EUROPEAN COMMISSION
HEALTH AND CONSUMERS DIRECTORATE-GENERAL

Public Health and Risk Assessment
Health information

NOTE TO HEALTH INFORMATION COMMITTEE

Subject: European action in the field of Rare Diseases

Rare diseases, including those of genetic origin, are life-threatening or chronically debilitating diseases which are of such low prevalence (less than 5 per 10 000) that special combined efforts are needed to address them so as to prevent significant morbidity or perinatal or early mortality or a considerable reduction in an individual's quality of life or socio-economic potential. It is estimated that between 5 000 and 8 000 distinct rare diseases exist today, affecting between 6% and 8% of the population in total. In other words, between 27 and 36 million people in the European Union (with 27 Member States) are affected by a rare disease. The specificities of rare diseases - limited number of patients and scarcity of relevant knowledge and expertise - single them out as a unique domain of very high European added-value. There is probably no other area in health where the collaboration between the 27 different national approaches can be as efficient and effective as rare diseases

Recent legal basis

- 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. Adopted 8th June 2009.

- Decision of the Commission creating a European Union Committee of Experts on Rare Diseases during 2009. Adopted 30th November 2009.

- Proposal of Directive of the European Parliament and of the Council on the application of patients' rights in cross-border healthcare providing for the development of European reference networks (ERNs) to be facilitated by the Member States. To be adopted End 2010 (tbc).

Main actions to implement these Council and Commission objectives

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)

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.). They will be analyzed by health authorities (Krakow, 13th May 2010) and in 16 national conferences (2010). The continuity of EUROPLAN after 2011 (under the form of a Joint Action) is under discussion.

Five Member States have adopted National Plan/Strategy for Rare Diseases (FR, BG, PT, GR and ES).

The project 'SOCIAL ECONOMIC BURDEN AND HEALTH-RELATED QUALITY OF LIFE IN PATIENTS WITH RARE DISEASES IN EUROPE (BURQOL-RD)' has been launched 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.

II. Adequate definition, codification and inventorying of rare diseases

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 database Orphanet will be supported using appropriate financial instruments. A Joint Action to support Orphanet launched in 2010 with appropriate budget to run activities according to objectives to obtain appropriate tools to serve the users and to support the documentation teams and having European governance.

III. 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 have been defined in 2008 by the Rare Diseases Task Force (RDTF) and endorsed by the Working Group on European Reference Networks (ERN). Essentially these criteria should permit to share knowledge that never will exists for 6 000 diseases in every one of the 27 Member States providing sufficient activity and capacity to provide relevant services and maintain quality of the services provided and 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 and demonstration of a multi-disciplinary approach. 10 pilot ERN have been selected by the Health Programme to test the concept. When the Directive on Cross-border healthcare will be in force, the ERN will have a solid legal basis.

IV. 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. The Commission will

establish publicly accessible platform for Rare Diseases patient registers defining criteria for register accreditation and qualification and the access to data or samples.

An evaluation of possible population screening (including neonatal screening) strategies for Rare Diseases has been launched in June 2009 concerning evaluation of population newborn screening practices for rare disorders in Member States of the European Union (18 months). It should produce:

- 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.

- 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.

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. The 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 has been launched last 19th March 2010.

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 plus Commission representatives. First meeting next 6th May 2010 in Brussels. 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.

Action for the Committee:

X	For information
	For comments
	For agreement