DISCUSSION
on Evidence synthesis

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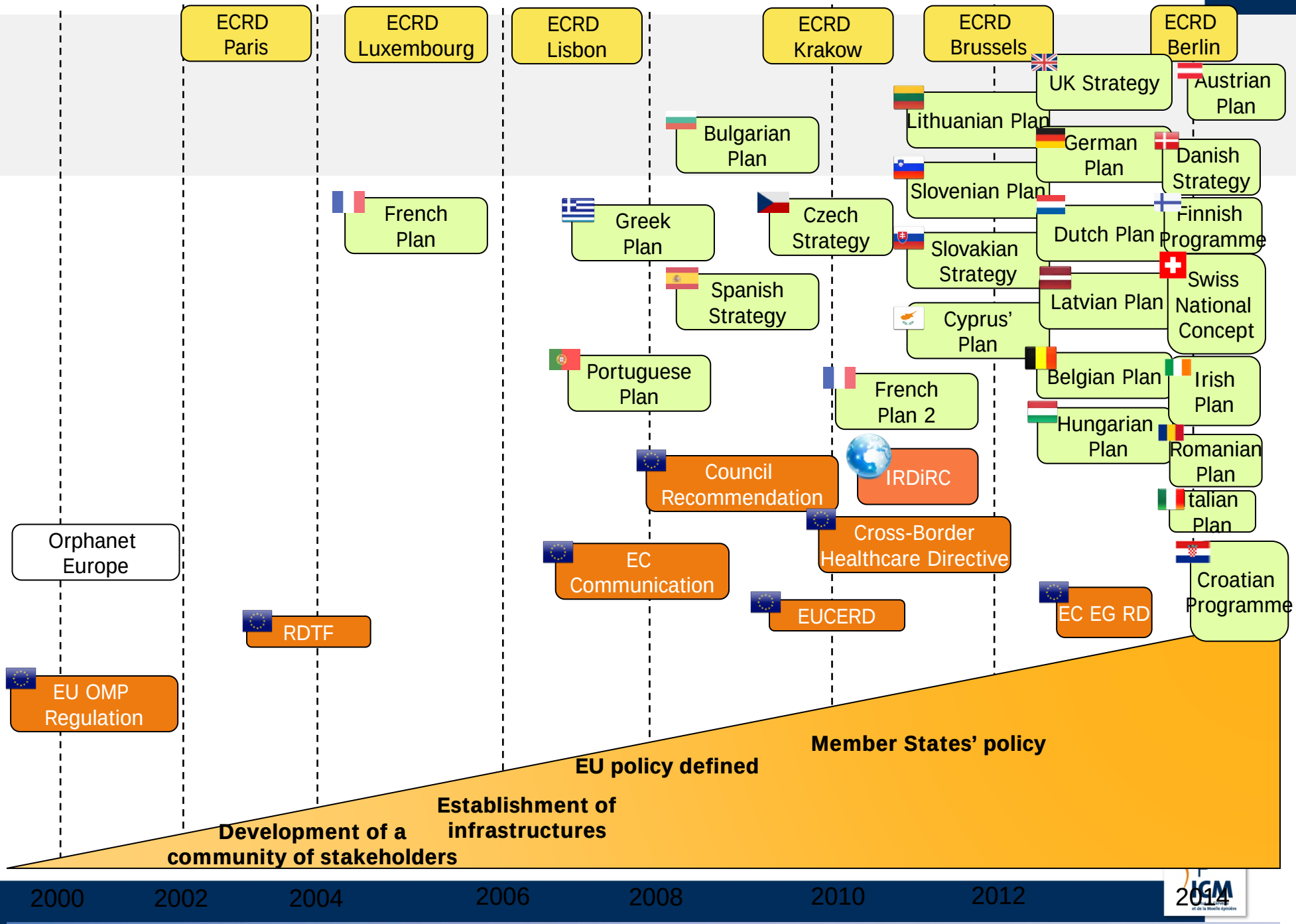
The Community of rare diseases
has a long history of close collaboration
between stakeholders to identify solutions
to problem caused by rarity


ECRD Paris
ECRD Luxembourg
ECRD Lisbon
ECRD Krakow
ECRD Brussels
ECRD Berlin
UK Strategy
Austrian Plan
Lithuanian Plan
Bulgarian Plan
German Plan
Danish Strategy
Slovenian Plan
French Plan
Greek Plan
Czech Strategy
Dutch Plan
Finnish Programme
Slovakian Strategy
Spanish Strategy
Latvian Plan
Swiss National Concept
Cyprus' Plan
Portuguese Plan
Belgian Plan
Irish Plan
French Plan 2
Hungarian Plan
Romanian Plan
IRDiRC
Council Recommendation
Italian Plan
Cross-Border Healthcare Directive
Orphanet Europe
EC Communication
Croatian Programme
EUCERD
EC EG RD
RDTF
EU OMP Regulation
Member States' policy
EU policy defined
Establishment of infrastructures
Development of a community of stakeholders
2000
2002
2004
2006
2008
2010
2012
2014

National Centres of Expertise and ERN

- **Recommendations on Quality Criteria for National Centres of Expertise**

- **Recommendations on European Reference Networks between Centres**

Establisment of ERN

European Reference Networks

Network

Member

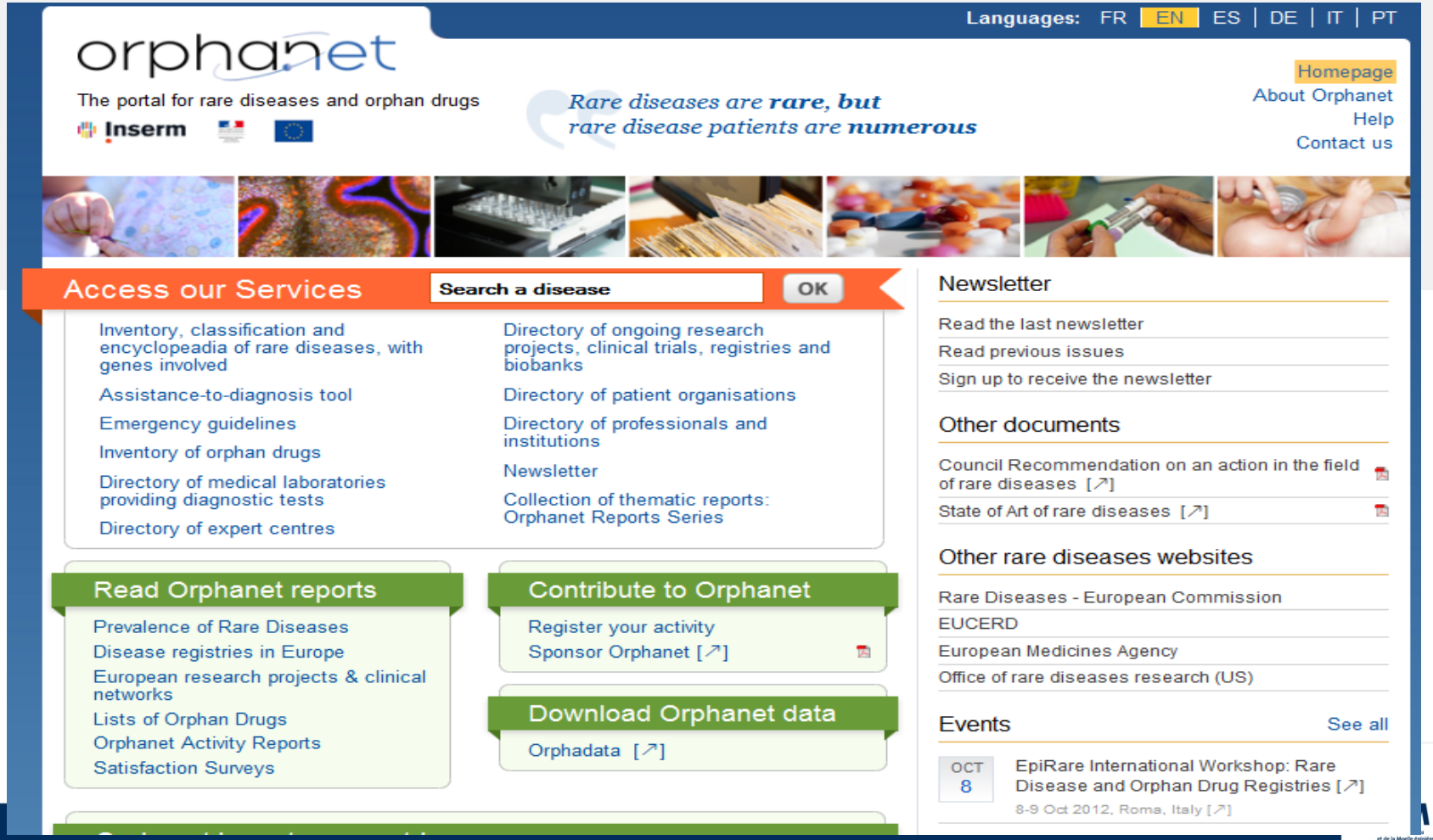
- **Cost-effective use of resources**
- **Need for highly specialised healthcare Centres demonstrating:**
  - Competence and experience
  - specific human resources
  - structural and equipment resources
  - appropriate organisation

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



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

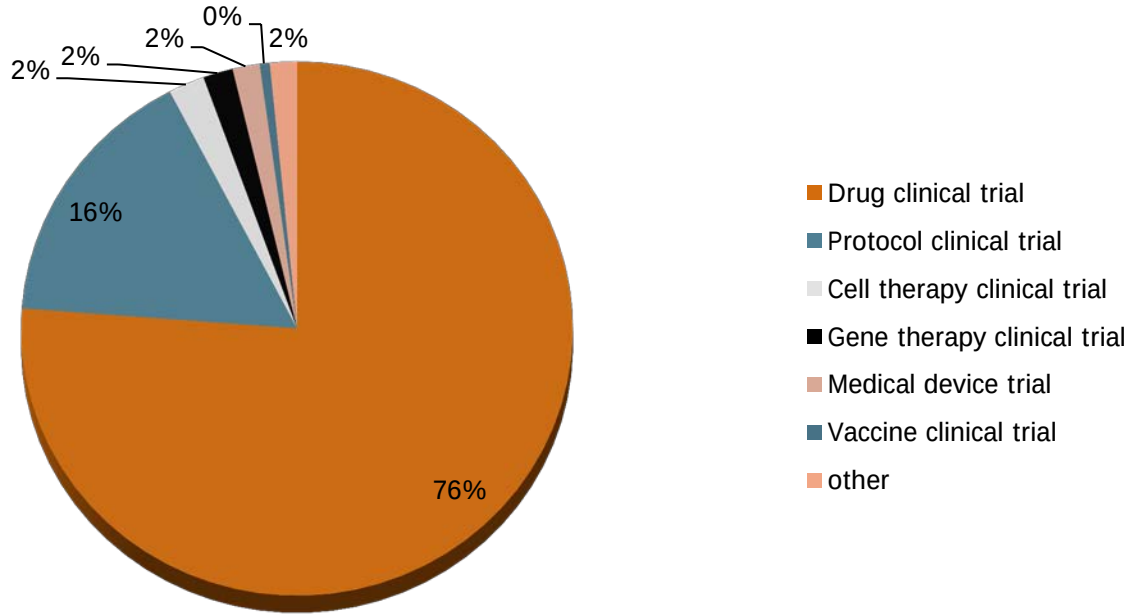
Support to the establishment of Disease Registries

- 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

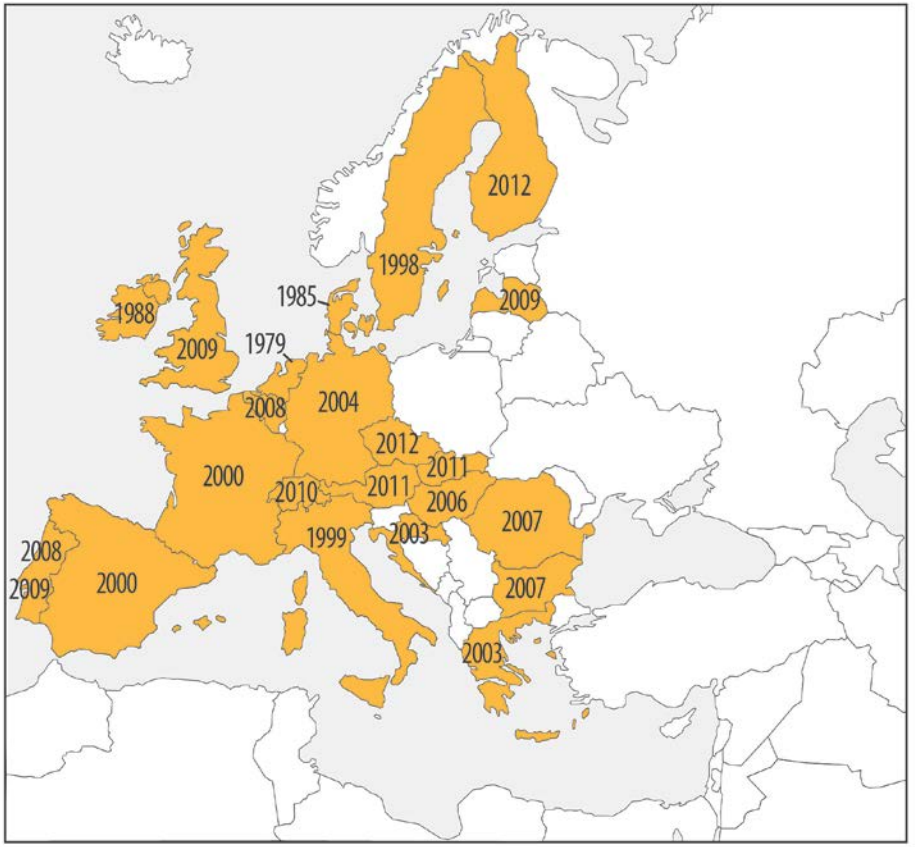
- 1829 ongoing national or international clinical trials for 828 diseases in 29 countries (Orphanet data – December 2016)

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 analized

- 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 scrutinity