Communication from the European Commission « Rare Diseases, Europe's challenge » and Council Recommandation on an action in the field of rare diseases

Executive Agency for Health and Consumers

EURO PLAN
European Project for Rare Diseases National Plans Development

EURORDIS
Rare Diseases Europe

Europe's overall strategy for rare diseases
Founding texts

- **Regulation of the European Parliament and of the Council of**
- **on orphan medicinal products**
  - ***Adopted on 16 December 1999***
- **Communication from the European Commission** to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions **: « Rare Diseases, Europe's challenge »**
  - ***Adopted on November 11th, 2008***
- **Council Recommandation on an action in the field of rare diseases**
  - ***Adopted on June 9th, 2009***
- **EUROPLAN selected indicators to evaluate the achievements of RD initiatives**
  - ***January 10th, 2010, Madrid***
- **EUROPLAN draft recommendations for the development of national plans for rare diseases : guidance document**
  - ***Currently discussed in 15 National conferences***

Rare Diseases, Europe’s challenge

- The European Union considers diseases to be rare when they affect not more than 5 per 10 000 persons;
- Between 5 000 and 8 000 different rare diseases affect between 27 and 36 million people in the European union;
- They are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity. Therefore they call for a global approach based on special and combined efforts.

- Actions undertaken aim at improving the access and equity to prevention, diagnosis and treatment for patients suffering from a rare disease throughout the European Union, according to the principles and overarching values of universality, access to good quality care, equity and solidarity endorsed by the Council conclusions on common values and principles in EU health systems in 2006.

Because of the limited number of patients
and scarcity of relevant knowledge and expertise

**Rare diseases are a unique domain
of very high European added-value**

National plans or strategies are to be established and implemented in Member States preferably by the end of 2013 at the latest.

- Council Recommendation (1) : Establish and implement plans or strategies for rare diseases at the appropriate level, or explore appropriate measures for rare diseases in other public health strategies, in order to aim to ensure that patients with rare diseases have access to high quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible effective orphan drugs

I. Plans and strategies in the field of rare diseases should

- Aim at guiding and structuring actions within the framework of national health and social systems;

- Integrate current and future initiatives at local, regional and national levels for a comprehensive approach;

- Define a limited number of priority actions with objectives and follow-up mechanisms;

- Take note of the development of guidelines and recommendations for their elaboration in the framework of the European Project for Rare Diseases National Plans Development (EUROPLAN),
- ***In order to be integrated into a common European effort.***

II. Adequate definition, codification and inventorying

- (2) Use a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons.
- (3) Ensure that rare diseases are adequately coded and traceable in all health information systems, encouraging an adequate recognition of the disease in the national healthcare and reimbursement systems based on the ICD, while respecting national procedures(ongoing revision of the ICD supported by the Commission).
- (4) Contribute actively to the development of the EU easily accessible and dynamic inventory diseases based on the Orphanet network and other existing networks.
- (5) Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance.

- ***In order to provide a framework for recognition of rare diseases and sharing of knowledge and expertise.***

III. Research on rare diseases

- ***For most severe rare diseases that would potentially be treatable, there is simply no current specific treatment :***
- (6) Identify ongoing research and research resources in the national and Community frameworks in order to establish the state of the art and improve the coordination of Community, national and regional programmes for rare diseases research.
- (7) Identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them, and promote interdisciplinary cooperative approaches to be complementarily addressed through national and Community programmes
- (8) Foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels, including the Community level.
- (9) Include provisions aimed at fostering research in the field of rare diseases.
- (10) Facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases and more generally with regard to the exchange of information and the sharing of expertise.

IV. Centres of expertise and European reference networks

- (11) Identify appropriate centres of expertise by the end of 2013, and consider supporting their creation.
- (12) Foster their participation in European reference networks respecting
- the national competences and rules with regard to their authorisation or recognition.
- (13) Organise healthcare pathways for patients through the
- establishment of cooperation with relevant experts and exchange of professionals and expertise within the country or from abroad when necessary.
- (14) Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.
- (15) Include the necessary conditions for the diffusion and mobility
- of expertise and knowledge in order to facilitate the treatment of patients in their proximity.
- (16) Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.

V. Gathering the expertise on rare diseases at European level

- (17) Gather national expertise on rare diseases and support the pooling of that expertise with European counterparts in order to support:
- (a) the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases; ex: helplines, respite care services and therapeutic recreation programmes;
- (b) adequate education and training for all health professionals to make them aware of the existence of these diseases and of resources available for their care;
- (c) the development of medical training in fields relevant to the diagnosis and management of rare diseases, such as genetics, immunology, neurology, oncology or paediatrics
- (d) the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences;
- (e) the sharing Member States' assessment reports on the therapeutic or clinical added value of orphan drugs at Community level where the relevant knowledge and expertise is gathered, in order to minimise delays in access to orphan drugs for rare disease patients.

VI. Empowerment of patient organisations

The WHO defined empowerment of patients as a "pre-requisite for health" and encouraged a "proactive partnership and patient self-care strategy to improve health outcomes and quality of life among the chronically ill".

The role of independent patient groups is crucial both in terms of direct support to individuals living with the disease, and in terms of the collective work they carry out to improve conditions for the community of rare disease patients as a whole and for the next generations.

Member States should aim to involve patients and patients' representatives in the policy process and seek to promote the activities of patient groups :

- (18) Consult patients and patients' representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases.
- (19) Promote the activities performed by patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking and outreach to very isolated patients.

VII. Sustainability

Actions should include appropriate provisions to ensure their sustainability over time.

- The development of research and healthcare infrastructures in the field of rare diseases requires long-lasting projects and therefore an appropriate financial effort to ensure their sustainability in the long term. This effort would notably maximise the synergy with the projects developed under the Second Community Health Programme, the Seventh Framework Programme for Research and Development and the successors of these programmes :
- (20) Together with the Commission, aim to ensure, through appropriate funding and cooperation mechanisms, the long-term sustainability of infrastructures developed in the field of information, research and healthcare for rare diseases.