European Policy in the field of Rare Disease to improve Patient Care and boost Research

Shenzhen, IRDiRC Conference 2014

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European Union Committee of Experts on Rare Diseases

A case for action

RD are individually rare
but patients are numerous
as there are many RD…..

Patients with RD deserve the same
attention as other patients

RD: a case for Health Care Policy

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Emergence of the concept of RD

- USA:
  - 1983:Orphan drug regulation
  - 1993: Office of Rare Diseases
- EU:
  - 1985 : Denmark – Rare Disorders Denmak
  - 1989: Sweden – Centres of reference and Agrenska
  - 1995:France – Office of Orphan drugs-
  - 1996: Orphanet
  - 1997: EURORDIS
  - 1998: Italy – Priority in Public Health
  - 2000 : EU Regulation on **Orphan Medicinal Products**

EU Public Health Policy for Rare Diseases

- 1999-2007:
  - Rare diseases as one of the priorities in **Public Health**
  - Target: improving knowledge and facilitating access to **information**
    - Orphanet Europe established in 2000

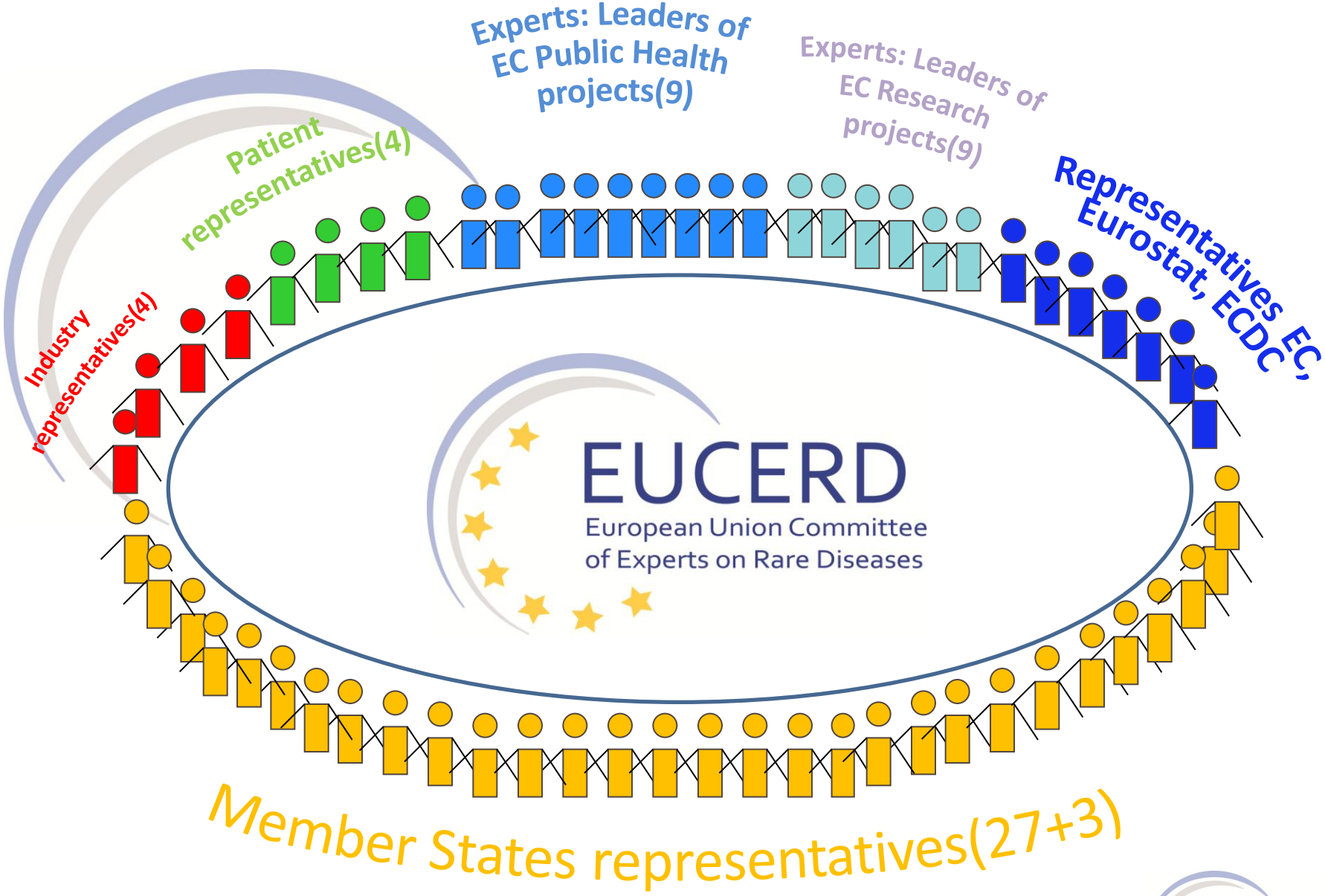
- 2008-2013:
  - Exchange of information via existing **networks**
  - Strategies and mechanisms to encourage transnational **cooperation**: European Reference networks of Centres of Expertise

EU policy in Public Health

- **11 Nov 2008: EC Communication** « Rare Diseases: Europe's challenge »

- **9 June 2009** « **Council Recommendation** on an action in the field of rare diseases »
  - All countries should have a plan or a strategy by the end of 2013

- **European Union Committee of Experts in Rare Diseases:**
  - 51 representatives of Minitries of Health, of patients, of experts, of Industry representatives



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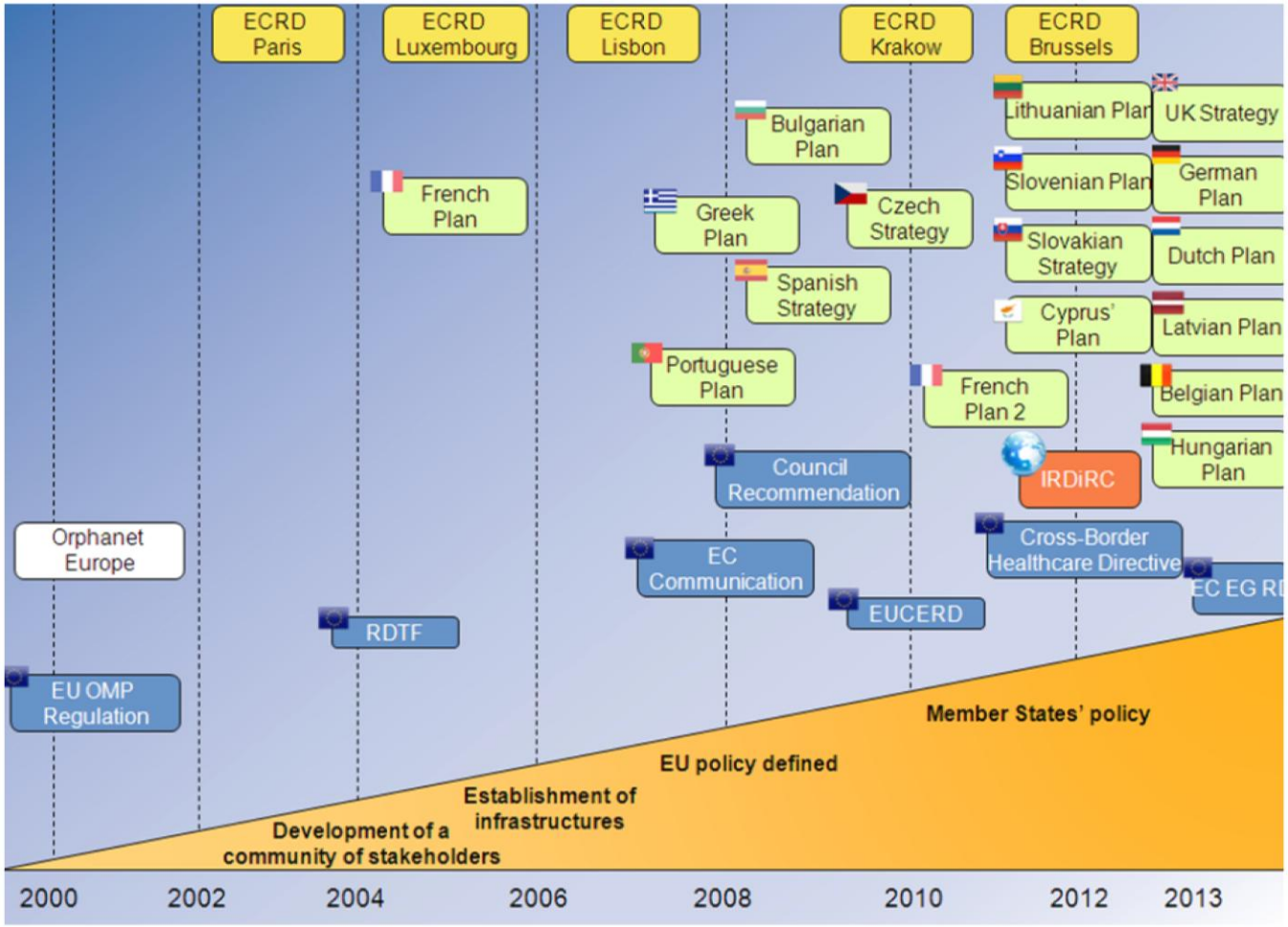
National Plans in 2014



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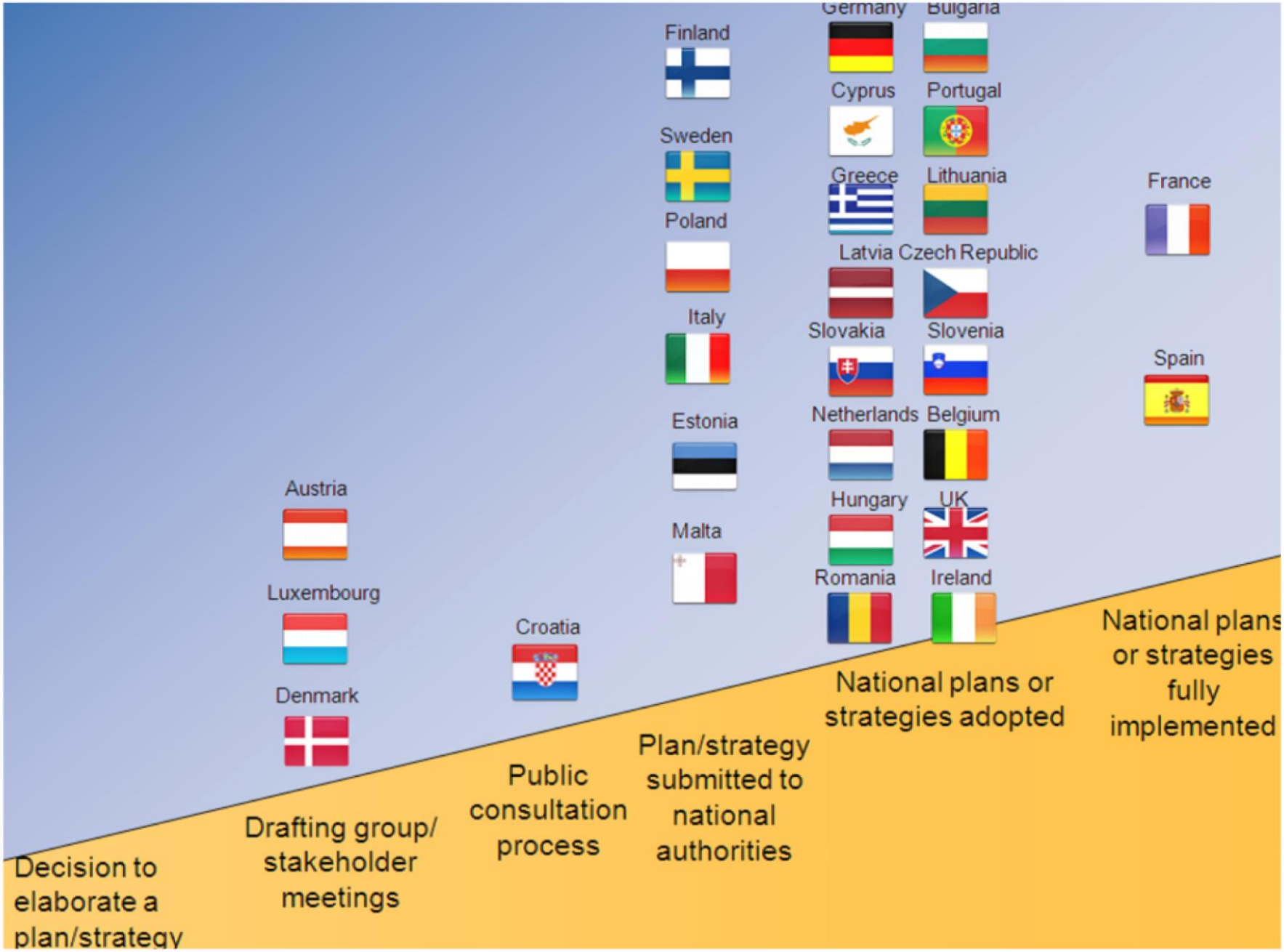
Finland
Germany
Bulgaria
Cyprus
Portugal
Sweden
Greece
Lithuania
France
Poland
Latvia
Czech Republic
Italy
Slovakia
Slovenia
Spain
Estonia
Netherlands
Belgium
Austria
Hungary
UK
Malta
Luxembourg
Romania
Ireland
Croatia
Denmark
National plans or strategies fully implemented
National plans or strategies adopted
Plan/strategy submitted to national authorities
Public consultation process
Drafting group/ stakeholder meetings
Decision to elaborate a plan/strategy

2014 EUCERD Report on the State of the Art of Rare Disease Activities in Europe


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2013 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

This work was financed by the EUCERD Joint Action: Working for Rare Diseases N° 2011 22 01

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RD: a case for Research Policy

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EU policy development in Research

- 2000: RD as priority diseases for funding
- 2004: Common call for proposals among voluntary countries
- 2011: Establishment of the International Rare Diseases Research Consortium (IRDiRC)
- 2013: EC funding for a European Platform for registries in the field of rare diseases
- 2014: Recommendation to use Orphacodes to code rare diseases in addition to ICD10

Empowered Patient Organisations EURORDIS a single voice in Europe

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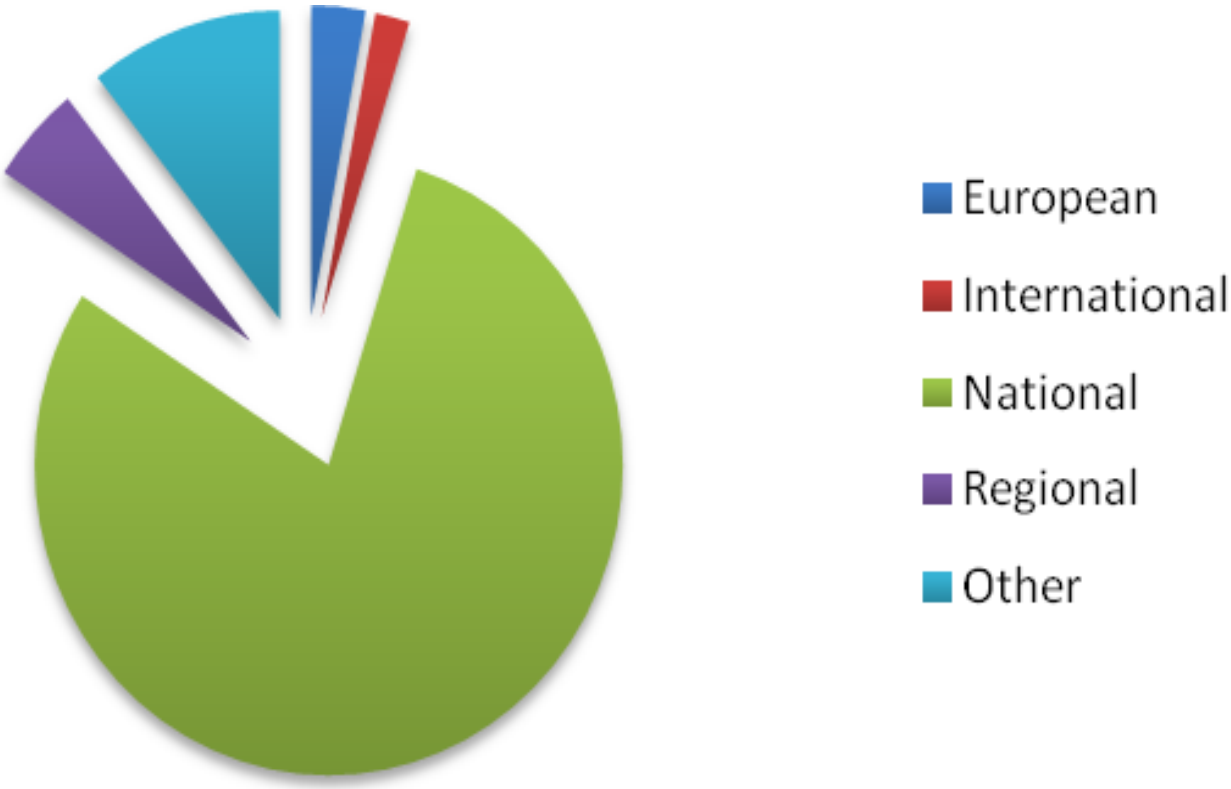
2,392 disease-specific patient organisations



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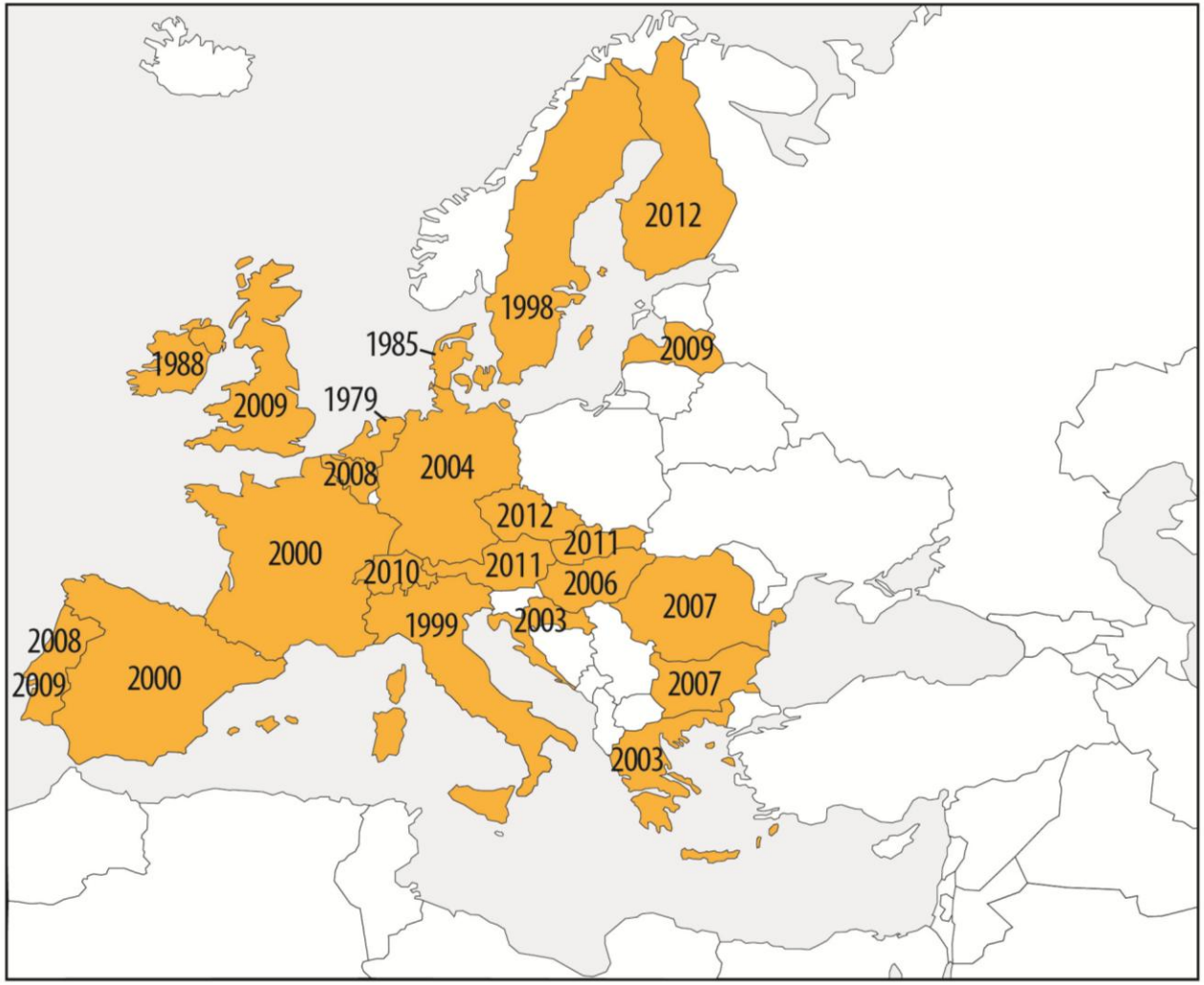
National alliance for rare disease patient organisations



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Major achievements so far

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National Centres of Expertise

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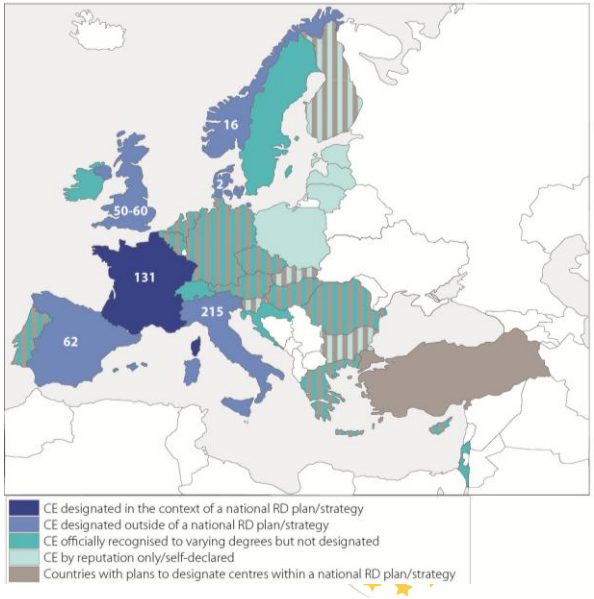
Establishment of Centres of Expertise in each country

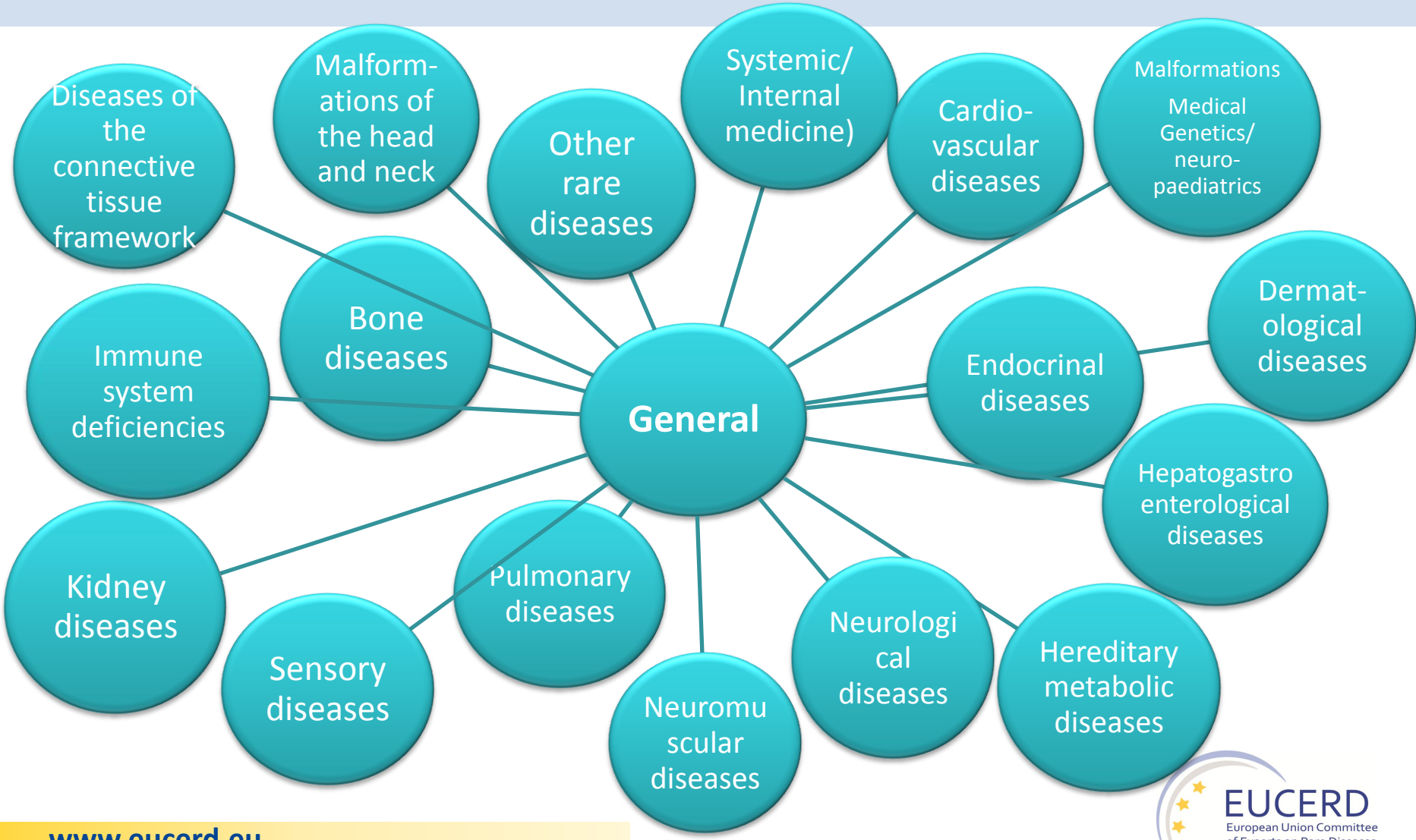
- EUCERD Recommendations on Quality Criteria for National Centres of Expertise
  - www.eucerd.eu
  - Process to identify them
  - Organisation of care
    - Quality of care
    - Good practice guidelines
    - Clinical research
    - Training/teaching

Experiences about CE to be shared

- How best to cover all RD patients' needs at national level in terms of disease coverage?
- Countries with past experience in Europe ->
  - Expertise officially identified at national level and/or established centres specialised in some RD/groups of RD:
    - **Denmark (2 centres)**
    - **France (131 centres)**
    - **Norway (16 centres)**
    - **Spain (62 centres)**
    - **UK (50-60 centres)**
    - ***Italy (215 regional centres)***



Organisation by medical area



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Better information to all citizens Orphanet as a Joint Action between European Countries

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Languages: FR | EN | ES | DE | IT | PT

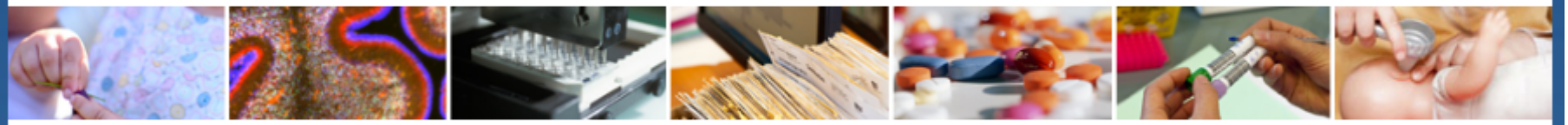
orphanet

The portal for rare diseases and orphan drugs

Inserm

*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 encyclopedia 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)

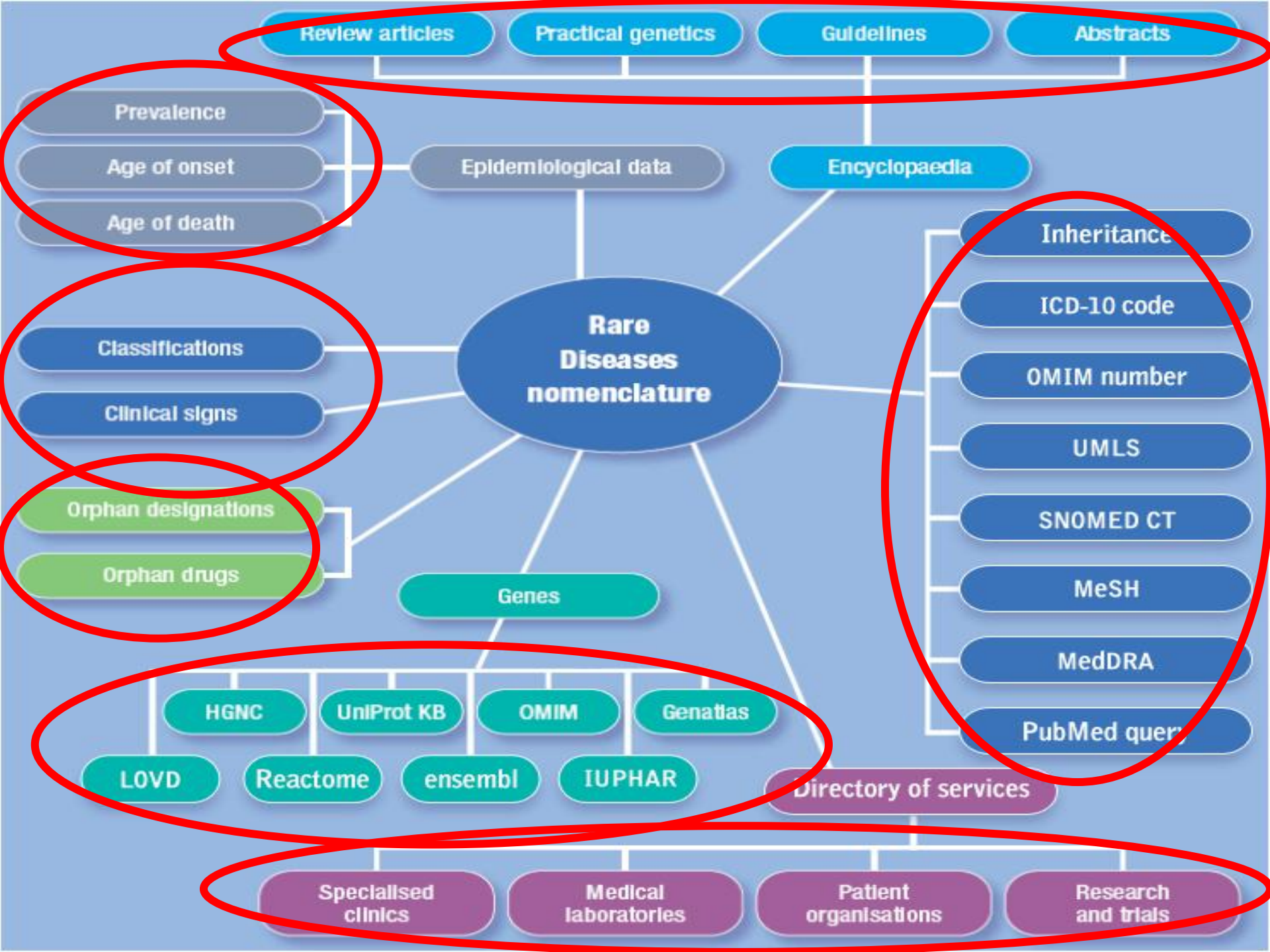
Events

See all

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

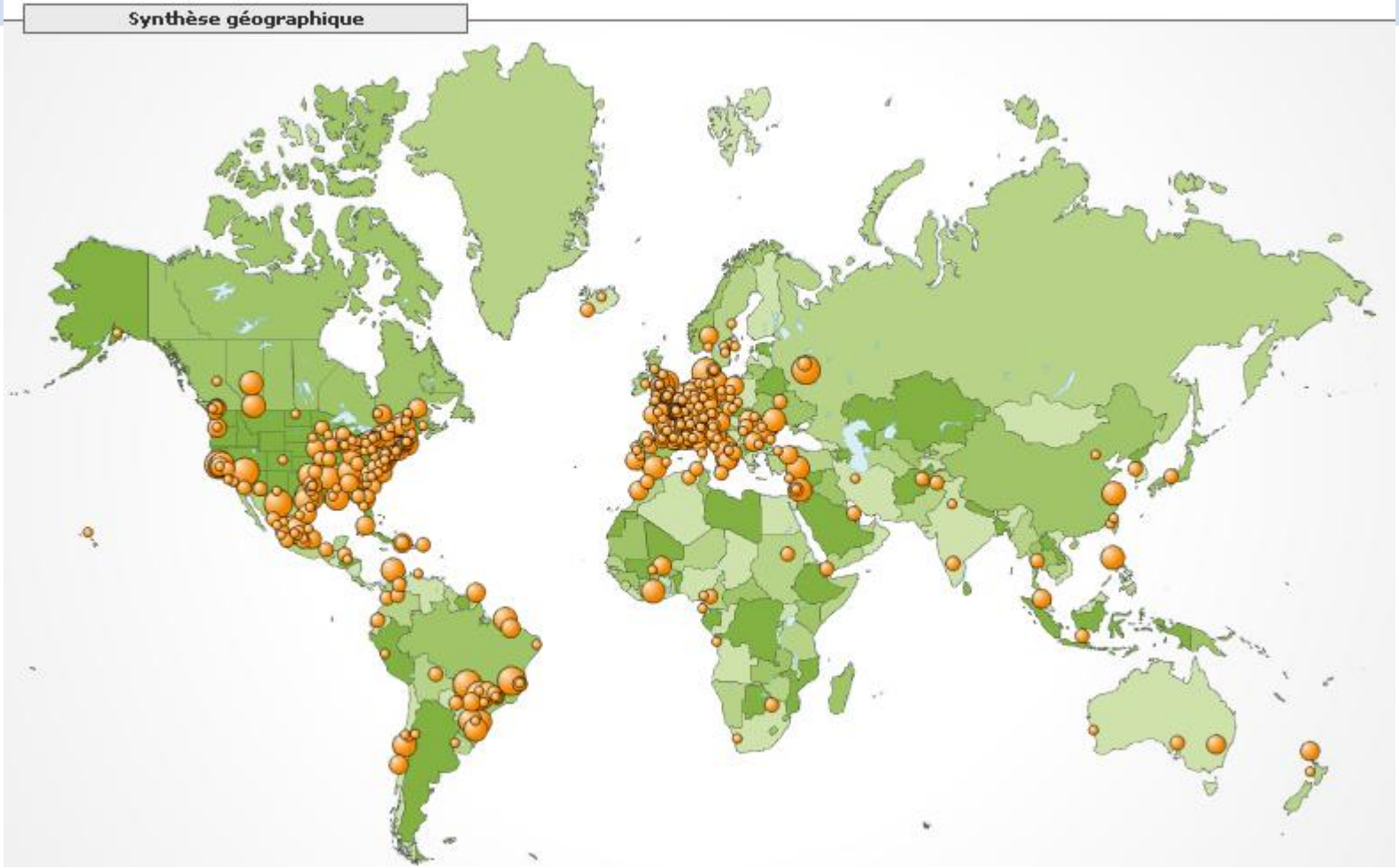
A relational database which allows multiple sorts of queries in 7 languages

Dutch, English, French, German, Italian, Spanish, Portuguese


Review articles
Practical genetics
Guidelines
Abstracts
Prevalence
Age of onset
Age of death
Epidemiological data
Encyclopaedia
Inheritance
ICD-10 code
OMIM number
UMLS
SNOMED CT
MeSH
MedDRA
PubMed query
Classifications
Clinical signs
Rare Diseases nomenclature
Orphan designations
Orphan drugs
Genes
HGNC
UniProt KB
OMIM
Genatlas
LOVD
Reactome
ensembl
IUPHAR
Directory of services
Specialised clinics
Medical laboratories
Patient organisations
Research and trials

>25 000 accesses daily from over 200 countries

2/3 Professionals 1/3 Patients and relatives



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The Orphanet consortium

2010

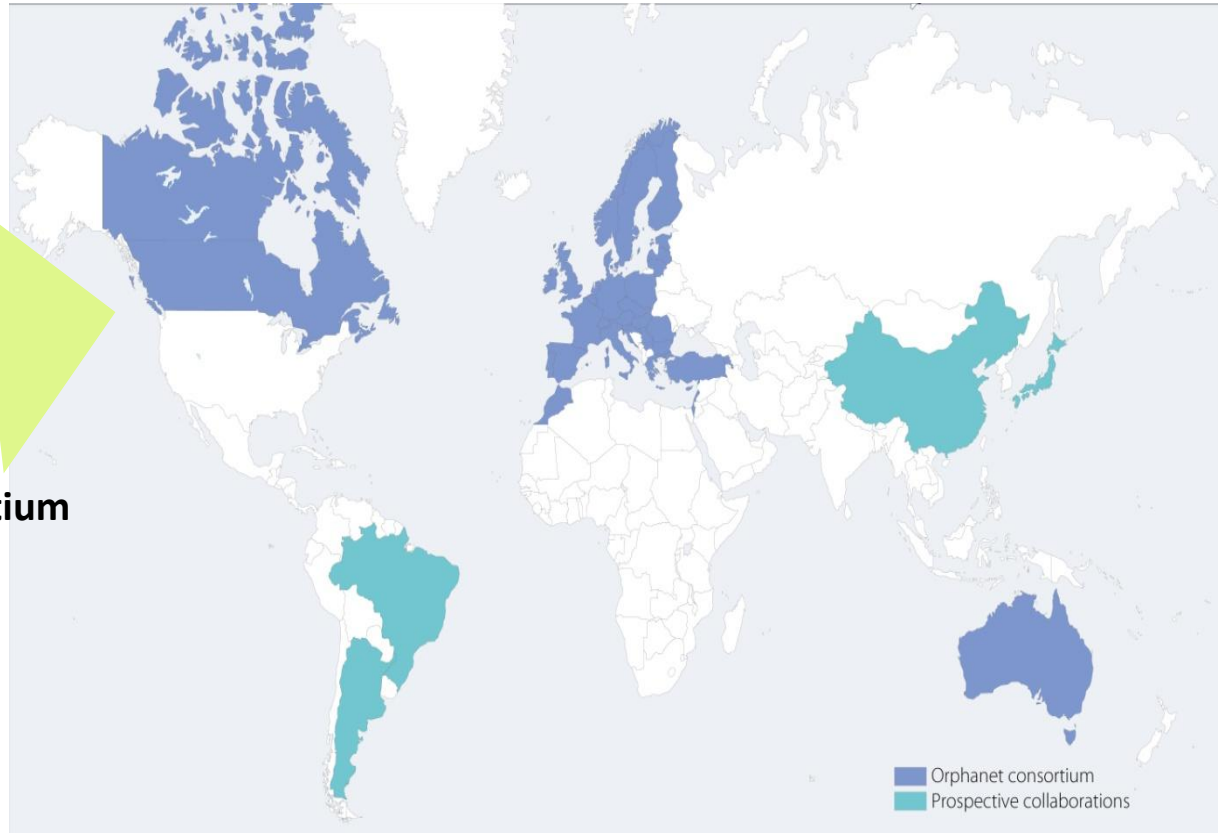
Orphanet consortium

2000

Orphanet Europe

1997

Orphanet France-INSERM



Partners are public institutions representing their country.
Partners dedicate resources to the Orphanet portal of rare diseases and orphan drugs.
National Orphanet teams work according to the **Orphanet Standard Operating Procedures**

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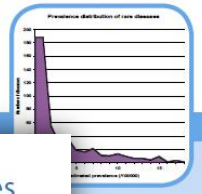
Orphanet Report Series

Rare Diseases collection

January 2011

Disease Registries in Europe



Orphanet Report Series

Rare Diseases collection

November 2011 | Number 1

Prevalence of rare diseases: Bibliographic data

Listed in alphabetical order of diseases

Orphanet Report Series

Orphan Drugs collection

January 2013

Lists of medicinal products for rare diseases in Europe

www.orpha.net

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Orphanet Report S

Rare Disea

December 2012 | Num

List of rare diseases

Listed in alphabetical order

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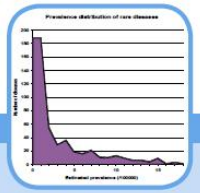
the field of rare diseases

www.orpha.net



Orphanet Report Series

Rare Diseases collection

November 2011 | Number 2

Prevalence of rare diseases: Bibliographic data

Listed in order of decreasing prevalence or number of published cases

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Orphanews to inform the Community:

- 16,000 registered readers
- 20 issues a year
- Political and Scientific news

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Conclusion

- Real impact of policies
- Dynamic at national level as well: adoption of plans or strategies
- Networking ,Orphanet, registries and strong patient organisations are the key success factors
- All RD face the same problems

Thank you for your attention