
Transgrediens et progrediens palmoplantar keratoderma	495	12	12
Transient congenital hypothyroidism*	178045	56	56
Transient erythroblastopenia of childhood	98871	13	12
Transient familial neonatal hyperbilirubinemia	2312	21	21
Transient hyperammonemia of the newborn	289877	<10	<10
Transient hypogammaglobulinemia of infancy	169139	<10	<10
Transient infantile hypertriglyceridemia and hepatosteatosis	300293	<10	<10
Transient myeloproliferative syndrome	420611	105	102
Transient neonatal diabetes mellitus	99886	37	37
Transient pseudohypoaldosteronism	93164	<10	<10
TRAPPC11-related limb-girdle muscular dystrophy R18	369840	<10	<10
Traumatic avascular necrosis	399175	<10	<10
Treacher-Collins syndrome	861	372	364
Trehalase deficiency	103909	<10	<10
Triatrial heart*	1463	34	32
Trichinellosis	863	<10	<10
Trichodental syndrome	3351	<10	<10
Tricho-dento-osseous syndrome	3352	<10	<10
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	65	65
Trichorhinophalangeal syndrome type 2	502	32	32
Trichorhinophalangeal syndrome*	324764	71	71
Trichothiodystrophy	33364	52	47
Tricuspid atresia	1209	307	287
Trigeminal neuralgia	221091	<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	16	15
Triose phosphate-isomerase deficiency	868	12	<10
Triphalangeal thumbs-brachyectrodactyly syndrome	2947	<10	<10
Triple A syndrome	869	100	95
Triploidy	3376	20	17
Trismus-pseudocamptodactyly syndrome	3377	<10	<10
Trisomy 10p	171929	<10	<10
Trisomy 12p	1699	32	32
Trisomy 13	3378	155	78
Trisomy 17p	261290	<10	<10
Trisomy 18	3380	229	115
Trisomy 18p	1715	18	16
Trisomy 1q	261344	25	24
Trisomy 20p	261318	11	11
Trisomy 4p	1738	24	22
Trisomy 5p	1742	18	18
Trisomy 8p	264450	21	21
Trisomy 8q	1752	18	18
Trisomy 9p	236	61	58
Trisomy X	3375	537	530
Tritanopia	88629	<10	<10
Tropical endomyocardial fibrosis	75565	<10	<10
Tropical pancreatitis	103918	153	153
Tropical spastic paraparesis	289326	51	48
True unicornuate uterus	180074	<10	<10
Truncus arteriosus	3384	283	262
TSH-secreting pituitary adenoma	91347	173	169
Tuberculosis*	3389	243	243
Tuberous sclerosis complex	805	3029	2971
Tubular aggregate myopathy	2593	29	26
Tubulinopathy-associated dysgyria	467166	27	26
Tubulointerstitial nephritis and uveitis syndrome	91500	187	183
Tufted angioma	1063	44	44
Tularemia	3392	<10	<10
Tumor necrosis factor receptor 1 associated periodic syndrome	32960	136	135

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Turner syndrome	881	**4609**	**4565**
Twin anemia-polycythemia sequence	617294	**<10**	**<10**
Twin to twin transfusion syndrome	95431	**26**	**21**
Typical nemaline myopathy	171436	**31**	**26**
Tyrosinemia type 1	882	**157**	**151**
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**
Ulna metaphyseal dysplasia syndrome	1837	**<10**	**<10**
Ulnar hemimelia	93320	**38**	**38**
Ulnar-mammary syndrome	3138	**44**	**44**
Unclassified myelodysplastic syndrome	98827	**36**	**36**
Unclassified myelodysplastic/ myeloproliferative disease	98825	**<10**	**<10**
Unclassified vasculitis	251328	**513**	**487**
Uncombable hair syndrome	1410	**18**	**18**
Undetermined colitis	103920	**3266**	**3256**
Undifferentiated carcinoma of the corpus uteri	213721	**<10**	**<10**
Undifferentiated connective tissue syndrome	90002	**866**	**854**
Undifferentiated embryonal sarcoma of the liver	178315	**<10**	**<10**
Undifferentiated pleomorphic sarcoma	2023	**<10**	**<10**
Unexplained long-lasting fever/ inflammatory syndrome	251332	**179**	**176**
Unicervical bicornuate uterus	180114	**<10**	**<10**
Unilateral aplasia of the Müllerian ducts*	180071	**<10**	**<10**
Unilateral polymicrogyria	268943	**142**	**142**
Univentricular heart	1464	**680**	**645**
Unspecified juvenile idiopathic arthritis	91140	**749**	**746**
Unspecified mitochondrial disorder*	254837	**1713**	**1576**
Unstable hemoglobin disease	99139	**40**	**40**
Upington disease	3408	**<10**	**<10**
Upper limb hypertrophy	295049	**<10**	**<10**
Upper limb mesomelic dysplasia	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**
Usher syndrome	886	**1415**	**1403**
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	**49**	**47**
VACTERL/VATER association	887	**852**	**811**
Vacuolar myopathy with sarcoplasmic reticulum protein aggregates	88635	**<10**	**<10**
Vaginal atresia	65681	**32**	**32**
Van den Ende-Gupta syndrome	2460	**<10**	**<10**
Van der Woude syndrome	888	**290**	**289**
Variant ABeta2M amyloidosis	314652	**<10**	**<10**
Variant Creutzfeldt-Jakob disease	576370	**<10**	**<10**
Vascular Ehlers-Danlos syndrome	286	**1016**	**976**
Vasculitis due to ADA2 deficiency	404553	**34**	**34**
Vasoproliferative tumor of the retina	353356	**41**	**40**
Vein of Galen aneurysmal malformation	1053	**121**	**110**
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	**219**	**218**
Verrucous hemangioma	464318	**28**	**28**
Very long chain acyl-CoA dehydrogenase deficiency	26793	**94**	**83**
VEXAS syndrome	596753	**242**	**219**
Vibratory urticaria	493342	**<10**	**<10**
Vici syndrome	1493	**<10**	**<10**
VIPoma	97282	**<10**	**<10**
Viral myositis	206991	**114**	**113**
Visual snow syndrome	420556	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
Vitamin B12-responsive methylmalonic acidemia	28	**66**	**63**
Vitamin B12-unresponsive methylmalonic acidemia	27	**118**	**104**
Vitamin K antagonist embryofetopathy	1914	**<10**	**<10**
Vocal cord and pharyngeal distal myopathy	600	**<10**	**<10**
Vogt-Koyanagi-Harada disease	3437	**246**	**243**
Von Hippel-Lindau disease	892	**584**	**563**
Von Willebrand disease	903	**10211**	**10136**
Vulvar carcinoma	494418	**<10**	**<10**
Vulvar intraepithelial neoplasia	137583	**<10**	**<10**
Vulvovaginal rhabdomyosarcoma	206492	**<10**	**<10**
Waardenburg syndrome	3440	**578**	**575**
Waardenburg-Shah syndrome	897	**54**	**49**
WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	466943	**28**	**28**
Wagner disease	898	**26**	**26**
WAGR syndrome	893	**68**	**61**
Waldenström macroglobulinemia	33226	**163**	**128**
Walker-Warburg syndrome	899	**27**	**16**
WARS2-related combined oxidative phosphorylation defect	572798	**<10**	**<10**
Warts-immunodeficiency-lymphedema-anogenital dysplasia syndrome	568056	**<10**	**<10**
Weaver syndrome	3447	**35**	**35**
Weill-Marchesani syndrome	3449	**30**	**30**
Well-differentiated fetal adenocarcinoma of the lung	284395	**<10**	**<10**
Wells syndrome	901	**<10**	**<10**
Werner syndrome	902	**22**	**19**
WHIM syndrome	51636	**14**	**13**
Whipple disease	3452	**99**	**98**
White forelock with malformations	2475	**<10**	**<10**
White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome	3207	**<10**	**<10**
White sponge nevus	171723	**<10**	**<10**
White-Sutton syndrome	468678	**58**	**57**
Whooping cough	1489	**<10**	**<10**
Wiedemann-Rautenstrauch syndrome	3455	**<10**	**<10**
Wiedemann-Steiner syndrome	319182	**162**	**161**
Wild type ABeta2M amyloidosis	85446	**<10**	**<10**
Wild type ATTR amyloidosis	330001	**3102**	**2543**
Wildervanck syndrome	3456	**<10**	**<10**
Williams syndrome	904	**1192**	**1169**
Williams-Campbell syndrome	411501	**<10**	**<10**
Wilson disease	905	**1238**	**1202**
Wilson-Turner syndrome	3459	**<10**	**<10**
Wiskott-Aldrich syndrome	906	**206**	**203**
Witteveen-Kolk syndrome	500163	**41**	**41**
Wolcott-Rallison syndrome	1667	**<10**	**<10**
Wolf-Hirschhorn syndrome	280	**126**	**115**
Wolfram syndrome	3463	**297**	**278**
Wolfram-like syndrome	411590	**41**	**41**
Woodhouse-Sakati syndrome	3464	**32**	**32**
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**
Wyburn-Mason syndrome	53719	**<10**	**<10**
X small rings	96201	**<10**	**<10**
Xanthoma disseminatum	158003	**<10**	**<10**
Xeroderma pigmentosum	910	**126**	**119**
Xeroderma pigmentosum variant	90342	**11**	**11**
Xeroderma pigmentosum-Cockayne syndrome complex	220295	**<10**	**<10**
XK aprosencephaly syndrome	3469	**<10**	**<10**
X-linked acrogigantism	300373	**56**	**55**
X-linked adrenal hypoplasia congenita	95702	**46**	**45**
X-linked adrenoleukodystrophy	43	**591**	**546**
X-linked centronuclear myopathy	596	**79**	**48**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked Charcot-Marie-Tooth disease type 1	101075	**185**	**184**
X-linked Charcot-Marie-Tooth disease type 2	101076	**36**	**34**
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	**326**	**320**
X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome	431140	**<10**	**<10**
X-linked cone dysfunction syndrome with myopia	90001	**14**	**14**
X-linked corneal dermoid	1661	**<10**	**<10**
X-linked creatine transporter deficiency	52503	**105**	**105**
X-linked distal spinal muscular atrophy type 3	139557	**<10**	**<10**
X-linked dominant chondrodysplasia punctata	35173	**59**	**57**
X-linked dominant chondrodysplasia, Chassaing-Lacombe type	163966	**<10**	**<10**
X-linked dyserythropoietic anemia with abnormal platelets and neutropenia	363727	**<10**	**<10**
X-linked Ehlers-Danlos syndrome	75497	**12**	**12**
X-linked epilepsy-learning disabilities-behavior disorders syndrome	85294	**21**	**21**
X-linked erythropoietic protoporphyria	443197	**16**	**14**
X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	480880	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked hereditary sensory and autonomic neuropathy with deafness	139583	**<10**	**<10**
X-linked hypophosphatemia	89936	**590**	**588**
X-linked ichthyosis syndrome*	281210	**<10**	**<10**
X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia	317476	**<10**	**<10**
X-linked immunoneurologic disorder	2571	**<10**	**<10**
X-linked intellectual disability due to GRIA3 mutations	364028	**<10**	**<10**
X-linked intellectual disability, Cabezas type	85293	**<10**	**<10**
X-linked intellectual disability, Cantagrel type	85277	**<10**	**<10**
X-linked intellectual disability, Hedera type	93952	**<10**	**<10**
X-linked intellectual disability, Najm type	163937	**20**	**20**
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	**15**	**15**
X-linked intellectual disability, Van Esch type	163976	**<10**	**<10**
X-linked intellectual disability-acromegaly-hyperactivity syndrome	85327	**<10**	**<10**
X-linked intellectual disability-ataxia-apraxia syndrome	85338	**<10**	**<10**
X-linked intellectual disability-cerebellar hypoplasia syndrome	137831	**28**	**28**
X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome	459070	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	1568	**<10**	**<10**
X-linked intellectual disability-epilepsy syndrome*	2076	**33**	**32**
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	**23**	**23**
X-linked intellectual disability-macrocephaly-macroorchidism syndrome	85320	**<10**	**<10**
X-linked intellectual disability-plagiocephaly syndrome	2898	**<10**	**<10**
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**
X-linked lissencephaly with abnormal genitalia	452	**<10**	**<10**
X-linked lymphoproliferative disease due to SH2D1A deficiency	538931	**16**	**16**
X-linked lymphoproliferative disease due to XIAP deficiency	538934	**29**	**29**
X-linked lymphoproliferative disease*	2442	**20**	**19**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked mandibulofacial dysostosis	1131	**<10**	**<10**
X-linked mendelian susceptibility to mycobacterial diseases	319605	**13**	**12**
X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	435938	**<10**	**<10**
X-linked myopathy with excessive autophagy	25980	**58**	**53**
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	**<10**	**<10**
X-linked progressive cerebellar ataxia	1175	**<10**	**<10**
X-linked recessive ocular albinism	54	**146**	**145**
X-linked reticulate pigmentary disorder	85453	**<10**	**<10**
X-linked retinal dysplasia	1852	**<10**	**<10**
X-linked retinoschisis	792	**432**	**430**
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	**<10**	**<10**
X-linked sideroblastic anemia and spinocerebellar ataxia	2802	**<10**	**<10**
X-linked skeletal dysplasia-intellectual disability syndrome	1436	**<10**	**<10**
X-linked spasticity-intellectual disability-epilepsy syndrome	3175	**<10**	**<10**
X-linked spinocerebellar ataxia type 4	85292	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Number of living cases
X-linked spondyloepimetaphyseal dysplasia	93349	**<10**	**<10**
Xp21 deletion syndrome	261476	**13**	**13**
Xp22.13p22.2 duplication syndrome	284180	**16**	**16**
Xp22.3 microdeletion syndrome	1643	**37**	**36**
Xq12-q13.3 duplication syndrome	314389	**<10**	**<10**
Xq21 microdeletion syndrome	1435	**<10**	**<10**
Xq25 microduplication syndrome	521258	**<10**	**<10**
Xq27.3q28 duplication syndrome	261483	**15**	**15**
XY type gonadal dysgenesis-associated anomalies syndrome	1770	**24**	**24**
Yellow nail syndrome	662	**56**	**55**
Yolk sac tumor	876	**<10**	**<10**
Young adult-onset distal hereditary motor neuropathy	314485	**15**	**15**
Young syndrome	3471	**<10**	**<10**
Young-onset Parkinson disease	2828	**201**	**180**
Yunis-Varon syndrome	3472	**<10**	**<10**
Zellweger syndrome	912	**55**	**22**
Zellweger-like syndrome without peroxisomal anomalies	50812	**<10**	**<10**
Zika virus disease	448237	**<10**	**<10**
Zimmermann-Laband syndrome	3473	**<10**	**<10**
Zygomycosis	73263	**<10**	**<10**

BNDMR
Banque Nationale de Données
Maladies Rares

NUMBER OF CASES PER OBSOLETE RARE DISEASE REGISTERED IN THE BNDMR

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE : Acrodysostosis with multiple hormone resistance	280651	**<10**	**<10**
OBSOLETE : ACTH-independent Cushing syndrome*	99893	**132**	**126**
OBSOLETE : Arnold-Chiari malformation type II	1136	**133**	**130**
OBSOLETE : Borderline epithelial tumor of ovary	206473	**<10**	**<10**
OBSOLETE : Congenital bowing of long bones	2292	**12**	**12**
OBSOLETE : Cushing syndrome*	553	**669**	**636**
OBSOLETE : Early-onset schizophrenia	96369	**168**	**166**
OBSOLETE : Familial ovarian cancer*	213517	**<10**	**<10**
OBSOLETE : Hemochromatosis type 4	139491	**320**	**314**
OBSOLETE : Isolated optic nerve hypoplasia/aplasia	137902	**34**	**34**
OBSOLETE : Leptomyelolipoma	268838	**59**	**59**
OBSOLETE : Lipoma associated with neurospinal dysraphism*	268832	**179**	**179**
OBSOLETE : MYH7-related late-onset scapuloperoneal muscular dystrophy	437572	**34**	**34**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE : Myostatin-related muscle hypertrophy	275534	**<10**	**<10**
OBSOLETE : Polyradiculoneuropathy associated with IgG/IgA/IgM monoclonal gammopathy without known antibodies	208981	**147**	**115**
OBSOLETE : Primary pigmented nodular adrenocortical disease	189439	**19**	**19**
OBSOLETE : Rare variants of adenocarcinoma of the corpus uteri	213574	**<10**	**<10**
OBSOLETE : Spondyloepimetaphyseal dysplasia with joint laxity	93359	**<10**	**<10**
OBSOLETE : Tuberculous meningitis	499004	**15**	**14**
OBSOLETE : Vibratory angioedema	493348	**<10**	**<10**
OBSOLETE: 3-Phosphoglycerate dehydrogenase deficiency*	422519	**<10**	**<10**
OBSOLETE: Abnormal origin or aberrant course of coronary artery*	95493	**74**	**73**
OBSOLETE: Acheiria, bilateral	295103	**<10**	**<10**
OBSOLETE: Acheiria, unilateral	295101	**40**	**40**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Acquired alimentary behavior disorder of infancy*	138118	**770**	**759**
OBSOLETE: Acquired hemophilia	73274	**316**	**254**
OBSOLETE: Acquired metabolic neuropathy	206616	**311**	**271**
OBSOLETE: Acrocephalosyndactyly*	946	**<10**	**<10**
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	**71**	**71**
OBSOLETE: Adult pulmonary Langerhans cell histiocytosis	99874	**35**	**34**
OBSOLETE: Adult-onset SAPHO syndrome	324982	**<10**	**<10**
OBSOLETE: Alpha-1-antichymotrypsin deficiency	93594	**<10**	**<10**
OBSOLETE: Amelia of lower limb, bilateral	295059	**<10**	**<10**
OBSOLETE: Amelia of upper limb, bilateral	295055	**<10**	**<10**
OBSOLETE: Amelia of upper limb, unilateral	295053	**<10**	**<10**
OBSOLETE: Amniotic bands*	1034	**153**	**151**
OBSOLETE: Aneurysm or dilatation of ascending aorta	95484	**133**	**133**
OBSOLETE: Antenatal Bartter syndrome	93604	**33**	**33**
OBSOLETE: Aortopulmonary coronary arterial course	99086	**<10**	**<10**
OBSOLETE: Apodia, unilateral	295105	**<10**	**<10**
OBSOLETE: Argyrophilic grain disease	97342	**<10**	**<10**
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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Atrichia-mental 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**
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 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: AymÚ-Gripp syndrome	477668	**<10**	**<10**
OBSOLETE: Basal epidermolysis bullosa simplex*	158665	**34**	**33**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Basement membrane disease*	93550	**<10**	**<10**
OBSOLETE: Behr syndrome	1239	**<10**	**<10**
OBSOLETE: Benign essential blepharospasm	93955	**110**	**109**
OBSOLETE: Benign exophthalmos syndrome	71269	**<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: Bowed tibiae-radial anomalies-osteopenia-fractures syndrome	3331	**<10**	**<10**
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: Brachydactyly*	294937	**114**	**112**
OBSOLETE: Bullous systemic lupus erythematosus	46489	**13**	**12**
OBSOLETE: Central polydactyly of toes	295010	**<10**	**<10**
OBSOLETE: Centripetalis recessive dystrophic epidermolysis bullosa	89841	**58**	**43**
OBSOLETE: Cerebrorenodigital syndrome	1396	**<10**	**<10**
OBSOLETE: Cervical dystonia	93962	**104**	**102**
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**
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	**14**	**14**
OBSOLETE: Classic mast cell leukemia	158796	**<10**	**<10**
OBSOLETE: Classic paraneoplastic limbic encephalitis	163898	**42**	**36**
OBSOLETE: Combined hyperlipidemia*	79211	**<10**	**<10**
OBSOLETE: Common variable immunodeficiency due to an intrinsic B cell defect	77303	**<10**	**<10**
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: Common variable immunodeficiency without known genetic defect	231205	**<10**	**<10**
OBSOLETE: Complete atrioventricular canal-left heart obstruction syndrome	99066	**<10**	**<10**
OBSOLETE: Congenital absence of both forearm and hand, unilateral	295093	**108**	**108**
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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Congenital absence of upper arm and forearm with hand present, unilateral	295085	**<10**	**<10**
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 cataract, Volkmann type	98983	**<10**	**<10**
OBSOLETE: Congenital cataract-ichthyosis syndrome	1376	**<10**	**<10**
OBSOLETE: Congenital hydronephrosis	2190	**1545**	**1533**
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	**31**	**31**
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**
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	**68**	**58**
OBSOLETE: Craniosynostosis-cataract syndrome	1530	**<10**	**<10**
OBSOLETE: Craniosynostosis-fibular aplasia syndrome	1533	**<10**	**<10**
OBSOLETE: Deletion 20p	1611	**<10**	**<10**
OBSOLETE: Deletion 4q	1625	**<10**	**<10**
OBSOLETE: Developmental delay-deafness syndrome, Hildebrand type	163988	**<10**	**<10**
OBSOLETE: Distal monosomy 20q	96152	**<10**	**<10**
OBSOLETE: Duplication 4q	1739	**<10**	**<10**
OBSOLETE: Early infantile epileptic encephalopathy without suppression burst	369894	**12**	**11**
OBSOLETE: Ehlers-Danlos syndrome type 1	90309	**82**	**82**
OBSOLETE: Ehlers-Danlos syndrome type 2	90318	**48**	**48**
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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
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	**45**	**45**
OBSOLETE: Facial asymmetry-temporal seizures syndrome	1167	**<10**	**<10**
OBSOLETE: Familial articular chondrocalcinosis type 1	99781	**<10**	**<10**
OBSOLETE: Familial capillary hemangioma	91415	**26**	**26**
OBSOLETE: Familial chondromalacia patellae	1428	**<10**	**<10**
OBSOLETE: Familial esophageal achalasia	99723	**<10**	**<10**
OBSOLETE: Familial hyperreninemic hypoaldosteronism type 1	99763	**<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	**52**	**52**
OBSOLETE: Familial idiopathic steroid-resistant nephrotic syndrome with minimal changes	93216	**11**	**11**
OBSOLETE: Familial juvenile hyperuricemic nephropathy type 1	209886	**53**	**50**
OBSOLETE: Familial lambdoid synostosis	3267	**<10**	**<10**
OBSOLETE: Familial parathyroid adenoma	99877	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Familial primary hypomagnesemia with normocalciuria and normocalcemia	34527	**<10**	**<10**
OBSOLETE: Familial segmental neurofibromatosis	79428	**92**	**92**
OBSOLETE: Familial spinal neurofibromatosis	79429	**<10**	**<10**
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	**14**	**14**
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**
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-Schüller-Christian disease	99873	**<10**	**<10**
OBSOLETE: Hashimoto-Pritzker syndrome	99872	**<10**	**<10**
OBSOLETE: Hemochromatosis type 5	447792	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Hemolytic anemia due to glyceraldehyde-3-phosphate dehydrogenase deficiency	248305	**<10**	**<10**
OBSOLETE: Hereditary motor and sensory neuropathy*	140450	**439**	**423**
OBSOLETE: High isolated anorectal malformation	171201	**505**	**501**
OBSOLETE: Hirsutism-skeletal dysplasia-intellectual disability syndrome	2156	**<10**	**<10**
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**
OBSOLETE: Hypopituitarism-short stature-skeletal anomalies syndrome	2626	**<10**	**<10**
OBSOLETE: Idiopathic hypersomnia with long sleep time	228315	**156**	**156**
OBSOLETE: Idiopathic hypersomnia without long sleep time	228318	**102**	**101**
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	**185**	**175**
OBSOLETE: Idiopathic steroid-sensitive nephrotic syndrome with minimal change	93207	**361**	**356**
OBSOLETE: Infantile axonal neuropathy	2679	**40**	**39**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Infundibulopelvic stenosis-multicystic kidney syndrome	1849	**<10**	**<10**
OBSOLETE: Inherited predisposition to essential thrombocythemia	225968	**<10**	**<10**
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	**131**	**128**
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 oxycephaly	63440	**62**	**61**
OBSOLETE: Isolated plagiocephaly	35098	**703**	**701**
OBSOLETE: Ito hypomelanosis	435	**305**	**302**
OBSOLETE: Junctional epidermolysis bullosa, non-Herlitz type	89840	**<10**	**<10**
OBSOLETE: Juvenile chronic recurrent multifocal osteomyelitis	2778	**105**	**105**
OBSOLETE: Juvenile-onset SAPHO syndrome	324989	**<10**	**<10**
OBSOLETE: Laminopathy type Decaudain-Vigouroux	137871	**12**	**11**
OBSOLETE: Laryngeal dyskinesia	93961	**17**	**16**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Letterer-Siwe disease	99870	**<10**	**<10**
OBSOLETE: Limb dystonia	93957	**41**	**40**
OBSOLETE: Limbic encephalitis with caspr2 antibodies	276402	**13**	**12**
OBSOLETE: Limbic encephalitis with DPP6 antibodies	329341	**<10**	**<10**
OBSOLETE: Limbic encephalitis with LGI1 antibodies	163908	**86**	**85**
OBSOLETE: Limbic encephalitis*	163892	**185**	**175**
OBSOLETE: Lissencephaly-demyelinating axonal neuropathy syndrome	101356	**<10**	**<10**
OBSOLETE: Localized epiphyseal dysplasia	1823	**<10**	**<10**
OBSOLETE: Low isolated anorectal malformation	171215	**1132**	**1113**
OBSOLETE: Low-grade ependymoma	251633	**<10**	**<10**
OBSOLETE: Lumbosacral spina bifida aperta	268388	**24**	**23**
OBSOLETE: Lumbosacral spina bifida cystica	268758	**17**	**17**
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: 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	**11**	**<10**
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**	**12**
OBSOLETE: Multiple fibroadenoma of the breast	50920	**321**	**319**
OBSOLETE: Multiple ventricular septal defects	99096	**12**	**12**
OBSOLETE: Mycobacterium xenopi infection	314946	**<10**	**<10**
OBSOLETE: Myopathy-growth delay-intellectual disability-hypospadias syndrome	2601	**<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	**76**	**74**
OBSOLETE: Non-herpetic acute limbic encephalitis	163924	**45**	**43**
OBSOLETE: Non-secreting chemodectoma	101106	**<10**	**<10**
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	**306**	**305**
OBSOLETE: Oligoarticular juvenile idiopathic arthritis without anti-nuclear antibodies	247846	**82**	**82**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Ophthalmoplegia-myalgia-tubular aggregates syndrome	2742	**<10**	**<10**
OBSOLETE: Osteochondritis of tarsal/metatarsal bone	2054	**<10**	**<10**
OBSOLETE: Otopalatodigital syndrome	669	**12**	**11**
OBSOLETE: Papillary fibroelastoma of the heart	208600	**<10**	**<10**
OBSOLETE: Paraneoplastic limbic encephalitis*	163895	**16**	**13**
OBSOLETE: Patella aplasia/hypoplasia, unilateral	295038	**<10**	**<10**
OBSOLETE: Pediatric Castleman disease	93682	**<10**	**<10**
OBSOLETE: Pediatric polyarteritis nodosa	93564	**<10**	**<10**
OBSOLETE: Pediatric Sjögren syndrome	93566	**<10**	**<10**
OBSOLETE: Pediatric systemic sclerosis	93567	**<10**	**<10**
OBSOLETE: Peripheral hypothyroidism*	226310	**22**	**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: Pitt-Hopkins-like syndrome	221150	**<10**	**<10**
OBSOLETE: Platelet function disease associated with renal insufficiency	99146	**<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: 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**
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	**69**	**68**
OBSOLETE: Radial hemimelia, bilateral	295071	**<10**	**<10**
OBSOLETE: Radial hemimelia, unilateral	295069	**<10**	**<10**
OBSOLETE: Radiation-induced hypopituitarism	95622	**<10**	**<10**
OBSOLETE: Rapidly progressive glomerulonephritis	280569	**55**	**55**
OBSOLETE: Rare idiopathic male infertility	98345	**29**	**29**
OBSOLETE: Rare major hypertriglyceridemia*	181425	**11**	**11**
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	**18**	**16**
OBSOLETE: Retrocerebellar cyst	269200	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis with anti-nuclear antibodies	247854	**67**	**67**
OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis without anti-nuclear antibodies	247861	**37**	**37**
OBSOLETE: Sakati-Nyhan syndrome	3128	**<10**	**<10**
OBSOLETE: Secondary acute transverse myelitis	139420	**12**	**12**
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	**13**	**11**
OBSOLETE: Sino-auricular heart block	1260	**<10**	**<10**
OBSOLETE: SLC5A6-CDG	521268	**<10**	**<10**
OBSOLETE: Solitary median maxillary central incisor syndrome	2286	**23**	**22**
OBSOLETE: Spastic diplegia, infantile type	1680	**64**	**64**
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**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome	94095	**<10**	**<10**
OBSOLETE: Spondylocostal dysostosis-hypospadias-intellectual disability syndrome	329252	**<10**	**<10**
OBSOLETE: Sporadic achalasia	99722	**<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	**114**	**112**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with minimal changes	93221	**93**	**91**
OBSOLETE: Sporadic Leigh syndrome	255199	**<10**	**<10**
OBSOLETE: Sporadic pheochromocytoma	276624	**156**	**151**
OBSOLETE: Sporadic secreting paraganglioma	276627	**24**	**24**
OBSOLETE: Squamous cell carcinoma of head and neck	67037	**<10**	**<10**
OBSOLETE: Superior celosomia	93942	**<10**	**<10**
OBSOLETE: Suprabasal epidermolysis bullosa simplex*	158661	**<10**	**<10**
OBSOLETE: Symbrachydactyly of hand and foot, unilateral	295136	**12**	**12**
OBSOLETE: TACI-related selective deficiency of IgA	99974	**<10**	**<10**
OBSOLETE: Terminal limb defects*	294929	**<10**	**<10**
OBSOLETE: Terminal transverse defects of arm	93937	**<10**	**<10**
OBSOLETE: Thoracolumbosacral spina bifida aperta	268384	**<10**	**<10**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
OBSOLETE: Tibial hemimelia, bilateral	295079	**<10**	**<10**
OBSOLETE: Tibial hemimelia, unilateral	295077	**<10**	**<10**
OBSOLETE: Total spina bifida cystica	268377	**15**	**15**
OBSOLETE: Total spina bifida cystica	268748	**<10**	**<10**
OBSOLETE: Toxic or/and iatrogenic neuropathy	206619	**208**	**180**
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: 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: Unclassified spondylometaphyseal dysplasia	163678	**<10**	**<10**
OBSOLETE: Univentricular heart with single atrio-ventricular valve	99069	**16**	**15**
OBSOLETE: Unknown leukodystrophy	84096	**120**	**109**
OBSOLETE: Upper thoracic spina bifida aperta	268740	**<10**	**<10**
OBSOLETE: Upper thoracic spina bifida cystica	268770	**<10**	**<10**
OBSOLETE: Vascular disruption sequence	3160	**<10**	**<10**
OBSOLETE: Vitiligo-associated autoimmune disease	247871	**14**	**14**
OBSOLETE: Von Hippel anomaly	98941	**21**	**20**

Rare disease	ORPHA Code	Number of cases in the BNDMR	Living cases
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	**26**	**26**
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 Opitz G/BBB syndrome	306597	**<10**	**<10**
OBSOLETE: X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome	83648	**<10**	**<10**

BNDMR

Banque Nationale de Données
Maladies Rares