# Rare diseases: European action

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PURPOSE: to establish national plans for rare diseases and ensure adequate definition of and research into such diseases.

ACT: Council Recommendation on an action in the field of rare diseases.

BACKGROUND: it is estimated that between 5 000 and 8 000 distinct rare diseases exist today, affecting between 6 % and 8 % of the population in the course of their lives. In particular, the total number of people affected by rare diseases in the EU is between 27 and 36 million. Because of their low prevalence, their specificity and the high total number of people affected, rare diseases call for a global approach based on special and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve the quality of life and socioeconomic potential of affected persons.

The Commission, in its White Paper 'Together for Health: A Strategic Approach for the EU 2008-2013' of 23 October 2007, which develops the EU Health Strategy, identified rare diseases as a priority for action.

CONTENT: the Council makes a number of recommendations to Member States, notably that they:

- establish and implement **plans or strategies for rare diseases** at the appropriate level, in order to aim to ensure that patients with rare diseases have access to high-quality care, and in particular: (i) elaborate and adopt a plan or strategy by the end of 2013, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems; (ii) take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies; (iii) define a limited number of priority actions; (iv) take note of the development of guidelines and recommendations for the elaboration of national action for rare diseases by relevant authorities at national level;
- use a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons;
- aim to ensure that rare diseases are adequately coded and traceable in all health information systems;
- contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network and other existing networks;
- identify **ongoing researchon rare diseases** and research resources in the national and Community frameworks;
- identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them;
- foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels;
- facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases;
- **identify appropriate centres of expertise** throughout their national territory by the end of 2013, and consider supporting their creation;
- foster the participation of centres of expertise in **European reference networks**;
- organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts;
- support the use of information and communication technologies, such as telemedicine, where it is necessary to ensure distant access to the specific healthcare needed;
- include in their plans or strategies the necessary conditions for the diffusion and mobility of expertise and knowledge;

- encourage centres of expertise to be based on a multidisciplinary approach to care;
- **gather national expertise on rare diseases** and support the pooling of that expertise with European counterparts in order to support: (i) the sharing of best practices on diagnostic tools and medical care; (ii) adequate education and training for all health professionals; (iii) the development of medical training in fields relevant to the diagnosis and management of rare diseases; (iv) the development of European guidelines on diagnostic tests or population screening; (v) the sharing Member States assessment reports on the therapeutic or clinical added value of orphan drugs at Community level;
- **consult patients** and patients representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases;
- promote the activities performed by patient organisations, such as awareness-raising, capacity-building and training;
- 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.

Lastly, the Council invites the Commission to produce, by the end of 2013 and in order to allow proposals in any possible future programme of Community action in the field of health, an implementation report on this recommendation, which should consider the extent to which the proposed measures are working effectively and the need for further action to improve the lives of patients affected by rare diseases and those of their families. The Council also asks to be informed of the follow-up to the Commission Communication on rare diseases on a regular basis.