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 November 2023

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The French National Rare Disease Registry (Banque Nationale de Données Maladies Rares, BNDMR) is a national epidemiological and public health tool. It is funded within the framework of the 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.

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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.

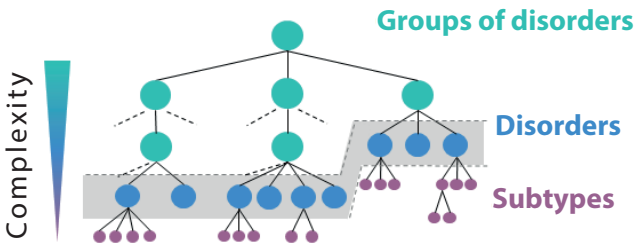

Figure 1 : Orphanet nomenclature classification levels representation

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 2021 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 01/11/2023, 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.

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.

In the absence of information on the "healthy carrier" status, patients are counted as sick and therefore are part of the displayed figures.

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.73)

The results are presented in a table containing three 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 time.

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	Cases in the BNDMR as of 01/11/2023
10q22.3q23.3 microdeletion syndrome	276413	**11**
10q22.3q23.3 microduplication syndrome	276422	**≤10**
11q22.2q22.3 microdeletion syndrome	444002	**≤10**
12q14 microdeletion syndrome	94063	**11**
12q15q21.1 microdeletion syndrome	289513	**≤10**
13q12.3 microdeletion syndrome	412035	**≤10**
14q11.2 microdeletion syndrome	261120	**≤10**
14q11.2 microduplication syndrome	261229	**≤10**
14q22q23 microdeletion syndrome	264200	**≤10**
14q24.1q24.3 microdeletion syndrome	401935	**≤10**
14q32 duplication syndrome	488280	**≤10**
15q overgrowth syndrome	314585	**132**
15q11.2 microdeletion syndrome	261183	**157**
15q11q13 microduplication syndrome	238446	**99**
15q13.3 microdeletion syndrome	199318	**107**
15q14 microdeletion syndrome	261190	**≤10**
16p11.2p12.2 microdeletion syndrome	261211	**47**
16p11.2p12.2 microduplication syndrome	261204	**39**
16p12.1p12.3 triplication syndrome	485405	**≤10**
16p13.11 microdeletion syndrome	261236	**44**
16p13.11 microduplication syndrome	261243	**49**
16p13.2 microdeletion syndrome	500055	**≤10**
16p13.3 microduplication syndrome	96078	**15**
16q24.1 microdeletion syndrome	352629	**≤10**
16q24.3 microdeletion syndrome	261250	**≤10**
17p11.2 microduplication syndrome	1713	**97**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
17p13.3 microduplication syndrome	217385	**28**
17q11.2 microduplication syndrome	139474	**14**
17q12 microdeletion syndrome	261265	**55**
17q12 microduplication syndrome	261272	**60**
17q21.31 microduplication syndrome	217340	**≤10**
17q23.1q23.2 microdeletion syndrome	261279	**≤10**
17q24.2 microdeletion syndrome	529962	**≤10**
19p13.12 microdeletion syndrome	254346	**≤10**
19p13.13 microdeletion syndrome	357001	**≤10**
19p13.3 microduplication syndrome	447980	**≤10**
19q13.11 microdeletion syndrome	217346	**≤10**
1p21.3 microdeletion syndrome	293948	**≤10**
1p31p32 microdeletion syndrome	401986	**≤10**
1p35.2 microdeletion syndrome	456298	**≤10**
1p36 deletion syndrome	1606	**216**
1q21.1 microdeletion syndrome	250989	**102**
1q21.1 microduplication syndrome	250994	**57**
1q41q42 microdeletion syndrome	250999	**≤10**
1q44 microdeletion syndrome	238769	**20**
20p12.3 microdeletion syndrome	261295	**≤10**
20p13 microdeletion syndrome	313781	**≤10**
20q11.2 microdeletion syndrome	444051	**≤10**
20q11.2 microduplication syndrome	363659	**≤10**
20q13.33 microdeletion syndrome	261311	**≤10**
21q22.11q22.12 microdeletion syndrome	261323	**23**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
22q11.2 deletion syndrome	567	**3133**
22q11.2 duplication syndrome	1727	**331**
2-aminoadipic 2-oxoadipic aciduria	79154	**≤10**
2-hydroxyglutaric aciduria*	19	**11**
2-methylbutyryl-CoA dehydrogenase deficiency	79157	**≤10**
2p13.2 microdeletion syndrome	363680	**≤10**
2p15p16.1 microdeletion syndrome	261349	**≤10**
2p21 microdeletion syndrome	163693	**≤10**
2q23.1 microdeletion syndrome	228402	**14**
2q23.1 microduplication syndrome	313947	**≤10**
2q24 microdeletion syndrome	1617	**22**
2q31.1 microdeletion syndrome	251014	**≤10**
2q32q33 microdeletion syndrome	251019	**≤10**
2q37 microdeletion syndrome	1001	**121**
3C syndrome	7	**≤10**
3-hydroxy-3-methylglutaric aciduria	20	**28**
3-hydroxy-3-methylglutaryl-CoA synthase deficiency	35701	**≤10**
3-hydroxyacyl-CoA dehydrogenase deficiency*	309127	**≤10**
3-hydroxyisobutyric aciduria	939	**≤10**
3M syndrome	2616	**72**
3MC syndrome	293843	**11**
3-methylcrotonyl-CoA carboxylase deficiency	6	**12**
3-methylglutaconic aciduria type 1	67046	**≤10**
3-methylglutaconic aciduria type 3	67047	**11**
3-methylglutaconic aciduria type 4	67048	**≤10**
3-methylglutaconic aciduria*	289902	**≤10**
3p25.3 microdeletion syndrome	435638	**≤10**
3q13 microdeletion syndrome	1621	**12**
3q26 microduplication syndrome	96095	**19**
3q26q27 microdeletion syndrome	356947	**≤10**
3q27.3 microdeletion syndrome	397695	**≤10**
3q29 microdeletion syndrome	65286	**41**
3q29 microduplication syndrome	251038	**17**
45,X/46,XY mixed gonadal dysgenesis	1772	**276**
46,XX disorder of sex development induced by endogenous maternal-derived androgen*	325093	**≤10**
46,XX disorder of sex development induced by exogenous maternal-derived androgen*	325099	**≤10**
46,XX disorder of sex development-anorectal anomalies syndrome	2973	**≤10**
46,XX gonadal dysgenesis	243	**78**
46,XX ovotesticular disorder of sex development	2138	**74**
46,XX testicular disorder of sex development	393	**61**
46,XY complete gonadal dysgenesis	242	**381**
46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	752	**27**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency	753	**56**
46,XY disorder of sex development due to isolated 17,20-lyase deficiency	90796	**≤10**
46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	168558	**≤10**
46,XY ovotesticular disorder of sex development	325345	**49**
46,XY partial gonadal dysgenesis	251510	**278**
47,XYY syndrome	8	**260**
48,XXXY syndrome	96263	**28**
48,XXYY syndrome	10	**61**
49,XXXXY syndrome	96264	**31**
49,XXXYY syndrome	261534	**≤10**
49,XYYYY syndrome	99330	**≤10**
4H leukodystrophy	289494	**63**
4p16.3 microduplication syndrome	96072	**≤10**
4q21 microdeletion syndrome	238750	**14**
4q25 proximal deletion syndrome	502437	**≤10**
5p13 microduplication syndrome	329802	**≤10**
5q14.3 microdeletion syndrome	228384	**12**
5q35 microduplication syndrome	228415	**≤10**
6p22 microdeletion syndrome	251046	**≤10**
6-phosphogluconate dehydrogenase deficiency	99135	**114**
6q terminal deletion syndrome	75857	**33**
6q16 microdeletion syndrome	171829	**17**
6q25 microdeletion syndrome	251056	**15**
7p22.1 microduplication syndrome	314034	**≤10**
7q11.23 microduplication syndrome	96121	**85**
7q31 microdeletion syndrome	251061	**12**
8p inverted duplication/deletion syndrome	96092	**13**
8p11.2 deletion syndrome	251066	**≤10**
8p23.1 duplication syndrome	251076	**22**
8p23.1 microdeletion syndrome	251071	**54**
8q12 microduplication syndrome	228399	**≤10**
8q21.11 microdeletion syndrome	284160	**≤10**
8q24.3 microdeletion syndrome	508488	**22**
9p13 microdeletion syndrome	324313	**≤10**
9q21.13 microdeletion syndrome	531151	**≤10**
9q31.1q31.3 microdeletion syndrome	401923	**≤10**
9q33.3q34.11 microdeletion syndrome	495818	**≤10**
AA amyloidosis	85445	**283**
AApoAIV amyloidosis	439232	**≤10**
Aarskog-Scott syndrome	915	**151**
Aase-Smith syndrome	916	**≤10**
ABeta2M amyloidosis*	439246	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Abetalipoproteinemia	14	**37**
Ablepharon macrostomia syndrome	920	**≤10**
Abnormal number of coronary ostia	99089	**≤10**
Abnormal origin of right or left pulmonary artery from the aorta	99050	**11**
Abnormal origin of the pulmonary artery*	1138	**≤10**
Abruzzo-Erickson syndrome	921	**≤10**
Absence deformity of leg-cataract syndrome	2310	**≤10**
Absence of fingerprints-congenital milia syndrome	1658	**≤10**
Absence of the pulmonary artery	980	**≤10**
Absence of uterine body	180142	**≤10**
Absent thumb-short stature-immunodeficiency syndrome	2951	**≤10**
Absent tibia-polydactyly-arachnoid cyst syndrome	3328	**≤10**
Acanthosis nigricans-insulin resistance-muscle cramps-acral enlargement syndrome	90301	**≤10**
Aceruloplasminemia	48818	**41**
Acetazolamide-responsive myotonia	99736	**≤10**
Achalasia-microcephaly syndrome	929	**13**
Acheiria	294983	**≤10**
Acheiropodia	931	**≤10**
Achondrogenesis	932	**≤10**
Achondroplasia	15	**717**
Achromatopsia	49382	**305**
Acinar cell carcinoma of pancreas	424046	**≤10**
Acquired aneurysmal subarachnoid hemorrhage	90065	**≤10**
Acquired angioedema with C1Inh deficiency	528663	**75**
Acquired angioedema*	91385	**56**
Acquired Creutzfeldt-Jakob disease*	454700	**≤10**
Acquired cutis laxa	228285	**≤10**
Acquired generalized lipodystrophy	79086	**64**
Acquired ichthyosis	454	**69**
Acquired idiopathic sideroblastic anemia	75564	**≤10**
Acquired methemoglobinemia	464453	**≤10**
Acquired monoclonal Ig light chain-associated Fanconi syndrome	91136	**24**
Acquired partial lipodystrophy	79087	**56**
Acquired prothrombin deficiency	26348	**≤10**
Acquired pseudoxanthoma elasticum	228247	**≤10**
Acquired purpura fulminans	49566	**33**
Acquired von Willebrand syndrome	99147	**319**
Acral peeling skin syndrome	263534	**19**
Acrocallosal syndrome	36	**18**
Acrocapitofemoral dysplasia	63446	**≤10**
Acrocardiofacial syndrome	2008	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Acrocraniofacial dysostosis	949	**≤10**
Acrodermatitis enteropathica	37	**26**
Acrodysostosis	950	**73**
Acrodysostosis with multiple hormone resistance	280651	**≤10**
Acrodysplasia scoliosis	2956	**≤10**
Acrofacial dysostosis*	364574	**≤10**
Acrofacial dysostosis, Kennedy-Teebi type	64542	**≤10**
Acrofacial dysostosis, Weyers type	952	**≤10**
Acrogeria	2500	**≤10**
Acrokeratoelastoidosis of Costa	38	**37**
Acrokeratosis verruciformis of Hopf	79151	**≤10**
Acromegaloid facial appearance syndrome	965	**≤10**
Acromegaly	963	**3103**
Acromelic dysplasia*	93436	**13**
Acromesomelic dysplasia*	93437	**≤10**
Acromesomelic dysplasia, Grebe type	2098	**≤10**
Acromesomelic dysplasia, Hunter-Thompson type	968	**≤10**
Acromesomelic dysplasia, Maroteaux type	40	**12**
Acromicric dysplasia	969	**38**
Acrootoocular syndrome	2980	**≤10**
Acropectoral syndrome	85203	**≤10**
Acropectorovertebral dysplasia	957	**≤10**
Acrorenal syndrome	971	**≤10**
Acro-renal-mandibular syndrome	958	**≤10**
Acro-renal-ocular syndrome	959	**≤10**
ACTH-dependent Cushing syndrome*	99892	**61**
ACTH-independent Cushing syndrome*	99893	**119**
Actinic lichen planus	254395	**≤10**
Actinic prurigo	330061	**≤10**
Actinomycosis	457095	**≤10**
Action myoclonus-renal failure syndrome	163696	**≤10**
Activated PI3K-delta syndrome	397596	**52**
Acute ackee fruit intoxication	73423	**≤10**
Acute adrenal insufficiency	95409	**150**
Acute annular outer retinopathy	284460	**≤10**
Acute bilirubin encephalopathy	529799	**≤10**
Acute disseminated encephalomyelitis	83597	**383**
Acute encephalopathy with biphasic seizures and late reduced diffusion	363549	**≤10**
Acute encephalopathy with inflammation-mediated status epilepticus*	363567	**≤10**
Acute erythroid leukemia	318	**≤10**
Acute fatty liver of pregnancy	243367	**≤10**
Acute generalized exanthematous pustulosis	293173	**259**
Acute hepatic porphyria*	95157	**11**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	217371	**≤10**
Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome	466794	**≤10**
Acute infantile liver failure-multisystemic involvement syndrome	370088	**≤10**
Acute inflammatory demyelinating polyradiculoneuropathy	98916	**516**
Acute intermittent porphyria	79276	**606**
Acute interstitial pneumonia	79126	**130**
Acute liver failure	90062	**33**
Acute lymphoblastic leukemia*	513	**87**
Acute macular neuroretinopathy	488239	**≤10**
Acute megakaryoblastic leukemia	518	**≤10**
Acute monoblastic/monocytic leukemia	514	**≤10**
Acute motor and sensory axonal neuropathy	98917	**726**
Acute motor axonal neuropathy	98918	**270**
Acute myeloblastic leukemia with maturation	98834	**≤10**
Acute myeloblastic leukemia without maturation	98833	**≤10**
Acute myeloid leukaemia with myelodysplasia-related features	86845	**≤10**
Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)	98829	**≤10**
Acute myeloid leukemia with minimal differentiation	98832	**≤10**
Acute myeloid leukemia*	519	**35**
Acute myelomonocytic leukemia	517	**23**
Acute necrotizing encephalopathy of childhood	263524	**≤10**
Acute pandysautonomia	231457	**≤10**
Acute panmyelosis with myelofibrosis	86843	**≤10**
Acute peripheral arterial occlusion	90064	**≤10**
Acute poisoning by drugs with membrane-stabilizing effect	43119	**≤10**
Acute promyelocytic leukemia	520	**≤10**
Acute pure sensory neuropathy	231450	**65**
Acute radiation syndrome	454831	**≤10**
Acute sensory ataxic neuropathy	231466	**75**
Acute transverse myelitis	139417	**322**
Acute tricyclic antidepressant poisoning	43117	**≤10**
Acute undifferentiated leukemia	98835	**≤10**
Acute zonal occult outer retinopathy	284454	**11**
Acyl-CoA dehydrogenase 9 deficiency	99901	**≤10**
Acyl-CoA dehydrogenase deficiency*	309120	**≤10**
Adamantinoma	55881	**≤10**
Adams-Oliver syndrome	974	**150**
Addison disease	85138	**1108**
Adducted thumbs-arthrogryposis syndrome, Christian type	2952	**12**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Adenine phosphoribosyltransferase deficiency	976	**58**
Adenocarcinoma of ovary	213504	**26**
Adenocarcinoma of the cervix uteri	213772	**≤10**
Adenocarcinoma of the esophagus	99976	**≤10**
Adenocarcinoma of the small intestine	104075	**≤10**
Adenohypophysitis	95512	**458**
Adenoma of pancreas	93292	**≤10**
Adenosarcoma of the corpus uteri	213600	**≤10**
Adenosine monophosphate deaminase deficiency	45	**37**
Adenylosuccinate lyase deficiency	46	**24**
Adenylosuccinate synthetase-like 1-related distal myopathy	482601	**≤10**
Adiposis dolorosa	36397	**18**
ADNP syndrome	404448	**50**
Adrenocortical carcinoma	1501	**307**
Adrenocortical carcinoma with pure aldosterone hypersecretion	231625	**≤10**
Adrenomyodystrophy	977	**≤10**
Adult acute respiratory distress syndrome	70578	**25**
Adult familial nephronophthisis-spastic quadriparesia syndrome	2666	**≤10**
Adult hepatocellular carcinoma	210159	**≤10**
Adult idiopathic neutropenia	2688	**304**
Adult neuronal ceroid lipofuscinosis	79262	**16**
ADULT syndrome	978	**26**
Adult T-cell leukemia/lymphoma	86875	**≤10**
Adult-onset autosomal dominant leukodystrophy	99027	**≤10**
Adult-onset autosomal recessive cerebellar ataxia	284289	**61**
Adult-onset autosomal recessive sideroblastic anemia	255132	**≤10**
Adult-onset cervical dystonia, DYT23 type	420492	**393**
Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy	329336	**61**
Adult-onset distal myopathy due to VCP mutation	329478	**46**
Adult-onset dystonia-parkinsonism	199351	**17**
Adult-onset foveomacular vitelliform dystrophy	99000	**81**
Adult-onset immunodeficiency with anti-interferon-gamma autoantibodies	306431	**163**
Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency	329314	**≤10**
Adult-onset nemaline myopathy	171442	**25**
Adult-onset Still disease	829	**649**
African iron overload	139507	**≤10**
African trypanosomiasis	3385	**≤10**
Agammaglobulinemia-microcephaly-craniosynostosis-severe dermatitis syndrome	83617	**≤10**
AGel amyloidosis	85448	**15**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Agenesis of the superior vena cava	99114	**≤10**
Aggressive primary cutaneous B-cell lymphoma*	178554	**≤10**
Aggressive systemic mastocytosis	98850	**60**
Agnathia-holoprosencephaly-situs inversus syndrome	990	**≤10**
AH amyloidosis	442582	**≤10**
AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	412069	**35**
Aicardi syndrome	50	**62**
Aicardi-Goutières syndrome	51	**202**
AICA-ribosiduria	250977	**≤10**
AL amyloidosis	85443	**2265**
Alacrimia-choreoathetosis-liver dysfunction syndrome	404454	**≤10**
Alagille syndrome	52	**494**
Åland Islands eye disease	178333	**≤10**
Alar cartilages hypoplasia-coloboma-telecanthus syndrome	2007	**≤10**
Alazami syndrome	319671	**13**
Albers-Schönberg osteopetrosis	53	**42**
Albinism-deafness syndrome	998	**≤10**
Alexander disease	58	**49**
ALG11-CDG	280071	**≤10**
ALG12-CDG	79324	**≤10**
ALG13-CDG	324422	**≤10**
ALG1-CDG	79327	**12**
ALG2-CDG	79326	**≤10**
ALG3-CDG	79321	**≤10**
ALG6-CDG	79320	**≤10**
ALG8-CDG	79325	**≤10**
ALG9-CDG	79328	**≤10**
Alkaptonuria	56	**106**
ALK-positive large B-cell lymphoma	364043	**≤10**
Allan-Herndon-Dudley syndrome	59	**53**
Allergic bronchopulmonary aspergillosis	1164	**294**
Alopecia antibody deficiency	1006	**≤10**
Alopecia totalis	700	**70**
Alopecia universalis	701	**110**
Alopecia-contractures-dwarfism-intellectual disability syndrome	1005	**≤10**
Alopecia-epilepsy-pyorrhea-intellectual disability syndrome	1008	**≤10**
Alopecia-intellectual disability syndrome	2850	**≤10**
Alopecia-intellectual disability-hypergonadotropic hypogonadism syndrome	1014	**≤10**
Alpers-Huttenlocher syndrome	726	**25**
Alpha delta granule deficiency	734	**113**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Alpha-1-antitrypsin deficiency	60	**1293**
Alpha-B crystallin-related late-onset myopathy	399058	**≤10**
Alpha-crystallinopathy*	98910	**≤10**
Alpha-dystroglycan-related limb-girdle muscular dystrophy R16	280333	**≤10**
Alpha-mannosidosis	61	**66**
Alpha-N-acetylgalactosaminidase deficiency	3137	**≤10**
Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3	62	**142**
Alpha-thalassemia	846	**405**
Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	98791	**14**
Alpha-thalassemia-myelodysplastic syndrome	231401	**≤10**
Alpha-thalassemia-X-linked intellectual disability syndrome	847	**49**
Alport syndrome	63	**2213**
Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	86818	**≤10**
Alström syndrome	64	**82**
Alternating hemiplegia of childhood	2131	**52**
Alternating hemiplegia*	209978	**69**
Alveolar echinococcosis	284	**13**
Alveolar soft tissue sarcoma	163699	**≤10**
Amelia of lower limb	294969	**≤10**
Amelia of upper limb	294967	**≤10**
Amelia*	294925	**≤10**
Ameloblastoma	314419	**≤10**
Amelocerebrohypohidrotic syndrome	1946	**11**
Amelogenesis imperfecta	88661	**727**
Amelo-onycho-hypohidrotic syndrome	1028	**≤10**
Aminoacylase deficiency*	308448	**≤10**
Aminopterin/methotrexate embryofetopathy	1908	**≤10**
Amoebic keratitis	67043	**13**
Amyotrophic lateral sclerosis	803	**12538**
Amyotrophic lateral sclerosis type 4	357043	**16**
Anal fistula	228113	**≤10**
Anaplastic astrocytoma	251589	**≤10**
Anaplastic ependymoma	251646	**≤10**
Anaplastic ganglioglioma	251957	**≤10**
Anaplastic large cell lymphoma	98841	**≤10**
Anaplastic oligoastrocytoma	251663	**≤10**
Anaplastic oligodendroglioma	251630	**≤10**
Anaplastic thyroid carcinoma	142	**≤10**
Anauxetic dysplasia	93347	**11**
Andersen-Tawil syndrome	37553	**85**
Androgen insensitivity syndrome*	754	**62**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Aneurysm of sinus of Valsalva	1054	**516**
Aneurysmal bone cyst	480553	**≤10**
Aneurysm-osteoarthritis syndrome	284984	**69**
Angelman syndrome	72	**758**
Angel-shaped phalango-epiphyseal dysplasia	63442	**≤10**
Angiocentric glioma	251671	**≤10**
Angioimmunoblastic T-cell lymphoma	86886	**≤10**
Angioma serpiginosum	95429	**12**
Angioosteohypertrophic syndrome	2346	**373**
Angioosteohypotrophic syndrome	75508	**≤10**
Angiosarcoma	263413	**≤10**
Angiostrongyliasis	74	**≤10**
Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome	69088	**≤10**
Aniridia-cerebellar ataxia-intellectual disability syndrome	1065	**25**
Aniridia-intellectual disability syndrome	1068	**≤10**
Aniridia-ptosis-intellectual disability-familial obesity syndrome	1067	**≤10**
Aniridia-renal agenesis-psychomotor retardation syndrome	1064	**≤10**
ANK3-related intellectual disability-sleep disturbance syndrome	356996	**≤10**
Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	1071	**61**
Ankylosing vertebral hyperostosis with tylosis	2206	**≤10**
Annular pancreas	675	**247**
Anoctamin-5-related limb-girdle muscular dystrophy R12	206549	**100**
Anodontia	99797	**≤10**
Anomalous aortic origin of the left coronary artery	541443	**25**
Anomalous aortic origin of the right coronary artery	541454	**30**
Anomalous origin of coronary artery from the pulmonary artery	541507	**74**
Anomaly of the mitral subvalvular apparatus	101932	**≤10**
Anophthalmia plus syndrome	1104	**≤10**
Anophthalmia/microphthalmia-esophageal atresia syndrome	77298	**≤10**
Anotia	93976	**39**
Antecubital pterygium syndrome	2987	**≤10**
Anterior cutaneous nerve entrapment syndrome	51890	**≤10**
Anterior segment developmental anomaly*	88632	**14**
Anterior urethral valve	435372	**18**
Anti-glomerular basement membrane disease	375	**120**
Anti-HLA hyperimmunization	2194	**≤10**
Anti-neutrophil cytoplasmic antibody-associated vasculitis*	156152	**808**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Anti-p200 pemphigoid	454710	**45**
Antisynthetase syndrome	81	**1081**
Antley-Bixler syndrome	83	**18**
Aorta coarctation	1457	**1133**
Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome	1110	**≤10**
Aortic arch interruption	2299	**137**
Aorto-ventricular tunnel	3400	**≤10**
Apert syndrome	87	**106**
Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome	1112	**≤10**
Aplasia cutis congenita	1114	**71**
Aplasia cutis congenita-intestinal lymphangiectasia syndrome	1116	**≤10**
Aplasia of lacrimal and salivary glands	86815	**≤10**
Apnea of prematurity	99981	**≤10**
Apodia	294986	**≤10**
Apolipoprotein A-I deficiency	425	**≤10**
Apparent mineralocorticoid excess	320	**≤10**
Aquagenic palmoplantar keratoderma	498359	**84**
Arachnodactyly-abnormal ossification-intellectual disability syndrome	1129	**≤10**
Arachnodactyly-intellectual disability-dysmorphism syndrome	1130	**≤10**
Arachnoid cyst	2356	**110**
Arachnoiditis	137817	**≤10**
Aregenerative anemia	101096	**32**
Argininemia	90	**33**
Argininosuccinic aciduria	23	**66**
Arnold-Chiari malformation type I	268882	**1175**
Arnold-Chiari malformation type II	1136	**129**
Aromatase deficiency	91	**≤10**
Aromatase excess syndrome	178345	**≤10**
Aromatic L-amino acid decarboxylase deficiency	35708	**15**
Arrhinia-choanal atresia-microphthalmia syndrome	1135	**≤10**
Arrhythmogenic right ventricular cardiomyopathy*	247	**822**
Arterial dissection-lentiginosis syndrome	1682	**190**
Arterial tortuosity syndrome	3342	**44**
Arthrochalasia Ehlers-Danlos syndrome	1899	**15**
Arthrogryposis multiplex congenita*	1037	**133**
Arthrogryposis syndrome*	109007	**565**
Arthrogryposis-hyperkeratosis syndrome, lethal form	1485	**≤10**
Arthrogryposis-like hand anomaly-sensorineural deafness syndrome	1144	**≤10**
Arthrogryposis-oculomotor limitation-electroretinal anomalies syndrome	1154	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Arthrogryposis-renal dysfunction-cholestasis syndrome	2697	**16**
Arthrogryposis-severe scoliosis syndrome	65720	**35**
ARX-related encephalopathy-brain malformation spectrum*	423655	**≤10**
ARX-related epileptic encephalopathy*	182079	**15**
Asbestos intoxication	2302	**225**
Ascher syndrome	1253	**≤10**
Asherman syndrome	137686	**≤10**
Aspartylglucosaminuria	93	**≤10**
Aspergillosis	1163	**167**
Astrocytoma*	94	**62**
Ataxia neuropathy spectrum*	254818	**268**
Ataxia with vitamin E deficiency	96	**56**
Ataxia-deafness-intellectual disability syndrome	1188	**≤10**
Ataxia-hypogonadism-choroidal dystrophy syndrome	1180	**≤10**
Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome	370022	**≤10**
Ataxia-oculomotor apraxia type 1	1168	**43**
Ataxia-oculomotor apraxia type 4	459033	**≤10**
Ataxia-pancytopenia syndrome	2585	**≤10**
Ataxia-tapetoretinal degeneration syndrome	1178	**≤10**
Ataxia-telangiectasia	100	**229**
Ataxia-telangiectasia-like disorder	251347	**≤10**
Atelosteogenesis type I	1190	**≤10**
Atelosteogenesis type III	56305	**≤10**
Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome	1192	**≤10**
Athyreosis	95713	**583**
Atopic keratoconjunctivitis	163934	**34**
Atresia of small intestine	1201	**213**
Atresia of urethra	105	**≤10**
Atrial septal aneurysm	99107	**≤10**
Atrial septal defect-atrioventricular conduction defects syndrome	1479	**≤10**
Atrial standstill	1344	**38**
Atrichia with papular lesions	86819	**≤10**
Atrioventricular defect-blepharophimosis-radial and anal defect syndrome	1352	**≤10**
Atrioventricular septal defect*	98722	**73**
Atrophic lichen planus	254449	**≤10**
Atrophoderma vermiculata	79100	**≤10**
Attenuated familial adenomatous polyposis	220460	**63**
ATTRV122I amyloidosis	85451	**247**
ATTRV30M amyloidosis	85447	**849**
Atypical autism	199627	**1850**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Atypical Fanconi syndrome-neonatal hyperinsulinism syndrome	544628	**≤10**
Atypical hemolytic uremic syndrome	2134	**643**
Atypical juvenile parkinsonism	391411	**≤10**
Atypical Norrie disease due to Xp11.3 microdeletion	261501	**≤10**
Atypical papilloma of choroid plexus	251902	**≤10**
Atypical Rett syndrome	3095	**252**
Atypical Werner syndrome	79474	**≤10**
Audiogenic seizures	166415	**≤10**
Auditory neuropathy-optic atrophy syndrome	542585	**18**
Auricular abnormalities-cleft lip with or without cleft palate-ocular abnormalities syndrome	77300	**35**
Auriculocondylar syndrome	137888	**15**
Autism spectrum disorder due to AUTS2 deficiency	352490	**31**
Autism spectrum disorder-epilepsy-arthrogryposis syndrome	370943	**≤10**
Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency	308410	**≤10**
Autism-facial port-wine stain syndrome	137911	**≤10**
Autoerythrocyte sensitization syndrome	324636	**≤10**
Autoimmune bullous skin disease*	79669	**38**
Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	420789	**≤10**
Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome	391487	**≤10**
Autoimmune hemolytic anemia*	98375	**868**
Autoimmune hemolytic anemia, cold type*	228312	**41**
Autoimmune hemolytic anemia, warm type	90033	**338**
Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome	444463	**28**
Autoimmune hepatitis	2137	**6330**
Autoimmune hypoparathyroidism	36913	**39**
Autoimmune interstitial lung disease-arthritis syndrome	444092	**112**
Autoimmune lymphoproliferative syndrome	3261	**111**
Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency	436159	**34**
Autoimmune lymphoproliferative syndrome with recurrent viral infections	275517	**≤10**
Autoimmune pancreatitis type 2	280315	**104**
Autoimmune pancreatitis*	103919	**42**
Autoimmune polyendocrinopathy type 1	3453	**82**
Autoimmune polyendocrinopathy type 2	3143	**56**
Autoimmune polyendocrinopathy type 3	227982	**≤10**
Autoimmune polyendocrinopathy*	282196	**≤10**
Autoimmune pulmonary alveolar proteinosis	747	**102**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Autoimmune thrombocytopenia*	71203	**130**
Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis	329173	**≤10**
Autosomal dominant adult-onset proximal spinal muscular atrophy	209335	**≤10**
Autosomal dominant aplasia and myelodysplasia	314399	**≤10**
Autosomal dominant brachyolmia	93304	**≤10**
Autosomal dominant centronuclear myopathy	169189	**38**
Autosomal dominant cerebellar ataxia type I*	94145	**26**
Autosomal dominant cerebellar ataxia type II*	208508	**37**
Autosomal dominant cerebellar ataxia type III*	94148	**89**
Autosomal dominant cerebellar ataxia type IV*	94149	**≤10**
Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	314404	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation	324611	**14**
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to TFG mutation	435819	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons	401964	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2*	64746	**900**
Autosomal dominant Charcot-Marie-Tooth disease type 2A1	99946	**104**
Autosomal dominant Charcot-Marie-Tooth disease type 2A2	99947	**92**
Autosomal dominant Charcot-Marie-Tooth disease type 2B	99936	**17**
Autosomal dominant Charcot-Marie-Tooth disease type 2C	99937	**27**
Autosomal dominant Charcot-Marie-Tooth disease type 2D	99938	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2DD	521414	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2E	99939	**23**
Autosomal dominant Charcot-Marie-Tooth disease type 2F	99940	**48**
Autosomal dominant Charcot-Marie-Tooth disease type 2G	99941	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2I	99942	**28**
Autosomal dominant Charcot-Marie-Tooth disease type 2J	99943	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2K	99944	**31**
Autosomal dominant Charcot-Marie-Tooth disease type 2L	99945	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2M	228179	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2N	228174	**32**
Autosomal dominant Charcot-Marie-Tooth disease type 2O	284232	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2Q	329258	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2U	397735	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2V	447964	**12**
Autosomal dominant Charcot-Marie-Tooth disease type 2Y	435387	**≤10**
Autosomal dominant Charcot-Marie-Tooth disease type 2Z	466768	**20**
Autosomal dominant childhood-onset proximal spinal muscular atrophy	363447	**33**
Autosomal dominant congenital benign spinal muscular atrophy	1216	**≤10**
Autosomal dominant cutis laxa	90348	**14**
Autosomal dominant deafness-onychodystrophy syndrome	79499	**≤10**
Autosomal dominant distal axonal motor neuropathy-myofibrillar myopathy syndrome	476093	**≤10**
Autosomal dominant dopa-responsive dystonia	98808	**44**
Autosomal dominant epidermolytic ichthyosis	312	**86**
Autosomal dominant epilepsy with auditory features	101046	**12**
Autosomal dominant focal dystonia, DYT25 type	329466	**22**
Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering	402003	**≤10**
Autosomal dominant generalized dystrophic epidermolysis bullosa	231568	**71**
Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form	79399	**≤10**
Autosomal dominant generalized epidermolysis bullosa simplex, severe form	79396	**39**
Autosomal dominant hyper-IgE syndrome	2314	**108**
Autosomal dominant hyperinsulinism due to Kir6.2 deficiency	276580	**≤10**
Autosomal dominant hyperinsulinism due to SUR1 deficiency	276575	**≤10**
Autosomal dominant hypophosphatemic rickets	89937	**24**
Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	457193	**18**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type A	100043	**91**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type B	100044	**11**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type C	100045	**≤10**

Number of cases per rare disease registered in the BNDMR as of 1st November 2023

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	100046	**≤10**
Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	93114	**≤10**
Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	324585	**≤10**
Autosomal dominant intermediate Charcot-Marie-Tooth disease*	90114	**36**
Autosomal dominant keratitis	2334	**≤10**
Autosomal dominant limb-girdle muscular dystrophy type 1A	266	**41**
Autosomal dominant macrothrombocytopenia	140957	**168**
Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	319581	**≤10**
Autosomal dominant mitochondrial myopathy with exercise intolerance	457050	**≤10**
Autosomal dominant multiple pterygium syndrome	65743	**≤10**
Autosomal dominant myoglobinuria	99846	**≤10**
Autosomal dominant neovascular inflammatory vitreoretinopathy	329211	**≤10**
Autosomal dominant nocturnal frontal lobe epilepsy	98784	**122**
Autosomal dominant optic atrophy and cataract	67036	**≤10**
Autosomal dominant optic atrophy and peripheral neuropathy	250932	**≤10**
Autosomal dominant optic atrophy plus syndrome	1215	**41**
Autosomal dominant optic atrophy*	98672	**413**
Autosomal dominant optic atrophy, classic form	98673	**361**
Autosomal dominant osteopetrosis type 1	2783	**24**
Autosomal dominant otospondylomegaepiphyseal dysplasia	166100	**≤10**
Autosomal dominant palmoplantar keratoderma and congenital alopecia	1010	**≤10**
Autosomal dominant polycystic kidney disease	730	**6574**
Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	88924	**66**
Autosomal dominant popliteal pterygium syndrome	1300	**11**
Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome	476119	**≤10**
Autosomal dominant primary hypomagnesemia with hypocalciuria	34528	**≤10**
Autosomal dominant prognathism	2964	**≤10**
Autosomal dominant progressive external ophthalmoplegia	254892	**26**
Autosomal dominant progressive nephropathy with hypertension	88659	**≤10**
Autosomal dominant proximal spinal muscular atrophy*	211037	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Autosomal dominant rhegmatogenous retinal detachment	209867	**≤10**
Autosomal dominant secondary polycythemia	247511	**≤10**
Autosomal dominant severe congenital neutropenia	486	**22**
Autosomal dominant spastic ataxia type 1	251282	**≤10**
Autosomal dominant spastic paraplegia type 10	100991	**36**
Autosomal dominant spastic paraplegia type 13	100994	**≤10**
Autosomal dominant spastic paraplegia type 17	100998	**≤10**
Autosomal dominant spastic paraplegia type 3	100984	**119**
Autosomal dominant spastic paraplegia type 31	101011	**26**
Autosomal dominant spastic paraplegia type 4	100985	**307**
Autosomal dominant spastic paraplegia type 41	320355	**≤10**
Autosomal dominant spastic paraplegia type 42	171863	**≤10**
Autosomal dominant spastic paraplegia type 6	100988	**≤10**
Autosomal dominant spastic paraplegia type 73	444099	**≤10**
Autosomal dominant spastic paraplegia type 8	100989	**16**
Autosomal dominant spastic paraplegia type 9A	447753	**≤10**
Autosomal dominant spastic paraplegia type 9B	447757	**≤10**
Autosomal dominant spondylocostal dysostosis	1797	**41**
Autosomal dominant thrombocytopenia with platelet secretion defect	466806	**≤10**
Autosomal dominant tubulointerstitial kidney disease	34149	**1006**
Autosomal dominant vitreoretinochoroidopathy	3086	**16**
Autosomal erythropoietic protoporphyria	79278	**271**
Autosomal recessive amelia	1027	**≤10**
Autosomal recessive anterior segment dysgenesis	519388	**16**
Autosomal recessive ataxia due to PEX10 deficiency	247815	**≤10**
Autosomal recessive ataxia due to ubiquinone deficiency	139485	**15**
Autosomal recessive ataxia, Beauce type	88644	**21**
Autosomal recessive axonal hereditary motor and sensory neuropathy*	91024	**227**
Autosomal recessive axonal neuropathy with neuromyotonia	324442	**≤10**
Autosomal recessive bestrophinopathy	139455	**27**
Autosomal recessive brachyolmia	448242	**≤10**
Autosomal recessive centronuclear myopathy	169186	**26**
Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	453521	**≤10**
Autosomal recessive cerebellar ataxia due to STUB1 deficiency	412057	**≤10**
Autosomal recessive cerebellar ataxia with late-onset spasticity	352641	**≤10**
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency	404499	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency	284282	**15**
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome*	404481	**≤10**
Autosomal recessive cerebellar ataxia-movement disorder syndrome	95434	**≤10**
Autosomal recessive cerebellar ataxia-psychomotor delay syndrome	284271	**≤10**
Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome	363429	**≤10**
Autosomal recessive cerebelloparenchymal disorder type 3	1170	**20**
Autosomal recessive cerebral atrophy	363969	**≤10**
Autosomal recessive Charcot-Marie-Tooth disease type 2X	466775	**≤10**
Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	101097	**11**
Autosomal recessive chorioretinopathy-microcephaly syndrome	2518	**≤10**
Autosomal recessive congenital ichthyosis*	281097	**123**
Autosomal recessive cutis laxa type 1	90349	**≤10**
Autosomal recessive cutis laxa type 2A	357058	**≤10**
Autosomal recessive cutis laxa type 2B	357064	**≤10**
Autosomal recessive dopa-responsive dystonia	101150	**30**
Autosomal recessive epidermolytic ichthyosis	512103	**≤10**
Autosomal recessive faciodigitogenital syndrome	1974	**18**
Autosomal recessive frontotemporal pachygyria	329329	**≤10**
Autosomal recessive generalized dystrophic epidermolysis bullosa, intermediate form	89842	**33**
Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	79408	**26**
Autosomal recessive generalized epidermolysis bullosa simplex	89838	**≤10**
Autosomal recessive hyperinsulinism due to Kir6.2 deficiency	79644	**≤10**
Autosomal recessive hyperinsulinism due to SUR1 deficiency	79643	**12**
Autosomal recessive hypophosphatemic rickets	289176	**17**
Autosomal recessive infantile hypercalcemia	300547	**146**
Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	217055	**≤10**
Autosomal recessive intermediate Charcot-Marie-Tooth disease type B	254334	**≤10**
Autosomal recessive intermediate Charcot-Marie-Tooth disease type C	369867	**≤10**
Autosomal recessive intermediate Charcot-Marie-Tooth disease*	268337	**144**
Autosomal recessive isolated optic atrophy	98676	**71**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Autosomal recessive lethal neonatal axonal sensorimotor polyneuropathy	538096	**≤10**
Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome	314572	**≤10**
Autosomal recessive lower motor neuron disease with childhood onset	206580	**≤10**
Autosomal recessive malignant osteopetrosis	667	**46**
Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency	319569	**≤10**
Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial JAK1 deficiency	574957	**≤10**
Autosomal recessive multiple pterygium syndrome	2990	**18**
Autosomal recessive myogenic arthrogryposis multiplex congenita	319332	**≤10**
Autosomal recessive optic atrophy, OPA7 type	227976	**≤10**
Autosomal recessive palmoplantar keratoderma and congenital alopecia	1366	**≤10**
Autosomal recessive polycystic kidney disease	731	**429**
Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity	437552	**≤10**
Autosomal recessive progressive external ophthalmoplegia	254886	**20**
Autosomal recessive secondary polycythemia not associated with VHL gene	247378	**≤10**
Autosomal recessive severe congenital neutropenia due to CSF3R deficiency	420702	**≤10**
Autosomal recessive severe congenital neutropenia due to CXCR2 deficiency	420699	**≤10**
Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	331176	**≤10**
Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency	423384	**≤10**
Autosomal recessive sideroblastic anemia	260305	**≤10**
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	98	**94**
Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome	254343	**≤10**
Autosomal recessive spastic paraplegia type 11	2822	**56**
Autosomal recessive spastic paraplegia type 14	100995	**≤10**
Autosomal recessive spastic paraplegia type 15	100996	**≤10**
Autosomal recessive spastic paraplegia type 18	209951	**≤10**
Autosomal recessive spastic paraplegia type 20	101000	**28**
Autosomal recessive spastic paraplegia type 21	101001	**≤10**
Autosomal recessive spastic paraplegia type 25	101005	**≤10**
Autosomal recessive spastic paraplegia type 26	101006	**≤10**
Autosomal recessive spastic paraplegia type 28	101008	**≤10**
Autosomal recessive spastic paraplegia type 35	171629	**11**
Autosomal recessive spastic paraplegia type 39	139480	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Autosomal recessive spastic paraplegia type 43	320370	**≤10**
Autosomal recessive spastic paraplegia type 44	320401	**≤10**
Autosomal recessive spastic paraplegia type 46	320391	**≤10**
Autosomal recessive spastic paraplegia type 48	306511	**≤10**
Autosomal recessive spastic paraplegia type 54	320380	**≤10**
Autosomal recessive spastic paraplegia type 56	320411	**≤10**
Autosomal recessive spastic paraplegia type 5A	100986	**21**
Autosomal recessive spastic paraplegia type 61	401780	**≤10**
Autosomal recessive spastic paraplegia type 62	401785	**≤10**
Autosomal recessive spastic paraplegia type 70	401835	**≤10**
Autosomal recessive spastic paraplegia type 76	488594	**≤10**
Autosomal recessive spastic paraplegia type 78	513436	**≤10**
Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome	95433	**≤10**
Autosomal recessive spondylocostal dysostosis	2311	**48**
Autosomal semi-dominant severe lipodystrophic laminopathy	280365	**12**
Autosomal spastic paraplegia type 30	101010	**33**
Autosomal spastic paraplegia type 58	397946	**≤10**
Autosomal spastic paraplegia type 72	401849	**≤10**
Autosomal systemic lupus erythematosus	300345	**55**
Axenfeld anomaly	98978	**47**
Axenfeld-Rieger syndrome	782	**144**
Axial mesodermal dysplasia spectrum	1834	**≤10**
Axial spondylometaphyseal dysplasia	168549	**≤10**
Axonal polyneuropathy associated with IgG/IgM/IgA monoclonal gammopathy	209004	**172**
Aymé-Gripp syndrome	1272	**≤10**
Azygos continuation of the inferior vena cava	99121	**≤10**
B4GALT1-CDG	79332	**≤10**
Babesiosis	108	**≤10**
Bacterial myositis	206994	**14**
Bacterial susceptibility due to TLR signaling pathway deficiency	183713	**≤10**
Bacterial toxic-shock syndrome	36234	**≤10**
Bainbridge-Ropers syndrome	352577	**35**
Balint syndrome	363746	**≤10**
Baller-Gerold syndrome	1225	**72**
Baló concentric sclerosis	228165	**≤10**
Bamforth-Lazarus syndrome	1226	**≤10**
Bangstad syndrome	1227	**≤10**
Bannayan-Riley-Ruvalcaba syndrome	109	**38**
BAP1-related tumor predisposition syndrome	289539	**20**
Baraitser-Winter cerebrofrontofacial syndrome	2995	**57**
Barber-Say syndrome	1231	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Bardet-Biedl syndrome	110	**615**
Baroreflex failure	443084	**≤10**
Barth syndrome	111	**50**
Bartsocas-Papas syndrome	1234	**≤10**
Bartter syndrome	112	**339**
Basel-Vanagaite-Smirin-Yosef syndrome	464738	**≤10**
Bathing suit ichthyosis	100976	**≤10**
Bazex-Dupré-Christol syndrome	113	**12**
B-cell chronic lymphocytic leukemia	67038	**23**
B-cell immunodeficiency-limb anomaly-urogenital malformation syndrome	567502	**≤10**
Becker muscular dystrophy	98895	**1220**
Becker nevus syndrome	64755	**32**
Beckwith-Wiedemann syndrome	116	**1361**
Beemer-Ertbruggen syndrome	1237	**≤10**
Behavioral variant of frontotemporal dementia	275864	**903**
Behçet disease	117	**2822**
Benign adult familial myoclonic epilepsy	86814	**21**
Benign cephalic histiocytosis	157997	**≤10**
Benign epithelial tumor of salivary glands	276148	**≤10**
Benign familial infantile epilepsy	306	**91**
Benign familial mesial temporal lobe epilepsy	163717	**34**
Benign familial neonatal epilepsy	1949	**115**
Benign familial neonatal-infantile seizures	140927	**49**
Benign focal seizures of adolescence	1544	**41**
Benign hereditary chorea	1429	**79**
Benign idiopathic neonatal seizures	64545	**37**
Benign infantile focal epilepsy with midline spikes and waves during sleep	166308	**63**
Benign infantile seizures associated with mild gastroenteritis	166305	**≤10**
Benign nocturnal alternating hemiplegia of childhood	209973	**≤10**
Benign non-familial infantile seizures*	166295	**≤10**
Benign occipital epilepsy	25968	**181**
Benign paroxysmal tonic upgaze of childhood with ataxia	1179	**≤10**
Benign paroxysmal torticollis of infancy	71518	**20**
Benign partial epilepsy of infancy with complex partial seizures	166299	**48**
Benign partial epilepsy with secondarily generalized seizures in infancy	166302	**24**
Benign partial infantile seizures*	166311	**27**
Benign recurrent intrahepatic cholestasis	65682	**92**
Benign Samaritan congenital myopathy	324581	**≤10**
Benign schwannoma	252164	**82**
Benign tumor of fallopian tubes	180237	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
BENTA disease	464336	**≤10**
Bernard-Soulier syndrome	274	**104**
Best vitelliform macular dystrophy	1243	**371**
Beta-ketothiolase deficiency	134	**58**
Beta-mannosidosis	118	**≤10**
Beta-propeller protein-associated neurodegeneration	329284	**32**
Beta-sarcoglycan-related limb-girdle muscular dystrophy R4	119	**39**
Beta-thalassemia	848	**1229**
Bethlem myopathy	610	**212**
Bickerstaff brainstem encephalitis	79138	**30**
Bicornuate uterus*	180134	**47**
Bietti crystalline dystrophy	41751	**21**
Bifid nose	2695	**≤10**
Bifid uvula	99771	**143**
Bifunctional enzyme deficiency	300	**11**
Bilateral microtia-deafness-cleft palate syndrome	140963	**≤10**
Bilateral polymicrogyria	268940	**191**
Bilateral striopallidodentate calcinosis	1980	**173**
Biliary atresia with splenic malformation syndrome	244283	**22**
Bilirubin encephalopathy*	415286	**13**
Biotinidase deficiency	79241	**44**
Biotin-thiamine-responsive basal ganglia disease	65284	**14**
Birdshot chorioretinopathy	179	**831**
Birt-Hogg-Dubé syndrome	122	**487**
Björnstad syndrome	123	**≤10**
Blackfan-Diamond anemia	124	**204**
Blake pouch cyst	98922	**32**
Blau syndrome	90340	**70**
Bleeding diathesis due to a collagen receptor defect	73271	**12**
Bleeding diathesis due to thromboxane synthesis deficiency	220443	**18**
Bleeding disorder due to CalDAG-GEFI deficiency	420566	**≤10**
Bleeding disorder due to P2Y12 defect	36355	**33**
Blepharo-cheilo-odontic syndrome	1997	**25**
Blepharophimosis-intellectual disability syndrome*	293642	**≤10**
Blepharophimosis-intellectual disability syndrome, MKB type	293707	**≤10**
Blepharophimosis-intellectual disability syndrome, Ohdo type	2728	**38**
Blepharophimosis-intellectual disability syndrome, SBBYS type	3047	**17**
Blepharophimosis-ptosis-epicanthus inversus syndrome	126	**211**
Blepharophimosis-ptosis-epicanthus inversus syndrome plus	572333	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome	2057	**≤10**
Blepharoptosis-myopia-ectopia lentis syndrome	1259	**≤10**
Blepharospasm-oromandibular dystonia syndrome	93964	**162**
Bloom syndrome	125	**17**
Blount disease	2768	**26**
Blue cone monochromatism	16	**85**
Blue diaper syndrome	94086	**≤10**
Blue rubber bleb nevus	1059	**83**
Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	91135	**≤10**
Bohring-Opitz syndrome	97297	**24**
Bone sarcoma*	223727	**≤10**
BOR syndrome	107	**484**
Borderline epithelial tumor of ovary	206473	**≤10**
Borjeson-Forssman-Lehmann syndrome	127	**28**
Bosley-Salih-Alorainy syndrome	69737	**≤10**
Bothnia retinal dystrophy	85128	**≤10**
Botulism	1267	**≤10**
Bowen-Conradi syndrome	1270	**≤10**
Brachydactyly type A1	93388	**40**
Brachydactyly type A2	93396	**22**
Brachydactyly type A4	93394	**15**
Brachydactyly type B	93383	**40**
Brachydactyly type C	93384	**91**
Brachydactyly type E	93387	**58**
Brachydactyly-arterial hypertension syndrome	1276	**18**
Brachydactyly-elbow wrist dysplasia syndrome	1275	**≤10**
Brachydactyly-mesomelia-intellectual disability-heart defects syndrome	1277	**≤10**
Brachydactyly-nystagmus-cerebellar ataxia syndrome	1246	**≤10**
Brachydactyly-preaxial hallux varus syndrome	1278	**≤10**
Brachydactyly-short stature-retinitis pigmentosa syndrome	166035	**≤10**
Brachydactyly-syndactyly, Zhao type	93409	**≤10**
Brachymorphism-onychodysplasia-dysphalangism syndrome	1292	**≤10**
Brachyolmia*	1293	**28**
Brachyolmia, Maroteaux type	93302	**≤10**
Brachyolmia-amelogenesis imperfecta syndrome	2899	**≤10**
Brachytelephalangic chondrodysplasia punctata	79345	**22**
Brachytelephalangy-dysmorphism-Kallmann syndrome	1295	**≤10**
Braddock syndrome	52047	**≤10**
Bradyopsia	75374	**≤10**
Brain calcification, Rajab type	178506	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Brain demyelination due to methionine adenosyltransferase deficiency	168598	**≤10**
Brain malformation-congenital heart disease-postaxial polydactyly syndrome	75389	**≤10**
Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	500150	**18**
Brain-lung-thyroid syndrome	209905	**29**
Branchiogenic deafness syndrome	50815	**22**
Branchio-oculo-facial syndrome	1297	**64**
Branchiootic syndrome	52429	**19**
Branchioskeletogenital syndrome	1299	**≤10**
BRESEK syndrome	85284	**≤10**
Brittle cornea syndrome	90354	**≤10**
Brody myopathy	53347	**19**
Bronchial neuroendocrine tumor	97287	**42**
Bronchiolitis obliterans with obstructive pulmonary disease	1303	**574**
Bronchogenic cyst	2357	**72**
Bronchopulmonary dysplasia	70589	**1547**
Brooke-Spiegler syndrome	79493	**84**
Brucellosis	1304	**≤10**
Bruck syndrome	2771	**23**
Brugada syndrome	130	**2749**
Budd-Chiari syndrome	131	**684**
Buerger disease	36258	**403**
Bulbospinal muscular atrophy of adult*	206707	**75**
Bulbospinal muscular atrophy of childhood*	206704	**≤10**
Bullous impetigo	36237	**≤10**
Bullous lichen planus	33408	**≤10**
Bullous pemphigoid	703	**3046**
Burkitt lymphoma	543	**18**
Burning mouth syndrome	353253	**≤10**
Burn-McKeown syndrome	1200	**≤10**
Buschke-Ollendorff syndrome	1306	**46**
Butterfly-shaped pigment dystrophy	99001	**≤10**
Butyrylcholinesterase deficiency	132	**23**
BVES-related limb-girdle muscular dystrophy	476084	**≤10**
C syndrome	1308	**11**
C11ORF73-related autosomal recessive hypomyelinating leukodystrophy	495844	**≤10**
CACH syndrome	135	**94**
CAD-CDG	448010	**≤10**
CADDS	369942	**≤10**
Caffey disease	1310	**26**
Calciphylaxis	280062	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Calpain-3-related limb-girdle muscular dystrophy D4	565909	**≤10**
Calpain-3-related limb-girdle muscular dystrophy R1	267	**472**
CAMOS syndrome	83472	**≤10**
Campomelia, Cumming type	1318	**≤10**
Campomelic dysplasia	140	**23**
Campomelic dysplasia and related disorders*	93439	**≤10**
Camptobrachydactyly	1319	**14**
Camptodactyly of fingers	295016	**67**
Camptodactyly syndrome, Guadalajara type 1	1327	**≤10**
Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	2848	**14**
Camptodactyly-fibrous tissue hyperplasia-skeletal anomalies syndrome	1321	**≤10**
Camptodactyly-tall stature-scoliosis-hearing loss syndrome	85164	**≤10**
Camptodactyly-taurinuria syndrome	1325	**≤10**
Camurati-Engelmann disease	1328	**42**
Canavan disease	141	**29**
Cancer-associated retinopathy	71505	**19**
CANOMAD syndrome	71279	**101**
Cantú syndrome	1517	**19**
Cap myopathy	171881	**19**
Cap polyposis	160148	**≤10**
Capillary malformation-arteriovenous malformation	137667	**786**
Carbamoyl-phosphate synthetase 1 deficiency	147	**51**
Carcinoid syndrome	100093	**≤10**
Cardiac anomalies-heterotaxy syndrome	137628	**79**
Cardiac diverticulum	1686	**≤10**
Cardiac-valvular Ehlers-Danlos syndrome	230851	**≤10**
Cardiocranial syndrome, Pfeiffer type	2872	**14**
Cardiofaciocutaneous syndrome	1340	**252**
Cardiogenic shock	97292	**≤10**
Cardiomyopathy-cataract-hip spine disease syndrome	1345	**≤10**
Cardiomyopathy-hypotonia-lactic acidosis syndrome	91130	**≤10**
Cardiospondylocarpofacial syndrome	3238	**≤10**
Carey-Fineman-Ziter syndrome	1358	**16**
Caribbean parkinsonism	97355	**≤10**
Carney complex	1359	**154**
Carney complex-trismus-pseudocamptodactyly syndrome	319340	**≤10**
Carney triad	139411	**≤10**
Carney-Stratakis syndrome	97286	**≤10**
Carnitine palmitoyl transferase 1A deficiency	156	**14**
Carnitine palmitoyltransferase II deficiency	157	**147**
Carnitine-acylcarnitine translocase deficiency	159	**16**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Carnosinase deficiency	1361	**≤10**
Caroli disease	53035	**150**
Caroli syndrome	480520	**20**
Carpenter syndrome	65759	**≤10**
Carpotarsal osteochondromatosis	2767	**≤10**
Cartilage-hair hypoplasia	175	**50**
Carvajal syndrome	65282	**≤10**
Castleman disease	160	**346**
Cataract-congenital heart disease-neural tube defect syndrome	314993	**≤10**
Cataract-glaucoma syndrome	162	**49**
Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	436174	**≤10**
Cataract-intellectual disability-hypogonadism syndrome	1387	**≤10**
Cataract-microcornea syndrome	1377	**26**
Cataract-nephropathy-encephalopathy syndrome	1380	**≤10**
Catastrophic antiphospholipid syndrome	464343	**185**
Catecholaminergic polymorphic ventricular tachycardia	3286	**338**
Catel-Manzke syndrome	1388	**≤10**
Cat-eye syndrome	195	**82**
Cathepsin A-related arteriopathy-strokes-leukoencephalopathy	575553	**≤10**
Cat-scratch disease	50839	**≤10**
Caudal duplication	1756	**≤10**
Caudal regression syndrome	3027	**97**
Caudal regression-sirenomelia spectrum*	444941	**≤10**
CCDC115-CDG	468684	**≤10**
CDKL5-deficiency disorder	505652	**54**
CEBPE-associated autoinflammation-immunodeficiency-neutrophil dysfunction syndrome	566067	**≤10**
Celiac artery compression syndrome	293208	**≤10**
Celiac disease-epilepsy-cerebral calcification syndrome	1459	**≤10**
CELSR1-related late-onset primary lymphedema	569816	**≤10**
Cenani-Lenz syndrome	3258	**≤10**
Central areolar choroidal dystrophy	75377	**22**
Central cloudy dystrophy of François	98972	**≤10**
Central congenital hypothyroidism*	226298	**64**
Central core disease	597	**352**
Central diabetes insipidus	178029	**1033**
Central nervous system calcification-deafness-tubular acidosis-anemia syndrome	3240	**≤10**
Central nervous system embryonal tumor*	251870	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Central neurocytoma	73256	**≤10**
Central polydactyly	295004	**≤10**
Central precocious puberty	759	**1956**
Central retinal vein occlusion	411527	**90**
Central serous chorioretinopathy	443079	**16**
Centronuclear myopathy*	595	**134**
Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome	504476	**503**
Cerebellar ataxia, Cayman type	94122	**≤10**
Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome	1171	**29**
Cerebellar ataxia-ectodermal dysplasia syndrome	1174	**≤10**
Cerebellar ataxia-hypogonadism syndrome	1173	**≤10**
Cerebellar hypoplasia-tapetoretinal degeneration syndrome	2246	**≤10**
Cerebellar-facial-dental syndrome	444072	**≤10**
Cerebral arteriovenous malformation	46724	**780**
Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	136	**580**
Cerebral autosomal recessive arteriopathy-subcortical infarcts-leukoencephalopathy	199354	**≤10**
Cerebral cortical dysplasia*	268950	**312**
Cerebral sinovenous thrombosis	329217	**437**
Cerebral visual impairment	447788	**≤10**
Cerebrocostomandibular syndrome	1393	**13**
Cerebrofacioarticular syndrome	314679	**≤10**
Cerebrofaciothoracic dysplasia	1394	**≤10**
Cerebrooculonasal syndrome	66625	**≤10**
Cerebrotendinous xanthomatosis	909	**61**
Cernunnos-XLF deficiency	169079	**≤10**
Cervical aortic arch	99079	**≤10**
Cervical dermoid cyst	141046	**138**
Cervical hypertrichosis-peripheral neuropathy syndrome	2218	**≤10**
Cervicofacial fibrochondroma	141067	**454**
Char syndrome	46627	**19**
Charcot-Marie-Tooth disease type 1*	65753	**1120**
Charcot-Marie-Tooth disease type 1A	101081	**2969**
Charcot-Marie-Tooth disease type 1B	101082	**230**
Charcot-Marie-Tooth disease type 1C	101083	**54**
Charcot-Marie-Tooth disease type 1D	101084	**≤10**
Charcot-Marie-Tooth disease type 1E	90658	**17**
Charcot-Marie-Tooth disease type 1F	101085	**12**
Charcot-Marie-Tooth disease type 2B1	98856	**22**
Charcot-Marie-Tooth disease type 2B2	101101	**21**
Charcot-Marie-Tooth disease type 2B5	228374	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Charcot-Marie-Tooth disease type 2H	101102	**19**
Charcot-Marie-Tooth disease type 2P	300319	**13**
Charcot-Marie-Tooth disease type 2R	397968	**≤10**
Charcot-Marie-Tooth disease type 2S	443073	**15**
Charcot-Marie-Tooth disease type 2T	495274	**≤10**
Charcot-Marie-Tooth disease type 4*	64749	**98**
Charcot-Marie-Tooth disease type 4A	99948	**29**
Charcot-Marie-Tooth disease type 4B1	99955	**≤10**
Charcot-Marie-Tooth disease type 4B2	99956	**≤10**
Charcot-Marie-Tooth disease type 4C	99949	**89**
Charcot-Marie-Tooth disease type 4D	99950	**18**
Charcot-Marie-Tooth disease type 4F	99952	**44**
Charcot-Marie-Tooth disease type 4G	99953	**24**
Charcot-Marie-Tooth disease type 4H	99954	**≤10**
Charcot-Marie-Tooth disease type 4J	139515	**15**
Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome	90103	**≤10**
CHARGE syndrome	138	**672**
CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	599082	**22**
Chédiak-Higashi syndrome	167	**24**
Cheilitis glandularis	1221	**≤10**
Cheirospondyloenchondromatosis	99647	**≤10**
Cherubism	184	**40**
Chilblain lupus	90280	**21**
CHILD syndrome	139	**≤10**
Childhood absence epilepsy	64280	**646**
Childhood-onset autosomal recessive myopathy with external ophthalmoplegia	363677	**≤10**
Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia	284324	**≤10**
Childhood-onset benign chorea with striatal involvement	494541	**≤10**
Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	500180	**≤10**
Childhood-onset nemaline myopathy	171439	**21**
Childhood-onset progressive contractures-limb-girdle weakness-muscle dystrophy syndrome	466921	**≤10**
Childhood-onset spasticity with hyperglycinemia	401866	**≤10**
CHIME syndrome	3474	**≤10**
Choanal atresia	137914	**265**
Choanal atresia-athelia-hypothyroidism-delayed puberty-short stature syndrome	589856	**≤10**
Cholangiocarcinoma	70567	**≤10**
Choledochal cyst	480501	**188**
Cholestasis-lymphedema syndrome	1414	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Cholestasis-pigmentary retinopathy-cleft palate syndrome	1415	**≤10**
Chondrodysplasia punctata*	93442	**57**
Chondrodysplasia with joint dislocations, gPAPP type	280586	**≤10**
Chondrodysplasia-disorder of sex development syndrome	1422	**≤10**
Chondromyxoid fibroma	404507	**≤10**
Chondrosarcoma	55880	**≤10**
Chordoid glioma	251674	**≤10**
Chordoma	178	**16**
Choreoacanthocytosis	2388	**13**
Choriocarcinoma of the central nervous system	252015	**≤10**
Choroid plexus carcinoma	251899	**≤10**
Choroid plexus tumor*	251896	**≤10**
Choroideremia	180	**199**
Christianson syndrome	85278	**28**
Chromophobe renal cell carcinoma	319303	**31**
Chronic acquired demyelinating polyneuropathy*	208974	**313**
Chronic actinic dermatitis	330064	**≤10**
Chronic beryllium disease	133	**15**
Chronic bilirubin encephalopathy	529808	**≤10**
Chronic cutaneous lupus erythematosus*	163531	**209**
Chronic diarrhea due to glucoamylase deficiency	103907	**≤10**
Chronic diarrhea with villous atrophy	1670	**≤10**
Chronic enteropathy associated with SLCO2A1 gene	468641	**≤10**
Chronic eosinophilic leukemia	168940	**≤10**
Chronic Epstein-Barr virus infection syndrome	2566	**45**
Chronic granulomatous disease	379	**265**
Chronic inflammatory demyelinating polyneuropathy	2932	**3692**
Chronic intestinal failure	294422	**160**
Chronic intestinal pseudoobstruction	2978	**402**
Chronic lymphoproliferative disorder of natural killer cells	512017	**11**
Chronic mucocutaneous candidiasis	1334	**25**
Chronic myeloid leukemia	521	**55**
Chronic myelomonocytic leukemia	98823	**≤10**
Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	324964	**691**
Chronic pneumonitis of infancy	91359	**104**
Chronic polyradiculoneuropathy*	208978	**1197**
Chronic relapsing inflammatory optic neuropathy	499085	**49**
Chronic respiratory distress with surfactant metabolism deficiency	217566	**≤10**
Chronic thromboembolic pulmonary hypertension	70591	**2419**
Chronic visceral acid sphingomyelinase deficiency	77293	**145**
Chudley-McCullough syndrome	314597	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Chuvash erythrocytosis	238557	**≤10**
Chylomicron retention disease	71	**24**
Chylous ascites	1160	**17**
CIDEC-related familial partial lipodystrophy	435651	**≤10**
CINCA syndrome	1451	**42**
Cirrhotic cardiomyopathy	57777	**≤10**
Citrullinemia type I	247525	**79**
Citrullinemia type II	247585	**≤10**
CLAPO syndrome	168984	**≤10**
Clark-Baraitser syndrome	600731	**≤10**
Class I glucose-6-phosphate dehydrogenase deficiency	466026	**410**
Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	90794	**2083**
Classic galactosemia	79239	**269**
Classic glucose transporter type 1 deficiency syndrome	71277	**215**
Classic hairy cell leukemia	58017	**≤10**
Classic Hodgkin lymphoma	391	**44**
Classic homocystinuria	394	**206**
Classic lissencephaly*	102009	**65**
Classic mycosis fungoides	2584	**15**
Classical Ehlers-Danlos syndrome	287	**464**
Classical-like Ehlers-Danlos syndrome type 1	230839	**31**
CLCN4-related X-linked intellectual disability syndrome	485350	**36**
Clear cell renal carcinoma	319276	**186**
Clear cell sarcoma of kidney	457246	**≤10**
Cleft hard palate	101023	**540**
Cleft lip and alveolus	141291	**870**
Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-hearing loss syndrome	508476	**≤10**
Cleft lip with or without cleft palate*	1991	**2415**
Cleft lip/palate	199306	**2322**
Cleft lip/palate-deafness-sacral lipoma syndrome	2003	**11**
Cleft lip/palate-ectodermal dysplasia syndrome	3253	**≤10**
Cleft lip/palate-intestinal malrotation-cardiopathy syndrome	2001	**≤10**
Cleft lip-retinopathy syndrome	1995	**≤10**
Cleft mitral valve	95465	**48**
Cleft palate*	2014	**4214**
Cleft palate-large ears-small head syndrome	2013	**≤10**
Cleft palate-lateral synechia syndrome	2016	**≤10**
Cleft palate-stapes fixation-oligodontia syndrome	2010	**≤10**
Cleft velum	99772	**1239**
Cleidocranial dysplasia	1452	**305**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Climatic droplet keratopathy	98958	**≤10**
CLIPPERS	284448	**≤10**
CLOVES syndrome	140944	**271**
CNTNAP2-related developmental and epileptic encephalopathy	163681	**1591**
COASY protein-associated neurodegeneration	397725	**≤10**
Coats disease	190	**270**
Coats plus syndrome	313838	**14**
Cobblestone lissencephaly without muscular or ocular involvement	352682	**≤10**
Cobblestone lissencephaly*	51577	**≤10**
Cocaine embryofetopathy	1911	**≤10**
Cocaine intoxication	90068	**≤10**
Cochlear nerve deficiency	502318	**18**
Cochleovestibular malformation	502305	**11**
Cockayne syndrome	191	**70**
CODAS syndrome	1458	**≤10**
Coenzyme Q10 deficiency*	35656	**23**
Coffin-Lowry syndrome	192	**139**
Coffin-Siris syndrome	1465	**335**
COG1-CDG	263508	**≤10**
COG2-CDG	435934	**≤10**
COG4-CDG	263501	**≤10**
COG5-CDG	263487	**≤10**
COG6-CGD	464443	**≤10**
COG7-CDG	79333	**≤10**
COG8-CDG	95428	**≤10**
Cogan syndrome	1467	**86**
Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	444077	**≤10**
Cohen syndrome	193	**94**
COL4A1-related familial vascular leukoencephalopathy	36383	**148**
Colchicine poisoning	31824	**≤10**
Cold agglutinin disease	56425	**221**
Cole-Carpenter syndrome	2050	**≤10**
Collagen type III glomerulopathy	84087	**≤10**
Coloboma of choroid and retina	98942	**286**
Coloboma of eye lens	98943	**≤10**
Coloboma of eyelid	98946	**13**
Coloboma of inferior eyelid	155889	**≤10**
Coloboma of iris	98944	**184**
Coloboma of macula	98945	**≤10**
Coloboma of macula-brachydactyly type B syndrome	1471	**≤10**
Coloboma of optic disc	98947	**79**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Coloboma of superior eyelid	155884	**12**
Colobomatous microphthalmia	98938	**261**
Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	435930	**≤10**
Colonic atresia	1198	**15**
Combined deficiency of factor V and factor VIII	35909	**35**
Combined hamartoma of the retina and retinal pigment epithelium	440727	**≤10**
Combined immunodeficiency due to CARMIL2 deficiency	542301	**≤10**
Combined immunodeficiency due to CD27 deficiency	238505	**≤10**
Combined immunodeficiency due to CD3gamma deficiency	169082	**≤10**
Combined immunodeficiency due to CD70 deficiency	538958	**≤10**
Combined immunodeficiency due to CRAC channel dysfunction	169090	**≤10**
Combined immunodeficiency due to DOCK8 deficiency	217390	**16**
Combined immunodeficiency due to GINS1 deficiency	505227	**≤10**
Combined immunodeficiency due to IL21R deficiency	357329	**≤10**
Combined immunodeficiency due to ITK deficiency	538963	**≤10**
Combined immunodeficiency due to LRBA deficiency	445018	**11**
Combined immunodeficiency due to MALT1 deficiency	397964	**≤10**
Combined immunodeficiency due to Moesin deficiency	504530	**≤10**
Combined immunodeficiency due to partial RAG1 deficiency	231154	**≤10**
Combined immunodeficiency due to STK4 deficiency	314689	**≤10**
Combined immunodeficiency due to ZAP70 deficiency	911	**≤10**
Combined immunodeficiency with facio-oculoskeletal anomalies	221139	**≤10**
Combined immunodeficiency with granulomatosis	157949	**160**
Combined immunodeficiency-enteropathy spectrum	436252	**31**
Combined malonic and methylmalonic acidemia	289504	**≤10**
Combined oxidative phosphorylation defect type 11	324535	**≤10**
Combined oxidative phosphorylation defect type 13	319514	**≤10**
Combined oxidative phosphorylation defect type 14	319519	**≤10**
Combined oxidative phosphorylation defect type 20	420728	**≤10**
Combined oxidative phosphorylation defect type 23	444013	**≤10**
Combined oxidative phosphorylation defect type 4	254925	**≤10**
Combined oxidative phosphorylation defect type 7	254930	**≤10**
Combined oxidative phosphorylation defect type 9	319509	**≤10**
Combined pancreatic lipase-colipase deficiency	309111	**≤10**
Combined pituitary hormone deficiencies, genetic forms	95494	**113**
Combined pulmonary fibrosis-emphysema syndrome	300564	**470**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Combined T and B cell immunodeficiency*	101972	**≤10**
Commissural lip fistula	141061	**≤10**
Common cystic lymphatic malformation*	458833	**141**
Common variable immunodeficiency	1572	**2580**
Common variable immunodeficiency	77303	**≤10**
Complement component 3 deficiency	280133	**≤10**
Complement hyperactivation-angiopathic thrombosis-protein-losing enteropathy syndrome	566175	**≤10**
Complete androgen insensitivity syndrome	99429	**227**
Complete atrioventricular septal defect	1329	**839**
Complete septate uterus	180126	**12**
Complex hereditary spastic paraplegia*	102013	**142**
Complex lethal osteochondrodysplasia	457378	**≤10**
Complex regional pain syndrome	83452	**380**
Complication after organ transplantation	306644	**131**
Complications after hematopoietic stem cell transplantation	90053	**≤10**
Composite hemangioendothelioma	458758	**≤10**
Composite lymphoma	168966	**≤10**
Conductive deafness-malformed external ear syndrome	3216	**58**
Conductive deafness-ptosis-skeletal anomalies syndrome	3236	**≤10**
Cone dystrophy with supernormal rod response	209932	**11**
Cone rod dystrophy	1872	**971**
Congenital abducens nerve palsy	440233	**≤10**
Congenital absence of both forearm and hand	294979	**82**
Congenital absence of both lower leg and foot	294981	**≤10**
Congenital absence of upper arm and forearm with hand present	294975	**≤10**
Congenital absence/hypoplasia of fingers excluding thumb, unilateral	973	**60**
Congenital achiasma	324353	**≤10**
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	90795	**105**
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	90793	**39**
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	90791	**40**
Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	95699	**19**
Congenital adrenal hyperplasia*	418	**568**
Congenital agenesis of the scrotum	495879	**≤10**
Congenital alpha2-antiplasmin deficiency	79	**≤10**
Congenital alveolar capillary dysplasia	210122	**≤10**
Congenital amegakaryocytic thrombocytopenia	3319	**18**
Congenital amyoplasia	488586	**15**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Congenital aortic valve stenosis	3093	**279**
Congenital aortopulmonary window	2037	**26**
Congenital atransferrinemia	1195	**13**
Congenital axonal neuropathy with encephalopathy	538101	**≤10**
Congenital bilateral absence of vas deferens	48	**145**
Congenital bile acid synthesis defect type 1	79301	**≤10**
Congenital bile acid synthesis defect type 2	79303	**11**
Congenital bile acid synthesis defect type 3	79302	**≤10**
Congenital bile acid synthesis defect type 4	79095	**≤10**
Congenital bowing of long bones	2292	**11**
Congenital cataract microcornea with corneal opacity	289499	**≤10**
Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome	1369	**≤10**
Congenital cataracts-facial dysmorphism-neuropathy syndrome	48431	**19**
Congenital central hypoventilation syndrome	661	**176**
Congenital cervical spinal stenosis	831	**15**
Congenital chloride diarrhea	53689	**17**
Congenital chronic diarrhea with protein-losing enteropathy	329242	**24**
Congenital chylothorax	264688	**49**
Congenital contractural arachnodactyly	115	**91**
Congenital cornea plana	53691	**≤10**
Congenital coronary artery aneurysm	95491	**13**
Congenital cystic eye	519384	**≤10**
Congenital diaphragmatic hernia	2140	**1828**
Congenital dyserythropoietic anemia type I	98869	**20**
Congenital dyserythropoietic anemia type II	98873	**35**
Congenital dyserythropoietic anemia type III	98870	**≤10**
Congenital dyserythropoietic anemia type IV	293825	**≤10**
Congenital dyserythropoietic anemia*	85	**15**
Congenital ectropion uveae	91491	**≤10**
Congenital enteropathy due to enteropeptidase deficiency	168601	**≤10**
Congenital enterovirus infection	292	**≤10**
Congenital Epstein-Barr virus infection	70596	**≤10**
Congenital epulis	157826	**≤10**
Congenital erythropoietic porphyria	79277	**40**
Congenital esophageal diverticulum	91358	**≤10**
Congenital eyelid retraction	99176	**≤10**
Congenital factor II deficiency	325	**47**
Congenital factor V deficiency	326	**529**
Congenital factor VII deficiency	327	**1947**
Congenital factor X deficiency	328	**280**
Congenital factor XI deficiency	329	**1415**
Congenital factor XII deficiency	330	**362**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Congenital factor XIII deficiency	331	**66**
Congenital fiber-type disproportion myopathy	2020	**168**
Congenital fibrinogen deficiency	335	**573**
Congenital fibrosis of extraocular muscles	45358	**57**
Congenital generalized lipodystrophy	528	**83**
Congenital Gerbode defect	99095	**≤10**
Congenital glaucoma	98976	**589**
Congenital heart block	60041	**304**
Congenital heart defect-round face-developmental delay syndrome	1355	**≤10**
Congenital hemangioma*	458775	**273**
Congenital hereditary endothelial dystrophy type I	98975	**14**
Congenital hereditary endothelial dystrophy type II	293603	**≤10**
Congenital hereditary facial paralysis-variable hearing loss syndrome	306530	**≤10**
Congenital herpes simplex virus infection	293	**≤10**
Congenital Horner syndrome	91413	**32**
Congenital hydrocephalus	2185	**123**
Congenital hypoplasia of thumb	294988	**58**
Congenital hypothyroidism due to maternal intake of antithyroid drugs	226313	**≤10**
Congenital hypothyroidism due to transplacental passage of TSH-binding inhibitory antibodies	95715	**30**
Congenital ichthyosis-microcephalus-tetraplegia syndrome	2271	**≤10**
Congenital infiltrating lipomatosis of the face	583097	**≤10**
Congenital insensitivity to pain with severe intellectual disability	453510	**≤10**
Congenital insensitivity to pain-anosmia-neuropathic arthropathy	88642	**29**
Congenital insensitivity to pain-hyperhidrosis-absence of C-fiber innervation	217399	**≤10**
Congenital intrauterine infection-like syndrome	1229	**≤10**
Congenital intrinsic factor deficiency	332	**22**
Congenital isolated ACTH deficiency	199296	**145**
Congenital isolated hyperinsulinism*	657	**571**
Congenital knee dislocation	295034	**≤10**
Congenital lactase deficiency	53690	**≤10**
Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type	70472	**≤10**
Congenital laryngeal cyst	141124	**33**
Congenital laryngeal palsy	137932	**132**
Congenital laryngeal web	2374	**13**
Congenital laryngomalacia	2373	**1018**
Congenital left ventricular aneurysm	1055	**≤10**
Congenital limbs-face contractures-hypotonia-developmental delay syndrome	562528	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Congenital lipoid adrenal hyperplasia due to STAR deficency	90790	**31**
Congenital lobar emphysema	1928	**112**
Congenital macroglossia	2430	**129**
Congenital megacalycosis	93109	**17**
Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	69063	**11**
Congenital mesoblastic nephroma	2665	**16**
Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome	391376	**≤10**
Congenital microcoria	566	**18**
Congenital microgastria	199293	**≤10**
Congenital mitral stenosis	99057	**35**
Congenital muscular alpha-dystroglycanopathy with brain and eye anomalies*	352687	**22**
Congenital muscular dystrophy due to LMNA mutation	157973	**68**
Congenital muscular dystrophy type 1B	98893	**≤10**
Congenital muscular dystrophy with cerebellar involvement	370959	**≤10**
Congenital muscular dystrophy with hyperlaxity	371007	**28**
Congenital muscular dystrophy with intellectual disability	370968	**24**
Congenital muscular dystrophy with intellectual disability and severe epilepsy	329178	**≤10**
Congenital muscular dystrophy without intellectual disability	370980	**46**
Congenital muscular dystrophy, Fukuyama type	272	**≤10**
Congenital muscular dystrophy, Ullrich type	75840	**198**
Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome	1875	**≤10**
Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome	486815	**≤10**
Congenital myasthenic syndrome	590	**617**
Congenital myopathy with cores*	172976	**126**
Congenital myopathy with excess of thin filaments	98904	**≤10**
Congenital myopathy with internal nuclei and atypical cores	319160	**≤10**
Congenital myopathy with myasthenic-like onset	424107	**17**
Congenital myopathy with reduced type 2 muscle fibers	544602	**13**
Congenital myopathy, Paradas type	199329	**≤10**
Congenital myotonia*	206973	**177**
Congenital nemaline myopathy*	457074	**20**
Congenital nephrotic syndrome, Finnish type	839	**89**
Congenital neuronal ceroid lipofuscinosis	168486	**≤10**
Congenital neutropenia-myelofibrosis-nephromegaly syndrome	369852	**≤10**
Congenital non-bullous ichthyosiform erythroderma	79394	**138**
Congenital oculomotor nerve palsy	440221	**22**
Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome	2772	**≤10**
Congenital pancreatic cyst	313906	**94**
Congenital panfollicular nevus	139414	**13**
Congenital partial pulmonary venous return anomaly	99124	**234**
Congenital patent ductus arteriosus aneurysm	99072	**≤10**
Congenital plasminogen activator inhibitor type 1 deficiency	465	**14**
Congenital portosystemic shunt	480531	**201**
Congenital prekallikrein deficiency	749	**≤10**
Congenital primary aphakia	83461	**≤10**
Congenital primary lymphedema of Gordon	569821	**≤10**
Congenital primary megaureter	617	**1097**
Congenital progressive bone marrow failure-B-cell immunodeficiency-skeletal dysplasia syndrome	508542	**≤10**
Congenital pseudoarthrosis of the clavicle	66630	**≤10**
Congenital pseudoarthrosis of the limbs	157808	**13**
Congenital ptosis	91411	**288**
Congenital pulmonary airway malformation	2444	**598**
Congenital pulmonary lymphangiectasia	2414	**31**
Congenital pulmonary sequestration	3161	**186**
Congenital pulmonary valvar stenosis	3189	**547**
Congenital pulmonary veins atresia or stenosis	3188	**53**
Congenital pulmonary venous return anomaly*	3090	**105**
Congenital radioulnar synostosis	3269	**57**
Congenital renal artery stenosis	97598	**111**
Congenital respiratory-biliary fistula	2040	**≤10**
Congenital reticular ichthyosiform erythroderma	281190	**≤10**
Congenital retinal arteriovenous communication	353334	**≤10**
Congenital rubella syndrome	290	**26**
Congenital short bowel syndrome	2301	**93**
Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome	369861	**≤10**
Congenital smooth muscle hamartoma	263435	**27**
Congenital sodium diarrhea	103908	**≤10**
Congenital stationary night blindness	215	**228**
Congenital stromal corneal dystrophy	101068	**≤10**
Congenital subglottic stenosis	141121	**92**
Congenital sucrase-isomaltase deficiency	35122	**69**
Congenital supravalvular mitral ring	99059	**≤10**
Congenital syphilis	499009	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Congenital systemic arteriovenous fistula	2039	**23**
Congenital temporomandibular joint ankylosis	210576	**≤10**
Congenital total pulmonary venous return anomaly	99125	**223**
Congenital toxoplasmosis	858	**34**
Congenital tracheal stenosis	141127	**110**
Congenital tracheomalacia	95430	**215**
Congenital tricuspid stenosis	95459	**≤10**
Congenital tricuspid valve dysplasia	555874	**34**
Congenital trigeminal anesthesia	231013	**≤10**
Congenital trochlear nerve palsy	98686	**30**
Congenital tufting enteropathy	92050	**55**
Congenital unguarded mitral orifice	99060	**22**
Congenital unilateral hypoplasia of depressor anguli oris	1166	**129**
Congenital varicella syndrome	291	**≤10**
Congenital velopharyngeal incompetence	2291	**191**
Congenital vertebral-cardiac-renal anomalies syndrome	521438	**≤10**
Congenital vertical talus	178382	**≤10**
Congenitally corrected transposition of the great arteries	216694	**804**
Congenitally uncorrected transposition of the great arteries	860	**1555**
Connective tissue disorder due to lysyl hydroxylase-3 deficiency	300284	**≤10**
Constitutional megaloblastic anemia with severe neurologic disease	319651	**≤10**
Constitutional mismatch repair deficiency syndrome	252202	**≤10**
Continuous spikes and waves during sleep	725	**529**
Contractures-developmental delay-Pierre Robin syndrome	436003	**14**
Contractures-ectodermal dysplasia-cleft lip/palate syndrome	1484	**≤10**
Cooks syndrome	1487	**≤10**
Cor triatriatum dexter	99098	**≤10**
Cor triatriatum sinister	99099	**≤10**
Corneal dystrophy-perceptive deafness syndrome	1490	**≤10**
Corneal endotheliitis	137602	**71**
Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome	352662	**≤10**
Cornelia de Lange syndrome	199	**394**
Corneodermatoosseous syndrome	3194	**≤10**
Coronary arterial fistula	2041	**33**
Coronary ostial stenosis or atresia	99087	**≤10**
Coronary sinus atresia	99118	**≤10**
Coronary sinus stenosis	99117	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Corpus callosum agenesis-abnormal genitalia syndrome	2508	**≤10**
Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome	52055	**≤10**
Corpus callosum agenesis-macrocephaly-hypertelorism syndrome	459074	**≤10**
Corpus callosum agenesis-neuronopathy syndrome	1496	**36**
Cortical blindness-intellectual disability-polydactyly syndrome	1389	**≤10**
Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation	300570	**22**
Corticobasal syndrome	454887	**217**
Corticosteroid-sensitive aseptic abscess syndrome	54251	**17**
Costello syndrome	3071	**80**
Cowden syndrome	201	**492**
Coxopodopatellar syndrome	1509	**45**
Cramp-fasciculation syndrome	581271	**133**
Crane-Heise syndrome	1512	**≤10**
Cranial meningocele	268820	**≤10**
Cranial neuralgia*	221109	**≤10**
Cranio-cervical dystonia with laryngeal and upper-limb involvement	420485	**14**
Craniodiaphyseal dysplasia	1513	**≤10**
Craniodigital-intellectual disability syndrome	1514	**≤10**
Cranioectodermal dysplasia	1515	**22**
Craniofacial conodysplasia	85168	**≤10**
Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome	459061	**≤10**
Craniofacial-deafness-hand syndrome	1529	**≤10**
Craniofrontonasal dysplasia	1520	**48**
Craniofrontonasal dysplasia-Poland anomaly syndrome	1521	**≤10**
Craniometadiaphyseal dysplasia, wormian bone type	85184	**≤10**
Craniometaphyseal dysplasia	1522	**23**
Cranio-osteoarthropathy	1525	**≤10**
Craniopharyngioma	54595	**1848**
Craniosynostosis, Boston type	1541	**≤10**
Craniosynostosis-anal anomalies-porokeratosis syndrome	85199	**≤10**
Craniosynostosis-Dandy-Walker malformation-hydrocephalus syndrome	1538	**≤10**
Craniosynostosis-dental anomalies	284149	**≤10**
Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome	171839	**≤10**
Craniosynostosis-intracranial calcifications syndrome	52054	**≤10**
Creatine deficiency syndrome*	79172	**18**
Crigler-Najjar syndrome	205	**50**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Crimean-Congo hemorrhagic fever	99827	**≤10**
Crisponi syndrome	1545	**24**
Criss-cross heart	1461	**≤10**
Crossed polysyndactyly	2935	**≤10**
Crouzon syndrome	207	**352**
Crouzon syndrome-acanthosis nigricans syndrome	93262	**22**
Cryoglobulinemic vasculitis	91138	**603**
Cryopyrin-associated periodic syndrome*	208650	**81**
Cryptococcosis	1546	**≤10**
Cryptogenic late-onset epileptic spasms	163708	**17**
Cryptogenic organizing pneumonia	1302	**466**
Cryptomicrotia-brachydactyly-excess fingertip arch syndrome	1547	**≤10**
Cryptorchidism-arachnodactyly-intellectual disability syndrome	1548	**≤10**
CTCF-related neurodevelopmental disorder	363611	**11**
Currarino syndrome	1552	**241**
Curry-Jones syndrome	1553	**≤10**
Cushing disease	96253	**2571**
Cushing syndrome due to ectopic ACTH secretion	99889	**131**
Cushing syndrome due to macronodular adrenal hyperplasia	189427	**620**
Cushing syndrome*	553	**654**
Cutaneous collagenous vasculopathy	280779	**≤10**
Cutaneous mastocytoma	79455	**307**
Cutaneous mastocytosis*	66646	**669**
Cutaneous neuroendocrine carcinoma	79140	**23**
Cutaneous pseudolymphoma	451607	**≤10**
Cutaneous small vessel vasculitis	889	**119**
Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome	1555	**≤10**
Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies	221145	**≤10**
Cutis laxa*	209	**82**
Cutis laxa-Marfanoid syndrome	171719	**≤10**
Cutis marmorata telangiectatica congenita	1556	**144**
Cyclic neutropenia	2686	**83**
Cylindrical spirals myopathy	171886	**≤10**
Cystadenoma of childhood	206470	**13**
Cystic echinococcosis	400	**≤10**
Cystic fibrosis	586	**8998**
Cystic fibrosis-gastritis-megaloblastic anemia syndrome	2575	**≤10**
Cystic hamartoma of lung and kidney	2111	**≤10**
Cystic leukoencephalopathy without megalencephaly	85136	**≤10**
Cysticercosis	1560	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Cystinosis	213	**196**
Cystinuria	214	**641**
Cystoid macular dystrophy	75381	**20**
Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk	137698	**≤10**
Czeizel-Losonci syndrome	2437	**≤10**
D-2-hydroxyglutaric aciduria	79315	**≤10**
Dacryocystitis-osteopoikilosis syndrome	1562	**11**
Dandy-Walker malformation-postaxial polydactyly syndrome	1566	**≤10**
Darier disease	218	**226**
DDX41-related hematologic malignancy predisposition syndrome	488647	**≤10**
De Barsy syndrome	2962	**14**
De novo thrombotic microangiopathy after kidney transplantation	244275	**25**
Deaf blind hypopigmentation syndrome, Yemenite type	3214	**≤10**
Deafness with labyrinthine aplasia, microtia, and microdontia	90024	**≤10**
Deafness-craniofacial syndrome	3241	**≤10**
Deafness-ear malformation-facial palsy syndrome	3232	**14**
Deafness-enamel hypoplasia-nail defects syndrome	3220	**15**
Deafness-encephaloneuropathy-obesity-valvulopathy syndrome	254898	**≤10**
Deafness-hypogonadism syndrome	90646	**≤10**
Deafness-infertility syndrome	94064	**≤10**
Deafness-intellectual disability syndrome, Martin-Probst type	85321	**≤10**
Deafness-lymphedema-leukemia syndrome	3226	**≤10**
Deafness-onychodystrophy syndrome*	3231	**≤10**
Deep dermatophytosis	397587	**≤10**
Deficiency in anterior pituitary function-variable immunodeficiency syndrome	293978	**34**
Dehydrated hereditary stomatocytosis	3202	**59**
Dejerine-Sottas syndrome	64748	**17**
Delayed membranous cranial ossification	3034	**≤10**
Delayed speech-facial asymmetry-strabismus-ear lobe creases syndrome	3038	**20**
Deletion 5q35	1627	**18**
Delta-beta-thalassemia	231237	**≤10**
Delta-sarcoglycan-related limb-girdle muscular dystrophy R6	219	**≤10**
Demodicidosis	283	**≤10**
DEND syndrome	79134	**≤10**
Dengue fever	99828	**≤10**
Dent disease	1652	**330**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Dental ankylosis	1077	**≤10**
Dentatorubral pallidoluysian atrophy	101	**12**
Dentin dysplasia	1653	**19**
Dentinogenesis imperfecta	49042	**362**
Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome	71267	**≤10**
Denys-Drash syndrome	220	**82**
Dermatitis herpetiformis	1656	**76**
Dermatofibrosarcoma protuberans	31112	**11**
Dermatoleukodystrophy	1659	**≤10**
Dermatomyositis	221	**1727**
Dermatosparaxis Ehlers-Danlos syndrome	1901	**≤10**
Dermochondrocorneal dystrophy	79149	**≤10**
Dermoid or epidermoid cyst of the central nervous system	530033	**15**
Dermoodontodysplasia	1660	**≤10**
Desbuquois syndrome	1425	**12**
Desminopathy	98909	**82**
Desmin-related myopathy with Mallory body-like inclusions	84132	**≤10**
Desmoid tumor	873	**21**
Desmoplastic infantile astrocytoma/ganglioglioma	251940	**≤10**
Desmoplastic small round cell tumor	83469	**≤10**
Desquamative interstitial pneumonia	98852	**268**
Developmental delay with autism spectrum disorder and gait instability	329195	**189**
Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	369891	**53**
Developmental malformations-deafness-dystonia syndrome	79107	**≤10**
Dextrocardia	1666	**24**
D-glyceric aciduria	941	**≤10**
Diabetic embryopathy	1926	**≤10**
DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	494444	**≤10**
Diaphanospondylodysostosis	66637	**≤10**
Diaphragmatic defect-limb deficiency-skull defect syndrome	2141	**≤10**
Diaphyseal medullary stenosis-bone malignancy syndrome	85182	**≤10**
Diastrophic dysplasia	628	**47**
Diazoxide-resistant diffuse hyperinsulinism*	165988	**≤10**
Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	276598	**≤10**
Diazoxide-resistant focal hyperinsulinism*	79298	**≤10**
Diazoxide-resistant hyperinsulinism*	276585	**12**
Diazoxide-sensitive diffuse hyperinsulinism*	165985	**30**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Didelphys uterus	180086	**75**
Didymosis aplasticosebacea	370046	**≤10**
Diencephalic-mesencephalic junction dysplasia	319192	**≤10**
Differentiated thyroid carcinoma	146	**68**
Diffuse alveolar hemorrhage	90060	**48**
Diffuse astrocytoma	251595	**≤10**
Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome	404437	**≤10**
Diffuse cutaneous mastocytosis	79456	**97**
Diffuse intrinsic pontine glioma	497188	**≤10**
Diffuse large B-cell lymphoma of the central nervous system	300849	**≤10**
Diffuse large B-cell lymphoma with chronic inflammation	300888	**≤10**
Diffuse large B-cell lymphoma*	544	**57**
Diffuse leptomeningeal melanocytosis	252031	**≤10**
Diffuse lymphatic malformation	141209	**134**
Diffuse neonatal hemangiomatosis	2123	**64**
Diffuse palmoplantar keratoderma with painful fissures	369999	**≤10**
Diffuse palmoplantar keratoderma-acrocyanosis syndrome	86918	**≤10**
Diffuse panbronchiolitis	171700	**18**
Digestive duplication	238	**17**
Digestive duplication cyst of the tongue	141071	**≤10**
Digital extensor muscle aplasia-polyneuropathy	2926	**≤10**
Dihydropyrimidine dehydrogenase deficiency	1675	**≤10**
Dihydropyrimidinuria	38874	**≤10**
Dilated cardiomyopathy with ataxia	66634	**21**
Dimethylglycine dehydrogenase deficiency	243343	**≤10**
Diphallia	227	**≤10**
Diphtheria	1679	**≤10**
Discoid lupus erythematosus	90281	**62**
Dislocation of the hip-dysmorphism syndrome	2412	**15**
Disorder of sex development-intellectual disability syndrome	2983	**≤10**
Dissecting cellulitis of the scalp	345	**≤10**
Disseminated peritoneal leiomyomatosis	71274	**≤10**
Disseminated superficial actinic porokeratosis	79152	**26**
Distal 16p11.2 microdeletion syndrome	261222	**133**
Distal 17p13.1 microdeletion syndrome	319171	**≤10**
Distal 17p13.3 microdeletion syndrome	261257	**14**
Distal 22q11.2 microdeletion syndrome	261330	**92**
Distal 22q11.2 microduplication syndrome	261337	**13**
Distal 7q11.23 microdeletion syndrome	254351	**12**
Distal 7q11.23 microduplication syndrome	261102	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Distal anoctaminopathy	399096	**49**
Distal arthrogryposis type 1	1146	**27**
Distal arthrogryposis type 10	251515	**≤10**
Distal arthrogryposis type 5D	329457	**≤10**
Distal arthrogryposis*	97120	**177**
Distal hereditary motor neuropathy type 1	139518	**49**
Distal hereditary motor neuropathy type 2	139525	**≤10**
Distal hereditary motor neuropathy type 5	139536	**≤10**
Distal hereditary motor neuropathy type 7	139589	**≤10**
Distal hereditary motor neuropathy*	53739	**319**
Distal hereditary motor neuropathy, Jerash type	139552	**≤10**
Distal limb deficiencies-micrognathia syndrome	1307	**≤10**
Distal monosomy 10p	1580	**22**
Distal monosomy 10q	96148	**41**
Distal monosomy 12p	280325	**19**
Distal monosomy 12q	96149	**≤10**
Distal monosomy 13q	1590	**34**
Distal monosomy 14q	96150	**17**
Distal monosomy 15q	1596	**49**
Distal monosomy 17q	1597	**13**
Distal monosomy 19p13.3	96129	**≤10**
Distal monosomy 1q	36367	**22**
Distal monosomy 3p	1620	**30**
Distal monosomy 4q	96145	**46**
Distal monosomy 6p	96125	**25**
Distal monosomy 7p	96126	**≤10**
Distal monosomy 7q36	1636	**23**
Distal monosomy 9p	1642	**44**
Distal myopathy with anterior tibial onset	178400	**23**
Distal myopathy with posterior leg and anterior hand involvement	63273	**23**
Distal myopathy, Welander type	603	**≤10**
Distal myotilinopathy	98911	**30**
Distal nebulin myopathy	399103	**16**
Distal renal tubular acidosis	18	**319**
Distal spinal muscular atrophy type 3	139547	**57**
Distal symphalangism	3248	**≤10**
Distal trisomy 10q	96102	**≤10**
Distal trisomy 11q	96103	**≤10**
Distal trisomy 13q	96105	**≤10**
Distal trisomy 14q	1705	**15**
Distal trisomy 16q	96106	**16**
Distal trisomy 17q	3379	**≤10**
Distal trisomy 18q	1716	**18**
Distal trisomy 19q	1717	**15**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Distal trisomy 1p36	96069	**≤10**
Distal trisomy 20q	96107	**≤10**
Distal trisomy 22q	96109	**15**
Distal trisomy 2p	96070	**≤10**
Distal trisomy 2q	96094	**≤10**
Distal trisomy 3p	96071	**≤10**
Distal trisomy 4q	96096	**≤10**
Distal trisomy 5q	96097	**≤10**
Distal trisomy 6p	1745	**≤10**
Distal trisomy 6q	96098	**≤10**
Distal trisomy 7p	96074	**≤10**
Distal trisomy 8q	96100	**≤10**
Distal trisomy 9q	96101	**≤10**
Distal Xq28 microduplication syndrome	293939	**15**
DITRA	404546	**≤10**
DK1-CDG	91131	**≤10**
DNA2-related mitochondrial DNA deletion syndrome	352470	**13**
DNAJB2-related Charcot-Marie-Tooth disease type 2	443950	**≤10**
DNAJB6-related limb-girdle muscular dystrophy D1	34516	**23**
DOCK2 deficiency	447737	**≤10**
Dominant hypophosphatemia with nephrolithiasis or osteoporosis	244305	**255**
Donnai-Barrow syndrome	2143	**11**
DONSON-related microcephaly-short stature-limb abnormalities spectrum	572761	**≤10**
DOORS syndrome	79500	**≤10**
Dopamine beta-hydroxylase deficiency	230	**≤10**
Dopa-responsive dystonia due to sepiapterin reductase deficiency	70594	**≤10**
Dopa-responsive dystonia*	255	**35**
Double outlet left ventricle	3427	**37**
Double outlet right ventricle	3426	**511**
Double uterus-hemivagina-renal agenesis syndrome	3411	**89**
Dowling-Degos disease	79145	**13**
Down syndrome	870	**5464**
DPAGT1-CDG	86309	**≤10**
DPM1-CDG	79322	**≤10**
DPM3-CDG	263494	**≤10**
Dravet syndrome	33069	**863**
Drug or radiation exposure-related interstitial lung disease	264978	**412**
Drug- or toxin-induced pulmonary arterial hypertension*	275786	**148**
Drug reaction with eosinophilia and systemic symptoms	139402	**985**
Drug-induced localized lipodystrophy	90157	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Drug-induced lupus erythematosus	231111	**31**
Drug-induced vasculitis	251325	**15**
Duane retraction syndrome	233	**149**
Dubin-Johnson syndrome	234	**32**
Dubowitz syndrome	235	**12**
Duchenne and Becker muscular dystrophy*	262	**366**
Duchenne muscular dystrophy	98896	**2100**
Duodenal atresia	1203	**109**
Duplication of the esophagus*	91357	**≤10**
Duplication of the pituitary gland	314621	**≤10**
Duplication of urethra	237	**52**
Dural sinus malformation	97339	**50**
Dyggve-Melchior-Clausen disease	239	**19**
DYRK1A-related intellectual disability syndrome	464306	**40**
Dysbetalipoproteinemia	412	**11**
Dyschondrosteosis-nephritis syndrome	1765	**≤10**
Dyschromatosis symmetrica hereditaria	41	**≤10**
Dyschromatosis universalis hereditaria	241	**≤10**
Dysembryoplastic neuroepithelial tumor	251946	**268**
Dysequilibrium syndrome	1766	**113**
Dysferlin-related limb-girdle muscular dystrophy R2	268	**228**
Dyskeratosis congenita	1775	**191**
Dysmorphism-cleft palate-loose skin syndrome	1779	**≤10**
Dysmorphism-conductive hearing loss-heart defect syndrome	289553	**≤10**
Dysmorphism-pectus carinatum-joint laxity syndrome	2104	**≤10**
Dysmorphism-short stature-deafness-disorder of sex development syndrome	2282	**≤10**
Dysosteosclerosis	1782	**≤10**
Dysostosis, Stanescu type	1798	**≤10**
Dysphagia lusoria	99082	**≤10**
Dysplasia epiphysealis hemimelica	1822	**22**
Dysplasia of head of femur, Meyer type	168621	**≤10**
Dysplastic cortical hyperostosis	2204	**≤10**
Dysraphism-cleft lip/palate-limb reduction defects syndrome	2476	**11**
Dyssegmental dysplasia, Silverman-Handmaker type	1865	**≤10**
Dysspondyloenchondromatosis	85198	**≤10**
Dystonia 16	210571	**13**
Dystonia 28	589618	**12**
Dystonia-aphonia syndrome	412217	**≤10**
Dystrophic epidermolysis bullosa pruriginosa	89843	**≤10**
Dystrophic epidermolysis bullosa*	303	**387**
Eales disease	40923	**44**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Early infantile epileptic encephalopathy	1934	**1113**
Early myoclonic encephalopathy	1935	**59**
Early-onset autosomal dominant Alzheimer disease	1020	**100**
Early-onset cerebellar ataxia with retained tendon reflexes	1177	**49**
Early-onset epilepsy-intellectual disability-brain anomalies syndrome	488635	**13**
Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation	289266	**31**
Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	411986	**≤10**
Early-onset generalized limb-onset dystonia	256	**279**
Early-onset myopathy with fatal cardiomyopathy	289377	**≤10**
Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome	439212	**≤10**
Early-onset non-syndromic cataract	91492	**1130**
Early-onset parkinsonism-intellectual disability syndrome	2379	**≤10**
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	496641	**≤10**
Early-onset progressive encephalopathy with migrant continuous myoclonus	1943	**19**
Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	500144	**≤10**
Early-onset schizophrenia	96369	**156**
Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	313772	**≤10**
Early-onset X-linked optic atrophy	98890	**29**
Ear-patella-short stature syndrome	2554	**25**
EAST syndrome	199343	**15**
Ebstein malformation of the tricuspid valve	1880	**344**
Ectasia of the right atrial appendage	99101	**≤10**
Ectodermal dysplasia with natal teeth, Turnpenny type	69083	**≤10**
Ectodermal dysplasia, trichoodontoonychial type	1818	**≤10**
Ectodermal dysplasia-sensorineural deafness syndrome	1883	**≤10**
Ectodermal dysplasia-skin fragility syndrome	158668	**≤10**
Ectodermal dysplasia-syndactyly syndrome	247820	**≤10**
Ectopia lentis-chorioretinal dystrophy-myopia syndrome	1884	**12**
Ectrodactyly-polydactyly syndrome	1892	**≤10**
EDICT syndrome	293936	**65**
EEC syndrome	1896	**124**
EEM syndrome	1897	**≤10**
Ehlers-Danlos syndrome*	98249	**1796**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Ehlers-Danlos/osteogenesis imperfecta syndrome	230857	**28**
Eisenmenger syndrome	97214	**340**
Elastofibroma dorsi	228243	**≤10**
Ellis Van Creveld syndrome	289	**55**
Emanuel syndrome	96170	**16**
Embryonal carcinoma	180226	**≤10**
Emery-Dreifuss muscular dystrophy	261	**213**
Emery-Nelson syndrome	1927	**≤10**
Enamel-renal syndrome	1031	**45**
Encephalitis lethargica	83600	**≤10**
Encephaloclastic disorder*	269190	**≤10**
Encephalocraniocutaneous lipomatosis	2396	**≤10**
Encephalopathy due to mitochondrial and peroxisomal fission defect	527276	**≤10**
Encephalopathy due to prosaposin deficiency	139406	**≤10**
Encephalopathy due to sulfite oxidase deficiency	833	**27**
Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	319678	**≤10**
Encircling double aortic arch	99075	**34**
Endocardial fibroelastosis	2022	**≤10**
Endometrial stromal sarcoma	213711	**≤10**
Endometrioid carcinoma of ovary	454723	**≤10**
Endophthalmitis	199323	**33**
Endosteal hyperostosis, Worth type	2790	**≤10**
Enlarged parietal foramina	60015	**20**
Enthesitis-related juvenile idiopathic arthritis	85438	**2605**
Eosinophilic angiocentric fibrosis	449566	**21**
Eosinophilic colitis	402035	**13**
Eosinophilic fasciitis	3165	**178**
Eosinophilic gastroenteritis	2070	**11**
Eosinophilic granulomatosis with polyangiitis	183	**1089**
Ependymal tumor*	301	**≤10**
Ependymoblastoma	251880	**≤10**
Ependymoma	251636	**47**
Epiblepharon	99169	**42**
Epidermal nevus syndrome	35125	**216**
Epidermodysplasia verruciformis	302	**30**
Epidermolysis bullosa acquisita	46487	**187**
Epidermolysis bullosa simplex with mottled pigmentation	79397	**≤10**
Epidermolysis bullosa simplex with muscular dystrophy	257	**≤10**
Epidermolysis bullosa simplex*	304	**313**
Epidermolytic nevus	497737	**20**
Epidermolytic palmoplantar keratoderma	2199	**62**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Epilepsy with myoclonic absences	86911	**277**
Epilepsy-microcephaly-skeletal dysplasia syndrome	1948	**≤10**
Epilepsy-telangiectasia syndrome	1951	**≤10**
Epileptic encephalopathy with global cerebral demyelination	353217	**≤10**
Epiphyseal stippling-osteoclastic hyperplasia syndrome	1952	**≤10**
Epiphysiolysis of the hip	399329	**≤10**
Episodic ataxia type 1	37612	**45**
Episodic ataxia type 3	79135	**≤10**
Episodic ataxia type 5	211067	**≤10**
Episodic ataxia type 6	209967	**≤10**
Episodic ataxia with slurred speech	401953	**≤10**
Epithelial basement membrane dystrophy	98956	**35**
Epithelial recurrent erosion dystrophy	293381	**≤10**
Epithelioid hemangioendothelioma	157791	**31**
Epstein-Barr virus-positive diffuse large B-cell lymphoma of the elderly	289661	**≤10**
Erdheim-Chester disease	35687	**294**
Ermine phenotype	999	**≤10**
Erosive pustular dermatosis of the scalp	222	**≤10**
Erythema elevatum diutinum	90000	**≤10**
Erythema multiforme major	502499	**131**
Erythroderma desquamativum	314	**≤10**
Erythrokeratoderma ''en cocardes''	315	**≤10**
Erythrokeratoderma variabilis progressiva*	308166	**≤10**
Erythrokeratodermia variabilis	317	**24**
Esophageal atresia	1199	**2318**
Esophageal duplication cyst	100047	**≤10**
Essential fructosuria	2056	**≤10**
Essential thrombocythemia	3318	**84**
Estrogen resistance syndrome	785	**≤10**
Ethylmalonic encephalopathy	51188	**≤10**
Euryblepharon	99172	**≤10**
Euthyroid dysprealbuminemic hyperthyroxinemia	597939	**≤10**
Euthyroid Graves orbitopathy	466682	**15**
Evans syndrome	1959	**500**
Exercise intolerance with lactic acidosis*	254843	**20**
Exercise-induced malignant hyperthermia	466650	**19**
Exfoliative ichthyosis	289586	**≤10**
Exostoses-anetodermia-brachydactyly type E syndrome	1962	**≤10**
Exstrophy-epispadias complex	322	**591**
Extensive peripapillary myelinated nerve fibers	440724	**14**
External auditory canal aplasia/hypoplasia	141074	**1389**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
External auditory canal atresia-vertical talus-hypertelorism syndrome	3023	**≤10**
Extracranial carotid artery aneurysm	494424	**11**
Extracutaneous mastocytoma	66662	**≤10**
Extragonadal germinoma	182127	**135**
Extragonadal teratoma	883	**100**
Extramammary Paget disease	2800	**≤10**
Extraneural perineurioma	100002	**≤10**
Extranodal nasal NK/T cell lymphoma	86879	**≤10**
Extrapelvic endometriosis	137820	**72**
Extraskeletal Ewing sarcoma	370334	**≤10**
Fabry disease	324	**1558**
Facial arteriovenous malformation*	156230	**420**
Facial dermoid cyst	141051	**399**
Facial diplegia with paresthesias	480701	**≤10**
Facial dysmorphism-anorexia-cachexia-eye and skin anomalies syndrome	1969	**≤10**
Facial dysmorphism-immunodeficiency-livedo-short stature syndrome	352712	**11**
Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering blebs syndrome	412022	**≤10**
Facial dysmorphism-shawl scrotum-joint laxity syndrome	1778	**≤10**
Facial onset sensory and motor neuronopathy	85162	**27**
Faciocardiorenal syndrome	1973	**≤10**
Facioscapulohumeral dystrophy	269	**3314**
Fallot complex-intellectual disability-growth delay syndrome	3304	**≤10**
Familial abdominal aortic aneurysm	86	**64**
Familial acute necrotizing encephalopathy	88619	**≤10**
Familial adenomatous polyposis	733	**359**
Familial adrenal hypoplasia with absent pituitary luteinizing hormone	95700	**≤10**
Familial Alzheimer-like prion disease	280397	**≤10**
Familial anetoderma	228277	**44**
Familial angiolipomatosis	199279	**≤10**
Familial aortic dissection	229	**302**
Familial articular hypermobility syndrome	2295	**682**
Familial atrial fibrillation	334	**117**
Familial atrial myxoma	615	**≤10**
Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease	436242	**≤10**
Familial atypical multiple mole melanoma syndrome	404560	**119**
Familial avascular necrosis of femoral head	86820	**≤10**
Familial benign chronic pemphigus	2841	**205**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Familial benign copper deficiency	1551	**≤10**
Familial bicuspid aortic valve	402075	**288**
Familial calcium pyrophosphate deposition	1416	**29**
Familial caudal dysgenesis	1768	**≤10**
Familial cavitary optic disc anomaly	464760	**≤10**
Familial cerebral cavernous malformation	221061	**476**
Familial cerebral saccular aneurysm	231160	**54**
Familial cervical artery dissection	36382	**196**
Familial Chilblain lupus	481662	**≤10**
Familial chylomicronemia syndrome	444490	**29**
Familial clubfoot with or without associated lower limb anomalies	199315	**80**
Familial cold urticaria	47045	**25**
Familial congenital mirror movements	238722	**27**
Familial congenital nasolacrimal duct obstruction	451612	**≤10**
Familial congenital palsy of trochlear nerve	91498	**≤10**
Familial cortical myoclonus	319189	**≤10**
Familial cutaneous collagenoma	53296	**≤10**
Familial developmental dysphasia	1799	**45**
Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	300751	**404**
Familial drusen	75376	**72**
Familial dysautonomia	1764	**18**
Familial dyskinesia and facial myokymia	324588	**≤10**
Familial encephalopathy with neuroserpin inclusion bodies	85110	**≤10**
Familial episodic pain syndrome	391384	**≤10**
Familial expansile osteolysis	85195	**≤10**
Familial exudative vitreoretinopathy	891	**134**
Familial focal epilepsy with variable foci	98820	**55**
Familial generalized lentiginosis	231040	**≤10**
Familial gestational hyperthyroidism	99819	**15**
Familial glucocorticoid deficiency	361	**29**
Familial hemophagocytic lymphohistiocytosis	540	**105**
Familial hyperaldosteronism type I	403	**91**
Familial hyperaldosteronism type II	404	**19**
Familial hyperaldosteronism type III	251274	**≤10**
Familial hyperaldosteronism*	235936	**49**
Familial hypercholanemia	238475	**23**
Familial hyperprolactinemia	397685	**19**
Familial hyperthyroidism due to mutations in TSH receptor	424	**93**
Familial hypoaldosteronism	427	**20**
Familial hypocalciuric hypercalcemia	405	**577**
Familial idiopathic dilatation of the right atrium	1677	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Familial infantile bilateral striatal necrosis	225154	**≤10**
Familial infantile myoclonic epilepsy	352582	**18**
Familial intestinal malrotation	508410	**≤10**
Familial isolated arrhythmogenic right ventricular dysplasia	217656	**1355**
Familial isolated clinodactyly of fingers	295014	**23**
Familial isolated congenital asplenia	101351	**26**
Familial isolated dilated cardiomyopathy	154	**6669**
Familial isolated hyperparathyroidism	99879	**186**
Familial isolated hypoparathyroidism	2238	**282**
Familial isolated pituitary adenoma	314777	**≤10**
Familial isolated restrictive cardiomyopathy	75249	**170**
Familial isolated trichomegaly	411788	**≤10**
Familial juvenile hypertrophy of the breast	180176	**48**
Familial keratoacanthoma	493	**≤10**
Familial long QT syndrome*	768	**2012**
Familial male-limited precocious puberty	3000	**26**
Familial median cleft of the upper and lower lips	401942	**≤10**
Familial Mediterranean fever	342	**2051**
Familial medullary thyroid carcinoma	99361	**≤10**
Familial melanoma	618	**337**
Familial mesial temporal lobe epilepsy with febrile seizures	165805	**≤10**
Familial mitral valve prolapse	741	**67**
Familial monosomy 7 syndrome	495930	**≤10**
Familial multinodular goiter	276399	**16**
Familial multiple lipomatosis	199276	**26**
Familial multiple meningioma	263662	**14**
Familial multiple nevi flammei	624	**≤10**
Familial or sporadic hemiplegic migraine	569	**207**
Familial ossifying fibroma	435329	**≤10**
Familial osteochondritis dissecans	251262	**≤10**
Familial ovarian cancer*	213517	**≤10**
Familial pancreatic carcinoma	1333	**148**
Familial papillary or follicular thyroid carcinoma	319487	**≤10**
Familial paroxysmal ataxia	97	**127**
Familial partial epilepsy*	309	**96**
Familial partial lipodystrophy*	98306	**27**
Familial partial lipodystrophy, Dunnigan type	2348	**386**
Familial partial lipodystrophy, Köbberling type	79084	**23**
Familial patent arterial duct	466729	**16**
Familial platelet disorder with associated myeloid malignancy	71290	**113**
Familial primary hyperparathyroidism*	2207	**565**
Familial primary localized cutaneous amyloidosis	353220	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Familial progressive cardiac conduction defect	871	**215**
Familial progressive hyper- and hypopigmentation	280628	**≤10**
Familial progressive hyperpigmentation	79146	**16**
Familial progressive retinal dystrophy-iris coloboma-congenital cataract syndrome	488197	**≤10**
Familial prostate cancer	1331	**15**
Familial reactive perforating collagenosis	79147	**≤10**
Familial recurrent peripheral facial palsy	2809	**21**
Familial renal glucosuria	69076	**36**
Familial scaphocephaly syndrome, McGillivray type	168624	**15**
Familial Scheuermann disease	3135	**32**
Familial short QT syndrome	51083	**49**
Familial sick sinus syndrome	166282	**14**
Familial spontaneous pneumothorax	2903	**34**
Familial steroid-resistant nephrotic syndrome with adrenal insufficiency	506334	**≤10**
Familial steroid-resistant nephrotic syndrome with sensorineural deafness	280406	**≤10**
Familial supernumerary nipples	2456	**≤10**
Familial temporal lobe epilepsy	98819	**39**
Familial thoracic aortic aneurysm and aortic dissection	91387	**835**
Familial thrombocytosis	71493	**17**
Familial thrombomodulin anomalies	3324	**≤10**
Familial thyroglossal duct cyst	93953	**101**
Familial thyroid dyshormonogenesis	95716	**408**
Familial tumoral calcinosis	53715	**38**
Familial vesicoureteral reflux	289365	**145**
Familial visceral myopathy	2604	**≤10**
Fanconi anemia	84	**304**
Fanconi-Bickel syndrome	2088	**23**
Farber disease	333	**≤10**
Farmer's lung disease	99906	**38**
FASTKD2-related infantile mitochondrial encephalomyopathy	166105	**≤10**
Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease	439854	**≤10**
Fatal familial insomnia	466	**≤10**
Fatal infantile cytochrome C oxidase deficiency	1561	**≤10**
Fatal infantile hypertonic myofibrillar myopathy	280553	**≤10**
Fatal infantile lactic acidosis with methylmalonic aciduria	17	**25**
Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	168566	**≤10**
FATCO syndrome	2492	**≤10**
Fatty acid hydroxylase-associated neurodegeneration	329308	**11**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Fatty acyl-CoA reductase 1 deficiency	438178	**≤10**
Febrile infection-related epilepsy syndrome	163703	**77**
Feingold syndrome	1305	**106**
Felty syndrome	47612	**≤10**
Female infertility due to oocyte meiotic arrest	488191	**≤10**
Female infertility due to zona pellucida defect	404466	**≤10**
Female restricted epilepsy with intellectual disability	101039	**111**
Femoral agenesis/hypoplasia	1987	**182**
Femoral-facial syndrome	1988	**≤10**
Femur-fibula-ulna complex	2019	**13**
Fetal akinesia deformation sequence	994	**14**
Fetal akinesia-cerebral and retinal hemorrhage syndrome	363409	**≤10**
Fetal alcohol syndrome	1915	**773**
Fetal and neonatal alloimmune thrombocytopenia	853	**27**
Fetal anticonvulsant syndrome*	370068	**≤10**
Fetal cytomegalovirus syndrome	294	**227**
Fetal hydantoin syndrome	1912	**≤10**
Fetal iodine syndrome	1910	**12**
Fetal lower urinary tract obstruction*	435365	**30**
Fetal parvovirus syndrome	295	**≤10**
Fetal valproate spectrum disorder	1906	**575**
Fever-associated acute infantile liver failure syndrome	464724	**≤10**
FG syndrome type 1	93932	**≤10**
FGFR2-related bent bone dysplasia	313855	**≤10**
Fibroblastic rheumatism	477650	**≤10**
Fibrochondrogenesis	2021	**≤10**
Fibrodysplasia ossificans progressiva	337	**158**
Fibronectin glomerulopathy	84090	**≤10**
Fibrosarcoma	2030	**≤10**
Fibrous dysplasia of bone	249	**2070**
Fibular aplasia-complex brachydactyly syndrome	2639	**≤10**
Fibular aplasia-ectrodactyly syndrome	1118	**26**
Fibular dimelia-diplopodia syndrome	1757	**≤10**
Fibular hemimelia	93323	**94**
Filippi syndrome	3255	**≤10**
Fingerprint body myopathy	97232	**≤10**
Finnish upper limb-onset distal myopathy	399086	**≤10**
First branchial cleft anomaly	141013	**190**
Fixed drug eruption	293812	**125**
Fixed subaortic stenosis	3092	**71**
FKRP-related limb-girdle muscular dystrophy R9	34515	**128**
Flat face-microstomia-ear anomaly syndrome	1968	**≤10**
Fleck corneal dystrophy	98970	**≤10**
FLNA-related X-linked myxomatous valvular dysplasia	555877	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Floating-Harbor syndrome	2044	**75**
Florid cemento-osseous dysplasia	83451	**≤10**
Focal acral hyperkeratosis	308013	**≤10**
Focal dermal hypoplasia	2092	**72**
Focal epilepsy-intellectual disability-cerebro-cerebellar malformation	352587	**23**
Focal facial dermal dysplasia	398166	**30**
Focal myositis	48918	**157**
Focal palmoplantar and gingival keratoderma	2200	**≤10**
Focal palmoplantar keratoderma with joint keratoses	370002	**≤10**
Foix-Alajouanine syndrome	79093	**≤10**
Foix-Chavany-Marie syndrome	2048	**≤10**
Follicular lymphoma	545	**21**
Folliculotropic mycosis fungoides	178512	**≤10**
Formiminoglutamic aciduria	51208	**≤10**
Fountain syndrome	3219	**≤10**
Fourth branchial cleft anomaly	141037	**86**
Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome	397618	**≤10**
Foveal hypoplasia-presenile cataract syndrome	2253	**22**
Fowler urethral sphincter dysfunction syndrome	2795	**≤10**
Fowler vasculopaty	221126	**≤10**
FOXG1 syndrome	561854	**49**
Fragile X syndrome	908	**2029**
Fragile X-associated tremor/ataxia syndrome	93256	**96**
Fraser syndrome	2052	**23**
Frasier syndrome	347	**25**
FRAXE intellectual disability	100973	**47**
FRAXF syndrome	100974	**≤10**
Free sialic acid storage disease	834	**≤10**
Freeman-Sheldon syndrome	2053	**45**
Fried syndrome	85335	**≤10**
Friedreich ataxia	95	**920**
Frontal fibrosing alopecia	254492	**≤10**
Frontofacionasal dysplasia	1791	**13**
Frontometaphyseal dysplasia	1826	**22**
Frontonasal arteriovenous malformation	141168	**30**
Frontonasal dysplasia*	250	**62**
Frontorhiny	391474	**≤10**
Frontotemporal dementia with motor neuron disease	275872	**401**
Frontotemporal dementia*	282	**530**
Fructose-1,6-bisphosphatase deficiency	348	**33**
Fryns syndrome	2059	**15**
FTH1-related iron overload	247790	**≤10**
Fuchs endothelial corneal dystrophy	98974	**410**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Fuchs heterochromic iridocyclitis	263479	**43**
Fucosidosis	349	**≤10**
Fuhrmann syndrome	2854	**≤10**
Fukutin-related limb-girdle muscular dystrophy R13	206554	**≤10**
Fulminant viral hepatitis	35063	**17**
Fumaric aciduria	24	**12**
Functioning gonadotropic adenoma	91348	**1076**
Functioning pituitary adenoma*	314753	**≤10**
Fundus albipunctatus	227796	**22**
Fungal keratitis	519930	**15**
Fungal myositis	207000	**≤10**
Furuncular myiasis	591	**≤10**
Gabriele-de Vries syndrome	506358	**13**
Galactokinase deficiency	79237	**≤10**
Galactose epimerase deficiency	79238	**≤10**
Galactosialidosis	351	**≤10**
Gallbladder neuroendocrine tumor	100086	**≤10**
Galloway-Mowat syndrome	2065	**40**
Gamma-aminobutyric acid transaminase deficiency	2066	**≤10**
Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5	353	**233**
Gangliocytoma	251937	**≤10**
Ganglioglioma	251949	**134**
Ganglioneuroblastoma	251877	**≤10**
Ganglioneuroma	251992	**11**
GAPO syndrome	2067	**≤10**
Gastrocutaneous syndrome	2069	**≤10**
Gastrointestinal stromal tumor	44890	**≤10**
Gastroschisis	2368	**262**
Gaucher disease	355	**486**
Geleophysic dysplasia	2623	**21**
Gemignani syndrome	2074	**≤10**
Generalized arterial calcification of infancy	51608	**20**
Generalized basaloid follicular hamartoma syndrome	168632	**≤10**
Generalized bulbospinal muscular atrophy*	206710	**57**
Generalized epilepsy with febrile seizures-plus	36387	**547**
Generalized epilepsy-paroxysmal dyskinesia syndrome	79137	**≤10**
Generalized essential telangiectasia	280774	**≤10**
Generalized glucocorticoid resistance syndrome	786	**11**
Generalized peeling skin syndrome	263543	**≤10**
Generalized pustular psoriasis	247353	**20**
Genetic hyperferritinemia without iron overload	254704	**173**
Genetic non-syndromic obesity	98267	**552**
Genetic recurrent myoglobinuria	99845	**15**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Genetic steroid-resistant nephrotic syndrome	656	**205**
Genetic transient congenital hypothyroidism	226316	**13**
Genitopalatocardiac syndrome	2075	**≤10**
Genitopatellar syndrome	85201	**≤10**
Genochondromatosis type 1	85197	**≤10**
Genochondromatosis type 2	93398	**≤10**
Geroderma osteodysplastica	2078	**≤10**
Gerstmann-Straussler-Scheinker syndrome	356	**≤10**
Ghosal hematodiaphyseal dysplasia	1802	**≤10**
Giant adenofibroma of the breast	180267	**68**
Giant axonal neuropathy	643	**13**
Giant cell arteritis	397	**4103**
Giant cell tumor of bone	363976	**≤10**
Gingival fibromatosis-facial dysmorphism syndrome	2025	**≤10**
Gingival fibromatosis-hypertrichosis syndrome	2026	**≤10**
Gingival fibromatosis-progressive deafness syndrome	2027	**≤10**
Gitelman syndrome	358	**672**
Glanzmann thrombasthenia	849	**352**
Glaucoma secondary to spherophakia/ectopia lentis and megalocornea	238763	**13**
Glaucoma-sleep apnea syndrome	2085	**≤10**
Glial tumor of neuroepithelial tissue with unknown origin*	251668	**≤10**
Glial tumor*	182067	**29**
Glioblastoma	360	**40**
Glioependymal/ependymal cyst	269197	**≤10**
Gliomatosis cerebri	251582	**≤10**
Global developmental delay-alopecia-macrocephaly-facial dysmorphism-structural brain anomalies syndrome	544488	**≤10**
Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome	488613	**≤10**
Global developmental delay-osteopenia-ectodermal defect syndrome	73223	**≤10**
Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	480898	**≤10**
Glomus tumor	391651	**≤10**
Glomuvenous malformation	83454	**66**
Glossopalatine ankylosis	141163	**≤10**
Glucagonoma	97280	**≤10**
Glucose-galactose malabsorption	35710	**≤10**
Glutaric acidemia type 3	35706	**≤10**
Glutaryl-CoA dehydrogenase deficiency	25	**146**
Glutathione synthetase deficiency	32	**15**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Glycerol kinase deficiency*	308993	**≤10**
Glycine encephalopathy	407	**96**
Glycogen storage disease due to acid maltase deficiency	365	**440**
Glycogen storage disease due to aldolase A deficiency	57	**≤10**
Glycogen storage disease due to glucose-6-phosphatase deficiency	364	**303**
Glycogen storage disease due to glycogen branching enzyme deficiency	367	**35**
Glycogen storage disease due to glycogen debranching enzyme deficiency	366	**206**
Glycogen storage disease due to glycogen synthase deficiency*	308520	**≤10**
Glycogen storage disease due to hepatic glycogen synthase deficiency	2089	**20**
Glycogen storage disease due to lactate dehydrogenase deficiency	2364	**≤10**
Glycogen storage disease due to LAMP-2 deficiency	34587	**41**
Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency	79240	**≤10**
Glycogen storage disease due to liver glycogen phosphorylase deficiency	369	**39**
Glycogen storage disease due to liver phosphorylase kinase deficiency	264580	**22**
Glycogen storage disease due to muscle and heart glycogen synthase deficiency	137625	**≤10**
Glycogen storage disease due to muscle glycogen phosphorylase deficiency	368	**327**
Glycogen storage disease due to muscle phosphofructokinase deficiency	371	**≤10**
Glycogen storage disease due to muscle phosphorylase kinase deficiency	715	**≤10**
Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	713	**≤10**
Glycogen storage disease due to phosphoglycerate mutase deficiency	97234	**≤10**
Glycogen storage disease due to phosphorylase kinase deficiency*	370	**116**
Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	263297	**≤10**
GM1 gangliosidosis	354	**104**
GM2 gangliosidosis*	309152	**14**
GM2 gangliosidosis, AB variant	309246	**≤10**
GM3 synthase deficiency	370933	**≤10**
GMPPB-related limb-girdle muscular dystrophy R19	363623	**≤10**
GMS syndrome	2090	**≤10**
GNAO1-related developmental delay-seizures-movement disorder spectrum	592564	**18**
Gnathodiaphyseal dysplasia	53697	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
GNB5-related intellectual disability-cardiac arrhythmia syndrome	542306	**≤10**
GNE myopathy	602	**90**
Goldberg-Shprintzen megacolon syndrome	66629	**≤10**
Goldmann-Favre syndrome	53540	**46**
Gollop-Wolfgang complex	1986	**≤10**
Gómez-López-Hernández syndrome	1532	**≤10**
Gonadoblastoma	206484	**≤10**
Gonococcal conjunctivitis	1482	**≤10**
Good syndrome	169105	**58**
Gordon syndrome	376	**18**
Gorham-Stout disease	73	**42**
Gorlin syndrome	377	**444**
Gorlin-Chaudhry-Moss syndrome	2095	**≤10**
GRACILE syndrome	53693	**≤10**
Graft versus host disease	39812	**68**
Graham Little-Piccardi-Lassueur syndrome	505	**≤10**
Grange syndrome	79094	**≤10**
Granular corneal dystrophy type I	98962	**15**
Granular corneal dystrophy type II	98963	**≤10**
Granulomatosis with polyangiitis	900	**1858**
Granulomatous mastitis	64722	**45**
Granulomatous slack skin	33111	**≤10**
Gray platelet syndrome	721	**23**
Greig cephalopolysyndactyly syndrome	380	**215**
GRFoma	97261	**≤10**
GRIN2B-related developmental delay, intellectual disability and autism spectrum disorder	589547	**29**
Griscelli syndrome	381	**25**
Growing teratoma syndrome	314613	**≤10**
Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome	391348	**≤10**
Growth delay due to insulin-like growth factor I resistance	73273	**32**
Growth delay due to insulin-like growth factor type 1 deficiency	73272	**42**
Growth delay-hydrocephaly-lung hypoplasia syndrome	3035	**≤10**
Grubben-de Cock-Borghgraef syndrome	2101	**≤10**
Guanidinoacetate methyltransferase deficiency	382	**32**
Guillain-Barré syndrome*	2103	**1649**
Guttmacher syndrome	2957	**≤10**
Gyrate atrophy of choroid and retina	414	**37**
H syndrome	168569	**16**
Haddad syndrome	99803	**21**
Hajdu-Cheney syndrome	955	**48**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Hallermann-Streiff syndrome	2108	**25**
Hallermann-Streiff-like syndrome	2109	**≤10**
Hallux varus-preaxial polysyndactyly syndrome	2110	**≤10**
HANAC syndrome	73229	**31**
Hand-foot-genital syndrome	2438	**≤10**
Harlequin ichthyosis	457	**23**
Harlequin syndrome	199282	**≤10**
Harrod syndrome	2115	**≤10**
Hartnup disease	2116	**≤10**
Hartsfield syndrome	2117	**≤10**
Heart defects-limb shortening syndrome	1354	**≤10**
Heart defect-tongue hamartoma-polysyndactyly syndrome	1338	**≤10**
Heart-hand syndrome type 2	1350	**≤10**
Heart-hand syndrome, Slovenian type	168796	**12**
Heavy chain disease	86864	**≤10**
HELLP syndrome	244242	**30**
Hemangioblastoma	252054	**≤10**
Hemifacial hyperplasia	141145	**29**
Hemifacial spasm	221083	**271**
Hemihyperplasia-multiple lipomatosis syndrome	276280	**≤10**
Hemimegalencephaly	99802	**93**
Hemimelia*	2130	**≤10**
Hemochromatosis type 2	79230	**60**
Hemochromatosis type 3	225123	**34**
Hemochromatosis type 4	139491	**319**
Hemoglobin C disease	2132	**179**
Hemoglobin C-beta-thalassemia syndrome	231242	**43**
Hemoglobin D disease	90039	**≤10**
Hemoglobin E disease	2133	**58**
Hemoglobin E-beta-thalassemia syndrome	231249	**51**
Hemoglobin Lepore-beta-thalassemia syndrome	330032	**≤10**
Hemoglobin M disease	330041	**≤10**
Hemoglobinopathy Toms River	280615	**≤10**
Hemolytic anemia due to glucophosphate isomerase deficiency	712	**≤10**
Hemolytic anemia due to glutathione reductase deficiency	90030	**≤10**
Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency	35120	**≤10**
Hemolytic anemia due to red cell pyruvate kinase deficiency	766	**119**
Hemolytic uremic syndrome with DGKE deficiency	357008	**≤10**
Hemophagocytic syndrome associated with an infection	158048	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Hemophilia A	98878	**8331**
Hemophilia B	98879	**1964**
Hemophilia*	448	**32**
Hennekam syndrome	2136	**26**
Hennekam-Beemer syndrome	2135	**≤10**
Heparin-induced thrombocytopenia	3325	**≤10**
Hepatic cystic hamartoma	386	**47**
Hepatic fibrosis-renal cysts-intellectual disability syndrome	2031	**≤10**
Hepatic veno-occlusive disease	890	**306**
Hepatitis delta	402823	**≤10**
Hepatoblastoma	449	**20**
Hepatocellular adenoma	54272	**85**
Hepatocellular carcinoma*	88673	**13**
Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	137681	**≤10**
Hepatoerythropoietic porphyria	95159	**≤10**
Hepatosplenic T-cell lymphoma	86882	**≤10**
Hereditary acrokeratotic poikiloderma	2907	**≤10**
Hereditary amyloidosis with primary renal involvement	85450	**54**
Hereditary angioedema with C1Inh deficiency	528623	**908**
Hereditary angioedema with normal C1Inh	528647	**351**
Hereditary angioedema*	91378	**61**
Hereditary arterial and articular multiple calcification syndrome	289601	**≤10**
Hereditary ATTR amyloidosis*	271861	**1247**
Hereditary breast and ovarian cancer syndrome	145	**974**
Hereditary breast cancer	227535	**35**
Hereditary cerebral hemorrhage with amyloidosis	85458	**≤10**
Hereditary chronic pancreatitis	676	**234**
Hereditary combined deficiency of vitamin K-dependent clotting factors	98434	**18**
Hereditary continuous muscle fiber activity	972	**12**
Hereditary coproporphyria	79273	**61**
Hereditary diffuse gastric cancer	26106	**≤10**
Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia	313808	**15**
Hereditary elliptocytosis	288	**128**
Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome	221043	**≤10**
Hereditary folate malabsorption	90045	**≤10**
Hereditary fructose intolerance	469	**193**
Hereditary geniospasm	53372	**≤10**
Hereditary gingival fibromatosis	2024	**11**
Hereditary hemorrhagic telangiectasia	774	**3471**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Hereditary hypercarotenemia and vitamin A deficiency	199285	**≤10**
Hereditary hyperekplexia	3197	**71**
Hereditary hyperferritinemia-cataract syndrome	163	**164**
Hereditary hypophosphatemic rickets with hypercalciuria	157215	**63**
Hereditary inclusion body myopathy type 4	324381	**≤10**
Hereditary inclusion body myopathy-joint contractures-ophthalmoplegia syndrome	79091	**≤10**
Hereditary isolated aplastic anemia	397692	**32**
Hereditary late-onset Parkinson disease	411602	**45**
Hereditary leiomyomatosis and renal cell cancer	523	**247**
Hereditary methemoglobinemia	621	**16**
Hereditary motor and sensory neuropathy type 5	64751	**≤10**
Hereditary motor and sensory neuropathy type 6	90120	**22**
Hereditary motor and sensory neuropathy with acrodystrophy	90119	**≤10**
Hereditary motor and sensory neuropathy, Okinawa type	90117	**≤10**
Hereditary mucoepithelial dysplasia	1839	**12**
Hereditary myopathy with early respiratory failure	178464	**78**
Hereditary myopathy with lactic acidosis due to ISCU deficiency	43115	**≤10**
Hereditary neurocutaneous malformation	1062	**≤10**
Hereditary neuroendocrine tumor of small intestine	456333	**≤10**
Hereditary neuropathy with liability to pressure palsies	640	**1226**
Hereditary neutrophilia	279943	**12**
Hereditary nonpolyposis colon cancer*	443909	**53**
Hereditary orotic aciduria	30	**≤10**
Hereditary painful callosities	79141	**15**
Hereditary palmoplantar keratoderma, Gamborg-Nielsen type	86923	**≤10**
Hereditary papillary renal cell carcinoma	47044	**12**
Hereditary pediatric Behçet-like disease	476102	**90**
Hereditary persistence of alpha-fetoprotein	168615	**≤10**
Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome	46532	**≤10**
Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	251380	**55**
Hereditary pheochromocytoma-paraganglioma	29072	**821**
Hereditary progressive mucinous histiocytosis	158025	**≤10**
Hereditary pulmonary alveolar proteinosis	264675	**20**
Hereditary renal hypouricemia	94088	**≤10**
Hereditary sensorimotor neuropathy with hyperelastic skin	280598	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Hereditary sensory and autonomic neuropathy due to TECPR2 mutation	320385	**≤10**
Hereditary sensory and autonomic neuropathy type 1	36386	**30**
Hereditary sensory and autonomic neuropathy type 1B	139564	**17**
Hereditary sensory and autonomic neuropathy type 2	970	**17**
Hereditary sensory and autonomic neuropathy type 4	642	**19**
Hereditary sensory and autonomic neuropathy type 5	64752	**22**
Hereditary sensory and autonomic neuropathy with deafness and global delay	139573	**≤10**
Hereditary sensory and autonomic neuropathy*	140471	**92**
Hereditary sensory neuropathy-deafness-dementia syndrome	456318	**≤10**
Hereditary site-specific ovarian cancer syndrome	213524	**28**
Hereditary spherocytosis	822	**1319**
Hereditary stomatocytosis*	98365	**19**
Hereditary thermosensitive neuropathy	84093	**≤10**
Hereditary thrombocytopenia with early-onset myelofibrosis	480851	**≤10**
Hereditary thrombocytopenia with normal platelets	268322	**82**
Hereditary thrombophilia due to congenital antithrombin deficiency	82	**30**
Hereditary xanthinuria	3467	**≤10**
Hermansky-Pudlak syndrome	79430	**84**
Herpes simplex virus encephalitis	1930	**91**
Herpes simplex virus stromal keratitis	137599	**152**
Herpetiform pemphigus	208524	**18**
Hidrotic ectodermal dysplasia	189	**115**
High bone mass osteogenesis imperfecta	314029	**≤10**
High myopia-sensorineural deafness syndrome	363396	**≤10**
High-grade astrocytoma*	251561	**≤10**
Hinman syndrome	84085	**151**
Hip dysplasia, Beukes type	2114	**12**
Hirschsprung disease	388	**1635**
Hirschsprung disease-ganglioneuroblastoma syndrome	2151	**≤10**
Hirschsprung disease-nail hypoplasia-dysmorphism syndrome	2153	**≤10**
His bundle tachycardia	3283	**≤10**
Histidinemia	2157	**≤10**
Histiocytic sarcoma	86896	**20**
Histiocytoid cardiomyopathy	137675	**≤10**
Histoplasmosis	390	**≤10**
HNRNPDL-related limb-girdle muscular dystrophy D3	55596	**≤10**
Hodgkin lymphoma*	98293	**31**
Holmes-Adie syndrome	454718	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Holocarboxylase synthetase deficiency	79242	**11**
Holoprosencephaly	2162	**130**
Holoprosencephaly-craniosynostosis syndrome	2163	**≤10**
Holoprosencephaly-postaxial polydactyly syndrome	2166	**≤10**
Holoprosencephaly-radial heart renal anomalies syndrome	3186	**≤10**
Holt-Oram syndrome	392	**293**
Holzgreve syndrome	2167	**≤10**
Homocystinuria due to methylene tetrahydrofolate reductase deficiency	395	**131**
Homocystinuria without methylmalonic aciduria	622	**35**
Homozygous familial hypercholesterolemia	391665	**54**
Horizontal gaze palsy with progressive scoliosis	2744	**12**
Hot water reflex epilepsy	166412	**≤10**
House allergic alveolitis	99907	**371**
Hoyeraal-Hreidarsson syndrome	3322	**≤10**
HSD10 disease	391417	**≤10**
HTRA1-related autosomal dominant cerebral small vessel disease	482077	**24**
Humeral agenesis/hypoplasia	294973	**≤10**
Humero-radial synostosis	3265	**21**
Humero-radio-ulnar synostosis	3266	**≤10**
Humero-ulnar synostosis	94056	**≤10**
Huntington disease	399	**3102**
Huntington disease-like 1	157941	**≤10**
Huntington disease-like 2	98934	**47**
Huntington disease-like 3	157946	**≤10**
Huntington disease-like syndrome due to C9ORF72 expansions	401901	**≤10**
Huntington disease-like syndrome*	158266	**≤10**
Huriez syndrome	384	**≤10**
Hutchinson-Gilford progeria syndrome	740	**15**
Hyaline body myopathy	53698	**≤10**
Hyaline fibromatosis syndrome	498474	**16**
Hydatidiform mole	99927	**≤10**
Hydranencephaly	2177	**≤10**
Hydroa vacciniforme	330058	**≤10**
Hydroa vacciniforme-like lymphoma	364039	**≤10**
Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome	2180	**≤10**
Hydrocephalus-obesity-hypogonadism syndrome	2183	**≤10**
Hydrocephaly-cerebellar agenesis syndrome	1397	**≤10**
Hydrocephaly-tall stature-joint laxity syndrome	2181	**≤10**
Hydrops fetalis	1041	**30**
Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome	528091	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Hyperammonemia due to N-acetylglutamate synthase deficiency	927	**28**
Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	401948	**≤10**
Hyperandrogenism due to cortisone reductase deficiency	168588	**≤10**
Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	209902	**≤10**
Hyperekplexia*	306773	**12**
Hyperekplexia-epilepsy syndrome	163985	**≤10**
Hypereosinophilic syndrome*	168956	**153**
Hypergonadotropic hypogonadism-cataract syndrome	2410	**≤10**
Hyper-IgE syndrome*	331223	**18**
Hyper-IgM syndrome with susceptibility to opportunistic infections	183663	**35**
Hyper-IgM syndrome without susceptibility to opportunistic infections	183666	**14**
Hyperimmunoglobulinemia D with periodic fever	343	**119**
Hyperinsulinism due to glucokinase deficiency	79299	**≤10**
Hyperinsulinism due to HNF1A deficiency	324575	**≤10**
Hyperinsulinism due to HNF4A deficiency	263455	**≤10**
Hyperinsulinism due to INSR deficiency	263458	**≤10**
Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	71212	**22**
Hyperinsulinism-hyperammonemia syndrome	35878	**43**
Hyperkalemic periodic paralysis	682	**109**
Hyperkeratosis lenticularis perstans	409	**≤10**
Hyperkeratosis-hyperpigmentation syndrome	1336	**≤10**
Hyperlysinemia	2203	**≤10**
Hypermethioninemia due to glycine N-methyltransferase deficiency	289891	**≤10**
Hypermethioninemia encephalopathy due to adenosine kinase deficiency	289290	**≤10**
Hypermobile Ehlers-Danlos syndrome	285	**2814**
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	415	**26**
Hyperostosis corticalis generalisata	3416	**≤10**
Hyperostosis cranialis interna	443098	**≤10**
Hyperparathyroidism-jaw tumor syndrome	99880	**27**
Hyperphalangy	295002	**≤10**
Hyperphenylalaninemia due to DNAJC12 deficiency	508523	**≤10**
Hyperphenylalaninemia due to tetrahydrobiopterin deficiency	238583	**127**
Hyperphosphatasia-intellectual disability syndrome	247262	**≤10**
Hyperprolinemia type 1	419	**20**
Hyperprolinemia type 2	79101	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Hypersensitivity pneumonitis*	31740	**498**
Hypertelorism-hypospadias-polysyndactyly syndrome	2211	**≤10**
Hypertelorism-microtia-facial clefting syndrome	2213	**≤10**
Hypertension due to gain-of-function mutations in the mineralocorticoid receptor	88660	**≤10**
Hypertrichosis lanuginosa congenita	2222	**15**
Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation	324525	**≤10**
Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome	363694	**≤10**
Hyperzincemia and hypercalprotectinemia	251523	**≤10**
Hypnic headache	276429	**≤10**
Hypobetalipoproteinemia*	31154	**17**
Hypocalcemic rickets*	289103	**72**
Hypocalcemic vitamin D-dependent rickets	289157	**134**
Hypocalcemic vitamin D-resistant rickets	93160	**48**
Hypochondroplasia	429	**382**
Hypocomplementemic urticarial vasculitis	36412	**94**
Hypodontia-dysplasia of nails syndrome	2228	**≤10**
Hypoglossia-hypodactyly syndrome	989	**27**
Hypogonadism-mitral valve prolapse-intellectual disability syndrome	2233	**≤10**
Hypogonadotropic hypogonadism-retinitis pigmentosa syndrome	2235	**≤10**
Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome	293967	**≤10**
Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome	528105	**≤10**
Hypohidrotic ectodermal dysplasia	238468	**882**
Hypohidrotic ectodermal dysplasia with immunodeficiency	98813	**≤10**
Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome	1882	**32**
Hypoinsulinemic hypoglycemia and body hemihypertrophy	293964	**≤10**
Hypokalemic periodic paralysis	681	**236**
Hypomandibular faciocranial dysostosis	1790	**≤10**
Hypomyelination neuropathy-arthrogryposis syndrome	2680	**≤10**
Hypomyelination of early myelinating structures	599376	**≤10**
Hypomyelination with atrophy of basal ganglia and cerebellum	139441	**11**
Hypomyelination with brain stem and spinal cord involvement and leg spasticity	363412	**≤10**
Hypomyelination-congenital cataract syndrome	85163	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Hypoparathyroidism-sensorineural deafness-renal disease syndrome	2237	**104**
Hypophosphatasia	436	**447**
Hypophosphatemic rickets*	437	**434**
Hypopigmentation-punctate palmoplantar keratoderma syndrome	324561	**≤10**
Hypoplasia of the mitral valve annulus	99058	**≤10**
Hypoplasminogenemia	722	**≤10**
Hypoplastic left heart syndrome	2248	**205**
Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome	293864	**≤10**
Hypoplastic right heart syndrome*	98723	**≤10**
Hypoplastic tibiae-postaxial polydactyly syndrome	3332	**≤10**
Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome	2250	**≤10**
Hypospadias-intellectual disability, Goldblatt type syndrome	2261	**≤10**
Hypothalamic adipsic hypernatraemia syndrome	443101	**≤10**
Hypothalamic hamartomas with gelastic seizures	86906	**58**
Hypothyroidism due to deficient transcription factors involved in pituitary development or function	226307	**≤10**
Hypothyroidism due to TSH receptor mutations	90673	**67**
Hypotonia-cystinuria syndrome	163690	**≤10**
Hypotonia-cystinuria type 1 syndrome*	238517	**≤10**
Hypotonia-failure to thrive-microcephaly syndrome	79507	**60**
Hypotonia-speech impairment-severe cognitive delay syndrome	371364	**11**
Hypotrichosis simplex	55654	**31**
Hypotrichosis simplex of the scalp	90368	**≤10**
Hypotrichosis with juvenile macular degeneration	1573	**≤10**
Hypotrichosis-deafness syndrome	330029	**≤10**
Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	69735	**≤10**
Hypoxanthine guanine phosphoribosyltransferase partial deficiency	79233	**≤10**
ICF syndrome	2268	**17**
Ichthyosis follicularis-alopecia-photophobia syndrome	2273	**17**
Ichthyosis hystrix of Curth-Macklin	79503	**≤10**
Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome	2269	**≤10**
Ichthyosis-hypotrichosis syndrome	91132	**≤10**
Ichthyosis-prematurity syndrome	88621	**16**
Ichthyosis-short stature-brachydactyly-microspherophakia syndrome	363992	**≤10**
Idiopathic achalasia	930	**86**
Idiopathic acute eosinophilic pneumonia	724	**11**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Idiopathic anterior uveitis	280914	**1148**
Idiopathic aplastic anemia	88	**828**
Idiopathic bilateral vestibulopathy	171684	**≤10**
Idiopathic bronchiectasis	60033	**1141**
Idiopathic camptocormia	1320	**216**
Idiopathic CD4 lymphocytopenia	228000	**63**
Idiopathic chronic eosinophilic pneumonia	2902	**119**
Idiopathic congenital hypothyroidism	95717	**574**
Idiopathic dropped head syndrome	447881	**16**
Idiopathic ductopenia	480512	**20**
Idiopathic eosinophilic myositis	247724	**≤10**
Idiopathic eosinophilic pneumonia*	182101	**20**
Idiopathic gastroparesis	558411	**127**
Idiopathic giant cell myocarditis	329874	**45**
Idiopathic hemiconvulsion-hemiplegia syndrome	86908	**54**
Idiopathic hypercalciuria	2197	**934**
Idiopathic hypereosinophilic syndrome	3260	**532**
Idiopathic hypersomnia	33208	**2885**
Idiopathic interstitial pneumonia*	98300	**368**
Idiopathic intracranial hypertension	238624	**34**
Idiopathic isolated micropenis	95707	**533**
Idiopathic juvenile osteoporosis	85193	**669**
Idiopathic localized lipodystrophy	90158	**≤10**
Idiopathic macular telangiectasia type 1	353344	**11**
Idiopathic macular telangiectasia type 3	353351	**≤10**
Idiopathic neonatal atrial flutter	45452	**≤10**
Idiopathic nephrotic syndrome*	357502	**846**
Idiopathic non-lupus full-house nephropathy	567544	**≤10**
Idiopathic panuveitis	280921	**748**
Idiopathic peliosis hepatis	480524	**139**
Idiopathic phalangeal acro-osteolysis	444316	**≤10**
Idiopathic pleuroparenchymal fibroelastosis	494428	**179**
Idiopathic posterior uveitis	280917	**423**
Idiopathic pulmonary artery dilatation	1676	**≤10**
Idiopathic pulmonary fibrosis	2032	**4312**
Idiopathic pulmonary hemosiderosis	99931	**54**
Idiopathic recurrent pericarditis	251307	**319**
Idiopathic spontaneous coronary artery dissection	458718	**81**
Idiopathic steroid-resistant nephrotic syndrome	567548	**246**
Idiopathic steroid-sensitive nephrotic syndrome	69061	**3887**
Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance	567546	**36**
Idiopathic trachyonychia	79153	**≤10**
Idiopathic uveal effusion syndrome	209956	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Idiopathic ventricular fibrillation, non Brugada type	228140	**666**
Idiopathic/heritable pulmonary arterial hypertension	422	**2931**
IgA pemphigus	555905	**16**
IgG4-related disease*	284264	**269**
IgG4-related systemic disease	596448	**801**
Ileal neuroendocrine tumor	100078	**≤10**
IMAGe syndrome	85173	**12**
Imerslund-Gräsbeck syndrome	35858	**50**
Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome	238569	**640**
Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections-lymphopenia syndrome	529977	**≤10**
Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	37042	**58**
Immune thrombocytopenia	3002	**7145**
Immune-mediated necrotizing myopathy	206569	**609**
Immunodeficiency by defective expression of MHC class I	34592	**≤10**
Immunodeficiency by defective expression of MHC class II	572	**30**
Immunodeficiency due to a classical component pathway complement deficiency	169147	**43**
Immunodeficiency due to a late component of complement deficiency	169150	**40**
Immunodeficiency due to CD25 deficiency	169100	**≤10**
Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	70592	**≤10**
Immunodeficiency due to selective anti-polysaccharide antibody deficiency	70593	**74**
Immunodeficiency with factor H anomaly	200421	**≤10**
Immunodeficiency with factor I anomaly	200418	**≤10**
Immunoglobulin A vasculitis	761	**2548**
Immunoglobulin heavy chain deficiency	169110	**≤10**
Immunotactoid glomerulopathy	97567	**≤10**
Immunotactoid or fibrillary glomerulopathy*	91137	**≤10**
Incessant infant ventricular tachycardia	45453	**≤10**
Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	52430	**28**
Inclusion body myositis	611	**1099**
Incontinentia pigmenti	464	**629**
Indeterminate cell histiocytosis	158019	**≤10**
Indolent primary cutaneous T-cell lymphoma	178548	**≤10**
Indolent systemic mastocytosis	98848	**1408**
Infant acute respiratory distress syndrome	70587	**37**
Infantile apnea	70590	**1685**
Infantile bilateral striatal necrosis*	1576	**≤10**
Infantile cerebellar-retinal degeneration	313850	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	402364	**≤10**
Infantile choroidocerebral calcification syndrome	1313	**≤10**
Infantile convulsions and choreoathetosis	31709	**12**
Infantile digital fibromatosis	199267	**≤10**
Infantile dystonia-parkinsonism	238455	**12**
Infantile epileptic-dyskinetic encephalopathy	364063	**33**
Infantile hemangioma of rare localization*	210589	**1923**
Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency	352563	**≤10**
Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome	522077	**≤10**
Infantile multisystem neurologic-endocrine-pancreatic disease	456312	**≤10**
Infantile myofibromatosis	2591	**16**
Infantile neuroaxonal dystrophy	35069	**62**
Infantile neuronal ceroid lipofuscinosis	79263	**31**
Infantile neurovisceral acid sphingomyelinase deficiency	77292	**29**
Infantile onset panniculitis with uveitis and systemic granulomatosis	251304	**≤10**
Infantile osteopetrosis with neuroaxonal dysplasia	85179	**≤10**
Infantile Refsum disease	772	**13**
Infantile spasms syndrome	3451	**1849**
Infantile spasms-broad thumbs syndrome	3173	**≤10**
Infantile-onset ascending hereditary spastic paralysis	293168	**≤10**
Infantile-onset autosomal recessive nonprogressive cerebellar ataxia	284332	**12**
Infantile-onset axonal motor and sensory neuropathy-optic atrophy-neurodegenerative syndrome	457205	**≤10**
Infantile-onset generalized dyskinesia with orofacial involvement	494526	**≤10**
Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression	391316	**≤10**
Infantile-onset periodic fever-panniculitis-dermatosis syndrome	500062	**≤10**
Infantile-onset pulmonary alveolar proteinosis-hypogammaglobulinemia	572428	**≤10**
Infantile-onset spinocerebellar ataxia	1186	**64**
Infantile-onset X-linked spinal muscular atrophy	1145	**12**
Infection-related hemolytic uremic syndrome	544482	**1770**
Infectious anterior uveitis	279922	**126**
Infectious epithelial keratitis	137593	**66**
Infectious panuveitis	279925	**50**
Infectious posterior uveitis	279919	**156**
Infective endocarditis	570762	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Inferior vena cava interruption without azygos continuation	99123	**≤10**
Inflammatory bowel disease-recurrent sinopulmonary infections syndrome	529980	**≤10**
Inflammatory myofibroblastic tumor	178342	**15**
Inflammatory myopathy with abundant macrophages	247718	**≤10**
Inflammatory pseudotumor of the liver	90003	**12**
Infundibulo-neurohypophysitis	238305	**≤10**
Inherited acute myeloid leukemia	319465	**≤10**
Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations	319462	**14**
Inherited congenital spastic tetraplegia	210141	**20**
Inherited Creutzfeldt-Jakob disease	282166	**≤10**
Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency	289548	**≤10**
Insulin autoimmune syndrome	411593	**≤10**
Insulinoma	97279	**23**
Insulin-resistance syndrome type A	2297	**22**
Insulin-resistance syndrome type B	2298	**≤10**
Intellectual disability, Birk-Barel type	166108	**≤10**
Intellectual disability, Wolff type	3080	**≤10**
Intellectual disability-alacrima-achalasia syndrome	289483	**≤10**
Intellectual disability-autism-speech apraxia-craniofacial dysmorphism syndrome	529965	**≤10**
Intellectual disability-brachydactyly-Pierre Robin syndrome	364577	**≤10**
Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome	508498	**≤10**
Intellectual disability-cataracts-calcified pinnae-myopathy syndrome	3042	**32**
Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	397709	**≤10**
Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	329224	**17**
Intellectual disability-developmental delay-contractures syndrome	3454	**16**
Intellectual disability-dysmorphism-hypogonadism-diabetes mellitus syndrome	3044	**≤10**
Intellectual disability-epilepsy-extrapyramidal syndrome	468620	**≤10**
Intellectual disability-expressive aphasia-facial dysmorphism syndrome	436151	**≤10**
Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	404440	**57**
Intellectual disability-facial dysmorphism-hand anomalies syndrome	370010	**≤10**
Intellectual disability-hypoplastic corpus callosum-preauricular tag syndrome	1495	**18**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome	457279	**13**
Intellectual disability-muscle weakness-short stature-facial dysmorphism syndrome	457365	**≤10**
Intellectual disability-myopathy-short stature-endocrine defect syndrome	3068	**≤10**
Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome	352530	**≤10**
Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome	513456	**18**
Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	369837	**≤10**
Intellectual disability-seizures-macrocephaly-obesity syndrome	369950	**≤10**
Intellectual disability-severe speech delay-mild dysmorphism syndrome	391372	**54**
Intellectual disability-short stature-hypertelorism syndrome	3074	**≤10**
Intellectual disability-spasticity-ectrodactyly syndrome	1891	**≤10**
Intellectual disability-strabismus syndrome	363528	**22**
Interatrial communication	1478	**932**
Intercalary limb defects*	294927	**≤10**
Intermediate atrioventricular septal defect	576242	**115**
Intermediate generalized junctional epidermolysis bullosa	79402	**11**
Intermediate nemaline myopathy	171433	**13**
Intermediate osteopetrosis	210110	**≤10**
Intermediate uveitis	279914	**445**
Internal carotid absence	981	**≤10**
Interstitial cystitis	37202	**≤10**
Interstitial granulomatous dermatitis with arthritis	79099	**≤10**
Interstitial lung disease due to ABCA3 deficiency	440402	**15**
Interstitial lung disease due to SP-C deficiency	440392	**48**
Interventricular septum aneurysm	99092	**≤10**
Intestinal lymphangiectasia*	36204	**27**
Intestinal polyposis syndrome*	104010	**62**
Intraductal papillary mucinous carcinoma of pancreas	424058	**38**
Intrahepatic cholestasis of pregnancy	69665	**115**
Intraneural perineurioma	100003	**≤10**
Intrauterine growth restriction-congenital multiple café-au-lait macules-increased sister chromatid exchange syndrome	508512	**16**
Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	436144	**≤10**
Intravascular large B-cell lymphoma	98839	**≤10**
Invasive mole	99925	**≤10**
Invasive non-typhoidal salmonellosis	324648	**≤10**
Inverse Klippel-Trénaunay syndrome	329324	**12**
Inverted duplicated chromosome 15 syndrome	3306	**105**
IRF2BPL-related regressive neurodevelopmental disorder-dystonia-seizures syndrome	597623	**≤10**
IRIDA syndrome	209981	**20**
Iridocorneal endothelial syndrome	64734	**≤10**
IRVAN syndrome	209943	**≤10**
Isaacs syndrome	84142	**120**
Ischiovertebral syndrome	85200	**≤10**
Isochromosomy Yp	98797	**≤10**
Isochromosomy Yq	98798	**≤10**
Isolated agammaglobulinemia	229717	**386**
Isolated agenesis of gallbladder	440987	**≤10**
Isolated amyelia	268868	**≤10**
Isolated anencephaly/exencephaly	1048	**≤10**
Isolated aniridia	250923	**490**
Isolated ankyloblepharon filiforme adnatum	91397	**≤10**
Isolated anterior cervical hypertrichosis	3387	**≤10**
Isolated arrhinia	1134	**≤10**
Isolated asymptomatic elevation of creatine phosphokinase	206599	**596**
Isolated ATP synthase deficiency	254913	**≤10**
Isolated autosomal dominant hypomagnesemia, Glaudemans type	199326	**≤10**
Isolated bilateral hemispheric cerebellar hypoplasia	269221	**≤10**
Isolated biliary atresia	30391	**1506**
Isolated blepharochalasis	519390	**≤10**
Isolated bone marrow mastocytosis	158778	**≤10**
Isolated cerebellar agenesis	1398	**249**
Isolated cerebellar vermis agenesis	269203	**≤10**
Isolated cerebellar vermis hypoplasia	199630	**75**
Isolated childhood apraxia of speech	209908	**67**
Isolated cleft lip	199302	**524**
Isolated complex I deficiency	2609	**77**
Isolated complex III deficiency	1460	**36**
Isolated congenital alacrima	91416	**12**
Isolated congenital anonychia	79143	**≤10**
Isolated congenital anosmia	88620	**≤10**
Isolated congenital auditory ossicle malformation	162526	**75**
Isolated congenital breast hypoplasia/aplasia	180188	**22**
Isolated congenital digital clubbing	217059	**≤10**
Isolated congenital ectropion	99171	**≤10**
Isolated congenital hepatic fibrosis	485426	**193**
Isolated congenital hypoglossia/aglossia	141152	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Isolated congenital hypogonadotropic hypogonadism	238666	**2034**
Isolated congenital megalocornea	91489	**38**
Isolated congenital microcephaly	199642	**465**
Isolated congenital nasal pyriform aperture stenosis	162516	**135**
Isolated congenital onychodysplasia	79144	**≤10**
Isolated congenital radial head dislocation	295032	**≤10**
Isolated congenital sclerocornea	91490	**40**
Isolated congenital syngnathia	141214	**≤10**
Isolated corpus callosum agenesis	200	**629**
Isolated cryptophthalmia	91396	**≤10**
Isolated cytochrome C oxidase deficiency	254905	**29**
Isolated Dandy-Walker malformation	217	**89**
Isolated delta-storage pool disease	248340	**105**
Isolated diffuse palmoplantar keratoderma*	307148	**67**
Isolated distichiasis	99177	**≤10**
Isolated ectopia lentis	1885	**149**
Isolated encephalocele	199647	**19**
Isolated focal cortical dysplasia	65683	**1148**
Isolated focal non-epidermolytic palmoplantar keratoderma	448264	**≤10**
Isolated focal palmoplantar keratoderma*	307846	**29**
Isolated follicle stimulating hormone deficiency	52901	**≤10**
Isolated foveal hypoplasia	519398	**60**
Isolated generalized anhidrosis with normal sweat glands	468666	**≤10**
Isolated glycerol kinase deficiency	408	**≤10**
Isolated hemihyperplasia	2128	**272**
Isolated hereditary congenital facial paralysis	306527	**21**
Isolated Klippel-Feil syndrome	2345	**149**
Isolated lissencephaly type 1 without known genetic defects	1084	**24**
Isolated megalopapilla	519402	**≤10**
Isolated mesenteric vein thrombosis	583861	**24**
Isolated microphthalmia-anophthalmia-coloboma*	2542	**138**
Isolated microspherophakia	519396	**≤10**
Isolated neonatal sclerosing cholangitis	480556	**31**
Isolated optic nerve hypoplasia/aplasia	137902	**32**
Isolated optic neuritis	499096	**200**
Isolated osteopoikilosis	166119	**≤10**
Isolated partial vaginal agenesis	96269	**12**
Isolated permanent neonatal diabetes mellitus	99885	**63**
Isolated Pierre Robin syndrome	718	**2072**
Isolated polycystic liver disease	2924	**467**
Isolated pulmonary capillaritis	264691	**≤10**
Isolated punctate palmoplantar keratoderma*	2338	**24**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Isolated right ventricular hypoplasia	439	**25**
Isolated spina bifida*	823	**504**
Isolated splenic vein thrombosis	583856	**≤10**
Isolated split hand-split foot malformation	2440	**373**
Isolated succinate-CoQ reductase deficiency	3208	**15**
Isolated thyroid-stimulating hormone deficiency	90674	**113**
Isolated thyrotropin-releasing hormone deficiency	238670	**≤10**
Isolated tracheoesophageal fistula	454750	**37**
Isolated unilateral hemispheric cerebellar hypoplasia	269218	**13**
Isotretinoin syndrome	2305	**≤10**
Isotretinoin-like syndrome	2306	**≤10**
Isovaleric acidemia	33	**98**
ISPD-related limb-girdle muscular dystrophy R20	352479	**≤10**
ITM2B amyloidosis	439254	**12**
Jackson-Weiss syndrome	1540	**≤10**
Jacobsen syndrome	2308	**75**
Jalili syndrome	1873	**≤10**
Jeavons syndrome	139431	**142**
Jervell and Lange-Nielsen syndrome	90647	**26**
Jessner lymphocytic infiltration of the skin	33314	**≤10**
Jeune syndrome	474	**93**
Johanson-Blizzard syndrome	2315	**≤10**
Johnson neuroectodermal syndrome	2316	**≤10**
Joubert syndrome	475	**436**
Joubert syndrome with hepatic defect	1454	**≤10**
Joubert syndrome with Jeune asphyxiating thoracic dystrophy	397715	**≤10**
Joubert syndrome with ocular defect	220493	**18**
Joubert syndrome with oculorenal defect	2318	**15**
Joubert syndrome with renal defect	220497	**14**
Junctional epidermolysis bullosa inversa	79405	**≤10**
Junctional epidermolysis bullosa with pyloric atresia	79403	**≤10**
Junctional epidermolysis bullosa*	305	**100**
Juvenile absence epilepsy	1941	**572**
Juvenile amyotrophic lateral sclerosis	300605	**18**
Juvenile dermatomyositis	93672	**320**
Juvenile glaucoma	98977	**73**
Juvenile Huntington disease	248111	**32**
Juvenile idiopathic arthritis*	92	**782**
Juvenile myelomonocytic leukemia	86834	**14**
Juvenile myoclonic epilepsy	307	**881**
Juvenile nasopharyngeal angiofibroma	289596	**18**
Juvenile neuronal ceroid lipofuscinosis	79264	**63**
Juvenile overlap myositis	329894	**≤10**
Juvenile Paget disease	2801	**19**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Juvenile polymyositis	93568	**≤10**
Juvenile polyposis syndrome	2929	**58**
Juvenile primary lateral sclerosis	247604	**≤10**
Juvenile temporal arteritis	26137	**≤10**
Juvenile xanthogranuloma	158000	**57**
Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	445062	**≤10**
Kabuki syndrome	2322	**534**
Kagami-Ogata syndrome	254519	**≤10**
Kallmann syndrome-heart disease syndrome	2326	**14**
Kaposi sarcoma	33276	**25**
Kaposiform hemangioendothelioma	2122	**26**
Karyomegalic interstitial nephritis	401996	**≤10**
Kasabach-Merritt syndrome	2330	**27**
Kawasaki disease	2331	**663**
KBG syndrome	2332	**436**
KCNQ2-related epileptic encephalopathy	439218	**114**
KDM5C-related syndromic X-linked intellectual disability	85279	**24**
Kearns-Sayre syndrome	480	**193**
Keipert syndrome	2662	**≤10**
Kennedy disease	481	**277**
Kenny-Caffey syndrome	2333	**13**
Keratinopathic ichthyosis*	281103	**34**
Keratocystic odontogenic tumor	447777	**≤10**
Keratoderma hereditarium mutilans	494	**≤10**
Keratoderma hereditarium mutilans with ichthyosis	79395	**≤10**
Keratosis follicularis spinulosa decalvans	2340	**≤10**
Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome	281201	**≤10**
Keratosis pilaris atrophicans*	498	**14**
Ketoacidosis due to monocarboxylate transporter-1 deficiency	438075	**≤10**
Keutel syndrome	85202	**≤10**
KID syndrome	477	**29**
Kidney tubulopathy-dilated cardiomyopathy syndrome	73224	**≤10**
Kienbock disease	97332	**≤10**
Kikuchi-Fujimoto disease	50918	**55**
Kimura disease	482	**15**
Kindler epidermolysis bullosa	2908	**26**
King-Denborough syndrome	99741	**≤10**
Kleefstra syndrome	261494	**176**
Kleine-Levin syndrome	33543	**304**
Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome	447974	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Kniest dysplasia	485	**29**
Knobloch syndrome	1571	**24**
Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome	2698	**≤10**
Kommerell diverticulum	99077	**≤10**
Koolen-De Vries syndrome	96169	**92**
Kosaki overgrowth syndrome	477831	**≤10**
Kostmann syndrome	99749	**≤10**
Krabbe disease	487	**97**
KRT1-related diffuse nonepidermolytic keratoderma	530838	**≤10**
Kufor-Rakeb syndrome	306674	**≤10**
Kuskokwim syndrome	1149	**≤10**
Kyphoscoliotic Ehlers-Danlos syndrome	536545	**63**
L1 syndrome	275543	**108**
L-2-hydroxyglutaric aciduria	79314	**25**
Lacrimoauriculodentodigital syndrome	2363	**40**
Lafora disease	501	**15**
Laing early-onset distal myopathy	59135	**58**
LAMA5-related multisystemic syndrome	521450	**≤10**
Lambert syndrome	1296	**≤10**
Lambert-Eaton myasthenic syndrome	43393	**182**
Lamb-Shaffer syndrome	530983	**47**
Lamellar ichthyosis	313	**361**
Laminin subunit alpha 2-related congenital muscular dystrophy	258	**202**
Landau-Kleffner syndrome	98818	**47**
Langer mesomelic dysplasia	2632	**≤10**
Langerhans cell histiocytosis	389	**1506**
Langerhans cell sarcoma	86897	**≤10**
Large congenital melanocytic nevus	626	**1210**
Laron syndrome	633	**42**
Laron syndrome with immunodeficiency	220465	**≤10**
Larsen syndrome	503	**75**
Larsen-like osseous dysplasia-short stature syndrome	2370	**≤10**
Larsen-like syndrome, B3GAT3 type	284139	**≤10**
Laryngeal abductor paralysis	2808	**16**
Laryngeal abductor paralysis-intellectual disability syndrome	2375	**≤10**
Laryngeal neuroendocrine tumor	100083	**≤10**
Laryngocele	2372	**≤10**
Laryngo-onycho-cutaneous syndrome	2407	**≤10**
Laryngotracheal angioma	137935	**44**
Laryngotracheoesophageal cleft	2004	**86**
Larynx atresia	1202	**32**
Late infantile neuronal ceroid lipofuscinosis	168491	**55**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Late-onset distal myopathy, Markesbery-Griggs type	98912	**24**
Late-onset focal dermal elastosis	228227	**≤10**
Late-onset isolated ACTH deficiency	199299	**222**
Late-onset junctional epidermolysis bullosa	79406	**≤10**
Late-onset retinal degeneration	67042	**11**
Lateral facial cleft*	141269	**13**
Lateral meningocele syndrome	2789	**≤10**
Lattice corneal dystrophy type I	98964	**18**
Laubry-Pezzi syndrome	99094	**143**
Laurence-Moon syndrome	2377	**≤10**
Laurin-Sandrow syndrome	2378	**≤10**
LCAT deficiency	650	**≤10**
Lead poisoning	330015	**≤10**
Leber congenital amaurosis	65	**501**
Leber hereditary optic neuropathy	104	**921**
Leber plus disease	99718	**54**
Ledderhose disease	199251	**≤10**
Left sided atrial isomerism	566862	**≤10**
Left ventricular noncompaction	54260	**797**
Legg-Calvé-Perthes disease	2380	**26**
Legionnaires disease	549	**≤10**
Legius syndrome	137605	**105**
Leigh syndrome with cardiomyopathy	70474	**≤10**
Leigh syndrome with leukodystrophy	255241	**16**
Leigh syndrome*	506	**135**
Leiomyosarcoma	64720	**22**
Leiomyosarcoma of the cervix uteri	213807	**≤10**
Leiomyosarcoma of the corpus uteri	213625	**≤10**
Leishmaniasis	507	**≤10**
Lemierre syndrome	137839	**≤10**
Lennox-Gastaut syndrome	2382	**976**
Lenz-Majewski hyperostotic dwarfism	2658	**≤10**
Leprechaunism	508	**≤10**
Leprosy	548	**33**
Leptomyelolipoma	268838	**49**
Leptospirosis	509	**≤10**
Léri-Weill dyschondrosteosis	240	**872**
Lesch-Nyhan syndrome	510	**62**
Lethal ataxia with deafness and optic atrophy	1187	**≤10**
Lethal congenital contracture syndrome	294965	**≤10**
Lethal congenital contracture syndrome type 1	1486	**≤10**
Lethal hemolytic anemia-genital anomalies syndrome	1046	**≤10**
Lethal infantile mitochondrial myopathy	254857	**25**
Lethal intrauterine growth restriction-cortical malformation-congenital contractures syndrome	2570	**≤10**
Lethal Larsen-like syndrome	2371	**≤10**
Lethal neonatal spasticity-epileptic encephalopathy syndrome	435845	**≤10**
Lethal occipital encephalocele-skeletal dysplasia syndrome	293925	**≤10**
Lethal osteosclerotic bone dysplasia	1832	**≤10**
Lethal polymalformative syndrome, Boissel type	210144	**12**
Lethal recessive chondrodysplasia	1423	**≤10**
Leukocyte adhesion deficiency	2968	**12**
Leukoencephalopathy with bilateral anterior temporal lobe cysts	139444	**≤10**
Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	137898	**≤10**
Leukoencephalopathy with calcifications and cysts	542310	**≤10**
Leukoencephalopathy with mild cerebellar ataxia and white matter edema	363540	**≤10**
Leukoencephalopathy-palmoplantar keratoderma syndrome	2386	**≤10**
Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome	314051	**≤10**
Leukonychia totalis	2387	**≤10**
Leukonychia totalis-acanthosis-nigricans-like lesions-abnormal hair syndrome	210133	**≤10**
Levocardia	95854	**≤10**
Leydig cell hypoplasia	755	**25**
L-ferritin deficiency	440731	**≤10**
Lhermitte-Duclos disease	65285	**≤10**
Lichen amyloidosis	49804	**≤10**
Lichen myxedematosus*	402007	**≤10**
Lichen planopilaris	525	**≤10**
Lichen planus pemphigoides	254478	**97**
Lichen planus pigmentosus	254463	**≤10**
Liddle syndrome	526	**32**
Li-Fraumeni syndrome	524	**53**
LIG4 syndrome	99812	**≤10**
Limbal stem cell deficiency	171673	**17**
Limb-girdle muscular dystrophy due to POMK deficiency	445110	**≤10**
Limb-girdle muscular dystrophy*	263	**637**
Limb-mammary syndrome	69085	**≤10**
Linear and whorled nevoid hypermelanosis	79150	**16**
Linear atrophoderma of Moulin	140933	**≤10**
Linear hypopigmentation and craniofacial asymmetry with acral, ocular and brain anomalies	589608	**≤10**
Linear IgA dermatosis	46488	**173**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Linear lichen planus	254379	**15**
Linear nevus sebaceus syndrome	2612	**183**
Linear verrucous nevus syndrome	2611	**206**
LIPE-related familial partial lipodystrophy	435660	**≤10**
Lipoblastoma	247762	**≤10**
Lipodystrophy due to peptidic growth factors deficiency	1979	**≤10**
Lipodystrophy-intellectual disability-deafness syndrome	50811	**≤10**
Lipoid proteinosis	530	**14**
Lipoma associated with neurospinal dysraphism*	268832	**146**
Lipomyelomeningocele	268835	**137**
Lipoprotein glomerulopathy	329481	**≤10**
Liposarcoma	69078	**26**
Lisch epithelial corneal dystrophy	98955	**≤10**
Lissencephaly due to LIS1 mutation	95232	**40**
Lissencephaly due to TUBA1A mutation	171680	**11**
Lissencephaly type 1 due to doublecortin gene mutation	2148	**56**
Lissencephaly type 3*	102011	**≤10**
Lissencephaly type 3-metacarpal bone dysplasia syndrome	86822	**≤10**
Lissencephaly with cerebellar hypoplasia type A	100011	**≤10**
Lissencephaly with cerebellar hypoplasia*	86823	**≤10**
Listeriosis	533	**≤10**
Livedoid vasculopathy	542643	**14**
Liver adenomatosis	566841	**≤10**
LMNA-related cardiocutaneous progeria syndrome	363618	**≤10**
Localized dystrophic epidermolysis bullosa	595356	**17**
Localized epidermolysis bullosa simplex	79400	**41**
Localized junctional epidermolysis bullosa	251393	**≤10**
Localized lichen myxedematosus*	86795	**≤10**
Localized lipodystrophy*	79088	**11**
Localized scleroderma	90289	**244**
Locked-in syndrome	2406	**≤10**
Loeys-Dietz syndrome	60030	**537**
Logopenic progressive aphasia	250831	**61**
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	5	**75**
Loose anagen syndrome	168	**≤10**
Low phospholipid-associated cholelithiasis	69663	**1262**
Lowe-Kohn-Cohen syndrome	2408	**≤10**
Lower limb hypertrophy	295051	**25**
Lower limb malformation-hypospadias syndrome	2487	**≤10**
Lower lip fistula	141064	**12**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Lower motor neuron syndrome with late-adult onset	276435	**35**
Low-grade astrocytoma*	251592	**≤10**
Lown-Ganong-Levine syndrome	844	**≤10**
Lowry-MacLean syndrome	2409	**≤10**
Lowry-Wood syndrome	1824	**≤10**
LRP5-related primary osteoporosis	498481	**≤10**
Lujan-Fryns syndrome	776	**72**
LUMBAR syndrome	83628	**39**
Lung agenesis-heart defect-thumb anomalies syndrome	1120	**≤10**
Lupus erythematosus panniculitis	90285	**26**
Lupus erythematosus tumidus	90283	**61**
Luscan-Lumish syndrome	597738	**≤10**
Lyme disease	91546	**173**
Lymphangioleiomyomatosis	538	**462**
Lymphatic filariasis	2035	**≤10**
Lymphedema-distichiasis syndrome	33001	**56**
Lymphoid interstitial pneumonia	79128	**151**
Lymphomatoid granulomatosis	86869	**≤10**
Lymphomatoid papulosis	98842	**≤10**
Lymphoproliferative syndrome*	238510	**13**
Lynch syndrome	144	**247**
Lysinuric protein intolerance	470	**49**
Lysosomal acid lipase deficiency	275761	**43**
Macrocephaly-developmental delay syndrome	397612	**41**
Macrocephaly-intellectual disability-autism syndrome	210548	**31**
Macrocephaly-intellectual disability-left ventricular non compaction syndrome	466791	**≤10**
Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	457485	**24**
Macrocephaly-short stature-paraplegia syndrome	2427	**≤10**
Macrocephaly-spastic paraplegia-dysmorphism syndrome	2429	**13**
Macrocystic lymphatic malformation	79489	**415**
Macrodactyly of fingers	295044	**14**
Macrodactyly of toes	295047	**23**
Macrophage activation syndrome	158061	**81**
Macrophagic myofasciitis	592	**514**
Macrosomia-microphthalmia-cleft palate syndrome	2432	**≤10**
Macrostomia-preauricular tags-external ophthalmoplegia syndrome	83619	**≤10**
Macrothrombocytopenia with mitral valve insufficiency	220448	**≤10**
Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome	487796	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Macular coloboma-cleft palate-hallux valgus syndrome	91494	**≤10**
Macular corneal dystrophy	98969	**15**
Maculopapular cutaneous mastocytosis	79457	**608**
Madras motor neuron disease	137867	**≤10**
Maffucci syndrome	163634	**17**
MAGEL2-related Prader-Willi-like syndrome	398069	**25**
MAGIC syndrome	324972	**21**
Majeed syndrome	77297	**≤10**
Mal de débarquement	210272	**≤10**
Mal de Meleda	87503	**20**
Malakoplakia	556	**≤10**
Malan overgrowth syndrome	420179	**30**
Malaria	673	**15**
Male hypergonadotropic hypogonadism-intellectual disability-skeletal anomalies syndrome	2234	**≤10**
Male infertility with azoospermia or oligozoospermia due to single gene mutation	399805	**53**
Male infertility with teratozoospermia due to single gene mutation	399808	**14**
Maligant granulosa cell tumor of the ovary	99915	**17**
Malignant atrophic papulosis	679	**≤10**
Malignant dysgerminomatous germ cell tumor of the ovary	99912	**≤10**
Malignant epithelial tumor of salivary glands	276145	**≤10**
Malignant germ cell tumor of the cervix uteri	213837	**≤10**
Malignant germ cell tumor of the vagina	206489	**≤10**
Malignant hyperthermia of anesthesia	423	**285**
Malignant melanoma of the mucosa	168999	**17**
Malignant migrating focal seizures of infancy	293181	**50**
Malignant non-dysgerminomatous germ cell tumor of ovary	206538	**≤10**
Malignant peripheral nerve sheath tumor	3148	**16**
Malignant peritoneal mesothelioma	168811	**≤10**
Malignant Sertoli-Leydig cell tumor of the ovary	99916	**15**
Malignant teratoma of ovary	398987	**≤10**
Malonic aciduria	943	**12**
Malposition of a coronary ostium	99090	**≤10**
MALT lymphoma	52417	**33**
MAN1B1-CDG	397941	**≤10**
Mandibular arteriovenous malformation	141174	**39**
Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome	363649	**≤10**
Mandibuloacral dysplasia	2457	**≤10**
Mandibulofacial dysostosis*	155899	**68**
Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome	357158	**≤10**
Mandibulofacial dysostosis-microcephaly syndrome	79113	**65**
Mantle cell lymphoma	52416	**15**
Maple syrup urine disease	511	**205**
Marburg acute multiple sclerosis	228157	**30**
Marcus-Gunn syndrome	91412	**38**
Marden-Walker syndrome	2461	**≤10**
Marfan syndrome	558	**7621**
Marfanoid habitus-autosomal recessive intellectual disability syndrome	2463	**30**
Marfanoid syndrome, De Silva type	2464	**≤10**
Marginal zone lymphoma*	300912	**11**
Marie Unna hereditary hypotrichosis	444	**≤10**
Marinesco-Sjögren syndrome	559	**20**
Marshall syndrome	560	**54**
Marshall-Smith syndrome	561	**14**
Martinique crinkled retinal pigment epitheliopathy	466718	**≤10**
Mast cell leukemia	98851	**≤10**
Mast cell sarcoma	66661	**≤10**
Maternal phenylketonuria	2209	**38**
Maternal uniparental disomy of chromosome 16	96185	**≤10**
Maternal uniparental disomy of chromosome 20	96186	**≤10**
Maternal uniparental disomy of chromosome 21	96187	**≤10**
Maternally-inherited diabetes and deafness	225	**302**
Matthew-Wood syndrome	2470	**≤10**
Maxillary arteriovenous malformation	141171	**40**
Maxillonasal dysplasia	1248	**170**
Mayer-Rokitansky-Küster-Hauser syndrome	3109	**705**
Mazabraud syndrome	57782	**16**
McCune-Albright syndrome	562	**464**
McDonough syndrome	2471	**≤10**
McKusick-Kaufman syndrome	2473	**≤10**
McLeod neuroacanthocytosis syndrome	59306	**13**
Meacham syndrome	3097	**≤10**
Meckel syndrome	564	**13**
Meconium aspiration syndrome	70588	**28**
Medial condensing osteitis of the clavicle	57196	**≤10**
Median cleft lip/mandibule	2006	**≤10**
Median cleft of the upper lip and maxilla	141239	**47**
Median facial cleft*	141234	**≤10**
Median nodule of the upper lip	2699	**≤10**
Medium chain acyl-CoA dehydrogenase deficiency	42	**259**
Medullary sponge kidney	1309	**189**
Medullary thyroid carcinoma	1332	**70**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Medulloblastoma	616	**204**
Medulloepithelioma of the central nervous system	251883	**≤10**
Meesmann corneal dystrophy	98954	**≤10**
Mega-cisterna magna	97252	**≤10**
Megaconial congenital muscular dystrophy	280671	**14**
Megacystis-megaureter syndrome	238637	**49**
Megacystis-microcolon-intestinal hypoperistalsis syndrome	2241	**≤10**
Megalencephalic leukoencephalopathy with subcortical cysts	2478	**36**
Megalencephaly	2477	**37**
Megalencephaly-capillary malformation-polymicrogyria syndrome	60040	**127**
Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	83473	**22**
Megalencephaly-severe kyphoscoliosis-overgrowth syndrome	457359	**≤10**
Megalocornea-intellectual disability syndrome	2479	**≤10**
MEGDEL syndrome	352328	**≤10**
MEHMO syndrome	85282	**≤10**
Meige disease	90186	**1765**
Meigs syndrome	314451	**≤10**
Melanoma and neural system tumor syndrome	252206	**≤10**
Melanoma of soft tissue	97338	**13**
MELAS	550	**801**
Melkersson-Rosenthal syndrome	2483	**22**
Melnick-Needles syndrome	2484	**18**
Melorheostosis	2485	**48**
Melorheostosis with osteopoikilosis	1879	**≤10**
MEND syndrome	401973	**≤10**
Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency	99898	**≤10**
Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency	319558	**≤10**
Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	319552	**≤10**
Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency	319595	**≤10**
Mendelian susceptibility to mycobacterial diseases*	748	**≤10**
Ménétrier disease	2494	**≤10**
Meningeal melanocytoma	252046	**≤10**
Meningioma	2495	**301**
Meningococcal meningitis	33475	**50**
Menke-Hennekam syndrome	592574	**≤10**
Menkes disease	565	**63**
Menstrual cycle-dependent periodic fever	498251	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
MEPAN syndrome	508093	**≤10**
MERRF	551	**91**
Mesial temporal lobe epilepsy with hippocampal sclerosis	99701	**700**
Mesoaxial synostotic syndactyly with phalangeal reduction	157801	**≤10**
Mesomelia-synostoses syndrome	2496	**≤10**
Mesomelic and rhizo-mesomelic dysplasia*	93438	**≤10**
Mesomelic dysplasia, Savarirayan type	85170	**≤10**
Metabolic myopathy due to lactate transporter defect	171690	**15**
Metachondromatosis	2499	**42**
Metachromatic leukodystrophy	512	**194**
Metaphyseal acroscyphodysplasia	1240	**≤10**
Metaphyseal anadysplasia	1040	**≤10**
Metaphyseal chondrodysplasia, Schmid type	174	**65**
Metaphyseal chondrodysplasia, Spahr type	2501	**≤10**
Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria	99646	**≤10**
Metaphyseal dysplasia, Braun-Tinschert type	85188	**≤10**
Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome	2504	**≤10**
Metatropic dysplasia	2635	**20**
Methanol poisoning	31825	**≤10**
Methimazole embryofetopathy	1923	**≤10**
Methotrexate toxicity	565782	**≤10**
Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	308425	**≤10**
Methylmalonic acidemia with homocystinuria	26	**191**
Methylmalonic acidemia without homocystinuria*	293355	**52**
Methylmalonic aciduria due to transcobalamin receptor defect	280183	**≤10**
Mevalonate kinase deficiency*	309025	**60**
Mevalonic aciduria	29	**24**
MGAT2-CDG	79329	**≤10**
Micro syndrome	2510	**30**
Microbrachycephaly-ptosis-cleft lip syndrome	2511	**15**
Microcephalic cortical malformations-short stature due to RTTN deficiency	468631	**≤10**
Microcephalic osteodysplastic dysplasia, Saul-Wilson type	85172	**≤10**
Microcephalic osteodysplastic primordial dwarfism type II	2637	**18**
Microcephalic osteodysplastic primordial dwarfism types I and III	2636	**≤10**
Microcephalic primordial dwarfism*	324761	**17**
Microcephalic primordial dwarfism-insulin resistance syndrome	436182	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Microcephaly-albinism-digital anomalies syndrome	2513	**≤10**
Microcephaly-brachydactyly-kyphoscoliosis syndrome	3433	**≤10**
Microcephaly-brain defect-spasticity-hypernatremia syndrome	2523	**≤10**
Microcephaly-capillary malformation syndrome	294016	**≤10**
Microcephaly-cardiac defect-lung malsegmentation syndrome	2516	**≤10**
Microcephaly-cardiomyopathy syndrome	2515	**≤10**
Microcephaly-cervical spine fusion anomalies syndrome	2522	**≤10**
Microcephaly-cleft palate-abnormal retinal pigmentation syndrome	2521	**≤10**
Microcephaly-complex motor and sensory axonal neuropathy syndrome	423894	**≤10**
Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrom	500159	**≤10**
Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome	457284	**≤10**
Microcephaly-deafness-intellectual disability syndrome	2533	**14**
Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome	457351	**≤10**
Microcephaly-lymphedema-chorioretinopathy syndrome	2526	**29**
Microcephaly-microcornea syndrome, Seemanova type	2528	**≤10**
Microcephaly-polymicrogyria-corpus callosum agenesis syndrome	171703	**≤10**
Microcephaly-seizures-intellectual disability-heart disease syndrome	2519	**≤10**
Microcephaly-short stature-intellectual disability-facial dysmorphism syndrome	423306	**≤10**
Microcephaly-thin corpus callosum-intellectual disability syndrome	397951	**≤10**
Microcystic lymphatic malformation	79490	**363**
Microcytic anemia with liver iron overload	83642	**≤10**
Microduplication Xp11.22p11.23 syndrome	217377	**17**
Microform holoprosencephaly	280200	**19**
Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	476126	**≤10**
Microlissencephaly	1083	**17**
Microlissencephaly-micromelia syndrome	50810	**≤10**
Microphthalmia with brain and digit anomalies	139471	**≤10**
Microphthalmia with limb anomalies	1106	**≤10**
Microphthalmia with linear skin defects syndrome	2556	**14**
Microphthalmia, Lenz type	568	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Microphthalmia-brain atrophy syndrome	77299	**≤10**
Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome	251279	**≤10**
Microscopic polyangiitis	727	**608**
Microspherophakia-metaphyseal dysplasia syndrome	2551	**≤10**
Microtia	83463	**826**
Microvillus inclusion disease	2290	**42**
Mid-dermal elastolysis	228299	**≤10**
Middle ear neuroendocrine tumor	100084	**≤10**
Middle East respiratory syndrome	576074	**≤10**
Midline cervical cleft	141288	**23**
Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis	93279	**32**
Miller Fisher syndrome	98919	**187**
Miller-Dieker syndrome	531	**99**
Mills syndrome	94091	**≤10**
Milroy disease	79452	**371**
MIRAGE syndrome	494433	**≤10**
Mirror polydactyly-vertebral segmentation-limbs defects syndrome	3004	**≤10**
Mirror-image polydactyly	498494	**≤10**
MiT family translocation renal cell carcinoma	319308	**≤10**
MITF-related melanoma and renal cell carcinoma predisposition syndrome	293822	**22**
Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	1933	**≤10**
Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy	255235	**≤10**
Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies	369897	**≤10**
Mitochondrial DNA depletion syndrome, encephalomyopathic form*	254803	**≤10**
Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency	279934	**≤10**
Mitochondrial DNA depletion syndrome, hepatocerebral form*	254871	**≤10**
Mitochondrial DNA depletion syndrome, hepatocerebrorenal form	363534	**≤10**
Mitochondrial DNA depletion syndrome, myopathic form	254875	**21**
Mitochondrial DNA-associated Leigh syndrome	255210	**21**
Mitochondrial DNA-related cardiomyopathy and hearing loss	1349	**≤10**
Mitochondrial DNA-related dystonia	254851	**≤10**
Mitochondrial DNA-related mitochondrial myopathy*	254788	**45**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Mitochondrial DNA-related progressive external ophthalmoplegia	663	**102**
Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	314637	**≤10**
Mitochondrial membrane protein-associated neurodegeneration	289560	**≤10**
Mitochondrial myopathy and sideroblastic anemia	2598	**≤10**
Mitochondrial myopathy with reversible cytochrome C oxidase deficiency	254864	**≤10**
Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	502423	**≤10**
Mitochondrial myopathy-lactic acidosis-deafness syndrome	2597	**39**
Mitochondrial neurogastrointestinal encephalomyopathy	298	**37**
Mitochondrial pyruvate carrier deficiency	447784	**≤10**
Mitochondrial trifunctional protein deficiency	746	**23**
Mitral atresia	1205	**34**
Mitral valve agenesis	99062	**≤10**
Mixed connective tissue disease	809	**1741**
Mixed cystic lymphatic malformation	458792	**246**
Mixed germ cell tumor	180234	**≤10**
Mixed phenotype acute leukemia	530995	**≤10**
Mixed-type autoimmune hemolytic anemia	90036	**50**
Miyoshi myopathy	45448	**48**
MMEP syndrome	3434	**≤10**
MME-related autosomal dominant Charcot Marie Tooth disease type 2	497757	**≤10**
Moderate and severe traumatic brain injury	90056	**19**
MODY	552	**875**
Moebius syndrome	570	**254**
Moebius syndrome-axonal neuropathy-hypogonadotropic hypogonadism syndrome	2560	**≤10**
Mohr-Tranebjaerg syndrome	52368	**≤10**
MOMO syndrome	2563	**≤10**
Monilethrix	573	**24**
Monoamine oxidase A deficiency	3057	**≤10**
Monoclonal mast cell activation syndrome	529468	**1483**
Monocytopenia with susceptibility to infections	228423	**35**
Monomelic amyotrophy	65684	**196**
Monosomy 13q14	1587	**12**
Monosomy 13q34	96168	**18**
Monosomy 18p	1598	**63**
Monosomy 18q	1600	**172**
Monosomy 21	574	**24**
Monosomy 22	96123	**13**
Monosomy 22q13.3	48652	**291**
Monosomy 5p	281	**199**
Monosomy 9p	261112	**32**
Monosomy 9q22.3	77301	**≤10**
Mooren ulcer	519408	**15**
More information	502434	**≤10**
Morgagni-Stewart-Morel syndrome	77296	**≤10**
Morning glory disc anomaly	35737	**75**
Morvan syndrome	83467	**≤10**
Mosaic genome-wide paternal uniparental disomy	329813	**≤10**
Mosaic trisomy 1	1692	**≤10**
Mosaic trisomy 12	1698	**13**
Mosaic trisomy 14	1703	**19**
Mosaic trisomy 15	1706	**12**
Mosaic trisomy 16	1708	**18**
Mosaic trisomy 2	1723	**≤10**
Mosaic trisomy 20	1724	**16**
Mosaic trisomy 22	96068	**15**
Mosaic trisomy 3	100071	**≤10**
Mosaic trisomy 5	96060	**≤10**
Mosaic trisomy 7	1747	**≤10**
Mosaic trisomy 8	96061	**40**
Mosaic trisomy 9	99776	**38**
Mosaic variegated aneuploidy syndrome	1052	**34**
Mounier-Kühn syndrome	3347	**14**
Mowat-Wilson syndrome	2152	**147**
Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	280679	**≤10**
Moyamoya disease	2573	**355**
Moyamoya disease with early-onset achalasia	401945	**≤10**
Moynahan syndrome	2574	**≤10**
MPI-CDG	79319	**14**
MT-ATP6-related mitochondrial spastic paraplegia	320360	**≤10**
Mucinous adenocarcinoma of ovary	398961	**≤10**
Mucinous cystadenocarcinoma of the pancreas	424053	**33**
Muckle-Wells syndrome	575	**92**
Mucocutaneous venous malformations	2451	**1626**
Mucolipidosis type II	576	**36**
Mucolipidosis type III	577	**42**
Mucolipidosis type IV	578	**11**
Mucopolysaccharidosis type 1	579	**293**
Mucopolysaccharidosis type 2	580	**204**
Mucopolysaccharidosis type 3	581	**209**
Mucopolysaccharidosis type 4	582	**198**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Mucopolysaccharidosis type 6	583	**58**
Mucopolysaccharidosis type 7	584	**12**
Mucous membrane pemphigoid	46486	**1301**
Mueller-Weiss syndrome	566943	**≤10**
Muenke syndrome	53271	**126**
Muir-Torre syndrome	587	**125**
Mulibrey nanism	2576	**≤10**
Müllerian aplasia and hyperandrogenism	247768	**≤10**
Müllerian aplasia*	73217	**≤10**
Müllerian derivatives-lymphangiectasia-polydactyly syndrome	1655	**≤10**
Müllerian duct anomalies-limb anomalies syndrome	2491	**≤10**
Multicentric carpo-tarsal osteolysis with or without nephropathy	2774	**≤10**
Multicentric osteolysis-nodulosis-arthropathy spectrum	371428	**≤10**
Multicentric reticulohistiocytosis	139436	**15**
Multicystic dysplastic kidney	1851	**2869**
Multifocal atrial tachycardia	3282	**11**
Multifocal lymphangioendotheliomatosis-thrombocytopenia syndrome	464321	**≤10**
Multifocal motor neuropathy	641	**827**
Multifocal pattern dystrophy simulating fundus flavimaculatus	99003	**21**
Multiloculated renal cyst	97366	**30**
Multiminicore myopathy	598	**143**
Multinodular goiter-cystic kidney-polydactyly syndrome	2091	**14**
Multiple acyl-CoA dehydrogenase deficiency	26791	**108**
Multiple benign circumferential skin creases on limbs	2505	**≤10**
Multiple carboxylase deficiency*	148	**≤10**
Multiple congenital anomalies-hypotonia-seizures syndrome	280633	**34**
Multiple congenital anomalies-hypotonia-seizures syndrome type 2	300496	**≤10**
Multiple endocrine neoplasia type 1	652	**455**
Multiple endocrine neoplasia type 2	653	**259**
Multiple endocrine neoplasia type 4	276152	**≤10**
Multiple endocrine neoplasia*	276161	**50**
Multiple epiphyseal dysplasia due to collagen 9 anomaly	166002	**≤10**
Multiple epiphyseal dysplasia type 1	93308	**≤10**
Multiple epiphyseal dysplasia type 4	93307	**≤10**
Multiple epiphyseal dysplasia type 5	93311	**≤10**
Multiple epiphyseal dysplasia*	251	**385**
Multiple epiphyseal dysplasia, Beighton type	166011	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia	166029	**≤10**
Multiple intestinal atresia	2300	**13**
Multiple metaphyseal dysplasia*	93430	**125**
Multiple mitochondrial dysfunctions syndrome type 1	401869	**≤10**
Multiple myeloma	29073	**424**
Multiple osteochondromas	321	**1070**
Multiple paragangliomas associated with polycythemia	324299	**≤10**
Multiple pterygium syndrome*	294060	**≤10**
Multiple sclerosis-ichthyosis-factor VIII deficiency syndrome	3151	**≤10**
Multiple self-healing squamous epithelioma	65748	**≤10**
Multiple sulfatase deficiency	585	**20**
Multiple symmetric lipomatosis	2398	**102**
Multiple synostoses syndrome	3237	**59**
Multiple system atrophy	102	**1083**
Multisystem inflammatory syndrome in children and adults	598363	**299**
Multisystemic smooth muscle dysfunction syndrome	404463	**≤10**
Muscle filaminopathy	171445	**69**
Muscle-eye-brain disease	588	**16**
Muscular atrophy-ataxia-retinitis pigmentosa-diabetes mellitus syndrome	2579	**≤10**
Muscular dystrophy, Selcen type	199340	**≤10**
Muscular glycogenosis*	206959	**101**
Muscular hypertrophy-hepatomegaly-polyhydramnios syndrome	324416	**≤10**
Musculocontractural Ehlers-Danlos syndrome	2953	**11**
Mutilating hereditary sensory neuropathy with spastic paraplegia	139578	**37**
Mutilating palmoplantar keratoderma with periorificial keratotic plaques	659	**≤10**
Myalgia-eosinophilia syndrome associated with tryptophan	2582	**≤10**
Myasthenia gravis	589	**6961**
MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome	498693	**≤10**
Mycophenolate mofetil embryopathy	268249	**≤10**
Mycoplasma encephalitis	83482	**≤10**
Mycosis fungoides and variants*	178566	**≤10**
Myelocystocele	268813	**≤10**
Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality	86841	**≤10**
Myelodysplastic syndrome*	52688	**76**
Myelodysplastic/myeloproliferative disease*	98275	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Myeloid/lymphoid neoplasm associated with FGFR1 rearrangement	168953	**≤10**
Myelomeningocele	93969	**592**
Myeloperoxidase deficiency	2587	**≤10**
Myeloproliferative neoplasm*	98274	**≤10**
MYH7-related late-onset scapuloperoneal muscular dystrophy	437572	**35**
MYH9-related disease	182050	**225**
Myhre syndrome	2588	**53**
Myoclonic epilepsy in non-progressive encephalopathies	86913	**28**
Myoclonic epilepsy of infancy	86909	**139**
Myoclonic-astatic epilepsy	1942	**482**
Myoclonus-cerebellar ataxia-deafness syndrome	2589	**≤10**
Myoclonus-dystonia syndrome	36899	**240**
Myopathic Ehlers-Danlos syndrome	536516	**≤10**
Myopathy and diabetes mellitus	2596	**≤10**
Myopic macular degeneration	178493	**26**
Myosclerosis	289380	**≤10**
Myostatin-related muscle hypertrophy	275534	**≤10**
Myotonia fluctuans	99734	**32**
Myotonia permanens	99735	**≤10**
Myotonic dystrophy*	206647	**114**
Myxopapillary ependymoma	251643	**≤10**
NAD(P)HX epimerase deficiency	555407	**≤10**
Naegeli-Franceschetti-Jadassohn syndrome	69087	**≤10**
Nager syndrome	245	**44**
Nail-patella syndrome	2614	**376**
Nail-patella-like renal disease	2613	**≤10**
Nance-Horan syndrome	627	**33**
Nanophthalmos	35612	**25**
Narcolepsy type 1	2073	**2521**
Narcolepsy type 2	83465	**931**
NARP syndrome	644	**75**
Nasal dermoid cyst	141103	**40**
Nasal dorsum fistula	141219	**153**
Nasal glial heterotopia	141112	**≤10**
Nasolacrimal duct cyst	141083	**25**
Nasopalpebral lipoma-coloboma syndrome	2399	**≤10**
Nasopharyngeal carcinoma	150	**≤10**
Nasu-Hakola disease	2770	**≤10**
Native American myopathy	168572	**37**
Navajo neurohepatopathy	255229	**≤10**
Naxos disease	34217	**≤10**
Necrobiosis lipoidica	542592	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Necrobiotic xanthogranuloma	158011	**13**
Necrotizing enterocolitis	391673	**93**
Necrotizing soft tissue infection	440368	**≤10**
Nelson syndrome	199244	**20**
Nemaline myopathy*	607	**114**
Neonatal acute respiratory distress due to SP-B deficiency	217563	**≤10**
Neonatal adrenoleukodystrophy	44	**15**
Neonatal alloimmune neutropenia	464370	**≤10**
Neonatal antiphospholipid syndrome	398097	**96**
Neonatal autoimmune hemolytic anemia	398109	**≤10**
Neonatal brainstem dysfunction	137929	**234**
Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome	79118	**≤10**
Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome	457185	**≤10**
Neonatal epileptic encephalopathy due to glutaminase deficiency	557064	**≤10**
Neonatal hemochromatosis	446	**24**
Neonatal hypoxic and ischemic brain injury	137577	**290**
Neonatal ichthyosis-sclerosing cholangitis syndrome	59303	**11**
Neonatal inflammatory skin and bowel disease	294023	**≤10**
Neonatal intrahepatic cholestasis due to citrin deficiency	247598	**≤10**
Neonatal lupus erythematosus	398124	**21**
Neonatal Marfan syndrome	284979	**28**
Neonatal osteosclerotic dysplasia*	93443	**≤10**
Neonatal scleroderma	398127	**≤10**
Neonatal severe primary hyperparathyroidism	417	**19**
Neovascular glaucoma	94058	**≤10**
Nephroblastoma	654	**340**
Nephrogenic diabetes insipidus	223	**192**
Nephrogenic syndrome of inappropriate antidiuresis	93606	**11**
Nephrogenic systemic fibrosis	137617	**≤10**
Nephronophthisis	655	**439**
Nephropathy-deafness-hyperparathyroidism syndrome	2668	**≤10**
Nephrosis-deafness-urinary tract-digital malformations syndrome	2669	**≤10**
Netherton syndrome	634	**115**
Neuhauser anomaly	99078	**19**
Neuhauser-Eichner-Opitz syndrome	2672	**≤10**
Neu-Laxova syndrome	2671	**≤10**
Neuralgic amyotrophy	2901	**729**
Neurenteric cyst	268865	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Neuroacanthocytosis*	263440	**≤10**
Neuroblastoma	635	**151**
Neurocutaneous melanocytosis	2481	**16**
Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	88639	**≤10**
Neurodegeneration with brain iron accumulation*	385	**46**
Neurodegenerative syndrome due to cerebral folate transport deficiency	217382	**17**
Neurodevelopmental delay-seizures-ophthalmic anomalies-osteopenia-cerebellar atrophy syndrome	529665	**≤10**
Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome	453499	**26**
Neuroendocrine carcinoma of pancreas	506098	**≤10**
Neuroendocrine cell hyperplasia of infancy	217560	**75**
Neuroendocrine neoplasm of appendix	100079	**≤10**
Neuroendocrine tumor of stomach	100075	**≤10**
Neuroendocrine tumor of the colon	100080	**≤10**
Neurofaciodigitorenal syndrome	2673	**≤10**
Neuroferritinopathy	157846	**14**
Neurofibroma	252183	**129**
Neurofibromatosis type 1	636	**12173**
Neurofibromatosis type 2	637	**721**
Neurofibromatosis type 6	2678	**221**
Neurofibromatosis-Noonan syndrome	638	**96**
Neurogenic arthrogryposis multiplex congenita	1143	**61**
Neuroleptic malignant syndrome	94093	**≤10**
Neurological conditions associated with aminoacylase 1 deficiency	137754	**≤10**
Neurolymphomatosis	206586	**31**
Neuromyelitis optica spectrum disorder	71211	**1208**
Neuronal ceroid lipofuscinosis*	216	**30**
Neuronal tumor*	251924	**≤10**
Neuropathy with hearing impairment	139512	**≤10**
Neurotrophic keratopathy	137596	**47**
Neutral lipid storage disease with ichthyosis	98907	**24**
Neutral lipid storage disease*	165	**18**
Neutral lipid storage myopathy	98908	**14**
Neutropenia-monocytopenia-deafness syndrome	2690	**≤10**
Neutrophil immunodeficiency syndrome	183707	**11**
NEVADA syndrome	370059	**≤10**
Nevus comedonicus syndrome	64754	**≤10**
Nevus of Ito	263432	**≤10**
Nevus of Ota	263425	**23**
New-onset refractory status epilepticus	363558	**30**
Nicolaides-Baraitser syndrome	3051	**56**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Niemann-Pick disease type C	646	**193**
Night blindness-skeletal anomalies-dysmorphism syndrome	1390	**≤10**
Nijmegen breakage syndrome	647	**≤10**
NK-cell enteropathy	263665	**≤10**
NKX6-2-related autosomal recessive hypomyelinating leukodystrophy	527497	**≤10**
NLRC4-related familial cold autoinflammatory syndrome	576349	**≤10**
NLRP12-associated hereditary periodic fever syndrome	247868	**≤10**
NMDA receptor encephalitis	217253	**165**
Nocardiosis	31204	**≤10**
Nodal marginal zone B-cell lymphoma	86867	**≤10**
Nodular cutaneous amyloidosis	137810	**≤10**
Nodular fasciitis	477742	**≤10**
Nodular lymphocyte predominant Hodgkin lymphoma	86893	**≤10**
Nodular neuronal heterotopia	2149	**452**
Nodular non-suppurative panniculitis	33577	**11**
Noma	2700	**≤10**
NON RARE IN EUROPE: Common mesentery	620	**15**
NON RARE IN EUROPE: Eosinophilic esophagitis	73247	**394**
NON RARE IN EUROPE: Periventricular leukomalacia	171676	**72**
Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome	231720	**≤10**
Non-acquired isolated growth hormone deficiency	631	**5262**
Non-acquired panhypopituitarism	90695	**1021**
Non-amyloid fibrillary glomerulopathy	97566	**31**
Non-amyloid monoclonal immunoglobulin deposition disease	86861	**170**
Non-distal monosomy 10q	1581	**42**
Non-distal monosomy 12q	96160	**≤10**
Non-distal trisomy 10q	1695	**≤10**
Non-distal trisomy 13q	1702	**16**
Non-epidermolytic palmoplantar keratoderma	2337	**19**
Non-functioning neuroendocrine tumor of pancreas	506075	**≤10**
Non-functioning paraganglioma	94080	**215**
Non-functioning pituitary adenoma	91349	**4663**
Non-histaminic angioedema*	658	**361**
Non-insulinoma pancreatogenous hypoglycemia syndrome	276608	**15**
Non-involuting congenital hemangioma	141179	**286**
Non-Langerhans cell histiocytosis*	157987	**19**
Non-papillary transitional cell carcinoma of the bladder	209989	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Non-paraneoplastic sensory ganglionopathy	208989	**30**
Non-progressive cerebellar ataxia with intellectual disability	314647	**35**
Non-recovering obstetric brachial plexus lesion	439202	**23**
Non-rhizomelic chondrodysplasia punctata*	176	**38**
Non-seminomatous germ cell tumor of testis	363494	**≤10**
Non-specific early-onset epileptic encephalopathy	442835	**318**
Non-specific interstitial pneumonia	91364	**1305**
Non-specific syndromic intellectual disability	528084	**1438**
Non-spherocytic hemolytic anemia due to hexokinase deficiency	90031	**≤10**
Non-syndromic anorectal malformation with anal stenosis	601008	**20**
Non-syndromic anorectal malformation with perineal fistula	600952	**155**
Non-syndromic anorectal malformation with pouch colon	601013	**≤10**
Non-syndromic anorectal malformation with rectal atresia	601018	**11**
Non-syndromic anorectal malformation with rectal stenosis	601023	**≤10**
Non-syndromic anorectal malformation with rectourethral fistula	600961	**78**
Non-syndromic anorectal malformation with rectovaginal fistula	601028	**19**
Non-syndromic anorectal malformation with rectovesical fistula	600984	**11**
Non-syndromic anorectal malformation with vestibular fistula	600993	**52**
Non-syndromic anorectal malformation without fistula	601002	**30**
Non-syndromic anorectal malformation*	557	**321**
Non-syndromic bicoronal craniosynostosis	35099	**171**
Non-syndromic bilambdoid and sagittal craniosynostosis	1516	**19**
Non-syndromic cloacal malformation	600998	**65**
Non-syndromic craniosynostosis*	139390	**111**
Non-syndromic genetic deafness	87884	**5593**
Non-syndromic male infertility due to sperm motility disorder	276234	**21**
Non-syndromic metopic craniosynostosis	3366	**974**
Non-syndromic pontocerebellar hypoplasia*	98523	**117**
Non-syndromic posterior hypospadias	95706	**1415**
Non-syndromic sagittal craniosynostosis	35093	**1862**
Noonan syndrome	648	**2573**
Noonan syndrome with multiple lentigines	500	**176**
Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	363972	**19**
Noonan syndrome-like disorder with loose anagen hair	2701	**75**
Norrie disease	649	**62**
North Carolina macular dystrophy	75327	**32**
NPHP3-related Meckel-like syndrome	3032	**≤10**
NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	600663	**≤10**
NUT midline carcinoma	443167	**≤10**
Oblique facial cleft*	141253	**≤10**
Occipital horn syndrome	198	**12**
Occipital pachygyria and polymicrogyria	280640	**21**
Occult macular dystrophy	247834	**67**
Ochoa syndrome	2704	**17**
Ocular albinism with congenital sensorineural deafness	352740	**≤10**
Ocular albinism with late-onset sensorineural deafness	1000	**≤10**
Ocular albinism*	284804	**178**
Ocular anomalies-axonal neuropathy-developmental delay syndrome	496790	**≤10**
Ocular cicatricial pemphigoid	99922	**54**
Ocular motor apraxia, Cogan type	1125	**53**
Oculoauriculofrontonasal syndrome	398156	**≤10**
Oculo-auriculo-vertebral spectrum	141132	**122**
Oculoauriculovertebral spectrum with radial defects	2549	**13**
Oculocerebral hypopigmentation syndrome, Cross type	2719	**≤10**
Oculocerebrocutaneous syndrome	1647	**≤10**
Oculocerebrofacial syndrome, Kaufman type	2707	**≤10**
Oculocerebrorenal syndrome of Lowe	534	**97**
Oculocutaneous albinism type 1	352731	**192**
Oculocutaneous albinism type 2	79432	**141**
Oculocutaneous albinism type 3	79433	**≤10**
Oculocutaneous albinism type 4	79435	**30**
Oculocutaneous albinism type 5	370091	**≤10**
Oculocutaneous albinism type 6	370097	**≤10**
Oculocutaneous albinism*	55	**1014**
Oculodentodigital dysplasia	2710	**62**
Oculofaciocardiodental syndrome	2712	**38**
Oculogastrointestinal muscular dystrophy	1876	**≤10**
Oculomaxillofacial dysostosis	1794	**≤10**
Oculoosteocutaneous syndrome	2713	**≤10**
Oculootodental syndrome	99806	**≤10**
Oculo-palato-cerebral syndrome	2714	**≤10**
Oculopharyngeal muscular dystrophy	270	**509**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Oculopharyngodistal myopathy	98897	**39**
Odontochondrodysplasia	166272	**≤10**
Odontomatosis-aortae esophagus stenosis syndrome	2724	**≤10**
Odontomicronychial dysplasia	1811	**13**
Odonto-onycho-dermal dysplasia	2721	**12**
Ogden syndrome	276432	**15**
Oguchi disease	75382	**≤10**
Okihiro syndrome	93293	**155**
Oligoarticular juvenile idiopathic arthritis	85410	**4088**
Oligoastrocytoma	251656	**≤10**
Oligocone trichromacy	75378	**≤10**
Oligodendroglioma	251627	**31**
Oligodontia	99798	**2244**
Oligomeganephronia	2260	**171**
Olivopontocerebellar atrophy-deafness syndrome	2732	**≤10**
Ollier disease	296	**330**
Omenn syndrome	39041	**21**
Omodysplasia	2733	**≤10**
Omphalocele	660	**197**
Omphalomesenteric cyst	490	**≤10**
Oncogenic osteomalacia	352540	**26**
Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome	2743	**≤10**
Opitz GBBB syndrome	2745	**50**
Opsismodysplasia	2746	**≤10**
Opsoclonus-myoclonus syndrome	1183	**101**
Optic atrophy-ataxia-peripheral neuropathy-global developmental delay syndrome	543470	**≤10**
Optic atrophy-intellectual disability syndrome	401777	**39**
Optic disc pit	519404	**≤10**
Optic pathway glioma	2086	**285**
Oral erosive lichen	31142	**37**
Oral-facial-digital syndrome with short stature and brachymesophalangy	508501	**≤10**
Orbital leiomyoma	52994	**≤10**
Ornithine transcarbamylase deficiency	664	**412**
Orofaciodigital syndrome type 1	2750	**56**
Orofaciodigital syndrome type 10	2756	**≤10**
Orofaciodigital syndrome type 2	2751	**≤10**
Orofaciodigital syndrome type 4	2753	**≤10**
Orofaciodigital syndrome type 5	2919	**≤10**
Orofaciodigital syndrome type 6	2754	**≤10**
Orofaciodigital syndrome type 9	141007	**≤10**
Orofaciodigital syndrome*	140997	**36**
Oromandibular dystonia	93958	**28**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Osgood-Schlatter disease	97335	**38**
Ossification anomalies-psychomotor developmental delay syndrome	73230	**≤10**
Osteochondritis dissecans	2764	**33**
Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome	2653	**≤10**
Osteochondrosis of the metatarsal bone	564003	**≤10**
Osteochondrosis of the tarsal bone	563991	**≤10**
Osteocraniostenosis	2763	**250**
Osteofibrous dysplasia	488265	**19**
Osteogenesis imperfecta	666	**3254**
Osteoglosphonic dysplasia	2645	**≤10**
Osteomesopyknosis	2777	**12**
Osteonecrosis of the jaw	399293	**≤10**
Osteopathia striata-cranial sclerosis syndrome	2780	**39**
Osteopathia striata-pigmentary dermopathy-white forelock syndrome	2779	**≤10**
Osteopetrosis and related disorders*	2781	**56**
Osteopetrosis with renal tubular acidosis	2785	**31**
Osteoporosis-macrocephaly-blindness-joint hyperlaxity syndrome	2787	**≤10**
Osteoporosis-oculocutaneous hypopigmentation syndrome	2786	**≤10**
Osteoporosis-pseudoglioma syndrome	2788	**17**
Osteosarcoma	668	**64**
Osteosclerosis-developmental delay-craniosynostosis syndrome	178377	**≤10**
Osteosclerosis-ichthyosis-premature ovarian failure syndrome	75325	**≤10**
Osteosclerotic metaphyseal dysplasia	500548	**≤10**
O'Sullivan-McLeod syndrome	99965	**≤10**
Otodental syndrome	2791	**13**
Otofaciocervical syndrome	2792	**≤10**
Otopalatodigital syndrome spectrum disorder*	364541	**≤10**
Otopalatodigital syndrome type 1	90650	**16**
Otopalatodigital syndrome type 2	90652	**≤10**
Otospondylomegaepiphyseal dysplasia	1427	**≤10**
Ovarian fibroma	314473	**≤10**
Ovarian fibrothecoma	314478	**≤10**
Ovarian hyperstimulation syndrome	64739	**≤10**
Overgrowth syndrome with 2q37 translocation	498488	**≤10**
Overgrowth-macrocephaly-facial dysmorphism syndrome	137634	**13**
Overhydrated hereditary stomatocytosis	3203	**≤10**
Overlap myositis	206572	**498**
Overlapping connective tissue disease*	251312	**18**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Pachydermoperiostosis	2796	**36**
Pachygyria-intellectual disability-epilepsy syndrome	2798	**57**
Pachyonychia congenita	2309	**70**
Paget disease of the nipple	180275	**≤10**
PAGOD syndrome	991	**≤10**
Pai syndrome	1993	**22**
Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	477993	**≤10**
Pallister-Hall syndrome	672	**63**
Palmoplantar keratoderma, Nagashima type	140966	**≤10**
Palmoplantar keratoderma-deafness syndrome	2202	**16**
Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome	538574	**≤10**
Palmoplantar keratoderma-spastic paralysis syndrome	2201	**≤10**
Pancytopenia due to IKZF1 mutations	317473	**≤10**
PANDAS	66624	**≤10**
Panhypophysitis	95513	**97**
Pantothenate kinase-associated neurodegeneration	157850	**29**
Papillary renal cell carcinoma	319298	**26**
Papilloma of choroid plexus	2807	**≤10**
Papillon-Lefèvre syndrome	678	**35**
Papular elastorrhexis	228264	**≤10**
Papular mucinosis of infancy	90395	**≤10**
Paracetamol poisoning	464458	**≤10**
Paramedian facial cleft*	155867	**13**
Paramedian nasal cleft	141242	**21**
Paramyotonia congenita of Von Eulenburg	684	**261**
Paraneoplastic pemphigus	63455	**40**
Paraneoplastic sensory ganglionopathy	208999	**11**
Paraneoplastic uveitis	279928	**≤10**
Paraparetic variant of Guillain-Barré syndrome	231445	**13**
Paraplegia-intellectual disability-hyperkeratosis syndrome	2824	**≤10**
Paratesticular adenocarcinoma	363478	**≤10**
Parathyroid carcinoma	143	**≤10**
PARC syndrome	2825	**≤10**
Parenteral nutrition-associated cholestasis	567983	**≤10**
Parietal foramina with clavicular hypoplasia	251290	**≤10**
Paris-Trousseau thrombocytopenia	851	**≤10**
Parkinson-dementia complex of Guam	90020	**15**
Parkinsonian-pyramidal syndrome	171695	**65**
Paroxysmal dyskinesia*	1431	**60**
Paroxysmal dystonia*	200037	**33**
Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	53583	**15**
Paroxysmal exertion-induced dyskinesia	98811	**≤10**
Paroxysmal extreme pain disorder	46348	**45**
Paroxysmal kinesigenic dyskinesia	98809	**118**
Paroxysmal nocturnal hemoglobinuria	447	**339**
Paroxysmal non-kinesigenic dyskinesia	98810	**28**
Partial androgen insensitivity syndrome	90797	**144**
Partial atrioventricular septal defect	1330	**540**
Partial bilateral aplasia of the Müllerian ducts*	180068	**≤10**
Partial chromosome Y deletion	1646	**68**
Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome	401959	**≤10**
Partial deep dermal and full thickness burns	90076	**≤10**
Partially involuting congenital hemangioma	458785	**22**
Partington syndrome	94083	**23**
Patella aplasia/hypoplasia	86789	**≤10**
Paternal 20q13.2q13.3 microdeletion syndrome	261304	**≤10**
Paternal uniparental disomy of chromosome 6	96191	**≤10**
Paternal uniparental disomy of chromosome 7	96192	**≤10**
Patterson-Stevenson-Fontaine syndrome	2439	**≤10**
Pauci-immune glomerulonephritis	93126	**228**
Pearson syndrome	699	**40**
Pectus excavatum-macrocephaly-dysplastic nails syndrome	2835	**≤10**
Pediatric arterial ischemic stroke	439175	**416**
Pediatric collagenous gastritis	487809	**≤10**
Pediatric multiple sclerosis	477738	**267**
Pediatric systemic lupus erythematosus	93552	**623**
Pediatric-onset Graves disease	525731	**812**
Peeling skin syndrome*	817	**23**
PEHO syndrome	2836	**≤10**
PEHO-like syndrome	99807	**≤10**
Pelizaeus-Merzbacher disease	702	**155**
Pelizaeus-Merzbacher-like disease	280270	**26**
Pellucid marginal degeneration	137672	**48**
Pelviscapular dysplasia	93333	**≤10**
Pelvis-shoulder dysplasia	2839	**≤10**
Pemphigoid gestationis	63275	**86**
Pemphigus erythematosus	79480	**12**
Pemphigus foliaceus	79481	**15**
Pemphigus vegetans	79479	**≤10**
Pemphigus vulgaris	704	**848**
Pendred syndrome	705	**363**
Penile agenesis	49	**≤10**
Penoscrotal transposition	2842	**≤10**
PENS syndrome	313936	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Pentalogy of Cantrell	1335	**≤10**
Pentasomy X	11	**≤10**
Pericardial and diaphragmatic defect	2847	**13**
Perineural cyst	65250	**123**
Perineurioma*	85102	**≤10**
Periodic fever-infantile enterocolitis-autoinflammatory syndrome	436166	**16**
Periodic paralysis with later-onset distal motor neuropathy	397750	**≤10**
Periodic paralysis with transient compartment-like syndrome	397755	**≤10**
Periodic paralysis*	206976	**61**
Periodontal Ehlers-Danlos syndrome	75392	**24**
Perioral myoclonia with absences	139426	**≤10**
Peripapillary staphyloma	519400	**≤10**
Peripartum cardiomyopathy	563	**≤10**
Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	163746	**12**
Peripheral dysostosis	1795	**≤10**
Peripheral motor neuropathy-dysautonomia syndrome	2400	**61**
Peripheral primitive neuroectodermal tumor	370348	**≤10**
Peripheral pulmonary stenosis	99084	**53**
Perivascular epithelioid cell neoplasm	595133	**≤10**
Perlman syndrome	2849	**≤10**
Peroxisomal acyl-CoA oxidase deficiency	2971	**≤10**
Peroxisome biogenesis disorder*	79189	**22**
Perrault syndrome	2855	**44**
Persistent combined dystonia*	391711	**32**
Persistent fifth aortic arch	99076	**≤10**
Persistent hyperplastic primary vitreous	91495	**285**
Persistent idiopathic facial pain	398147	**≤10**
Persistent left superior vena cava connecting through coronary sinus to left-sided atrium	99109	**17**
Persistent left superior vena cava connecting to the roof of left-sided atrium	99111	**≤10**
Persistent Müllerian duct syndrome	2856	**39**
Persistent placoid maculopathy	97341	**≤10**
Persistent polyclonal B-cell lymphocytosis	300324	**≤10**
Peters anomaly	708	**207**
Peters plus syndrome	709	**46**
Peutz-Jeghers syndrome	2869	**131**
PFAPA syndrome	42642	**2060**
Pfeiffer syndrome	710	**113**
PGM1-CDG	319646	**≤10**
PGM3-CDG	443811	**≤10**
PHACE syndrome	42775	**124**
Phacoanaphylactic uveitis	209959	**≤10**
Phakomatosis pigmentokeratotica	2874	**17**
Phakomatosis pigmentovascularis	2875	**57**
Phalangeal microgeodic syndrome	352636	**≤10**
Pharyngeal-cervical-brachial variant of Guillain-Barré syndrome	231426	**≤10**
Phenobarbital embryopathy	1919	**≤10**
Phenylketonuria	716	**2942**
PHIP-related behavioral problems-intellectual disability-obesity-dysmorphic features syndrome	589905	**≤10**
Phocomelia, Schinzel type	2879	**≤10**
Phosphoenolpyruvate carboxykinase deficiency	2880	**≤10**
Phosphoribosylpyrophosphate synthetase superactivity	3222	**≤10**
Photosensitive epilepsy	166409	**63**
Phyllodes tumor of the breast	180261	**17**
Piebald trait-neurologic defects syndrome	2885	**≤10**
Piebaldism	2884	**98**
Pierpont syndrome	487825	**11**
Pierre Robin syndrome-faciodigital anomaly syndrome	2888	**71**
Pierson syndrome	2670	**≤10**
PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis	568062	**≤10**
Pigeon-breeder lung disease	99908	**81**
Pigmented paravenous retinochoroidal atrophy	251295	**≤10**
Pili bifurcati	720	**≤10**
Pilocytic astrocytoma	251612	**67**
Pilodental dysplasia-refractive errors syndrome	2892	**≤10**
Pilomatrixoma	91414	**67**
Pineal tumor of neuroepithelial tissue*	251905	**≤10**
Pineoblastoma	251909	**≤10**
Pinnae fistula or cyst	155838	**172**
Pitt-Hopkins syndrome	2896	**197**
Pituicytoma	251623	**17**
Pituitary adenoma*	99408	**497**
Pituitary apoplexy	95613	**464**
Pituitary carcinoma	300385	**19**
Pituitary deficiency due to empty sella turcica syndrome	91354	**151**
Pituitary deficiency due to Rathke cleft cysts	91350	**651**
Pituitary dermoid and epidermoid cysts	91351	**146**
Pituitary gigantism	99725	**≤10**
Pituitary stalk interruption syndrome	95496	**1079**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Pityriasis rubra pilaris	2897	**26**
PLA2G6-associated neurodegeneration*	329303	**≤10**
PLAA-associated neurodevelopmental disorder	521426	**≤10**
Placental insufficiency	439167	**≤10**
Placental site trophoblastic tumor	99928	**≤10**
Plague	707	**≤10**
Plasma cell leukemia	454714	**≤10**
Plasmacytoma	86855	**≤10**
Plastic bronchitis	439881	**≤10**
Platyspondylic dysplasia, Torrance type	85166	**≤10**
PLCG2-associated antibody deficiency and immune dysregulation	300359	**≤10**
PLEC-related intermediate epidermolysis bullosa simplex without extracutaneous involvement	79401	**≤10**
Plectin-related limb-girdle muscular dystrophy R17	254361	**≤10**
Pleomorphic xanthoastrocytoma	251607	**≤10**
Pleural empyema	449266	**23**
Pleural mesothelioma	50251	**≤10**
Pleuro-pericardial cyst	99131	**≤10**
Pleuropulmonary blastoma	64742	**23**
PLIN1-related familial partial lipodystrophy	280356	**17**
Plummer-Vinson syndrome	54028	**≤10**
PMM2-CDG	79318	**116**
PMP22-RAI1 contiguous gene duplication syndrome	477817	**≤10**
PMP2-related Charcot-Marie-Tooth disease type 1	476394	**12**
Pneumococcal meningitis	55655	**66**
Pneumoconiosis*	182098	**86**
Pneumocystosis	723	**19**
Pneumonia caused by Pseudomonas aeruginosa infection	90066	**≤10**
POEMS syndrome	2905	**158**
POGLUT1-related limb-girdle muscular dystrophy R21	480682	**≤10**
Poikiloderma with neutropenia	221046	**≤10**
Poland syndrome	2911	**445**
Poliomyelitis	2912	**92**
Polyarteritis nodosa	767	**538**
Polyarticular juvenile idiopathic arthritis*	404580	**85**
Polycythemia vera	729	**104**
Polycythemia*	98427	**22**
Polydactyly of a biphalangeal thumb	93339	**153**
Polydactyly of a triphalangeal thumb	93336	**≤10**
Polydactyly of an index finger	93337	**≤10**
Polydactyly-myopia syndrome	2917	**≤10**
Polyendocrine-polyneuropathy syndrome	453533	**≤10**
Polyglucosan body myopathy type 1	397937	**≤10**
Polyglucosan body myopathy type 2	456369	**≤10**
Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	500533	**≤10**
Polymicrogyria due to TUBB2B mutation	300573	**≤10**
Polymicrogyria with optic nerve hypoplasia	250972	**≤10**
Polymicrogyria*	35981	**275**
Polymyalgia rheumatica	93569	**1329**
Polymyositis	732	**985**
Polyneuropathy associated with IgM monoclonal gammapathy with anti-MAG	639	**1187**
Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	171848	**≤10**
Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome	2928	**≤10**
Polyradiculoneuropathy associated with IgG/IgA/IgM monoclonal gammopathy without known antibodies	208981	**146**
Polyrrhinia	141091	**≤10**
Polysyndactyly	93338	**71**
Polyvalvular heart disease syndrome	228410	**≤10**
POMGNT1-related limb-girdle muscular dystrophy R15	206564	**≤10**
POMT1-related limb-girdle muscular dystrophy R11	86812	**17**
POMT2-related limb-girdle muscular dystrophy R14	206559	**≤10**
Pontine autosomal dominant microangiopathy with leukoencephalopathy	477749	**≤10**
Pontine tegmental cap dysplasia	269229	**≤10**
Pontocerebellar hypoplasia type 1	2254	**24**
Pontocerebellar hypoplasia type 10	411493	**≤10**
Pontocerebellar hypoplasia type 2	2524	**42**
Pontocerebellar hypoplasia type 3	97249	**≤10**
Pontocerebellar hypoplasia type 4	166063	**≤10**
Pontocerebellar hypoplasia type 6	166073	**≤10**
Pontocerebellar hypoplasia type 7	284339	**≤10**
Pontocerebellar hypoplasia type 9	369920	**≤10**
Popliteal pterygium syndrome*	294963	**≤10**
Porencephaly	2940	**81**
Porencephaly-cerebellar hypoplasia-internal malformations syndrome	2941	**≤10**
Porencephaly-microcephaly-bilateral congenital cataract syndrome	306547	**≤10**
Porokeratosis of Mibelli	735	**28**
Porokeratosis plantaris palmaris et disseminata	737	**11**
Porphyria cutanea tarda	101330	**2148**
Porphyria due to ALA dehydratase deficiency	100924	**≤10**
Porphyria variegata	79473	**304**
Porphyria*	738	**26**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Portosinusoidal vascular disease	596937	**2289**
Postaxial acrofacial dysostosis	246	**11**
Postaxial polydactyly type A	93334	**82**
Postaxial polydactyly type B	93335	**107**
Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome	420584	**≤10**
Postaxial tetramelic oligodactyly	2730	**14**
Posterior column ataxia-retinitis pigmentosa syndrome	88628	**≤10**
Posterior cortical atrophy	54247	**67**
Posterior meningocele	268810	**21**
Posterior polymorphous corneal dystrophy	98973	**27**
Posterior urethral valve	93110	**1709**
Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome	572013	**≤10**
Postinfectious vasculitis	48435	**42**
Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	477673	**≤10**
Postpartum psychosis	443173	**≤10**
Postpoliomyelitis syndrome	2942	**288**
Post-traumatic pituitary deficiency	95619	**2641**
Postural orthostatic tachycardia syndrome due to NET deficiency	443236	**≤10**
Potassium-aggravated myotonia*	612	**≤10**
Potocki-Shaffer syndrome	52022	**20**
PPARG-related familial partial lipodystrophy	79083	**18**
Prader-Willi syndrome	739	**2089**
Prader-Willi-like syndrome*	398073	**29**
Precursor B-cell acute lymphoblastic leukemia	99860	**38**
Precursor T-cell acute lymphoblastic leukemia	99861	**≤10**
Predisposition to invasive fungal disease due to CARD9 deficiency	457088	**≤10**
Predisposition to severe viral infection due to IRF7 deficiency	574918	**≤10**
Predominantly large-vessel vasculitis*	156140	**43**
Predominantly medium-vessel vasculitis*	156143	**31**
Predominantly small-vessel vasculitis*	156146	**56**
Preeclampsia	275555	**21**
Premature closure of the arterial duct	95486	**≤10**
Prenatal-onset spinal muscular atrophy with congenital bone fractures	486811	**≤10**
Prepubertal anorexia nervosa	525738	**475**
Pressure-induced localized lipoatrophy	90160	**≤10**
Primary anetoderma	228272	**30**
Primary angiitis of the central nervous system	140989	**60**
Primary autoimmune enteropathy	522037	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Primary biliary cholangitis	186	**3848**
Primary biliary cholangitis/primary sclerosing cholangitis and autoimmune hepatitis overlap syndrome	562639	**66**
Primary bone dysplasia with multiple joint dislocations*	93441	**21**
Primary central nervous system lymphoma	46135	**≤10**
Primary ciliary dyskinesia	244	**1188**
Primary ciliary dyskinesia-retinitis pigmentosa syndrome	247522	**≤10**
Primary congenital hypothyroidism*	226295	**226**
Primary cutaneous aggressive epidermotropic CD8+ T-cell lymphoma	178528	**≤10**
Primary cutaneous follicle center lymphoma	178540	**≤10**
Primary cutaneous marginal zone B-cell lymphoma	178536	**≤10**
Primary cutaneous peripheral T-cell lymphoma not otherwise specified	86885	**≤10**
Primary cutis verticis gyrata*	671	**≤10**
Primary dystonia, DYT13 type	98807	**12**
Primary dystonia, DYT2 type	99657	**≤10**
Primary dystonia, DYT21 type	306734	**19**
Primary dystonia, DYT27 type	464440	**129**
Primary dystonia, DYT4 type	98805	**≤10**
Primary dystonia, DYT6 type	98806	**21**
Primary early-onset glaucoma*	156005	**≤10**
Primary eosinophilic gastrointestinal disease*	402029	**≤10**
Primary erythromelalgia	90026	**155**
Primary essential cutis verticis gyrata	357220	**≤10**
Primary failure of tooth eruption	412206	**45**
Primary familial polycythemia	90042	**≤10**
Primary Fanconi renotubular syndrome	3337	**60**
Primary hemophagocytic lymphohistiocytosis*	158038	**≤10**
Primary hepatic neuroendocrine carcinoma	100085	**≤10**
Primary hyperaldosteronism-seizures-neurological abnormalities syndrome	369929	**≤10**
Primary hypereosinophilic syndrome	314950	**109**
Primary hypergonadotropic hypogonadism-partial alopecia syndrome	2232	**≤10**
Primary hyperoxaluria	416	**305**
Primary hypertrophic osteoarthropathy*	248095	**≤10**
Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement	2196	**≤10**
Primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement	31043	**37**
Primary hypomagnesemia with hypercalciuria and nephrocalcinosis*	306516	**23**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Primary hypomagnesemia with secondary hypocalcemia	30924	**40**
Primary hypomagnesemia-refractory seizures-intellectual disability syndrome	564178	**≤10**
Primary hypophysitis*	95506	**40**
Primary immunodeficiency syndrome due to LAMTOR2 deficiency	90023	**≤10**
Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency	75391	**≤10**
Primary immunodeficiency with post-measles-mumps-rubella vaccine viral infection	431166	**≤10**
Primary intestinal lymphangiectasia	90362	**73**
Primary intrahepatic lithiasis	480506	**35**
Primary intraocular lymphoma	279904	**≤10**
Primary intraosseous venous malformation	140436	**47**
Primary laryngeal lymphangioma	137926	**18**
Primary lateral sclerosis	35689	**465**
Primary mediastinal large B-cell lymphoma	98838	**≤10**
Primary membranoproliferative glomerulonephritis	54370	**721**
Primary membranous glomerulonephritis	97560	**1447**
Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	391408	**≤10**
Primary myelofibrosis	824	**19**
Primary non-essential cutis verticis gyrata	357225	**≤10**
Primary oculocerebral lymphoma	279897	**15**
Primary orthostatic hypotension*	182058	**≤10**
Primary orthostatic tremor	238606	**96**
Primary pediatric heart tumor	875	**19**
Primary peritoneal carcinoma	168829	**≤10**
Primary pigmented nodular adrenocortical disease	189439	**17**
Primary progressive aphasia*	95432	**49**
Primary progressive apraxia of speech	314566	**≤10**
Primary progressive freezing gait	75567	**≤10**
Primary pulmonary hypoplasia	2257	**49**
Primary pulmonary lymphoma	2420	**≤10**
Primary renal tubular acidosis*	314822	**16**
Primary sclerosing cholangitis	171	**2565**
Primary short bowel syndrome*	365563	**≤10**
Primary Sjögren syndrome	289390	**8210**
Primary syringomyelia	99856	**663**
Primary tethered cord syndrome	268861	**239**
Primary unilateral adrenal hyperplasia	231580	**809**
Primitive portal vein thrombosis	854	**3943**
Proboscis lateralis	141099	**≤10**
Progeria-short stature-pigmented nevi syndrome	2959	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Progeroid and marfanoid aspect-lipodystrophy syndrome	300382	**≤10**
Progeroid features-hepatocellular carcinoma predisposition syndrome	435953	**≤10**
Progeroid syndrome, Petty type	2963	**≤10**
Progressive bifocal chorioretinal atrophy	75373	**≤10**
Progressive cavitating leukoencephalopathy	139447	**≤10**
Progressive cerebello-cerebral atrophy	247198	**21**
Progressive cone dystrophy	1871	**443**
Progressive deafness with stapes fixation	3235	**18**
Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	457212	**≤10**
Progressive familial intrahepatic cholestasis	172	**385**
Progressive hemifacial atrophy	1214	**62**
Progressive multifocal leukoencephalopathy	217260	**≤10**
Progressive muscular atrophy	454706	**42**
Progressive myoclonic epilepsy type 1	308	**102**
Progressive myoclonic epilepsy type 7	435438	**≤10**
Progressive myoclonic epilepsy type 8	424027	**≤10**
Progressive myoclonic epilepsy type 9	457265	**≤10**
Progressive myoclonic epilepsy with dystonia	352596	**≤10**
Progressive myoclonic epilepsy*	98261	**31**
Progressive non-fluent aphasia	100070	**282**
Progressive non-infectious anterior vertebral fusion	2062	**≤10**
Progressive osseous heteroplasia	2762	**18**
Progressive pseudorheumatoid arthropathy of childhood	1159	**22**
Progressive scapulohumeroperoneal distal myopathy	447977	**≤10**
Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	228012	**≤10**
Progressive supranuclear palsy	683	**888**
Progressive symmetric erythrokeratodermia	316	**13**
Prolactinoma	2965	**5876**
Prolidase deficiency	742	**≤10**
Proliferating trichilemmal cyst	492	**≤10**
Properdin deficiency	2966	**≤10**
Propionic acidemia	35	**143**
Proteasome-associated autoinflammatory syndrome	324977	**≤10**
Protein S acquired deficiency	26349	**≤10**
Proteus syndrome	744	**141**
Proteus-like syndrome	2969	**≤10**
Proton-pump inhibitor-responsive esophageal eosinophilia	411696	**≤10**
Proximal 16p11.2 microdeletion syndrome	261197	**172**
Proximal 16p11.2 microduplication syndrome	370079	**100**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Proximal myopathy with focal depletion of mitochondria	521305	**≤10**
Proximal myotonic myopathy	606	**728**
Proximal renal tubular acidosis	47159	**53**
Proximal spinal muscular atrophy	70	**2197**
Proximal symphalangism	3250	**15**
Proximal Xq28 duplication syndrome	1762	**47**
PrP systemic amyloidosis	397606	**≤10**
Prune belly syndrome	2970	**81**
PRUNE1-related neurological syndrome	544469	**≤10**
Pseudoachondroplasia	750	**79**
Pseudodiastrophic dysplasia	85174	**≤10**
Pseudohypoaldosteronism type 1	756	**103**
Pseudohypoaldosteronism type 2	757	**46**
Pseudohypoaldosteronism*	444916	**25**
Pseudohypoparathyroidism type 1A	79443	**280**
Pseudohypoparathyroidism type 1B	94089	**163**
Pseudohypoparathyroidism type 1C	79444	**≤10**
Pseudohypoparathyroidism type 2	94090	**≤10**
Pseudohypoparathyroidism with Albright hereditary osteodystrophy*	457059	**33**
Pseudohypoparathyroidism without Albright hereditary osteodystrophy*	457062	**≤10**
Pseudomyxoma peritonei	26790	**≤10**
Pseudoprogeria syndrome	2985	**≤10**
Pseudopseudohypoparathyroidism	79445	**108**
Pseudounicornuate uterus	180079	**≤10**
Pseudo-von Willebrand disease	52530	**22**
Pseudoxanthoma elasticum	758	**541**
Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa	436274	**≤10**
Psoriasis-related juvenile idiopathic arthritis	85436	**433**
Psychogenic movement disorders	71519	**189**
Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome	505242	**≤10**
PTEN hamartoma tumor syndrome*	306498	**34**
Pterygium colli-intellectual disability-digital anomalies syndrome	2988	**≤10**
Ptosis-strabismus-ectopic pupils syndrome	2999	**≤10**
Ptosis-vocal cord paralysis syndrome	2997	**≤10**
Pudendal neuralgia	60039	**≤10**
Pulmonary agenesis	984	**58**
Pulmonary alveolar microlithiasis	60025	**16**
Pulmonary arterial hypertension associated with chronic hemolytic anemia*	275828	**≤10**
Pulmonary arterial hypertension associated with congenital heart disease*	275803	**426**
Pulmonary arterial hypertension associated with connective tissue disease*	275798	**404**
Pulmonary arterial hypertension associated with HIV infection*	275808	**42**
Pulmonary arterial hypertension associated with portal hypertension*	275813	**209**
Pulmonary arterial hypertension associated with schistosomiasis*	275823	**≤10**
Pulmonary arteriovenous malformation	2038	**47**
Pulmonary artery coming from patent ductus arteriosus	99049	**≤10**
Pulmonary artery hypoplasia	99083	**23**
Pulmonary atresia with ventricular septal defect	1207	**546**
Pulmonary atresia-intact ventricular septum syndrome	1208	**324**
Pulmonary blastoma	64741	**≤10**
Pulmonary capillary hemangiomatosis	199241	**≤10**
Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome	210136	**24**
Pulmonary hypertension owing to lung disease and/or hypoxia*	275837	**571**
Pulmonary hypertension with unclear multifactorial mechanism*	275844	**173**
Pulmonary interstitial glycogenosis	217557	**≤10**
Pulmonary nodular lymphoid hyperplasia	60026	**≤10**
Pulmonary non-tuberculous mycobacterial infection	411703	**153**
Pulmonary valve agenesis*	982	**56**
Pulmonary valve agenesis-intact ventricular septum-persistent ductus arteriosus syndrome	99048	**≤10**
Pulmonary valve agenesis-tetralogy of Fallot-absence of ductus arteriosus syndrome	101206	**18**
Pulmonary venoocclusive disease	31837	**264**
Punctate inner choroidopathy	580951	**≤10**
Punctate palmoplantar keratoderma type 1	79501	**12**
Punctate palmoplantar keratoderma type 2	79502	**≤10**
PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	438213	**35**
Pure autonomic failure	441	**44**
Pure hair and nail ectodermal dysplasia	69084	**43**
Pure hereditary spastic paraplegia*	102012	**353**
Pure mitochondrial myopathy	254854	**487**
Pure or complex hereditary spastic paraplegia*	320335	**61**
Purine nucleoside phosphorylase deficiency	760	**≤10**
Pustulosis palmaris et plantaris	163927	**≤10**
Pycnodysostosis	763	**69**
PYCR2-related microcephaly-progressive leukoencephalopathy	481152	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Pyknoachondrogenesis	3003	**≤10**
Pyle disease	3005	**≤10**
Pyoderma gangrenosum	48104	**26**
Pyoderma gangrenosum-acne-suppurative hidradenitis syndrome	289478	**≤10**
Pyogenic arthritis-pyoderma gangrenosum-acne syndrome	69126	**14**
Pyomyositis	764	**≤10**
Pyridoxal phosphate-responsive seizures	79096	**18**
Pyridoxine-dependent epilepsy	3006	**63**
Pyruvate carboxylase deficiency	3008	**≤10**
Pyruvate dehydrogenase deficiency	765	**141**
Q fever	781	**≤10**
QRICH1-related intellectual disability-chondrodysplasia syndrome	580940	**≤10**
QRSL1-related combined oxidative phosphorylation defect	570491	**≤10**
Quadricuspid aortic valve	542568	**≤10**
Quebec platelet disorder	220436	**45**
Quinquaud folliculitis decalvans	346	**≤10**
Rabson-Mendenhall syndrome	769	**≤10**
Radial deficiency-tibial hypoplasia syndrome	1121	**≤10**
Radial hemimelia	93321	**86**
Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome	2252	**≤10**
Radial ray hypoplasia-choanal atresia syndrome	3026	**≤10**
Radiation myelitis	90021	**24**
Radiation-induced plexopathy	521123	**44**
Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome	71289	**≤10**
Radioulnar synostosis-microcephaly-scoliosis syndrome	3268	**≤10**
Ramsay Hunt syndrome	3020	**12**
RAPADILINO syndrome	3021	**≤10**
Rapidly involuting congenital hemangioma	141184	**195**
Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	293987	**39**
Rapid-onset dystonia-parkinsonism	71517	**17**
Rare adenocarcinoma of the breast	213528	**12**
Rare combined vascular malformation*	458837	**388**
Rare cutaneous lupus erythematosus*	535	**250**
Rare hereditary thrombophilia*	217454	**≤10**
Rare isolated myopia	98619	**295**
Rare non-syndromic intellectual disability	101685	**15942**
Rare variants of adenocarcinoma of the corpus uteri	213574	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
RARS-related autosomal recessive hypomyelinating leukodystrophy	438114	**≤10**
RAS-associated autoimmune leukoproliferative disease	268114	**≤10**
Rasmussen subacute encephalitis	1929	**100**
Rat-bite fever	31205	**≤10**
Ravine syndrome	99852	**61**
Reactive arthritis	29207	**472**
Reading seizures	166433	**≤10**
Recessive dystrophic epidermolysis bullosa inversa	79409	**15**
Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome	280384	**≤10**
Recessive mitochondrial ataxia syndrome	94125	**18**
Recessive X-linked ichthyosis	461	**175**
Recombinant 8 syndrome	96167	**≤10**
Rectal duplication	171220	**≤10**
Recurrent hepatitis C virus induced liver disease in liver transplant recipients	90052	**≤10**
Recurrent idiopathic neuroretinitis	499103	**≤10**
Recurrent infection due to specific granule deficiency	169142	**≤10**
Recurrent infections associated with rare immunoglobulin isotypes deficiency	183675	**439**
Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome	480864	**33**
Recurrent respiratory papillomatosis	60032	**96**
Reducing body myopathy	97239	**≤10**
Reflex epilepsy*	310	**21**
Refractory anemia	98826	**≤10**
Refractory anemia with excess blasts	86839	**≤10**
Refractory celiac disease	398063	**25**
Refsum disease	773	**26**
Regional odontodysplasia	83450	**≤10**
Regressive spondylometaphyseal dysplasia	448267	**≤10**
Reis-Bücklers corneal dystrophy	98961	**22**
Relapsing fever	91547	**208**
Relapsing polychondritis	728	**529**
Renal agenesis	411709	**2818**
Renal caliceal diverticuli-deafness syndrome	2838	**≤10**
Renal cell carcinoma*	217071	**28**
Renal coloboma syndrome	1475	**165**
Renal dysplasia	93108	**2345**
Renal hypoplasia	93101	**3069**
Renal medullary carcinoma	319319	**≤10**
Renal nutcracker syndrome	71273	**13**
Renal tubular dysgenesis	3033	**41**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Renal tubulopathy-encephalopathy-liver failure syndrome	254902	**≤10**
Renin-angiotensin-aldosterone system-blocker-induced angioedema	100057	**644**
Renpenning syndrome	3242	**36**
RERE-related neurodevelopmental syndrome	494344	**≤10**
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha	566231	**≤10**
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta	566243	**33**
Resistance to thyrotropin-releasing hormone syndrome	99832	**≤10**
Respiratory bronchiolitis-interstitial lung disease syndrome	79127	**134**
Restrictive dermopathy	1662	**≤10**
Reticular dysgenesis	33355	**≤10**
Reticular dystrophy of the retinal pigment epithelium	99002	**149**
Reticulate acropigmentation of Kitamura	178307	**≤10**
Retinal arterial tortuosity	75326	**12**
Retinal capillary malformation	71213	**≤10**
Retinal dystrophy-optic nerve edema-splenomegaly-anhidrosis-migraine headache syndrome	313800	**≤10**
Retinal macular dystrophy type 2	319640	**28**
Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	247691	**≤10**
Retinitis pigmentosa	791	**4210**
Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome	3085	**≤10**
Retinitis punctata albescens	52427	**24**
Retinoblastoma	790	**210**
Retinopathy of prematurity	90050	**301**
Rett syndrome	778	**745**
Reversible cerebral vasoconstriction syndrome	284388	**≤10**
Revesz syndrome	3088	**≤10**
Reye syndrome	3096	**13**
Reynolds syndrome	779	**37**
RFT1-CDG	244310	**≤10**
Rhabdoid tumor	69077	**26**
Rhabdomyosarcoma	780	**99**
Rhabdomyosarcoma of the cervix uteri	213802	**≤10**
Rheumatic fever	3099	**54**
Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	85408	**1122**
Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	85435	**518**
Rhizomelic chondrodysplasia punctata	177	**19**
Rhombencephalosynapsis	59315	**33**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
RHYNS syndrome	140976	**≤10**
Riboflavin transporter deficiency	97229	**32**
Richieri Costa-Pereira syndrome	3102	**≤10**
Rieger anomaly	91483	**34**
Right aortic arch	99081	**66**
Right inferior vena cava connecting to left-sided atrium	99119	**≤10**
Right sided atrial isomerism	97548	**48**
Right superior vena cava connecting to left-sided atrium	99110	**≤10**
Rigid spine syndrome	97244	**47**
Ring chromosome 1 syndrome	1437	**≤10**
Ring chromosome 10 syndrome	1438	**≤10**
Ring chromosome 11 syndrome	96175	**≤10**
Ring chromosome 12 syndrome	1439	**≤10**
Ring chromosome 13 syndrome	96176	**≤10**
Ring chromosome 14 syndrome	1440	**12**
Ring chromosome 15 syndrome	96177	**≤10**
Ring chromosome 16 syndrome	96178	**≤10**
Ring chromosome 17 syndrome	1441	**≤10**
Ring chromosome 18 syndrome	1442	**26**
Ring chromosome 19 syndrome	1443	**≤10**
Ring chromosome 2 syndrome	96171	**≤10**
Ring chromosome 20 syndrome	1444	**42**
Ring chromosome 21 syndrome	1445	**17**
Ring chromosome 22 syndrome	1446	**25**
Ring chromosome 3 syndrome	96172	**≤10**
Ring chromosome 4 syndrome	1447	**≤10**
Ring chromosome 7 syndrome	1449	**≤10**
Ring chromosome 8 syndrome	1450	**≤10**
Ring chromosome 9 syndrome	96173	**≤10**
Ring chromosome Y syndrome	261529	**≤10**
Ring dermoid of cornea	91481	**31**
Rippling muscle disease	97238	**≤10**
Roberts syndrome	3103	**≤10**
Robin sequence-oligodactyly syndrome	3104	**15**
Robinow syndrome	97360	**61**
Roch-Leri mesosomatous lipomatosis	529	**16**
Roifman syndrome	353298	**≤10**
Rolandic epilepsy	1945	**622**
Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	163727	**≤10**
Rolandic epilepsy-speech dyspraxia syndrome	163721	**12**
Romano-Ward syndrome	101016	**298**
Rosaï-Dorfman disease	158014	**108**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Rosette-forming glioneuronal tumor	251975	**≤10**
Rothmund-Thomson syndrome	2909	**42**
Rotor syndrome	3111	**≤10**
Roussy-Lévy syndrome	3115	**≤10**
Rubella panencephalitis	83616	**≤10**
Rubinstein-Taybi syndrome	783	**444**
Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome	397927	**≤10**
S-adenosylhomocysteine hydrolase deficiency	88618	**≤10**
Saethre-Chotzen syndrome	794	**156**
Saldino-Mainzer syndrome	140969	**≤10**
Sandhoff disease	796	**40**
Sandifer syndrome	71272	**≤10**
Sanjad-Sakati syndrome	2323	**≤10**
SAPHO syndrome	793	**275**
Sarcoidosis	797	**9024**
Sarcoma of the corpus uteri*	213620	**≤10**
SATB2-associated syndrome	576278	**51**
SCALP syndrome	370052	**≤10**
Scalp-ear-nipple syndrome	2036	**≤10**
Scapuloperoneal spinal muscular atrophy	431255	**17**
Schimke immuno-osseous dysplasia	1830	**28**
Schinzel-Giedion syndrome	798	**23**
Schistosomiasis	1247	**13**
Schizencephaly	799	**91**
Schneckenbecken dysplasia	3144	**≤10**
Schnitzler syndrome	37748	**58**
Schnyder corneal dystrophy	98967	**≤10**
Schöpf-Schulz-Passarge syndrome	50944	**≤10**
Schwannomatosis	93921	**452**
Schwartz-Jampel syndrome	800	**16**
Scimitar syndrome	185	**138**
Scleredema	352763	**≤10**
Scleroderma*	801	**1183**
Scleromyxedema	167635	**≤10**
Sclerosing cholangitis*	447771	**221**
Sclerosteosis	3152	**≤10**
Scott syndrome	806	**39**
Sebocystomatosis	841	**20**
Seborrhea-like dermatitis with psoriasiform elements	168606	**≤10**
Seckel syndrome	808	**37**
Second branchial cleft anomaly	141022	**321**
Secondary erythromelalgia	529864	**36**
Secondary hypereosinophilic syndrome	314962	**135**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Secondary hypoparathyroidism due to impaired parathormon secretion	140286	**275**
Secondary intestinal lymphangiectasia	90363	**≤10**
Secondary non-traumatic avascular necrosis	399180	**≤10**
Secondary pulmonary alveolar proteinosis	420259	**≤10**
Secondary pulmonary hemosiderosis	99930	**≤10**
Secondary sclerosing cholangitis	447774	**156**
Secondary short bowel syndrome	95427	**1319**
Secondary syringomyelia	99857	**318**
Segmental odontomaxillary dysplasia	67039	**≤10**
Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome	137608	**≤10**
Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	314662	**31**
Seizures-scoliosis-macrocephaly syndrome	466926	**≤10**
Selective IgM deficiency	331235	**35**
Self-healing papular mucinosis	90397	**≤10**
Self-improving collodion baby	281122	**≤10**
Semantic dementia	100069	**235**
Semicircular canal dehiscence syndrome	420402	**≤10**
Senior-Boichis syndrome	84081	**17**
Senior-Loken syndrome	3156	**79**
Sensorineural deafness with dilated cardiomyopathy	217622	**≤10**
Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	70595	**30**
Sepsis in premature infants	90051	**≤10**
Septate uterus*	180122	**32**
Septate vagina	180154	**32**
Septo-optic dysplasia spectrum	3157	**417**
Serine biosynthesis pathway deficiency, infantile/juvenile form	583595	**≤10**
SERKAL syndrome	139466	**≤10**
Serous cystadenocarcinoma of pancreas	424073	**137**
Serpiginous choroiditis	35686	**53**
Serrated polyposis syndrome	157798	**≤10**
SETD2-related microcephaly-severe intellectual disability-multiple congenital anomalies syndrome	597743	**≤10**
Severe achondroplasia-developmental delay-acanthosis nigricans syndrome	85165	**≤10**
Severe acute respiratory syndrome	140896	**29**
Severe autosomal recessive macrothrombocytopenia	438207	**≤10**
Severe combined immunodeficiency due to adenosine deaminase deficiency	277	**19**
Severe combined immunodeficiency due to CARD11 deficiency	357237	**≤10**
Severe combined immunodeficiency due to complete RAG1/2 deficiency	331206	**15**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Severe combined immunodeficiency due to CORO1A deficiency	228003	**≤10**
Severe combined immunodeficiency due to DCLRE1C deficiency	275	**37**
Severe combined immunodeficiency due to FOXN1 deficiency	169095	**≤10**
Severe combined immunodeficiency*	183660	**62**
Severe congenital nemaline myopathy	171430	**35**
Severe congenital neutropenia*	42738	**615**
Severe dermatitis-multiple allergies-metabolic wasting syndrome	369992	**≤10**
Severe disseminated cytomegalovirus infection in immunocompetent patients	35062	**≤10**
Severe early-childhood-onset retinal dystrophy	364055	**79**
Severe early-onset axonal neuropathy due to MFN2 deficiency	90118	**15**
Severe early-onset pulmonary alveolar proteinosis due to MARS deficiency	440427	**31**
Severe generalized junctional epidermolysis bullosa	79404	**16**
Severe hereditary thrombophilia due to congenital protein C deficiency	745	**63**
Severe hereditary thrombophilia due to congenital protein S deficiency	743	**36**
Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome	467176	**≤10**
Severe intellectual disability and progressive spastic paraplegia	280763	**27**
Severe intellectual disability-epilepsy-anal anomalies-distal phalangeal hypoplasia	94066	**≤10**
Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	363686	**≤10**
Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	397933	**20**
Severe intellectual disability-progressive spastic diplegia syndrome	404473	**41**
Severe lateral tibial bowing with short stature	324307	**≤10**
Severe microbrachycephaly-intellectual disability-athetoid cerebral palsy syndrome	1236	**≤10**
Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome	369939	**≤10**
Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	397593	**≤10**
Severe neonatal-onset encephalopathy with microcephaly	209370	**72**
Severe neurodegenerative syndrome with lipodystrophy	363400	**≤10**
Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract	500545	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Severe oculo-renal-cerebellar syndrome	2715	**≤10**
Severe primary trimethylaminuria	468726	**≤10**
Severe X-linked intellectual disability, Gustavson type	3078	**≤10**
Severe X-linked mitochondrial encephalomyopathy	238329	**≤10**
Sex cord-stromal tumor of testis	363489	**≤10**
Sézary syndrome	3162	**≤10**
Sheehan syndrome	91355	**108**
Sheldon-Hall syndrome	1147	**18**
Shigellosis	810	**≤10**
Shone complex	99063	**171**
Short bowel syndrome*	104008	**657**
Short chain acyl-CoA dehydrogenase deficiency	26792	**139**
Short rib-polydactyly syndrome*	1505	**≤10**
Short rib-polydactyly syndrome, Beemer-Langer type	93268	**≤10**
Short rib-polydactyly syndrome, Majewski type	93269	**≤10**
Short rib-polydactyly syndrome, Saldino-Noonan type	93270	**≤10**
Short rib-polydactyly syndrome, Verma-Naumoff type	93271	**≤10**
Short stature due to GHSR deficiency	314811	**≤10**
Short stature due to partial GHR deficiency	314802	**28**
Short stature, Brussels type	2867	**≤10**
Short stature-advanced bone age-early-onset osteoarthritis syndrome	435804	**24**
Short stature-brachydactyly-obesity-global developmental delay syndrome	464288	**≤10**
Short stature-delayed bone age due to thyroid hormone metabolism deficiency	171706	**≤10**
Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome	314394	**≤10**
Short stature-optic atrophy-Pelger-Huët anomaly syndrome	391677	**≤10**
Short stature-pituitary and cerebellar defects-small sella turcica syndrome	85442	**≤10**
Short stature-skeletal dysplasia-retinal degeneration-intellectual disability-sensorineural hearing loss syndrome	589442	**≤10**
Short stature-valvular heart disease-characteristic facies syndrome	2868	**≤10**
Short stature-webbed neck-heart disease syndrome	2865	**≤10**
SHORT syndrome	3163	**22**
Short-limb skeletal dysplasia with severe combined immunodeficiency	935	**≤10**
SHOX-related short stature	314795	**170**
Shprintzen-Goldberg syndrome	2462	**44**
Shwachman-Diamond syndrome	811	**103**
Sialidosis type 1	812	**≤10**
Sialidosis type 2	87876	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Sialidosis*	309294	**≤10**
Sialuria	3166	**≤10**
Sickle cell anemia	232	**12271**
Sickle cell-beta-thalassemia disease syndrome	251359	**1417**
Sickle cell-hemoglobin C disease syndrome	251365	**3031**
Sickle cell-hemoglobin D disease syndrome	251370	**19**
Sickle cell-hemoglobin E disease syndrome	251375	**36**
Silent sinus syndrome	71276	**≤10**
Sillence syndrome	3168	**≤10**
Silver-Russell syndrome	813	**785**
SIM1-related Prader-Willi-like syndrome	398079	**≤10**
Simple cryoglobulinemia	91139	**115**
Simpson-Golabi-Behmel syndrome	373	**73**
Sinding-Larsen-Johansson disease	97337	**≤10**
Singleton-Merten dysplasia	85191	**≤10**
Sinoatrial node dysfunction and deafness	324321	**≤10**
Sitosterolemia	2882	**13**
Situs ambiguus	157769	**15**
Situs inversus totalis	101063	**61**
Sjögren-Larsson syndrome	816	**50**
Skeletal dysplasia-epilepsy-short stature syndrome	1858	**≤10**
Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome	508533	**≤10**
Skeletal Ewing sarcoma	319	**62**
Skin fragility-woolly hair-palmoplantar keratoderma syndrome	293165	**≤10**
SLC35A1-CDG	238459	**≤10**
SLC35A2-CDG	356961	**≤10**
SLC39A8-CDG	468699	**≤10**
Slender bone dysplasia*	93440	**≤10**
Small cell carcinoma of the bladder	284400	**≤10**
Small cell carcinoma of the ovary	370396	**≤10**
Small cell lung cancer	70573	**27**
Smith-Lemli-Opitz syndrome	818	**86**
Smith-Magenis syndrome	819	**365**
Smith-McCort dysplasia	178355	**≤10**
Smoldering systemic mastocytosis	158775	**25**
Sneddon syndrome	820	**51**
Sodium channelopathy-related small fiber neuropathy	306577	**413**
Soft tissue sarcoma*	3394	**≤10**
Solid pseudopapillary carcinoma of pancreas	424065	**22**
Solitary bone cyst	83468	**≤10**
Solitary fibrous tumor/hemangiopericytoma	2126	**≤10**
Solitary rectal ulcer syndrome	209964	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Somatomammotropinoma	314769	**331**
Somatotropic adenoma*	96256	**44**
Sorsby pseudoinflammatory fundus dystrophy	59181	**≤10**
Sotos syndrome	821	**573**
Southeast Asian ovalocytosis	98868	**≤10**
Spasmus nutans	279882	**≤10**
Spastic ataxia with congenital miosis	1182	**≤10**
Spastic ataxia*	316226	**123**
Spastic ataxia-corneal dystrophy syndrome	2572	**≤10**
Spastic ataxia-dysarthria due to glutaminase deficiency	557056	**≤10**
Spastic paraparesis-deafness syndrome	2815	**28**
Spastic paraplegia type 2	99015	**80**
Spastic paraplegia type 7	99013	**161**
Spastic paraplegia-facial-cutaneous lesions syndrome	2819	**≤10**
Spastic paraplegia-glaucoma-intellectual disability syndrome	2818	**≤10**
Spastic paraplegia-intellectual disability-nystagmus-obesity syndrome	521390	**≤10**
Spastic paraplegia-nephritis-deafness syndrome	2820	**≤10**
Spastic paraplegia-neuropathy-poikiloderma syndrome	2821	**≤10**
Spastic paraplegia-optic atrophy-neuropathy and spastic paraplegia-optic atrophy-neuropathy-related disorder*	431320	**≤10**
Spastic paraplegia-optic atrophy-neuropathy syndrome	320406	**≤10**
Spastic paraplegia-precocious puberty syndrome	2826	**≤10**
Spastic paraplegia-severe developmental delay-epilepsy syndrome	464282	**≤10**
Spastic tetraplegia-retinitis pigmentosa-intellectual disability syndrome	3011	**≤10**
Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome	447997	**≤10**
SPECC1L-related hypertelorism syndrome	1519	**28**
Specific language disorder*	211053	**60**
Specific learning disability*	211047	**1822**
Spectrin-associated autosomal recessive cerebellar ataxia	352403	**≤10**
Spina bifida aperta	268369	**95**
Spina bifida cystica*	268744	**≤10**
Spina bifida-hypospadias syndrome	3176	**≤10**
Spinal arteriovenous metameric syndrome	53721	**21**
Spinal atrophy-ophthalmoplegia-pyramidal syndrome	1217	**≤10**
Spinal cord injury	90058	**38**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Spinal muscular atrophy associated with central nervous system anomaly*	207012	≤10
Spinal muscular atrophy with respiratory distress type 1	98920	71
Spinal muscular atrophy with respiratory distress type 2	404521	30
Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	2590	≤10
Spindle cell hemangioma	210584	≤10
Spinocerebellar ataxia type 1	98755	101
Spinocerebellar ataxia type 10	98761	≤10
Spinocerebellar ataxia type 11	98767	≤10
Spinocerebellar ataxia type 13	98768	≤10
Spinocerebellar ataxia type 14	98763	37
Spinocerebellar ataxia type 15/16	98769	15
Spinocerebellar ataxia type 17	98759	12
Spinocerebellar ataxia type 19/22	98772	30
Spinocerebellar ataxia type 2	98756	146
Spinocerebellar ataxia type 20	101110	≤10
Spinocerebellar ataxia type 21	98773	15
Spinocerebellar ataxia type 23	101108	≤10
Spinocerebellar ataxia type 25	101111	≤10
Spinocerebellar ataxia type 26	101112	≤10
Spinocerebellar ataxia type 27	98764	33
Spinocerebellar ataxia type 28	101109	26
Spinocerebellar ataxia type 29	208513	42
Spinocerebellar ataxia type 3	98757	233
Spinocerebellar ataxia type 30	211017	≤10
Spinocerebellar ataxia type 31	217012	≤10
Spinocerebellar ataxia type 34	1955	≤10
Spinocerebellar ataxia type 36	276198	20
Spinocerebellar ataxia type 4	98765	≤10
Spinocerebellar ataxia type 42	458803	≤10
Spinocerebellar ataxia type 5	98766	17
Spinocerebellar ataxia type 6	98758	50
Spinocerebellar ataxia type 7	94147	75
Spinocerebellar ataxia type 8	98760	≤10
Spinocerebellar ataxia with axonal neuropathy type 1	94124	16
Spinocerebellar ataxia with axonal neuropathy type 2	64753	50
Spinocerebellar ataxia with epilepsy	254881	14
Spinocerebellar ataxia-dysmorphism syndrome	1185	≤10
Spinocerebellar degeneration-corneal dystrophy syndrome	3177	≤10
Splenic marginal zone lymphoma	86854	≤10
Split cord malformation	573278	45

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Split hand-split foot-deafness syndrome	71271	≤10
SPONASTRIME dysplasia	93357	≤10
Spondylocamptodactyly syndrome	3180	≤10
Spondylocarpotarsal synostosis	3275	22
Spondylodysplastic dysplasia*	93434	12
Spondylodysplastic Ehlers-Danlos syndrome	536471	12
Spondyloenchondrodysplasia	1855	≤10
Spondyloepimetaphyseal dysplasia congenita, Strudwick type	93346	30
Spondyloepimetaphyseal dysplasia with joint laxity	93359	≤10
Spondyloepimetaphyseal dysplasia with multiple dislocations	93360	≤10
Spondyloepimetaphyseal dysplasia, aggrecan type	171866	11
Spondyloepimetaphyseal dysplasia, Isidor type	370015	≤10
Spondyloepimetaphyseal dysplasia, matrilin-3 type	156728	≤10
Spondyloepimetaphyseal dysplasia, Missouri type	93356	≤10
Spondyloepimetaphyseal dysplasia, PAPSS2 type	93282	≤10
Spondyloepiphyseal dysplasia and spondyloepimetaphyseal dysplasia*	253	91
Spondyloepiphyseal dysplasia congenita	94068	114
Spondyloepiphyseal dysplasia tarda	93284	71
Spondyloepiphyseal dysplasia with metatarsal shortening	137678	≤10
Spondyloepiphyseal dysplasia, Maroteaux type	263482	≤10
Spondyloepiphyseal dysplasia, Reardon type	163662	≤10
Spondyloepiphyseal dysplasia, Stanescu type	459051	≤10
Spondylo-megaepiphyseal-metaphyseal dysplasia	228387	≤10
Spondylometaphyseal dysplasia*	254	38
Spondylometaphyseal dysplasia, 'corner fracture' type	93315	≤10
Spondylometaphyseal dysplasia, Golden type	168544	≤10
Spondylometaphyseal dysplasia, Kozlowski type	93314	18
Spondylometaphyseal dysplasia, Schmidt type	93316	25
Spondylometaphyseal dysplasia, Sedaghatian type	93317	≤10
Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	85167	≤10
Spondylo-ocular syndrome	85194	22
Spondyloperipheral dysplasia-short ulna syndrome	1856	≤10
Spontaneous intracranial hypotension	443180	≤10
Spontaneous periodic hypothermia	29822	13
Sporadic adult-onset ataxia of unknown etiology	247234	119
Sporadic Creutzfeldt-Jakob disease	204	12
Sporadic fetal brain disruption sequence	1665	≤10
Sporadic pheochromocytoma/secreting paraganglioma	276621	1146
Sprengel deformity	3181	27

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Squamous cell carcinoma of the cervix uteri	213767	**≤10**
Squamous cell carcinoma of the colon	423994	**≤10**
Squamous cell carcinoma of the esophagus	99977	**≤10**
Squamous cell carcinoma of the lip	502366	**≤10**
Squamous cell carcinoma of the nasal cavity and paranasal sinuses	500464	**≤10**
Squamous cell carcinoma of the oral cavity	502363	**≤10**
Squamous cell carcinoma of the oropharynx	500478	**≤10**
Squamous cell carcinoma of the penis	398058	**≤10**
Squamous cell carcinoma of the rectum	424002	**≤10**
SRD5A3-CDG	324737	**≤10**
SSR4-CDG	370927	**≤10**
Stapes ankylosis with broad thumbs and toes	140917	**≤10**
Staphylococcal necrotizing pneumonia	36238	**≤10**
Staphylococcal scalded skin syndrome	36236	**12**
Stargardt disease	827	**1350**
Startle epilepsy	166427	**≤10**
STAT3-related early-onset multisystem autoimmune disease	438159	**14**
Steatocystoma multiplex-natal teeth syndrome	3184	**≤10**
Steel syndrome	438117	**≤10**
Steinert myotonic dystrophy	273	**6499**
Sterile multifocal osteomyelitis with periostitis and pustulosis	210115	**77**
Sternal cleft	2017	**≤10**
Steroid-responsive encephalopathy associated with autoimmune thyroiditis	83601	**17**
Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	95455	**487**
Stickler syndrome	828	**817**
Stiff person spectrum disorder	3198	**88**
Stiff skin syndrome	2833	**≤10**
STING-associated vasculopathy with onset in infancy	425120	**14**
Stormorken-Sjaastad-Langslet syndrome	3204	**13**
Straddling or overriding tricuspid valve	95461	**≤10**
Striate palmoplantar keratoderma	50942	**≤10**
Stromme syndrome	506307	**≤10**
STT3B-CDG	370924	**≤10**
Sturge-Weber syndrome	3205	**459**
Stüve-Wiedemann syndrome	3206	**15**
STXBP1-related encephalopathy	599373	**32**
Subacute cutaneous lupus erythematosus	163525	**134**
Subacute inflammatory demyelinating polyneuropathy	206594	**191**
Subacute sclerosing leukoencephalitis	2806	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Subaortic stenosis-short stature syndrome	3191	**≤10**
Subcorneal pustular dermatosis	48377	**≤10**
Subcortical band heterotopia	99796	**80**
Subcutaneous panniculitis-like T-cell lymphoma	86884	**≤10**
Subependymal giant cell astrocytoma	251618	**≤10**
Submucosal cleft palate	155878	**119**
Succinic semialdehyde dehydrogenase deficiency	22	**51**
Succinyl-CoA:3-oxoacid CoA transferase deficiency	832	**15**
Sudden infant death-dysgenesis of the testes syndrome	168593	**≤10**
SUNCT syndrome	57145	**≤10**
Superficial epidermolytic ichthyosis	455	**14**
Superficial fibromatosis*	199257	**≤10**
Superficial pemphigus*	46485	**245**
Superficial siderosis	247245	**≤10**
Superior limbic keratoconjunctivitis	88633	**≤10**
Supernumerary breasts	180182	**≤10**
Supratip dysplasia	466695	**≤10**
Supravalvular aortic stenosis	3193	**104**
SURF1-related Charcot-Marie-Tooth disease type 4	391351	**≤10**
Susac syndrome	838	**80**
Susceptibility to respiratory infections associated with CD8alpha chain mutation	169085	**≤10**
Susceptibility to viral and mycobacterial infections due to STAT1 deficiency	391311	**38**
Sweet syndrome	3243	**45**
Sydenham chorea	306731	**11**
Symbrachydactyly of hands and feet	1570	**23**
Symmetrical thalamic calcifications	1314	**≤10**
Sympathetic ophthalmia	79098	**34**
Symphalangism with multiple anomalies of hands and feet	3246	**≤10**
Symptomatic form of Coffin-Lowry syndrome in female carriers	276630	**≤10**
Symptomatic form of fragile X syndrome in female carriers	449291	**28**
Symptomatic form of hemochromatosis type 1	465508	**232**
Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers	206546	**215**
Syndactyly type 1	93402	**108**
Syndactyly type 2	93403	**53**
Syndactyly type 3	93404	**22**
Syndactyly type 4	93405	**17**
Syndactyly type 5	93406	**13**
Syndactyly type 6	295012	**≤10**
Syndactyly type 8	2498	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Syndactyly-camptodactyly and clinodactyly of fifth fingers-bifid toes syndrome	357332	**≤10**
Syndactyly-nystagmus syndrome due to 2q31.1 microduplication	294026	**≤10**
Syndactyly-telecanthus-anogenital and renal malformations syndrome	140952	**≤10**
Syndromic diarrhea	84064	**48**
Syndromic microphthalmia type 5	178364	**≤10**
Syndromic orbital border hypoplasia	98606	**≤10**
Syndromic recessive X-linked ichthyosis	281090	**32**
Syndromic X-linked intellectual disability 7	85274	**≤10**
SYNGAP1-related developmental and epileptic encephalopathy	544254	**69**
Synovial sarcoma	3273	**≤10**
Syringocystadenoma papilliferum	840	**≤10**
Syringomyelia*	3280	**329**
Systemic capillary leak syndrome	188	**31**
Systemic diseases with anterior uveitis*	280926	**145**
Systemic diseases with panuveitis*	280933	**95**
Systemic Epstein-Barr virus-positive T-cell lymphoproliferative disease of childhood	364033	**≤10**
Systemic lupus erythematosus	536	**11149**
Systemic mastocytosis with associated hematologic neoplasm	98849	**52**
Systemic mastocytosis*	2467	**820**
Systemic primary carnitine deficiency	158	**81**
Systemic sclerosis	90291	**7090**
Systemic-onset juvenile idiopathic arthritis	85414	**1277**
T+ B+ severe combined immunodeficiency*	397802	**≤10**
TAFRO syndrome	457077	**≤10**
Takayasu arteritis	3287	**716**
Tako-Tsubo cardiomyopathy	66529	**124**
Tall stature-intellectual disability-renal anomalies syndrome	500095	**≤10**
Tangier disease	31150	**≤10**
TARP syndrome	2886	**11**
Tarsal-carpal coalition syndrome	1412	**≤10**
Tatton-Brown-Rahman syndrome	404443	**58**
Tay-Sachs disease	845	**74**
T-B- severe combined immunodeficiency*	317419	**≤10**
T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta	169160	**≤10**
T-B+ severe combined immunodeficiency due to gamma chain deficiency	276	**38**
T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	169154	**17**
T-B+ severe combined immunodeficiency due to JAK3 deficiency	35078	**27**
T-B+ severe combined immunodeficiency*	317416	**≤10**
TBCK-related intellectual disability syndrome	488632	**≤10**
T-cell immunodeficiency with epidermodysplasia verruciformis	324294	**≤10**
T-cell large granular lymphocyte leukemia	86872	**39**
T-cell/histiocyte rich large B cell lymphoma	300857	**≤10**
Telecanthus-hypertelorism-strabismus-pes cavus syndrome	3293	**≤10**
Telethonin-related limb-girdle muscular dystrophy R7	34514	**≤10**
TELO2-related intellectual disability-neurodevelopmental disorder	488642	**≤10**
TEMPI syndrome	284227	**≤10**
Temple syndrome	254516	**49**
Temple-Baraitser syndrome	420561	**≤10**
Temtamy preaxial brachydactyly syndrome	363417	**≤10**
Temtamy syndrome	1777	**11**
Tenosynovial giant cell tumor	66627	**15**
Terminal osseous dysplasia-pigmentary defects syndrome	88630	**≤10**
Terrien marginal degeneration	519410	**≤10**
Tessier number 4 facial cleft	141258	**≤10**
Tessier number 5 facial cleft	141261	**≤10**
Tessier number 6 facial cleft	141265	**≤10**
Tessier number 7 facial cleft	141276	**69**
Testicular agenesis	325124	**191**
Testicular regression syndrome	983	**162**
Testicular seminomatous germ cell tumor	842	**≤10**
Tetraamelia-multiple malformations syndrome	3301	**≤10**
Tetragametic chimerism	199310	**≤10**
Tetralogy of Fallot	3303	**2729**
Tetramelic monodactyly	2564	**≤10**
Tetrasomy 12p	884	**83**
Tetrasomy 18p	3307	**48**
Tetrasomy 21	96055	**≤10**
Tetrasomy 9p	3310	**22**
Tetrasomy X	9	**17**
Thakker-Donnai syndrome	1780	**45**
Thalidomide embryopathy	3312	**≤10**
Thanatophoric dysplasia	2655	**25**
Theca steroid-producing cell malignant tumor of ovary, not further specified	99917	**≤10**
Thiamine-responsive encephalopathy	199348	**≤10**
Thiamine-responsive megaloblastic anemia syndrome	49827	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Thickened earlobes-conductive deafness syndrome	2405	**≤10**
Thiel-Behnke corneal dystrophy	98960	**14**
Thiemann disease, familial form	3314	**≤10**
Third branchial cleft anomaly	141030	**19**
THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	363444	**≤10**
Thomas syndrome	3316	**≤10**
Thomsen and Becker disease	614	**437**
Thoracic dysplasia-hydrocephalus syndrome	1861	**≤10**
Thoracic outlet syndrome	97330	**28**
Thoraco-abdominal enteric duplication	1759	**≤10**
Thoracolaryngopelvic dysplasia	3317	**≤10**
Thoracomelic dysplasia	1803	**≤10**
Thrombocytopenia with congenital dyserythropoietic anemia	67044	**≤10**
Thrombocytopenia-absent radius syndrome	3320	**52**
Thrombomodulin-related bleeding disorder	436169	**≤10**
Thrombotic microangiopathy*	93573	**478**
Thrombotic thrombocytopenic purpura	54057	**785**
Thumb deformity-alopecia-pigmentation anomaly syndrome	2251	**≤10**
Thygeson superficial punctate keratitis	519406	**≤10**
Thymic aplasia	83471	**≤10**
Thymic carcinoma	99868	**≤10**
Thymic neuroendocrine tumor	97289	**≤10**
Thymic-renal-anal-lung dysplasia	3326	**≤10**
Thymoma	99867	**65**
Thyroid ectopia	95712	**1119**
Thyroid hemiagenesis	95719	**45**
Thyroid hypoplasia	95720	**59**
Thyroid lymphoma	97285	**≤10**
Thyrotoxic periodic paralysis	79102	**12**
Tibial aplasia-ectrodactyly syndrome	3329	**26**
Tibial hemimelia	93322	**33**
Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome	988	**15**
Tibial muscular dystrophy	609	**72**
Tietz syndrome	42665	**≤10**
Timothy syndrome	65283	**13**
Titin-related limb-girdle muscular dystrophy R10	140922	**61**
TMEM165-CDG	314667	**≤10**
TMEM199-CDG	466703	**≤10**
TMEM70-related mitochondrial encephalo-cardio-myopathy	1194	**18**
Tolosa-Hunt syndrome	64686	**21**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
TOR1AIP1-related limb-girdle muscular dystrophy	424261	**≤10**
Toriello-Carey syndrome	3338	**≤10**
Toriello-Lacassie-Droste syndrome	3339	**≤10**
Torsade-de-pointes syndrome with short coupling interval	51084	**27**
Torticollis-keloids-cryptorchidism-renal dysplasia syndrome	3341	**≤10**
Townes-Brocks syndrome	857	**159**
Toxic maculopathy due to antimalarial drugs	279894	**14**
Toxocariasis	3343	**14**
Tracheal agenesis	3346	**14**
Tracheobronchopathia osteochondroplastica	3348	**14**
TRAF7-associated heart defect-digital anomalies-facial dysmorphism-motor and speech delay syndrome	592570	**≤10**
Transaldolase deficiency	101028	**≤10**
Transcobalamin deficiency	859	**≤10**
Transgrediens et progrediens palmoplantar keratoderma	495	**≤10**
Transient congenital hypothyroidism*	178045	**49**
Transient erythroblastopenia of childhood	98871	**≤10**
Transient familial neonatal hyperbilirubinemia	2312	**26**
Transient hyperammonemia of the newborn	289877	**≤10**
Transient hypogammaglobulinemia of infancy	169139	**≤10**
Transient infantile hypertriglyceridemia and hepatosteatosis	300293	**≤10**
Transient myeloproliferative syndrome	420611	**102**
Transient neonatal diabetes mellitus	99886	**35**
Transient pseudohypoaldosteronism	93164	**≤10**
TRAPPC11-related limb-girdle muscular dystrophy R18	369840	**≤10**
Traumatic avascular necrosis	399175	**≤10**
Treacher-Collins syndrome	861	**352**
Trehalase deficiency	103909	**≤10**
Triatrial heart*	1463	**27**
Trichinellosis	863	**≤10**
Trichodental syndrome	3351	**≤10**
Tricho-dento-osseous syndrome	3352	**≤10**
Trichofolliculoma	864	**≤10**
Trichomegaly-retina pigmentary degeneration-dwarfism syndrome	3363	**≤10**
Tricho-retino-dento-digital syndrome	1264	**≤10**
Trichorhinophalangeal syndrome type 1 and 3	77258	**56**
Trichorhinophalangeal syndrome type 2	502	**30**
Trichorhinophalangeal syndrome*	324764	**69**
Trichothiodystrophy	33364	**50**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Tricuspid atresia	1209	**283**
Trigeminal neuralgia	221091	**≤10**
Trigonocephaly-broad thumbs syndrome	3365	**≤10**
Trigonocephaly-short stature-developmental delay syndrome	3369	**≤10**
TRIM32-related limb-girdle muscular dystrophy R8	1878	**15**
Triose phosphate-isomerase deficiency	868	**12**
Triphalangeal thumb-polysyndactyly syndrome	2950	**≤10**
Triphalangeal thumbs-brachyectrodactyly syndrome	2947	**≤10**
Triple A syndrome	869	**96**
Triploidy	3376	**19**
Trismus-pseudocamptodactyly syndrome	3377	**≤10**
Trisomy 10p	171929	**≤10**
Trisomy 12p	1699	**32**
Trisomy 13	3378	**141**
Trisomy 17p	261290	**≤10**
Trisomy 18	3380	**201**
Trisomy 18p	1715	**16**
Trisomy 1q	261344	**23**
Trisomy 20p	261318	**≤10**
Trisomy 4p	1738	**23**
Trisomy 5p	1742	**18**
Trisomy 8p	264450	**20**
Trisomy 8q	1752	**17**
Trisomy 9p	236	**59**
Trisomy X	3375	**514**
Tritanopia	88629	**≤10**
Tropical pancreatitis	103918	**139**
Tropical spastic paraparesis	289326	**47**
True unicornuate uterus	180074	**≤10**
Truncus arteriosus	3384	**261**
TSH-secreting pituitary adenoma	91347	**163**
Tuberculosis	3389	**177**
Tuberculous meningitis	499004	**15**
Tuberous sclerosis complex	805	**2879**
Tubular aggregate myopathy	2593	**26**
Tubulinopathy-associated dysgyria	467166	**21**
Tubulointerstitial nephritis and uveitis syndrome	91500	**164**
Tufted angioma	1063	**33**
Tularemia	3392	**≤10**
Tumor necrosis factor receptor 1 associated periodic syndrome	32960	**139**
Turner syndrome	881	**4443**
Twin to twin transfusion syndrome	95431	**19**
Typical nemaline myopathy	171436	**31**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Tyrosinemia type 1	882	**148**
Tyrosinemia type 2	28378	**12**
Tyrosinemia type 3	69723	**≤10**
Uhl anomaly	3403	**≤10**
Ulbright-Hodes syndrome	3404	**≤10**
Ulerythema ophryogenesis	3406	**≤10**
Ulnar hemimelia	93320	**39**
Ulnar-mammary syndrome	3138	**40**
Unclassified myelodysplastic syndrome	98827	**31**
Unclassified myelodysplastic/myeloproliferative disease	98825	**≤10**
Unclassified vasculitis	251328	**460**
Uncombable hair syndrome	1410	**17**
Undetermined colitis	103920	**2992**
Undifferentiated carcinoma of the corpus uteri	213721	**≤10**
Undifferentiated connective tissue syndrome	90002	**737**
Undifferentiated pleomorphic sarcoma	2023	**≤10**
Unexplained long-lasting fever/inflammatory syndrome	251332	**120**
Unicervical bicornuate uterus	180114	**≤10**
Unilateral aplasia of the Müllerian ducts*	180071	**≤10**
Unilateral polymicrogyria	268943	**134**
Univentricular heart	1464	**592**
Unspecified juvenile idiopathic arthritis	91140	**732**
Unspecified mitochondrial disorder*	254837	**1592**
Unstable hemoglobin disease	99139	**38**
Upper limb hypertrophy	295049	**≤10**
Upper limb mesomelic dysplasia	2497	**≤10**
Upper tract urothelial carcinoma	598216	**≤10**
Urachal cyst	488	**≤10**
Urachal diverticulum	431347	**≤10**
Urachal sinus	431344	**≤10**
Urban-Rogers-Meyer syndrome	3409	**≤10**
Uremic pruritus	94059	**≤10**
Usher syndrome	886	**1315**
Uterine cervical aplasia and agenesis	180145	**≤10**
Uterine hypoplasia	180139	**≤10**
Uveal coloboma-cleft lip and palate-intellectual disability	1473	**≤10**
Uveal melanoma	39044	**≤10**
VACTERL with hydrocephalus	3412	**46**
VACTERL/VATER association	887	**771**
Vacuolar myopathy with sarcoplasmic reticulum protein aggregates	88635	**≤10**
Vaginal atresia	65681	**32**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Van den Ende-Gupta syndrome	2460	**≤10**
Van der Woude syndrome	888	**285**
Variant ABeta2M amyloidosis	314652	**≤10**
Variant Creutzfeldt-Jakob disease	576370	**≤10**
Vascular Ehlers-Danlos syndrome	286	**953**
Vasculitis due to ADA2 deficiency	404553	**33**
Vasoproliferative tumor of the retina	353356	**38**
Vein of Galen aneurysmal malformation	1053	**114**
Velo-facial-skeletal syndrome	3424	**≤10**
Ventilator-induced diaphragmatic dysfunction	505395	**≤10**
Ventricular extrasystoles with syncopal episodes-perodactyly-Robin sequence syndrome	3201	**≤10**
Ventriculomegaly-cystic kidney disease	443988	**≤10**
Verloove Vanhorick-Brubakk syndrome	3429	**≤10**
Vernal keratoconjunctivitis	70476	**201**
Verrucous hemangioma	464318	**≤10**
Very long chain acyl-CoA dehydrogenase deficiency	26793	**88**
VEXAS syndrome	596753	**126**
Vibratory angioedema	493348	**≤10**
Vici syndrome	1493	**≤10**
VIPoma	97282	**≤10**
Viral myositis	206991	**110**
Vitamin B12-responsive methylmalonic acidemia	28	**65**
Vitamin B12-unresponsive methylmalonic acidemia	27	**114**
Vitamin K antagonist embryofetopathy	1914	**≤10**
Vocal cord and pharyngeal distal myopathy	600	**≤10**
Vogt-Koyanagi-Harada disease	3437	**206**
Von Hippel-Lindau disease	892	**544**
Von Willebrand disease	903	**9244**
Vulvar carcinoma	494418	**≤10**
Vulvovaginal rhabdomyosarcoma	206492	**≤10**
Waardenburg syndrome	3440	**547**
Waardenburg-Shah syndrome	897	**50**
WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	466943	**25**
Wagner disease	898	**25**
WAGR syndrome	893	**61**
Waldenström macroglobulinemia	33226	**153**
Walker-Warburg syndrome	899	**26**
WARS2-related combined oxidative phosphorylation defect	572798	**≤10**
Warts-immunodeficiency-lymphedema-anogenital dysplasia syndrome	568056	**≤10**
Weaver syndrome	3447	**28**
Weill-Marchesani syndrome	3449	**31**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
Weiss-Kruszka Syndrome	502430	**≤10**
Well-differentiated fetal adenocarcinoma of the lung	284395	**≤10**
Wells syndrome	901	**≤10**
Werner syndrome	902	**19**
WHIM syndrome	51636	**≤10**
Whipple disease	3452	**89**
White forelock with malformations	2475	**≤10**
White matter hypoplasia-corpus callosum agenesis-intellectual disability syndrome	3207	**≤10**
White sponge nevus	171723	**≤10**
White-Sutton syndrome	468678	**55**
Whooping cough	1489	**≤10**
Wiedemann-Rautenstrauch syndrome	3455	**≤10**
Wiedemann-Steiner syndrome	319182	**150**
Wild type ABeta2M amyloidosis	85446	**≤10**
Wild type ATTR amyloidosis	330001	**2619**
Wildervanck syndrome	3456	**≤10**
Williams syndrome	904	**1138**
Williams-Campbell syndrome	411501	**≤10**
Wilson disease	905	**1185**
Wilson-Turner syndrome	3459	**≤10**
Wiskott-Aldrich syndrome	906	**189**
Witteveen-Kolk syndrome	500163	**38**
Wolcott-Rallison syndrome	1667	**≤10**
Wolf-Hirschhorn syndrome	280	**117**
Wolfram syndrome	3463	**282**
Wolfram-like syndrome	411590	**40**
Woodhouse-Sakati syndrome	3464	**27**
Woolly hair	170	**≤10**
Woolly hair nevus	79414	**≤10**
Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia	166277	**≤10**
Wound myiasis	165955	**≤10**
Wyburn-Mason syndrome	53719	**≤10**
X small rings	96201	**≤10**
Xanthoma disseminatum	158003	**≤10**
Xeroderma pigmentosum	910	**113**
Xeroderma pigmentosum variant	90342	**11**
Xeroderma pigmentosum-Cockayne syndrome complex	220295	**≤10**
X-linked acrogigantism	300373	**68**
X-linked adrenal hypoplasia congenita	95702	**45**
X-linked adrenoleukodystrophy	43	**557**
X-linked centronuclear myopathy	596	**76**
X-linked Charcot-Marie-Tooth disease type 1	101075	**176**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
X-linked Charcot-Marie-Tooth disease type 2	101076	**36**
X-linked Charcot-Marie-Tooth disease type 3	101077	**≤10**
X-linked Charcot-Marie-Tooth disease type 4	101078	**≤10**
X-linked Charcot-Marie-Tooth disease type 5	99014	**≤10**
X-linked Charcot-Marie-Tooth disease*	64747	**299**
X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome	431140	**≤10**
X-linked cone dysfunction syndrome with myopia	90001	**14**
X-linked corneal dermoid	1661	**≤10**
X-linked creatine transporter deficiency	52503	**98**
X-linked distal spinal muscular atrophy type 3	139557	**≤10**
X-linked dominant chondrodysplasia punctata	35173	**54**
X-linked dominant chondrodysplasia, Chassaing-Lacombe type	163966	**≤10**
X-linked dyserythropoietic anemia with abnormal platelets and neutropenia	363727	**≤10**
X-linked Ehlers-Danlos syndrome	75497	**12**
X-linked epilepsy-learning disabilities-behavior disorders syndrome	85294	**19**
X-linked erythropoietic protoporphyria	443197	**14**
X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	480880	**≤10**
X-linked hereditary sensory and autonomic neuropathy with deafness	139583	**≤10**
X-linked hypophosphatemia	89936	**549**
X-linked ichthyosis syndrome*	281210	**≤10**
X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia	317476	**≤10**
X-linked immunoneurologic disorder	2571	**≤10**
X-linked intellectual disability due to GRIA3 mutations	364028	**≤10**
X-linked intellectual disability, Cabezas type	85293	**≤10**
X-linked intellectual disability, Cantagrel type	85277	**≤10**
X-linked intellectual disability, Hedera type	93952	**≤10**
X-linked intellectual disability, Najm type	163937	**19**
X-linked intellectual disability, Nascimento type	163956	**≤10**
X-linked intellectual disability, Shashi type	85286	**≤10**
X-linked intellectual disability, Siderius type	85287	**≤10**
X-linked intellectual disability, Snyder type	3063	**14**
X-linked intellectual disability, Van Esch type	163976	**≤10**
X-linked intellectual disability-acromegaly-hyperactivity syndrome	85327	**≤10**
X-linked intellectual disability-ataxia-apraxia syndrome	85338	**≤10**
X-linked intellectual disability-cerebellar hypoplasia syndrome	137831	**27**
X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome	459070	**≤10**
X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	1568	**≤10**
X-linked intellectual disability-epilepsy syndrome*	2076	**31**
X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome	85317	**≤10**
X-linked intellectual disability-hypogonadism-ichthyosis-obesity-short stature syndrome	3055	**≤10**
X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	85329	**≤10**
X-linked intellectual disability-hypotonia-movement disorder syndrome	457260	**21**
X-linked intellectual disability-macrocephaly-macroorchidism syndrome	85320	**≤10**
X-linked intellectual disability-psychosis-macroorchidism syndrome	3077	**≤10**
X-linked intellectual disability-retinitis pigmentosa syndrome	85332	**≤10**
X-linked intellectual disability-short stature-overweight syndrome	457240	**≤10**
X-linked lissencephaly with abnormal genitalia	452	**≤10**
X-linked lymphoproliferative disease due to SH2D1A deficiency	538931	**13**
X-linked lymphoproliferative disease due to XIAP deficiency	538934	**23**
X-linked lymphoproliferative disease*	2442	**19**
X-linked mandibulofacial dysostosis	1131	**≤10**
X-linked mendelian susceptibility to mycobacterial diseases	319605	**13**
X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	435938	**≤10**
X-linked myopathy with excessive autophagy	25980	**58**
X-linked myopathy with postural muscle atrophy	178461	**≤10**
X-linked myotubular myopathy-abnormal genitalia syndrome	456328	**≤10**
X-linked non progressive cerebellar ataxia	314978	**≤10**
X-linked osteoporosis with fractures	391330	**≤10**
X-linked progressive cerebellar ataxia	1175	**≤10**
X-linked recessive ocular albinism	54	**133**
X-linked reticulate pigmentary disorder	85453	**≤10**
X-linked retinal dysplasia	1852	**≤10**
X-linked retinoschisis	792	**388**
X-linked scapuloperoneal muscular dystrophy	431272	**≤10**
X-linked severe congenital neutropenia	86788	**≤10**
X-linked sideroblastic anemia	75563	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
X-linked sideroblastic anemia and spinocerebellar ataxia	2802	**≤10**
X-linked skeletal dysplasia-intellectual disability syndrome	1436	**≤10**
X-linked spasticity-intellectual disability-epilepsy syndrome	3175	**≤10**
X-linked spinocerebellar ataxia type 4	85292	**≤10**
X-linked spondyloepimetaphyseal dysplasia	93349	**≤10**
Xp21 deletion syndrome	261476	**≤10**
Xp22.13p22.2 duplication syndrome	284180	**16**
Xp22.3 microdeletion syndrome	1643	**34**
Xq12-q13.3 duplication syndrome	314389	**≤10**
Xq21 microdeletion syndrome	1435	**≤10**
Xq25 microduplication syndrome	521258	**≤10**
Xq27.3q28 duplication syndrome	261483	**15**
XY type gonadal dysgenesis-associated anomalies syndrome	1770	**23**
Yellow nail syndrome	662	**51**
Yolk sac tumor	876	**≤10**
Young adult-onset distal hereditary motor neuropathy	314485	**13**
Young syndrome	3471	**≤10**
Young-onset Parkinson disease	2828	**180**
Yunis-Varon syndrome	3472	**≤10**
Zellweger syndrome	912	**50**
Zellweger-like syndrome without peroxisomal anomalies	50812	**≤10**
Zika virus disease	448237	**≤10**
Zimmermann-Laband syndrome	3473	**≤10**
Zygomycosis	73263	**≤10**

BNDMR
Banque Nationale de Données Maladies Rares

NUMBER OF CASES PER OBSOLETE RARE DISEASE REGISTERED IN THE BNDMR

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: 3-Phosphoglycerate dehydrogenase deficiency*	422519	**≤10**
OBSOLETE: Abnormal origin or aberrant course of coronary artery*	95493	**74**
OBSOLETE: Acheiria, bilateral	295103	**≤10**
OBSOLETE: Acheiria, unilateral	295101	**40**
OBSOLETE: Acquired alimentary behavior disorder of infancy*	138118	**772**
OBSOLETE: Acquired hemophilia	73274	**313**
OBSOLETE: Acquired metabolic neuropathy	206616	**316**
OBSOLETE: Acrocephalosyndactyly*	946	**≤10**
OBSOLETE: Acute cutaneous lupus erythematosus*	163528	**60**
OBSOLETE: Adactyly of foot	435623	**17**
OBSOLETE: Adactyly of foot, bilateral	295118	**≤10**
OBSOLETE: Adactyly of foot, unilateral	295116	**≤10**
OBSOLETE: Adactyly of hand*	294931	**71**
OBSOLETE: Adult pulmonary Langerhans cell histiocytosis	99874	**35**
OBSOLETE: Adult-onset SAPHO syndrome	324982	**≤10**
OBSOLETE: Alpha-1-antichymotrypsin deficiency	93594	**≤10**
OBSOLETE: Amelia of lower limb, bilateral	295059	**≤10**
OBSOLETE: Amelia of upper limb, bilateral	295055	**≤10**
OBSOLETE: Amelia of upper limb, unilateral	295053	**≤10**
OBSOLETE: Amniotic bands*	1034	**149**
OBSOLETE: Aneurysm or dilatation of ascending aorta	95484	**133**
OBSOLETE: Aortopulmonary coronary arterial course	99086	**≤10**
OBSOLETE: Apodia, unilateral	295105	**≤10**
OBSOLETE: Argyrophilic grain disease	97342	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: Arterial hypertension due to renal artery stenosis secondary to vasculitis	97599	**≤10**
OBSOLETE: Arthrogryposis due to muscular dystrophy	1155	**≤10**
OBSOLETE: Aseptic osteitis	57194	**≤10**
OBSOLETE: Atrichia-mental and growth delay syndrome	1211	**≤10**
OBSOLETE: ATR-X-related syndrome*	263355	**≤10**
OBSOLETE: Atypical hemolytic uremic syndrome with B factor anomaly	93578	**≤10**
OBSOLETE: Atypical hemolytic uremic syndrome with C3 anomaly	93575	**≤10**
OBSOLETE: Atypical hemolytic uremic syndrome with H factor anomaly	93579	**≤10**
OBSOLETE: Atypical hemolytic uremic syndrome with I factor anomaly	93580	**≤10**
OBSOLETE: Atypical hemolytic uremic syndrome with MCP/CD46 anomaly	93576	**≤10**
OBSOLETE: Auriculoocular anomalies-cleft lip syndrome	71270	**≤10**
OBSOLETE: Autoimmune enteropathy type 2	103916	**≤10**
OBSOLETE: Autosomal dominant childhood-onset cortical cataract	306561	**≤10**
OBSOLETE: Autosomal dominant focal dystonia, DYT7 type	93963	**≤10**
OBSOLETE: Autosomal dominant Opitz G/BBB syndrome	306588	**≤10**
OBSOLETE: Autosomal dominant optic atrophy and late-onset deafness	255117	**≤10**
OBSOLETE: Autosomal dominant spastic paraplegia type 9	100990	**≤10**
OBSOLETE: Autosomal recessive optic atrophy, OPA6 type	99012	**14**
OBSOLETE: AymÚ-Gripp syndrome	477668	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: Basal epidermolysis bullosa simplex*	158665	**34**
OBSOLETE: Basement membrane disease*	93550	**≤10**
OBSOLETE: Behr syndrome	1239	**≤10**
OBSOLETE: Benign essential blepharospasm	93955	**109**
OBSOLETE: Benign exophthalmos syndrome	71269	**≤10**
OBSOLETE: Blaichman syndrome	1250	**≤10**
OBSOLETE: Blepharophimosis-epicanthus inversus-ptosis due to a point mutation syndrome	261572	**≤10**
OBSOLETE: Blepharophimosis-radioulnar synostosis syndrome	1256	**≤10**
OBSOLETE: Bowed tibiae-radial anomalies-osteopenia-fractures syndrome	3331	**≤10**
OBSOLETE: Brachydactyly of fingers	294996	**≤10**
OBSOLETE: Brachydactyly of fingers, bilateral	295130	**≤10**
OBSOLETE: Brachydactyly of fingers, unilateral	295128	**≤10**
OBSOLETE: Brachydactyly of toes	294998	**≤10**
OBSOLETE: Brachydactyly of toes, unilateral	295132	**≤10**
OBSOLETE: Brachydactyly*	294937	**112**
OBSOLETE: Bullous systemic lupus erythematosus	46489	**13**
OBSOLETE: Central polydactyly of toes	295010	**≤10**
OBSOLETE: Centripetalis recessive dystrophic epidermolysis bullosa	89841	**57**
OBSOLETE: Cerebrorenodigital syndrome	1396	**≤10**
OBSOLETE: Cervical dystonia	93962	**99**
OBSOLETE: Cervicofacial lymphatic malformation	137923	**≤10**
OBSOLETE: Choristoma	91353	**≤10**
OBSOLETE: Chronic pain requiring intraspinal analgesia	95426	**≤10**
OBSOLETE: CINCA syndrome with NLRP3 mutations	93365	**≤10**
OBSOLETE: Circumscribed lymphatic malformation	217410	**13**
OBSOLETE: Classic paraneoplastic limbic encephalitis	163898	**40**
OBSOLETE: Combined hyperlipidemia*	79211	**≤10**
OBSOLETE: Common variable immunodeficiency due to an intrinsic T cell defect	99831	**≤10**
OBSOLETE: Common variable immunodeficiency due to TNFR deficiency	183672	**≤10**
OBSOLETE: Complete atrioventricular canal-left heart obstruction syndrome	99066	**≤10**
OBSOLETE: Congenital absence of both forearm and hand, unilateral	295093	**111**
OBSOLETE: Congenital absence of both lower leg and foot, bilateral	295099	**≤10**
OBSOLETE: Congenital absence of both lower leg and foot, unilateral	295097	**≤10**
OBSOLETE: Congenital absence of thigh and lower leg with foot present, unilateral	295089	**≤10**
OBSOLETE: Congenital absence of upper arm and forearm with hand present, unilateral	295085	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: Congenital absence/hypoplasia of fingers excluding thumb	294990	**11**
OBSOLETE: Congenital absence/hypoplasia of fingers excluding thumb, bilateral	295114	**≤10**
OBSOLETE: Congenital absence/hypoplasia of thumb, bilateral	295112	**≤10**
OBSOLETE: Congenital absence/hypoplasia of thumb, unilateral	295110	**≤10**
OBSOLETE: Congenital adrenal hypoplasia of maternal cause	95701	**≤10**
OBSOLETE: Congenital aortic valve insufficiency	95449	**25**
OBSOLETE: Congenital cataract, Volkmann type	98983	**≤10**
OBSOLETE: Congenital cataract-ichthyosis syndrome	1376	**≤10**
OBSOLETE: Congenital hydronephrosis	2190	**1548**
OBSOLETE: Congenital liver hemangioma	238691	**30**
OBSOLETE: Congenital muscular dystrophy-muscle hypertrophy-severe intellectual disability syndrome	329206	**≤10**
OBSOLETE: Congenital nasal pyriform aperture stenosis with holoprosencephaly	162521	**31**
OBSOLETE: Congenital sucrase-isomaltase deficiency with minimal starch tolerance	306446	**≤10**
OBSOLETE: Congenital sucrase-isomaltase deficiency with starch and lactose intolerance	306474	**≤10**
OBSOLETE: Congenital sucrase-isomaltase deficiency with starch intolerance	306436	**25**
OBSOLETE: Congenital sucrase-isomaltase deficiency without sucrose intolerance	306486	**≤10**
OBSOLETE: Congenital valvular dysplasia	1864	**18**
OBSOLETE: Coronary artery intramyocardial course	99085	**≤10**
OBSOLETE: Corticobasal degeneration	278	**70**
OBSOLETE: Craniosynostosis-cataract syndrome	1530	**≤10**
OBSOLETE: Craniosynostosis-fibular aplasia syndrome	1533	**≤10**
OBSOLETE: Deletion 20p	1611	**≤10**
OBSOLETE: Deletion 4q	1625	**≤10**
OBSOLETE: Developmental delay-deafness syndrome, Hildebrand type	163988	**≤10**
OBSOLETE: Distal monosomy 20q	96152	**≤10**
OBSOLETE: Duplication 4q	1739	**≤10**
OBSOLETE: Early infantile epileptic encephalopathy without suppression burst	369894	**12**
OBSOLETE: Ehlers-Danlos syndrome type 1	90309	**79**
OBSOLETE: Ehlers-Danlos syndrome type 2	90318	**47**
OBSOLETE: Ehlers-Danlos syndrome type 7A	99875	**≤10**
OBSOLETE: Ehlers-Danlos syndrome type 7B	99876	**≤10**
OBSOLETE: Ehlers-Danlos syndrome, fibronectinemic type	75501	**≤10**
OBSOLETE: Embryonary disorganization syndrome	1664	**≤10**
OBSOLETE: Eosinophilic granuloma	99871	**≤10**
OBSOLETE: Epidermal nevus-vitamin D-resistant rickets syndrome	2694	**≤10**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: Epidermolysis bullosa simplex superficialis	89839	**≤10**
OBSOLETE: Epimetaphyseal skeletal dysplasia	1819	**≤10**
OBSOLETE: Epithelio-exfoliative colitis-deafness syndrome	103912	**≤10**
OBSOLETE: Erythromelalgia	1956	**44**
OBSOLETE: Facial asymmetry-temporal seizures syndrome	1167	**≤10**
OBSOLETE: Familial articular chondrocalcinosis type 1	99781	**≤10**
OBSOLETE: Familial capillary hemangioma	91415	**25**
OBSOLETE: Familial chondromalacia patellae	1428	**≤10**
OBSOLETE: Familial esophageal achalasia	99723	**≤10**
OBSOLETE: Familial hyperreninemic hypoaldosteronism type 1	99763	**≤10**
OBSOLETE: Familial hyperreninemic hypoaldosteronism type 2	99764	**≤10**
OBSOLETE: Familial hypospadias	440	**280**
OBSOLETE: Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	93217	**≤10**
OBSOLETE: Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	93213	**52**
OBSOLETE: Familial idiopathic steroid-resistant nephrotic syndrome with minimal changes	93216	**12**
OBSOLETE: Familial juvenile hyperuricemic nephropathy type 1	209886	**52**
OBSOLETE: Familial lambdoid synostosis	3267	**≤10**
OBSOLETE: Familial parathyroid adenoma	99877	**≤10**
OBSOLETE: Familial primary hypomagnesemia with normocalciuria and normocalcemia	34527	**≤10**
OBSOLETE: Familial segmental neurofibromatosis	79428	**91**
OBSOLETE: Familial spinal neurofibromatosis	79429	**≤10**
OBSOLETE: Femoral agenesis/hypoplasia, bilateral	295067	**≤10**
OBSOLETE: Femoral agenesis/hypoplasia, unilateral	295065	**≤10**
OBSOLETE: Fibular hemimelia, bilateral	295083	**≤10**
OBSOLETE: Fibular hemimelia, unilateral	295081	**14**
OBSOLETE: Follicular atrophoderma-basal cell carcinoma	79459	**≤10**
OBSOLETE: Generalized epilepsy and praxis-induced seizures	99649	**≤10**
OBSOLETE: Genetic primary hypomagnesemia with hypocalciuria*	306519	**≤10**
OBSOLETE: Genetic primary hypomagnesemia with normocalciuria*	306522	**≤10**
OBSOLETE: Giant infantile hemangioma	210592	**29**
OBSOLETE: Glycogen storage disease due to acid maltase deficiency, adult onset	308604	**≤10**
OBSOLETE: Glycogen storage disease due to acid maltase deficiency, juvenile onset	308573	**≤10**
OBSOLETE: Graft rejection after lung transplantation	91128	**≤10**
OBSOLETE: Hashimoto-Pritzker syndrome	99872	**≤10**
OBSOLETE: Hemochromatosis type 5	447792	**≤10**
OBSOLETE: Hemolytic anemia due to glyceraldehyde-3-phosphate dehydrogenase deficiency	248305	**≤10**
OBSOLETE: Hereditary motor and sensory neuropathy*	140450	**448**
OBSOLETE: High isolated anorectal malformation	171201	**498**
OBSOLETE: Hirsutism-skeletal dysplasia-intellectual disability syndrome	2156	**≤10**
OBSOLETE: Humeral agenesis/hypoplasia, unilateral	295061	**≤10**
OBSOLETE: Hyperlipoproteinemia type 5	70470	**≤10**
OBSOLETE: Hyperphalangy, bilateral	295142	**≤10**
OBSOLETE: Hypertrichotic osteochondrodysplasia	2765	**≤10**
OBSOLETE: Hypertrophic cardiomyopathy due to intensive athletic training	217601	**≤10**
OBSOLETE: Hypopituitarism-short stature-skeletal anomalies syndrome	2626	**≤10**
OBSOLETE: Idiopathic hypersomnia with long sleep time	228315	**154**
OBSOLETE: Idiopathic hypersomnia without long sleep time	228318	**102**
OBSOLETE: Idiopathic recurrent and disabling cutaneous herpes	35061	**≤10**
OBSOLETE: Idiopathic steroid-sensitive nephrotic syndrome with diffuse mesangial proliferation	93209	**≤10**
OBSOLETE: Idiopathic steroid-sensitive nephrotic syndrome with focal segmental hyalinosis	93206	**186**
OBSOLETE: Idiopathic steroid-sensitive nephrotic syndrome with minimal change	93207	**360**
OBSOLETE: Infantile axonal neuropathy	2679	**39**
OBSOLETE: Infundibulopelvic stenosis-multicystic kidney syndrome	1849	**≤10**
OBSOLETE: Inherited predisposition to essential thrombocythemia	225968	**≤10**
OBSOLETE: Intellectual disability-hypotonia-skin hyperpigmentation syndrome	3050	**≤10**
OBSOLETE: Intellectual disability-microcephaly-phalangeal-facial abnormalities syndrome	3067	**≤10**
OBSOLETE: Intellectual disability-microcephaly-unusual facies syndrome	3313	**≤10**
OBSOLETE: Intellectual disability-unusual facies syndrome	3043	**≤10**
OBSOLETE: Intermediate isolated anorectal malformation	171208	**126**
OBSOLETE: Intracranial aneurysms-multiple congenital anomalies syndrome	1057	**≤10**
OBSOLETE: Isolated cloverleaf skull syndrome	2343	**≤10**
OBSOLETE: Isolated facial myokymia	221106	**≤10**
OBSOLETE: Isolated oxycephaly	63440	**57**
OBSOLETE: Isolated plagiocephaly	35098	**649**
OBSOLETE: Ito hypomelanosis	435	**298**
OBSOLETE: Junctional epidermolysis bullosa, non-Herlitz type	89840	**≤10**
OBSOLETE: Juvenile chronic recurrent multifocal osteomyelitis	2778	**101**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: Juvenile-onset SAPHO syndrome	324989	**≤10**
OBSOLETE: Laminopathy type Decaudain-Vigouroux	137871	**12**
OBSOLETE: Laryngeal dyskinesia	93961	**17**
OBSOLETE: Letterer-Siwe disease	99870	**≤10**
OBSOLETE: Limb dystonia	93957	**41**
OBSOLETE: Limbic encephalitis with caspr2 antibodies	276402	**20**
OBSOLETE: Limbic encephalitis with DPP6 antibodies	329341	**≤10**
OBSOLETE: Limbic encephalitis with LGI1 antibodies	163908	**96**
OBSOLETE: Limbic encephalitis*	163892	**190**
OBSOLETE: Lissencephaly-demyelinating axonal neuropathy syndrome	101356	**≤10**
OBSOLETE: Localized epiphyseal dysplasia	1823	**≤10**
OBSOLETE: Low isolated anorectal malformation	171215	**1101**
OBSOLETE: Low-grade ependymoma	251633	**≤10**
OBSOLETE: Lymphedema praecox	77241	**≤10**
OBSOLETE: Lymphedema tarda	77242	**≤10**
OBSOLETE: Lymphocytic colitis	65279	**≤10**
OBSOLETE: Lymphomatous meningitis	329998	**≤10**
OBSOLETE: Maculopapular lupus rash	90287	**≤10**
OBSOLETE: Madelung deformity	35688	**26**
OBSOLETE: Madelung deformity, bilateral	295223	**13**
OBSOLETE: Maternally-inherited mitochondrial hypertrophic cardiomyopathy	255225	**≤10**
OBSOLETE: MECP2 duplication syndrome	85281	**11**
OBSOLETE: Metastatic spermatocytic seminoma	99866	**≤10**
OBSOLETE: Microcephaly-seizures-developmental delay syndrome	228418	**≤10**
OBSOLETE: Microcornea-corectopia-macular hypoplasia syndrome	2535	**≤10**
OBSOLETE: Microphthalmia-cataract syndrome	2543	**20**
OBSOLETE: Multicentric Castleman disease	93686	**13**
OBSOLETE: Multiple fibroadenoma of the breast	50920	**318**
OBSOLETE: Multiple ventricular septal defects	99096	**12**
OBSOLETE: Mycobacterium xenopi infection	314946	**≤10**
OBSOLETE: Myopathy-growth delay-intellectual disability-hypospadias syndrome	2601	**≤10**
OBSOLETE: Natal teeth-intestinal pseudoobstruction-patent ductus syndrome	1654	**≤10**
OBSOLETE: Neuroepithelioma	2677	**≤10**
OBSOLETE: Neurogenic palpebral tumor	98593	**≤10**
OBSOLETE: Non-distal monosomy 7p	96136	**≤10**
OBSOLETE: Non-dystrophic myopathy with collagen 6 anomaly*	206659	**76**
OBSOLETE: Non-herpetic acute limbic encephalitis	163924	**38**
OBSOLETE: Non-secreting chemodectoma	101106	**≤10**
OBSOLETE: Not NOTCH3-related small vessel disease of the brain	77304	**≤10**
OBSOLETE: Ocular coloboma*	194	**32**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: Oculocerebral dysplasia	2705	**≤10**
OBSOLETE: Oligoarticular juvenile idiopathic arthritis with anti-nuclear antibodies	247839	**321**
OBSOLETE: Oligoarticular juvenile idiopathic arthritis without anti-nuclear antibodies	247846	**80**
OBSOLETE: Ophthalmoplegia-myalgia-tubular aggregates syndrome	2742	**≤10**
OBSOLETE: Osteochondritis of tarsal/metatarsal bone	2054	**≤10**
OBSOLETE: Otopalatodigital syndrome	669	**11**
OBSOLETE: Papillary fibroelastoma of the heart	208600	**≤10**
OBSOLETE: Paraneoplastic limbic encephalitis*	163895	**16**
OBSOLETE: Patella aplasia/hypoplasia, unilateral	295038	**≤10**
OBSOLETE: Pediatric Castleman disease	93682	**≤10**
OBSOLETE: Pediatric polyarteritis nodosa	93564	**≤10**
OBSOLETE: Pediatric Sjögren syndrome	93566	**≤10**
OBSOLETE: Pediatric systemic sclerosis	93567	**≤10**
OBSOLETE: Peripheral hypothyroidism*	226310	**25**
OBSOLETE: Peripheral resistance to thyroid hormones	97927	**≤10**
OBSOLETE: Peters anomaly-cataract syndrome	101033	**≤10**
OBSOLETE: Phosphoenolpyruvate carboxykinase 2 deficiency	79317	**≤10**
OBSOLETE: Pitt-Hopkins-like syndrome	221150	**≤10**
OBSOLETE: Platelet function disease associated with renal insufficiency	99146	**≤10**
OBSOLETE: Polydactyly of a biphalangeal thumb, unilateral	295144	**≤10**
OBSOLETE: Polydactyly of a triphalangeal thumb, bilateral	295150	**≤10**
OBSOLETE: Polymicrogyria-turricephaly-hypogenitalism syndrome	2925	**≤10**
OBSOLETE: Polysyndactyly, bilateral	295161	**≤10**
OBSOLETE: Polysyndactyly, unilateral	295159	**≤10**
OBSOLETE: Postaxial polydactyly of fingers*	294942	**26**
OBSOLETE: Postaxial polydactyly of toes	295008	**≤10**
OBSOLETE: Postaxial polydactyly of toes, unilateral	295179	**≤10**
OBSOLETE: Postaxial polydactyly type B, bilateral	295169	**≤10**
OBSOLETE: Postsurgical hypopituitarism	95621	**≤10**
OBSOLETE: Posttraumatic diabetes insipidus	95625	**≤10**
OBSOLETE: Posttraumatic hypopituitarism	95623	**≤10**
OBSOLETE: Preaxial polydactyly of fingers*	294939	**36**
OBSOLETE: Preaxial polydactyly of toes	295006	**≤10**
OBSOLETE: Preaxial polydactyly of toes, bilateral	295177	**≤10**
OBSOLETE: Primary parathyroid hyperplasia	99878	**62**
OBSOLETE: Pulmonary vein atresia	99126	**≤10**
OBSOLETE: Radial hemimelia, bilateral	295071	**≤10**
OBSOLETE: Radial hemimelia, unilateral	295069	**≤10**
OBSOLETE: Radiation-induced hypopituitarism	95622	**≤10**
OBSOLETE: Rapidly progressive glomerulonephritis	280569	**54**
OBSOLETE: Rare idiopathic male infertility	98345	**29**

Rare Disease	ORPHA code	Cases in the BNDMR as of 01/11/2023
OBSOLETE: Rare major hypertriglyceridemia*	181425	**11**
OBSOLETE: Recessive aplasia cutis congenita of limbs	1115	**≤10**
OBSOLETE: Recessive hereditary methemoglobinemia type 2	139380	**≤10**
OBSOLETE: Renier-Gabreels-Jasper syndrome	93975	**12**
OBSOLETE: Retrocerebellar cyst	269200	**≤10**
OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis with anti-nuclear antibodies	247854	**76**
OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis without anti-nuclear antibodies	247861	**39**
OBSOLETE: Sakati-Nyhan syndrome	3128	**≤10**
OBSOLETE: Secondary acute transverse myelitis	139420	**12**
OBSOLETE: Secondary ciliary dyskinesia	91365	**≤10**
OBSOLETE: Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency	35123	**≤10**
OBSOLETE: Short stature-heart defect-craniofacial anomalies syndrome	1088	**≤10**
OBSOLETE: Shoulder and girdle defects-familial intellectual disability syndrome	2580	**≤10**
OBSOLETE: Shy-Drager syndrome	98932	**≤10**
OBSOLETE: Single ventricular septal defect	99097	**12**
OBSOLETE: Sino-auricular heart block	1260	**≤10**
OBSOLETE: SLC5A6-CDG	521268	**≤10**
OBSOLETE: Solitary median maxillary central incisor syndrome	2286	**23**
OBSOLETE: Spastic diplegia, infantile type	1680	**65**
OBSOLETE: Split foot	294994	**≤10**
OBSOLETE: Split foot, bilateral	295126	**≤10**
OBSOLETE: Split hand	294992	**≤10**
OBSOLETE: Split hand or/and split foot malformation*	294935	**11**
OBSOLETE: Split hand, bilateral	295122	**≤10**
OBSOLETE: Split hand, unilateral	295120	**≤10**
OBSOLETE: Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome	94095	**≤10**
OBSOLETE: Spondylocostal dysostosis-hypospadias-intellectual disability syndrome	329252	**≤10**
OBSOLETE: Sporadic achalasia	99722	**≤10**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation	93222	**≤10**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	93220	**≤10**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	93218	**115**
OBSOLETE: Sporadic idiopathic steroid-resistant nephrotic syndrome with minimal changes	93221	**91**
OBSOLETE: Sporadic Leigh syndrome	255199	**≤10**
OBSOLETE: Sporadic pheochromocytoma	276624	**153**
OBSOLETE: Sporadic secreting paraganglioma	276627	**26**
OBSOLETE: Squamous cell carcinoma of head and neck	67037	**≤10**
OBSOLETE: Superior celosomia	93942	**≤10**
OBSOLETE: Suprabasal epidermolysis bullosa simplex*	158661	**≤10**
OBSOLETE: Symbrachydactyly of hand and foot, unilateral	295136	**12**
OBSOLETE: TACI-related selective deficiency of IgA	99974	**≤10**
OBSOLETE: Terminal limb defects*	294929	**≤10**
OBSOLETE: Terminal transverse defects of arm	93937	**≤10**
OBSOLETE: Tibial hemimelia, bilateral	295079	**≤10**
OBSOLETE: Tibial hemimelia, unilateral	295077	**≤10**
OBSOLETE: Toxic or/and iatrogenic neuropathy	206619	**210**
OBSOLETE: Tracheo-esophageal fistula-hypospadias syndrome	2042	**≤10**
OBSOLETE: Tricho-oculo-dermo-vertebral syndrome	3354	**≤10**
OBSOLETE: Trochlear nerve palsy	99664	**≤10**
OBSOLETE: Truncal dystonia	93956	**≤10**
OBSOLETE: Ulnar hemimelia, bilateral	295073	**≤10**
OBSOLETE: Ulnar hemimelia, unilateral	295075	**≤10**
OBSOLETE: Unclassified metaphyseal chondrodysplasia	90345	**≤10**
OBSOLETE: Unclassified overlapping connective tissue disease	251316	**≤10**
OBSOLETE: Unclassified spondylometaphyseal dysplasia	163678	**≤10**
OBSOLETE: Univentricular heart with single atrio-ventricular valve	99069	**16**
OBSOLETE: Unknown leukodystrophy	84096	**124**
OBSOLETE: Vascular disruption sequence	3160	**≤10**
OBSOLETE: Vitiligo-associated autoimmune disease	247871	**13**
OBSOLETE: Von Hippel anomaly	98941	**21**
OBSOLETE: Xeroderma pigmentosum complementation group A	276249	**≤10**
OBSOLETE: Xeroderma pigmentosum complementation group B	276252	**≤10**
OBSOLETE: Xeroderma pigmentosum complementation group C	276255	**25**
OBSOLETE: Xeroderma pigmentosum complementation group D	276258	**≤10**
OBSOLETE: Xeroderma pigmentosum complementation group E	276261	**≤10**
OBSOLETE: X-linked acrogigantism due to Xq26 microduplication	448372	**≤10**
OBSOLETE: X-linked dominant intellectual disability-epilepsy syndrome	93951	**≤10**
OBSOLETE: X-linked intellectual disability, Raynaud type	3061	**≤10**
OBSOLETE: X-linked Opitz G/BBB syndrome	306597	**≤10**
OBSOLETE: X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome	83648	**≤10**
OBSOLETE: X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome	83648	**≤10**

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