

The European Commission's science and knowledge service

Joint Research Centre

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Unit F.1 – 'Health in Society'



I have no actual or potential conflict of interest in relation to this presentation.

**Directorate-General
Joint Research Centre**

**Directorate-General
Health and Food Safety**

European Platform on Rare Diseases Registration (EU RD Platform)

Knowledge generation centre for rare diseases

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European Platform on Rare Diseases Registration (EU RD Platform)

Knowledge generation centre for rare diseases

- **Facilitating data exchange between RD registries**

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European Platform on Rare Diseases Registration (EU RD Platform)

Knowledge generation centre for rare diseases

- **Facilitating data exchange between RD registries**
- **Reaching the critical number of patients for**
 - **studies (epidemiological, clinical, translational, pharmacological, etc.)**
 - **research**

European Platform on Rare Diseases Registration

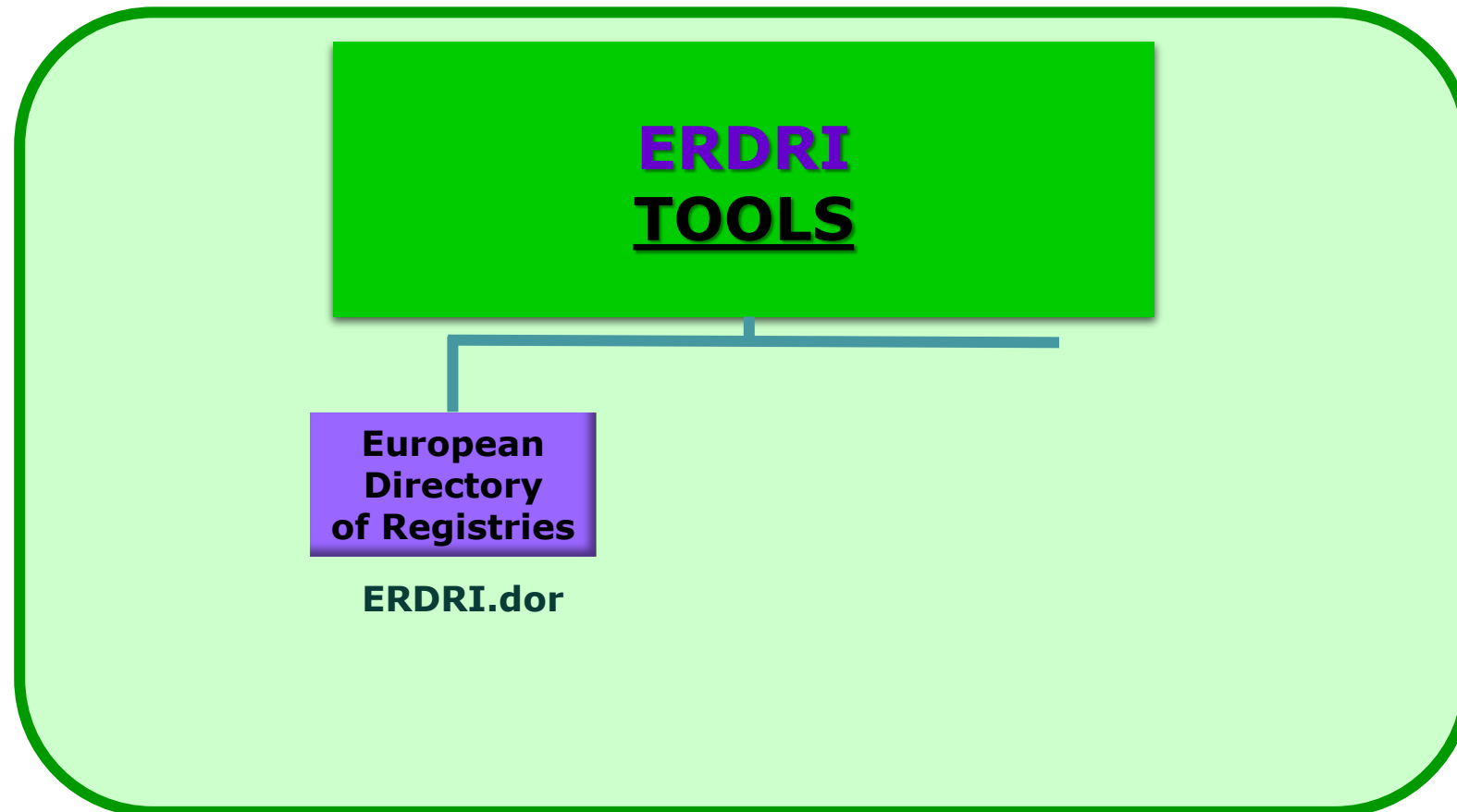
European Rare Disease Registry Infrastructure

ERDRI

In collaboration with EUCERD-JA WP 'Registries', University of Frankfurt

European Platform on Rare Diseases Registration

INTEROPERABILITY for RD data collection and exchange



European Platform on Rare Diseases Registration

ERDRI.dor – The European Directory of RD registries

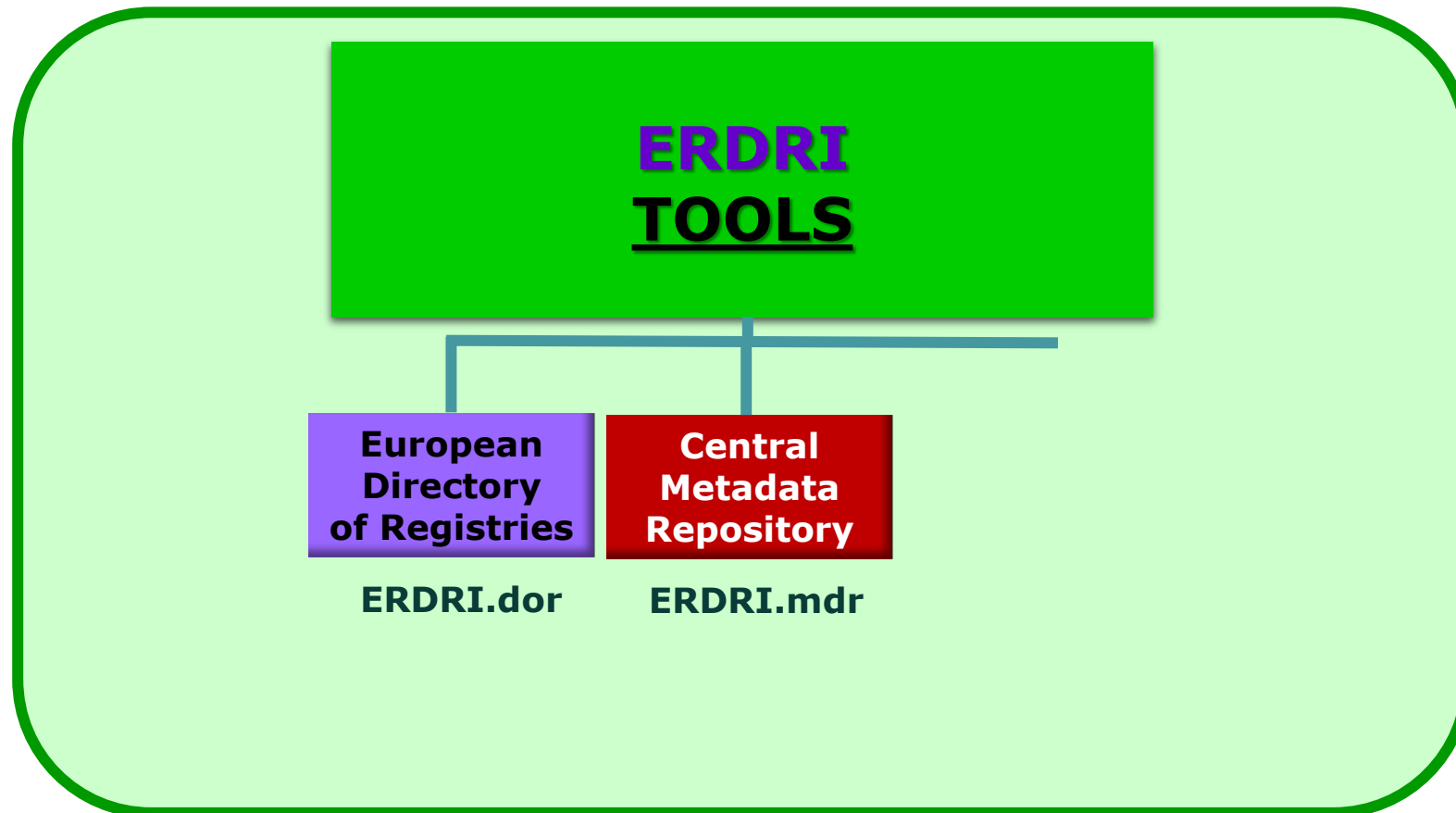
- List of participating RD registries
- Descriptive information
 - specific rare disease addressed
 - scope
 - operating institution
 - contact information, etc.)
 -
 - = characteristics of registries (27)

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



European Platform on Rare Diseases Registration

INTEROPERABILITY for RD data collection and exchange



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

Consequence: use of common definitions for data elements

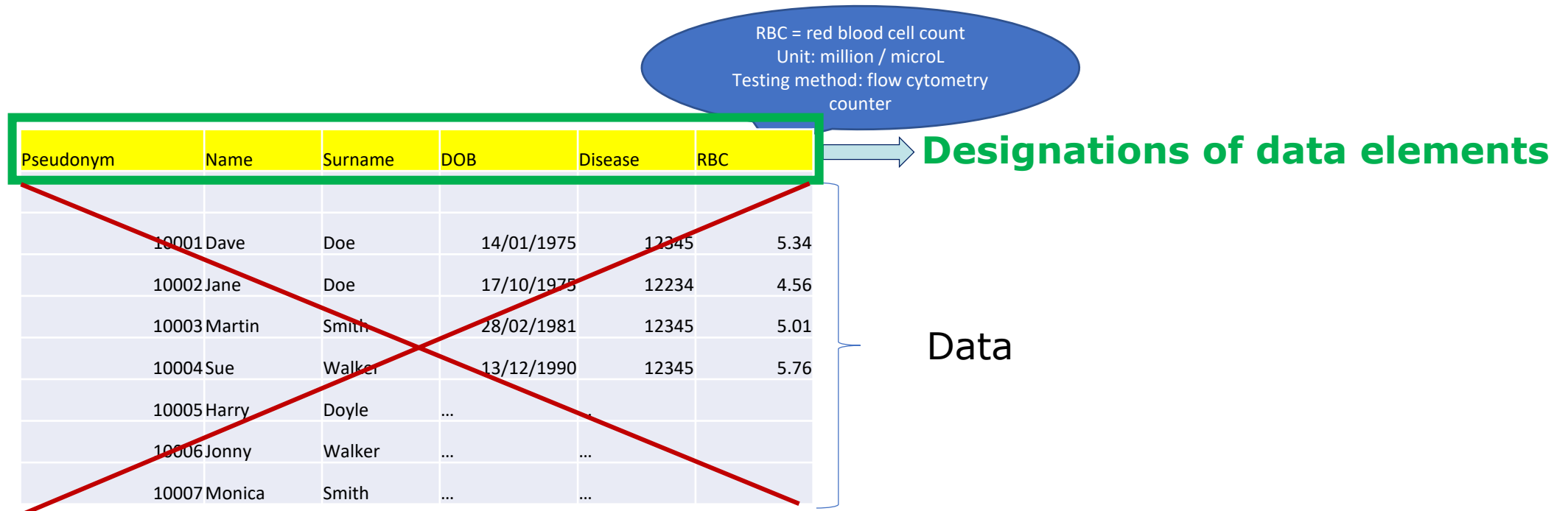


terms are understood in the same way by all data providers + data users

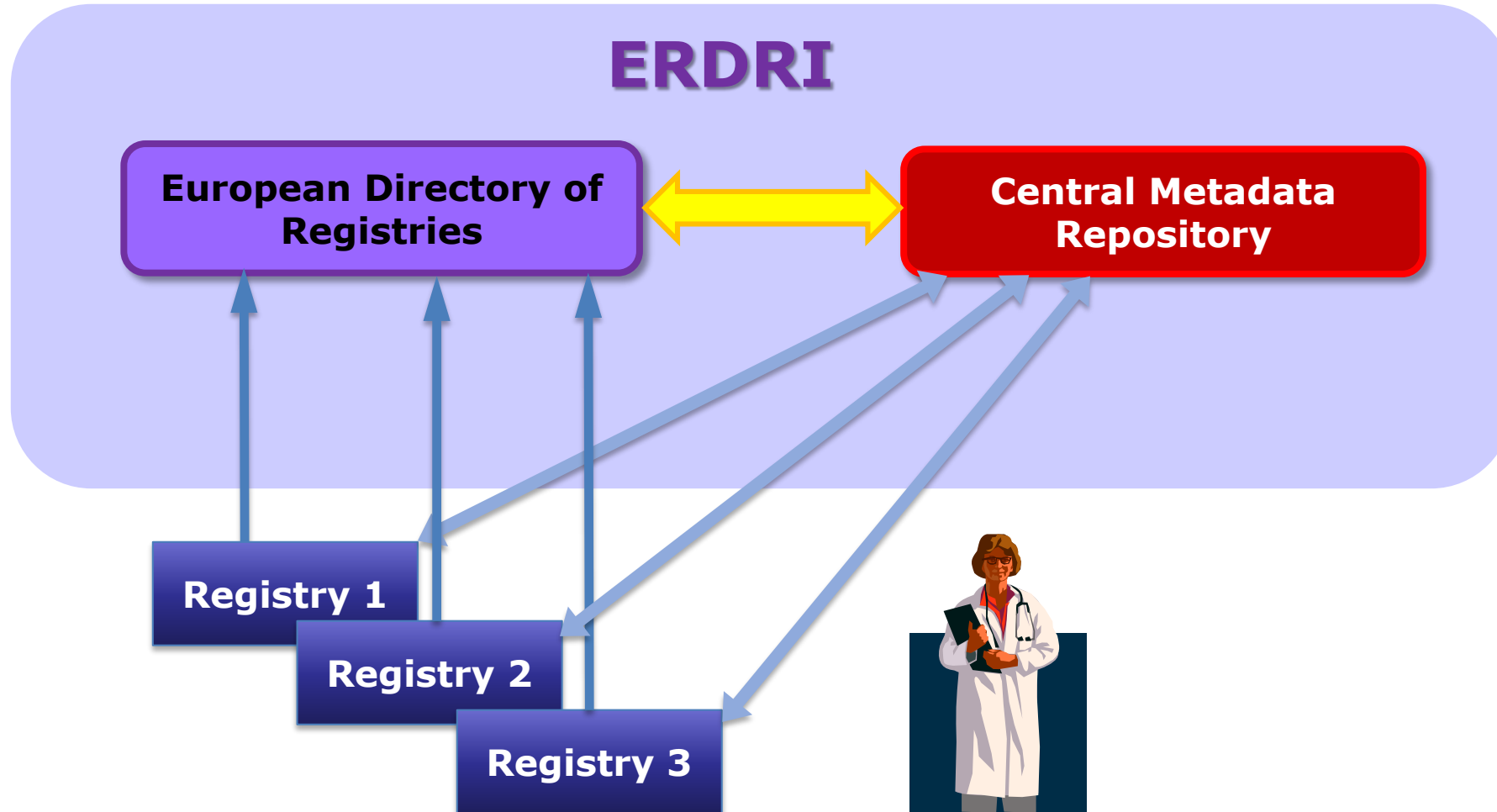
ERDRI.mdr = **thesaurus** of terms used by participating registries
common format

European Platform on Rare Diseases Registration

ERDRI.mdr – The Central Metadata Repository

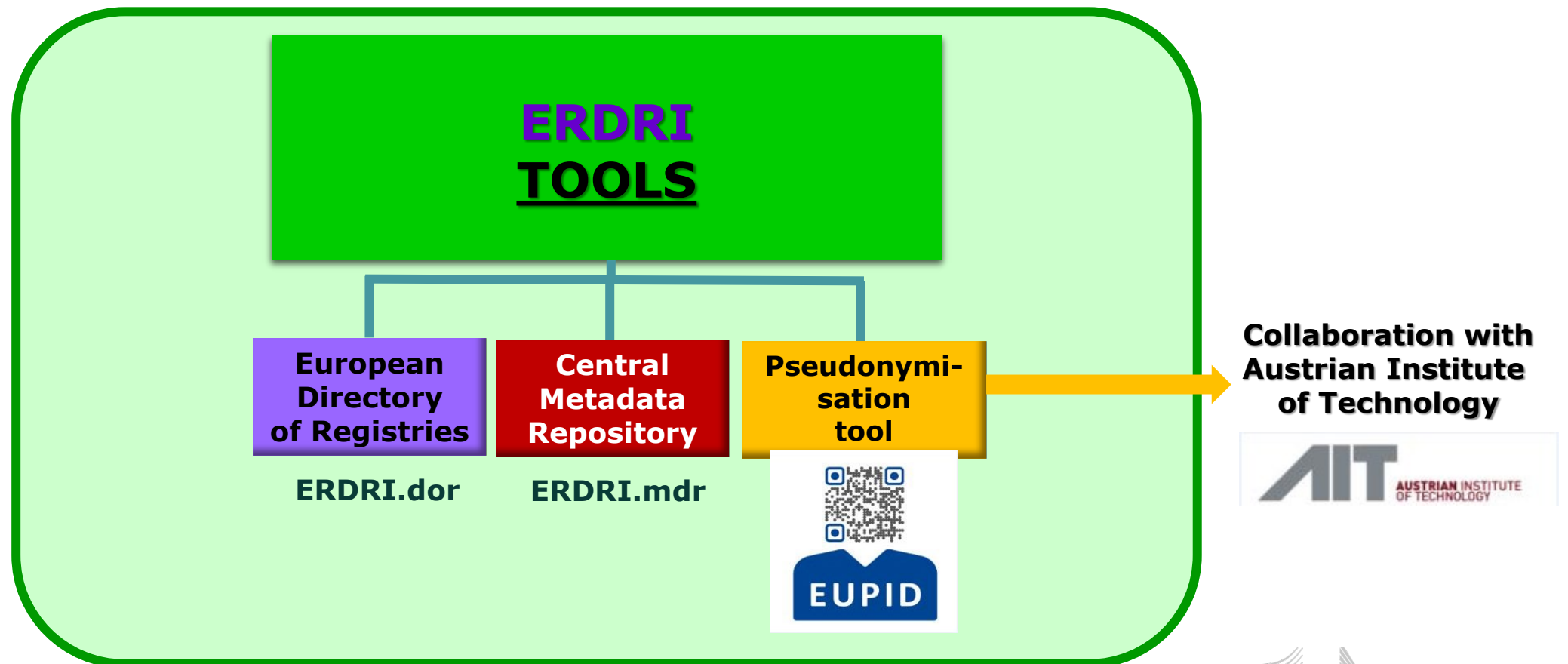


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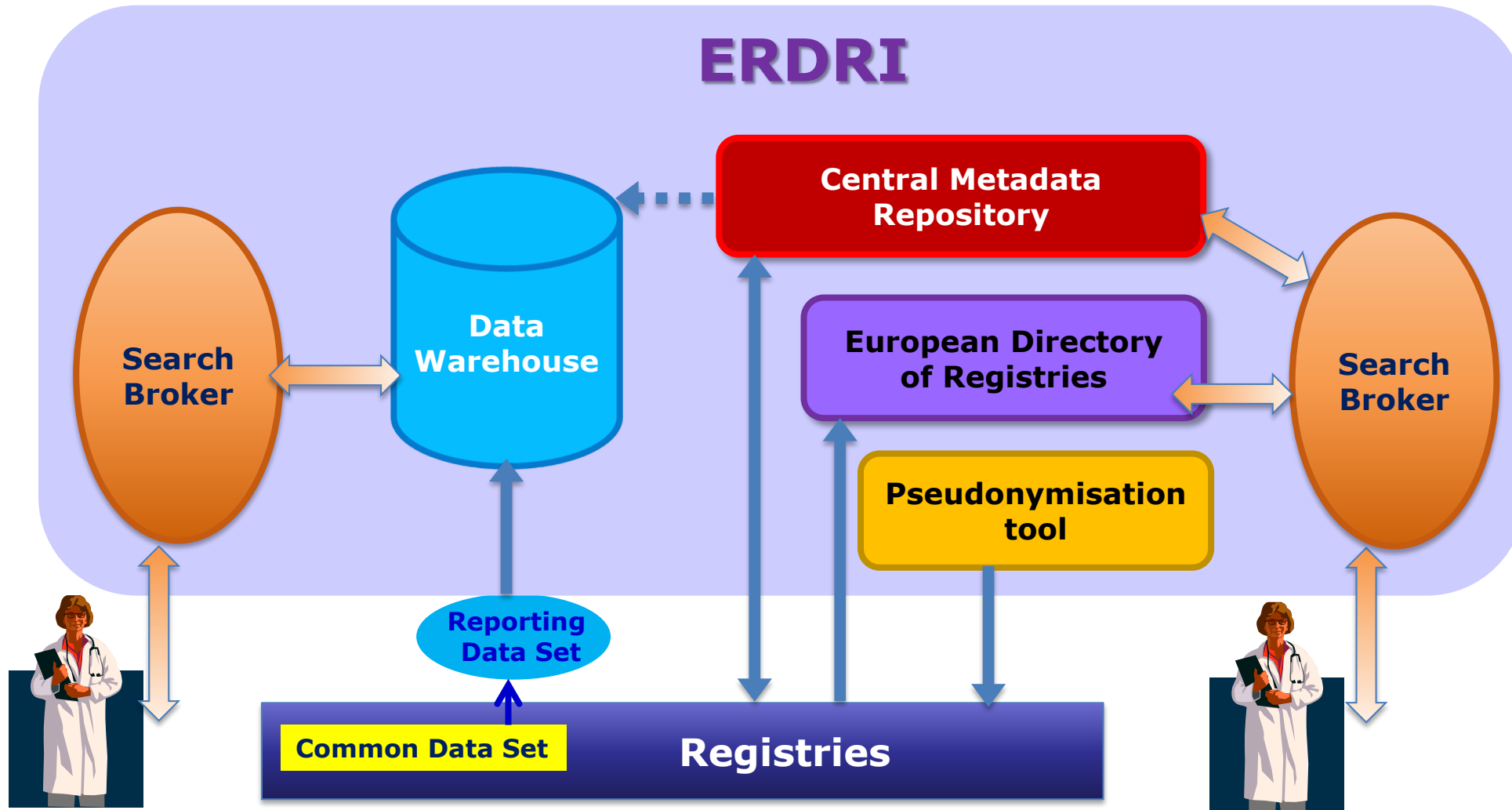


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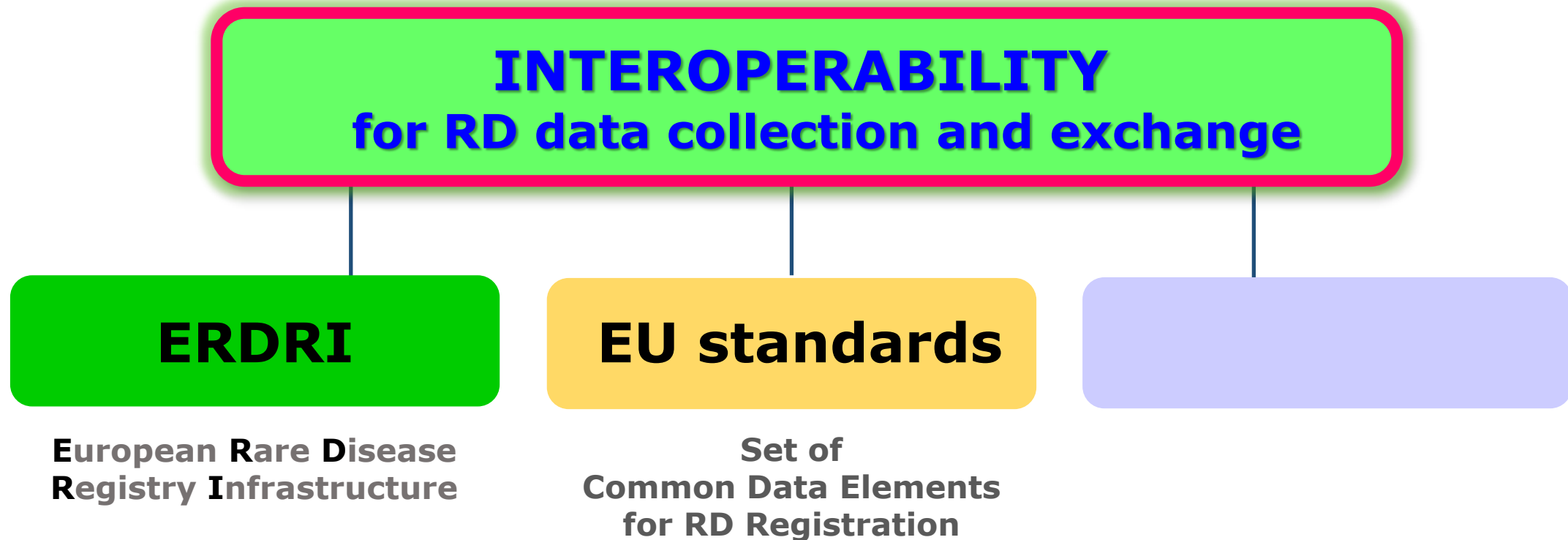
INTEROPERABILITY for RD data collection and exchange



European Platform on Rare Diseases Registration



European Platform on Rare Diseases Registration



European Platform on Rare Diseases Registration

INTEROPERABILITY for RD data collection and exchange

ERDRI

**European Rare Disease
Registry Infrastructure**

EU standards

**Set of
Common Data Elements
for RD Registration**

Training

- ❑ JRC ERDRI Training Workshop - MDR, DoR
 - session 1 – February 2018
 - session 2 – March 2018
- ❑ JRC ERDRI Training Workshop – pseudonymisation
 - March 2018

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)

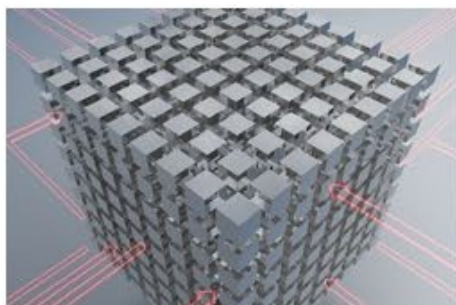


European standards for data collection and data sharing



Trainings
Events
Latest news

Data repository



European RD Registry Data Warehouse

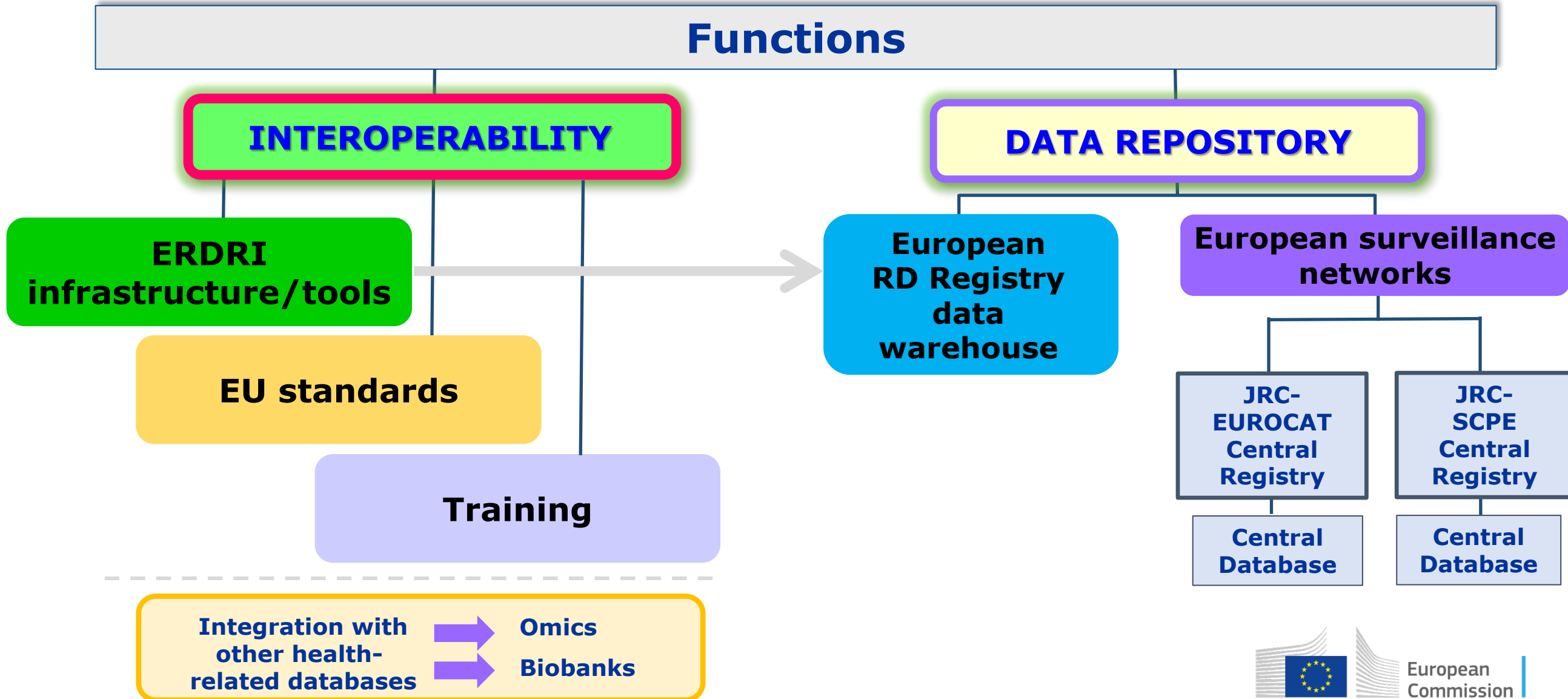


Surveillance of Congenital Anomalies in Europe



Surveillance of Cerebral Palsy in Europe

European Platform on Rare Diseases Registration



European Platform on Rare Diseases Registration

JRC F.1 'Health in Society'

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