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**REPORT FROM THE COMMISSION TO THE EUROPEAN PARLIAMENT, THE  
COUNCIL, THE EUROPEAN ECONOMIC AND SOCIAL COMMITTEE AND THE  
COMMITTEE OF THE REGIONS**

**Implementation report on the Commission Communication on Rare Diseases: Europe's  
challenges [COM(2008) 679 final] and Council Recommendation of 8 June 2009 on an  
action in the field of rare diseases (2009/C 151/02)**

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# 1. Introduction

## a. Policy framework

Rare diseases affect between **27 and 36 million people in the European Union** and are a key health policy priority due to the limited number of patients and scarcity of relevant knowledge and expertise regarding particular diseases.

Patients with rare diseases often spend years of uncertainty waiting for their disease to be diagnosed, and for an appropriate treatment to be found. The medical expert who can diagnose such a rare disease may practice in another region or indeed in another Member State. Scientific knowledge on the specific rare disease is likely to be insufficient and scattered.

This is why the European Union dimension and the co-operation between Member States can make a difference, for example in pooling together knowledge and expertise, in fostering research and co-operation and in granting the authorisation of the best possible medicines for the whole European Union. EU action on rare diseases provides high added value.

To achieve this goal in 2008, the Commission adopted a **Communication on Rare Diseases: Europe's challenge**<sup>1</sup>, setting out an overall strategy to support Member States in diagnosing, treating and caring for EU citizens with rare diseases. The Communication focuses on three main areas: i) improving the recognition and visibility of rare diseases; ii) supporting policies on rare diseases in the Member States for a coherent overall strategy, and iii) developing cooperation, coordination and regulation for rare diseases at EU level.

Alongside the Communication, a **Council Recommendation on action in the field of rare diseases**<sup>2</sup> was adopted a few months later, calling on Member States to put national strategies in place. The Recommendation focuses on i) definition, codification and inventory of rare diseases, ii) research, iii) European reference networks, iv) gathering expertise at EU level, v) empowerment of patient organisations, and vi) sustainability.

Article 13 of Directive 2011/24/EU<sup>3</sup> on the application of patients' rights in cross-border healthcare also addresses rare diseases. It states that the Commission shall support Member States, in particular by making health professionals aware of the tools available to assist the diagnosis of rare diseases, and by making stakeholders aware of the possibilities offered by Regulation 883/2004<sup>4</sup> for referral of rare disease patients to other Member States.

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<sup>1</sup> COM(2008) 679 final of 11 November 2008

<sup>2</sup> OJ C 151, 03/07/2009, p. 7–10

<sup>3</sup> OJ L 88, 04/04/2011, p. 45–65

<sup>4</sup> <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2004:166:0001:0123:en:PDF>

Rare Diseases were identified, for the first time, as a priority field for public health action in the European Union, in the Commission Communication of 24 November 1993<sup>5</sup> on the framework for action in the field of public health. This was followed by providing support for several projects as well as by setting up the **Rare Diseases Task Force**.

**The Orphan Medicinal Products Regulation** (Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products)<sup>6</sup> established criteria for orphan designation in the EU and delivered a set of incentives (e.g. 10-year market exclusivity, protocol assistance, access to the Centralised Procedure for Marketing Authorisation) to encourage research, development and marketing of medicines to treat, prevent or diagnose rare diseases.

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.

## **b. Bases of the report and methodology**

In the Communication and the Council Recommendation, the Commission was called upon to report on the implementation of the strategy. In order to collect information about the situation at national level, the Commission sent out an electronic questionnaire to Member States. Eighteen countries provided the requested information. Answers from Member States, together with information collected by the EUCERD Joint Action and published as “Report on the State of the Art of Rare Diseases Activities in Europe”<sup>7</sup>, served as the main source of information for this Implementation report.

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

### **a. European Commission activities**

In order to support Member 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

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<sup>5</sup> <http://aei.pitt.edu/5792/>

<sup>6</sup> <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2000:018:0001:0005:en:PDF>

<sup>7</sup> [http://www.eucerd.eu/?page\\_id=15](http://www.eucerd.eu/?page_id=15)

rare diseases" which served as a basis for the adoption of the "EUCERD Recommendations on Core Indicators for Rare Disease National Plans/Strategies"<sup>8</sup>.

Some EUROPLAN activities, especially those related to technical assistance to Member States with particular difficulties in the preparation of their national plan or strategy, are covered further by a specific work package of the EUCERD Joint Action.

Through this work package, the Commission continues to support the preparation of national plans in the countries where such plans are not yet in place.

The EUCERD Joint Action covers a 42-month period (March 2012 – August 2015). It supports Member States in the development of strategies, mapping the provision of specialised social services and integration of rare diseases into mainstream social policies, as well as supporting implementation of a coding and classification of rare diseases. The Joint Action also provides support for the production of OrphaNews Europe<sup>9</sup> and the annual State of the Art report of Rare Disease Activities in Europe.

## **b. Situation in the Member States**

**Objectives for action:** In the Council Recommendation Member States committed themselves to adopt a plan or strategy to address rare diseases as soon as possible and by the end of 2013 at the latest.

In 2009, a focus on rare diseases was relatively new and innovative in most Member States and only a few had national plans in place. These were Bulgaria, France, Portugal and Spain.

By the first quarter of 2014, 16 **Member States had national plans or strategies in place to address rare diseases**. Seven further countries are well advanced in the development of their plans/strategies.

**Member States with an adopted National plan or strategy for rare diseases:** Belgium, Bulgaria, Cyprus, Czech Republic, France, Germany, Greece, Hungary, Lithuania, Netherlands, Portugal, Romania, Slovakia, Slovenia, Spain, United Kingdom.

**Member States in advanced stage of preparation of the National plan or strategy for rare diseases:** Austria, Croatia, Denmark, Finland, Ireland, Italy, Poland.

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<sup>8</sup> [http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD\\_Recommendations\\_Indicators\\_adopted.pdf](http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_Indicators_adopted.pdf)

<sup>9</sup> <http://www.orpha.net/actor/cgi-bin/OAhome.php?Ltr=EuropaNews>

Countries vary considerably in the level of implementation of their plans. This is partially due to the fact that several countries such as the UK, Germany, the Netherlands and Belgium only recently adopted their plans/strategies. Only one country, France, has already finished implementing the first plan and adopted a second national plan.

Most Member States have no dedicated budget for the implementation of national plans. Funding is usually provided as part of overall health spending. Countries do provide occasional budgets for the implementation of specific projects. Some countries reported that budgets are under additional strain as a result of the economic crisis.

Despite their comprehensiveness and inter-sectorial approach, all plans were adopted at the level of the Ministry of Health. In the Czech Republic, in addition, the plan was also endorsed by the Prime Minister.

The scope of the rare diseases plans differs between countries. For example, while rare cancers are an important part of the rare diseases spectrum, several plans/strategies do not cover this group of diseases. This is true for Germany, France, Belgium, Denmark and Portugal. Denmark does not consider infectious diseases as rare diseases.

Fourteen countries have run information campaigns to raise awareness on rare diseases. Germany, Croatia, Cyprus and Latvia are currently preparing their campaigns.

Monitoring and evaluating national plans are important aspects of this initiative and the EU co-funded the EUROPLAN project<sup>10</sup> - and subsequently the EUCERD Joint Action<sup>11</sup> – to provide a framework to support Member States in their efforts to develop and implement their national plans.

Other countries with plans in place (Croatia, France, Lithuania, Portugal and Spain) base their monitoring strategy on EUROPLAN indicators. Bulgaria and Slovakia have no monitoring strategy. In the remaining countries monitoring strategies are under development.

### **3. Definition, codification and inventorying of rare diseases**

#### **a. European Commission activities**

*Objectives for action:* Establishing a clear definition of rare diseases is a prerequisite for effective actions in this field. In Article 3 of the Council Recommendation, 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**. It is also important to rapidly improve the codification of rare diseases in the healthcare systems. Member States

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<sup>10</sup> [http://www.europlanproject.eu/\\_newsite\\_986989/index.html](http://www.europlanproject.eu/_newsite_986989/index.html)

<sup>11</sup> <http://ec.europa.eu/eahc/projects/database.html?prjno=20112201>

have agreed to aim at ensuring that rare diseases are adequately coded and traceable in health information systems as well as to contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network<sup>12</sup>.

Examples of the **situation in various Member States as regards the rare diseases definition:**

- **Sweden:** diseases or disorders which affect fewer than 100 people per million, and which lead to a marked degree of disability;
- **Finland:** uses the definition of no more than 1 in 2000 affected persons and of severe/debilitating disease;
- **Denmark:** has not established an official definition of rare disease. Danish health authorities tend to define rare disease as those affecting no more than 500-1,000 patients in the Danish population;
- **Estonia:** has no approved official definition of rare diseases. Stakeholders, however, accept the EU definition from the Regulation on Orphan medicinal products;
- **Belgium:** defines rare diseases as life-threatening or chronically debilitating diseases which are of such low prevalence that special combined efforts are needed to address them. As a guide, low prevalence means affecting less than 5 per 10,000 individuals in the European Union Community.

## **b. Member States activities**

Member States with adopted plans or strategies comply with the EU definition for the Community level policy. Those without plans in place usually do not have any official definition of rare disease.

Currently **all Member States are using the International Classification of Diseases ICD-9 or ICD-10 systems** in which most rare diseases are absent. Recently some of the Member States decided to introduce ORPHA codes (rare diseases codification system developed by the Orphanet database) in their health statistics systems in parallel to the ICD nomenclature or as a pilot project. The EUCERD joint action is providing input into the WHO ICD-11 draft in order to ensure the presence of rare diseases in international nomenclatures.

In order to collect and make available information on rare diseases, the Commission is supporting the **Orphanet Joint Action**<sup>13</sup> through the EU Health Programme, involving all Member States, either as associated or collaborating partners. Orphanet is a relational database available in seven languages which aims to link together information on over 6,000 diseases and allows for multiple queries. Each country also has its own entry page in their national language.

<sup>12</sup> <http://www.orpha.net/consor/cgi-bin/index.php>

<sup>13</sup> <http://ec.europa.eu/eahc/projects/database.html?prjno=20102206>



## 4. Research on rare diseases

### a. European Commission activities

**Objectives for action:** under point 5.12 of the Communication and under article 3 of the Council Recommendation, Member States and the Commission are called upon to seek to improve the coordination of Community, national and regional programmes for rare diseases research. 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)**<sup>14</sup>. With a total budget of **over €620 million**, these projects span across several disease areas such as neurology, immunology, cancer, pneumology, and dermatology<sup>15</sup>. Through its research policy activities, the European Commission has also been a driving force in the launch of initiatives aiming to better coordinate research at European and international levels.

#### Examples of national programmes for research in rare diseases

In **Germany** in September 2010, a new call for proposals for the possible extension of the 10 networks which started in 2008 and the creation of new networks was published. After the evaluation of 39 proposals by a review board of international rare disease experts, the Federal Ministry of Education and Research has selected 12 networks for funding starting in 2012 with more than €21million for three years. Additional funding of rare disease research is ongoing in other funding initiatives such as the National Genome Research Network (NGFN), Innovative Therapies, Regenerative Medicine, Molecular Diagnostics, Clinical Trials and others, representing about €20 million per year.

In **France** calls for research projects are steered by the French National Agency for Research (basic research) or by the Ministry of Health (clinical research) or by both (translational research). Calls for projects in Social Sciences are also available. Patients' associations also provide research funds. Basic, clinical and translational research is constantly supported across the board with no national priority on a specific rare disease. An amount of €51 million was allotted for research during the second national plan 2011-2014.

In **Croatia** there is a lack of detailed data on funds devoted for research on rare diseases. It is estimated that around 4% of current research projects in Croatia can be related to rare diseases.

The EU-funding strategy for rare diseases research has focused on the understanding of the underlying causes for these diseases and on diagnosis, prevention and treatment. The strategy is illustrated by the FP7 health calls in 2012 and 2013, which featured several topics on rare diseases<sup>16</sup>. EU-funded collaborative research brings together multidisciplinary teams representing universities, research organisations, SMEs, industry and patient organisations

<sup>14</sup> This figure refers to research funded in the Health Theme of the FP7 Cooperation Programme (2007-2013).

<sup>15</sup> A recent publication including EU funding for rare disease research can be found on: [http://ec.europa.eu/research/health/pdf/rare-diseases-how-europe-meeting-challenges\\_en.pdf](http://ec.europa.eu/research/health/pdf/rare-diseases-how-europe-meeting-challenges_en.pdf)

<sup>16</sup> The rare disease topics in the 2012 and 2013 FP7 Health Calls were: *'Support for international rare disease research; Clinical utility of -omics for better diagnosis of rare diseases; Databases, biobanks and clinical 'bio-informatics' hub for rare diseases; Preclinical and clinical development of orphan drugs; Observational trials in rare diseases; Best practice and knowledge sharing in clinical management of rare diseases* (2012) and *Development of imaging technologies for therapeutic interventions in rare diseases; New methodologies for clinical trials for small population groups* (2013).

from across Europe and beyond. Collaborative research on European and international level is especially important in an area such as rare diseases, characterised by small patient populations and scarce resources. The EU has also funded over 100 individual fellowships, grants and training networks in this field<sup>17</sup>.

The EU-funded ERA-NET project **E-RARE-2**<sup>18</sup> aims to develop and strengthen the coordination of national and regional research programmes. One of its main activities is the launch of Joint Transnational Calls. These calls have involved funding agencies from 13 EU Member States<sup>19</sup> as well as Turkey, Israel, Switzerland, and Canada. Together with its predecessor, E-RARE-2 has funded over 60 research projects.

In collaboration with its national and international partners, the European Commission spearheaded the launch of the **International Rare Diseases Research Consortium (IRDiRC)**<sup>20</sup> 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. At the end of 2013, IRDiRC had over 35 member organisations from four continents committed to work together towards the initiative's goals.

The EU's strong commitment to rare disease research and to IRDiRC is set to continue through Horizon 2020, the EU Framework Programme for Research and Innovation funding for the period 2014-2020. Over the next seven-year period, the EU will continue to fund research in rare diseases for the benefit of patients in Europe and across the world.

**Patient registries and databases** constitute important instruments to serve research in the field of rare diseases and to improve patient care and healthcare planning. They help to pool data in order to achieve a sufficient sample size for epidemiological and/or clinical research. They are also vital to assessing the feasibility of clinical trials, to facilitating the planning of appropriate trials and to supporting the enrolment of patients. They can also be used for the measurement of quality, safety, efficacy and efficiency of a treatment. An overview of issues surrounding the establishment, governance and financing of academic registries was published by Orphanet<sup>21</sup>.

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. A minority of them are managed by

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<sup>17</sup> These activities have been funded through the FP7 People (Marie Curie Actions) and Ideas (European Research Council) programmes. Read more on: <http://ec.europa.eu/research/mariecurieactions> and on: <http://erc.europa.eu>

<sup>18</sup> Read more on the E-RARE-2 website: <http://www.e-rare.eu>

<sup>19</sup> Austria, Belgium, France, Germany, Greece, Hungary, Italy, Latvia, the Netherlands, Poland, Portugal, Romania and Spain.

<sup>20</sup> Read more on the IRDiRC website: <http://www.irdirc.org>

<sup>21</sup> <http://www.orpha.net/orphacom/cahiers/docs/GB/Registries.pdf>

pharmaceutical or biotech companies, while others are being run by patient organisations. The lack of interoperability between rare diseases registries is severely jeopardising the registries' potential.

This is why 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 for all stakeholders, to support new and existing registries in view of their interoperability, to provide IT tools to maintain data collection and to host activities of the surveillance networks.

### **b. Member States activities**

Some countries have specific funding programmes for research in the field of rare diseases. Amongst the countries which have established both on-going or finalised specific rare disease research funding programmes/calls are: Austria, France, Germany, Hungary, Italy, the Netherlands, Portugal, Spain and the United Kingdom.

Many other countries support rare disease projects through generalised research funding programmes. A few countries (such as France, Germany, Italy, the Netherlands and Spain) also have, or have had, specific initiatives and incentives in place to boost R&D in the field of orphan medicinal products and other innovative therapies at national level.

## **5. Centres of expertise and European reference networks for rare diseases**

### **a. European Commission activities**

Directive 2011/24/EU on the application of patients' rights in cross-border healthcare (2011)<sup>22</sup> sets the rules for patients' rights to access to safe and good quality treatment across EU borders and reimbursement rules. The Directive provides a firm basis for increased cooperation between national health authorities. Some provisions address rare diseases. Article 12 foresees enhanced cooperation of Member States including the criteria and conditions for **European Reference Networks** and for healthcare providers.

The Directive aims to identify already established centres of expertise and to encourage voluntary participation of healthcare providers in the future European Reference Networks. On 10<sup>th</sup> March 2014 the Commission adopted a list of criteria and conditions that the European Reference Networks must fulfil and the conditions and criteria required from healthcare providers wishing to become a Member of a European Reference Network<sup>23,24</sup>.

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<sup>22</sup> <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2011:088:0045:0065:en:PDF>

<sup>23</sup> OJ L 147, 17/05/2014, p. 71–78

<sup>24</sup> OJ L 147, 17/05/2014, p. 79–87

Before the adoption of Directive 2011/24/EU, the Commission supported **10 specific pilot European Reference Networks for Rare Diseases** through the EU Health Programme. Experience gained by these projects helped to design a legal framework and will serve future European Reference Networks.

List of **pilot European Reference Networks for Rare Diseases**

- **Dyscerne**: European Network of Centres of Reference for Dysmorphology
- **ECORN CF**: European Centres of Reference Network for Cystic Fibrosis
- **PAAIR**: Patient Associations and Alpha1 International Registry,
- **EPNET** European Porphyria Network,
- **EN-RBD** European Network of Rare Bleeding Disorders, Paediatric Hodgkins Lymphoma Network
- **NEUROPED**: European Network of Reference for Rare Paediatric Neurological Diseases
- **EURO HISTIO NET**: A reference network for Langerhans cell histiocytosis and associated syndrome in EU
- **TAG**: Together Against Genodermatoses
- **CARE NMD**: Dissemination and Implementation of the Standards of Care for Duchene muscular Dystrophy in Europe

## **b. Member States activities**

Member States have taken very different approaches in the organisation of the centres of expertise within their healthcare systems. Some countries have formally designated centres of expertise for rare diseases: France, Denmark, Spain and the United Kingdom. Italy has regionally designated centres of expertise for rare diseases.

Designation criteria vary from country to country, sometimes even from region to region within a country, even if these criteria are often in line with EUCERD recommendations on quality criteria for centres of expertise for rare diseases in Member States<sup>25</sup>.

A number of countries have centres of expertise for rare diseases which, while not officially designated, are acknowledged by authorities to varying degrees: Austria, Belgium, Croatia, Czech Republic, Cyprus, Germany, Greece, Hungary, Ireland, the Netherlands, Sweden, and Slovenia.

A number of countries have centres of expertise for rare diseases which are recognised by reputation only, sometimes self-declared as centres of expertise: Bulgaria, Estonia, Finland, Latvia, Lithuania, Portugal, Poland, Romania, and the Slovak Republic.

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<sup>25</sup> [http://www.eucerd.eu/?post\\_type=document&p=1224](http://www.eucerd.eu/?post_type=document&p=1224)

## 6. Gathering the expertise on rare diseases at European level

**Objectives for action:** under Section V of the Council Recommendation, Member States are called upon to **gather national expertise on rare diseases and support its pooling**.

The majority of Member States support the pooling of expertise with European counterparts in order to foster the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases. Several of them have organised education and training for health professionals to make them aware about existing resources available.

In order to support this process, the Commission recently co-funded the **Rare Best Practices project**<sup>26</sup>. This is a four-year project (January 2013-December 2016) co-funded by the Seventh Framework Programme for Innovation and Technological Development (FP7). The main aims of the project include: elaborating standards and transparent reliable procedures for the development and evaluation of clinical practice guidelines for rare diseases and establishing consensus on an innovative methodology.

## 7. Empowerment of patients' organisations

### a. European Commission activities

**Objectives for action:** under Article 6 of the Council Recommendation, Member States are called on to consult patients' organisations on the policies in the field of rare diseases and to promote the activities of these organisations.

The participation of patients' organisations in all aspects of the development of rare diseases policy is very important for identifying patients' needs. The Commission is supporting this approach at EU level by involving European umbrella patients' organisations in various actions such as expert groups and committees.

The Commission has also provided operating grants to patients' organisations via the EU Health Programme.

### b. Member States activities

An increasing number of national alliances of rare disease patient organisations have been created in Europe. According to Orphanet, by the end of 2013 there were **2512 rare diseases specific patients' organisations** including 2161 national, 213 regional, 72 European and 61 international organisations.

All Member States which replied to the questionnaire engage in active dialogue with rare diseases patients' organisations, mainly by consulting patients and patients' representatives on the policies in the field of rare diseases.

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<sup>26</sup> <http://www.rarebestpractices.eu/>

## 8. Governance and European coordination

**Objectives for action:** the Communication under point 7 states that the Commission should be assisted by an advisory committee on rare diseases.

Such a Committee was set up by Commission Decision of 30 November 2009 establishing a European Union Committee of Experts on Rare Diseases (2009/872/EC)<sup>27</sup>. The Committee's work resulted in the adoption of five sets of recommendations and an opinion, along with the publication of a bi-monthly newsletter and an annual report on the State of the Art of Rare Diseases Activities in Europe which describes activities at Member State, EU and global levels.

The Committee was recently replaced by the Commission expert group on rare diseases<sup>28</sup> in line with provisions of the Framework for Commission expert groups: horizontal rules and public register<sup>29</sup>.

The expert group is composed of Member States' representatives, as well as representatives of patients' organisations, European associations of producers of products or service providers, European professional associations or scientific societies and individual experts. The main task of the expert group is to advise the Commission in the implementation of Union actions on rare diseases including drawing up of legal instruments, policy documents, guidelines and recommendations.

## 9. Actions to increase high-quality health care for rare diseases

### a. Regulation on Orphan medicinal products

In response to a public health concern and in order to stimulate the research and development of orphan medicines, the EU adopted the Orphan Regulation which aims to provide incentives for the development of orphan medicinal products. Regulation (EC) No 141/2000<sup>30</sup> of the European Parliament and of the Council establishes a centralised procedure for the designation of orphan medicinal products and puts in place incentives for the research, marketing and development of medicines for rare diseases.

As of January 2014, more than **90 orphan medicines have been authorised** by the European Commission. Equally important, the European Commission has designated more than 1000 products as orphan medicinal products<sup>31</sup>. The sponsors developing these products benefit from incentives such as protocol assistance. This assistance should facilitate the development and authorisation of innovative medicines for the benefit of the patients.

In recent years, the number of designations has increased while the number of authorisations has remained stable (7 authorisations in 2013 versus 10 authorisations in 2012).

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<sup>27</sup> <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2009:315:0018:0021:EN:PDF>

<sup>28</sup> [http://ec.europa.eu/health/rare\\_diseases/docs/dec\\_expert\\_group\\_2013\\_en.pdf](http://ec.europa.eu/health/rare_diseases/docs/dec_expert_group_2013_en.pdf)

<sup>29</sup> [http://ec.europa.eu/transparency/regexpert/PDF/C\\_2010\\_EN.pdf](http://ec.europa.eu/transparency/regexpert/PDF/C_2010_EN.pdf)

<sup>30</sup> <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2000:018:0001:0005:en:PDF>

<sup>31</sup> [http://ec.europa.eu/health/human-use/orphan-medicines/index\\_en.htm](http://ec.europa.eu/health/human-use/orphan-medicines/index_en.htm)

## **b. Facilitating access to orphan medicinal products**

Despite these incentives, authorised orphan medicinal products are not available in all EU Members and access for patients is not equal in all EU Member States. Moreover, important delays in availability have been observed. A project has therefore been initiated by Member States and the Commission to coordinate investments in the evaluation of new medicines and in the exchange of information and knowledge<sup>32</sup>.

Although decisions on pricing and reimbursement are an exclusive national competence, the Member States are faced with important and common challenges to provide affordable and sustainable access to valuable medicines for patients with obvious unmet medical needs. Meeting these challenges can pose even more of a problem when limited numbers of patients are concerned and possible treatments to meet the unmet medical needs are scarce and expensive, as is often the case with rare diseases and orphan medicinal products.

### **Working Group "Mechanism of Coordinated Access to Orphan Medicinal Products" under the Process on Corporate Responsibility in the Field of Pharmaceuticals**

The main objective of the working group<sup>33</sup> was to examine how to provide "real life access" to orphan medicinal products for patients suffering from rare diseases. The main recommendation of the group was to develop a coordinated mechanism between volunteering Member States and sponsors to evaluate the value of an orphan medicinal product, which could be based on a transparent value framework, in order to support the exchange of information aimed at enabling informed decisions at Member State level on pricing and reimbursement. This should lead to more rational prices for payers, more predictable market conditions for industry and more equitable access for patients.<sup>34</sup>

## **c. Population screening for rare diseases**

**Objectives for action:** In the Communication under point 5, the Commission committed to **evaluating current population screening** (including neonatal screening) strategies for rare diseases.

The Commission commissioned a report on the practices of newborn screening for rare disorders implemented in all the EU Member States including the number of centres, an estimation of the number of infants screened and the number of disorders included in the newborn screening as well as reasons for the selection of these disorders<sup>35</sup>. The majority of Member States covered by this report has a body which oversees newborn screening. The

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<sup>32</sup>[http://ec.europa.eu/enterprise/sectors/healthcare/competitiveness/process\\_on\\_corporate\\_responsibility/platform\\_access/index\\_en.htm#h2-2](http://ec.europa.eu/enterprise/sectors/healthcare/competitiveness/process_on_corporate_responsibility/platform_access/index_en.htm#h2-2)

<sup>33</sup>[http://ec.europa.eu/enterprise/sectors/healthcare/competitiveness/process\\_on\\_corporate\\_responsibility/platform\\_access/index\\_en.htm#h2-2](http://ec.europa.eu/enterprise/sectors/healthcare/competitiveness/process_on_corporate_responsibility/platform_access/index_en.htm#h2-2)

<sup>34</sup> After the group concluded their work in 2013, a few working group members pursued the discussions on the initiative of the Medicines Evaluation Committee (MEDEV, an informal group of experts from statutory health insurance institutions in Europe) with a view to put the group's conclusions into practice and set up pilot projects.

<sup>35</sup> <http://ec.europa.eu/eahc/news/news104.html>

**numbers of diseases screened vary substantially between Member States, from one in Finland to 29 in Austria.**

On the basis of this report, the EU Committee of experts on rare diseases adopted an opinion on potential areas of European collaboration in the field of newborn screening<sup>36</sup>.

## **10. Global dimension of the rare diseases policy**

**Objectives for action:** The Communication aims at **fostering cooperation on rare diseases at an international level** with all interested countries and in close collaboration with the World Health Organisation.

The European Union and its Member States are regarded as leaders in the development of actions for rare diseases. Actions undertaken by the EU and in Member States have influenced developments in this field in non-European countries, and political and technical developments in European Union also had a significant impact on other countries' rare diseases policy.

Several specific actions have been taken up by non-European countries, sometimes as a result of being visible on the Internet, as in the case of Orphanet which publishes on-line information in seven languages and has gained importance as a truly global source of information. Other initiatives are supporting global international organisations in their work related to rare diseases, such as for example the EUCERD Joint Action involvement in the ICD-10 update. The International Rare Disease Research Consortium<sup>37</sup> is an excellent example of international cooperation initiated by the European Commission.

The European Commission policy on rare diseases has as well benefited from policy achievements in other countries.

## **11. Conclusions and proposals for the future**

According to the Council recommendation, the implementation report 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 European Union has come a long way in fostering co-operation to improve the lives of people suffering from a rare disease since the adoption of the Commission Communication in 2008 and the Council Recommendation in 2009.

By and large the objectives of the Communication and the Council Recommendation have been **reached. Both have served to strengthen the cooperation** between the European Union, the Member States and all the relevant stakeholders.

The Commission has fostered the exchange of experiences to help Member States develop their national plans or strategies for rare diseases.

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<sup>36</sup> [http://www.eucerd.eu/wp-content/uploads/2013/07/EUCERD\\_NBS\\_Opinion\\_Adopted.pdf](http://www.eucerd.eu/wp-content/uploads/2013/07/EUCERD_NBS_Opinion_Adopted.pdf)

<sup>37</sup> Read more on the IRDiRC website: <http://www.irdirc.org>



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 a significant number are close to adopting a plan. Supporting Member States in this endeavour remains the key priority for the Commission's work in this area.

Despite such encouraging progress, there is still a long way to go to ensure that people suffering from a rare disease can obtain the right diagnosis and best possible treatment throughout the EU. There are still Member States who do not yet have a national plan or strategy. In those Member States that do have a national plan or strategy in place, implementation has mostly started only recently and needs to be monitored.

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
- Make use of the Directive 2011/24/EU on the application of patients' rights in cross-border healthcare to bring together **European Reference Networks** on rare diseases. 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.

The views expressed by Members States and stakeholders, in the framework of the Commission expert group on rare diseases, will also be taken into account.