I+D y Comercialización de Fármacos para Enfermedades Raras

El papel de los registros y plataformas ómicas de enfermedades raras en el desarrollo y autorización de medicamentos huérfanos

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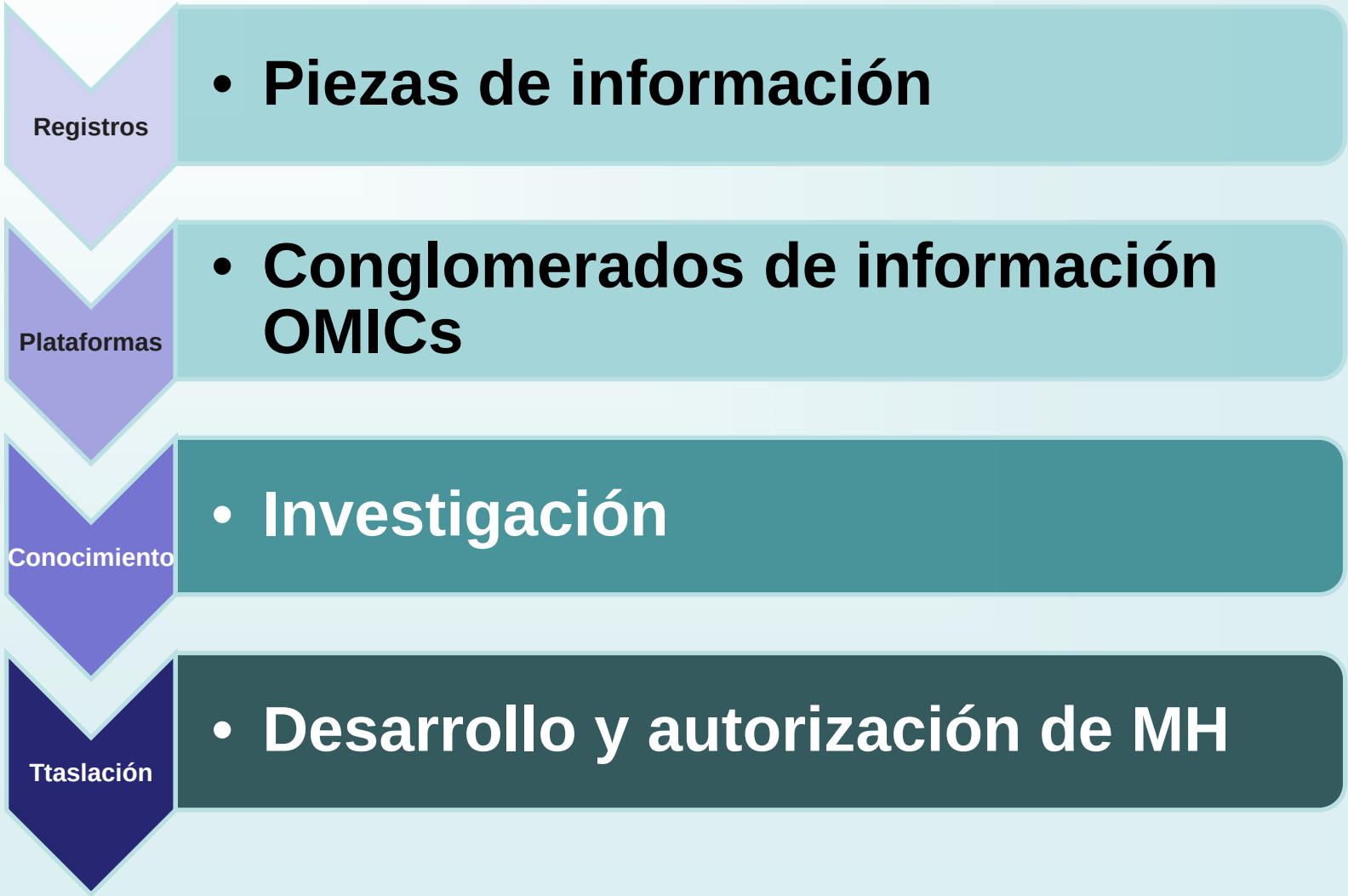
Puntos Clave



Registros: Definición

- **Los registros son herramientas epidemiológicas**
- **Basadas en métodos observacionales**
- **Utilizan la recogida sistemática de datos sobre el comienzo y desarrollo de la enfermedad**
- **Objetivo de favorecer la**
  - **Investigación etiológica**
  - **Investigación clínica**
  - **Contribuyen a la planificación de los recursos socio-sanitarios**

Registro ≠ Base de datos

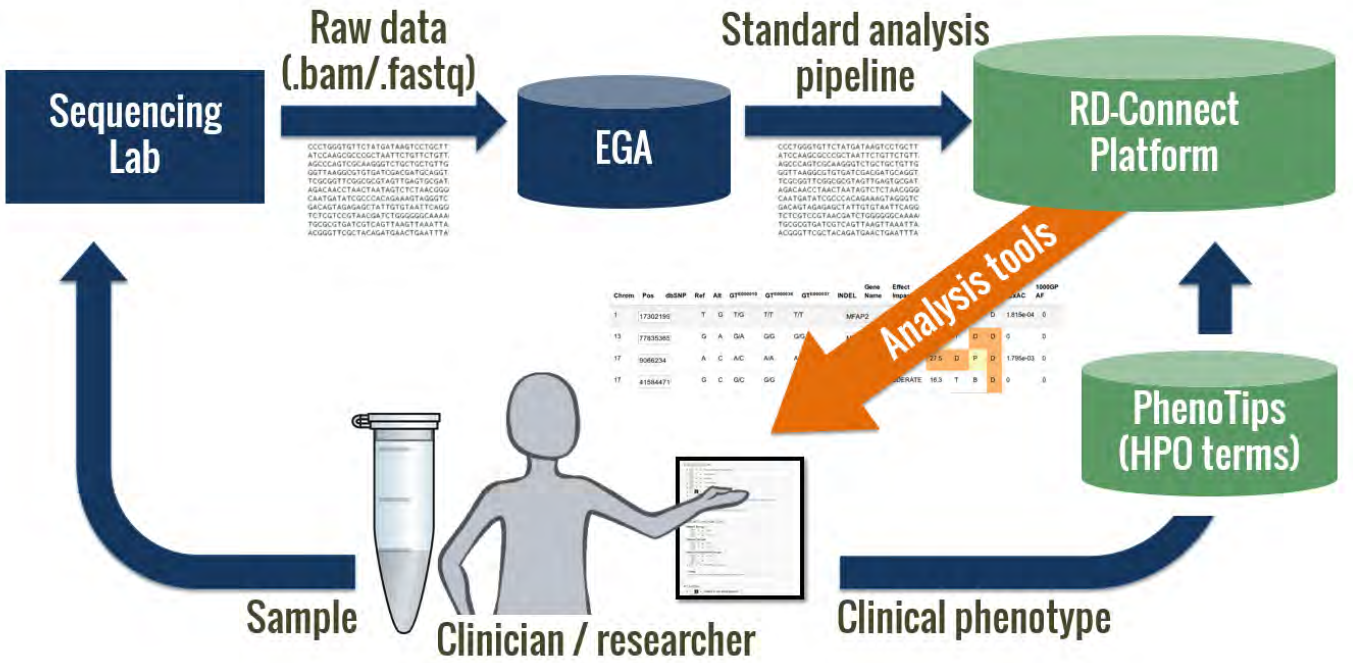
RD Connect

Derrotar a los Silos de información Compartir datos para investigar y mejorar los análisis

RD Connect

Análisis genómico y descubrir nuevos genes	Colección de datos de fenotipo normalizados	Catálogos de biobancos	Unión de datos a lo largo de los recursos

Datos Omics, datos clínicos y muestras biológicas individuales de pacientes de EERR



Variantes que causan enfermedad pueden ser identificadas usando la **Plataforma de análisis genómico**

Las muestras pueden ser localizables en un **Catálogo de Muestras**

Registros de datos en el directorio del **ID-Cards directory** - registros y biobancos

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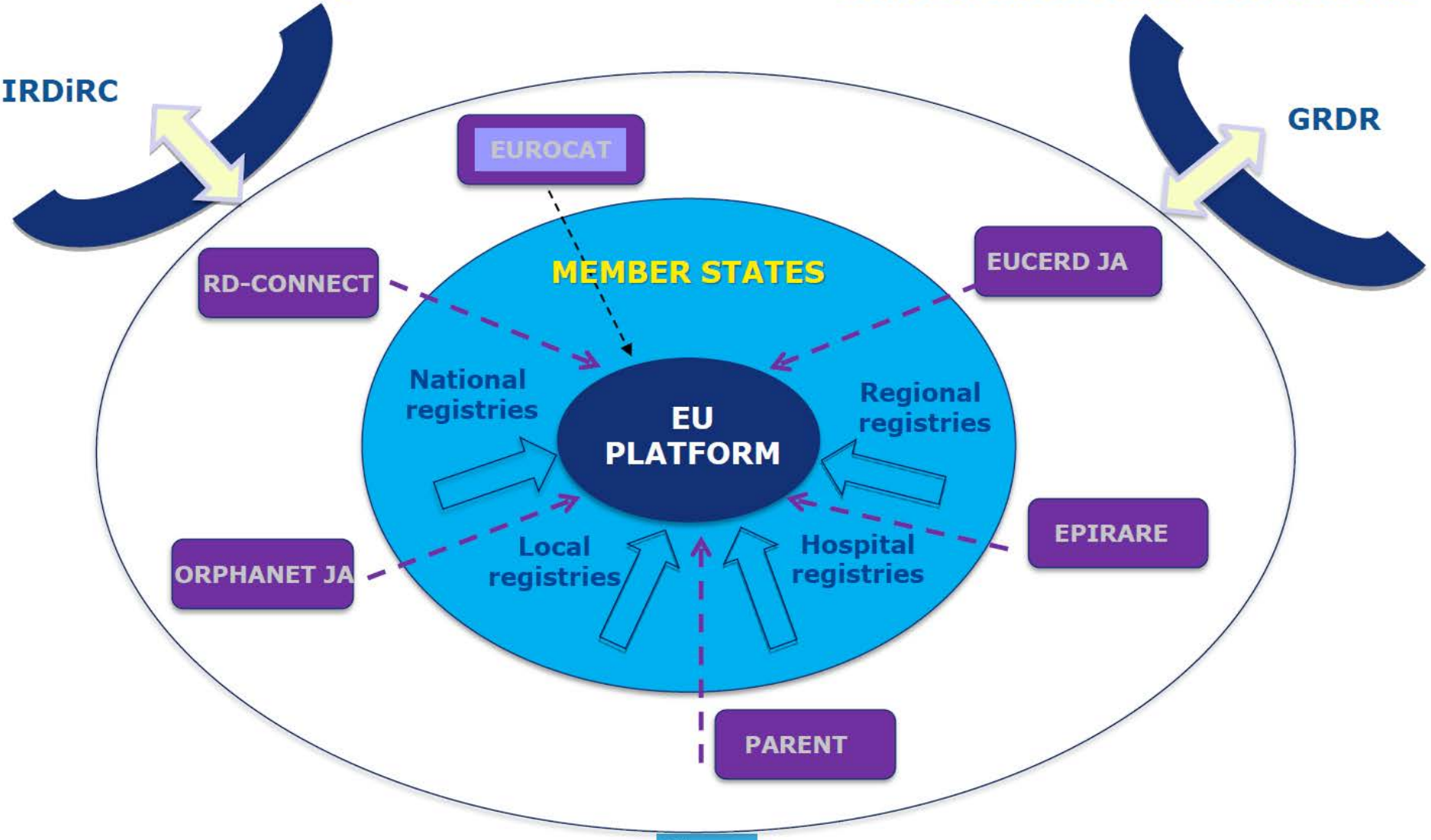
European Commission

Building the Platform: collaborations and interactions



Joint Research Centre

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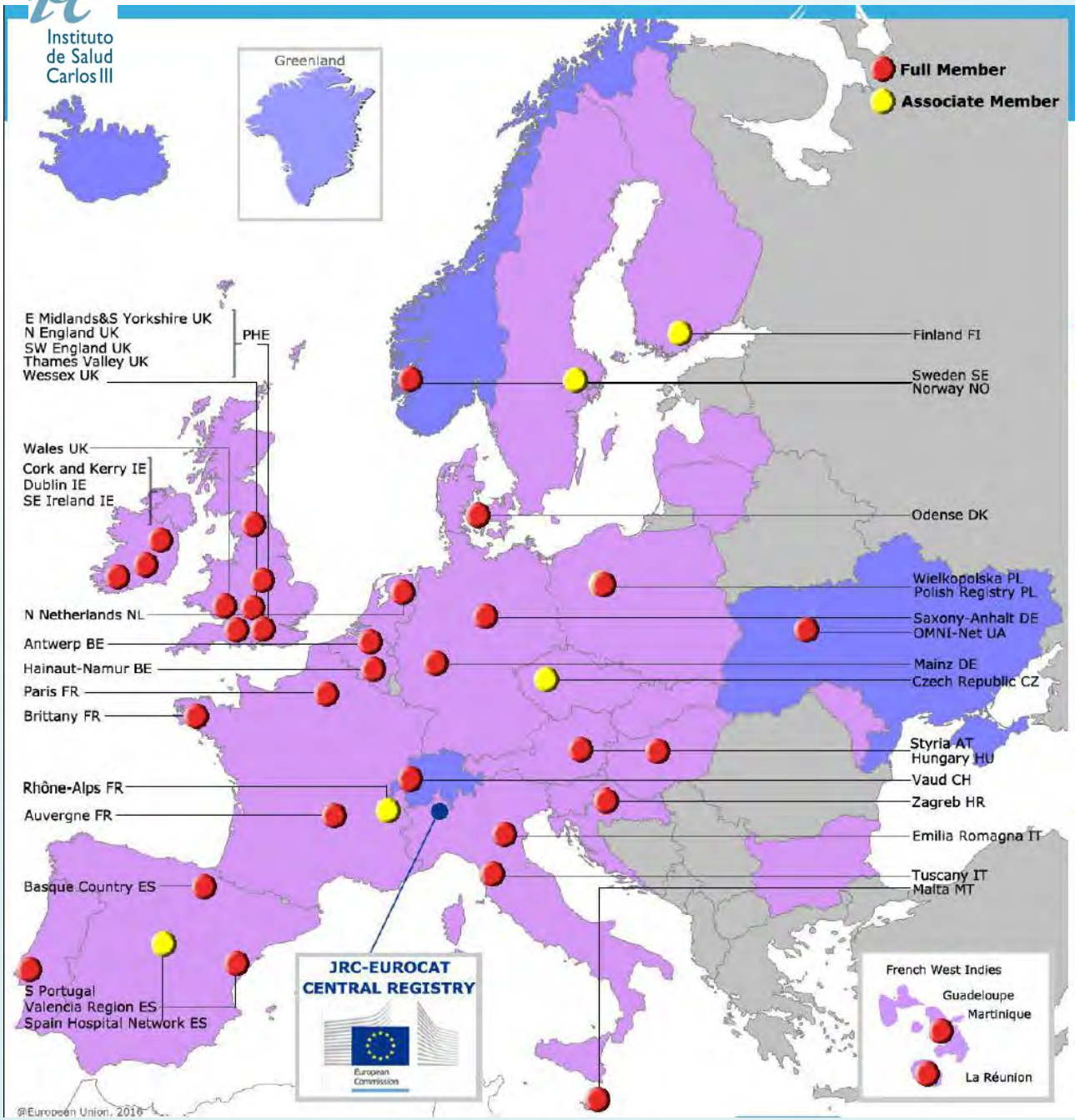


FULL MEMBERS

- 32 registries in 18 countries
- population-based registries transmitting case data on all congenital anomaly (CA) cases in their region.

ASSOCIATE MEMBERS

- 6 registries in 6 countries
- transmit an aggregate file containing the total number of cases in each congenital anomaly subgroup by type of birth.

Global Rare Diseases Patient Registry Data Repository

GRDR

Registros de Pacientes

1. Los pacientes proporcionana información en salud y resultados de tests a través de los DCE

2. Se asigna un Global Unique Patient ID (GUID);

3. Los datos del pacientes son unidos a sus mustras biológicas

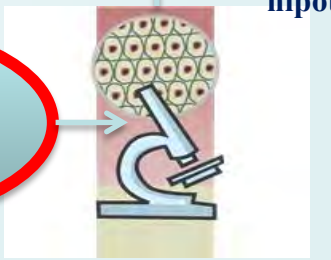
Nuevos Registros

Registros existentes

GUID

Repositorio Datos agregados y de-identificados

Bio especímenes



4. GRDR agrega información clínica codificaday datos de las muestras

5. Datos codificados y su utildiad para estudios

6. Los investigadores identificabn possible hipòtesis de trabajo

7. Notificación a los pacientes de posibles proyectos

NIH National Center for Advancing Translational Sciences

Pacientes

Investigadores

CCAA

PACIENTES

PROFESIONALES

ADMINISTRACIÓN

Registro de pacientes

INVESTIGACIÓN

Registro de Base Poblacional

PLANIFICACIÓN EN SALUD

Historia natural de la enfermedad

Seguimiento

Ensayos clínicos (reclutamiento)

Muestras biológicas

REGISTRO NACIONAL DE ENFERMEDADES RARAS

Políticas de salud

Prevalencia

Incidencia

Mortalidad

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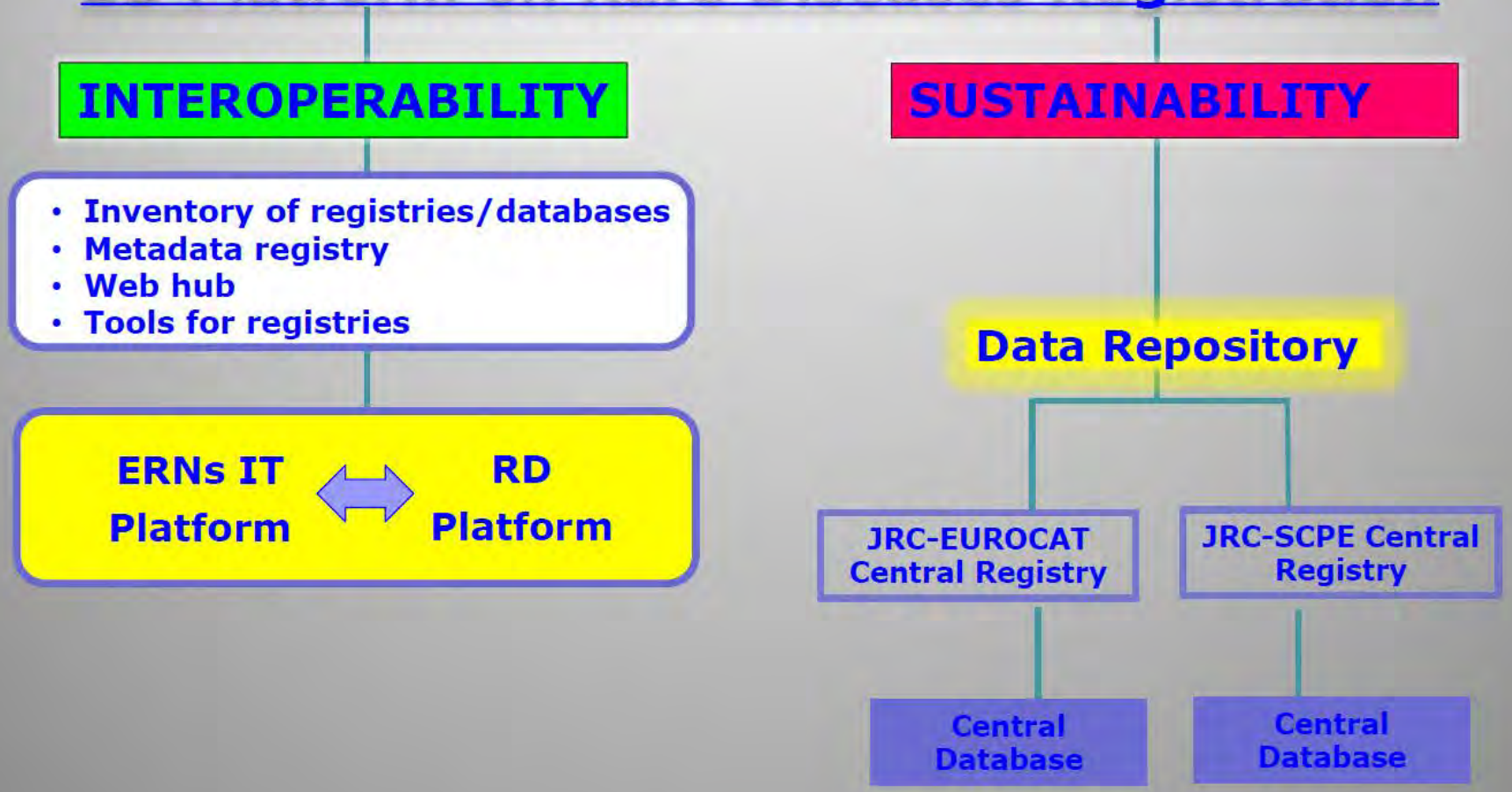
EU Platform on Rare Diseases Registration



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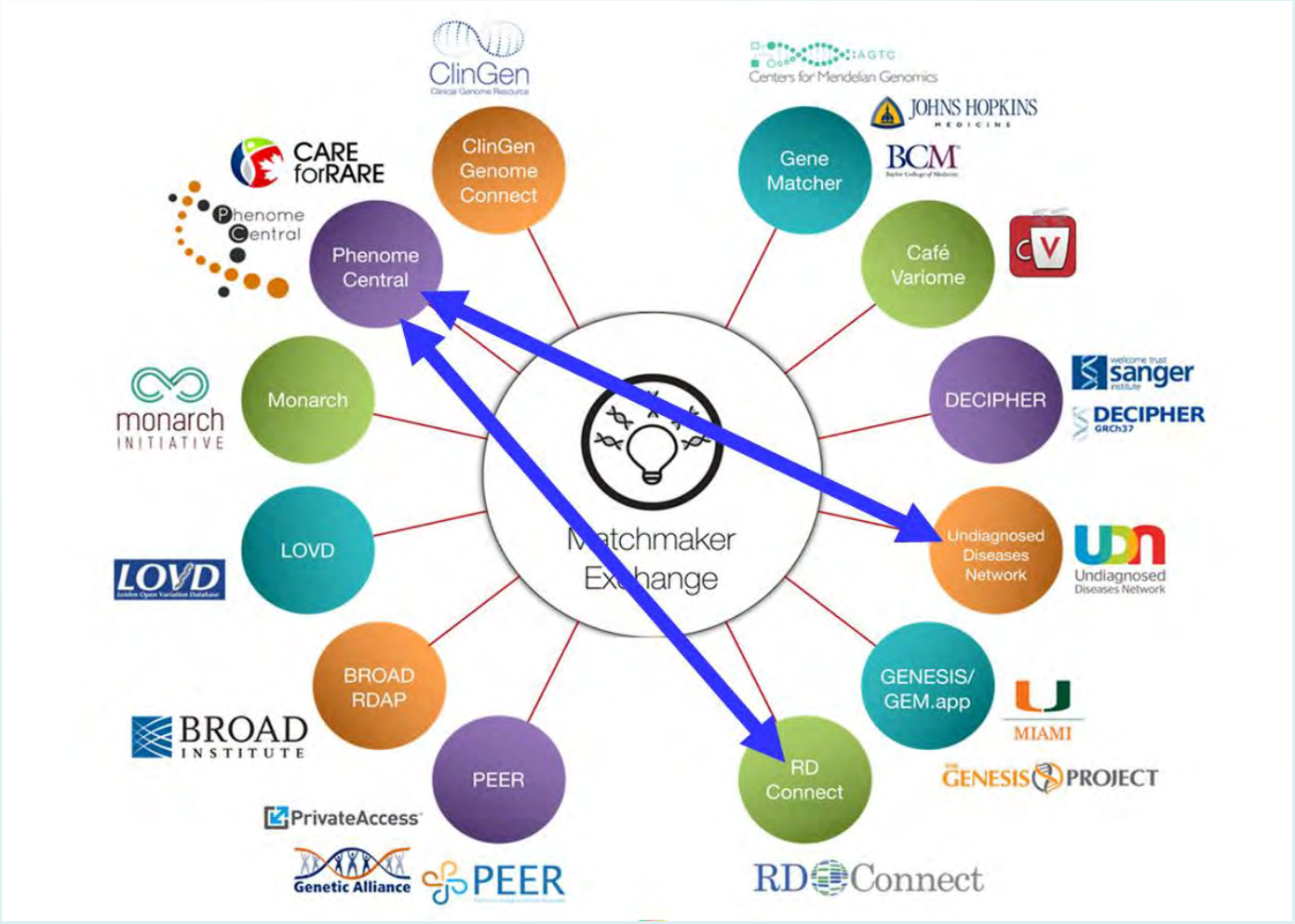
Interoperabilidad en el marco de la Plataforma MatchMaker Exchange



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2013 EURORDIS POLICY FACT SHEET – RARE DISEASE PATIENT REGISTRIES

RARE DISEASE PATIENT REGISTRIES

EURORDIS Rare Diseases Europe

Rare Disease Patient Registries represent a fundamental research effort upon which a number of critical activities are based. They constitute key instruments for increasing knowledge on Rare Diseases (RD) by pooling data for fundamental and clinical research, epidemiological research, and real-life post-marketing observational studies[1]. They broadly support health and social service planning by playing a pivotal role in healthcare organisation. They also represent a necessary infrastructure for the implementation of the European Reference Networks for rare diseases, and as such they represent a top priority[2] for the RD community at a National, European and International level. Furthermore, Patient Registries are one of the main pillars of the current EU policy framework on National Plans for RD[3]. EURORDIS holds Patient Registries as an advocacy priority and is actively participating in the major EU projects[4, 5, 6]. In the field, shaping and implementing an EU coordinated strategy on registries that will be patient-centred.

WHY ARE REGISTRIES SO IMPORTANT FOR RD PATIENTS?

It has been demonstrated that Patient Registries are a major determinant for successful translational research in the field of RD. Where well-implemented registries and active patient organizations exist, the likelihood for developing a treatment for the disease in question is increased[7]. Furthermore, the consistent longitudinal collection of patient data facilitates the creation of standards of care and dramatically improves patient outcomes and life expectancy even in the absence of new therapies. These compelling arguments for Rare Disease Patient Registries as indispensable infrastructure tools for translating basic and clinical research into therapeutic solutions have elevated their status to a major priority for all stakeholders, making them a building block of any sound policy on RD.

HOW TO ADDRESS THE ISSUE?

Being very thorough research infrastructures, Patient Registries require significant time and human resources as well as conspicuous financial investment and sustainability plans. Standard operating procedures and common resources or platforms for centralizing new or existing registries should be developed. Collaborative efforts at all levels are paramount to establish and manage Rare Disease Patient Registries and derive from them meaningful outcomes in the most efficient manner and for the uppermost benefit of patients. Nevertheless, no uniform, accepted standards currently govern the collection, organisation, or availability of data collected by Rare Disease Patient Registries. The existence of conspicuous data-sharing barriers creates a compelling argument for developing globally accepted definitions, classifications, data standards and favourable and congruent policies and resources facilitating data sharing and pooling.

DEVELOPING RARE DISEASE PATIENT REGISTRIES: THE 10 MAJOR PRINCIPLES PROMOTED BY EURORDIS[8]

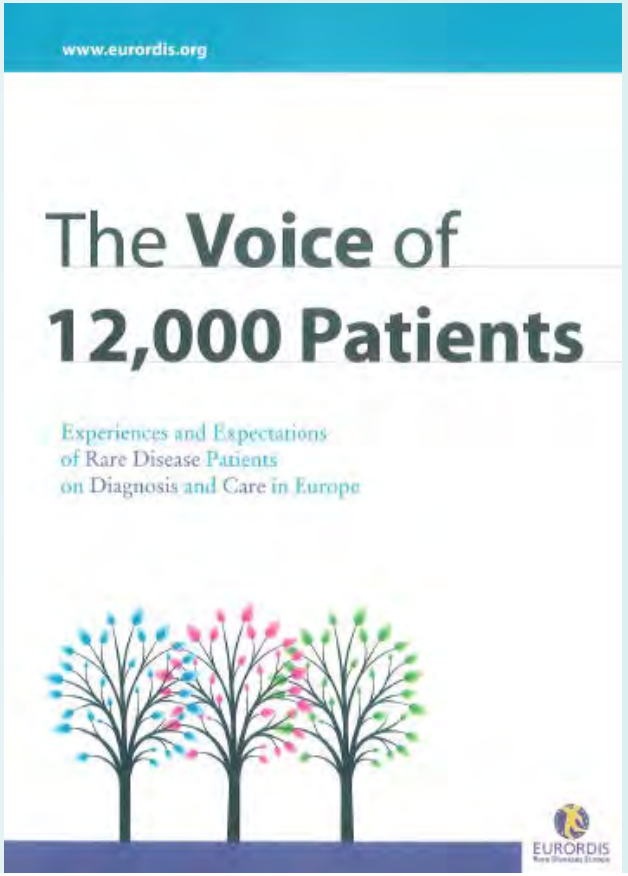
* Patient Registries should be recognised as a global priority in the field of Rare Diseases.
The priority for establishing and supporting them has become more and more compelling and urgent for all stakeholders in order to achieve their different but complementary goals aimed at augmenting knowledge and developing new therapeutic breakthrough in the field of RD.

* Rare Disease Patient Registries should encompass the widest geographic scope possible. The benefits of international collaboration and maximization of limited resources are most obvious for rare diseases, especially the very rare ones, due to the low individual prevalence and the scarcity of information related to each of them.

* Rare Disease Patient Registries should be centred on a disease or group of diseases rather than a therapeutic intervention. Disease focused registries ensure higher quality and completeness of data and allow fulfilling post-marketing requirements, especially for regulators and payers, [illegible] long-term outcomes and cost-effectiveness of new medi[illegible]

* Interoperability and harmonization between R[illegible] Patient Registries should be consistently pur[illegible] become a compelling necessity to develop globally accep[illegible] classifications, data standards and favourable a[illegible] policies and resources facilitating data sharing and p[illegible] standard operating procedures and common resource[illegible] for centralizing new or existing registries should be dev[illegible]

* A minimum set of Common Data Elements (CDI[illegible] consistently used in all Rare Disease Patient Re[illegible] would facilitate the standardization of data (same d[illegible] entry procedures), harmonization (facilitation of data [illegible] comparison), and interoperability (shared quality assur[illegible] security standards).



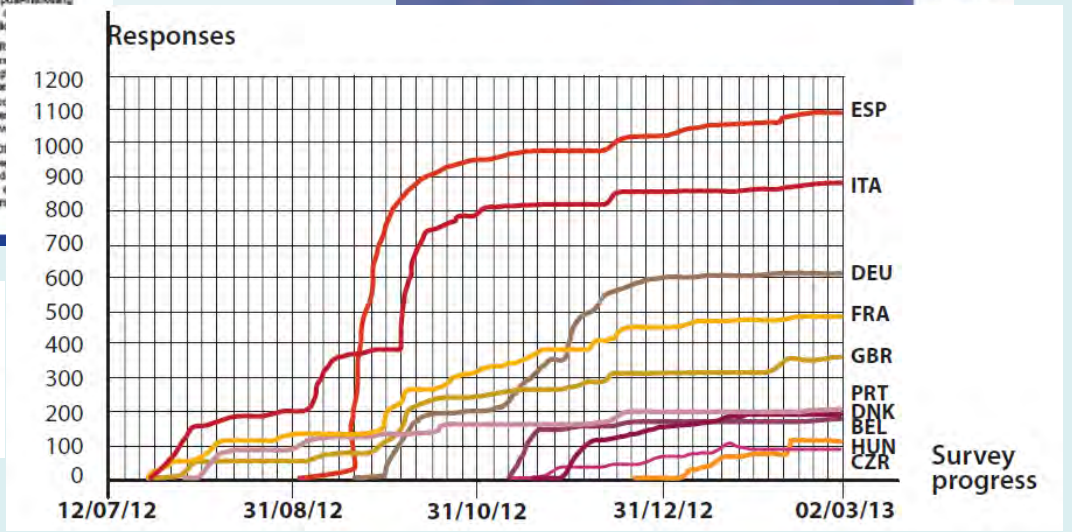


Figure 4.
Access to survey per language

Registros: Medicamentos Huérfanos

- **Informan y permiten**
  - **El análisis de riesgos, tendencias y variabilidad geográfica**
  - **Optimizan la inversión de recursos**
- **Promueven la innovación**
  - **Nuevas hipótesis: Etiología vs mecanismos**
  - **Relación causa-efecto: Medicina personalizada**
- **Promueve la investigación**
  - **Medicina centrada en el paciente y ensayos clínicos**
  - **Estudiar la historia natural de la enfermedad**
  - **Medidas de coste-efectividad e impacto del MH**
  - **Control de los efectos secundarios**

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GRACIAS

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