



COUNCIL OF  
THE EUROPEAN UNION



## Council Recommendation on action in the field of rare diseases

*2947th EMPLOYMENT, SOCIAL POLICY, HEALTH AND  
CONSUMER AFFAIRS Council meeting*

*Luxembourg, 9 June 2009*

"THE COUNCIL OF THE EUROPEAN UNION,

Having regard to the Treaty establishing the European Community, and in particular the second subparagraph of Article 152(4) thereof,

Having regard to the proposal from the Commission,

Having regard to the opinion of the European Parliament<sup>1</sup>,

Having regard to the opinion of the European Economic and Social Committee<sup>2</sup>,

Whereas:

- (1) Rare diseases are a threat to the health of EU citizens insofar as they are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity. Despite their rarity, there are so many different types of rare diseases that millions of people are affected.

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<sup>1</sup> Legislative resolution of 23 April 2009 (not yet published in the OJ).

<sup>2</sup> Opinion of 25 February 2009 (not yet published in the OJ).

# P R E S S

- (2) The principles and overarching values of universality, access to good quality care, equity and solidarity, as endorsed in the Council conclusions on common values and principles in EU health systems of 2 June 2006, are of paramount importance for patients with rare diseases.
- (3) The Community Action Programme on Rare Diseases, including genetic diseases, was adopted for the period 1 January 1999 to 31 December 2003<sup>3</sup>. This programme defined the prevalence for a rare disease as affecting no more than 5 per 10 000 persons in the EU. A more refined definition based on updated scientific review, taking into account both prevalence and incidence, will be developed using the Second Community Health Programme<sup>4</sup> resources.
- (4) Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products<sup>5</sup> provides that a medicinal product shall be designated as an "orphan medicinal product" when intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting not more than 5 in 10 000 persons in the Community when the application is made.
- (5) 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 other words, although rare diseases are characterised by low prevalence for each of them, the total number of people affected by rare diseases in the EU is between 27 and 36 million. Most of them suffer from less frequently-occurring diseases affecting one in 100 000 people or less. These patients are particularly isolated and vulnerable.
- (6) 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 socio-economic potential of affected persons.
- (7) Rare diseases were one of the priorities of the Community's Sixth Framework Programme for Research and Development<sup>6</sup> and continue to be a priority for action in its Seventh Framework Programme for Research and Development<sup>7</sup>, as developing new diagnostics and treatments for rare disorders, as well as performing epidemiological

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<sup>3</sup> Decision No 1295/1999/EC of the European Parliament and of the Council of 29 April 1999 adopting a programme of Community action on rare diseases within the framework for action in the field of public health (1999 to 2003) (OJ L 155, 22.6.1999, p. 1). Decision repealed by Decision No 1786/2002/EC (OJ L 271, 9.10.2002, p. 1).

<sup>4</sup> Decision No 1350/2007/EC of the European Parliament and of the Council of 23 October 2007 establishing a second programme of Community action in the field of health (2008-2013) (OJ L 301, 20.11.2007, p. 3).

<sup>5</sup> OJ L 18, 22.1.2000, p. 1.

<sup>6</sup> Decision No 1513/2002/EC of the European Parliament and of the Council of 27 June 2002 concerning the Sixth Framework Programme of the European Community for research, technological development and demonstration activities, contributing to the creation of the European Research Area and to innovation (2002 to 2006) (OJ L 232, 29.8.2002, p. 1).

<sup>7</sup> Decision No 1982/2006/EC of the European Parliament and of the Council of 18 December 2006 concerning the Seventh Framework Programme of the European Community for research, technological development and demonstration activities (2007-2013) (OJ L 412, 30.12.2006, p. 1).

research on those disorders, require multi-country approaches in order to increase the number of patients for each study.

- (8) 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.
- (9) In order to improve the coordination and coherence of national, regional and local initiatives addressing rare diseases and cooperation between research centres, relevant national actions in the field of rare diseases could be integrated into plans or strategies for rare diseases.
- (10) According to the Orphanet database, of the thousands of known rare diseases for which a clinical identification is possible, only 250 of them have a code in the existing International Classification of Diseases (ICD) (10th version). An appropriate classification and codification of all rare diseases is necessary in order to give them the necessary visibility and recognition in national health systems.
- (11) In 2007 the World Health Organisation (WHO) launched the process of revision of the 10th version of the ICD in order to adopt the new, 11th version of this classification at the World Health Assembly in 2014. The WHO has appointed the Chair of the EU Rare Diseases Task Force as the Chair of the Topic Advisory Group on Rare Diseases in order to contribute to this process of revision, providing proposals for codification and classification of rare diseases.
- (12) The implementation of a common identification of rare diseases by all the Member States would strongly reinforce the contribution of the EU in this Topic Advisory Group and would facilitate cooperation at Community level in the field of rare diseases.
- (13) In July 2004, a Commission High Level Group on Health Services and Medical Care was established to bring together experts from all Member States to work on practical aspects of collaboration between national health systems in the EU. One of this High Level Group's working groups is focusing on European Reference Networks (ERNs) for rare diseases. Some criteria and principles for ERNs have been developed, including their role in tackling rare diseases. ERNs could also serve as research and knowledge centres, treating patients from other Member States and ensuring the availability of subsequent treatment facilities where necessary.
- (14) The Community added value of ERNs is particularly high for rare diseases by reason of the rarity of these conditions, which implies both a limited number of patients and a scarcity of expertise within a single country. Gathering expertise at European level is therefore paramount in order to ensure equal access to accurate information, appropriate and timely diagnosis and high quality care for rare disease patients.
- (15) In December 2006 an expert group of the European Union Rare Diseases Task Force issued a report "Contribution to policy shaping: For a European collaboration on health services and medical care in the field of rare diseases" to the High Level Group on Health Services and Medical Care. The expert group report outlines, inter alia, the importance of identifying centres of expertise and the roles that such centres should fulfil. It is also agreed that, in principle and where possible, expertise should travel rather than patients themselves. Some measures called for in the report are included in this Recommendation.

- (16) Cooperation and knowledge sharing between centres of expertise has proven to be a very efficient approach to dealing with rare diseases in Europe.
- (17) The centres of expertise could follow a multidisciplinary approach to care, in order to address the complex and diverse conditions implied by rare diseases.
- (18) The specificities of rare diseases – a limited number of patients and a scarcity of relevant knowledge and expertise – single them out as a unique domain of very high added value of action at Community level. This added value can especially be achieved through gathering national expertise on rare diseases which is scattered throughout the Member States.
- (19) It is of utmost importance to ensure an active contribution of the Member States to the elaboration of some of the common instruments foreseen in the Commission Communication on rare diseases: Europe's challenges of 11 November 2008, especially on diagnostics and medical care and European guidelines on population screening. This could be also the case for the assessment reports on the therapeutic added value of orphan medicinal products, which could contribute to accelerating the price negotiation at national level, thereby reducing delays for access to orphan drugs for rare diseases patients.
- (20) 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"<sup>8</sup>. In this sense, 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.
- (21) Member States should aim to involve patients and patients' representatives in the policy process and seek to promote the activities of patient groups.
- (22) 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,

HEREBY RECOMMENDS that Member States:

## **I. Plans and strategies in the field of rare diseases**

- (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, and in particular:

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<sup>8</sup> <http://www.euro.who.int/Document/E88086.pdf>

- (a) elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems;
- (b) take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies for a comprehensive approach;
- (c) define a limited number of priority actions within their plans or strategies, with objectives and follow-up mechanisms;
- (d) take note of the development of guidelines and recommendations for the elaboration of national action for rare diseases by relevant authorities at national level in the framework of the ongoing European Project for Rare Diseases National Plans Development (EUROPLAN) selected for funding over the period 2008-2011 in the first programme of Community action in the field of public health<sup>9</sup>.

## **II. Adequate definition, codification and inventorying of rare diseases**

- (2) Use for the purposes of Community-level policy work a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons.
- (3) Aim to 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.
- (4) 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 as referred to in the Commission Communication on rare diseases.
- (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.

## **III. Research on rare diseases**

- (6) Identify ongoing research and research resources in the national and Community frameworks in order to establish the state of the art, assess the research landscape in the area of rare diseases, 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.

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<sup>9</sup> Decision No 1786/2002/EC of the European Parliament and of the Council of 23 September 2002 adopting a programme of Community action in the field of public health (2003-2008) (OJ L 271, 9.10.2002, p. 1).

- (8) Foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels, including the Community level.
- (9) Include in their plans or strategies 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 for rare diseases**

- (11) Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.
- (12) Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.
- (13) Organise healthcare pathways for patients suffering from rare diseases 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, in their plans or strategies, 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;
  - (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**

- (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**

- (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.

HEREBY INVITES the Commission:

- (1) 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 addressed to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions and based on the information provided by the Member States, 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.
- (2) To inform the Council of the follow-up to the Commission Communication on rare diseases on a regular basis.

Done at

*For the Council  
The President"*

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