Basic information

2008/0218(CNS) CNS - Consultation procedure Recommendation	Procedure completed
Rare diseases: European action **Subject** 4.20.01 Medicine, diseases	

Key players

European Parliament

Committee responsible	Rapporteur	Appointed
ENVI Environment, Public Health and Food Safety	TRAKATELLIS Antonios (PPE-DE)	04/12/2008

Committee for opinion	Rapporteur for opinion	Appointed
ITRE Industry, Research and Energy	GROSSETÊTE Françoise (PPE-DE)	02/12/2008

Council of the European Union

Council configuration	Meetings	Date
Employment, Social Policy, Health and Consumer Affairs	2947	2009-06-08
Employment, Social Policy, Health and Consumer Affairs	2916	2008-12-16

European Commission

Commission DG	Commissioner
Health and Food Safety	VASSILIOU Androulla

Key events

Date	Event	Reference	Summary
11/11/2008	Legislative proposal published	COM(2008)0726	Summary
04/12/2008	Committee referral announced in Parliament		
16/12/2008	Debate in Council		Summary
31/03/2009	Vote in committee		Summary
03/04/2009	Committee report tabled for plenary, 1st reading/single reading	A6-0231/2009	
23/04/2009	Decision by Parliament	T6-0288/2009	Summary

23/04/2009	Results of vote in Parliament		
23/04/2009	Debate in Parliament		
08/06/2009	Act adopted by Council after consultation of Parliament		
08/06/2009	End of procedure in Parliament		
03/07/2009	Final act published in Official Journal		

Technical information

Procedure reference	2008/0218(CNS)
Procedure type	CNS - Consultation procedure
Procedure subtype	Legislation
Legislative instrument	Recommendation
Stage reached in procedure	Procedure completed
Committee dossier	ENVI/6/69817

Documentation gateway

European Parliament

Document type	Committee	Reference	Date	Summary
Committee draft report		PE420.052	04/02/2009	
Amendments tabled in committee		PE421.262	04/03/2009	
Committee opinion	ITRE	PE418.380	10/03/2009	
Committee report tabled for plenary, 1st reading/single reading		A6-0231/2009	03/04/2009	
Text adopted by Parliament, 1st reading/single reading		T6-0288/2009	23/04/2009	Summary

European Commission

Document type	Reference	Date	Summary
Document attached to the procedure	COM(2008)0679	11/11/2008	Summary
Legislative proposal	COM(2008)0726	11/11/2008	Summary
Document attached to the procedure	SEC(2008)2712	11/11/2008	
Document attached to the procedure	SEC(2008)2713	11/11/2008	
Commission response to text adopted in plenary	SP(2009)3507	25/06/2009	
Follow-up document	C(2009)9181	30/11/2009	
	COM(2014)0548		

Follow-up document		05/09/2014	Summary

Other institutions and bodies

Institution/body	Document type	Reference	Date	Summary
EESC	Economic and Social Committee: opinion, report	CES0346/2009	25/02/2009	

Additional information

Source	Document	Date
European Commission	EUR-Lex	

Final act

EP/Council Recommendation 2009/0703(02)
OJ C 151 03.07.2009, p. 0007 Summary

Rare diseases: European action

2008/0218(CNS) - 05/09/2014 - Follow-up document

The Commission adopted a report on the implementation report on the Commission Communication on Rare Diseases: Europe's challenges and Council Recommendation on an action in the field of rare diseases.

Rare diseases affect between **27 and 36 million people in the European Union** and are a key health policy priority for the EU.

With a view to enhancing the European dimension and the co-operation between Member States in this area, to achieve this goal in 2008, the Commission adopted a Communication on Rare Diseases, setting out an overall strategy to support Member States in diagnosing, treating and caring for EU citizens with rare diseases. Alongside the Communication, a Council Recommendation on action in the field of rare diseases was adopted a few months later, calling on Member States to put national strategies in place.

Directive 2011/24/EUon the application of patients' rights in cross-border healthcare also addresses rare diseases as does the Orphan Medicinal Products Regulation (Regulation (EC) No 141/2000. The **Rare Diseases Task Force** was also set up within the Commission.

Member States committed to use for the purposes of Community-level policy work a common definition of a **rare disease** as a **disease affecting no more than 5 per 10 000 persons**.

This report presents an **overview of the implementation** of the rare diseases strategy to date and takes stock of achievements and lessons learnt. It seeks to draw conclusions on the extent to which the measures foreseen in the Commission Communication and the Council Recommendation have been put in place and the need for further action to improve the lives of

patients affected by rare diseases and their families.

Plans and strategies: in order to support Members States in the process of developing national plans and strategies, the European Commission co-financed the **EUROPLAN** project from the EU Health Programme.

The project, running from April 2008 to March 2011, involved representatives of national health authorities of **21 Member States**, and brought together 57 associated and collaborating partners from 34 countries. One of the deliverables was a report on **indicators for monitoring** the implementation and evaluating the impact of a National Plan or Strategy for rare diseases.

This has supported a significant number of Member States to put in place dedicated plans to address rare diseases: 16 Member States now have rare diseases plans (as compared to only 4 in 2008) and seven further countries are well advanced in the development of their plans/strategies.

The EU has funded close to **120 collaborative research projects** relevant to rare diseases through its Seventh Framework Programme for Innovation and Technological Development (FP7). With a total budget of over EUR 620 million, these projects span across several disease areas such as neurology, immunology, cancer, pneumology, and dermatology.

In collaboration with its national and international partners, the European Commission spearheaded the launch of the **International Rare Diseases Research Consortium (IRDiRC)** in early 2011. Its key objective is to deliver, by 2020, 200 new therapies for rare diseases and the means to diagnose most of them by stimulating, better coordinating, and maximising output of rare disease research on a global level. As of January 2014 there were 588 rare diseases registries distributed as follows: 62 European, 35 global, 423 national, 65 regional and 3 undefined. Most of the registries are established in public and academic institutions.

It should be noted that the European Commission's Joint Research Centre is currently developing a **European Platform on Rare Diseases Registration.** The main objectives for this platform are to provide a central access point for information on **rare diseases patients' registries.**

Future proposals: despite such encouraging progress, there is **still a long way to go**. There are still Member States who do not yet have a national plan or strategy. This is why action on rare diseases features prominently in the new Health Programme and the new EU Research and Innovation Programme Horizon 2020.

The following actions are envisaged to continue supporting Member States:

- maintain the EU's coordinative role in the development of the EU policy on rare diseases and to support Member States in their activities on the national level;
- continue to support the development of high quality National Rare Diseases Plans/Strategies in the European Union;
- provide continued support for the International Rare Disease Research Consortium and initiatives developed under its umbrella;
- continue to ensure proper codification of rare diseases;
- work further to decrease inequalities between patients with rare diseases and patients suffering from more common disorders and to support initiatives promoting equal access to diagnosis and treatment;
- continue to promote patients empowerment in all aspects of rare diseases policy development;
- continue activities increasing public awareness about rare diseases and EU activity in this field;
- support the development of the tools facilitating cooperation and interoperability of the European Reference Networks for rare diseases;
- stimulate development and use of eHealth solutions in the area of rare diseases;
- implement and continue support for the European Platform on rare diseases registration;
- continue playing a global role in the rare diseases initiative and collaborating with important international stakeholders.

Rare diseases: European action

2008/0218(CNS) - 11/11/2008 - Document attached to the procedure

Rare diseases are considered as such in the EU when they affect not more than 5 per 10 000 persons. This nevertheless means than between 5 000 and 8 000 different rare diseases affect or will affect an estimated 29 million people in the EU. The lack of specific health policies for rare diseases and the scarcity of the expertise, translate into delayed diagnosis and difficult access to care. This results in additional physical, psychological and intellectual impairments, inadequate or even harmful treatments and loss of confidence in the health care system, despite the fact that some rare diseases are compatible with a normal life if diagnosed on time and properly managed. Misdiagnosis and non-diagnosis are the main hurdles to improving life-quality for thousands of rare disease patients.

The national healthcare services for diagnosis, treatment and rehabilitation of people with rare diseases differ significantly depending on their availability and quality.

Depending on the Member State and/or region where they live, EU citizens have unequal access to expert services and available care options.

Objectives: the Communication sets out an overall Community strategy for support to Member States in ensuring effective and efficient recognition, prevention, diagnosis, treatment, care, and research for rare diseases in Europe. The Communication orients the operational actions in three main fields of work:

- improving Recognition and Visibility on Rare Diseases: the key to improving overall strategies for rare diseases is to ensure that they are recognised, so that all the other linked actions can follow appropriately. The existing definition of rare diseases in the EU was adopted by the Community action programme on rare diseases 1999-2003 as those diseases presenting a prevalence not more than 5 per 10 000 persons in the EU. The same definition is set out in Regulation (EC) 141/2000 and, accordingly used by the European Commission for the designation of orphan drugs. The EU will maintain the current definition. The Commission aims to put in place a thorough coding and classification system at European level, which will provide the framework for better sharing knowledge and understanding rare diseases as a scientific and public health issue across the EU;

- supporting policies on rare diseases in Member States: the Commission proposes that Member States base themselves on common approach for addressing rare diseases, based on existing best practice, through the adoption of a Council Recommendation. (For the Commission's proposal for

a Council Recommendation, please see summary of the same date.)

- developing European cooperation on rare diseases: Community action will help Member States to achieve efficiency in bringing together and organise the scarce resources in the area of rare diseases, and can help patients and professionals to collaborate across Member States in order to

share and coordinate expertise and information. The establishment of an EU dynamic inventory of rare diseases will contribute to tackle some of the main causes of neglecting the issue of rare diseases including the ignorance of which diseases are rare. The Commission will ensure that this information continues to be available at European level, building in particular on the Orphanetdatabase;

- disease information networks: priorities for action regarding the existing (or future) specific disease information networks are, inter alia, to guarantee the exchange of information via existing European information networks; to develop strategies and mechanisms for exchanging information between stakeholders; and to develop comparable epidemiological data at EU level.

Operational actions to develop European cooperation and improve access to high quality health care for rare diseases: the Communication proposes that this will be done, in particular through development of national/regional centres of expertise and establishing EU reference networks. It also discusses access to specialised social services, and access to Orphan Drugs. With regard to the latter, the Commission will set up a working party to exchange knowledge between Member States and European authorities on the scientific assessment of the clinical added value of orphan medicines. The paper goes on to describe proposed compassionate use programmes, incentives for Orphan Drug development, e-Health, the evaluation of current population screening (including neonatal screening) strategies for rare diseases and of potential new ones, quality management of diagnostic laboratories, and primary prevention. It discusses registries and databases, noting that a key issue will also be to ensure the long-term sustainability of data collection systems. With regard to research and development, the communication notes that the development of therapies faces three hurdles: the lack of understanding of underlying patho-physiological mechanisms, the lack of support of early phases of clinical development and the lack of opportunity/cost perception from the pharmaceutical industry. The paper outlines proposed action in this area.

Rare diseases: European action

2008/0218(CNS) - 16/12/2008

The Council held a first exchange of views on the proposal for a Council Recommendation on a European action in the field of rare diseases.

All the delegations that took the floor congratulated the Commission on this initiative. Several Ministers mentioned their national action plans and asked that the resources and structures provided be taken into account when the Recommendation is adopted. The Ministers agreed that the particular nature of rare diseases made this an area in which Europe could bring substantial added value.

Recall that the European Union initiated a policy in this area with the adoption in 1999 of the Regulation on orphan medicinal products. In tandem with its Communication "Rare Diseases: Europe's challenges" (COM(2008)0679), which sets out an overall Community strategy, the Commission proposes that, in the Recommendation, the Council should adopt a common approach to combating rare diseases that is based on existing best practice.

Rare diseases: European action

2008/0218(CNS) - 08/06/2009 - Final act

PURPOSE: to establish national plans for rare diseases and ensure adequate definition of and research into such diseases.

ACT: Council Recommendation on an action in the field of rare diseases.

BACKGROUND: 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 particular, the total number of people affected by rare diseases in the EU is between 27 and 36 million. Because of their low prevalence, their specificity and the high total number of people affected, rare diseases call for a global approach based on special and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve the quality of life and socioeconomic potential of affected persons.

The Commission, in its White Paper 'Together for Health: A Strategic Approach for the EU 2008-2013' of 23 October 2007, which develops the EU Health Strategy, identified rare diseases as a priority for action.

CONTENT: the Council makes a number of recommendations to Member States, notably that they:

- establish and implement **plans or strategies for rare diseases** at the appropriate level, in order to aim to ensure that patients with rare diseases have access to high-quality care, and in particular: (i) elaborate and adopt a plan or strategy by the end of 2013, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems; (ii) take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies; (iii) define a limited number of priority actions; (iv) take note of the development of guidelines and recommendations for the elaboration of national action for rare diseases by relevant authorities at national level;
- use a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons;
- aim to ensure that rare diseases are adequately coded and traceable in all health information systems;
- contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network and other existing networks;
- identify **ongoing researchon rare diseases** and research resources in the national and Community frameworks;
- identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them;
- foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels;
- facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases;
- **identify appropriate centres of expertise** throughout their national territory by the end of 2013, and consider supporting their creation;
- foster the participation of centres of expertise in **European reference networks**;

- organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts;
- support the use of information and communication technologies, such as telemedicine, where it is necessary to ensure distant access to the specific healthcare needed;
- include in their plans or strategies the necessary conditions for the diffusion and mobility of expertise and knowledge;
- encourage centres of expertise to be based on a multidisciplinary approach to care;
- **gather national expertise on rare diseases** and support the pooling of that expertise with European counterparts in order to support: (i) the sharing of best practices on diagnostic tools and medical care; (ii) adequate education and training for all health professionals; (iii) the development of medical training in fields relevant to the diagnosis and management of rare diseases; (iv) the development of European guidelines on diagnostic tests or population screening; (v) the sharing Member States′ assessment reports on the therapeutic or clinical added value of orphan drugs at Community level;
- **consult patients** and patients′ representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases;
- promote the activities performed by patient organisations, such as awareness-raising, capacity-building and training;
- together with the Commission, aim to ensure, through appropriate funding and cooperation mechanisms, the long-term **sustainability** of infrastructures developed in the field of information, research and healthcare for rare diseases.

Lastly, the Council invites the Commission to produce, by the end of 2013 and in order to allow proposals in any possible future programme of Community action in the field of health, an implementation report on this recommendation, which should consider the extent to which the proposed measures are working effectively and the need for further action to improve the lives of patients affected by rare diseases and those of their families. The Council also asks to be informed of the follow-up to the Commission Communication on rare diseases on a regular basis.

Rare diseases: European action

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

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

PROPOSED ACT: Council Recommendation.

CONTENT: it is estimated that between 5 000 and 8 000 distinct rare diseases exist today, affecting between 6% and 8% of the population in the course of their lives i.e between 27 and 36 million people in the EU. Most of them suffer from less frequently-occurring diseases affecting one in 100 000 people or less. Because of their low prevalence and their specificity, rare diseases call for a global approach based on special and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve quality of life and socio-economic potential of affected persons.

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

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

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

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

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

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

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

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

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

Rare diseases: European action

2008/0218(CNS) - 23/04/2009 - Text adopted by Parliament, 1st reading/single reading

The European Parliament adopted by 383 votes to 116, with 15 abstentions, a legislative resolution amending, under the consultation procedure, the proposal for a Council recommendation on a European action in the field of rare diseases. Parliament wants financial support at European level for this Recommendation. It also wants Member States to adopt a comprehensive and integrated strategy, by the end of 2010 (rather than 2011).

Recommendations to Member States are amended as follows:

- national plans must also ensure the **rehabilitation** for those living with the disease;
- priority actions within the national plan must contain substantial and clearly designated funding;
- Member States must declare whether they have any specialised centres and compile a catalogue of experts.

New recommendations to Member States are inserted in the text. Member States should:

- declare whether they have any specialised centres and compile a catalogue of experts;
- encourage efforts to avoid rare diseases which are **hereditary**, and which will lead finally to the eradication of those rare diseases, through: genetic counselling of carrier parents; and where appropriate and not contrary to existing national laws and always on a voluntary basis, through pre-implantation selection of healthy embryos;
- 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;
- establish at the national level multi- stakeholder advisory groups to guide governments in the setting up and implementation of national action plans for rare diseases;
- 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 funding 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;
- 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;
- foster knowledge-sharing between researchers, laboratories and research projects in the EU and similar institutions in third countries, to bring global benefits not only to the EU but also to poorer countries;
- make use of the possibilities offered by Regulation (EC) No 141/2000 on orphan medicinal products;
- 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;
- encourage, possibly with **EU 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;
- European guidelines on population screening and diagnostic tests should include 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;
- there must be structural support for investment in the **Orphanet database**;
- funding for patient organisations which is not directly linked to single pharmaceutical companies should be provided. Member States should facilitate patient access to information existing at European level concerning medicines, or treatment centres in Member States or third countries providing medical care specifically suited to their illnesses;
- Member States should 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.

A **new recommendation to the Commission** is inserted in the proposal. The Commission should support, in a sustainable way, "Orphanet", a European website and "one-stop shop" providing the following information: on the existence of specific research into rare diseases, the findings thereof and their availability to patients; on available medicines for each rare disease; on the treatment existing in each Member State for each rare disease; on existing specialist medical centres in Member States or third countries for each rare disease.

Lastly, the Parliament states that the **implementation report** must be produced before the end of 2012. Accordingly. the Commission must produce, by 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.