




RARE DISEASE AND ORPHAN DRUG REGISTRIES

2nd International Workshop

21-22 October 2013


Italian National Institute of Health



Co-funded by the  

A three year project co-funded by the European Commission within the framework of the EU program of Community Action in the field of Public Health

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In Europe a disease is considered rare when it affects no more than 5 individuals among 10,000 persons

News


- October 21-22, 2013
2nd edition of the International Workshop "RARE DISEASE AND ORPHAN DRUG REGISTRIES"
National Institute of Health
Rome (Italy)
- RARE DISEASES AND ORPHAN DRUGS - An International Journal of Public Health**
[download the Call for paper submission]
[visit the website]


[Home](#) > Rome, 21-22 October 2013

RARE DISEASE AND ORPHAN DRUG REGISTRIES

2nd International Workshop

21-22 October 2013
Italian National Institute of Health – Viale Regina Elena, 299 - Rome (Italy)

 [Program](#)

 [Abstract Book](#)

PETITION TO THE EUROPEAN PARLIAMENT

Special Provisions for rare diseases in the current proposal for a General Data Protection Regulation

EPIRARE (European Platform for Rare Disease Registries)

EPIRARE general information

Building Consensus and Synergies
European Platform for Rare Disease Registries

EPIRARE is a project
funded by the EU Commission (DG SANCO)

Starting date: April 2011

Duration: 30 months (April 2011 – October 2013)

Funding modalities: cofinanced 60% of total cost by European Commission

Coordinator of the project:

**National Centre for Rare Diseases, Italian National
Institute of Health – Rome, Italy**

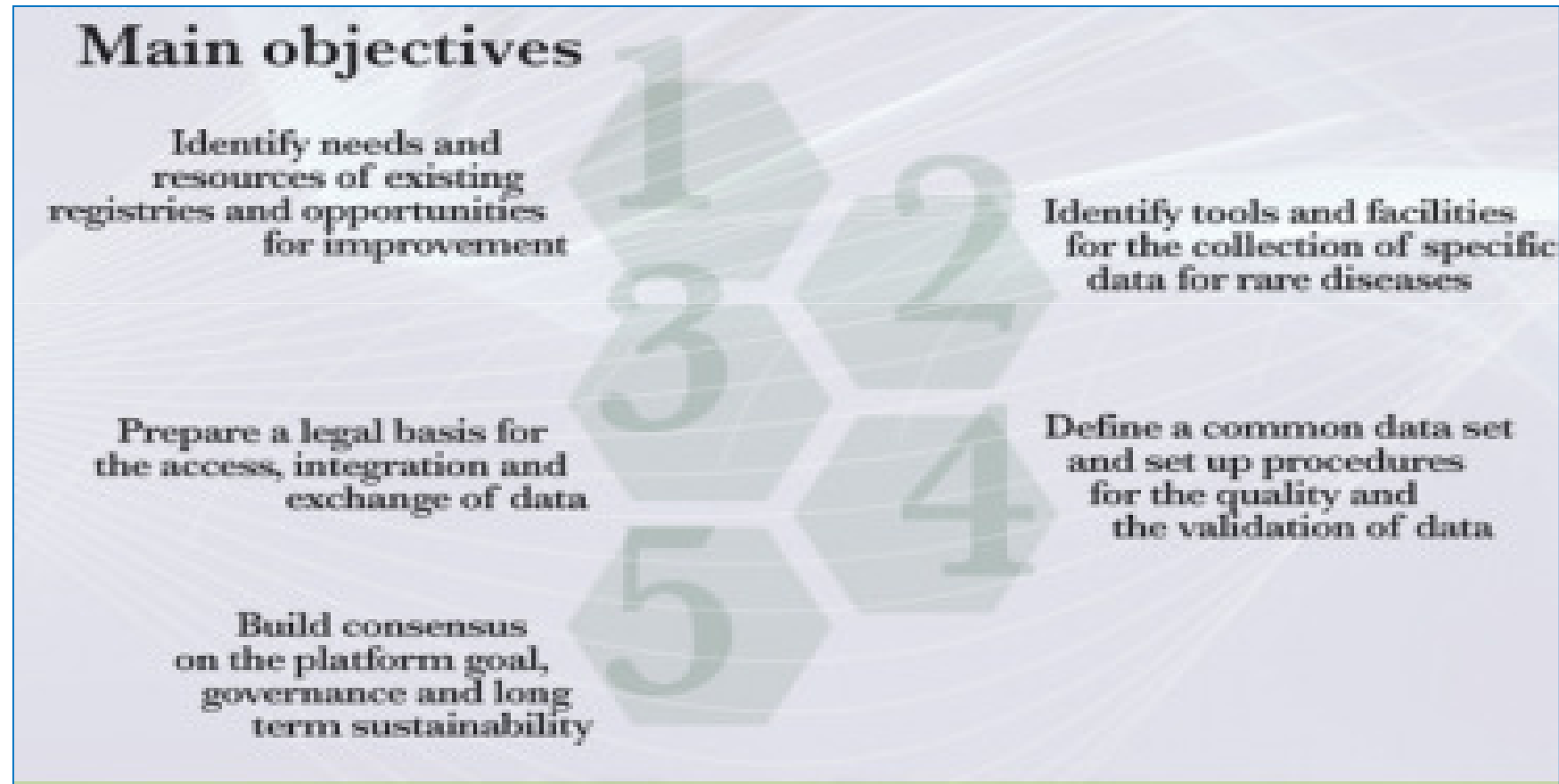
Partners

23 partners in
14 countries in
3 continents



Aims

- To prepare a **European Platform** for the Registration of Rare Diseases Patients
- To ensure the **quality and best use of the registered data**



Background

- More than 500 RD registries in the EU (Orphanet and other sources)
- No uniform standards for the storing and management of data
- Registries set up for 20% of rare diseases, more registries for the same RD
- Need to increase data sharing and exchange
- Need to harmonise existing regulations (EU, national and regional)

The EU Council Recommendation on Rare Diseases (adopted in June 2009) recommends, among other topics, the implementation of registers and databases for epidemiological purposes.

Therefore, many initiatives may appear in the coming years

EPIRARE activities

- **Survey of registries** - conditions and needs
- Survey of registries - data elements definitions and formats
- **Survey of patients expectations**
- Consultation/survey of industry expectations
- Informal Consultation/Information of national policy-makers
- **Liaison with Countries with established or planned national registries** (BE, BG, DE, ES, FR, IT)
- **Liaison with US ORDR, RD-Connect (IRDiRC)**
- Participation in EU initiatives and RD Committees

EPIRARE activities

- An analysis has been carried out on the **current directive on Personal Data Protection** and on the **draft Regulation discussed in the EU Parliament**



- A document has been prepared on the needs of research and public health for international RD registries



A “**petition**”, requesting amendments to the draft regulation, has been prepared for the attention of the MEPs, and supporting a request for an audit in the Parliament

The screenshot shows the EPIRARE website header with the logo and navigation menu. The main content area features a dark banner with the text "PETITION TO THE EUROPEAN PARLIAMENT" and "Special Provisions for rare diseases in the current proposal for a General Data Protection Regulation". Below the banner, there is a list of languages for the petition and a brief description of the petition's purpose.

Co-funded by the European Commission and the Executive Agency for Health and Consumers.

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PETITION TO THE EUROPEAN PARLIAMENT

Special Provisions for rare diseases in the current proposal for a General Data Protection Regulation

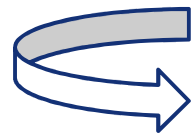
Home » Petition

Petični/ Pétition/ Petition/ Αίτηση / Petizione / Peticije/ Peticiju / Petición/ Petition

Following the concerns raised by the report introducing the Parliamentary discussion on the General Data Protection Regulation, we have published a petition ("[Don't Stop Research on Rare Diseases!](#)") to the European Parliament and to the EU Council.

Read more on the reasons for this petition in your preferred language here:

EPIRARE



SURVEY

addressed to all RD registers in Europe
(developed 2011, implemented 2012)

- 80% in favour of an EU platform
- 73% in favour of an EU portal

Main services expected (39% - 69%)

- **Technological tools**

(IT tools, networking tools)

- **Specific expert advice**

(legal, quality, privacy, ethics)

- **Resources**

(model documents, quality control systems,
access to useful data)

Needs of identified stakeholders' groups

Registry holders

Services reducing collateral workload of a registry
Services facilitating patient management
Assistance to start and run a registry
Funding
Motivation of professionals providing data
Communication and visibility
Networking

Patients

Improved care for RD patients
Information
Social inclusion

Pharmaceutical Industry

One stop shop for accessing information on diseases
Finding a disease specific registry
Recruitment of patients for clinical trials

Registers Survey VS Patient Survey :

Aims of a register

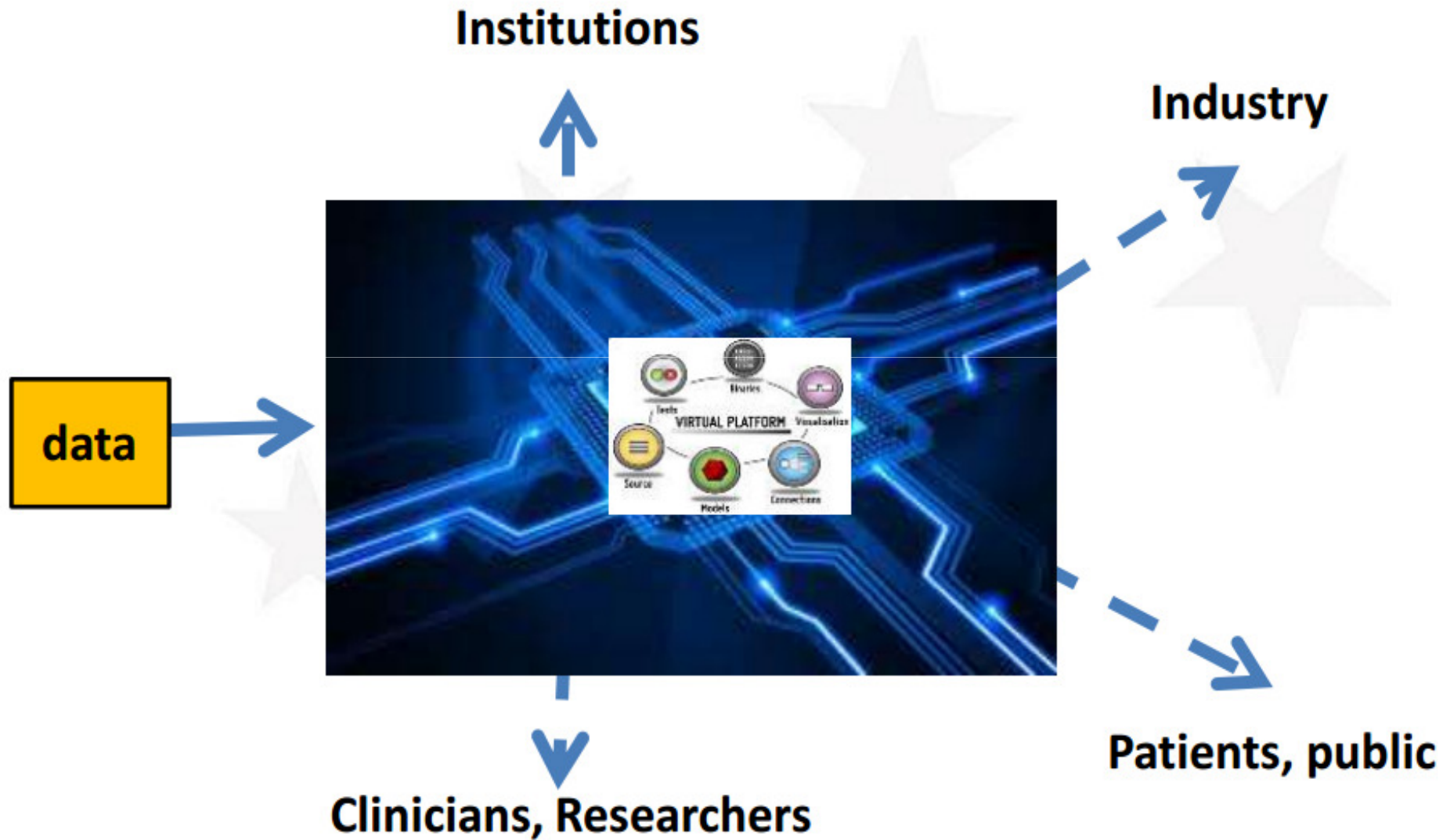
Register reality

1. **Epidemiological research**
2. **Clinical research**
3. **Natural history of the disease**
4. Disease surveillance
5. **Treatment evaluation (efficacy/safety)**
6. Genetic mutations database
7. **Healthcare/Social Services planning**

Patient expectations

1. **Healthcare/Social Services planning**
2. **Treatment evaluation (efficacy/safety)**
3. **Natural history of the disease**
4. **Epidemiological research**
5. Clinical research
6. Disease surveillance
7. Genetic mutations database

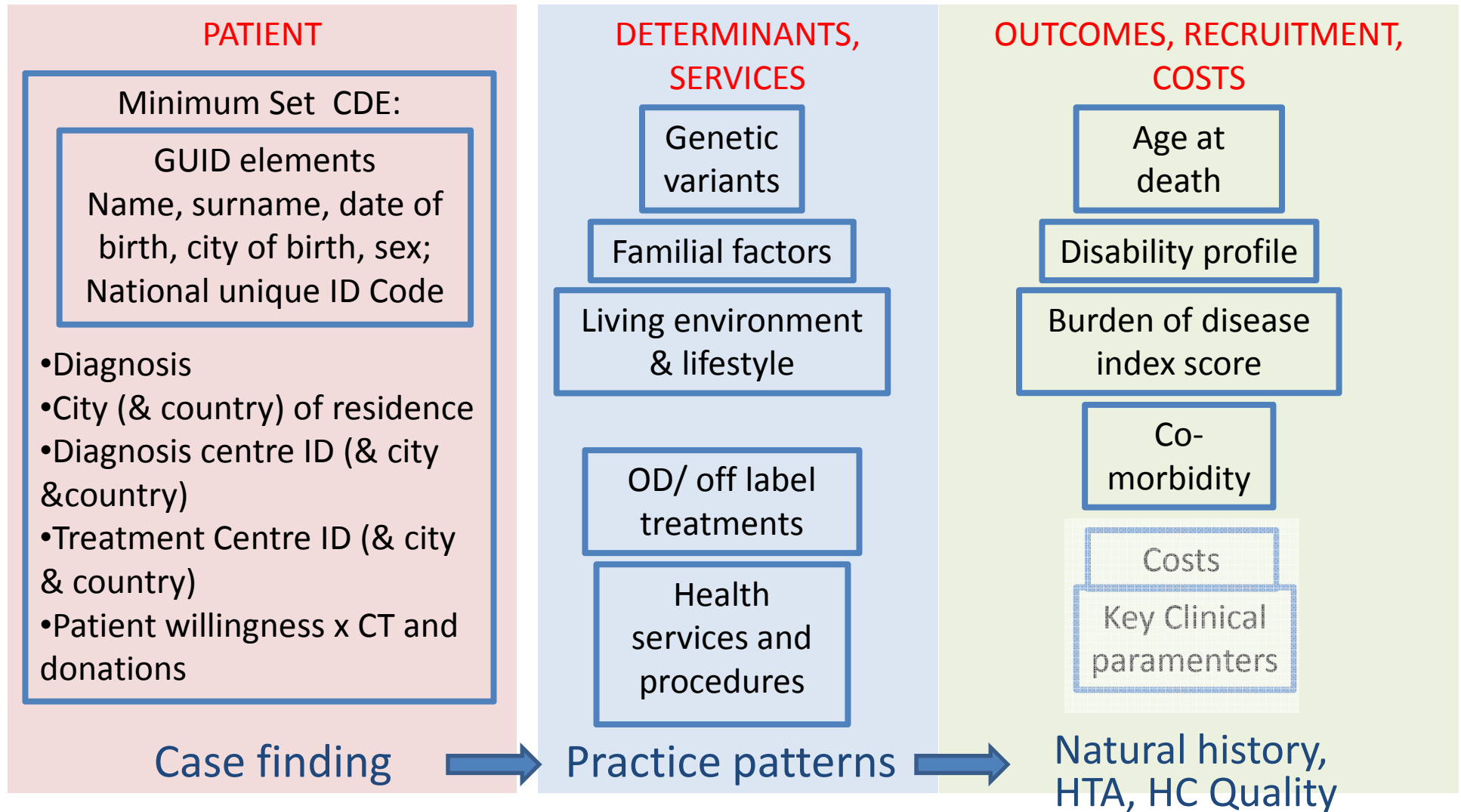
The EPIRARE Platform concept



The EPIRARE Platform Data Repository - prerequisites

- Multidisease
- Multipurpose
- Multistakeholder
- Multisource

The EPIRARE platform – Sets of Common Data Elements



Common Data Elements (CDEs) among RD Registries: results from the EPIRARE survey on Common Data Elements

Epirare project carried out a **web-based survey to a list of regional, national and international RD and orphan drug registries.**

The survey *inquired on more than 50 data elements likely to be collected by registries:*

- registry aims,
- patients' personal data,
- clinical and genetic disease data and comorbidity,
- data on: diagnostic and treating Centres, orphan and common drugs and other treatments
- patients' interest in clinical trials and biological donations
- transplantations,
- link to biobanks.

Common Data Elements (CDEs) among RD Registries: results from the EPIRARE survey on Common Data Elements

Results:

Registries may be able to make estimates on: incidence (52%) and prevalence (52%), life expectancy (59%), survival time (48%), diagnostic delay (45.5%), patient migration for diagnosis (52%) and for treatment (48%), patient willingness to be contacted for donating biological sample (38%) and participating in clinical trials (29%), link to biobanks (20%), disability (7%), quality of life (13%), orphan drug use (29%), its effectiveness (33%) and its safety (20%), current drugs effectiveness (27%), information in other types of treatments (58%), surgery (48%), cognitive (5%), physic (15%) and other rehabilitation (27%).

Conclusion: the survey show that **most Registries currently collect data as proposed by the Epirare questionnaire** and may be used in their current form to be shared within the platform to support public health policies and research.

**Development and maintenance of the
European Platform on Rare Diseases
Registration**

**Directorate-General
Health and Consumers
(DG SANCO)**

**Directorate-General
Joint Research Centre
(DG JRC)**
(Institute for Health and Consumer Protection)

**Principal goal of
the Platform**

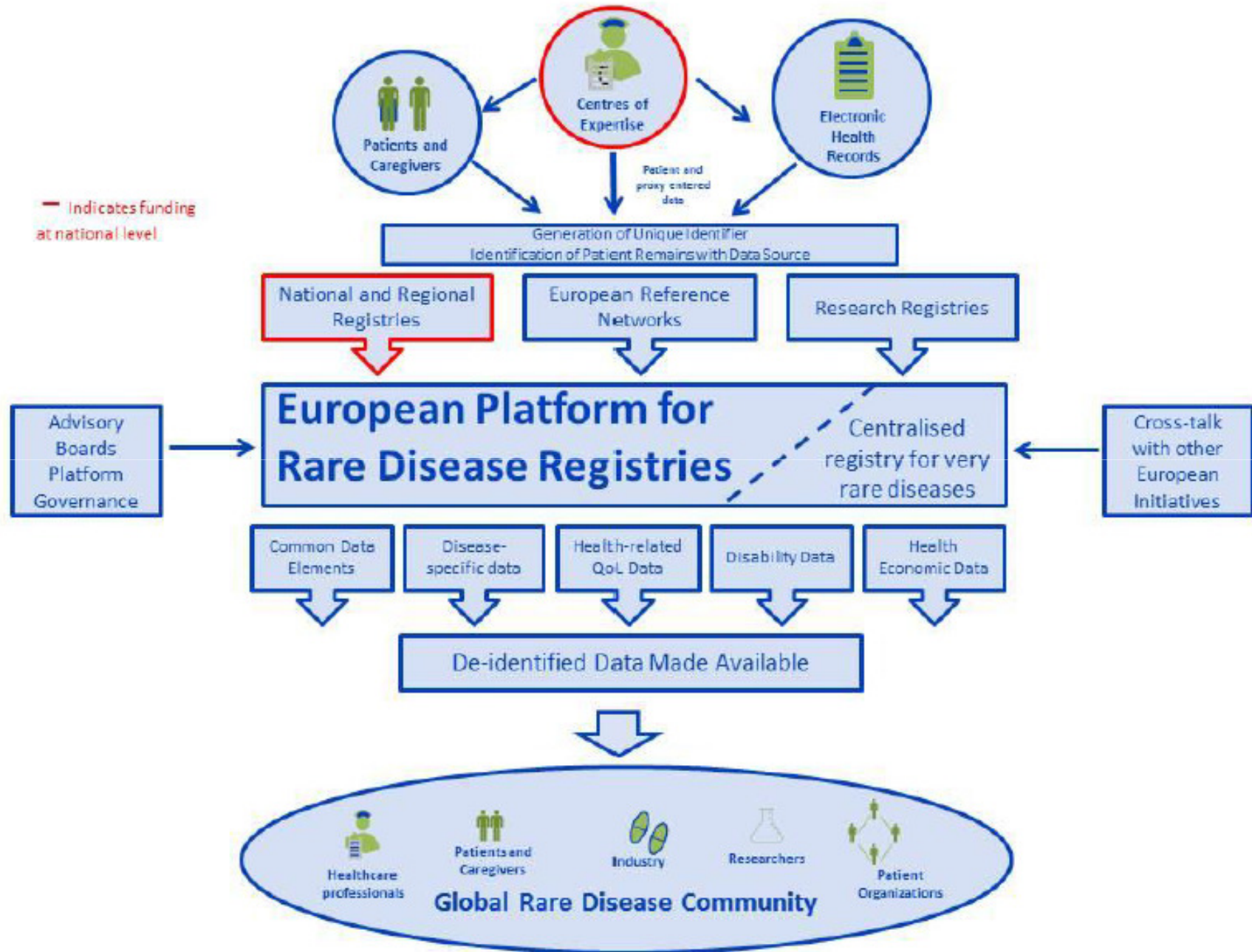
**To enable data analysis within and
across many rare diseases and to
facilitate clinical trials and other
studies and research**

Main Objectives

1. Promote **interoperability** of existing registries
2. **Support for new registries**
3. Act as a **hub** providing access to all data collection in the field of RD
4. Provide **IT tools** to maintain already existing selected data collection

Activities will be developed

- Taking into account outcomes of the projects funded by the Health Programme and Framework Research Programmes
EPIRARE, RD-CONNECT, PARENT JA, EUCERD JA, ORPHANET JA
- Ensuring collaboration with national registries
- Establishing collaboration with IRDiRC
- Establishing collaboration with the Global Rare Diseases Patient Registry and Data Repository (GRDR)
- Establishing collaboration with the Cancer Registries (ENCR)



THE INTERNATIONAL AND EUROPEAN CONTEXT FOR RARE DISEASE RESEARCH

European Union action in the field of Rare Diseases

- **Adoption of national plans and strategies in rare diseases in all Member States before the end of 2013.**
- Adoption of Implementing Decisions of the Cross-border healthcare Directive before the end of 2013.
- **Strength dissemination and visibility of the rare diseases database ORPHANET:** description of 6000 diseases.
- Contribution to the **revision of ICD** (International Classification of Diseases) version 11, during 2015/2016 classifying and codifying the maximum number of rare diseases.
- Creation of the **European Platform on Rare Diseases Registration** located in Ispra (Italy) during 2014. More than 600 registers in the EU.
- **Creation of an ERIC** (European Research Infrastructure Consortium) to support the European development on rare diseases during 2014/2015
- **Improving accessibility to orphan medicinal products in the UE**
- **Supporting EURORDIS** to reinforce patient's empowerment
- To analyse feasibility of a common approach on neonatal screening for some rare disorders.
- Creation of **IRDIRC** (International Rare Diseases Research Consortium) for transatlantic cooperation (EU, USA, Canada, Australia, South Korea, Japan, ...)
- Future Joint Action on Rare Tumours (2015-2017).

Instruments to implement these goals

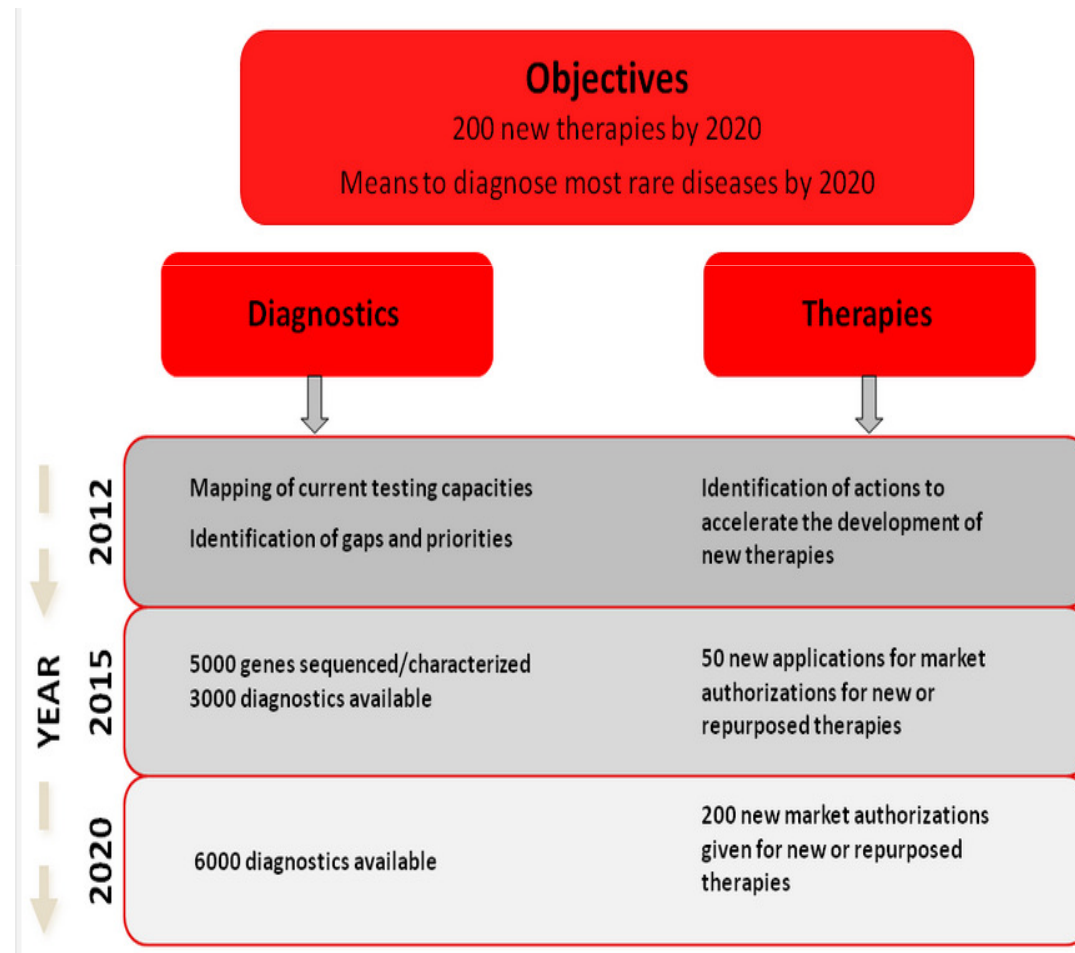
- **3rd European Union Health Programme 2014-2019**
 - **Research Programme Horizon 2020**
 - **Structural Funds 2014-2019**
-
- **The European Union Committee of Experts on Rare Diseases (EUCERD)**

International Consortium for rare diseases research - IRDiRC

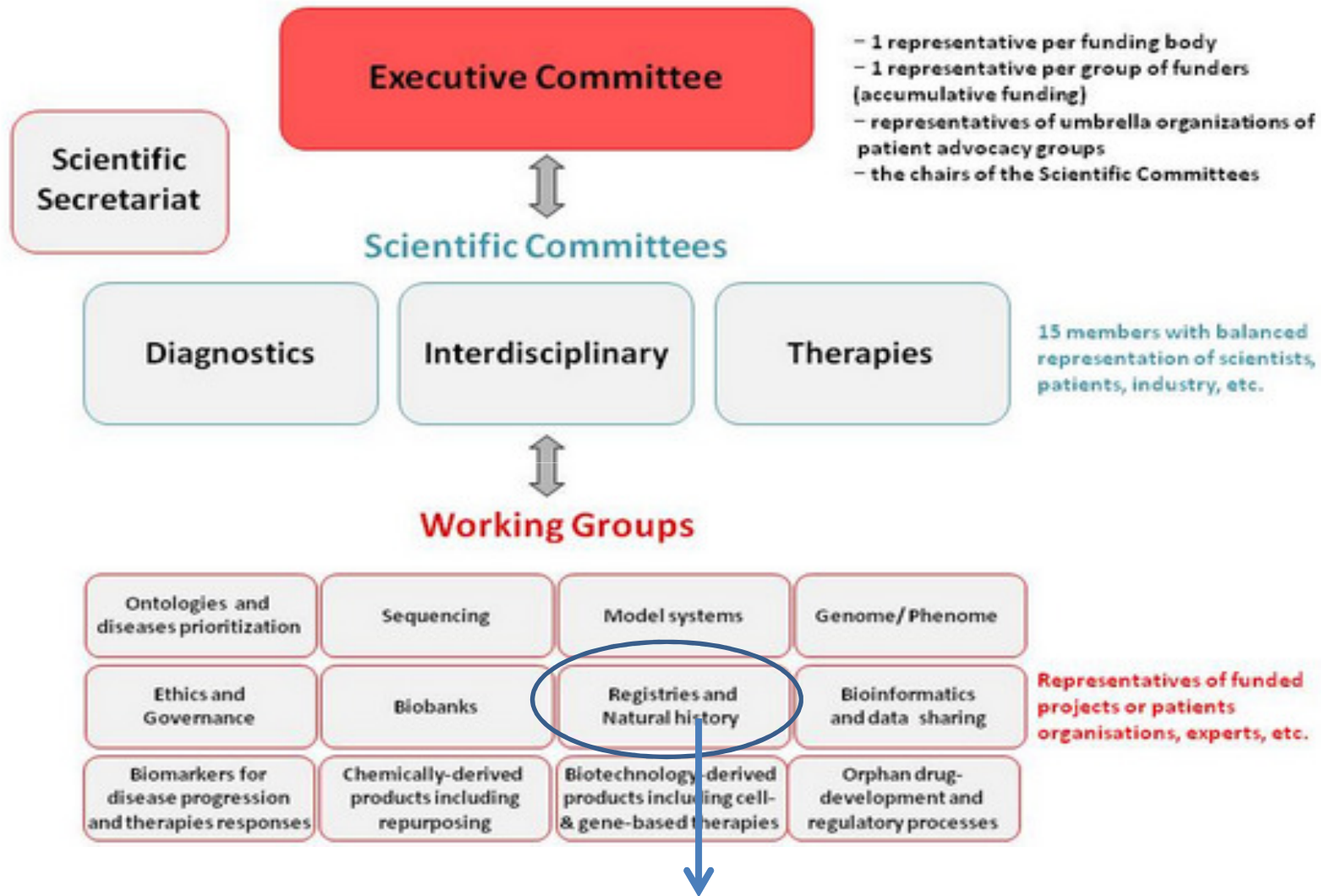
Two main objectives by the year 2020:

- to deliver **200 new therapies** for rare diseases
- to **diagnose most rare diseases**.

A number of grand challenges are being addressed through collaborative actions to reach these **2020 goals**.

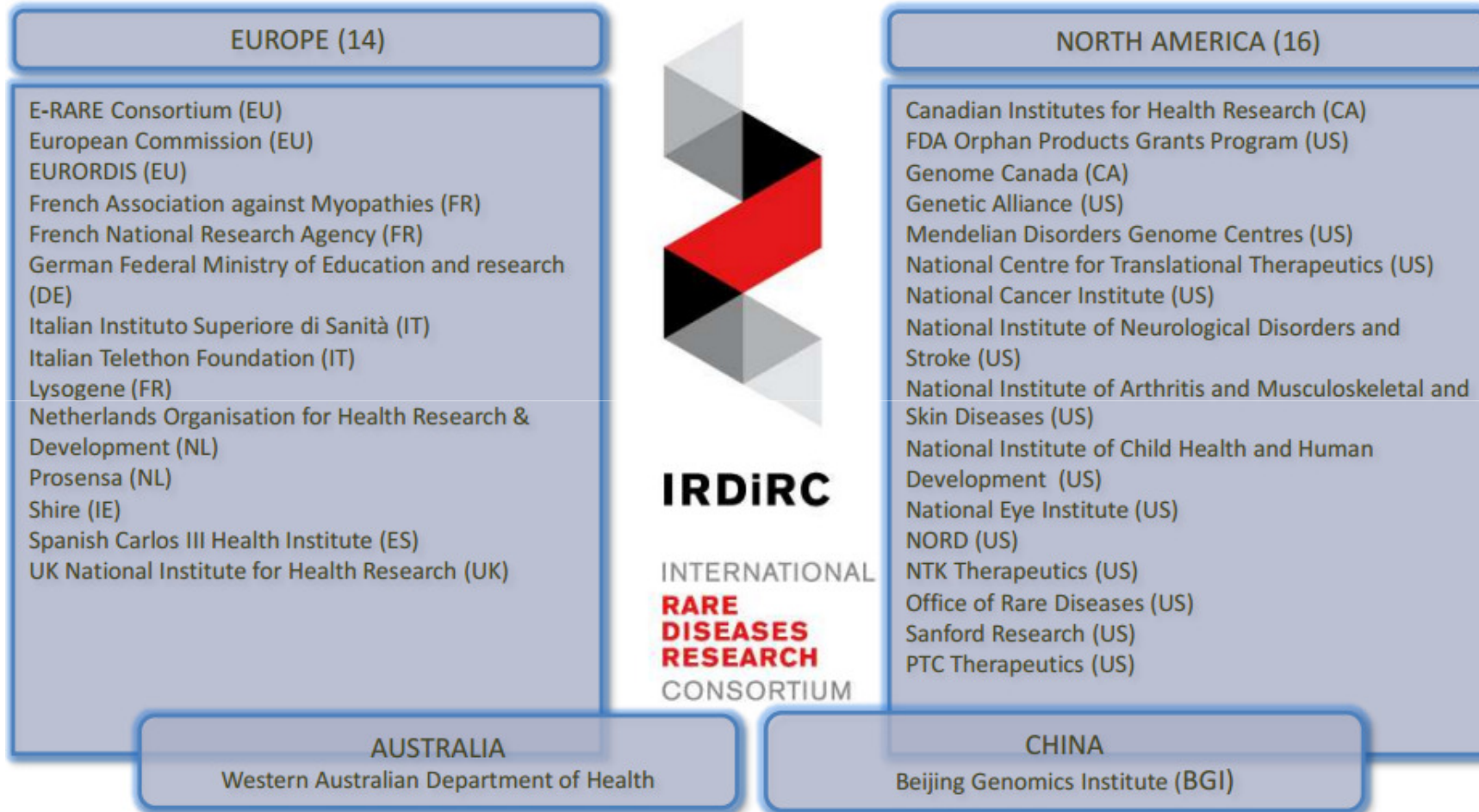


IRDIRC



RD-CONNECT: An integrated platform connecting databases, registries, biobanks for rare disease research

IRDiRC Executive: 32 committed members



Members are composed of funding body/organisation investing a minimum of \$10 million US over 5 years in research projects/programmes contributing towards IRDiRC objectives

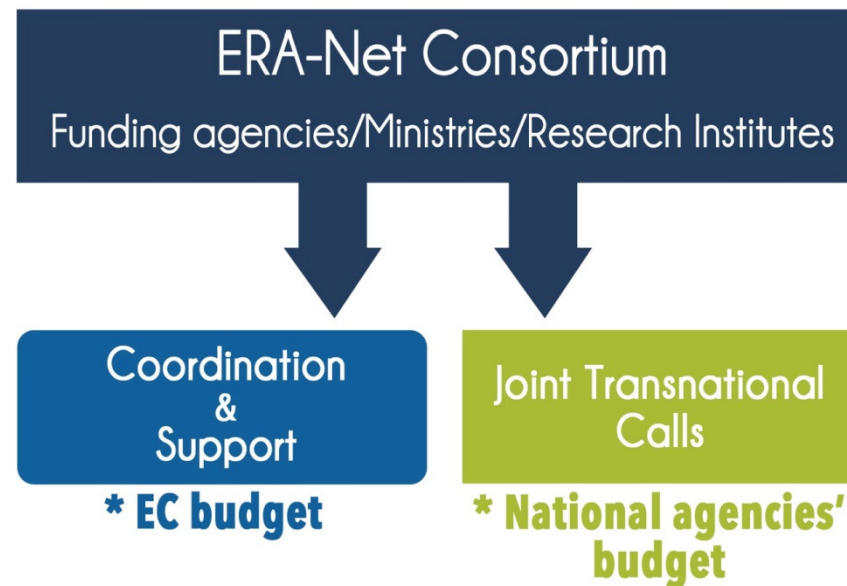
Small funders may form a group of funders

E-Rare

ERA-Net for Research Programmes on Rare Diseases

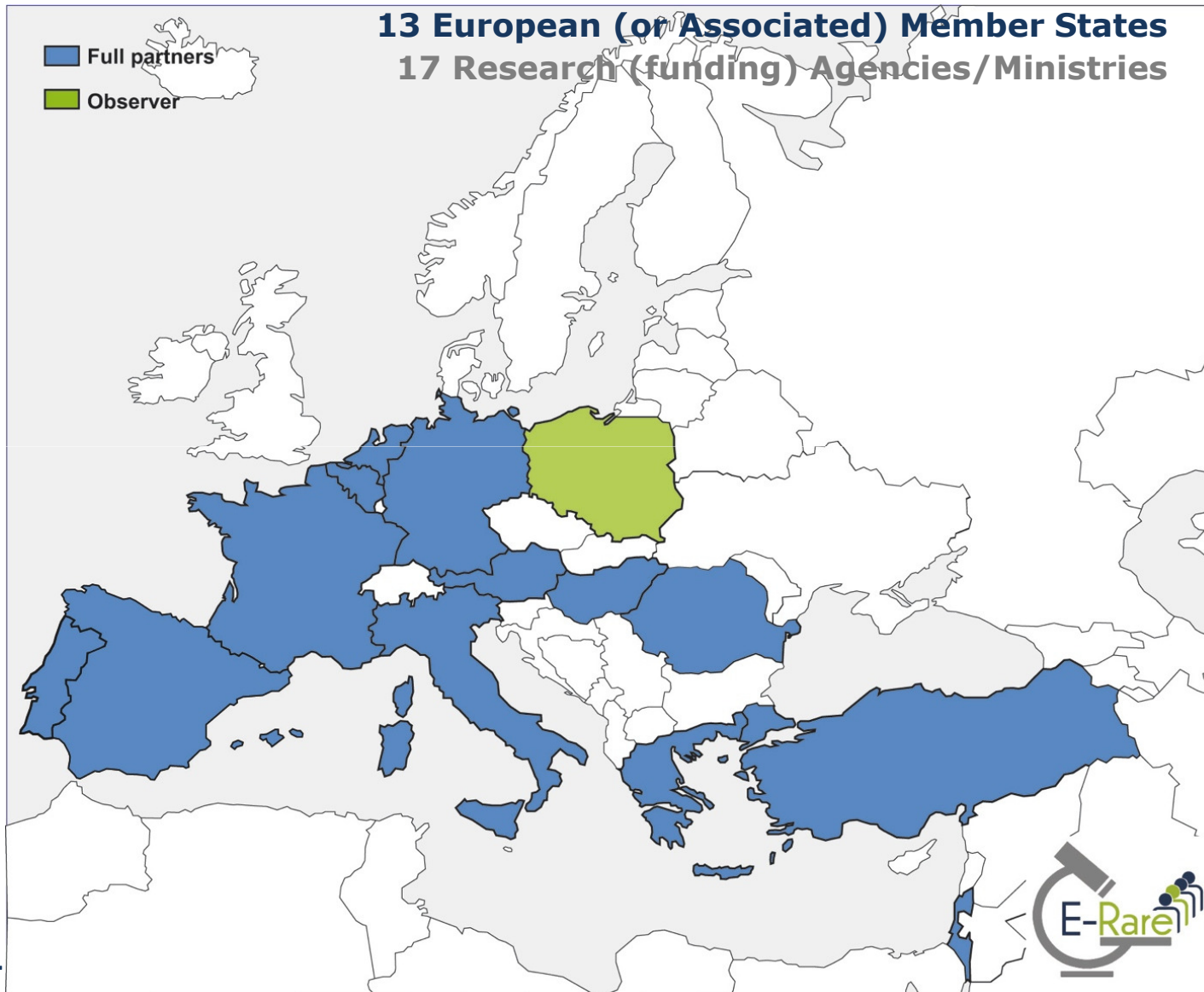
What is an ERA-Net?

ERA-Net
=
European **R**esearch **A**rea **N**etwork



E-Rare Consortium (2010-2014)

ERA-Net for Research Programmes on Rare Diseases





E-Rare

(2010 -2014)

Coordination & Support

- ▶ Harmonization of national research programmes on rare diseases
- ▶ Enlargement and Increase of international collaboration
- ▶ Development of common policies
- ▶ Implementation of transnational research activities (JTCs)
- ▶ Other joint activities

*** EC budget**

Joint Transnational Calls

- ▶ Yearly joint calls
- ▶ Two "general" joint calls
- ▶ Two "focused" joint calls
- ▶ Open to any funding body

*** National agencies' budget**



Other EU Research Infrastructures

- **EATRIS** European Advanced Translational Research Infrastructure in Medicine
Translational research



- **ECRIN** European Clinical Research Infrastructure Network
Clinical studies and biotherapies



- **BBMRI** Biobanking and Biomolecular Resources Research Infrastructure
Research Biobanks

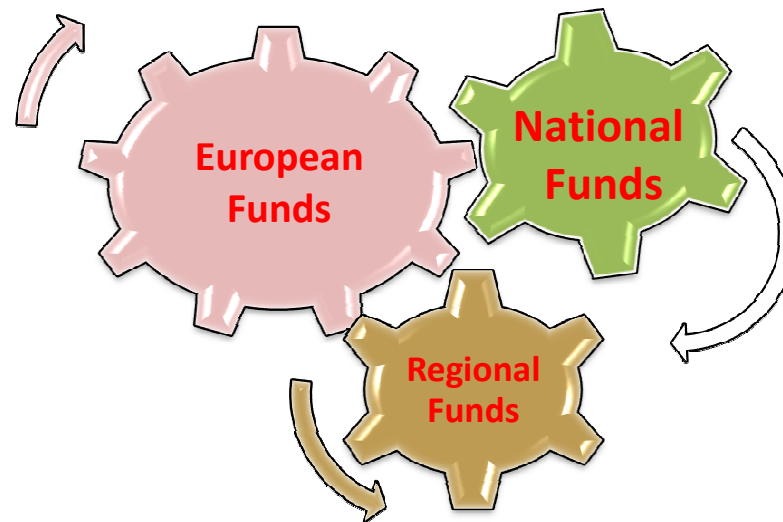


European Strategy Forum on Research Infrastructures



ESFRI identified **48 projects** of new research infrastructures so far, ESFRI will more focus on their implementation for the next few years

Development and Implementation of EU Research Infrastructures



RARE DISEASE REGISTRY PLATFORMS

Global Rare Diseases Patient Registry

 U.S. Department of Health & Human Services
NATIONAL INSTITUTE OF HEALTH *NIH... Turning Discovery into health*
ORDR Office of Rare Diseases Research
of the NATIONAL CENTER FOR ADVANCING TRANSLATIONAL SCIENCES

GRDR Global Rare Diseases Patient Registry and Data Repository

- The Office of Rare Diseases Research (ORDR)/NIH has launched a **pilot program** to establish a **Global Rare Diseases Patient Registry and Data Repository (GRDR)** to collect patient clinical information without personal identifiers **for research**.

The goal of GRDR is to enable analyses of data across many rare diseases and to **facilitate clinical trials and other studies**.

- The GRDR will also develop the capability to link patients' data and medical information to **donated biospecimens** by using a Voluntary Global Unique Patient Identifier (GUID). The identifier will enable the **creation of an interface between the patient registries that are linked to biorepositories and the Rare Disease Human Biospecimens/Biorepositories (RD-HUB)** <http://biospecimens.ordr.info.nih.gov/>.



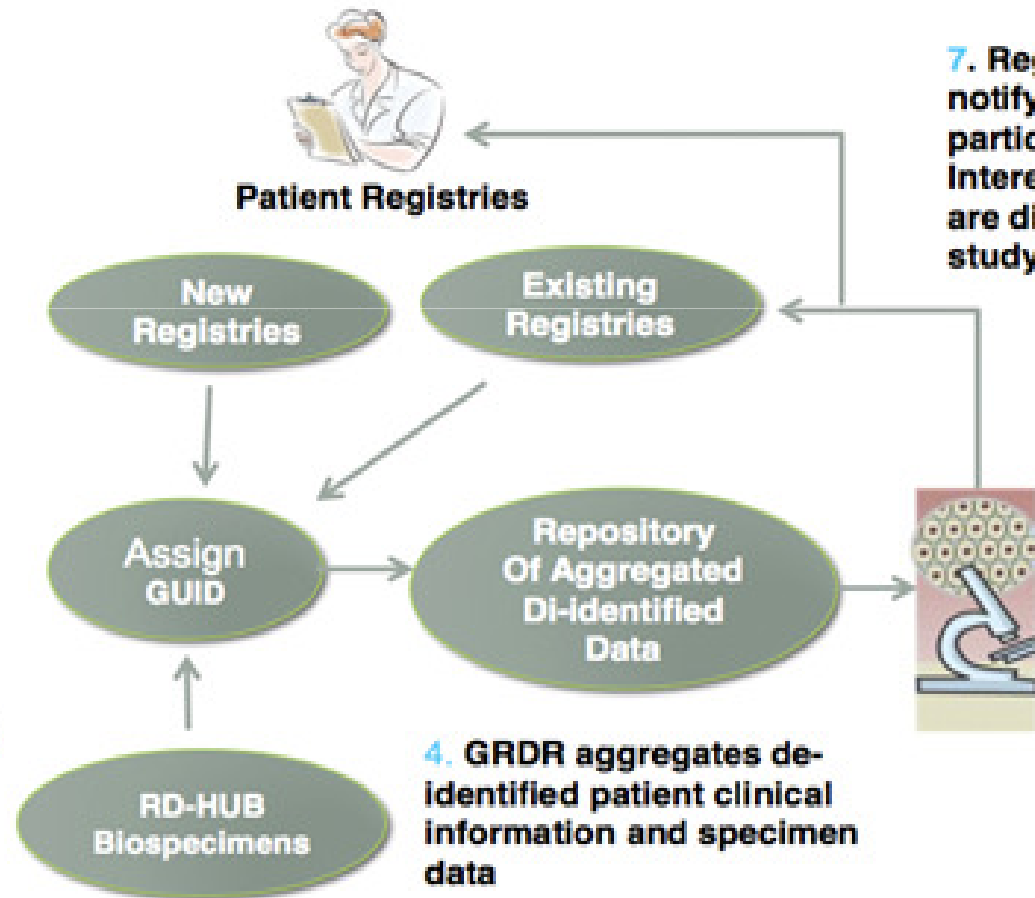
*Rare Diseases have no borders! They don't affect individuals,
they affect entire families*

GRDR

1. Patients Provide health information and test results

2. A Global Unique Patient ID (GUID) is assigned and patient data is mapped to common data elements (CDE's)

3. Patient data linked to biospecimens via the GUID interfacing with RD-HUB



7. Registry owners notify identified participants. Interested patients are directed to the study contact

6. Researchers can identify potential study participants and submit a contact request to the original registry owner

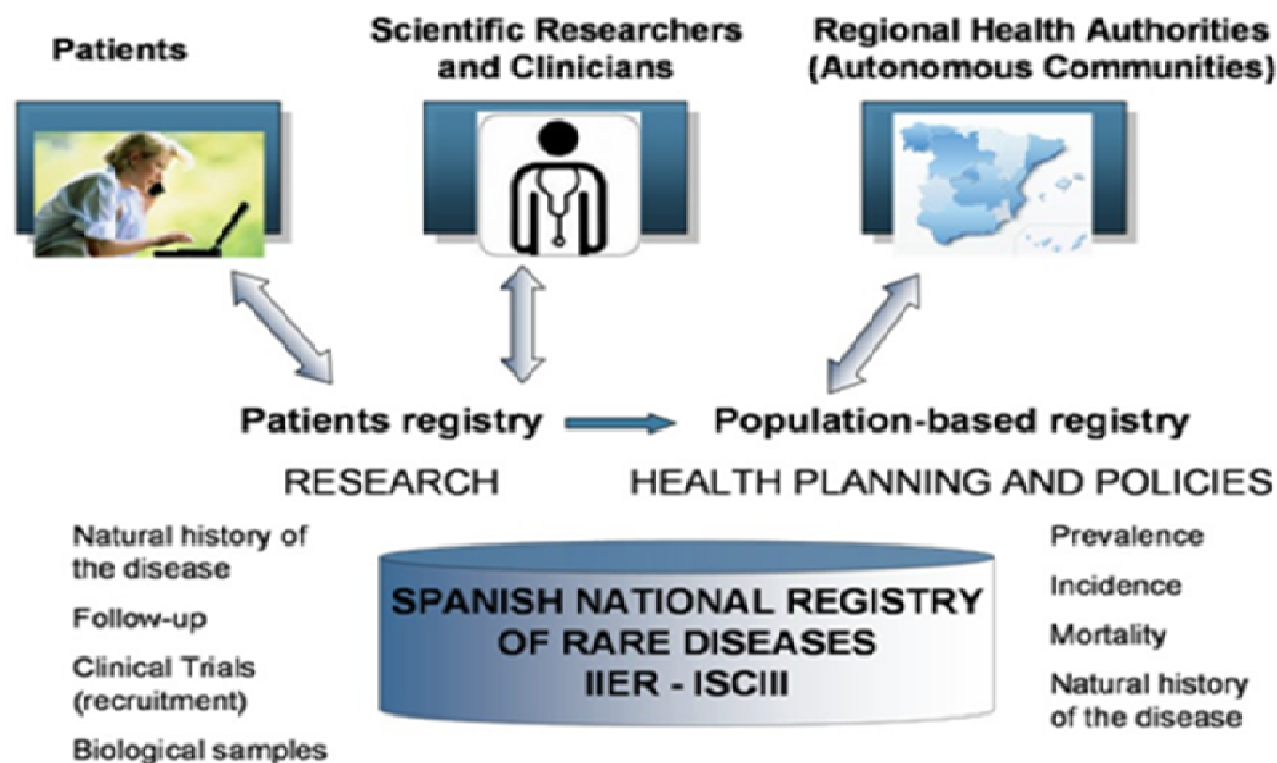
5. De-identified registry data is available to researchers for studies and clinical trials

The Spanish National Rare Diseases Registry

SpainRDR is a project financed by the Institute of Health Carlos III (ISCIII) within the scope of the IRDiRC . Coordination: Institute of rare diseases Research (IIER).

Aims to build the National Rare Diseases Registry in Spain based on the input of two different strategies:

- **Patient registries** addressed to patient outcome research
- **Population-based registries** addressed to epidemiological research and social and health systems planning



The system of French National Rare Diseases Registries BaMaRa-BNDMR and RaDiCo

Plan National Maladies Rares

Axis A

IMPROVE THE QUALITY OF PATIENT'S CARE

A1- Improve the access to diagnosis and care

A-1- 4 Coordinate the «rare diseases» structures with a shared information system

BaMaRa - Banque Nationale de Données Maladies Rares

Axis B

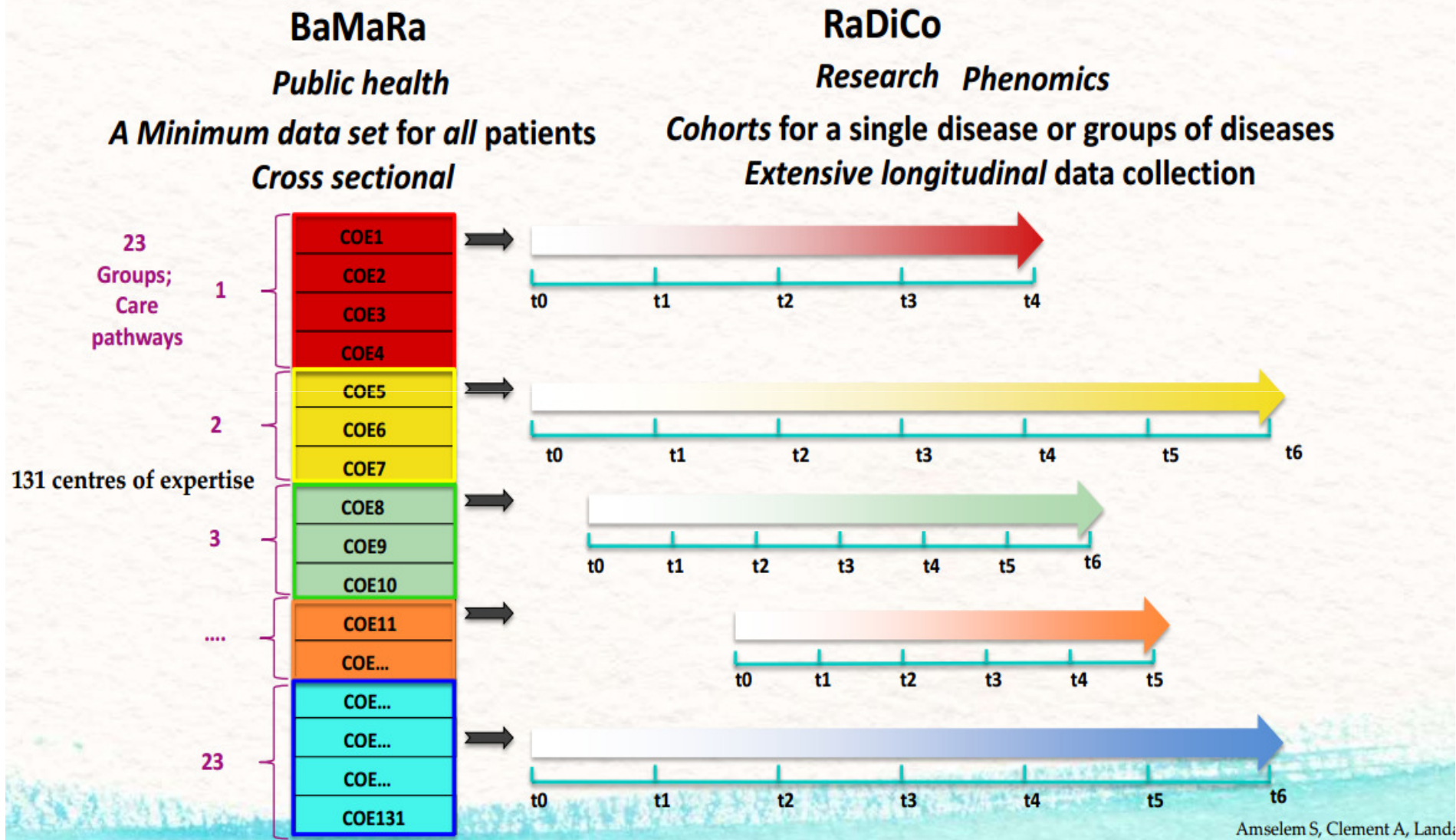
DEVELOP RESEARCH ON RARE DISEASES

B-1 Create a national structure to impulse research with public and private partners

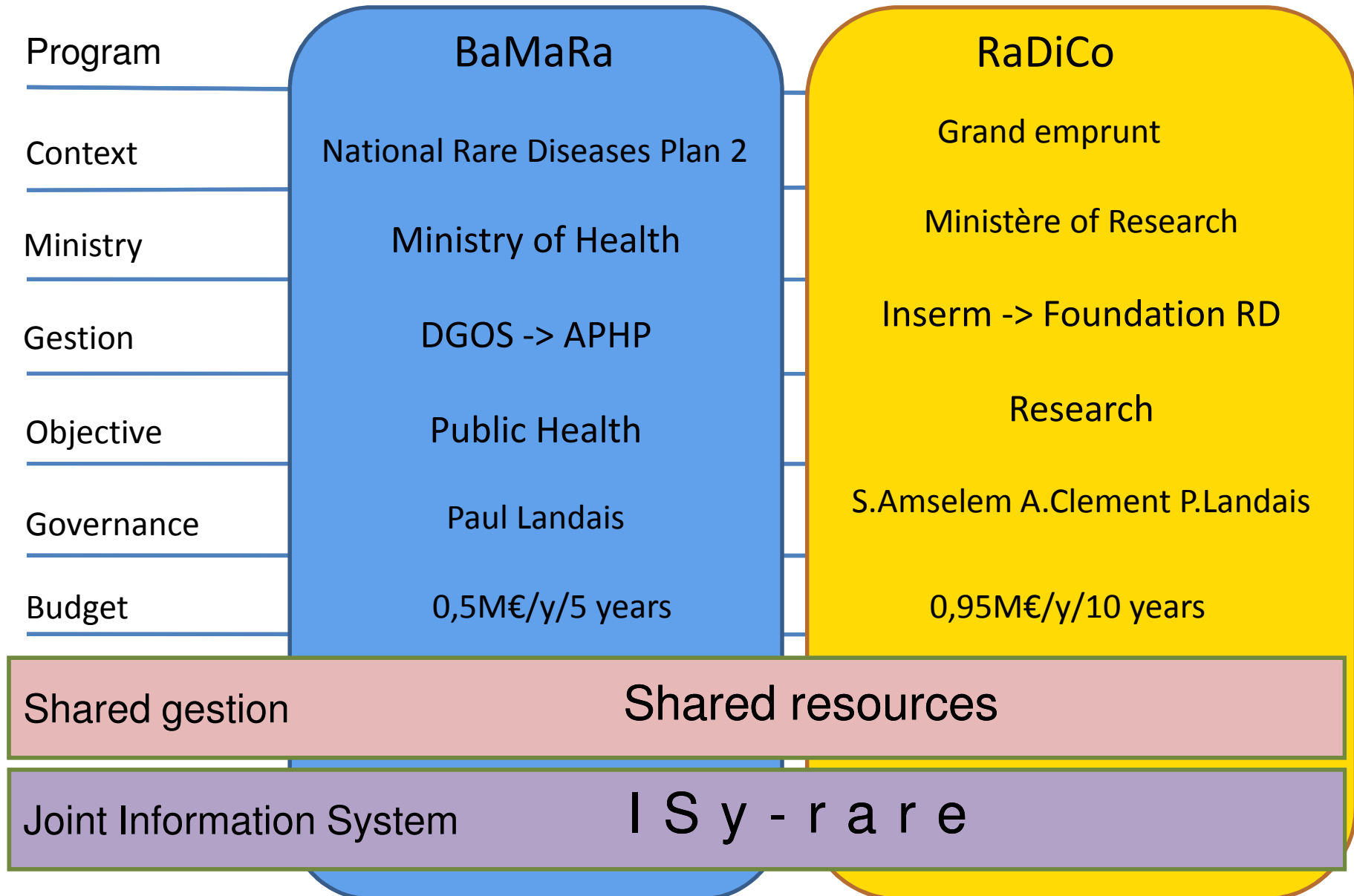
B-1-1 Create a foundation dedicated to rare diseases

RaDiCo - Rare Diseases Cohorts

The French experience: a shared information system



A joint information system for BaMaRa and RaDiCo



The Italian National Rare Diseases Registry

IDENTITY CARD

- **Name:** National Registry of Rare Diseases (NRRD)
- **Date of establishment:** 2001
- **Location:** National Center for Rare Diseases (Istituto Superiore di Sanità)
- **Funding support:** Public funds (Italian MoH+ISS)
- **Geographical coverage:** National (90% of Italian regions)

THE MINISTERIAL DECREE 2001, n. 279 establishes:

1) **Italian National Network for RD** for

- Prevention
- Surveillance
- Diagnosis
- Treatment

The network is made of Centres for Rare Diseases where patients are diagnosed and treated completely free of charge

2) **National Registry of Rare Diseases** as an important tool for evaluating the impact of RD on health services and on general population

3) **List of RD**

National Registry of Rare Diseases: OBJECTIVES

- **Planning and evaluating health care programmes** (services utilization, patients' mobility...)
- **Diseases surveillance** (estimation of prevalence and incidence, geographical and temporal distribution of RD)

