



Orphanet Report Series

Rare Diseases collection

January 2011

Disease Registries in Europe

www.orpha.net



Table of contents

Methodology	3
Summary	4
1- Distribution of registries by country	4
2- Distribution of registries by coverage	5
3- Distribution of registries by institution	5
4- Network registries	5
Distribution of registries by country	6
European registries	28
International registries	30
Network registries	31



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.

The concept of coverage (regional, national, european or international) that is associated with patient registries reflects the area of collection of a single physical database, to which several clinical entities are contributing.

The notion of networks corresponds to the relationship existing between several databases (or patient registries), whether a coordinating database exists or not.

Cancer registries are listed only if they belong to the network RARECARE or focus on a rare form of cancer. American registries are listed only if they cover the European area and focus on a rare disease uncollected in Europe.

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

Summary

1- Distribution of registries by country

COUNTRY	REGIONAL	NATIONAL	EUROPEAN	GLOBAL	NOT DEFINED	TOTAL
AT - Austria	1	12	2	0	0	15
BE - Belgium	2	16	1	1	0	20
BG - Bulgaria	0	4	0	0	0	4
CH - Switzerland	1	5	1	2	0	9
CY - Cyprus	0	1	0	0	0	1
CZ - Czech Republic	0	4	0	0	0	4
DE - Germany	4	57	20	5	0	86
DK - Denmark	1	3	0	0	0	4
EE - Estonia	0	2	0	0	0	2
ES - Spain	4	25	3	0	0	32
FI - Finland	0	5	0	0	0	5
FR - France	17	92	11	1	1	122
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	6	0	0	0	10
IL - Israel	0	2	0	0	0	2
IS - Iceland	0	2	0	0	0	2
IT - Italy	9	35	2	5	0	51
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	6	3	5	0	15
NO - Norway	0	3	1	0	0	4
PL - Poland	3	6	0	0	0	9
PT - Portugal	1	8	0	0	0	9
RO - Romania	0	2	0	0	0	2
RS - Serbia	0	4	0	0	0	4
SE - Sweden	0	15	0	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	38	6	1	0	58
US - United States of America	0	0	0	6	0	6
TOTAL	61	373	50	29	1	514

2- Distribution of registries by coverage

COVERAGE	NUMBER OF REGISTRIES
REGIONAL	61
NATIONAL	373
EUROPEAN	50
GLOBAL	29
NOT DEFINED	1
TOTAL	514

3- Distribution of registries by institution

INSTITUTION	NUMBER OF REGISTRIES
ACADEMIC	490
PATIENT ORGANISATION	8
PRIVATE COMPANY	16
TOTAL	514

4- Network registries

NETWORKS	NUMBER OF NETWORKS
TOTAL	9

Distribution of registries by country

AT - AUSTRIA (15 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Austrian acromegaly registry	National	Academia
Austrian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
Austrian brain tumor registry	National	Academia
Austrian cancer registry - contributes to the RARECARE project	National	Academia
Austrian chronic myeloid leukemia registry	National	Academia
Austrian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Academia
Austrian Haemophilia Registry	National	Academia
Austrian Huntington disease registry	National	Academia
Austrian myeloma registry	National	Academia
Austrian registry for inborn errors of metabolism	National	Academia
Austrian severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Academia
EMSA-SG: central patient registry of the European multiple system atrophy network	European	Academia
ENRAH: European alternating hemiplegia and rare epilepsies registry in childhood	European	Academia
MDS: Austrian myelodysplastic syndromes patient registry	National	Academia
Styrian registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia

BE - BELGIUM (20 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
ACROBEL: the Belgian registry on acromegaly, epidemiology and quality of care	National	Academia
Antwerpen registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Belgian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
Belgian cystic fibrosis patient registry (BMR-RBM) - contributes to the EUROCARE CF and the ECFS registries	National	Academia
Belgian familial adenomatous polyposis registry	National	Academia
Belgian Neuromuscular Disease Registry	National	Academia
Belgian patient database for Wilson disease - contributes to the EuroWilson registry	National	Academia
Belgian patient registry for Duchenne and Becker muscular dystrophy - part of the TREAT-NMD network	National	Academia

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

BG - BULGARIA (4 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Bulgarian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Bulgarian registry of patients with thalassaemia	National	Academia
Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Bulgaria - part of the TREAT-NMD network	National	Academia
The Bulgarian genetic registry of monogenic disorders	National	Academia

CH - SWITZERLAND (9 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
BH4: international patient registry of tetrahydrobiopterin deficiency	Global	Academia
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Switzerland - contributes to the TREAT-NMD network	National	Academia
PFAPA Registry: Periodic fever aphthous stomatitis, pharyngitis and adenopathy patient registry	European	Academia
PNH Registry: Paroxysmal Nocturnal Hemoglobinuria registry	Global	Industry
SIOLD: Swiss registry for Interstitial and Orphan Lung Diseases	National	Academia

Swiss alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
Swiss cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Academia
Swiss registry of biliary atresia - contributes to the EBAR registry	National	Academia
Vaud registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia

CY - CYPRUS (1 registry)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Cyprian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Academia

CZ - CZECH REPUBLIC (4 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Czech cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Academia
Czech severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
Duchenne and Becker muscular dystrophy patient registry in the Czech Republic and Slovakia - contributes to the TREAT-NMD network	National	Academia
Spinal muscular atrophy patient registry in the Czech Republic - part of the TREAT-NMD network	National	Academia

DE - GERMANY (86 registries)

DE-ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
AID-NET : Clinical Registry and Biobank (DNA/RNA/serum) for autoinflammatory syndromes (children)	National	Academia
Ataxia-Telangiectasia patient registry - contributes to the ESID Database	National	Academia
Central Cutaneous Lymphoma Registry	National	Academia
Centre Saxony-Anhalt registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
CMMR: Central Malignant Melanoma Registry in germany	National	Academia
CompERA-XL: International, prospective registry for the documentation of first-line and maintenance therapy in patients with pulmonary hypertension	European	Academia
Conn Registry: German registry of primary aldosteronism	National	Academia
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	Global	Academia
CURE-Net : National registry for congenital uro-rectal malformations	National	Academia

CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	European	Academia
DÖSAK tumor registry for documentation of tumors of the face and jaws in germany, austria and switzerland	European	Academia
Duchenne and Becker muscular dystrophy and spinal muscular atrophy patient registries in Austria and Germany - part of the TREAT-NMD network	National	Academia
EBAR: European Biliary Atresia Registry	European	Academia
EHDN: European Huntington's disease registry	European	Academia
EHDN: neuroacanthocytosis patient registry	Global	Academia
EKRS: Saarland Cancer Registry - contributes to the RARECARE Project	Regional	Academia
ENETS: European Neuroendocrine Tumour Registry	European	Academia
EPICURE-bank: European Epilepsy Brain Bank	European	Academia
ESID: European registry of primary immunodeficiencies	Global	Academia
EU-RHAB: European Rhabdoid Registry	European	Academia
EURIPFREG: European idiopathic pulmonary fibrosis registry	European	Academia
EURIPIDES: European Registry for ICD and CRT devices in pediatrics and adults with congenital heart disease	European	Academia
EUROFA: European Friedreich Ataxia Registry	European	Academia
European Alport registry	European	Academia
EUROSCA-R: European patient registry on spinocerebellar ataxias	European	Academia
EUTOS: European chronic myeloid leukemia patient registry (collaboration between the European LeukemiaNet and Novartis Europe)	European	Academia
GeNeMove: German database for wilson disease	National	Academia
German acromegaly registry	National	Academia
German adrenal tumors registry	National	Academia
German AID (Autoinflammatory disorders) registry -subproject AID-NET	National	Academia
German alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
German childhood cancer registry (Partner of ACCIS: Automated Childhood Cancer Information System)	National	Academia
German cystic fibrosis registry - contributes to the EURO CARE CF registry	National	Academia
German Epilepsy Registry	National	Academia
German Fanconi anemia registry	National	Academia
German gastrointestinal stromal tumor registry	National	Academia
German Haemophilia Registry (DHR)	National	Academia
German mucopolysaccharidosis patient registry	National	Academia
German multiple endocrine neoplasia type 1 (MEN 1) registry	National	Academia
German national case collection of familial pancreatic cancer	National	Academia
German paroxysmal nocturnal hemoglobinuria registry	National	Academia

German pituitary tumors registry	National	Academia
German registry for congenital heart defects - part of the competence network for congenital heart defects	National	Academia
German registry for congenital thrombocytopenia	National	Academia
German registry for Morbus Adamantiades-Behçet e.V.	National	Academia
German registry for papulosis atrophicans maligna	National	Academia
German registry for patients with pulmonary hypertension	National	Academia
German registry of congenital dyserythropoietic anemias (CDA)	National	Academia
German severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
German vasculitis registry	National	Academia
GMALL-registry and biobank: registry for adult patients with acute lymphoblastic leukemia or related diseases	National	Academia
GOLD.net : Registry and biobank for Diffus Parenchymal Lung Disease	National	Academia
HepNet: German hepatocellular carcinoma (HCC) registry	National	Academia
International pheochromocytoma and paraganglioma registry	Global	Academia
INVM (Isolated Noncompaction of Ventricular Myocardium) registry	National	Academia
KINDLERNET: Central patient registry Kindler syndrome	European	Academia
LCH: German Langerhans cell histiocytosis registry	National	Academia
Lupus nephritis registry (established by the german paediatric nephrology association)	National	Academia
Mainz registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
MAISTHRO-Registry : multicentric thrombophilia registry (MAIn-ISar-THROmbose-Register)	National	Academia
MDS: German myelodysplastic syndromes patient registry	National	Academia
MEFOPA: registry for patients with rare Mendelian forms of Parkinson's Disease	European	Academia
National FKRP-patient registry germany - part of the TREAT-NMD network	National	Academia
National nephrogenic systemic fibrosis registry	National	Academia
National PSHN registry (german society for paediatric nephrology GPN)	National	Academia
National registry for Blackfan-Diamond disease	National	Academia
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	European	Academia
NET-Registry: German neuroendocrine gastrointestinal tumors	National	Academia
Neuromyelitis optica patient registry	National	Academia
NIRK: national central registry for ichthyoses and related keratinization disorders	National	Academia
NIRK: patient registry for autosomal recessive congenital ichthyosis	National	Academia
NKR: German registry for adrenocortical carcinoma	National	Academia

Patient registry for primary hyperoxaluria	National	Academia
Patient registry of the German Network for Systemic Scleroderma	National	Academia
PID-NET: National registry of primary immunodeficiencies	National	Academia
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	European	Academia
RAMEDIS : Rare Metabolic Diseases Database	National	Academia
RegiSCAR: European registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples	European	Academia
Registry for children with congenital limb malformations	National	Academia
Registry for congenital melanocytic nevi and neurocutaneous melanocytosis	National	Academia
Registry for Merkel Cell Carcinoma	National	Academia
Registry for patients with mitochondrial diseases (mitoREGISTER) - subproject of mitoNET	National	Academia
Registry for Patients with WT1 Mutation Associated Diseases	National	Academia
RetDis Database (blood or DNA samples and associated clinical descriptions of patients and families with inherited eye diseases)	European	Academia
STER: FVII deficiency treatment international registry	Global	Academia
Von Hippel-Lindau registry	Regional	Academia

DK - DENMARK (4 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Danish cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Danish malignant hyperthermia registry - contributes to the European Malignant Hyperthermia Group (EMHG)	National	Academia
Funen county registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Mendelian cytogenetics network online database	National	Academia

EE - ESTONIA (2 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Estonian cancer registry - contributes to the RARECARE project	National	Academia
Estonian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

ES - SPAIN (32 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Asturias registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Barcelona birth defects registry - contributes to the EUROCAT network	Regional	Academia
Budd-Chiari syndrome (BCS) and hepatic vascular diseases registry	National	Academia
ERCUSYN: European registry on Cushing's syndrome	European	Academia
EUGINDAT-PIADATABASE: European primary inherited aminoacidurias database	European	Academia
GIRMOGEN_PRO Database: mental retardation with genetics etiology registry in Spain	National	Academia
MOLDIAG-PACA: patient registry of pancreatic cancer	European	Academia
REA: Acromegaly Spanish registry	National	Academia
RECOMINA: patient registry of microscopic colitis and study of the environmental risk factors	National	Academia
REDIP: Spanish registry of primary immunodeficiencies - contributes to the ESID European registry	National	Academia
Registry for rare disorders in Extremadura (Spain)	Regional	Academia
Registry of congenital anomalies of the Basque Country (Spain) - contributes to the EUROCAT network	Regional	Academia
REHAP: Spanish Registry of Pulmonary Arterial Hypertension	National	Academia
RETEGEP: Spanish Registry of Gastroenteropancreatic Endocrine Tumors	National	Academia
Spanish alpha-1 antitrypsin deficiency registry (REDAAT) - contributes to the Alpha One International Registry (AIR)	National	Academia
Spanish cystic fibrosis patient registry - contributes to the EUROCAT CF registry	National	Academia
Spanish Familial Adenomatous Polyposis Registry	National	Academia
Spanish Gaucher's disease registry	National	Academia
Spanish Overgrowth Syndrome Registry	National	Academia
Spanish patient registry for spinal muscular atrophy - part of the TREAT-NMD network	National	Academia
Spanish patient registry of hereditary angioedema	National	Academia
Spanish patient registry of hereditary retinal dystrophy	National	Academia
Spanish patient registry of myelodysplastic syndromes	National	Academia
Spanish patient registry of rare diseases: multiple endocrine neoplasia, acromegaly and enteropancreatic endocrine tumors.	National	Academia
Spanish patient registry of transmissible spongiform encephalopathies	National	Academia
Spanish registry of Duchenne muscular dystrophy - part of the TREAT-NMD network	National	Academia
Spanish registry of metabolic hereditary diseases	National	Academia
Spanish registry of patients with fragile X syndrome	National	Academia

Spanish registry of POEMS syndrome patients (Osteosclerotic myeloma)	National	Academia
Spanish Registry of Rare Diseases	National	Academia
Spanish registry of renal hereditary diseases	National	Academia
Spanish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia

FI - FINLAND (5 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Finland - contributes to the TREAT-NMD network	National	Patient organisation
Finnish cancer registry - contributes to the RARECARE project	National	Patient organisation
Finnish patient registry on Fabry disease	National	Academia
Finnish registry of congenital anomalies - contributes to the EUROCAT network	National	Academia
The Finnish registry of visual impairment	National	Academia

FR - FRANCE (122 registries)

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

EHN - EURO-HISTIO-NET: European registry of Langerhans Cell Histiocytosis	European	Academia
ENET Registry: European Neuro-Endocrine Tumors Group	National	Academia
EPI-EPNET: European hepatic and erythropoietic porphyrias registry	European	Academia
EPIMAD: registry of chronic inflammatory intestine diseases in North-West	Regional	Academia
Escort-Hu: European sickle cell disease cohort- hydroxyurea	European	Industry
Establishment of children and adolescents cohort in Behcet disease in France	National	Academia
EURECHINOREG: European registry of alveolar echinococcosis	European	Academia
European prospective registry of children born to mothers affected by the antiphospholipids syndrome	European	Academia
EUROTRAPS: European patient registry on TRAPS syndrome	European	Academia
FranceCoag: French prospective cohort of patients affected with haemophilia or severe form of other hereditary hemorrhagic diseases except platelet disorders	National	Academia
French acromegaly registry	National	Academia
French addictive acute intoxications cohort	National	Academia
French amyotrophic lateral sclerosis patient registry	National	Academia
French atypical sarcoidosis clinical forms registry	National	Academia
French auto-immunity and Rituximab (AIR) registry: prospective study of patients treated with Rituximab	National	Academia
French bradykinin idiopathic angioneurotic edema and oestrogen-sensitive registry	National	Academia
French central hypoventilation syndrome registry - will contribute to the European CHS registry	National	Academia
French certified patient registry for Langerhans cell histiocytosis and biological collection	National	Academia
French certified registry of glycogen storage disease type 2	National	Academia
French certified registry of patients affected by Gaucher disease	National	Academia
French certified registry of patients affected by thalassemia	National	Academia
French cohort creation in retinitis pigmentosa	National	Academia
French cohort for auto-inflammatory diseases	National	Academia
French cohort in genetic microcephalies	National	Academia
French cohort in primary ciliary dyskinesia	National	Academia
French cohort of acquired autoimmune haemolytic anemia	National	Academia
French cohort of Castleman's disease	National	Academia
French cohort of common variable immunodeficiency with hypogammaglobulinemia in adults (CVID)	National	Academia
French cohort of focal dystonia families	National	Academia
French cohort of idiopathic pulmonary fibrosis	National	Academia
French cohort of inflammatory bowel disease (IBD)	National	Academia
French cohort of rare diabetes (monogenic forms and syndromic forms)	National	Academia
French cohort of rhombencephalosynapsis	National	Academia

French cohort of Usher syndrome	National	Academia
French Cohorts in Sneddon syndrome and suspected Sneddon syndrome livedo	National	Academia
French constitutive hematologic diseases registry	National	Academia
French cystic fibrosis cohort	National	Academia
French cystic fibrosis patient registry	National	Patient organisation
French cystinosis registry	National	Academia
French epidemiological registry of esophageal atresia	National	Academia
French familial cardiac malformations registry	National	Academia
French observatory of biliary atresia	National	Academia
French observatory of gastric linitis plastica	National	Academia
French observatory of primary biliary cirrhosis	National	Academia
French observatory of primitive sclerosing cholangitis	National	Academia
French patient registry affected by genetic deafness in France	National	Academia
French Patient registry in chorioretinopathy, birdshot type	National	Academia
French pediatric registry of rituximab treated patients affected by severe systemic diseases - contributes to the French AIR registry	National	Academia
French prospective cohort of childhood care for autoimmune haemolytic anemia and Evans syndrome	National	Academia
French prospective follow-up cohort of child affected by autoimmune haemolytic anemia (AHAI), Evans syndrome and thrombocytopenic autoimmune purpura (ATP)	National	Academia
French registry for capillary leak syndromes	National	Academia
French registry for macrophagic myofasciitis	National	Academia
French registry for right arrhythmogenic ventricular dysplasia (ARVC/D)	National	Academia
French registry of autosomal recessive polycystic kidney disease	National	Academia
French registry of cases of spontaneous periodic hypothermia	National	Academia
French registry of child handicap and perinatal observatory	National	Academia
French registry of child hematological malignancies	National	Academia
French registry of child hemolytic uremic syndrome	National	Academia
French registry of children solid tumors	National	Academia
French registry of corticosteroid-sensitive aseptic abscess	National	Academia
French registry of familial and premature prostate cancers (before 50 years)	National	Academia
French registry of generalized resistance to thyroid hormone	National	Academia
French registry of hereditary dyslipidemia in children: familial combined dyslipidemias	National	Academia
French registry of Iron overload genetic rare diseases, non-related to the HFE gene	National	Academia
French registry of Kabuki syndrome	National	Academia

French registry of Marshall's syndrome with periodic fever	National	Academia
French registry of neuromuscular diseases from reference centres	National	Academia
French registry of patients affect by Leber amaurosis and retinitis pigmentosa to assess the clinical trial in gene therapy	National	Academia
French registry of pregnant women carriers of anti-SSA antibodies	National	Academia
French registry of rare genetic metabolism disorders of steroids - contributing to the international RGSDC registry	National	Academia
French registry of rare hypersomnias	National	Academia
French registry of rare pulmonary hypertension (HTAP)	National	Academia
French registry of tetrahydrobiopterin deficiencies	National	Academia
French severe chronic neutropenia certified patient registry - contributes to the SCN international registry (SCNIR)	National	Academia
French sickle cell anemia registry	National	Academia
French Still disease patient registry	National	Academia
French Williams syndrome cohort	National	Academia
FROG: FRENch Observatory on Gaucher disease	National	Industry
GENEPSO: French epidemiological cohort of BRCA systemic mutations carriers	National	Academia
Gironde registry of hematological malignancies	Regional	Academia
GMF: French registry of myelodysplastic syndromes and leukemia chemo- and radio-induced	National	Academia
GTE: French registry of endocrine tumors	National	Academia
Idiopathic pulmonary fibrosis: Cohort studies for evaluation of pronostic factors, therapeutic evaluation	National	Academia
INFEVERS: European registry of mutations involved in familial mediterranean fever (FMF) and hereditary autoinflammatory disorders	European	Academia
International FKR (Fukutin-Related Protein) defects registry - part of TREAT-NMD network	Global	Academia
ITINERAIR-HTAP: French cohort of adult with pulmonary arterial hypertension	National	Industry
ITINERAIR-pediatrie: French cohort of children with pulmonary arterial hypertension	National	Industry
ITINERAIR-scleroderma: French pulmonary arterial hypertension screening cohort of patients with scleroderma	National	Industry
LEA: children and adolescents with acute leukemia : propective cohort in France	National	Academia
Left ventricular noncompaction French registry	National	Academia
Mesothelioma cohort in Seine Saint-Denis and Val de Marne	Regional	Academia
Myotonic dystrophy patient registry in France - part of the TREAT-NMD network	National	Academia
Paris registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
PHA1-NET: PseudoHypoAldosteronism type 1 cohort	National	Academia
Primary central nervous system tumors registry of Gironde	Regional	Academia

Regional registry of thyroid cancers in Rhône-Alpes	Regional	Academia
Registry and pronostic cohort of cutaneous lymphomas in Aquitaine	Regional	Academia
Registry for digestive cancers in Burgundy	Regional	Academia
Registry of digestive tumors in Calvados (province of France)	Regional	Academia
Registry of observed trichinellosis cases in France yearly	National	Academia
Registry of the network studying thrombotic microangiopathies	National	Academia
Rhône-Alpes registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Rhône-Alpes registry of systemic mastocytosis	Regional	Academia
SYRENE: Rett syndrome network - French database of clinical and genetic aspects of Rett syndrome	National	Academia
VALID: cohort creation on Budd-Chiari syndrome, hepatic venoocclusive disease, hepatoportal sclerosis and portal vein thrombosis	European	Academia
Vedrop registry of chronic cholestasis patient with vitamin E deficiency	European	Industry

GR - GREECE (2 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Greek cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Greek severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Academia

HR - CROATIA (1 registry)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Croatian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

HU - HUNGARY (3 registries)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Hungary - contributes to the TREAT-NMD network	National	Academia
Hungarian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Hungarian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia

IE - IRELAND (9 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
CFRI: The Cystic Fibrosis Registry of Ireland - contributes to the EUROCAT network	National	Academia
Dublin registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Galway registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Irish myelodysplastic syndromes specific registry	National	Academia
Irish registry for Bernard-Soulier syndrome	National	Academia
Irish registry of amyotrophic lateral sclerosis and motor neurone disease	National	Academia
Irish registry of Hurler syndrome	National	Academia
Irish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
South East of Ireland registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Academia
South of Ireland registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia

IL - ISRAEL (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Israeli cystic fibrosis patient registry - contributes to the EUROCAT network	National	Academia
Israeli severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia

IS - ICELAND (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Icelandic cancer registry - contributes to the RARECARE project	National	Patient organisation
Icelandic cystic fibrosis patient registry - contributes to the EUROCAT network	National	Academia

IT - ITALY (51 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Arrhythmogenic right ventricular cardiomyopathy/dysplasia: clinical registry and database, evaluation of therapies, DNA banking	National	Academia
Campania registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Italy - contributes to the TREAT-NMD network	National	Academia

Emilia Romagna registry of congenital anomalies (IMER) - contributes to the EUROCAT registry	Regional	Academia
EUROFEVER: European registry for autoinflammatory diseases	Global	Academia
European registry of congenital dyserythropoietic anemia	European	Academia
FMF: Italian registry for familial mediterranean fever in the young	National	Academia
Friedreich's ataxia Italian patient registry	National	Academia
GLATIT: Glanzmann thrombasthenia Italian registry	National	Academia
HAE-registry: European hereditary angioedema patient registry	European	Academia
IBAHC: Italian registry for alternating hemiplegia of childhood	National	Academia
International registry of bone fragility fractures in the young	Global	Academia
International Registry of Rare Bleeding Disorders (RBDD)	Global	Academia
International registry of recurrent and familial hemolytic uremic syndrome / thrombotic thrombocytopenic purpura	Global	Academia
International registry on thrombotic thrombocytopenic purpura (TTP)	Global	Academia
Italian alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
Italian cystic fibrosis patient registry - contributes to the EUROCARE CF registry	National	Academia
Italian genetic movement disorders registry	National	Academia
Italian Li-Fraumeni syndrome registry	National	Academia
Italian neuroblastoma registry	National	Academia
Italian registry for cri du chat syndrome (monosomy 5p)	National	Academia
Italian registry for hereditary multiple exostoses	National	Academia
Italian registry for MYH9-related thrombocytopenia	National	Academia
Italian registry of adult patients affected by familial mediterranean fever	National	Academia
Italian registry of congenital nephrotic syndromes	National	Academia
Italian registry of Creutzfeldt-Jakob disease and correlated syndromes	National	Academia
Italian registry of diffuse infiltrative pneumopathies	National	Academia
Italian registry of hemophilia centre (AICE)	National	Academia
Italian registry of hypertrophic cardiomyopathy in Anderson-Fabry disease	National	Academia
Italian registry of Legionellosis	National	Academia
Italian registry of maturity onset diabetes of the young (MODY)	National	Academia
Italian registry of muscle channel-diseases	National	Academia
Italian registry of myotonic dystrophies	National	Academia
Italian registry of patients and families affected by Pseudoxanthoma Elasticum	National	Academia
Italian registry of skeletal dysplasia	National	Academia
Italian retinoblastoma registry	National	Academia

Italian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
North-east Italy registry of neurofibromatosis	Regional	Academia
North-East of Italy registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Regional registry for neuromuscular disorders	Regional	Academia
Registry of inherited bleeding disorders in Emilia-Romagna region	Regional	Academia
Registry of pregnant patients affected by essential thrombocythemia	National	Academia
RIAF: Fanconi's anemia Italian registry	National	Academia
RIAT: Ataxia teleangiectasia Italian registry	National	Academia
RICH: Italian registry of infants with congenital hypothyroidism	National	Academia
RIMM: Italian registry for myelofibrosis with myeloid metaplasia	National	Academia
RISMD: Italian myelodysplastic syndromes registry	National	Academia
Sicilian registry of congenital anomalies (ISMAC) - contributes to the EUROCAT network	Regional	Academia
Tuscany registry of congenital anomalies - contributes to the EUROCAT network	Regional	Academia
Venetian registry of rare diseases	Regional	Academia
V-RIAT: variant Ataxia telangiectasia Italian registry	National	Academia

LT - LITHUANIA (1 registry)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Lithuanian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

LU - LUXEMBOURG (1 registry)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Luxembourgers cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

LV - LATVIA (1 registry)

ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Latvian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

MK - REPUBLIC OF MACEDONIA (1 registry)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Macedonian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

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

NL - NETHERLANDS (15 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
CONCOR: Dutch registry of patients with a congenital heart malformation	National	Academia
Duchenne and Becker muscular dystrophy patient registry in the Netherlands - part of the TREAT-NMD network	National	Academia
Dutch alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
Dutch cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Dutch severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
ECARUCA: cytogenetic and clinical database on rare chromosomal disorders	European	Academia
EPCOT: European prospective cohort on thrombophilia	European	Academia
Nephrotic syndrome registry	Global	Academia
North Netherlands registry of congenital anomalies - contributes to the EURO CAT network	Regional	Academia
PAN research: Prospective amyotrophic lateral sclerosis (ALS) study Netherlands	National	Academia
The international Pompe registry	Global	Industry
The International Collaborative Gaucher Group (ICGG) Gaucher registry	Global	Industry
The international Fabry registry	Global	Industry
The international Mps I registry	Global	Industry
X-ALD: X-linked adrenoleukodystrophy database	European	Academia

NO - NORWAY (4 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
EURADRENAL: European patient registry on autoimmune Addison's disease (sera, DNA and RNA)	European	Academia
Norwegian cancer registry - contributes to the RARECARE project	National	Academia

Norwegian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Norwegian severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia

PL - POLAND (9 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Cracow cancer registry - contributes to the RARECARE project	Regional	Academia
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Poland - contributes to the TREAT-NMD network	National	Academia
Kielce cancer registry - contributes to the RARECARE project	Regional	Academia
Polish cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Polish registry of primary immunodeficiencies - contributes to the ESID European registry	National	Academia
Polish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
Polish Silver-Russell syndrome patient registry	National	Academia
PRCM: Polish registry of congenital malformations - contributes to the EUROCAT network	National	Academia
Warsaw cancer registry - contributes to the RARECARE project	Regional	Academia

PT - PORTUGAL (9 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Duchenne and Becker muscular dystrophy patient registry in Portugal - contributes to the TREAT-NMD network	National	Academia
Portugues centre for study and registry of congenital anomalies (CERAC) - contributes to the EUROCAT network	National	Academia
Portuguese cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Portuguese Fabry registry	National	Academia
Portuguese registry of biliary atresia - contributes to the EBAR registry	National	Academia
Portuguese registry of primary immunodeficiency diseases (REPORID)	National	Academia
Portuguese Rett syndrome registry	National	Academia
Portuguese severe chronic neutropenia patient registry - contributes to the SCN international registry (SCNIR)	National	Academia
Southern Portugal cancer registry - contributes to the RARECARE project	Regional	Academia

RO - ROMANIA (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Romanian biliary atresia registry	National	Academia
Romanian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

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

SE - SWEDEN (18 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
FOS : Fabry Outcome Survey	Global	Industry
HOS : Hunter Outcome Survey	Global	Industry
IOS : Icatibant Outcome Survey for hereditary angioedema	Global	Industry
National registry on bronchopulmonary dysplasia	National	Academia
SPAHR: Swedish Pulmonary Arterial Hypertension Registry	National	Academia
SWEDCON: Swedish Registry of Congenital Heart Disease	National	Academia
Swedish Acute Lymphoblastic Leukemia Registry	National	Academia
Swedish Acute Myelogenous Leukemia Registry	National	Academia
Swedish alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
Swedish Childhood Cancer Registry	National	Academia
Swedish Chronic Myeloid Leukemia Registry	National	Academia
Swedish cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Swedish database on Usher syndrome	National	Academia
Swedish Multiple Myeloma Registry	National	Academia
Swedish Polyposis Registry	National	Academia
Swedish Registry for Familial Amyloid Polyneuropathy	National	Academia
Swedish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
SWEDROP: Swedish Registry for Retinopathy of Prematurity	National	Academia

SI - SLOVENIA (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Slovenian cancer registry - contributes to the RARECARE project	National	Academia
Slovenian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

SK - SLOVAKIA (2 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Slovakian cancer registry - contributes to the RARECARE project	National	Academia
Slovakian cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia

TR - TURKEY (4 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
Database setup for the visualisation and examination of oral ulcers in Behcet disease patients	National	Academia
Duchenne and Becker muscular dystrophy and spinal muscular dystrophy patient registries in Turkey - contributes to the TREAT-NMD network	National	Academia
Turkish cystic fibrosis patient registry - contributes to the EURO CARE CF registry	National	Academia
Turkish severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia

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

UK - UNITED KINGDOM (58 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
AOMIC: adult onset myositis immunogenetic collaboration	National	Academia
BPOLD: British Paediatric Orphan Lung Disease Registry	National	Academia
CARIS - Welsh registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Academia
CAROB - congenital anomalies registry for Oxfordshire, Berkshire & Buckinghamshire - part of the BINOCAR and EUROCAT network	Regional	Academia

CRANE: database of patients with cleft lip and/or cleft palate in England and Wales	National	Academia
Duchenne and Becker muscular dystrophy patient registry in United Kingdom and Ireland - part of the TREAT-NMD network	National	Patient organisation
EBV associated NK/T cell diseases registry	National	Academia
EHDN: registry of juvenile Huntington's disease	Global	Academia
EMSYCAR - East Midlands & South Yorkshire congenital anomalies registry - part of BINOCAR and EUROCAT network	Regional	Academia
English alpha-1 antitrypsin deficiency registry - contributes to the Alpha One International Registry (AIR)	National	Academia
English and Irish Fanconi anemia registry	National	Academia
English central hypoventilation syndrome registry - will contribute to the European CHS	National	Academia
English cystic fibrosis database	National	Academia
English cystic fibrosis patient registry - contributes to the EURO CARE CF and ECFS registries	National	Academia
English cystinosis registry	National	Academia
English dyskeratosis congenita registry	National	Academia
English Gaucher registry	National	Academia
English hereditary angioedema patient registry - part of the HAE European registry	National	Academia
English hyperoxaluria registry	National	Academia
English juvenile dermatomyositis registry and repository	National	Academia
English mucopolysaccharidosis registry	National	Patient organisation
English phenylketonuria registry	National	Academia
English registry for lymphangiomyomatosis	National	Academia
English registry of biliary atresia - contributes to the EBAR registry	National	Academia
English registry of primary immunodeficiencies - contributes to the ESID European registry	National	Academia
English registry of syndromes with abnormal vertebral segmentation	National	Academia
English registry of Wolf-Hirschhorn syndrome	National	Academia
English severe chronic neutropenia registry - contributes to the SCN international registry (SCNIR)	National	Academia
EUHASS: European haemophilia safety surveillance - pharmacovigilance program monitoring the safety of treatments for people with inherited bleeding disorders in Europe	European	Academia
EUMDS: European Registry for Myelodysplastic Syndromes - part of EuroLeukemiaNet (ELN)	European	Academia
EURODSD: European disorders of sexual development registry	European	Academia
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	European	Academia
European Prader-Willi syndrome database	European	Academia

EUROWILSON: European clinical database for Wilson disease	European	Academia
Glasgow registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Academia
LCH: English Langerhans cell histiocytosis registry	National	Academia
Merseyside and Cheshire registry of congenital anomalies -part of BINOCAR and EUROCAT network	Regional	Academia
Myotonic dystrophy patient registry in United Kingdom - part of the TREAT-NMD network	National	Academia
National Congenital Anomaly System (NCAS) - part of BINOCAR and EUROCAT network	Regional	Academia
NDSCR - National Down syndrome cytogenetic registry - part of BINOCAR and EUROCAT network	National	Academia
NHD: the national haemophilia database	National	Academia
NHR: National Haemoglobinopathy Registry	National	Academia
NorCAS - Northern registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Academia
Regional spinocerebellar ataxia registry	Regional	Academia
SCAR - Scottish registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Academia
Spinal muscular atrophy patient registry in United Kingdom and Ireland - part of the TREAT-NMD network	National	Academia
Spinocerebellar ataxia type 1 registry	National	Academia
SWCAR - South West congenital anomalies registry - part of BINOCAR and EUROCAT network	Regional	Academia
The regional paediatric cardiology database	Regional	Academia
UK Paediatric ITP (Immune Thrombocytopenic Purpura) Registry	National	Academia
UK renal rare disease registry	National	Academia
UKAITPR: United Kingdom adult idiopathic thrombocytopenic purpura registry	National	Academia
UKCCCR: English familial ovarian cancer patient registry	National	Academia
UKESR: United Kingdom Evans Syndrome Registry	National	Academia
UKFITPR: United Kingdom familial idiopathic thrombocytopenic purpura (ITP) Registry	National	Academia
United Kingdom neuromyelitis optica registry	National	Academia
WANDA - Wessex registry of antenatally detected anomalies - part of BINOCAR and EUROCAT network	Regional	Academia
West Midlands registry of congenital anomalies - part of BINOCAR and EUROCAT network	Regional	Academia

US - UNITED STATES (6 registries)		
ENGLISH LABEL OF THE ACTIVITY	COVERAGE	INSTITUTION
CMDIR: congenital muscular dystrophy international registry	Global	Academia
International Friedreich Ataxia Research Alliance (FARA) registry	Global	Patient organisation
International Morquio A registry	Global	Patient organisation
International Rare Genetic Steroid Disorders Consortium (RGSDC) registry	Global	Academia
International registry for primary hyperoxaluria	Global	Academia
THAOS: transthyretin amyloidosis outcomes survey	Global	Industry

European registries

EUROPEAN REGISTRIES (50 registries)		
ENGLISH LABEL OF THE ACTIVITY	COORDINATION	INSTITUTION
EMSA-SG: central patient registry of the European multiple system atrophy network	AT	Academia
ENRAH: European alternating hemiplegia and rare epilepsies registry in childhood	AT	Academia
EUNEFRON: registry of the European network for the study of orphan nephropathies	BE	Academia
PFAPA Registry: Periodic fever aphtous stomatitis, pharyngitis and adenopathy patient registry	CH	Academia
CompERA-XL: International, prospective registry for the documentation of first-line and maintenance therapy in patients with pulmonary hypertension	DE	Academia
CWS-SoTiSaR: A registry for soft tissue sarcoma and other soft tissue tumours in children, adolescents, and young adults	DE	Academia
DÖSAK tumor registry for documentation of tumors of the face and jaws in germany, austria and switzerland	DE	Academia
EBAR: European Biliary Atresia Registry	DE	Academia
EHDN: European Huntington's disease registry	DE	Academia
ENETS: European Neuroendocrine Tumour Registry	DE	Academia
EPICURE-bank: European Epilepsy Brain Bank	DE	Academia
EU-RHAB: European Rhabdoid Registry	DE	Academia
EURIPFREG: European idiopathic pulmonary fibrosis registry	DE	Academia
EURIPIDES: European Registry for ICD and CRT devices in pediatrics and adults with congenital heart disease	DE	Academia
EUROFA: European Friedreich Ataxia Registry	DE	Academia
European Alport registry	DE	Academia
EUROSCA-R: European patient registry on spinocerebellar ataxias	DE	Academia
EUTOS: European chronic myeloid leukemia patient registry (collaboration between the European LeukemiaNet and Novartis Europe)	DE	Academia
KINDLERNET: Central patient registry Kindler syndrome	DE	Academia
MEFOPA: registry for patients with rare Mendelian forms of Parkinson's Disease	DE	Academia
NCL-Registry: International neuronal ceroid lipofuscinoses patient registry	DE	Academia
PODONET: Registry for Steroid-Resistant Nephrotic Syndrome (SRNS) patients	DE	Academia
RegiSCAR: European registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples	DE	Academia

RetDis Database (blood or DNA samples and associated clinical descriptions of patients and families with inherited eye diseases)	DE	Academia
ERCUSYN: European registry on Cushing's syndrome	ES	Academia
EUGINDAT-PIADATABASE: European primary inherited aminoacidurias database	ES	Academia
MOLDIAG-PACA: patient registry of pancreatic cancer	ES	Academia
Cystadane post marketing registry of patient with homocystinuria	FR	Industry
EHN - EURO-HISTIO-NET: European registry of Langerhans Cell Histiocytosis	FR	Academia
EPI-EPNET: European hepatic and erythropoietic porphyrias registry	FR	Academia
Escort-Hu: European sickle cell disease cohort- hydroxyurea	FR	Industry
EU-CHS: European central hypoventilation syndrome registry	FR	Academia
EURECHINOREG: European registry of alveolar echinococcosis	FR	Academia
European prospective registry of children born to mothers affected by the antiphospholipids syndrome	FR	Academia
EUOTRAPS: European patient registry on TRAPS syndrome	FR	Academia
INFEVERS: European registry of mutations involved in familial mediterranean fever (FMF) and hereditary autoinflammatory disorders	FR	Academia
VALID: cohort creation on Budd-Chiari syndrome, hepatic venoocclusive disease, hepatoportal sclerosis and portal vein thrombosis	FR	Academia
Vedrop registry of chronic cholestasis patient with vitamin E deficiency	FR	Industry
EUHASS: European haemophilia safety surveillance - pharmacovigilance program monitoring the safety of treatments for people with inherited bleeding disorders in Europe	GB	Academia
EUMDS: European Registry for Myelodysplastic Syndromes - part of EuroLeukemiaNet (ELN)	GB (+ IT)	Academia
EURODSD: European disorders of sexual development registry	GB	Academia
EUROPAC: the European registry of hereditary pancreatitis and familial pancreatic cancer	GB	Academia
European Prader-Willi syndrome database	GB	Academia
EUROWILSON: European clinical database for Wilson disease	GB	Academia
European registry of congenital dyserythropoietic anemia	IT	Academia
HAE-registry: European hereditary angioedema patient registry	IT	Academia
ECARUCA: cytogenetic and clinical database on rare chromosomal disorders	NL	Academia
EPCOT: European prospective cohort on thrombophilia	NL	Academia
X-ALD: X-linked adrenoleukodystrophy database	NL	Academia
EURADRENAL: European patient registry on autoimmune Addison's disease (sera, DNA and RNA)	NO	Academia
EURADRENAL: European patient registry and biobank on autoimmune Addison's disease	NO	Academic

International registries

INTERNATIONAL REGISTRIES (29 registries)		
ENGLISH LABEL OF THE ACTIVITY	COORDINATION	INSTITUTION
Pediatric granulomatous arthritis international registry	BE	Academia
BH4: international patient registry of tetrahydrobiopterin deficiency	CH	Academia
PNH Registry: Paroxysmal Nocturnal Hemoglobinuria registry	CH	Industry
CPT-SIOP-Registry : International Registry for Choroid Plexus Tumors	DE	Academia
EHDN: neuroacanthocytosis patient registry	DE	Academia
ESID: European registry of primary immunodeficiencies	DE	Academia
International pheochromocytoma and paraganglioma registry	DE	Academia
STER: FVII deficiency treatment international registry	DE	Academia
International FKR (Fukutin-Related Protein) defects registry - part of TREAT-NMD network	FR	Academia
EHDN: registry of juvenile Huntington's disease	GB	Academia
EUROFEVER: European registry for autoinflammatory diseases	IT	Academia
International registry of bone fragility fractures in the young	IT	Academia
International Registry of Rare Bleeding Disorders (RBDD)	IT	Academia
International registry of recurrent and familial hemolytic uremic syndrome / thrombotic thrombocytopenic purpura	IT	Academia
International registry on thrombotic thrombocytopenic purpura (TTP)	IT	Academia
Nephrotic syndrome registry	NL	Academia
The international Pompe registry	NL	Industry
The International Collaborative Gaucher Group (ICGG) Gaucher registry	NL	Industry
The international Fabry registry	NL	Industry
The international Mps I registry	NL	Industry
FOS : Fabry Outcome Survey	SE	Industry
HOS : Hunter Outcome Survey	SE	Industry
IOS : Icatibant Outcome Survey for hereditary angioedema	SE	Industry
CMDIR: congenital muscular dystrophy international registry	US	Academia
International Friedreich Ataxia Research Alliance (FARA) registry	US	Patient organisation
International Morquio A registry	US	Patient organisation
International Rare Genetic Steroid Disorders Consortium (RGSDC) registry	US	Academia
International registry for primary hyperoxaluria	US	Academia
THAOS: transthyretin amyloidosis outcomes survey	US	Industry

Network registries

NETWORK REGISTRIES (9 registries)		
ENGLISH LABEL OF THE ACTIVITY	COORDINATION	COVERAGE
SCNIR: severe chronic neutropenia international registry	DE	Global
EU-CHS: European central hypoventilation syndrome registry	FR	European
HAE-registry: European hereditary angioedema patient registry	IT	European
RARECARE: surveillance of rare cancers in Europe	IT	European
PAAIR: Patient's Association and Alpha-1 International Registry network	NL	Global
BINOCAR: British Isles network of congenital anomaly registries	UK	National
ECFS: European Cystic Fibrosis Society patient registry	UK	European
EUROCAT: European surveillance of congenital anomalies	UK	European
TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases (registries)	UK	European

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

Editor-in-chief: Ségolène Aymé • Editor of the report: Nicolas Doulet • Visual design : Céline Angin • Photography : Patrice Latron / Inserm

The correct form when quoting this document is:

« Disease Registries in Europe », Orphanet Report Series, *Rare Diseases collection*, January 2011
<http://www.orpha.net/orphacom/cahiers/docs/GB/Registries.pdf>