

**Directorate-General
Joint Research Centre**

**Directorate-General
Health and Food Safety**

European Platform on Rare Diseases Registration

Knowledge generation centre for rare diseases

Joint Research Centre, Directorate F – Health, Consumers and Reference Materials
Unit F.1 – Health in Society

Why an EU Platform on RD Registration?

Rare Diseases:

- ✓ Low prevalence: less than 5 per 10,000 persons (EU)
- ✓ > 6,000 distinct rare diseases
- ✓ **30 million people affected in the EU**
- ✓ Limited number of patients per center/country
- ✓ Patients scattered across EU
- ✓ Scattered knowledge and expertise on diagnosis, treatment



- ✓ Extreme fragmentation of data sources (600-1000 registries) across EU Member States

Why an EU Platform on RD Registration?

➤ **To cope with the extreme fragmentation of data sources across EU Member States**

- Many RD registries exist, but
- the lack of interoperability severely limits the registries' potential
 - no standardised data collection for most RDs

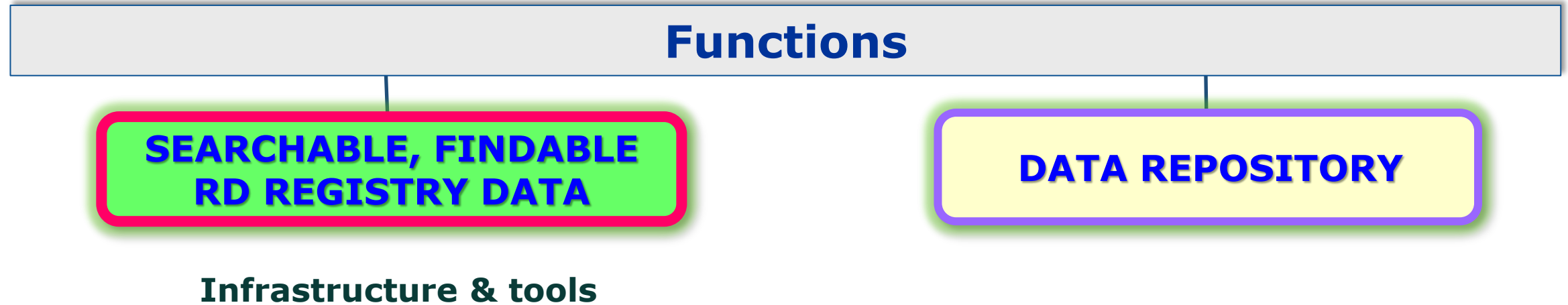
➤ **Benefits:**

Reach the critical number of patients for

- **studies (epidemiological, clinical, translational, pharmacological, etc.)**
- **research**



European Platform on Rare Diseases Registration



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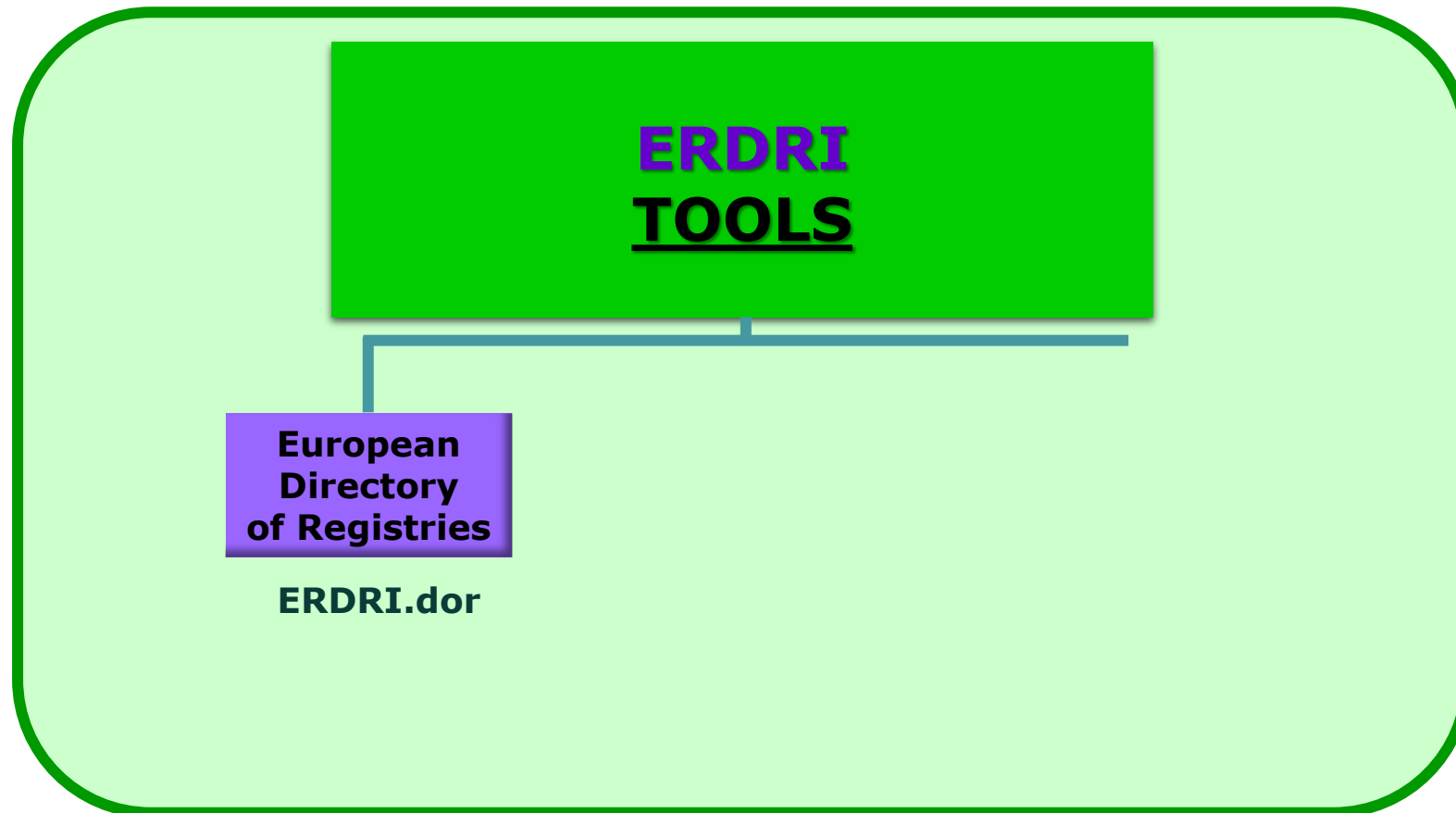
**SEARCHABLE, FINDABLE RD REGISTRY DATA
INTEROPERABILITY**

ERDRI

**European Rare Disease
Registry Infrastructure**

European Platform on Rare Diseases Registration

INTEROPERABILITY



European Platform on Rare Diseases Registration

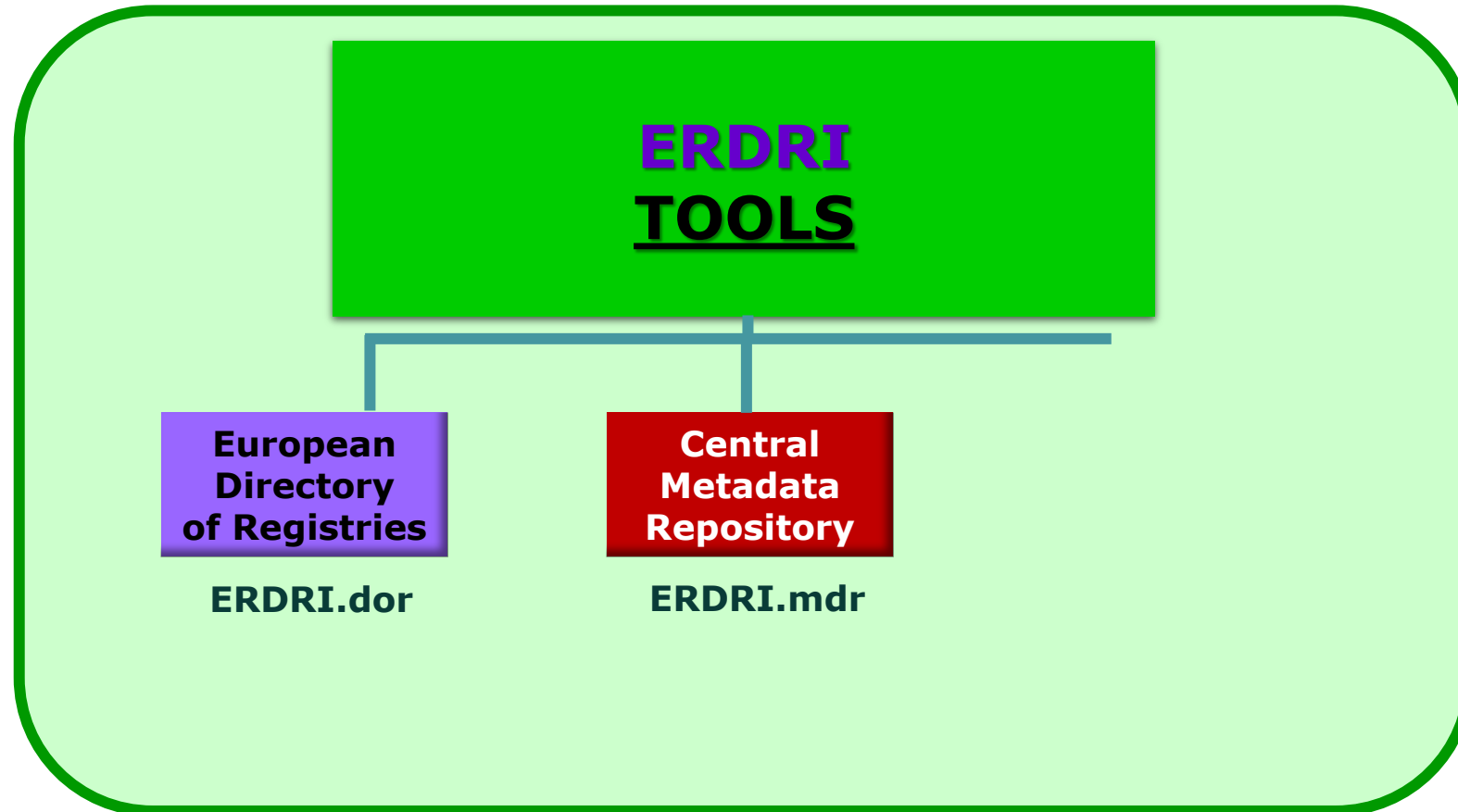
ERDRI.dor – The European Directory of RD registries

- List of participating RD registries
- Descriptive information
 - specific rare disease addressed
 - scope
 - operating institution
 - contact information, etc.
 -
 - = characteristics of registries (27)

**CORE COMPONENT OF
ERDRI / EU RD PLATFORM**



European Platform on Rare Diseases Registration



European RD Registry Infrastructure (ERDRI)

ERDRI.mdr – The Central Metadata Repository

Semantic interoperability

Central Metadata Repository ERDRI.mdr

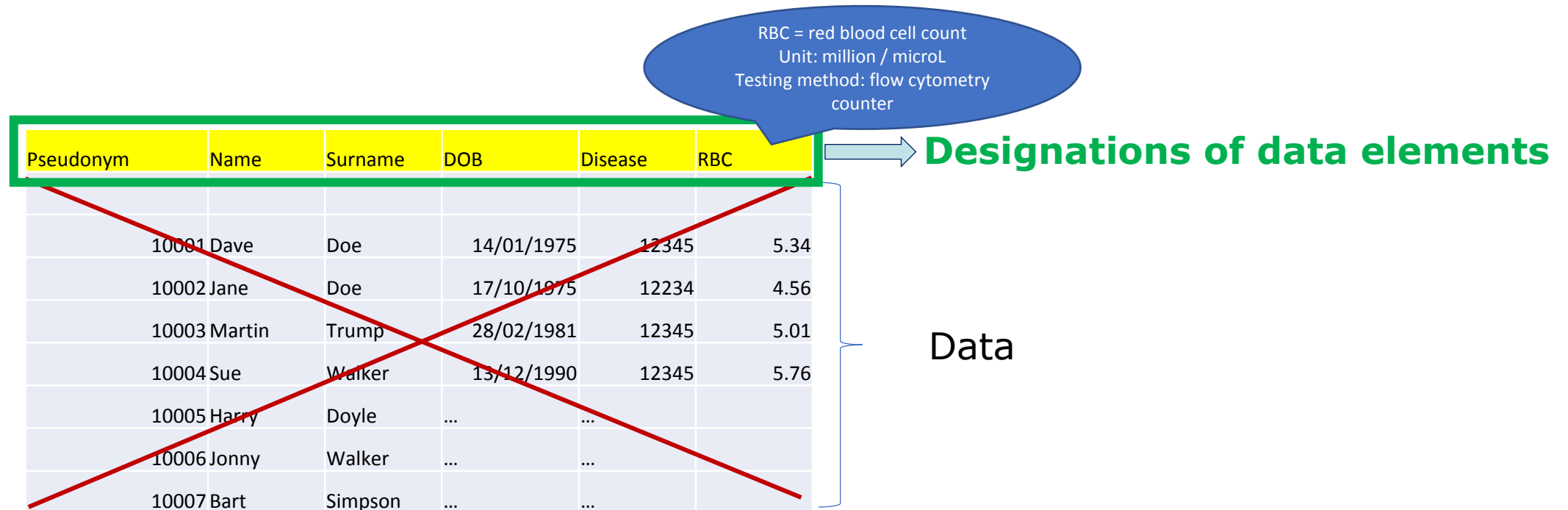


collection of metadata on all data elements collected by participating registries

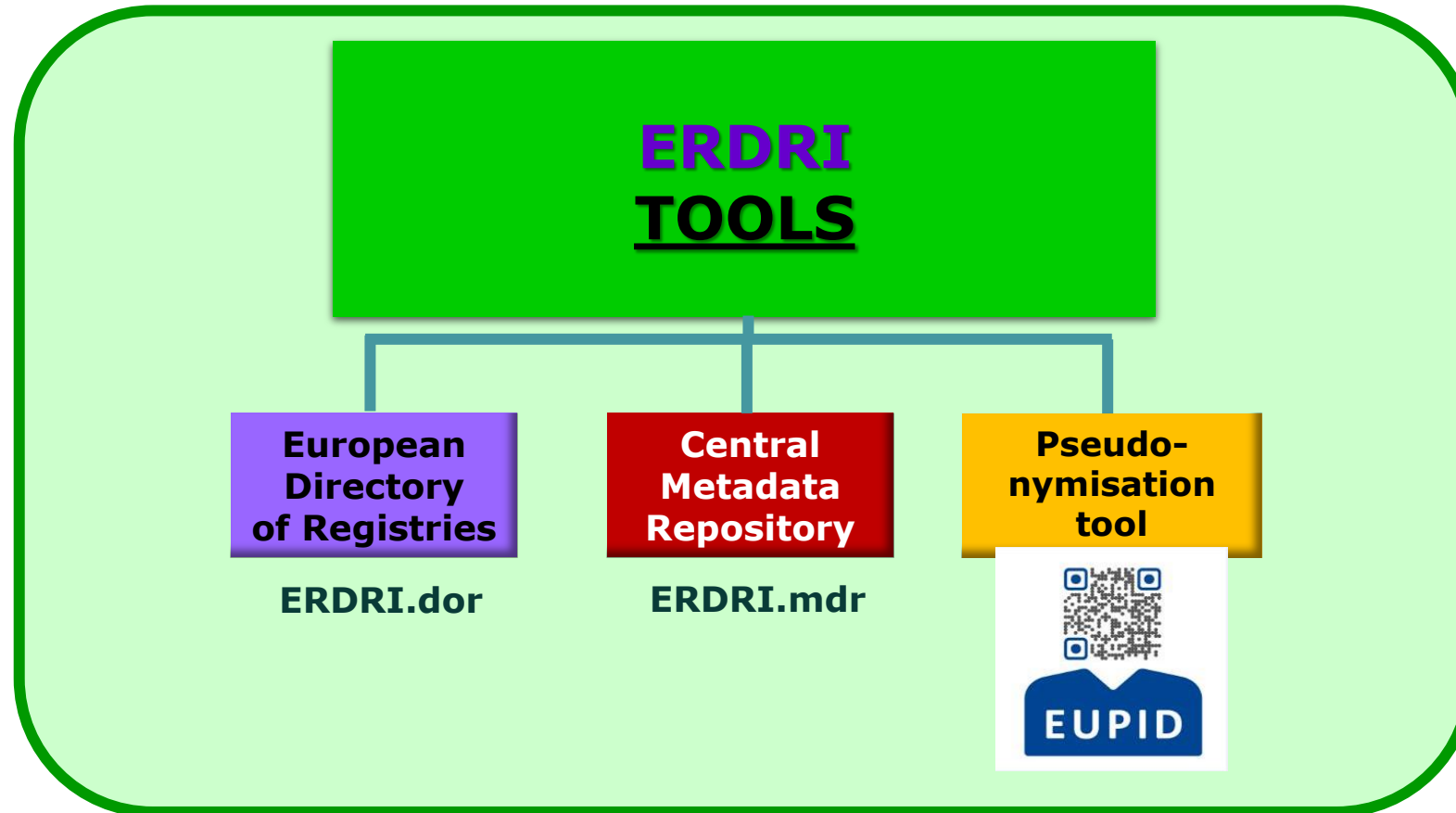
- Designation
- Definition
- Measurement unit + range

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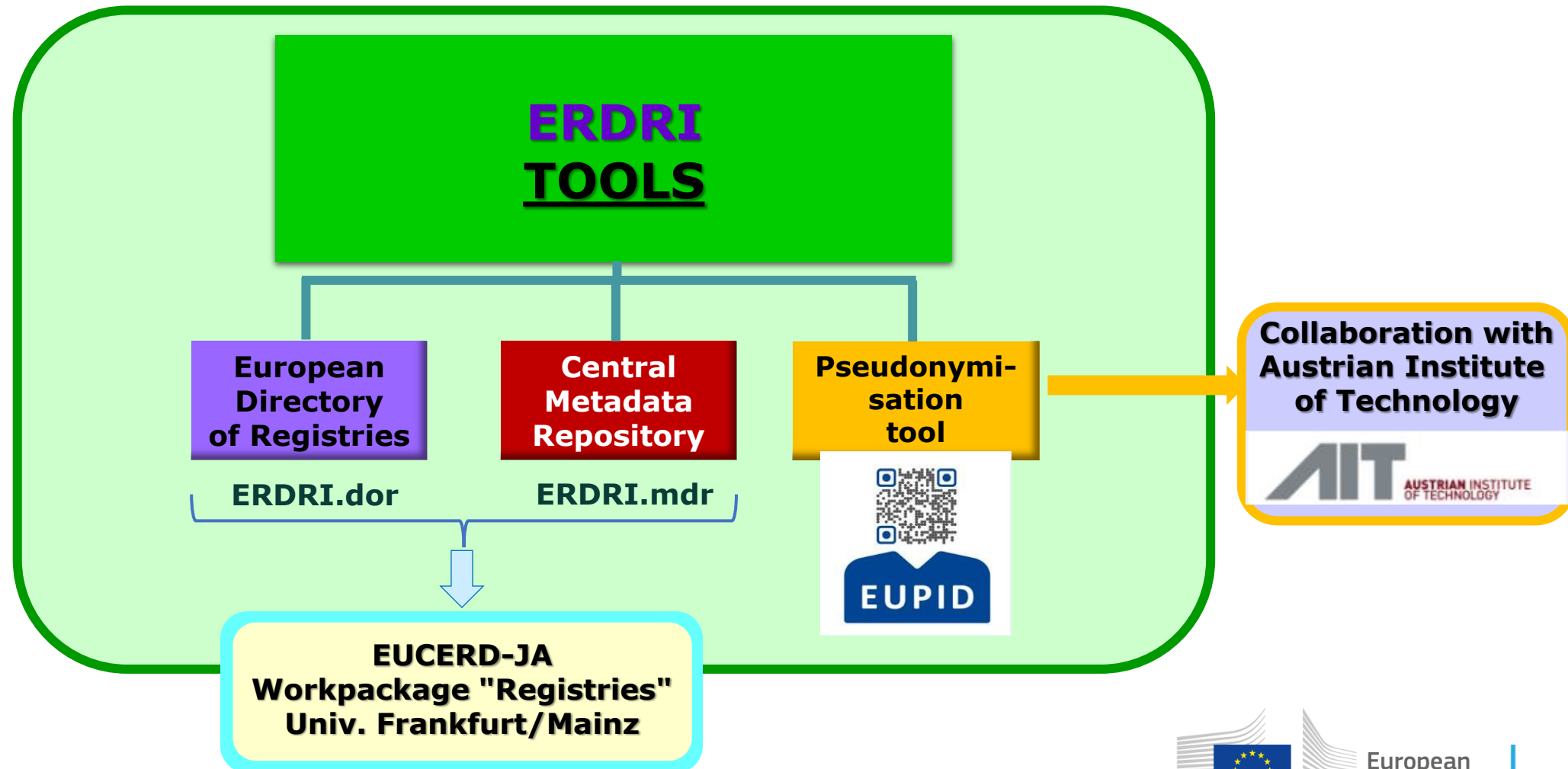
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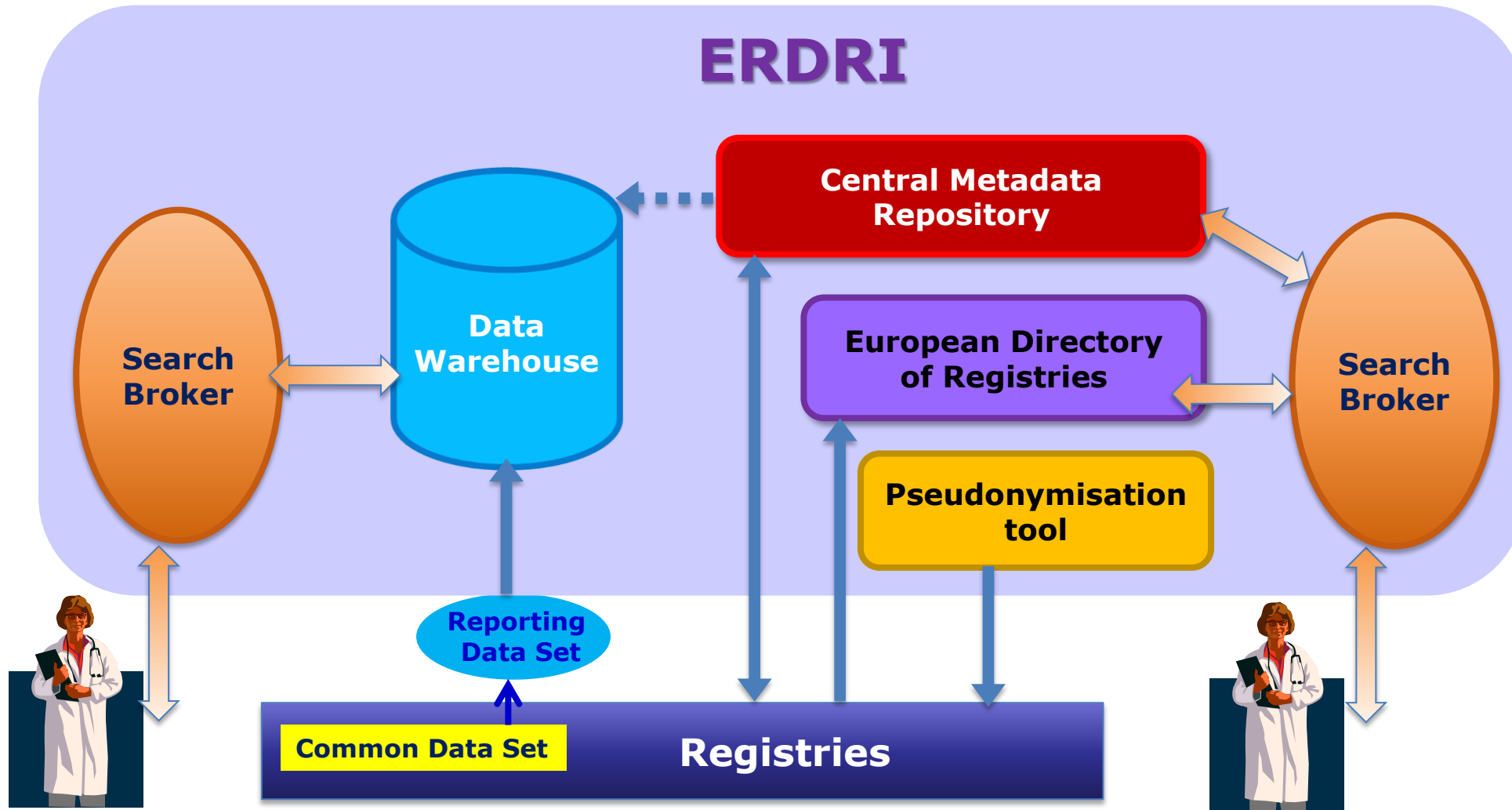
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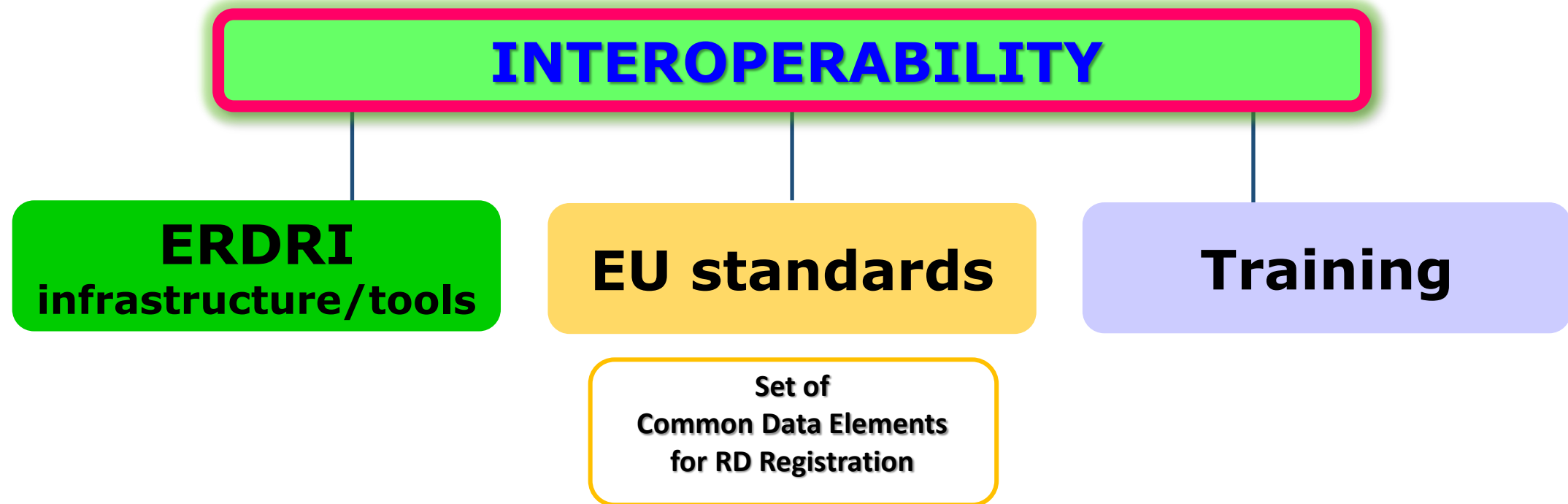
European Platform on Rare Diseases Registration

Makes RD patient data FAIR

**ERDRI provides the infrastructure and tools
to make registries' data**

F FINDABLE
A ACCESSIBLE
I INTEROPERABLE
R REUSABLE

European Platform on Rare Diseases Registration



European Platform on Rare Diseases Registration (EU RD Platform)

Providing a central access point for information on rare disease patients' registries to all stakeholders

Searchable, findable rare disease patient data



European Rare Disease Registry Infrastructure (ERDRI)

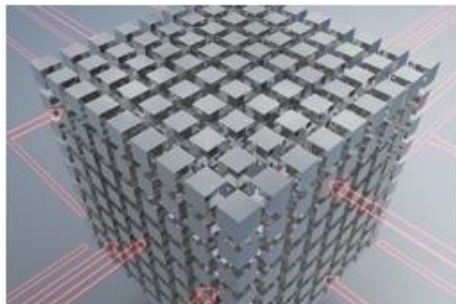


European standards for data collection and data sharing



Trainings
Events
Latest news

Data repository



European RD Registry Data Warehouse



Surveillance of Congenital Anomalies in Europe



Surveillance of Cerebral Palsy in Europe

Questionnaire to Member States on EU RD Platform

Responses received from 14 MS:

Austria, Belgium, Bulgaria, Cyprus, Croatia, Czech Republic,
France, Germany, Italy, Latvia, Lithuania, Malta, Slovakia, Spain

Thank you!

Questionnaire to Member States on EU RD Platform

Purpose of the survey completely different
from the survey on Orphanet

❑ Orphanet: funding

❑ EU RD Platform: { - interest in using the Platform
- dissemination of information
about the Platform

Questionnaire to Member States on EU RD Platform

Different purposes

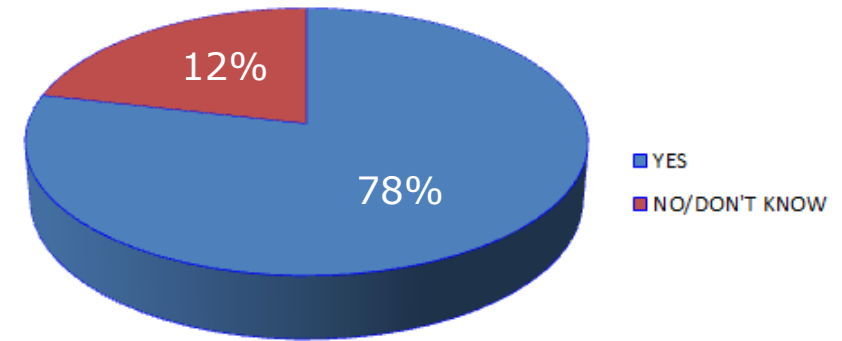
- ❑ Orphanet: diseases
- ❑ EU RD Platform: patients in RD registries

Questionnaire to Member States on EU RD Platform

- Q1.** My **country** would like to **join** the EU RD Platform, to use the infrastructure and services offered, which include **contributing** to the research and studies on rare diseases at the EU level
- Q2.** My country would like to **help registries** at local/regional/national level in my country to **join** the EU RD Platform by making them aware of the existence of the EU RD Platform and informing the Joint Research Centre of the European Commission about the registries interested to become part of it
- Q3.** My country might envisage **allocation of resources** to the registries at local/regional/national level in my country which join the EU RD Platform and thus **use the European standards** set by the EU RD Platform
- Q4.** My country might envisage **support for the registries** at local/regional/national level in my country **which join** the EU RD Platform (such as co-organising with the European Commission JRC education programmes for the registries' staff or other programmes)
- Q5.** My country would be interested to **sign a Memorandum of Understanding** with the European Commission JRC on the use of the EU RD Platform

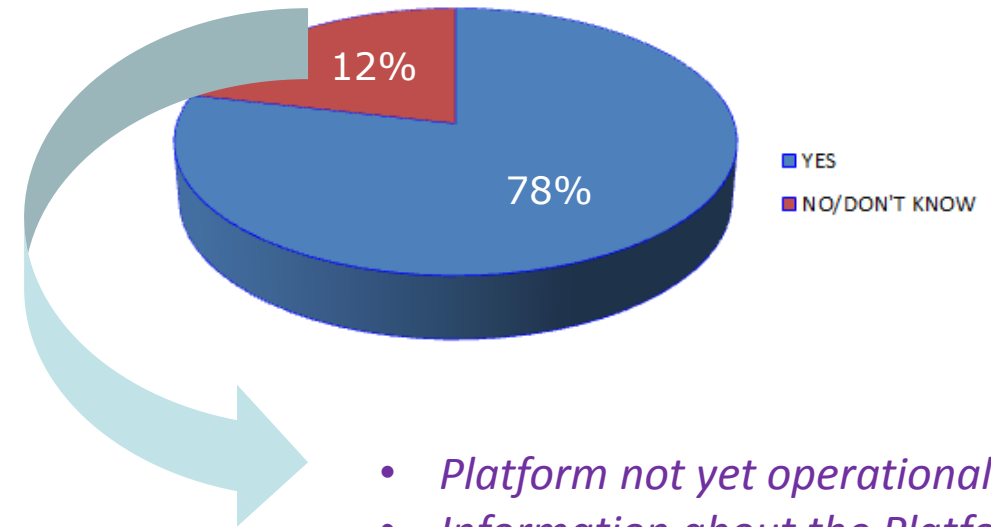
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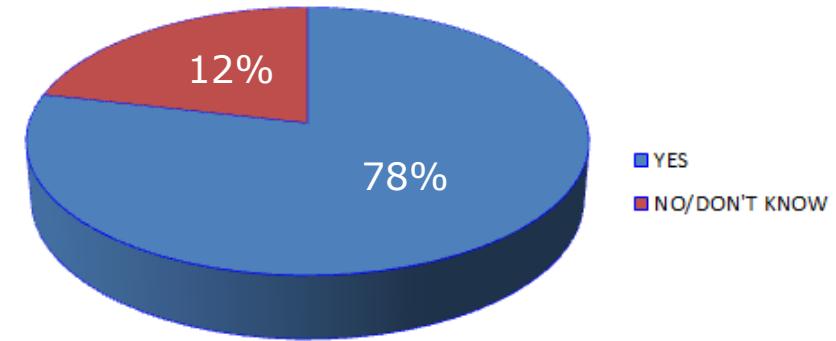
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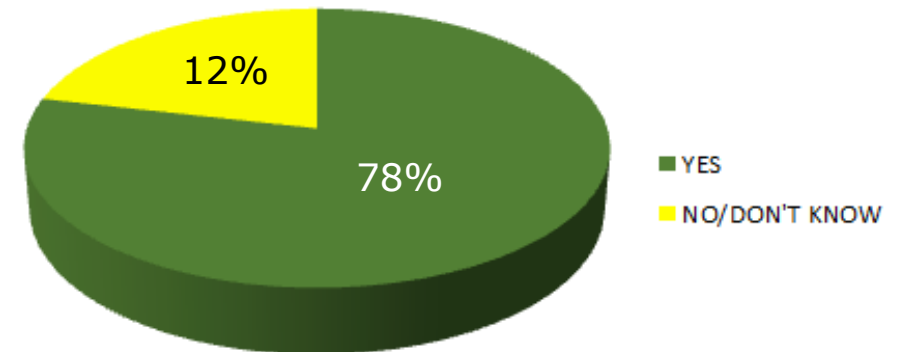
- *Platform not yet operational*
- *Information about the Platform*
- *Conditions for access?*
- *Financial consequences?*

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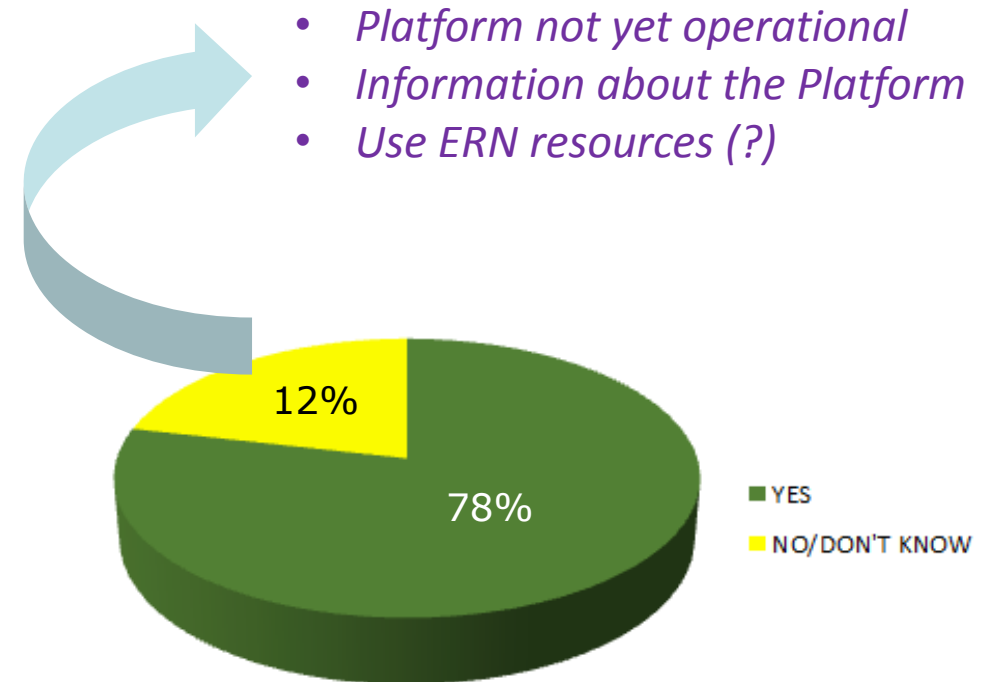


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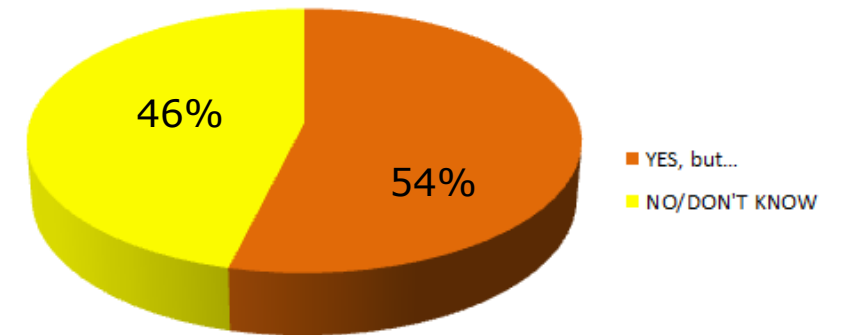
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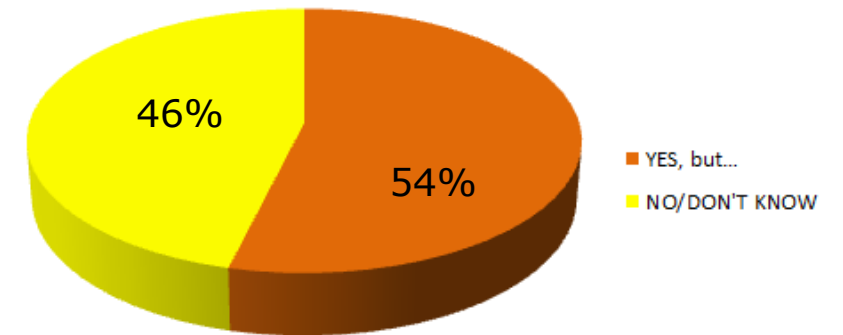
Questionnaire to Member States on EU RD Platform

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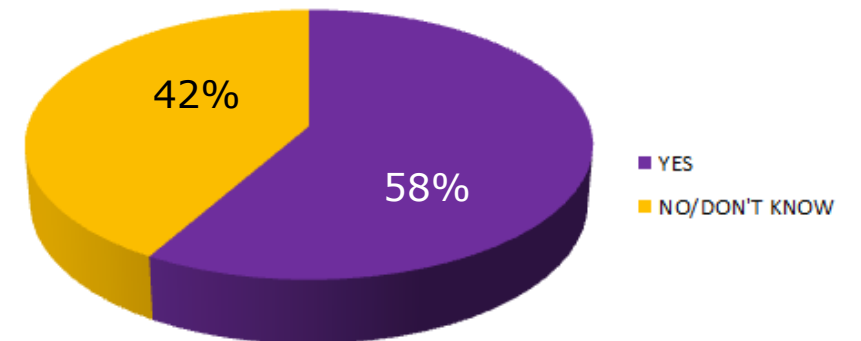


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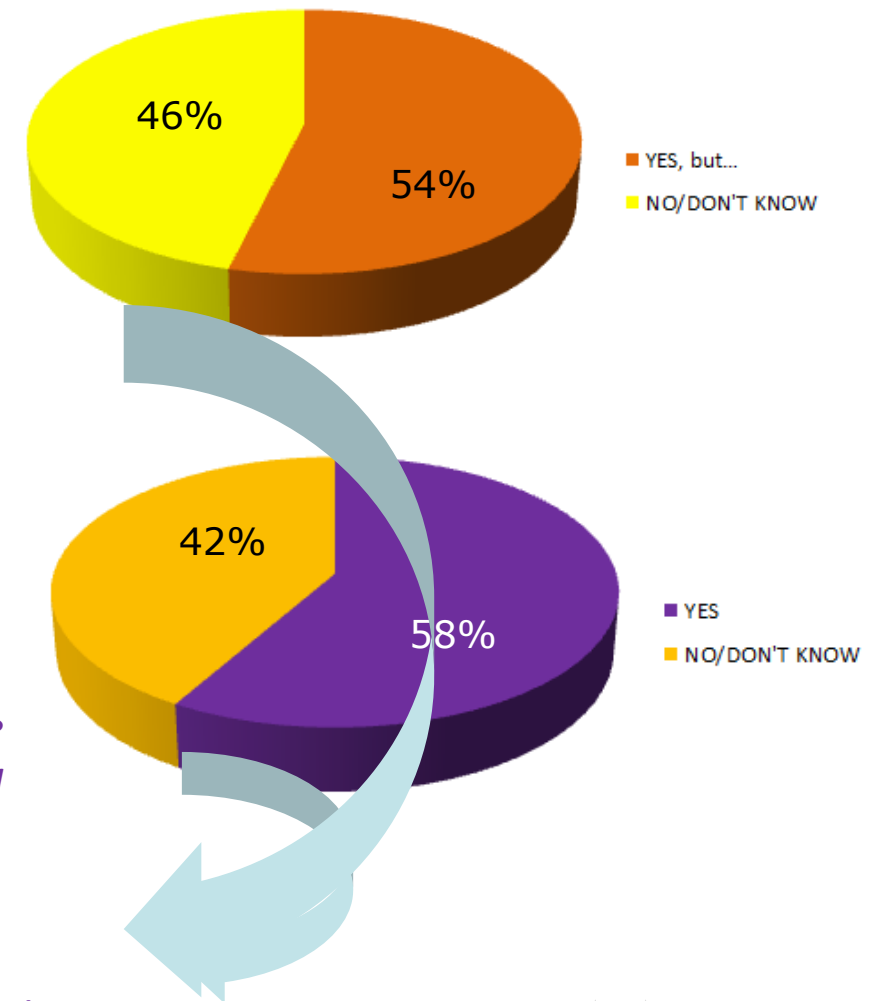


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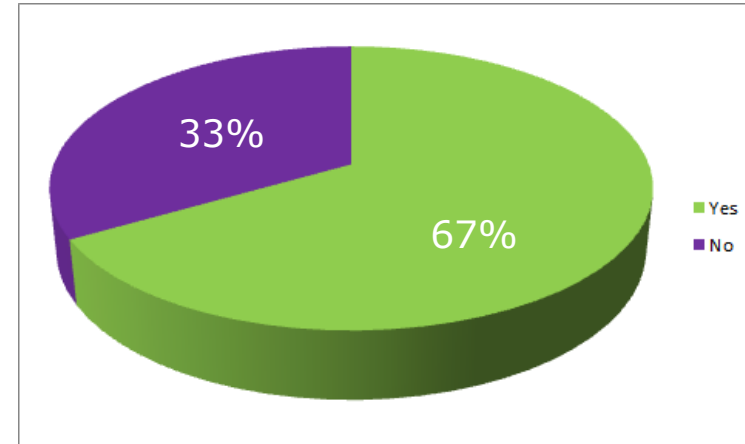
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- *No financial allocation in place*
- *Decision at regional/local level*
- *Platform not yet operational*
- *Use ERN resources (?)*
- *Additional financial support*
- *No national plans for RD registries*



Questionnaire to Member States on EU RD Platform

Q5. My country would be interested to **sign a Memorandum of Understanding** with the European Commission JRC on the use of the EU RD Platform



Questionnaire to Member States on EU RD Platform

EU RD Platform

- ❖ **all components are available free of charge to all interested registries**
- ❖ **is the answer to the needs expressed over years by the RD community (registries, national authorities, patients, regulators)**

Questionnaire to Member States on EU RD Platform

General comments:

- ❖ **majority of responders interested in using the EU RD Platform**
- ❖ **questionnaire released at an early timepoint, before the launch of the EU RD Platform**

European Platform on Rare Diseases Registration

**The EU RD Platform will be launched
on the Rare Disease Day
28 February 2019**