Overview of current Centres of Reference on rare diseases

in the EU

ANNEXES

Report to the High Level Group
on Health Services and Medical Care

ANNEX 1

List of members of the working group from the Task Force of Rare Diseases

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ANNEX 2

UK National Specialist Commissioning Advisory Group (NSCAG) Services: http://www.advisorybodies.doh.gov.uk/NSCAG/services.htm

SERVICE	Disease(s)	ADDRESS
Amyloidosis	Amyloidosis	Royal Free Hospital, London
Bladder exstrophy	Bladder exstrophy	Great Ormond Street Hospital, London
		Manchester Children's Hospital, Manchester
Choriocarcinoma	Choriocarcinoma	Charing Cross Hospital, London
		Weston Park Hospital, Sheffield
Craniofacial surgery	Craniosynostosis Craniofacial dystosis Orbital dystopia Encephacoele Craniofacial clefts	Great Ormond Street Hospital, London
		Birmingham Children's Hospital, Birmingham
		Radcliffe Infirmary, Oxford
		Royal Liverpool Children's Hospital (Alder Hey), Liverpool
Deep brain stimulation for severe Parkinson's disease	Parkinson's disease	Radcliffe Hospital, Oxford
		Charing Cross Hospital, London
		Queen Elizabeth Hospital, Birmingham
		Frenchay Hospital, Bristol
		Walton Unit for Neurology & Neurosurgery, Liverpool
		King's College Hospital, London
		The National Hospital for Neurology & Neurosurgery, London
		Newcastle
		Sheffield
		Salford
Epidermolysis bullosa service	Epidermolysis bullosa	Birmingham Children's Hospital, Birmingham
		Great Ormond Street Hospital, London
		Guy's & St. Thomas' Hospital, London
		Birmingham Heartlands & Solihull Hospitals, Birmingham
Liver disease service for children	Rare liver diseases inc: Acute liver failure Biliary atresia Chronic liver disease Hepatitis A, B and C Metabolic liver disease Neonatal hepatitis	Birmingham Children's Hospital
		King's College Hospital, London
		St. James' University Hospital, Leeds
Lysosomal storage disorders	Lysosomal storage disorders e.g. Hurler's syndrome Gaucher's disease	Addenbrookes Hospital, Cambridge
		Royal Free Hospital, London
		Great Ormond Street Hospital, London
		Central Manchester and Manchester Children's Hospital, Manchester
		Hope Hospital, Salford
		University College Hospital, London
Ophthalmic pathology	Diseases of the eye and ocular adjoining tissues	Institute of Ophthalmology, University College Hospital, London
		Royal Liverpool University Hospital, Liverpool
		Manchester Royal Infirmary, Manchester
		Royal Hallamshire Hospital, Sheffield
Primary malignant bone tumours	Osteosarcoma Chondrosarcoma Ewing's sarcoma	Avon Orthopaedic Centre, North Bristol NHS Trust
		Nuffield Orthopaedic Centre, Oxford
		Newcastle upon Tyne Hospitals NHS Trust, Newcastle
		Robert Jones & Agnes Hunt Hospital, Oswestry
		Royal National Orthopaedic Hospital, Stanmore
		Royal Orthopaedic Hospital, Birmingham
		University College London Hospitals Foundation NHS Trust, London
		Royal Orthopaedic Hospital, Birmingham
		University College London Hospitals Foundation NHS Trust , London
Pseudomyxoma peritonei surgery	Pseudomyxoma peritonei	North Hampshire Hospital, Basingstoke
		Christie Hospital, Manchester

SERVICE	Disease(s)	ADDRESS
Pulmonary hypertension	Pulmonary hypertension	Royal Free Hospital, London
		Freeman Hospital, Newcastle
		Royal Brompton Hospital, London
		Royal Hallamshire Hospital, Sheffield
		Hammersmith Hospital, London
		Papworth Hospital, Cambridge
		Great Ormond Street Hospital, London
Pulmonary thromboendarterectomy	Pulmonary hypertension	Papworth Hospital, Cambridge
Rare Neuromuscular diseases	Limb girdle muscular dystrophies Congenital muscular dystrophies Congenital myasthenic syndromes Muscle channelopathies	Institute of Human Genetics, Newcastle
		University Medical School (lab services), Newcastle
		John Radcliffe Hospital, Oxford
		Churchill Hospital (lab services), Oxford
		Hammersmith Hospital, London
		Guy's Hospital (lab services), London
		National Hospital for Neurology and Neurosurgery, London (Queens Square)
Reconstructive surgery in adolescents for congenital malformation of the female genital tract	Rare congenital conditions involving malformation of the female genital tract	Queen Charlotte's and Chelsea Hospital, Du Cane Road, London
Retinoblastoma	Retinoblastoma	St. Bartholomew's Hospital, London
		Birmingham Children's Hospital, Birmingham
Severe combined immunodeficiency and related disorders (SCIDS)	SCIDs	Great Ormond Street Hospital, London
		Royal Victoria Infirmary, Newcastle
Severe intestinal failure	Crohn's Short bowel syndrome Severe pancreatitis	St. Mark's Hospital, London
		Hope Hospital, Salford
Stem cell transplantation for juvenile idiopathic arthritis and related conditions	Juvenile idiopathic arthritis	Great Ormond Street Hospital, London
		Freeman Hospital, Newcastle

ANNEX 3

Centres of reference in Belgium in the field of rare diseases

CENTRES OF REFERENCE FOR NEUROMUSCULAR DISEASES

- Neuromuscular Centre of Reference AZ-VUB	AZ VUB
- Neuromuscular Referral Center	Antwerpen
- Neuromuscular Centre of Reference UZ Leuven	UZ Leuven
- Neuromuscular Centre of Reference UZ Gent	UZ Gent
- Centre de Référence neuromusculaire Liègeois	CHR Citadelle
- Centre de Référence neuromusculaire	UCL Saint Luc

CENTRES OF REFERENCE FOR HEREDITARY METABOLIC DISEASES

- Centre of reference for hereditary metabolic diseases	CEMA-UZ-Leuven
- Center for Metabolic Disease	UZ Leuven Gasthuisberg
- Center for Metabolic Disease	UZ Gent
- Centrum Erfelijke Metabole Aandoeningen	UZ Antwerpen

CENTRES OF REFERENCE OFR CREUTZFELD JACOB DISEASE

- Université de liège CHU Sart Tilman
- Katholieke Universiteit Leuven Gasthuisberg
- Université catholique de Louvain, cliniques universitaires Saint-Luc
- Vrije Universiteit Brussel
- Université libre de Bruxelles Hopital Erasme
- Universiteit Gent UZ de Pintelaan
- Universitaire Instelling Antwerpen

AGREED CENTRES FOR DIAGNOSE AND STUDY OF GENETIC DISEASES

The characteristics and amount of these centers are regulated by law

- Centre de génétique de l'ULB - Campus Erasme - Route de Lennik, 808 - 1070 Bruxelles
- Centre de génétique de l'UCL - Tour Vésale 5220 - Avenue Mounier, 52 - 1200 Bruxelles
- Centre Universitaire Wallon de génétique - CHU du Sart Tilman - 4000 Liège
- Institut de pathologie et de génétique - Allée des Templiers, 4 - 6280 Loverval
- Kliniek Universitair Ziekenhuis - Centrum voor menselijke erfelijkheid - 3000 Leuven
- Centrum Medische Genetica - UI Antwerpen - 2610 Wilrijk
- Centrum voor Medische Genetica - UZ Gent - 9000 Gent

ANNEX 4

French Centres of Reference for rare diseases established in 2004 and expected number at the end of the designation process.

MEDICAL AREA	EXPECTED NUMBER	ALREADY DESIGNATED CENTRES
Autoimmune dis. + rare systemic	5 to 7	Centre of reference for necrotising vascularitis and systemic sclerodermia
		Centre of reference for Lupus and antiphospholipid Syndromes
Cardiovascular	3 to 4	Centre of reference for hereditary cardiac diseases
		Centre of reference for genetic cardiac rythm anomalies
Developmental Anomalies	4	Centre of reference for developmental anomalies
		Centre of reference for genetic developmental anomalies
Dermatology	3 to 4	Centre of reference fo acquired toxic and immunologic epidermolysis bullosa
		Centre of reference for genetic dermatologic diseases
Endocrine dis.	3 to 4	Centre of reference for Prader-Willi syndrome
Hepato-gastro-Enterology	3 to 4	Centre of reference for intestinal diseases
		Centre of reference for inflamatory biliary diseases
Hematologic, non Malignant	5 to 7	Centre of reference of genetic diseases of the erythrocyte and of erythropoiesis, except sickel cell disease
		Centre of reference for sickel cell disease
		Centre of reference for porphyrias
Metabolic dis	3 to 4	Centre of reference for inborn errors of metabolism
		Centre of reference for renal and metabolic disorders
		Centre of reference for paediatric metabolic disorders
Neurological dis.	3 to 4	Centre of reference for Huntington disease
		Centre of reference for lysosomal disorders with a neurological expression
		Centre of reference for neuromuscular diseases and amyotrophic lateral sclerosis
Neuro-muscular	5 to 7	Centre of reference for neuromuscular disorders
		Centre of reference for neuromuscular disorders
Pulmonary dis.	5 to 7	Centre of reference for severe pulmonary hypertension
		Centre of reference for Ondine syndrome
		Centre of reference for orphan pulmanory diseases
Ophthalmologic	1 to 2	Centre of reference for sensorial diseases
		Centre of reference of genetic ophthalmologic disorders
Renal dis.	3 to 4	Centre of reference for renal hereditary disorders
Bone diseases	1 to 2	Centre of reference for constitutional bone diseases
Immune dis.	1 to 2	Centre of reference for hereditary immune deficits
Conjonctive tissue	3 to 4	Centre of reference for Marfan syndrome
		Centre of reference for Rendu-Osler disease
Deafness	1 to 2	Centre of reference for congenital and hereditary Deafness
Others	?	Centre of reference for neurofibromatosis
Total	**80 to 100**	**34**

ANNEX 5

Groups of Rare Diseases for which 228 Italian Regional Centres were established by Official Regional Decisions following the Governmental Regulation on Rare Diseases (D.M. 271/2001)

Infectious and parasitic diseases
Neoplasms
Endocrine glands, nutritional, metabolic diseases and immune disorders
Diseases of the blood and hemopoietic organs
Diseases of the nervous system and sense organs
Diseases of the circulatory system
Diseases of the digestive system
Diseases of the genitourinary system
Diseases of the skin and subcutaneous tissue
Diseases of the musculoskeletal system and connective tissue
Congenital malformations
Certain conditions originating in the perinatal period
Symptoms, signs not elsewhere classified

ANNEX 6

National referral centres for rare disease
designated by the National Board of Health of Denmark

Clinic for rare disease, Rigshospitalet, Copenhagen

Galactosaemia
Neurofibromatosis Recklinghausen
Osteogenesis imperfecta
Marfan syndrome
Prader -Willi syndrome
Ehlers- Danlos syndrome
Craniofacial anomalies
Bladderexstrophy
Myelomeningocele/ spina bifida

Centre for rare disease, Århus Universitetshospital, Skejby Sygehus, Århus.

Morbus Wilson
Spielmeyer-Vogt and other neuronale ceroid-lipofuscinosis
Neurofibromatosis Recklinghausen
Osteogenesis imperfecta
Marfan syndrome
Prader -Willi syndrome
Ehlers- Danlos syndrome
Craniofacial anomalies
Bladderexstrophy
Myelomeningocele/ spina bifida

Both centres have expertise and accept patients with several other rare diseases

ANNEX 7

Questionnaire Orphanet

Return this form by:

Fax *: 0161 276 4058*

Post *: Emma Gillaspy, European Projects Office, The Nowgen Centre, Grafton Street, Manchester, M13 9WU*

E-mail *: emma.gillaspy@cmmc.nhs.uk*

SPECIALISED OUTPATIENT CLINIC FOR RARE DISEASES*

Form to be filled in by the consulting physician
(Fill in a separate form for each different address and type of outpatient clinic)

* A disease is considered to be rare when it affects less than 1 person in 2,000.

Name of the Outpatient Clinic

Type of Outpatient Clinic

☐ Genetic Counselling Clinic ☐ Cytogenetics Clinic ☐ Dysmorphology Clinic

☐ Specialised Clinic (please give details of the rare disease or group of rare diseases treated).
Details:

Patients Treated at the Clinic

☐ Adults only ☐ Children only ☐ Adults and Children

Consulting Physician(s):
For genetic counselling, cytogenetic and dysmorphology outpatient clinics, please only indicate the name of the **coordinating doctor.**
For specialised outpatient clinics, please indicate **two names maximum.**

☐ Mr ☐ Mrs☐ Ms

First name: Middle initial: Last name:

Title : ☐ Dr ☐ Prof ☐ Other

Professional degree: ☐ MD ☐ PhD.....☐ Other

Additional information only for Orphanet team (not for online publication)
Phone number (direct line or mobile): .....
Personal e-mail:

☐ Mr ☐ Mrs☐ Ms

First name: Middle initial: Last name:

Title : ☐ Dr ☐ Prof ☐ Other

Professional degree: ☐ MD ☐ PhD.....☐ Other

Additional information only for Orphanet team (not for online publication)
Phone number (direct line or mobile): .....
Personal e-mail:

Address of the Outpatient Clinic

Acronym of the service :

Full name if the service:

Address of the service:

Secretary phone number : Fax number:

Secretary e-mail :

Website URL: http://

Affiliation: ☐ Private Hospital ☐ NHS Trust Hospital
☐ University ☐ University Funded Research Institute
☐ NHS Funded Research Institute ☐ Public or Charity Funded Research Institute
☐ Pharmaceutical Company ☐ Private Company
☐ Biotechnology Institute

Status : ☐ Public institution ☐ Private/Independent ☐ Private non for profit

Setting : ☐ Research setting ☐ Clinical setting

Department/ Institution (if applicable):
Acronym :

Full name of department:

Director's name of the department:

Hosting institution's name:

PLEASE INDICATE INFORMED CONSENT FOR RELEASE OF THIS INFORMATION ON THE ORPHANET WEBSITE BY SIGNING THE BOX BELOW.

According to the electronic data protection law, your consent is required before diffusion of any nominative information. You have all rights to access, opposition to, or correction of these data. Phone numbers and e-mail addresses may be not available to the public. You need only to contact the director of the Orphanet database, Dr Ségolène Aymé. The file composed from this information is declared at the French electronic data protection Committee.

I agree to the terms and conditions for publication of this information on the ORPHANET database website.

Date: Signature:

Return this form by:

Fax : *0161 276 4058*
Post : *Emma Gillaspy, European Projects Office, The Nowgen Centre, Grafton Street, Manchester, M13 9WL*
E-Mail : *emma.gillaspy@cmmc.nhs.uk*

ACTIVITY OF THE SPECIALISED OUTPATIENT CLINIC

Form to filled in by the consulting Physician, intended for the scientific committee
(fill in a separate form for each type of outpatient clinic)

Name of the rare disease or group of rare diseases:

Data related to the whole team:

1) **Publications and communications (conferences, etc.) over the past five years of the consulting physician(s)**

- Number of publications on the disease(s)
- Number of communications on the disease(s)

2) Number of patients

- Total number of patients seen
- Number of new cases last year

3) What technical tools do you have in your institution to establish the diagnosis?

4) Is your clinic multi-disciplinary?

5) Do you have a research program financed by a public or private agency for this (or these) disease(s)?

6) To what learned societies do you belong?

7) Are you a member of the scientific committee of a patient support group?

8) Are you officially a reference centre?

ANNEX 8

National networks of expert centres in the field of rare diseases

NATIONAL FRENCH NETWORKS

- Réseau national pour la prise en charge clinique et génétique des parangliomes et des phéochromocytomes SDH-dépendants
- Mise en place d'un réseau français pour le dépistage familial de l'hémochromatose génétique - Recherche de gène modificateurs
- Réseau bio-clinique de recherche et d'étude de l'Hyperthermie Maligne Peranesthésique et du Central Core Disease
- Réseau de recherche clinique et biologique sur le Syndrome d'Evans et l'anémie hémolytique autoimmune chez l'enfant
- Réseau des dystrophies musculaires d'Emery_Dreifuss et autres pathologies de l'enveloppe nucléaire
- Réseau de recherche sur la maladie de Pompe (Glycogénose de type II) - du dépistage à la thérapie -
- Réseau de recherche sur les formes autosomiques récessives de la maladie de Charcot-Marie-Tooth
- Réseau de soin des maladies vasculaires du foie - Syndrome de Budd chiari et thrombose porte.
- Slérose en plaques et maladies inflammatoires du système nerveux Ile de France Sud Est
- Réseau de recherche sur les maladies musculaires des canaux ioniques et apparentées
- Diagnostic immunologique, enzymologique et moléculaire du syndrome de Netherton et des ichtyoses lamellaires
- Réseau des dysmorphies musculaires d'Emery Dreifuss et autres nucléopathies
- Étude de cohorte des déficits immunitaires primitifs avec hypogammaglobulimémie de l?adulte
- Réseau de recherche dans la sclérose latérale amyotrophique familiale
- Réseau clinique de recherche sur la Néoplasie endocrinienne multiple de type 1
- Surdités héréditaires : isolement de gènes responsables, descriptions cliniques et physiopathologies de certaines formes
- Cavernomes héréditaires : spectre clinique, identification des gènes et exploration des mécanismes physio-pathologiques
- Réseau européen des registres de surveillance des anomalies congénitales rares
- Pseudohermaphrodisme masculin et autres troubles de développement sexuel
- Association Française pour le Dépistage et la Prévention des Handicaps de l'Enfant
- Réseau sur les Maladies Héréditaires de la Production et des Fonctions Plaquettaires
- Maladies génétiques rares de l'érythropoïèse et de la membrane érythrocytaire
- Maladie de Hirschsprung : étude clinique et moléculaire d'une malformation d'origine multigénique
- Réseau d'étude des retards mentaux et déficits cognitifs monogéniques
- Réseau de recherche sur les syndromes néphrotiques cortico-résistants
- Analyse phénotypique et génotypique des patients du registre français des neutropénies congénitales
- Dystrophie myotonique de Steinert : de la clinique à la thérapie
- Maladies de surcharge lysosomales : prévalence, histoire naturelle et thérapeutiques émergentes
- Réseau d'Innovation Thérapeutique en Oncologie Pédiatrique (réseau ITCC)
- Réseau Myopathies des ceintures - Limb-girdle-muscular dystrophies - LGMD
- Réseau pour le syndrome Marfan et les pathologies proches non syndromiques
- Hypertension artérielle pulmonaire primitive et secondaire, physio-pathologie et thérapeutique
- Réseau européen de centres de référence sur la porphyrie aiguë intermittente
- Réseau Neurofibromatose
- Réseau France Coagulation
- Réseau des Mastocytoses
- Réseau sur les ichtyoses
- Réseau bio-clinique de recherche et d'étude du syndrome de Lowe
- Réseau sur les maladies inflammatoires récurrentes héréditaires
- Réseau européen de recherche sur les acidoses tubulaires rénales
- Compréhension et traitement des atteintes neurologiques dans les mucopolysaccharidoses
- Réseau de recherche sur la maladie de Gilles de la Tourette
- Association francophone pour l'étude de la maladie de Wilson
- Encéphalite herpétique : étude épidémiologique, clinique, virologique et génétique
- Réseau Européen pour l'étude des tumeurs de la surrénale
- Réseau de recherche génétique sur la sclérose en plaques
- Réseau COMETE sur les tumeurs endocrines de la surrénale
- Réseau des Dystrophies musculaires congénitales - DMC
- Réseau national de recherche sur l'Aplasie Müllérienne
- Physiopathologie de la myasthénie
- Genostem
- Réseau national sur les dystonies
- Groupe d'étude des histiocytoses
- Réseau sur les neurofibromatoses
- Groupe français des myélodysplasies
- Réseau d'étude du complexe de Carney
- Groupe de recherche sur les Epilepsies
- Réseau Huntington de langue française
- Maladies génétiques rares de la peau
- Groupe d'étude français des vascularites
- Maladies mitochondriales humaines
- Réseau PHA 1
- Réseau des dysplasies du squelette
- Réseau national « Maladies Mitochondriales »
- Réseau sur les atrophies optiques dominantes
- Réseau de recherche sur le Syndrome de Brugada
- Réseau sur les leucémies aiguës a mégacaryocytes
- Groupe d'étude francophones des tumeurs endocrines
- Réseau sur les dégénérescences spino-cérébelleuses
- Réseau sur les maladies osseuses constitutionnelles
- Réseau sur les épidermolyses bulleuses héréditaires

- Eurowilson - registre européen de la maladie de Wilson
- Réseau d'étude des anomalies du développement cortical
- Réseau de recherche sur le groupe des Congenital Disorders of Glycosylation CDG

NATIONAL GERMAN NETWORKS

- Informationszentrum Epilepsie
- Deutsches MITONET e.V.
- Kompetenznetz Parkinson
- Muskeldystrophie-Netzwerk
- NCL-Stiftung
- LEUKONET
- METABNET
- CJD-Netz
- Myelin Projekt Deutschland e.V.
- Deutsches Down-Syndrom InfoCenter
- Deutsche Gesellschaft für Angioödeme e.V.
- Deutsches Netzwerk für erbliche Bewegungsstörungen
- Kompetenznetz Pädiatrische Onkologie und Hämatologie
- Netzwerk Hypophysen- und Nebennierenerkrankungen e.V.
- European integrated project on spinocerebellar ataxias
- Deutsche Gesellschaft für Hämatologie und Onkologie e.V.
- EU-Projekt Rare Lung Diseases - Seltene Lungenerkrankungen
- Netzwerk für Ichthyosen und verwandte Verhornungsstörungen
- Kompetenznetz entzündlich rheumatischer Systemerkrankungen
- European Network on Tropheryma whipplei infection
- Kompetenznetzwerk Epidermolysis bullosa (EB-Netz)
- Netzwerk für Angeborene Störungen der Blutbildung
- Deutsches Netzwerk für Systemische Sklerodermie
- Kompetenznetz "Akute und chronische Leukämien"

NATIONAL SPANISH NETWORKS

- Epidemiología, fisiopatología y caracterización clínica y molecular de las distrofias hereditarias de retina
- Aplicaciones de la biología molecular y celular al diagnóstico y tratamiento de enfermos con anemia de Fanconi
- Identificación de factores de riesgo y caracterización de arbovirosis y robovirosis en España
- Red de Investigación Cooperativa sobre las enfermedades de la cadena mitocondrial en España
- Mieloma múltiple y otras gammapatías. De la génesis a la terapéutica
- Red Nacional de Investigación Cooperativa sobre la Anemia de Fanconi
- Instituto de Investigación de Enfermedades Raras de Base Genética
- Red Nacional de Investigación en Hepatología y Gastroenterología
- Grupo de Investigación en Retraso Mental de Origen Genético
- Red Epidemiológica de Investigación sobre Enfermedades Raras
- Bases genéticas y moleculares de los trastornos de la audición
- Red Española para el estudio de las enfermedades hepáticas
- Mecanismos humorales y celulares implicados en las artritis
- Red Española de Mastocitosis
- Gimferrer's iron study team
- Centro Nacional de Microbiología
- Red de Genotipación y Psiquiatría Genética
- Red para la investigación de la Brucelosis
- Centro de Investigación de Enfermedades Neurológicas
- Biología, clínica y terapia de las ataxias cerebelosas
- Red de Centros de Genética Clínica y Molecular
- Red de Enfermedades Metabólicas Hereditarias

NATIONAL ITALIAN NETWORKS

- Alleanza per le malattie rare - Università Policlinico Federico II Napoli
- Associazione Italiana Ematologia Oncologia Pediatrica
- Gruppo Italiano studi epidemiologici in Dermatologia (GISED)
- Network Italiano Acondroplasia
- Network Italiano Fibrosi Cistica
- Network Italiano Promozione acido folico per la prevenzione di difetti congeniti
- Network Nazionale alfa - 1 – antitripsina
- Network delle malattie metaboliche
- Network delle malattie lisosomiali
- Pediatric Rheumatology International Trials Organisation (PRINTO)
- Registro Campania Difetti Congeniti
- Registro IMER (Indagine Malformazioni Congenite Emilia Romagna)
- Registro ISMAC (Indagine Siciliana Malformazioni Congenite)
- Registro Italiano Anemia di Fanconi
- Registro Italiano della Febbre Mediterranea Familiare
- Registro italiano Malattie Mitocondriali
- Registro Italiano Sclerosi Laterale amiotrofica
- Registro Lombardia Malformazioni Congenite
- Registro Malattie Rare Regione Marche
- Registro Malattie Rare Regione Veneto
- Registro nazionale degli assuntori di ormone della crescita
- Registro nazionale degli ipotiroidei congeniti
- Registro nazionale della Legionellosi

ANNEX 9

Examples of European reference networks funded by DG Sanco

- Rare pulmonary diseases - Establishment of diagnostic criteria and reference/training centers
- Paediatric rheumatic diseases A European information network on
- Severe Chronic Neutropenia: European network on the epidemiology, pathophysiology and treatment
- Congenital Anaemias: European Network for Rare and Congenital Anaemias
- Metabolic diseases: Ongoing transfer of expertise on prevention, diagnosis and treatment of common complications in adults with rare metabolic diseases
- Rare pulmonary diseases - Establishment of diagnostic criteria and reference/training centres
- Immunodeficiencies: Information Network
- Network of Public Health Institutions on Rare Diseases: European Network on centres working on epidemiology, public health indicators and quality of life of select rare diseases

Examples of European reference networks funded by DG Research (FP5 and 6)

- European Skeletal Dysplasia network
- Wilson disease: network for early clinical trials
- Rare hereditary neurological disorders: research network
- Rare autoimmune disorders: research network
- Rare genetic skin disorders: research network
- Prader-Willi syndrome: research network
- Rare diseases of nuclear organisation: research network
- Cystic fibrosis: research network
- Mitochondrial diseases: research network
- European network for research on alternating hemiplegia in childhood
- Task force in Europe for drug development for the young (TEDDY)
- Service for the support of the European Orphan medicine market (EuOrphan)

Examples of other EC funded networks

DG Information Society and Media has also funded a number of initiatives under the theme of e-health and telemedicine

ANNEX 10

European countries with centres of reference for rare diseases
Characteristics of their programme

Country	Definition Rare Disease	Reference centres since	Number of Centres
England	1 / 50,000	1990	30
Sweden	1 / 10,000	1990	75
Denmark	1 / 10,000	2001	108
Italy	1 / 2,000	2001	228
France	1 / 2,000	2004	34 to date 100 planned

ANNEX 11

European Population at 1 January 2003 n millions of inhabitants

COUNTRY	POPULATION
Germany (D)	82,5
Turkey (TR)	70,2
France	59,6
United-Kingdom (UK)	59,3
Italy (I)	57,3
Spain (E)	40,7
Poland (PL)	38,2
Romania (RO)	21,8
Netherlands (NL)	16,2
Greece (EL)	11
Belgium (B)	10,4
Portugal (P)	10,4
Czech Republic (CZ)	10,2
Hungary (HU)	10,1
Sweden (S)	8,9
Austria (A)	8,1
Bulgaria (BG)	7,8
Denmark (DK)	5,4
Slovakia (SK)	5,4
Finland (FIN)	5,2
Ireland (IRL)	4
Lithuania (LT)	3,5
Latvia (LV)	2,3
Slovenia (SI)	2
Estonia (EE)	1,4
Cyprus (CY)	0,7
Luxembourg (L)	0,4
Malta (MT)	0,4

Population in 2005 453.6 without candidate countries
553.4 with candidate countries

Source: Eurostat

ANNEX 12

LIST OF CENTRES OF REFERENCE IN UK BY TYPE OF CENTRE

DISEASE ORIENTED CENTRES

- Amyloidosis
- Bladder exstrophy
- Choriocarcinoma
- Epidermolysis bullosa service for adults
- Epidermolysis bullosa service for children
- Liver disease service for children
- Lysosomal storage disorders*
- Ocular oncology
- Ophthalmic pathology
- Personality disorder
- Primary malignant bone tumours*
- Pulmonary hypertension
- Rare neuromuscular diseases
- Retinoblastoma
- Severe combined immunodeficiency and related disorders (SCIDS)
- Severe intestinal failure

TECHNOLOGY ORIENTED CENTRES

- Craniofacial surgery
- Deep brain stimulation for severe Parkinson's Disease
- Extra-corporeal membrane oxygenation (ECMO) for adults
- Extra-corporeal membrane oxygenation (ECMO) for children*
- Extra-corporeal membrane oxygenation (ECMO) for neonates and infants
- Heart and lung transplantation service for adults and children
- Liver transplantation service for adults and children
- Mental health services for deaf children & adolescents (inpatient)
- Pancreas transplantation
- Pseudomyxoma peritonei surgery
- Pulmonary thromboendarterectomy
- Reconstructive surgery in adolescents for congenital malformation of the female genital tract
- Secure forensic mental health service for young people
- Small bowel transplantation for adults and children
- Stem cell transplantation for juvenile idiopathic arthritis and related connective tissue disorders
- Ventricular assist devices for adults
- Ventricular assist devices / extra-corporeal membrane oxygenation (bridge to transplant) for children

ANNEX 13
Distribution of specialised clinics in 13 European countries as collected by Orphanet (August 05)

Type of clinics	Germany	Austria	Belgium	Cyprus	Spain	Finland	France	Greece	Italy	Portugal	Romania	Switzer.	UK
Amyloidosis clinic							4		2	1			1
Angiomas and vascular malformations clinic	1						16		1				
Congenital anomalies of limbs clinic			1				2		3				
Facial anomalies clinic							11			1		1	
Antirabies clinic							1		1				
Constitutional aplasias clinic							1		1				
Behcet disease clinic	2								2	1			
CADASIL clinic							1		2				
Cardiogenetics clinic		1	2				4		3	1		2	
Genetic counselling clinic	38		6	1	22	7	115	2	65	8	1	1	27
Creutzfeldt-Jakob clinic			1				1						
Cutis laxa clinic			1				3		1				
Cytogenetics clinic					13		57		33	1		1	
Dysmorphology clinic					10		61		25	5		2	27
Ehlers-Danlos clinic			1				6		3				
Epidermolysis bullosa clinic	1	1	1				4		2				4
Fabry disease clinic		1					3		1	1		1	
Fibromuscular dysplasia clinic							1						
Gaucher disease clinic							3		1				
Genodermatosis clinic	1				1		11		2				
Sjögren syndrome clinic	3						3		1				
Pediatric haematology clinic							6		2				
Hemoglobinopathies clinic		1			1		9		3	1			
Hemophilia and other coagulation defects clinic	1				1		50		2	2			
Pediatric hepatology clinic							7						3

Type of clinics	Germany	Austria	Belgium	Cyprus	Spain	Finland	France	Greece	Italy	Portugal	Romania	Switzer.	UK
Langerhans cell histiocytosis clinic							6						
Huntington disease clinic		1			1		22		4	1			
Malignant hyperthermia clinic							3		1			1	
Leukodystrophies clinic							3		4				
Lymphedema clinic							2						
Cutaneous lymphoma clinic							7		1	1			
Scalp diseases clinic							1						
Phosphocalcic metabolism diseases clinic	1						2		2			1	
Lysosomal diseases clinic							6		2				6
Metabolic diseases clinic					3		15		8	3		3	
Mitochondrial diseases clinic									3				
Neurodegenerative diseases clinic							6		6	1			
Neurometabolic diseases clinic	1						6						
Neuromuscular diseases clinic	1						74		3	3		1	7
Ophthalmology clinic			2				6		3				4
Bone diseases clinic							11		3				
Peroxysomal diseases clinic							1						
Memory disorders clinic							10						
Sleep disorders clinic		1	1				22						
Vascular liver diseases clinic					1		1						
Marfan syndrome clinic	1						8		2	1			
Vascular medicine clinic							2		1				
Melanoma clinic		1					7						
Premature menopause clinic							5						
Moebius syndrome clinic					1		1						
Movement disorders clinic	1		1				9		1				
Cystic fibrosis clinic	5		1		2		50	1	3				
Pediatric Nephrology clinic			1		2		26		1	1		2	
Neurofibromatosis type 1 clinic					1		29		4				
Neurofibromatosis type 2 clinic			1		1		2		1				

Type of clinics	Germany	Austria	Belgium	Cyprus	Spain	Finland	France	Greece	Italy	Portugal	Romania	Switzer.	UK
Neuroimmunology clinic							2						
Neuropaediatrics and infantile epilepsy clinic	3				1		24		4	3		2	
Congenital neutropenias clinic	1	1	2		2		2	2	2	1			2
Oncogenetics clinic	1	1			2		72		11				
Pediatric oncohaematology clinic	1	2					22		2				
Pediatric Oncology clinic	1						6		1				
Osteogenesis imperfecta clinic			1				4		1				
Osteopetrosis clinic							1		1	1			
Phenylketonuria clinic							11						
Porphyria clinic							1		2				
Prader-Willi clinic		1	1				3		1				
Pseudoxanthoma elasticum clinic			1				3		1				
Rendu-Osler syndrome clinic							11						
Retinitis pigmentosa clinic			1				4		2				
Scleroderma clinic							1		1	1			
Amyotrophic lateral sclerosis clinic							17		1	1			
Tuberous sclerosis clinic	6		1				2						
Spina Bifida clinic	1						15		2	1			
Deafness clinic							8		1	1		1	
Paraneoplastic neurologic syndromes clinic		1					3						
Rare Parkinsonian syndromes clinic							6		2				
Teratology clinic							3		2				
Hereditary cardiac rythm defects clinic			2				5		2				
Child and adolescent bone tumor clinic		1					1		1				
Turner syndrome clinic			1				3		1				
Vasculitis clinic	3						1		3				
Von Hippel-Lindau disease clinic			1				11		1				
Primary immunodeficiencies	3	1			2		1	1	3			1	3
Congenital anaemias	8		4		10		3		11	3			18

ANNEX 14

List of types of specialised clinics for rare diseases as identified by Orphanet in Europe

Area	Format of clinics	N° diseases	Target
bone diseases	Bone diseases clinic	group	
	Osteogenesis imperfecta clinic	single	
	Osteopetrosis clinic	single	
cardiology	Hereditary cardiac rythm defects clinic	group	
	Vascular medicine clinic	group	
	Vasculitis	group	
	Fibromuscular dysplasia clinic	Single	
dermatology	Angiomas and vascular malformations clinic	group	
	Genodermatosis clinic	group	
	Scalp diseases clinic	group	
	Cutis laxa clinic	single	
	Epidermolysis bullosa clinic	Single	
	Melanoma clinic	single	
	Neurofibromatosis type 1 clinic	single	
dysmorphology	Congenital anomalies of limbs	group	
	Facial anomalies clinic	group	
endocrinology	Prader-Willi clinic	Single	
	Turner syndrome clinic	Single	
general	Teratology	group	
genetics	Cytogenetics clinic	General	
	Dysmorphology clinic	General	
	Genetic counselling clinic	general	
	Cardiogenetics clinic	group	
gynecology	Premature menopause clinic	group	
haematology	Pediatric haematology clinic	group	paediatrics
	Constitutional aplasias clinic	single	
	Hemoglobinopathies clinic	single	
	Hemophilia and other coagulation defects clinic	single	
	Congenital anaemais	group	
	Porphyria clinic	Single	
hepatology	Biliary tract inflamatory diseases	group	
	Pediatric hepatology clinic	group	paediatrics
	Vascular liver diseases clinic	group	
immunology	Congenital neutropenias	group	paediatrics
infectious	Antirabies clinic	single	
Internal medicine	Systemic diseases clinic	group	
	Amyloidosis	Single	
	Behcet	single	
	CADASIL clinic	single	
	Cystic fibrosis clinic	single	
	Ehlers-Danlos clinic	Single	
	Malignant hyperthermia clinic	single	
	Marfan syndrome clinic	single	
	Pseudoxanthoma elasticum clinic	Single	
	Rendu-Osler syndrome clinic	Single	
	Scleroderma clinic	Single	
	Sjögren syndrome clinic	single	
	Tuberous sclerosis clinic	Single	

Area	Format of clinics	N° diseases	Target
	Von Hippel-Lindau disease clinic	Single	
metabolic	Lysosomal diseases clinic	group	
	Metabolic diseases clinic	group	paediatrics
	Phosphocalcic metabolism diseases clinic	group	
	Fabry disease clinic	Single	
	Gaucher disease clinic	Single	
	Phenylketonuria clinic	Single	
	Peroxysomal diseases clinic	group	
nephrology	Pediatric Nephrology clinic	group	paediatrics
neurology	Leukodystrophies clinic	group	
	Memory disorders clinic	group	
	Movement disorders clinic	group	
	Neurodegenerative diseases clinic	group	
	Neuroimmunology clinic	group	
	Neurometabolic diseases clinic	group	
	Neuromuscular diseases clinic	group	
	Neuropaediatrics and infantile epilepsy clinic	group	paediatrics
	Paraneoplastic neurologic syndromes clinic	group	
	Sleep disorders clinic	group	
	Amyotrophic lateral sclerosis clinic	single	
	Creutzfeldt-Jakob clinic	single	
	Friedreich ataxia clinic	single	
	Huntington disease clinic	single	
	Moebius syndrome clinic	single	
	Rare Parkinsonian syndromes clinic	single	
neurosurgery	Spina Bifida clinic	Single	
oncology	Child and adolescent bone tumor clinic	group	paediatrics
	Oncogenetics clinic	group	
	Pediatric oncohaematology clinic	group	paediatrics
	Pediatric Oncology clinic	group	paediatrics
	Cutaneous lymphoma clinic	single	
	Langerhans cell histiocytosis clinic	single	paediatrics
ophthalmology	Ophthalmology clinic	group	
	Retinitis pigmentosa clinic	Single	
otorhinolaryngol	Deafness clinic	group	
	Neurofibromatosis type 2 clinic	single	
pneumology	Pulmonary rare diseases clinic	group	
	Ondine syndrome clinic	single	paediatrics
	Severe pulmonary hypertension clinic	single	
surgery	Sexual ambiguity clinic	single	paediatrics
other	Lymphedema clinic	group	

ANNEX 15

Specialised clinics in Orphanet ordered by number of rare diseases covered

Type of clinics	N° diseases
Genetic counselling clinic	2352
Dysmorphology clinic	2304
Metabolic diseases clinic	250
Neuropaediatrics and infantile epilepsy clinic	190
Cytogenetics clinic	168
Genodermatosis clinic	148
Bone diseases, congenital/hereditary clinic	101
Neuromuscular diseases clinic	94
Rare and congenital anaemias	93
Ophthalmology clinic	57
Congenital anomalies of limbs	43
Paediatric Nephrology clinic	43
Paediatric oncohaematology clinic	38
Paediatric Oncology clinic	37
Paediatric hepatology clinic	32
Scalp diseases clinic	32
Lysosomal diseases clinic	31
Neurometabolic diseases clinic	31
Deafness clinic	26
Facial anomalies clinic	25
Teratology clinic	23
Haemophilia and other coagulation defects clinic	22
Oncogenetics clinic	18
Peroxysomal diseases clinic	16
Angiomas and vascular malformations clinic	15
Neurodegenerative diseases clinic	15
Paediatric haematology clinic	14
Vasculitis clinic	14
Vascular medicine clinic	13
Mitochondrial diseases clinic	12
Leukodystrophies clinic	10
Cardiogenetics clinic	9
Congenital neutropenias	9
Rare Parkinsonian syndromes clinic	8
Hereditary cardiac rythm defects clinic	8
Child and adolescent bone tumour clinic	8
Lymphedema clinic	7
Phosphocalcic metabolism diseases clinic	7
Movement disorders clinic	7
Neuroimmunology clinic	7
Ehlers-Danlos clinic	6
Osteopetrosis clinic	5
Amyotrophic lateral sclerosis clinic	5
Epidermolysis bullosa clinic	4
Memory disorders clinic	4
Spina Bifida clinic	4
Creutzfeldt-Jakob clinic	3
Cutaneous lymphoma clinic	3
Vascular liver diseases clinic	3
Marfan syndrome clinic	3
Retinitis pigmentosa clinic	3
Paraneoplastic neurologic syndromes clinic	3
Constitutional aplasias clinic	2
Sleep disorders clinic	2
Phenylketonuria clinic	2
Scleroderma clinic	2
Amyloidosis clinic	1
Antirabies clinic	1
Behcet clinic	1
CADASIL clinic	1
Cutis laxa clinic	1
Fabry disease clinic	1
Fibromuscular dysplasia clinic	1
Gaucher disease clinic	1
Sjögren syndrome clinic	1
Langerhans cell histiocytosis clinic	1
Huntington disease clinic	1
Malignant hyperthermia clinic	1
Melanoma clinic	1
Premature menopause clinic	1
Moebius syndrome clinic	1
Cystic fibrosis clinic	1
Neurofibromatosis type 1 clinic	1
Neurofibromatosis type 2 clinic	1
Osteogenesis imperfecta clinic	1
Porphyria clinic	1
Prader-Willi clinic	1
Pseudoxanthoma elasticum clinic	1
Rendu-Osler syndrome clinic	1
Tuberous sclerosis clinic	1
Turner syndrome clinic	1
Von Hippel-Lindau disease clinic	1

ANNEX 16

Examples of priority groups of rare diseases for ECR

Main issue

TYPES OF RARE DISEASES	SPECIALTY	DIAGNOSIS	TREATMENT
Amyloidosis	internal medicine		yes
Amyotrophic lateral sclerosis	neurology		yes
Angiomas and vascular malformations	dermatology		yes
Behcet syndrome	internal medicine		yes
CADASIL syndrome	internal medicine	yes	yes
Child and adolescent bone tumor	oncology		yes
Congenital anaemias	haematology	yes	
Congenital anomalies of limbs	dysmorphology	yes	yes
Congenital neutropenias	immunology		yes
Congenital/genetic deafness	otorhinolaryngology	yes	
Constitutional aplasias	haematology	yes	yes
Cutaneous lymphoma	oncology		yes
Cystic fibrosis	internal medicine	yes	yes
Dysmorphology/mental disability	genetics	yes	
Ehlers-Danlos	internal medicine	yes	
Facial anomalies / cleft palate/lip	dysmorphology	yes	
Genetic dermatosis	dermatology	yes	
Hemoglobinopathies	haematology	yes	yes
Hemophilia, coagulation defects clinic	haematology	yes	yes
Hereditary/congenital bone diseases	bone diseases	yes	yes
Huntington disease	neurology	yes	yes
Langerhans cell histiocytosis	oncology		
Leukodystrophies	neurology	yes	
Lymphedema clinic	other		yes
Lysosomal diseases	metabolism	yes	yes
Malignant hyperthermia	internal medicine	yes	
Marfan syndrome clinic	internal medicine	yes	yes
Metabolic diseases	metabolism	yes	yes
Moebius syndrome	neurology		
Neurofibromatosis	dermatology	yes	
Neuroimmunologic diseases	neurology	yes	
Neurometabolic diseases	neurology	yes	
Neuromuscular diseases	neurology	yes	yes
Neuropaediatrics and infantile epilepsy	neurology	yes	yes
Paraneoplastic neurologic syndromes	neurology		
Paediatric hepatology	hepatology		yes
Paediatric Nephrology	nephrology	yes	yes
Paediatric oncohaematology	oncology		yes
Paediatric Oncology	oncology		yes
Peroxysomal diseases	metabolism	yes	yes
Porphyria	haematology	yes	
Prader-Willi	endocrinology	yes	yes
Premature menopause	gynaecology	yes	
Primary immuno deficiencies	immunology	yes	yes
Prion diseases	neurology		
Pseudoxanthoma elasticum	internal medicine	yes	
Rare genetic cardiac diseases	cardiology	yes	yes
Rare memory disorders	neurology		
Rare movement disorders	neurology	yes	yes
Rare neurodegenerative diseases	neurology	yes	yes
Rare ophthalmologic disorders	ophthalmology	yes	yes
Rare Parkinsonian syndromes	neurology		yes
Rare pulmonary diseases	pneumology		yes
Rare vascular disorders	cardiology	yes	

TYPES OF RARE DISEASES	SPECIALTY	DIAGNOSIS	TREATMENT
Rendu-Osler syndrome	internal medicine	yes	yes
Scleroderma	internal medicine		
Sexual ambiguity	paediatric surgery	yes	yes
Sjögren syndrome	internal medicine		yes
Sleep disorders	neurology		
Spina Bifida	neurosurgery		yes
Systemic diseases	internal medicine	yes	yes
Teratology	general	yes	
Tuberous sclerosis	internal medicine	yes	
Turner syndrome	endocrinology		
Vascular liver diseases	hepatology		
Von Hippel-Lindau disease	internal medicine	yes	yes

ANNEX 17
Examples of rare diseases that may be treated with a costly drug

Achondroplasia
Acquired autoimmune haemolytic anemia
Acromegaly
Acute erythroblastic leukemia
Acute lymphoblastic leukemia
Acute megacaryoblastic leukemia
Acute monoblastic leukemia
Acute myeloblastic leukemia with maturation
Acute myeloblastic leukemia without maturation
Acute myelomonocytic leukemia
Acute promyelocytic leukemia
Adrenocortical carcinoma
Alpha-1 antitrypsin deficiency
Alveolar echinococcosis
Amyloidosis
Amyotrophic lateral sclerosis
Anaplastic large cell lymphoma
Anterior pituitary insufficiency, familial
Antithrombin deficiency, congenital
Aspergillosis
Barrett esophagus
Beta-thalassemia
Burkitt lymphoma
Carbamoylphosphate synthetase deficiency
Carnitine uptake deficiency
Castleman disease
Charcot-Marie-Tooth disease, type 1
Citrullinemia
Congenital neutropenia
Congenital short bowel
Cryoglobulinemia
Cutaneous lymphoma
Cystic fibrosis
Cysticercosis
Cystinosis
Cystinuria
Dermatitis herpetiformis
Digitalic compounds poisoning
Ethylene glycol poisoning
Fabry disease
Familial adenomatous polyposis, autosomal dominant
Fibrosarcoma
Focal dystonia
Follicular lymphoma
Gastrointestinal stromal tumor
Gaucher disease
Gelineau disease
Glioblastoma
Glycogen storage disease type 2
Graft versus host disease
Granulomatous disease, chronic
Hairy cell leukemia (HCL)
Hemophilia
Heparin-induced thrombocytopenia
Hepatitis, chronic autoimmune
Hodgkin disease
Homocystinuria due to cystathionine beta-synthase deficiency
Hypogonadotropic hypogonadism without anosmia, X linked
Immunodeficiency, common variable
Jessner's benign lymphocytic infiltration of the skin
Juvenile arthritis, idiopathic
Kaposi's sarcoma
Kawasaki disease
Langerhans cell histiocytosis
Large B cell diffuse lymphoma
Lennox-Gastaut syndrome
Leprechaunism
Leukemia, chronic myeloid
Lipodystrophy, Berardinelli type
Lymphangioma
Lymphoblastic lymphoma
Malignant hyperthermia susceptibility
Mesothelioma
Methylcobalamin deficiency type cbl E
Methylcobalamin deficiency type cbl G
Methylenetetrahydrofolate reductase deficiency
Methylmalonicaciduria - homocystinuria
Microscopic polyangiitis
Mucopolysaccharidosis type 1
Myasthenia gravis
Myelodysplastic syndromes
Myeloma, multiple
N-acetylglutamate synthetase deficiency
Nanism due to growth hormone isolated deficiency
Nanism due to growth hormone isolated deficiency with x linked hypogammaglobulinemia
Nanism due to growth hormone isolated deficiency with x linked hypogammaglobulinemia
Nanism due to growth hormone qualitative anomaly
Nanism due to growth hormone resistance
Oligodendroglioma
Ornithine carbamoyltransferase deficiency
Osteosarcoma
Parathyroid carcinoma
Patent ductus arteriosus
Pheochromocytoma
Polyarteritis nodosa
Polycythemia vera
Polymyositis
Porphyria
Prader-Willi syndrome
Precocious puberty, gonadotropin-dependant
Primary biliary cirrhosis
Pulmonary arterial hypertension
Pure autonomic failure
Rabson-Mendenhall syndrome
Renal carcinoma, familial
Severe myoclonic epilepsy of infancy (SMEI)
Sexual precocity, familial, gonadotropin-independent, male-limited
Sickle cell anemia
Sjögren syndrome
Thrombocythemia, essential
Thrombocytopenic purpura, autoimmune
Turner syndrome
Tyrosinemia type 1
Ulcerative colitis
Waldenström macroglobulinemia
Willebrand disease
Wilson disease