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# REPORT

on the proposal for a Council recommendation on a European action in the field of rare diseases
(COM(2008)0726 – C6-0455/2008 – 2008/0218(CNS))

Committee on the Environment, Public Health and Food Safety

Rapporteur: Antonios Trakatellis

### *Symbols for procedures*

| | |
|---|---|
| * | Consultation procedure<br>*majority of the votes cast* |
| **I | Cooperation procedure (first reading)<br>*majority of the votes cast* |
| **II | Cooperation procedure (second reading)<br>*majority of the votes cast, to approve the common position*<br>*majority of Parliament's component Members, to reject or amend the common position* |
| *** | Assent procedure<br>*majority of Parliament's component Members except in cases covered by Articles 105, 107, 161 and 300 of the EC Treaty and Article 7 of the EU Treaty* |
| ***I | Codecision procedure (first reading)<br>*majority of the votes cast* |
| ***II | Codecision procedure (second reading)<br>*majority of the votes cast, to approve the common position*<br>*majority of Parliament's component Members, to reject or amend the common position* |
| ***III | Codecision procedure (third reading)<br>*majority of the votes cast, to approve the joint text* |

(The type of procedure depends on the legal basis proposed by the Commission.)

### *Amendments to a legislative text*

In amendments by Parliament, amended text is highlighted in ***bold italics*.** In the case of amending acts, passages in an existing provision that the Commission has left unchanged, but that Parliament wishes to amend, are highlighted in **bold**. Any deletions that Parliament wishes to make in passages of this kind are indicated thus: [...]. Highlighting in *normal italics* is an indication for the relevant departments showing parts of the legislative text for which a correction is proposed, to assist preparation of the final text (for instance, obvious errors or omissions in a given language version). Suggested corrections of this kind are subject to the agreement of the departments concerned.

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## DRAFT EUROPEAN PARLIAMENT LEGISLATIVE RESOLUTION

**on the proposal for a Council recommendation on a European action in the field of rare diseases**
**(COM(2008)0726 – C6-0455/2008 – 2008/0218(CNS))**

**(Consultation procedure)**

*The European Parliament*,

– having regard to the Commission proposal to the Council (COM(2008)0726),

– having regard to Article 152(4), second subparagraph of the EC Treaty, pursuant to which the Council consulted Parliament (C6-0455/2008),

– having regard to Rule 51 of its Rules of Procedure,

– having regard to the report of the Committee on the Environment, Public Health and Food Safety and the opinion of the Committee on Industry, Research and Energy (A6-0231/2009),

1. Approves the Commission proposal as amended;

2. Calls on the Commission to alter its proposal accordingly, pursuant to Article 250(2) of the EC Treaty;

3. Calls on the Council to notify Parliament if it intends to depart from the text approved by Parliament;

4. Asks the Council to consult Parliament again if it intends to amend the Commission proposal substantially;

5. Instructs its President to forward its position to the Council and Commission.

**Amendment 1**

**Proposal for a recommendation**
**Recital 1**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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. | (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***, but since there are so many different types of rare disease, the total number of people affected is quite*** |

***high***.

**Amendment 2**

**Proposal for a recommendation**
**Recital 2**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (2) A Community Action Programme on Rare Diseases, including genetic diseases, was adopted for the period 1 January 1999 to 31 December 2003. This programme defined the prevalence for a rare disease as affecting no more than 5 per 10 000 persons in the European Union. | (2) A Community Action Programme on Rare Diseases, including genetic diseases, was adopted for the period 1 January 1999 to 31 December 2003. This programme defined the prevalence for a rare disease as affecting no more than 5 per 10 000 persons in the European Union***, a number to be judged on a statistical basis subject to a scientific review***. |

*Justification*

*More flexibility is required to avoid dilemmas arising from diseases with incidence 5,1 or 5,2 etc. per 10 000 persons.*

**Amendment 3**

**Proposal for a recommendation**
**Recital 2 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(2a) Based on this statistical incidence, rare diseases should be meticulously catalogued and reviewed regularly by a scientific committee to determine the need for possible additions.*** |

*Justification*

*It is an absolute necessity to compile the catalogue of rare diseases in the European Union.*

**Amendment 4**

**Proposal for a recommendation**
**Recital 4**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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. | (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, ***although rare diseases are characterised by low prevalence for each one of them, the total number of people affected is quite high, ranging*** 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. |

**Amendment 5**

**Proposal for a recommendation**
**Recital 5**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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. | (5) Because of their low prevalence***,*** their specificity ***and the high total number of cases***, rare diseases call for a global approach based on special and combined efforts***, including in partnership with third countries such as the United States,*** to prevent significant morbidity or avoidable premature mortality, and to improve quality of life and socio-economic potential of affected persons ***in developed and developing countries.*** |

*Justification*

*The specificities of rare diseases - limited number of patients and scarcity of relevant knowledge and expertise - means that cooperation with countries outside the EU can bring benefits, not just to the EU but also in knowledge sharing with poorer and developing countries.*

**Amendment 6**

**Proposal for a recommendation**
**Recital 8**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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. | (8) In order to improve the coordination and coherence of national, regional and local initiatives ***and cooperation between research centres*** addressing rare diseases, all relevant national actions in the field of rare diseases should be integrated into national plans for rare diseases. |

**Amendment 7**

**Proposal for a recommendation**
**Recital 13**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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 expertise at European level is therefore paramount to ensure equal access to high quality care to rare disease patients. | (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 expertise at European level is therefore paramount to ensure equal access ***to accurate information, appropriate and timely diagnosis and*** to high quality care to rare disease patients. |

**Amendment 8**

**Proposal for a recommendation**
**Recital 14 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(14a) On 12 October 2008 the Pharmaceutical Forum adopted its final report, which proposes guidelines to enable Member States, stakeholders and*** |

***the Commission to step up their efforts to guarantee easier and swifter access to orphan medicinal products within the European Union.***

*Justification*

*Following the conclusions of the Pharmaceutical Forum, the Member States have given a political undertaking to eliminate bottlenecks in respect of access to orphan medicinal products via the Community approval procedure. The guidelines set out in the Pharmaceutical Forum's final report need to be given specific mention in the proposal for a recommendation.*

**Amendment 9**

**Proposal for a recommendation**
**Recital 20**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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. | (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***, but also at EU level in terms of pan-EU patient support networks for specific rare diseases***. |

*Justification*

*Because of the rarity of these diseases, many patient organisations are working in pan-European networks to communicate and support each other across the EU 27.*

**Amendment 10**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 1 – introductory part**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| 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 | 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: | orphan drugs***, as well as rehabilitation and habilitation for those living with the disease*** throughout their national territory on the basis of equity and solidarity throughout the EU, and in particular: |

*Justification*

*It is important to note that many sufferers of rare diseases will be forced to live with their ailment for many years. For this reason, research and other activities in this area should also seek to ensure the social integration of those suffering from rare diseases in accordance with Article 26 of the UN Convention on the Rights of Persons with Disabilities.*

**Amendment 11**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 1 – point 1**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (1) elaborate and adopt a comprehensive and integrated strategy, by the end of ***2010***, aimed at guiding and structuring all relevant actions in the field of rare diseases in the form of a national plan for rare diseases; |

*Justification*

*It is important for Member States to elaborate and adopt by the end of 2010 a comprehensive and integrated strategy aimed at guiding and structuring all relevant actions in the field of rare diseases in the form of a national plan for rare diseases;*

**Amendment 12**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 1 – point 3**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (3) define a limited number of priority actions within the national plan for rare diseases, with concrete objectives, clear deadlines***, substantial and clearly designated funding***, management structures and regular reports; |

**Amendment 13**

**Proposal for a recommendation**
**Paragraph 1 – point 3 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(3a) Declare whether they have any specialised centres and compile a catalogue of experts;*** |

**Amendment 14**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 1 – point 5**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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. | (5) include in the national plans provisions designed to ensure equitable access to high quality care, including diagnostics, ***primary preventive measures,*** treatments and orphan drugs, ***together with rehabilitation and habilitation for those living with the disease for the benefit of*** 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 ***following the principles agreed in the High Level Pharmaceutical Forum paper entitled "Improving access to orphan medicines for all affected EU citizens"***. |

*Justification*

*It is important to note that many sufferers of rare diseases will be forced to live with their ailment for many years. For this reason, research and other activities in this area should also seek to ensure the social integration of those suffering from rare diseases in accordance with Article 26 of the UN Convention on the Rights of Persons with Disabilities.*

**Amendment 15**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 1 – point 5 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(5a) encourage efforts to avoid rare diseases which are hereditary, and which will lead finally to the eradication of those rare diseases, through:*** |
| | ***(a) genetic counselling of carrier parents; and*** |
| | ***(b) where appropriate and not contrary to existing national laws and always on a voluntary basis, through pre-implantation selection of healthy embryos.*** |

**Amendment 16**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 1 – point 5 b (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(5b) provide for exceptional measures within the national plans in relation to making available medicinal products which have no marketing authorisation, when there is a real public health need;and, in the absence of appropriate and available therapeutic alternatives in a Member State, and when the risk/benefit balance is presumed to be positive, ensure that patients affected by rare diseases have access to the medicinal products in question.*** |

*Justification*

*The national plans also need to provide for the possibility of providing and reimbursing temporary access to treatments or medicinal products which are in the process of securing marketing authorisation, or which do not have such authorisation, in extremely specific cases where the use of these products could be beneficial to patients suffering from a rare disease. This possibility exists via the compassionate use ('Autorisation Temporaire d'Utilisation' - ATU) procedure.*

**Amendment 17**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 1 – point 5 c (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(5c) establish at the national level multi-stakeholder advisory groups comprising all interested stakeholders to guide governments in the setting up and implementation of national action plans for rare diseases. These should ensure that governments are well-informed and that the decisions taken at national level reflect the views and needs of society.*** |

*Justification*

*In order to minimise the risk of loosing expertise on such a matter of specific nature due to changes in government and administration, there should be standing committees comprising all interested and experienced parties i.e. patients , treating physicians, payers, academia and industry. These parties should offer guidance for the political decision takers and policy makers in the set up and implementation of national action plans.*

**Amendment 18**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 1 – point 5 d (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(5d) encourage treatments for rare diseases to be funded at national level. Where Member States may not wish or may not be able to have Centres of Excellence, this central national fund should be used to ensure that patients can travel to a Centre in another country. However, it is also vital that this separate budget is annually reviewed and adapted on the basis of the knowledge about patients needing treatment in that given year, and about eventual new therapies to be added. This should be done with the input of the multi-stakeholder advisory committees.*** |

*Justification*

*Treating centres or hospitals should be encouraged in diagnosing and treating patients with rare disease . If they have to pay from their own budgets, they might not be able to bare the financial burden. Funding at national level can give the opportunity to the patients to have access to the appropriate medical care.*

**Amendment 19**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 2 – point 1**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (1) implement a European Union common definition of rare diseases as those diseases affecting no more than 5 per 10 000 persons; | (1) implement a European Union common definition of rare diseases as those diseases affecting no more than 5 per 10 000 persons***, as a number for the whole European Union, but it is very important to know the exact distribution for each Member State***; |

**Amendment 20**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 2 – point 4**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (4) support at national or regional level specific disease information networks, registries and databases. | (4) support***, in particular by financial means,*** at ***European,*** national or regional level specific disease information networks, registries and databases***, including regularly-updated information, which is accessible to the public, on the internet*** . |

**Amendment 21**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 3 – point 3**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (3) foster participation of national | (3) foster participation of national |

| | |
|---|---|
| researchers and laboratories in research projects on rare diseases funded at Community level; | researchers and laboratories in research projects on rare diseases funded at Community level ***and make use of the possibilities offered by Regulation (EC) No 141/2000 on orphan medicinal products***; |

**Amendment 22**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 3 – point 3 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(3a) foster knowledge-sharing and cooperation between researchers, laboratories and research projects in the European Union and similar institutions in third countries, to bring global benefits not only to the European Union but also to poorer and developing countries, which are less well placed to provide resources for research into rare diseases;*** |

*Justification*

*The specificities of rare diseases - limited number of patients and scarcity of relevant knowledge and expertise - means that cooperation with countries outside the EU can bring benefits, not just to the EU but also in knowledge sharing with poorer and developing countries.*

**Amendment 23**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 3 – point 4**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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) 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, ***and rehabilitation and habilitation*** |

***programmes for the duration of a rare disease as well as research on diagnostic tests and tools***;

*Justification*

*It is important to note that many sufferers of rare diseases will be forced to live with their ailment for many years. For this reason, research and other activities in this area should also seek to ensure the social integration of those suffering from rare diseases in accordance with Article 26 of the UN Convention on the Rights of Persons with Disabilities.*

**Amendment 24**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 3 – point 4 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(4a) provide adequate and long-term funding, for example through public-private partnerships, so as to support research efforts at national and European level and guarantee the sustainability thereof;*** |

**Amendment 25**

**Proposal for a recommendation**
**Paragraph 4 – point 1**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (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; ***help compile catalogues of rare diseases and rare diseases experts;*** |

**Amendment 26**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 4 – point 3**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (3) organise ***European*** healthcare pathways for ***those*** patients ***suffering from rare diseases*** through the establishment of cooperation with relevant experts within the country or from abroad when necessary; cross-border healthcare, including mobility of patients ***and expertise through data-mobility support***, 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; |

*Justification*

*It is important to organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts within the country or from abroad when necessary.*

**Amendment 27**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 4 – point 5**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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) 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***, by involving patients in the activitiesof these centres***; |

**Amendment 28**

**Proposal for a recommendation**
**Paragraph 4 – point 5 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(5a) encourage, possibly with European Union funding or co-funding, centres and hospitals of expertise to create specific training for professionals in certain rare diseases and allow them to acquire relevant expertise;*** |

*Justification*

*It is essential to train more professionals in the field of rare diseases in order to combat them efficiently.*

**Amendment 29**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 5 – point 1 – subpoint b**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (b) European guidelines on population screening and diagnostic tests; | (b) European guidelines on population screening and diagnostic tests***, including genetic tests like heterozygote testing and polar body diagnosis, ensuring high-quality testing and appropriate genetic counselling while respecting ethical diversity in the Member States***; |

**Amendment 30**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 5 – point 1 – subpoint c**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (c) ***sharing*** Member *States'* 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; | (c) ***establishing*** Member *States'* assessment reports on the ***clinical*** added value of orphan drugs at EU level ***within the EMEA where the relevant European knowledge and expertise is gathered***, in order to minimise delays for access to |

orphan drugs for rare disease patients;

**Amendment 31**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 5 – point 1 – subpoint c a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(ca) structural support for investment in the Orphanet database to ensure ease of access to information concerning rare diseases;*** |

**Amendment 32**

**Proposal for a recommendation**
**Recommendations to Member States – Paragraph 6**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| 6. Empowerment of patient organisations | 6. Empowerment of ***independent*** 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; | (1) take action to ensure that patients and ***independent*** 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; | (2) support the activities performed by ***independent*** patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking, outreach to very isolated patients; |
| | ***(2a) ensure that funding for patient organisations which is not directly linked to single pharmaceutical companies is provided;*** |
| | ***(2b) facilitate patient access to information existing at European level*** |

| | |
|---|---|
| | ***concerning medicines, treatments or treatment centres in the Member States or third countries providing medical care specifically suited to their illnesses;*** |
| (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). | (3) include in the national plans for rare diseases provisions on the support to and the consultation of ***independent*** patient organisations as referred to in paragraphs (1) and (2)***; ensure that national plans provide for the identification of national or regional centres of expertise and for the compilation of catalogues of experts on rare diseases;*** |

*Justification*

*It is important to facilitate patient access to information existing at European level concerning medicines, treatments or treatment centres in the Member States or third countries providing medical care specifically suited to their illnesses.*

**Amendment 33**

**Proposal for a recommendation**
**Recommendations to the Commission – paragraph -1 (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(-1) To support, in a sustainable way, "Orphanet", a European website and "one-stop shop" providing the following information:*** |
| | ***(a) on the existence of specific research into rare diseases, the findings thereof and their availability to patients,*** |
| | ***(b) on available medicines for each rare disease,*** |
| | ***(c) on the treatment existing in each Member State for each rare disease,*** |
| | ***(d) on existing specialist medical centres in Member States or third countries for each rare disease.*** |

**Amendment 34**

**Proposal for a recommendation**
**Recommendations to the Commission – Paragraph 1**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| 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***. | 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 ***2012, the year in which it will propose the implementing actions covering inter alia: a) the budgetary measures necessary for the Community Programme on Rare Diseases to be effective; b) the creation of relevant networks of centres of expertise; c) the collection of epidemiological data on rare diseases; d) the mobility of experts and professionals; e) the mobility of patients; and f) consideration of the need for other actions to improve the lives of patients affected by rare diseases and those of their families.*** |

## EXPLANATORY STATEMENT

### General introduction and appraisal of the Proposal for a Council Recommendation

This Council Recommendation is welcome because a concerted action in the field of rare diseases at the European Union level and at the national level is an **absolute necessity**. This is because although the incidence for each rare disease is very low, millions of people are affected across the European Union because these diseases are measured in thousands.

**However** this Proposal at hand is really insufficient as in its current state and it is not possible to produce a viable programme out of it. This is because it does not describe, at least in general terms, **the necessary funding** from EU and the co funding by EU and Member States or other organisations. Thus cannot effectively promote certain essential aspects on rare diseases i.e. **creation of networks of centres of expertise, cataloguing of diseases, special research required etc.**

The text asks for an implementation proposal from the Commission five years after the adoption, which is a long period during which nothing can be practically done as there is no funding provided.

Therefore as a Rapporteur, I propose that the Commission be asked to **provide the implementation proposal** at the latest by the end of 2012, since by this date the required data from Member States on expert centres and expertise on rare diseases will be at hand (2011).

In this implementation specific mention should be made for funding/co funding etc. in the areas of:
a.) the collection of epidemiological data and compiling a catalogue of rare diseases, as this is necessary in order to have a clear picture of the field of these diseases in the EU;
b.) the formation of relevant networks;
c.) the creation of expert centres in Member States which lack such centres;
d.) the creation of special training courses in the existing centres for professionals to acquire expertise;
e.) the mobilisation of experts and professionals in order to create the necessary conditions for advancing existing knowledge;
f.) research on diagnostic tools and tests on rare diseases and especially on genetic ones.

We should considering this proposal for a Council Recommendation as a roadmap to create helpful conditions in the field of rare diseases, and we should understand that it is of a general nature but I would like to stress once more that in order to be efficiently and successfully applied, the Proposal needs to be more precise and definite in the calendar (years) of implementation.

**Important aspects to be mentioned**
**in the Commission's proposal for implementation**

1. As it is evident from the Council Recommendation, the field of rare diseases in the European Union is **not well defined** :
-neither as a registry cataloguing the diseases one by one,
- nor as far as the accuracy of the statistical data is concerned.
This is evident from the great **spread** in the number of patients (27 to 36 million people) and the number of rare diseases (5 000 to 10 000 rare diseases).

2. It is also important to note that we should approach the subject with **flexibility** because a definition of rare disease as affecting less than 5 persons per 10 000 is **too restrictive**, as you may have numbers slightly above this ratio and be confronted with the dilemma of not considering as a rare disease one if the ratio is for example 5,1 . Also, a rare disease may have a different distribution from one Member State to another.

3. Due to the great number of affected persons but the low incidence for each disease, a concerted action across the European Union level is of absolute necessity.
For this reason, I believe that it is appropriate that the final implementation proposal should try to **compile the specialized centres and hospitals** for some of these diseases, as well as the **expertise** existing in each country and **connect them in a network.**

4. It is also obvious that to combat rare diseases, one need to regulate **mobility of patients** because there is not all the expertise in each Member State.
Therefore as long as there are no expertise and centres which can take care of the patients, this should be corrected by their mobility. Here the issue of funding should be left for consideration to the Member States.

5. **Mobility of professionals** is another very important aspect, i.e.:
a.) mobility of experts across the European Union in order to help the creation of new centres in other Member States
b.) mobility of professionals in order to acquire expertise on certain rare diseases in existing centres across the European Union. This action requires the creation of training courses and clinical practice in order to train more experts on these diseases.
Therefore the mission of these centres will not only be to combat rare diseases but to create the necessary environment for transmission of knowledge.

6. Finally because many of the rare diseases are of hereditary nature, **research and innovation** is absolutely essential in order to enhance the **diagnostic ability through genetic tests**.

7. There are cases for which genetic tests exist and can be applied to **pre-implantation procedures** in order to avoid these diseases in embryos and consequently eradicate some of these diseases. This should be done in concerted action with genetic counselling and networks of information. This is in practice in some Member States for certain diseases, and it should be considered and taken into account as the application of these actions will eradicate in the end some of these diseases.

10.3.2009

# OPINION OF THE COMMITTEE ON INDUSTRY, RESEARCH AND ENERGY

for the Committee on the Environment, Public Health and Food Safety

on the proposal for a Council recommendation on a European action in the field of rare diseases
(COM(2008)0726 – C6-0455/2008 – 2008/0218(CNS))

Rapporteur: Françoise Grossetête

## SHORT JUSTIFICATION

Rare diseases affect some 30 million Europeans. Each disease affects a small number of individuals, sometimes fewer than a dozen in any given country. The average is one in 2000. Over 7000 rare diseases are known to exist today, and two new pathologies are described every week in medical publications. Eighty per cent of rare diseases are genetic in origin, and 65% are paediatric.

It is therefore essential that rare diseases be made a priority at European level, in order to deal properly with the needs of several million citizens suffering from misdiagnosis and thus receiving no treatment to cure their diseases.

Unlike other public health or research issues, what is at stake here is not merely making up for lost time with regard to a neglected problem, but devising tools or permanent arrangements which will enable the specific features of these diseases to be taken into due account for all time to come.

Their rarity spawns specific challenges in scientific, economic and organisational terms, and these challenges cannot be tackled with the tools that are generally applied. What is called for is a global, consistent and sustainable approach, which can only be achieved by simultaneously mobilising all those concerned: politicians, institutions, patients' associations, health professionals, sufferers' associations, carers, researchers and industry.

The tiny numbers involved mean that sufferers cannot be dealt with at local or regional level, while the illnesses are far too numerous to be taught to health professionals; the result is that expert knowledge is as rare as the diseases. Since no EU Member State is capable of combating rare diseases on its own, access to information, diagnostics, carers and research needs to be organised into coherent and coordinated networks - usable by all those involved - of methodologies and shared approaches at national and European level. The Europe-wide approach is therefore essential when it comes to organising the fight against rare diseases and

to mutualising and implementing resources and wherewithal.

On 16 December 1999, the European Parliament and the Council of Ministers adopted Regulation (EC) No 141/2000 on orphan medicinal products, which urges the pharmaceutical and biotechnology industry to develop and market orphan medicinal products (tax incentives, protocol assistance, 10 years' exclusivity, etc.). A Committee for orphan medicinal products (COMP) was set up within the European Agency for the Evaluation of Medicinal Products (EMEA), with the remit of examining applications for designation and assisting the Commission in discussions concerning orphan medicinal products.

In October 2008, 569 medicinal products had been granted 'orphan' designation in Europe, whereof 54 have already received Community marketing authorisation; almost 3 million patients are concerned.

The adoption of the European Regulation on orphan medicinal products created a favourable framework for companies involved in developing these products, in the shape of the aforementioned tax incentives, commercial exclusivity, etc., and has proved a boost to work on developing orphan medicinal products.

The pharmaceutical industry is disinclined to develop such products under normal market conditions: certain illnesses are so rare that the costs of development and marketing the product would never be recouped by potential sales.

There are many examples of how progress made as a result of work on rare diseases has greatly benefited less-rare diseases and will continue to do so. The range of therapeutic approaches is multiplying: genetic and cellular therapies, new molecules, 'à la carte' treatments, etc. The innovative therapies which are developed for rare diseases are a foretaste of the ever more personalised medicine of tomorrow.

In economic terms, the explosive growth of opportunities for clinical development in the field of rare diseases opens up fresh prospects, and here too, all available means to exploit these opportunities need to be coordinated at European level. Taken individually, these diseases do not constitute viable markets. Despite the existence of European measures encouraging the development of orphan medicinal products, the pharmaceutical industry is even less willing to take risks in the initial stages of developing treatments (from preclinical to phase II) than it is with more common diseases.

For the industry, the relationship between financial risks and profit prospects becomes attractive only late on in the process of developing new treatments. Between the proof of concept and the investment of the substantial private funding required before the marketing authorisation stage is reached, rare diseases have to go through a 'death valley' much longer than that faced by commoner diseases, with their much greater market potential.

It is therefore necessary, on the one hand, to construct tailor-made development tools and to optimise the development cycle for treatments so as to minimise costs, and, on the other, to invent new tools for sharing risks between private and public funding, which will enable these stages to be funded. At European level, developing networks and stepping up collaboration are unquestionably major levers in attaining this end. If measures are not taken quickly, there is a real risk that certain treatments will never see the light of day and that the only treatments

which will be marketed will come from the other side of the Atlantic, at prohibitive prices.

The proposal for a Council recommendation on a European action in the field of rare diseases has a particularly important role to play with regard to the drafting and adoption of national rare diseases strategies and plans. In all the Member States these strategies and plans should have the same scope; they should be coordinated between the various Member States, and be consistently and effectively linked into Community policies.

## AMENDMENTS

The Committee on Industry, Research and Energy calls on the Committee on the Environment, Public Health and Food Safety, as the committee responsible, to incorporate the following amendments in its report:

**Amendment 1**

**Proposal for a recommendation**
**Recital 5**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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. | (5) Because of their low prevalence and their specificity, rare diseases call for a global approach based on special and combined efforts***, including in partnership with third countries such as the United States,*** to prevent significant morbidity or avoidable premature mortality, and to improve quality of life and socio-economic potential of affected persons ***in developed and developing countries.*** |

*Justification*

*The specificities of rare diseases - limited number of patients and scarcity of relevant knowledge and expertise - means that cooperation with countries outside the EU can bring benefits, not just to the EU but also in knowledge sharing with poorer and developing countries.*

**Amendment 2**

**Proposal for a recommendation**
**Recital 14 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(14a) On 12 October 2008 the Pharmaceutical Forum adopted its final report, which proposes guidelines to enable Member States, stakeholders and the Commission to step up their efforts to guarantee easier and swifter access to orphan medicinal products within the European Union.*** |

*Justification*

*Following the conclusions of the Pharmaceutical Forum, the Member States have given a political undertaking to eliminate bottlenecks in respect of access to orphan medicinal products via the Community approval procedure. The guidelines set out in the Pharmaceutical Forum's final report need to be given specific mention in the proposal for a recommendation.*

**Amendment 3**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 1 – point 1**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (1) elaborate and adopt a comprehensive and integrated strategy, by the end of ***2010***, aimed at guiding and structuring all relevant actions in the field of rare diseases in the form of a national plan for rare diseases; |

*Justification*

*It is important for Member States to elaborate and adopt by the end of 2010 a comprehensive and integrated strategy aimed at guiding and structuring all relevant actions in the field of rare diseases in the form of a national plan for rare diseases;*

**Amendment 4**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 1 – point 3**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (3) define a limited number of priority actions within the national plan for rare diseases, with concrete objectives, clear deadlines, ***substantial and clearly identified funding,*** management structures and regular reports; |

**Amendment 5**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 1 – point 5 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(5a) provide for exceptional measures within the national plans when it comes to making available medicinal products which have no marketing authorisation, when there is a real public health need. In the absence of appropriate and available therapeutic alternatives in a Member State, and when the risk/benefit balance is presumed to be positive, patients affected by rare diseases shall have access to the medicinal products in question.*** |

*Justification*

*The national plans also need to provide for the possibility of providing and reimbursing temporary access to treatments or medicinal products which are in the process of securing marketing authorisation, or which do not have such authorisation, in extremely specific cases where the use of these products could be beneficial to patients suffering from a rare disease. This possibility exists via the compassionate use ('Autorisation Temporaire d'Utilisation' - ATU) procedure.*

**Amendment 6**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 2 – point 4**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (4) support at national or regional level specific disease information networks, registries and databases. | (4) support***, in particular by financial means,*** at ***European,*** national or regional level specific disease information networks, registries and databases. |

**Amendment 7**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 3 – point 3 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(3a) put in place adequate, long-term funding, by means , for example, of public/private partnerships, to support research at national and European level, so as to guarantee its viability;*** |

**Amendment 8**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 3 – point 3 b (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(3b) foster knowledge-sharing and cooperation between researchers, laboratories and research projects in the European Union and similar institutions in third countries, to bring global benefits not only to the European Union but also to poorer and developing countries, which are less well placed to provide resources for research into rare diseases;*** |

*Justification*

*The specificities of rare diseases - limited number of patients and scarcity of relevant knowledge and expertise - means that cooperation with countries outside the EU can bring*

*benefits, not just to the EU but also in knowledge sharing with poorer and developing countries.*

**Amendment 9**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 4 – point 3**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (3) organise ***European*** healthcare pathways for ***those*** patients ***suffering from rare diseases*** 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; |

*Justification*

*It is important to organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts within the country or from abroad when necessary.*

**Amendment 10**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 4 – point 5**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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) 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 ***and involving the patients in the activities, management and evaluation of the centres***. |

**Amendment 11**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 5 – point 1 – point c**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| (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; | (c) sharing ***Member States'*** assessment reports on the ***clinical*** added value of orphan drugs at EU level ***within the European Medicines Agency, which brings together European expertise in this field***, in order to minimise delays for access to orphan drugs for rare disease patients; |

*Justification*

*The EMEA is the most appropriate platform for reducing the waiting times and centralising the evaluation reports.*

**Amendment 12**

**Proposal for a recommendation**
**Recommendations to Member States – paragraph 6 – point 2 a (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(2a) facilitate patient access to information existing at European level concerning medicines, treatments or treatment centres in the Member States or third countries providing medical care specifically suited to their illnesses;*** |

*Justification*

*It is important to facilitate patient access to information existing at European level concerning medicines, treatments or treatment centres in the Member States or third countries providing medical care specifically suited to their illnesses.*

**Amendment 13**

**Proposal for a recommendation**
**Recommendations to the Commission – paragraph -1 (new)**

| *Text proposed by the Commission* | *Amendment* |
|---|---|
| | ***(-1) To support, in a sustainable way, "Orphanet", a European website and "one-stop shop" providing the following information:*** |
| | ***(a) on the existence of specific research into rare diseases, the findings thereof and their availability to patients,*** |
| | ***(b) on available medicines for each rare disease,*** |
| | ***(c) on the treatment existing in each Member State for each rare disease,*** |
| | ***(d) on existing specialist medical centres in Member States or third countries for each rare disease.*** |

## PROCEDURE

| | |
|---|---|
| **Title** | European action in the field of rare diseases |
| **References** | COM(2008)0726 – C6-0455/2008 – 2008/0218(CNS) |
| **Committee responsible** | ENVI |
| **Opinion by**<br>Date announced in plenary | ITRE<br>4.12.2008 |
| **Rapporteur**<br>Date appointed | Françoise Grossetête<br>2.12.2008 |
| **Discussed in committee** | 11.2.2009 |
| **Date adopted** | 9.3.2009 |
| **Result of final vote** | +: 43<br>–: 0<br>0: 0 |
| **Members present for the final vote** | Jan Březina, Giles Chichester, Dragoş Florin David, Pilar del Castillo Vera, Den Dover, Lena Ek, Norbert Glante, Umberto Guidoni, Fiona Hall, David Hammerstein, Rebecca Harms, Erna Hennicot-Schoepges, Mary Honeyball, Romana Jordan Cizelj, Werner Langen, Anne Laperrouze, Pia Elda Locatelli, Eluned Morgan, Reino Paasilinna, Atanas Paparizov, Francisca Pleguezuelos Aguilar, Anni Podimata, Herbert Reul, Teresa Riera Madurell, Paul Rübig, Andres Tarand, Catherine Trautmann, Claude Turmes, Nikolaos Vakalis, Adina-Ioana Vălean, Dominique Vlasto |
| **Substitute(s) present for the final vote** | Alexander Alvaro, Pilar Ayuso, Ivo Belet, Françoise Grossetête, Marie-Noëlle Lienemann, Erika Mann, Vittorio Prodi, Esko Seppänen, Vladimir Urutchev, Lambert van Nistelrooij |
| **Substitute(s) under Rule 178(2) present for the final vote** | Elisabetta Gardini, Ulrike Rodust |

## PROCEDURE

| | |
|---|---|
| **Title** | European action in the field of rare diseases |
| **References** | COM(2008)0726 – C6-0455/2008 – 2008/0218(CNS) |
| **Date of consulting Parliament** | 13.11.2008 |
| **Committee responsible**<br>Date announced in plenary | ENVI<br>4.12.2008 |
| **Committee(s) asked for opinion(s)**<br>Date announced in plenary | ITRE<br>4.12.2008 |
| **Rapporteur(s)**<br>Date appointed | Antonios Trakatellis<br>4.12.2008 |
| **Discussed in committee** | 10.2.2009 |
| **Date adopted** | 31.3.2009 |
| **Result of final vote** | +: 42<br>–: 7<br>0: 0 |
| **Members present for the final vote** | Adamos Adamou, Georgs Andrejevs, Margrete Auken, Liam Aylward, Pilar Ayuso, Irena Belohorská, Maria Berger, Johannes Blokland, John Bowis, Frieda Brepoels, Martin Callanan, Magor Imre Csibi, Chris Davies, Avril Doyle, Edite Estrela, Jill Evans, Anne Ferreira, Karl-Heinz Florenz, Matthias Groote, Françoise Grossetête, Gyula Hegyi, Dan Jørgensen, Christa Klaß, Eija-Riitta Korhola, Urszula Krupa, Peter Liese, Jules Maaten, Marios Matsakis, Linda McAvan, Péter Olajos, Miroslav Ouzký, Vittorio Prodi, Dagmar Roth-Behrendt, Guido Sacconi, María Sornosa Martínez, Antonios Trakatellis, Thomas Ulmer, Åsa Westlund, Glenis Willmott |
| **Substitute(s) present for the final vote** | Nicodim Bulzesc, Christofer Fjellner, Milan Gaľa, Johannes Lebech, Miroslav Mikolášik, Hartmut Nassauer, Alojz Peterle, Bart Staes, Robert Sturdy |
| **Substitute(s) under Rule 178(2) present for the final vote** | Søren Bo Søndergaard |