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Proposal for a

COUNCIL RECOMMENDATION

on a European action in the field of rare diseases

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THE COUNCIL OF THE EUROPEAN UNION,

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

Having regard to the proposal from the Commission¹,

Having regard to the opinion of the European Parliament²,

Having regard to the opinion of the European Economic and Social Committee³,

Having regard to the opinion of the Committee of the Regions⁴,

Whereas:

- (1) Rare diseases are a threat to the health of European citizens insofar as they are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity.
- (2) A Community Action Programme on Rare Diseases, including genetic diseases, was adopted for the period 1 January 1999 to 31 December 2003⁵. This programme defined the prevalence for a rare disease as affecting no more than 5 per 10 000 persons in the European Union.
- (3) Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December, 1999, on orphan medicinal products 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.
- (4) 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, between 27 and 36 million people in the European Union. Most of them suffer from less frequently-occurring diseases affecting one in 100 000 people or less.
- (5) 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.

¹ OJ C , , p. .

² OJ C , , p. .

³ OJ C , , p. .

⁴ OJ C , , p. .

⁵ 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).

- (6) Rare diseases were one of the priorities of the Community's Sixth Framework Programme for research, technological development and demonstration activities (2002-2006), and continue to be a priority for action in the new Seventh Framework Programme for research, technological development and demonstration activities (2007-2013)⁶ as developing new diagnostics and treatments for rare disorders, as well as performing epidemiological research on those disorders, requires multi-country approaches to increase the number of patients for each study.
- (7) The Commission, in its White Paper "Together for Health: A Strategic Approach for the EU 2008-2013" of 23 October 2007 developing the EU Health Strategy⁷ identified rare diseases as a priority for action.
- (8) In order to improve the coordination and coherence of national, regional and local initiatives addressing rare diseases, all relevant national actions in the field of rare diseases should be integrated into national plans for rare diseases.
- (9) According to the database Orphanet, on the 5 863 rare diseases known and for which a clinical identification is possible, only 250 rare diseases have a code in the existing International Classification of Diseases (10th version). An appropriate classification and codification of all the rare diseases is necessary for giving them the necessary visibility and recognition in the national health systems.
- (10) The World Health Organisation (hereinafter "WHO") launched in 2007 the process of revision of the 10th version of the International Classification of Diseases in order to adopt in the World Health Assembly in 2014 the new 11th version of this classification. The WHO has appointed the European Union Rare Diseases Task Force as Topic Advisory Group on Rare Diseases in order to contribute to this process of revision providing proposals for codification and classification of rare diseases.
- (11) The implementation of a common identification of rare diseases by all the Member States would highly reinforce the contribution of the European Union in this Topic Advisory Group and would facilitate the cooperation at Community level in the field of rare diseases.
- (12) In July 2004, a High Level Group on health services and medical care was established⁸ to bring together experts from all Member States to work on practical aspects of collaboration between national health systems in the EU. One of the working groups of this High Level Group is focusing on European Reference Networks (ERNs) for rare diseases⁹. Some principles have been developed, including their role in tackling rare diseases, and some criteria that such centres should fulfil. ERNs should also serve as research and knowledge centres, treating patients from other Member States and ensuring the availability of subsequent treatment facilities where necessary.
- (13) The Community added-value of European reference networks is particularly high for rare diseases by reason of the rarity of these conditions, which implies both limited number of patients and scarcity of expertise within a single country. Gathering

⁶ <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2006:412:0001:0041:EN:PDF>

⁷ See http://ec.europa.eu/health/ph_overview/strategy/health_strategy_en.htm

⁸ In response to the High-level reflection process the Commission adopted a Communication on patient mobility and healthcare developments in the EU - COM(2004) 301, 20.4.2004 - and established a mechanism for taking forward the work set out in the Communication.

⁹ http://ec.europa.eu/health/ph_threats/non_com/rare_8_en.htm

expertise at European level is therefore paramount to ensure equal access to high quality care to rare disease patients.

- (14) 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”¹⁰ to the EC’s High Level Group on Health Services and Medical Care. The expert group report outlines, inter alia, the importance of identifying centres of expertise at national and regional level and the roles that such centres should fulfil. Some measures called for in the report are included in this recommendation.
- (15) Cooperation and knowledge sharing between national and regional centres of expertise proved to be the most efficient approach to deal with rare diseases in Europe.
- (16) The national and regional centres of expertise should follow a multidisciplinary approach to care which would integrate medical and social aspects, in order to address the complex and diverse conditions implied by rare diseases.
- (17) The specificities of rare diseases – limited number of patients and 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.
- (18) 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 Communication of the Commission on rare diseases, especially European reference opinions on diagnostics and medical care and European guidelines on population screening. This should be also the case for the assessment reports on the therapeutic added value of orphan medicinal products, which could contribute to accelerate the price negotiation at national level, thereby reducing delays for access to orphan drugs for rare diseases patients.
- (19) 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”¹¹. In this sense, the role of 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.
- (20) Patients and patients’ representatives should therefore be involved at all steps of the policy and decision-making processes. Their activities should be actively promoted and supported, including financially, in each Member State.
- (21) 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 Community Health Programme (2008-2013), the Community 7th Framework Programme for Research, Technological Development and Demonstration Activities (2007-2013) and the successors of these programmes.

¹⁰ http://ec.europa.eu/health/ph_threats/non_com/docs/contribution_policy.pdf

¹¹ <http://www.euro.who.int/Document/E88086.pdf>

HEREBY RECOMMENDS THAT MEMBER STATES:

1. 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, and in particular:
 - (1) elaborate and adopt a comprehensive and integrated strategy, by the end of 2011, aimed at guiding and structuring all relevant actions in the field of rare diseases in the form of a national plan for rare diseases;
 - (2) take action to ensure that all current and future initiatives at regional and national levels are integrated into their national plan;
 - (3) define a limited number of priority actions within the national plan for rare diseases, with concrete objectives, clear deadlines, management structures and regular reports;
 - (4) support 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 2007-2010 in the Public Health Programme;
 - (5) include in the national plans provisions designed to ensure equitable access to high quality care, including diagnostics, treatments and orphan drugs, to all rare disease patients throughout their national territory with a view to ensuring equitable access to quality care on the basis of equity and solidarity throughout the European Union.
2. Adequate definition, codification and inventorying of rare diseases
 - (1) implement a European Union common definition of rare diseases as those diseases affecting no more than 5 per 10 000 persons;
 - (2) ensure that rare diseases are adequately coded and traceable in all health information systems, contributing to an adequate recognition of the disease in the national health care and reimbursement systems;
 - (3) contribute actively to the establishment of the European Union dynamic inventory of rare diseases as referred to in the Communication;
 - (4) support at national or regional level specific disease information networks, registries and databases.
3. Research on rare diseases
 - (1) identify ongoing research projects and existing research resources in order to establish the state of the art in the area of rare diseases;
 - (2) identify needs and priorities for basic, clinical and translational research in the field of rare diseases, as well as priorities for social research;
 - (3) foster participation of national researchers and laboratories in research projects on rare diseases funded at Community level;
 - (4) include in the national plan for rare diseases provisions aimed at fostering research, including public health and social research, in the field of rare

diseases, especially with a view to the development of tools such as transversal infrastructures as well as disease-specific projects.

4. Centres of expertise and European reference networks for rare diseases
 - (1) 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 notably by including in their national plan for rare diseases provisions on the creation of national or regional centres of expertise;
 - (2) foster the participation of national or regional centres of expertise into European reference networks and provide adequate, long-term public funding in order to ensure their sustainability and thus continuity of care for patients.
 - (3) organise healthcare pathways for patients through the establishment of cooperation with relevant experts within the country or from abroad when necessary; cross-border healthcare, including mobility of patients, health professionals and providers and provision of services through information and communication technologies should be supported where it is necessary to ensure universal access to the specific healthcare needed;
 - (4) ensure that national or regional centres of expertise are based on a multidisciplinary approach to care when addressing complex and diverse conditions such as rare diseases; and promote the integration of medical and social levels within the centres;
 - (5) ensure that national or regional centres of expertise 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.
5. Gathering at European level the expertise on rare diseases
 - (1) 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 and recommendations 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, in order to minimise delays for access to orphan drugs for rare disease patients;
6. Empowerment of patient organisations
 - (1) take action to ensure that patients and patients' representatives are duly consulted at all steps of the policy and decision-making processes in the field of rare diseases, including for the establishment and management of centres of expertise and of European reference networks and for the elaboration of national plans;
 - (2) support the activities performed by patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking, outreach to very isolated patients;

- (3) include in the national plans for rare diseases provisions on the support to and the consultation of patient organisations as referred to in paragraphs (1) and (2).

7. Sustainability

- (1) ensure through appropriate funding mechanisms the long-term sustainability of research infrastructures, such as biobanks, databases and registries, and of healthcare infrastructures such as centres of expertise, as well as European reference networks for rare diseases;
- (2) 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;
- (3) 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.

HEREBY INVITES THE COMMISSION:

1. To produce an implementation report on this Recommendation addressed to the Council, the European Parliament, the European Economic and Social Committee and the Committee of the Regions 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, to consider the extent to which the proposed measures are working effectively, and to consider the need for further action.
2. To inform the Council on the follow-up of the Communication of the Commission on rare diseases on a regular basis.

Done at Brussels,

*For the Council
The President*