Opinion of the European Economic and Social Committee on the Proposal for a Council Recommendation on a European action in the field of rare diseases

COM(2008) 726 final — 2008/0218 (CNS)

(2009/C 218/18)

On 28 November 2008, the Council decided to consult the European Economic and Social Committee, under Article 262 of the Treaty establishing the European Community, on the

'*Proposal for a Council Recommendation on a European action in the field of rare diseases*'

The Section for Employment, Social Affairs and Education, which was responsible for preparing the Committee's work on the subject, adopted its opinion on 4 February 2009. The rapporteur was Ms CSER.

At its 451st plenary session, held on 25 and 26 February 2009 (meeting of 25 February 2009), the European Economic and Social Committee adopted the following opinion by 162 votes to 4, with 8 abstentions.

1. Conclusions and recommendations

1.1 The EESC welcomes the proposal for a Council Recommendation, and endorses the harmonised EU approach to identifying, defining and classifying rare diseases.

1.2 The EESC agrees that national and regional centres of expertise should be designated and their participation in European Reference Networks encouraged and fostered.

1.3 The EESC endorses the support given to coordinated research currently being carried out into rare diseases and the steps to promote both this research and coordination projects aimed at making the best use of limited funding, together with a move towards closer international cooperation.

1.4 As regards pooling expertise at European level, the EESC recommends that intellectual property rights be taken into account and suitable guarantees offered.

1.5 The EESC supports the idea of drawing up national plans, but feels that 2011 is too early for the plans to be prepared in the requisite detail.

1.6 The EESC is pleased that national and regional centres are to be identified by 2011; however, this will depend on the national plans being adequately prepared.

1.7 The EESC recommends that coordination and information flows be established across Europe, and that shared and standardised technical terminology be developed. It would also be useful to prepare a handbook to facilitate dialogue between different professional cultures, covering the particular features of the sector in hand.

1.8 The EESC recommends that a special communication and reporting system be developed to enable a reference network and 'mobile service' to function in such a way that all concerned have access to the information they need.

1.9 The EESC is pleased that sociological research is also to play a role in pinpointing the needs arising in connection with rare diseases.

1.10 The EESC recommends that all Member States establish their own centres for rare diseases, able to play a part in coordinating research and medical institutions, healthcare providers and governments.

1.11 The EESC recommends that national centres for rare diseases deal with tasks relating to data compilation, accreditation, methodology and coordination.

1.12 The EESC recommends making national strategies on rare diseases an integral part of national public health programmes.

1.13 The EESC recommends developing long-term sources of funding rather than using project-based funding, in order to ensure a more efficient, productive use of resources and to protect patients' rights more effectively.

1.14 The EESC recommends looking into the involvement of patients' associations, professional organisations, other civil society organisations and the social partners here, and analysing and evaluating scope for using reference centres located in other Member States, taking patients' rights and interests into account.

1.15 The EESC recommends further analysis of healthcare professionals' commitment and activities in this field, involving professional organisations, civil society organisations and the social partners with a view to ensuring that the requisite guarantees are in place.

1.16 In order to reduce inequalities in healthcare, the EESC suggests looking at conditions for ensuring a balanced use of resources, given that the recommendation sets the goal of providing healthcare for patients suffering from all rare diseases.

1.17 The EESC supports the creation of an EU Advisory Committee on Rare Diseases (EUACRD), and recommends that, in addition to representatives of the Member States, the healthcare industry, patients' organisations and experts, the social partners and other organisations of the civil society, should be involved; otherwise it will not be possible to develop a national strategies, which is one of the prerequisites for implementing the recommendation.

1.18 The EESC recommends that international health policy support the initiative of the European Day of rare diseases, with a view to it becoming an international event.

1.19 The EESC agrees that a report on implementation should be compiled five years after adoption of the recommendation, but insists that the requisite changes be made during implementation, taking patients' rights into account. The EESC would like to be involved in the continuous evaluation of such implementation.

2. **General comments**

2.1 Background

2.1.1 Rare diseases, including genetic diseases, were the subject of a Community action programme covering the period from 1 January 1999 to 31 December 2003 (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)); in this, rare diseases are defined as illnesses which do not affect more than 5 out of every 10 000 people in the EU. Regulation (EC) No 1431/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products is also based on this definition.

2.1.2 Because of their low prevalence and their very specific nature, rare diseases call for a global approach based on special, combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve the quality of life and socio-economic potential of those affected.

2.1.3 The group working on the European reference network on rare diseases set up by the European Commission will have to develop basic principles and treatment, as well as criteria for the European reference centres. These issues are also dealt with in the 6th and 7th R&D Framework Programmes.

2.1.4 In 2014 the World Health Organisation (WHO) is planning to adopt the 11th version of the International Classification of Diseases, which likewise includes rare diseases. The WHO has requested that the EU's task force on rare diseases act as an advisory body in the codification and classification of rare diseases.

2.1.5 A uniform definition of rare diseases in all Member States would significantly enhance the EU's contribution to its collaboration with the WHO, and give the EU a stronger role to play in solving health problems in the rest of the world.

2.1.6 The European Health Strategy, adopted in 2007, sets high-quality diagnosis, treatment and information as key priorities for persons affected by rare diseases.

3. **Specific comments**

3.1 Rare diseases: definition and occurrence

3.1.1 Rare diseases call for a global approach based on special, combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve the quality of life and socio-economic potential of those affected.

3.1.2 As much as 6 % of the EU's total population is affected at some time during their lives by between 5 000 and 8 000 distinct rare diseases; in other words, 29-36 million Europeans are already or will be affected by a rare disease.

3.1.3 The frequency of most rare diseases is extremely low – 1 in every 100 000 persons or even lower. Patients with very rare diseases and their families are particularly isolated and vulnerable.

3.1.4 The age at which the first symptoms appear varies considerably; about half of rare diseases appear at birth or during childhood, whereas the remaining half can appear during adulthood. Most rare diseases are genetic diseases, but they can also result from environmental exposure during pregnancy or later in life, often in combination with genetic susceptibility. Some are rare forms or rare complications of common diseases.

3.2 Lack of recognition and awareness of rare diseases

3.2.1 Rare diseases differ widely in severity and in expression. Persons suffering from rare diseases have a significantly lower life expectancy. Many such diseases are complex, degenerative and chronically debilitating, whilst others are compatible with a normal life - if diagnosed in time and managed and/or treated properly. Several disabilities often co-exist, with many functional consequences. These disabilities enhance the feeling of isolation, possibly resulting in discrimination and reducing any educational, professional and social opportunities.

3.3 Lack of policies on rare diseases in the Member States

3.3.1 Although rare diseases heavily contribute to morbidity and mortality, they are invisible in health care information systems due to the lack of appropriate coding and classification systems. The lack of specific health policies for rare diseases and the scarcity of expertise translate into delayed diagnosis and difficult access to care. National healthcare services for the diagnosis, treatment and rehabilitation of people with rare diseases vary significantly in respect of availability and quality. People from Member States and/or regions within Member States have unequal access to expert services and to orphan drugs.

3.3.2 There are wide variations in the resources available in each Member State for research, diagnosis and treatment, and the dispersal of such resources means that they are not used efficiently, as a result of which many patients are either treated too late or not at all.

3.3.3 Particular expertise is required in the diagnosis and treatment of rare diseases. Due to insufficient resources, there are major discrepancies, and many patients suffer the consequences of incomplete or inaccurate diagnoses.

3.3.4 Given the specific nature of rare diseases – the low numbers of patients and the lack of relevant knowledge and skills – international cooperation is essential and offers added value. Probably no other area of public health offers so much potential for effective, valuable cooperation between the 27 Member States with their differing approaches, as has been acknowledged by decision-makers at European and national level, as well as by all parties concerned. Pooling limiting resources could help achieve better results. Data compilation practices, in terms of the type of data compiled and timing, vary from one country to another; there are also differences in terms of notification requirements. In some cases, such requirements apply to the entire population; in others, there is only sporadic compilation of data. Consistent data and information are vital for formulating and implementing health policies which are both effective in ensuring prevention and financially viable; they also contribute to research at national and EU level. Equally important is providing those concerned with access to the relevant data and information.

3.3.5 It is especially important to improve quality of life for patients suffering from rare diseases and their families, and to ensure that they are suitably integrated into society and labour markets, given that their lives are a constant struggle to overcome physical and mental challenges and the differences between them and other people.

3.3.6 In the EU Member States there are numerous NGOs and civil society initiatives to inform patients suffering from rare diseases, disseminate existing scientific and clinical knowledge, and improve access to affordable and appropriate treatments and medicines, all of which ultimately contribute to the socio-economic integration of such patients. These civil society initiatives are not backed up by enough resources, do not benefit from coordinated, balanced government support, and are not part of an organised network; as a result, patients' rights are constantly undermined. There is no systematic cooperation between patients, their families, civil society organisations, specialists and the social partners. There are sizeable inequalities and accumulated difficulties in terms of the care provided and access thereto.

3.3.7 The diagnosis and treatment of rare diseases are extremely costly procedures. For treatments requiring new technologies or high levels of specific expenditure, it is essential for each Member State to set and apply the highest possible ceiling for funding.

3.3.8 In 2008 the European Commission published a communication on rare diseases, preparation for which involved a wide-ranging consultation procedure completed in February of the same year.

3.3.9 Responses to the consultation procedure confirmed the need for Community-wide measures. The proposed Council recommendation focuses on three areas:

— identifying and codifying rare diseases, and creating a European system to codify and classify rare diseases to support recognition of each one. In the course of preparing a new version of the International Classification of Diseases, the Commission is collaborating with the WHO, given that compared to elsewhere in the world, it is in the EU that various types of rare diseases are most likely to be identified;

— setting fundamental principles and policy guidelines for use in the formulation of national action plans; supporting and encouraging the development of national health policies on rare diseases, aimed at securing equal access to prevention, diagnosis, treatment and rehabilitation, as well as the general accessibility of such services.

— The Council Recommendation set out in the draft Communication envisages the following:

  — Member States are to draw up national plans on rare diseases;

  — mechanisms enabling the definition, codification and classification of rare diseases are also to be put in place;

  — research into rare diseases is to be promoted, for example through cross-border cooperation, making full use of EU research cooperation potential;

  — centres of expertise should be identified and their participation in European Reference Networks encouraged;

  — overall statistics should be compiled on expertise on rare diseases in the Member States;

  — measures should be taken to ensure the involvement of patients and organisations representing them; and

  — closer cooperation is needed in all fields where Community action offers added value in developing joint policy guidelines and ensuring their mutual recognition throughout Europe. This could involve specific measures relating to research, reference centres, access to information, incentives to develop orphan drugs, screening, etc., as elements constituting a minimum common strategy on rare diseases (e.g. pilot programmes, research and development, and steps to monitor implementation of Regulation (EC) No 141/2000 on orphan drugs).

3.3.10 The purpose of the Communication is to support the development of a comprehensive common European strategy to ensure effective recognition, prevention, diagnosis, treatment and research in the field of rare diseases, strengthen cooperation between Member States and provide back-up for European information networks and patients' organisations. A high level of human health protection must be ensured in the definition and implementation of all Community policies and activities. This will in turn contribute to the overarching goal - an improvement in the health situation, and therefore an increase in Healthy Life Years, a key Lisbon Strategy indicator. However, for this to happen, it is vital to ensure greater consistency between Community programmes and initiatives such as the EU's Community public health programmes, the research and development framework programmes, the strategy on orphan drugs, the directive on cross-border healthcare, and other current and future measures at national and EU level.

3.3.11 The proposal for a Council recommendation suggests there is a need for Member States to draw up comprehensive, integrated national strategies on rare diseases by the end of 2011 and involve patients and patients' representatives in all stages of policy and decision-making processes. Their activities should be actively promoted and supported, especially financially.

3.3.12 The EESC is in favour of drawing up comprehensive, integrated national strategies, but feels that the timing should be reconsidered to enable the strategy to take patients' interests into account. For this to happen, centres for rare diseases should be set up in the Member States to work on methodology, data compilation, accreditation and coordination.

3.3.13 In the interests of ensuring an information flow at EU level and promoting research, as well as identifying and developing reference centres, shared and standardised technical terminology must be adopted and developed together with diagnostic and therapeutic protocols. The recognition of such protocols and terminology would not only benefit patients but also healthcare professionals and providers; it would therefore be useful to prepare a practical handbook, to facilitate dialogue between different professional cultures on rare diseases, their diagnosis and treatment.

3.3.14 Identification and development of a European reference network and provision of a 'mobile service' requires specific communication activities and a reporting system so that everyone concerned genuinely has access to the relevant information.

3.3.15 Given that this new set-up for research structures and the provision of services is likely to generate intellectual property, it is vital to take appropriate measures to ensure legal protection.

3.3.16 The EESC welcomes the first European Day of rare diseases held on 29 February 2008, and backs the initiative to introduce a world day of rare diseases, thus setting in motion an international development which would help considerably to boost the effectiveness of research and treatment. The EESC feels that it is absolutely vital to communicate effectively, to promote intercultural dialogue, above all to overcome linguistic barriers, and to remedy shortcomings in technical conditions so that those concerned (patients, their dependents, healthcare service providers, civil society organisations and the social partners) have access to adequate and accurate information.

3.3.17 In several of its previous opinions, the Committee emphasised the key role played by civil society and the social partners in preserving the values of the Community, and in applying such values to deliver genuine improvements. It therefore feels it is vital for stakeholders from organised civil society and the social partners to be given an appropriate role to play in achieving the objectives set out in the Communication on rare diseases. Given that civil society and the social partners generate the resources used to fund public health expenditure, they should be given a strategic role in distributing such resources.

3.3.18 In order to reduce inequalities in healthcare, and in view of the exceptional nature of the expenditure involved, the EESC suggests looking at conditions for making balanced use of resources, given that the recommendation sets the goal of providing healthcare for patients suffering from all rare diseases. The availability of resources varies from one Member State to another, and there are huge discrepancies between the number of persons theoretically entitled to treatment and those who actually receive it.

3.3.19 The EESC is in favour of coordinated research and of steps to identify and establish reference centres, given that this would be an excellent opportunity for the EU to help solve international health problems, in keeping with the objective set out in the White Paper entitled 'Together for Health: A strategic approach for the EU 2008-2013', namely that the EU should play a more effective international role.

3.3.20 Setting up the EUACRD permanent advisory committee is an important step towards achieving this objective. Alongside Member States' representatives, experts, patients' organisations and representatives of the healthcare industry, the EESC recommends that civil society and the social partners be involved in the work of the advisory committee on a permanent basis. Without their involvement, it will not be possible to devise a national strategy, which is one of the prerequisites for implementing the recommendation.

Brussels, 25 February 2009.

The President
of the European Economic and Social Committee
Mario SEPI