Rare diseases: European action

2008/0218(CNS) - 11/11/2008 - Legislative proposal

PURPOSE: on establishing national plans for rare diseases and adequate definition of and research on such diseases.

PROPOSED ACT: Council Recommendation.

CONTENT: 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 i.e between 27 and 36 million people in the EU. Most of them suffer from less frequently-occurring diseases affecting one in 100 000 people or less. Because of their low prevalence and their specificity, rare diseases call for a global approach based on special and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve quality of life and socio-economic potential of affected persons.

The Commission wishes to address certain recommendations to **Member States**:

- **establish national plans for rare diseases** in order to ensure to patients with rare diseases universal access to high quality care, including diagnostics, treatments and orphan drugs throughout their national territory on the basis of equity and solidarity throughout the EU. This includes the following: elaborate a comprehensive strategy, by the end of 2011, aimed at structuring all relevant actions in the field of rare diseases in the form of a national plan for rare diseases; define a limited number of priority actions within the national plan, with concrete objectives, clear deadlines, management structures and regular reports; support the development of guidelines for the elaboration of national action for rare diseases in the framework of the ongoing European Project for Rare Diseases National Plans Development (EUROPLAN) selected for funding over the period 2007-2010 in the Public Health Programme; include in the national plans provisions designed to ensure equitable access to high quality care;

- **adequate definition, codification and inventorying of rare diseases**. This includes the following: implement a EU common definition of rare diseases as those **diseases affecting no more than 5 per 10 000 persons**; ensure that rare diseases are adequately coded and traceable in all health information systems; contribute to the establishment of the EU dynamic inventory of rare diseases; support specific disease information networks, registries and databases;

- **research on rare diseases**. Member States should: identify research projects and research resources to establish the state of the art in the area of rare diseases; identify needs and priorities for basic, clinical and translational research, as well as priorities for social research; foster participation of national researchers and laboratories in research projects funded at Community level; include in the national plan provisions aimed at fostering research, especially with a view to the development of tools such as transversal infrastructures as well as disease-specific projects;

- **centres of expertise and European reference networks for rare diseases**: Member States should: identify national or regional centres of expertise throughout their national territory by the end of 2011, and foster the creation of centres of expertise where they do not exist; foster the participation of centres of expertise into European reference networks and provide adequate, long-term public funding; organise healthcare pathways for patients through the establishment of cooperation with relevant experts within the country or from abroad; cross-border healthcare should be supported; ensure that centres of expertise are based on a multidisciplinary approach to care, and that they adhere to the standards defined by the European reference networks for rare diseases taking into due account the needs and expectations of patients and professionals;

- **gathering at European level the expertise on rare diseases**. Member States should: ensure mechanisms to gather national expertise on rare diseases and pool it together with European counterparts in order to support the development of: (a) common protocols such as European reference opinions on diagnostic tools, medical care, education and social care; (b) European guidelines on population screening and diagnostic tests; (c) sharing Member State's assessment reports on the therapeutic added value of orphan drugs at EU level;

- **empowerment of patient organisations**: Member States should: take action to ensure that patients are duly consulted at all steps of the policy and decision-making processes; support the activities performed by patient organisations, such as awareness raising, capacity-building and training;

- **sustainability:** Member States should: ensure through appropriate funding mechanisms the long-term sustainability of research infrastructures and of healthcare infrastructures, as well as European reference networks for rare diseases; cooperate with other Member States to address the need for sustainability of European-wide research infrastructures, common to all Member States and common to the highest possible number of rare diseases; include in the national plan for rare diseases provisions on the need for addressing the issue of financial sustainability for activities in the field of rare diseases.

The Commission is invited to produce an implementation report on the Recommendation on the basis of the information provided by the Member States, not later than in the end of the fifth year after the date of adoption of this Recommendation, and inform the Council on the follow-up of the Communication of the Commission on rare diseases on a regular basis.