

# 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

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

### ■ Based on Article 152 of the EU Treaty

■ A Community action programme on RD, including genetic diseases, was adopted for the period of 1 January 1999 to 31 December 2003. Aim: 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.

■ For the period 2008-2013 the Commission has adopted the [White Paper “Together for Health: A Strategic Approach for the EU 2008-2013”](#). 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 it is suggested to put forward EC-level structured cooperation mechanisms to advise the Commission and to promote cooperation between the Member States.

■ As a consequence Rare diseases are now one of the priorities in the [EU Health Programme 2008-2013](#). In the DG SANCO 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,
- 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.

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

- **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.
- In the current Research 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

## ***The Commission Communication and a proposal for a 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 152 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 152 on public health except for the few areas where measures or incentive measures may be adopted (see Article 152.4): they are not binding for member states, but do have a political weight, and aim at preparation of legislation at National level.**

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

### **Actions to improve information, identification and knowledge on 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**
- **The EU will establish since 2009 an 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**

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*New priorities after Commission Communication and Council Recommendation*

*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 recognised as WHO Advisory Group on Rare Diseases.**
- **5 860 rare diseases listed in Orphanet but only 250 having a code in the ICD-10**

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

### **Action to support implementation of National Plans for 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 on 2007). Appropriate international conferences will be organised (18<sup>th</sup> November 2008 in Paris, 21<sup>st</sup> May 2009 in Prague, 29<sup>th</sup> July 2009 in Stockholm)**

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

### **To improve prevention, diagnosis and care of patients with Rare Diseases**

- **Development of e-Health in the field of RD using on-line and electronic tools**
- **Creation of a help line unique EU-wide number for information and social services on rare diseases (e.g. a 116 number)**
- **To launch a European series Patient Leaflets on some rare diseases in all the EU languages**
- **Provide support toward harmonization of quality testing and counselling for rare genetic diseases through initiatives such as EuroGentest**
- **An evaluation of possible population screening (including neonatal screening) strategies for Rare Diseases will be launched**

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

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

**Call for Tender to be 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"**

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

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

- **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 (Commission, EMEA)**
- **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/EUACRD European collaborative scientific assessment (Commission, EMEA)**
- **A method for the assessment of the Clinical Added Value of Orphan Drugs should perform a common scientific assessment of the CAV for each Orphan Drug and deliver an opinion document (Commission, EMEA)**

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

### **Actions to develop national/regional centres of expertise and establish European Reference Networks**

- **To repertory in an EU list the existing Centres of Expertise identified throughout the Member States;**
- **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***

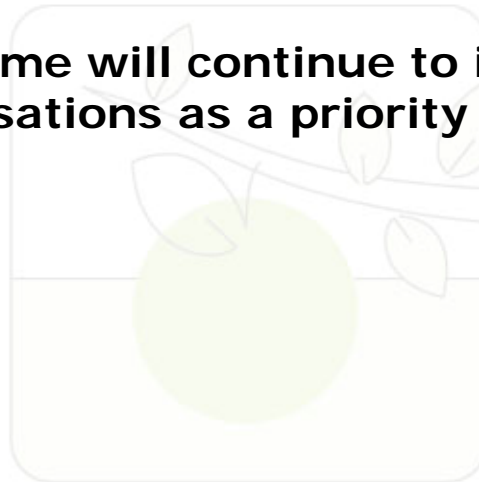
### **Actions to gather at European level the limited and scattered expertise on rare diseases**

- **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**
- **The Commission will establish publicly accessible EU Register of Rare Diseases patient registers databases and biobanks defining criteria for register accreditation and qualification and the access to data or samples.**
- **Specific support to further research into biomarkers should also be given to encourage long-term follow-up, and the acquisition of robust evidence on clinical effectiveness**

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

**Actions to empower patients with Rare Diseases at individual and collective level**

- **The Health Programme will continue to integrate the support to the patient's organisations as a priority for action**



## *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***

### **Actions to coordinate the policies and initiatives at EU level**

- **An EU Committee of Experts on Rare Disease (EUECRD) will be created, by Commission Decision, in order to advise the European Commission.**
- **The European Conferences on Rare Diseases will be organised every two or three years. Funds will be provided by the Health Programme (Commission).**
- **Every four years the Commission should produce an Implementation report on the Commission Communication and Council Recommendation addressed to the Council, the Parliament, the Social and Economic Committee and the Committee of the Regions (Commission)**

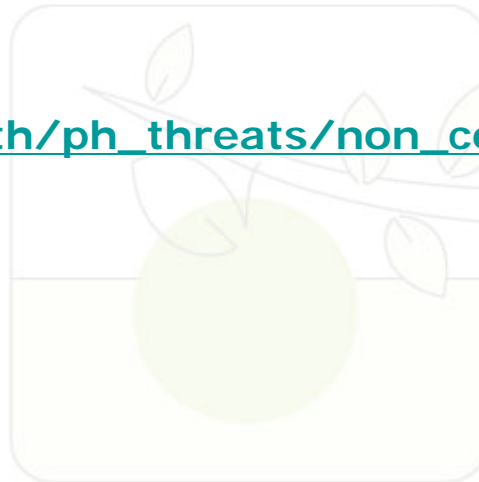
## *Future legal basis for the developments of the EU Public Health Policy*

- **Proposal for a 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).
- **Directive of the European Parliament and of the Council on the application of patients' rights in cross-border healthcare** 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.
- Decision of the Commission creating a **European Union Committee of Experts on Rare Diseases** during 2009. To be composed by 56 members representing Member States, patient's organisations, industry, FP Projects, Health Programme projects, etc.
- This committee will replace the *European Commission Task Force on Rare Diseases* (36 members representing EU funded projects and member states), The RDTF publishes a monthly electronic newsletter on the EC's Rare Diseases actions: [ORPHANews Europe](http://www.orpha.net/actor/EuropaNews/2006/060316.html)
- <http://www.orpha.net/actor/EuropaNews/2006/060316.html>

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

***Web site***

[http://ec.europa.eu/health/ph\\_threats/non\\_com/rare\\_diseases\\_en.htm](http://ec.europa.eu/health/ph_threats/non_com/rare_diseases_en.htm)



## EU historical support to RD research

### Previous programmes:

Promote cooperation, collaboration and knowledge building

### FP5: Total 47 RD projects, € 64 million

• R&D: Friedreich's Ataxia; Peroxisomal diseases; Myopathies; Congenital disorders of glycosylation; Juvenile systemic lupus erythematosus; Cystic fibrosis

• INFRA: European network of DNA, cell and tissue banks on rare diseases

• Biotech: Gene therapy for inherited diseases; Genechip technology for detection of molecularly heterogeneous genetic diseases

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# **PRIORITY 1: Life Sciences, Genomics and Biotechnology for Health**

- **Four calls for proposals in Priority 1**
- **59 RD relevant projects selected for funding (including Priority 8, ERA-Net scheme, and Science and Society)**
- **Global budget for RD: around € 230 million**
- **<http://cordis.europa.eu/lifescihealth/major/rare-diseases-projects-1.htm>**

# Rare Diseases $\neq$ necessary keyword to identify relevant topics in the work programme

## **Advanced genomics and its applications for health**

- Fundamental knowledge and basic tools
- Biotechnology for health

## **Combating major diseases**

- Applications-orientated genomic approaches to medical Knowledge
  - Cardiovascular disease, diabetes and rare diseases
  - Brain and combating diseases of the nervous system
- Combating cancer

## **+ Actions across priority 1**

<http://cordis.europa.eu/lifescihealth/major/rare-diseases-projects-1.htm>

## Aim:

identify the future needs of the  
rare diseases scientific community

Results: recommendations to the EC

### 1. types of projects:

collaborative research projects  
coordination/networking  
emerging teams



## 2. Priorities: open priorities:

- **infrastructures:** identification of genes and haplotyping, protein pathways, animal models, data management, biobanks
- **natural history of diseases (throughout Europe)**
- **mendelian phenotypes of common diseases (including modifier genes)**
- **physiopathology and mechanism of (groups of) diseases (including disease course and modifier genes)**
- **pre-clinical and early clinical studies (including phase 1 and phase 2 clinical trials)**
- **therapeutic interventions:** gene therapies, cell therapies, drugs, devices
- **social sciences (social perception/daily experience/impact of early diagnosis/genetic counselling)**



New  
FP7

## Main elements:

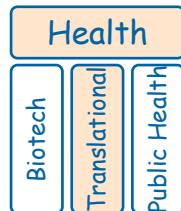


- Duration increased to seven years
- Annual average budget increase  
€ 4.47 (FP6) to € 7.22 billion (FP7)
- Frontier research (~ € 1.1 billion per year)
- New structure:  
Cooperation - Ideas - People - Capacities
- Joint Technology Initiatives
  - ⇒ Arising from European Technology Platforms in fields of major European interest
  - ⇒ "Strategic Research Agenda" defined by stakeholders
  - ⇒ Innovative Medicines Initiative
- Outsourcing

## Health: 3 sub-themes

### Sub-theme 2: Translating research for human health

- Integrating biological data and processes: large-scale data gathering, systems biology
- Research on the brain and related diseases, human development and ageing
- Translational research in infectious diseases: drug resistance, HIV/AIDS, malaria, tuberculosis, Hepatitis, new and re-emerging epidemics, etc.)
- Translational research in major diseases: cancer; cardiovascular disease; diabetes/obesity; **rare diseases**; other chronic diseases (e.g. rheumatic, respiratory, musculo-skeletal diseases and arthritis)



# Rare diseases: topic 1

## HEALTH-2007-2.4.4-1: Natural course and pathophysiology of rare diseases.

Support will be given to innovative, multidisciplinary projects investigating the **natural course** (on a Europe-wide scale) and **pathophysiology** of non-infectious, non-malignant rare diseases in the following six areas: **endocrine, immunological and metabolic diseases**; diseases principally affecting the **genitourinary** tract; diseases principally affecting the **digestive** system; diseases principally affecting the **respiratory** system. Attention should be given to the development of adequate **models** (*in vitro* and animal models). The acquired knowledge will provide the bases for future development of diagnostic, therapeutic and potentially preventive approaches. Research on disorders affecting the nervous system is excluded. Children health and ageing aspects should be taken into consideration whenever appropriate.

**Funding scheme:** Collaborative projects (small or medium-scale focused research projects).



# Rare diseases: topic 2

## **HEALTH-2007-2.4.4-2: Research capacity-building in the field of rare diseases.**

This action is dedicated to developing a team- and project-building platform to help researchers in the field of rare diseases setting up efficient, multidisciplinary teams tackling rare diseases research challenges. This action should offer the opportunity for potential multinational teams to exchange ideas and strategies, for instance through workshops or teleconferences, with a view to structuring future research proposals. Of particular importance is that EU27 is fairly represented. This action is not intended to provide direct support to individual consortia for preparation of their specific project

**Funding scheme:** Coordination and support action (coordination or support action).

# Calls for proposals rare diseases

Other parts of the Theme: other potential entry points e.g.



Networking biobanking initiatives across Europe: developing standards and norms for existing and future human sample biobanks (CP).

High throughput molecular diagnostics in individual patients for genetic diseases with heterogeneous clinical presentation (CP).