# EU Policy on Rare Diseases

## Steering Committee of the European Partnership for Action Against Cancer

## Berlin, 21 March 2012

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# 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 benefit so much from collaboration at EU level. The reduced number of patients for these diseases and the need to mobilise resources require a co-ordinated European approach to be efficient.

# Legal basis for the developments of the EU Policy on rare diseases

**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.

# Legal basis for the developments of the EU Policy on rare diseases

**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.

**Decision of the Commission creating a European Union Committee of Experts on Rare Diseases**. To be composed by 51 members representing Member States, patient's organisations, industry, FP Projects, Health Programme projects, etc. Adopted 30th November 2009.

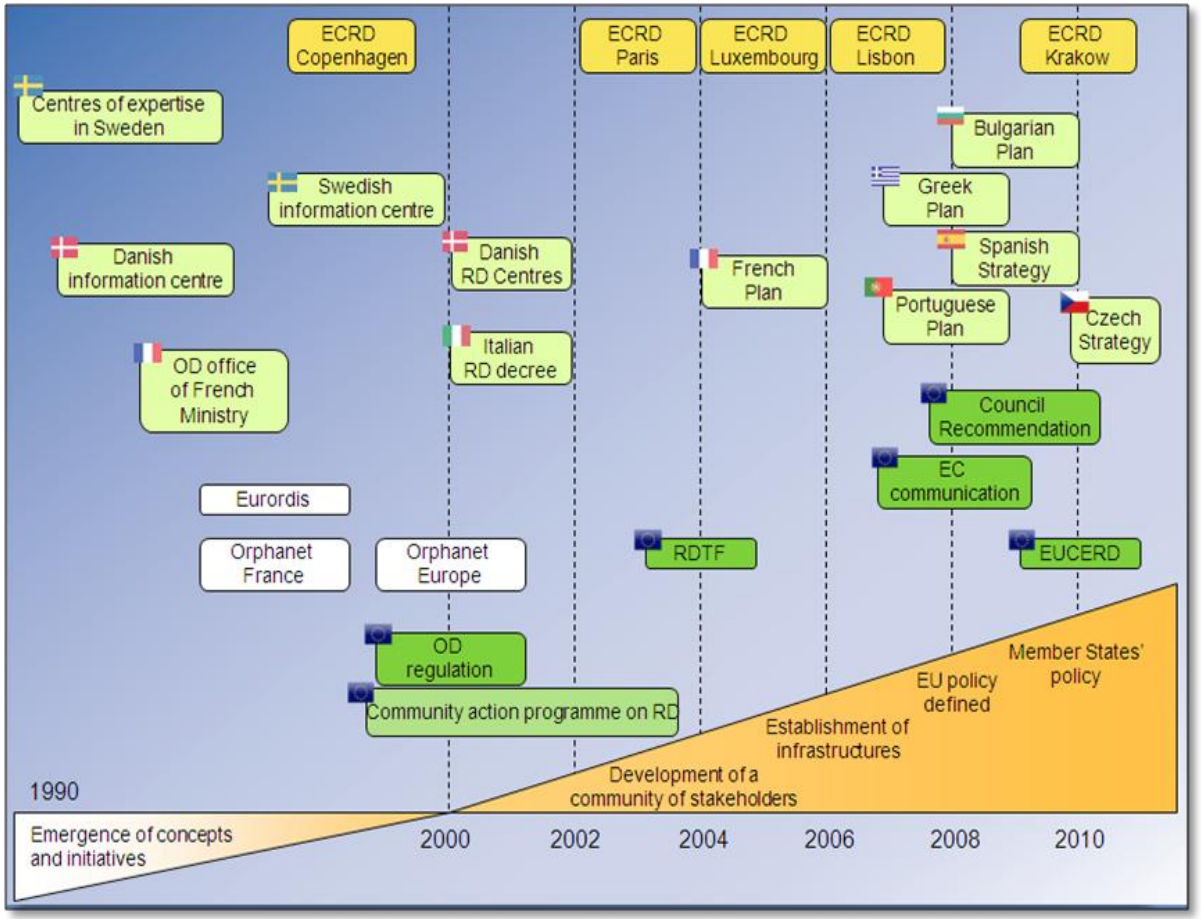
# Legal basis for the developments of the EU Policy on rare diseases

**Directive of the European Parliament and of the Council** of 9 March 2011 on the application of **patients' rights in cross-border healthcare** (2011/24/EU) provides for the development of European reference networks (ERNs) by Commission and Member States. The ERN can improve the access to diagnosis and the provision of high-quality healthcare to patients who have conditions requiring a particular concentration of resources or expertise, especially for rare diseases. Deadline for transposition the 23th of October of 2013.

**Directive 95/46/EC** of the European Parliament and of the Council of 24 October 1995 on the protection of individuals with regard to the processing of personal data on the free movement of such data. (Data Protection Directive).

**Directive 2005/28/EC** laying down principles and detailed guidelines for good clinical practice as regards investigational medicinal products for human use, as well as the requirements for authorisation of the manufacturing or importation of such products ("**clinical trials**")

# Emergence of concepts and initiatives surrounding rare diseases in Europe




From: Aymé S., Rodwell C., eds., "2011 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases - Part I: Overview of Rare Disease Activities in Europe and Key Developments in 2010", July 2011.

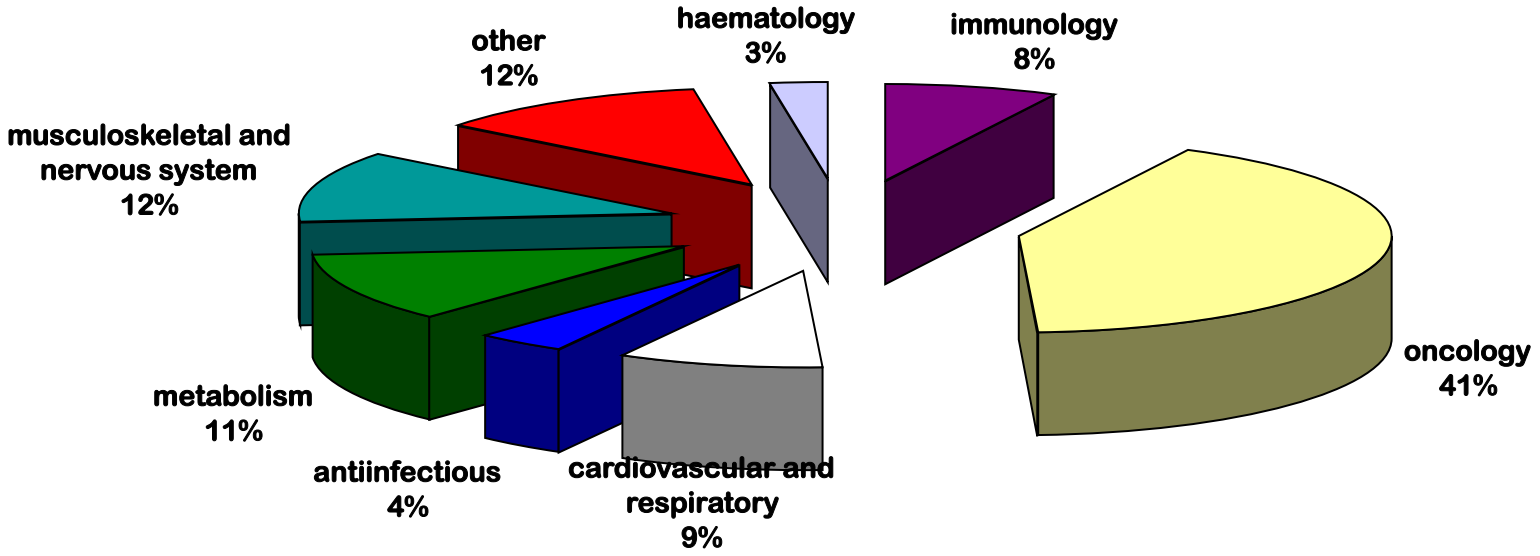
# Why an orphan regulation?

- Rare diseases -> developing and marketing cost would not be recovered by the expected sales
- Persons suffering from rare conditions deserve same quality of treatment as other patients
- Pharmaceutical industry does not develop medicines for rare diseases under normal market conditions
- Objective:
- - provide incentives that stimulate research and development (push)
- - modify market conditions (pull)

# Main incentives for orphan designation

- Economic / marketing
- Fee reduction / exemption
- Extended incentives for SMEs (post authorisation)
- Market exclusivity
- Product development
- Protocol assistance
- Community marketing authorisation
- National incentives (EC inventory)

# Distribution of designations



The Commission Communication and the Council Recommendation on rare diseases – Main priorities

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.

The Commission is assisted by an EU Committee of Experts on Rare Diseases (EUCERD) to advise on implementation of the Communication and the Recommendation.

The Committee is 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).

*http://www.eucerd.eu/*

# Adequate definition, codification and inventorying of rare diseases

- ICD -10 revision
- Information for patients and professionals
- Orphanet

EU Investing in research on rare diseases (RD) for more than 2 decades

FP5: 47 RD-relevant projects, € 64 million

ftp://ftp.cordis.europa.eu/pub/lifescihealth/docs/reprint_rec48300_rare_dis_060207.pdf

FP6: 59 RD-relevant projects; global budget: € 230 million

http://cordis.europa.eu/lifescihealth/major/rare-diseases-projects-1.htm


In the 4 first years of FP7: 50 projects relevant to rare diseases were supported in the Health theme, for a global budget of more than 237 million euro (including 27 projects for 110 million euro under section 2.4.4).

In 2011, 108 million euro earmarked for rare diseases in health research calls. Topics include:

- **Omics for rare diseases**
- **Preclinical/clinical development of orphan drugs**
- **Observational trials**
- **Best practice and knowledge sharing in the clinical management**

*http://ec.europa.eu/research/health/medical-research/rare-diseases/index_en.html*

# International Rare Disease Research Consortium (IRDiRC)

Launched in April 2011 by initiative of the European Commission, Health Directorate, DG Research and Innovation and the US National Institutes of Health, with the help of Canadian Institutes of Health Research, and interest from other research funding agencies in France, Germany, Italy, Spain and the United Kingdom.

By the year 2020, to deliver 200 new therapies for rare diseases and diagnostic tools for most rare diseases,

establish and provide access to harmonised data and samples,

perform the molecular and clinical characterisation of rare diseases, boost translational, preclinical and clinical research,

streamline ethical and regulatory procedures.

# IRDiRC objectives & milestones

Directorate for Research and Innovation

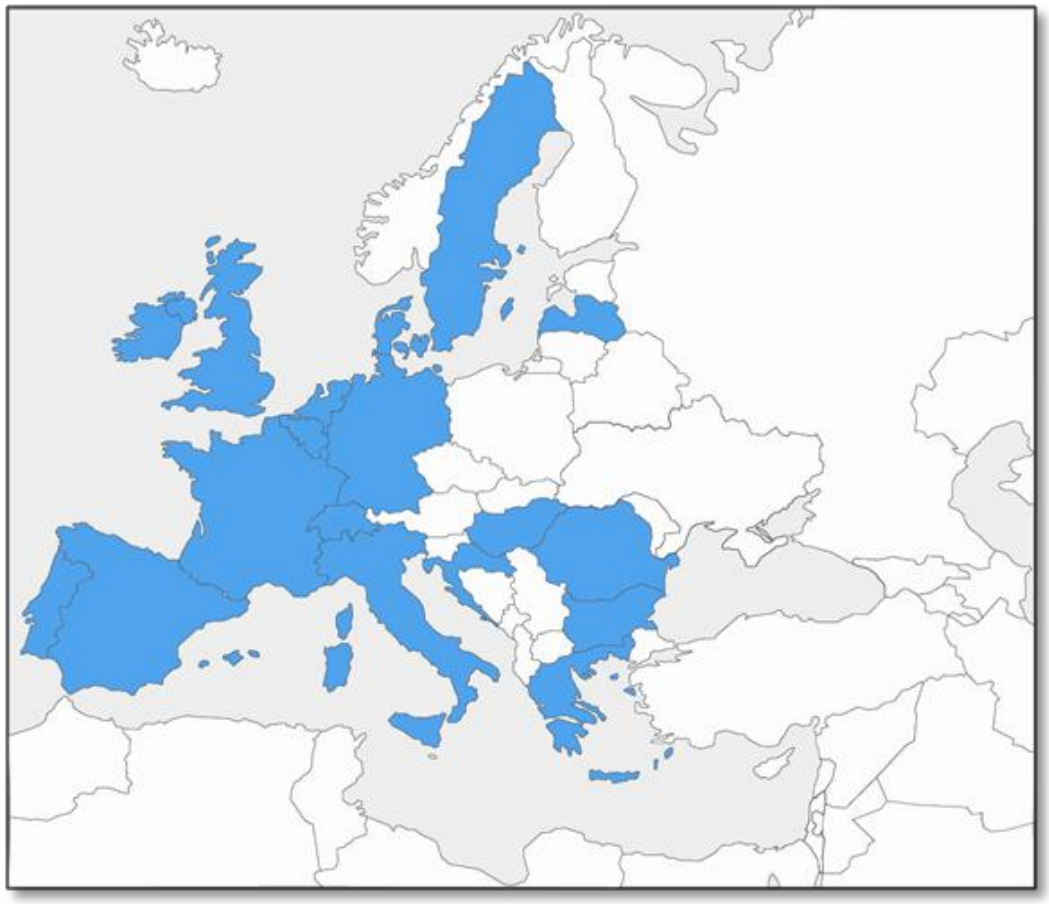
## Objectives

200 new therapies

Means to diagnose most rare diseases

| YEAR | Diagnostics | Therapies |
|---|---|---|
| 2012 | Mapping of sequencing & characterisation efforts<br>Identified & prioritised gaps in sequencing & diagnostics | Prioritisation of 100 new or repurposed therapies |
| 2015 | 5000 sequenced/characterised<br>3000 diagnostics | 50 new applications for market authorizations for new or repurposed therapies |
| 2020 | 6000 diagnostics | 200 new market authorizations given for new or repurposed therapies |

# Countries in Europe with a national alliance for rare disease patient organizations



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# Pilot European Reference networks

**Dyscerne**: European Network of Centres of Reference for Dysmorphology (ended)

**ECORN CF**: European Centres of Reference Network for Cystic Fibrosis (ended)

**PAAIR**: Patient Associations and Alpha1 International Registry (PAAIR) (ended)

**EPNET**: European Porphyria Network - providing better healthcare for patients and their families (ended)

**EN-RBD**: Establishment of a European Network of Rare Bleeding Disorders (ended)

**Paediatric Hodgkins Lymphoma Network**: European-wide organisation of quality controlled treatment (on-going)

**NEUROPED**: European Network of Reference for Rare Paediatric Neurological Diseases (ended)

**EURO HISTIO NET**: A reference network for Langerhans cell histiocytosis and associated syndrome in EU (on-going)

**TAG**: Improving Health Care and Social Support for Patients and Family affected by Severe Genodermatoses – TogetherAgainstGenodermatoses (on-going)

**CARE NMD**: Dissemination and Implementation of the Standards of Care for Duchene muscular Dystrophy in Europe (including Eastern countries) (on-going)

# Directive on the application of patients' rights in cross-border healthcare

The Directive intends to clarify patients' rights to access safe and good quality healthcare in another Member State (MS), and be reimbursed for it.

Increase transparency by making mandatory for MS and healthcare providers to make public comprehensive and accurate information on the services, the possible treatment options, the prices, and the quality and safety of the services they provide

This Directive will increase cooperation between national health authorities:

National Contact Points

Cross-border recognition of prescriptions

EU structures to implement projects on European reference, eHealth and health technology assessment networks

# Art 12. ERN

Art. 12 of the Directive notably foresees enhanced cooperation of Member States in the area of European reference networks (ERN).

Main goal is to facilitate improvements in the diagnosis and treatment of certain diseases of conditions across the EU:

By the delivery of high-quality, accessible and cost-effective healthcare

for patients suffering of medical conditions which could require a particular concentration of expertise or resources, particularly in medical domains where expertise is rare.

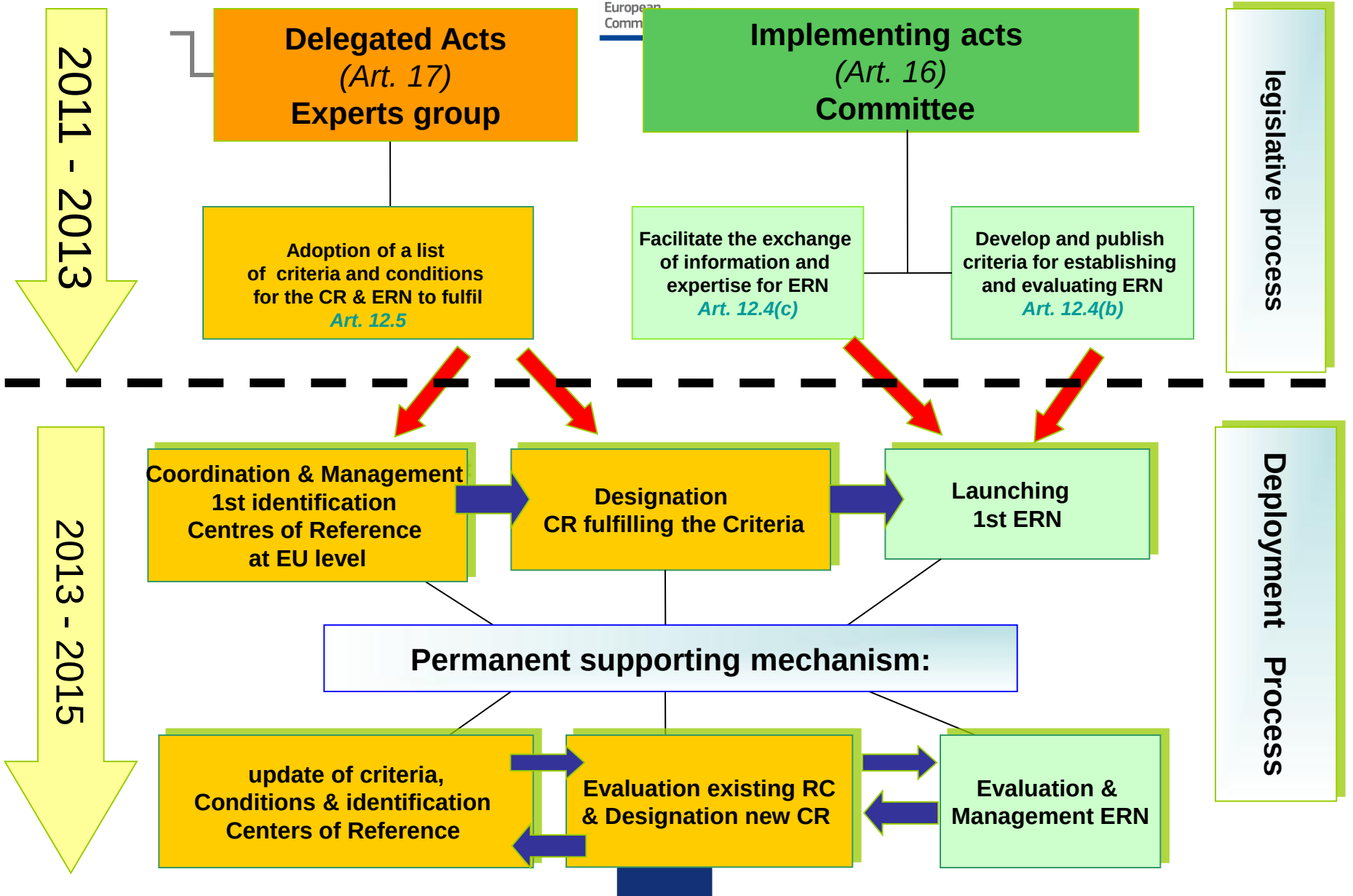
# Article 12 : ERN

The Commission shall support MS in the development of ERN between healthcare providers and Centres of expertise in the Member States

Participation in the ERN shall be voluntary. Its members shall participate and contribute to the networks' activities in accordance with the MS legislation where the members are established.

ERN shall be open to new healthcare providers which might wish to join them, provided that such healthcare providers fulfil all the required conditions and criteria

# Milestones and timeline for the implementation (ERN)



# EUCERD recommendation

Recommendations for Centres of Expertise adopted unanimously by the European Union Committee of Experts on Rare Diseases

Adopted on 24 of October 2011

# Directorate for Health and Consumers priorities on rare diseases

Web site

Public health actions

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

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