



# Orphanet Report Series

*Rare Diseases collection*

November 2010

## European collaborative research projects

funded by DG Research and by E-Rare

in the field of rare diseases

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## European clinical networks

funded by DG Sanco and contributing to clinical research

in the field of rare diseases

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## Methodology

This report is based on data extracted from the Orphanet database in October 2010.

Networks in Orphanet are defined as a group of coordinated activities with financing or an official designation.

### 1) European Commission Directorate General for Research 5th, 6th and 7th Framework Programme funded projects

Research on rare diseases has been supported by the European Commission for more than twenty years through Framework Programmes for research, technological development and demonstration activities. Research projects in the field of rare diseases funded by the 5th Framework Programme (FP5 1998-2002), 6th Framework Programme (FP6 2002-2006) and 7th Framework Programme (FP7 2007-2013) are registered in the Orphanet database. This data has been collected with the help of the CORDIS<sup>1</sup> website.

During the Fifth Framework Programme (FP5 1998-2002) the thematic programme “Improving the quality of life and management of living resources” included, amongst other topics, fundamental and clinical research in the field of rare diseases. Support was provided for multinational research into rare diseases, applying advances in modern technology to diagnosis, treatment, prevention and surveillance through epidemiology. 47 projects were funded for about €64 million in total.

Under the subsequent Sixth Framework Programme (FP6 2002-2006), one of the seven thematic areas supported projects with a focus on “Life sciences, genomics and biotechnology for health”. This thematic area stimulated and sustained multidisciplinary research to exploit the full potential of genome information to underpin applications to human health. In the field of applications, the emphasis was on research aimed at bringing basic knowledge through to the application stage (translational approach), to allow real, consistent and coordinated medical progress at European level and to improve the quality of life. This thematic area was twofold, one of the aspects being the fight against major diseases, including rare diseases. FP6 saw a significant increase in the funding for rare disease

projects: around €230 million for a total of 59 projects, also including an ERA-Net project (E-Rare). Overall this allowed for the mobilisation of researchers to tackle the fragmentation of research and the production of new knowledge, but also a better coordination of research at EU level, and the fostering of the dialogue with all stakeholders, including patients.

The Seventh Framework Programme of the European Union for research, technological development and demonstration activities (FP7, 2007-2013) is composed of four main specific programmes – “Cooperation”, “Ideas”, “People” and “Capacities” – including cross-cutting issues such as support for SMEs, international cooperation, the contribution of research to EU policy, and the inclusion of societal considerations. Rare disease research features under the heading of the Health theme, one of ten themes proposed under the specific programme on “Cooperation”. This specific programme is designed to gain or strengthen leadership in key scientific and technological areas by supporting trans-national cooperation between universities, industry, research centres, public authorities and stakeholders across the European Union and the rest of the world. Specifically, the focus for rare disease research collaborative in FP7 is on pan-European studies of natural history, pathophysiology, and the development of preventive, diagnostic and therapeutic interventions. This sector includes rare Mendelian phenotypes of common diseases. Supported projects should help identify and mobilise the critical mass of expertise in order (i) to shed light on the course and/or mechanisms of rare diseases, or (ii) to test diagnostic, preventive and/or therapeutic approaches, to alleviate the negative impact of the disease on the quality of life of the patients and their families, as appropriate depending on the level of knowledge concerning the specific (group of) disease(s) under study.

### 2) E-Rare funded projects

E-Rare<sup>2</sup> is an FP6 funded ERA-Net for research programmes on rare diseases. The ERA-net scheme aims to step up the cooperation and coordination of research activities carried out at national or regional level in the Member States and Associated States through

<sup>1</sup> [http://cordis.europa.eu/home\\_en.html](http://cordis.europa.eu/home_en.html)

<sup>2</sup> <http://www.e-rare.eu/>

the networking of research activities conducted at national or regional level, and the mutual opening of national and regional research programmes. The scheme aims to help develop a European Research Area by improving the coherence and coordination across Europe of such research programmes. The scheme will also enable national systems to take on tasks collectively that they would not have been able to tackle independently. Both networking and mutual opening require a progressive approach. The ERA-NET scheme therefore has a long-term perspective that must also allow for the different way that research is organised in different Member States and Associated States.

E-Rare is a network of ten partners responsible for the development and management of national/regional research programmes on rare diseases. This project helps develop synergies among the national and/or regional research programmes on rare diseases in participating countries, to establish a common research policy on rare diseases and to coordinate their national/regional research programmes, notably through the setting up of joint strategic activities and transnational calls for proposals.

E-Rare has already launched two calls for proposals. A first transnational call for proposals was launched by E-Rare in 2007<sup>3</sup>: six E-Rare partners (France, Germany, Italy, Israel, Spain and Turkey) participated in the call and 13 projects were selected for funding. The second joint transnational call<sup>4</sup> was launched at the end of 2008/beginning of 2009: ten countries participated in the call (France, Germany, Israel, Spain, Turkey, the Netherlands, Portugal, Italy, Austria and Greece). 16 transnational research consortia with 75 participating research teams from 10 countries were funded via this call for a total research budget of €9.6 million.

### **3) European Commission Directorate General for Health and Consumers funded projects**

The Community action programme on rare diseases, including genetic diseases, was adopted by the European Commission for the period 1 January 1999 to 31 December 2007. The aim of the programme was

to contribute, in co-ordination with other Community measures, to ensuring a high level of health protection in relation to rare diseases. As a first EU effort in this area, specific attention was given to improving knowledge and facilitating access to information about these diseases. Rare diseases are now one of the priorities in the second programme of Community action in the field of health (2008-13)<sup>5</sup>. According to the DG Public Health Work Plans for the implementation of the Public Health Programme, the two main lines of action are the exchange of information via existing European information networks on rare diseases, and the development of strategies and mechanisms for information exchange and co-ordination at EU level to encourage continuity of work and trans-national co-operation.

In the field of rare diseases DG Public Health prioritises networks<sup>6</sup>, which centralise information on as many rare diseases as possible - not just a specific group or a single disease - to improve information, monitoring and surveillance.

Various projects were supported in the framework of the Programme for Community Action on Rare Diseases<sup>7</sup> for 1 January 1999 to 31 December 2003 and the EU Public Health Programme 2003-2008<sup>8</sup> in order to improve the exchange of information via existing European information networks on rare diseases, to promote better classification, to develop strategies and mechanisms for exchanging information between people affected by a rare disease, volunteers and professionals, to define relevant health indicators and develop comparable epidemiological data at EU level, and to support an exchange of best practise and develop measures for patient groups and also aid the development of European Reference Networks of Centres of Expertise and the identification of rare diseases.

<sup>3</sup> <http://www.e-rare.eu/Announcements/1st-Joint-Call.html>

<sup>4</sup> <http://www.e-rare.eu/Announcements/2nd-Joint-Call.html>

<sup>5</sup> [http://ec.europa.eu/health/programme/policy/2008-2013/index\\_en.htm](http://ec.europa.eu/health/programme/policy/2008-2013/index_en.htm)

<sup>6</sup> [http://ec.europa.eu/health/rare\\_diseases/projects/index\\_en.htm](http://ec.europa.eu/health/rare_diseases/projects/index_en.htm)

<sup>7</sup> [http://eur-lex.europa.eu/pri/en/oj/dat/1999/L\\_155/L\\_15519990622en00010005.pdf](http://eur-lex.europa.eu/pri/en/oj/dat/1999/L_155/L_15519990622en00010005.pdf)

<sup>8</sup> [http://ec.europa.eu/health/programme/policy/2003-2008/index\\_en.htm](http://ec.europa.eu/health/programme/policy/2003-2008/index_en.htm)

## Summary

### 1- Number of networks for which a country is coordinator or participant

COUNTRY	COORDINATION	PARTICIPATION	TOTAL
AT - Austria	8	39	47
BE - Belgium	10	69	79
BG - Bulgaria	0	6	6
CH - Switzerland	2	66	68
CY - Cyprus	0	8	8
CZ - Czech Republic	0	30	30
DE - Germany	39	164	203
DK - Denmark	6	50	56
EE - Estonia	0	9	9
ES - Spain	14	82	96
FI - Finland	2	29	31
FR - France	48	150	198
GB - Great Britain	23	127	150
GR - Greece	2	26	28
HR - Croatia	0	5	5
HU - Hungary	1	21	22
IE - Ireland	0	26	26
IL - Israel	0	32	32
IS - Iceland	1	7	8
IT - Italy	27	137	164
LT - Lithuania	0	5	5
LV - Latvia	0	6	6
LU - Luxembourg	1	4	1
MK - Republic of Macedonia	0	2	2
NL - Netherlands	19	96	115
NO - Norway	2	31	33
PL - Poland	0	28	28
PT - Portugal	0	32	32
RO - Romania	0	8	8
RS - Serbia	0	7	7
SE - Sweden	8	74	82
SI - Slovenia	0	13	13
SK - Slovakia	0	5	5
TR - Turkey	0	17	17
NON EUROPEAN COUNTRIES	0	151	151

## 2- Number of networks for which non European countries are participants

NON EUROPEAN COUNTRIES	NUMBER OF PARTICIPANTS
AFRICA	48
AMERICA	43
ASIA	29
EUROPA (not EU 27MS)	14
MIDDLE EAST (not IL)	7
OCEANIA	10
<b>TOTAL</b>	<b>151</b>

## 3- Geographical coverage of networks

COVERAGE	NUMBER OF NETWORKS
EUROPEAN	145
GLOBAL	68
<b>TOTAL</b>	<b>213</b>

## 4- Distribution of networks by type of call

FUNDING BODIES	NUMBER OF NETWORKS
DG SANCO	23
E-RARE CONSORTIUM	29
FP5 - DG RESEARCH	5
FP6 - DG RESEARCH	69
FP7 - DG RESEARCH	87
<b>TOTAL</b>	<b>213</b>

## List of networks

FP5 FUNDED NETWORKS (5 NETWORKS)			
1998 - 2002			
NETWORK ACRONYM / TITLE	PARTICIPATING COUNTRIES	COORDINATING COUNTRY	CONCERNED DISEASES
EMSA-SG: the European Multiple System Atrophy study group	AT, CN, DE, DK, ES, FR, GB, IL, IT, PT, SE, SI, US	AT	Multiple system atrophy
ESDN: European Skeletal Dysplasia Network	BE, CH, DE, FI, FR, GB	GB	Rare bone disease
EUROCRAN: European collaboration on craniofacial anomalies - Eurocleft clinical network	DK, GB, NO, SE	SE	Cranial malformation
SIOPEN-R-NET: International society of paediatric oncology European neuroblastoma research network	AT, BE, CH, CZ, DK, ES, FR, HU, IL, IT, NO, PL, PT, SE	AT	Neuroblastoma
WHIPPLE'S DISEASE: European network on <i>Tropheryma whipplei</i> infection	AT, BE, DE, FR, IT	DE	Whipple disease

FP6 FUNDED NETWORKS (69 NETWORKS)			
2003 - 2006			
NETWORK ACRONYM / TITLE	PARTICIPATING COUNTRIES	COORDINATING COUNTRY	CONCERNED DISEASES
ANTEPRION: development of a pre-clinical blood test for prion diseases	DE, ES, FR, GB, IL, NL, SE, SK	NL	Transmissible spongiform encephalopathies
ANTIMAL: development of new drugs for the treatment of malaria	BE, BF, CH, DE, ES, FR, GA, GB, IT, KE, NL, UA, ZA, ZW	GB	Malaria
AUTOROME: from immune responses in rare autoimmune diseases to novel therapeutic intervention strategies-a personalized medicine approach	CH, DE, FR, IE, IL, NL	DE	Stiff-man syndrome
			Systemic autoimmune disease
			Vasculitis
BIOMALPAR: biology and pathology of the malaria parasite	CH, CM, DE, FR, GB, GR, IN, IT, ML, NG, NL, SD, SE, UG	FR	Malaria
BNE: BrainNet Europe II: European brain tissue bank - Network for clinical neuroscience and basic research	AT, DE, ES, FI, FR, GB, GR, HU, IT, NL, SE	DE	Rare neurologic disease
CILMALVAC: The tetrahymena system as an innovative approach to malaria antigene expression	DE, DK, GB	DE	Malaria

CLINIGENE: European network for the advancement of clinical gene transfer and therapy	AT, CH, CZ, DE, ES, FI, FR, GB, IL, IT, PT, SE, US	FR	-
CONTICANET: Connective Tissue Cancers Network	BE, DE, ES, FR, GB, IE, IT, NL, SI	FR	Soft tissue sarcomas
CSI-LTB: the role of chromosome stability in persistence, latency and reactivation of mycobacterium tuberculosis	CH, GB, IN, IT	CH	Tuberculosis
EMVDA: the European malaria vaccine development association	CH, DE, DK, GB, NL, SE, TZ	DK	Malaria
ENRAH: European Network for Research on Alternating Hemiplegia in Childhood	AT, BE, CZ, DE, ES, FR, GB, IT, NL	AT	Alternating hemiplegia of childhood
EUGINDAT: European genomics initiative on disorders of plasma membrane amino acid transporters	CH, DE, ES, FI, IL, IT	ES	Amino acid transport disease
EUMITOCOMBAT: rational treatment strategies combating mitochondrial oxidative phosphorylation (OXPHOS) disorders	CZ, DE, ES, FI, FR, GB, IT, NL, SE	NL	Mitochondrial Oxidative Phosphorylation Disorder
EURAMY: systemic amyloidoses in Europe	BE, DE, FR, GB, IT, NL, PT, SE	SE	Amyloidosis
EURAPS: autoimmune polyendocrine syndrome type I - a rare disorder of childhood as a model for autoimmunity	AU, CH, CN, DE, EE, FI, GB, IE, IT, NO, SE	SE	Autoimmune polyendocrinopathy type 1
EUREGENE: European renal genome project	BE, CH, DE, DK, FI, FR, GB, IT	DE	Rare renal disease
EURHAVAC: European network for harmonisation of malaria vaccine development	DK	DK	Malaria
EUROBONET: European bone tumors network	BE, CH, DE, DK, ES, FI, GB, HU, IT, NL, NO, SE	NL	Bone sarcoma
			Rare bone tumor
EUROCARE CF: European coordination action for research in cystic fibrosis	BE, CZ, DE, FR, GB, IL, IT, NL, PL, PT, SE	GB	Cystic fibrosis
EUROGENTEST: Network for test development, harmonization, validation and standardization	BE, CH, CZ, DE, EC, ES, FI, FR, GB, HU, IE, IT, ME, NL, PL, PT, RS, SE	BE	-
EUROGLYCANET: European network on Congenital Disorders of Glycosylation	BE, BG, CH, CZ, DE, DK, ES, FR, GB, GR, HR, IL, IT, NL, PL, PT	BE	CDG syndrome
EURO-IRON1: abnormal iron distribution in humans	AT, DE, FR, GB, IL, IT	FR	Rare hemochromatosis
EUROPEAN LEUKEMIA NET: Strengthen and develop scientific and technological excellence in research and therapy of leukemia	AT, BE, BY, CH, CY, CZ, DE, DK, ES, FI, FR, GB, GR, HR, HU, IE, IL, IT, LT, ME, NL, NO, PL, RO, RS, RU, SE, TR	DE	Acute lymphoblastic leukemia
			Acute myeloid leukemia
			Chronic B-cell lymphocytic leukemia
			Chronic myeloproliferative disease
			Myelodysplastic syndromes

EUROSCA: European integrated project on spinocerebellar ataxias	BE, DE, ES, FR, GB, HU, IT, NL, PL	DE	Autosomal dominant cerebellar ataxia
EUROWILSON: European network on Wilson disease	AT, DE, FR, GB, HU, IT, NL, PL	GB	Wilson disease
EVI-GENORET: functional genomics of the retina in health and disease	BE, CH, DE, ES, FR, GB, GR, IE, IT, NL, PT, SE	FR	Genetic retinal dystrophy
FASTEST-TB: Development and clinical evaluation of fast tests for tuberculosis diagnosis	BE, DE, IN, MX, NG, TR	DE	Tuberculosis
GENESKIN: European network on rare genetic skin diseases	AT, BE, CH, DE, ES, FR, GB, HU, IE, IT, NL, SE	IT	genetic skin diseases
HDLOMICS: functional genomics of inborn errors and therapeutic interventions in high density lipoprotein (HDL) metabolism	CH, DE, DK, GR, NL, SE	DE	HDL metabolism disorder
HUE-MAN: Towards the development of an effective enzyme replacement therapy for human alpha- mannosidosis	BE, CZ, DE, DK, FR, NO	DE	Alpha-mannosidosis
HUMALMAB: human monoclonal antibodies as tools for malaria research and therapy	CH, DK, GB, TZ	DK	Malaria
IMMUNOPRION: strains, species and immunology in prion diseases	BE, CH, FR, GB, NL	FR	Transmissible spongiform encephalopathies
LEISHMED: Monitoring risk factors of spreading of Leishmaniasis around the Mediterranean Basin	BE, CH, CY, CZ, DE, DK, ES, FR, GB, GR, IL, IT, JO, MA, PT, TN, TR	BE	Leishmaniasis
LYMPHANGIOGENOMICS: genome-wide discovery and functional analysis of novel genes in lymphangiogenesis	AT, BE, CH, DE, FI, FR, SE	FI	Lymphedema
MALARIA AGE EXPOSURE: age of exposure and immunity to malaria in infants	AU, ES, GB, IN, IT	ES	Malaria
MANASP: Development of novel management strategies for invasive aspergillosis	DE, FR, IE, IT, SE	DE	Allergic bronchopulmonary aspergillosis Aspergillosis
MILD-TB: immunogenicity of Mycobacterium tuberculosis lipids in the non- replicating status of latency	CH, CI, FR, IT	IT	Tuberculosis
MITOCIRCLE: mitochondrial diseases: from bedside to genome to bedside	FR, GB, IT, NL	NL	Mitochondrial disease
MMR-RELATED CANCER: prevention, diagnosis and molecular characterisation of mismatch repair defect-related hereditary cancers of the digestive system	DK, FI, NL, PT	NL	Rare digestive tumor
MM-TB: molecular markers of M. tuberculosis early interactions with host phagocytes	FR, GB, IT	IT	Tuberculosis
MOLDIAG-PACA: Novel molecular diagnostic tools for the prevention and diagnosis of pancreatic cancer	DE, EE, ES, GB, IE, IT, SE	DE	Familial pancreatic cancer

MPCM: pathogenic role of micro-vesiculation in cerebral malaria	AU, CA, CH, FR, GB, MW	FR	Malaria
MYASTAID: European network on myasthenia	BE, DE, DK, FR, GR, IL, NL	FR	Myasthenia gravis
MYORES: European Muscle Development Network	AU, CH, CZ, DE, ES, FR, GB, IL, IT, US	FR	Neuromuscular disease
NEOTIM: innate and adaptive immunity in clinical and experimental mycobacterial infection in neonates and infants	BE, DE, FR, GB, IT, SE	SE	Tuberculosis
NEUPROCF: new diagnostic and prognostic biomarkers in cystic fibrosis	DE, FR, GB, PL, PT, SE	FR	Cystic fibrosis
NEUROKCNQPATHIES: cell biology of rare monogenic neurological KCNQ disorders	DE, DK, ES, GB, IT, NO	ES	Benign familial neonatal seizures
			Episodic ataxia type 1
			Familial paroxysmal ataxia
			Nonsyndromic genetic deafness
NEUROPRION: European network dedicated to research on prion diseases	AT, BE, CH, DE, DK, ES, FR, FI, GB, GR, IE, IL, IT, NL, NO, SE	FR	Transmissible spongiform encephalopathies
NEWTBDRUGS: New Drugs for Persistent Tuberculosis	DE, GB, SE	DE	Tuberculosis
NM4TB: new medicines for tuberculosis	CH, DE, DK, FR, GB, HU, IT, RU, SE, SK, ZA	CH	Tuberculosis
PEROXISOMES: Integrated project to decipher the biological function of peroxisomes in health and disease	AT, BE, DE, FI, FR, IT, NL, PT, SE	AT	Peroxisomal disease
PNSEURONET: European network on Paraneoplastic Neurological Syndromes	AT, CZ, DE, ES, FR, GB, IT, NL, NO, SI	IT	Paraneoplastic neurologic syndromes
PRIBOMAL: pre-clinical studies towards an affordable, safe and efficacious two-component paediatric malaria vaccine	DE, DK, NL, SE	NL	Malaria
PROTHETS: prognosis and therapeutic targets in the "Ewing" family of tumors	AT, DE, ES, FI, FR, IT, RU	IT	Ewing sarcoma
			Neuroepithelioma
			Primitive neuroectodermal tumor
PULMOTENSION: Functional genomics and therapy of lung vascular remodelling	AT, BE, CH, DE, DK, FI, FR, GB, GR, IE, IT	DE	Rare pulmonary hypertension
PWS: Prader-Willi Syndrom: a model linking gene expression, obesity and mental health	AT, BE, DE, FR, GB, IL, NL, SE	GB	Prader-Willi syndrome
RATSTREAM: European project on the characterisation of transgenic rat models for neurodegenerative and psychiatric diseases: Automated home cage analyses, live imaging and treatment	BE, CH, DE, FR, NL	DE	Familial Parkinson disease
			Huntington disease
			Spinocerebellar ataxia type 17

SARS/FLU VACCINE: development of a combined Influenza/SARS vaccine	AT, CZ, DE, SI	AT	Severe acute respiratory syndrome
SCRIN-SILICO: finding promising drug candidates against tuberculosis with multidisciplinary protocol based non-conventional search	CZ, DE, HU, IT	HU	Tuberculosis
SERO-TB: development of a specific serological kit for the diagnosis of TB	DK, ES, ET, TR	DK	Tuberculosis
SPASTICMODELS: genetic models of chronic neuronal degeneration causing hereditary spastic paraplegia	DE, DK, GB, IT	IT	Familial spastic paraplegia
STEM-HD: embryonic stem cells for therapy and exploration of mechanisms in Huntington disease	BE, FR, GB, IL, IT	FR	Huntington disease
TAMAHUD: identification of early disease markers, novel pharmacologically tractable targets and small molecule phenotypic modulators in Huntington's Disease'	DE, ES, GB, IT	IT	Huntington disease
TB TREATMENT MARKER: establishing a TB treatment efficacy marker	DK, GW	DK	Tuberculosis
TB-DRUG OLIGOCOLOR: development of a molecular platform for the simultaneous detection of Mycobacterium tuberculosis resistance to rifampicin and fluoroquinolones	AR, BE, CO, NL, SE	BE	Tuberculosis
THERAPEUSKIN: European network on gene therapy of hereditary epidermolysis bullosa	CH, DE, FR, GB	FR	Hereditary epidermolysis bullosa
TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases	BE, CH, DE, ES, FI, FR, GB, HU, IT, NL, SE	GB	Neuromuscular disease
VACCINES4TB: genome- and HLA-wide scanning and validation of cytotoxic CD8 T cell responses against Mycobacterium tuberculosis	DE, DK, NL	DK	Tuberculosis
VITAL: development of optimised recombinant idiotypic vaccines for subset-specific immunotherapy of B cell lymphomas	GB, IT, NL, NO, SE	IT	B-cell non-Hodgkin lymphoma

**FP7 FUNDED NETWORKS (87 NETWORKS)**

2007-2013

NETWORK ACRONYM / TITLE	PARTICIPATING COUNTRIES	COORDINATING COUNTRY	CONCERNED DISEASES
03K: Oral off-patent oncology drugs for kids	FR, GB, IT	FR	-
AAVEYE: gene therapy for inherited severe photoreceptor diseases	CH, EE, GB, IT	IT	Optic neuropathy
			Leber congenital amaurosis
			Retinitis pigmentosa
ARTEMIP: The safety pharmacology of artemisinins when used to reverse pathophysiology of malaria in pregnancy	CH, GB, NL, PT, SE	GB	Malaria
BIG-HEART: bench-to-beside integrated approach to familial hypertrophic cardiomyopathy: to the heart of the disease	DE, GB, IT, NL	GB	Familial hypertrophic cardiomyopathy
BIO-NMD: identifying and validating pre-clinical biomarkers for diagnostics and therapeutics of neuromuscular disorders	DE, FR, IT, NL, SE, US	FR	Bethlem myopathy
			Congenital muscular dystrophy, Ullrich type
			Muscular dystrophy, Duchenne type
BRAINCAV: Nonhuman adenovirus vectors for gene transfer to the brain	DE, ES, FR, GB, IT, PT	FR	Mucopolysaccharidosis type 7
CARDIOGENET: definition of a genetic network involved in congenital heart disease	DE, FR, GB, IT, NL	IT	Congenital heart malformation
CHAGASEPINET: Comparative epidemiology of genetic lineages of <i>Trypanosoma cruzi</i>	BE, ES, FR, GB, SE	GB	Trypanosomiasis
CHD PLATFORM: Establishment of a European parent- and patient-oriented information and communication platform on Congenital Heart Defects	DE, ES, GB, NO, SE	DE	Congenital heart malformation
CHEARTED: gene-environment interactions in heart development	AU, BE, DE, FR, GB, NL, NO	NL	Congenital heart malformation
CHERISH: Improving diagnoses of mental retardation in children in Central Eastern Europe and Central Asia through genetic characterisation and bioinformatics/-statistics	AR, CY, CZ, EE, IT, LT, PL, RU	IT	Rare intellectual deficit
CRIMALDDI: Coordination, rationalisation and integration of antimalarial drug discovery initiatives	CH, CM, DE, FR, GB, IT, ZA	GB	Malaria
CRUMBS IN SIGHT: restoring Mueller glia cell photoreceptor interactions with Crumbs	DE, FR, GB, NL	NL	Congenital Leber amaurosis
			Retinitis pigmentosa

CUREHLH: European initiative to improve knowledge, treatment and survival of haemophagocytic syndromes in children	DE, FR, IT, SE	DE	Hemophagocytic syndrome
DIRECT: Disseminate research funded by EC improving Treatment options for children suffering from cancer	AT, DE	AT	Rare tumor
EFACTS - EUROFA: European Friedreich's Ataxia Consortium for Translational Studies	AT, BE, DE, ES, FR, GB, IT, US	BE	Friedreich's ataxia
EMPERIE: European management platform for emerging and re-emerging infectious disease entities	DE, ES, FR, GB, HK, NL	NL	Rare infectious diseases
ENAROMATIC: European network for advanced research on olfaction for malaria transmitting insect control	CH, DE, FR, GB, GR, HU, IT, NG, US	GR	Malaria
ENCE CF-LAM-LTX: European networks of centres of expertise for CF (Cystic Fibrosis), LAM (Lymphangiomyomatosis), and LTX (Lung Transplantation)	AT, CZ, DE, FR, GB	DE	Cystic fibrosis
			Lymphangiomyomatosis
EPOKS: European patient organizations and knowledge society in the field of rare diseases	FR, GB, IE, PT	FR	-
ESI-TBVI: Establishment, strategy and initial activities of the tuberculosis vaccine initiative : coordination of european efforts with global research initiatives	DE, DK, GB, NL	NL	Tuberculosis
EUCILIA: Pathophysiology of rare diseases due to ciliary dysfunction: nephronophthisis, oral-facial-digital type 1 and bardet-biedl syndromes	DE, GB, IT	IT	Nephronophthisis-associated ciliopathy
			Oral-facial-digital syndrome type 1
			Retinal ciliopathy due to mutation in Bardet-Biedl gene
EUCLYD: A European consortium for Lysosomal disorders	DE, IT, NL, SE	IT	Lysosomal disease
EUCO-NET: European network for global cooperation in the field of AIDS & TB	AR, BE, BR, CO, DE, IN, IT, RU, ZA	DE	Tuberculosis
EUNEFRON: the European network for the study of orphan nephropathies	BE, CH, DE, DK, FR, GB, IT, NL	BE	Rare renal disease
EURADRENAL: pathophysiology and natural course of autoimmune adrenal failure in Europe	CH, DE, GB, IT, NO, PL, SE	NO	Autoimmune polyendocrinopathy type 1
			Autoimmune polyendocrinopathy type 2
			Chronic adrenal insufficiency acquired
EURIPFNET: European IPF network: natural course, pathomechanisms and novel treatment options in idiopathic pulmonary fibrosis	AT, DE, FR, GB, IT	DE	Idiopathic pulmonary fibrosis (UIP)

EURODSD: European collaborative study on disorders of sex development	DE, FR, GB, IT, NL, SE	DE	Disorder of sex development of endocrine origin
			Malformative disorder of sex development
EURO-GENE-SCAN: European genetic disease diagnostics	DE, GB, PL, SE	SE	-
EURO-PADNET: the pathophysiology and natural course of patients with Primary Antibody Deficiencies (PAD)	CZ, DE, FR, GB, IT, NL, SE	GB	Primary Antibody Deficiencies
EUROTRAPS: natural course, pathophysiology, models for early diagnosis, prevention and innovative treatment of TNF Receptor Associated Periodic Syndrome (TRAPS) with application for all hereditary recurrent fevers	AT, DE, FR, GB, IL, IT	FR	Periodic fever syndrome
EVIMALAR: Towards the establishment of a permanent European Virtual Institute dedicated to Malaria Research (EVIMaLaR)	AU, CH, CM, DE, DK, FR, GB, GR, IN, IT, NG, NL, PT, SD, SE, UG	GB	Malaria
FAST-XDR-DETECT: Development of a two-approach plate system for the fast and simultaneous detection of MDR and XDR M. tuberculosis	AR, BE, CO, DE, LV, SE	BE	Tuberculosis
FIGHT-MG: Myasthenias, a group of immune mediated neurological diseases: from etiology to therapy.	CH, DE, FR, GR, IL, IT, NO	FR	Myasthenia gravis
GEN2PHEN: genotype-to-phenotype databases: A holistic solution	BE, CH, DE, ES, FI, FR, GB, GR, IN, IS, NL, PT, SE, ZA	GB	-
HOMITB: Host and microbial molecular dissection of pathogenesis and immunity in tuberculosis	BE, DE, FR, GB, IT, MA, SE	SE	Tuberculosis
INHERITANCE: INtegrated HEart Research In TrANslational genetics of dilated Cardiomyopathies in Europe	DE, DK, ES, FR, IT, NL, SE	IT	Dilated cardiomyopathy
LEISHDNAVAX: development of a DNA vaccine for visceral leishmaniasis	DE, GB, IL, IN, TN	GB	Leishmaniasis
LEISHDRUG: targeting the Leishmania kinome for the development of novel anti-parasitic strategies	DE, ES, FR, IL, IT, KR, TN, UY	FR	Leishmaniasis
LEUKOTREAT: Therapeutic challenge in Leukodystrophies: Translational and ethical research towards clinical trials	AT, DE, ES, FR, GB, IT, NL	FR	Leukodystrophy
LOULLA&PHILLA: Development of 6-mercaptopurine and Methotrexate oral liquid formulations for the maintenance treatment of Acute Lymphoblastic Leukemia in children	DE, DK, FR, IE	FR	Acute lymphoblastic leukemia
LUPAS: Luminescent polymers for in vivo imaging of amyloid signatures	CH, DE, FR, NO, SE	SE	Transmissible spongiform encephalopathies
			Familial amyloid polyneuropathy

MALACTRES: Multi-drug resistance in malaria under combination therapy: Assessment of specific markers and development of innovative, rapid and simple diagnostics	BE, BF, GB, NG, NL, TZ	NL	Malaria
MALSIG: Signalling in life cycle stages of malaria parasites	DE, FR, GB, IN, IT, NL	GB	Malaria
MALVECBLOK: Population biology and molecular genetics of vectorial capacity in Anopheles gambiae: targeting reproductive behaviour and immunity for transmission-refractory interventions	FR, IT, KE, ML, NL	FR	Malaria
MEFOPA: European Project on Mendelian Forms of Parkinson's Disease	AE, BE, CH, DE, DK, ES, FR, HU, GB, GR, IT, NL, NO, PT, SE	DE	Genetic Parkinson disease
MEPHITIS: Targeting protein synthesis in the apicoplast and cytoplasm of plasmodium	AU, ES, FR, IN, IT, PT	ES	Malaria
MISMATCH2MODEL: Characterization and quantitative modeling of DNA mismatch repair and its role in the maintenance of genomic stability and cancer avoidance	CH, DE, DK, FR, NL	NL	Familial nonpolyposis colon cancer
MITOTARGET: mitochondrial dysfunction in neurodegenerative diseases: towards new therapeutics		FR	Amyotrophic lateral sclerosis
			Familial spastic paraplegia
			Huntington disease
MYELINET: Myelin orphan diseases in health	AT, BE, CH, CY, DE, DK, ES, FR, GB, IL, IT, NO, PL, RS, SI, TR	AT	Leukodystrophy
NANOTRYP: exploiting nanobodies in development of new diagnostic tools and treatment methods for trypanosomiasis		BE	American trypanosomiasis
			African trypanosomiasis
NATT: New approaches to target tuberculosis	BE, DE, GB, IN, SE	BE	Tuberculosis
NEMO: Treatment of Neonatal seizures with medication off-patent: evaluation of efficacy and safety of bumetanide	DE, FI, FR, GB, IE, SE, US	GB	Neonatal epilepsy syndrome
NEURO.GSK3: GSK-3 in neuronal plasticity and neurodegeneration: basic mechanisms and pre-clinical assessment	BE, DE, ES, GB, IL, PL	BE	Frontotemporal dementia
NEUROSIS: efficacy and safety of inhaled budesonide in very preterm infants at risk for bronchopulmonary dysplasia	CZ, DE, FR, GB, IL, IT, NL	DE	Bronchopulmonary dysplasia
NEUROXSYS: Genomic regulatory systems of human X-linked neurological diseases	DE, FR, GB, NO	NO	-

NEWTBVAC: Discovery and preclinical development of new generation tuberculosis vaccines	BE, CH, DE, DK, ES, FR, GB, IT, KR, NL	NL	Tuberculosis
NGIDD: Neuron-Glia interactions in nerve development and disease	DE, IL, IT, NL	NL	-
NIMBL: Nuclease Immune Mediated Brain and Lupus-like conditions (NIMBL): natural history, pathophysiology, diagnostic and therapeutic modalities with application to other disorders of autoimmunity	ES, GB, IT, NL, US	GB	Aicardi-Goutieres syndrome
			Cerebroretinal vasculopathy
NMD-CHIP: development of targeted DNA-chips for high throughput diagnosis of neuromuscular disorders	BE, DE, FR, HU, IT, NL, SE	FR	Neuromuscular disease
NOSTRESS: Unraveling the molecular mechanism of nitrosative stress resistance in tuberculosis	AR, ES, GB, IN	ES	Tuberculosis
NOVELPID: the genetic dissection of herpes simplex encephalitis (HSE) in children	FR	FR	Herpetic encephalitis
NOVSEC-TB: novel secretion systems of Mycobacterium tuberculosis and their role in host-pathogen interaction	CH,FR, IE, IT, NL, ZA	FR	Tuberculosis
OPTIMALVAC: Initiative on optimizing malaria vaccine lab assays evaluation	BE, CH, DE, DK, ES, FR, GB, NL, SE, US	DE	Malaria
PEMPHIGUS: from autoimmunity to disease	CH, DE, FR, IT	DE	Paraneoplastic pemphigus
			Pemphigus vulgaris
PRATH: Preclinical study of Recombinant human Anti-C5 for the Treatment of atypical HUS	FR, GB, IT	IT	Atypical hemolytic uremic syndrome
PREGVAX: Plasmodium Vivax infection in pregnancy	BR, CO, ES, GT, IN, IT, PG, SE	ES	Malaria
PREMALSTRUCT: Structural analysis of the CSA binding interactions involved during pregnancy associated malaria	DE, DK, FR, IN, FR	FR	Malaria
PSYCHCNVS: copy number variations conferring risk of psychiatric disorders in children	GE, GB, IS, MK, RS, RU, UA	IS	Rare pediatric psychiatric disease
RAPSODI: pre-clinical studies of a PSA-based human vaccine candidate targeting visceral, cutaneous and mucocutaneous leishmaniasis and development of the associated procedures for further clinical trials	ES, FR, IN, TN, PE	FR	Leishmaniasis
RDCVF: Rod-derived Cone Viability Factor	DE, FR, PT	FR	Retinitis pigmentosa
REDMAL: Clinical development of a Pfs48/45-based malaria transmission blocking vaccine	DK, GB, IN, NL, TZ	NL	Malaria

RESPECT: Relating expectations and needs to participation and empowerment of children in clinical trials	BE, DE, IT, LU, SE, SI	SE	-
SETTREN: Schistosoma Epigenetics - Targets, Regulation, New Drugs	BR, DE, FR, SE	FR	Schistosomiasis
STOPLATENT-TB: Latent tuberculosis: new tools for the detection and clearance of dormant mycobacterium tuberculosis	CO, ES, FR, GB, IT, MX	ES	Tuberculosis
STOPPAM: Strategies to prevent pregnancy-associated malaria	BJ, DK, FR, NL, SE, TZ	FR	Malaria
SYSCILIA: A systems biology approach to dissect cilia function and its disruption in human genetic disease	DE, FR, IE, IT, NL, US	NL	Retinal ciliopathy
			Nephronophthisis-associated ciliopathy
SYSTEMTB: Systems biology of Mycobacterium tuberculosis	CH, CZ, DE, ES, FR, GB, IT, NL, PL	ES	Tuberculosis
TB PAN-NET: Pan-European network for the study and clinical management of drug resistant tuberculosis	BE, CH, DE, DK, GB, HR, EE, FI, FR, IT, LT, LV, NO, PL, RO, SE, US	IT	Tuberculosis
TB-EURO-GEN: Genetic analysis of the host-pathogen interaction in tuberculosis	DE, GB, SE	GB	Tuberculosis
TBSUSGENT: Sustaining research momentum over the coming decades: mentoring the next generation of researchers for tuberculosis	CA, DE, GB, IN, IT, ZA	GB	Tuberculosis
TB-VIR: Mycobacterium tuberculosis W-Beijing genetic diversity and differential virulence and host immune responses	CH, DE, ES, FR, KR	FR	Tuberculosis
TECHGENE: High throughput molecular diagnostics in individual patients for genetic diseases with heterogeneous clinical presentation	BE, CZ, DE, EE, ES, FR, IT, NL	NL	Rare genetic disease
TM-REST: A new platform for fast molecular detection of MDR and XDR resistant strains of M. tuberculosis and of drug resistant malaria	AL, BG, CH, DE, GB, IT	IT	Tuberculosis
TRANSMALARIABLOC: Malaria transmission blocking by vaccines, drugs and immune mosquitoes: efficacy assessment and targets	BF, FR, GB, GR, IT, UG	GB	Malaria
TREATRUSH: Fighting blindness of Usher syndrome: diagnosis, pathogenesis and retinal treatment (TreatRetUsher)	CH, DE, FR, GB, IT, NL, US	FR	Usher syndrome
TRYPOBASE: nucleobase derivatives as drugs against trypanosomal diseases	CH, ES, IN, SE, UY	ES	American trypanosomiasis
			African trypanosomiasis

## E-RARE CONSORTIUM FUNDED NETWORKS (29 NETWORKS)

Calls of 2007 and 2009

NETWORK ACRONYM / TITLE	PARTICIPATING COUNTRIES	COORDINATING COUNTRY	CONCERNED DISEASES
CAV-4-MPS: understanding and treating neurodegeneration caused by mucopolysaccharidoses	DE, ES, FR	ES	Mucopolysaccharidosis
CRANIRARE: an integrated clinical and scientific approach for craniofacial malformations	DE, FR, TR	DE	Cranial malformation
CUREFXS: targeting Rho-signalling, a new therapeutic avenue in fragile-X syndrome	DE, ES, FR, IT, NL	ES	Fragile X syndrome
ELA2-CN: congenital neutropenia with ELA-2 mutations (ELA2-CN), identification of (epi)genetic co-factors and molecular pathways underlying clinical heterogeneity	DE, IL, NL, TR	DE	Autosomal dominant severe congenital neutropenia
			Neutropenia cyclic
EMINA: European multidisciplinary initiative on neuroacanthocytosis	AT, DE, FR, NL, TR	DE	Neuroacanthocytosis
EPINOSTICS: "autoimmune liver diseases" epitope peptide mapping - The entry to novel and innovative diagnostic and therapeutic applications	DE, ES, FR, IS, IT, GB, GR	DE	Chronic autoimmune hepatitis
ERMION: European research project on Medelian inherited optic neuropathy	DE, FR, IT	FR	Genetic optic neuropathy
EUPAPNET: European pulmonary alveolar proteinosis network: molecular determinants of causes, variability and outcome	DE, IT, NL	IT	Pulmonary alveolar proteinosis
EUROBNFS: Benign Familial Neonatal Seizures (BNFS) as a disease model for human idiopathic epilepsies: expansion of genotype-phenotype correlations and insights into novel disease mechanisms	DE, ES, FR, IT	IT	Benign familial neonatal seizures
EURO-CGD: genetics and pathogenesis of chronic granulomatous disease and development of new gene transfer therapeutic approaches	IT	IT	Chronic granulomatous disease
EUROGEBETA: European network on genetics, pathophysiology and translational research into rare pancreatic beta-cell insufficiency diseases	ES, FR, IL, IT	FR	Rare pancreatic beta cell insufficiency diseases
EURORETT: European network on Rett syndrome	DE, ES, FR, IS, IT	FR	Rett syndrome
EUROSPA: European & Mediterranean network on spastic paraplegias	DE, FR, IS, IT	FR	Familial spastic paraplegia

GETHERTHAL: improvements of vector technology and safety for the gene therapy of thalassemia	DE, FR, GR	GR	Alpha-thalassemia
			Beta-thalassemia
HAE III: genetics, pathophysiology, and therapy of hereditary angioedema type III	DE, FR, IT	DE	Hereditary angioedema type 3
HMA-IRON: towards improved diagnosis and treatment of rare inherited microcytic hypochromic anemias related to iron metabolism	DE, ES, FR, IT	FR	Sideroblastic anemia
			Microcytic anemia with liver iron overload
HSCR: International Hirschsprung disease consortium	ES, FR, IT, HK, NL, US	FR	Hirschsprung disease
KINDLERNET: International Kindler syndrome network	DE, FR, IT	DE	Poikiloderma of Kindler
MLC-TEAM: megalencephalic leukoencephalopathy with subcortical cysts: from molecular basis to search for therapy	ES, NL	ES	Megalencephaly - cystic leukodystrophy
MTMPATHIES: myotubularinopathies: common molecular mechanism and tissue specificity	FR, IT	FR	X-linked centronuclear myopathy
NEMMYOP: functional characterisation of the nemaline myopathy in a murine model with the nebulin mutation: moving from basic understanding towards therapeutic interventions	DE, FR, NL	FR	Nemaline myopathy
NEUTRONET: inherited inhibition of inborn immunity: an integrated molecular genetic approach to discover novel human gene defects	AT, DE, FR, TR	DE	Rare genetic immune disease
NSEURONET: European network on Noonan syndrome and related disorders	DE, FR, IT, NL	IT	Noonan syndrome and Noonan-related syndrome
OSTEOPETR: new genes and therapeutic approaches to osteopetrosis	DE, FR, IT	IT	Osteopetrosis
PODONET: consortium for clinical, genetic and experimental research into hereditary diseases of the podocyte	DE, FR, IT, TR	DE	Familial idiopathic steroid-resistant nephrotic syndrome
			Sporadic idiopathic steroid-resistant nephrotic syndrome
REVERTANTEB: identification of revertant mosaicism in epidermolysis bullosa and subsequently using the revertant keratinocytes in a pre-clinical mouse model suitable to test revertant cell therapy	DE, ES, NL	NL	Hereditary epidermolysis bullosa

RISCA : Prospective study of individuals at risk for spinocerebellar ataxia type 1, type 2, type 3 and type 6 (SCA1, SC2, SCA3, SCA6)	DE, ES, FR, IT	DE	Spinocerebellar ataxia type 1
			Spinocerebellar ataxia type 2
			Spinocerebellar ataxia type 3
			Spinocerebellar ataxia type 6
RHORCOD: comprehensive analysis of rod-cone photoreceptor degeneration associated with Rhodopsin gene mutations	AT, DE, FR, IL, IT, PT	FR	Congenital stationary night blindness
			Retinitis pigmentosa
WHIMPATH: understanding the WHIM syndrome and search for new therapies: molecular analysis of CXCR4 in leukocyte trafficking and activation	ES, FR, IT	IT	WHIM syndrome

### DG SANCO FUNDED NETWORKS (23 NETWORKS)

2003 - 2008

NETWORK ACRONYM / TITLE	PARTICIPATING COUNTRIES	COORDINATING COUNTRY	CONCERNED DISEASES
Alzheimer Europe: European database on rare forms of dementia	AT, DE, FR, GB, LU, NL, RO	LU	Rare dementia
Dyscerne: a European network of centres of expertise for dysmorphology	BE, BG, CH, CY, CZ, DE, DK, ES, FI, FR, GB, GR, IE, IL, IT, LV, MK, NL, NO, PL, PT, RO, SE, SI, SK, TR	GB	Rare developmental defect during embryogenesis
ECORN-CF - European centres of reference network for cystic fibrosis	BE, CZ, DE, GB, GR, LT, NL, PL, RO, SE	DE	Cystic fibrosis
ENERCA: European network for rare and congenital anaemias	BE, CH, CY, DE, ES, FR, GB, GR, IT, NL, PL, PT, RO, RS, SE	ES	Rare constitutional anemia
EPI: European Porphyria Initiative	BE, CH, CZ, DE, DK, ES, FI, FR, GB, HU, IE, IL, IT, NL, NO, PL, SE	FR	Porphyria
EPNET: European network on centre of reference on porphyria	BE, CH, CZ, DE, FI, ES, FR, GB, HU, IE, IT, NL, NO, PL, SE	FR	Porphyria
ERCUSYN: European registry on Cushing's syndrome	AT, BE, BG, CH, CZ, DE, EE, ES, FR, GB, GR, HU, IE, IL, IR, IS, IT, LV, NL, NO, PL, PT, RO, SE, SI, TR	ES	Cushing syndrome
EU-CHS: European network for central hypoventilation syndromes: optimizing health care to patients	DE, FR, IT, PL, SE	FR	Ondine syndrome
EUHASS: European Haemophilia Safety Surveillance	BE, GB, IT, NL	GB	Hemophilia

EUROCAT: European surveillance of congenital anomalies	AT, BE, BG, CH, CY, DE, DK, ES, FI, FR, GB, HR, HU, IE, IT, LU, MT, NL, NO, PL, PT, SI	GB	Congenital anomalies
EUROFEVER: PReS European network of registries for autoinflammatory diseases in childhood	BE, DE, FR, GB, IT, NL, TR	IT	Behcet disease
			Blau syndrome
			Familial Mediterranean fever
			Muckle-Wells syndrome
			Marshall's syndrome with periodic fever
Partial mevalonate kinase deficiency with recurrent fever +/- hyperIgD			
EURO HISTIO NET: study group on histiocytosis	AT, ES, FR, GB, IT	FR	Langerhans cell histiocytosis
EUROMUSCLENET: European information network on muscle diseases	BE, DE, DK, ES, FI, GB, IT, NL	DE	Neuromuscular disease
European network of paediatric Hodgkin's lymphoma - European-wide organisation of quality controlled treatment	AT, CZ, DE, DK, ES, FR, GB, IE, NO, PL, SE	DE	Hodgkin lymphoma
IDR: ImmunoDeficiency Resource	AT, CA, DE, DK, ES, FI, FR, GB, IE, IT, NL, NO, NZ, SE, US	FI	Primary immunodeficiency
NEUROPED: European network of reference for rare paediatric neurological diseases	AT, BE, CZ, DE, DK, ES, FR, GB, IT, NL, NO, PT, SI	AT	Alternating hemiplegia of childhood
			Childhood-onset epilepsy syndrome
			Congenital hypothalamic hamartoma syndrome
			Infantile epilepsy syndrome
			Narcolepsy without cataplexy
			Narcolepsy-cataplexy
			Neonatal epilepsy syndrome
Sturge-Weber syndrome			
NF-EUROPE: European Federation For Neurofibromatosis Associations	BE, BG, CH, DE, DK, ES, FI, FR, IT, NL, NO, PT, SE	DE	Neurofibromatosis - Noonan syndrome
			Neurofibromatosis type 1
			Neurofibromatosis type 2
			Neurofibromatosis type 6
PAAIR: Patient's Association and Alpha-1 International Registry network	AT, BE, CH, CZ, DE, DK, EE, ES, GB, IT, LT, LV, NL, PL, SE	NL	Alpha-1 antitrypsin deficiency

PRINTO: Paediatric Rheumatology International Trials Organisation	AT, BE, CH, CZ, DE, DK, ES, FI, FR, GB, GR, HU, IE, IL, IT, LU, LV, NL, PT, SE, SK, TR	IT	-
RARECARE: surveillance of rare cancers in Europe	AT, BE, CH, DE, EE, ES, FI, FR, GB, HR, IE, IS, IT, MT, NL, NO, PL, PT, SE, SI, SK	IT	Rare tumor
RBDD: Rare Bleeding Disorders Database network	BE, BR, CH, CN, CO, DE, DK, EG, ES, FR, GB, GR, HU, IN, IQ, IR, IT, KW, NO, NZ, OM, PA, PE, PL, PT, RU, RS, SI, TH, TR, US, VE, VN	IT	Combined deficiency of factor V and factor VIII
			Congenital factor II deficiency
			Congenital factor V deficiency
			Congenital factor VII deficiency
			Congenital factor X deficiency
			Congenital factor XI deficiency
			Congenital factor XIII deficiency
			Congenital fibrinogen deficiency
Congenital vitamin K-dependent coagulation factors deficiency			
SCNIR: severe chronic neutropenia international registry	AT, BE, CZ, DE, ES, FR, GB, GR, HU, IE, IL, IT, NL, NO, PL, PT, RS, SE, TR	DE	Severe chronic neutropenia
TAG: TogetherAgainstGenodermatoses - improving health care and social support for patients and family affected by severe genodermatoses	CY, FR, GR, IT, PT, RO, SI, TR	FR	Familial palmoplantar keratoderma
			Genetic ichthyosis
			Hereditary epidermolysis bullosa
			Neurofibromatosis type 1
			Xeroderma pigmentosum

For any questions or comments, please contact us: [contact.orphanet@inserm.fr](mailto:contact.orphanet@inserm.fr)

Editor-in-chief: Ségolène Aymé • Editors of the report: Nicolas Doulet, Charlotte Rodwell • Visual design: Céline Angin

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