

**Proposal for a Commission Communication to the
European Parliament, the Council, the Economic and
Social Committee and the Committee of the Regions on
a European action on rare diseases**

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- **The concept of “rare disease” (RD) emerged in 1978 with the publication of an article**

(Holzman NA. Rare diseases, common problems: recognition and management. Pediatrics, 1978; 62(6): 1056-1060).

stating that rare diseases, though diverse, have common problems of being recognised by physicians and problems of being effectively managed as knowledge about each is very limited and little clinical research in the field exists.

- **In the early 1980’s, rare diseases gained recognition as a political issue with the fight of patient advocacy groups to obtain a set of incentives for the development of therapeutic products in the USA.**
- **In the early 1990’s the debate cantered around research challenges 1990s when rare diseases, which are mostly genetic, became instrumental for mapping human genes.**

- **More recently, the public health dimension of rare diseases has been recognised by the European Commission and by several individual countries and consequently specific action plans were developed.**

Legal basis for the developments of 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 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.**

- **Rare diseases are now one of the priorities in the [EU Public Health Programme 2003-2008](#). According to 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, and the development of strategies and mechanisms for information exchange and co-ordination at EU level to encourage continuity of work and trans-national co-operation.**

- **For the period 2008/2013 a new Public Health Programme should replace the existing Public Health Programme.**

New strand 'Generation of knowledge'.

No 'Diseases' strand.

Future legal basis for the developments of Health indicators in the EU Public Health Policy

In 2007 the Commission plans to adopt a new Health Strategy. This will be an ambitious project and one of DG SANCO's top priorities for the coming year. It aims to:

- **Set a clear strategic framework which covers the broad range of work done within DG SANCO, as well as in other parts of the Commission, and includes some new initiative**
 - **Set broad objectives for a 10 year timeframe, with a 5 year mid-term review**
- **Enable the closest possible cooperation with Member States to improve health in Europe in the decade to come**
- **Focus on key health issues, on mainstreaming health in all policies, and on global health issues address key challenges such as pandemics and other threats, demographic change, new technologies, health inequalities, chronic disease burden, globalisation, patient and health professional mobility(etc)**
 - **Need of a Commission Communication in the field of rare diseases**

Ongoing DG SANCO priorities on rare diseases

- 1. EU Projects identifying rare diseases and assessing prevalence**
- 2. EU Projects supporting cooperation between rare diseases organisations**
- 3. EU Projects creating networks of action for rare diseases**
- 4. European Conferences on Rare Diseases**
- 5. The European Commission Task Force on Rare Diseases**
- 6. EU action to improve classification and codification of rare diseases in the next ICD-11 (International Classification of Diseases)**
- 7. Contribute to the Orphan drugs strategy**
- 8. Contribute to the European Community Framework Programme (FP7)**
- 9. Pilot reference networks (centres of reference) for rare diseases**
- 10. A proposal for a Communication and a proposal for a Council Recommendation on rare diseases**

EU Projects creating networks of action for rare diseases

- 1. A priority for action is to guarantee**
- 2. the exchange of information via existing European information networks on rare diseases,**
- 3. to promote better classification,**
- 4. to develop strategies and mechanisms for exchanging information between people affected by a rare disease, volunteers and professionals,**
- 5. to define relevant health indicators**
- 6. to develop comparable epidemiological data at EU level,**
- 7. to support an exchange of best practise and develop measures for patient groups.**
- 8. Registries and databases constitute a strong priority to guarantee patient's information of quality and a solid information basis permitting an efficient monitoring, research and knowledge management of all rare diseases.**

EU Projects creating networks of action for rare diseases

- **A solid benchmarking could be established with successful EU Public Health Programme ongoing projects, having World relevance in the area [ORPHANET, EUROCAT (Surveillance of congenital anomalies in Europe), ENERCA (European Network for Rare Congenital Anaemias, or EAIS (European Autism Information System)] with**
- **EU FP6 ongoing projects [EUROWILSON, RBDD (Rare Bleeding Disorders Database) or EUROSCA].**
- **These project outputs should also supports specific international consensus conferences such as the Consensus Conference on Primary Immunodeficiency in 2006 or the European Haemophilia Consortium Conferences.**

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.**

DG SANCO priorities on rare diseases

Pilot reference networks (centres of reference) for rare diseases

- **DG SANCO has established the High Level Group on Health Services and Medical Care as a means of taking forward the recommendations made by the reflection process on patient mobility. One of the Working Groups of this High Level Group refers to reference networks (centres of reference).**
- **In 2006 the Rare Diseases Task Force Working Group on centres of reference has submitted a report 'Contribution to policy shaping: For a European collaboration on health services and medical rare in the field of rare diseases' updating the information about Centres of Reference in Europe. The report details the use of the concept of centres of reference in Europe as well as the respective functions.**

Pilot reference networks (centres of reference) for rare diseases

The suggested criteria to be fulfilled by the European centres of reference 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.**

Pilot reference networks (centres of reference) for rare diseases

The Work Plan 2006 for the implementation of the EU public health programme, introduces as a priority in the area of rare diseases: to develop European Networks of Centres of Reference for Rare Diseases. According to this priority some Projects have been selected for funding:

- **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 Porphyrria 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.**

The European Commission Task Force on Rare Diseases

- **The main objectives of the Task Force on Rare Diseases are to identify morbidity and mortality indicators for rare diseases, set up a common framework in the field of public health for rare diseases, and to produce an electronic newsletter.**
- **To better emphasize the importance of and action in the field of rare diseases, the European Commission set up an advisory structure: the Task Force on Rare Diseases (RDTF).**
- **The Task Force is assisted by a Scientific Secretariat which was set up to contribute to the development of public health action in the field of rare diseases**
- **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>**

A proposal for a 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).**

DG SANCO priorities on rare diseases

A proposal for a Communication and a proposal for a Council Recommendation on rare diseases

- Recommendations are without **legal force** but are negotiated and voted on according to the **appropriate procedure**. Recommendations differ from **regulations, directives and decisions**, in that they are not binding for Member States. Though without legal force, they do have a political weight.
- The Recommendation is an instrument of indirect action aiming at preparation of legislation in Member States, differing from the Directive only by the absence of obligatory power.
- In order to coordinate the position of the Member States in respect of this important field, it is considered that a Recommendation is the appropriate legal instrument. In the case of the proposed Communication on rare diseases, it is considered necessary to accompany that Communication with a Proposal for a Council Recommendation on rare diseases covering the areas of:

DG SANCO priorities on rare diseases
A proposal for a Communication and a proposal for a Council Recommendation on rare diseases

- **Common definition of rare diseases in the EU**
- **Establishment of an inventory of RD**
- **Dissemination of appropriate information and Support to information networks**
- **Necessity of national plans for rare diseases in the EU Member States and European guidelines for the elaboration of the national plans for rare diseases**
- **Common databases and medical protocol for the identification of rare diseases**
- **Common approach for a better codification and classification of rare diseases in the process of revision of the International Classification of Diseases**
- **Creation of a EU Working Group of rare diseases as advisory group for WHO in this revision process**
- **Common approach to the support of patient's organisations**

DG SANCO priorities on rare diseases

A proposal for a Communication and a proposal for a Council Recommendation on rare diseases

- **Common definition of rare diseases in the EU**
- **Development of national/regional centres of reference and establish EU reference networks**
- **Development of e-Health in the field of RD**
- **Availability and accessibility of accurate diagnostic tests, including genetic tests**
- **Evaluation of population screening (including neonatal screening) strategies for RD**
- **Primary preventive measures when possible**
- **Best practices on RD care**
- **Equal access to orphan drugs**
- **Orphan Medical devices and orphan diagnostics**
- **Health Technology Assessment of Orphan Drugs**
- **Coordinated compassionate use programme**
- **Specialised social services**

DG SANCO priorities on rare diseases
A proposal for a Communication and a proposal for a Council Recommendation on rare diseases

- **Supporting databases, registries, repositories and biobanks**
- **Biomarkers**
- **Data protection**
- **Networks of research for RD**
- **Coordination between MS funding agencies**
- **Intensifying Research**
- **Common approach to the empowerment of patient organisations**
- **Development of health indicators in the field of RD**
- **Organisation of European Conferences on RD**
- **Creation of the EU Advisory Committee on RD**
- **Rare Diseases in the EU budget**
- **Establishment of a Community Agency for RD**
- **Regular report on the situation of RD in the EU**

DG SANCO priorities on rare diseases
A proposal for a Communication and a proposal for a Council Recommendation on rare diseases

June 2007: First Draft in consultation with Task Force

August 2007: Final Draft

September 2007: Process of consultation starts with specialised bodies

November 2007-February 2008: Public consultation

March 2008: Impact Assessment

March-October 2008: Discussion in the European Parliament, Council, Economic and Social Committee and Committee of the Regions

End 2008: Expected adoption of the Communication under French Presidency of the Council

DG SANCO priorities on rare diseases

Web site

- http://ec.europa.eu/health/ph_threats/non_com/rare_diseases_en.htm

■ **Public consultation**

Responses to this consultation, focussed around the specific questions identified in the text above, should be sent to the Commission by 14 February 2008, by email to

sanco-rarediseases-consultation@ec.europa.eu,

or by post to:

European Commission

Health and Consumer Protection Directorate-General

Rare Diseases consultation

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