



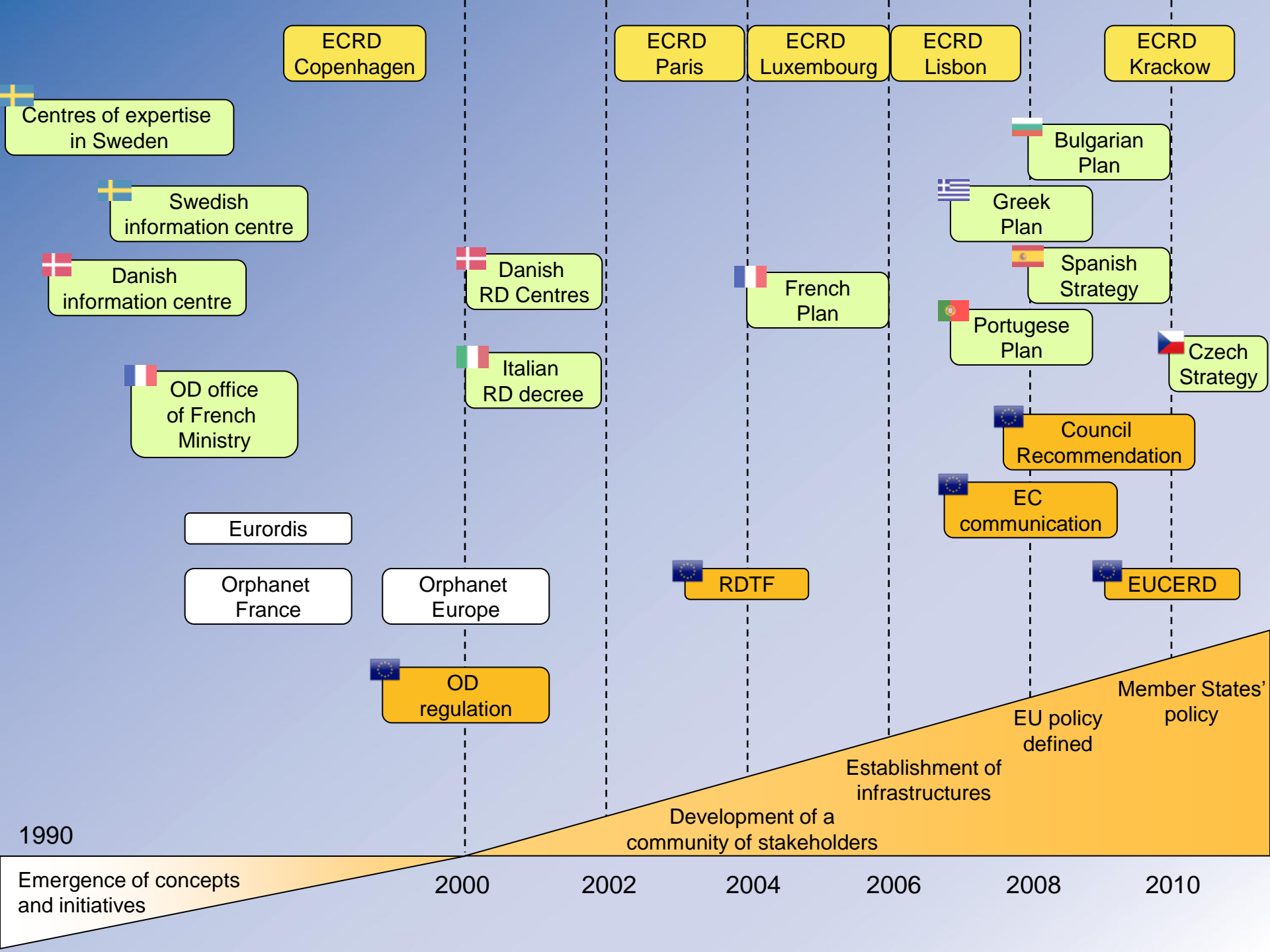
# **Developments in the Area of Rare Diseases**



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# EUCERD Mission

- Aid the European Commission with the preparation and implementation of Community activities in the field of rare diseases
- Cooperate and consult with the specialised bodies in **Member States**, the relevant **European authorities** in the fields of research and public health action and other **relevant stakeholders** acting in the field
- **Foster exchanges** of relevant experience, policies and practices between these parties



# EU policy development in Public Health

January 2004

- **Rare Disease Task Force** ([www.rdtf.org](http://www.rdtf.org))
  - Working groups on Coding and Classification, on Indicators and on Standards of Care
  - OrphaNews Europe: 12,000 readers every two weeks

11 November 2008

- **EC Communication** « Rare Diseases: Europe's challenge »

9 June 2009

- « **Council Recommendation** on an action in the field of rare diseases »

30 November 2009

- **European Union Committee of Experts in Rare Diseases:**
  - 51 representatives of MS, of EC DGs, of patients, of experts, of the Industry
  - Chair: S. Aymé – Vice-Chairs: Kate Bushby, Yann Le Cam, Helena Kaariainen

# EUCERD Responsibilities

- Monitoring, evaluating and disseminating the results of measures taken at Community and national level
- Contributing to the preparation of Commission reports on the implementation of the Commission Communication and the Council Recommendation
- Delivering opinions, recommendations or reports to the Commission either at the latter's request or on its own initiative
- Assisting the Commission in international cooperation on matters relating to rare diseases
- Assisting the Commission in drawing up guidelines, recommendations



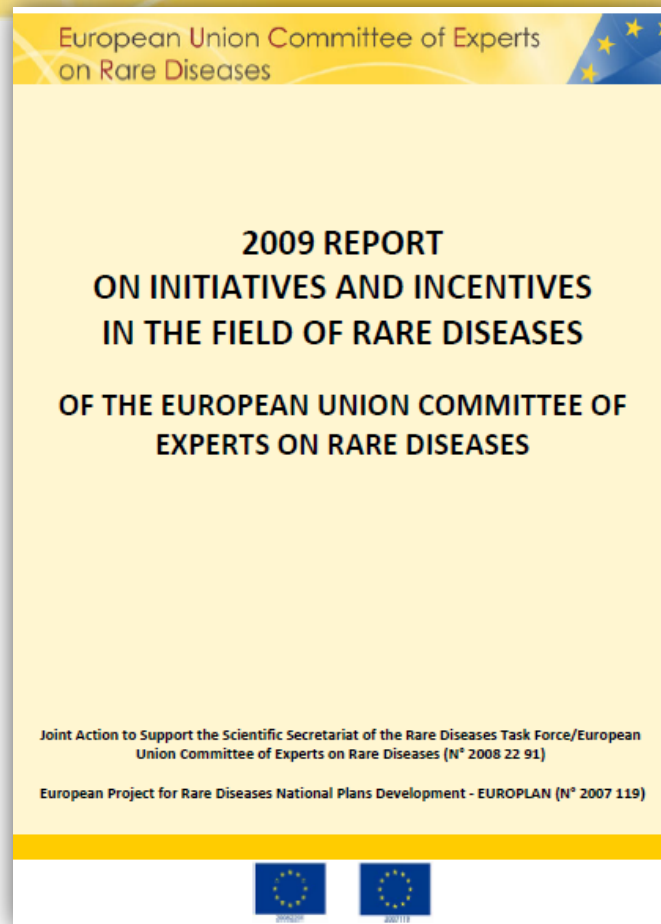
# OrphaNews Europe



- Currently financed by the Joint Action and AFM
- Over 80 issues since June 2005
- Published twice a month
- Over 12 000 subscribed readers
- 10-15 articles on policy/projects/social developments per newsletter
- 20-25 scientific articles per newsletter
- News from 25 countries in 2010
- Satisfaction survey in 2010 – 98% satisfaction of respondents
- Developments foreseen: RSS, full text search options, personalised subscription



# Report on initiatives and incentives 2009



[www.eucerd.eu](http://www.eucerd.eu)

# National Centres of Expertise and European Reference Networks





# Political context

- Centres of expertise highlighted in
  - *Communication « Rare Diseases: Europe's challenge » of 11 November 2008*
  - *Recommendations of the Council of 8 June 2009*
- Strong agreement that CE should be established in the frame of national policy for RD
- No analysis so far of past experience in MS where there are CE in place

# Conclusion on Centers of Expertise



- Without CE of national level, no possibility for networks
- Respect responsibilities of MS for organisation, financing and delivery of healthcare
- Difficulties:
  - diversity of health care systems and economies
  - Large/ medium/ small size countries

# Concepts in place for European Reference Networks



- Networking is about collaboration and sharing
- Current networks are networks of experts, not of centres
- Sharing expertise
  - Case management /Tele-expertise
  - Production of standards of care/ information packages / training / education
  - Multidisciplinary clinical research
- Sharing tools
  - ICT tools for sharing expertise
  - ICT tools for disseminating
  - Databases /cohorts/ biobanks

# Key documents

- Overview of Current Centres of Reference on Rare Diseases in the EU - September 2005
- RDTF Report: Centres of Reference for Rare Diseases in Europe – State-of-the-Art – September 2006
- Assessing the European Added-Value of European Reference Networks – March 2008
- Centres of Expertise and European Reference Networks - December 2010
- Preliminary Analysis of the Outcomes and Experiences of Pilot European Reference Networks – February 2011

# Patient Registries and Indicators



# Objectives

- State of Art of registration of patient data in Europe
- Issue recommendations on registry establishment, management and funding
- Identify indicators which could be derived from registries to monitor health outcomes
  - In Europe and in Member States where possible
  - To follow-up on trends

# Key Documents

- EUCERD Reports
  - Health Indicators for Rare Diseases: State of the art and future directions - March 2008
  - Registries as sources of Health Indicators for rare diseases - March 2011 (in preparation)
- Orphanet Report Series
  - Disease Registries in Europe – January 2011

# Rare Diseases in Health Information Systems: Coding and Classification Issue





# Aims and Achievements

- Aims
  - to introduce RD in the next edition of the International Classification of Diseases
  - To cross reference the Orphanet nomenclature with MedDRA, MeSH and SNOMED-CT
- Achievements
  - ICD 11 will incorporate RD: Alpha draft for April 2011 and Beta draft for 2012
  - Cross referencing achieved by Dec 2011
  - Orphanet nomenclature as a freely accessible resource by April 2011

# EUCERD Road Map



# Activities in 2011

- **Maintain activities already funded**
  - **Workshops planned**
    - Centres of Expertise : Luxembourg, 21-22 March 2011
    - Patient Registries Versus Product Registries : London, EMA, Autumn 2011
    - Coding, classification and sources of clinical data: Montpellier, 4 November 2011 (Conference open to the Public)
  - **Reports to be published**
    - Centres of Expertise in Europe: Possible models for expert care
    - Disease registries: a field for public-private partnership
    - State of Art of Rare Diseases in Europe in 2011

# Areas for Action

- Implementation of plans and strategies
- Adequate definition, codification and inventorying of RD
- Development of R&D: precompetitive platforms / public/private partnership
- Promotion of Expert Centres and European collaboration
- Social services for patients
- Ensure quality services in the field of testing
- Develop training courses on transversal themes
- Develop benchmarking
- Improve communication and dissemination

Thank you for your attention!

