Orphanet Report Series

Rare Diseases collection

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Disease Registries in Europe

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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 authorisation is usually granted at a time when evidence is still limited although already somewhat convincing.

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

Cancer registries are listed only if they belong to the network RARECARE or focus on a rare form of cancer.

The report includes data about EU countries and surrounding countries participating to the Orphanet consortium.

The diseases covered by each register are provided with Orpha number which is their identifier in the Orphanet nomenclature.

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

List of rare diseases that are covered by the listed registries

ORPHA Number	Disease name
19	2-hydroxyglutaric aciduria
79157	2-methylbutyryl-CoA dehydrogenase deficiency
35701	3-hydroxy 3-methylglutaryl-CoA synthase deficiency
67046	3-methylglutaconic aciduria type 1
2982	46,XX disorder of sex development
243	46,XX gonadal dysgenesis
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
22	4-hydroxybutyric aciduria
13	6-pyruvoyl-tetrahydropterin synthase deficiency
96121	7q11.23 microduplication syndrome
918	ABCD syndrome
988	Absent tibia - polydactyly
48818	Aceruloplasminemia
932	Achondrogenesis
93296	Achondrogenesis type 2
15	Achondroplasia
49382	Achromatopsia
218439	Acquired cardiac rythm disease
79356	Acrokeratoderma
963	Acromegaly
99892	ACTH-dependent Cushing syndrome
318	Acute erythroid leukemia
79276	Acute intermittent porphyria
79126	Acute interstitial pneumonia
513	Acute lymphoblastic leukemia
518	Acute megacaryoblastic leukemia
514	Acute monoblastic leukemia
519	Acute myeloid leukemia
517	Acute myelomonocytic leukemia
35889	Acute opioid poisoning
520	Acute promyelocytic leukemia
85138	Addison disease
45	Adenosine monophosphate deaminase deficiency
100091	Adrenal/paraganglial tumor
1501	Adrenocortical carcinoma
2688	Adult idiopathic neutropenia
206583	Adult polyglucosan body disease

ORPHA Number	Disease name
829	Adult Still's disease
978	ADULT syndrome
99027	Adult-onset autosomal dominant leukodystrophy
99000	Adult-onset foveomacular vitelliform dystrophy
51	Aicardi-Goutieres syndrome
52	Alagille syndrome
53	Albers-Schönberg osteopetrosis
665	Albright hereditary osteodystrophy
58	Alexander disease
56	Alkaptonuria
1164	Allergic bronchopulmonary aspergillosis
726	Alpers syndrome
98791	Alpha thalassemia - intellectual deficit syndrome
847	Alpha thalassemia - X-linked intellectual deficit
60	Alpha-1 antitrypsin deficiency
98910	Alpha-crystallinopathy
61	Alpha-mannosidosis
846	Alpha-thalassemia
231401	Alpha-thalassemia - myelodysplastic syndrome
63	Alport syndrome
64	Alström syndrome
2131	Alternating hemiplegia of childhood
284	Alveolar echinococcosis
99756	Alveolar rhabdomyosarcoma
1023	Ambras syndrome
98902	Amish nemaline myopathy
69	Amyloidosis
803	Amyotrophic lateral sclerosis
754	Androgen insensitivity syndrome
72	Angelman syndrome
77	Aniridia
1065	Aniridia - cerebellar ataxia - intellectual deficit
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate
77298	Anophthalmia/microphthalmia - esophageal atresia
96346	Anorectal malformation
80	Antiphospholipid syndrome
81	Antisynthetase syndrome

ORPHA Number	Disease name
2299	Aortic arch interruption
87	Apert syndrome
320	Apparent mineralocorticoid excess
35704	Arginine:glycine amidinotransferase deficiency
90	Argininemia
23	Argininosuccinic aciduria
1136	Arnold-Chiari malformation type II
247	Arrhythmogenic right ventricular dysplasia
94	Astrocytoma
100	Ataxia-telangiectasia
1201	Atresia of small intestine
1344	Atrial cardiomyopathy with heart block
99103	Atrial septal defect, ostium secundum type
2134	Atypical hemolytic uremic syndrome
93575	Atypical hemolytic uremic syndrome with C3 anomaly
93579	Atypical hemolytic uremic syndrome with H factor anomaly
93580	Atypical hemolytic uremic syndrome with I factor anomaly
3095	Atypical Rett syndrome
79669	Autoimmune bullous skin disease
98375	Autoimmune hemolytic anemia
3261	Autoimmune lymphoproliferative syndrome
3453	Autoimmune polyendocrinopathy type 1
71203	Autoimmune thrombocytopenia
93665	Autoinflammatory syndrome
33110	Autosomal agammaglobulinemia
169189	Autosomal dominant centronuclear myopathy
99	Autosomal dominant cerebellar ataxia
99939	Autosomal dominant Charcot-Marie-Tooth disease type 2E
228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M
98808	Autosomal dominant dopa-responsive dystonia
98853	Autosomal dominant Emery-Dreifuss muscular dystrophy
73229	Autosomal dominant familial hematuria - retinal arteriolar tortuosity - contractures
2314	Autosomal dominant hyper IgE syndrome
89937	Autosomal dominant hypophosphatemic rickets
100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B

ORPHA Number	Disease name
2334	Autosomal dominant keratitis
503	Autosomal dominant Larsen syndrome
266	Autosomal dominant limb-girdle muscular dystrophy type 1A
264	Autosomal dominant limb-girdle muscular dystrophy type 1B
265	Autosomal dominant limb-girdle muscular dystrophy type 1C
90635	Autosomal dominant nonsyndromic sensorineural deafness type DFNA
3212	Autosomal dominant optic atrophy and congenital deafness
98673	Autosomal dominant optic atrophy, classic type
2783	Autosomal dominant osteopetrosis type 1
2790	Autosomal dominant osteosclerosis, Worth type
486	Autosomal dominant severe congenital neutropenia
1797	Autosomal dominant spondylocostal dysostosis
3086	Autosomal dominant vitreoretinochoroidopathy
88919	Autosomal recessive Alport syndrome
1027	Autosomal recessive amelia
88644	Autosomal recessive ataxia, Beauce type
169186	Autosomal recessive centronuclear myopathy
1172	Autosomal recessive cerebellar ataxia
217046	Autosomal recessive childhood-onset cortical cataract
89838	Autosomal recessive epidermolysis bullosa simplex
169446	Autosomal recessive hyper IgE syndrome
79644	Autosomal recessive hyperinsulinism due to Kir6.2 deficiency
79643	Autosomal recessive hyperinsulinism due to SUR1 deficiency
267	Autosomal recessive limb girdle muscular dystrophy type 2A
280333	Autosomal recessive limb-girdle muscular dystrophy - dystroglycanopathy type C7
268	Autosomal recessive limb-girdle muscular dystrophy type 2B
353	Autosomal recessive limb-girdle muscular dystrophy type 2C
62	Autosomal recessive limb-girdle muscular dystrophy type 2D
119	Autosomal recessive limb-girdle muscular dystrophy type 2E
219	Autosomal recessive limb-girdle muscular dystrophy type 2F
34514	Autosomal recessive limb-girdle muscular dystrophy type 2G
1878	Autosomal recessive limb-girdle muscular dystrophy type 2H
34515	Autosomal recessive limb-girdle muscular dystrophy type 2I
140922	Autosomal recessive limb-girdle muscular dystrophy type 2J
86812	Autosomal recessive limb-girdle muscular dystrophy type 2K
206549	Autosomal recessive limb-girdle muscular dystrophy type 2L
206554	Autosomal recessive limb-girdle muscular dystrophy type 2M
206559	Autosomal recessive limb-girdle muscular dystrophy type 2N
667	Autosomal recessive malignant osteopetrosis
2990	Autosomal recessive multiple pterygium syndrome
90636	Autosomal recessive nonsyndromic sensorineural deafness type DFNB
731	Autosomal recessive polycystic kidney disease
2512	Autosomal recessive primary microcephaly
99013	Autosomal recessive spastic paraplegia type 7
2311	Autosomal recessive spondylocostal dysostosis
98152	Autosomal uniparental disomy
109	Bannayan-Riley-Ruvalcaba syndrome
110	Bardet-Biedl syndrome
111	Barth syndrome
98895	Becker muscular dystrophy
116	Beckwith-Wiedemann syndrome
117	Behcet disease
86814	Benign adult familial myoclonic epilepsy
306	Benign familial infantile seizures
71518	Benign paroxysmal torticollis of infancy
274	Bernard-Soulier syndrome
1243	Best disease
848	Beta-thalassemia
231230	Beta-thalassemia associated with another hemoglobin anomaly
231214	Beta-thalassemia major
610	Bethlem myopathy
1848	Bilateral renal agenesis
30391	Biliary atresia
122	Birt-Hogg-Dube syndrome
124	Blackfan-Diamond disease
157980	Bladder cancer
86870	Blastic NK-cell lymphoma
90340	Blau syndrome
126	Blepharophimosis - epicanthus inversus - ptosis
3047	Blepharophimosis-intellectual deficit syndrome, SBBYS type
125	Bloom syndrome
16	Blue cone monochromatism
223727	Bone sarcoma
107	BOR syndrome
93383	Brachydactyly type B
1297	Branchio-oculo-facial syndrome
97287	Bronchial endocrine tumor
1303	Bronchiolitis obliterans with obstructive pulmonary disease
70589	Bronchopulmonary dysplasia
2771	Bruck syndrome
130	Brugada syndrome
131	Budd-Chiari syndrome
543	Burkitt lymphoma
99001	Butterfly-shaped pigment dystrophy
136	CADASIL syndrome
1310	Caffey disease
280062	Calciphylaxis
140	Campomelic dysplasia
141	Canavan disease
171881	Cap myopathy
137667	Capillary malformation-arteriovenous malformation
147	Carbamoylphosphate synthetase deficiency
37553	Cardiodysrythmic potassium-sensitive periodic paralysis
167848	Cardiomyopathy
97286	Carney-Stratakis syndrome
157	Carnitine palmitoyl transferase II deficiency
93973	Carpenter-Waziri syndrome
160	Castleman disease
98985	Cataract with Y-shaped suture opacities
98986	Cataract, Coppock-like
1377	Cataract-microcornea syndrome
717	Catecholamine-producing tumor
3286	Catecholaminergic polymorphic ventricular tachycardia
195	Cat-eye syndrome
3027	Caudal regression sequence
169464	CD59 deficiency
75377	Central areolar choroidal dystrophy
597	Central core disease
595	Centronuclear myopathy
909	Cerebrotendinous xanthomatosis
169079	Cernunnos-XLF deficiency
98989	Cerulean cataract
90658	Charcot-Marie-Tooth disease type 1E
101085	Charcot-Marie-Tooth disease type 1F
98856	Charcot-Marie-Tooth disease type 2B1
138	CHARGE syndrome
167	Chediak-Higashi syndrome
55880	Chondrosarcoma
2388	Choreoacanthocytosis
179	Chorioretinopathy, Birdshot type

ORPHA Number	Disease name
180	Choroideremia
68335	Chromosomal anomaly
379	Chronic granulomatous disease
2978	Chronic intestinal pseudo-obstruction
521	Chronic myeloid leukemia
98274	Chronic myeloproliferative disease
93971	Chudley-Lowry-Hoar syndrome
187	Citrullinemia
247525	Citrullinemia type I
394	Classical homocystinuria
1991	Cleft lip with or without cleft palate
2001	Cleft lip/palate - intestinal malrotation - cardiopathy
2014	Cleft palate
191	Cockayne syndrome
192	Coffin-Lowry syndrome
1466	COFS syndrome
31824	Colchicine poisoning
1198	Colonic atresia
35909	Combined deficiency of factor V and factor VIII
169082	Combined immunodeficiency due to CD3gamma deficiency
169090	Combined immunodeficiency due to CRAC channel dysfunction
911	Combined immunodeficiency due to ZAP70 deficiency
1572	Common variable immunodeficiency
77303	Common variable immunodeficiency due to an intrinsic B cell defect
99831	Common variable immunodeficiency due to an intrinsic T cell defect
1329	Complete atrioventricular canal
1872	Cone rod dystrophy
3091	Congenital abnormal systemic venous return
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral
418	Congenital adrenal hyperplasia
97556	Congenital and infantile nephrotic syndrome
2040	Congenital bronchobiliary fistula
48431	Congenital cataracts - facial dysmorphism - neuropathy
115	Congenital contractural arachnodactyly
2140	Congenital diaphragmatic hernia
137	Congenital disorder of glycosylation
85	Congenital dyserythropoietic anemia
79277	Congenital erythropoietic porphyria
327	Congenital factor VII deficiency
329	Congenital factor XI deficiency
331	Congenital factor XIII deficiency
2020	Congenital fiber-type disproportion myopathy
335	Congenital fibrinogen deficiency
45358	Congenital fibrosis of extraocular muscles
88991	Congenital heart malformation
442	Congenital hypothyroidism
651	Congenital idiopathic nystagmus
210163	Congenital lethal myopathy, Compton-North type
68378	Congenital limb malformation
98905	Congenital multicore myopathy with external ophthalmoplegia
97242	Congenital muscular dystrophy
157973	Congenital muscular dystrophy due to LMNA mutation
34520	Congenital muscular dystrophy with integrin deficiency
272	Congenital muscular dystrophy, Fukuyama type
75840	Congenital muscular dystrophy, Ullrich type
199329	Congenital myopathy, Paradas type
839	Congenital nephrotic syndrome, Finnish type
79394	Congenital nonbullous ichthyosiform erythroderma
465	Congenital plasminogen activator inhibitor type 1 deficiency
2416	Congenital primary lymphedema
2444	Congenital pulmonary airway malformation of the lung
2414	Congenital pulmonary lymphangiectasia
3090	Congenital pulmonary venous return anomaly
215	Congenital stationary night blindness
858	Congenital toxoplasmosis
169826	Congenital vitamin K-dependent coagulation factors deficiency
860	Congenitally uncorrected transposition of the great arteries
2445	Conotruncal heart malformations
101987	Constitutional neutropenia
98990	Coralliform cataract
199	Cornelia de Lange syndrome
54251	Corticosteroid-sensitive aseptic abscesses
1529	Craniofacial-deafness-hand syndrome
204	Creutzfeldt-Jakob disease
2935	Crossed polysyndactyly
96253	Cushing disease
553	Cushing syndrome
79140	Cutaneous neuroendocrine carcinoma
2686	Cyclic neutropenia
586	Cystic fibrosis
213	Cystinosis
95702	Cytomegalic congenital adrenal hypoplasia
137678	Czech dysplasia, metatarsal type
218	Darier disease
3214	Deaf blind hypopigmentation syndrome, Yemenite type
1578	Dehydratase deficiency
79134	DEND syndrome
1652	Dent disease
101	Dentatorubral-pallidoluysian atrophy
220	Denys-Drash syndrome
221	Dermatomyositis
86920	Dermatopathia pigmentosa reticularis
98909	Desminopathy
84132	Desmin-related myopathy with Mallory body-like inclusions
873	Desmoid disease
83469	Desmoplastic small round cell tumor
1666	Dextrocardia
90060	Diffuse alveolar hemorrhage
544	Diffuse large B-cell lymphoma
31828	Digitalis poisoning
1146	Digitotalar dysmorphism
226	Dihydropteridine reductase deficiency
79166	Disorder of amino acid absorption and transport
83004	Disorder of sex development of endocrine origin
178400	Distal myopathy with anterior tibial onset
600	Distal myopathy with vocal cord weakness
602	Distal myopathy, Nonaka type
18	Distal renal tubular acidosis
70594	Dopa responsive dystonia due to sepiapterin reductase deficiency
255	Dopa-responsive dystonia
79145	Dowling-Degos disease
870	Down syndrome
263494	DPM3-CDG syndrome
33069	Dravet syndrome
139402	Drug rash with eosinophilia and systemic symptoms
262	Duchenne and Becker muscular dystrophy
98896	Duchenne muscular dystrophy
1203	Duodenal atresia
1775	Dyskeratosis congenita
199351	Dystonia-parkinsonism, Paisan-Ruiz type
303	Dystrophic epidermolysis bullosa
256	Early onset torsion dystonia
1020	Early-onset autosomal dominant Alzheimer disease
2379	Early-onset parkinsonism - intellectual deficit
1880	Ebstein malformation
1896	EEC syndrome
90309	Ehlers-Danlos syndrome type 1
99875	Ehlers-Danlos syndrome type 7A

ORPHA Number	Disease name
99876	Ehlers-Danlos syndrome type 7B
287	Ehlers-Danlos syndrome, classic type
285	Ehlers-Danlos syndrome, hypermobility type
1900	Ehlers-Danlos syndrome, kyphoscoliotic type
286	Ehlers-Danlos syndrome, vascular type
261	Emery-Dreifuss muscular dystrophy
877	Endocrine tumor
100092	Enteropancreatic endocrine tumor
85438	Enthesitis-related arthritis
301	Ependymal tumor
302	Epidermodysplasia verruciformis
158681	Epidermolysis bullosa simplex with circinate migratory erythema
79397	Epidermolysis bullosa simplex with mottled pigmentation
257	Epidermolysis bullosa simplex with muscular dystrophy
158684	Epidermolysis bullosa simplex with pyloric atresia
79396	Epidermolysis bullosa simplex, Dowling-Meara type
79401	Epidermolysis bullosa simplex, Ogna type
312	Epidermolytic ichthyosis
2199	Epidermolytic palmoplantar keratoderma
79355	Erythrokeratoderma
317	Erythrokeratodermia variabilis
79278	Erythropoietic protoporphyria
1199	Esophageal atresia
3318	Essential thrombocythemia
31826	Ethylene glycol poisoning
1959	Evans syndrome
319	Ewing sarcoma
324	Fabry disease
269	Facioscapulohumeral dystrophy
733	Familial adenomatous polyposis
85447	Familial amyloid polyneuropathy
334	Familial atrial fibrillation
86820	Familial avascular necrosis of femoral head
2841	Familial benign chronic pemphigus
211	Familial cylindromatosis
75376	Familial drusen
891	Familial exudative vitreoretinopathy
540	Familial hemophagocytic lymphohistiocytosis
424	Familial hyperthyroidism due to mutations in TSH receptor
31043	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis
440	Familial hypospadias
656	Familial idiopathic steroid-resistant nephrotic syndrome
93217	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis
93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis
217656	Familial isolated arrhythmogenic right ventricular dysplasia
154	Familial isolated dilated cardiomyopathy
155	Familial isolated hypertrophic cardiomyopathy
209886	Familial juvenile hyperuricemic nephropathy type 1
523	Familial leiomyomatosis
768	Familial long QT syndrome
342	Familial mediterranean fever
618	Familial melanoma
88632	Familial ocular anterior segment mesenchymal dysgenesis
569	Familial or sporadic hemiplegic migraine
213517	Familial ovarian cancer
1333	Familial pancreatic carcinoma
97	Familial paroxysmal ataxia
2348	Familial partial lipodystrophy, Dunnigan type
871	Familial progressive cardiac conduction defect
1331	Familial prostate cancer
51083	Familial short QT syndrome
2903	Familial spontaneous pneumothorax
91387	Familial thoracic aortic aneurysm and aortic dissection
84	Fanconi anemia
1561	Fatal infantile cytochrome C oxidase deficiency
1987	Femoral agenesis/hypoplasia
994	Fetal akinesia deformation sequence
337	Fibrodysplasia ossificans progressiva
249	Fibrous dysplasia of bone
2092	Focal dermal hypoplasia
1866	Focal dystonia
48918	Focal myositis
545	Follicular lymphoma
2253	Foveal hypoplasia - presenile cataract
908	Fragile X syndrome
93256	Fragile X-associated tremor/ataxia syndrome
347	Frasier syndrome
2053	Freeman-Sheldon syndrome
95	Friedreich ataxia
24	Fumaric aciduria
63443	Gastric cancer
36273	Gastric linitis plastica
44890	Gastrointestinal stromal tumor
2368	Gastroschisis
355	Gaucher disease
51608	Generalized arterial calcification of infancy
228429	Generalized congenital lipodystrophy with myopathy
36387	Generalized epilepsy with febrile seizures-plus context
171876	Generalized pseudohypoaldosteronism type 1
3221	Generalized resistance to thyroid hormone
271835	Genetic digestive tract tumor
183497	Genetic neuromuscular disease
98497	Genetic peripheral neuropathy
358	Gitelman syndrome
849	Glanzmann thrombasthenia
182067	Glial tumor
360	Glioblastoma
25	Glutaric acidemia type 1
26791	Glutaric acidemia type 2
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
34587	Glycogen storage disease due to LAMP-2 deficiency
368	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
374	Goldenhar syndrome
53540	Goldmann-Favre syndrome
1986	Gollop-Wolfgang complex
375	Goodpasture syndrome
377	Gorlin syndrome
53693	GRACILE syndrome
3274	Granulomatous arthritis of childhood
380	Greig cephalopolysyndactyly syndrome
79476	Griscelli disease
73272	Growth delay due to insulin-like growth factor I deficiency
73273	Growth delay due to insulin-like growth factor I resistance
2102	GTP cyclohydrolase I deficiency
382	Guanidinoacetate methyltransferase deficiency
168569	H syndrome
99803	Haddad syndrome
457	Harlequin ichthyosis
875	Heart tumor of the child
1350	Heart-hand syndrome type 2
168796	Heart-hand syndrome, Slovenian type
178330	Heinz body anemia
2130	Hemimelia
139491	Hemochromatosis type 4

ORPHA Number	Disease name
2132	Hemoglobin C disease
2133	Hemoglobin E disease
68364	Hemoglobinopathy
158032	Hemophagocytic syndrome
448	Hemophilia
98879	Hemophilia B
761	Henoch-Schönlein purpura
890	Hepatic veno-occlusive disease
79124	Hepatic veno-occlusive disease - immunodeficiency
449	Hepatoblastoma
88673	Hepatocellular carcinoma
33402	Hepatocellular carcinoma, childhood-onset
64743	Hepatoportal sclerosis
91378	Hereditary angioedema
145	Hereditary breast and ovarian cancer syndrome
30925	Hereditary central diabetes insipidus
676	Hereditary chronic pancreatitis
79273	Hereditary coproporphyria
3197	Hereditary hyperekplexia
157215	Hereditary hypophosphatemic rickets with hypercalciuria
178464	Hereditary myopathy with early respiratory failure
43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency
640	Hereditary neuropathy with liability to pressure palsies
144	Hereditary nonpolyposis colon cancer
79357	Hereditary palmoplantar keratoderma
46532	Hereditary persistence of fetal hemoglobin - beta-thalassemia
29072	Hereditary pheochromocytoma-paraganglioma
685	Hereditary spastic paraplegia
82	Hereditary thrombophilia due to congenital antithrombin deficiency
71291	Hereditary vascular retinopathy
183678	Hermansky-Pudlak syndrome with neutropenia
231500	Hermansky-Pudlak syndrome with pulmonary fibrosis
231512	Hermansky-Pudlak syndrome without pulmonary fibrosis
1930	Herpetic encephalitis
189	Hidrotic ectodermal dysplasia
388	Hirschsprung disease
98293	Hodgkin lymphoma
391	Hodgkin lymphoma, classical
93970	Holmes-Gang syndrome
2162	Holoprosencephaly
392	Holt-Oram syndrome

ORPHA Number	Disease name
395	Homocystinuria due to methylenetetrahydrofolate reductase deficiency
622	Homocystinuria without methylmalonic aciduria
3322	Hoyeraal-Hreidarsson syndrome
399	Huntington disease
98934	Huntington disease-like 2
93473	Hurler syndrome
740	Hutchinson-Gilford progeria syndrome
2182	Hydrocephalus with stenosis of aqueduct of Sylvius
927	Hyperammonemia due to N-acetylglutamate synthetase deficiency
101089	Hyper-IgM syndrome type 2
101090	Hyper-IgM syndrome type 3
101092	Hyper-IgM syndrome type 5
682	Hyperkalemic periodic paralysis
415	Hyperornithinemia-hyperammonemia-homocitrullinuria
3416	Hyperostosis corticalis generalisata
416	Hyperoxaluria
238583	Hyperphenylalaninemia
31740	Hypersensitivity pneumonitis
98813	Hypohidrotic ectodermal dysplasia with immunodeficiency
681	Hypokalemic periodic paralysis
436	Hypophosphatasia
2248	Hypoplastic left heart syndrome
3332	Hypoplastic tibiae - post axial polydactyly
90673	Hypothyroidism due to TSH receptor mutations
1573	Hypotrichosis with juvenile macular degeneration
2268	ICF syndrome
79354	Ichthyosis
79503	Ichthyosis hystrix of Curth-Macklin
60033	Idiopathic bronchiectasis
182101	Idiopathic eosinophilic pneumonia
33208	Idiopathic hypersomnia
98482	Idiopathic inflammatory myopathy
98300	Idiopathic interstitial pneumonia
85193	Idiopathic juvenile osteoporosis
747	Idiopathic pulmonary alveolar proteinosis
2032	Idiopathic pulmonary fibrosis
99931	Idiopathic pulmonary hemosiderosis
69061	Idiopathic steroid-sensitive nephrotic syndrome
3002	Immune thrombocytopenic purpura
34592	Immunodeficiency by defective expression of HLA class 1
572	Immunodeficiency by defective expression of HLA class 2

ORPHA Number	Disease name
169150	Immunodeficiency due to a late component of complements deficiency
169147	Immunodeficiency due to an early component of complement deficiency
169100	Immunodeficiency due to CD25 deficiency
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency
200421	Immunodeficiency with factor H anomaly
200418	Immunodeficiency with factor I anomaly
68367	Inborn errors of metabolism
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
611	Inclusion body myositis
464	Incontinentia pigmenti
238455	Infantile dystonia-parkinsonism
35069	Infantile neuroaxonal dystrophy
79263	Infantile neuronal ceroid lipofuscinosis
1186	Infantile onset spinocerebellar ataxia
772	Infantile Refsum disease
90003	Inflammatory pseudotumor of the liver
79361	Inherited epidermolysis bullosa
85295	Intellectual deficit, X-linked - choreoathetosis - abnormal behavior
85287	Intellectual deficit, X-linked, Siderius type
1478	Interauricular communication
171433	Intermediate nemaline myopathy
182095	Interstitial lung disease
264735	Interstitial lung disease specific to adulthood
104010	Intestinal polyposis
79159	Isobutyryl-CoA dehydrogenase deficiency
1048	Isolated anencephaly/exencephaly
2542	Isolated anophthalmia - microphthalmia
557	Isolated anorectal malformation
238666	Isolated congenital hypogonadotropic hypogonadism
408	Isolated glycerol kinase deficiency
137902	Isolated optic nerve hypoplasia
718	Isolated Pierre Robin syndrome
823	Isolated spina bifida
33	Isovaleric acidemia
435	Ito hypomelanosis
90647	Jervell and Lange-Nielsen syndrome
475	Joubert syndrome
93972	Juberg-Marsidi syndrome
93672	Juvenile dermatomyositis
2929	Juvenile gastrointestinal polyposis
248111	Juvenile Huntington disease
92	Juvenile idiopathic arthritis
79264	Juvenile neuronal ceroid lipofuscinosis
93568	Juvenile polymyositis
2322	Kabuki syndrome

ORPHA Number	Disease name
2331	Kawasaki disease
480	Kearns-Sayre syndrome
481	Kennedy disease
494	Keratoderma hereditarium mutilans
50942	Keratosis palmoplantaris striata
477	KID syndrome
2908	Kindler syndrome
99978	Klatskin tumor
33543	Kleine-Levin syndrome
485	Kniest dysplasia
99749	Kostmann syndrome
275543	L1 syndrome
501	Lafora disease
313	Lamellar ichthyosis
98301	Laminopathy
137871	Laminopathy type Decaudain-Vigouroux
2632	Langer mesomelic dysplasia
502	Langer-Giedion syndrome
389	Langerhans cell histiocytosis
626	Large congenital melanocytic nevus
633	Laron syndrome
220465	Laron syndrome with immunodeficiency
1202	Larynx atresia
2377	Laurence-Moon syndrome
65	Leber congenital amaurosis
54260	Left ventricular noncompaction
2380	Legg-Calvé-Perthes disease
549	Legionellosis
506	Leigh syndrome
240	Léri-Weill dyschondrosteosis
33108	Lethal multiple pterygium syndrome
1662	Lethal restrictive dermopathy
99842	Leukocyte adhesion deficiency type I
99843	Leukocyte adhesion deficiency type II
755	Leydig cell hypoplasia
526	Liddle syndrome
524	Li-Fraumeni syndrome
99812	LIG4 syndrome
263	Limb-girdle muscular dystrophy
69085	Limb-mammary syndrome
89844	Lissencephaly syndrome, Norman-Roberts type
79400	Localized epidermolysis bullosa simplex
60030	Loeys-Dietz syndrome type 1
91546	Lyme disease
538	Lymphangioleiomyomatosis
33001	Lymphedema - distichiasis
171898	Lymphoid hemopathy
223735	Lymphoma
592	Macrophagic myofasciitis
97977	Malformative disorder of sex development
679	Malignant atrophic papulosis
3399	Malignant germ-cell tumor
423	Malignant hyperthermia
168999	Malignant melanoma of the mucosa
943	Malonic aciduria
52417	MALT lymphoma
90153	Mandibuloacral dysplasia with type A lipodystrophy
90154	Mandibuloacral dysplasia with type B lipodystrophy
52416	Mantle cell lymphoma
558	Marfan syndrome
559	Marinesco-Sjögren syndrome
42642	Marshall's syndrome with periodic fever
2466	MASA syndrome
98292	Mastocytosis
562	McCune-Albright syndrome
2473	McKusick-Kaufman syndrome
59306	McLeod neuroacanthocytosis syndrome
3097	Meacham syndrome
2006	Median cleft lip/mandibule
616	Medulloblastoma
98954	Meesmann corneal dystrophy
97338	Melanoma of soft part
51013	Melanoma-pancreatic cancer syndrome
550	MELAS syndrome
54370	Membranoproliferative glomerulonephritis
748	Mendelian susceptibility to mycobacterial diseases
551	MERRF syndrome
50251	Mesothelioma
31825	Methanol poisoning
2169	Methylcobalamin deficiency type cblE
2170	Methylcobalamin deficiency type cblG
26	Methylmalonic acidemia with homocystinuria
280183	Methylmalonic aciduria due to transcobalamin receptor defect
83642	Microcytic anemia with liver iron overload
2538	Microgastria - limb reduction defect
1083	Microlissencephaly
2543	Microphthalmia - cataract
568	Microphthalmia, Lenz type
58220	Microscopic colitis
83463	Microtia
171439	Mild nemaline myopathy
531	Miller-Dieker syndrome
68380	Mitochondrial disease
35698	Mitochondrial DNA depletion syndrome
45448	Miyoshi myopathy
552	MODY syndrome
573	Monilethrix
567	Monosomy 22q11
281	Monosomy 5p
98503	Motor neuron disease
2451	Mucocutaneous venous malformations
79213	Mucopolysaccharidosis
587	Muir-Torre syndrome
148	Multiple carboxylase deficiency
68341	Multiple congenital anomalies/dysmorphic syndrome
652	Multiple endocrine neoplasia type 1
29073	Multiple myeloma
321	Multiple osteochondromas
228145	Multiple sclerosis variant
102	Multiple system atrophy
588	Muscle eye brain disease
171445	Muscle filaminopathy
71864	Muscular channelopathy
98473	Muscular dystrophy
199340	Muscular dystrophy, Selcen type
589	Myasthenia gravis
52688	Myelodysplastic syndromes
824	Myelofibrosis with myeloid metaplasia
171895	Myeloid hemopathy
2587	Myeloperoxidase deficiency
182050	MYH9-related thrombocytopenia
36899	Myoclonic dystonia 11
275534	Myostatin-related muscle hypertrophy
98911	Myotilin-related myofibrillar myopathy without spheroid body
206647	Myotonic dystrophy
69087	Naegeli-Franceschetti-Jadassohn syndrome
627	Nance-Horan syndrome
83465	Narcolepsy without cataplexy
2073	Narcolepsy-cataplexy
150	Nasopharyngeal carcinoma
607	Nemaline myopathy
44	Neonatal adrenoleukodystrophy
654	Nephroblastoma
223	Nephrogenic diabetes insipidus
137617	Nephrogenic systemic fibrosis
634	Netherton syndrome
3388	Neural tube defect
635	Neuroblastoma
2481	Neurocutaneous melanocytosis
385	Neurodegeneration with brain iron accumulation
98534	Neurodegenerative disease with dementia
636	Neurofibromatosis type 1
637	Neurofibromatosis type 2
93921	Neurofibromatosis type 3
2678	Neurofibromatosis type 6

ORPHA Number	Disease name
163746	Neurologic Waardenburg-Shah syndrome
68381	Neuromuscular disease
71211	Neuromyelitis optica
216	Neuronal ceroid lipofuscinosis
139512	Neuropathy with hearing impairment
183707	Neutrophil immunodeficiency syndrome
2691	Nevo syndrome
77293	Niemann-Pick disease type B
646	Niemann-Pick disease type C
647	Nijmegen breakage syndrome
86867	Nodal marginal zone B-cell lymphoma
94080	Non secreting paraganglioma
79399	Non-Dowling-Meara generalized epidermolysis bullosa simplex
547	Non-Hodgkin lymphoma
157987	Non-Langerhans cell histiocytosis
91364	Nonspecific interstitial pneumonia
91492	Non-syndromic congenital cataract
87884	Nonsyndromic genetic deafness
648	Noonan syndrome
649	Norrie disease
98991	Nuclear cataract
1000	Ocular albinism - late-onset sensorineural deafness
194	Ocular coloboma
79431	Oculocutaneous albinism type 1A
79434	Oculocutaneous albinism type 1B
79432	Oculocutaneous albinism type 2
79433	Oculocutaneous albinism type 3
79435	Oculocutaneous albinism type 4
270	Oculopharyngeal muscular dystrophy
75382	Oguchi disease
46484	Oligodendroglial tumor
660	Omphalocele
661	Ondine syndrome
2086	Optic pathway glioma
664	Ornithine transcarbamylase deficiency
139039	Orofacial clefting syndrome
666	Osteogenesis imperfecta
2781	Osteopetrosis
2785	Osteopetrosis with renal tubular acidosis
2788	Osteoporosis - pseudoglioma
668	Osteosarcoma
64739	Ovarian hyperstimulation syndrome
93460	Overgrowth syndrome
2309	Pachyonychia congenita
2202	Palmoplantar keratoderma-deafness syndrome
98589	Palpebral malignant melanoma
217074	Pancreatic carcinoma
180824	Pancreatic tumor
677	Pancreatoblastoma
157850	Pantothenate-kinase-associated neurodegeneration
2807	Papilloma of choroid plexus
678	Papillon-Lefèvre syndrome
684	Paramyotonia congenita of Von Eulenburg
31827	Paraquat poisoning
90307	Parkes Weber syndrome
447	Paroxysmal nocturnal hemoglobinuria
1330	Partial atrioventricular canal
98992	Partial congenital cataract
706	Patent arterial duct
93566	Pediatric Sjögren syndrome
93552	Pediatric systemic lupus erythematosus
93567	Pediatric systemic sclerosis
705	Pendred syndrome
99885	Permanent neonatal diabetes mellitus
79189	Peroxisome biogenesis disorder-Zellweger syndrome spectrum
708	Peters anomaly
709	Peters-plus syndrome
2869	Peutz-Jeghers syndrome
716	Phenylketonuria
2896	Pitt-Hopkins syndrome
99408	Pituitary adenoma
85166	Platyspondylic dysplasia, Torrance type
64742	Pleuropulmonary blastoma
2905	POEMS syndrome
2911	Poland syndrome
732	Polymyositis
79358	Porokeratosis
738	Porphyria
101330	Porphyria cutanea tarda
79473	Porphyria variegata
854	Portal vein thrombosis
2918	Postaxial polydactyly
98993	Posterior polar cataract
98913	Postsynaptic congenital myasthenic syndromes
52022	Potocki-Shaffer syndrome
739	Prader-Willi syndrome
2922	Preaxial polydactyly
99860	Precursor B-cell acute lymphoblastic leukemia
186	Primary biliary cirrhosis
46135	Primary central nervous system lymphoma
244	Primary ciliary dyskinesia
541	Primary cutaneous CD30+ T-cell lymphoproliferative disease
542	Primary cutaneous lymphoma
101997	Primary immunodeficiency
90023	Primary immunodeficiency syndrome due to p14 deficiency
35689	Primary lateral sclerosis
168803	Primary peritoneal tumor
2420	Primary pulmonary lymphoma
171	Primary sclerosing cholangitis
1871	Progressive cone dystrophy
172	Progressive familial intrahepatic cholestasis
2762	Progressive osseous heteroplasia
2966	Properdin deficiency
35	Propionic acidemia
606	Proximal myotonic myopathy
70	Proximal spinal muscular atrophy
756	Pseudohypoaldosteronism type 1
757	Pseudohypoaldosteronism type 2
758	Pseudoxanthoma elasticum
182090	Pulmonary arterial hypertension
98984	Pulverulent cataract
69084	Pure hair and nail ectodermal dysplasia
760	Purine nucleoside phosphorylase deficiency
764	Pyomyositis
207119	Qualitative or quantitative defects of FKRP
3025	Radial ray agenesis
3022	Rapp-Hodgkin syndrome
102002	Rare ataxia
93419	Rare bone disease
180250	Rare breast tumor
101945	Rare bronchopulmonary tumor
218436	Rare cardiac rythm disease
71281	Rare central nervous system and retinal vascular disease
183651	Rare constitutional anemia
68383	Rare constitutional medullar aplasia
98059	Rare digestive tumor
280275	Rare disease
101953	Rare dyslipidemia
68363	Rare dystonia
101998	Rare epilepsy
97966	Rare eye disease
96210	Rare genetic deafness
183625	Rare genetic diabetes mellitus
98053	Rare genetic disease
101435	Rare genetic eye disease
158300	Rare genetic hematologic disease
183757	Rare genetic intellectual deficit
98056	Rare genetic renal disease
97992	Rare hematologic disease
248308	Rare hemorrhagic disorder
248315	Rare hemorrhagic disorder due to a coagulation factors defect
68334	Rare hemorrhagic disorder due to a constitutional coagulation factors defect

ORPHA Number	Disease name
71202	Rare hemorrhagic disorder due to a constitutional platelet anomaly
275729	Rare hemorrhagic disorder due to a constitutional thrombocytopenia
101943	Rare hepatic and biliary tract tumor
220489	Rare hereditary hemochromatosis
217454	Rare hereditary thrombophilia
104012	Rare inflammatory bowel disease
87277	Rare intellectual deficit
102003	Rare movement disorder
182064	Rare neuroinflammatory or neuroimmunological disease
98026	Rare odontologic disease
213500	Rare ovarian cancer
68402	Rare parkinsonian disorder
181415	Rare primary hyperaldosteronism
101944	Rare pulmonary disease
71198	Rare pulmonary hypertension
93626	Rare renal disease
93603	Rare renal tubular disease
93619	Rare renal tumor
280342	Rare rheumatological disease of childhood
71209	Rare soft tissue tumor
98057	Rare tumor
180151	Rare vaginal malformation
101938	Rare vascular liver disease
94125	Recessive mitochondrial ataxic syndrome
461	Recessive X-linked ichthyosis
169142	Recurrent infection due to specific granule deficiency
183675	Recurrent infections associated with rare immunoglobulin isotypes deficiency
169467	Recurrent Neisseria infections due to factor D deficiency
60032	Recurrent respiratory papillomatosis
97239	Reducing body myopathy
773	Refsum disease
774	Rendu-Osler-Weber disease
71862	Retinal dystrophy
791	Retinitis pigmentosa
52427	Retinitis punctata albescens
790	Retinoblastoma
90050	Retinopathy of prematurity
139455	Retinopathy, Burgess-Black type
778	Rett syndrome
69077	Rhabdoid tumor
284130	Rheumatoid arthritis
177	Rhizomelic chondrodysplasia punctata
59315	Rhombencephalosynapsis
97244	Rigid spine syndrome
97238	Rippling muscle disease
3103	Roberts syndrome
97360	Robinow syndrome
101016	Romano-Ward syndrome
2909	Rothmund-Thomson syndrome
783	Rubinstein-Taybi syndrome
797	Sarcoidosis
85146	Scapuloperoneal amyotrophy
1830	Schimke immuno-osseous dysplasia
799	Schizencephaly
801	Scleroderma
3152	Sclerosteosis
841	Sebocystomatosis
70595	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis
3157	Septo-optic dysplasia
250805	Serpinopathy
183660	Severe combined immunodeficiency
277	Severe combined immunodeficiency due to adenosine deaminase deficiency
169160	Severe combined immunodeficiency T- B+ due to CD3delta/CD3epsilon/CD3zeta
169157	Severe combined immunodeficiency T- B+ due to CD45 deficiency
169154	Severe combined immunodeficiency T- B+ due to IL-7Ralpha deficiency
35078	Severe combined immunodeficiency T- B+ due to JAK3 deficiency
171430	Severe congenital nemaline myopathy
42738	Severe congenital neutropenia
83618	Severe dilated cardiomyopathy due to lamin A/C mutation
169095	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy
3162	Sezary's syndrome
1147	Sheldon-Hall syndrome
35123	Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency
140941	Short stature due to primary acid-labile subunit deficiency
314795	Shox-related short stature
811	Shwachman-Diamond syndrome
3166	Sialuria
166282	Sick sinus syndrome
232	Sickle cell anemia
275752	Sickle cell disease and related diseases
251355	Sickle cell disease associated with an other hemoglobin anomaly
158014	Sinus histiocytosis with massive lymphadenopathy
93974	Smith-Fineman-Myers syndrome
818	Smith-Lemli-Opitz syndrome
819	Smith-Magenis syndrome
820	Sneddon syndrome
3394	Soft tissue sarcoma
59181	Sorsby's fundus dystrophy
821	Sotos syndrome
98920	Spinal muscular atrophy with respiratory distress
64753	Spinocerebellar ataxia with axonal neuropathy type 2
86854	Splenic marginal zone lymphoma
2440	Split hand-split foot malformation
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type
29822	Spontaneous periodic hypothermia
84271	Sporadic idiopathic steroid-resistant nephrotic syndrome
93220	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis
827	Stargardt disease
273	Steinert myotonic dystrophy
36426	Stevens-Johnson syndrome
828	Stickler syndrome
455	Superficial epidermolytic ichthyosis
169085	Susceptibility to respiratory infections associated with CD8alpha chain mutation
178364	Syndromic microphthalmia type 5
188	Systemic capillary leak syndrome
536	Systemic lupus erythematosus
2467	Systemic mastocytosis
240266	Systemic non-Langerhans cell histiocytosis
90291	Systemic sclerosis
883	Teratoma
842	Testicular seminoma
3000	Testotoxicosis
3303	Tetralogy of Fallot
1860	Thanatophoric dwarfism type I
86846	Therapy related acute myeloid leukemia and myelodysplastic syndrome
614	Thomsen and Becker disease
496	Thost-Unna palmoplantar keratoderma
93573	Thrombotic microangiopathy
54057	Thrombotic thrombocytopenic purpura
99867	Thymoma
100087	Thyroid tumor
79102	Thyrotoxic periodic paralysis
3329	Tibial aplasia - ectrodactyly
609	Tibial muscular dystrophy
42665	Tietz syndrome
65283	Timothy syndrome
95455	Toxic epidermal necrolysis
859	Transcobalamin II deficiency
99886	Transient neonatal diabetes mellitus
56970	Transmissible spongiform encephalopathies
85451	Transthyretin-related familial amyloid cardiomyopathy
32960	TRAPS syndrome

ORPHA Number	Disease name
861	Treacher-Collins syndrome
863	Trichinellosis
2947	Triphalangeal thumbs - brachyectrodactyly
3378	Trisomy 13
3380	Trisomy 18
88629	Tritanopia
3384	Truncus arteriosus
805	Tuberous sclerosis
182130	Tumor of endocrine glands
68347	Tumor of hematopoietic and lymphoid tissues
53715	Tumoral calcinosis
881	Turner syndrome
90038	Typical hemolytic uremic syndrome
171436	Typical nemaline myopathy
308	Unverricht-Lundborg disease
83001	Urogenital tract malformation
886	Usher syndrome
180062	Uterovaginal malformation
39044	Uveal melanoma
178338	UV-sensitive syndrome
211237	Vascular tumor
52759	Vasculitis
1480	Ventricular septal defect
28	Vitamin B12-responsive methylmalonic acidemia
79310	Vitamin B12-responsive methylmalonic acidemia type cblA
79311	Vitamin B12-responsive methylmalonic acidemia type cblB
27	Vitamin B12-unresponsive methylmalonic acidemia
892	Von Hippel-Lindau disease
903	Von Willebrand disease
894	Waardenburg syndrome type 1
895	Waardenburg syndrome type 2
896	Waardenburg syndrome type 3
897	Waardenburg-Shah syndrome
893	WAGR syndrome
899	Walker-Warburg syndrome
3447	Weaver syndrome
900	Wegener granulomatosis
902	Werner syndrome
51636	WHIM syndrome
171723	White sponge nevus
904	Williams syndrome
905	Wilson disease
906	Wiskott-Aldrich syndrome
280	Wolf-Hirschhorn syndrome
3463	Wolfram syndrome
65282	Woolly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome
910	Xeroderma pigmentosum
43	X-linked adrenoleukodystrophy
47	X-linked agammaglobulinemia
88917	X-linked Alport syndrome
596	X-linked centronuclear myopathy
1497	X-linked complicated corpus callosum dysgenesis
306617	X-linked complicated spastic paraplegia type 1
52503	X-linked creatine transporter deficiency
98863	X-linked Emery-Dreifuss muscular dystrophy
101088	X-linked hyper-IgM syndrome
89936	X-linked hypophosphatemia
37042	X-linked immune dysregulation - polyendocrinopathy - enteropathy
2442	X-linked lymphoproliferative disease
25980	X-linked myopathy with excessive autophagy
178461	X-linked myopathy with postural muscle atrophy
54	X-linked recessive ocular albinism
792	X-linked retinoschisis
662	Yellow nail syndrome
876	Yolk sac tumor
2828	Young adult-onset Parkinsonism
98912	ZASP-related myofibrillar myopathy
912	Zellweger syndrome
98995	Zonular cataract

Summary

1- Distribution of registries by country

COUNTRY	REGIONAL	NATIONAL	EUROPEAN	GLOBAL	NOT DEFINED	TOTAL
AT - Austria	1	14	2	0	1	18
BE - Belgium	2	16	2	1	0	21
BG - Bulgaria	0	8	0	0	0	8
CH - Switzerland*	1	5	1	1	0	8
CY - Cyprus	0	1	0	0	0	1
CZ - Czech Republic	0	4	0	0	0	4
DE - Germany	9	64	25	7	0	105
DK - Denmark	1	3	0	0	0	4
EE - Estonia	0	2	0	0	0	2
ES - Spain	4	33	2	0	0	39
FI - Finland	0	7	0	0	0	7
FR - France	18	93	12	5	2	130
GR - Greece	0	2	0	0	0	2
HR - Croatia*	0	1	0	0	0	1
HU - Hungary	0	3	0	0	0	3
IE - Ireland	4	7	0	0	0	11
IL - Israel*	0	2	0	0	0	2
IS - Iceland*	0	2	0	0	0	2
IT - Italy	7	46	3	7	0	63
LT - Lithuania	0	1	0	0	0	1
LU - Luxembourg	0	1	0	0	0	1
LV - Latvia	0	1	0	0	0	1
MK - Republic of Macedonia*	0	1	0	0	0	1
MT - Malta	0	1	0	0	0	1
NL - Netherlands	1	11	3	6	0	21
NO - Norway*	0	4	3	0	0	7
PL - Poland	3	5	1	0	0	9
PT - Portugal	1	11	0	0	0	12
RO - Romania	0	2	0	0	0	2
RS - Serbia*	0	4	0	0	0	4
SE - Sweden	0	14	1	3	0	18
SI - Slovenia	0	2	0	0	0	2
SK - Slovakia	0	2	0	0	0	2
TR - Turkey*	0	4	0	0	0	4
UA - Ukraine*	0	1	0	0	0	1
UK - United Kingdom	13	45	7	5	0	70
TOTAL	65	423	62	35	3	588

*surrounding countries participating to the Orphanet consortium

2- Distribution of registries by coverage

COVERAGE	NUMBER OF REGISTRIES
Regional	65
National	423
European	62
Global	35
Not defined	3
TOTAL	588

3- Distribution of registries by affiliation

AFFILIATION	NUMBER OF REGISTRIES
Public	495
Private non-for-profit	44
Private for-profit	49
TOTAL	588

Distribution of registries by country

AT - AUSTRIA (18 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Austrian acromegaly registry	National	Public
Austrian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	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	Private for-profit
Austrian GIST registry	National	Private non-for-profit
Austrian Haemophilia Registry	National	Public
Austrian Huntington disease registry	National	Private for-profit
Austrian myeloma registry	National	Private non-for-profit
Austrian registry for inborn errors of metabolism	National	Public
Austrian severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
Children cardiology registry	National	Public
EB Registry	Not defined	Public
EMSA-SG: central patient registry of the European multiple system atrophy network	European	Public
ENRAH: European alternating hemiplegia and rare epilepsies registry in childhood	European	Private non-for-profit
Lung- and pleura cancer registry	National	Public
MDS: Austrian myelodysplastic syndromes patient registry	National	Public
Styrian registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public

BE - BELGIUM (21 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
ACROBEL: the Belgian registry on acromegaly, epidemiology and quality of care	National	Private for-profit
Antwerpen registry of congenital anomalies - contributes to the EUROCAT network	Regional	Private for-profit
Belgian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Belgian cystic fibrosis patient registry (BMR-RBM) - contributes to the EUROCARE CF and the ECFS registries	National	Public
Belgian familial adenomatous polyposis registry	National	Private non-for-profit
Belgian Neuromuscular Disease Registry	National	Private non-for-profit

Belgian patient database for Wilson disease - contributes to the EuroWilson registry	National	Public
Belgian patient registry for rare bleeding disorders - contributes to the RBDD international registry	National	Public
Belgian registry of primary immunodeficiencies - contributes to the ESID European registry	National	Private for-profit
Belgian rituximab therapy registry for immune anemia and thrombocytopenia	National	Public
Belgian severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public
Belgian sickle cell anemia registry	National	Public
Belgian systemic sclerosis cohort	National	Public
ENRAH: Belgian contribution to European registry for alternating hemiplegia in childhood	National	Private non-for-profit
EUNEFRON: registry of the European network for the study of orphan nephropathies	European	Public
EURECHINOREG: Belgian contribution to the European registry of human alveolar echinococcosis	National	Public
EUROGLYCANET - International patient registry and cohort for congenital disorders of glycosylation	European	Private for-profit
Haemoglobinopathies - database	National	Private for-profit
Hainault and Namur registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
LCH: Belgian Langerhans cell histiocytosis registry	National	Public
Pediatric granulomatous arthritis international registry	Global	Private for-profit

BG - BULGARIA (9 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Bulgaria - part of the TREAT-NMD network	National	Private for-profit
National registry of adult patients with chronic myeloid leukemia - BG	National	Public
National registry of patients with Gaucher disease - BG	National	Public
National registry of patients with mucopolysaccharidosis type II (MPS2) - BG	National	Public
National registry of patients with phenylketonuria - BG	National	Public
National registry of patients with primary immunodeficiencies (PID) - BG	National	Public
National registry of patients with thalassaemia major - BG	National	Public
National registry of patients with Wilson disease - BG	National	Public

CH - SWITZERLAND (8 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Switzerland - contributes to the TREAT-NMD network	National	Public
Perihilar Cholangiocarcinoma International Registry	Global	Public
PFAPA Registry: Periodic fever aphtous stomatitis, pharyngitis and adenopathy patient registry	European	Public
Swiss alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Swiss cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Swiss registries for Interstitial and Orphan Lung Diseases (SIOLD Registries)	National	Public
Swiss registry of biliary atresia - contributes to the EBAR registry	National	Public
Vaud registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public

CY - CYPRUS (1 registry)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Cyprian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

CZ - CZECH REPUBLIC (4 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Czech cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Czech severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
Duchenne and Becker muscular dystrophy patient registry in the Czech Republic and Slovakia - contributes to the TREAT-NMD network	National	Private for-profit
Spinal muscular atrophy patient registry in the Czech Republic - part of the TREAT-NMD network	National	Private for-profit

DE - Germany (105 registries)		
DE-ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
AID-NET : German registry for autoinflammatory syndromes (children)	National	Public
ALS registry Nordrhein-Westfalen	Regional	Public
ALS registry Rheinland-Pfalz	Regional	Public
ALS registry swabia	Regional	Public
Ataxia-Telangiectasia patient registry - contributes to the ESID Database	National	Public

CEDATA-GPGE: Registry of Children with Inflammatory Bowel Disease in Germany and Austria	European	Public
Central Cutaneous Lymphoma Registry	National	Public
Centre Saxony-Anhalt registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Child liver tumor registry	National	Public
Childrens IBD registry in Saxony	Regional	Public
CMMR: Central Malignant Melanoma Registry in Germany	National	Public
CompERA-XL: International, prospective registry for the documentation of first-line and maintenance therapy in patients with pulmonary hypertension	European	Public
Conn Registry: German registry of primary aldosteronism	National	Public
Core documentation of rheumatic children in Germany	National	Public
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	Global	Public
CURE-Net : National registry for congenital uro-rectal malformations	National	Public
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	European	Public
DÖSAK tumor registry for documentation of tumors of the face and jaws in Germany, austria and switzerland	European	Public
Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Austria and Germany - part of the TREAT-NMD network	National	Public
EBAR: European Biliary Atresia Registry	European	Public
EHDN: European Huntington's disease registry	European	Public
EHDN: neuroacanthocytosis patient registry	Global	Public
EKRS: Saarland Cancer Registry - contributes to the RARECARE Project	Regional	Private non-for-profit
ENETS: European Neuroendocrine Tumour Registry	European	Public
EPICURE-registry: European Epilepsy Brain patient Registry	European	Public
Epidemiological cancer registry Baden-Württemberg	Regional	Public
ESID: European registry of primary immunodeficiencies	Global	Public
EU-RHAB: European Rhabdoid Tumor Registry	European	Public
eurIPFreg: European idiopathic pulmonary fibrosis registry	European	Public
EURIPIDES: European Registry for ICD and CRT devices in pediatrics and adults with congenital heart disease	European	Private non-for-profit
EUROFA - EFACT: European Friedreich Ataxia Registry	European	Public
European Alport therapy registry	European	Public
European patient registry for intoxication type metabolic diseases (E-IMD)	European	Public
EUROSCA-R: European patient registry on spinocerebellar ataxias	European	Public
EUTOS: European chronic myeloid leukemia patient registry (collaboration between the European LeukemiaNet and Novartis Europe)	European	Public
FACE - National registry for Robin sequence	National	Public

GeNeMove: German database for wilson disease	National	Public
German acromegaly registry	National	Public
German adrenal tumors registry	National	Private non-for-profit
German AID (Autoinflammatory disorders) registry -subproject AID-NET	National	Public
German alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
German calciphylaxis registry	National	Public
German childhood cancer registry (Partner of ACCIS: Automated Childhood Cancer Information System)	National	Private non-for-profit
German cystic fibrosis registry - contributes to the EUROCARE CF registry	National	Public
German Epilepsy Registry	National	Public
German Fanconi anemia registry	National	Public
German gastrointestinal stromal tumor registry	National	Public
German Haemophilia Registry (DHR)	National	Public
German marginal zone lymphoma registry	National	Public
German mucopolysaccharidosis patient registry	National	Public
German multiple endocrine neoplasia type 1 (MEN 1) registry	National	Public
German national case collection of familial pancreatic cancer	National	Public
German paroxysmal nocturnal hemoglobinuria registry	National	Public
German pituitary tumors registry	National	Private non-for-profit
German registry for congenital heart defects - part of the competence network for congenital heart defects	National	Private non-for-profit
German registry for congenital thrombocytopenia	National	Public
German registry for Morbus Adamantiades-Behçet e.V.	National	Private non-for-profit
German registry for papulosis atrophicans maligna	National	Public
German registry for patients with pulmonary hypertension	National	Public
German registry of congenital dyserythropoietic anemias (CDA)	National	Public
German severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
German vasculitis registry	National	Public
GMALL-registry: registry for adult patients with acute lymphoblastic leukemia or related diseases	National	Public
GOLDnet: Registry for Diffus Parenchymal Lung Disease	National	Public
HepNet: German hepatocellular carcinoma (HCC) registry	National	Public
International pheochromocytoma and paraganglioma registry	Global	Public
INVM (Isolated Noncompaction of Ventricular Myocardium) registry	National	Private for-profit
Kids Lung Register: International register and biobank for rare lung diseases	European	Public
KINDLERNET: Central patient registry Kindler syndrome	European	Public

LBL Registry: Registry for children with lymphoblastic lymphoma	National	Public
LCH: German Langerhans cell histiocystosis registry	National	Private non-for-profit
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
MDS: German myelodysplastic syndromes patient registry	National	Public
MEFOPA: registry for patients with rare Mendelian forms of Parkinson's Disease	European	Public
National FKRP-patient registry Germany - part of the TREAT-NMD network	National	Public
National nephrogenic systemic fibrosis registry	National	Public
National registry for Blackfan-Diamond disease	National	Public
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	European	Public
NET-Registry: German neuroendocrine gastrointestinal tumors	National	Public
Neuromyelitis optica patient registry	National	Public
NHL-BFM Registry 2012: Registry of the NHL-BFM study group for all subtypes of Non-Hodgkin Lymphoma diagnosed in children and adolescents	European	Public
NIRK: national central registry for ichthyoses and related keratinization disorders	National	Public
NIRK: patient registry for autosomal recessive congenital ichthyosis	National	Public
NKR: German registry for adrenocortical carcinoma	National	Public
OSTEOPETR: International registry of patients suffering from osteopetrosis	Global	Public
Patient registry for primary hyperoxaluria - contributes to the OxalEurope-Network	National	Public
Patient registry of the German Network for Systemic Scleroderma	National	Public
PID-NET: National registry of primary immunodeficiencies	National	Public
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	European	Public
Prospective inception cohort for juvenile Systemic Sclerosis	Global	Public
PSHN Registry - Purpura Schoenlein-Henoch-Nephritis registry of the German society for paediatric nephrology (GPN)	National	Public
RAMEDIS : Rare Metabolic Diseases Database	National	Public

RegiSCAR: International registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples - patient registry	European	Public
Register for rare myeloproliferative neoplasms	European	Public
Registry for congenital melanocytic nevi and neurocutaneous melanocytosis	National	Private non-for-profit
Registry for Merkel Cell Carcinoma	National	Public
Registry for patients with mitochondrial diseases (mitoREGISTER) - subproject of mitoNET	National	Public
Registry for Patients with WT1 Mutation Associated Diseases	National	Public
Registry for rare tumor in children	National	Public
RetDis Database: clinical descriptions of patients and families with inherited eye diseases	European	Public
STER: FVII deficiency treatment international registry	Global	Public
Tumor Registry of Lymphatic Neoplasia (TNL-Registry): Epidemiological registry describing treatment reality and therapy modalities of patients with malignant Lymphatic Systemic Diseases (Non-Hodgkin`s Lymphoma, Chronic Lymphocytic Leukemia and Multiple Myeloma) requiring therapy	National	Private for-profit
Von Hippel-Lindau registry	Regional	Public

DK - DENMARK (4 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Danish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Danish malignant hyperthermia registry - contributes to the European Malignant Hyperthermia Group (EMHG)	National	Public
Funen county registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Mendelian cytogenetics network online database	National	Public

EE - ESTONIA (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Estonian cancer registry - contributes to the RARECARE project	National	Public
Estonian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public

ES - SPAIN (39 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Asturias registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Barcelona birth defects registry - contributes to the EUROCAT network	Regional	Public

Budd-Chiari syndrome (BCS) and hepatic vascular diseases registry	National	Public
Database of Thrombotic Microangiopathies and Dense Deposit Disease	National	Public
ERCUSYN: European registry on Cushing's syndrome	European	Public
Fanconi anemia patient registry database	National	Public
MOLDIAG-PACA: patient registry of pancreatic cancer	European	Public
REA: Acromegaly Spanish registry	National	Public
RECOMINA: patient registry of microscopic colitis and study of the environmental risk factors	National	Private non-for-profit
REDIP: Spanish registry of primary immunodeficiencies - contributes to the ESID European registry	National	Public
Registro MEN: Spanish registry of multiple endocrine neoplasia	National	Public
Registry for rare disorders in Extremadura (Spain)	Regional	Public
Registry of congenital anomalies of the Basque Country (Spain) - contributes to the EUROCAT network	Regional	Public
REHAP: Spanish Registry of Pulmonary Arterial Hypertension	National	Public
RETEGEP: Spanish Registry of Gastroenteropancreatic Endocrine Tumors	National	Private non-for-profit
RETEGEP: Spanish Registry of Gastroenteropancreatic Neuroendocrine Tumors	National	Private non-for-profit
REWBA: Spanish Registry of Wolfram, Bardet-Biedl and Alstrom syndromes	National	Public
Spanish alpha-1 antitrypsin deficiency registry (REDAAT) - contributes to the Alpha One International Registry (AIR)	National	Public
Spanish Familial Adenomatous Polyposis Registry	National	Public
Spanish Gaucher's disease registry	National	Private non-for-profit
Spanish Overgrowth Syndrome Registry	National	Public
Spanish patient registry for spinal muscular atrophy - part of the TREAT-NMD network	National	Public
Spanish patient registry of ataxias	National	Private non-for-profit
Spanish patient registry of hereditary angioedema	National	Public
Spanish patient registry of hereditary retinal dystrophy	National	Public
Spanish patient registry of McArdle disease	National	Public
Spanish patient registry of myelodysplasic syndromes	National	Public
Spanish patient registry of primary tubulopathies	National	Public
Spanish patient registry of rare diseases: multiple endocrine neoplasia, acromegaly and enteropancreatic endocrine tumors.	National	Public
Spanish patient registry of transmissible spongiform encephalopathies	National	Public
Spanish Registry of Ataxias and Spinocerebellar Neurodegenerative Diseases	National	Public
Spanish Registry of Cushing Syndrome	National	Private non-for-profit
Spanish registry of Duchenne muscular dystrophy - part of the TREAT-NMD network	National	Public

Spanish registry of metabolic hereditary diseases	National	Private non-for-profit
Spanish registry of patients with fragile X syndrome	National	Public
Spanish registry of POEMS syndrome patients (Osteosclerotic myeloma)	National	Public
Spanish Registry of Rare Diseases	National	Public
Spanish registry of renal hereditary diseases	National	Public
Spanish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

FI - FINLAND (7 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Finnish cancer registry - contributes to the RARECARE project	National	Public
Finnish Hematology Register and Biobank - FHRB	National	Public
Finnish IPF registry	National	Public
Finnish patient registry on Fabry disease	National	Public
Finnish TREAT-NMD Patient Registry	National	Private non-for-profit
Register of Congenital Malformations	National	Public
The Finnish Register of Visual Impairment	National	Private non-for-profit

FR - FRANCE (130 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Aquitaine registry of mesothelioma	Regional	Public
Auvergne registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Bas-Rhin registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Basse Normandie registry of hematological malignancies	Regional	Public
BLAU registry: French pediatric granulomatous arthritis registry	National	Public
Breast and other gynecological cancers registry of Côte-d'Or	Regional	Public
CEREDIH: French primary immunodeficiencies registry	National	Public
CoF-AT study: a French cohort on ataxia-telangiectasia	National	Public
Cohort of patients affected by Marfan or related syndrome	National	Public
Cohort of patients with hereditary dystrophies of retina	Not defined	Public
Côte d'Or registry of hematological malignancies	Regional	Public
Cystadane post marketing registry of patient with homocystinuria	European	Private for-profit
D[4]/Phenodent: French registry of patients affected by rare odontologic diseases	National	Public
Duchenne and Becker muscular dystrophy patient registry in France - part of the TREAT-NMD network	National	Public
EDMUS - NOMADMUS: French cohort of Devic's neuromyelitis optica and related neurological disorders	National	Public

EDMUS: European Database for Multiple Sclerosis and other related diseases	European	Public
EHN - EURO-HISTIO-NET: European registry of Langerhans Cell Histiocytosis	European	Public
ENET Registry: European Neuro-Endocrine Tumors Group	National	Public
EPI-EPNET: European hepatic and erythropoietic porphyrias registry	European	Public
EPIMAD: registry of chronic inflammatory intestine diseases in North-West	Regional	Public
Escort-Hu: European sickle cell disease cohort- hydroxyurea	European	Private for-profit
Establishment of children and adolescents cohort in Behcet disease in France	National	Public
EU-CHS: European central hypoventilation syndrome registry	European	Public
EURECHINOREG: European registry of alveolar echinococcosis	European	Public
European multicenters SCLS (systemic capillary leak syndromes) registry	European	Public
European prospective registry of children born to mothers affected by the antiphospholipids syndrome	European	Public
EUROTRAPS: European patient registry on TRAPS syndrome	European	Public
FranceCoag: French prospective cohort of patients affected with haemophilia or severe form of other hereditary hemorrhagic diseases except platelet disorders	National	Public
French acromegaly registry	National	Public
French addictive acute intoxications cohort	National	Public
French amyotrophic lateral sclerosis patient registry	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 bradykinic idiopathic angioneurotic edema and oestrogen-sensitive registry	National	Public
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	Public
French certified registry of patients affected by Gaucher disease	National	Public
French certified registry of patients affected by thalassemia	National	Public
French cohort creation in retinitis pigmentosa	National	Public
French cohort for auto-inflammatory diseases	National	Public
French cohort in genetic microcephalies	National	Public
French cohort in primary ciliary dyskinesia	National	Public
French cohort of acquired autoimmune haemolytic anemia	National	Public
French cohort of Castleman's disease	National	Public
French cohort of common variable immunodeficiency with hypogammaglobulinemia in adults (CVID)	National	Public

French cohort of focal dystonia famillies	National	Public
French cohort of idiopathic pulmonary fibrosis	National	Public
French cohort of inflammatory bowel disease (IBD)	National	Public
French cohort of rare diabetes (neonatal diabetes, monogenic, atypical and syndromic forms)	National	Public
French cohort of rhombencephalosynapsis	National	Public
French cohort of Usher syndrome	National	Public
French Cohorts in Sneddon syndrome and suspected Sneddon syndrome livedo	National	Public
French constitutive hematologic diseases registry	National	Public
French cystic fibrosis cohort and CFTR-RD cohort	National	Public
French cystic fibrosis patient registry	National	Public
French cystinosis registry	National	Public
French epidemiological registry of esophageal atresia	National	Public
French familial cardiac malformations registry	National	Public
French national patients registry on rare peritoneal tumor RENAPE	Not defined	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 pediatric registry of rituximab treated patients affected by severe systemic diseases - contributes to the French AIR registry	National	Public
French prospective follow-up cohort of child affected by autoimmune haemolytic anemia (AHAI), Evans syndrome and thrombocytopenic autoimmune purpura (ATP)	National	Public
French register of the SDH-related hereditary paraglioma	National	Public
French registry for macrophagic myofasciitis	National	Public
French registry for right arrythmogenic ventricular dysplasia (ARVC/D)	National	Public
French registry of autosomal recessive polycystic kidney disease	National	Public
French registry of cases of spontaneous periodic hypothermia	National	Public
French registry of child atypical hemolytic uremic syndrome	National	Public
French registry of child hematological malignancies	National	Public
French registry of children solid tumors	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 patients affect by Leber amaurosis and retinitis pigmentosa to assess the clinical trial in gene therapy	National	Public
French registry of rare genetic metabolism disorders of steroids - contributing to the international RGSDC registry	National	Public
French registry of rare hypersomnias	National	Public
French registry of rare pulmonary hypertension (HTAP)	National	Public
French registry of tetrahydrobiopterin deficiencies	National	Public
French severe chronic neutropenia certified patient registry - contributes to the SCN international registry (SCNIR)	National	Public
French sickle cell anemia registry	National	Public
French Still disease patient registry	National	Public
French Williams syndrome cohort	National	Private for-profit
FROG: FRench Observatory on Gaucher disease	National	Private for-profit
GENEPSO: French epidemiological cohort of BRCA systemic mutations carriers	National	Public
Gironde registry of hematological malignancies	Regional	Public
GMF: French registry of myelodysplastic syndromes and leukemia chemo- and radio-induced	National	Public
GTE: French registry of endocrine tumors	National	Public
Idiopathic pulmonary fibrosis: Cohort studies for evaluation of pronostic factors, therapeutic evaluation	National	Public
Infevers : The registry of Hereditary Auto-inflammatory Disorders Mutations	Global	Public
ITINERAIR-HTAP: French cohort of adult with pulmonary arterial hypertension	National	Private for-profit
ITINERAIR-pediatrie: French cohort of children with pulmonary arterial hypertension	National	Private for-profit
ITINERAIR-scleroderma: French pulmonary arterial hypertension screening cohort of patients with scleroderma	National	Private for-profit
KAWA-NET: epidemiological database of Kawasaki disease in France	National	Public
LEA: children and adolescents with acute leukemia : propective cohort in France	National	Public
Left ventricular noncompaction French registry	National	Public
Mesothelioma cohort in Seine Saint-Denis and Val de Marne	Regional	Public
Myotonic dystrophy patient registry in France - part of the TREAT-NMD network	National	Public
National database for the study and follow-up of paediatric rare tumors.	National	Public
Paris registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public

PGRx : Immune thrombocytopenic purpura (ITP)	Global	Private non-for-profit
PGRx : Lupus	Global	Private non-for-profit
PGRx : Myositis	Global	Private non-for-profit
PGRx: Suspected rheumatoid arthritis	Global	Private non-for-profit
PHA1-NET: PseudoHypoAldosteronism type 1 cohort	National	Public
POLA: French patient registry of high level oligodendroglioma	National	Public
Primary central nervous system tumors registry of Gironde	Regional	Public
Regional registry of thyroid cancers in Rhône-Alpes	Regional	Public
Registre multicentrique à vocation nationale des mésothéliomes pleuraux (registre qualifié)	National	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 (province of France)	Regional	Public
Registry of heriditary Haemochromatosis of the Languedoc Roussillon (including rare forms non-HFE hemochromatosis)	Regional	Public
Registry of observed trichinellosis cases in France yearly	National	Public
Registry of the network studying thrombotic microangiopathies	National	Public
Rhône-Alpes registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Rhône-Alpes registry of systemic mastocytosis	Regional	Public
SYRENE: Rett syndrome network - French database of clinical and genetic aspects of Rett syndrome	National	Public
VALID: cohort creation on Budd-Chiari syndrome, hepatic venooclusive disease, hepatoportal sclerosis and portal vein thrombosis	European	Public
Vedrop registry of chronic cholestasis patient with vitamin E deficiency	European	Private for-profit

GR - GREECE (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Greek cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Greek severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Public

HR - CROATIA (1 registry)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Croatian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

HU - HUNGARY (3 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Hungary - contributes to the TREAT-NMD network	National	Private for-profit
Hungarian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Hungarian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

IE - IRELAND (11 registries)		
ENGLISH LABEL OF THE ACTIVITY	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
Galway registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Irish myelodysplastic syndromes specific registry	National	Public
Irish registry for Bernard-Soulier syndrome	National	Public
Irish registry of amyotrophic lateral sclerosis and motor neurone disease	National	Public
Irish registry of Hurler syndrome	National	Public
Irish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
National Irish alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Private non-for-profit
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
South of Ireland registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia

IL - ISRAEL (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Israeli cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Israelian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

IS - ICELAND (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Icelander cancer registry - contributes to the RARECARE project	National	Public
Icelander cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

IT - ITALY (63 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
AICE: Italian registry of hemophilia centre	National	Public
AIR: Italian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry	National	Public
Arrhythmogenic right ventricular cardiomyopathy/ dysplasia: clinical registry and database, evaluation of therapies	National	Public
Campania registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Italy - contributes to the TREAT-NMD network	National	Private non-for-profit
Enroll-HD - A Prospective Registry Study in a Global HD Cohort	Global	Public
EUROFEVER: European registry for autoinflammatory diseases	Global	Public
EURO-WABB: Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes	European	Public
EuroWilson: Registry and network tio improve the management of Wilson Disease	European	Public
FMF: Italian registry for familial mediterranean fever in the young	National	Public
Friedreich's ataxia Italian patient registry	National	Private non-for-profit
GLATIT: Glanzmann thrombasthenia Italian registry	National	Public
HAE-registry: European hereditary angioedema patient registry	European	Public
IBAHC: Italian registry for alternating hemiplegia of childhood	National	Private non-for-profit
International registry of bone fragility fractures in the young	Global	Public
International Registry of congenital dyserythropoietic anemia II	Global	Public
International Registry of Rare Bleeding Disorders (RBDD)	Global	Private non-for-profit
International registry of recurrent and familial hemolytic uremic syndrome/thrombotic thrombocytopenic purpura	Global	Private non-for-profit
IPERN: Registro di pazienti affetti da atrite giovanile idiopatica trattati con anti-TNF	National	Public
ISMAC: Sicilian registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public

Italian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Italian genetic movement disorders registry	National	Public
Italian HLH Registry	National	Public
Italian LCH Registry	National	Public
Italian Li-Fraumeni syndrome registry	National	Public
Italian neuroblastoma registry	National	Public
Italian registry for Cri du Chat syndrome (monosomy 5p)	National	Public
Italian registry for hereditary multiple exostoses	National	Public
Italian registry for MYH9-related thrombocytopenia	National	Public
Italian registry for patients with Shwachman Diamond Syndrome	National	Public
Italian registry of adult patients affected by familial mediterranean fever	National	Public
Italian registry of congenital nephrotic syndromes	National	Public
Italian registry of Creutzfeldt-Jakob disease and correlated syndromes	National	Public
Italian registry of diffuse infiltrative pneumopathies	National	Public
Italian registry of hemolytic uremic syndrome	National	Public
Italian registry of hypertrophic cardiomyopathy in Anderson-Fabry disease	National	Private non-for-profit
Italian registry of Legionellosis	National	Public
Italian registry of membranoproliferative glomerulonephritis	National	Private non-for-profit
Italian registry of muscle channel-diseases	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
MITOCON: National registry mitochondrial disease	National	Public
MODY: Italian registry of maturity onset diabetes of the young	National	Public
North-east Italy registry of neurofibromatosis	Regional	Public
North-East of Italy registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
RAM-NET: Italian registry of patients with neuromuscular diseases - contributes to Treat-NMD european network	National	Private non-for-profit
Regional registry for neuromuscular disorders	Regional	Public
Registry of inherited bleeding disorders in Emilia Romagna region	Regional	Public
Registry of pregnant patients affected by essential thrombocythemia	National	Public
RIAF: Fanconi's anemia Italian registry	National	Public
RIAT: Ataxia teleangiectasia Italian registry	National	Public
RIMM: Italian registry for myelofibrosis with myeloid metaplasia	National	Public

RISMD: Italian myelodysplastic syndromes registry	National	Private non-for-profit
RNIC: National Registry of Infant with Congenital Hypothyroidism	National	Public
SCNIR: Italian severe chronic neutropenia registry - contributes to the SCN international registry	National	Public
Telethon-UILDM registry CMD (Congenital Muscular Dystrophy) - contributes to european Treat-NMD network	National	Public
Telethon-UILDM registry FSHD (Facioscapulohumeral dystrophy) - contributes to european Treat-NMD network	National	Public
Telethon-UILDM registry LGMD (Limb Girdle Muscular Dystrophy) - contributes to european Treat-NMD network	National	Public
TTP: International registry on thrombotic thrombocytopenic purpura	Global	Private non-for-profit
Tuscany registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
V-RIAT: variant Ataxia telangiectasia Italian registry	National	Public

LT - LITHUANIA (1 registry)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Lithuanian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

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

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

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

MT - MALTA (1 registry)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Maltese cancer registry - contributes to the RARECARE project	National	Public

NL - NETHERLANDS (21 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
CONCOR: Dutch registry of patients with a congenital heart malformation	National	Private for-profit
DDRMD - Dutch Diagnosis Registration Metabolic Diseases	National	Public
Duchenne and Becker muscular dystrophy patient registry in the Netherlands - part of the TREAT-NMD network	National	Public
Dutch alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Dutch cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private non-for-profit
Dutch patient registry for Fabry disease	National	Public
Dutch patient registry for Gaucher disease	National	Public
Dutch patient registry for Niemann-Pick Disease Type B	National	Public
Dutch patient registry for Niemann-Pick Disease Type C	National	Public
Dutch severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
ECARUCA: cytogenetic and clinical database on rare chromosomal disorders	European	Public
EPCOT: European prospective cohort on thrombophilia	European	Private for-profit
International Dystrophic Epidermolysis Bullosa Patient Registry	Global	Public
Nephrotic syndrome registry	Global	Public
North Netherlands registry of congenital anomalies - contributes to the EUROCAT network	Regional	Public
PAN research: Prospective amyotrophic lateral sclerosis (ALS) study Netherlands	National	Public
The intenational Pompe registry	Global	Private for-profit
The International Collaborative Gaucher Group (ICGG) Gaucher registry	Global	Private for-profit
The international Fabry registry	Global	Private for-profit
The international Mps I registry	Global	Private for-profit
X-ALD: X-linked adrenoleukodystrophy database	European	Public

NO - NORWAY (7 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
EURADRENAL: European patient registry on autoimmune Addison's disease (sera, DNA and RNA)	European	Public
European Porphyria Registry (EPR)	European	Public
HUE-MAN patient registry on alpha mannosidosis	European	Public

Norwegian Porphyria Registry	National	Public
Norwegian cancer registry - contributes to the RARECARE project	National	Public
Norwegian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Norwegian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

PL - POLAND (9 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Cracow cancer registry - contributes to the RARECARE project	Regional	Public
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Poland - contributes to the TREAT-NMD network	National	Public
EHDN: Observational Study of the European Huntington's Disease Network	European	Public
Kielce 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 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
Warsaw cancer registry - contributes to the RARECARE project	Regional	Public

PT - PORTUGAL (12 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Adrenal tumor national registry	National	Public
Duchenne and Becker muscular dystrophy patient registry in Portugal - contributes to the TREAT-NMD network	National	Public
Pituitary tumors national registry	National	Public
Portuguese cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit
Portuguese Fabry registry	National	Public
Portuguese registry for alpha-1 antitrypsin deficiency	National	Private non-for-profit
Portuguese registry of biliary atresia - contributes to the EBAR registry	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

RO - ROMANIA (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Romanian biliary atresia registry	National	Public
Romanian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private non-for-profit

RS - SERBIA (4 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
Serbian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Serbian registry of hemophilia and von Willebrand disease patients	National	Public
Serbian registry of patients with rare bleeding disorders - contributes to the RBDD international registry	National	Public
Serbian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

SE - SWEDEN (18 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COVERAGE**	**AFFILIATION**
FOS : Fabry Outcome Survey	Global	Private for-profit
HOS : Hunter Outcome Survey	Global	Private for-profit
IOS : Icatibant Outcome Survey for hereditory angioedema	Global	Private for-profit
National registry on bronchopulmonary dysplasia	National	Public
SPAHR: Swedish Pulmonary Arterial Hypertension Registry	National	Public
SWEDCON: Swedish Registry of Congenital Heart Disease	National	Public
Swedish Acute Lymphoblastic Leukemia Registry	National	Public
Swedish Acute Myelogenous Leukemia Registry	National	Public
Swedish alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
Swedish and Finnish registry of CADASIL patients	European	Public
Swedish Childhood Cancer Registry	National	Public
Swedish Chronic Myeloid Leukemia Registry	National	Public
Swedish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Swedish Multiple Myeloma Registry	National	Public
Swedish Polyposis Registry	National	Public

Swedish Registry for Familial Amyloid Polyneuropathy	National	Public
Swedish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
SWEDROP: Swedish Registry for Retinopathy of Prematurity	National	Private for-profit

SI - SLOVENIA (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Slovenian cancer registry - contributes to the RARECARE project	National	Private for-profit
Slovenian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public

SK - SLOVAKIA (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Slovakian cancer registry - contributes to the RARECARE project	National	Public
Slovakian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Private for-profit

TR - TURKEY (4 registries)		
ENGLISH LABEL OF THE ACTIVITY	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	Private for-profit
Turkish cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Public
Turkish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public

UA - UKRAINE (1 registry)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
Spinal muscular atrophy patient registry in Ukraine - part of the TREAT-NMD network	National	Private for-profit

UK - UNITED KINGDOM (70 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	AFFILIATION
AOMIC: adult onset myositis immunogenetic collaboration	National	Public
Batten Disease Neuronal Ceroid Lipofuscinosis (NCL) Patient Registry	National	Public

BPOLD: British Paediatric Orphan Lung Disease Registry	National	Public
CARIS - Welsh registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public
CAROBB - congenital anomalies registry for Oxfordshire, Berkshire & Buckinghamshire - part of the BINOCAR and EUROCAT network	Regional	Public
CRANE: patients registry with cleft lip and/or cleft palate in England and Wales	National	Public
Donors of rare blood types database (part of ENERCA)	Global	Public
DRN 377: Clinical Register for Transient Neonatal Diabetes	National	Public
DYSCERNE's dysmorphology diagnostic system (DDS)	European	Public
EBV associated NK/T cell malignancies registry	National	Public
EHDN: registry of juvenile Huntington's disease	Global	Public
EMSYCAR - East Midlands & South Yorkshire congenital anomalies registry - part of BINOCAR and EUROCAT network	Regional	Public
English alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Public
English central hypoventilation syndrome registry - will contribute to the European CHS	National	Public
English cystic fibrosis database	National	Public
English cystic fibrosis patient registry - contributes to the EUROCARE CF and ECFS registries	National	Public
English cystinosis registry	National	Public
English hereditary angioedema patient registry - part of the HAE European registry	National	Public
English hyperoxaluria registry	National	Public
English juvenile dermatomyositis registry and repository	National	Public
English mucopolysaccharidosis registry	National	Public
English phenylketonuria registry	National	Public
English registry for lymphangioleiomyomatosis	National	Public
English registry of biliary atresia - contributes to the EBAR registry	National	Public
English registry of syndromes with abnormal vertebral segmentation	National	Public
English registry of Wolf-Hirschhorn syndrome	National	Public
English severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Public
EUMDS: European Registry for Myelodysplastic Syndromes - part of EuroLeukemiaNet (ELN)	European	Public
EURODSD: European disorders of sexual development registry (FINISHED)	European	Public
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	European	Public
European Prader-Willi syndrome database	European	Public
EURO-WABB: European patient-based data collection registry on rare diabetes syndromes (RDS)	European	Public

EUROWILSON: European clinical database for Wilson disease	European	Public
Familial Ovarian Cancer Register (FOCR)	National	Public
Glasgow registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public
Global FKRP (Fukutin-Related Protein) defects registry - part of TREAT-NMD Alliance	Global	Public
Great Ormond Street Hospital Congenital Melanocytic Naevus Registry	National	Public
Hunter Outcome Survey (HOS): patient registry	Global	Public
I-DSD: European disorders of sexual development registry	Global	Public
LCH: English Langerhans cell histiocytosis registry	National	Public
Merseyside and Cheshire registry of congenital anomalies -part of BINOCAR and EUROCAT network	Regional	Public
Myotonic dystrophy patient registry in United Kingdom - part of the TREAT-NMD network	National	Public
National Congenital Anomaly System (NCAS) - part of BINOCAR and EUROCAT network	Regional	Public
NDSCR - National Down syndrome cytogenetic registry - part of BINOCAR and EUROCAT network	National	Public
NHD: the national haemophilia database	National	Private non-for-profit
NHR: National Haemoglobinopathy Registry	National	Private for-profit
NorCAS - Northern registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public
Regional spinocerebellar ataxia registry	Regional	Public
SCAR - Scottish registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public
SWCAR - South West congenital anomalies registry - part of BINOCAR and EUROCAT network	Regional	Public
The Alström syndrome UK (ASUK) Clinical Research Database	National	Public
The National Chronic Granulomatous Disease Registry	National	Public
The regional paediatric cardiology database	Regional	Public
UK & Ireland Fanconi Anaemia Registry	National	Public
UK and Ireland Duchenne and Becker muscular dystrophy patient registry (part of the TREAT-NMD network)	National	Private non-for-profit
UK and Ireland Spinal muscular atrophy (SMA) patient registry (part of the TREAT-NMD network)	National	Public
UK Dyskeratosis Congenita (DC) registry	National	Public
UK Huntington disease registry (collaborating with the EHDN/Euro HD Registry)	National	Public
UK Myotonic dystrophy type I patient registry (part of the TREAT-NMD network)	National	Public
UK Neurofibromatosis 2 (NF2) Patient Registry	National	Public
UK Paediatric ITP (Immune Thrombocytopenic Purpura) Registry	National	Public
UK renal rare disease registry	National	Private for-profit

UK Thrombotic Thrombocytopenia Purpura (UKTTP) Registry	National	Public
UKAITPR: United Kingdom adult idiopathic thrombocytopenic purpura registry	National	Public
UKCCCR: English familial ovarian cancer patient registry	National	Public
UKESR: United Kingdom Evans Syndrome Registry	National	Public
UKFITPR: United Kingdom familial idiopathic thrombocytopenic purpura (ITP) Registry	National	Public
United Kingdom neuromyelitis optica registry	National	Public
WANDA - Wessex registry of antenatally detected anomalies - part of BINOCAR and EUROCAT network	Regional	Public
West Midlands registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Public

European registries

EUROPEAN REGISTRIES (62 registries)		
ENGLISH LABEL OF THE ACTIVITY	**COORDINATION**	**AFFILIATION**
CEDATA-GPGE: Registry of Children with Inflammatory Bowel Disease in Germany and Austria	DE	Public
CompERA-XL: International, prospective registry for the documentation of first-line and maintenance therapy in patients with pulmonary hypertension	DE	Public
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	DE	Public
Cystadane post marketing registry of patient with homocystinuria	FR	Private for-profit
DÖSAK tumor registry for documentation of tumors of the face and jaws in germany, austria and switzerland	DE	Public
DYSCERNE's dysmorphology diagnostic system (DDS)	GB	Public
EBAR: European Biliary Atresia Registry	DE	Public
ECARUCA: cytogenetic and clinical database on rare chromosomal disorders	NL	Public
EDMUS: European Database for Multiple Sclerosis and other related diseases	FR	Public
EHDN: European Huntington's disease registry	DE	Public
EHDN: Observational Study of the European Huntington's Disease Network	PL	Public

EHN - EURO-HISTIO-NET: European registry of Langerhans Cell Histiocytosis	FR	Public
EMSA-SG: central patient registry of the European multiple system atrophy network	AT	Public
ENETS: European Neuroendocrine Tumour Registry	DE	Public
ENRAH: European alternating hemiplegia and rare epilepsies registry in childhood	AT	Private non-for-profit
EPCOT: European prospective cohort on thrombophilia	NL	Private for-profit
EPICURE-registry: European Epilepsy Brain patient Registry	DE	Public
EPI-EPNET: European hepatic and erythropoietic porphyrias registry	FR	Public
ERCUSYN: European registry on Cushing's syndrome	ES	Public
Escort-Hu: European sickle cell disease cohort- hydroxyurea	FR	Private for-profit
EU-CHS: European central hypoventilation syndrome registry	FR	Public
EUMDS: European Registry for Myelodysplastic Syndromes - part of EuroLeukemiaNet (ELN)	GB	Public
EUNEFRON: registry of the European network for the study of orphan nephropathies	BE	Public
EURADRENAL: European patient registry on autoimmune Addison's disease (sera, DNA and RNA)	NO	Public
EURECHINOREG: European registry of alveolar echinococcosis	FR	Public
EU-RHAB: European Rhabdoid Tumor Registry	DE	Public
eurIPFreg: European idiopathic pulmonary fibrosis registry	DE	Public
EURIPIDES: European Registry for ICD and CRT devices in pediatrics and adults with congenital heart disease	DE	Private non-for-profit
EURODSD: European disorders of sexual development registry (FINISHED)	GB	Public
EUROFA - EFACT: European Friedreich Ataxia Registry	DE	Public
EUROGLYCANET - International patient registry and cohort for congenital disorders of glycosylation	BE	Private for-profit
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	GB	Public
European Alport therapy registry	DE	Public
European multicenters SCLS (systemic capillary leak syndromes) registry	FR	Public
European patient registry for intoxication type metabolic diseases (E-IMD)	DE	Public
European Porphyria Registry (EPR)	NO	Public
European Prader-Willi syndrome database	GB	Public
European prospective registry of children born to mothers affected by the antiphospholipids syndrome	FR	Public
EUROSCA-R: European patient registry on spinocerebellar ataxias	DE	Public
EUROTRAPS: European patient registry on TRAPS syndrome	FR	Public
EURO-WABB: European patient-based data collection registry on rare diabetes syndromes (RDS)	GB	Public

EURO-WABB: Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes	IT	Public
EUROWILSON: European clinical database for Wilson disease	GB	Public
EuroWilson: Registry and network tio improve the management of Wilson Disease	IT	Public
EUTOS: European chronic myeloid leukemia patient registry (collaboration between the European LeukemiaNet and Novartis Europe)	DE	Public
HAE-registry: European hereditary angioedema patient registry	IT	Public
HUE-MAN patient registry on alpha mannosidosis	NO	Public
Kids Lung Register: International register and biobank for rare lung diseases	DE	Public
KINDLERNET: Central patient registry Kindler syndrome	DE	Public
MEFOPA: registry for patients with rare Mendelian forms of Parkinson's Disease	DE	Public
MOLDIAG-PACA: patient registry of pancreatic cancer	ES	Public
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	DE	Public
NHL-BFM Registry 2012: Registry of the NHL-BFM study group for all subtypes of Non-Hodgkin Lymphoma diagnosed in children and adolescents	DE	Public
PFAPA Registry: Periodic fever aphtous stomatitis, pharyngitis and adenopathy patient registry	CH	Public
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	DE	Public
RegiSCAR: International registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples - patient registry	DE	Public
Register for rare myeloproliferative neoplasms	DE	Public
RetDis Database: clinical descriptions of patients and families with inherited eye diseases	DE	Public
Swedish and Finnish registry of CADASIL patients	SE	Public
VALID: cohort creation on Budd-Chiari syndrome, hepatic venooclusive disease, hepatoportal sclerosis and portal vein thrombosis	FR	Public
Vedrop registry of chronic cholestasis patient with vitamin E deficiency	FR	Private for-profit
X-ALD: X-linked adrenoleukodystrophy database	NL	Public

International registries

INTERNATIONAL REGISTRIES (43 registries)		
ENGLISH LABEL OF THE ACTIVITY	COORDINATION	AFFILIATION
CMDIR: congenital muscular dystrophy international registry	US	Private for-profit
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	DE	Public
Donors of rare blood types database (part of ENERCA)	GB	Public
EHDN: neuroacanthocytosis patient registry	DE	Public
EHDN: registry of juvenile Huntington's disease	GB	Public
Enroll-HD - A Prospective Registry Study in a Global HD Cohort	IT	Public
ESID: European registry of primary immunodeficiencies	DE	Public
EUROFEVER: European registry for autoinflammatory diseases	IT	Public
FARA: International Friedreich Ataxia Research Alliance registry	US	Private for-profit
FOS : Fabry Outcome Survey	SE	Private for-profit
Global FKRP (Fukutin-Related Protein) defects registry - part of TREAT-NMD Alliance	GB	Public
HOS : Hunter Outcome Survey	SE	Private for-profit
Hunter Outcome Survey (HOS): patient registry	GB	Public
I-DSD: European disorders of sexual development registry	GB	Public
IFAR: International Fanconi Anemia patient Registry	US	Public
Infevers : The registry of Hereditary Auto-inflammatory Disorders Mutations	FR	Public
International Dystrophic Epidermolysis Bullosa Patient Registry	NL	Public
International Morquio A registry	US	Private for-profit
International Pachyonychia Congenita Research Registry (IPCRR)	US	Private non-for-profit
International pheochromocytoma and paraganglioma registry	DE	Public
International Rare Genetic Steroid Disorders Consortium (RGSDC) registry	US	Private for-profit
International registry for primary hyperoxaluria	US	Private for-profit
International registry of bone fragility fractures in the young	IT	Public
International Registry of congenital dyserythropoietic anemia II	IT	Public
International Registry of Rare Bleeding Disorders (RBDD)	IT	Private non-for-profit
International registry of recurrent and familial hemolytic uremic syndrome/thrombotic thrombocytopenic purpura	IT	Private non-for-profit
IOS : Icatibant Outcome Survey for hereditory angioedema	SE	Private for-profit
Nephrotic syndrome registry	NL	Public
OSTEOPETR: International registry of patients suffering from osteopetrosis	DE	Public
Pediatric granulomatous arthritis international registry	BE	Private for-profit

Perihilar Cholangiocarcinoma International Registry	CH	Public
PGRx : Immune thrombocytopenic purpura (ITP)	FR	Private non-for-profit
PGRx : Lupus	FR	Private non-for-profit
PGRx : Myositis	FR	Private non-for-profit
PGRx: Suspected rheumatoid arthritis	FR	Private non-for-profit
Prospective inception cohort for juvenile Systemic Sclerosis	DE	Public
STER: FVII deficiency treatment international registry	DE	Public
THAOS: transthyretin amyloidosis outcomes survey	US	Private for-profit
The intenational Pompe registry	NL	Private for-profit
The International Collaborative Gaucher Group (ICGG) Gaucher registry	NL	Private for-profit
The international Fabry registry	NL	Private for-profit
The international Mps I registry	NL	Private for-profit
TTP: International registry on thrombotic thrombocytopenic purpura	IT	Private non-for-profit
THAOS: transthyretin amyloidosis outcomes survey	US	Industry

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