Orphanet Report Series

Rare Diseases collection

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Rare Disease Registries in Europe

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From science to health

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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.

List of rare diseases that are covered by the listed registries

Orpha Number	Disease name
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
988	Absent tibia-polydactyly syndrome
48818	Aceruloplasminemia
15	Achondroplasia
49382	Achromatopsia
101963	Acquired chronic primary adrenal insufficiency
73274	Acquired hemophilia
163391	Acrodermatitis continua of Hallopeau
79356	Acrokeratoderma
963	Acromegaly
99892	ACTH-dependent Cushing syndrome
318	Acute erythroid leukemia
293173	Acute generalized exanthematous pustulosis
79276	Acute intermittent porphyria
79126	Acute interstitial pneumonia
513	Acute lymphoblastic leukemia
518	Acute megakaryoblastic leukemia
514	Acute monoblastic 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
2666	Adult familial nephronophthisis-spastic quadriparesia syndrome
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 development anomaly
375	Anti-glomerular basement membrane disease
2299	Aortic arch interruption
87	Apert syndrome
320	Apparent mineralocorticoid excess
90	Argininemia
23	Argininosuccinic aciduria
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
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
34516	Autosomal dominant limb-girdle muscular dystrophy type 1D
34149	Autosomal dominant tubulointerstitial kidney disease
79278	Autosomal erythropoietic protoporphyria
281217	Autosomal ichthyosis syndrome
281097	Autosomal recessive congenital ichthyosis
34515	Autosomal recessive limb-girdle muscular dystrophy type 2I
731	Autosomal recessive polycystic kidney disease
2512	Autosomal recessive primary microcephaly
782	Axenfeld-Rieger syndrome
110	Bardet-Biedl syndrome
111	Barth syndrome

67038	B-cell chronic lymphocytic leukemia
86852	B-cell prolymphocytic leukemia
98895	Becker muscular dystrophy
116	Beckwith-Wiedemann syndrome
117	Behçet disease
848	Beta-thalassemia
231214	Beta-thalassemia major
610	Bethlem myopathy
30391	Biliary atresia
415286	Bilirubin encephalopathy
179	Birdshot chorioretinopathy
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
136	CADASIL
280062	Calciphylaxis
137667	Capillary malformation-arteriovenous malformation syndrome
147	Carbamoyl-phosphate synthetase 1 deficiency
56044	Carcinoma of gallbladder and extrahepatic biliary tract
167848	Cardiomyopathy
97286	Carney-Stratakis syndrome
160	Castleman disease
464343	Catastrophic antiphospholipid syndrome
717	Catecholamine-producing tumor
3027	Caudal regression sequence
86870	CD4+/CD56+ hematodermic neoplasm
597	Central core disease
595	Centronuclear myopathy
166	Charcot-Marie-Tooth disease
55880	Chondrosarcoma
180	Choroideremia
379	Chronic granulomatous disease
2932	Chronic inflammatory demyelinating polyneuropathy
521	Chronic myeloid leukemia
101959	Chronic primary adrenal insufficiency
247525	Citrullinemia type I
391	Classic Hodgkin lymphoma
394	Classic homocystinuria
1991	Cleft lip with or without cleft palate
2014	Cleft palate
31824	Colchicine poisoning
468672	Colobomatous macrophthalmia-microcornea syndrome
1198	Colonic atresia
35909	Combined deficiency of factor V and factor VIII
1572	Common variable immunodeficiency
1329	Complete atrioventricular canal
1872	Cone rod dystrophy
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral
418	Congenital adrenal hyperplasia
2040	Congenital bronchobiliary fistula
2140	Congenital diaphragmatic hernia
137	Congenital disorder of glycosylation
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
442	Congenital hypothyroidism
68378	Congenital limb malformation
1928	Congenital lobar emphysema
69063	Congenital membranous nephropathy due to maternal anti-neutral endopeptidase alloimmunization
97242	Congenital muscular dystrophy
370953	Congenital muscular dystrophy due to dystroglycanopathy
590	Congenital myasthenic syndrome
97245	Congenital myopathy
98904	Congenital myopathy with excess of thin filaments
206973	Congenital myotonia
79394	Congenital non-bullous ichthyosiform erythroderma
2444	Congenital pulmonary airway malformation
2414	Congenital pulmonary lymphangiectasia
3161	Congenital pulmonary sequestration
3090	Congenital pulmonary venous return anomaly
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
319651	Constitutional megaloblastic anemia with severe neurologic disease
101987	Constitutional neutropenia
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
1666	Dextrocardia
146	Differentiated thyroid carcinoma
90060	Diffuse alveolar hemorrhage
544	Diffuse large B-cell lymphoma
31828	Digitalis poisoning
226	Dihydropteridine reductase deficiency
309830	Disorder of catecholamine synthesis
309830	Disorder of catecholamine synthesis
79175	Disorder of gamma-aminobutyric acid metabolism
309819	Disorder of pterin metabolism
79167	Disorder of urea cycle metabolism and ammonia detoxification
599	Distal myopathy

18	Distal renal tubular acidosis
255	Dopa-responsive dystonia
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
1775	Dyskeratosis congenita
303	Dystrophic epidermolysis bullosa
256	Early-onset generalized limb-onset dystonia
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
183	Eosinophilic granulomatosis with polyangiitis
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
319	Ewing sarcoma
883	Extragonadal 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 predisposition cancer syndrome
540	Familial hemophagocytic lymphohistiocytosis
656	Familial idiopathic steroid-resistant nephrotic syndrome
93217	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis
93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis
209886	Familial juvenile hyperuricemic nephropathy 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
84	Fanconi anemia
101039	Female restricted epilepsy with intellectual disability
1987	Femoral agenesis/hypoplasia
249	Fibrous dysplasia of bone
48918	Focal myositis
1866	Focal, segmental or multifocal dystonia
86902	Follicular dendritic cell sarcoma
545	Follicular lymphoma
51208	Formiminoglutamic aciduria
347	Frasier syndrome
95	Friedreich ataxia
227796	Fundus albipunctatus
352	Galactosemia
314022	Gastric adenocarcinoma and proximal polyposis of the stomach
36273	Gastric linitis plastica
100092	Gastroenteropancreatic neuroendocrine tumor
44890	Gastrointestinal stromal tumor
2368	Gastroschisis
355	Gaucher disease
36387	Generalized epilepsy with febrile seizures-plus
247353	Generalized pustular psoriasis
3221	Generalized resistance to thyroid hormone
101960	Genetic chronic primary adrenal insufficiency
183497	Genetic neuromuscular disease
435554	Genetic precocious puberty
183592	Genetic renal tubular disease
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
2102	GTP cyclohydrolase I deficiency
168569	H syndrome
99803	Haddad syndrome
73229	HANAC syndrome
457	Harlequin ichthyosis
2130	Hemimelia
139491	Hemochromatosis type 4
68364	Hemoglobinopathy
158032	Hemophagocytic syndrome
448	Hemophilia
98878	Hemophilia A
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 coproporphyria
26106	Hereditary diffuse gastric cancer
2024	Hereditary gingival fibromatosis
774	Hereditary hemorrhagic telangiectasia
523	Hereditary leiomyomatosis and renal cell cancer
79357	Hereditary palmoplantar keratoderma
29072	Hereditary pheochromocytoma-paraganglioma
264675	Hereditary pulmonary alveolar proteinosis
213524	Hereditary site-specific ovarian cancer syndrome
86896	Histiocytic sarcoma
2162	Holoprosencephaly
395	Homocystinuria due to methylene tetrahydrofolate reductase deficiency
622	Homocystinuria without methylmalonic aciduria
56970	Human prion disease
399	Huntington disease
2182	Hydrocephalus with stenosis of the aqueduct of Sylvius
927	Hyperammonemia due to N-acetylglutamate synthase deficiency
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
31740	Hypersensitivity pneumonitis
2248	Hypoplastic left heart syndrome
3332	Hypoplastic tibiae-postaxial polydactyly 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
90003	IgG4-related hepatopathy
49041	IgG4-related retroperitoneal fibrosis
3002	Immune thrombocytopenic purpura
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
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
182095	Interstitial lung disease
264735	Interstitial lung disease specific to adulthood
104010	Intestinal polyposis syndrome
1048	Isolated anencephaly/exencephaly
557	Isolated anorectal malformation
30391	Isolated biliary atresia
238666	Isolated congenital hypogonadotropic hypogonadism
718	Isolated Pierre Robin syndrome
823	Isolated spina bifida
2440	Isolated split hand-split foot malformation
33	Isovaleric acidemia
474	Jeune syndrome
220497	Joubert syndrome with renal defect
92	Juvenile idiopathic arthritis
248111	Juvenile Huntington disease
93568	Juvenile polymyositis
2322	Kabuki syndrome
2331	Kawasaki disease
2908	Kindler syndrome
99978	Klatskin tumor
33543	Kleine-Levin syndrome
313	Lamellar ichthyosis
98301	Laminopathy
389	Langerhans cell histiocytosis
264955	Langerhans cell histiocytosis in childhood and adulthood
264750	Langerhans cell histiocytosis specific to adulthood
264724	Langerhans cell histiocytosis specific to childhood
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	Lymphangioleiomyomatosis
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 partial seizurzs of infancy
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 cblE
2170	Methylcobalamin deficiency type cblG
26	Methylmalonic acidemia with homocystinuria
79284	Methylmalonic acidemia with homocystinuria type cblF
79282	Methylmalonic acidemia with homocystinuria, type cblC
369955	Methylmalonic acidemia with homocystinuria, type cblJ
280183	Methylmalonic aciduria due to transcobalamin receptor defect
83642	Microcytic anemia with liver iron overload
568	Microphthalmia, Lenz type
98555	Microphthalmia-anophthalmia-coloboma
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
79213	Mucopolysaccharidosis
579	Mucopolysaccharidosis type 1
580	Mucopolysaccharidosis type 2
641	Multifocal motor neuropathy
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
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency
877	Neuroendocrine tumor
636	Neurofibromatosis type 1
637	Neurofibromatosis type 2
93921	Neurofibromatosis type 3
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
86867	Nodal marginal zone B-cell lymphoma
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
94080	Non-secreting paraganglioma
91364	Non-specific interstitial pneumonia
498467	Non-syndromic postaxial polydactyly
498464	Non-syndromic preaxial polydactyly
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
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
668	Osteosarcoma
213500	Ovarian cancer
93460	Overgrowth syndrome
677	Pancreatoblastoma
2807	Papilloma of choroid plexus
31827	Paraquat poisoning
143	Parathyroid carcinoma
447	Paroxysmal nocturnal hemoglobinuria
1330	Partial atrioventricular canal
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

738	Porphyria
101330	Porphyria cutanea tarda
79473	Porphyria variegata
854	Portal vein thrombosis
294942	Postaxial polydactyly of fingers
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
172	Progressive familial intrahepatic cholestasis
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 retardation due to S-adenosylhomocysteine hydrolase deficiency
306498	PTEN hamartoma tumor syndrome
1578	Pterin-4-alpha-carbinolamine dehydratase deficiency
182090	Pulmonary arterial hypertension
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
102002	Rare ataxia
93419	Rare bone disease
68411	Rare bone tumor
180250	Rare breast tumor
101945	Rare bronchopulmonary tumor
218436	Rare cardiac rhythm disease
167848	Rare cardiomyopathy
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
98059	Rare digestive tumor
101953	Rare dyslipidemia
68363	Rare dystonia
101998	Rare epilepsy
97966	Rare eye disease
180821	Rare gastroesophageal tumor
183625	Rare genetic diabetes mellitus
98053	Rare genetic disease
101435	Rare genetic eye disease
158300	Rare genetic hematologic disease
98056	Rare genetic renal 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
220489	Rare hereditary hemochromatosis
217454	Rare hereditary thrombophilia
68367	Rare inborn errors of metabolism
104012	Rare inflammatory bowel disease
87277	Rare intellectual disability
180257	Rare malignant breast tumor
68329	Rare maxillo-facial surgical disease
98062	Rare nervous system tumor
98006	Rare neurologic disease
98026	Rare odontologic disease
98061	Rare otorhinolaryngologic tumor
181415	Rare primary hyperaldosteronism
101944	Rare pulmonary disease
71198	Rare pulmonary hypertension
93626	Rare renal disease
93603	Rare renal tubular disease
280342	Rare rheumatological disease of childhood
89826	Rare skin disease
79386	Rare skin tumor or hamartoma
71209	Rare soft tissue tumor
98023	Rare systemic or rheumatologic disease
280342	Rare systemic or rheumatological disease of childhood
248361	Rare thrombotic disorder due to a constitutional coagulation factors defect
98057	Rare tumor
104011	Rare tumor of intestine
180824	Rare tumor of pancreas
182114	Rare urogenital tumor
180151	Rare vaginal malformation
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
1848	Renal agenesis, bilateral
71862	Retinal dystrophy
791	Retinitis pigmentosa
790	Retinoblastoma
90050	Retinopathy of prematurity
778	Rett syndrome
69077	Rhabdoid tumor
85408	Rheumatoid factor-negative juvenile idiopathic arthritis
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis

59315	Rhombencephalosynapsis
140976	RHYNS syndrome
1440	Ring chromosome 14 syndrome
158014	Rosaï-Dorfman disease
2909	Rothmund-Thomson syndrome
140969	Saldino-Mainzer syndrome
797	Sarcoidosis
799	Schizencephaly
801	Scleroderma
3156	Senior-Loken syndrome
42738	Severe congenital neutropenia
745	Severe hereditary thrombophilia due to congenital protein C deficiency
3162	Sézary syndrome
811	Shwachman-Diamond syndrome
232	Sickle cell anemia
275752	Sickle cell disease and related diseases
813	Silver-Russell syndrome
816	Sjogren-Larsson syndrome
820	Sneddon syndrome
86854	Splenic marginal zone lymphoma
247234	Sporadic adult-onset ataxia of unknown etiology
84271	Sporadic idiopathic steroid-resistant nephrotic syndrome
93220	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis
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
188	Systemic capillary leak syndrome
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
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
68347	Tumor of hematopoietic and lymphoid tissues
90038	Typical hemolytic-uremic syndrome
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
1667	Wolcott-Rallison syndrome
3463	Wolfram syndrome
910	Xeroderma pigmentosum
178461	X-linked myopathy with postural muscle atrophy
431272	X-linked scapuloperoneal muscular dystrophy
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	GLOBAL	TOTAL
AT - Austria	6	27	4	1	38
BE - Belgium	2	14	2	2	20
BG - Bulgaria	0	8	0	0	8
CH - Switzerland*	1	13	2	4	20
CY - Cyprus	0	2	0	0	2
CZ - Czech Republic	0	5	0	0	5
DE - Germany	13	94	18	24	149
DK - Denmark	1	4	0	0	5
EE - Estonia	0	3	0	0	3
ES - Spain	12	45	3	0	60
FI - Finland	0	7	0	0	7
FR - France	19	103	11	6	143
GR - Greece	0	3	0	0	3
HR - Croatia	0	2	0	0	2
HU - Hungary	0	6	0	0	6
IE - Ireland	3	13	0	1	17
IL - Israel*	0	2	0	0	2
IS - Iceland*	0	2	0	0	2
IT - Italy	13	60	4	6	83
LT - Lithuania	0	1	0	0	1
LU - Luxembourg	0	1	0	0	1
LV - Latvia	0	1	0	0	1
MK - Republic of Macedonia*	0	1	0	0	1
MT - Malta	0	2	0	0	2
NL - Netherlands	1	16	5	10	32
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	14	0	3	17
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
UK - United Kingdom	1	48	13	12	74
TOTAL	80	535	69	69	753

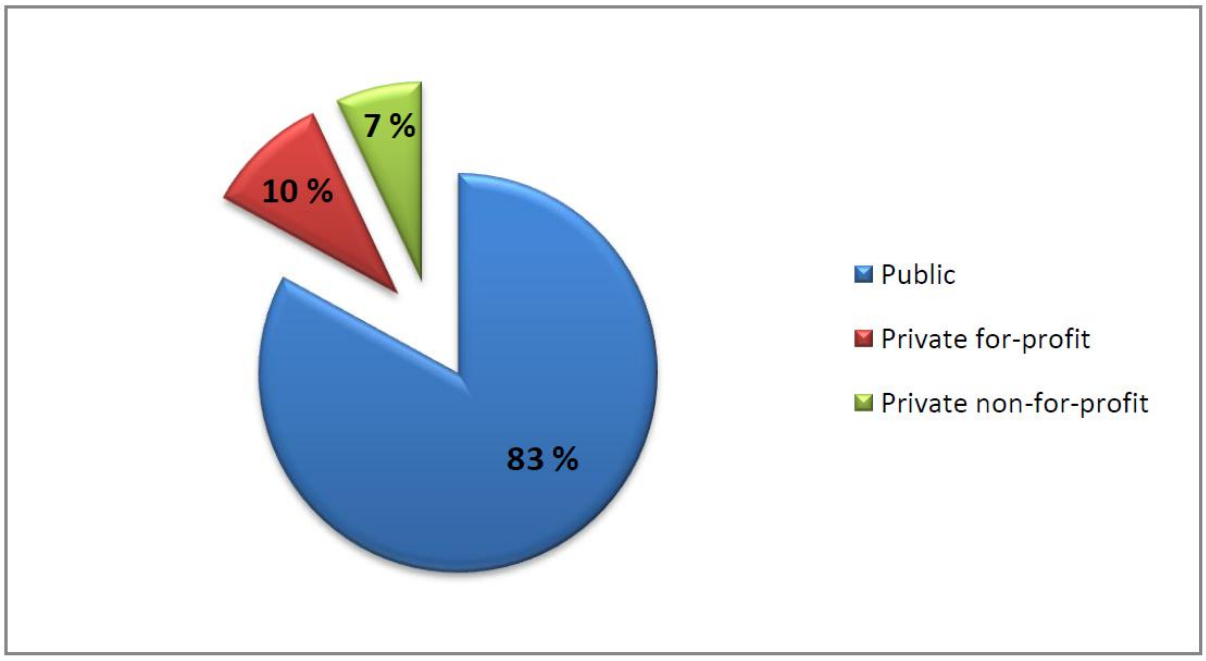
*neighbour countries participating to the Orphanet consortium

2- Distribution of registries by coverage

COVERAGE	NUMBER OF REGISTRIES *
Regional	80
National	535
European	69
Global	69
TOTAL	753

*all the registries, including heads of registries networks and its nodes are counted

3- Distribution of registries by affiliation



Distribution of registries by country

AT - AUSTRIA (38 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 acromegaly registry	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 - contributes to the RARECARE project	National	Public
Austrian chronic myeloid leukemia registry	National	Private non-for-profit
Austrian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Austrian GIST registry	National	Private non-for-profit
Austrian Hemophilia Registry - Registry for patients with inborn coagulation defects	National	Public
Austrian myeloma registry	National	Private non-for-profit
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
Ceprotin Treatment Registry	National	Public
Cleft registry of the Austrian Cleft Plate 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	European	Public
ERN-PAEDCAN Partner: Paediatric Rare Tumours Networks – European Registry [PARTNER]	European	Public
Innsbruck registry for Adamantiades-Behcet disease: Retrospective and prospective data collection	Regional	Public
International aHUS registry	Global	Public
MDS: Austrian myelodysplastic syndromes patient 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 (20 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Antwerpen registry of congenital anomalies - contributes to the EUROCAT network	Regional	Private for-profit
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	Private for-profit
Belgian cystic fibrosis patient registry (BMR-RBM) - contributes to the EUROCARE CF and the ECFS registries	National	Public
Belgian familial adenomatous polyposis registry	National	Private non-for-profit
Belgian Neuromuscular Disease Registry	National	Public
Belgian patient database for Wilson disease - contributes to the EuroWilson registry (terminated)	National	Private non-for-profit
Belgian registry of primary immunodeficiencies - contributes to the ESID European registry	National	Private for-profit
Belgian severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
Belgian sickle cell anemia registry	National	Public
Belgian systemic sclerosis cohort	National	Public
Central Registry of Rare Diseases	National	Public
ENETS: European Neuroendocrine Tumor Registry	European	Private for-profit
ENRAH: Belgian contribution to European registry for alternating hemiplegia in childhood	National	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	Global	Private for-profit
Haemoglobinopathies - database	National	Private for-profit
Hainault and Namur registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Pediatric granulomatous arthritis international registry	Global	Private for-profit

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	Private for-profit
National registry of adult patients with chronic myeloid leukemia	National	Public
National registry of patients with Gaucher disease	National	Public
National registry of patients with mucopolysaccharidosis type II (MPS2)	National	Public
National registry of patients with phenylketonuria	National	Public
National registry of patients with primary immunodeficiencies (PID)	National	Public
National registry of patients with thalassaemia major	National	Public
National registry of patients with Wilson disease	National	Public

CH - SWITZERLAND (20 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
COST Action BM1105 Patient Registry - GnRH Network	European	Public
EUROCAT (Switzerland, Vaud) - Registry of congenital malformations of canton Vaud	Regional	Public
Hereditary TTP Registry	Global	Public
Intercontinental Cooperative ITP Study Group Registry	Global	Public
Juvenile Inflammatory Rheumatism (JIR) Cohorte (JIR-cohorte)	European	Private non-for-profit
Paediatric registry for congenital pulmonary malformations	National	Public
Perihilar Cholangiocarcinoma International Registry	Global	Public
PNH Registry: Paroxysmal Nocturnal Hemoglobinuria registry	Global	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 non-for-profit
Swiss Childhood Cancer Registry	National	Public
Swiss Cleft Lip and Palate Registry	National	Public
Swiss patient registry for Duchenne/Becker Muscular Dystrophy and Spinal Muscular Atrophy - contributes to the TREAT - NMD network	National	Public
Swiss Primary Biliary Cholangitis Cohort Study	National	Private non-for-profit
Swiss Primary Ciliary Dyskinesia Registry - contributing to the International PCD Registry	National	Public
Swiss Primary Sclerosing Cholangitis Cohort Study	National	Private non-for-profit
Swiss registries for Interstitial and Orphan Lung Diseases (SIOLD Registries)	National	Public
Swiss Registry for Neuromuscular Disorders	National	Private non-for-profit
Swiss registry of biliary atresia - contributes to the EBAR registry	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 EUROCARE CF registry	National	Private for-profit
NMDcy: Cypriot Neuromuscular Diseases - contributes to TREAT-NMD	National	Private for-profit

CZ - CZECH REPUBLIC (5 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Autoimmune neuromuscular disease	National	Private for-profit
Czech cystic fibrosis patient registry - contributes to the EUROCARE 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	Private for-profit
Spinal muscular atrophy patient registry in the Czech Republic - part of the TREAT- NMD network	National	Private for-profit

DE - Germany (149 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
ALS registry Nordrhein-Westfalen	Regional	Public
ALS registry Rheinland-Pfalz	Regional	Public
ALS registry Swabia	Regional	Public
ARegPKD - an international registry study for autosomal recessive polycystic kidney disease	Global	Public
Ataxia-Telangiectasia patient registry - contributes to the ESID Database	National	Public
Bayern population based cancer registry	Regional	Public
Bremen cancer registry	Regional	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
Child liver tumor registry	National	Public
Childrens 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	National	Public
Conn Registry: German registry of primary aldosteronism	National	Public

Cooperative European Paediatric Renal Transplant Initiative registry	Global	Private non-for-profit
Core documentation of children and adolescents with rheumatic diseases	National	Private non-for-profit
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	Global	Public
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	Public
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	Global	Public
DCLLSG registry - 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	European	Public
Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Austria and Germany - part of the TREAT-NMD network	National	Public
DYSTRACT: German Dystonia Registry	National	Public
EBAR: European Biliary Atresia Registry	European	Public
EHDN: neuroacanthocytosis patient registry	Global	Public
EIMD: European registry and network for intoxication type metabolic diseases (contributes to U-IMD)	European	Public
EKRS: Saarland Cancer Registry - contributes to the RARECARE Project	Regional	Private non-for-profit
ENETS: European Neuroendocrine Tumour Registry	European	Public
Enroll-HD Registry	Global	Private non-for-profit
Epidemiological cancer registry Baden-Württemberg	Regional	Public
ERN [ERN-LUNG] – Rare Respiratory Disease Registry	European	Public
ERN [MetabERN] - UIMD: Unified European Registry for Inherited Metabolic Disorders	European	Public
ERN [ERKNet] - ERK-REG: ERKNet Registry for Rare Kidney Diseases	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	Public
eurIPFreg: 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	National	Public
FACE - National registry for Robin sequence	National	Public

FungiScope - Global Rare Fungal Infection Registry	Global	Public
GeNeMove: German database for wilson disease	National	Public
German acromegaly registry	National	Public
German adrenal tumors registry	National	Private non-for-profit
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 EuCalNet	National	Public
German central registry for Sickle cell disease	National	Public
German childhood cancer registry (Partner of ACCIS: Automated Childhood Cancer Information System)	National	Public
German CMT-patient registry Germay – part of the TREAT-NMD network	National	Public
German cystic fibrosis registry - contributes to the EUROCARE CF registry	National	Public
German epilepsy registry	National	Public
German Fanconi anemia registry	National	Public
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	Public
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	Public
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	Private non-for-profit
German registry for congenital heart defects - part of the competence network for congenital heart defects	National	Private non-for-profit
German registry for congenital thrombocytopenia	National	Public
German Registry for Incidental Gallbladder Carcinoma	National	Private non-for-profit
German registry for LCH (Langerhans Cell Histiocytosis) in childhood	National	Public
German registry for Morbus Adamantiades-Behçet e.V.	National	Private non-for-profit
German registry for neonates, infants, children, adolescents, and adults with newly diagnosed and/or relapsed neuroblastic tumors - 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

German vasculitis registry	National	Public
Germany patient registry for membranoproliferative glomerulonephritis	National	Public
GMALL-registry: registry for adult patients with acute lymphoblastic leukemia or related diseases	National	Public
GOLDnet: Registry for Diffus Parenchymal Lung Disease	National	Public
GPOH-MET Registry: Registry for children and adolescents with malignant endocrine tumour	National	Public
GPOH-registry sickle cell disease	National	Private non-for-profit
Hamburg cancer registry	Regional	Public
HepNet: German hepatocellular carcinoma (HCC) registry	National	Public
Hunter Outcome Survey (HOS): patient registry	Global	Public
INSIGHTS-IPF Investigating Significant Health Trends in Idiopathic Pulmonary Fibrosis	National	Public
iNTD (International Working Group on Neurotransmitter Related Disorders) Registry (contributes to U-IMD)	Global	Public
International Collaborative Gaucher Group (ICGG) Gaucher Registry	Global	Public
International pheochromocytoma and paraganglioma registry	Global	Public
INVM (Isolated Noncompaction of Ventricular Myocardium) registry	National	Private for-profit
Kids Lung Register: International register and biobank for rare lung diseases	Global	Public
KINDLERNET: Central patient registry Kindler syndrome	Global	Public
LBL Registry: Registry for children with lymphoblastic lymphoma	National	Public
Lupus nephritis registry	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: German myelodysplastic syndromes patient registry	National	Public
MEFOPA: registry for patients with rare Mendelian forms of Parkinson's Disease	European	Public
Mucopolysaccharidosis type 1 (MPS I) Registry	Global	Public
Mykke : German registry for children and adolescents with myocarditis	National	Public
Nationa CMT-patient registry germany - part of the TREAT-NMD network	National	Public
National FKRP-patient registry germany - part of the TREAT-NMD network	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 retroperitoneal fibrosis	National	Public
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	Global	Public
NeoExNET registry: patient registry for neuroendocrine tumors	National	Public
Nephronophthisis registry for patients in Germany, Austria and Switzerland	European	Public
NET-Registry: German neuroendocrine gastrointestinal tumors	National	Public

Neuro-Heart-Registry: Registry for patients with heart involment in neurovascular diseases	National	Public
Neuromyelitis optica patient registry	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	Global	Public
NIRK: national central registry for ichthyoses and related keratinization disorders	National	Public
NIRK: patient registry for autosomal recessive congenital ichthyosis	National	Public
NKR: German registry for adrenocortical carcinoma	National	Public
OSTEOPETR: International registry of patients suffering from osteopetrosis	Global	Public
Patient registry for primary hyperoxaluria - contributes to the OxalEurope-Network	National	Public
Patient registry for retinal degeneration PRO RETINA e.V	National	Private-non-for profit
Patient registry for primary hyperoxaluria - contributes to the OxalEurope-Network	National	Public
Patient registry of the German Network for Systemic Scleroderma	National	Public
PID-NET: German National Registry for Primary Immunodeficiencies (PID)	National	Public
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	Global	Public
Pompe Disease Registry	Global	Public
PROGNOSIS : The Prospective German Non-CF-Bronchiectasis Registry - contributes to EMBARC registry	National	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	Global	Public
Register for rare myeloproliferative neoplasms	Global	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 mitochondrial diseases (mitoREGISTER) - subproject of mitoNET	National	Public
Registry for Patients with WT1 Mutation Associated Diseases	National	Public
RetDis Database: clinical descriptions of patients and families with inherited eye diseases	Global	Public
Schleswig-Holstein cancer registry	National	Public
STEP Registry: Registry for rare tumors in children and adolescents	National	Public
STER: FVII deficiency treatment international registry	Global	Public
TIRCON: NBIA (Neurodegeneration with Brain Iron Accumulation) patient registry	Global	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	Private for-profit
Von Hippel-Lindau registry	Regional	Public

DK - DENMARK (5 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Danish cystic fibrosis patient registry - contributes to the EUROCARE 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 EUROCAT 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 - contributes to the RARECARE project	National	Public
Estonian cystic fibrosis patient registry - contributes to the EUROCARE 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 (60 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
aHUS/C3G: Database of atypical hemolytic uremic syndrome and C3 glomerulonephritis	National	Public
ECEMC: Registry of the Spanish Collaborative Study of Congenital Malformations	National	Public
Epidemiologic Registry of Patients Diagnosed With Acute Myeloid Leukemia	National	Private non-for-profit
ERCUSYN: European registry on Cushing's syndrome	European	Public
EUROMAC: Registry of patients affected by McArdle Disease	European	Public
EURO-WABB: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome, Bardet-Biedl Syndrome and Other Rare Diabetes Syndromes	European	Public
Fanconi anemia patient registry database	National	Public
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 Myopathies 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
PanGenFAM: Familial Pancreatic Cancer Registry	National	Public
Population registry of rare diseases and congenital anomalies of Cantabria	Regional	Public
Population registry of rare diseases of Balearic Islands	Regional	Public
Population registry of rare diseases of Navarra	Regional	Public
RACAV: Registry of congenital anomalies of the Basque Country - contributes to the EUROCAT network	Regional	Public
Rare disease registry of Aragon	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	Private non-for-profit
ReeR: Spanish National Registry of Rare Diseases	National	Public
REeRIGG4: Spanish IgG4-related Disease Registry	National	Public
Registro MEN: Spanish registry of multiple endocrine neoplasia, pheochromocitomas and paragangliomas	National	Public
Registry for rare diseases in Andalusia	Regional	Public
Registry for rare diseases in Extremadura	Regional	Public
REHAP: Spanish Registry of Pulmonary Arterial Hypertension	National	Public
REHEVASC: Spanish registry for hepatic vascular diseases	National	Public
REHIPED - Spanish Registry for Pediatric Pulmonary Hypertension	National	Public
RenalTube: Spanish patient registry of primary tubulopathies	National	Public
REPA: Spanish registry of alveolar proteinosis	National	Public
REPHem-SEHOP: Paediatric Haemoglobinopathies Spanish Registry	National	Public
RERGA: Registry for rare diseases in Galicia	Regional	Public
RETEGEP: Spanish Registry of Gastroenteropancreatic Neuroendocrine Tumors	National	Private non-for-profit
REWBA: Spanish Registry of Wolfram, Bardet-Biedl and Alstrom syndromes	National	Public
SIER: Information System on rare diseases in the Region of Murcia	Regional	Public
SIER-CV: Information System on rare diseases in Valencian Community	Regional	Public
SIERMA: Information system on rare diseases in Madrid	Regional	Public
SIERrm: Information System on rare diseases in the Region of Murcia	Regional	Public
Spanish alpha-1 antitrypsin deficiency registry (REDAAT) - contributes to the Alpha One International Registry (AIR)	National	Private non-for-profit
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	Private non-for-profit
Spanish patient registry of hereditary retinal dystrophy	National	Public
Spanish patient registry of myelodysplasic syndromes	National	Private non-for-profit
Spanish patient registry of rare diseases: multiple endocrine neoplasia, acromegaly and enteropancreatic endocrine tumors.	National	Public
Spanish patient registry of transmissible spongiform encephalopathies	National	Public
Spanish Registry of Cushing Syndrome - contributes to ERCUSYN	National	Private non-for-profit
Spanish registry of Duchenne muscular dystrophy - part of the TREAT-NMD network	National	Public
Spanish registry of lymphangioleiomyomatosis	National	Public
Spanish registry of patients with McArdle disease	National	Private for-profit
Spanish registry of pulmonary Langerhans cell histiocytosis	National	Public
Spanish registry of rare diseases	National	Public
Spanish registry of renal hereditary diseases	National	Public
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
FHRB: Finnish Hematology Register and Biobank	National	Public
Finnish IPF registry	National	Public
Finnish patient registry on Fabry disease - contributing to the international Fabry registry and Shire registry	National	Private for-profit

Finnish TREAT-NMD Patient Registry	National	Private non-for-profit
Registry of Congenital Malformations	National	Public
The Finnish Registry of Visual Impairment	National	Private non-for-profit

FR - FRANCE (143 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: CÖhort Angioedema Berinert	National	Public
CoF-AT study: a French cohort on ataxia-telangiectasia	National	Public
Cohort of patients with hereditary dystrophies of retina	National	Public
Côte d'Or registry of hematological malignancies	Regional	Public
D[4]/Phenodent: French registry of patients affected by rare odontologic diseases	National	Public
DM Scope: Myotonic dystrophy patient registry in France - part of the TREAT-NMD network	National	Public
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
ENET Registry: European Neuro-Endocrine Tumors Group	European	Public
EPI-EPNET: European hepatic and erythropoietic porphyrias registry	European	Public
EPIMAD: registry of chronic inflammatory intestine diseases in North-West	Regional	Public
Escort-Hu: European sickle cell disease cohort- hydroxyurea	European	Private for-profit
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
FranceCoag: French prospective cohort of patients affected with haemophilia or severe form of other hereditary hemorrhagic diseases except platelet disorders	National	Public
French acromegaly registry	National	Public
French addictive acute intoxications cohort	National	Public
French atypical sarcoïdosis 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	Public
French central hypoventilation syndrome registry - contributes 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	Public
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 Castleman's disease	National	Public
French cohort of common variable immunodeficiency with hypogammaglobulinemia in adults (CVID)	National	Public
French cohort of focal dystonia famillies	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 cohort and CFTR-RD cohort	National	Public
French cystic fibrosis patient registry	National	Public
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	Public
RE-LAM-CE : French National Lymphangioleiomyomatosis Registry	National	Public
French national patients registry on rare peritoneal tumor RENAPE	National	Public
BNDMR: French National Registry for Rare Diseases	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	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 registry of Amyotrophic Lateral Sclerosis	National	Public
French registry of the SDH-related hereditary paraglioma	National	Public
French registry for macrophagic myofasciitis	National	Public

French registry for right arrythmogenic ventricular dysplasia (ARVC/D)	National	Public
French Registry of Atypical Hemolytic Uremic Syndrome (aHUS) in Children	National	Public
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 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 Still disease patient registry	National	Public
French West Indies registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
FROG: FRench Observatory on Gaucher disease	National	Private for-profit
GENEPSO: French epidemiological cohort of BRCA systemic mutations carriers	National	Public
Gironde registry of hematological malignancies	Regional	Public
GMF: French registry of myelodysplastic syndromes and leukemia chemo- and radio-induced	National	Public
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	Private for-profit
ITINERAIR-pediatrie: French cohort of children with pulmonary arterial hypertension	National	Private for-profit
ITINERAIR-scleroderma: French pulmonary arterial hypertension screening cohort of patients with scleroderma	National	Private for-profit
KAWA-NET: epidemiological database of Kawasaki disease in France	National	Public
Congenital anomalies registry of Reunion Island - contributes to the EUROCAT network	Regional	Public
Laminopathies and emerinopathies patient registry	National	Public
LEA: children and adolescents with acute leukemia : propective cohort in France	National	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
Myotonic dystrophy patient registry in France - part of the TREAT-NMD network	National	Public
RaDiCo-EURBIO-Alport: Study of the natural history of Alport Syndrome by establishment of an international database	European	Public
RaDiCo-ECYSCO: European cystinosis cohort	European	Public

RaDiCo-GenIDA: International social network for data collection on the natural history of rare monogenic forms of intellectual disabilities	Global	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		
RaDiCo-FARD: National cohort for the evaluation of individual burden in the course of rare skin diseases	National	Public
RaDiCo-ACOStill: National cohort on adult and childhood onset Still disease	National	Public
RaDiCo-COBBALT: National cohort on Bardet-Bield syndrome and Alström syndrome	National	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-PID: French National cohort on Idiopathic Interstitial Pneumonia from infancy to elderly	National	Public
RaDiCo-RETICO: National cohort on inherited retinal dystrophies	National	Public
RaDiCo-MPS: National cohort on Mucopolysaccharidosis in the era of specific therapeutics	National	Public
RaDiCo-PP: National cohort on periodic paralysis : clinical, genetic and medico-economic studies involving m-health monitoring tools	National	Public
RaDiCo-DCP: National cohort on Primary Ciliary Dyskinesia (PCD) : Identification of specific severity criteria and phenotype-genotype correlation study	National	Public
RaDiCo-COLPAC: National cohort on the epidemiology, clinical and genetic heterogeneity of Low Phospholipid-Associated Cholelithiasis (LPAC) syndrome	National	Public
RaDiCo-SED-VASC: National cohort on vascular Ehlers-Danlos syndrome	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	Private for-profit
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx): Immune thrombocytopenic purpura (ITP)	Global	Private non-for-profit
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Lupus	Global	Private non-for-profit
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Myositis	Global	Private non-for-profit
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
Regional registry of thyroid cancers in Rhône-Alpes	Regional	Public
Registry and pronostic cohort of cutaneous lymphomas in Aquitaine	Regional	Public
Registry for digestive cancers in Burgundy	Regional	Public
Registry for Patients with Digital Ulcers Associated with Systemic Sclerosis (DU/SSc)	Global	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	Private non-for-profit
Rhône-Alpes registry of systemic mastocytosis	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

VALDIG: cohort creation on Budd-Chiari syndrome, hepatic venooclusive disease, hepatoportal sclerosis and portal vein thrombosis	Global	Public

GR - GREECE (3 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Greek cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Greek severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
National Registry for Haemoglobinopathies in Greece (NRHG)	National	Private for-profit

HR - CROATIA (2 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Croatian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Registry for neuromuscular disease	National	Private for-profit

HU - HUNGARY (6 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
DMD registry - contributes to the TREAT-NMD network	National	Public
Hungarian Angioedema Register	National	Public
Hungarian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Hungarian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
National NF Register	National	Private for-profit
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 EUROCARE CF registry	National	Public
Dublin registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Hunter Outcome Survey (HOS): patient registry	Global	Public
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	Private non-for profit
National Cleft Database - contributes to EUROCAT	National	Public
National Haemophilia Register	National	Public
National Severe Immunodeficiency Registry - contributes to ESID registry	National	Public
Rare Kidney Disease Registry & BioResource	National	Public
South East of Ireland registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public
South of Ireland registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Sudden Cardiac Death in the Young Registry	National	Public
The Irish Rare Kidney Disease Registry and Biobank	National	Public

IL - ISRAEL (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Israelian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Israelian 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**
Icelander cancer registry - contributes to the RARECARE project	National	Public
Icelander cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

IT - ITALY (83 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
AICE: Italian registry of hemophilia centre	National	Public
Arrhythmogenic right ventricular cardiomyopathy/dysplasia: clinical registry and database, evaluation of therapies	National	Public
Behçet's disease registry	National	Public
Campania registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Calabria 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	European	Public
EUROFEVER: European registry for autoinflammatory diseases	European	Public
EUROWILSON: Registry and network to improve the management of Wilson Disease	European	Public
FMF: Italian registry for familial mediterranean fever in the young	National	Public
Friedreich's ataxia Italian patient registry	National	Private non-for-profit
GLATIT: Glanzmann thrombasthenia Italian registry	National	Public
HAE-registry: European hereditary angioedema patient registry	European	Public

IBAHC: Italian registry for alternating hemiplegia of childhood	National	Public
INNCB Myasthenia gravis registry	National	Public
International registry of bone fragility fractures in the young	Global	Public
International Registry of congenital dyserythropoietic anemia II	Global	Public
International Registry of Rare Bleeding Disorders (RBDD)	Global	Public
International registry of recurrent and familial hemolytic uremic syndrome/thrombotic thrombocytopenic purpura	Global	Private non-for-profit
IPER: Registry of Juvenile idiopathic arthritis 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 genetic movement disorders registry	National	Public
Italian HLH Registry	National	Public
Italian LCH Registry	National	Public
Italian Li-Fraumeni syndrome registry	National	Public
Italian myelodysplastic syndromes registry	National	Public
FISM_registry: Italian Network MDS Registry	National	Public
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	Public
Italian Registry of adult patients affected by familial mediterranean fever	National	Public
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	Private non-for-profit
Italian Registry of Kernicterus and hyperbilirubinemia	National	Public
Italian registry of Legionellosis	National	Public
Italian Registry of membranoproliferative glomerulonephritis	National	Private non-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
LAZIO Lymphomas Database	Regional	Private non-for-profit
MITOCON: National registry mitochondrial disease	National	Public
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 non-for-profit
Rare Diseases Registry - Veneto Region	Regional	Public
Regional registry for neuromuscular disorders	Regional	Public
SIMaRRP: Regional Registry Rare Diseases of Puglia Region	Regional	Private non-for-profit
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 non-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 non-for-profit
Rett Syndrome Database	National	Public
RIAF: Fanconi's anemia Italian registry	National	Public
RIAT: Ataxia teleangiectasia Italian registry	National	Public
RIMM: Italian registry for myelofibrosis with myeloid metaplasia	National	Public
Ring14 Clinical database	Global	Private non-for-profit
RIPID: Italian registry of diffuse infiltrative pneumopathies	National	Public
RNIC: National Registry of Infant 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	Private for-profit
Telethon-UILDM registry CMD (Congenital Muscular Dystrophy) - contributes to european Treat-NMD network	National	Public
Telethon-UILDM registry FSHD (Facioscapulohumeral dystrophy) - contributes to european Treat-NMD network	National	Public
Telethon-UILDM registry LGMD (Limb Girdle Muscular Dystrophy) - contributes to european Treat-NMD network	National	Public
The Italian multi-region thalassaemia registry	National	Private non-for-profit
The Italian registry of Hereditary Angioedema	National	Private non-for-profit
Transthyretin amyloid neuropathy (TTR-FAP) italian registry	National	Private non-for-profit
TTP: International registry on thrombotic thrombocytopenic purpura	Global	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	Private for-profit

LU - LUXEMBOURG (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Luxembourgers cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

LV - LATVIA (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Latvian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

MK - REPUBLIC OF MACEDONIA (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Macedonian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

MT - MALTA (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Malta congenital anomalies registry (MCAR) - contributes to EUROCAT	National	Public
Maltese cancer registry - contributes to the RARECARE project	National	Public

NL - NETHERLANDS (32 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
ALS database	National	Public
CONCOR: Dutch registry of patients with a congenital heart malformation	National	Private for-profit
DDRMD: Dutch Diagnosis Registration Metabolic Diseases	National	Public
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 cystic fibrosis patient registry - contributes to the European ECFS patient registry	National	Private non-for-profit
Dutch Neuromuscular Database CRAMP: Computer Registry of All Myopathies and Polyneuropathies	National	Public
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
ECARUCA: cytogenetic and clinical database on rare chromosomal disorders	European	Public
EPCOT: European prospective cohort on thrombophilia	European	Public
ESPN/ERA-EDTA Registry: European Registry for Children on Renal Replacement	European	Public

Therapy		
EUROCAT Northern Netherlands – contributes to the international EUROCAT network	Regional	Public
European Parathyroid Tumor Registry	European	Public
FSHD registry	National	Public
Galactosemia Patient Registry - part of the Galactosemia Network	National	Public
International Dystrophic Epidermolysis Bullosa Patient Registry	Global	Public
Membranous Nephropathy Registry	Global	Public
PedNet: Haemophilia Registry	Global	Private non-for-profit
PHARMACHILD patient registry: Long-term pharmacovigilance for adverse effects in childhood arthritis, focusing on immune modulatory drugs (part of PRINTO network)	National	Public
RD5000 database: Dutch Registry for Inherited Retinal Dystrophies	National	Public
Registry NXT - Genzyme's Rare Disease Registries	Global	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)	Global	Public
The ALD Connect Patient Portal	Global	Public
The intenational Pompe registry	Global	Private for-profit
The International Collaborative Gaucher Group (ICGG) Gaucher registry	Global	Private for-profit
The international Fabry registry	Global	Private for-profit
The international MPS I registry	Global	Private for-profit

NO - NORWAY (7 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
EURADRENAL: European patient registry on autoimmune Addison's disease (sera, DNA and RNA)	European	Public
European Porphyria Registry (EPR)	European	Public
HUE-MAN patient registry on alpha mannosidosis	European	Public
Norvegian Porphyria Registry	National	Public
Norwegian cancer registry - contributes to the RARECARE project	National	Public
Norwegian cystic fibrosis patient registry - contributes to the EUROCARE 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
POLtube: Polish Registry of Inherited Tubulopathies	National	Public
Polish Registry of Patients with Neuromuscular Diseases contributes to the TREAT-	National	Public

NMD network		
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 - contributes to EURO-WABB	National	Public

PT - PORTUGAL (16 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
RORA: Açores regional cancer patient registry	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
RORENO: North regional cancer registry	Regional	Public
Pituitary tumors national registry	National	Public
Portuguese cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Portuguese Fabry patient registry - contributing to the international Fabry registry	National	Public
Portuguese registry for alpha-1 antitrypsin deficiency	National	Private non-for-profit
Portuguese registry for bronchiectasis	National	Private non-for-profit
REPORID: Portuguese registry of primary immunodeficiency diseases	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
ROG: Vila Nova de Gaia regional cancer registry	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 EUROCARE CF registry	National	Private non-for-profit

RS - SERBIA (4 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Serbian cystic fibrosis patient registry - contributes to the EUROCARE 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 (17 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
FOS : Fabry Outcome Survey	Global	Private for-profit
HOS : Hunter Outcome Survey	Global	Private for-profit
IOS : Icatibant Outcome Survey for hereditory angioedema	Global	Private for-profit
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	Public
Swedish Chronic Myeloid Leukemia Registry	National	Public
Swedish cystic fibrosis patient registry - contributes to the EUROCARE 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	Private for-profit

SI - SLOVENIA (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Slovenian cancer registry - contributes to the RARECARE project	National	Private for-profit
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	National	Private for-profit

registries in Turkey - contributes to the TREAT-NMD network		
Turkish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Turkish pediatric atypical hemolytic uremic syndrome registry	National	Private for-profit
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	Private for-profit

UK - UNITED KINGDOM (74 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
AHEAD: ADVATE Hemophilia A Outcome Database	National	Public
Batten Disease Neuronal Ceroid Lipofuscinosis (NCL) Patient Registry	National	Public
BPOLD: British Paediatric Orphan Lung Disease Registry	National	Public
CARIS: Welsh registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public
CCRN 1055: Osteonecrosis of the Jaw (ONJ) Case Registry	Global	Private for-profit
CCRN 1076: A multi-centre safety registry for malaria patients treated with EurartesimTM	National	Private for-profit
Atypical hemolytic-Uremic Syndrome (aHUS) Registry	Global	Private for-profit
CCRN 2421: A registry study to characterize genetic and pathway biomarkers in Noonan syndrome and other RASopathy patients	National	Public
CCRN 2536: 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: Patients with axial spondyloarthritis: multicountry registry of clinical characteristics, including radiographic progression, and burden of disease over 5 years in reallife setting	Global	Private for-profit
CCRN 995: A European multi-centre, multi-country, post-authorisation, observational study (registry) of patients with chronic adrenal insufficiency	European	Private for-profit
ChILD-EU Database and Observational Study for Childhood Interstitial Lung Diseases	European	Public
CRANE: patients registry with cleft lip and/or cleft palate in England and Wales	National	Public
DRN 377: Clinical Register for Transient Neonatal Diabetes	National	Public
EBV associated NK/T cell malignancies registry	National	Public
EHDN: registry of juvenile Huntington's disease	Global	Public
EHR: European Haemoglobinopathy Registry	European	Public
EIMD: European registry and network for intoxication type metabolic diseases	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 EUROCARE CF and ECFS registries	National	Public
English hereditary angioedema patient registry - part of the HAE European registry	National	Public
English phenylketonuria registry	National	Public
English registry for lymphangioleiomyomatosis	National	Public

English registry of syndromes with abnormal vertebral segmentation	National	Public
English registry of Wolf-Hirschhorn syndrome	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	European	Public
EURO WILSON: Creating a European Clinical Database and designing randomised controlled clinical trials for Wilson disease	European	Puublic
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
EURO-WABB: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome, Bardet-Biedl Syndrome and Other Rare Diabetes Syndromes	European	Public
FOCR: Familial Ovarian Cancer Register	National	Public
Family History of Bowel Cancer Registry	National	Public
Global FKRP registry (Global Fukutin-Related Protein defects registry) - Part of TREAT-NMD Alliance	National	Public
Great Ormond Street Hospital Congenital Melanocytic Naevus Registry	National	Public
I-DSD: International Disorders of Sex Development registry	Global	Public
International A-T Registry	Global	Public
International Niemann-Pick Disease Registry	Global	Public
LCH: English Langerhans cell histiocytosis registry	National	Public
MCRN078: European Registry of Anti-Epileptic Drug Use in Patients with Lennox-Gastaut Syndrome	European	Public
Myotonic dystrophy patient registry in United Kingdom - part of the TREAT-NMD network	National	Public
NCARDRS: National Congenital Anomaly and Rare Disease Registration Service	National	Public
NHD : National haemophilia database	National	Public
NHR: National Haemoglobinopathy Registry	National	Public
Registry for Patients with Niemann-Pick Type C Disease	Global	Public
ASUK: The Alström syndrome UK Clinical Research Database	National	Public
The Global aHUS Registry	Global	Private for-profit
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	Global	Public
The MTM and CNM Registry - The Myotubular and Centronuclear Myopathy Patient Registry	Global	Public
The National Chronic Granulomatous Disease Registry	National	Public
TuberOus SClerosis registry to increase disease Awareness (TOSCA)	Global	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	Private non for-profit
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 fascioscapulohumeral muscular dystrophy patient registry (part of TREAT-NMD network)	National	Public
UK Huntington disease registry (collaborating with the EHDN/Euro HD Registry)	National	Public
UK mucopolysaccharidosis registry	National	Private non for-profit
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
RaDaR: UK Renal Rare Disease Registry - Part of OXAL Europe	National	Public
UK Thrombotic Thrombocytopenia Purpura (UKTTP) Registry	National	Public
UKAITPR: United Kingdom adult idiopathic thrombocytopenic purpura registry	National	Public
UKESR: United Kingdom Evans Syndrome Registry	National	Public
UKFITPR: United Kingdom familial idiopathic thrombocytopenic purpura (ITP) Registry	National	Public
United Kingdom neuromyelitis optica registry	National	Public
United Kingdom Primary Sjogren's Syndrome Registry	National	Public

European registries

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

EUROPEAN REGISTRIES (60 registries) *		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COORDINATION**	**AFFILIATION**
Central data registry of the European Competence Network on Mastocytosis (ECMN)	AT	Public
EMSA-SG: Central patient registry of the European multiple system atrophy network	AT	Public
Thromboreductin-Registry	AT	Private for-profit
ENETS: European Neuroendocrine Tumour Registry	BE	Public
EUNEFRON: registry of the European network for the study of orphan nephropathies	BE	Public
COST Action BM1105 Patient Registry - GnRH Network	CH	Public
Juvenile Inflammatory Rheumatism Cohorte (JIR-cohorte)	CH	Public
CEDATA-GPGE: Registry of Children with Inflammatory Bowel Disease in Germany and Austria	DE	Public
DÖSAK tumor registry for documentation of tumors of the face and jaws in Germany, Austria and Switzerland	DE	Public
EBAR: European Biliary Atresia Registry	DE	Public
EIMD: European registry and network for intoxication type metabolic diseases (contributes to U-IMD)	DE	Public
ERN [ERN-LUNG] – Rare Respiratory Disease Registry	DE	Public
ERN [MetabERN] - UIMD: Unified European Registry for Inherited Metabolic Disorders	DE	Public
ERN [ERKNet] - ERK-REG: ERKNet Registry for Rare Kidney Diseases	DE	Public
EU-RHAB: European Rhabdoid Tumor Registry	DE	Public
ESID: European registry of primary immunodeficiencies	DE	Public
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
MEFOPA: registry for patients with rare Mendelian forms of Parkinson's Disease	DE	Public
Nephronophthisis registry for patients in Germany, Austria and Switzerland	DE	Public
ERCUSYN: European registry on Cushing's syndrome	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
Escort-Hu: European sickle cell disease cohort- hydroxyurea	FR	Private for-profit
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
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
HAE-registry: European hereditary angioedema patient registry	IT	Public
ERN [PAEDCAN] - PARTNER: Paediatric Rare Tumours Networks – European Registry	IT	Public
EUROFEVER: European registry for autoinflammatory diseases	IT	Public
EUROWILSON: Registry and network to improve the management of Wilson Disease	IT	Public
E-HOD: European Network and Registry for Homocystinurias and Methylation Defects (contributes to U-IMD)	NL	Public
EPCOT: European prospective cohort on thrombophilia	NL	Public
ERN [Endo-ERN] – EuRRECa: European Registries for Rare Endocrine Conditions	NL	Public
ESPN/ERA-EDTA Registry: European Registry for Children on Renal Replacement Therapy	NL	Public
European Parathyroid Tumor Registry	NL	Public
EURADRENAL: European patient registry on autoimmune Addison's disease (sera, DNA and RNA)	NO	Public
European Porphyria Registry (EPR)	NO	Public
HUE-MAN patient registry on alpha mannosidosis	NO	Public
CCRN 995: A European multi-centre, multi-country, post-authorisation, observational study (registry) of patients with chronic adrenal insufficiency	UK	Private for-profit
EHR: European Haemoglobinopathy Registry	UK	Public
ERNEST - European Registry for Myeloproliferative Neoplasms towards a better understanding of Epidemiology, Survival and Treatment	UK	Private for-profit
EUMDS: European Registry for Myelodysplastic Syndromes	UK	Public
EURODSD: European disorders of sexual development registry	UK	Public
EuroMyositis registry	UK	Public
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	UK	Public
European Prader-Willi syndrome database	UK	Public
EURO-WABB: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome, Bardet-Biedl Syndrome and Other Rare Diabetes Syndromes	UK	Public

International registries

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

INTERNATIONAL REGISTRIES (58 registries) *		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COORDINATION**	**AFFILIATION**
EUROGLYCANET - International patient registry and cohort for congenital disorders of glycosylation	BE	Private for-profit
Pediatric granulomatous arthritis international registry	BE	Private for-profit
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
ARegPKD - an international registry study for autosomal recessive polycystic kidney disease	DE	Public
CERTAIN: Cooperative European Paediatric Renal Transplant Initiative registry	DE	Private non-for-profit
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	DE	Public
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	DE	Public
Enroll-Huntington's disease Registry	DE	Private non-for-profit
Fabry Disease Registry	DE	Public
FungiScope - Global Rare Fungal Infection Registry	DE	Public
iNTD (International Working Group on Neurotransmitter Related Disorders) Registry (contributes to U-IMD)	DE	Public
International pheochromocytoma and paraganglioma registry	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	Public
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
Pompe Disease Registry	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
STER: FVII deficiency treatment international registry	DE	Public
TIRCON: NBIA (Neurodegeneration with Brain Iron Accumulation) patient registry	DE	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx): Immune thrombocytopenic purpura (ITP)	FR	Private non-for-profit
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx): Lupus	FR	Private non-for-profit
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx): Myositis	FR	Private non-for-profit

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 venooclusive disease, hepatoportal sclerosis and portal vein thrombosis	FR	Public
Hunter Outcome Survey (HOS): patient registry	IE	Public
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 non-for-profit
Ring14 Clinical database	IT	Private non-for-profit
International Dystrophic Epidermolysis Bullosa Patient Registry	NL	Public
Membranous Nephropathy Registry	NL	Public
PedNet Haemophilia Registry	NL	Private non-for-profit
Registry NXT - Genzyme's Rare Disease Registries	NL	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	Public
The ALD Connect Patient Portal	NL	Public
The International Collaborative Gaucher Group (ICGG) Gaucher registry	NL	Private for-profit
The international MPS I registry	NL	Private for-profit
FOS : Fabry Outcome Survey	SE	Private for-profit
IOS : Icatibant Outcome Survey for hereditory angioedema	SE	Private for-profit
CCRN 1055: Osteonecrosis of the Jaw (ONJ) Case Registry	UK	Private for-profit
CCRN 1093: Atypical HemolyticUremic Syndrome (aHUS Registry)	UK	Private for-profit
CCRN 3119: Patients with axial spondyloarthritis: multicountry registry of clinical characteristics, including radiographic progression, and burden of disease over 5 years in reallife setting	UK	Private for-profit
EHDN: Registry of juvenile Huntington's disease	UK	Public
I-DSD: International Disorders of Sex Development registry	UK	Public
International A-T Registry	UK	Public
International Niemann-Pick Disease Registry	UK	Public
The MTM and CNM Registry - The Myotubular and Centronuclear Myopathy Patient Registry	UK	Public
TuberOus SClerosis registry to increase disease Awareness (TOSCA)	UK	Private for-profit

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

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