



European Union action in the field of Rare Diseases

Burgos, 26th July 2010

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Legal basis for the developments of the EU Public Health Policy

■ Based on new Article 168 (former 152) of the EU Treaty

■ A Community action programme on Rare Diseases, including genetic diseases, was adopted for the period of 1 January 1999 to 31 December 2003 with the aim of ensuring a high level of health protection in relation to RD. As the first EU effort in this area, specific attention was given to improving knowledge and facilitating access to information about these diseases.

■ Orphan Medicinal Product Regulation (Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products, was proposed to set up the criteria for orphan designation in the EU and describes the incentives (e.g. 10-year market exclusivity, protocol assistance, access to the Centralised Procedure for Marketing Authorisation) to encourage the research, development and marketing of medicines to treat, prevent or diagnose rare diseases.

■ For the period 2008-2013 the Commission has adopted the White Paper COM(2007) 630 final "Together for Health: A Strategic Approach for the EU 2008-2013" of 23 October 2007 developing the EU Health Strategy. Actions under Objective 1 of this EU Strategy cover a Communication on European Action in the Field of Rare Diseases and in point 4.1 of this EU Strategy it is suggested to put forward EC-level structured cooperation mechanisms to advise the Commission and to promote cooperation between the Member States.

Legal basis for the developments of the EU Public Health Policy

- As a consequence Rare diseases are now one of the priorities in the [Second EU Health Programme 2008-2013](#). According to the DG SANCO Work Plans for the implementation of the Public Health Programme, main lines of action and priorities are chosen every year. **Work Plan for 2011 in process of adoption.**
- In the current Framework Programme, the FP7, the Health Theme of the "Cooperation" Specific Programme, is designed to support multinational collaborative research in different forms. The main focus of the Health theme in the rare diseases area are Europe-wide studies of natural history, pathophysiology, and the development of preventive, diagnostic and therapeutic interventions.
- Commission Communication COM (2008) 679/2 to the European Parliament, the Council, the Economic and Social Committee and the Committee of the Regions on Rare diseases: Europe's challenges creating an integrated approach for the EU action in the field of rare diseases. **Adopted 11th November 2008.**
- Council Recommendation on a European action in the field of rare diseases recommending actions at national level to implement the EU action (e.g. National Plans for Rare Diseases). **Adopted 8th June 2009.**

Future legal basis for the developments of the EU Public Health Policy

■ **Decision of the Commission creating a European Union Committee of Experts on Rare Diseases during 2009. To be composed by 51 members representing Member States, patient's organisations, industry, FP Projects, Health Programme projects, etc.**

Adopted 30th November 2009.

■ **Directive of the European Parliament and of the Council on the application of patients' rights in cross-border healthcare (COM(2008)414) provides for the development of European reference networks (ERNs) to be facilitated by the Member States. The ERN for Rare Diseases will have a strategic role in the improvement of quality treatment for all patients throughout the European Union as called by the patients' organisations. To be adopted End 2011 (tbc).**

The Commission Communication and the Council Recommendation on rare diseases

- **There is probably no other area in public health in which 27 national approaches could be considered to be so inefficient and ineffective as with rare diseases. The reduced number of patients for these diseases and the need to mobilise resources could be only efficient if done in a coordinated European way.**
- **Article 168.6 provides for the adoption by qualified majority by the Council of Recommendations, on the basis of Commission proposals, for the purposes set out in that article.**
- **These Recommendations are the only legislative tool provided for in Article 168 on public health except for the few areas where measures or incentive measures may be adopted (see Article 168.4).**

The Commission Communication and the Council Recommendation on rare diseases

I. Plans and strategies in the field of rare diseases

Calls on the MS to elaborate and adopt a plan or strategy by the end of 2013.

II. Adequate definition, codification and inventorying of rare diseases

Evokes the common definition of a rare disease as a condition affecting no more than 5 per 10 000 persons; aims to ensure that rare diseases are adequately coded and traceable in all health information systems based on the ICD and in respect of national procedures; and encourages MS to contribute actively to the inventory of rare diseases based on the Orphanet network.

III. Research on rare diseases

Calls for the identification and fostering of rare disease research at all levels.

IV. Centres of expertise and European reference networks for rare diseases

Asks the MS to identify and facilitate networks of expertise based on a multidisciplinary approach to care, and foster the diffusion and mobility of expertise and knowledge.

The Commission Communication and the Council Recommendation on rare diseases

V. Gathering the expertise on rare diseases at European level

MS should share best practices, develop medical training relevant to the diagnosis and management of rare diseases, coordinate European guidelines, and, to minimise the delay in access to orphan drugs, MS should share clinical/therapeutic added-value assessment reports at the Community level.

VI. Empowerment of patient organisations

MS should consult patient representatives on policy development; facilitate patient access to updated information on rare diseases; promote patient organisation activities.

VII. Sustainability

Long-term sustainability in the field of information, research and healthcare of infrastructures must be ensured.

New priorities after Commission Communication and Council Recommendation

I. Plans and strategies in the field of rare diseases

- **The Member States are invited to establish national or regional action plans for RD before 2013 in order to implement the actions suggested in the Commission Communication and the Council Recommendation and to provide an annual report on the progress made toward this objective**
- **The Commission will provide European guidelines for the elaboration of these action plans for RD (EUROPLAN Project selected for funding for the period 2008-2011). Appropriate conferences will be organised (twelve national conferences scheduled during 2010). Continuity of EUROPLAN (under the form of a Joint Action scheduled)**
- **A national plan/strategy (NP/NS) can be defined as the sum of integrated and comprehensive health policy actions for RD, to be developed and implemented at national level. A plan has: a) well specified objectives and b) actions that have to be supported by a budget, implemented within a time frame, evaluated with specific indicators (EUROPLAN Definition).**

New priorities after Commission Communication and Council Recommendation

I. Plans and strategies in the field of rare diseases

- **This definition includes two central concepts of the Council Recommendation on RD. '*Integrated*' refers to the fact that strategies should be developed in a way to identify complementarities, maximize synergies and avoid duplications. '*Comprehensive*' refers to the fact that the actions foreseen in the plan should fulfill all main patients' needs (e.g. quality of care but also social services and centres of expertise).**
- **Definition of common and harmonised indicators to appraise and evaluate the national plan/strategy (NP/NS) of rare diseases.**
- **The EUROPLAN Recommendations also includes the international dimension not possible to fill at national level (e.g. classification and codification, reference networks, orphan drugs, research, etc.). To be analysed by health authorities (Krakow, 13th May 2010) and national conferences (2010). EUROPLAN final conference (Rome, March 2011).**
- **Continuity of EUROPLAN after 2011 (under the form of a Joint Action) scheduled.**

***New priorities after Commission Communication and Council Recommendation
Plans and strategies in the field of rare diseases***

■ **Six Member States have adopted National Plan/Strategy for Rare Diseases:**

[French National Plan on Rare Diseases \(2005-2008\)](#)

The Plan 2005-2008 is [under evaluation](#) (concluded) before the adoption of a new Plan 2010-2014.

[Bulgarian National Plan on Rare Diseases 2009-2013](#)

[Greek National Plan on Rare Diseases 2008-2012](#)

[Portuguese National Programme for Rare Diseases 2008-2015](#)

[Rare Diseases Strategy of the Spanish National Health System](#)

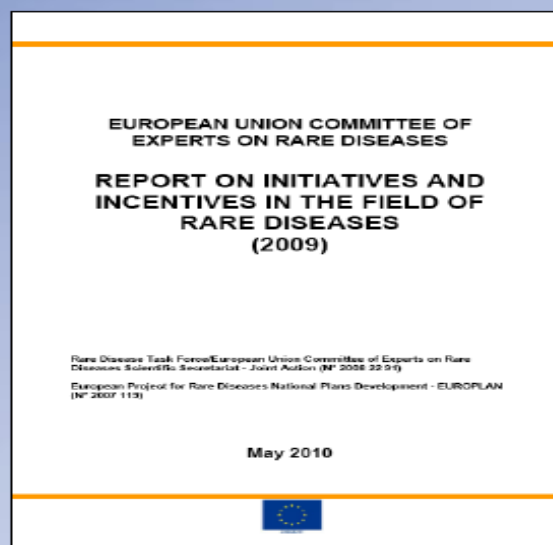
[Czech Republic National Strategy for Rare diseases 2011-2013](#)

■ **Six are in an advanced stage for adoption: Belgium, Italy (framework law and national plan), Romania, Austria, United Kingdom**

■ **Eighth have (+ or -) structures in place for the definition of a plan: Luxembourg, Poland, Cyprus, Finland, Lithuania, Malta, Norway, Sweden**

***New priorities after Commission Communication and Council Recommendation
Plans and strategies in the field of rare diseases***

Report on initiatives and incentives 2009



www.eucerd.eu

I. Plans and strategies in the field of rare diseases

From the side of the economic costs of rare diseases the Commission selected in 2009 the project

SOCIAL ECONOMIC BURDEN AND HEALTH-RELATED QUALITY OF LIFE IN PATIENTS WITH RARE DISEASES IN EUROPE (BURQOL-RD)

- **The general objective of this project is to develop a disease based model capable of quantifying the socioeconomic burden and Health-Related Quality of Life (HRQOL) for patients in Europe with rare diseases (RD) and their caregivers.**
- **The model will incorporate information on medical resources related to RD (e.g. hospitalisation, consultations, drugs), on non-medical resources (e.g. walking sticks, wheelchairs, modifications to house and car), services (e. g. home care, transportation), informal care by relatives, productivity losses, intangible costs as well as the overall HRQOL (utilities). HRQL will be measured both in RD patients and their caregivers.**
- **This information will be collected with a standardized pre-tested questionnaire from a sample of patients registered in RD associations. The disease information that will be collected will include disease duration and self-assessed disease severity.**
- **Accordingly, the mean annual costs will be estimated from a society perspective.**

Consortium led by the **Canary Islands Foundation for Research and Health** with the London School of Economics, The Swedish Institute for Health Economics, Università Commerciale Luigi Bocconi and others.

I. Plans and strategies in the field of rare diseases

From the side of the socioeconomic needs the Commission has selected and supported during last years three extensive surveys performed by EURORDIS

- ***The Voice of 12,000 Patients***”, was officially launched on March 3 2009. The book, published by **EURORDIS**, presents the conclusions of the **EurordisCare 2** and **EurordisCare 3** surveys on the experience and expectations on diagnoses and access to care of 12 000 patients representing 18 rare diseases and 24 European countries.
- It details the methodology, the overall results, results by country and by disease, and how the surveys have contributed to the development of policies and actions in favour of better diagnosis and care for rare disease patients.

I. Plans and strategies in the field of rare diseases

From the side of the socioeconomic needs the Commission has selected and supported during last years three extensive surveys performed by EURORDIS

- **25% of patients reported waiting between 5 and 30 years from the time of first symptoms to a confirmatory diagnosis of their disease.**
- **40% of patients were initially misdiagnosed leading to severe consequences such as inappropriate medical interventions, including surgery and psychological treatment.**
- **In 33% of cases, the diagnosis was announced in unsatisfactory terms or conditions.**
- **In 12.5% of cases, it was announced in unacceptable ones.**
- **The genetic nature of the disease was not communicated to the patient or family in 25% of cases. This is paradoxical, given the genetic origin of most rare diseases.**
- **Genetic counselling was only provided in 50% of cases.**
- **25% of patients had to travel to a different region to obtain a diagnosis and 2% had to travel to a different country.**

New priorities after Commission Communication and Council Recommendation

II. Adequate definition, codification and inventorying of rare diseases

- The database Orphanet will be supported using appropriate financial instruments



The screenshot shows the Orphanet website interface. At the top, there are language links: Français, English, Español, Deutsch, Italiano. Below this is the Orphanet logo and the tagline "The portal for rare diseases and orphan drugs". A navigation bar contains links for Rare diseases, Orphan drugs, Clinics, Diagnostic tests, Research and trials, Patient organisations, Directory of resources, and Education and media. The main content area is divided into three sections: SIMPLE SEARCH, OTHER SEARCH OPTION(S), and ORPHANET DATA. The SIMPLE SEARCH section has a search box and an "OK" button. The OTHER SEARCH OPTION(S) section lists links for Orphan drugs, Patient organisations, Research and trials, Clinics, Diagnostic tests, and Directory of resources. The ORPHANET DATA section shows statistics: Diseases (7242), Clinics (3017), Laboratories (2838), Professionals (10136), and Daily visitors (22558). Below the main content area, there are four boxes: RARE DISEASES, RESOURCES DIRECTORY, SERVICES FOR PROFESSIONALS, and SERVICES FOR PATIENTS. The RARE DISEASES box lists links for information about a disease, alphabetical list, search by clinical sign, emergency guidelines, patient encyclopaedia, professional encyclopaedia, and about rare diseases. The RESOURCES DIRECTORY box lists links for clinics, reference centres, clinical laboratories, research projects, registries / databases, professionals, patient organisations, EuroGentest, and register your activity. The SERVICES FOR PROFESSIONALS box lists links for professional encyclopaedia, search by clinical sign, emergency guidelines, Orphanet Journal Of Rare Diseases, newsletters, education tools, register your activity, and OrphanXchange. The SERVICES FOR PATIENTS box lists links for patient encyclopaedia, information about a disease, patient organisations, clinics, participate in clinical research, contact other patients / families, training sessions, and newsletters. The NEWS box lists links for Orphanet's 10th birthday, conference presentation highlights, EC Rare Disease webpages, and French National Plan for Rare Diseases.

Improve the quality of medical care for Rare Diseases.

Provide adapted services for the rare diseases community

[About Orphanet](#) | [Quality charter](#)
[Register your activity](#)

RARE DISEASES

- > Information about a disease
- > Alphabetical list
- > Search by clinical sign
- > Emergency guidelines
- > Patient encyclopaedia
- > Professional encyclopaedia
- > About Rare Diseases

RESOURCES DIRECTORY

- > Clinics
- > Reference centres
- > Clinical laboratories
- > Research projects
- > Registries / databases
- > Professionals
- > Patient organisations
- > EuroGentest
- > Register your activity

ORPHAN DRUGS

Services for professionals

- > Professional encyclopaedia
- > Search by clinical sign
- > Emergency guidelines
- > Orphanet Journal Of Rare Diseases
- > Newsletters
- > Education tools
- > Register your activity
- > OrphanXchange

Services for patients

- > Patient encyclopaedia
- > Information about a disease
- > Patient organisations
- > Clinics
- > Participate in clinical research
- > Contact other patients / families
- > Training sessions
- > Newsletters

NEWS

- > [Orphanet's 10th birthday](#)
- > [Conference presentation highlights](#)
- > EC Rare Disease webpages
- > [French National Plan for Rare Diseases](#)

New priorities after Commission Communication and Council Recommendation

II. Adequate definition, codification and inventorying of rare diseases

Revision of the International Classification of Diseases (ICD)

- The WHO has launched the process of revision of the International Classification of Diseases (ICD) -10 to prepare the new ICD-11 which should be ready around 2015. The EC is very involved on the process from the side of the Rare Diseases.
- Discussions on the revision and improvement of the ICD will be also launched for the mental health disorders.
- The EU Task Force on Rare Diseases cooperates with the WHO Topic Advisory Group on Rare Diseases.
- 5 838 rare diseases listed in Orphanet but only 240 having an explicit code in the ICD-10
- An international Workshop on Classification and Codification on Rare Diseases will be organised by the Commission and WHO (Luxembourg, October 2010)

New priorities after Commission Communication and Council Recommendation

III. Research on rare diseases

- **Assess research landscape, and improve coordination of Community, national and regional programmes**
- **Identify needs and priorities, and promote interdisciplinary co-operative approaches**
- **Foster participation of national researchers in RD research projects**
- **Include in national plans or strategies provisions aimed at fostering research in the field of RD**
- **Facilitate the development of RD research cooperation with 3rd countries active in RD research, incl. exchange of information and sharing of expertise.**

New priorities after Commission Communication and Council Recommendation

III. Research on rare diseases

First Call for Proposals FP7

HEALTH-2007-2.4.4-1: Natural course and pathophysiology of rare diseases. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 3 million).

HEALTH-2007-2.4.4-2: Research capacity-building in the field of rare diseases. Coordination or support action.

→ 11 projects (10 CP + 1 SA) supported for a global budget of € 30 million

Third Call for Proposals FP7

HEALTH-2009-2.4.4-1: Rare neurological diseases. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 6 million).

HEALTH-2009-2.4.4-2: Preclinical development of substances with a clear potential as orphan drugs. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 3 million).

→ 9 projects (7 under 2.4.4-1 and 2 under 2.4.4-2) selected for funding for a global budget of € 45 million – negotiation being finalised

Fourth Call for Proposals FP7

HEALTH.2010.2.4.4-1: Clinical development of substances with a clear potential as orphan drugs. Collaborative projects (small or medium-scale focused research projects; max EC contribution € 6 million).

HEALTH.2010.2.4.4-2: ERA-Net on rare diseases. Coordinating action (max EC contribution € 2 million).

→ Evaluation January/February 2010; entry into negotiations May/June 2010

New priorities after Commission Communication and Council Recommendation

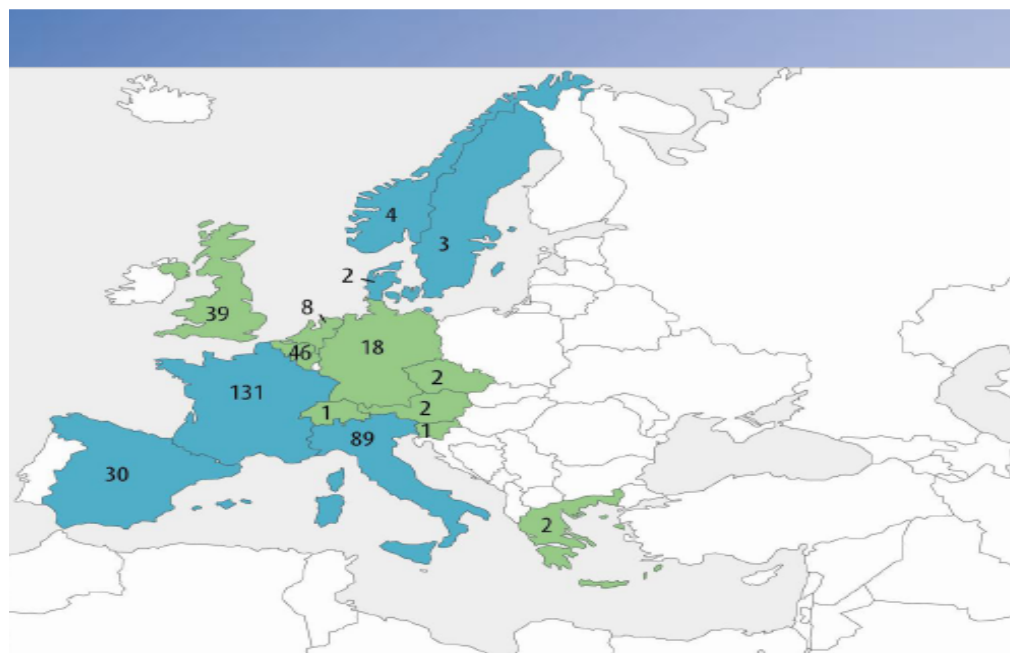
III. Research on rare diseases

E-Rare: ERA-Net for research programmes on rare diseases

- EC FP6 - funded project (2 Mi.€)
- Coordination action, **4 years** (start date: June 2006)
- Objectives: coordinate national or regional research programmes on RD and develop joint and strategic activities to:
 - Harmonize and develop synergies between national research programmes on rare diseases
 - Develop common research policy on rare diseases
 - Implement transnational research funding activities (Joint Calls)

***New priorities after Commission Communication and Council Recommendation
IV. Centres of expertise and European reference networks for rare diseases***

Designated centres of expertise: Positive effect documented in Denmark, France, Norway



Countries with official RD centres:

- Denmark
- France
- Sweden
- Norway
- Italy
- Spain

Countries with official expert centres:

- Belgium
- Austria
- Czech Republic
- Germany
- Greece
- Netherlands
- Slovenia
- Switzerland
- UK

***New priorities after Commission Communication and Council Recommendation
IV. Centres of expertise and European reference networks for rare diseases***

Some suggested criteria by the EU Task Force on Rare Diseases to be fulfilled by the European reference networks are:

- Sufficient activity and capacity to provide relevant services and maintain quality of the services provided
- Capacity to provide expert advice, diagnosis or confirmation of diagnosis, to produce and adhere to good practice guidelines and to implement outcome measures and quality control
- Demonstration of a multi-disciplinary approach;
- High level of expertise and experience documented through publications, grants or honorific positions, teaching and training activities
- Strong contribution to research
- Involvement in epidemiological surveillance, such as registries
- Close links and collaboration with other expert centres at national and international level and capacity to network
- Close links and collaboration with patients associations where they exist.
- Appropriate arrangements for referrals of patients from other Member States established within a framework.
- Appropriate capacities to diagnose, to follow-up and manage patients with evidence of good outcomes so far as applicable.

***New priorities after Commission Communication and Council Recommendation
IV. Centres of expertise and European reference networks for rare diseases***

The Work Plan 2006 for the implementation of the EU public health programme, introduced for the first time as a priority in the area of rare diseases: to develop European Networks of Centres of Reference for Rare Diseases. According to this priority 10 Projects have been selected for funding between 2006 and 2009:

- **European Centres of Reference Network for Cystic Fibrosis** with the Klinikum der Johann Wolfgang Goethe-Universität (DE) as Project Leader,
- **European Network of Centres of Reference for Dysmorphology** with The University of Manchester (UK) as Project Leader,
- **Patient Associations and Alpha1 International Registry** with the Stichting Alpha1 International Registry (NL) as Project Leader,
- **European Porphyria Network: providing better healthcare for patients and their families** with the Assistance Publique - Hôpitaux de Paris (FR) as Project Leader,
- **Establishment of a European Network of Rare Bleeding Disorders**, with the Università degli Studi di Milano (IT) as Project Leader.
- **European network of paediatric Hodgkin's lymphoma – European-wide organisation of quality controlled treatment** with the University of Leipzig (D) as Project Leader.
- **European Network of Reference for Rare Paediatric Neurological Diseases (NEUROPED)** with the European Network for Research on Alternating Hemiplegia (AT) as Project Leader.
- **A reference network for Langerhans cell histiocytosis and associated syndrome in EU** with Assistance Publique Hôpitaux de Paris (FR) as Project Leader.
- **Improving Health Care and Social Support for Patients and Family affected by Severe Genodermatoses – TogetherAgainstGenodermatoses (TAG)** with Fondation René Touraine (FR) as Project Leader.
- **European Reference Network of expert centres in rare anaemias (ENERCA 3)** with Hospital Clínic de Barcelona (ES) as Project Leader.

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

An evaluation of possible population screening (including neonatal screening) strategies for Rare Diseases will be launched

Call for Tender launched in June 2009 concerning evaluation of population newborn screening practices for rare disorders in Member States of the European Union (18 months)

***Deliverable 1:* "Report on the practices of NBS for rare diseases implemented in all the Member States including number of centres, estimation of the number of infants screened and the number of disorders included in the NBS as well as reasons for the selection of these disorders". The study includes the necessary tables which list all screening requirements and outputs useful to adopt future decisions in a comparative basis.**

***Deliverable 2:* "Expert opinion on the development of European policies in the field of newborn screening for rare diseases". This expert opinion will also discuss the existing barriers and propose solutions to be implemented, if feasible, at the EU level.**

***Deliverable 3:* "Set up of a European Union Network of experts on Newborn Screening and organization of a Final European Experts Consensus Workshop on Newborn Screening"**

A consortium leaded by the Istituto Superiore di Sanità (Italy) is working about.

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

- **Joint Action between the Commission and the Member States to be launched in June 2010 concerning the support to the EUROCAT (European Congenital Anomalies Surveillance Network).**
- **There are very few rare diseases for which a primary prevention is possible. Still, primary preventive measures for rare diseases will be taken when possible (e.g. prevention of neural tube defects by Folic Acid supplementation). Action in this field should be the topic for a debate at EU level led by the Commission aiming to determine for which rare diseases primary preventive measures may be successful.**

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

Actions to ensure equal access to all EU patients to orphan drugs

Between April 2000 and April 2010 a total of 1 113 applications were submitted for designation as orphan medicinal products and the Committee for Orphan Medicinal Products (COMP) adopted 724 positive opinions and 16 negative opinions representing 62 marketing authorisations.

Cancer treatment was the most-represented therapeutic area for which the COMP adopted positive orphan-designation opinions (46%). Almost two-thirds of designated orphan medicinal products were for conditions affecting children and the COMP took on average 66 days to evaluate applications— the same as in the previous year.

- **To explore additional incentives at national or European level to strengthen research into rare diseases and development of orphan medicinal products, and Member State familiarity with these products.**
- **A European guideline should clarify responsibilities in a Compassionate Use situation**
- **The Commission should present, a report to the Council and the Parliament identifying bottlenecks on orphan drugs access (delays, marketing, access, reimbursement, prices, etc.) proposing the necessary legislative modifications in order to guarantee equal access to orphan drugs throughout the EU on the basis of a COMP/EUCERD ²⁵ European collaborative scientific assessment (Commission, EMEA)**

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

The EU dilemma (I)

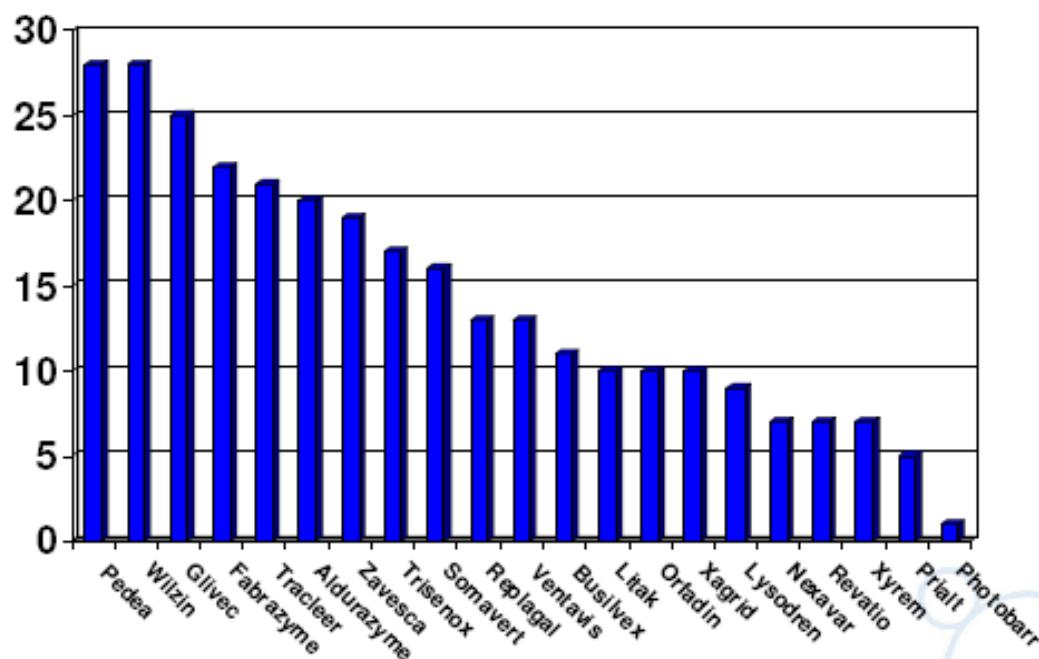
The regional divide

- *One* standard for drug approval
 - *One* application, *one* assessment
 - *One* decision valid in 27 EU + 3 EFTA countries
-
- “single payer”, but:
 - 30+ different HTA methodologies and interpretations
 - 30+ independent decisions about whether the medicine should be paid for

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

Availability by country- no.of countries



New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

- **Development of a medicine due to**
 - Low number of potential patients
 - Absence of patient registers
 - Lack of national centers and expertise
 - Heterogeneity of the diseases
 - Novel technologies/logistic requirements
- remains difficult and risky in spite of increased incentives**

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

How to improve equal access?

- **Exchange knowledge amongst MS on scientific assessment of the clinical added value of OD**
 - Improve connection between the NA and the EU processes
 - Improve and share the know-how to assess the clinical value
 - Common(non-binding)clinical added value assessment reports to facilitate the national pricing and reimbursement decisions ?
 - Central EU assessment procedure?

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

- EU level awareness and expertise on OD
 - Patient registers, centers of expertise and networks of centers of expertise can facilitate generation of additional data on benefits of the medicine in real life settings.



New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

Actions to ensure equal access to all EU patients to orphan drugs

- **A method for the assessment of the Clinical Added Value of Orphan Medicinal Products should perform a common scientific assessment of the CAV for each Orphan Drug and deliver an opinion document.**
- **Call for Tender tender EAHC/Health/2010/05 concerning the creation of a mechanism for the exchange of knowledge between Member States and European authorities on the scientific assessment of the clinical added value for orphan medicines (Launched in March 2010).**
- **The aim of these common assessment reports for CAVOD should be to provide a well-informed opinion on the place of the product with the authorised therapeutic indication in the therapeutic strategy of the rare condition, to the best of current knowledge. For this purpose detailed discussions with the Member States and interested parties, particularly the EMA and the COMP, on the best way to establish such mechanism are necessary.**

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level



Diagnostic tests are part of quality healthcare

Orphanet and EuroGentest provide information on available tests in Europe and surrounding countries



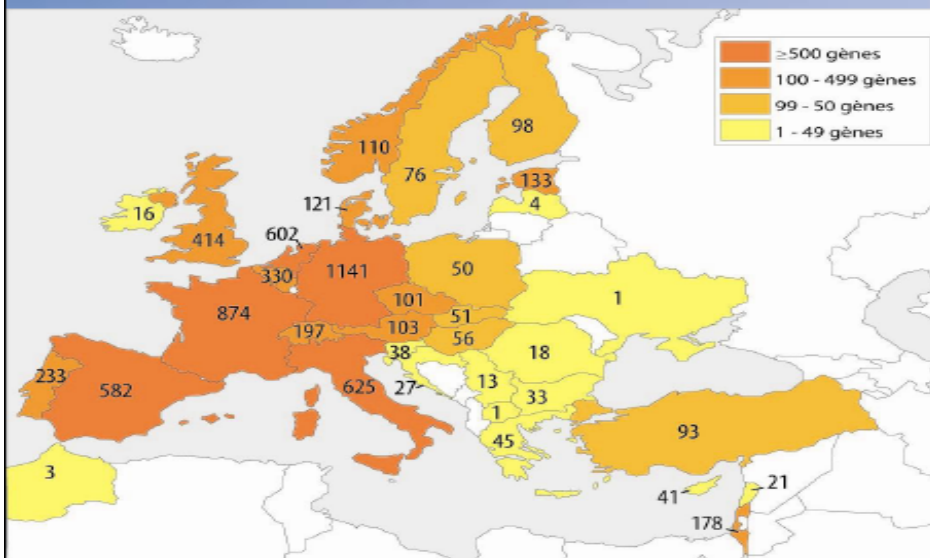
New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level

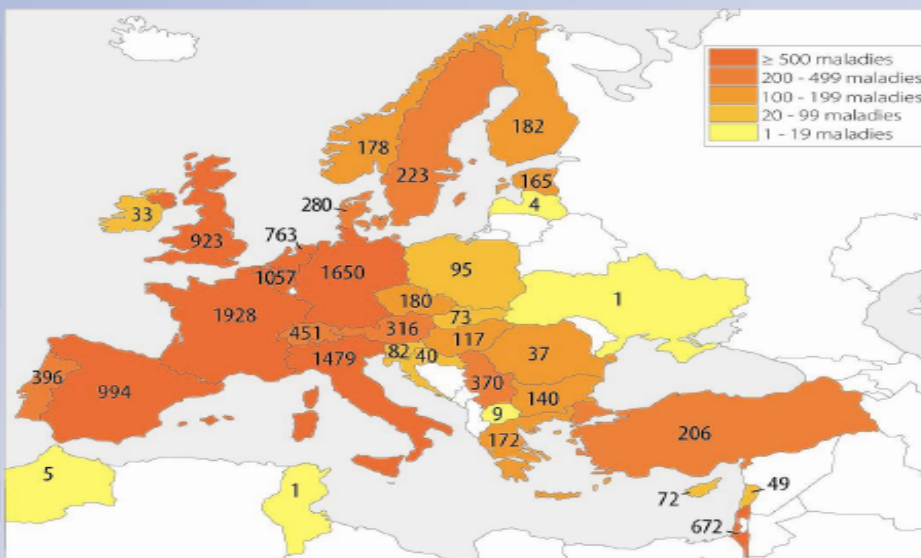


Major progresses in gene identification translated into diagnostic tests

Number of genes tested by country



Number of diseases tested by country



New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level



395 Registries as strategic tools

Number of patient registries per country

• France	103	• Ireland	9
• Germany	51	• Portugal	7
• Great Britain	50	• Switzerland	6
• Italy	47	• Greece	2
• Spain	28	• Bulgaria	4
• Belgium	19	• Denmark	3
• Netherlands	10	• Romania	2
• Austria	13		
		• Orphanet Report Series on Orphanet front page	

New priorities after Commission Communication and Council Recommendation

V. Gathering the expertise on rare diseases at European level



60 International Patient Registries around a medicinal product

- Cystic fibrosis
- Alpha 1 anti-trypsin
- Bleeding disorders
- Langerhans cell histiocytosis
- Severe chronic neutropenia
- Biliary atresia
- Neuromuscular diseases
- Wilson disease
- Fanconi anemia
- Pulmonary hypertension
- Metabolic diseases: Gaucher, Fabry, Pompe, MPS1...
- Ondine syndrome
- Primary immunodeficiencies
- Retinal dystrophies
- Huntington disease

New priorities after Commission Communication and Council Recommendation

VI. Empowerment of patient organisations

- **The Health Programme will continue to integrate the support to the patient's organisations as a priority for action.**
- **Operational Grants selected for EURORDIS in 2008 and 2009.**
- **The Council Recommendation suggests to Member States to do the same.**

New priorities after Commission Communication and Council Recommendation

VII. Sustainability

- **The Third Health Programme 2013-2017 (tbc) should integrate sustainability of rare diseases resources as a priority.**
- **The Member States should use facilities provided to health infrastructures in the Structural Funds (2007-2013) regulations.**
- **Continuity of efforts on rare diseases research in the FP8 integrating a sustainable dimension.**
- **Sustainability in the National Plans/Strategies for Rare Diseases.**

New priorities after Commission Communication and Council Recommendation

Actions to develop the international cooperation on rare diseases

- **An international cooperation framework on rare diseases with other countries (e.g. US, Canada, Japan, Singapore, Australia,...) will be adopted (Commission)**
- **A proposal of resolution on an international action in the field of rare diseases will be submitted by the European Commission to the World Health Assembly (Commission)**

New priorities after Commission Communication and Council Recommendation

Governance and monitoring

- **The Commission should be assisted by an EU Committee of Experts on Rare Diseases (EUCERD) to advise on implementation of the Communication and the Recommendation. The Committee will be chaired by the European Commission and will be assisted by a Scientific Secretariat, supported through the Health Programme. Composed by 51 members representing Member States, patient's organisations, Pharmaceutical industry, FP Projects, Health Programme projects and ECDC + 12 Commission representatives (SANCO, RTD, ENTR, EMA, COMP). **First meeting next 2nd December 2010 (tbc).****
- **This committee will replace the existing EU Rare Diseases Task Force.**
- **The organisation of a European Rare Diseases day (29 February, a rare day and 28th February in 2010) and European conferences (**the last European Conference on Rare Diseases, Krakow, 13-15 May 2010 and next one in Brussels 2012**) to raise awareness of professionals and of the general public will also be encouraged.**

DG SANCO priorities on rare diseases

Web site

http://ec.europa.eu/health/rare_diseases/policy/index_en.htm

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