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 ORPHAcode which is the unique identifier in the Orphanet nomenclature. Data in this report reflects the last update made in the Orphanet database by the registry holders and may not depict changes made to the registries since.

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

ORPHA code	Disease name
567	22q11.2 deletion syndrome
325055	46,XX disorder of gonadal development
2982	46,XX disorder of sex development
98078	46,XX disorder of sex development induced by androgens excess
90776	46,XX disorder of sex development induced by fetal androgens excess
2973	46,XX disorder of sex development-anorectal anomalies syndrome
2138	46,XX ovotesticular disorder of sex development
325118	46,XY disorder of gonadal development
98085	46,XY 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
168563	46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome
13	6-pyruvoyl-tetrahydropterin synthase deficiency
180142	Absence of uterine body
48818	Aceruloplasminemia
15	Achondroplasia
49382	Achromatopsia
101963	Acquired chronic primary adrenal insufficiency
599480	Acquired hemophilia A
599485	Acquired hemophilia B
464453	Acquired methemoglobinemia
95502	Acquired pituitary hormone deficiency
163931	Acrodermatitis continua of Hallopeau
79356	Acrokeratoderma
963	Acromegaly
99892	ACTH-dependent Cushing syndrome
318	Acute erythroid leukemia
293173	Acute generalized exanthematous pustulosis
98916	Acute inflammatory demyelinating polyradiculoneuropathy
79276	Acute intermittent porphyria
79126	Acute interstitial pneumonia
513	Acute lymphoblastic leukemia
518	Acute megakaryoblastic leukemia
514	Acute monoblastic/monocytic leukemia
519	Acute myeloid leukemia
517	Acute myelomonocytic leukemia
35889	Acute opioid poisoning
520	Acute promyelocytic leukemia
309120	Acyl-CoA dehydrogenase deficiency
85138	Addison disease
45	Adenosine monophosphate deaminase deficiency
100091	Adrenal/paraganglial tumor
1501	Adrenocortical carcinoma
181412	Adrenogenital syndrome
139399	Adrenomyeloneuropathy
86875	Adult T-cell leukemia/lymphoma
829	Adult-onset Still disease
300846	Aggressive B-cell non-Hodgkin lymphoma
86873	Aggressive NK-cell leukemia
1164	Allergic bronchopulmonary aspergillosis
60	Alpha-1-antitrypsin deficiency
61	Alpha-mannosidosis
846	Alpha-thalassemia
275745	Alpha-thalassemia and related disorders
847	Alpha-thalassemia-X-linked intellectual disability syndrome
63	Alport syndrome
64	Alström syndrome
2131	Alternating hemiplegia of childhood
284	Alveolar echinococcosis
67043	Amoebic keratitis
803	Amyotrophic lateral sclerosis
228113	Anal fistula
251630	Anaplastic oligodendroglioma
754	Androgen insensitivity syndrome
72	Angelman syndrome
1069	Aniridia-absent patella syndrome
1065	Aniridia-cerebellar ataxia-intellectual disability syndrome
96346	Anorectal malformation
88632	Anterior segment developmental anomaly
375	Anti-glomerular basement membrane disease
2299	Aortic arch interruption
87	Apert syndrome
182040	Aplastic anemia
320	Apparent mineralocorticoid excess
90	Argininemia
23	Argininosuccinic aciduria

35708	Aromatic L-amino acid decarboxylase deficiency
247	Arrhythmogenic right ventricular cardiomyopathy
1163	Aspergillosis
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
220460	Attenuated familial adenomatous polyposis
85447	ATTRV30M amyloidosis
2134	Atypical hemolytic uremic syndrome
79669	Autoimmune bullous skin disease
98375	Autoimmune hemolytic anemia
2137	Autoimmune hepatitis
3261	Autoimmune lymphoproliferative syndrome
436159	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency
3453	Autoimmune polyendocrinopathy type 1
3143	Autoimmune polyendocrinopathy type 2
747	Autoimmune pulmonary alveolar proteinosis
71203	Autoimmune thrombocytopenia
93665	Autoinflammatory syndrome
99	Autosomal dominant cerebellar ataxia
94145	Autosomal dominant cerebellar ataxia type I
94148	Autosomal dominant cerebellar ataxia type III
98352	Autosomal dominant disease with diffuse palmoplantar keratoderma as a major feature
2314	Autosomal dominant hyper-IgE syndrome
266	Autosomal dominant limb-girdle muscular dystrophy type 1A
730	Autosomal dominant polycystic kidney disease
34149	Autosomal dominant tubulointerstitial kidney disease
79278	Autosomal erythropoietic protoporphyria
281217	Autosomal ichthyosis syndrome
281097	Autosomal recessive congenital ichthyosis
101150	Autosomal recessive dopa-responsive dystonia
300547	Autosomal recessive infantile hypercalcemia
731	Autosomal recessive polycystic kidney disease
2512	Autosomal recessive primary microcephaly
300345	Autosomal systemic lupus erythematosus
98152	Autosomal uniparental disomy
782	Axenfeld-Rieger syndrome
67038	B-cell chronic lymphocytic leukemia
86852	B-cell prolymphocytic leukemia
110	Bardet-Biedl syndrome
111	Barth syndrome
112	Bartter syndrome
98895	Becker muscular dystrophy
116	Beckwith-Wiedemann syndrome
117	Behçet disease
848	Beta-thalassemia
275749	Beta-thalassemia and related diseases
231214	Beta-thalassemia major
610	Bethlem myopathy
415286	Bilirubin encephalopathy
179	Birdshot chorioretinopathy
122	Birt-Hogg-Dubé syndrome
124	Blackfan-Diamond anemia
90340	Blau syndrome
16	Blue cone monochromatism
223727	Bone sarcoma
168598	Brain demyelination due to methionine adenosyltransferase deficiency
352649	Brain dopamine-serotonin vesicular transport disease
97287	Bronchial neuroendocrine tumor
1303	Bronchiolitis obliterans with obstructive pulmonary disease
70589	Bronchopulmonary dysplasia
131	Budd-Chiari syndrome
543	Burkitt lymphoma
329931	C3 glomerulonephritis
140	Campomelic dysplasia
137667	Capillary malformation-arteriovenous malformation
147	Carbamoyl-phosphate synthetase 1 deficiency
56044	Carcinoma of gallbladder and extrahepatic biliary tract
1359	Carney complex
97286	Carney-Stratakis syndrome
3027	Caudal regression sequence
86870	CD4+/CD56+ hematodermic neoplasm
597	Central core disease
595	Centronuclear myopathy
136	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy
79158	Cerebral organic aciduria
166	Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy
1422	Chondrodysplasia-disorder of sex development syndrome
55880	Chondrosarcoma
251896	Choroid plexus tumor
180	Choroideremia
182	Chromomycosis
168940	Chronic eosinophilic leukemia
2932	Chronic inflammatory demyelinating polyneuropathy
521	Chronic myeloid leukemia
86830	Chronic myeloproliferative disease, unclassifiable
86829	Chronic neutrophilic leukemia
324964	Chronic nonbacterial osteomyelitis/Chronic

	recurrent multifocal osteomyelitis
101959	Chronic primary adrenal insufficiency
247525	Citrullinemia type I
466026	Class I glucose-6-phosphate dehydrogenase deficiency
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form
71277	Classic glucose transporter type 1 deficiency syndrome
391	Classic Hodgkin lymphoma
394	Classic homocystinuria
79163	Classic organic aciduria
1991	Cleft lip with or without cleft palate
2014	Cleft palate
31824	Colchicine poisoning
468672	Colobomatous macrophthalmia-microcornea syndrome
1198	Colonic atresia
35909	Combined deficiency of factor V and factor VIII
445018	Combined immunodeficiency due to LRBA deficiency
1572	Common variable immunodeficiency
1329	Complete atrioventricular septal defect
1872	Cone rod dystrophy
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral
418	Congenital adrenal hyperplasia
48	Congenital bilateral absence of vas deferens
2140	Congenital diaphragmatic hernia
137	Congenital disorder of glycosylation
98873	Congenital dyserythropoietic anemia type II
325	Congenital factor II deficiency
326	Congenital factor V deficiency
327	Congenital factor VII deficiency
328	Congenital factor X deficiency
329	Congenital factor XI deficiency
331	Congenital factor XIII deficiency
2020	Congenital fiber-type disproportion myopathy
335	Congenital fibrinogen deficiency
174590	Congenital hypogonadotropic hypogonadism
442	Congenital hypothyroidism
657	Congenital isolated hyperinsulinism
1928	Congenital lobar emphysema
69063	Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization
97242	Congenital muscular dystrophy
370953	Congenital muscular dystrophy due to dystroglycanopathy
75840	Congenital muscular dystrophy, Ullrich type
98904	Congenital myopathy with excess of thin filaments
206973	Congenital myotonia
480531	Congenital portosystemic shunt
2444	Congenital pulmonary airway malformation
3161	Congenital pulmonary sequestration
3090	Congenital pulmonary venous return anomaly
2040	Congenital respiratory-biliary fistula
3091	Congenital systemic veins anomaly
93583	Congenital thrombotic thrombocytopenic purpura
858	Congenital toxoplasmosis
169826	Congenital vitamin K-dependent coagulation factors deficiency
860	Congenitally uncorrected transposition of the great arteries
2445	Conotruncal heart malformations
293830	Constitutional dyserythropoietic anemia
319651	Constitutional megaloblastic anemia with severe neurologic disease
252202	Constitutional mismatch repair deficiency syndrome
101987	Constitutional neutropenia
54251	Corticosteroid-sensitive aseptic abscess syndrome
98038	Cranial malformation
208650	Cryopyrin-associated periodic syndrome
553	Cushing syndrome
79140	Cutaneous neuroendocrine carcinoma
400	Cystic echinococcosis
586	Cystic fibrosis
213	Cystinosis
214	Cystinuria
397587	Deep dermatophytosis
1652	Dent disease
220	Denys-Drash syndrome
221	Dermatomyositis
98909	Desminopathy
873	Desmoid tumor
83469	Desmoplastic small round cell tumor
98553	Developmental defect of the eye
1666	Dextrocardia
98043	Diaphragmatic or abdominal wall malformation
146	Differentiated thyroid carcinoma
90060	Diffuse alveolar hemorrhage
497188	Diffuse intrinsic pontine glioma
544	Diffuse large B-cell lymphoma
31828	Digitalis poisoning
226	Dihydropteridine reductase deficiency
309830	Disorder of catecholamine synthesis
79175	Disorder of gamma-aminobutyric acid metabolism
309819	Disorder of pterin metabolism
90771	Disorder of sex development
79167	Disorder of urea cycle metabolism and ammonia detoxification
1642	Distal monosomy 9p

18	Distal renal tubular acidosis
404546	DITRA
34516	DNAJB6-related limb-girdle muscular dystrophy D1
244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis
255	Dopa-responsive dystonia
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency
230	Dopamine beta-hydroxylase deficiency
870	Down syndrome
33069	Dravet syndrome
139402	Drug rash with eosinophilia and systemic symptoms
262	Duchenne and Becker muscular dystrophy
98896	Duchenne muscular dystrophy
1203	Duodenal atresia
268	Dysferlin-related limb-girdle muscular dystrophy R2
303	Dystrophic epidermolysis bullosa
256	Early-onset generalized limb-onset dystonia
91492	Early-onset non-syndromic cataract
1880	Ebstein malformation of the tricuspid valve
158668	Ectodermal dysplasia-skin fragility syndrome
98249	Ehlers-Danlos syndrome
289	Ellis Van Creveld syndrome
261	Emery-Dreifuss muscular dystrophy
85438	Enthesitis-related juvenile idiopathic arthritis
301	Ependymal tumor
302	Epidermodysplasia verruciformis
46487	Epidermolysis bullosa acquisita
304	Epidermolysis bullosa simplex
257	Epidermolysis bullosa simplex with muscular dystrophy
79355	Erythrokeratoderma
1199	Esophageal atresia
3318	Essential thrombocythemia
31826	Ethylene glycol poisoning
1959	Evans syndrome
883	Extragonadal teratoma
324	Fabry disease
141229	Facial cleft
269	Facioscapulohumeral dystrophy
733	Familial adenomatous polyposis
334	Familial atrial fibrillation
404560	Familial atypical multiple mole melanoma syndrome
313846	Familial cutaneous telangiectasia and oropharyngeal cancer predisposition syndrome
93587	Familial cystic renal disease
540	Familial hemophagocytic lymphohistiocytosis
235936	Familial hyperaldosteronism
93372	Familial hypocalciuric hypercalcemia type 1
342	Familial Mediterranean fever
99361	Familial medullary thyroid carcinoma
618	Familial melanoma
569	Familial or sporadic hemiplegic migraine
213517	Familial ovarian cancer
1333	Familial pancreatic carcinoma
319487	Familial papillary or follicular thyroid carcinoma
31043	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement
1331	Familial prostate cancer
231108	Familial rhabdoid tumor
84	Fanconi anemia
101039	Female restricted epilepsy with intellectual disability
1987	Femoral agenesis/hypoplasia
34515	FKRP-related limb-girdle muscular dystrophy R9
1866	Focal, segmental or multifocal dystonia
86902	Follicular dendritic cell sarcoma
545	Follicular lymphoma
51208	Formiminoglutamic aciduria
908	Fragile X syndrome
347	Frasier syndrome
95	Friedreich ataxia
98535	Frontotemporal degeneration with dementia
227796	Fundus albipunctatus
519930	Fungal keratitis
228119	Fusariosis
352	Galactosemia
314022	Gastric adenocarcinoma and proximal polyposis of the stomach
36273	Gastric linitis plastica
100092	Gastroenteropancreatic neuroendocrine neoplasm
44890	Gastrointestinal stromal tumor
2368	Gastroschisis
355	Gaucher disease
77259	Gaucher disease type 1
77260	Gaucher disease type 2
77261	Gaucher disease type 3
36387	Generalized epilepsy with febrile seizures-plus
247353	Generalized pustular psoriasis
101960	Genetic chronic primary adrenal insufficiency
183497	Genetic neuromuscular disease
183573	Genetic overgrowth/obesity syndrome
435554	Genetic precocious puberty
34526	Genetic primary hypomagnesemia
183592	Genetic renal tubular disease
656	Genetic steroid-resistant nephrotic syndrome
358	Gitelman syndrome
849	Glanzmann thrombasthenia
360	Glioblastoma
25	Glutaryl-CoA dehydrogenase deficiency

407	Glycine encephalopathy
365	Glycogen storage disease due to acid maltase deficiency
367	Glycogen storage disease due to glycogen branching enzyme deficiency
366	Glycogen storage disease due to glycogen debranching enzyme deficiency
284426	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency
137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency
99849	Glycogen storage disease due to muscle beta-enolase deficiency
368	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
371	Glycogen storage disease due to muscle phosphofructokinase deficiency
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency
97234	Glycogen storage disease due to phosphoglycerate mutase deficiency
370	Glycogen storage disease due to phosphorylase kinase deficiency
602	GNE myopathy
377	Gorlin syndrome
53693	GRACILE syndrome
2102	GTP cyclohydrolase I deficiency
168569	H syndrome
99803	Haddad syndrome
73229	HANAC syndrome
319247	Hantavirus pulmonary syndrome
163596	Hb Bart's hydrops fetalis
2130	Hemimelia
139491	Hemochromatosis type 4
2132	Hemoglobin C disease
90039	Hemoglobin D disease
2133	Hemoglobin E disease
330041	Hemoglobin M disease
68364	Hemoglobinopathy
280615	Hemoglobinopathy Toms River
766	Hemolytic anemia due to red cell pyruvate kinase deficiency
158032	Hemophagocytic syndrome
448	Hemophilia
98878	Hemophilia A
98879	Hemophilia B
340	Hemorrhagic fever-renal syndrome
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
324381	Hereditary inclusion body myopathy type 4
523	Hereditary leiomyomatosis and renal cell cancer
621	Hereditary methemoglobinemia
443909	Hereditary nonpolyposis colon cancer
79357	Hereditary palmoplantar keratoderma
29072	Hereditary pheochromocytoma-paraganglioma
264675	Hereditary pulmonary alveolar proteinosis
94088	Hereditary renal hypouricemia
213524	Hereditary site-specific ovarian cancer syndrome
98365	Hereditary stomatocytosis
84085	Hinman syndrome
388	Hirschsprung disease
86896	Histiocytic sarcoma
390	Histoplasmosis
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
168956	Hypereosinophilic syndrome
343	Hyperimmunoglobuline mia D with periodic fever
289891	Hypermethioninemia due to glycine N-methyltransferase deficiency
289290	Hypermethioninemia encephalopathy due to adenosine kinase deficiency
415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
99880	Hyperparathyroidism-jaw tumor syndrome
238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency
31740	Hypersensitivity pneumonitis
437	Hypophosphatemic rickets
2248	Hypoplastic left heart syndrome
79354	Ichthyosis
60033	Idiopathic bronchiectasis
182101	Idiopathic eosinophilic pneumonia
33208	Idiopathic hypersomnia
98482	Idiopathic inflammatory myopathy
98300	Idiopathic interstitial pneumonia
357502	Idiopathic nephrotic syndrome
2032	Idiopathic pulmonary fibrosis
99931	Idiopathic pulmonary hemosiderosis

284264	IgG4-related disease
49041	IgG4-related retroperitoneal fibrosis
3002	Immune thrombocytopenia
98290	Immunodeficiency-associated lymphoproliferative disease
761	Immunoglobulin A vasculitis
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
611	Inclusion body myositis
178557	Indolent primary cutaneous B-cell lymphoma
238455	Infantile dystonia-parkinsonism
772	Infantile Refsum disease
1186	Infantile-onset spinocerebellar ataxia
90003	Inflammatory pseudotumor of the liver
140162	Inherited cancer-predisposing syndrome
319462	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations
79361	Inherited epidermolysis bullosa
252190	Inherited nervous system cancer-predisposing syndrome
319328	Inherited renal cancer-predisposing syndrome
71862	Inherited retinal disorder
1478	Interatrial communication
86900	Interdigitating dendritic cell sarcoma
37202	Interstitial cystitis
182095	Interstitial lung disease
264735	Interstitial lung disease specific to adulthood
306504	Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome
104010	Intestinal polyposis syndrome
1048	Isolated anencephaly/exencephaly
250923	Isolated aniridia
30391	Isolated biliary atresia
2542	Isolated microphthalmia-anophthalmia-coloboma
718	Isolated Pierre Robin syndrome
2924	Isolated polycystic liver disease
823	Isolated spina bifida
2440	Isolated split hand-split foot malformation
33	Isovaleric acidemia
474	Jeune syndrome
220497	Joubert syndrome with renal defect
305	Junctional epidermolysis bullosa
92	Juvenile idiopathic arthritis
86834	Juvenile myelomonocytic leukemia
2322	Kabuki syndrome
2331	Kawasaki disease
2908	Kindler epidermolysis bullosa
99978	Klatskin tumor
33543	Kleine-Levin syndrome
99749	Kostmann syndrome
59135	Laing early-onset distal myopathy
258	Laminin subunit alpha 2-related congenital muscular dystrophy
98301	Laminopathy
389	Langerhans cell histiocytosis
86897	Langerhans cell sarcoma
626	Large congenital melanocytic nevus
633	Laron syndrome
1202	Larynx atresia
65	Leber congenital amaurosis
54260	Left ventricular noncompaction
549	Legionnaires disease
137605	Legius syndrome
2382	Lennox-Gastaut syndrome
509	Leptospirosis
158687	Lethal acantholytic erosive disorder
68356	Leukodystrophy
524	Li-Fraumeni syndrome
263	Limb-girdle muscular dystrophy
163892	Limbic encephalitis
69663	Low phospholipid-associated cholelithiasis
137631	Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome
538	Lymphangioleiomyomatosis
223735	Lymphoma
144	Lynch syndrome
309337	Lysosomal glycogen storage disease
592	Macrophagic myofasciitis
163634	Maffucci syndrome
679	Malignant atrophic papulosis
423	Malignant hyperthermia of anesthesia
168999	Malignant melanoma of the mucosa
293181	Malignant migrating focal seizures of infancy
398043	Malignant tumor of penis
52417	MALT lymphoma
52416	Mantle cell lymphoma
558	Marfan syndrome
300912	Marginal zone lymphoma
559	Marinesco-Sjögren syndrome
98292	Mastocytosis
3097	Meacham syndrome
2006	Median cleft lip/mandibule
1332	Medullary thyroid carcinoma
616	Medulloblastoma
97338	Melanoma of soft tissue
31825	Methanol poisoning
2169	Methylcobalamin deficiency type 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
1083	Microlissencephaly

98555	Microphthalmia-anophthalmia-coloboma
568	Microphthalmia, Lenz type
83463	Microtia
169808	Mild hemophilia A
169799	Mild hemophilia B
494433	MIRAGE syndrome
68380	Mitochondrial disease
217613	Mitochondrial disease with dilated cardiomyopathy
169805	Moderate hemophilia A
169796	Moderate hemophilia B
552	MODY
3057	Monoamine oxidase A deficiency
83467	Morvan syndrome
98503	Motor neuron disease
575	Muckle-Wells syndrome
79213	Mucopolysaccharidosis
579	Mucopolysaccharidosis type 1
580	Mucopolysaccharidosis type 2
247768	Müllerian aplasia and hyperandrogenism
641	Multifocal motor neuropathy
68341	Multiple congenital anomalies/dysmorphic syndrome
652	Multiple endocrine neoplasia type 1
653	Multiple endocrine neoplasia type 2
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
589	Myasthenia gravis
2583	Mycetoma
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
391673	Necrotizing enterocolitis
607	Nemaline myopathy
654	Nephroblastoma
223	Nephrogenic diabetes insipidus
137617	Nephrogenic systemic fibrosis
655	Nephronophthisis
3388	Neural tube defect
635	Neuroblastoma
2481	Neurocutaneous melanocytosis
385	Neurodegeneration with brain iron accumulation
306719	Neurodegenerative disease with chorea
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency
877	Neuroendocrine neoplasm
636	Neurofibromatosis type 1
637	Neurofibromatosis type 2
2678	Neurofibromatosis type 6
35705	Neurometabolic disorder due to serine deficiency
68381	Neuromuscular disease
71211	Neuromyelitis optica spectrum disorder
216	Neuronal ceroid lipofuscinosis
77292	Niemann-Pick disease type A
77293	Niemann-Pick disease type B
646	Niemann-Pick disease type C
647	Nijmegen breakage syndrome
240760	Nijmegen breakage syndrome-like disorder
467	Non-acquired combined pituitary hormone deficiency
631	Non-acquired isolated growth hormone deficiency
90695	Non-acquired panhypopituitarism
94080	Non-functioning paraganglioma
547	Non-Hodgkin lymphoma
157987	Non-Langerhans cell histiocytosis
91364	Non-specific interstitial pneumonia
557	Non-syndromic anorectal malformation
498467	Non-syndromic postaxial polydactyly
498464	Non-syndromic preaxial polydactyly
182121	Non-syndromic urogenital tract malformation of male
182124	Non-syndromic urogenital tract malformation of male and female
648	Noonan syndrome
98733	Noonan syndrome and Noonan-related syndrome
3032	NPHP3-related Meckel-like syndrome
2704	Ochoa syndrome
1125	Ocular motor apraxia, Cogan type
157962	Oculoauricular syndrome, Schorderet type
534	Oculocerebrorenal syndrome of Lowe
296	Ollier disease
660	Omphalocele
661	Ondine syndrome
1183	Opsoclonus-myoclonus syndrome
2086	Optic pathway glioma
289899	Organic aciduria
664	Ornithine transcarbamylase deficiency
139039	Orofacial clefting syndrome
666	Osteogenesis imperfecta
399293	Osteonecrosis of the jaw
2781	Osteopetrosis and related disorders
668	Osteosarcoma
213500	Ovarian cancer
93460	Overgrowth syndrome
85112	Palmoplantar keratoderma-XX sex reversal-predisposition

	to squamous cell carcinoma syndrome
677	Pancreatoblastoma
73260	Paracoccidioidomycosis
31827	Paraquat poisoning
143	Parathyroid carcinoma
447	Paroxysmal nocturnal hemoglobinuria
1330	Partial atrioventricular septal defect
93126	Pauci-immune glomerulonephritis
602659	Pediatric cancer
33402	Pediatric hepatocellular carcinoma
93552	Pediatric systemic lupus erythematosus
49	Penile agenesis
206976	Periodic paralysis
2855	Perrault syndrome
709	Peters plus syndrome
2869	Peutz-Jeghers syndrome
42642	PFAPA syndrome
716	Phenylketonuria
99408	Pituitary adenoma
50251	Pleural mesothelioma
64742	Pleuropulmonary blastoma
723	Pneumocystosis
2911	Poland syndrome
729	Polycythemia vera
79358	Porokeratosis
101330	Porphyria cutanea tarda
79473	Porphyria variegata
98253	Postinfectious encephalitis
739	Prader-Willi syndrome
398073	Prader-Willi-like syndrome
99860	Precursor B-cell acute lymphoblastic leukemia
101958	Primary adrenal insufficiency
186	Primary biliary cholangitis
364526	Primary bone dysplasia
46135	Primary central nervous system lymphoma
244	Primary ciliary dyskinesia
541	Primary cutaneous CD30+ T-cell lymphoproliferative disease
542	Primary cutaneous lymphoma
3337	Primary Fanconi renotubular syndrome
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
306498	PTEN hamartoma tumor syndrome
1578	Pterin-4 alpha-carbinolamine dehydratase deficiency
182090	Pulmonary arterial hypertension
411703	Pulmonary non-tuberculous mycobacterial infection
163927	Pustulosis palmaris et plantaris
69126	Pyogenic arthritis-pyoderma gangrenosum-acne syndrome
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
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome
165711	Rare abdominal surgical disease
98050	Rare allergic disease
102002	Rare ataxia
93419	Rare bone disease
68411	Rare bone tumor
180250	Rare breast tumor
101945	Rare bronchopulmonary tumor
97929	Rare cardiac disease
167848	Rare cardiomyopathy
68335	Rare chromosomal anomaly
98028	Rare circulatory system disease
88991	Rare congenital non-syndromic heart malformation
183651	Rare constitutional anemia
535	Rare cutaneous lupus erythematosus
68361	Rare deafness
93890	Rare developmental defect during embryogenesis
139030	Rare developmental defect with connective tissue involvement
101952	Rare diabetes mellitus
181376	Rare diabetes mellitus type 2
98059	Rare digestive tumor
280275	Rare disease
98032	Rare disease in anaesthesiology
98066	Rare disease in plastic and reconstructive surgery
98067	Rare disease in surgical orthopedic
108999	Rare disorder due to toxic effects
506207	Rare disorder potentially indicated for transplant
101953	Rare dyslipidemia
68363	Rare dystonia
97978	Rare endocrine disease

101998	Rare epilepsy
97935	Rare gastroenterologic disease
101936	Rare gastroesophageal disease
180821	Rare gastroesophageal tumor
183625	Rare genetic diabetes mellitus
98053	Rare genetic disease
391799	Rare genetic dystonia
101435	Rare genetic eye disease
158300	Rare genetic hematologic disease
71859	Rare genetic neurological disorder
307052	Rare genetic parkinsonian disorder
98056	Rare genetic renal disease
96344	Rare gynecologic or obstetric disease
97992	Rare hematologic disease
248308	Rare hemorrhagic disorder
248315	Rare hemorrhagic disorder due to a coagulation factors defect
68334	Rare hemorrhagic disorder due to a constitutional coagulation factors defect
71202	Rare hemorrhagic disorder due to a constitutional platelet anomaly
275729	Rare hemorrhagic disorder due to a constitutional thrombocytopenia
248326	Rare hemorrhagic disorder due to a platelet anomaly
101943	Rare hepatic and biliary tract tumor
57146	Rare hepatic disease
183518	Rare hereditary ataxia
220489	Rare hereditary hemochromatosis
217454	Rare hereditary thrombophilia
98004	Rare immune disease
68367	Rare inborn errors of metabolism
68416	Rare infectious disease
98047	Rare infertility
104012	Rare inflammatory bowel disease
87277	Rare intellectual disability
117569	Rare intestinal disease
180257	Rare malignant breast tumor
68329	Rare maxillo-facial surgical disease
250908	Rare neoplastic disease
98062	Rare nervous system tumor
98006	Rare neurologic disease
98026	Rare odontologic disease
97966	Rare ophthalmic disorder
98036	Rare otorhinolaryngologic disease
98061	Rare otorhinolaryngologic tumor
101937	Rare pancreatic disease
181415	Rare primary hyperaldosteronism
101944	Rare pulmonary disease
71198	Rare pulmonary hypertension
93626	Rare renal disease
93619	Rare renal tumor
97955	Rare respiratory disease
89826	Rare skin disease
79386	Rare skin tumor or hamartoma
71209	Rare soft tissue tumor
97965	Rare surgical cardiac disease
98065	Rare surgical neurologic disease
97962	Rare surgical thoracic disease
98023	Rare systemic or rheumatologic disease
280342	Rare systemic or rheumatological disease of childhood
52662	Rare teratologic disease
248361	Rare thrombotic disorder due to a constitutional coagulation factors defect
98057	Rare tumor
104011	Rare tumor of intestine
180824	Rare tumor of pancreas
101433	Rare urogenital disease
182114	Rare urogenital tumor
180151	Rare vaginal malformation
68362	Rare vascular disease
101938	Rare vascular liver disease
211237	Rare vascular tumor
268114	RAS-associated autoimmune leukoproliferative disease
461	Recessive X-linked ichthyosis
90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients
97239	Reducing body myopathy
773	Refsum disease
1848	Renal agenesis, bilateral
217071	Renal cell carcinoma
93545	Renal or urinary tract malformation
566243	Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta
791	Retinitis pigmentosa
790	Retinoblastoma
90050	Retinopathy of prematurity
778	Rett syndrome
69077	Rhabdoid tumor
780	Rhabdomyosarcoma
213802	Rhabdomyosarcoma of the cervix uteri
213615	Rhabdomyosarcoma of the corpus uteri
85408	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis
59315	Rhombencephalosynapsis
140976	RHYNS syndrome
1440	Ring chromosome 14 syndrome
158014	Rosaï-Dorfman disease
2909	Rothmund-Thomson syndrome
88618	S-adenosylhomocysteine hydrolase deficiency
140969	Saldino-Mainzer syndrome
797	Sarcoidosis

449280	Scedosporiosis
799	Schizencephaly
93921	Schwannomatosis
801	Scleroderma
3156	Senior-Loken syndrome
139466	SERKAL syndrome
157798	Serrated polyposis syndrome
42738	Severe congenital neutropenia
169802	Severe hemophilia A
169793	Severe hemophilia B
745	Severe hereditary thrombophilia due to congenital protein C deficiency
325546	Sex chromosome disorder of sex development
3162	Sézary syndrome
90038	Shiga toxin-associated hemolytic uremic syndrome
811	Shwachman-Diamond syndrome
232	Sickle cell anemia
275752	Sickle cell disease and related diseases
1047	Sideroblastic anemia
813	Silver-Russell syndrome
816	Sjögren-Larsson syndrome
319	Skeletal Ewing sarcoma
820	Sneddon syndrome
268369	Spina bifida aperta
90058	Spinal cord injury
94147	Spinocerebellar ataxia type 7
64753	Spinocerebellar ataxia with axonal neuropathy type 2
247234	Sporadic adult-onset ataxia of unknown etiology
276621	Sporadic pheochromocytoma/secreting paraganglioma
826	Sporotrichosis
500481	Squamous cell carcinoma of salivary glands
494547	Squamous cell carcinoma of the hypopharynx
494550	Squamous cell carcinoma of the larynx
500464	Squamous cell carcinoma of the nasal cavity and paranasal sinuses
502363	Squamous cell carcinoma of the oral cavity
500478	Squamous cell carcinoma of the oropharynx
300579	Staphylococcal toxemia
827	Stargardt disease
438159	STAT3-related early-onset multisystem autoimmune disease
273	Steinert myotonic dystrophy
36426	Stevens-Johnson syndrome
95455	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum
3198	Stiff person spectrum disorder
168593	Sudden infant death-dysgenesis of the testes syndrome
98557	Syndromic aniridia
182228	Systemic autoimmune disease
188	Systemic capillary leak syndrome
536	Systemic lupus erythematosus
2467	Systemic mastocytosis
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
32960	Tumor necrosis factor receptor 1 associated periodic syndrome
182130	Tumor of endocrine glands
68347	Tumor of hematopoietic and lymphoid tissues
363472	Tumor of testis and paratestis
881	Turner syndrome
88950	UMOD-related autosomal dominant tubulointerstitial kidney disease
117594	Unclassified rare disease
99139	Unstable hemoglobin disease
83001	Urogenital tract malformation
886	Usher syndrome
180062	Uterovaginal malformation
39044	Uveal melanoma
887	VACTERL/VATER association
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
166078	Von Willebrand disease type 1
166081	Von Willebrand disease type 2
166096	Von Willebrand disease type 3
206492	Vulvovaginal rhabdomyosarcoma
893	WAGR syndrome
899	Walker-Warburg syndrome
904	Williams syndrome
905	Wilson disease

1667	Wolcott-Rallison syndrome
3463	Wolfram syndrome
43	X-linked adrenoleukodystrophy
452	X-linked lissencephaly with abnormal genitalia
178461	X-linked myopathy with postural muscle atrophy
456328	X-linked myotubular myopathy-abnormal genitalia syndrome
431272	X-linked scapuloperoneal muscular dystrophy
910	Xeroderma pigmentosum
876	Yolk sac tumor
3471	Young syndrome
73263	Zygomycosis

Summary

1- *Distribution of registries by country*

The regional, national, European, and international categories below correspond to the geographical coverage of the registries, i.e. the geographical distribution of patients included in a registry.

COUNTRY	REGIONAL	NATIONAL	EUROPEAN	INTERNATIONAL**	TOTAL
Austria	7	26	4	2	39
Belgium	4	16	4		24
Bulgaria		11			11
Croatia		2			2
Cyprus		3			3
Czech Republic		10			10
Denmark	1	5			6
Estonia		2	1		3
Finland		7			7
France	21	101	18	6	146
Germany	7	111	24	23	165
Greece		4			4
Hungary		5			5
Iceland*		2			2
Ireland	3	14	1		18
Israel*		2			2
Italy	11	70	6	8	95
Latvia		1			1
Lithuania		1			1
Luxembourg		1			1
Malta		2			2
Netherlands (the)	1	15	13	12	41
Norway*		4	4		8
Poland	3	8			11
Portugal	5	11			16
Republic of North Macedonia*		1			1
Romania		2			2
Serbia*		4			4
Slovakia		2			2
Slovenia		2			2
Spain	12	39	5	1	57
Sweden		15	2	4	21
Switzerland*	2	18	1	7	28
Turkey*		5			5
Ukraine*		1			1
United Kingdom of Great Britain and Northern Ireland (the)	1	38	14	13	66
TOTAL	**78**	**561**	**97**	**76**	**812**

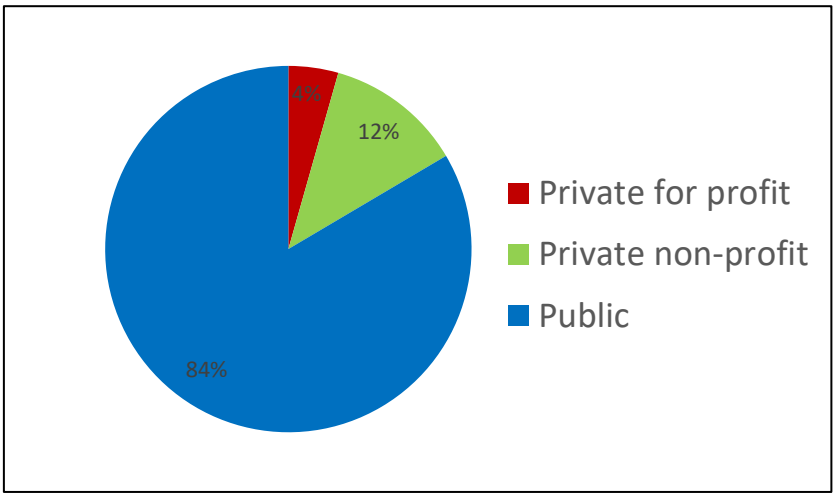
*Non-European countries participating in the Orphanet network.
**Beyond Europe.

2- *Distribution of registries by coverage*

COVERAGE	NUMBER OF REGISTRIES*
European	97
International**	76
National	561
Regional	78
TOTAL	**812**

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

3- *Distribution of registries by affiliation*



Regional and national registries

The following table lists regional and national RD-related registries, including national registries part of or participating in larger European or international registries. For the list of European and international registries coordinated in a particular country, please see tables "European registries" and "International registries" below.

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

REGIONAL/NATIONAL REGISTRIES (639 registries)

AUSTRIA (33 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
ASCIS - The Austrian Spinal Cord Injury Study	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	Public
Austrian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Austrian GIST registry	National	Public
Austrian Hemophilia Registry - Registry for patients with inborn coagulation defects	National	Public
Austrian myeloma registry	National	Public
Austrian registry for inborn errors of metabolism	National	Public
Austrian Registry on Acquired Hemophilia	National	Public
Austrian severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
Bone and soft tissue tumor registry Graz	Regional	Public
Ceprotin Treatment Registry	National	Public
Cleft registry of the Austrian Cleft Palate Craniofacial Association	National	Public
Disorders of Cornification Innsbruck	Regional	Public
Innsbruck registry for Adamantiades-Behcet disease: Retrospective and prospective data collection	Regional	Public
MDS: Austrian myelodysplastic syndromes patient registry - contributes to the European Myelodysplastic Syndromes (MDS) Registry	National	Public
NF-10 - Prospective collection of potentially prognostically relevant data in patients with indolent non-follicular B-cell lymphoma	National	Public
Non-Interventional, web-based Registry for Histiocytic Disorders	National	Public
Registry for Philadelphia chromosome-positive acute lymphoblastic leukemia in childhood and adolescence	National	Public

Registry for relapsing acute lymphoblastic leukemia in childhood and adolescence	National	Public
Registry of the NHL-BFM study group for all subtypes of Non-Hodgkin lymphoma in children and adolescents	National	Public
Registry of the University Clinic of Oral and Maxillofacial Surgery Salzburg	Regional	Public
Severe Immune Cytopenia Registry	Regional	Public
Skin cancer syndrome registry Graz	Regional	Public
Styrian registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Upshaw-Schulman-Syndrome registry	National	Public

BELGIUM (20 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Antwerpen registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Belgian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Belgian contribution to the international rare bleeding disorders registry (RBDD)	National	Public
Belgian Cystic Fibrosis patient Registry (BCFR) - contributes to the EUROCARE CF and the ECFS registries	National	Public
Belgian familial adenomatous polyposis registry	National	Public
Belgian Lynch syndrome registry	National	Public
Belgian Neuromuscular Diseases Registry (BNMDR)	National	Public
Belgian registry of primary immunodeficiencies - contributes to the ESID European registry	National	Public
Belgian severe chronic neutropenia patient registry - contributes to the Severe Chronic Neutropenia International Registry (SCNIR)	National	Public
Belgian sickle cell anemia registry	National	Public
Central Registry of Rare Diseases (CRRD)	National	Public
ERN [Endo-ERN] - EuRRECa: European Registries for Rare Endocrine Conditions - Belgian contribution (Ghent university hospital)	National	Public
ERN [Endo-ERN] - EuRRECa: European Registries for Rare Endocrine Conditions - Belgian contribution (HUDERF)	National	Public
Haemoglobinopathies - database	National	Public
Hainaut and Namur registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
I-DSD: International Disorders of Sex Development registry - BE - Flanders contribution (Ghent university hospital)	Regional	Public
I-DSD: International Disorders of Sex Development registry - BE - HUDERF	Regional	Public
Registry for Hirschsprung Disease of the BELAPS (Belgian Association of Pediatric Surgery)	National	Private for profit
The Belgian Systemic Sclerosis Cohort - BSSC	National	Public
The PRECISE LAAO Registry: Preprocedural Assessment of Anatomy and Device Sizing With CT and 3D Simulation in Left Atrial Appendage Occlusion	National	Private for profit

BULGARIA (11 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Bulgarian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public

Bulgarian registry of patients with hereditary angioedema	National	Public
Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Bulgaria - part of the TREAT-NMD network	National	Public
National registry of adult patients with chronic myeloid leukemia - BG	National	Private non-profit
National registry of patients with Gaucher disease - BG	National	Private non-profit
National registry of patients with mucopolysaccharidosis type II (MPS2) - BG	National	Private non-profit
National registry of patients with phenylketonuria - BG	National	Private non-profit
National registry of patients with primary immunodeficiencies (PID) - BG	National	Private non-profit
National registry of patients with thalassaemia major - BG	National	Private non-profit
National registry of patients with Wilson disease - BG	National	Private non-profit
The Bulgarian genetic registry of monogenic disorders	National	Public

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

CYPRUS (3 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Cyprian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
NMDcy: Cypriot Neuromuscular Diseases - contributes to TREAT-NMD	National	Public
Registry for Haemoglobinopathies	National	Public

CZECH REPUBLIC (10 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Autoimmune neuromuscular disease	National	Public
Czech cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Czech national Diamond-Blackfan Anemia Registry	National	Public
Czech National Programme for Haemophilia	National	Public
Czech National Rare Renal cancers	National	Public
Czech severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
Duchenne and Becker muscular dystrophy patient registry in the Czech Republic and Slovakia - contributes to the TREAT-NMD network	National	Public
National Register of Congenital Malformations	National	Public
Registry of Myasthenia Gravis	National	Public
Spinal muscular atrophy patient registry in the Czech Republic - part of the TREAT-NMD network	National	Public

DENMARK (6 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
Danish registry for neuromuscular diseases	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

ESTONIA (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Estonian cancer registry - contributes to the RARECARENet project	National	Public
Estonian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public

FINLAND (7 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Finnish cancer registry - contributes to the RARECARE project	National	Public
Finnish IPF registry	National	Public
Finnish patient registry on Fabry disease - contributing to the international Fabry registry and Shire registry	National	Public
Finnish TREAT-NMD Patient Registry	National	Public
Register of Congenital Malformations	National	Public
The Finnish Hematology Registry and Clinical Biobank (FHRB Biobank)	National	Public
The Finnish Register of Visual Impairment, Näkövammarekisteri	National	Public

FRANCE (122 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
Breast and other gynecological cancers registry of Côte-d'Or	Regional	Public
Bretagne registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
CEREDIH: French primary immunodeficiencies registry	National	Public
Cobra: CÖhort Angioedema Berinert	National	Public
Cohort of patients with hereditary dystrophies of retina	National	Public
Cohorte française de patients atteints de mucoviscidose et cohorte CFTR-RD	National	Private non-profit
Côte d'Or registry of hematological malignancies	Regional	Public

D[4]/Phenodent: French registry of patients affected by rare odontologic diseases	National	Public
Digestive Tumor Registry of Finistère	Regional	Public
DM Scope: Myotonic dystrophy patient registry in France - part of the TREAT-NMD network	National	Private non-profit
Duchenne and Becker muscular dystrophy patient registry in France - part of the TREAT-NMD network	National	Public
EDMUS - NOMADMUS: French cohort of Devic's neuromyelitis optica and related neurological disorders	National	Public
EPIMAD: registry of chronic inflammatory intestine diseases in North-West	Regional	Public
Establishment of children and adolescents cohort in Behcet disease in France	National	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	Private non-profit
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	Private for profit
French central hypoventilation syndrome registry - will contribute to the European CHS registry	National	Public
French certified patient registry for Langerhans cell histiocytosis	National	Public
French certified registry of glycogen storage disease type 2	National	Private for profit
French certified registry of patients affected by Gaucher disease	National	Public
French cohort creation in retinitis pigmentosa	National	Public
French cohort for auto-inflammatory diseases	National	Public
French cohort in genetic microcephalies	National	Public
French cohort in primary ciliary dyskinesia	National	Public
French cohort of acquired autoimmune haemolytic anemia	National	Public
French cohort of common variable immunodeficiency with hypogammaglobulinemia in adults (CVID)	National	Public
French cohort of focal dystonia 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 patient registry	National	Private non-profit
French cystinosis registry	National	Public
French Disease Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes - contributes to EURO-WABB	National	Public

French epidemiological registry of esophageal atresia	National	Public
French familial cardiac malformations registry	National	Public
French National FSHD Registry	National	Private non-profit
French National Lymphangioleiomyomatosis Registry (RE-LAM-CE)	National	Private non-profit
French national patients registry on rare peritoneal tumor RENAPE	National	Private non-profit
French National Registry for Rare Diseases (BNDMR)	National	Public
French observatory of biliary atresia	National	Public
French observatory of gastric linitis plastica	National	Public
French observatory of primary biliary cirrhosis	National	Public
French observatory of primitive sclerosing cholangitis	National	Public
French patient registry affected by genetic deafness in France	National	Public
French patient registry in chorioretinopathy, birdshot type	National	Public
French prospective cohort follow-up of children under the age of 18 with autoimmune cytopenia	National	Public
French Register of Amyotrophic Lateral Sclerosis	National	Public
French register of the SDH-related hereditary paraglioma	National	Public
French registry for macrophagic myofasciitis	National	Public
French Registry of Atypical Hemolytic Uremic Syndrome (aHUS) in Children	National	Private non-profit
French registry of child hematological malignancies	National	Public
French registry of corticosteroid-sensitive aseptic abscess	National	Public
French registry of familial and premature prostate cancers (before 50 years)	National	Public
French registry of generalized resistance to thyroid hormone	National	Public
French registry of hereditary dyslipidemia in children: familial combined dyslipidemias	National	Public
French registry of Iron overload genetic rare diseases, non-related to the HFE gene	National	Public
French registry of Kabuki syndrome	National	Public
French registry of Marshall's syndrome with periodic fever	National	Public
French registry of neuromuscular diseases from reference centres	National	Public
French registry of rare genetic metabolism disorders of steroids - contributing to the international RGSDC registry	National	Public
French registry of rare hypersomnias	National	Public
French registry of rare pulmonary hypertension (HTAP)	National	Public
French registry of tetrahydrobiopterin deficiencies	National	Public
French severe chronic neutropenia certified patient registry - contributes to the SCN international registry (SCNIR)	National	Public
French sickle cell anemia registry	National	Public
French West Indies registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
FROG: FRench Observatory on Gaucher disease	National	Public
GENEPSO: French BRCA1 and 2 carrier cohort study	National	Private non-profit
Gironde registry of hematological malignancies	Regional	Public

GMF: French registry of myelodysplastic syndromes and leukemia chemo- and radio-induced	National	Private for profit
GTE: French registry of endocrine tumors	National	Public
Idiopathic pulmonary fibrosis: Cohort studies for evaluation of pronostic factors, therapeutic evaluation	National	Public
ITINERAIR-HTAP: French cohort of adult with pulmonary arterial hypertension	National	Public
ITINERAIR-pediatrie: French cohort of children with pulmonary arterial hypertension	National	Public
ITINERAIR-scleroderma: French pulmonary arterial hypertension screening cohort of patients with scleroderma	National	Public
KAWA-NET: KAWASAKI disease registry	National	Public
La Réunion Fench Island registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Laminopathies and emerinopathies patient registry	National	Public
LEA - Multicentric prospective cohort of children and adolescents malignant hemopathies	National	Public
Leukofrance database and biobank	National	Public
Marne-Ardennes thyroid cancer registry	Regional	Public
Mesothelioma cohort in Seine Saint-Denis and Val de Marne	Regional	Public
Motor Function Measure database (Neuromuscular Diseases)	National	Public
Multicenter national registry of pleural mesothelioma (qualified register)	National	Public
NaThalY: National Thalassemia Registry	National	Public
National database for the study and follow-up of paediatric rare tumors	National	Public
National registry of children solid tumors	National	Public
Paris registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
PHA1-NET: PseudoHypoAldosteronism type 1 cohort	National	Public
POLA: French patient registry of high level oligodendroglioma	National	Public
Primary central nervous system tumors registry of Gironde	Regional	Public
RaDiCo-AC-OEIL: National cohort on congenital defects of the eye : natural history, genetic determinisms and improved ocular and extra-ocular outcome prediction for better patient management	National	Public
RaDiCo-ACOSTILL: National cohort on adult and childhood onset Still disease	National	Public
RaDiCo-COBBALT: National cohort on Bardet-Bield syndrome and Alström syndrome	National	Public
RaDiCo-COLPAC: National cohort on the epidemiology, clinical and genetic heterogeneity of Low Phospholipid-Associated Cholelithiasis (LPAC) syndrome	National	Public
RaDiCo-DCP: National cohort on Primary Ciliary Dyskinesia (PCD) : Identification of specific severity criteria and phenotype-genotype correlation study	National	Public
RaDiCo-FARD: National cohort for the evaluation of individual burden in the course of rare skin diseases	National	Public
RaDiCo-MARFAN: National cohort on Marfan syndrome and apparent diseases	National	Public
RaDiCo-MPS: National cohort on Mucopolysaccharidosis in the era of specific therapeutics	National	Public
RaDiCo-PID: National cohort on Idiopathic Interstitial Pneumonia from infancy to elderly	National	Public
RaDiCo-PP: National cohort on periodic paralysis : clinical, genetic and medico-economic studies involving m-health monitoring tools	National	Public
RaDiCo-RETICO: National cohort on inherited retinal dystrophies	National	Public

RaDiCo-SED-VASC: National cohort on vascular Ehlers-Danlos syndrome	National	Public
Regional registry of thyroid cancers in Rhône-Alpes	Regional	Public
Registry and pronostic cohort of cutaneous lymphomas in Aquitaine	Regional	Public
Registry for digestive cancers in Burgundy	Regional	Public
Registry of digestive tumors in Calvados	Regional	Public
Registry of hereditary Haemochromatosis of the Languedoc Roussillon (including rare forms non-HFE hemochromatosis)	Regional	Public
Registry of the network studying thrombotic microangiopathies	National	Public
Rhône-Alpes registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
SYRENE: Rett syndrome network - French database of clinical and genetic aspects of Rett syndrome	National	Public

GERMANY (118 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
ALS registry swabia	Regional	Public
Ataxia-Telangiectasia patient registry - contributes to the ESID Database	National	Public
Bayern population based cancer registry	National	Public
Bremen cancer registry	National	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
CMMR: Central Malignant Melanoma Registry in germany	National	Public
Common cancer registry of Berlin, Brandenburg, Mecklenburg-Western Pomerania, Saxony-Anhalt and the free states Saxony and Thuringia	Regional	Public
Conn Registry: German registry of primary aldosteronism	National	Private non-profit
CONNECT-GENERATE: National registry for autoimmune encephalitis	National	Public
Core documentation of children and adolescens with rheumatic diseases	National	Private non-profit
CURE-Net : National registry for congenital uro-rectal malformations	National	Public
CUSTODES - Cushing syndrome registry: therapy and outcome in germany - contributes to the ERCUSYN - European Register on Cushing's Syndrome	National	Private non-profit
DCLLSG registry of the German CLL Study Group - Long term observation of patients with CLL, B-PLL, T-PLL, SLL, T/ NK-LGL and Richter transformation	National	Public
EARCO Registry (German contact point)	National	Public
EKRS: Saarland Cancer Registry - contributes to the RARECARE Project	Regional	Public
Epidemiological cancer registry Baden-Württemberg	Regional	Public
ESNEK - Rare paediatric neurological disease registry Germany	National	Public
Fabry Disease Registry -DE	National	Private for profit
FOrMe registry: German Focal Segmental Glomerulosclerosis and Minimal Change Disease Registry	National	Public
GAIN: Patient Registry of the German Network for the Research and Therapy Optimization of Patients with Multi-Organ Autoimmune Diseases	National	Public

GeNeMove: German database for wilson disease	National	Public
German Acanthamoeba keratitis registry	National	Public
German acromegaly registry	National	Private for profit
German adrenal tumors registry	National	Public
German alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
German central registry for Sickle cell disease	National	Public
German childhood cancer registry (Partner of ACCIS: Automated Childhood Cancer Information System, Member of IACR and ENCR)	National	Public
German CMT-patient registry germany - part of the TREAT-NMD network	National	Public
German cystic fibrosis registry - contributes to the EUROCARE CF registry	National	Private non-profit
German DM registry: Patient registry for myotonic dystrophy (DM)	National	Public
German DMD- and SMA-patient registry : Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Austria and Germany - part of the TREAT-NMD network	National	Public
German epilepsy registry	National	Public
German Fanconi anemia registry	National	Private non-profit
German FKRP-patient registry - part of the TREAT-NMD network	National	Public
German follicular lymphoma registry	National	Private non-profit
German FSHD registry: Patient registry for Facioscapulohumeral dystrophy (FSHD)	National	Public
German gastrointestinal stromal tumor registry	National	Public
German Haemophilia Registry (DHR)	National	Public
German IBM registry: Patient registry for aquired or sporadic inclusion body myositis	National	Public
German marginal zone lymphoma registry	National	Private non-profit
German mucopolysaccharidosis patient registry	National	Public
German multiple endocrine neoplasia type 1 (MEN 1) registry	National	Public
German national case collection of familial pancreatic cancer	National	Private non-profit
German PAM/MFM registry: Patient registry for hereditary protein aggregate myopathies (PAM) / myofibrillar myopathies (MFM)	National	Public
German paroxysmal nocturnal hemoglobinuria registry	National	Public
German patient registry HLH (hemophagocytic lymphohistiocytosis)	National	Public
German pituitary tumors registry	National	Public
German register for hereditary nephrolithiasis	National	Public
German registry for congenital heart defects - part of the competence network for congenital heart defects	National	Public
German registry for congenital thrombocytopenia	National	Public
German Registry for Incidental Gallbladder Carcinoma	National	Private non-profit
German registry for Langerhans Cell Histiocytosis in childhood (LCH-REG-DE 2013)	National	Public
German registry for Morbus Adamantiades-Behçet e.V.	National	Public
German registry for neonates, infants, children, adolescents, and adults with newly diagnosed and/or relapsed neuroblastic tumors (NB Registry 2016) - partner of the INRG Database	National	Public

German registry for papulosis atrophicans maligna	National	Public
German ROP (retinopathy of prematurity) Registry	National	Private non-profit
German severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
Germany patient registry for membranoproliferative glomerulonephritis	National	Public
GeVas: German Vasculitis Registry	National	Public
GMALL-registry: registry for adult patients with acute lymphoblastic leukemia or related diseases	National	Public
GOLDnet: Registry for Diffuse Parenchymal Lung Disease	National	Public
GPOH-MET Registry: Registry for children and adolescents with malignant endocrine tumour	National	Private non-profit
GPOH-registry sickle cell disease	National	Private non-profit
Hamburg cancer registry	National	Public
Hantavirus Registry - HantaReg: Gathers Knowledge on Epidemiology, Clinical Course, Prognostic Factors and Molecular Characteristics for Hantavirus Infections and Their Complications	National	Public
HepNet: German hepatocellular carcinoma (HCC) registry	National	Public
HerediCaRe - National register for the evaluation and improvement of risk-adapted prevention of hereditary breast and ovarian cancer	National	Public
Hunter Outcome Survey (HOS): patient registry - DE	National	Private for profit
INSIGHTS-IPF Investigating Significant Health Trends in Idiopathic Pulmonary Fibrosis	National	Public
International Collaborative Gaucher Group (ICGG) Gaucher Registry - DE	National	Private for profit
INVM (Isolated Noncompaction of Ventricular Myocardium) registry	National	Public
LBL Registry: Registry for children with lymphoblastic lymphoma	National	Public
Leptospirosis Registry - LeptoScope: Gathers Knowledge on Epidemiology, Clinical Course, Prognostic Factors and Molecular Characteristics for Invasive Leptospirosis Disease	National	Public
Lupus nephritis registry (established by the german paediatric nephrology association)	National	Public
Mainz registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
MAISTHRO-Registry : multicentric thrombophilia registry (MAIn-ISar-THROmbose-Register)	National	Public
MCR - Munich Cancer Registry	Regional	Public
MDS Registry: German myelodysplastic syndromes patient registry	National	Public
mitoREGISTRY: Registry for patients with mitochondrial diseases - subproject of mitoNET	National	Public
MPN Registry: German MPN Register and Biomaterial Bank for BCR-ABL1-negative myeloid neoplasias	National	Private non-profit
Mucopolysaccharidosis type 1 (MPS I) Registry - DE	National	Private for profit
Mykke : German registry for children and adolescents with myocarditis	National	Public
National Cancer-predisposing syndrome registry	National	Public
National database for echinococcosis	National	Public
National MRKH patient registry	National	Public
National nephrogenic systemic fibrosis registry	National	Public
National patient registry rare histiocytic disorders - part of the International Rare Histiocytic Disorders Registry (IRHDR)	National	Public

National registry for Blackfan-Diamond disease	National	Public
National Registry for Mycotic keratitis	National	Public
National registry for retroperitoneal fibrosis	National	Private non-profit
NeoExNET registry: patient registry for neuroendocrine tumors	National	Public
NET-Registry: German neuroendocrine gastrointestinal tumors	National	Private for profit
Neuro-Heart-Registry: Registry for patients with heart involment in neurovascular diseases	National	Public
Neuromyelitis optica patient registry	National	Public
Neutropenia biobank - contributes to the SCN international registry (SCNIR)	National	Public
NIRK: national central registry for ichthyoses and related keratinization disorders	National	Public
NKR: German registry for adrenocortical carcinoma	National	Private non-profit
NPC-2016 - A multicenter registry for nasopharyngeal cancer in children, adolescents and young adults	National	Private non-profit
NPC-2016-Registry: Multi-centre registry for nasopharyngeal carcinoma in children, adolescents and young adults	National	Private non-profit
Patient registry for primary hyperoxaluria - contributes to the OxalEurope-Network	National	Public
Patient registry for retinal degeneration PRO RETINA e.V	National	Private non-profit
Patient registry of the German Network for Systemic Scleroderma	National	Private non-profit
PID-NET: German National Registry for Primary Immunodeficiencies (PID) - contributes to the ESID registry	National	Public
Pompe Disease Registry - DE	National	Private for profit
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
Rare anaemia Registry	National	Private non-profit
Registry and biobank for rare thyroid and parathyroid carcinoma	National	Public
Registry for C3 Glomerulopathy and Immune complex-mediated MPGN	National	Public
Registry for congenital melanocytic nevi and neurocutaneous melanocytosis	National	Public
Registry for Merkel Cell Carcinoma	National	Public
Registry for Patients with WT1 Mutation Associated Diseases	National	Private non-profit
Schleswig-Holstein cancer registry	National	Public
STEP Registry: Registry for rare tumors in children and adolescents	National	Private non-profit
The FOrMe Registry : The German Focal Segmental Glomerulosclerosis and Minimal Change Disease Registry	National	Public

GREECE (4 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Greek cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Greek severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public

Hellenic Neuromuscular Disorders Registry	National	Private non-profit
National Registry for Haemoglobinopathies in Greece (NRHG)	National	Private non-profit

HUNGARY (5 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
DMD registry - Hungary - contributes to the TREAT-NMD network	National	Public
Hungarian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Hungarian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
National NF Register	National	Public
SMA registry Hungary - contributes to the TREAT-NMD network	National	Public

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	Public

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
Epidermolysis Bullosa Registry Ireland	National	Private non-profit
Hunter Outcome Survey (HOS): patient registry	National	Private for profit
Irish ENS@T Biobank and Registry - contributes to ENS@T registry	National	Public
Irish Galactosemia Registry - contributes to European Galactosemia Patient Registry	National	Public
Irish Paediatric Surveillance Registry	National	Public
Irish Registry for homocystinurias and methylation defects - contributes to E-HOD	National	Public
Irish registry of amyotrophic lateral sclerosis and motor neurone disease - contributes to Euro-MOTOR	National	Public
Irish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
National Alpha-1 Antitrypsin Deficiency Registry - contributes to the Alpha One International Registry (AIR)	National	Public
National Cleft Database - contributes to EUROCAT	National	Public
National Haemophilia Register - Ireland	National	Public
National Severe Immunodeficiency Registry - contributes to ESID registry	National	Public
South East of Ireland registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public
South of Ireland registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
The Irish Rare Kidney Disease Registry and Biobank	National	Public

ISRAEL (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Israeli cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Israelian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

ITALY (81 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
AICE: Italian registry of hemophilia centre	National	Public
Angelman Syndrome Italian Registry (RISA)	National	Private non-profit
Arrhythmogenic right ventricular cardiomyopathy/dysplasia: clinical registry and database, evaluation of therapies	National	Public
Behçet's disease registry	National	Private non-profit
Campania registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
DMD / BMD Italy Patient Registry	National	Public
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Italy - contributes to the TREAT-NMD network	National	Public
EIMD: European registry and network for intoxication type metabolic diseases	National	Public
FMF: Italian registry for familial mediterranean fever in the young	National	Public
Friedreich's ataxia Italian patient registry	National	Public
GLATIT: Glanzmann thrombasthenia Italian registry	National	Public
GLUT1 deficiency syndrome registry	National	Private non-profit
IBAHC: Italian registry for alternating hemiplegia of childhood	National	Public
INNCB MG Registry	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	National	Public
Italian genetic movement disorders registry	National	Public
Italian HLH Registry - Hemophagocytic lymphohistiocytosis	National	Public
Italian LCH Registry - Langerhans cell histiocytosis	National	Public
Italian Li-Fraumeni syndrome registry	National	Public
Italian myelodysplastic syndromes registry	National	Public
Italian Network MDS Registry (FISM_registry)	National	Private non-profit
Italian neuroblastoma registry	National	Public
Italian register multiple endocrine neoplasia	National	Public
Italian registry for MYH9-related thrombocytopenia	National	Public
Italian Registry for patients with Shwachman Diamond Syndrome	National	Private non-profit

Italian Registry of adult patients affected by familial mediterranean fever	National	Public
Italian registry of Alport syndrome	National	Private non-profit
Italian Registry of Congenital Coagulopathies	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	Public
Italian Registry of Kernicterus and hyperbilirubinemia	National	Public
Italian registry of Legionellosis	National	Public
Italian Registry of Mastocytosis	National	Public
Italian Registry of membranoproliferative glomerulonephritis	National	Private non-profit
Italian registry of muscular channelopathy	National	Public
Italian registry of myotonic dystrophies	National	Public
Italian registry of patients and families affected by Pseudoxanthoma Elasticum	National	Public
Italian registry of skeletal dysplasia	National	Public
Italian retinoblastoma registry	National	Public
Lymphomas Database	Regional	Public
MITOCON: National registry mitochondrial disease	National	Private non-profit
MODY: Italian registry of maturity onset diabetes of the young	National	Public
Multiple Osteochondromas Registry - REM	National	Private non-profit
National Registry of Congenital Malformations	Regional	Public
National Registry of Rare Diseases	National	Public
North-east Italy registry of neurofibromatosis	Regional	Public
Osteogenesis Imperfecta Registry - ROI	National	Private non-profit
Paroxysmal Nocturnal Hemoglobinuria Registry	National	Public
RAM-NET: Italian registry of patients with neuromuscular diseases - contributes to Treat-NMD european network	National	Private non-profit
Rare disease registry EXAMPLE	National	Public
Rare Diseases Registry - Veneto Region	Regional	Public
Regional Registry Rare Diseases of Puglia Region (SIMaRRP)	Regional	Public
Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes - contributing to EURO-WABB	Natonal	Public
Registry of Ehlers-Danlos syndrome - RED	National	Private non-profit
Registry of inherited bleeding disorders in Emilia Romagna region	Regional	Public
Registry of Neurofibromatosis	National	Public
Registry of Ollier disease and Maffucci syndrome - ROM	National	Private non-profit
Registry of pregnant patients affected by essential thrombocythemia	National	Public
Registry of steroid-resistant nephrotic syndrome	National	Private for profit

RESIDRAS - National Registry of Dravet Syndrome and other Syndromes correlated with genes on SCN1A and PCDH19	National	Private non-profit
Rett Syndrome Database	National	Public
RIAF: Fanconi's Anemia Italian Registry	National	Private non-profit
RIAT: Ataxia teleangiectasia Italian registry	National	Public
RIMM: Italian registry for myelofibrosis with myeloid metaplasia	National	Public
RIPID - Italian registry of diffuse infiltrative pneumopathies	National	Public
RNIC: National Registry of patients with Congenital Hypothyroidism	National	Public
SCNIR: Italian severe chronic neutropenia registry - contributes to the SCN international registry	National	Public
Sicilian Registry of Thalassemia and Hemoglobinopathies (Re.S.T.E.)	Regional	Public
Telethon-UILDM registry CMD (Congenital Muscular Dystrophy) - contributes to european Treat-NMD network	National	Private non-profit
Telethon-UILDM registry FSHD (Facioscapulohumeral dystrophy) - contributes to european Treat-NMD network	National	Private non-profit
Telethon-UILDM registry LGMD (Limb Girdle Muscular Dystrophy) - contributes to european Treat-NMD network	National	Private non-profit
The Italian multi-region thalassaemia registry	National	Private non-profit
The Italian Registry of Hereditary Angioedema	National	Private non-profit
Transthyretin amyloid neuropathy (TTR-FAP) italian registry	National	Private non-profit
Tuscan Registry of Congenital Anomalies	Regional	Public
Tuscan Registry of Rare Diseases	Regional	Public
V-RIAT: variant Ataxia telangiectasia Italian registry	National	Public

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

LITHUANIA (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Lithuanian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public

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

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

NETHERLANDS (THE) (16 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
ALS database	National	Public
CONCOR: Dutch registration of adult patients with congenital heart disease	National	Public
DDRMD - Dutch Diagnosis Registration Metabolic Diseases	National	Public
Duchenne and Becker muscular dystrophy patient registry in the Netherlands - part of the TREAT-NMD network	National	Public
Dutch alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Dutch CF Registry - contributes to the European Cystic Fibrosis Society Patient Registry (ECFSPR)	National	Private non-profit
Dutch Neuromuscular Database CRAMP: Computer Registry of All Myopathies and Polyneuropathies	National	Private non-profit
Dutch patient registry for Fabry disease	National	Public
Dutch patient registry for Gaucher disease	National	Public
Dutch patient registry for Niemann-Pick Disease Type B	National	Public
Dutch patient registry for Niemann-Pick Disease Type C	National	Public
Dutch severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
EUROCAT Northern Netherlands - contributes to the international EUROCAT network	Regional	Public
FSHD registry	National	Private non-profit
HemoNED: Dutch Hemophilia Registry	National	Public
RD5000 database: Dutch Registry for Inherited Retinal Dystrophies	National	Private non-profit

NORWAY (4 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
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

POLAND (11 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Cracow cancer registry - contributes to the RARECARE project	Regional	Public
Kielce cancer registry - contributes to the RARECARE project	Regional	Public
Mazovian Cancer Registry - contributes to the RARECARE project	Regional	Public
Polish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Polish Registry of Inherited Tubulopathies (POLtube)	National	Public
Polish Registry of Patients with Neuromuscular Diseases contributes to the TREAT-NMD network	National	Public
Polish registry of primary immunodeficiencies - contributes to the ESID European registry	National	Public

Polish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
PRCM: Polish registry of congenital malformations - contributes to the EUROCAT network	National	Public
Registry for Wolfram syndrome, Astrom syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes contributing do EURO-WABB	National	Public
The national Polish pediatric IgAN registry	National	Public

PORTUGAL (16 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Açores regional cancer patient registry (RORA)	Regional	Public
Adrenal tumor national registry	National	Public
Central regional cancer patient registry	Regional	Public
Duchenne and Becker muscular dystrophy patient registry in Portugal - contributes to the TREAT-NMD network	National	Public
North regional cancer registry (RORENO)	Regional	Public
Pituitary tumors national registry	National	Public
Portuguese cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Portuguese Fabry patient registry - contributing to the international Fabry registry	National	Private for profit
Portuguese registry for alpha-1 antitrypsin deficiency	National	Public
Portuguese registry for bronchiectasis	National	Public
Portuguese registry of primary immunodeficiency diseases (REPORID)	National	Public
Portuguese Rett syndrome registry	National	Public
Portuguese severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
RENAC - Portuguese registry of congenital anomalies (contributes to the EUROCAT network)	National	Public
Southern Portugal cancer registry - contributes to the RARECARE project	Regional	Public
Vila Nova de Gaia regional cancer registry (ROG)	Regional	Public

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

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	Public

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

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

SLOVENIA (2 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
Slovenian cancer registry - contributes to the RARECARE project	National	Public
Slovenian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public

SPAIN (51 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	COVERAGE	AFFILIATION
ADPKD registry: Spanish registry for autosomal dominant polycystic kidney disease	National	Private non-profit
aHUS/C3G: Atypical Hemolytic Uremic Syndrome (aHUS) and C3 Glomerulopathy (C3G) Database	National	Public
EARCO Registry (Spanish contact point)	National	Public
ECEMC: Registry of the Spanish Collaborative Study of Congenital Malformations	National	Public
Epidemiologic Registry of Patients Diagnosed With Acute Myeloid Leukemia - ES	National	Private non-profit
EURO-WABB: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome, Bardet-Biedl Syndrome and Other Rare Diabetes Syndromes - ES	National	Public
MYO-SPAIN Platform: Patient registry on idiopathic inflammatory myopathy in Spain	National	Private for profit
NMD-ES: Spanish Registry of Neuromuscular Diseases	National	Public
PanGen-FAM: Spanish registry of hereditary pancreatic cancer	National	Public
Population registry of rare diseases and congenital anomalies of Cantabria (Spain)	Regional	Public
Population registry of rare diseases of Balearic Islands (Spain)	Regional	Public
Population registry of rare diseases of Navarra (Spain)	Regional	Public
RACAV: Registry of congenital anomalies of the Basque Country (Spain) - contributes to the EUROCAT network	Regional	Public
Rare disease registry of Aragon (Spain)	Regional	Public
Rare Diseases Registry of the Basque Country	Regional	Public
REDAPED: Spanish registry of ataxias and degenerative spastic paraparesis	National	Public

REDIP: Spanish registry of primary immunodeficiencies - contributes to the ESID European registry	National	Public
REEG: Spanish Gaucher's disease registry	National	Public
ReeR: Spanish National Registry of Rare Diseases	National	Public
REeRIGG4: Spanish IgG4-related Disease Registry	National	Private non-profit
Registro MEN: Spanish registry of multiple endocrine neoplasia, pheochromocitomas and paragangliomas	National	Private for profit
Registry for rare diseases in Andalusia (Spain)	Regional	Public
Registry for rare diseases in Extremadura (Spain)	Regional	Public
REHAP: Spanish Registry of Pulmonary Arterial Hypertension	National	Private for profit
REHem-AR-SEHOP: Spanish Registry of Hemoglobinopathies and Rare Anemias from the Spanish Society of Paediatric Haematology and Oncology	National	Public
REHEVASC: Spanish registry for hepatic vascular diseases	National	Private non-profit
REHIPED - Spanish Registry for Pediatric Pulmonary Hypertension	National	Public
REPA: Spanish registry of alveolar proteinosis	National	Public
RERGA: Registry for rare diseases in Galicia (Spain)	Regional	Public
RETEGEP: Spanish Registry of Gastroenteropancreatic Neuroendocrine Tumors	National	Private for profit
REWBA: Spanish Registry of Wolfram, Bardet-Biedl and Alstrom syndromes	National	Public
SIER-CV: Information System on rare diseases in Valencian Community (Spain)	Regional	Public
SIERMA: Information system on rare diseases in Madrid (Spain)	Regional	Public
SIERrm: Information System on rare diseases in the Region of Murcia (Spain)	Regional	Public
SIOPE Diffuse Intrinsic Pontine Glioma (DIPG) Registry (Spanish contact point)	National	Private non-profit
Spanish Academy of Dermatology and Venereology (AEDV) / Rare Diseases Registry ISCIII: Registry of xeroderma pigmentoso	National	Private non-profit
Spanish Academy of Dermatology and Venereology (AEDV) Registry of primary cutaneous lymphoma	National	Private non-profit
Spanish alpha-1 antitrypsin deficiency registry (REDAAT) - contributes to the Alpha One International Registry (AIR)	National	Public
Spanish Overgrowth Syndrome Registry	National	Public
Spanish patient registry for spinal muscular atrophy - part of the TREAT-NMD network	National	Public
Spanish patient registry of ataxias	National	Public
Spanish patient registry of hereditary retinal dystrophy	National	Public
Spanish patient registry of myelodysplasic syndromes	National	Public
Spanish patient registry of transmissible spongiform encephalopathies	National	Public
Spanish Registry of Cushing Syndrome - contributes to ERCUSYN	National	Private for profit
Spanish registry of Duchenne muscular dystrophy - part of the TREAT-NMD network	National	Public
Spanish Registry of Patients with Fanconi Anemia	National	Public
Spanish registry of patients with McArdle disease	National	Public
Spanish registry of pulmonary Langerhans cell histiocytosis	National	Public
Spanish Rheumatology Society Lupus Registry	National	Private non-profit

Spanish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

SWEDEN (15 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
National registry on bronchopulmonary dysplasia	National	Public
SPAHR: Swedish Pulmonary Arterial Hypertension Registry	National	Public
SWEDCON: Swedish Registry of Congenital Heart Disease	National	Public
Swedish Acute Lymphoblastic Leukemia Registry	National	Public
Swedish Acute Myelogenous Leukemia Registry	National	Public
Swedish alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Swedish Childhood Cancer Registry	National	Private non-profit
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 registry on Usher syndrome	National	Public
Swedish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
SWEDROP: Swedish Registry for Retinopathy of Prematurity	National	Public

SWITZERLAND (20 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
CAKUT cohort study - Geneva	Regional	Public
EARCO Registry : European Alpha-1 Research Collaboration (Swiss contact point)	National	Public
EUROCAT VAUD Switzerland - Registry of congenital malformations of canton Vaud	Regional	Public
Paediatric registry for congenital pulmonary malformations	National	Public
SACHER: Swiss Adult Congenital HEart disease Registry	National	Private for profit
Swiss Autoimmune Hepatitis Cohort Study	National	Private non-profit
Swiss Childhood Cancer Registry	National	Public
Swiss Cleft Lip and Palate Registry	National	Public
Swiss Diabetes Registry (SwissDiab)	National	Public
Swiss Hemophilia Registry	National	Public
Swiss Pompe Registry	National	Private for profit
Swiss Primary Biliary Cholangitis Cohort Study	National	Private for profit
Swiss Primary Ciliary Dyskinesia Registry (CH-PCD)- contributing to the International PCD Registry	National	Public
Swiss Primary Sclerosing Cholangitis Cohort Study	National	Public

Swiss Rare Disease Registry	National	Public
Swiss registries for Interstitial and Orphan Lung Diseases (SIOLD Registries)	National	Public
Swiss Registry for Neuromuscular Disorders	National	Public
Swiss registry of biliary atresia	National	Public
Swiss SLE Cohort Study	National	Public
SwissNET - Registry for Neuroendocrine Tumours in Switzerland	National	Public

TURKEY (5 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Database setup for the visualisation and examination of oral ulcers in Behcet disease patients	National	Public
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Turkey - contributes to the TREAT-NMD network	National	Public
Turkish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Turkish pediatric atypical hemolytic uremic syndrome registry	National	Public
Turkish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

UKRAINE (1 registry)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Spinal muscular atrophy patient registry in Ukraine - part of the TREAT-NMD network	National	Public

UNITED KINGDOM OF GREAT BRITAIN AND NORTHERN IRELAND (THE) (39 registries)		
ENGLISH LABEL OF THE REGISTRY/COHORT	**COVERAGE**	**AFFILIATION**
Batten Disease Neuronal Ceroid Lipofuscinosis (NCL) Patient Registry	National	Public
British Isles Network of Congenital Anomaly Registers	National	Public
CRANE: Cleft Registry and Audit Network - England, Wales & Northern Ireland	National	Public
DRN 377: Clinical Register for Transient Neonatal Diabetes	National	Private non-profit
EIMD: European registry and network for intoxication type metabolic diseases	National	Public
English cystic fibrosis database	National	Private non-profit
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 registry for lymphangioleiomyomatosis	National	Public
English registry of biliary atresia	National	Public
English severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
Familial Ovarian Cancer Register (FOCR)	National	Public
Great Ormond Street Hospital Congenital Melanocytic Naevus Registry	National	Public
Juvenile Dermatomyositis Cohort Biomarker Study and Repository	National	Public

Myotonic dystrophy patient registry in United Kingdom - part of the TREAT-NMD network	National	Public
National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)	National	Public
National haemophilia database (NHD)	National	Public
NDSCR - National Down syndrome cytogenetic registry - part of BINOCAR and EUROCAT network	National	Public
NHR: National Haemoglobinopathy Registry	National	Public
North Star database	National	Public
SWCAR - South West congenital anomalies registry - part of BINOCAR and EUROCAT network	Regional	Public
The UK Paediatric ITP Registry	National	Public
The United Kingdom Facioscapulohumeral Muscular Dystrophy Patient Registry	National	Public
The United Kingdom National Registry for Myotonic Dystrophy	National	Public
The United Kingdom Thrombotic Thrombocytopenic Purpura Registry (UK TTP Registry)	National	Public
UK and Ireland Vasculitis registry (UKIVAS)	National	Public
UK facioscapulohumeral muscular dystrophy patient registry (part of the TREAT-NMD network)	National	Public
UK mucopolysaccharidosis registry	National	Public
UK Myotonic dystrophy type I patient registry (part of the TREAT-NMD network)	National	Public
UK national Acromegaly patient register	National	Public
UK Paediatric ITP (Immune Thrombocytopenic Purpura) Registry	National	Public
UK Registry for Central Hypoventilation Syndrome (CHS)	National	Public
UK Renal Rare Disease Registry (RaDaR) - Part of OXAL Europe	National	Public
UK Thrombotic Thrombocytopenia Purpura (UKTTP) Registry	National	Public
UKAITPR: UK Adult ITP Registry	National	Public
UKESR: United Kingdom Evans Syndrome Registry	National	Private for profit
UKFITPR: United Kingdom familial idiopathic thrombocytopenic purpura (ITP) Registry	National	Private for profit
United Kingdom neuromyelitis optica registry	National	Public
United Kingdom Primary Sjogren's Syndrome Registry	National	Public

European registries

**This table lists only the coordinating entity of the European registries. For regional or national participants of these European registries, please see table "Regional and national registries" above.*

EUROPEAN REGISTRIES (97 registries)*

AUSTRIA - Coordination (4 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
Central data registry of the European Competence Network on Mastocytosis (ECMN)	Public
EMSA-SG: central patient registry of the European multiple system atrophy network	Public
ENRAH: European alternating hemiplegia and rare epilepsies registry in childhood	Public
Thromboreductin-Registry	Private for profit

BELGIUM - Coordination (4 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
ENETS: European Neuroendocrine Tumour Registry	Public
ERN [EpiCARE] - EPICARE-GRANT: An Operational EPICARE Registry for all Rare and Complex Epilepsies	Public
EUNEFRON: registry of the European network for the study of orphan nephropathies	Public
EUROGLYCANET - International patient registry and cohort for congenital disorders of glycosylation	Public

ESTONIA - Coordination (1 registry)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
Estonian Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes - contribuiting to EURO-WABB	Public

FRANCE - Coordination (18 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
Cloud-R HAE Register Hereditary Angiodema	Public
EDMUS: European Database for Multiple Sclerosis and other related diseases	Public
ENETS Registry: European Neuro-Endocrine Tumors Group	Public
ERN [EURO-NMD] - EURO-NMD Registry: Patient centered and interoperable registry hub for Rare Neuromuscular Diseases	Public
ERN [EYE] - REDgistry: An interoperable sustainable European Rare Eye Disease Registry	Public
ERN [ITHACA] - ILIAD Rare Diseases patient registry: an International Library of Intellectual disability and Anomalies of Development	Public
ERN [Skin] - ERN-Skin REGISTRY: Interoperable ERN on Rare and Undiagnosed Skin Disorders	Public
ERN [VASCERN] - VASCERN Registries	Public
EU-CHS: European central hypoventilation syndrome registry	Public
EURECHINOREG: European registry of alveolar echinococcosis	Public
EurêClark registry (The European Clarkson's syndrome registry)	Public
European LeukoDataBase	Public

European Society for Blood and Marrow Transplant Society Registry (EBMT registry)	Public
EURORETT: European network on Rett syndrome (registry)	Public
RaDiCo-ECYSCO: European cystinosis cohort	Public
RaDiCo-EURBIO-Alport: Study of the natural history of Alport Syndrome by establishment of an international database	Public
RaDiCo-IDMet: National and European cohort on Imprinting Disorders and their metabolic consequences	Public
SCETIDE - Stem CEll Transplant for primary Immune Deficiencies in Europe	Public

GERMANY - Coordination (24 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
AML-BFM Register 2017: Registry for children and adolescents with acute myeloid leukemia	Private non-profit
CEDATA-GPGE: Registry of Children with Inflammatory Bowel Disease in Germany and Austria	Public
CERTAIN-LI: Cooperative European Paediatric TransplAnt INitiative LIver	Private non-profit
CERTAIN: Cooperative European Paediatric Renal Transplant Initiative registry	Private non-profit
EBAR: European Biliary Atresia Registry	Public
EIMD: European registry and network for intoxication type metabolic diseases	Public
EMCL-REGISTRY: European Mantle cell lymphoma registry	Private non-profit
ERN [ERKNet] - ERKReg: The European Rare Kidney Disease Registry	Public
ERN [ERN-LUNG] : REGISTRY WAREHOUSE - RD Registry Data Warehouse	Public
ERN [MetabERN] - UIMD: Unified European Registry for Inherited Metabolic DisordersUIMD: Unified European Registry for Inherited Metabolic Disorders	Public
ERN [RARE-LIVER] - R-LIVER: Registry for Rare Liver Diseases of the ERN on hepatological diseases	Public
ERN [RND] - ERN-RND Registry: The ERN-RND Rare Neurological Disease Registry	Public
ESID: European registry of primary immunodeficiencies	Public
EU-RHAB: European Rhabdoid Tumor Registry	Private non-profit
eurIPFreg: European idiopathic pulmonary fibrosis registry	Public
EUROFA - EFACT: European Friedreich Ataxia Registry	Public
European Alport registry	Public
European chILD-registry and biobank of the european network for children's interstitial lung diseases (chILD-EU).	Public
European registry of sporadic degenerative ataxia with adult onset	Public
EUROSCA-R: European patient registry on spinocerebellar ataxias	Public
KiRaFe: ChildrenRegister for Congenital Malformations	Private non-profit
Nephronophthisis registry for patients in germany, austria and switzerland	Private for profit
Registry of the European Working Group (EWOG) for Severe Aplastic Anemia (SAA) in children and adolescents	Public
Registry of the European Working Group (EWOG) of Myelodysplastic Syndromes (MDS) and Juvenile Myelomonocytic Leukemia (JMML) in childhood	Public

IRELAND - Coordination (1 registry)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
UKIVAS - UK and Ireland registry of autoimmune vasculitides	Public

ITALY - Coordination (6 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
ERN [EURACAN] - STARTER: STarting an Adult Rare Tumor European Registry	Public
ERN [ReCONNET] - TogethERN ReCONNET: a European Registry Infrastructure for data harmonization in rare and complex connective tissue and musculoskeletal diseases	Public
ERN [RITA] - MERITA: A METADATA REGISTRY FOR THE ERN RITA	Public
ERN [PAEDCAN] - PARTNER: Paediatric Rare Tumours Networks - European Registry	Public
EUROFEVER: PReS European network of registries for autoinflammatoy diseases in childhood	Public
HAE-registry: European hereditary angioedema patient registry	Public

NETHERLANDS (THE) - Coordination (13 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
Diffuse Intrinsic Pontine Glioma (DIPG) Registry	Private non-profit
E-HOD - European network and registry for homocystinurias and methylation defects	Public
E-HOD - European network and registry for homocystinurias and methylation defects (contributes to U-IMD)	Public
EPCOT: European prospective cohort on thrombophilia	Public
ERN [BOND] - EuRR-Bone: European Registry for Rare Bone and Mineral Conditions	Public
ERN [CRANIO] - ERN CRANIO registry	Public
ERN [ERNICA] - ERNICA-registry for improving care	Public
ERN [eUROGEN] - ERN eUROGEN Registry for rare urogenital diseases	Public
ERN [GENTURIS] - GENTURIS registry: The ERN Genetic Tumour Risk Syndromes Registry	Public
ERN [GUARD-Heart] - The Heart-Core Registry: a Gateway to Uncommon and Rare Diseases of the Heart	Public
ESPN/ERA-EDTA Registry: European Registry for Children on Renal Replacement Therapy	Public
European Parathyroid Tumor Registry	Public
PAAIR: Patient's Association and Alpha-1 International Registry network (FINALISED)	Public

NORWAY - Coordination (4 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
EURADRENAL: European patient registry on autoimmune Addison's disease (sera, DNA and RNA)	Public
EURADRENAL: pathophysiology and natural course of autoimmune adrenal failure in Europe (registries)	Public
European Porphyria Registry (EPR)	Public
HUE-MAN patient registry on alpha mannosidosis	Public

SPAIN - Coordination (5 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
EARCO Registry	Public
ERCUSYN: European registry on Cushing's syndrome	Public
ERN [EuroBloodNet] - ENROL: European Rare Blood Disorders Platform	Public
ERN [TRANSPLANT-CHILD] - PETER: PaEdiatric Transplantation European Registry	Public
EUROMAC: Registry of patients affected by McArdle Disease	Public

SWEDEN - Coordination (2 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
EUROCRINE: European Surgical Registry for Rare Endocrine Tumours	Public
Swedish and Finnish registry of CADASIL patients	Public

SWITZERLAND - Coordination (1 registry)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
Juvenile Inflammatory Rheumatism (JIR) Cohorte (JIRcohorte)	Private non-profit

UNITED KINGDOM OF GREAT BRITAIN AND NORTHERN IRELAND (THE) - Coordination (14 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
ECARUCA: European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations	Public
ECFS: European Cystic Fibrosis Society patient registry	Public
EHR: European Haemoglobinopathy Registry	Public
ERN [Endo-ERN] - EuRRECa: European Registries for Rare Endocrine Conditions	Public
EUMDS: European Registry for Myelodysplastic Syndromes - part of EuroLeukemiaNet (ELN)	Public
EURO-WABB: An EU Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes	Public
EUROCARE CF: European cystic fibrosis registry	Public
EUROCAT: European surveillance of congenital anomalies	Public
EuroMyositis registry	Public
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	Public
European Prader-Willi syndrome database	Public
EUROWILSON: European network on Wilson disease (registries)	Public
UK and Ireland Duchenne and Becker muscular dystrophy patient registry (part of the TREAT-NMD network)	Public
UK and Ireland Spinal muscular atrophy (SMA) patient registry (part of the TREAT-NMD network)	Public

International registries coordinated in Europe

**This table lists only the coordinating entity of the international registries. For regional or national participants of these European registries, please see table "Regional and national registries" above.*

INTERNATIONAL REGISTRIES (76 registries)*

AUSTRIA - Coordination (2 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
EBCare Registry	Public
International aHUS registry	Public

FRANCE - Coordination (6 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Immune thrombocytopenic purpura (ITP)	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Lupus	Public
Franco-Canadian program for the surveillance and pharmacoepidemiological evaluation of risk factors for rare diseases (PGRx) : Myositis	Public
RaDiCo-GenIDA: International social network for data collection on the natural history of rare monogenic forms of intellectual disabilities	Public
Registry for Patients with Digital Ulcers Associated with Systemic Sclerosis (DU/SSc)	Public
VALDIG: cohort creation on Budd-Chiari syndrome, hepatic venooclusive disease, hepatoportal sclerosis and portal vein thrombosis	Public

GERMANY - Coordination (23 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
ADPedKD - International, longitudinal registry including ADPKD patients followed up from childhood.	Private non-profit
ARegPKD - International Registry Study on Autosomal Recessive Polycystic Kidney Disease	Public
Blue cone monochromatism - Patient registry	Private non-profit
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	Private non-profit
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	Private non-profit
DÖSAK tumor registry for documentation of tumors of the face and jaws in germany, austria and switzerland	Public
Enroll-HD Registry	Private non-profit
FungiScope - Global Rare Fungal Infection Registry	Public
iNTD (International Working Group on Neurotransmitter Related Disorders) Registry	Private non-profit
iNTD Network - International Working Group on Neurotransmitter Related Disorders	Private non-profit
International registry for Primary Ciliary Dyskinesia	Public
Kids Lung Register: International register and biobank for rare lung diseases	Public
KINDLERNET: Central patient registry Kindler syndrome	Public
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	Private for profit

NHL-BFM Registry 2012: Registry of the NHL-BFM study group for all subtypes of Non-Hodgkin Lymphoma diagnosed in children and adolescents	Public
OSTEOPETR: International registry of patients suffering from osteopetrosis	Public
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	Public
RegiSCAR: International registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples - patient registry	Public
Register for rare myeloproliferative neoplasms	Public
RetDis Database: clinical descriptions of patients and families with inherited eye diseases	Public
SCNIR: Severe Chronic Neutropenia International Registry	Public
STER: FVII deficiency treatment international registry	Public
TIRCON: NBIA (Neurodegeneration with Brain Iron Accumulation) patient registry	Public

ITALY - Coordination (8 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
Blue cone monochromatism patient registry (BCM Registry)	Private non-profit
International registry of bone fragility fractures in the young	Public
International Registry of congenital dyserythropoietic anemia II	Public
International Registry of Rare Bleeding Disorders (RBDD)	Public
International registry of recurrent and familial hemolytic uremic syndrome/thrombotic thrombocytopenic purpura	Private non-profit
Ring14 Clinical database	Public
The Global Registry of Hereditary Angioedema Type I and II	Private non-profit
TTP: International registry on thrombotic thrombocytopenic purpura	Public

NEHERLANDS (THE) - Coordination (12 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	**AFFILIATION**
Galactosemia Patient Registry - part of the Galactosemia Network	Public
International Dystrophic Epidermolysis Bullosa Patient Registry	Private non-profit
Membranous Nephropathy Registry	Public
PedNet Haemophilia Registry	Private non-profit
PHARMACHILD patient registry: Long-term pharmacovigilance for adverse effects in childhood arthritis, focusing on immune modulatory drugs (part of PRINTO network)	Public
Registry NXT - Genzyme's Rare Disease Registries	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	Private non-profit
The intenational Pompe registry	Private for profit
The International Collaborative Gaucher Group (ICGG) Gaucher registry	Private for profit
The international Fabry registry	Private for profit
The international Mps I registry	Private for profit

SPAIN - Coordination (1 registry)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
RenalTube: Database of patients with primary tubulopathies	Public

SWEDEN - Coordination (4 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
FOS : Fabry Outcome Survey	Public
HOS : Hunter Outcome Survey	Public
International registry for biologics in systemic lupus erythematosus (IRBIS)	Public
IOS : Icatibant Outcome Survey for hereditory angioedema	Public

SWITZERLAND - Coordination (7 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
COST Action BM1105 Patient Registry - GnRH Network	Public
Hereditary TTP Registry	Public
Intercontinental Cooperative ITP Study Group Registry	Public
International Registry of Porto-Systemic Shunts (IRCPSS)	Public
IRASPEN Registry - International Rare and Severe Psoriasis Expert Network	Public
Perihilar Cholangiocarcinoma International Registry	Public
PNH Registry: Paroxysmal Nocturnal Hemoglobinuria registry	Private for profit

UNITED KINGDOM OF GREAT BRITAIN AND NORTHERN IRELAND (THE) - Coordination (13 registries)	
ENGLISH LABEL OF THE REGISTRY/COHORT	AFFILIATION
Ataxia-Telangiectasia International Registry (A-T Society)	Public
Global FKRP registry (Global Fukutin-Related Protein defects registry) - Part of TREAT-NMD Alliance	Public
I-DSD: International Disorders of Sex Development registry	Public
I-DSD: International Disorders of Sex Development registry - GB	Public
International A-T Registry	Public
International Niemann-Pick Disease Registry	Public
Registry for Patients with Niemann-Pick Type C Disease	Public
The Global aHUS Registry	Private for profit
The Global FKRP Patient Registry	Public
The MTM and CNM Registry - The Myotubular and Centronuclear Myopathy Patient Registry	Public
TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases (registries)	Public
TuberOus SClerosis registry to increase disease Awareness (TOSCA)	Private for profit
International GNE Myopathy Patient Registry	Public

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

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