Registries in Europe - European Platform on Rare Disease Registration (EU RD Platform)

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European Commission's Strategy for Rare Diseases

Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on **"Rare** Diseases: **Europe's challenges"** (2008)

1. To improve recognition and visibility on rare diseases

2. To support policies on rare diseases in the EU Member States

3. To develop European cooperation, coordination and regulation for rare diseases

European Commission

Why an EU Platform on RD Registration?

➢ To <u>cope with the extreme fragmentation</u> of data sources across EU Member States

Hundreds of RD registries exist, but

- no standardised data collection for most RDs

- the lack of interoperability severely limits the registries' potential

Source: EURORDIS

European Commission

Why an EU Platform on RD Registration?

➢ To reach <u>interoperability between registries</u>

- ▪ Standardised data collection and exchange

- ▪ Semantic interoperability

- ▪ Make data FAIR

  - Findable
  - Accessible
  - Interoperable
  - Reusable

- ▪ Data linkage

- ▪ Data transfer

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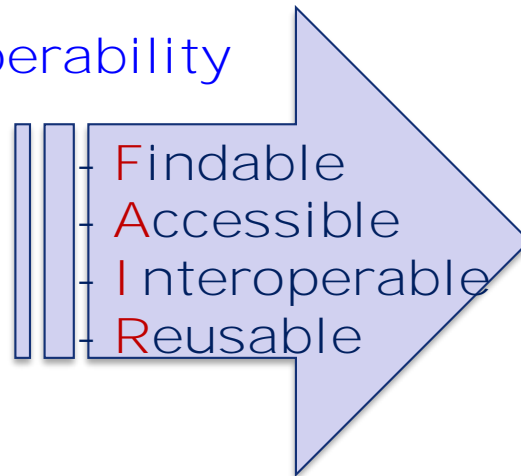
Why an EU Platform on RD Registration?

➢ To reach the <u>critical number</u> of patients for

- Studies

- Research

➢ Use of data
  for all possible purposes, advancing knowledge on RD

- clinical aspects   - evaluation of prognosis
- natural history   - epidemiology
- therapy   - basic research
- quality of care   - policy - health policy
                    etc.

European Platform on Rare Disease Registration (EU RD Platform)

Searchable, findable rare disease registry data

European Rare Disease Registry Infrastructure (ERDRI)
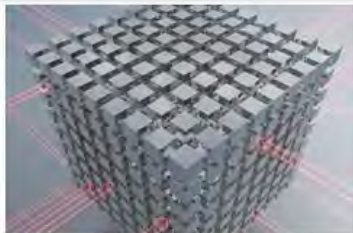
European standards for data collection and data sharing

Trainings, Resources and Latest news

Data repository

European RD Registry Data Warehouse

Surveillance of Congenital Anomalies in Europe

Surveillance of Cerebral Palsy in Europe

https://eu-rd-platform.jrc.ec.europa.eu

ERDRI
European Rare Disease Registry Infrastructure

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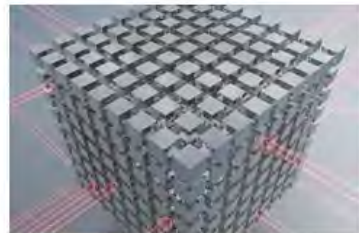
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European Platform on Rare Disease Registration (EU RD Platform)

- Unified source of information about RD registries and their data
- The Platform for RD REGISTRIES, bringing them together

- Overview of RD registries

European Directory of Registries

European Directory of Registries ERDRI.dor

European Rare Disease Registry Infrastructure (ERDRI)

European Directory of Registries (ERDRI.dor)

Overview of rare disease registries in Europe including their characteristics

RD registries/ERNs:

- overview of registries with characteristics
- searchable catalogue
- contact details

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General information

Name of the registry *

Acronym

Medical area *

Type of Registry *
- [ ] Epidemiology
- [ ] Clinical Registry
- [ ] Basic Research
- [ ] Pharmacological Research
- [ ] Patient Registry
- [ ] Healthcare planning
- [ ] Economic evaluation
- [ ] Other type

If other type of registry, please explain

Registry is member of
- [ ] Eurocat

Registry is member of a European Reference Network

Please input your ERN here ▾

Description *

Short description of the registry's scope

(500 characters remaining)

Website

Sponsors

Rare disease *

Search	
Input by orphacode	Enter a list of orphacodes in the textbox below. Separate codes by either a new line, blanks, commas or semicolons and then press 'Add' to process the list.

+ Add

Name of the disease

Structure

Inclusion and exclusion criteria

(1,000 characters remaining)

Recruitment area *

Name of the recruitment area *
(Country, state or region)

Recruitment start/end date
DD/MM/YYYY

to

Current number of cases *

at

Data source *

- [ ] University hospital
- [ ] Non university hospital
- [ ] Physician
- [ ] Other
- [ ] Patient/test person
- [ ] Family of patient
- [ ] Research Institution

If other data source, please explain

Number of Data Elements collected by the registry *

Technical solution (software) used *
eg. Excel, OSSE, Redcap, Oracle, Access etc.

Data management

Link to the privacy policy

Ethical review committee

Availability for future collaborations/studies

ean
nission

Registry information

Institution *

Facility

Department *

Street & number *

Postcode City *

Country *

Country (please specify)

Responsible for the registry

First name *

Last name *

Title

Academic title/degree

Position

E-Mail address *

Phone number *

Biobanks

Biobank name	Registry of biobanks	Biobank identifier	**+**

Additional components

Name of the component	Component described in	Relationship component-registry	**+**

European Platform on Rare Disease Registration (EU RD Platform)

- The Platform for RD METADATA
  (all data elements collected by RD registries)

- Basis for semantic interoperability between RD registries

Central Metadata Repository

Central Metadata Repository ERDRI .mdr

= Collection of metadata
on all data elements collected
by participating registries

Central Metadata Repository ERDRI.mdr

Designations of data elements

+ Definition
+ Measurement unit

Pseudonym	Name	Surname	DOB	Disease	RBC
10001	Dave	Doe	14/01/1975	12345	5.34
10002	Jane	Doe	17/10/1975	12234	4.56
10003	Martin	Trump	28/02/1981	12345	5.01
10004	Sue	Walker	13/12/1990	12345	5.76
10005	Harry	Doyle	...	...	
10006	Jonny	Walker	...	...	
10007	Bart	Simpson	...	...	

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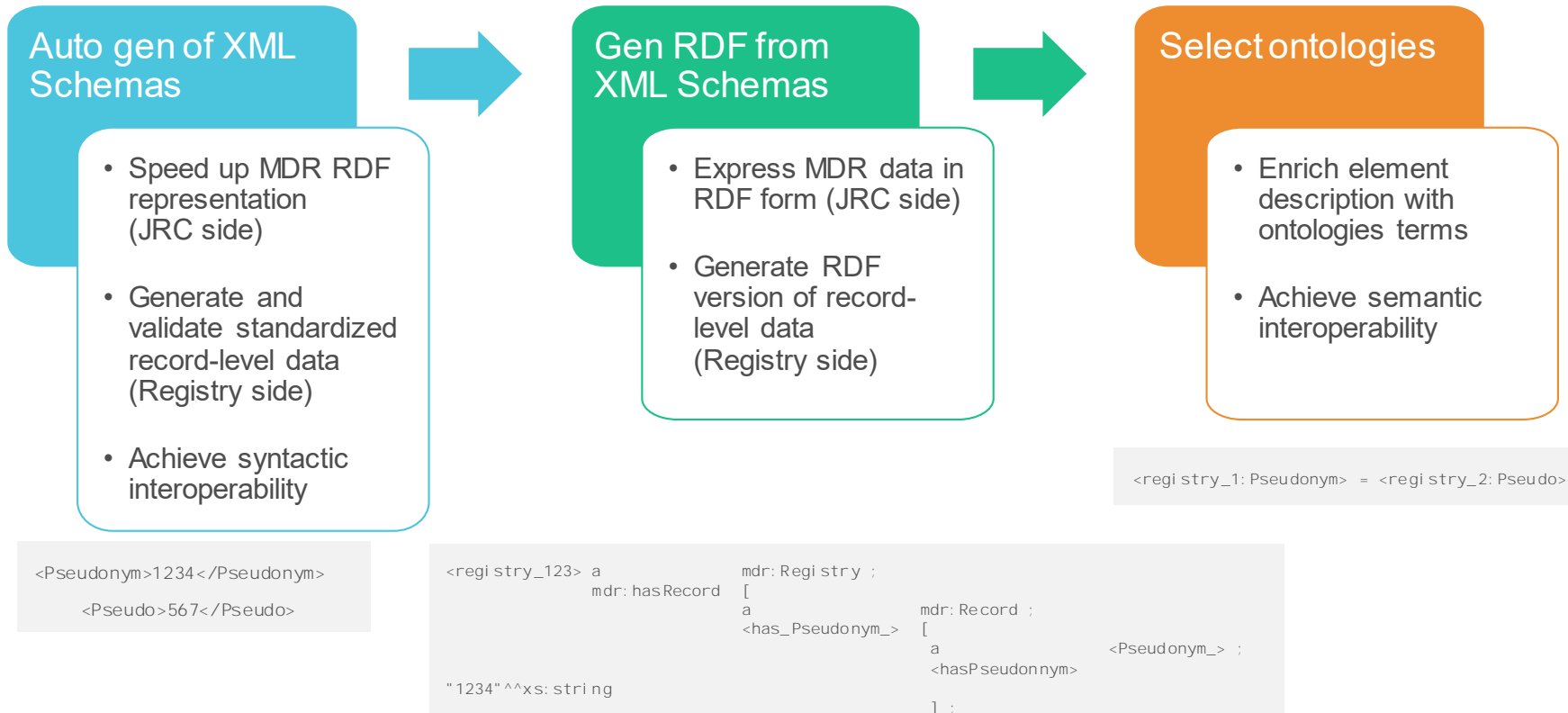
Central Metadata Repository ERDRI .mdr

- repository of
  metadata

- data dictionnaries
- ontologies
- semantic interoperability

FAI Rification of the MDR

Auto gen of XML Schemas

- Speed up MDR RDF representation (JRC side)

- Generate and validate standardized record-level data (Registry side)

- Achieve syntactic interoperability

Gen RDF from XML Schemas

- Express MDR data in RDF form (JRC side)

- Generate RDF version of record-level data (Registry side)

Select ontologies

- Enrich element description with ontologies terms

- Achieve semantic interoperability

```
<registry_1:Pseudonym> = <registry_2:Pseudo>
```

```
<Pseudonym>1234</Pseudonym>

    <Pseudo>567</Pseudo>
```

```
<registry_123> a              mdr:Registry ;
            mdr:hasRecord  [
                           a              mdr:Record ;
                           <has_Pseudonym_>  [
                            a                 <Pseudonym_> ;
                            <hasPseudonnym>
                           ] ;
"1234"^^xs:string
```

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European Platform on Rare Disease Registration (EU RD Platform)

- The Platform for EU-wide unified PSEUDONYMISATION system for RD patients

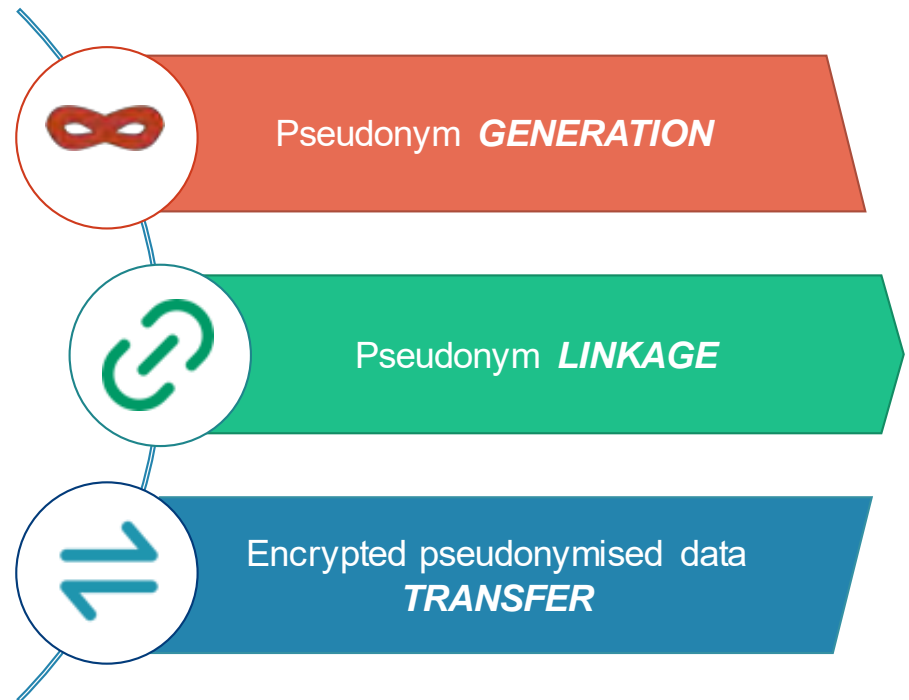## SPIDER pseudonymisation services

SPIDER pseudonymisation services

Pseudonym **GENERATION**

Pseudonym **LINKAGE**

Encrypted pseudonymised data **TRANSFER**

Spider

Share

Network

Data sharing **policy enforcement**

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European Platform on Rare Disease Registration (EU RD Platform)

- Ensures DISCOVERABILITY of DATA from participating RD registries

- Registries remain owners of their data and decide about use of the data

ERDRI Search tool

Search broker
ERDRI .sebro

Search broker
ERDRI .sebro

DOR

./erdridor

MDR

./mdr
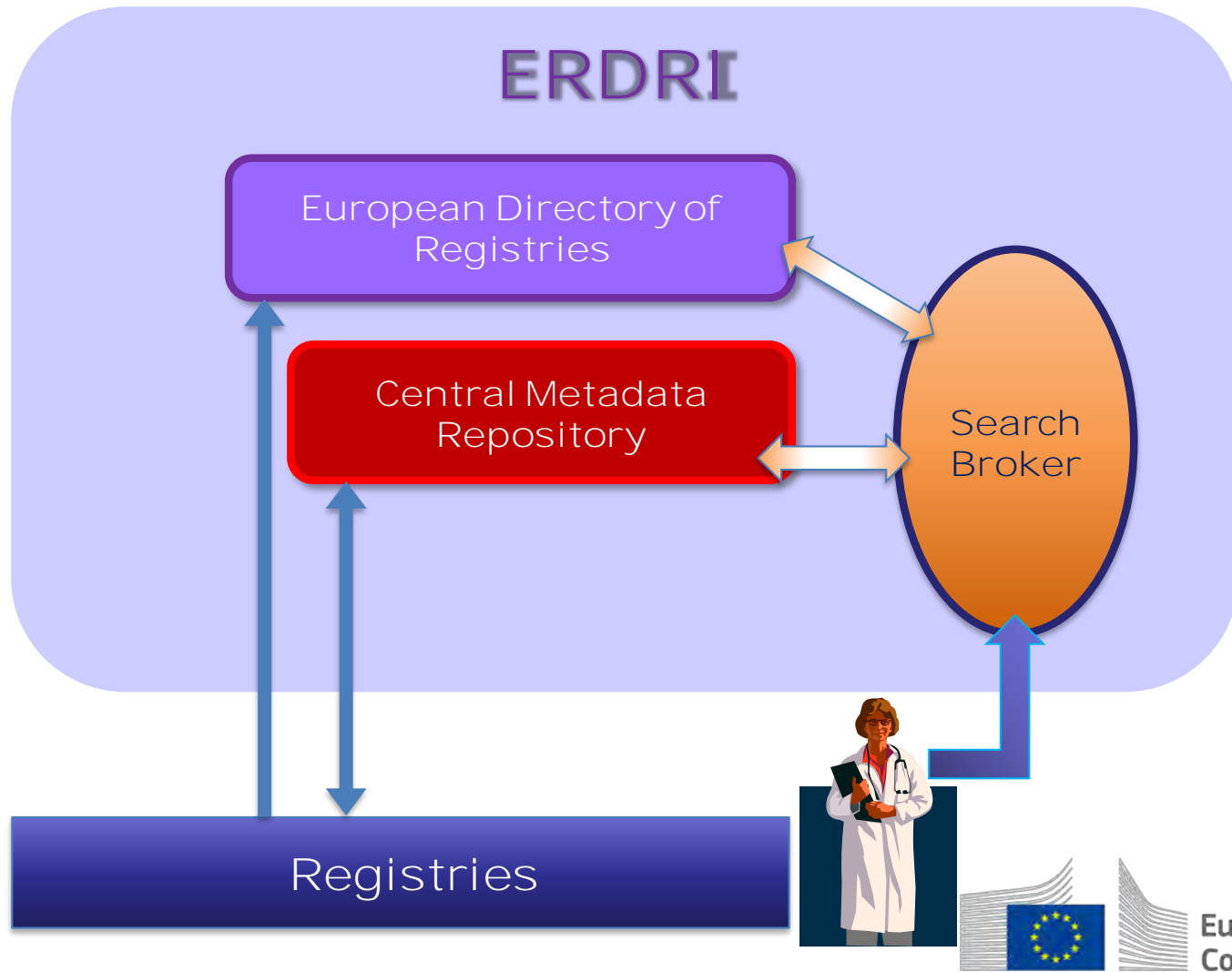
Search Broker

./sebro
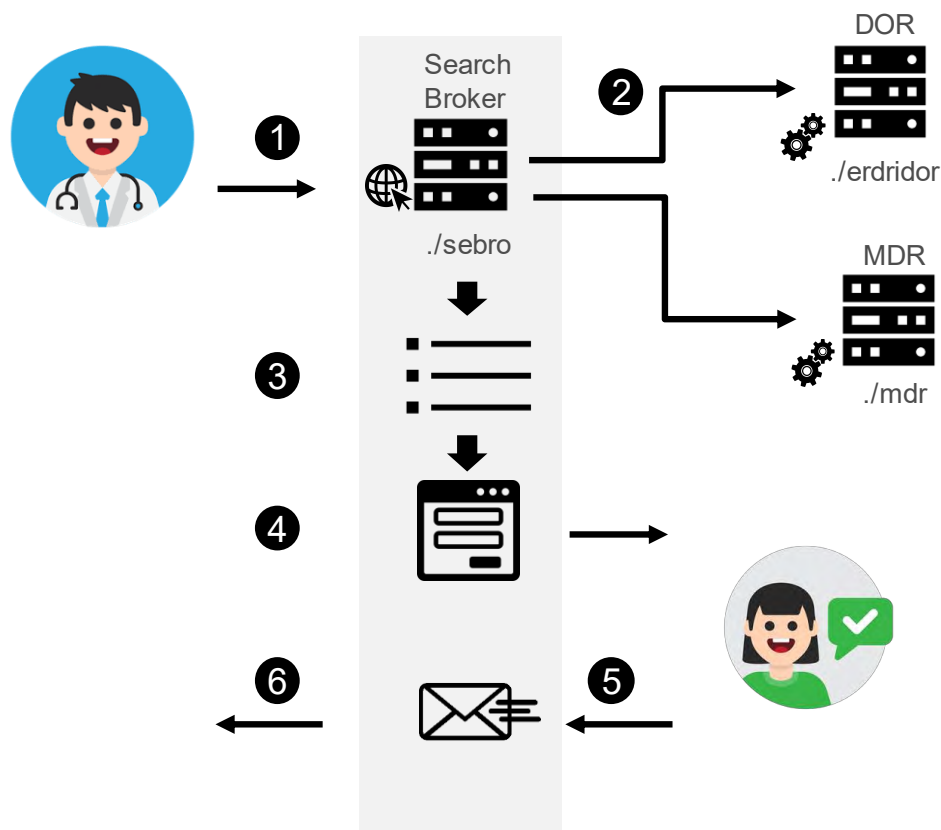
1. *Researcher inserts keywords*
   - *metadata elements name, rare disease, type of registry..*
2. *DOR and MDR are queried*
3. *Registries matching the request are identified*
4. *The EU RD Platform contacts the registries*
5. *Feedback from registries to the EU RD Platform*
6. *Researcher contacted by the EU RD Platform*

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ERDRI.sebro
central-level and record-level,
distributed ("federated")
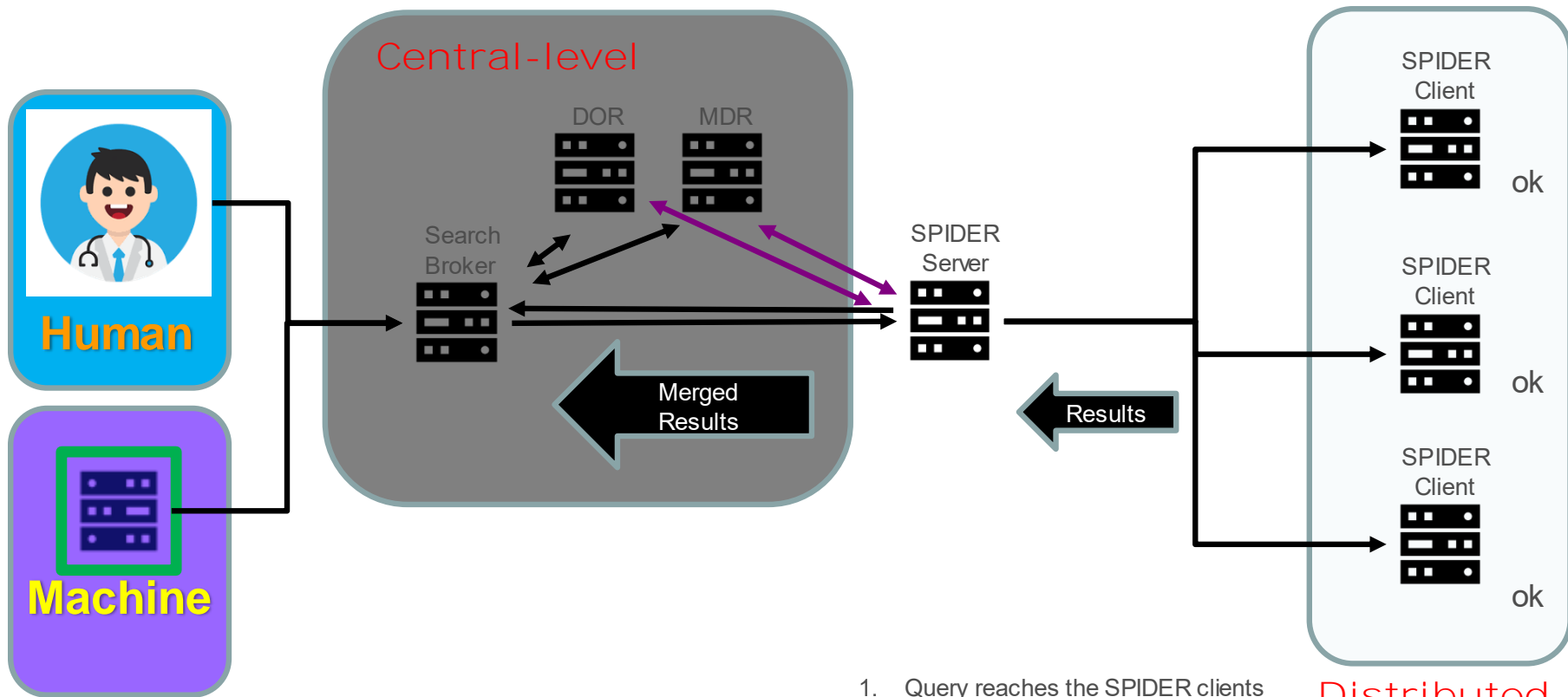
e.g. Count the number of female patients
where disease = Brugada syndrome and year of diagnosis = 2021

1. Query reaches the SPIDER clients
2. SPIDER clients compute the results
3. Results are merged and provided as a result

EU RD Platform - search function
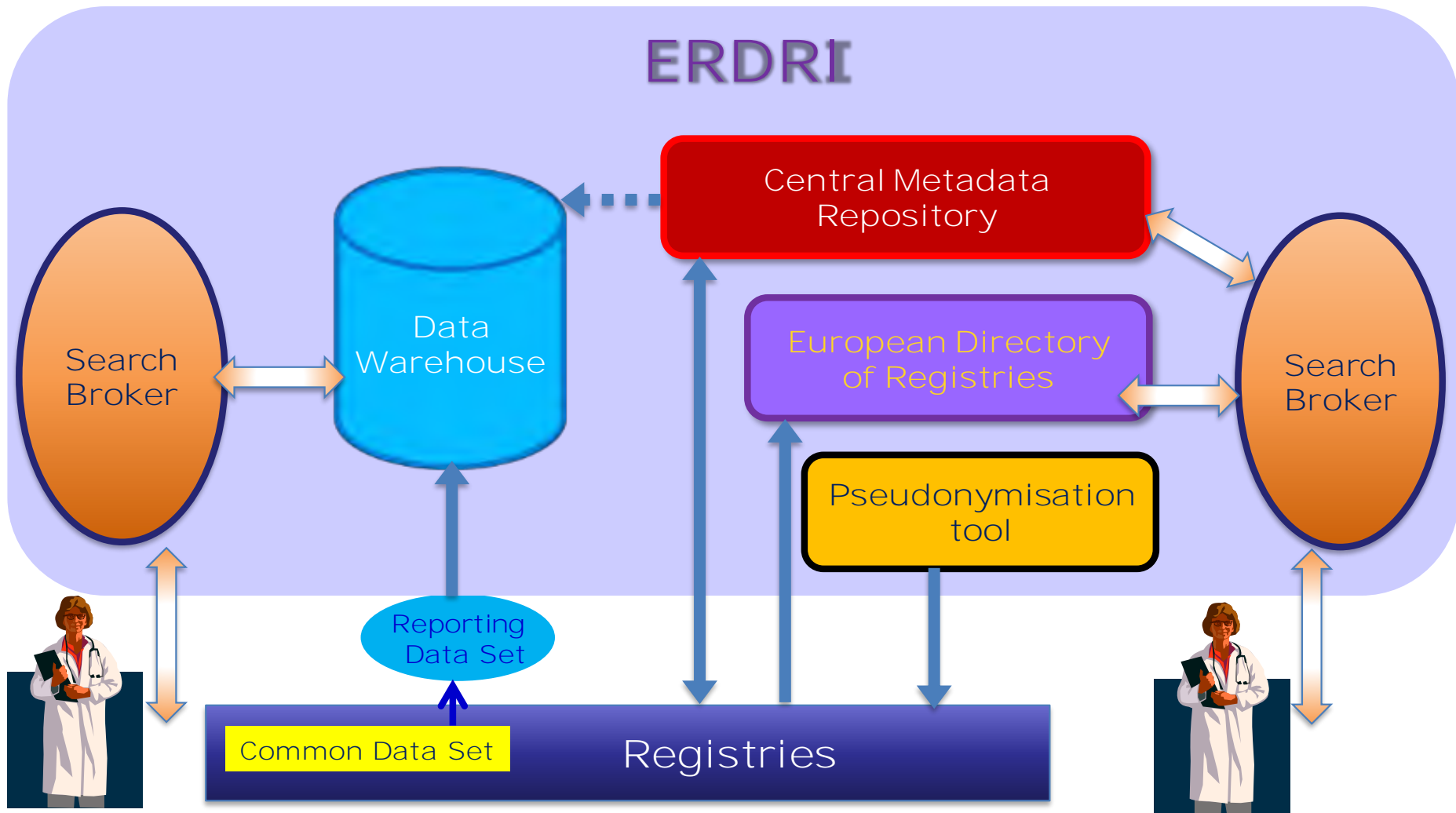
European Platform on Rare Disease Registration (EU RD Platform)

- Provides STANDARDS FOR DATA COLLECTION
  - Set of common data elements for RD registration

  In collaboration with ERNs/EJP-RD
  - Domain-specific common data elements for ERNs

SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION

GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
1. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	• String	https://eu-rd-platform.jrc.ec.europa.eu/erdri/eupid-intro
2. Personal information	2.1.	Date of birth	Patient's date of birth	• Date (dd/mm/yyyy)	
	2.2.	Sex	Patient's sex at birth	• Female • Male • Undetermined • Foetus (Unknown)	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	• Alive • Dead • Lost in follow-up • Opted-out	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	• Date (dd/mm/yyyy)	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	• Date (dd/mm/yyyy)	

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5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
	5.2.	Age at diagnosis	Age at which diagnosis was made	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
6 Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis retained by the specialised centre	Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code	http://www.orphadata.org/cgi-bin/inc/product1.inc.php
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code	http://www.hgvs.org
	6.3	Undiagnosed case	How the undiagnosed case is defined	• Phenotype (HPO) • Genotype (HGVS)	
7. Research	7.1.	Agreement to be contacted for research purposes	Patient's permission exists for being contacted for research purposes	• YES • NO	
	7.2.	Consent to the reuse of data	Patient's consent exists for his/her data to be reused for other research purposes	• YES • NO	
	7.3.	Biological sample	Patient's biological sample available for research	• YES • NO	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	• YES (if appropriate use link) • NO	https://directory.bbmri-eric.eu
8.Disability	8.1.	Classification of functioning/disability	Patient's disability profile according to International Classification of Functioning and Disability (ICF)	• Disability profile / Score	http://www.who.int/classifications/icf/whodasii/en/

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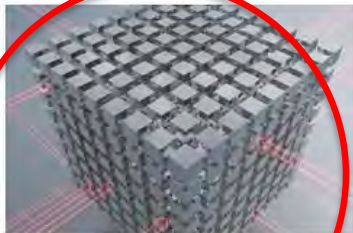**European Rare Disease Registry Infrastructure (ERDRI)**

European standards for data collection and data sharing

Trainings, Resources and Latest news

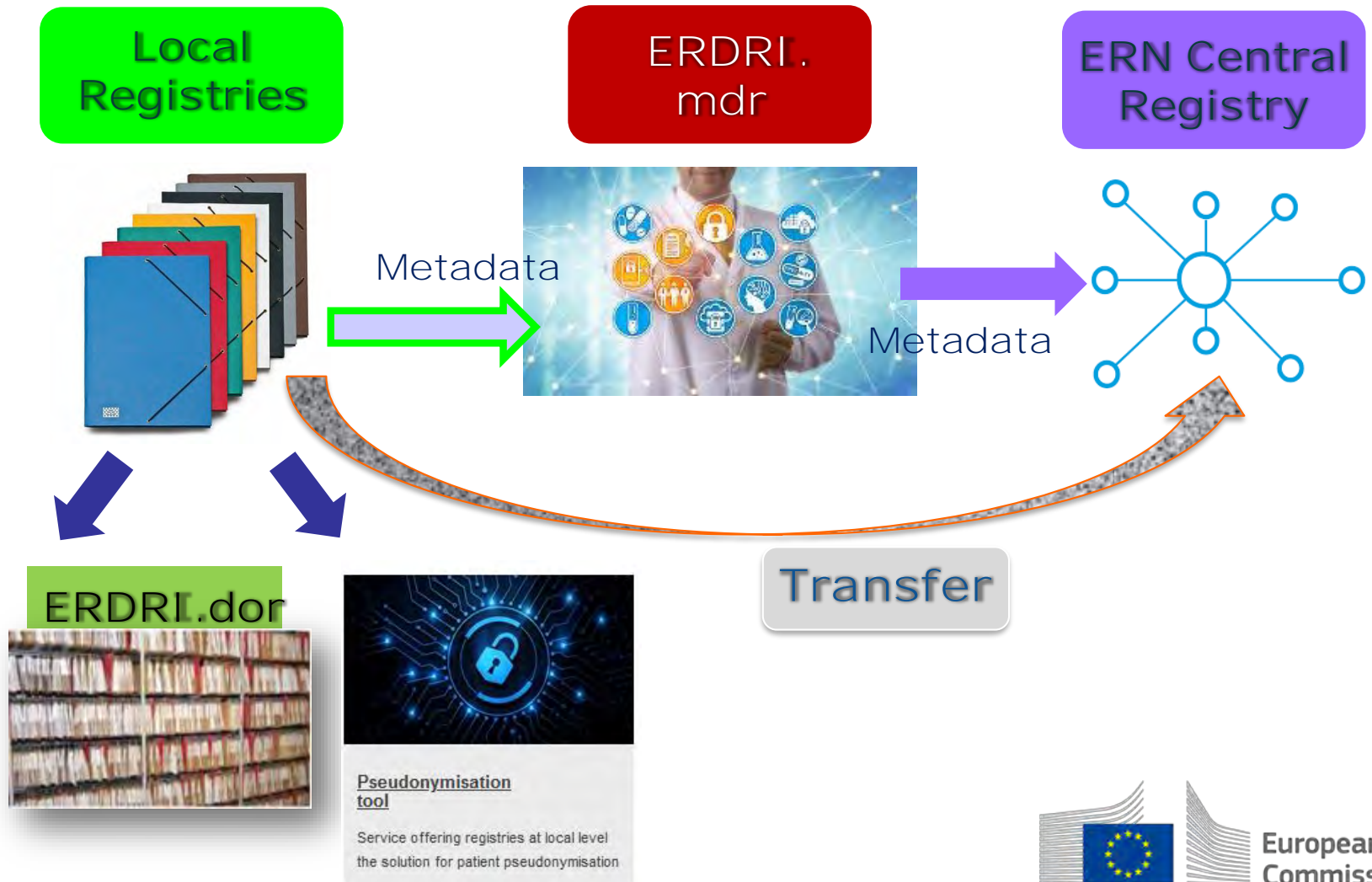## Data repository

European RD Registry Data Warehouse
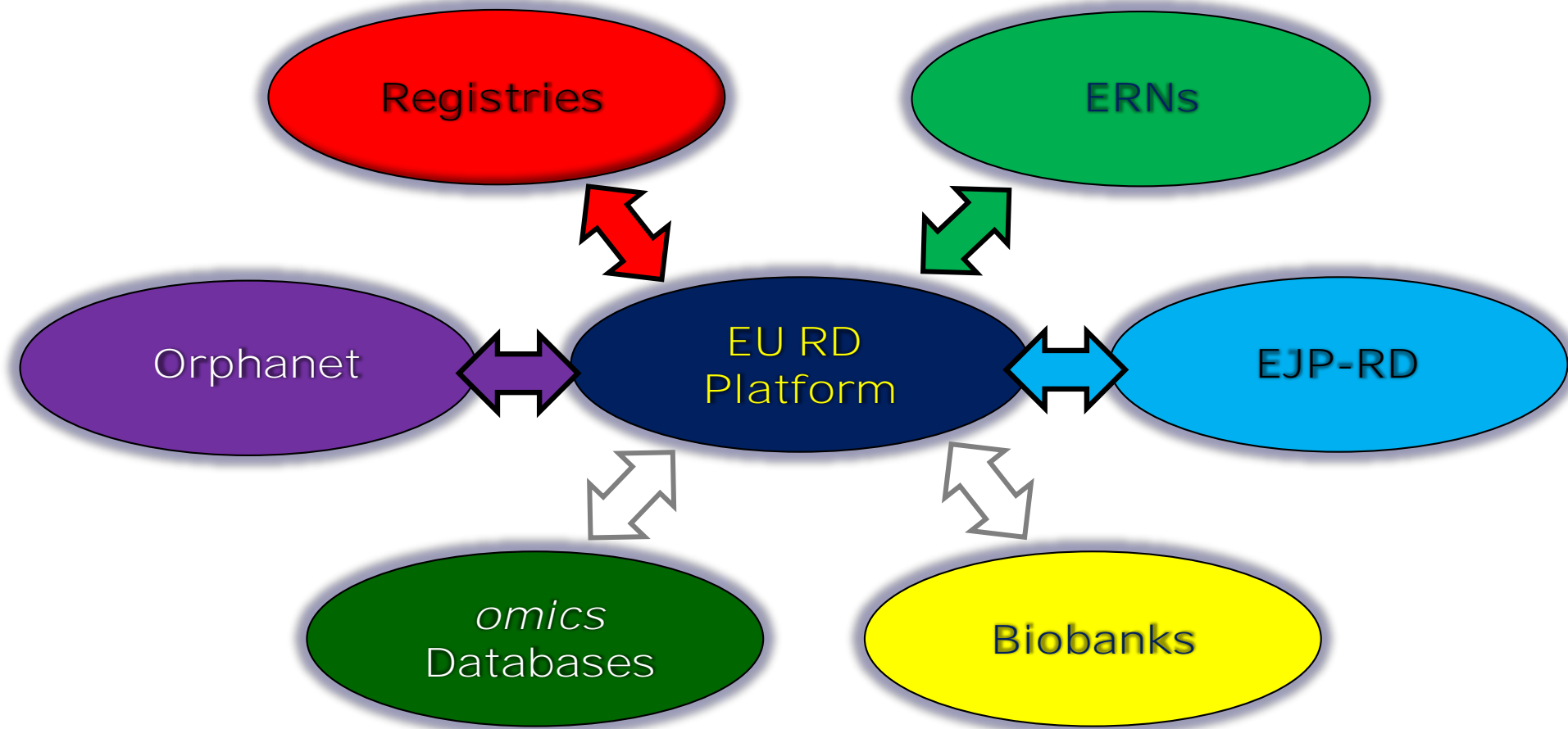
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Building the ERN central registry based on pre-existing registries

Join the EU RD Platform
knowledge generation centre for rare diseases

https://eu-rd-platform.jrc.ec.europa.eu