Advancement of Treatments
FOR RARE DISEASES

The European Platform on Rare Disease Registration – EU RD Platform: Connecting data, accelerating care pathways and enabling research

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Advancement of Treatments for Rare Diseases
16 -17 June | The Cyprus Institute of Neurology & Genetics Nicosia, Cyprus

Organisers: THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
DEPUTY MINISTRY OF RESEARCH, INNOVATION AND DIGITAL POLICY
REPUBLIC OF CYPRUS
CY2026.EU
CYPRUS PRESIDENCY OF THE COUNCIL OF THE EU

European Commission

EU RD Platform

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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”**

(2008/0679)

Directive of the European Parliament and of the Council on the application **of patients’ rights in cross-border healthcare**

(2011/24/EU)

Council Recommendation of 8 June 2009 on **an action in the field of rare diseases**

(2009/C 151/02)

Setting the Direction improve coordination, knowledge, and patient outcomes across Member States.”

EU RD Platform

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

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UNMET NEEDS

High fragmentation of data sources

Need for critical number of patients / cohorts

Lack of interoperability between registries

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Copes with the fragmentation
of data sources across EU MS

Facilitates reaching **critical number** of patients for:

Studies

- epidemiological
- clinical
- translational
- pharmacolgical
- therapies

Supports **interoperability between registries:**

- Standardised data collection and exchange
- Semantic interoperability
- Make data FAIR
- Data linkage
- Data transfer

Is **a knowledge generation centre**
for RDs

→ reach critical numbers → Research, studies

for all possible purposes, advancing knowledge on RD

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European Rare Disease Registry Infrastructure (ERDRI)

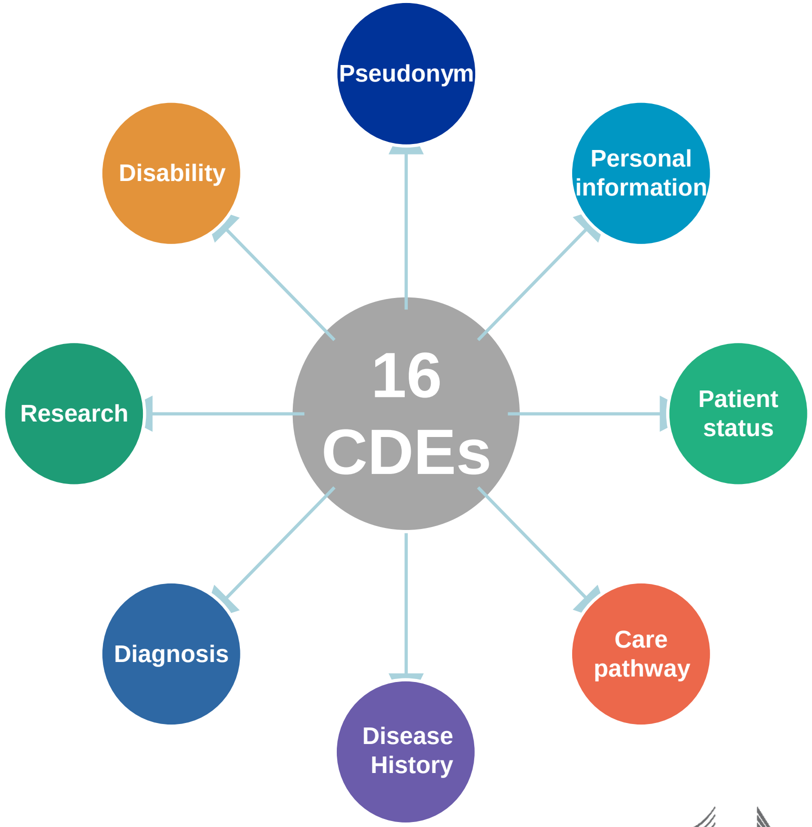
European standards for data collection and data sharing

Trainings, Resources and Latest news

eurocat

Surveillance of Congenital Anomalies in Europe

SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION



European Directory of Registries (ERDRI.dor)

Overview of rare disease registries in Europe including their characteristics

Central Metadata Repository (ERDRI.mdr)

Database containing the data elements used by rare disease registries

Search Broker (ERDRI.sebro)

ERDRI.sebro allows researchers to retrieve metatata of interest

SPIDER

Pseudonymisation tool (ERDRI.spider)

Service offering registries at local level the solution for patient pseudonymisation

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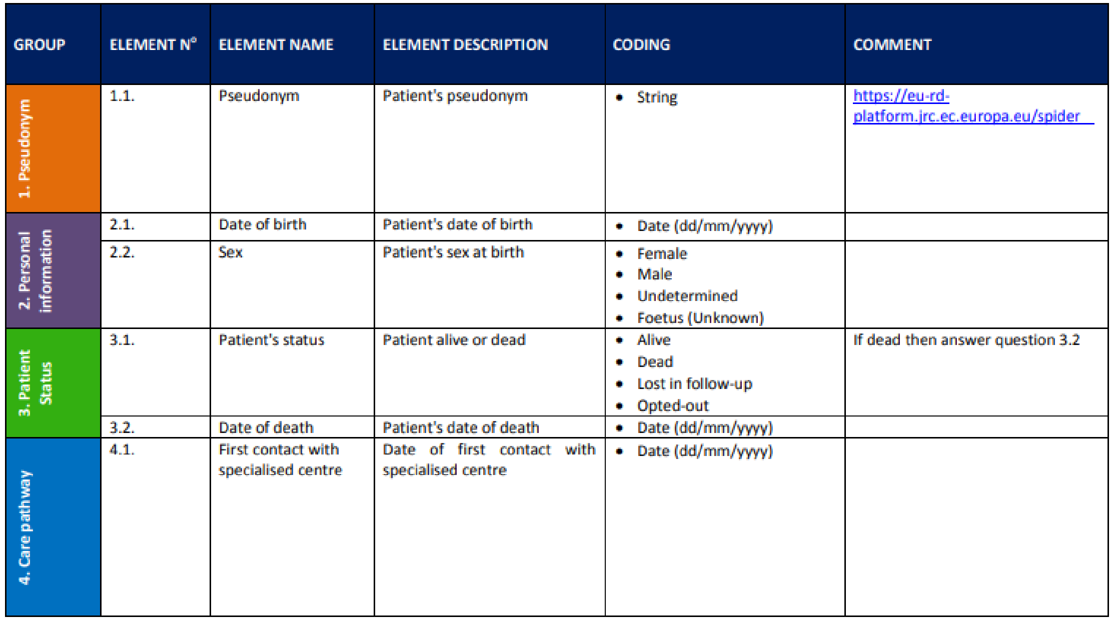
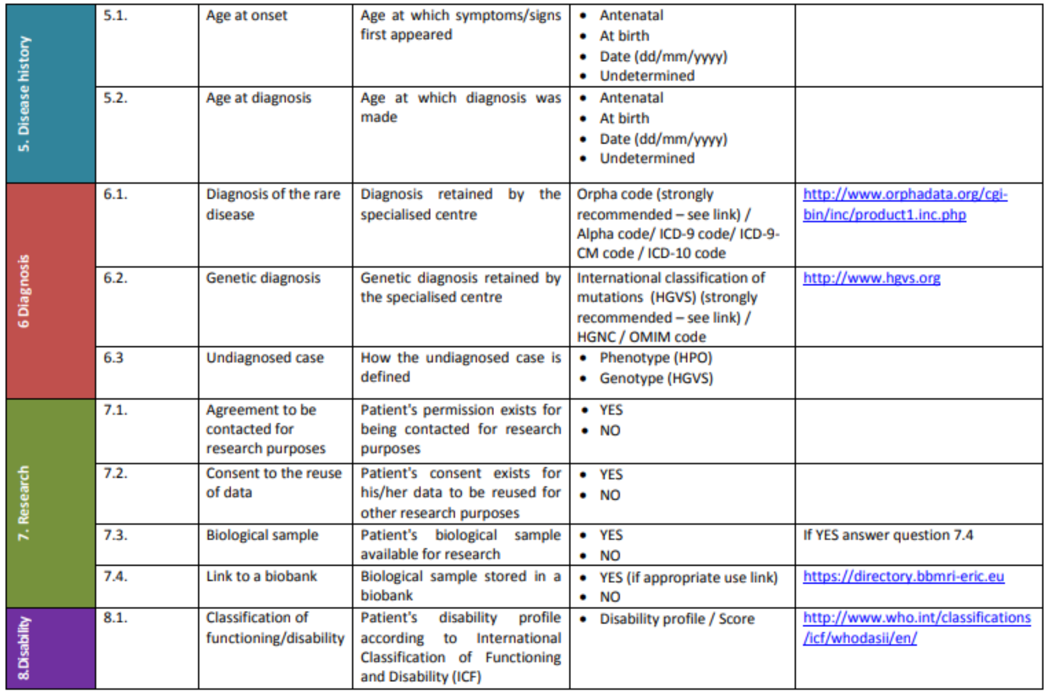
SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION



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/spider
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)	



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/

https://eu-rd-platform.jrc.ec.europa.eu/system/files/public/CDS/EU_RD_Platform_CDS_Final.pdf
available in 22 official EU languages

EU RD Platform

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European Rare Disease Registry Infrastructure (ERDRI)

European standards for data collection and data sharing

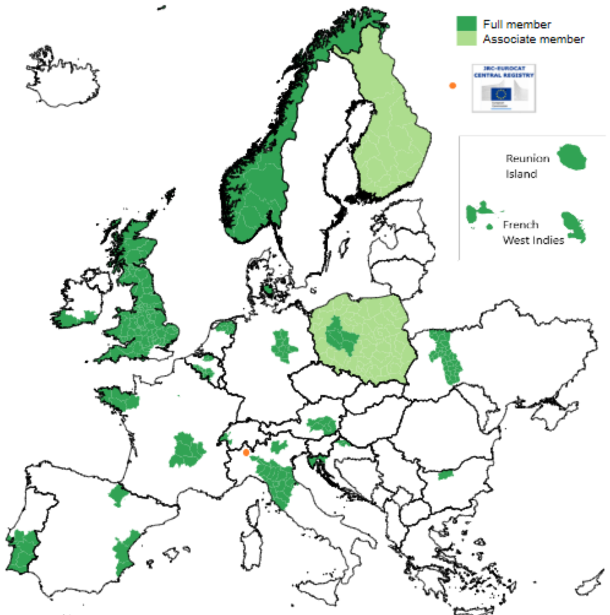
Trainings, Resources and Latest news

eurocat

Surveillance of Congenital Anomalies in Europe

European network of Congenital Anomaly Registries

- 40 Full registries
- > 650,000 individual cases
- **Surveillance** and statistical monitoring
- JRC-owned software for case registration and analysis



Administrative boundaries: © EuroGeographics

Role of the Central Registry:

- To maintain and further develop the **Central Database**
- To **securely manage the data** from all registries
- To **analyse data** with respect to data quality and routine statistical monitoring
- To maintain **relationship with single registries**
- To support and participate in the **coordinating activities**
- To organise **meetings** (annual network meetings, Management Committee and various Working Groups) and trainings
- To **disseminate network's results** (website, reports, JRC-EUROCAT communication)

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European Rare Disease Registry Infrastructure (ERDRI)

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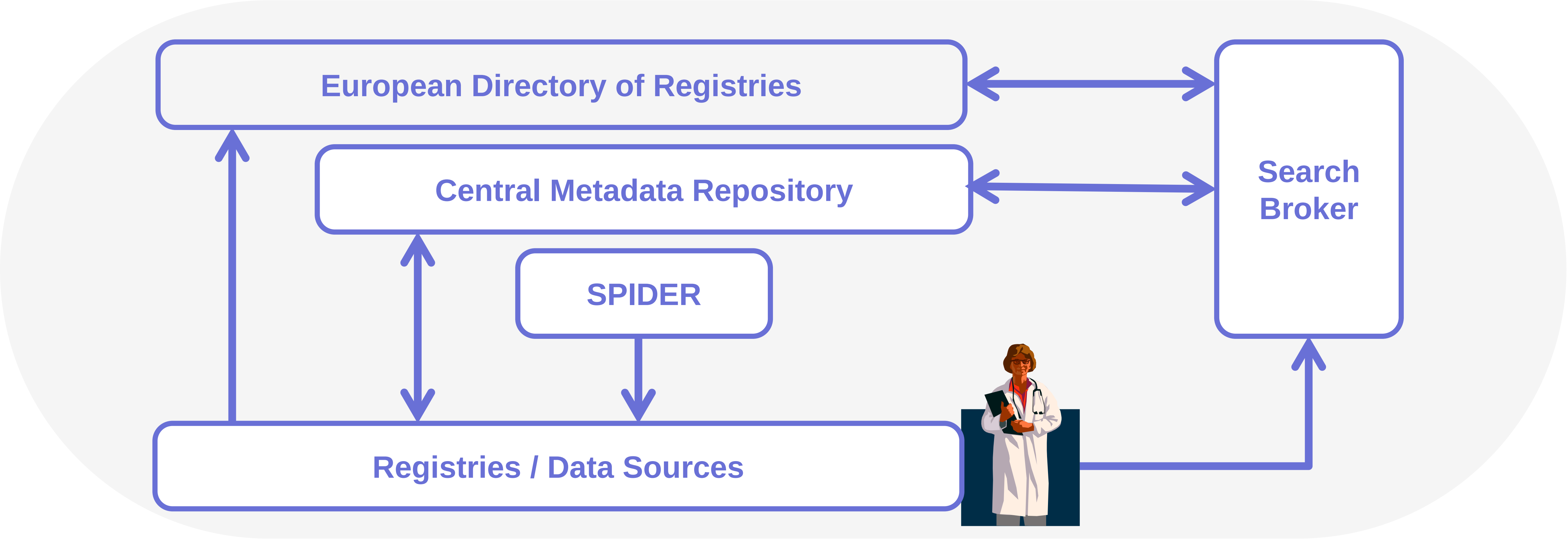

European Directory of Registries

Central Metadata Repository

SPIDER

Search Broker

Registries / Data Sources

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European Rare Disease Registry Infrastructure (ERDRI)

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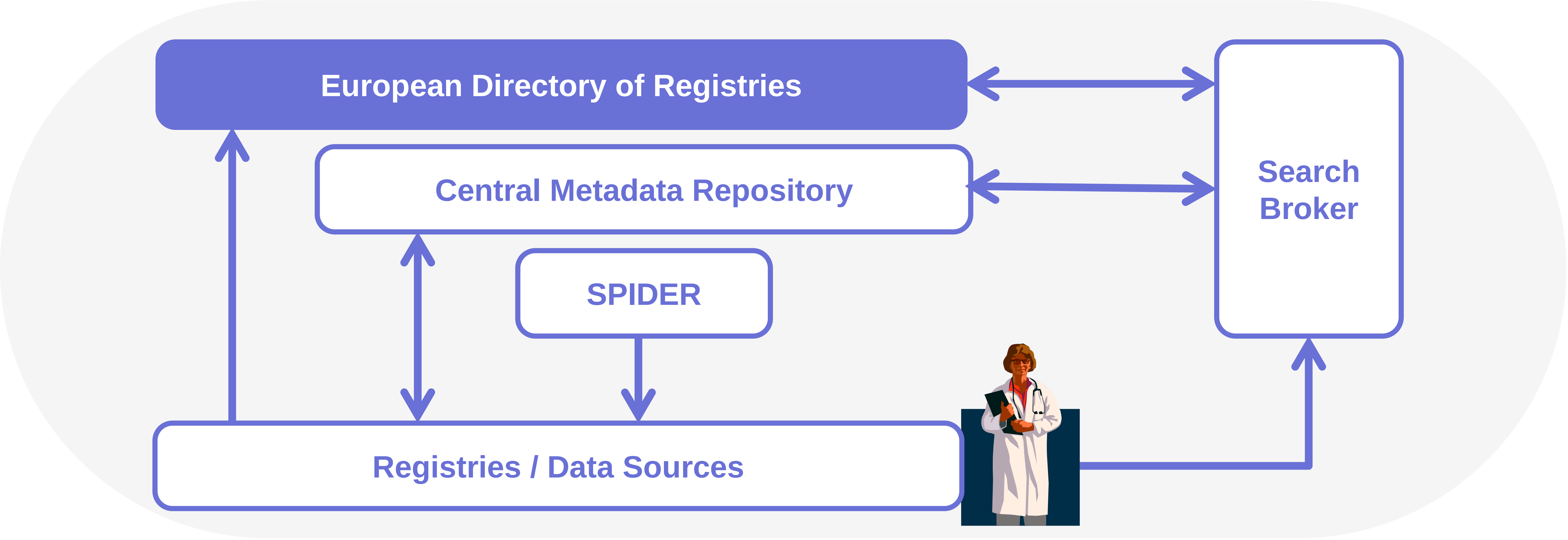

Participating registries: key characteristics and descriptions across **42 data fields**
Data entry by registry owners

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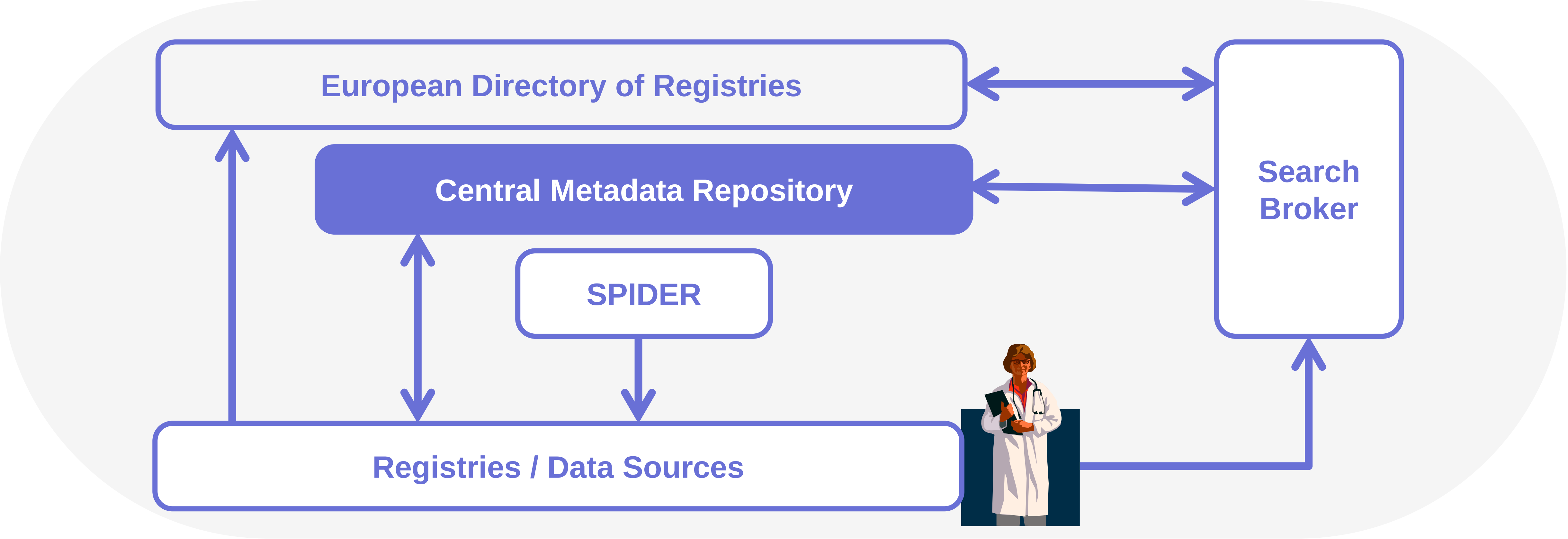

European Directory of Registries

Central Metadata Repository

SPIDER

Search Broker

Registries / Data Sources

Metadata collection: semantic interoperability |data elements | definitions | descriptions | units of measurement
Data entry by registry owners according to specific validation rules and upload methods

ERDRI statistics

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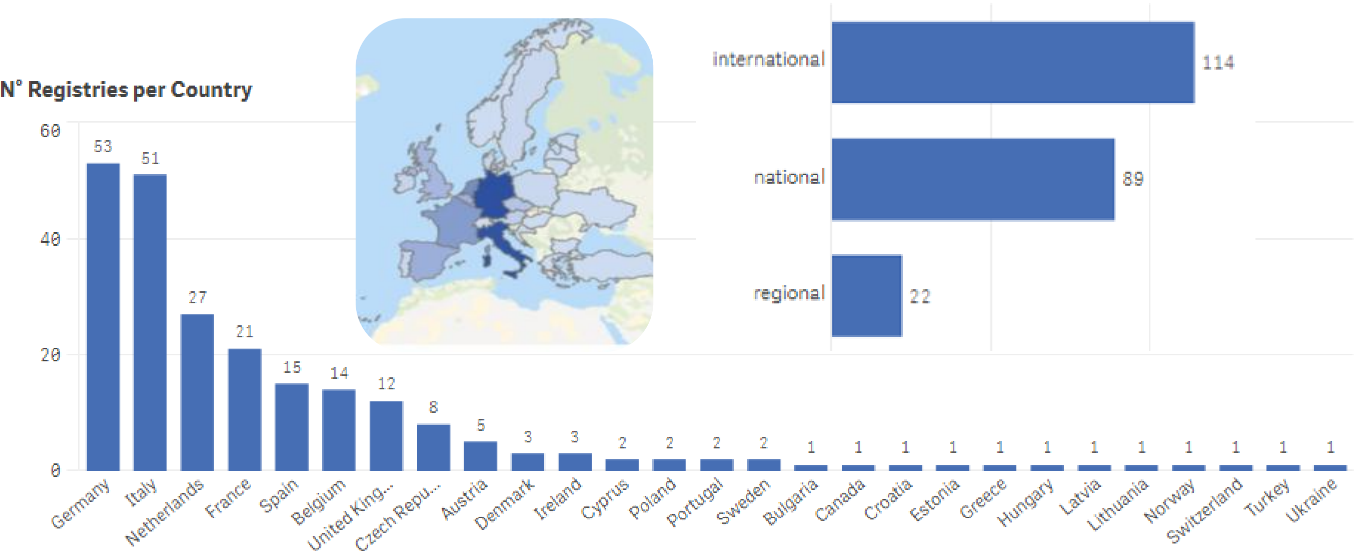
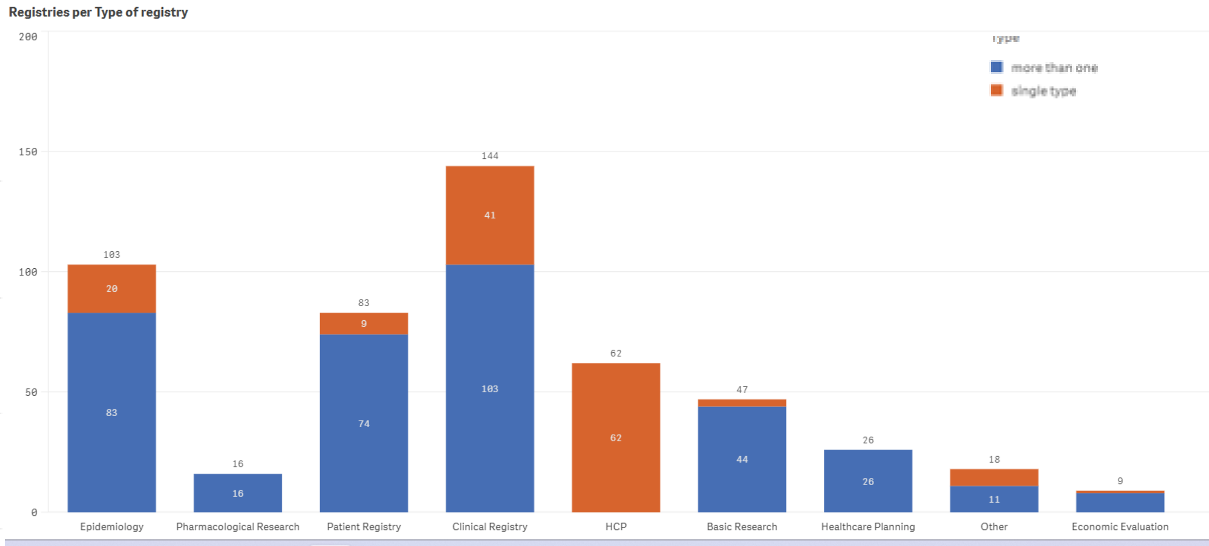
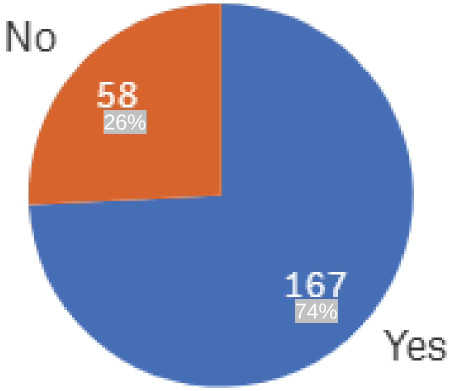
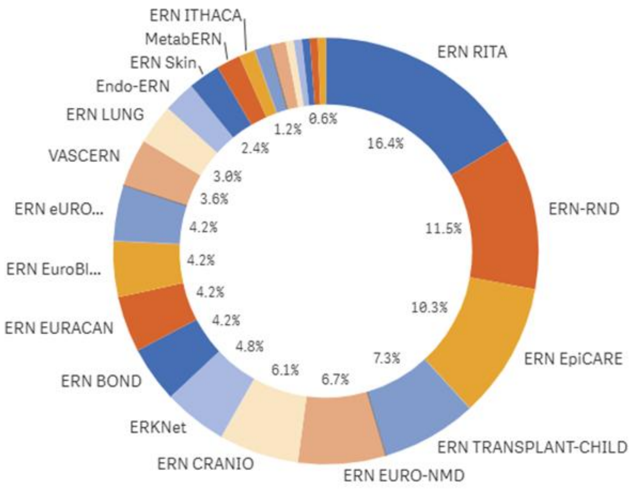


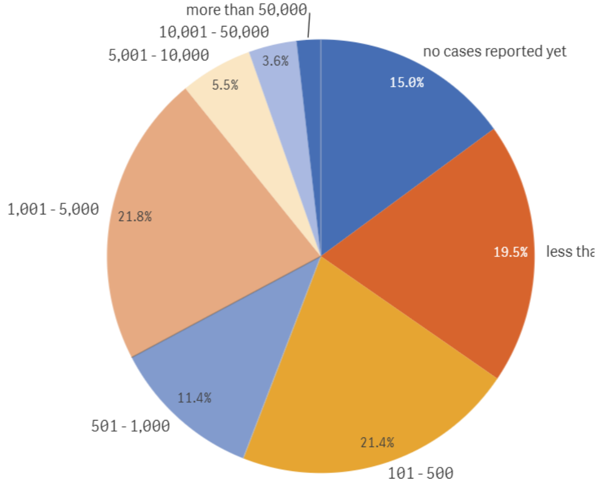




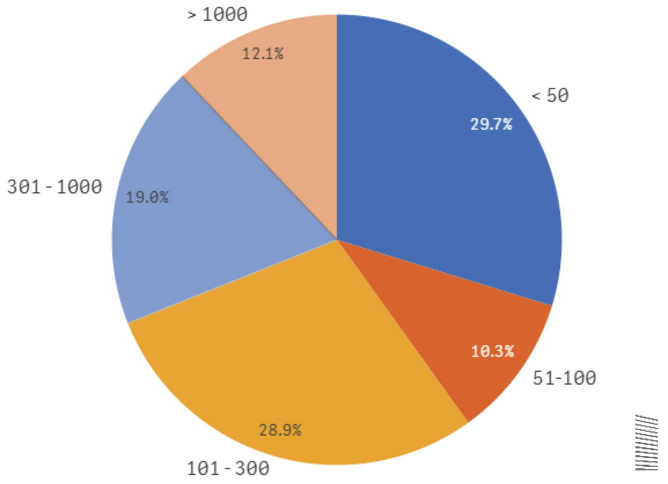
ERN vs non-ERN



Percentage of Registries per ERN

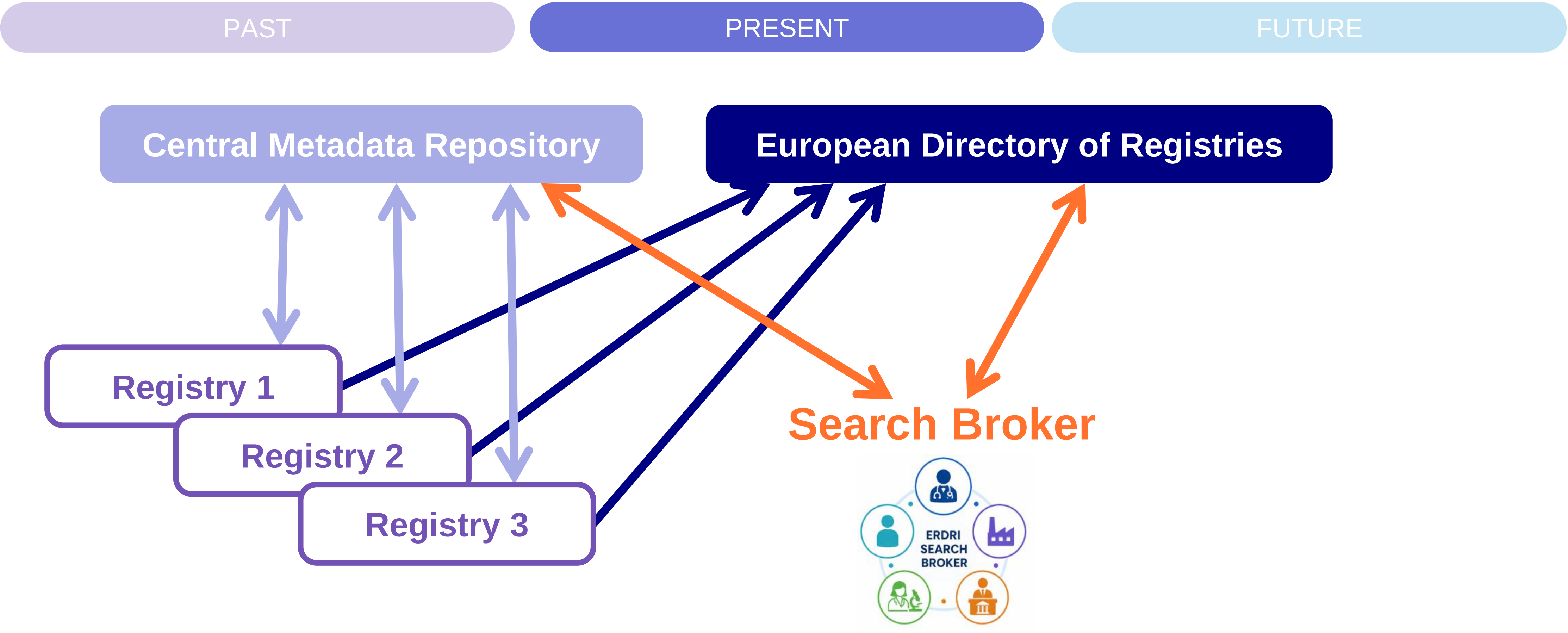


% of registires by range of cases reported



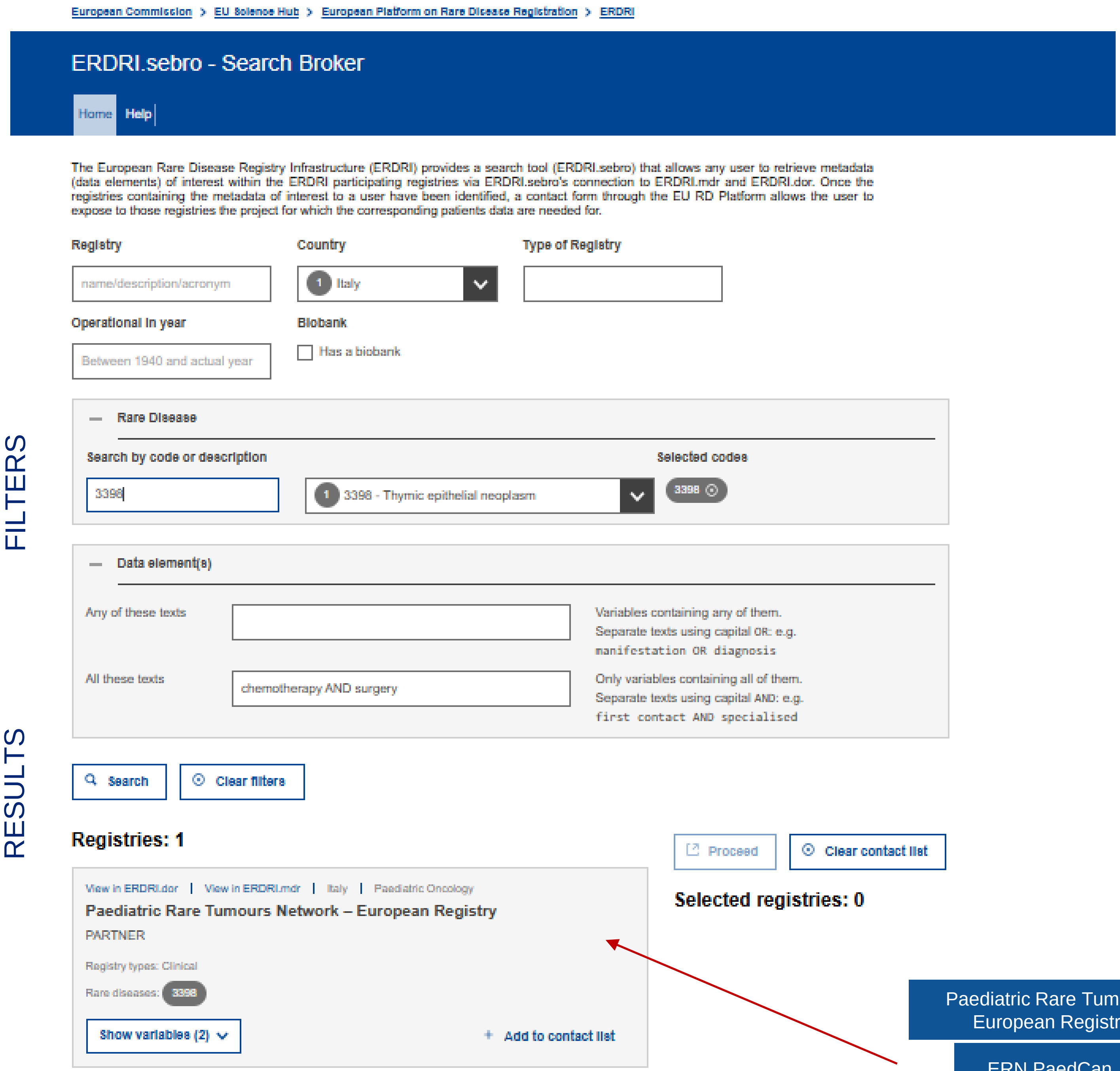
% of registries by data element range

ERDRI: Search tool



FILTERS

RESULTS


European Commission > EU Science Hub > European Platform on Rare Disease Registration > ERDRI

ERDRI.sebro - Search Broker

Home | Help

The European Rare Disease Registry Infrastructure (ERDRI) provides a search tool (ERDRI.sebro) that allows any user to retrieve metadata (data elements) of interest within the ERDRI participating registries via ERDRI.sebro's connection to ERDRI.mdr and ERDRI.dor. Once the registries containing the metadata of interest to a user have been identified, a contact form through the EU RD Platform allows the user to expose to those registries the project for which the corresponding patients data are needed for.

Registry: name/description/acronym
Country: 1 Italy
Type of Registry:
Operational in year: Between 1940 and actual year
Biobank: ☐ Has a biobank

Rare Disease

Search by code or description: 3398
3398 - Thymic epithelial neoplasm
Selected codes: 3398

Data element(s)

Any of these texts:
Variables containing any of them. Separate texts using capital OR: e.g. `manifestation OR diagnosis`

All these texts: chemotherapy AND surgery
Only variables containing all of them. Separate texts using capital AND: e.g. `first contact AND specialised`

Registries: 1

Selected registries: 0

View in ERDRI.dor | View in ERDRI.mdr | Italy | Paediatric Oncology

Paediatric Rare Tumours Network – European Registry

PARTNER

Registry types: Clinical

Rare diseases: 3398

Paediatric Rare Tumors - European Registry

ERN PaedCan

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

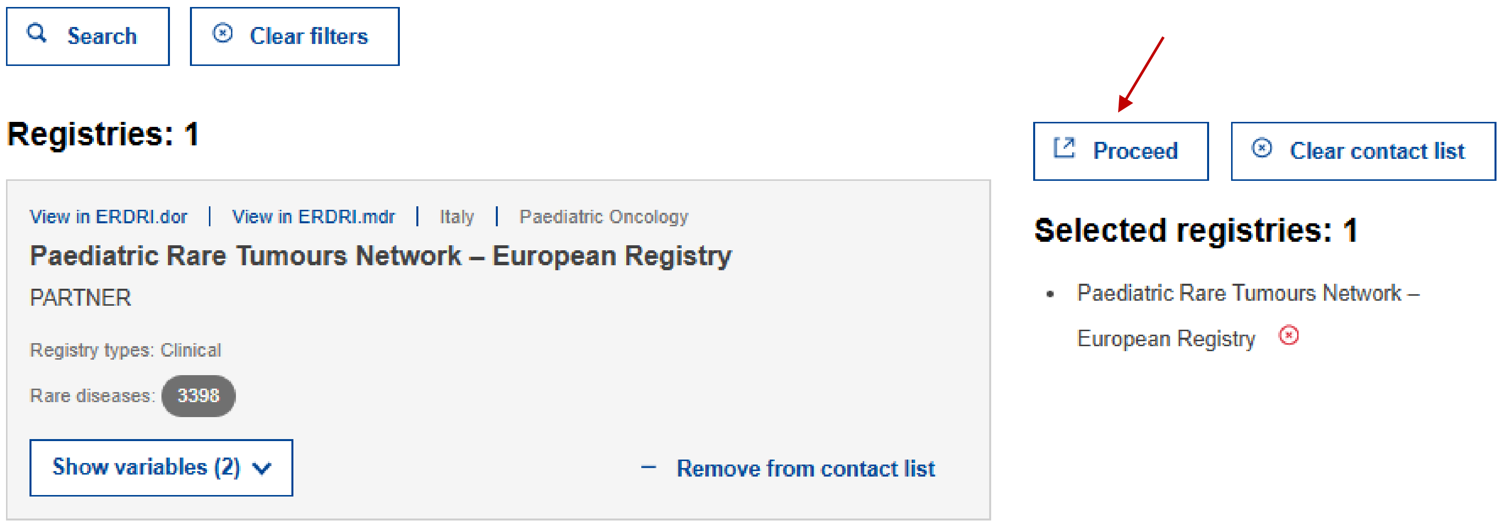
User inserts keywords

DOR and MDR are queried

Registries matching the request are identified

DOR: >70 var

MDR: >20 var



Registries: 1

View in ERDRI.dor | View in ERDRI.mdr | Italy | Paediatric Oncology

Paediatric Rare Tumours Network – European Registry

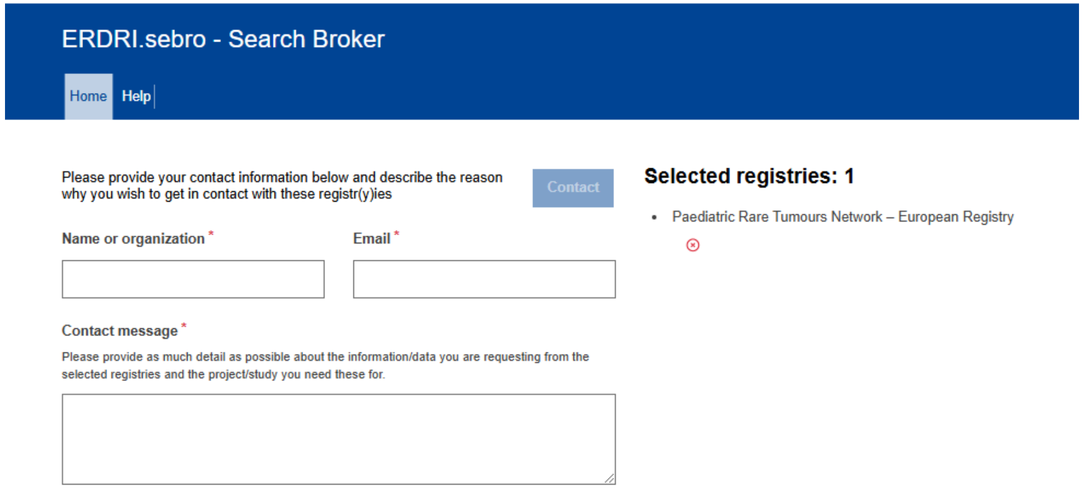
PARTNER

Registry types: Clinical

Rare diseases: 3398

Selected registries: 1

- Paediatric Rare Tumours Network – European Registry



Please provide your contact information below and describe the reason why you wish to get in contact with these registr(y)ies

Name or organization *

Email *

Contact message *

Please provide as much detail as possible about the information/data you are requesting from the selected registries and the project/study you need these for.

Selected registries: 1

- Paediatric Rare Tumours Network – European Registry

Request/study proposal /cohort building contact with the registries

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European Rare Disease Registry Infrastructure (ERDRI)

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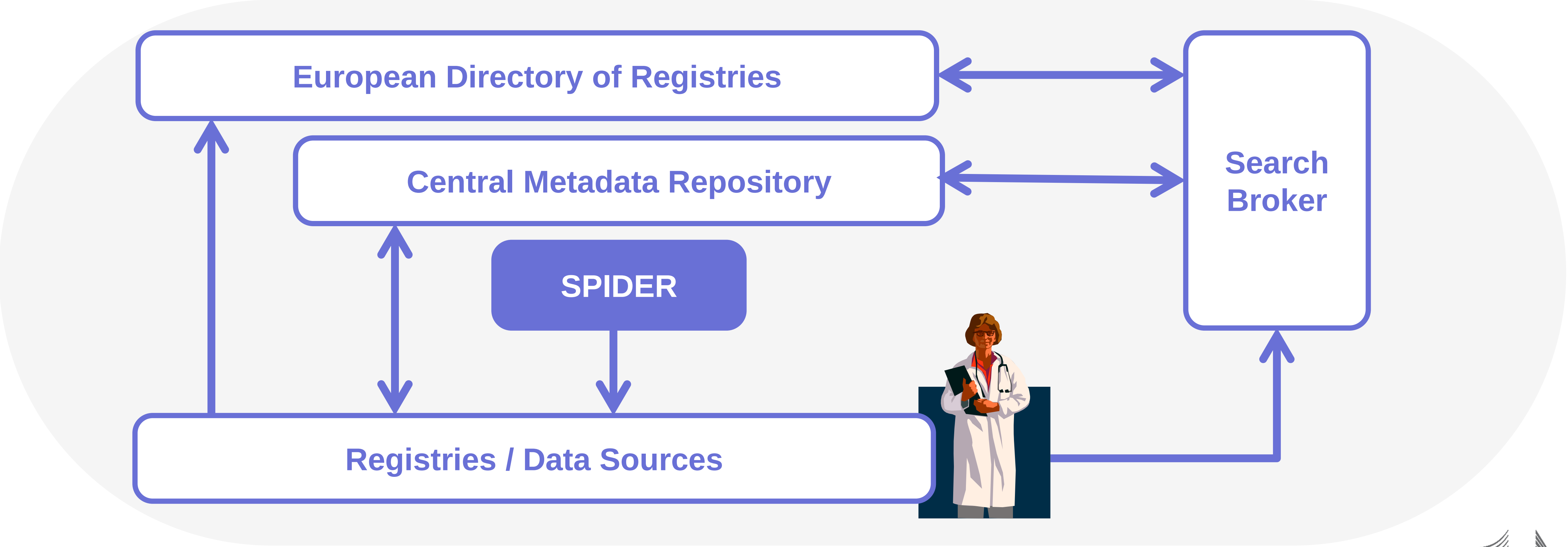

ERDRI: SPIDER

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PSEUDONYMISATION TOOL
Service offering registries at local level the solution for patient pseudonymisation.

SPIDER READY:
87 registries /centres

- ERNs
- sub-networks

Does NOT have access to patient data!

Pseudonym *GENERATION*

Pseudonym *LINKAGE*

Encrypted pseudonymised data *TRANSFER*

policy enforcement

SPIDER-ready registries/HCPs

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1. Pseudonym Generation

2. Pseudonym Linkage

3. Encrypted Transfer

policy enforcement

Meet prerequisites to access ERDRI tools

Have a SPIDER client

Have a cryptographic archive file

European Commission > EU Science Hub > European Platform on Rare Disease Registration > ERDRI

ERDRI

European Rare Disease Registry Infrastructure (ERDRI)

Web based client

- Provided for free
- Open source

SPIDER HOMEPAGE

CryptoArchive.p12

- Generated just once by the registry owner

SPIDER

Spider | Share | Network

Training & Support

Curated Educational Tracks

Onboarding ERDRI.dor & ERDRI.mdr | Preparation rfo (Use of SPIDER

Scheduled Delivery

General and specific ERN trainings delivered in 2024, 2025 and 2026 (CRANIO | eUrogen | RND | EURACAN)

Materials & Documentation: F.A.Q.S | TUTORIALS | GUIDES

https://eu-rd-platform.jrc.ec.europa.eu/training-resources-and-latest-news/resources_en

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

Dedicated support team: EU-RD-Platform@ec.europa.eu

SPIDER Use - Registries/HCPs

Spider Share Network

87/225 (39%)

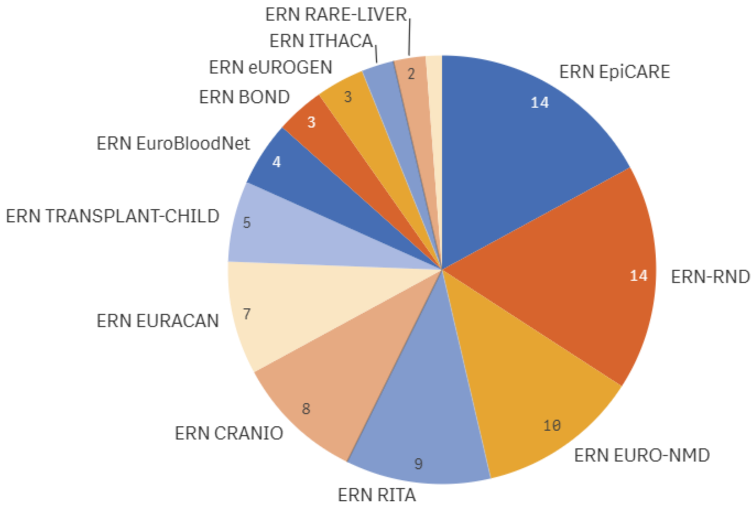
- ERN registries/HCPs with issued SPIDER cryprographic archive 79/167 (47%) + 8 non ERN
- declared at least one «allowed user»: 79/87

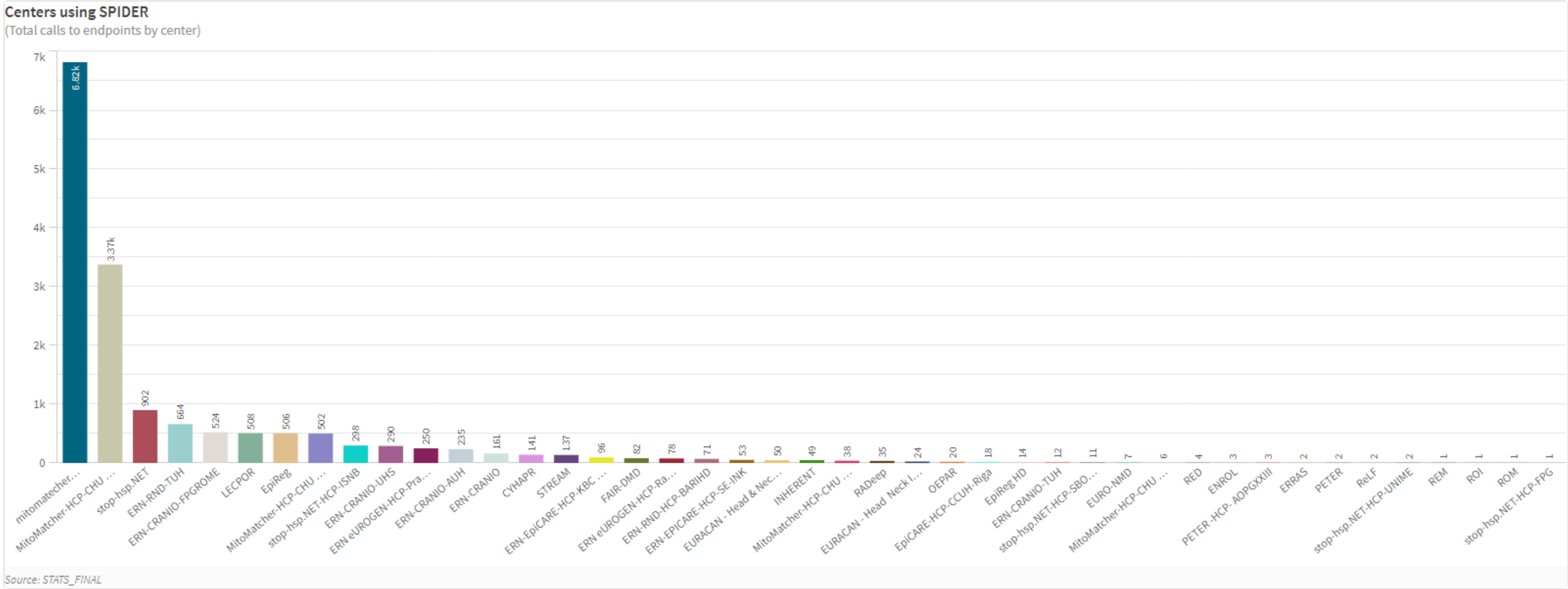
43

registries/HCPs using SPIDER
Euro-NMD | RND | CRANIO| EPICARE | EURACAN | eUrogen | Eurobloodnet | Transplant Child | BOND

***Data for one year**
*out of all 225 ERDRI registries)

European Reference Networks
EpiREG




Centers using SPIDER
(Total calls to endpoints by center)

Source: STATS_FINAL

EU RD Platform

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Strategic policy context

European Health Data Space (EHDS)

- Key pillar of the European Health Union and first common EU data space in a specific area.
- Leverages existing health data for regulatory, policy and clinical making.
- JRC contribution: data collection, standardisation, quality, privacy and security.

“Choose Europe for Life Sciences”

Communication from the Commission :To the European parliament, the Council, the European Economic and Social committee and the committee of the regions: Choose Europe for life sciences: A strategy to position the EU as the world’s most attractive place for life sciences by 2030

Optimising the R&I ecosystem to promote a globally competitive life science sector.

Reinforcing European R&I

- The creation of new knowledge
- Successful models for R&I ecosystems
- **European Platform on Rare Disease Registration / EU RD Platform** : mentioned directly in the strategy under **“Reinforcing European R&I”**

EU RD Platform: future role

Data • Quality • Interoperability

From registry readiness to EHDS integration and EU-wide impact

1 **Secondary use**

Use of data for research and public-interest purposes.

2 **EHDS data ecosystem**

Data holders ↔ **Data users**

3 **Preparing ERN/RD registry data for integration into the EHDS**

Strategic opportunity for ERN/RD registries

- Reuse ERDRI metadata and avoid duplication | Build sets of high-quality metadata records
- mapped or aligned to Health-DCAT-AP → discoverable in EHDS catalogues.
- Establish ERN registry descriptions as the rare disease metadata gold standard
- Shape EHDS implementation from within | Be ready when EHDS comes into force in 2029

Future direction: preparing ERN/RD registry data for integration in the EHDS


2Cu

Thank you