

**REPORT OF THE RESEARCH TASK FORCE ON RARE DISEASES
DG SANCO, LUXEMBOURG NOV 13th 2008**

**PRESENTATION ON THE COMMISSION COMMUNICATION ON A EUROPEAN ACTION IN
THE AREA OF RARE DISEASES (INCLUDING GENETIC DISEASES)**

The Commission has planned during 2007 to adopt a new Health Strategy, which includes the need for a Commission Communication in the field of Rare Diseases (RD). The Impact Assessment and Strategy White Paper have been developed and on November 11th 2008, the first approved draft of the Communication was released, ready to be discussed and approved by the European Council.

After Council approval, the document will need to be discussed in the Parliament, with a target date of June 2009 under the Czech Presidency.

This document is of great value for the rare disease field since it highlights the need to improve research in RD, the need to fill the scarcity of expertise which is responsible for delayed diagnosis, and the inadequacy of treatment which complicates the already complex life of patients.

Therefore the objectives of this Communication are:

- 1) to improve recognition and visibility of RD by placing a common coding and classification system throughout Europe by disseminating knowledge and information on RD
- 2) to support policies on RD in the Member States to create a common strategy by: developing national plans; fostering research; identifying national and a regional centres of expertise which will be encouraged to participate in the European Reference Network; gathering and pooling information at European level to create information networks in order to develop comparable epidemiological data; supporting exchange of best practice. This is intended to facilitate access to orphan drugs. A working group will be set up to exchange knowledge between member state and EU authorities on the scientific assessment of the clinical added value of orphan medicines. The Commission will evaluate existing guidelines for the compassionate use of orphan drugs and incentives for orphan drug development. A major interest was expressed for telemedicine and e-health.
- 3) to develop cooperation, coordination and regulation for RD.

The actions to achieve these goals will involve the revision of coding and classification of RD, the dissemination of knowledge and information regarding RD, and the creation of disease information networks with major efforts toward the development of comparable epidemiological data and the exchange of best practice. Important also will be the development of national/regional centre of expertise and the establishment of EU reference networks.

The Communication stresses the need to extend access to orphan drugs, and this will be evaluated by a working group directed to exchange between the Member States and Authorities on the scientific assessment of the added value of orphan drugs.

The need for a proper compassionate use of Orphan drugs was also highlighted and EMEA is invited to revise the guidelines to ensure a common approach across the Community.

E-health programmes are encouraged for dissemination and for a better follow up of patients. Screening approaches and establishment of quality management of diagnostic laboratories are fundamental.

An EU Advisory Commission on RD will be formed by a representative of the National Institute of Health of the member state, and representatives of patients' associations (EURORDIS), DG SANCO, DG Research, DG Enterprise and EMEA.

A very active discussion followed the presentation of the Communication highlighting the need to take into consideration the patient's needs and the sustainability of the programme. This should be one of the major roles of the Advisory Committee.

THE EUROPEAN RESEARCH NETWORK:

This project aims to define a consensus on what a Centre of Expertise (CE) must be and what it should do. A consensus has been reached that a CE must have the appropriate capacity to diagnose and manage patients, that it should be multidisciplinary, capable of producing and adhering to good clinical practice, able to provide expert advice, and be a centre of attraction linked to other countries.

Accordingly, a European Research Network (ERN) was recommended to overcome limited experience, maximise cost-effectiveness, share knowledge and training opportunities and provide small countries with a full range of expert services interacting with public health networks to generate guidelines and produce information for professionals and patients.

It was proposed that an ERN should be established through calls for proposals and a competitive process, although a series of questions were raised about how to ensure strategic relevance through a limited number of centres in the network, how to sustain relevant actions, how to liaise with E-Medicine issues, and whether EUROPLAN might be of help?

EUROPLAN is a project coordinating 23 countries and 29 partners coupled to EURORDIS and aimed at collecting information regarding initiatives undertaken in each state, and evaluating indicators for monitoring the impact of RD in each country. EUROPLAN analyses case studies to identify successful experience, generates recommendations including guidance to develop national plans, and sets up national conferences to transfer recommendations to stakeholders (europplan@iss.it).

ORPHANET

Dr. S. Ayme presented the most recent achievements of Orphanet (www.orpha.net). Orphanet is indeed a platform of inestimable value for the specialist and for the stakeholder interested in rare diseases. Recently, it has further improved the information in the identity card of each disease with epidemiological data, mode of inheritance and codes, and many more options to find appropriate expert resources such as clinics, reference centres and medical labs. The site represents a unique resource to look for nomenclature and classifications of the over 5000 RD.

HEALTH INDICATORS FOR RD AND CLASSIFICATION OF RD

A very active discussion took place on health indicators and classification of RD. The need for health indicators for RD is crucial for assessing the present status of RD and the monitoring of health policies in this field.

Indicators for RD have the aim of measuring RD globally and individually as public health issues for visibility/advocacy, to identify targets for interventions, to allocate appropriate resources, to enable surveillance of status and trends to measure the impact of prevention, diagnosis/ screening and care, to identify etiological and modifying factors, to analyse geographically differences and changes over time, to document the influence of health policy measures, and to guide new research initiatives.

The discussion is still ongoing and a lot of work is still needed.

OUTCOME FOR PROPOSALS IN 2008

An Operating Grant has been awarded to EURORDIS to create a joint action to organise specific networks for RD. This programme was successful since it has demonstrated a very good and extended network of collaborations. Other calls will be published in the future.

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