



# European Union action in the field of Rare Diseases

**Krakow, 14-15th May 2010**

**Antoni Montserrat Moliner  
Policy Officer for Rare Diseases  
European Commission  
DG SANCO Health Information Unit**

## ***Legal basis for the developments of the EU Public Health Policy***

### **■ Based on new Article 168 (former 152) of the EU Treaty**

**■ A Community action programme on Rare Diseases, including genetic diseases, was adopted for the period of 1 January 1999 to 31 December 2003 with the aim of ensuring a high level of health protection in relation to RD. As the first EU effort in this area, specific attention was given to improving knowledge and facilitating access to information about these diseases.**

**■ Orphan Medicinal Product Regulation (Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products, was proposed to set up the criteria for orphan designation in the EU and describes the incentives (e.g. 10-year market exclusivity, protocol assistance, access to the Centralised Procedure for Marketing Authorisation) to encourage the research, development and marketing of medicines to treat, prevent or diagnose rare diseases.**

**■ For the period 2008-2013 the Commission has adopted the [White Paper COM\(2007\) 630 final "Together for Health: A Strategic Approach for the EU 2008-2013"](#) of 23 October 2007 developing the EU Health Strategy. Actions under Objective 1 of this EU Strategy cover a Communication on European Action in the Field of Rare Diseases and in point 4.1 of this EU Strategy it is suggested to put forward EC-level structured cooperation mechanisms to advise the Commission and to promote cooperation between the Member States.**

## ***Legal basis for the developments of the EU Public Health Policy***

- **As a consequence Rare diseases are now one of the priorities in the [Second EU Health Programme 2008-2013](#). According to the DG SANCO Work Plans for the implementation of the Public Health Programme, main lines of action and priorities are choosed very year. **Work Plan for 2011 adopted 16th December 2009.****
- **In the current Framework Programme, the FP7, the Health Theme of the "Cooperation" Specific Programme, is designed to support multinational collaborative research in different forms. The main focus of the Health theme in the rare diseases area are Europe-wide studies of natural history, pathophysiology, and the development of preventive, diagnostic and therapeutic interventions.**
- **Commission Communication COM (2008) 679/2 to the European Parliament, the Council, the Economic and Social Committee and the Committee of the Regions on Rare diseases: Europe's challenges creating an integrated approach for the EU action in the field of rare diseases. **Adopted 11th November 2008.****
- **Council Recommendation on a European action in the field of rare diseases recommending actions at national level to implement the EU action (e.g. National Plans for Rare Diseases). **Adopted 8th June 2009.****

## ***Future legal basis for the developments of the EU Public Health Policy***

■ **Decision of the Commission creating a European Union Committee of Experts on Rare Diseases during 2009. To be composed by 51 members representing Member States, patient's organisations, industry, FP Projects, Health Programme projects, etc.**

**Adopted 30th November 2009.**

■ **Directive of the European Parliament and of the Council on the application of patients' rights in cross-border healthcare (COM(2008)414) provides for the development of European reference networks (ERNs) to be facilitated by the Member States. The ERN for Rare Diseases will have a strategic role in the improvement of quality treatment for all patients throughout the European Union as called by the patients' organisations. To be adopted End 2010 (tbc).**

## ***The Commission Communication and the Council Recommendation on rare diseases***

- **There is probably no other area in public health in which 27 national approaches could be considered to be so inefficient and ineffective as with rare diseases. The reduced number of patients for these diseases and the need to mobilise resources could be only efficient if done in a coordinated European way.**
- **Article 168.6 provides for the adoption by qualified majority by the Council of Recommendations, on the basis of Commission proposals, for the purposes set out in that article.**
- **These Recommendations are the only legislative tool provided for in Article 168 on public health except for the few areas where measures or incentive measures may be adopted (see Article 168.4).**

## ***The Commission Communication and the Council Recommendation on rare diseases***

### ***I. Plans and strategies in the field of rare diseases***

**Calls on the MS to elaborate and adopt a plan or strategy by the end of 2013.**

### ***II. Adequate definition, codification and inventorying of rare diseases***

**Evokes the common definition of a rare disease as a condition affecting no more than 5 per 10 000 persons; aims to ensure that rare diseases are adequately coded and traceable in all health information systems based on the ICD and in respect of national procedures; and encourages MS to contribute actively to the inventory of rare diseases based on the Orphanet network.**

### ***III. Research on rare diseases***

**Calls for the identification and fostering of rare disease research at all levels.**

### ***IV. Centres of expertise and European reference networks for rare diseases***

**Asks the MS to identify and facilitate networks of expertise based on a multidisciplinary approach to care, and foster the diffusion and mobility of expertise and knowledge.**

## ***The Commission Communication and the Council Recommendation on rare diseases***

### ***V. Gathering the expertise on rare diseases at European level***

**MS should share best practices, develop medical training relevant to the diagnosis and management of rare diseases, coordinate European guidelines, and, to minimise the delay in access to orphan drugs, MS should share clinical/therapeutic added-value assessment reports at the Community level.**

### ***VI. Empowerment of patient organisations***

**MS should consult patient representatives on policy development; facilitate patient access to updated information on rare diseases; promote patient organisation activities.**

### ***VII. Sustainability***

**Long-term sustainability in the field of information, research and healthcare of infrastructures must be ensured.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***I. Plans and strategies in the field of rare diseases***

- **The Member States are invited to establish national or regional action plans for RD before 2013 in order to implement the actions suggested in the Commission Communication and the Council Recommendation and to provide an annual report on the progress made toward this objective**
- **The Commission will provide European guidelines for the elaboration of these action plans for RD (EUROPLAN Project selected for funding for the period 2008-2011). Appropriate conferences will be organised (twelve national conferences scheduled during 2010). Continuity of EUROPLAN (under the form of a Joint Action scheduled)**
- **A national plan/strategy (NP/NS) can be defined as the sum of integrated and comprehensive health policy actions for RD, to be developed and implemented at national level. A plan has: a) well specified objectives and b) actions that have to be supported by a budget, implemented within a time frame, evaluated with specific indicators (EUROPLAN Definition).**

## ***New priorities after Commission Communication and Council Recommendation***

### ***I. Plans and strategies in the field of rare diseases***

- **This definition includes two central concepts of the Council Recommendation on RD. ‘*Integrated*’ refers to the fact that strategies should be developed in a way to identify complementarities, maximize synergies and avoid duplications. ‘*Comprehensive*’ refers to the fact that the actions foreseen in the plan should fulfill all main patients’ needs (e.g. quality of care but also social services and centres of expertise).**
- **Definition of common and harmonised indicators to appraise and evaluate the national plan/strategy (NP/NS) of rare diseases.**
- **The EUROPLAN Recommendations also includes the international dimension not possible to fill at national level (e.g. classification and codification, reference networks, orphan drugs, research, etc.). To be analysed by health authorities (Krakow, 13th May 2010) and national conferences (2010). EUROPLAN final conference (Rome, March 2011).**
- **Continuity of EUROPLAN after 2011 (under the form of a Joint Action) scheduled.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***I. Plans and strategies in the field of rare diseases***

■ **Five Member States have adopted National Plan/Strategy for Rare Diseases:**

**[French National Plan on Rare Diseases \(2005-2008\)](#)**

The Plan 2005-2008 is **[under evaluation](#)** (concluded) before the adoption of a new Plan 2010-2014.

**[Bulgarian National Plan on Rare Diseases 2009-2013](#)**

**[Greek National Plan on Rare Diseases 2008-2012](#)**

**[Portuguese National Programme for Rare Diseases 2008-2015](#)**

**[Rare Diseases Strategy of the Spanish National Health System](#)**

■ **Four are in an advanced stage for adoption: Belgium (in 2010), Italy (framework law and national plan), Romania, Czech Republic**

■ **Eighth have (+ or -) structures in place for the definition of a plan: United Kingdom, Germany, The Netherlands, Cyprus, Luxembourg, Hungary, Austria, Poland.**

## ***I. Plans and strategies in the field of rare diseases***

**From the side of the economic costs of rare diseases the Commission selected in 2009 the project**

### **SOCIAL ECONOMIC BURDEN AND HEALTH-RELATED QUALITY OF LIFE IN PATIENTS WITH RARE DISEASES IN EUROPE (BURQOL-RD)**

- **The general objective of this project is to develop a disease based model capable of quantifying the socioeconomic burden and Health-Related Quality of Life (HRQOL) for patients in Europe with rare diseases (RD) and their caregivers.**
- **The model will incorporate information on medical resources related to RD (e.g. hospitalisation, consultations, drugs), on non-medical resources (e.g. walking sticks, wheelchairs, modifications to house and car), services (e. g. home care, transportation), informal care by relatives, productivity losses, intangible costs as well as the overall HRQOL (utilities). HRQL will be measured both in RD patients and their caregivers.**
- **This information will be collected with a standardized pre-tested questionnaire from a sample of patients registered in RD associations. The disease information that will be collected will include disease duration and self-assessed disease severity.**
- **Accordingly, the mean annual costs will be estimated from a society perspective.**

## ***I. Plans and strategies in the field of rare diseases***

**From the side of the socioeconomic needs the Commission has selected and supported during last years three extensive surveys performed by EURORDIS**

- ***The Voice of 12,000 Patients***”, was officially launched on March 3 2009. The book, published by **EURORDIS**, presents the conclusions of the **EurordisCare 2** and **EurordisCare 3** surveys on the experience and expectations on diagnoses and access to care of 12 000 patients representing 18 rare diseases and 24 European countries.
- It details the methodology, the overall results, results by country and by disease, and how the surveys have contributed to the development of policies and actions in favour of better diagnosis and care for rare disease patients.

## ***I. Plans and strategies in the field of rare diseases***

**From the side of the socioeconomic needs the Commission has selected and supported during last years three extensive surveys performed by EURORDIS**

- **25% of patients reported waiting between 5 and 30 years from the time of first symptoms to a confirmatory diagnosis of their disease.**
- **40% of patients were initially misdiagnosed leading to severe consequences such as inappropriate medical interventions, including surgery and psychological treatment.**
- **In 33% of cases, the diagnosis was announced in unsatisfactory terms or conditions.**
- **In 12.5% of cases, it was announced in unacceptable ones.**
- **The genetic nature of the disease was not communicated to the patient or family in 25% of cases. This is paradoxical, given the genetic origin of most rare diseases.**
- **Genetic counselling was only provided in 50% of cases.**
- **25% of patients had to travel to a different region to obtain a diagnosis and 2% had to travel to a different country.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***II. Adequate definition, codification and inventorying of rare diseases***

- **The EU definition of rare disease based on a prevalence of less than 5 per 10,000 is maintained**
- **An EU or an international project exploring an incidence based definition of rare diseases will be launched in 2010.**
- **The EU will contribute to the ongoing process of revision of the ICD (International Classification of Diseases) in order to ensure appropriate codification and classification of rare diseases in the future ICD-11. A working group will be supported for all the period of this revision**
- **The EU will establish a dynamic Inventory of Rare Diseases to be periodically updated**
- **The database Orphanet will be supported using appropriate financial instruments**
- **The support to the disease information networks through the Health Programme and the FP7 should be pursued**

## New priorities after Commission Communication and Council Recommendation

### II. Adequate definition, codification and inventorying of rare diseases

The database Orphanet will be supported using appropriate financial instruments



The screenshot shows the Orphanet website interface. At the top, there are language options: Français, English, Español, Deutsch, Italiano. The main header includes the Orphanet logo, the tagline 'The portal for rare diseases and orphan drugs', and navigation links for Homepage, Site map (XML), Help, and Contact us. Below the header is a navigation menu with categories: Rare diseases, Orphan drugs, Clinics, Diagnostic tests, Research and trials, Patient organisations, Directory of resources, and Education and media.

The main content area features a search section with a 'SIMPLE SEARCH' box containing the text 'Search a disease' and an 'OK' button. To the right, there are 'OTHER SEARCH OPTION(S)' including links for Orphan drugs, Patient organisations, Research and trials, Clinics, Diagnostic tests, and Directory of resources.

On the right side, there is a table titled 'ORPHANET DATA' with the following statistics:

ORPHANET DATA	
Diseases	7242
Clinics	3017
Laboratories	2838
Professionals	10136
Daily visitors	22558

Below the search section, there are several content blocks:

- RARE DISEASES**: Information about a disease, Alphabetical list, Search by clinical sign, Emergency guidelines, Patient encyclopaedia, Professional encyclopaedia, About Rare Diseases.
- RESOURCES DIRECTORY**: Clinics, Reference centres, Clinical laboratories, Research projects, Registries / databases, Professionals, Patient organisations, EuroCentest, Register your activity.
- ORPHAN DRUGS**: (Section header only).
- Services for professionals**: Professional encyclopaedia, Search by clinical sign, Emergency guidelines, Orphanet Journal Of Rare Diseases, Newsletters, Education tools, Register your activity, OrphanXchange.
- Services for patients**: Patient encyclopaedia, Information about a disease, Patient organisations, Clinics, Participate in clinical research, Contact other patients / families, Training sessions, Newsletters.
- NEWS**: Orphanet's 10th birthday, Conference presentation highlights, EC Rare Disease webpages, French National Plan for Rare Diseases.

In the center-right, there is a large image of a person's hands holding a medical tube. Text overlaid on the image reads: 'Improve the quality of medical care for Rare Diseases. Provide adapted services for the rare diseases community. About Orphanet | Quality charter Register your activity'.

## II. Adequate definition, codification and inventorying of rare diseases

### ■ Comprehensive list of rare diseases: 5,838

- Identity card + genes
- Unique Orpha number
  - Stable what ever is the evolution of knowledge
  - Linked to parent and child disease in every classification
- Files available on request
- Suitable to code clinical activity / lab activity in information systems

### ■ Classifications of rare diseases

- List of all published classifications
- Visualisation of each classification
- Possibility to click at any level to have detailed information

### ■ Methodology:

- Collection of published classifications of specific groups of rare diseases
  - By clinical presentation
  - By mechanism
  - By aetiology
- Building up of a comprehensive classification of Orphanet entries to serve the needs of Health Care information systems: [the Orphanet classification of rare diseases](#)
  - Based on
    - Litterature (Text books, articles)
    - Expert opinion of working groups
  - Peer-reviewed by the International editorial board of Orphanet
  - Regularly updated

# Orpha nomenclature of diseases

## :: Acute lymphoblastic leukaemia

### Identity card

<b>Orpha number:</b>	ORPHA513	<b>Synonym(s):</b>	-
<b>Prevalence of rare diseases:</b>	1-9 / 100 000		
<b>Inheritance:</b>	-		
<b>Age of onset:</b>	Childhood		
<b>ICD 10 code:</b>	C91.0		
<b>MIM number:</b>	<a href="#">151390</a> [ <a href="#">↗</a> ] <a href="#">151442</a> [ <a href="#">↗</a> ] <a href="#">186770</a> [ <a href="#">↗</a> ] <a href="#">186855</a> [ <a href="#">↗</a> ] <a href="#">186921</a> [ <a href="#">↗</a> ] <a href="#">186990</a> [ <a href="#">↗</a> ] <a href="#">187040</a> [ <a href="#">↗</a> ] <a href="#">247640</a> [ <a href="#">↗</a> ] <a href="#">600185</a> [ <a href="#">↗</a> ]		

**Unique Orpha number**

- Stable what ever is the evolution of knowledge
- Linked to parent and child diseases in every classification

**Indexation with ICD10 codes: 2,143 diseases**  
**Indexation with MIM codes: 3,087 diseases**

### SUMMARY

Acute lymphoblastic leukaemia (ALL) is a malignant proliferation of lymphoid cells blocked at an early stage of differentiation and accounts for 75% of all cases of childhood leukaemia. About 2,000 children in the United States and 1,500 children in Europe are diagnosed with ALL each year. The disease is most common between 2 and 5 years of age, but can occur at any age. It is a distressing illness for the child and the family. Many organ systems are affected. Signs and symptoms include enlarged lymph nodes and bone pain. Most cases show chromosomal and genetic abnormalities, occurring spontaneously in genes playing important regulatory roles in controlling the lymphoid cell population. The most common ALL translocation, the t(12;21), appears to have good prognostic implications. ALL is biologically heterogeneous and morphologic, immunologic, cytogenetic, biochemical, and molecular genetic characterisation of leukaemia lymphoblasts is needed to establish the diagnosis or to exclude other possible causes of bone marrow failure and, finally, to classify ALL subtypes. Biological findings include hyperleukocytosis due to circulating lymphoblasts, anaemia and thrombocytopenia. Diagnosis is established by bone marrow biopsy revealing leukaemic cell infiltration. The chemotherapy protocols adopted by international cooperative groups have four main objectives: induction with the aim of complete remission, preventative therapy to avoid central nervous system involvement, consolidation/relapse induction, and maintenance

**Epidemiology**

- Indexed with class of **prevalence: 2,959 diseases**
- Indexed with mode of **inheritance: 2,846 diseases**
- Indexed with age of **onset: 2,760 diseases**

<b>Additional information</b>
<b>Further information on this disease</b>
<ul style="list-style-type: none"> <li>&gt; Gene(s)</li> <li>&gt; Publications in PubMed [<a href="#">↗</a>]</li> <li>&gt; Other website(s)</li> </ul>
<b>Health care resources for this disease</b>
<ul style="list-style-type: none"> <li>&gt; Diagnostic tests</li> <li>&gt; Patient organisations</li> <li>&gt; Medical products</li> </ul>
<b>Research activities on this disease</b>
<ul style="list-style-type: none"> <li>&gt; Research projects</li> <li>&gt; Clinical trials</li> <li>&gt; Registries/databases</li> </ul>

**Direct link to PubMed: 3,313 diseases**

**Files available on request**  
**Suitable to code clinical activity / lab activity**



Rare  
diseases

Orphan  
drugs

Clinics

Diagnostic  
tests

Research  
and trials

Patient  
organisations

Directory of  
resources

**Other  
information**

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About rare  
diseases

About orphan  
drugs

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## Orphanet Reports Series

Orphanet reports are a serie of texts covering topics relevant to all rare diseases. New reports are regularly put online and some of these texts are periodically updated. Acrobat Reader is required to read the documents

To install it, [download Acrobat Reader](#) [[↗](#)]

### [Prevalence or reported number of published cases listed in alphabetical order of disease](#)

November 2009 - n°1



### [Diseases listed by decreasing prevalence or number of published cases](#)

November 2009 - n°2



### [Patient Registries in Europe](#)

October 2009

## ***New priorities after Commission Communication and Council Recommendation***

### ***II. Adequate definition, codification and inventorying of rare diseases***

#### ***Revision of the International Classification of Diseases (ICD)***

- **The WHO has launched the process of revision of the International Classification of Diseases (ICD) -10 to prepare the new ICD-11 which should be ready around 2015. The EC is very involved on the process from the side of the Rare Diseases.**
- **Discussions on the revision and improvement of the ICD will be also launched for the mental health disorders.**
- **The EU Task Force on Rare Diseases cooperates with the WHO Topic Advisory Group on Rare Diseases.**
- **5 838 rare diseases listed in Orphanet but only 240 having an explicit code in the ICD-10**
- **An international Workshop on Classification and Codification on Rare Diseases will be organised by the Commission and WHO (Luxembourg, October 2010)**

## ***New priorities after Commission Communication and Council Recommendation***

### ***II. Adequate definition, codification and inventorying of rare diseases***

#### **■ Joint Action to support Orphanet launched in 2010.**

- Orphanet as a tool at country level
  - Identification of national teams
  - National support to teams
  - Governance of national project /inside national strategy
- Policy decision
  - on languages and management of translations
  - on priority products
  - On collaboration to be established with other organisations
- Appropriate budget to run activities according to objectives
- Appropriate tools to serve the users and to support the documentation teams
- European governance with funding bodies

## ***New priorities after Commission Communication and Council Recommendation***

### ***III. Research on rare diseases***

- **Assess research landscape, and improve coordination of Community, national and regional programmes**
- **Identify needs and priorities, and promote interdisciplinary co-operative approaches**
- **Foster participation of national researchers in RD research projects**
- **Include in national plans or strategies provisions aimed at fostering research in the field of RD**
- **Facilitate the development of RD research cooperation with 3rd countries active in RD research, incl. exchange of information and sharing of expertise.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***III. Research on rare diseases***

#### ***First Call for Proposals FP7***

**HEALTH-2007-2.4.4-1: Natural course and pathophysiology of rare diseases. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 3 million).**

**HEALTH-2007-2.4.4-2: Research capacity-building in the field of rare diseases. Coordination or support action.**

→ 11 projects (10 CP + 1 SA) supported for a global budget of € 30 million

#### ***Third Call for Proposals FP7***

**HEALTH-2009-2.4.4-1: Rare neurological diseases. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 6 million).**

**HEALTH-2009-2.4.4-2: Preclinical development of substances with a clear potential as orphan drugs. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 3 million).**

→ 9 projects (7 under 2.4.4-1 and 2 under 2.4.4-2) selected for funding for a global budget of € 45 million – negotiation being finalised

#### ***Fourth Call for Proposals FP7***

**HEALTH.2010.2.4.4-1: Clinical development of substances with a clear potential as orphan drugs. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 6 million).**

**HEALTH.2010.2.4.4-2: ERA-Net on rare diseases. Coordinating action (max EC contribution € 2 million).**

→ Evaluation January/February 2010; entry into negotiations May/June 2010

***New priorities after Commission Communication and Council Recommendation  
IV. Centres of expertise and European reference networks for rare diseases***

**Some suggested criteria by the EU Task Force on Rare Diseases to be fulfilled by the European reference networks are:**

- **Sufficient activity and capacity to provide relevant services and maintain quality of the services provided**
- **Capacity to provide expert advice, diagnosis or confirmation of diagnosis, to produce and adhere to good practice guidelines and to implement outcome measures and quality control**
- **Demonstration of a multi-disciplinary approach;**
- **High level of expertise and experience documented through publications, grants or honorific positions, teaching and training activities**
- **Strong contribution to research**
- **Involvement in epidemiological surveillance, such as registries**
- **Close links and collaboration with other expert centres at national and international level and capacity to network**
- **Close links and collaboration with patients associations where they exist.**
- **Appropriate arrangements for referrals of patients from other Member States established within a framework.**
- **Appropriate capacities to diagnose, to follow-up and manage patients with evidence of good outcomes so far as applicable.**

## ***New priorities after Commission Communication and Council Recommendation IV. Centres of expertise and European reference networks for rare diseases***

**The Work Plan 2006 for the implementation of the EU public health programme, introduced for the first time as a priority in the area of rare diseases: to develop European Networks of Centres of Reference for Rare Diseases. According to this priority 10 Projects have been selected for funding between 2006 and 2009:**

- **European Centres of Reference Network for Cystic Fibrosis with the Klinikum der Johann Wolfgang Goethe-Universität (DE) as Project Leader,**
- **European Network of Centres of Reference for Dysmorphology with The University of Manchester (UK) as Project Leader,**
- **Patient Associations and Alpha1 International Registry with the Stichting Alpha1 International Registry (NL) as Project Leader,**
- **European Porphyria Network: providing better healthcare for patients and their families with the Assistance Publique - Hôpitaux de Paris (FR) as Project Leader,**
- **Establishment of a European Network of Rare Bleeding Disorders, with the Università degli Studi di Milano (IT) as Project Leader.**
- **European network of paediatric Hodgkin's lymphoma – European-wide organisation of quality controlled treatment with the University of Leipzig (D) as Project Leader.**
- **European Network of Reference for Rare Paediatric Neurological Diseases (NEUROPED) with the European Network for Research on Alternating Hemiplegia (AT) as Project Leader.**
- **A reference network for Langerhans cell histiocytosis and associated syndrome in EU with Assistance Publique Hôpitaux de Paris (FR) as Project Leader.**
- **Improving Health Care and Social Support for Patients and Family affected by Severe Genodermatoses – TogetherAgainstGenodermatoses (TAG) with Fondation René Touraine (FR) as Project Leader.**
- **European Reference Network of expert centres in rare anaemias (ENERCA 3) with Hospital Clínic de Barcelona (ES) as Project Leader.**

## ***New priorities after Commission Communication and Council Recommendation IV. Centres of expertise and European reference networks for rare diseases***

### **Actions to develop national/regional centres of expertise and establish EU reference networks**

- **To repertory in an EU list the existing Centres of Expertise identified throughout the Member States by the end of 2010;**
- **To establish a procedure for designation and accreditation methodology of EU Reference Networks for Rare Diseases (according to the future Directive on Cross-border health care);**
- **To provide adequate, long-term public funding to Centres of Expertise in order to ensure their sustainability and continuity of care for patients;**
- **To provide adequate, long-term public funding to European Reference Networks Centres of Expertise in order to ensure their sustainability and continuity of care for patients;**
- **To recommend inclusion in the National Plan for Rare Diseases provisions on the recognition and funding of Centres of expertise and their participation in European Reference Networks;**
- **To recommend the adoption of national initiatives in the National Plans for Rare Diseases on specialised social services;**
- **To provide financial support to networks of specialised social service;**

## ***New priorities after Commission Communication and Council Recommendation***

### ***V. Gathering the expertise on rare diseases at European level***

- **The Health Programme and the FP7 will continue to support, in a coordinated way, registries, databases and biobanks on rare diseases with appropriate financial tools for a sustainable funding when necessary (proposals, operational grants, better coordination with FP7).**
- **The Commission will establish publicly accessible platform for Rare Diseases patient registers, databases and biobanks maybe defining criteria for register accreditation and qualification and the access to data or samples.**
- **A solid benchmarking could be established with successful EU Public Health Programme ongoing projects, having World relevance in the area [EUROCAT (Surveillance of congenital anomalies in Europe), ENERCA (European Network for Rare Congenital Anaemias, or EAIS (European Autism Information System)] or EU FP6 ongoing projects [EUROWILSON, RBDD (Rare Bleeding Disorders Database) or EUROSCA for ataxias].**
- **These project outputs should also supports specific international consensus conferences such as the Consensus Conference on Primary Immunodeficiency, the European Haemophilia Consortium Conferences, the Haemoglobinopathies Conference, etc.**
- **To analyse feasibility of the creation of a public-private foundation for RD, the European Research Foundation for Rare Diseases.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***V. Gathering the expertise on rare diseases at European level***

**An evaluation of possible population screening (including neonatal screening) strategies for Rare Diseases will be launched**

**Call for Tender launched in June 2009 concerning evaluation of population newborn screening practices for rare disorders in Member States of the European Union (18 months)**

***Deliverable 1:* "Report on the practices of NBS for rare diseases implemented in all the Member States including number of centres, estimation of the number of infants screened and the number of disorders included in the NBS as well as reasons for the selection of these disorders". The study includes the necessary tables which list all screening requirements and outputs useful to adopt future decisions in a comparative basis.**

***Deliverable 2:* "Expert opinion on the development of European policies in the field of newborn screening for rare diseases". This expert opinion will also discuss the existing barriers and propose solutions to be implemented, if feasible, at the EU level.**

***Deliverable 3:* "Set up of a European Union Network of experts on Newborn Screening and organization of a Final European Experts Consensus Workshop on Newborn Screening"**

**A consortium leaded by the Istituto Superiore di Sanità (Italy) is working about.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***V. Gathering the expertise on rare diseases at European level***

- **Joint Action between the Commission and the Member States to be launched in June 2010 concerning the support to the EUROCAT (European Congenital Anomalies Surveillance Network).**
- **There are very few rare diseases for which a primary prevention is possible. Still, primary preventive measures for rare diseases will be taken when possible (e.g. prevention of neural tube defects by Folic Acid supplementation). Action in this field should be the topic for a debate at EU level led by the Commission aiming to determine for which rare diseases primary preventive measures may be successful.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***V. Gathering the expertise on rare diseases at European level***

#### **Actions to ensure equal access to all EU patients to orphan drugs**

**Between April 2000 and April 2010 a total of 1 113 applications were submitted for designation as orphan medicinal products and the Committee for Orphan Medicinal Products (COMP) adopted 760 positive opinions and 16 negative opinions representing 60 marketing authorisations.**

**Cancer treatment was the most-represented therapeutic area for which the COMP adopted positive orphan-designation opinions (46%). Almost two-thirds of designated orphan medicinal products were for conditions affecting children and the COMP took on average 66 days to evaluate applications— the same as in the previous year.**

- **To explore additional incentives at national or European level to strengthen research into rare diseases and development of orphan medicinal products, and Member State familiarity with these products.**
- **A European guideline should clarify responsibilities in a Compassionate Use situation**
- **The Commission should present, a report to the Council and the Parliament identifying bottlenecks on orphan drugs access (delays, marketing, access, reimbursement, prices, etc.) proposing the necessary legislative modifications in order to guarantee equal access to orphan drugs throughout the EU on the basis of a COMP/EUCERD European collaborative scientific assessment (Commission, EMEA)**

## ***New priorities after Commission Communication and Council Recommendation***

### ***V. Gathering the expertise on rare diseases at European level***

#### **Actions to ensure equal access to all EU patients to orphan drugs**

- **A method for the assessment of the Clinical Added Value of Orphan Medicinal Products should perform a common scientific assessment of the CAV for each Orphan Drug and deliver an opinion document.**
- **Call for Tender tender EAHC/Health/2010/05 concerning the creation of a mechanism for the exchange of knowledge between Member States and European authorities on the scientific assessment of the clinical added value for orphan medicines (**Launched in March 2010**).**
- **The aim of these common assessment reports for CAVOD should be to provide a well-informed opinion on the place of the product with the authorised therapeutic indication in the therapeutic strategy of the rare condition, to the best of current knowledge. For this purpose detailed discussions with the Member States and interested parties, particularly the EMA and the COMP, on the best way to establish such mechanism are necessary.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***VI. Empowerment of patient organisations***

- **The Health Programme will continue to integrate the support to the patient's organisations as a priority for action.**
- **Operational Grants selected for EURORDIS in 2008 and 2009.**
- **The Council Recommendation suggests to Member States to do the same.**

## ***New priorities after Commission Communication and Council Recommendation***

### ***VII. Sustainability***

- **The Third Health Programme 2013-2017 (tbc) should integrate sustainability of rare diseases resources as a priority.**
- **The Member States should use facilities provided to health infrastructures in the Structural Funds (2007-2013) regulations.**
- **Continuity of efforts on rare diseases research in the FP8 integrating a sustainable dimension.**
- **Sustainability in the National Plans/Strategies for Rare Diseases.**

## ***New priorities after Commission Communication and Council Recommendation***

### **Actions to develop the international cooperation on rare diseases**

- **An international cooperation framework on rare diseases with other countries (e.g. US, Canada, Japan, Singapore, Australia,...) will be adopted (Commission)**
- **A proposal of resolution on an international action in the field of rare diseases will be submitted by the European Commission to the World Health Assembly (Commission)**

## ***New priorities after Commission Communication and Council Recommendation***


### **Governance and monitoring**

- **The Commission should be assisted by an EU Committee of Experts on Rare Diseases (EUCERD) to advise on implementation of the Communication and the Recommendation. The Committee will be chaired by the European Commission and will be assisted by a Scientific Secretariat, supported through the Health Programme. Composed by 51 members representing Member States, patient's organisations, Pharmaceutical industry, FP Projects, Health Programme projects and ECDC + 12 Commission representatives (SANCO, RTD, ENTR, EMA, COMP). **First meeting next October 2010 (tbc).****
- **This committee will replace the existing EU Rare Diseases Task Force.**
- **The organisation of a European Rare Diseases day (29 February, a rare day and 28th February in 2010) and European conferences (**the European Conference on Rare Diseases, Krakow, 13-15 May 2010**) to raise awareness of professionals and of the general public will also be encouraged.**

## ***DG SANCO priorities on rare diseases***

### ***Web site***

[http://ec.europa.eu/health/rare\\_diseases/policy/index\\_en.htm](http://ec.europa.eu/health/rare_diseases/policy/index_en.htm)



Contact point at DG SANCO  
[antoni.montserrat@ec.europa.eu](mailto:antoni.montserrat@ec.europa.eu)

Contact point at DG RTD  
[catherine.berens@ec.europa.eu](mailto:catherine.berens@ec.europa.eu)