



DISCUSSION on Evidence synthesis

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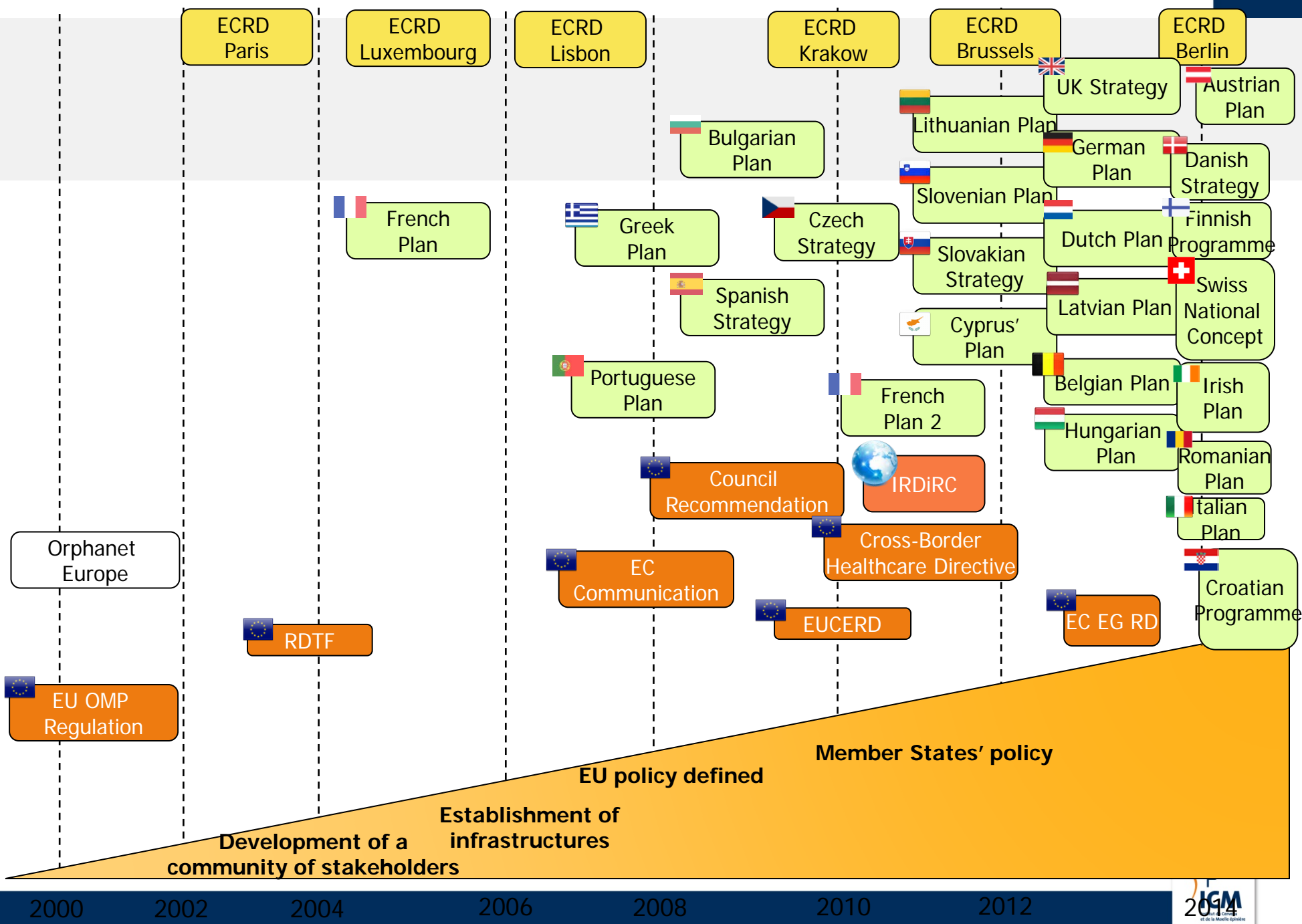
INSERM, Paris, France

Brain and Spine Institute, Salpêtrière Hospital

Workshop on Small population Studie
EMA March 2017

▶ The Community of rare diseases has a long history of close collaboration between stakeholders to identify solutions to problem caused by rarity





National Centres of Expertise and ERN

- ▶ **Recommendations on Quality Criteria for National Centres of Expertise**
- ▶ **Recommendations on European Reference Networks between Centres**



Establishment of ERN

- ▶ **Cost-effective use of resources**
- ▶ **Need for highly specialised healthcare Centres demonstrating:**
 - ↪ Competence and experience
 - ↪ specific human resources
 - ↪ structural and equipment resources
 - ↪ appropriate organisation

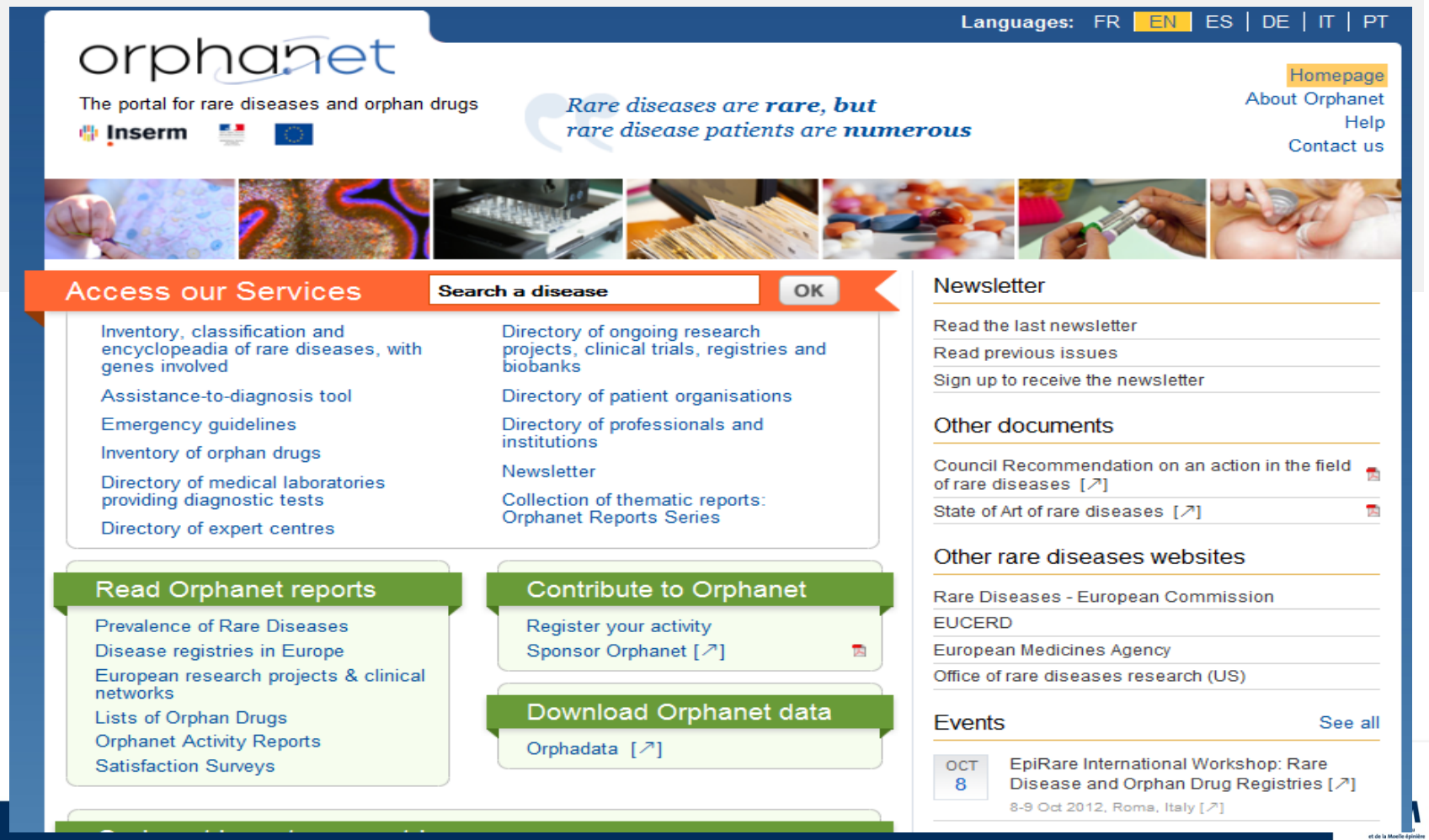
Scope of 22 European Networks for RD

Rare Bone Disorders
Rare craniofacial anomalies and ENT disorders
Rare Congenital Malformations and Rare Intellectual Disability
Rare inherited and congenital anomalies
Rare Endocrine Conditions
Rare Kidney Diseases Reference Network
Rare Neurological Diseases
Rare Neuromuscular Diseases
Rare and Complex Epilepsies
Rare Respiratory Diseases

Rare Skin Disorders Rare Eye Diseases
Rare Diseases of the Heart
Rare Hereditary Metabolic Disorders
Rare Hematological Diseases
Rare Hepatological Diseases
Rare Connective Tissue and Musculoskeletal Diseases
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases
Rare Multisystemic Vascular Diseases
Rare Adult Cancers (solid tumors)
Paediatric Cancer
Genetic Tumour Risk Syndromes

Dissemination of Information

WWW.ORPHA.NET



The screenshot shows the Orphanet website homepage. At the top, there's a navigation bar with language options: FR, EN (highlighted), ES, DE, IT, PT. The Orphanet logo is on the left, with the tagline 'The portal for rare diseases and orphan drugs' and logos for Inserm, France, and the European Union. A quote on the right says 'Rare diseases are rare, but rare disease patients are numerous'. Below this is a row of seven images related to rare diseases. The main content area is divided into several sections: 'Access our Services' with a search bar and a list of services; 'Read Orphanet reports' with a list of reports; 'Contribute to Orphanet' with a list of contribution options; 'Download Orphanet data' with a link to Orphadata; 'Newsletter' with links to read the last newsletter, previous issues, and sign up; 'Other documents' with links to Council Recommendation and State of Art of rare diseases; 'Other rare diseases websites' with links to various organizations; and 'Events' with a link to see all events and a specific event for October 8th.

orphanet
The portal for rare diseases and orphan drugs
Inserm France European Union

Rare diseases are rare, but rare disease patients are numerous

Homepage
About Orphanet
Help
Contact us

Access our Services Search a disease OK

- Inventory, classification and encyclopaedia of rare diseases, with genes involved
- Assistance-to-diagnosis tool
- Emergency guidelines
- Inventory of orphan drugs
- Directory of medical laboratories providing diagnostic tests
- Directory of expert centres
- Directory of ongoing research projects, clinical trials, registries and biobanks
- Directory of patient organisations
- Directory of professionals and institutions
- Newsletter
- Collection of thematic reports: Orphanet Reports Series

Read Orphanet reports

- Prevalence of Rare Diseases
- Disease registries in Europe
- European research projects & clinical networks
- Lists of Orphan Drugs
- Orphanet Activity Reports
- Satisfaction Surveys

Contribute to Orphanet

- Register your activity
- Sponsor Orphanet [↗]

Download Orphanet data

- Orphadata [↗]

Newsletter

- Read the last newsletter
- Read previous issues
- Sign up to receive the newsletter

Other documents

- Council Recommendation on an action in the field of rare diseases [↗]
- State of Art of rare diseases [↗]

Other rare diseases websites

- Rare Diseases - European Commission
- EUCERD
- European Medicines Agency
- Office of rare diseases research (US)

Events See all

- OCT 8 EpiRare International Workshop: Rare Disease and Orphan Drug Registries [↗]
8-9 Oct 2012, Roma, Italy [↗]

Dissemination of political and scientific information

- 20 issues yearly
- 25,000 readers
- Political news
- Scientific news



Codification of Rare Diseases

- ▶ Rare Diseases will be in ICD 11 to be published in 2018
- ▶ Release by Orphanet of an Inventory of Rare Diseases with classification
 - ↳ Multiple hierarchies
 - ↳ Linearisations by specialty
- ▶ Recommendation of the Expert Group on RD to use the Orphanet nomenclature in health information systems

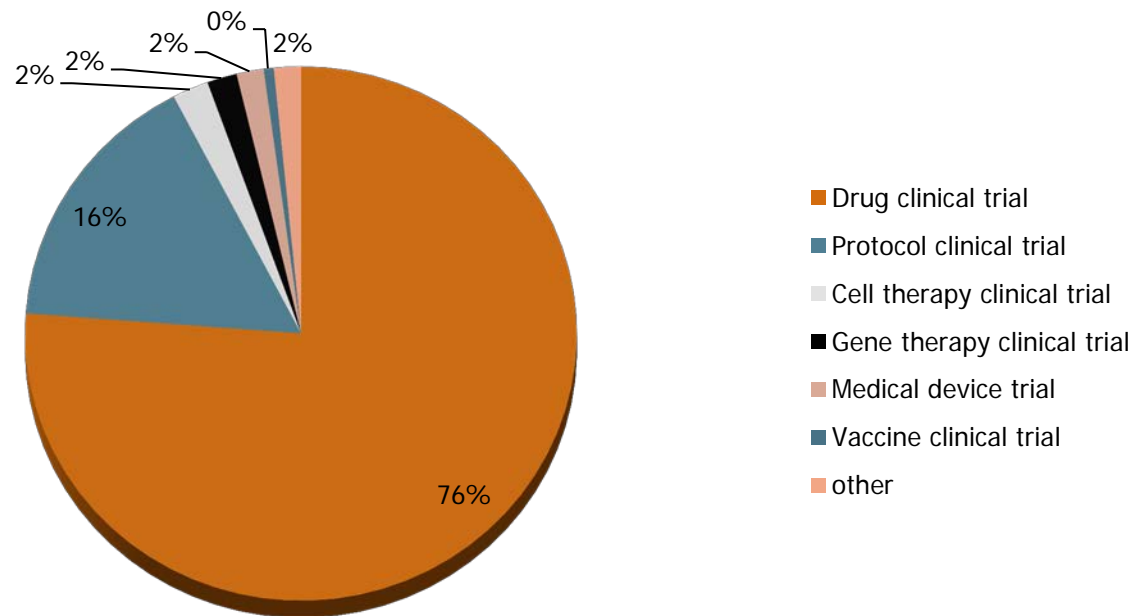
Support to the establishment of Disease Registries

- ▶ Registries are key element of national plans/strategies
- ▶ Need for disease registries
 - ↳ For clinical research purpose
 - ↳ For monitoring interventions
- ▶ Need to code rare diseases in electronic health records

Funding of R&D since 2000

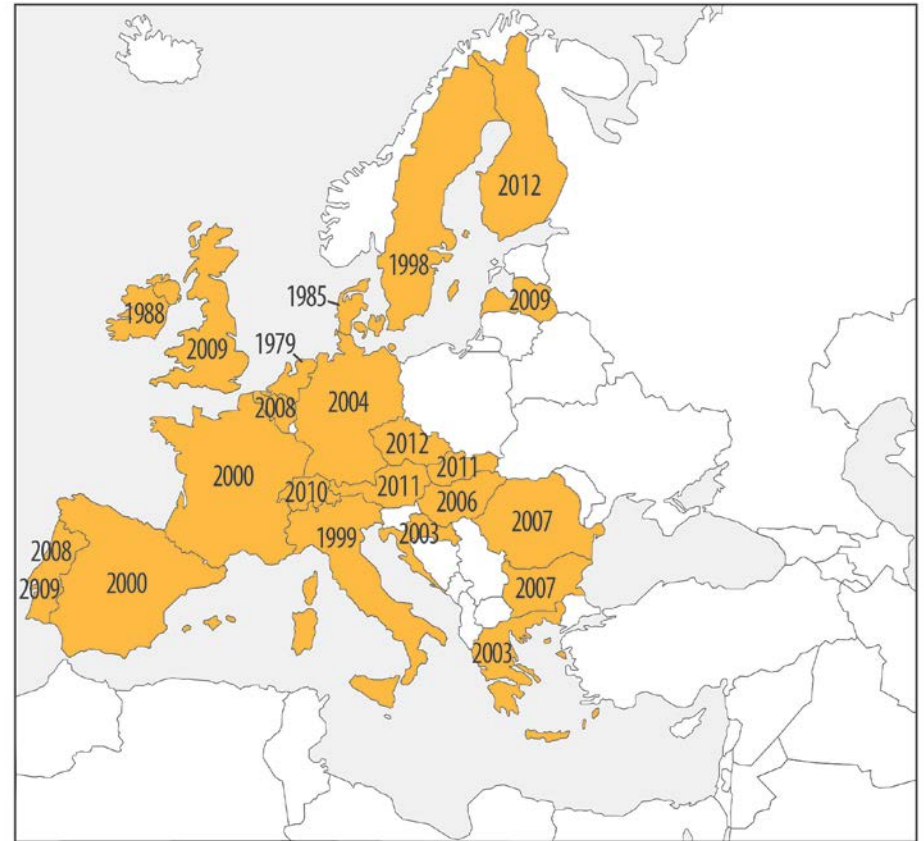
► 1829 ongoing national or international clinical trials for 828 diseases in 29 countries (Orphanet data – December 2016)

Percentage of clinical trials by category



Key role played by > 2,300 Patient organisations

- ▶ Very articulate and knowledgeable patients
- ▶ Involved at all level from Research to Services and in regulatory and political processes
- ▶ Coordinated by EURORDIS in Europe
- ▶ Organised at country level



Conclusion

- ▶ **Very dedicated community**
- ▶ **Progresses go in the right direction**
 - ↳ Far too slow for patients
 - ↳ Major inequality between countries
- ▶ **New experiment of healthcare organisation**
- ▶ **Partnership between countries**
- ▶ **Pragmatism and proactivism**

Data are accumulated but not analyzed

- ▶ Chronic diseases mean stages with different mechanisms
- ▶ Markers of evolution +++
- ▶ Multi criteria analysis
- ▶ More efforts on analysis (open access)
- ▶ More money on exploitation than collection

CONCLUSION

- ▶ **Learn more from experience**

- ↪ Workshop on past trial designs by homogenous group of diseases

- ▶ **Trials cannot be the first attempt to document the natural history**

- ▶ **More pragmatism to deliver for patients**



You are under scrutiny

