Session 5. Global rare disease policies and programs

A global approach to patient registries
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Institute of Health Carlos III, Spain

International Conference of Orphan Drugs and Rare Diseases - ICORD
Cape Town, South Africa, Oct, 19th-22th, 2016

RARE
Rare Diseases
Conference 2016

ICORD
International Conference on
Rare Diseases & Orphan Drugs

Global approach

- **National**
  - SpainRDR
  - Australia, France, Italy
  - …

- **Supranational**
  - TREAT-NMD
  - RD-CONNECT
  - GRDR
  - JRC European Platform

Registries are not

- Databases
- A collection of cases
- A proof of concepts

Registries are

- Information systems
- Based on observational study designs
- Several aims and rules
- Long-term perspectives

What do we consider if we want to build a patient registry?

- **Full Cooperation (*NHS & SS; Researchers; Industry and Patient Organizations*)**
- **Main challeging topics**
  - **Case ascertainment /sources of information**
  - **Reliabble data & completeness**
  - **Validity**
  - **Sustainability**

SpainRDR: A Spanish national experience and also an IRDiRC project

Institute of Rare Diseases Research

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RARE TUMOR IN CHILDREN
Francisco Javier Alonso

Instituto de Salud Carlos III

NHS Rare Diseases Strategy 2014

1. **Rare Diseases Information**
2. **Prevention**
3. **Health Care**
4. **Therapies**
5. **Social care**
6. **Research**
7. **Training**

Estrategia en
Enfermedades Raras del
Sistema Nacional de Salud

SANIDAD 2013

MINISTERIO DE SANIDAD, SERVICIOS SOCIALES E IGUALDAD

Patients
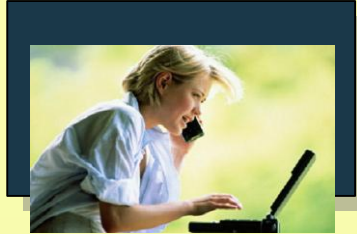
Scientific Researchers and Clinicians

Regional Health Authorities (Autonomous Communities)

Patients registry → **Population-based registry**

RESEARCH   HEALTH PLANNING AND POLICIES

Natural history of the disease

Follow-up

Clinical Trials (recruitment)

Biological samples

SPANISH NATIONAL REGISTRY OF RARE DISEASES IIER - ISCIII

Prevalence

Incidence

Mortality

Natural history of the disease

Data

- Reliable and interoperable
  - Common data elements
  - Phenotype – Ontologies: HPO & ORDO
  - Classifications – SNOMED-CT; ICD10-CM
  - Clinical terminology- UMLS
  - GUID/PUID
- Completeness – To have data is costly

Defining a quality assessment framework

Quality Improvement

Quality Control

Define Purpose

Report Study Findings

Develop Study Plan

Implement Improvements (SOPs improvement)

Design Protocol (SOPs development)

Evaluate Improvements

Conduct Study

Identify Improvement Opportunities

Analyze Study Data

Analyze Study Procedures

QR · QA · QC · QAss · QInd
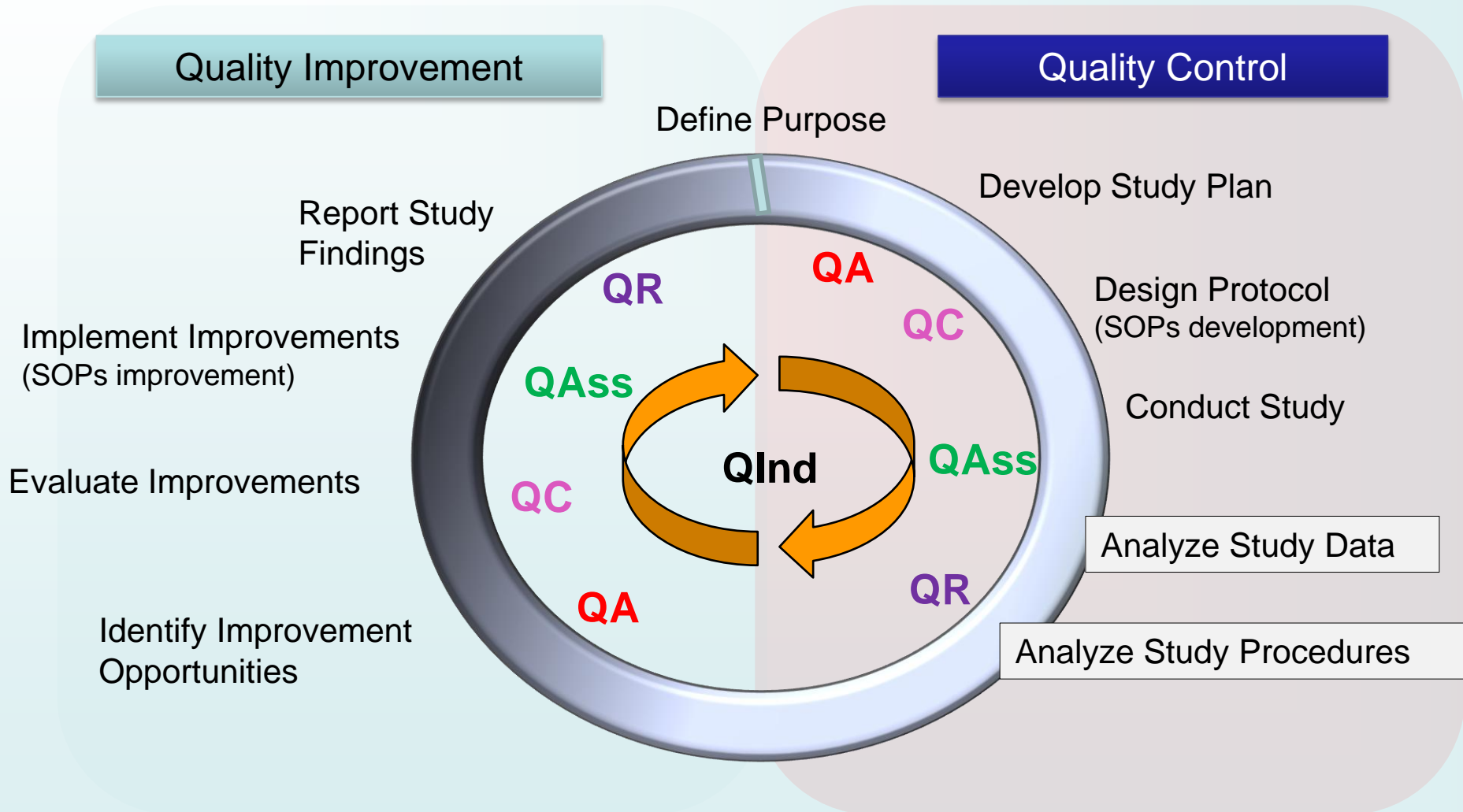
Create tools for analyses *vs* to use standard tools

Training on-line

Sustainability

- **ELSI framework**
- **Full Cooperation**
- **Full stakeholders engagement**

Patient registries: current collaborations

Spanish National Rare Diseases Registry

- α-1-antitripsin deficiency
- LAM
- DSD
- Epiderm. bullosa
- Wolfram syndrome
- Other ...
- Pulmonary histiocytosis

- ❑ Pediatric interstitial lung diseases
- ❑ Lymphangioleiomyomatosis
- ❑ Alveolar proteinosis
- ❑ Alpha-1 antitrypsin deficiency
- ❑ Traqueal stenosis
- ❑ Sarcoidosis
- ❑ Pulmonary histiocytosis
- ❑ Epidermolysis bullosa
- ❑ Disorders of sex development
- ❑ Congenital suprarenal hyperplasia
- ❑ Bradikinin mediated angioedema
- ❑ Wolfram syndrome
- ❑ Cystinosis
- ❑ Congenital anemias
- ❑ Duchenne disease
- ❑ Hereditary ataxias
- ❑ Hereditary spastic paraplegia

French registry approach

An integrated approach for Rare Diseases

Source: Choquet R, Landais P, BaMaRa 2013

TREAT-NMD
Neuromuscular Network
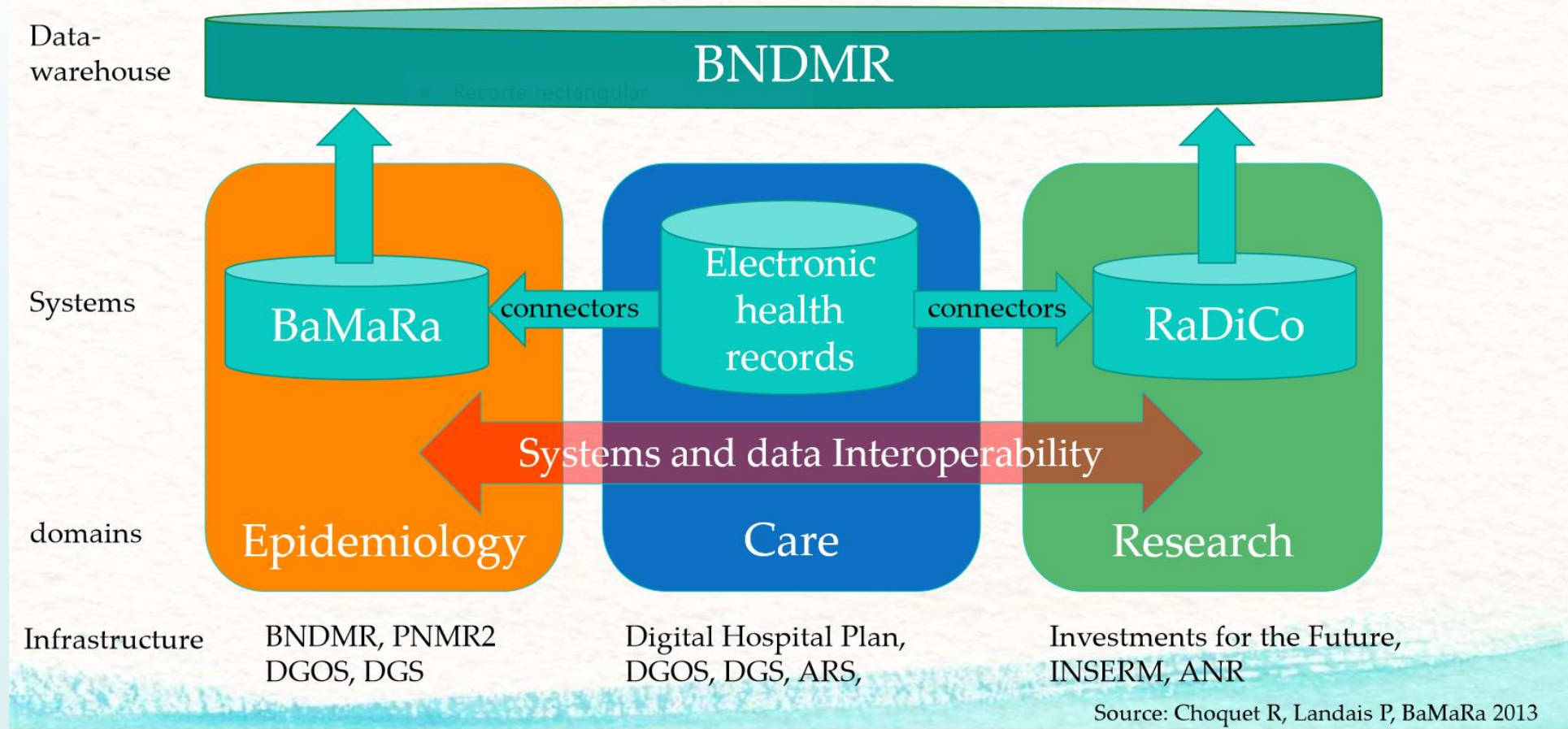
About TREAT-NMD
All about the network

Resources
Tools and infrastructure
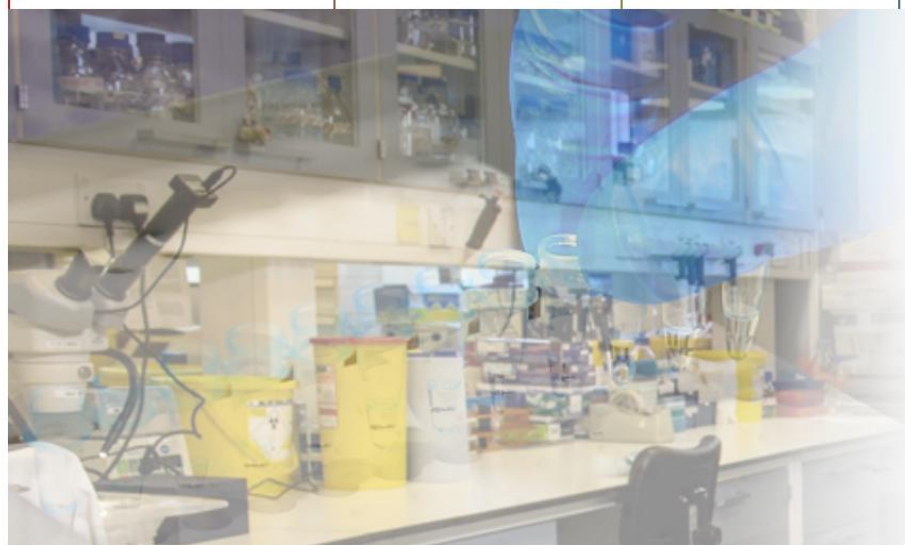
Research
Scientific and clinical

Care
Global best practice

Industry
Service overview

Disease Information

Advancing diagnosis, care and treatment for those living with **neuromuscular diseases** around the world...

NEWS   *C3 Announces RFA to Fund LGMD2A*   *Thursday, 20th October 2016*

Post marketing surveillance

Collection of safety and efficacy data once a drug has received conditional marketing approval

Disease information

Myotubular and centronuclear myopathy disease section now live!

Imperatives of DMD

Latest!!
Find out about DMD care in 24 languages - including Slovak & Korean!

News   Events

Cellular Mechanism Discovery for SMA
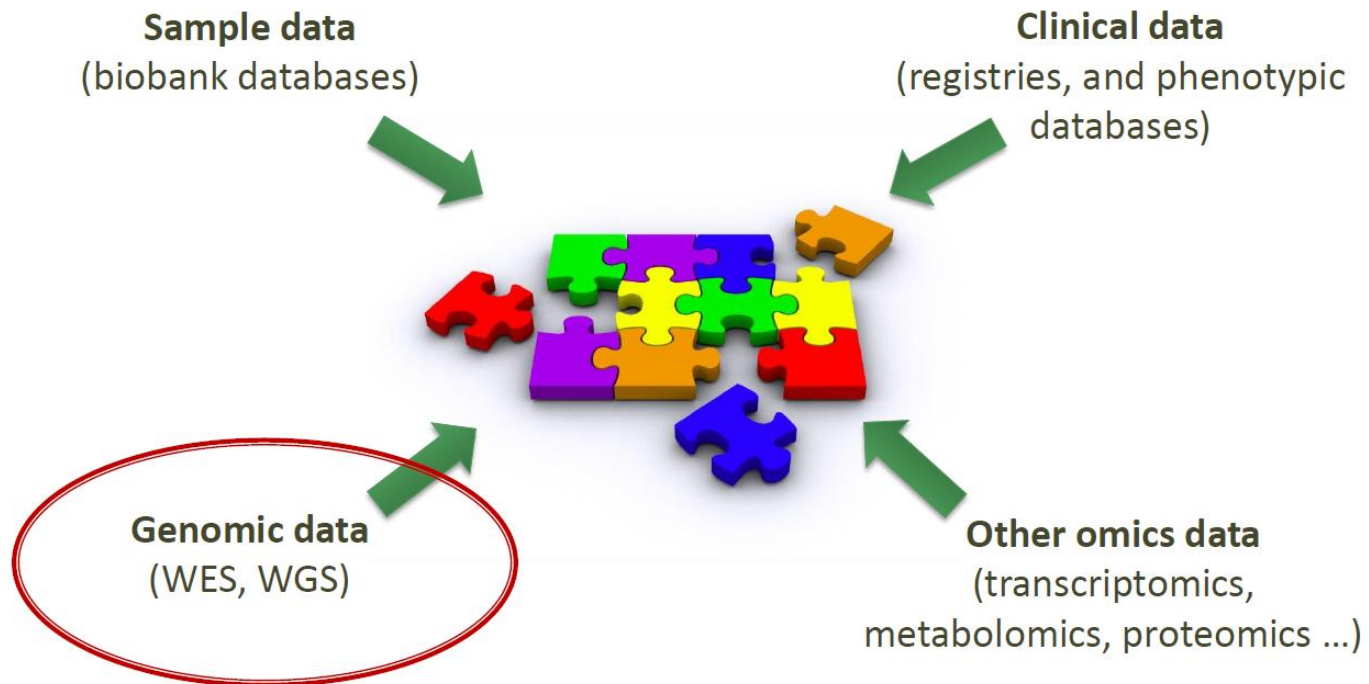Scientists University of Cologne uncover cellular mechanisms of SMA...

RD-CONNECT

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Collaborating to Advance Rare Diseases Research

NCATS and Harvard are collaborating to advance rare diseases research to benefit patients.

More...

Work with Us

Find out more about how your organization can collaborate with the GRDR to advance rare diseases research.

Henrietta Hyatt-Knorr, M.A. ✉

The NIH/NCATS GRDR® Program

The aim of the GRDR program is to develop a Web-based resource that aggregates, secures and stores de-identified patient information from different registries for rare diseases, all in one place.

Access NCATS Expertise & Resources

Find NCATS Programs & Initiatives

About the GRDR

Find out how GRDR experts are creating a resource for rare diseases researchers

Common Data Elements

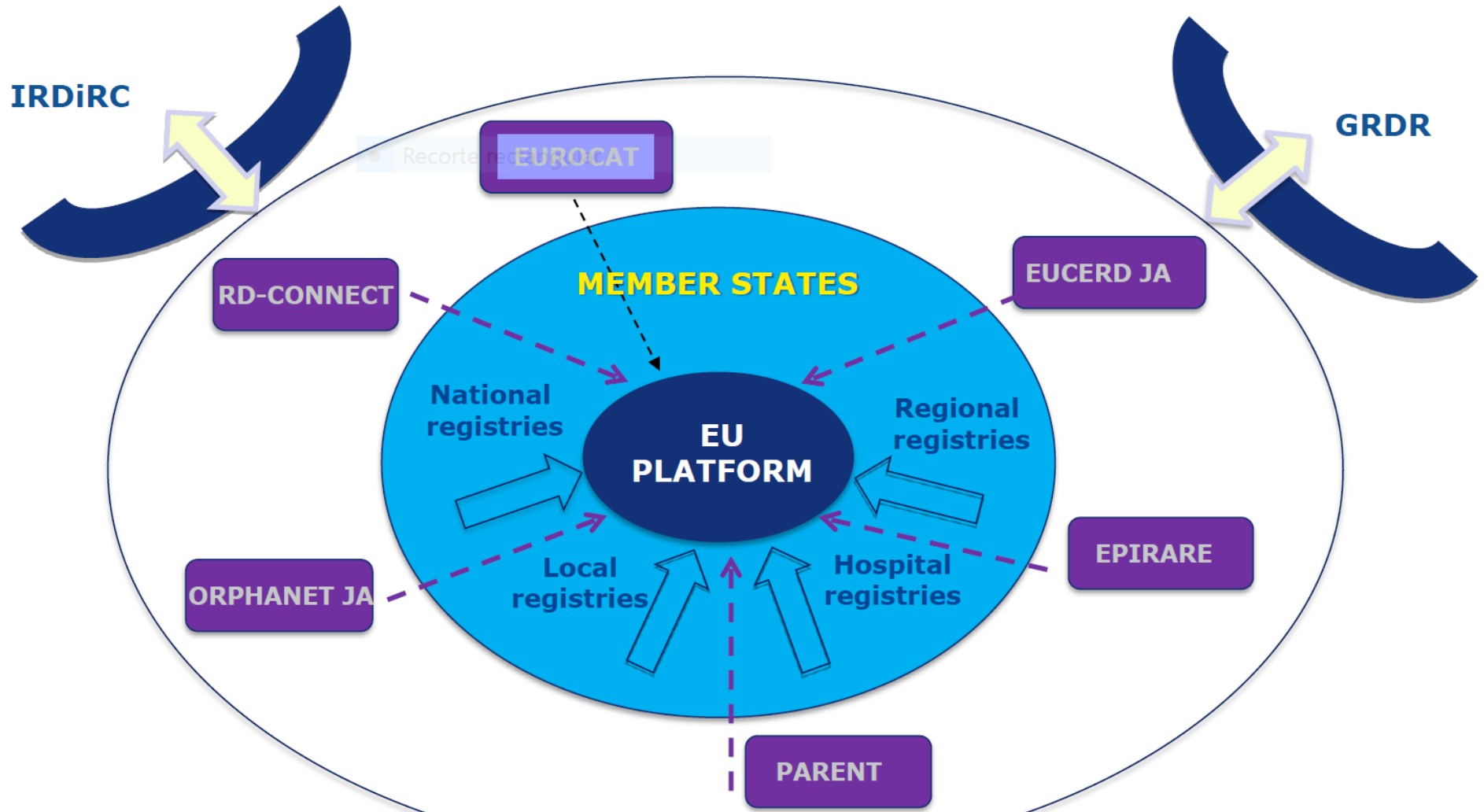Learn about CDEs and why they are crucial to global patient registries.

GRDR Resources

Get more information about the GRDR and access data submission forms.

Building the Platform: collaborations and interactions

Summary

- **RD Data sharing is challenging but also it is the future**
- **Recognize different approaches**
  - **To assess the best convenient**
- **Considering challeging topics**
  - **Case ascertainment/sources of information**
  - **Quality plan: reliability data & completeness**
  - **Interoperable**
  - **Validity**
  - **ELSI - PUID**
  - **Sustainability**

Thank you

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