



Orphanet Report Series

Rare Diseases collection

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Rare Disease Registries in Europe

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Table of contents

Methodology	3
List of rare diseases that are covered by the listed registries (by alphabetical order)	4
Summary	14
1- <i>Distribution of registries by country</i>	14
2- <i>Distribution of registries by coverage</i>	15
3- <i>Distribution of registries by affiliation</i>	15
List of registries by country.....	16
European registries.....	48
International registries coordinated in Europe	52



Methodology

Patient registries and databases constitute key instruments to develop clinical research in the field of rare diseases (RD), to improve patient care and healthcare planning. They are the only way to pool data in order to achieve a sufficient sample size for epidemiological and/or clinical research. They are vital to assess the feasibility of clinical trials, to facilitate the planning of appropriate clinical trials and to support the enrolment of patients.

Registries of patients treated with orphan drugs are particularly relevant as they allow the gathering of evidence on the effectiveness of the treatment and on its possible side effects, keeping in mind that marketing authorization is usually granted at a time when evidence is still limited although already somewhat convincing.

This report gathers the information collected by Orphanet so far, regarding systematic collections of data for a specific disease or a group of diseases.

The report includes data about EU countries and other countries participating to the [Orphanet network](#). The diseases or groups of diseases covered by each register are provided with their Orpha number which is the unique identifier in the Orphanet nomenclature. Data in this report reflects the last update made in the Orphanet database by the registry holders and may not depict changes made to the registries since.

List of rare diseases that are covered by the listed registries (by alphabetical order)

ORPHA code	Disease name
567	22q11.2 deletion syndrome
2982	46,XX disorder of sex development
98078	46,XX disorder of sex development induced by androgens excess
2138	46,XX ovotesticular disorder of sex development
752	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
753	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency
325351	46,XY disorder of sex development of endocrine origin
168558	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
13	6-pyruvoyl-tetrahydropterin synthase deficiency
48818	Aceruloplasminemia
15	Achondroplasia
49382	Achromatopsia
101963	Acquired chronic primary adrenal insufficiency
73274	Acquired hemophilia
79356	Acrokeratoderma
963	Acromegaly
99892	ACTH-dependent Cushing syndrome
318	Acute erythroid leukemia
293173	Acute generalized exanthematous pustulosis
98916	Acute inflammatory demyelinating polyradiculoneuropathy
79276	Acute intermittent porphyria

79126	Acute interstitial pneumonia
513	Acute lymphoblastic leukemia
518	Acute megakaryoblastic leukemia
514	Acute monoblastic/monocytic leukemia
519	Acute myeloid leukemia
517	Acute myelomonocytic leukemia
35889	Acute opioid poisoning
520	Acute promyelocytic leukemia
309120	Acyl-CoA dehydrogenase deficiency
85138	Addison disease
45	Adenosine monophosphate deaminase deficiency
100091	Adrenal/paraganglial tumor
1501	Adrenocortical carcinoma
139399	Adrenomyeloneuropathy
86875	Adult T-cell leukemia/lymphoma
829	Adult-onset Still disease
300846	Aggressive B-cell non-Hodgkin lymphoma
86873	Aggressive NK-cell leukemia
1164	Allergic bronchopulmonary aspergillosis
60	Alpha-1-antitrypsin deficiency
61	Alpha-mannosidosis
846	Alpha-thalassemia
63	Alport syndrome
64	Alström syndrome
2131	Alternating hemiplegia of childhood

284	Alveolar echinococcosis
803	Amyotrophic lateral sclerosis
251630	Anaplastic oligodendroglioma
754	Androgen insensitivity syndrome
72	Angelman syndrome
1069	Aniridia-absent patella syndrome
1065	Aniridia-cerebellar ataxia-intellectual disability syndrome
96346	Anorectal malformation
88632	Anterior segment developmental anomaly
375	Anti-glomerular basement membrane disease
2299	Aortic arch interruption
87	Apert syndrome
320	Apparent mineralocorticoid excess
90	Argininemia
23	Argininosuccinic aciduria
35708	Aromatic L-amino acid decarboxylase deficiency
247	Arrhythmogenic right ventricular cardiomyopathy
94	Astrocytoma
1168	Ataxia-oculomotor apraxia type 1
100	Ataxia-telangiectasia
370109	Ataxia-telangiectasia variant
251347	Ataxia-telangiectasia-like disorder
1201	Atresia of small intestine
85447	ATTRV30M amyloidosis
2134	Atypical hemolytic uremic syndrome
98375	Autoimmune hemolytic anemia

2137	Autoimmune hepatitis
3261	Autoimmune lymphoproliferative syndrome
3453	Autoimmune polyendocrinopathy type 1
3143	Autoimmune polyendocrinopathy type 2
747	Autoimmune pulmonary alveolar proteinosis
71203	Autoimmune thrombocytopenia
93665	Autoinflammatory syndrome
99	Autosomal dominant cerebellar ataxia
98352	Autosomal dominant disease with diffuse palmoplantar keratoderma as a major feature
266	Autosomal dominant limb-girdle muscular dystrophy type 1A
730	Autosomal dominant polycystic kidney disease
34149	Autosomal dominant tubulointerstitial kidney disease
79278	Autosomal erythropoietic protoporphyria
281217	Autosomal ichthyosis syndrome
281097	Autosomal recessive congenital ichthyosis
101150	Autosomal recessive dopa-responsive dystonia
731	Autosomal recessive polycystic kidney disease
2512	Autosomal recessive primary microcephaly
300345	Autosomal systemic lupus erythematosus
782	Axenfeld-Rieger syndrome
67038	B-cell chronic lymphocytic leukemia
86852	B-cell prolymphocytic leukemia
110	Bardet-Biedl syndrome
111	Barth syndrome
112	Bartter syndrome
98895	Becker muscular dystrophy
116	Beckwith-Wiedemann syndrome
117	Behçet disease

274	Bernard-Soulier syndrome
848	Beta-thalassemia
231214	Beta-thalassemia major
610	Bethlem myopathy
415286	Bilirubin encephalopathy
179	Birdshot chorioretinopathy
122	Birt-Hogg-Dubé syndrome
124	Blackfan-Diamond anemia
90340	Blau syndrome
16	Blue cone monochromatism
223727	Bone sarcoma
168598	Brain demyelination due to methionine adenosyltransferase deficiency
352649	Brain dopamine-serotonin vesicular transport disease
97287	Bronchial neuroendocrine tumor
1303	Bronchiolitis obliterans with obstructive pulmonary disease
70589	Bronchopulmonary dysplasia
131	Budd-Chiari syndrome
543	Burkitt lymphoma
329931	C3 glomerulonephritis
280062	Calciophylaxis
137667	Capillary malformation-arteriovenous malformation
147	Carbamoyl-phosphate synthetase 1 deficiency
56044	Carcinoma of gallbladder and extrahepatic biliary tract
97286	Carney-Stratakis syndrome
464343	Catastrophic antiphospholipid syndrome
3027	Caudal regression sequence
86870	CD4+/CD56+ hematodermic neoplasm
597	Central core disease
595	Centronuclear myopathy

79158	Cerebral organic aciduria
166	Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy
55880	Chondrosarcoma
251896	Choroid plexus tumor
180	Choroideremia
379	Chronic granulomatous disease
2932	Chronic inflammatory demyelinating polyneuropathy
521	Chronic myeloid leukemia
324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis
101959	Chronic primary adrenal insufficiency
247525	Citrullinemia type I
466026	Class I glucose-6-phosphate dehydrogenase deficiency
71277	Classic glucose transporter type 1 deficiency syndrome
391	Classic Hodgkin lymphoma
394	Classic homocystinuria
79163	Classic organic aciduria
1991	Cleft lip with or without cleft palate
2014	Cleft palate
31824	Colchicine poisoning
468672	Colobomatous macropthalmia-microcornea syndrome
1198	Colonic atresia
35909	Combined deficiency of factor V and factor VIII
1572	Common variable immunodeficiency
1329	Complete atrioventricular septal defect
1872	Cone rod dystrophy
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral
418	Congenital adrenal hyperplasia

3319	Congenital amegakaryocytic thrombocytopenia
48	Congenital bilateral absence of vas deferens
2140	Congenital diaphragmatic hernia
137	Congenital disorder of glycosylation
85	Congenital dyserythropoietic anemia
98873	Congenital dyserythropoietic anemia type II
327	Congenital factor VII deficiency
329	Congenital factor XI deficiency
331	Congenital factor XIII deficiency
2020	Congenital fiber-type disproportion myopathy
335	Congenital fibrinogen deficiency
174590	Congenital hypogonadotropic hypogonadism
442	Congenital hypothyroidism
1928	Congenital lobar emphysema
69063	Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization
97242	Congenital muscular dystrophy
370953	Congenital muscular dystrophy due to dystroglycanopathy
75840	Congenital muscular dystrophy, Ullrich type
590	Congenital myasthenic syndrome
97245	Congenital myopathy
98904	Congenital myopathy with excess of thin filaments
206973	Congenital myotonia
2444	Congenital pulmonary airway malformation
2414	Congenital pulmonary lymphangiectasia
3161	Congenital pulmonary sequestration
3090	Congenital pulmonary venous return anomaly
2040	Congenital respiratory-biliary fistula

3091	Congenital systemic veins anomaly
93583	Congenital thrombotic thrombocytopenic purpura
169826	Congenital vitamin K-dependent coagulation factors deficiency
860	Congenitally uncorrected transposition of the great arteries
2445	Conotruncal heart malformations
293830	Constitutional dyserythropoietic anemia
319651	Constitutional megaloblastic anemia with severe neurologic disease
252202	Constitutional mismatch repair deficiency syndrome
101987	Constitutional neutropenia
98038	Cranial malformation
208650	Cryopyrin-associated periodic syndrome
553	Cushing syndrome
79140	Cutaneous neuroendocrine carcinoma
400	Cystic echinococcosis
586	Cystic fibrosis
213	Cystinosis
397587	Deep dermatophytosis
1652	Dent disease
220	Denys-Drash syndrome
221	Dermatomyositis
98909	Desminopathy
873	Desmoid tumor
83469	Desmoplastic small round cell tumor
98553	Developmental defect of the eye
1666	Dextrocardia
98043	Diaphragmatic or abdominal wall malformation
146	Differentiated thyroid carcinoma
90060	Diffuse alveolar hemorrhage
497188	Diffuse intrinsic pontine glioma
544	Diffuse large B-cell lymphoma

31828	Digitalis poisoning
226	Dihydropteridine reductase deficiency
309830	Disorder of catecholamine synthesis
79175	Disorder of gamma-aminobutyric acid metabolism
309819	Disorder of pterin metabolism
90771	Disorder of sex development
79167	Disorder of urea cycle metabolism and ammonia detoxification
599	Distal myopathy
18	Distal renal tubular acidosis
34516	DNAJB6-related limb-girdle muscular dystrophy D1
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency
230	Dopamine beta-hydroxylase deficiency
870	Down syndrome
33069	Dravet syndrome
139402	Drug rash with eosinophilia and systemic symptoms
262	Duchenne and Becker muscular dystrophy
98896	Duchenne muscular dystrophy
1203	Duodenal atresia
303	Dystrophic epidermolysis bullosa
91492	Early-onset non-syndromic cataract
1880	Ebstein malformation
98249	Ehlers-Danlos syndrome
289	Ellis Van Creveld syndrome
261	Emery-Dreifuss muscular dystrophy
85438	Enthesitis-related juvenile idiopathic arthritis
301	Ependymal tumor
302	Epidermodysplasia verruciformis
257	Epidermolysis bullosa simplex with muscular dystrophy
79355	Erythrokeratoderma

1199	Esophageal atresia
3318	Essential thrombocythemia
31826	Ethylene glycol poisoning
1959	Evans syndrome
883	Extragenital teratoma
324	Fabry disease
141229	Facial cleft
269	Facioscapulohumeral dystrophy
733	Familial adenomatous polyposis
404560	Familial atypical multiple mole melanoma syndrome
313846	Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome
540	Familial hemophagocytic lymphohistiocytosis
93372	Familial hypocalciuric hypercalcemia type 1
342	Familial Mediterranean fever
99361	Familial medullary thyroid carcinoma
618	Familial melanoma
569	Familial or sporadic hemiplegic migraine
213517	Familial ovarian cancer
1333	Familial pancreatic carcinoma
319487	Familial papillary or follicular thyroid carcinoma
31043	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement
1331	Familial prostate cancer
231108	Familial rhabdoid tumor
84	Fanconi anemia
101039	Female restricted epilepsy with intellectual disability
1987	Femoral agenesis/hypoplasia
853	Fetal and neonatal alloimmune thrombocytopenia
34515	FKRP-related limb-girdle muscular dystrophy R9

1866	Focal, segmental or multifocal dystonia
86902	Follicular dendritic cell sarcoma
545	Follicular lymphoma
51208	Formiminoglutamic aciduria
2795	Fowler urethral sphincter dysfunction syndrome
347	Frasier syndrome
95	Friedreich ataxia
98535	Frontotemporal degeneration with dementia
227796	Fundus albipunctatus
519930	Fungal keratitis
352	Galactosemia
314022	Gastric adenocarcinoma and proximal polyposis of the stomach
36273	Gastric linitis plastica
100092	Gastroenteropancreatic neuroendocrine neoplasm
44890	Gastrointestinal stromal tumor
2368	Gastroschisis
355	Gaucher disease
77259	Gaucher disease type 1
77260	Gaucher disease type 2
77261	Gaucher disease type 3
36387	Generalized epilepsy with febrile seizures-plus
101960	Genetic chronic primary adrenal insufficiency
183497	Genetic neuromuscular disease
435554	Genetic precocious puberty
34526	Genetic primary hypomagnesemia
183592	Genetic renal tubular disease
656	Genetic steroid-resistant nephrotic syndrome
358	Gitelman syndrome
849	Glanzmann thrombasthenia
360	Glioblastoma

25	Glutaryl-CoA dehydrogenase deficiency
407	Glycine encephalopathy
365	Glycogen storage disease due to acid maltase deficiency
367	Glycogen storage disease due to glycogen branching enzyme deficiency
366	Glycogen storage disease due to glycogen debranching enzyme deficiency
284426	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency
137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency
99849	Glycogen storage disease due to muscle beta-enolase deficiency
368	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
371	Glycogen storage disease due to muscle phosphofructokinase deficiency
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency
97234	Glycogen storage disease due to phosphoglycerate mutase deficiency
370	Glycogen storage disease due to phosphorylase kinase deficiency
602	GNE myopathy
377	Gorlin syndrome
53693	GRACILE syndrome
900	Granulomatosis with polyangiitis
721	Gray platelet syndrome
2102	GTP cyclohydrolase I deficiency
168569	H syndrome
99803	Haddad syndrome
73229	HANAC syndrome
163596	Hb Bart's hydrops fetalis

2130	Hemimelia
139491	Hemochromatosis type 4
68364	Hemoglobinopathy
766	Hemolytic anemia due to red cell pyruvate kinase deficiency
158032	Hemophagocytic syndrome
448	Hemophilia
98878	Hemophilia A
98879	Hemophilia B
890	Hepatic veno-occlusive disease
449	Hepatoblastoma
88673	Hepatocellular carcinoma
64743	Hepatoportal sclerosis
91378	Hereditary angioedema
100050	Hereditary angioedema type 1
100051	Hereditary angioedema type 2
145	Hereditary breast and ovarian cancer syndrome
227535	Hereditary breast cancer
676	Hereditary chronic pancreatitis
79273	Hereditary coproporphria
26106	Hereditary diffuse gastric cancer
2024	Hereditary gingival fibromatosis
774	Hereditary hemorrhagic telangiectasia
523	Hereditary leiomyomatosis and renal cell cancer
443909	Hereditary nonpolyposis colon cancer
79357	Hereditary palmoplantar keratoderma
29072	Hereditary pheochromocytoma-paranglioma
264675	Hereditary pulmonary alveolar proteinosis
94088	Hereditary renal hypouricemia
213524	Hereditary site-specific ovarian cancer syndrome
98365	Hereditary stomatocytosis

388	Hirschsprung disease
86896	Histiocytic sarcoma
2162	Holoprosencephaly
395	Homocystinuria due to methylene tetrahydrofolate reductase deficiency
3322	Hoyeraal-Hreidarsson syndrome
56970	Human prion disease
399	Huntington disease
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius
927	Hyperammonemia due to N-acetylglutamate synthase deficiency
343	Hyperimmunoglobulin emia D with periodic fever
289891	Hypermethioninemia due to glycine N-methyltransferase deficiency
289290	Hypermethioninemia encephalopathy due to adenosine kinase deficiency
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
99880	Hyperparathyroidism-jaw tumor syndrome
238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency
31740	Hypersensitivity pneumonitis
437	Hypophosphatemic rickets
2248	Hypoplastic left heart syndrome
79354	Ichthyosis
60033	Idiopathic bronchiectasis
182101	Idiopathic eosinophilic pneumonia
33208	Idiopathic hypersomnia
98482	Idiopathic inflammatory myopathy
98300	Idiopathic interstitial pneumonia
2032	Idiopathic pulmonary fibrosis
99931	Idiopathic pulmonary hemosiderosis
284264	IgG4-related disease

49041	IgG4-related retroperitoneal fibrosis
3002	Immune thrombocytopenia
98290	Immunodeficiency-associated lymphoproliferative disease
761	Immunoglobulin A vasculitis
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
611	Inclusion body myositis
178557	Indolent primary cutaneous B-cell lymphoma
238455	Infantile dystonia-parkinsonism
772	Infantile Refsum disease
90003	Inflammatory pseudotumor of the liver
140162	Inherited cancer-predisposing syndrome
319462	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations
79361	Inherited epidermolysis bullosa
252190	Inherited nervous system cancer-predisposing syndrome
319328	Inherited renal cancer-predisposing syndrome
71862	Inherited retinal disorder
1478	Interatrial communication
86900	Interdigitating dendritic cell sarcoma
37202	Interstitial cystitis
182095	Interstitial lung disease
264735	Interstitial lung disease specific to adulthood
104010	Intestinal polyposis syndrome
1048	Isolated anencephaly/exencephaly
250923	Isolated aniridia
557	Isolated anorectal malformation
30391	Isolated biliary atresia

2542	Isolated microphthalmia-anophthalmia-coloboma
718	Isolated Pierre Robin syndrome
2924	Isolated polycystic liver disease
823	Isolated spina bifida
2440	Isolated split hand-split foot malformation
33	Isovaleric acidemia
474	Jeune syndrome
220497	Joubert syndrome with renal defect
248111	Juvenile Huntington disease
92	Juvenile idiopathic arthritis
2322	Kabuki syndrome
2331	Kawasaki disease
2908	Kindler syndrome
99978	Klatskin tumor
33543	Kleine-Levin syndrome
99749	Kostmann syndrome
59135	Laing early-onset distal myopathy
258	Laminin subunit alpha 2-related congenital muscular dystrophy
98301	Laminopathy
389	Langerhans cell histiocytosis
86897	Langerhans cell sarcoma
626	Large congenital melanocytic nevus
1202	Larynx atresia
65	Leber congenital amaurosis
54260	Left ventricular noncompaction
549	Legionellosis
137605	Legius syndrome
2382	Lennox-Gastaut syndrome
68356	Leukodystrophy
524	Li-Fraumeni syndrome
263	Limb-girdle muscular dystrophy
69663	Low phospholipid-associated cholelithiasis
538	Lymphangiomyomatosis
223735	Lymphoma

144	Lynch syndrome
309337	Lysosomal glycogen storage disease
592	Macrophagic myofasciitis
673	Malaria
679	Malignant atrophic papulosis
423	Malignant hyperthermia of anesthesia
168999	Malignant melanoma of the mucosa
293181	Malignant migrating focal seizures of infancy
398043	Malignant tumor of penis
52417	MALT lymphoma
52416	Mantle cell lymphoma
558	Marfan syndrome
300912	Marginal zone lymphoma
559	Marinesco-Sjögren syndrome
98292	Mastocytosis
3097	Meacham syndrome
2006	Median cleft lip/mandibule
1332	Medullary thyroid carcinoma
616	Medulloblastoma
97338	Melanoma of soft tissue
31825	Methanol poisoning
2169	Methylcobalamin deficiency type cbIE
2170	Methylcobalamin deficiency type cbIG
26	Methylmalonic acidemia with homocystinuria
79284	Methylmalonic acidemia with homocystinuria type cbIF
79282	Methylmalonic acidemia with homocystinuria, type cbIC
369955	Methylmalonic acidemia with homocystinuria, type cbIJ
280183	Methylmalonic aciduria due to transcobalamin receptor defect
83642	Microcytic anemia with liver iron overload

98555	Microphthalmia-anophthalmia-coloboma
568	Microphthalmia, Lenz type
83463	Microtia
68380	Mitochondrial disease
217613	Mitochondrial disease with dilated cardiomyopathy
225700	Mitochondrial disease with epilepsy
217587	Mitochondrial disease with hypertrophic cardiomyopathy
225703	Mitochondrial disease with peripheral neuropathy
206966	Mitochondrial myopathy
552	MODY
3057	Monoamine oxidase A deficiency
98503	Motor neuron disease
575	Muckle-Wells syndrome
79213	Mucopolysaccharidoses
579	Mucopolysaccharidosis type 1
580	Mucopolysaccharidosis type 2
641	Multifocal motor neuropathy
68341	Multiple congenital anomalies/dysmorphic syndrome
652	Multiple endocrine neoplasia type 1
247698	Multiple endocrine neoplasia type 2A
247709	Multiple endocrine neoplasia type 2B
276152	Multiple endocrine neoplasia type 4
29073	Multiple myeloma
321	Multiple osteochondromas
228145	Multiple sclerosis variant
102	Multiple system atrophy
588	Muscle-eye-brain disease
71864	Muscular channelopathy
98473	Muscular dystrophy
247798	MUTYH-related attenuated familial adenomatous polyposis
589	Myasthenia gravis

52688	Myelodysplastic syndrome
98274	Myeloproliferative neoplasm
182050	MYH9-related disease
593	Myofibrillar myopathy
206647	Myotonic dystrophy
2073	Narcolepsy type 1
83465	Narcolepsy type 2
150	Nasopharyngeal carcinoma
607	Nemaline myopathy
654	Nephroblastoma
223	Nephrogenic diabetes insipidus
137617	Nephrogenic systemic fibrosis
655	Nephronophthisis
3388	Neural tube defect
263440	Neuroacanthocytosis
635	Neuroblastoma
2481	Neurocutaneous melanocytosis
385	Neurodegeneration with brain iron accumulation
306719	Neurodegenerative disease with chorea
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency
877	Neuroendocrine neoplasm
636	Neurofibromatosis type 1
637	Neurofibromatosis type 2
2678	Neurofibromatosis type 6
35705	Neurometabolic disorder due to serine deficiency
68381	Neuromuscular disease
71211	Neuromyelitis optica
216	Neuronal ceroid lipofuscinosis
77292	Niemann-Pick disease type A
77293	Niemann-Pick disease type B
646	Niemann-Pick disease type C
647	Nijmegen breakage syndrome
240760	Nijmegen breakage syndrome-like disorder

467	Non-acquired combined pituitary hormone deficiency
631	Non-acquired isolated growth hormone deficiency
94080	Non-functioning paraganglioma
547	Non-Hodgkin lymphoma
157987	Non-Langerhans cell histiocytosis
91364	Non-specific interstitial pneumonia
498467	Non-syndromic postaxial polydactyly
498464	Non-syndromic preaxial polydactyly
165704	Non-syndromic urogenital tract malformation
648	Noonan syndrome
98733	Noonan syndrome and Noonan-related syndrome
3032	NPHP3-related Meckel-like syndrome
1125	Ocular motor apraxia, Cogan type
157962	Oculoauricular syndrome, Schorderet type
534	Oculocerebrorenal syndrome of Lowe
660	Omphalocele
661	Ondine syndrome
2086	Optic pathway glioma
289899	Organic aciduria
664	Ornithine transcarbamylase deficiency
139039	Orofacial clefting syndrome
666	Osteogenesis imperfecta
399293	Osteonecrosis of the jaw
2781	Osteopetrosis and related disorders
668	Osteosarcoma
213500	Ovarian cancer
93460	Overgrowth syndrome
677	Pancreatoblastoma
31827	Paraquat poisoning
143	Parathyroid carcinoma
851	Paris-Trousseau thrombocytopenia
447	Paroxysmal nocturnal hemoglobinuria

1330	Partial atrioventricular septal defect
93126	Pauci-immune glomerulonephritis
33402	Pediatric hepatocellular carcinoma
93552	Pediatric systemic lupus erythematosus
206976	Periodic paralysis
709	Peters plus syndrome
2869	Peutz-Jeghers syndrome
42642	PFAPA syndrome
716	Phenylketonuria
99408	Pituitary adenoma
50251	Pleural mesothelioma
64742	Pleuropulmonary blastoma
2911	Poland syndrome
79358	Porokeratosis
101330	Porphyria cutanea tarda
79473	Porphyria variegata
739	Prader-Willi syndrome
99860	Precursor B-cell acute lymphoblastic leukemia
186	Primary biliary cholangitis
46135	Primary central nervous system lymphoma
244	Primary ciliary dyskinesia
541	Primary cutaneous CD30+ T-cell lymphoproliferative disease
542	Primary cutaneous lymphoma
416	Primary hyperoxaluria
101997	Primary immunodeficiency
35689	Primary lateral sclerosis
168807	Primary malignant peritoneal tumor
54370	Primary membranoproliferative glomerulonephritis
824	Primary myelofibrosis
168803	Primary peritoneal tumor
2420	Primary pulmonary lymphoma
171	Primary sclerosing cholangitis

854	Primitive portal vein thrombosis
35	Propionic acidemia
606	Proximal myotonic myopathy
70	Proximal spinal muscular atrophy
756	Pseudohypoaldosteronism type 1
757	Pseudohypoaldosteronism type 2
97593	Pseudohypoparathyroidism
758	Pseudoxanthoma elasticum
88618	Psychomotor delay due to S-adenosylhomocysteine hydrolase deficiency
306498	PTEN hamartoma tumor syndrome
1578	Pterin-4 alpha-carbinolamine dehydratase deficiency
182090	Pulmonary arterial hypertension
69126	Pyogenic arthritis-pyoderma gangrenosum-acne syndrome
207090	Qualitative or quantitative defects of collagen 6
207085	Qualitative or quantitative defects of dystrophin
209188	Qualitative or quantitative defects of emerin
207119	Qualitative or quantitative defects of FKRP
207052	Qualitative or quantitative defects of sarcoglycan
209053	Qualitative or quantitative defects of titin
93321	Radial hemimelia
71289	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome
165711	Rare abdominal surgical disease
98050	Rare allergic disease
102002	Rare ataxia
93419	Rare bone disease
68411	Rare bone tumor
180250	Rare breast tumor

101945	Rare bronchopulmonary tumor
97929	Rare cardiac disease
218436	Rare cardiac rhythm disease
167848	Rare cardiomyopathy
68335	Rare chromosomal anomaly
98028	Rare circulatory system disease
88991	Rare congenital non-syndromic heart malformation
183651	Rare constitutional anemia
535	Rare cutaneous lupus erythematosus
68361	Rare deafness
93890	Rare developmental defect during embryogenesis
139030	Rare developmental defect with connective tissue involvement
101952	Rare diabetes mellitus
98059	Rare digestive tumor
280275	Rare disease
98032	Rare disease in anaesthesiology
98066	Rare disease in plastic and reconstructive surgery
98067	Rare disease in surgical orthopedic
108999	Rare disorder due to toxic effects
506207	Rare disorder potentially indicated for transplant
101953	Rare dyslipidemia
68363	Rare dystonia
97978	Rare endocrine disease
101998	Rare epilepsy
97935	Rare gastroenterologic disease
101936	Rare gastroesophageal disease
180821	Rare gastroesophageal tumor
183625	Rare genetic diabetes mellitus
98053	Rare genetic disease
391799	Rare genetic dystonia
101435	Rare genetic eye disease

158300	Rare genetic hematologic disease
307052	Rare genetic parkinsonian disorder
96344	Rare gynecologic or obstetric disease
97992	Rare hematologic disease
248308	Rare hemorrhagic disorder
248315	Rare hemorrhagic disorder due to a coagulation factors defect
68334	Rare hemorrhagic disorder due to a constitutional coagulation factors defect
71202	Rare hemorrhagic disorder due to a constitutional platelet anomaly
275729	Rare hemorrhagic disorder due to a constitutional thrombocytopenia
101943	Rare hepatic and biliary tract tumor
57146	Rare hepatic disease
183518	Rare hereditary ataxia
220489	Rare hereditary hemochromatosis
217454	Rare hereditary thrombophilia
98004	Rare immune disease
68367	Rare inborn errors of metabolism
68416	Rare infectious disease
98047	Rare infertility
104012	Rare inflammatory bowel disease
87277	Rare intellectual disability
117569	Rare intestinal disease
180257	Rare malignant breast tumor
68329	Rare maxillo-facial surgical disease
250908	Rare neoplastic disease
98062	Rare nervous system tumor
98006	Rare neurologic disease
98026	Rare odontologic disease
97966	Rare ophthalmic disorder
98036	Rare otorhinolaryngologic disease

98061	Rare otorhinolaryngologic tumor
101937	Rare pancreatic disease
181415	Rare primary hyperaldosteronism
101944	Rare pulmonary disease
71198	Rare pulmonary hypertension
93626	Rare renal disease
93619	Rare renal tumor
97955	Rare respiratory disease
89826	Rare skin disease
79386	Rare skin tumor or hamartoma
71209	Rare soft tissue tumor
138221	Rare sucking/swallowing disorder
97965	Rare surgical cardiac disease
98065	Rare surgical neurologic disease
97962	Rare surgical thoracic disease
98023	Rare systemic or rheumatologic disease
280342	Rare systemic or rheumatological disease of childhood
52662	Rare teratologic disease
248361	Rare thrombotic disorder due to a constitutional coagulation factors defect
98057	Rare tumor
104011	Rare tumor of intestine
180824	Rare tumor of pancreas
101433	Rare urogenital disease
182114	Rare urogenital tumor
180151	Rare vaginal malformation
68362	Rare vascular disease
101938	Rare vascular liver disease
211237	Rare vascular tumor
268114	RAS-associated autoimmune leukoproliferative disease
461	Recessive X-linked ichthyosis

90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients
60032	Recurrent respiratory papillomatosis
97239	Reducing body myopathy
773	Refsum disease
1848	Renal agenesis, bilateral
566243	Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta
791	Retinitis pigmentosa
790	Retinoblastoma
90050	Retinopathy of prematurity
778	Rett syndrome
69077	Rhabdoid tumor
85408	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis
59315	Rhombencephalosynapsis
140976	RHYNS syndrome
1440	Ring chromosome 14 syndrome
158014	Rosai-Dorfman disease
2909	Rothmund-Thomson syndrome
140969	Saldino-Mainzer syndrome
797	Sarcoidosis
799	Schizencephaly
93921	Schwannomatosis
801	Scleroderma
3156	Senior-Loken syndrome
42738	Severe congenital neutropenia
745	Severe hereditary thrombophilia due to congenital protein C deficiency
3162	Sézary syndrome
90038	Shiga toxin-associated hemolytic uremic syndrome
811	Shwachman-Diamond syndrome
232	Sickle cell anemia

275752	Sickle cell disease and related diseases
1047	Sideroblastic anemia
813	Silver-Russell syndrome
816	Sjögren-Larsson syndrome
319	Skeletal Ewing sarcoma
820	Sneddon syndrome
64753	Spinocerebellar ataxia with axonal neuropathy type 2
247234	Sporadic adult-onset ataxia of unknown etiology
276621	Sporadic pheochromocytoma/secretory paraganglioma
826	Sporotrichosis
500481	Squamous cell carcinoma of salivary glands
494547	Squamous cell carcinoma of the hypopharynx
494550	Squamous cell carcinoma of the larynx
500464	Squamous cell carcinoma of the nasal cavity and paranasal sinuses
502363	Squamous cell carcinoma of the oral cavity
500478	Squamous cell carcinoma of the oropharynx
827	Stargardt disease
273	Steinert myotonic dystrophy
36426	Stevens-Johnson syndrome
95455	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum
98557	Syndromic aniridia
182228	Systemic autoimmune disease
188	Systemic capillary leak syndrome
536	Systemic lupus erythematosus
158	Systemic primary carnitine deficiency
90291	Systemic sclerosis
85414	Systemic-onset juvenile idiopathic arthritis
86872	T-cell large granular lymphocyte leukemia

171918	T-cell non-Hodgkin lymphoma
86871	T-cell prolymphocytic leukemia
842	Testicular seminomatous germ cell tumor
3303	Tetralogy of Fallot
86846	Therapy related acute myeloid leukemia and myelodysplastic syndrome
49827	Thiamine-responsive megaloblastic anemia syndrome
67044	Thrombocytopenia with congenital dyserythropoietic anemia
3320	Thrombocytopenia-absent radius syndrome
93573	Thrombotic microangiopathy
54057	Thrombotic thrombocytopenic purpura
100100	Thymic tumor
99867	Thymoma
100088	Thyroid carcinoma
100087	Thyroid tumor
99886	Transient neonatal diabetes mellitus
3378	Trisomy 13
3380	Trisomy 18
3384	Truncus arteriosus
805	Tuberous sclerosis complex
32960	Tumor necrosis factor receptor 1 associated periodic syndrome
68347	Tumor of hematopoietic and lymphoid tissues
363472	Tumor of testis and paratestis
881	Turner syndrome
88950	UMOD-related autosomal dominant tubulointerstitial kidney disease
117594	Unclassified rare disease
83001	Urogenital tract malformation
886	Usher syndrome
180062	Uterovaginal malformation
39044	Uveal melanoma

286	Vascular Ehlers-Danlos syndrome
52759	Vasculitis
28	Vitamin B12-responsive methylmalonic acidemia
27	Vitamin B12-unresponsive methylmalonic acidemia
892	Von Hippel-Lindau disease
903	Von Willebrand disease
893	WAGR syndrome
899	Walker-Warburg syndrome
904	Williams syndrome
905	Wilson disease
906	Wiskott-Aldrich syndrome
1667	Wolcott-Rallison syndrome
3463	Wolfram syndrome
43	X-linked adrenoleukodystrophy
178461	X-linked myopathy with postural muscle atrophy
431272	X-linked scapulo-peroneal muscular dystrophy
910	Xeroderma pigmentosum
876	Yolk sac tumor
3471	Young syndrome
2828	Young-onset Parkinson disease
73263	Zygomycosis

Summary

1- Distribution of registries by country

COUNTRY	REGIONAL	NATIONAL	EUROPEAN	INTERNATIONAL**	TOTAL
AT - Austria	6	26	2	3	37
BE - Belgium	2	15	3	2	22
BG - Bulgaria	0	8	0	0	8
CH - Switzerland*	1	18	1	5	25
CY - Cyprus	0	2	0	0	2
CZ - Czechia	0	5	0	0	5
DE - Germany	10	103	19	26	158
DK - Denmark	1	4	0	0	5
EE - Estonia	0	3	0	0	3
ES - Spain	11	48	4	1	64
FI - Finland	0	7	0	0	7
FR - France	19	104	21	6	150
GB - United Kingdom of Great Britain and Northern Ireland (the)	0	47	14	15	76
GR - Greece	0	3	0	0	3
HR - Croatia	0	2	0	0	2
HU - Hungary	0	5	0	0	5
IE - Ireland	3	14	0	0	17
IL - Israel*	0	2	0	0	2
IS - Iceland*	0	2	0	0	2
IT - Italy	13	64	5	7	89
LT - Lithuania	0	1	0	0	1
LU - Luxembourg	0	1	0	0	1
LV - Latvia	0	1	0	0	1
MK - Republic of North Macedonia*	0	1	0	0	1
MT - Malta	0	2	0	0	2
NL - Netherlands (the)	1	15	11	11	38
NO - Norway*	0	4	3	0	7
PL - Poland	3	7	0	0	10
PT - Portugal	5	11	0	0	16
RO - Romania	0	2	0	0	2
RS - Serbia*	0	4	0	0	4
SE - Sweden	0	16	1	1	18
SI - Slovenia	0	2	0	0	2
SK - Slovakia	0	2	0	0	2
TR - Turkey*	0	5	0	0	5
UA - Ukraine*	0	1	0	0	1
TOTAL	75	557	84	77	793

*Neighbour countries participating to the Orphanet network.

**Beyond Europe.

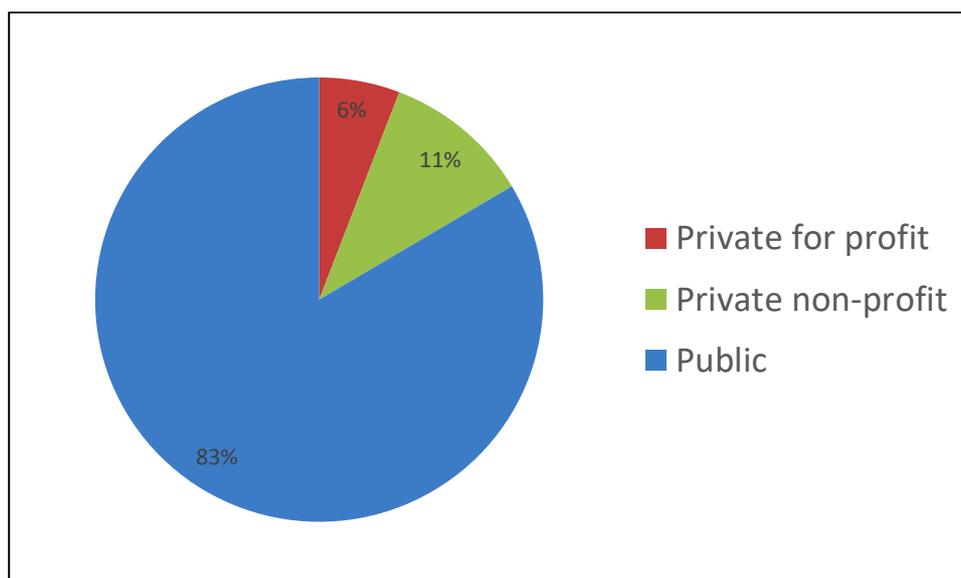
2- Distribution of registries by coverage

COVERAGE	NUMBER OF REGISTRIES*
Regional	75
National	557
European	84
International**	77
TOTAL	793

*All registries, including heads of registries networks and their nodes, are counted.

**Beyond Europe.

3- Distribution of registries by affiliation



List of registries by country

Please note that some ERN registries are still being developed and not yet functional, they are listed only for reference.

AT - Austria (37 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
AGMT (Arbeitsgemeinschaft medikamentöse Tumortherapie): ALL-Registry and Biobank	National	Public
AGMT (Arbeitsgemeinschaft medikamentöse Tumortherapie): Austrian Registry on Hypomethylating Agents in Myeloid Neoplasms	National	Public
AGMT (Arbeitsgemeinschaft medikamentöse Tumortherapie): Head and neck tumor registry Austria	National	Public
AGMT (Arbeitsgemeinschaft medikamentöse Tumortherapie): MBC-Registry (Metastatic breast cancer in Austria)	National	Public
AGMT (Arbeitsgemeinschaft medikamentöse Tumortherapie): PTCL registry (T-cell lymphoma in Austria)	National	Public
Austrian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Austrian brain tumor registry	National	Public
Austrian cancer registry	National	Public
Austrian chronic myeloid leukemia registry	National	Public
Austrian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Austrian GIST registry	National	Public
Austrian Hemophilia Registry - Registry for patients with inborn coagulation defects	National	Public
Austrian myeloma registry	National	Public
Austrian registry for inborn errors of metabolism	National	Public
Austrian Registry on Acquired Hemophilia	National	Public
Austrian severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
Bone and soft tissue tumor registry Graz	Regional	Public
Central data registry of the European Competence Network on Mastocytosis (ECMN)	European	Public
Ceprotrin Treatment Registry	National	Public
Cleft registry of the Austrian Cleft Palate Craniofacial Association	National	Public
Disorders of Cornification Innsbruck	Regional	Public
EB Registry Austria	National	Public
EMSA-SG: central patient registry of the European multiple system atrophy network	International	Public
ENRAH: European alternating hemiplegia and rare epilepsies registry in childhood	International	Public

Innsbruck registry for Adamantiades-Behcet disease: Retrospective and prospective data collection	Regional	Public
International aHUS registry	International	Public
MDS: Austrian myelodysplastic syndromes patient registry - contributes to the European Myelodysplastic Syndromes (MDS) Registry	National	Public
NF-10 - Prospective collection of potentially prognostically relevant data in patients with indolent non-follicular B-cell lymphoma	National	Public
Non-Interventional, web-based Registry for Histiocytic Disorders	National	Public
Registry for Philadelphia chromosome-positive acute lymphoblastic leukemia in childhood and adolescence	National	Public
Registry for relapsing acute lymphoblastic leukemia in childhood and adolescence	National	Public
Registry of the NHL-BFM study group for all subtypes of Non-Hodgkin Lymphoma in children and adolescents	National	Public
Registry of the University Clinic of Oral and Maxillofacial Surgery Salzburg	Regional	Public
Skin cancer syndrome registry Graz	Regional	Public
Styrian registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Thromboreductin-Registry	European	Private for profit
Upshaw-Schulman-Syndrome registry	National	Public
BE - Belgium (22 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Antwerpen registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Belgian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Belgian contribution to the international rare bleeding disorders registry (RBDD)	National	Public
Belgian Cystic Fibrosis patient Registry (BCFR) - contributes to the EUROCAT CF and the ECFS registries	National	Public
Belgian familial adenomatous polyposis registry	National	Public
Belgian familial Lynch syndrome registry	National	Public
Belgian Neuromuscular Diseases Registry (BNMDR)	National	Public
Belgian registry of primary immunodeficiencies - contributes to the ESID European registry	National	Public
Belgian severe chronic neutropenia patient registry - contributes to the Severe Chronic Neutropenia International Registry (SCNIR)	National	Public
Belgian sickle cell anemia registry	National	Public
Belgian systemic sclerosis cohort	National	Public
Central Registry of Rare Diseases (CRRD)	National	Public

ENETS: European Neuroendocrine Tumour Registry	European	Public
ENRAH: Belgian contribution to European registry for alternating hemiplegia in childhood	National	Public
ERN [EpiCARE] - EPICARE-GRANT: An Operational EPICARE Registry for all Rare and Complex Epilepsies	European	Public
EUNEFRON: registry of the European network for the study of orphan nephropathies	European	Public
EURECHINOREG: Belgian contribution to the European registry of human alveolar echinococcosis	National	Public
EUROGLYCANET - International patient registry and cohort for congenital disorders of glycosylation	International	Public
Haemoglobinopathies - database	National	Public
Hainaut and Namur registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
National Prospective Multicentre Registry on Hirschsprung's Disease	National	Private for profit
Pediatric granulomatous arthritis international registry	International	Public
BG - Bulgaria (8 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Bulgaria - part of the TREAT-NMD network	National	Public
National registry of adult patients with chronic myeloid leukemia - BG	National	Private not-for-profit
National registry of patients with Gaucher disease - BG	National	Private not-for-profit
National registry of patients with mucopolysaccharidosis type II (MPS2) - BG	National	Private not-for-profit
National registry of patients with phenylketonuria - BG	National	Private not-for-profit
National registry of patients with primary immunodeficiencies (PID) - BG	National	Private not-for-profit
National registry of patients with thalassaemia major - BG	National	Private not-for-profit
National registry of patients with Wilson disease - BG	National	Private not-for-profit
CH - Switzerland (25 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
COST Action BM1105 Patient Registry - GnRH Network	International	Public
EUROCAT VAUD Switzerland - Registry of congenital malformations of canton Vaud	Regional	Public
Hereditary TTP Registry	International	Public
Intercontinental Cooperative ITP Study Group Registry	International	Public

Juvenile Inflammatory Rheumatism Cohorte (JIRcohort)	European	Private not-for-profit
Paediatric registry for congenital pulmonary malformations	National	Public
Perihilar Cholangiocarcinoma International Registry	International	Public
PNH Registry: Paroxysmal Nocturnal Hemoglobinuria registry	International	Private for profit
SACHER: Swiss Adult Congenital HEart disease Registry	National	Private for profit
Swiss alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Swiss Autoimmune Hepatitis Cohort Study	National	Private not-for-profit
Swiss Childhood Cancer Registry	National	Public
Swiss Cleft Lip and Palate Registry	National	Public
Swiss Diabetes Registry (SwissDiab)	National	Public
Swiss Hemophilia Registry	National	Public
Swiss Pompe Registry	National	Private for profit
Swiss Primary Biliary Cholangitis Cohort Study	National	Private for profit
Swiss Primary Ciliary Dyskinesia Registry - contributing to the International PCD Registry	National	Public
Swiss Primary Sclerosing Cholangitis Cohort Study	National	Public
Swiss Rare Disease Registry	National	Public
Swiss registries for Interstitial and Orphan Lung Diseases (SIOLD Registries)	National	Public
Swiss Registry for Neuromuscular Disorders	National	Public
Swiss registry of biliary atresia - contributes to the EBAR registry	National	Public
Swiss SLE Cohort Study	National	Public
SwissNET - Registry for Neuroendocrine Tumours in Switzerland	National	Public
CY - Cyprus (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Cyprian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
NMDcy: Cypriot Neuromuscular Diseases - contributes to TREAT-NMD	National	Public
CZ - Czechia (5 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Autoimmune neuromuscular disease	National	Public
Czech cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Czech severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

Duchenne and Becker muscular dystrophy patient registry in the Czech Republic and Slovakia - contributes to the TREAT-NMD network	National	Public
Spinal muscular atrophy patient registry in the Czech Republic - part of the TREAT-NMD network	National	Public
DE - Germany (158 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
ADPedKD - International, longitudinal registry including ADPKD patients followed up from childhood.	International	Private not-for-profit
ALS registry Nordrhein-Westfalen	Regional	Public
ALS registry Rheinland-Pfalz	Regional	Public
ALS registry swabia	Regional	Public
ARegPKD - International Registry Study on Autosomal Recessive Polycystic Kidney Disease	International	Public
Ataxia-Telangiectasia patient registry - contributes to the ESID Database	National	Public
Bayern population based cancer registry	National	Public
Blue cone monochromatism - Patient registry	International	Private not-for-profit
BMFS: Network for Congenital Bone Marrow Failure Syndromes (patient registry)	National	Public
Bremen cancer registry	National	Public
CEDATA-GPGE: Registry of Children with Inflammatory Bowel Disease in Germany and Austria	European	Public
Central Cutaneous Lymphoma Registry	National	Public
Centre Saxony-Anhalt registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
CERTAIN-LI: Cooperative European Paediatric TransplAnt INitiative Liver	European	Private not-for-profit
CERTAIN: Cooperative European Paediatric Renal Transplant Initiative registry	European	Private not-for-profit
Child liver tumor registry	National	Public
Children IBD registry in Saxony	Regional	Public
CMMR: Central Malignant Melanoma Registry in Germany	National	Public
Common cancer registry of Berlin, Brandenburg, Mecklenburg-Western Pomerania, Saxony-Anhalt and the free states Saxony and Thuringia	Regional	Public
Conn Registry: German registry of primary aldosteronism	National	Private not-for-profit
Core documentation of children and adolescents with rheumatic diseases	National	Private not-for-profit
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	International	Private not-for-profit
CURE-Net : National registry for congenital uro-rectal malformations	National	Public

CUSTODES - Cushing syndrome registry: therapy and outcome in germany - contributes to the ERCUSYN - European Register on Cushing's Syndrome	National	Private not-for-profit
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	International	Private not-for-profit
DCLLSG registry of the German CLL Study Group - Long term observation of patients with CLL, B-PLL, T-PLL, SLL, T/ NK-LGL and Richter transformation	National	Public
DÖSAK tumor registry for documentation of tumors of the face and jaws in Germany, Austria and Switzerland	International	Public
DYSTRACT: German Dystonia Registry	National	Public
E-IMD : European registry and network for Intoxication type Metabolic Diseases	International	Public
EBAR: European Biliary Atresia Registry	European	Public
EHDN: neuroacanthocytosis patient registry	International	Public
EIMD: European registry and network for intoxication type metabolic diseases	National	Public
EKRS: Saarland Cancer Registry - contributes to the RARECARE Project	Regional	Public
Enroll-HD Registry	International	Private not-for-profit
Epidemiological cancer registry Baden-Württemberg	Regional	Public
ERN [ERKNet] - ERK-REG: ERKNet Registry for Rare Kidney Diseases	European	Public
ERN [ERN-LUNG] : REGISTRY WAREHOUSE - RD Registry Data Warehouse	European	Public
ERN [MetabERN] - UIMD: Unified European Registry for Inherited Metabolic Disorders	European	Public
ERN [RARE-LIVER] - R-LIVER: Registry for Rare Liver Diseases of the ERN on hepatological diseases	European	Public
ERN [RND] - ERN-RND Registry: The ERN-RND Rare Neurological Disease Registry	European	Public
ESID: European registry of primary immunodeficiencies	European	Public
ESNEK - Rare paediatric neurological disease registry Germany	National	Public
EU-RHAB: European Rhabdoid Tumor Registry	European	Private not-for-profit
EuCaNet: International Calciphylaxis Registry	European	Private for profit
eurlPFreg: European idiopathic pulmonary fibrosis registry	European	Public
EUROFA - EFACT: European Friedreich Ataxia Registry	European	Public
European Alport registry	European	Public
European chILD-registry and biobank of the European network for children's interstitial lung diseases (chILD-EU).	European	Public
European registry of sporadic degenerative ataxia with adult onset	European	Public
EUROSCA-R: European patient registry on spinocerebellar ataxias	European	Public

EXCITING Registry: Registry for Exploring Clinical and Epidemiological Characteristics of Interstitial Lung Diseases	National	Public
Fabry Disease Registry -DE	National	Private for profit
FACE - National registry for Robin sequence	National	Public
FungiScope - Global Rare Fungal Infection Registry	International	Public
GEKID: Association of the epidemiologic cancer registries	National	Public
GeNeMove: German database for Wilson disease	National	Public
German acromegaly registry	National	Private for profit
German adrenal tumors registry	National	Public
German AID (Autoinflammatory disorders) registry - subproject AID-NET	National	Public
German alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
German calciphylaxis registry -contributes to the international calciphylaxis registry EuCaNet	National	Private for profit
German central registry for Sickle cell disease	National	Public
German childhood cancer registry (Partner of ACCIS: Automated Childhood Cancer Information System, Member of IACR and ENCR)	National	Public
German CMT-patient registry germany - part of the TREAT-NMD network	National	Public
German cystic fibrosis registry - contributes to the EURO CARE CF registry	National	Private not-for-profit
German DM registry: Patient registry for myotonic dystrophy (DM)	National	Public
German DMD- and SMA-patient registry : Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Austria and Germany - part of the TREAT-NMD network	National	Public
German epilepsy registry	National	Public
German Fanconi anemia registry	National	Private not-for-profit
German FKRP-patient registry - part of the TREAT-NMD network	National	Public
German FSHD registry: Patient registry for Facioscapulohumeral dystrophy (FSHD)	National	Public
German gastrointestinal stromal tumor registry	National	Public
German Haemophilia Registry (DHR)	National	Public
German IBM registry: Patient registry for aquired or sporadic inclusion body myositis	National	Public
German marginal zone lymphoma registry	National	Private not-for-profit
German mucopolysaccharidosis patient registry	National	Public
German multiple endocrine neoplasia type 1 (MEN 1) registry	National	Public
German national case collection of familial pancreatic cancer	National	Private not-for-profit

German PAM/MFM registry: Patient registry for hereditary protein aggregate myopathies (PAM) / myofibrillar myopathies (MFM)	National	Public
German paroxysmal nocturnal hemoglobinuria registry	National	Public
German patient registry HLH (hemophagocytic lymphohistiocytosis)	National	Public
German pituitary tumors registry	National	Public
German registry for congenital heart defects - part of the competence network for congenital heart defects	National	Public
German registry for congenital thrombocytopenia	National	Public
German Registry for Incidental Gallbladder Carcinoma	National	Private not-for-profit
German registry for Langerhans Cell Histiocytosis in childhood (LCH-REG-DE 2013)	National	Public
German registry for Morbus Adamantiades-Behçet e.V.	National	Public
German registry for neonates, infants, children, adolescents, and adults with newly diagnosed and/or relapsed neuroblastic tumors (NB Registry 2016) - partner of the INRG Database	National	Public
German registry for papulosis atrophicans maligna	National	Public
German severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
Germany patient registry for membranoproliferative glomerulonephritis	National	Public
GeVas: German Vasculitis Registry	National	Public
GMALL-registry: registry for adult patients with acute lymphoblastic leukemia or related diseases	National	Public
GPOH-MET Registry: Registry for children and adolescents with malignant endocrine tumour	National	Private not-for-profit
GPOH-registry sickle cell disease	National	Private not-for-profit
Hamburg cancer registry	National	Public
HepNet: German hepatocellular carcinoma (HCC) registry	National	Public
Hunter Outcome Survey (HOS): patient registry - DE	National	Private for profit
INSIGHTS-IPF Investigating Significant Health Trends in Idiopathic Pulmonary Fibrosis	National	Public
iNTD (International Working Group on Neurotransmitter Related Disorders) Registry	International	Private not-for-profit
iNTD Network - International Working Group on Neurotransmitter Related Disorders	International	Private not-for-profit
International Collaborative Gaucher Group (ICGG) Gaucher Registry - DE	National	Private for profit
International registry for Primary Ciliary Dyskinesia	International	Public
INVM (Isolated Noncompaction of Ventricular Myocardium) registry	National	Public
Kids Lung Register: International register and biobank for rare lung diseases	International	Public

KINDLERNET: Central patient registry Kindler syndrome	International	Public
LBL Registry: Registry for children with lymphoblastic lymphoma	National	Public
Lupus nephritis registry (established by the german paediatric nephrology association)	National	Public
Mainz registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
MAISTHRO-Registry : multicentric thrombophilia registry (MAIn-ISar- THROmbose-Register)	National	Public
MCR - Munich Cancer Registry	Regional	Public
MDS Registry: German myelodysplastic syndromes patient registry	National	Public
mitoREGISTRY: Registry for patients with mitochondrial diseases - subproject of mitoNET	National	Public
Mucopolysaccharidosis type 1 (MPS I) Registry - DE	National	Private for profit
Mykke : German registry for children and adolescents with myocarditis	National	Public
National Cancer-predisposing syndrome registry	National	Public
National database for echinococcosis	National	Public
National MRKH patient registry	National	Public
National nephrogenic systemic fibrosis registry	National	Public
National patient registry rare histiocytic disorders - part of the International Rare Histiocytic Disorders Registry (IRHDR)	National	Public
National registry for Blackfan-Diamond disease	National	Public
National Registry for Mycotic keratitis	National	Public
National registry for retroperitoneal fibrosis	National	Private not-for-profit
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	International	Private for profit
NeoExNET registry: patient registry for neuroendocrine tumors	National	Public
Nephronophthisis registry for patients in Germany, Austria and Switzerland	International	Private for profit
NET-Registry: German neuroendocrine gastrointestinal tumors	National	Private for profit
Neuro-Heart-Registry: Registry for patients with heart involment in neurovascular diseases	National	Public
Neuromyelitis optica patient registry	National	Public
Neutropenia biobank - contributes to the SCN international registry (SCNIR)	National	Public
NHL-BFM Registry 2012: Registry of the NHL-BFM study group for all subtypes of Non-Hodgkin Lymphoma diagnosed in children and adolescents	International	Public
NIRK: national central registry for ichthyoses and related keratinization disorders	National	Public
NKR: German registry for adrenocortical carcinoma	National	Private not-for-profit
NPC-2016 - A multicenter registry for nasopharyngeal cancer in children, adolescents and young adults	National	Private not-for-profit

OSTEOPETR: International registry of patients suffering from osteopetrosis	International	Public
Patient registry for primary hyperoxaluria - contributes to the OxalEurope-Network	National	Public
Patient registry for retinal degeneration PRO RETINA e.V	National	Private not-for-profit
Patient registry of the German Network for Systemic Scleroderma	National	Private not-for-profit
PID-NET: German National Registry for Primary Immunodeficiencies (PID) - partner of the ESID registry	National	Public
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	International	Public
Pompe Disease Registry - DE	National	Private for profit
PROGNOSIS : The Prospective German Non-CF-Bronchiectasis Registry - contributes to EMBARC registry	European	Public
PSHN Registry - Purpura Schoenlein-Henoch-Nephritis registry of the German society for paediatric nephrology (GPN)	National	Public
RAMEDIS : Rare Metabolic Diseases Database	National	Public
RegiSCAR: International registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples - patient registry	International	Public
Register for rare myeloproliferative neoplasms	International	Public
Registry and biobank for rare thyroid and parathyroid carcinoma	National	Public
Registry for C3 Glomerulopathy and Immune complex-mediated MPGN	National	Public
Registry for congenital melanocytic nevi and neurocutaneous melanocytosis	National	Public
Registry for Merkel Cell Carcinoma	National	Public
Registry for Patients with WT1 Mutation Associated Diseases	National	Private not-for-profit
RetDis Database: clinical descriptions of patients and families with inherited eye diseases	International	Public
Schleswig-Holstein cancer registry	National	Public
SCNIR: Severe Chronic Neutropenia International Registry	International	Public
STEP Registry: Registry for rare tumors in children and adolescents	National	Private not-for-profit
STER: FVII deficiency treatment international registry	International	Public
TIRCON: NBIA (Neurodegeneration with Brain Iron Accumulation) patient registry	International	Public
Tumor Registry of Lymphatic Neoplasia (TNL-Registry): Epidemiological registry describing treatment reality and therapy modalities of patients with malignant Lymphatic Systemic Diseases (Non-Hodgkin's Lymphoma, Chronic Lymphocytic Leukemia and Multiple Myeloma) requiring therapy	National	Public
DK - Denmark (5 registries)		

ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Danish cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Danish malignant hyperthermia registry - contributes to the European Malignant Hyperthermia Group (EMHG)	National	Public
Funen county registry of congenital anomalies - contributes to the EURO CAT network	Regional	Public
Mendelian cytogenetics network online database	National	Public
Nordic Database for Rare Diseases	National	Public
EE - Estonia (3 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Estonian cancer registry	National	Public
Estonian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Estonian Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes - contributing to EURO-WABB	National	Public
ES - Spain (64 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
ADPKD registry: Spanish registry for autosomal dominant polycystic kidney disease	National	Private not-for-profit
aHUS/C3G: Atypical Hemolytic Uremic Syndrome (aHUS) and C3 Glomerulopathy (C3G) Database	National	Public
EARCO Registry (Spanish contact point)	National	Public
ECEMC: Registry of the Spanish Collaborative Study of Congenital Malformations	National	Public
Epidemiologic Registry of Patients Diagnosed With Acute Myeloid Leukemia - ES	National	Private not-for-profit
ERCUSYN: European registry on Cushing's syndrome	European	Public
ERN [EuroBloodNet] - ENROL: European Rare Blood Disorders Platform	European	Public
ERN [TRANSPLANT-CHILD] - PETER: PaEdiatric Transplantation European Registry	European	Public
EURO-WABB: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome, Bardet-Biedl Syndrome and Other Rare Diabetes Syndromes - ES	National	Public
EUROMAC: Registry of patients affected by McArdle Disease	European	Public
MYO-SPAIN Platform: Patient registry on idiopathic inflammatory myopathy in Spain	National	Private for profit
NMD-ES: Amyotrophic Lateral Sclerosis Registry	National	Public

NMD-ES: Congenital Myasthenic Syndrome Registry	National	Public
NMD-ES: Hereditary Neuropathies Registry	National	Public
NMD-ES: Inclusion Body Myositis Registry	National	Public
NMD-ES: Inflammatory Neuropathies Registry	National	Public
NMD-ES: Mitochondrial Diseases Registry	National	Public
NMD-ES: Myasthenia Gravis Registry	National	Public
NMD-ES: Myopathies and Muscular Dystrophies Registry	National	Public
NMD-ES: Myopathies due to collagen deficiency VI Registry	National	Public
NMD-ES: Pompe Disease Registry	National	Public
NMD-ES: Spanish Registry of Neuromuscular Diseases	National	Public
PanGen-FAM: Spanish registry of hereditary pancreatic cancer	National	Public
Population registry of rare diseases and congenital anomalies of Cantabria (Spain)	Regional	Public
Population registry of rare diseases of Balearic Islands (Spain)	Regional	Public
Population registry of rare diseases of Navarra (Spain)	Regional	Public
RACAV: Registry of congenital anomalies of the Basque Country (Spain) - contributes to the EUROCAT network	Regional	Public
Rare disease registry of Aragon (Spain)	Regional	Public
REDAPED: Spanish registry of ataxias and degenerative spastic paraparesis	National	Public
REDIP: Spanish registry of primary immunodeficiencies - contributes to the ESID European registry	National	Public
REEG: Spanish Gaucher's disease registry	National	Public
ReeR: Spanish National Registry of Rare Diseases	National	Public
REeRIGG4: Spanish IgG4-related Disease Registry	National	Private not-for-profit
Registro MEN: Spanish registry of multiple endocrine neoplasia, pheochromocitomas and paragangliomas	National	Private for profit
Registry for rare diseases in Andalusia (Spain)	Regional	Public
Registry for rare diseases in Extremadura (Spain)	Regional	Public
REHAP: Spanish Registry of Pulmonary Arterial Hypertension	National	Private for profit
REHem-AR-SEHOP: Spanish Registry of Hemoglobinopathies and Rare Anemias from the Spanish Society of Paediatric Haematology and Oncology	National	Public
REHEVASC: Spanish registry for hepatic vascular diseases	National	Private not-for-profit
REHIPED - Spanish Registry for Pediatric Pulmonary Hypertension	National	Public
RenalTube: Database of patients with primary tubulopathies	International	Public
REPA: Spanish registry of alveolar proteinosis	National	Public

RERGA: Registry for rare diseases in Galicia (Spain)	Regional	Public
RETEGEP: Spanish Registry of Gastroenteropancreatic Neuroendocrine Tumors	National	Private for profit
REWBA: Spanish Registry of Wolfram, Bardet-Biedl and Alstrom syndromes	National	Public
SIER-CV: Information System on rare diseases in Valencian Community (Spain)	Regional	Public
SIERMA: Information system on rare diseases in Madrid (Spain)	Regional	Public
SIERrm: Information System on rare diseases in the Region of Murcia (Spain)	Regional	Public
SIOPE Diffuse Intrinsic Pontine Glioma (DIPG) Registry (Spanish contact point)	National	Private not-for-profit
SpainRDR: Spanish Rare Disease Registries Research Network (patient registries)	National	Public
Spanish alpha-1 antitrypsin deficiency registry (REDAAT) - contributes to the Alpha One International Registry (AIR)	National	Public
Spanish Overgrowth Syndrome Registry	National	Public
Spanish patient registry for spinal muscular atrophy - part of the TREAT-NMD network	National	Public
Spanish patient registry of ataxias	National	Public
Spanish patient registry of hereditary retinal dystrophy	National	Public
Spanish patient registry of myelodysplastic syndromes	National	Public
Spanish patient registry of transmissible spongiform encephalopathies	National	Public
Spanish Registry of Cushing Syndrome - contributes to ERCUSYN	National	Private for profit
Spanish registry of Duchenne muscular dystrophy - part of the TREAT-NMD network	National	Public
Spanish Registry of Patients with Fanconi Anemia	National	Public
Spanish registry of patients with McArdle disease	National	Public
Spanish registry of pulmonary Langerhans cell histiocytosis	National	Public
Spanish Rheumatology Society Lupus Registry	National	Private not-for-profit
Spanish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
FI - Finland (7 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Finnish cancer registry - contributes to the RARECARE project	National	Public
Finnish IPF registry	National	Public
Finnish patient registry on Fabry disease - contributing to the international Fabry registry and Shire registry	National	Public
Finnish TREAT-NMD Patient Registry	National	Public
Register of Congenital Malformations	National	Public

The Finnish Hematology Registry and Clinical Biobank (FHRB Biobank)	National	Public
The Finnish Register of Visual Impairment, Näkövammarekisteri	National	Public
FR - France (150 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Aquitaine registry of mesothelioma	Regional	Public
Auvergne registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Basse Normandie registry of hematological malignancies	Regional	Public
BLAU registry: French pediatric granulomatous arthritis registry	National	Public
Breast and other gynecological cancers registry of Côte-d'Or	Regional	Public
CEREDIH: French primary immunodeficiencies registry	National	Public
Cloud-R HAE Register Hereditary Angiodema	European	Public
Cobra: COhort Angioedema Berinert	National	Public
CoF-AT study: a French cohort on ataxia-telangiectasia	National	Private not-for-profit
Cohort of patients with hereditary dystrophies of retina	National	Public
Cohorte française de patients atteints de mucoviscidose et cohorte CFTR-RD	National	Private not-for-profit
Côte d'Or registry of hematological malignancies	Regional	Public
D[4]/Phenodent: French registry of patients affected by rare odontologic diseases	National	Public
Digestive Tumor Registry of Finistère	Regional	Public
DM Scope: Myotonic dystrophy patient registry in France - part of the TREAT-NMD network	National	Private not-for-profit
Duchenne and Becker muscular dystrophy patient registry in France - part of the TREAT-NMD network	National	Public
EDMUS - NOMADMUS: French cohort of Devic's neuromyelitis optica and related neurological disorders	National	Public
EDMUS: European Database for Multiple Sclerosis and other related diseases	European	Public
ENETS Registry: European Neuro-Endocrine Tumors Group	European	Public
EPIMAD: registry of chronic inflammatory intestine diseases in North-West	Regional	Public
ERN [EURO-NMD] - EURO-NMD Registry: Patient centered and interoperable registry hub for Rare Neuromuscular Diseases	European	Public
ERN [EYE] - REDgistry: An interoperable sustainable European Rare Eye Disease Registry	European	Public
ERN [ITHACA] - ILIAD Rare Diseases patient registry: an International Library of Intellectual disability and Anomalies of Development	European	Public

ERN [PAEDCAN] - PARTNER: Paediatric Rare Tumours Networks - European Registry	European	Public
ERN [Skin] - ERN-Skin REGISTRY: Interoperable ERN on Rare and Undiagnosed Skin Disorders	European	Public
ERN [VASCERN] - VASCERN Registries	European	Public
Escort-Hu: European sickle cell disease cohort- hydroxyurea	European	Public
Establishment of children and adolescents cohort in Behcet disease in France	National	Public
EU-CHS: European central hypoventilation syndrome registry	European	Public
EURECHINOREG: European registry of alveolar echinococcosis	European	Public
EurêClark registry (The European Clarkson's syndrome registry)	European	Public
European LeukoDataBase	European	Public
European prospective registry of children born to mothers affected by the antiphospholipids syndrome	European	Public
European Society for Blood and Marrow Transplant Society Registry (EBMT registry)	European	Public
EURORETT: European network on Rett syndrome (registry)	European	Public
FranceCoag: French prospective cohort of patients affected with haemophilia or severe form of other hereditary hemorrhagic diseases except platelet disorders	National	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Immune thrombocytopenic purpura (ITP)	International	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Lupus	International	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Myositis	International	Public
French acromegaly registry	National	Private not-for-profit
French addictive acute intoxications cohort	National	Public
French atypical sarcoidosis clinical forms registry	National	Public
French auto-immunity and Rituximab (AIR) registry: prospective study of patients treated with Rituximab	National	Public
French case registry for IgG4-related disease	National	Private for profit
French central hypoventilation syndrome registry - will contribute to the European CHS registry	National	Public
French certified patient registry for Langerhans cell histiocytosis	National	Public
French certified registry of glycogen storage disease type 2	National	Private for profit
French certified registry of patients affected by Gaucher disease	National	Public

French certified registry of patients affected by thalassemia	National	Public
French cohort creation in retinitis pigmentosa	National	Public
French cohort for auto-inflammatory diseases	National	Public
French cohort in primary ciliary dyskinesia	National	Public
French cohort of acquired autoimmune haemolytic anemia	National	Public
French cohort of common variable immunodeficiency with hypogammaglobulinemia in adults (CVID)	National	Public
French cohort of focal dystonia families	National	Public
French cohort of idiopathic pulmonary fibrosis	National	Public
French cohort of inflammatory bowel disease (IBD)	National	Public
French cohort of rare diabetes (neonatal diabetes, monogenic, atypical and syndromic forms)	National	Public
French cohort of rhombencephalosynapsis	National	Public
French cohort of Usher syndrome	National	Public
French Cohorts in Sneddon syndrome and suspected Sneddon syndrome livedo	National	Public
French constitutive hematologic diseases registry	National	Public
French cystic fibrosis patient registry	National	Private not-for-profit
French cystinosis registry	National	Public
French Disease Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes - contributes to EURO-WABB	National	Public
French epidemiological registry of esophageal atresia	National	Public
French familial cardiac malformations registry	National	Public
French National FSHD Registry	National	Private not-for-profit
French National Lymphangiomyomatosis Registry (RE-LAM-CE)	National	Private not-for-profit
French national patient registry on rare peritoneal tumor RENAPE	National	Private not-for-profit
French National Registry for Rare Diseases (BNDMR)	National	Public
French observatory of biliary atresia	National	Public
French observatory of gastric linitis plastica	National	Public
French observatory of primary biliary cirrhosis	National	Public
French observatory of primitive sclerosing cholangitis	National	Public
French patient registry affected by genetic deafness in France	National	Public
French patient registry in chorioretinopathy, birdshot type	National	Public
French prospective cohort follow-up of children under the age of 18 with autoimmune cytopenia	National	Public

French Register of Amyotrophic Lateral Sclerosis	National	Public
French register of the SDH-related hereditary paraglioma	National	Public
French registry for macrophagic myofasciitis	National	Public
French registry for right arrhythmogenic ventricular dysplasia (ARVC/D)	National	Public
French Registry of Atypical Hemolytic Uremic Syndrome (aHUS) in Children	National	Private not-for-profit
French registry of child hematological malignancies	National	Public
French registry of familial and premature prostate cancers (before 50 years)	National	Public
French registry of generalized resistance to thyroid hormone	National	Public
French registry of hereditary dyslipidemia in children: familial combined dyslipidemias	National	Public
French registry of Iron overload genetic rare diseases, non-related to the HFE gene	National	Public
French registry of Kabuki syndrome	National	Public
French registry of Marshall's syndrome with periodic fever	National	Public
French registry of neuromuscular diseases from reference centres	National	Public
French registry of patients affect by Leber amaurosis and retinitis pigmentosa to assess the clinical trial in gene therapy	National	Public
French registry of rare genetic metabolism disorders of steroids - contributing to the international RGSDC registry	National	Public
French registry of rare hypersomnias	National	Public
French registry of rare pulmonary hypertension (HTAP)	National	Public
French registry of tetrahydrobiopterin deficiencies	National	Public
French severe chronic neutropenia certified patient registry - contributes to the SCN international registry (SCNIR)	National	Public
French sickle cell anemia registry	National	Public
French West Indies registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
FROG: FRENCH Observatory on Gaucher disease	National	Public
GENEPSO: French BRCA1 and 2 carrier cohort study	National	Private not-for-profit
Gironde registry of hematological malignancies	Regional	Public
GMF: French registry of myelodysplastic syndromes and leukemia chemo- and radio-induced	National	Private for profit
GTE: French registry of endocrine tumors	National	Public
Idiopathic pulmonary fibrosis: Cohort studies for evaluation of pronostic factors, therapeutic evaluation	National	Public
ITINERAIR-HTAP: French cohort of adult with pulmonary arterial hypertension	National	Public
ITINERAIR-pediatrie: French cohort of children with pulmonary arterial hypertension	National	Public

ITINERAIR-scleroderma: French pulmonary arterial hypertension screening cohort of patients with scleroderma	National	Public
KAWA-NET: KAWASAKI disease registry	National	Public
La Réunion French Island registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Laminopathies and emerinopathies patient registry	National	Public
LEA - Multicentric prospective cohort of children and adolescents malignant hemopathies	National	Public
LeukoFrance database and biobank	National	Public
Marne-Ardenne thyroid cancer registry	Regional	Public
Mesothelioma cohort in Seine Saint-Denis and Val de Marne	Regional	Public
Motor Function Measure database (Neuromuscular Diseases)	National	Public
Multicenter national registry of pleural mesothelioma (qualified register)	National	Public
National database for the study and follow-up of paediatric rare tumors	National	Public
National registry of children solid tumors	National	Public
Paris registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
PHA1-NET: PseudoHypoAldosteronism type 1 cohort	National	Public
POLA: French patient registry of high level oligodendroglioma	National	Public
Primary central nervous system tumors registry of Gironde	Regional	Public
RaDiCo-AC-OEIL: National cohort on congenital defects of the eye : natural history, genetic determinisms and improved ocular and extra-ocular outcome prediction for better patient management	National	Public
RaDiCo-ACOSTILL: National cohort on adult and childhood onset Still disease	National	Public
RaDiCo-COBALT: National cohort on Bardet-Biedl syndrome and Alström syndrome	National	Public
RaDiCo-COLPAC: National cohort on the epidemiology, clinical and genetic heterogeneity of Low Phospholipid-Associated Cholelithiasis (LPAC) syndrome	National	Public
RaDiCo-DCP: National cohort on Primary Ciliary Dyskinesia (PCD) : Identification of specific severity criteria and phenotype-genotype correlation study	National	Public
RaDiCo-ECYSCO: European cystinosis cohort	European	Public
RaDiCo-EURBIO-Alport: Study of the natural history of Alport Syndrome by establishment of an international database	European	Public
RaDiCo-FARD: National cohort for the evaluation of individual burden in the course of rare skin diseases	National	Public
RaDiCo-GenIDA: International social network for data collection on the natural history of rare monogenic forms of intellectual disabilities	International	Public

RaDiCo-IDMet: National and European cohort on Imprinting Disorders and their metabolic consequences	European	Public
RaDiCo-MARFAN: National cohort on Marfan syndrome and apparent diseases	National	Public
RaDiCo-MPS: National cohort on Mucopolysaccharidosis in the era of specific therapeutics	National	Public
RaDiCo-PID: National cohort on Idiopathic Interstitial Pneumonia from infancy to elderly	National	Public
RaDiCo-PP: National cohort on periodic paralysis : clinical, genetic and medico-economic studies involving m-health monitoring tools	National	Public
RaDiCo-RETICO: National cohort on inherited retinal dystrophies	National	Public
RaDiCo-SED-VASC: National cohort on vascular Ehlers-Danlos syndrome	National	Public
Regional registry of thyroid cancers in Rhône-Alpes	Regional	Public
Registry for digestive cancers in Burgundy	Regional	Public
Registry for Patients with Digital Ulcers Associated with Systemic Sclerosis (DU/SSc)	International	Public
Registry of digestive tumors in Calvados	Regional	Public
Registry of hereditary Haemochromatosis of the Languedoc Roussillon (including rare forms non-HFE hemochromatosis)	Regional	Public
Registry of the network studying thrombotic microangiopathies	National	Public
Rhône-Alpes registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
SCETIDE - Stem Cell Transplant for primary Immune Deficiencies in Europe	European	Public
SYRENE: Rett syndrome network - French database of clinical and genetic aspects of Rett syndrome	National	Public
The global aHUS Registry	National	Private for profit
VALDIG: cohort creation on Budd-Chiari syndrome, hepatic venoocclusive disease, hepatoportal sclerosis and portal vein thrombosis	International	Public
GB - United Kingdom of Great Britain and Northern Ireland (the) (76 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Ataxia-Telangiectasia International Registry (A-T Society)	International	Public
Batten Disease Neuronal Ceroid Lipofuscinosis (NCL) Patient Registry	National	Public
BPOLD: British Paediatric Orphan Lung Disease Registry	National	Public
CCRN 1055: Osteonecrosis of the Jaw (ONJ) Case Registry	International	Private for profit
CCRN 1076: A multi-centre safety registry for malaria patients treated with Eurartesim TM	National	Private for profit
CCRN 2421 (Noonan/RASopathy patients): A registry study to characterize genetic and	International	Public

pathway biomarkers in Noonan syndrome and other RASopathy patients - UK		
CCRN 2536 (Idiopathic pulmonary fibrosis): Post-Authorisation Safety Study of Esbriet® (Pirfenidone): A Prospective Observational Registry to Evaluate Long-Term Safety in a Real-World Setting	National	Private for profit
CCRN 3119 (Spondyloarthritis): Patients with axial spondyloarthritis: multicountry registry of clinical characteristics, including radiographic progression, and burden of disease over 5 years in real life setting	International	Private for profit
CCRN 995 (European Adrenal Insufficiency): A European multi-centre, multi-country, post-authorisation, observational study (registry) of patients with chronic adrenal insufficiency - UK	European	Private for profit
CRANE: Cleft Registry and Audit Network - England, Wales & Northern Ireland	National	Public
DRN 377: Clinical Register for Transient Neonatal Diabetes	National	Private not-for-profit
EBV associated NK/T cell malignancies registry	National	Private not-for-profit
ECARUCA: European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations	European	Public
ECFS: European Cystic Fibrosis Society patient registry	European	Public
EHDN: registry of juvenile Huntington's disease	International	Private not-for-profit
EHR: European Haemoglobinopathy Registry	European	Public
EIMD: European registry and network for intoxication type metabolic diseases	National	Public
ERN [Endo-ERN] - EuRRECa: European Registries for Rare Endocrine Conditions	European	Public
English alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
English cystic fibrosis database	National	Public
English cystic fibrosis patient registry - contributes to the EURO CARE CF and ECFS registries	National	Public
English hereditary angioedema patient registry - part of the HAE European registry	National	Public
English registry for lymphangiomyomatosis	National	Public
English severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
ERNEST - European Registry for Myeloproliferative Neoplasms towards a better understanding of Epidemiology, Survival and Treatment	European	Private for profit
EUMDS: European Registry for Myelodysplastic Syndromes - part of EuroLeukemiaNet (ELN)	European	Public
EURO-WABB: An EU Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes	European	Public
EURO-WABB: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom	National	Public

Syndrome, Bardet-Biedl Syndrome and Other Rare Diabetes Syndromes - UK		
EUROCF: European cystic fibrosis registry	International	Public
EUROCAT: European surveillance of congenital anomalies	European	Public
EURODSD: European disorders of sexual development registry	European	Public
EuroMyositis registry	European	Public
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	European	Public
European Prader-Willi syndrome database	European	Public
EUROWILSON: European network on Wilson disease (registries)	European	Public
Familial Ovarian Cancer Register (FOCR)	National	Public
Family History of Bowel Cancer Registry	National	Public
Global FKR registry (Global Fukutin-Related Protein defects registry) - Part of TREAT-NMD Alliance	International	Public
Great Ormond Street Hospital Congenital Melanocytic Naevus Registry	National	Public
I-DSD: International Disorders of Sex Development registry	International	Public
International A-T Registry	International	Public
International Niemann-Pick Disease Registry	International	Public
MCRN078 (E2080-E044-401): European Registry of Anti-Epileptic Drug Use in Patients with Lennox-Gastaut Syndrome - UK	National	Private for profit
Myotonic dystrophy patient registry in United Kingdom - part of the TREAT-NMD network	National	Public
National Congenital Anomaly and Rare Disease Registration Service (NCARDS)	National	Public
National haemophilia database (NHD)	National	Public
NHR: National Haemoglobinopathy Registry	National	Public
North Star database	National	Public
Registry for Patients with Niemann-Pick Type C Disease	International	Public
The Global aHUS Registry	National	Private for profit
The Global Registry for COL6-related dystrophies	International	Public
The International GNE Myopathy Registry - Hereditary Inclusion Body Myopathy-Disease Monitoring (HIBM-DMP): A Combined Registry and Prospective Observational Natural History Study to Assess HIBM Disease (part of the TREAT-NMD network) - UK	National	Public
The MTM and CNM Registry - The Myotubular and Centronuclear Myopathy Patient Registry	International	Public
The National Chronic Granulomatous Disease Registry	National	Public
The UK Paediatric ITP Registry	National	Public

TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases (registries)	International	Public
TuberOus SCLerosis registry to increase disease Awareness (TOSCA)	International	Private for profit
UK & Ireland Fanconi Anaemia Registry	National	Public
UK and Ireland Duchenne and Becker muscular dystrophy patient registry (part of the TREAT-NMD network)	National	Public
UK and Ireland Juvenile Dermatomyositis Cohort Biomarker Study and Repository	National	Public
UK and Ireland Spinal muscular atrophy (SMA) patient registry (part of the TREAT-NMD network)	National	Public
UK and Ireland Vasculitis registry (UKIVAS)	National	Public
UK facioscapulohumeral muscular dystrophy patient registry (part of the TREAT-NMD network)	National	Public
UK Huntington disease registry (collaborating with the EHDN/Euro HD Registry)	National	Public
UK mucopolysaccharidosis registry	National	Public
UK Myotonic dystrophy type I patient registry (part of the TREAT-NMD network)	National	Public
UK national Acromegaly patient register	National	Public
UK Paediatric ITP (Immune Thrombocytopenic Purpura) Registry	National	Public
UK Registry for Central Hypoventilation Syndrome (CHS)	National	Public
UK Renal Rare Disease Registry (RaDaR) - Part of OXAL Europe	National	Public
UK Thrombotic Thrombocytopenia Purpura (UKTTP) Registry	National	Public
UKAITPR: United Kingdom adult idiopathic thrombocytopenic purpura registry	National	Private for profit
UKESR: United Kingdom Evans Syndrome Registry	National	Private for profit
UKFITPR: United Kingdom familial idiopathic thrombocytopenic purpura (ITP) Registry	National	Private for profit
United Kingdom neuromyelitis optica registry	National	Public
United Kingdom Primary Sjogren's Syndrome Registry	National	Public
GR - Greece (3 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Greek cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Greek severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
National Registry for Haemoglobinopathies in Greece (NRHG)	National	Private not-for-profit
HR - Croatia (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION

Croatian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Registry for neuromuscular disease	National	Public
HU - Hungary (5 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
DMD registry - Hungary - contributes to the TREAT-NMD network	National	Public
Hungarian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Hungarian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
National NF Register	National	Public
SMA registry Hungary - contributes to the TREAT-NMD network	National	Public
IE - Ireland (17 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
CFRI: The Cystic Fibrosis Registry of Ireland - contributes to the EURO CARE CF registry	National	Public
Dublin registry of congenital anomalies - contributes to the EURO CAT network	Regional	Public
Hunter Outcome Survey (HOS): patient registry	National	Private for profit
Irish ENS@T Biobank and Registry - contributes to ENS@T registry	National	Public
Irish Galactosemia Registry - contributes to European Galactosemia Patient Registry	National	Public
Irish Registry for homocystinurias and methylation defects - contributes to E-HOD	National	Public
Irish registry of amyotrophic lateral sclerosis and motor neurone disease - contributes to Euro-MOTOR	National	Public
Irish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
National Alpha-1 Antitrypsin Deficiency Registry - contributes to the Alpha One International Registry (AIR)	National	Public
National Cleft Database - contributes to EURO CAT	National	Public
National Haemophilia Register - Ireland	National	Public
National Severe Immunodeficiency Registry - contributes to ESID registry	National	Public
South East of Ireland registry of congenital anomalies - part of BINOCAR and EURO CAT network	Regional	Public
South of Ireland registry of congenital anomalies - contributes to the EURO CAT network	Regional	Public
Sudden Cardiac Death in the Young Registry	National	Private not-for-profit
The Irish Rare Kidney Disease Registry and Biobank	National	Public

UKIVAS - UK and Ireland registry of autoimmune vasculitides	National	Public
IL - Israel (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Israeli cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Israeli severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
IS - Iceland (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Iceland cancer registry - contributes to the RARECARE project	National	Public
Iceland cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
IT - Italy (89 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
AICE: Italian registry of hemophilia centre	National	Public
Angelman Syndrome Italian Registry (RISA)	National	Private not-for-profit
Arrhythmogenic right ventricular cardiomyopathy/dysplasia: clinical registry and database, evaluation of therapies	National	Public
Behçet's disease registry	National	Private not-for-profit
Campania registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Congenital Malformations Registry	Regional	Public
DMD / BMD Italy Patient Registry	National	Public
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Italy - contributes to the TREAT-NMD network	National	Public
EIMD: European registry and network for intoxication type metabolic diseases	National	Public
ERN [EURACAN] - STARTER: Starting an Adult Rare Tumor European Registry	European	Public
ERN [ReCONNET] - TogethERN ReCONNET: a European Registry Infrastructure for data harmonization in rare and complex connective tissue and musculoskeletal diseases	European	Public
ERN [RITA] - MERITA: A METADATA REGISTRY FOR THE ERN RITA	European	Public
EUROFEVER: European registry for autoinflammatory diseases	European	Public
EUROWILSON: Registry and network to improve the management of Wilson Disease	National	Public
FMF: Italian registry for familial mediterranean fever in the young	National	Public
Friedreich's ataxia Italian patient registry	National	Public

GLATIT: Glanzmann thrombasthenia Italian registry	National	Public
GLUT1 deficiency syndrome registry	National	Private not-for-profit
HAE-registry: European hereditary angioedema patient registry	European	Public
IBAHC: Italian registry for alternating hemiplegia of childhood	National	Public
INNCB MG Registry	National	Public
International registry of bone fragility fractures in the young	International	Public
International Registry of congenital dyserythropoietic anemia II	International	Public
International Registry of Rare Bleeding Disorders (RBDD)	International	Public
International registry of recurrent and familial hemolytic uremic syndrome/thrombotic thrombocytopenic purpura	International	Private not-for-profit
IIPERN: Registry of JIA patients treated with anti-TNF	National	Public
ISMAC: Sicilian registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Italian Alpha-1 antitrypsin Deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Italian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Italian HLH Registry - Hemophagocytic lymphohistiocytosis	National	Public
Italian LCH Registry - Langerhans cell histiocytosis	National	Public
Italian Li-Fraumeni syndrome registry	National	Public
Italian myelodysplastic syndromes registry	National	Public
Italian Network MDS Registry (FISM registry)	National	Private not-for-profit
Italian neuroblastoma registry	National	Public
Italian register multiple endocrine neoplasia	National	Public
Italian registry for MYH9-related thrombocytopenia	National	Public
Italian Registry for patients with Shwachman Diamond Syndrome	National	Private not-for-profit
Italian Registry of adult patients affected by familial mediterranean fever	National	Public
Italian registry of Alport syndrome	National	Private not-for-profit
Italian registry of Creutzfeldt-Jakob disease and correlated syndromes	National	Public
Italian registry of Growth Hormone	National	Public
Italian registry of hemolytic uremic syndrome	National	Public
Italian registry of hypertrophic cardiomyopathy in Anderson-Fabry disease	National	Public
Italian Registry of Kernicterus and hyperbilirubinemia	National	Public
Italian registry of Legionellosis	National	Public
Italian Registry of membranoproliferative glomerulonephritis	National	Private not-for-profit

Italian registry of muscular channelopathy	National	Public
Italian registry of myotonic dystrophies	National	Public
Italian registry of patients and families affected by Pseudoxanthoma Elasticum	National	Public
Italian registry of skeletal dysplasia	National	Public
Italian retinoblastoma registry	National	Public
Lymphomas Database	Regional	Public
MITOCON: National registry mitochondrial disease	National	Private not-for-profit
MODY: Italian registry of maturity onset diabetes of the young	National	Public
National registry of Rare Diseases	National	Public
North-east Italy registry of neurofibromatosis	Regional	Public
North-East of Italy registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
RAM-NET: Italian registry of patients with neuromuscular diseases - contributes to Treat-NMD european network	National	Private not-for-profit
Rare Diseases Registry - Veneto Region	Regional	Public
Regional registry for neuromuscular disorders	Regional	Private not-for-profit
Regional Registry Rare Diseases of Puglia Region (SIMaRRP)	Regional	Public
Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes - contributing to EURO-WABB	National	Public
Registry of inherited bleeding disorders in Emilia Romagna region	Regional	Public
Registry of pregnant patients affected by essential thrombocythemia	National	Public
Registry of steroid-resistant nephrotic syndrome	National	Private for profit
REM: Multiple Osteochondromas Registry	National	Public
RESIDRAS - National Registry of Dravet Syndrome and other Syndromes correlated with genes on SCN1A and PCDH19	National	Private not-for-profit
Rett Syndrome Database	National	Public
RIAF: Fanconi's Anemia Italian Registry	National	Private not-for-profit
RIAT: Ataxia teleangiectasia Italian registry	National	Public
RIMM: Italian registry for myelofibrosis with myeloid metaplasia	National	Public
Ring14 Clinical database	International	Public
RIPID - Italian registry of diffuse infiltrative pneumopathies	National	Public
RNIC: National Registry of patients with Congenital Hypothyroidism	National	Public
ROI: Osteogenesis Imperfecta Registry	National	Public
SCNIR: Italian severe chronic neutropenia registry - contributes to the SCN international registry	National	Public
Sicilian Registry of Thalassemia and Hemoglobinopathies (Re.S.T.E.)	Regional	Public

Telethon-UILDM registry CMD (Congenital Muscular Dystrophy) - contributes to european Treat-NMD network	National	Private not-for-profit
Telethon-UILDM registry FSHD (Facioscapulohumeral dystrophy) - contributes to european Treat-NMD network	National	Private not-for-profit
Telethon-UILDM registry LGMD (Limb Girdle Muscular Dystrophy) - contributes to european Treat-NMD network	National	Private not-for-profit
The Global Registry of Hereditary Angioedema Type I and II	International	Private not-for-profit
The Italian multi-region thalassaemia registry	National	Private not-for-profit
The Italian Registry of Hereditary Angioedema	National	Private not-for-profit
Transthyretin amyloid neuropathy (TTR-FAP) italian registry	National	Private not-for-profit
TTP: International registry on thrombotic thrombocytopenic purpura	International	Public
Tuscan Registry of Congenital Anomalies	Regional	Public
Tuscan Registry of Rare Diseases	Regional	Public
V-RIAT: variant Ataxia telangiectasia Italian registry	National	Public
LT - Lithuania (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Lithuanian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
LU - Luxembourg (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Luxembourgers cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
LV - Latvia (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Latvian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
MK - Republic of North Macedonia (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Macedonian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
MT - Malta (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
MALTA CONGENITAL ANOMALIES REGISTRY (MCAR) - it contributes to EUROCAT	National	Public

Maltese cancer registry - contributes to the RARECARE project	National	Public
NL - Netherlands (the) (38 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
ALS database	National	Public
CONCOR: Dutch registration of adult patients with congenital heart disease	National	Public
DDRMD - Dutch Diagnosis Registration Metabolic Diseases	National	Public
Diffuse Intrinsic Pontine Glioma (DIPG) Registry	European	Private not-for-profit
Duchenne and Becker muscular dystrophy patient registry in the Netherlands - part of the TREAT-NMD network	National	Public
Dutch alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Dutch CF Registry - contributes to the European Cystic Fibrosis Society Patient Registry (ECFSPR)	National	Private not-for-profit
Dutch Neuromuscular Database CRAMP: Computer Registry of All Myopathies and Polyneuropathies	National	Private not-for-profit
Dutch patient registry for Fabry disease	National	Public
Dutch patient registry for Gaucher disease	National	Public
Dutch patient registry for Niemann-Pick Disease Type B	National	Public
Dutch patient registry for Niemann-Pick Disease Type C	National	Public
Dutch severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
E-HOD - European network and registry for homocystinurias and methylation defects (contributes to U-IMD)	European	Public
EPCOT: European prospective cohort on thrombophilia	European	Public
ERN [BOND] - EuRR-Bone: European Registry for Rare Bone and Mineral Conditions	European	Public
ERN [CRANIO] - ERN CRANIO registry	European	Public
ERN [ERNICA] - ERNICA-registry for improving care	European	Public
ERN [eUROGEN] - ERN eUROGEN Registry for rare urogenital diseases	European	Public
ERN [GENTURIS] - GENTURIS registry: The ERN Genetic Tumour Risk Syndromes Registry	European	Public
ERN [GUARD-Heart] - The Heart-Core Registry: a Gateway to Uncommon and Rare Diseases of the Heart	European	Public
ESPN/ERA-EDTA Registry: European Registry for Children on Renal Replacement Therapy	European	Public
EUROCAT Northern Netherlands - contributes to the international EUROCAT network	Regional	Public

European Parathyroid Tumor Registry	European	Public
FSHD registry	National	Private not-for-profit
Galactosemia Patient Registry - part of the Galactosemia Network	International	Public
International Dystrophic Epidermolysis Bullosa Patient Registry	International	Private not-for-profit
Membranous Nephropathy Registry	International	Public
PedNet Haemophilia Registry	International	Private not-for-profit
PHARMACHILD patient registry: Long-term pharmacovigilance for adverse effects in childhood arthritis, focusing on immune modulatory drugs (part of PRINTO network)	International	Public
RD5000 database: Dutch Registry for Inherited Retinal Dystrophies	National	Private not-for-profit
Registry NXT - Genzyme's Rare Disease Registries	International	Private for profit
STRIVE: A Long-term, Multi-center, Longitudinal Post-marketing, Observational Registry to Assess Long Term Safety and Effectiveness of HUMIRA® (Adalimumab) in Children With Moderately to Severely Active Polyarticular or Polyarticular-course Juvenile Idiopathic Arthritis (JIA) - NL	National	Public
The ALD Connect Patient Portal	International	Private not-for-profit
The International Pompe registry	International	Private for profit
The International Collaborative Gaucher Group (ICGG) Gaucher registry	International	Private for profit
The International Fabry registry	International	Private for profit
The International Mps I registry	International	Private for profit
NO - Norway (7 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
EURADRENAL: pathophysiology and natural course of autoimmune adrenal failure in Europe (registries)	European	Public
European Porphyria Registry (EPR)	European	Public
HUE-MAN patient registry on alpha mannosidosis	European	Public
Norwegian Porphyria Registry	National	Public
Norwegian cancer registry - contributes to the RARECARE project	National	Public
Norwegian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Norwegian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
PL - Poland (10 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Cracow cancer registry - contributes to the RARECARE project	Regional	Public

Kielce cancer registry - contributes to the RARECARE project	Regional	Public
Mazovian Cancer Registry - contributes to the RARECARE project	Regional	Public
Polish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Polish Registry of Inherited Tubulopathies (POLtube)	National	Public
Polish Registry of Patients with Neuromuscular Diseases contributes to the TREAT-NMD network	National	Public
Polish registry of primary immunodeficiencies - contributes to the ESID European registry	National	Public
Polish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
PRCM: Polish registry of congenital malformations - contributes to the EUROCAT network	National	Public
Registry for Wolfram syndrome, Astrom syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes contributing do EURO-WABB	National	Public
PT - Portugal (16 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Açores regional cancer patient registry (RORA)	Regional	Public
Adrenal tumor national registry	National	Public
Central regional cancer patient registry	Regional	Public
Duchenne and Becker muscular dystrophy patient registry in Portugal - contributes to the TREAT-NMD network	National	Public
North regional cancer registry (RORENO)	Regional	Public
Pituitary tumors national registry	National	Public
Portuguese cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Portuguese Fabry patient registry - contributing to the international Fabry registry	National	Private for profit
Portuguese registry for alpha-1 antitrypsin deficiency	National	Public
Portuguese registry for bronchiectasis	National	Public
Portuguese registry of primary immunodeficiency diseases (REPORID)	National	Public
Portuguese Rett syndrome registry	National	Public
Portuguese severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
RENAC - Portuguese registry of congenital anomalies (contributes to the EUROCAT network)	National	Public
Southern Portugal cancer registry - contributes to the RARECARE project	Regional	Public
Vila Nova de Gaia regional cancer registry (ROG)	Regional	Public

RO - Romania (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Romanian biliary atresia registry	National	Public
Romanian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
RS - Serbia (4 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Serbian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Serbian registry of hemophilia and von Willebrand disease patients	National	Public
Serbian registry of patients with rare bleeding disorders - contributes to the RBDD international registry	National	Public
Serbian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
SE - Sweden (18 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
EUROCRINE: European Surgical Registry for Rare Endocrine Tumours	European	Public
FOS : Fabry Outcome Survey	National	Public
HOS : Hunter Outcome Survey	National	Public
IOS : Icatibant Outcome Survey for hereditary angioedema	International	Public
National registry on bronchopulmonary dysplasia	National	Public
SPAHR: Swedish Pulmonary Arterial Hypertension Registry	National	Public
SWEDCON: Swedish Registry of Congenital Heart Disease	National	Public
Swedish Acute Lymphoblastic Leukemia Registry	National	Public
Swedish Acute Myelogenous Leukemia Registry	National	Public
Swedish alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Swedish Childhood Cancer Registry	National	Private not-for-profit
Swedish Chronic Myeloid Leukemia Registry	National	Public
Swedish cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Public
Swedish Multiple Myeloma Registry	National	Public
Swedish Polyposis Registry	National	Public
Swedish Registry for Familial Amyloid Polyneuropathy	National	Public

Swedish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
SWEDROP: Swedish Registry for Retinopathy of Prematurity	National	Public
SI - Slovenia (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Slovenian cancer registry - contributes to the RARECARE project	National	Public
Slovenian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
SK - Slovakia (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
National cancer registry (contributes to the RARECARE project)	National	Public
Slovak cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
TR - Turkey (5 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Database setup for the visualisation and examination of oral ulcers in Behcet disease patients	National	Public
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Turkey - contributes to the TREAT-NMD network	National	Public
Turkish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Turkish pediatric atypical hemolytic uremic syndrome registry	National	Public
Turkish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
UA - Ukraine (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Spinal muscular atrophy patient registry in Ukraine - part of the TREAT-NMD network	National	Public

European registries

*In this table only heads of European registries are listed in order to avoid double-counting.

EUROPEAN REGISTRIES (84 registries)*		
ENGLISH LABEL OF THE REGISTRY/COHORT	COORDINATION	AFFILIATION
Central data registry of the European Competence Network on Mastocytosis (ECMN)	AT	Public
Thromboreductin-Registry	AT	Private for profit
ENETS: European Neuroendocrine Tumour Registry	BE	Public
ERN [EpiCARE] - EPICARE-GRANT: An Operational EPICARE Registry for all Rare and Complex Epilepsies	BE	Public
EUNEFRON: registry of the European network for the study of orphan nephropathies	BE	Public
Juvenile Inflammatory Rheumatism Cohorte (JIRcohort)	CH	Private not-for-profit
CEDATA-GPGE: Registry of Children with Inflammatory Bowel Disease in Germany and Austria	DE	Public
CERTAIN-LI: Cooperative European Paediatric TransplAnt INitiative Liver	DE	Private not-for-profit
CERTAIN: Cooperative European Paediatric Renal Transplant Initiative registry	DE	Private not-for-profit
EBAR: European Biliary Atresia Registry	DE	Public
ERN [ERKNet] - ERK-REG: ERKNet Registry for Rare Kidney Diseases	DE	Public
ERN [ERN-LUNG] : REGISTRY WAREHOUSE - RD Registry Data Warehouse	DE	Public
ERN [MetabERN] - UIMD: Unified European Registry for Inherited Metabolic DisordersUIMD: Unified European Registry for Inherited Metabolic Disorders	DE	Public
ERN [RARE-LIVER] - R-LIVER: Registry for Rare Liver Diseases of the ERN on hepatological diseases	DE	Public
ERN [RND] - ERN-RND Registry: The ERN-RND Rare Neurological Disease Registry	DE	Public
ESID: European registry of primary immunodeficiencies	DE	Public
EU-RHAB: European Rhabdoid Tumor Registry	DE	Public
EuCalNet: International Calciphylaxis Registry	DE	Private for profit
eurIPFreg: European idiopathic pulmonary fibrosis registry	DE	Public
EUROFA - EFACT: European Friedreich Ataxia Registry	DE	Public
European Alport registry	DE	Public
European chILD-registry and biobank of the european network for children's interstitial lung diseases (chILD-EU)	DE	Public
European registry of sporadic degenerative ataxia with adult onset	DE	Public
EUROSCA-R: European patient registry on spinocerebellar ataxias	DE	Public

PROGNOSIS : The Prospective German Non-CF-Bronchiectasis Registry - contributes to EMBARC registry	DE	Public
ERCUSYN: European registry on Cushing's syndrome	ES	Public
ERN [EuroBloodNet] - ENROL: European Rare Blood Disorders Platform	ES	Public
ERN [TRANSPLANT-CHILD] - PETER: PaEdiatric Transplantation European Registry	ES	Public
EUROMAC: Registry of patients affected by McArdle Disease	ES	Public
Cloud-R HAE Register Hereditary Angiodema	FR	Public
EDMUS: European Database for Multiple Sclerosis and other related diseases	FR	Public
ENETS Registry: European Neuro-Endocrine Tumors Group	FR	Public
ERN [EURO-NMD] - EURO-NMD Registry: Patient centered and interoperable registry hub for Rare Neuromuscular Diseases	FR	Public
ERN [EYE] - REDgistry: An interoperable sustainable European Rare Eye Disease Registry	FR	Public
ERN [ITHACA] - ILIAD Rare Diseases patient registry: an International Library of Intellectual disability and Anomalies of Development	FR	Public
ERN [PAEDCAN] - PARTNER: Paediatric Rare Tumours Networks - European Registry	IT	Public
ERN [Skin] - ERN-Skin REGISTRY: Interoperable ERN on Rare and Undiagnosed Skin Disorders	FR	Public
ERN [VASCERN] - VASCERN Registries	FR	Public
Escort-Hu: European sickle cell disease cohort-hydroxyurea	FR	Public
EU-CHS: European central hypoventilation syndrome registry	FR	Public
EURECHINOREG: European registry of alveolar echinococcosis	FR	Public
EurêClark registry (The European Clarkson's syndrome registry)	FR	Public
European LeukoDataBase	FR	Public
European prospective registry of children born to mothers affected by the antiphospholipids syndrome	FR	Public
European Society for Blood and Marrow Transplant Society Registry (EBMT registry)	FR	Public
EURORETT: European network on Rett syndrome (registry)	FR	Public
RaDiCo-ECYSCO: European cystinosis cohort	FR	Public
RaDiCo-EURBIO-Alport: Study of the natural history of Alport Syndrome by establishment of an international database	FR	Public
RaDiCo-IDMet: National and European cohort on Imprinting Disorders and their metabolic consequences	FR	Public
SCETIDE - Stem Cell Transplant for primary Immune Deficiencies in Europe	FR	Public
CCRN 995 (European Adrenal Insufficiency): A European multi-centre, multi-country, post-authorisation, observational study (registry) of patients with chronic adrenal insufficiency - UK	GB	Private for profit
ECARUCA: European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations	GB	Public

ECFS: European Cystic Fibrosis Society patient registry	GB	Public
EHR: European Haemoglobinopathy Registry	GB	Public
ERN [Endo-ERN] - EuRRECa: European Registries for Rare Endocrine Conditions	GB	Public
ERNEST - European Registry for Myeloproliferative Neoplasms towards a better understanding of Epidemiology, Survival and Treatment	GB	Private for profit
EUMDS: European Registry for Myelodysplastic Syndromes - part of EuroLeukemiaNet (ELN)	GB	Public
EURO-WABB: An EU Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes	GB	Public
EUROCAT: European surveillance of congenital anomalies	GB	Public
EURODSD: European disorders of sexual development registry	GB	Public
EuroMyositis registry	GB	Public
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	GB	Public
European Prader-Willi syndrome database	GB	Public
EUROWILSON: European network on Wilson disease (registries)	GB	Public
ERN [EURACAN] - STARTER: Starting an Adult Rare Tumor European Registry	IT	Public
ERN [ReCONNET] - TogethERN ReCONNET: a European Registry Infrastructure for data harmonization in rare and complex connective tissue and musculoskeletal diseases	IT	Public
ERN [RITA] - MERITA: A METADATA REGISTRY FOR THE ERN RITA	IT	Public
EUROFEVER: European registry for autoinflammatory diseases	IT	Public
HAE-registry: European hereditary angioedema patient registry	IT	Public
Diffuse Intrinsic Pontine Glioma (DIPG) Registry	NL	Private not-for-profit
E-HOD - European network and registry for homocystinurias and methylation defects	NL	Public
EPCOT: European prospective cohort on thrombophilia	NL	Public
ERN [BOND] - EuRR-Bone: European Registry for Rare Bone and Mineral Conditions	NL	Public
ERN [CRANIO] - ERN CRANIO registry	NL	Public
ERN [ERNICA] - ERNICA-registry for improving care	NL	Public
ERN [eUROGEN] - ERN eUROGEN Registry for rare urogenital diseases	NL	Public
ERN [GENTURIS] - GENTURIS registry: The ERN Genetic Tumour Risk Syndromes Registry	NL	Public
ERN [GUARD-Heart] - The Heart-Core Registry: a Gateway to Uncommon and Rare Diseases of the Heart	NL	Public
ESPN/ERA-EDTA Registry: European Registry for Children on Renal Replacement Therapy	NL	Public
European Parathyroid Tumor Registry	NL	Public

EURADRENAL: pathophysiology and natural course of autoimmune adrenal failure in Europe (registries)	NO	Public
European Porphyria Registry (EPR)	NO	Public
HUE-MAN patient registry on alpha mannosidosis	NO	Public
EUROCRINE: European Surgical Registry for Rare Endocrine Tumours	SE	Public

International registries coordinated in Europe

*In this table only heads of International registries are listed in order to avoid double-counting.

INTERNATIONAL REGISTRIES (77 registries)*		
ENGLISH LABEL OF THE REGISTRY/COHORT	COORDINATION	AFFILIATION
EMSA-SG: central patient registry of the European multiple system atrophy network	AT	Public
ENRAH: European alternating hemiplegia and rare epilepsies registry in childhood	AT	Public
International aHUS registry	AT	Public
EUROGLYCANET - International patient registry and cohort for congenital disorders of glycosylation	BE	Public
Pediatric granulomatous arthritis international registry	BE	Public
COST Action BM1105 Patient Registry - GnRH Network	CH	Public
Hereditary TTP Registry	CH	Public
Intercontinental Cooperative ITP Study Group Registry	CH	Public
Perihilar Cholangiocarcinoma International Registry	CH	Public
PNH Registry: Paroxysmal Nocturnal Hemoglobinuria registry	CH	Private for profit
ADPedKD - International, longitudinal registry including ADPKD patients followed up from childhood.	DE	Private not-for-profit
ARegPKD - International Registry Study on Autosomal Recessive Polycystic Kidney Disease	DE	Public
Blue cone monochromatism - Patient registry	DE	Private not-for-profit
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	DE	Private not-for-profit
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	DE	Private not-for-profit
DÖSAK tumor registry for documentation of tumors of the face and jaws in germany, austria and switzerland	DE	Public
E-IMD : European registry and network for Intoxication type Metabolic Diseases	DE	Public
EHDN: neuroacanthocytosis patient registry	DE	Public
Enroll-HD Registry	DE	Private not-for-profit
FungiScope - Global Rare Fungal Infection Registry	DE	Public
iNTD (International Working Group on Neurotransmitter Related Disorders) Registry	DE	Private not-for-profit
iNTD Network - International Working Group on Neurotransmitter Related Disorders	DE	Private not-for-profit
International registry for Primary Ciliary Dyskinesia	DE	Public
Kids Lung Register: International register and biobank for rare lung diseases	DE	Public
KINDLERNET: Central patient registry Kindler syndrome	DE	Public
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	DE	Private for profit

Nephronophthisis registry for patients in germany, austria and switzerland	DE	Private for profit
NHL-BFM Registry 2012: Registry of the NHL-BFM study group for all subtypes of Non-Hodgkin Lymphoma diagnosed in children and adolescents	DE	Public
OSTEOPETR: International registry of patients suffering from osteopetrosis	DE	Public
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	DE	Public
RegiSCAR: International registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples - patient registry	DE	Public
Register for rare myeloproliferative neoplasms	DE	Public
RetDis Database: clinical descriptions of patients and families with inherited eye diseases	DE	Public
SCNIR: Severe Chronic Neutropenia International Registry	DE	Public
STER: FVII deficiency treatment international registry	DE	Public
TIRCON: NBIA (Neurodegeneration with Brain Iron Accumulation) patient registry	DE	Public
RenalTube: Database of patients with primary tubulopathies	ES	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Immune thrombocytopenic purpura (ITP)	FR	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Lupus	FR	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Myositis	FR	Public
RaDiCo-GenIDA: International social network for data collection on the natural history of rare monogenic forms of intellectual disabilities	FR	Public
Registry for Patients with Digital Ulcers Associated with Systemic Sclerosis (DU/SSc)	FR	Public
VALDIG: cohort creation on Budd-Chiari syndrome, hepatic venoocclusive disease, hepatoportal sclerosis and portal vein thrombosis	FR	Public
Ataxia-Telangiectasia Society	GB	Public
CCRN 1055: Osteonecrosis of the Jaw (ONJ) Case Registry	GB	Private for profit
CCRN 2421 (Noonan/RASopathy patients): A registry study to characterize genetic and pathway biomarkers in Noonan syndrome and other RASopathy patients - UK	GB	Public
CCRN 3119 (Spondyloarthritis): Patients with axial spondyloarthritis: multicountry registry of clinical characteristics, including radiographic progression, and burden of disease over 5 years in real-life setting	GB	Private for profit
EHDN: registry of juvenile Huntington's disease	GB	Private not-for-profit
EUROCARE CF: European cystic fibrosis registry	GB	Public
Global FKRP registry (Global Fukutin-Related Protein defects registry) - Part of TREAT-NMD Alliance	GB	Public
I-DSD: International Disorders of Sex Development registry	GB	Public

International A-T Registry	GB	Public
International Niemann-Pick Disease Registry	GB	Public
Registry for Patients with Niemann-Pick Type C Disease	GB	Public
The Global Registry for COL6-related dystrophies	GB	Public
The MTM and CNM Registry - The Myotubular and Centronuclear Myopathy Patient Registry	GB	Public
TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases (registries)	GB	Public
TuberOus SCLerosis registry to increase disease Awareness (TOSCA)	GB	Private for profit
International registry of bone fragility fractures in the young	IT	Public
International Registry of congenital dyserythropoietic anemia II	IT	Public
International Registry of Rare Bleeding Disorders (RBDD)	IT	Public
International registry of recurrent and familial hemolytic uremic syndrome/thrombotic thrombocytopenic purpura	IT	Private not-for-profit
Ring14 Clinical database	IT	Public
The Global Registry of Hereditary Angioedema Type I and II	IT	Private not-for-profit
TTP: International registry on thrombotic thrombocytopenic purpura	IT	Public
Galactosemia Patient Registry - part of the Galactosemia Network	NL	Public
International Dystrophic Epidermolysis Bullosa Patient Registry	NL	Private not-for-profit
Membranous Nephropathy Registry	NL	Public
PedNet Haemophilia Registry	NL	Private not-for-profit
PHARMACHILD patient registry: Long-term pharmacovigilance for adverse effects in childhood arthritis, focusing on immune modulatory drugs (part of PRINTO network)	NL	Public
Registry NXT - Genzyme's Rare Disease Registries	NL	Private for profit
The ALD Connect Patient Portal	NL	Private not-for-profit
The International Pompe registry	NL	Private for profit
The International Collaborative Gaucher Group (ICGG) Gaucher registry	NL	Private for profit
The International Fabry registry	NL	Private for profit
The International Mps I registry	NL	Private for profit
IOS : Icatibant Outcome Survey for hereditary angioedema	SE	Public

Please note that all data presented in this report are available for download at www.orphadata.org

For any questions or comments, please contact us: contact.orphanet@inserm.fr

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