

The European Commission's science and knowledge service

Joint Research Centre



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andri.papadopoulou@ec.europa.eu

European Rare Disease Registry Infrastructure (ERDRI)

part of the EU RD Platform

Andri PAPADOPOULOU

Joint Research Centre - JRC F.1.

Haemophilia Registries Workshop

London, 08 June 2018

The JRC within the Commission



JRC's Mission

***“ As the science and knowledge service
of the Commission our mission is to support
EU policies with independent evidence
throughout the whole policy cycle ”***



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



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

European Commission's Strategy for Rare Diseases

Implementation report on the Commission Communication on Rare Diseases: Europe's challenges (2008) and Council Recommendation on an action in the field of rare diseases (2009) [COM(2014) 548 final]

"This is why the European Commission's JRC is currently developing a European Platform on Rare Diseases Registration. The main objectives for this platform are to **provide a central access point for information** on rare diseases patients' registries for all stakeholders, **support new and existing registries in view of their interoperability**, **provide IT tools** to maintain data collection and **host activities of the surveillance networks**."

Why an EU Platform on RD Registration?

- To cope with the extreme fragmentation of data sources across EU Member States
- Benefits:
Reach the critical number of patients for
 - studies (epidemiological, clinical, translational, pharmacological, etc.)
 - research
- Solution:
 - make registries interoperable



The JRC is developing the EU RD Platform in collaboration with DG SANTE - AA

European Platform on Rare Diseases Registration

INTEROPERABILITY WORKSHOPS

Compatibility of procedures for improving use of RD registry data, information exchange, data sharing

Immediate needs – priority areas:

- ❖ Common data set
- ❖ Semantic interoperability

European Platform on Rare Diseases Registration

Need no.1 Set of Common Data Elements for RD Registration

Working Group

JRC – EU RD Platform

- EUCERD Joint Action
- EPIRARE
- RD-CONNECT

Discussed with

- ORPHANET
- BBMRI

16 elements
(14+2 for ID)



European Platform on Rare Diseases Registration

EUROPEAN PLATFORM ON RARE DISEASES REGISTRATION (EU RD Platform)

SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION

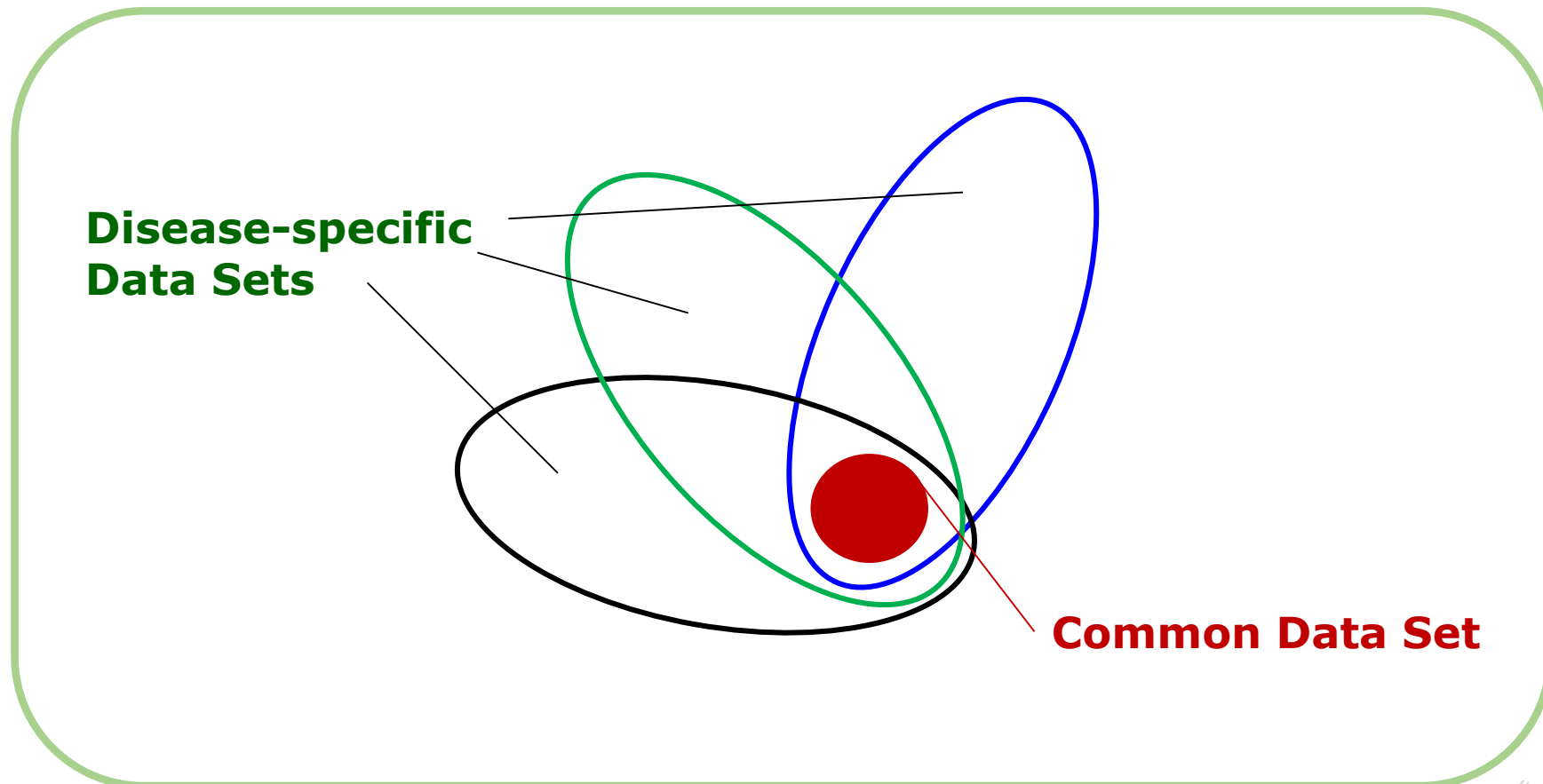
GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
1. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	<ul style="list-style-type: none">String	The JRC is working on providing a pseudonymisation tool to the registries
2. Personal information	2.1.	Date of birth	Patient's date of birth	<ul style="list-style-type: none">Date (dd/mm/yyyy)	
	2.2.	Sex	Patient's sex at birth	<ul style="list-style-type: none">FemaleMaleUndeterminedFoetus (Unknown)	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	<ul style="list-style-type: none">AliveDeadLost in follow-upOpted-out	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	<ul style="list-style-type: none">Date (dd/mm/yyyy)	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	<ul style="list-style-type: none">Date (dd/mm/yyyy)	

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GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	<ul style="list-style-type: none"> Antenatal At birth Date (dd/mm/yyyy) Undetermined 	
	5.2.	Age at diagnosis	Age at which diagnosis was made	<ul style="list-style-type: none"> 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	<ul style="list-style-type: none"> 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	<ul style="list-style-type: none"> 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	<ul style="list-style-type: none"> YES NO 	
	7.3.	Biological sample	Patient's biological sample available for research	<ul style="list-style-type: none"> YES NO 	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	<ul style="list-style-type: none"> 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)	<ul style="list-style-type: none"> Disability profile / Score 	http://www.who.int/classifications/icf/whodasii/en/

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Data sets



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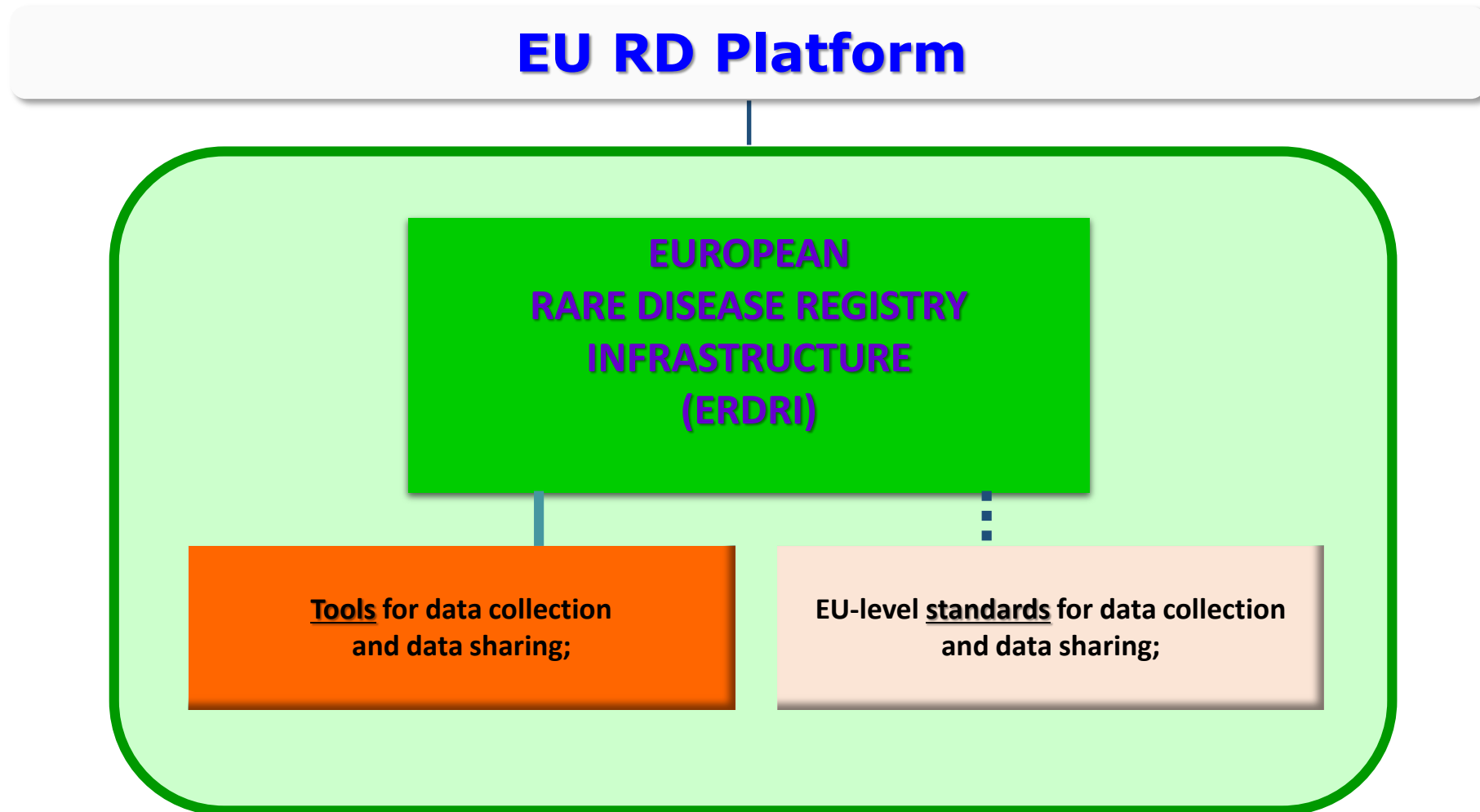
EU RD Platform

Collaboration with EUCERD-JA – WP "Registries"
University of Frankfurt

INTEROPERABILITY

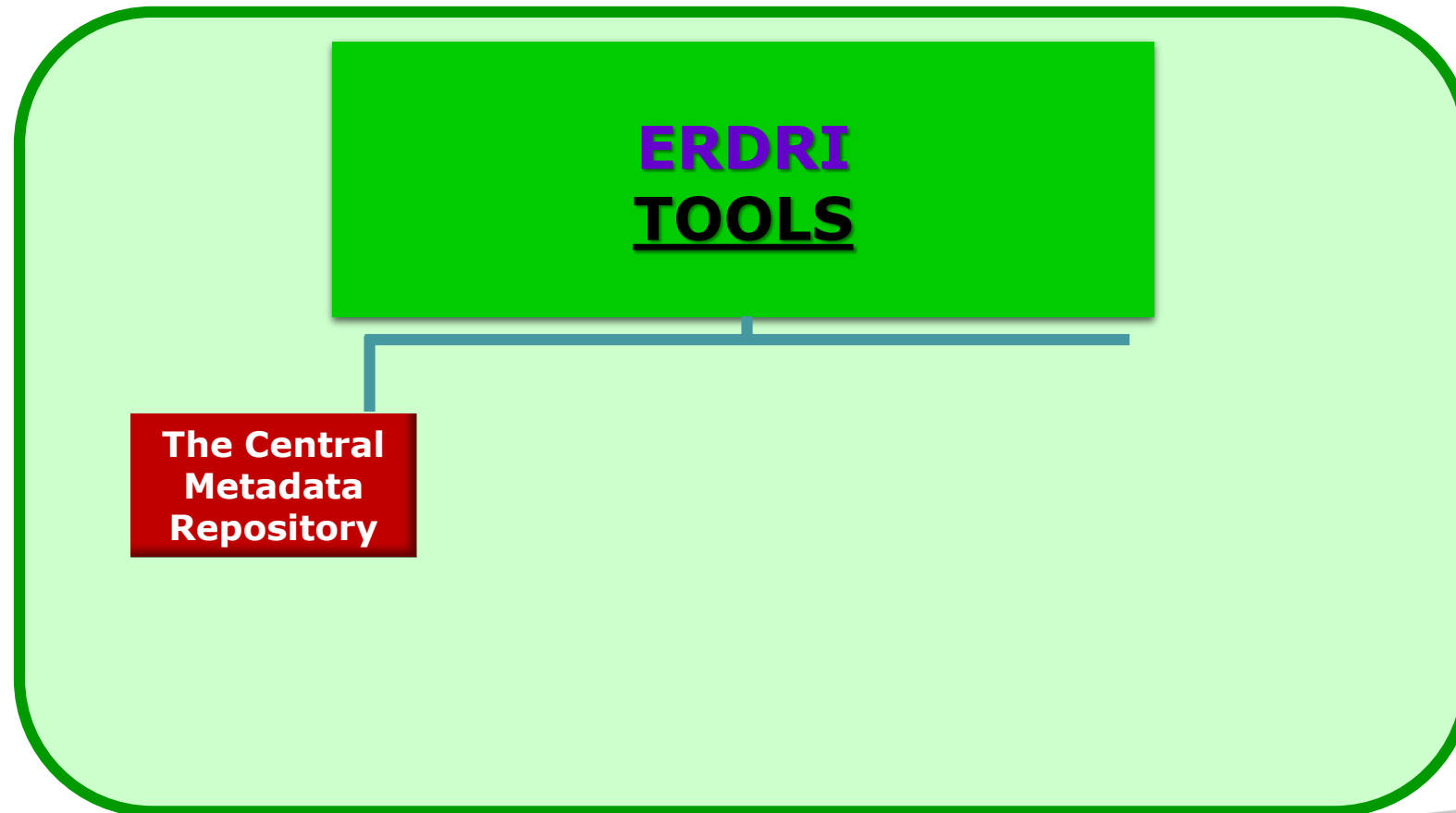
**EUROPEAN
RARE DISEASE REGISTRY
INFRASTRUCTURE
(ERDRI)**

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Need no.2 Semantic interoperability



European RD Registry Infrastructure (ERDRI)

ERDRImdr – The Central Metadata Repository

Semantic interoperability

Central Metadata Repository

ERDRI.mdr

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collection of metadata on all data elements
collected by participating registries

- Designation
- Definition
- Measurement unit + range

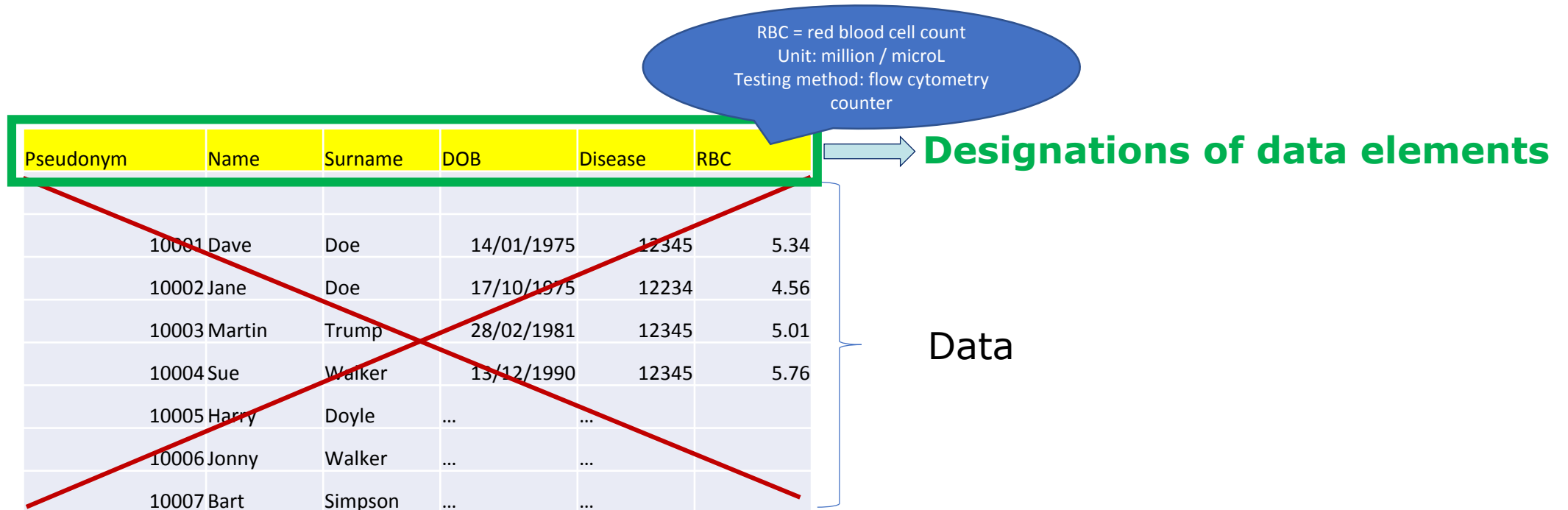
Consequence: use of common definitions for data elements

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

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

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ERDRImdr – The Central Metadata Repository



European RD Registry Infrastructure (ERDRI)

ERDRImdr – The Central Metadata Repository

Semantic interoperability

Central Metadata Repository

ERDRI.mdr

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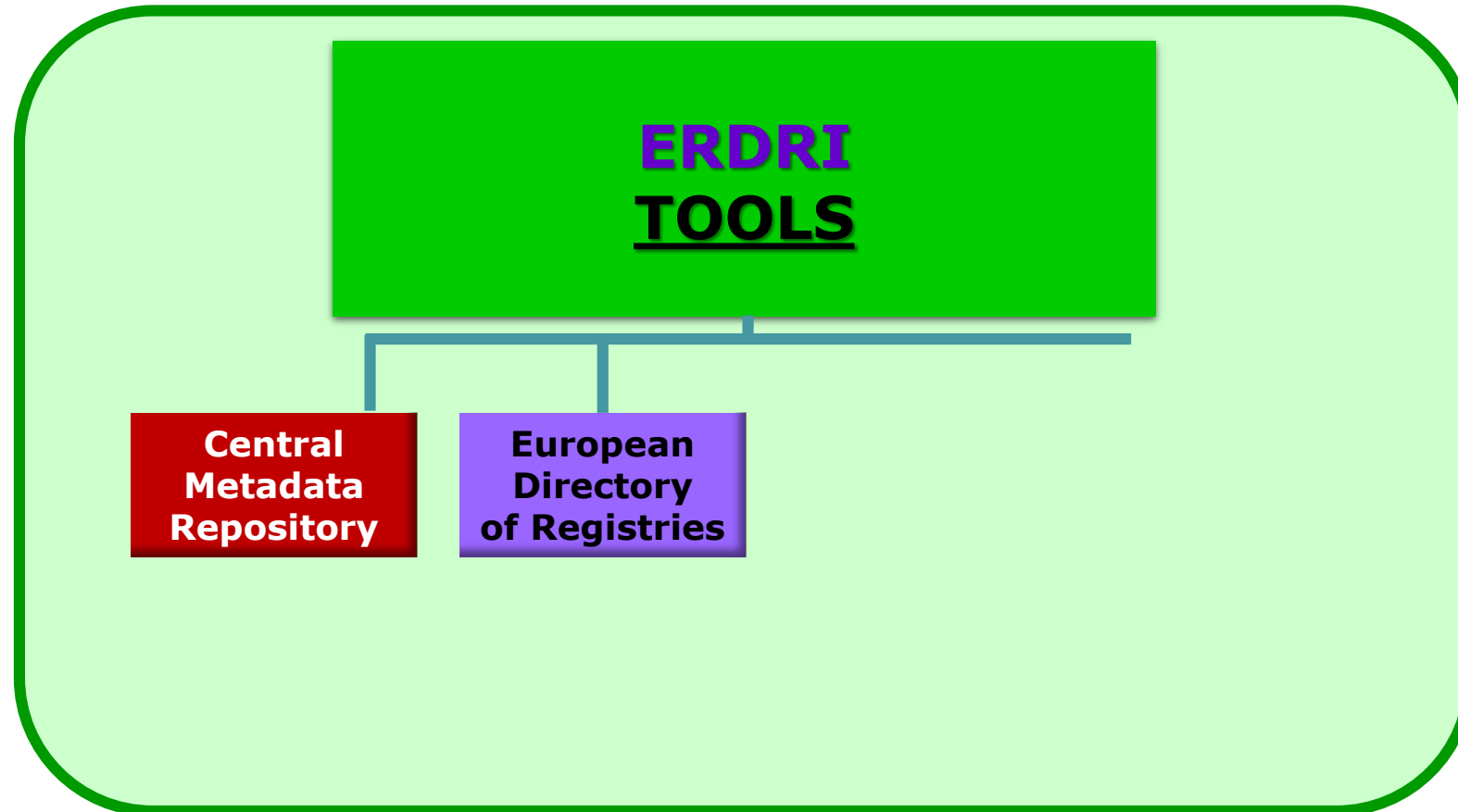
collection of metadata on all data elements collected by participating registries

- Designation
- Definition
- Measurement unit + range



Method (software) for importing registries' metadata into ERDRI.mdr

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ERDRIdor – The European Directory of RD registries University of Mainz

➤ List of participating RD registries

➤ Descriptive information

- (- specific rare disease addressed
- scope
- operating institution
- contact information, etc.)

➤ List of the data elements collected by the registries according to the MDR:
registry-specific data scheme

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



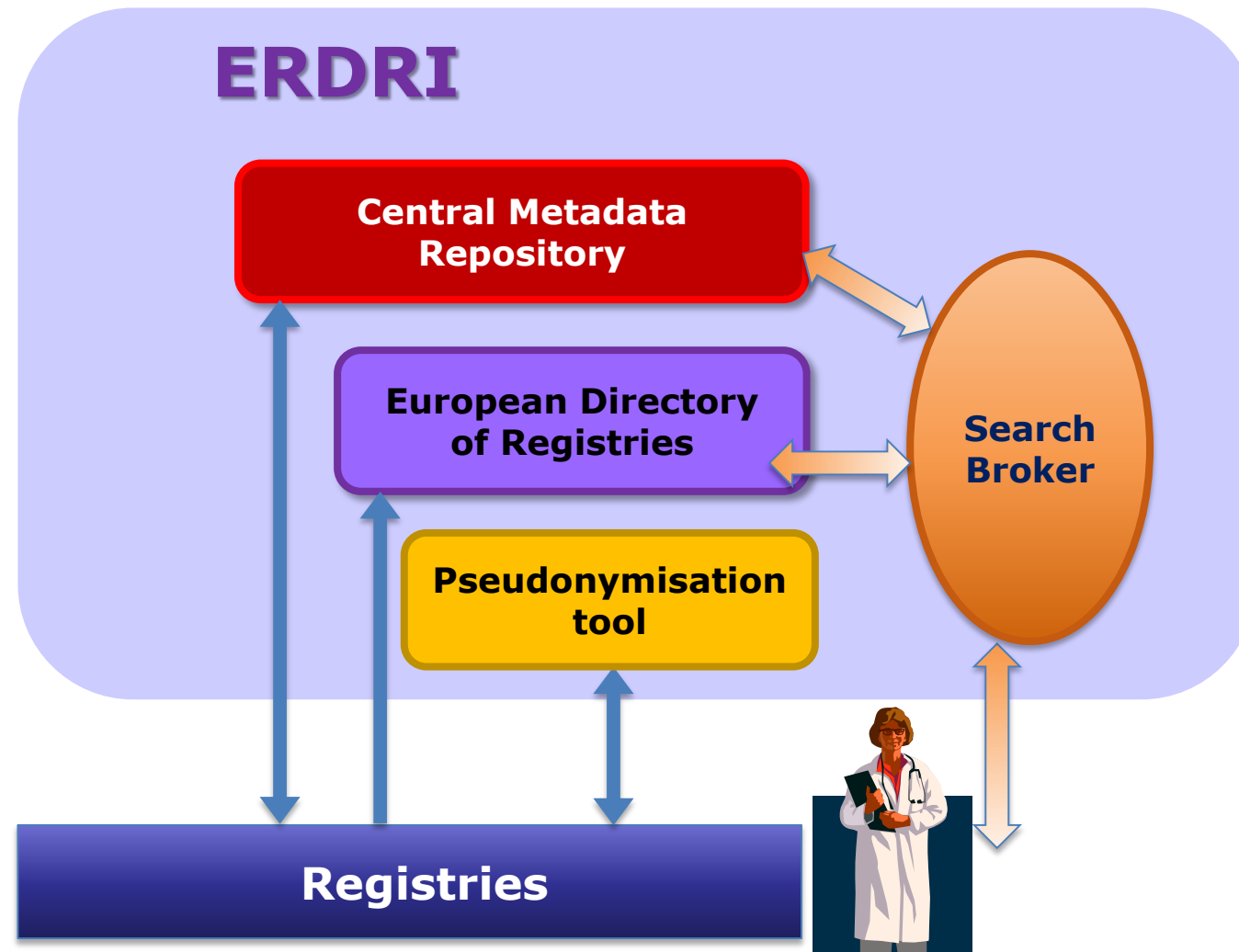
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ERDRI - functions

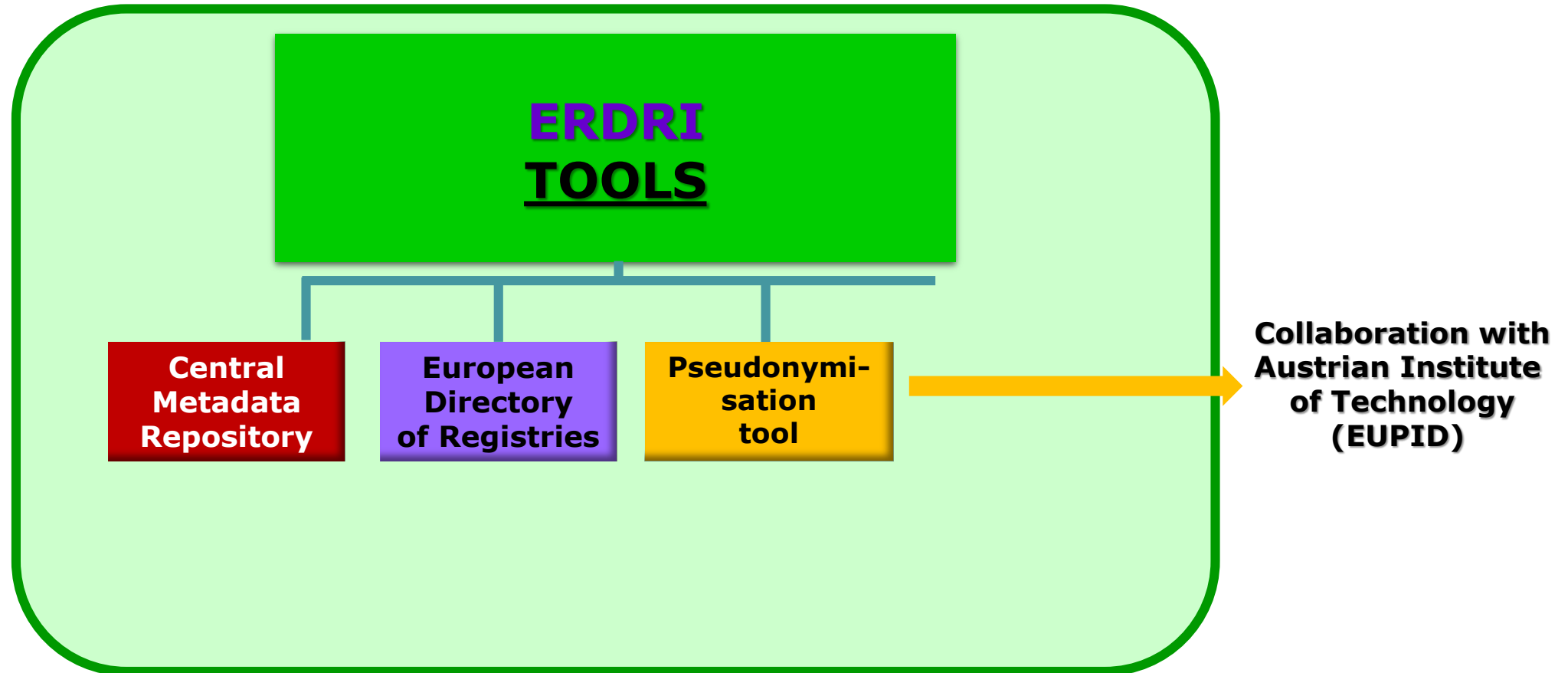
➤ **PROVIDES** the infrastructure and tools

❖ To make registries' data **SEARCHABLE, FINDABLE, interoperable, reusable**
(if accessible – it depends on the data owner!)

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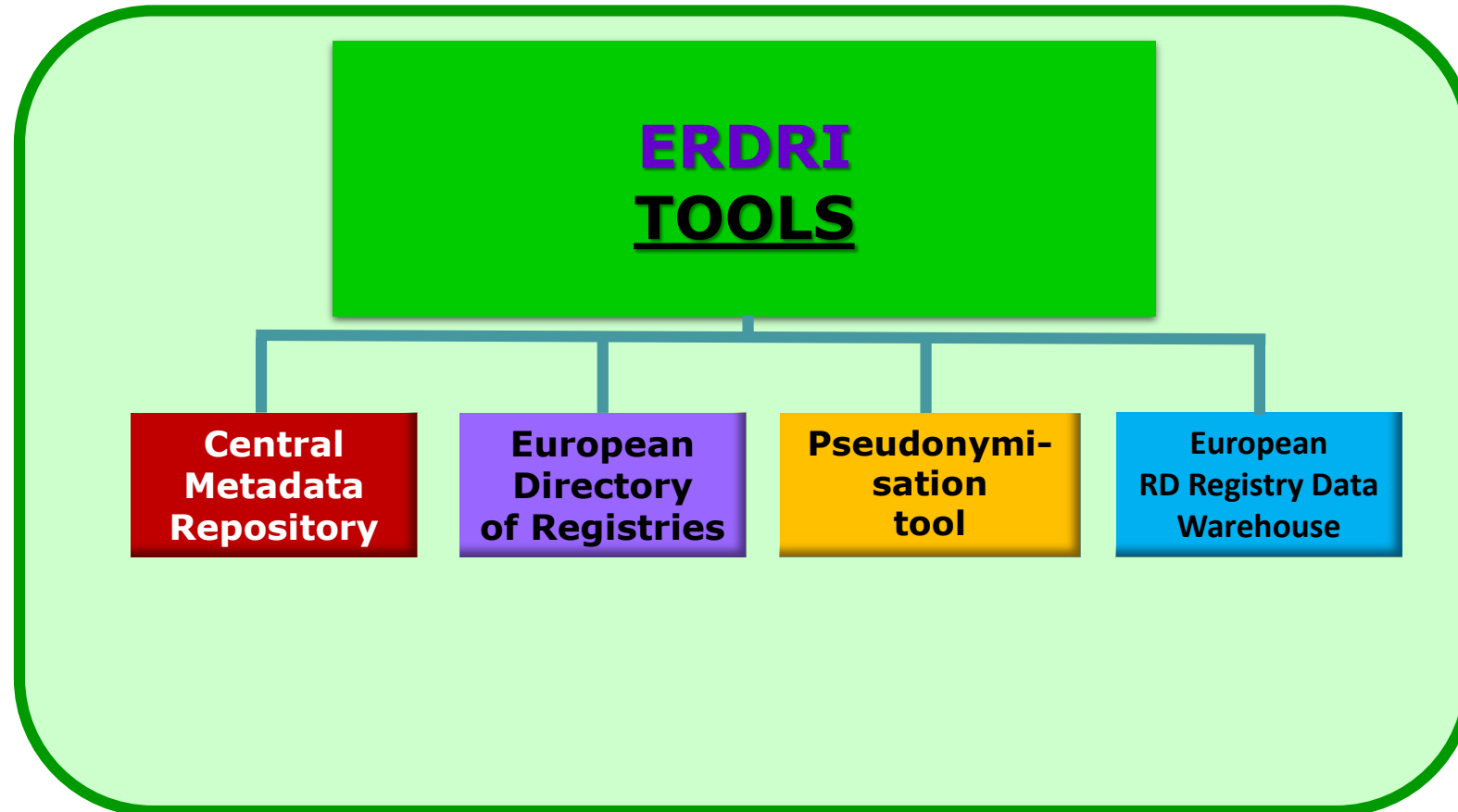


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ERDRI - functions

- **PROVIDES** the infrastructure and tools
 - ❖ To make registries' data **SEARCHABLE, FINDABLE, interoperable, reusable** (if accessible – it depends on the data owner!)
- **DOES NOT**
 - ❖ Store identifiable patient data
 - ❖ Host whole registries / their data

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European RD Registry Data Warehouse

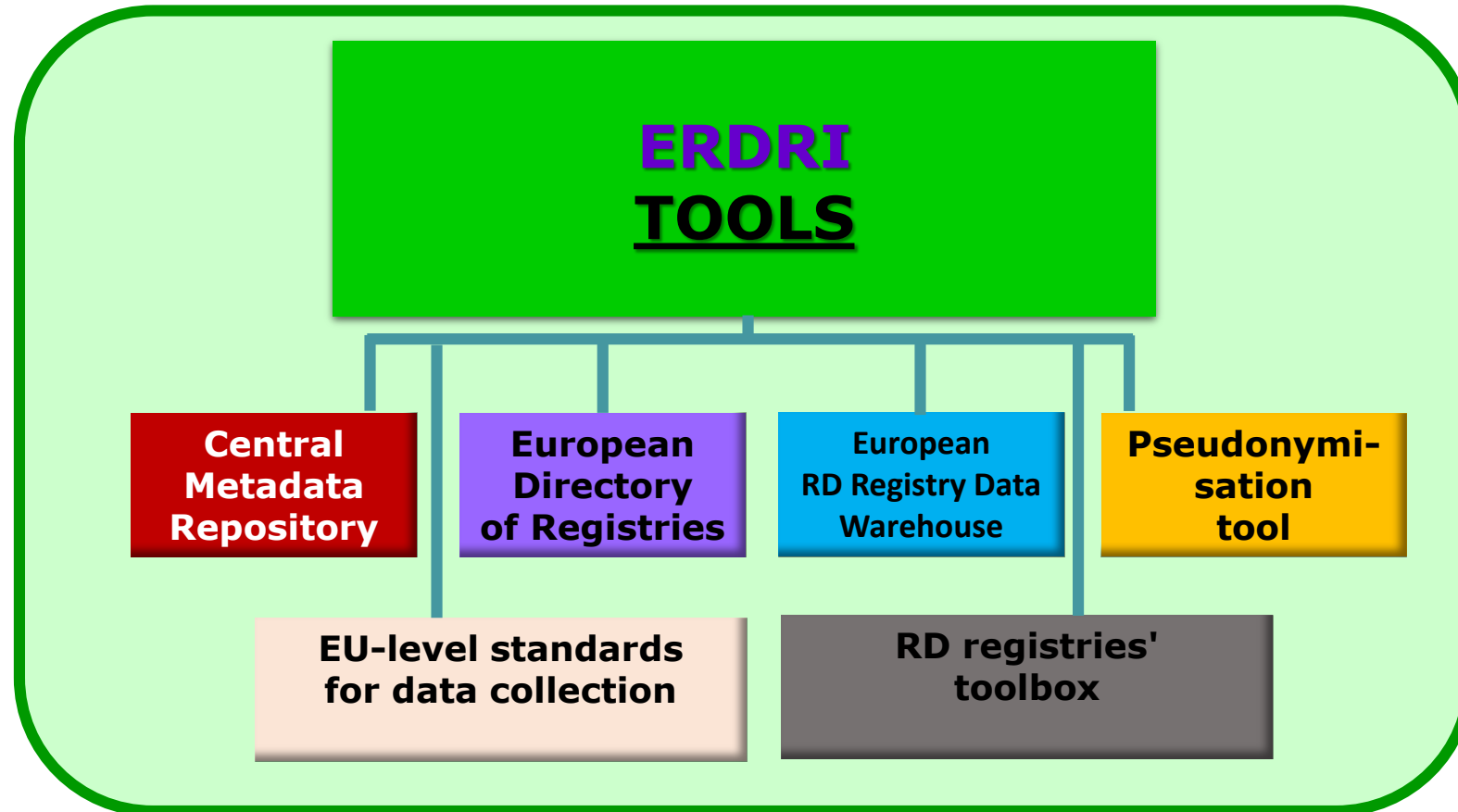
- Aggregated data
- Reporting data set (RDS):

Subset of Common Data Set without patient identification

Added value for all stakeholders:

Selected European RD data publicly accessible

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European RD Registry Infrastructure (ERDRI)

Interoperability provided by ERDRI

Benefits

Interoperability significantly improves registries' performance:

- helps to be part of an integrated resource
- maximises the utility of participating registries
- enables use of data across the registries
- provides accelerated communication
- enables automated data searching/finding
- enables studies and research, extended use and re-use of existing data for various purposes



**Thank You
for Your Attention**

www.jrc.ec.europa.eu

