Opportunities for integration of the European Platform on Rare Disease Registration (EU RD Platform) with HMA/EMA catalogues

DG Joint Research Centre (JRC) F1
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HMA/EMA multi-stakeholder workshop on patient registries
Amsterdam, 13.02.2024

Why an EU RD Platform?

30 million people affected by RDs

- To cope with the fragmentation of data sources across EU MS
- To reach the critical number of patients for
  - Studies
  - Research
  - Building patient cohorts
- To reach interoperability between registries
  - Standards for data collection and data exchange
  - Semantic interoperability
  - FAIR data
  - Data linkage
  - Data transfer

Source: EURORDIS

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

European Commission

EU RD Platform: same aims as HMA/EMA catalogues

- Ensuring **DISCOVERABILITY** of DATA from participating RD registries. Making registries' **DATA SEARCHABLE AND FINDABLE**

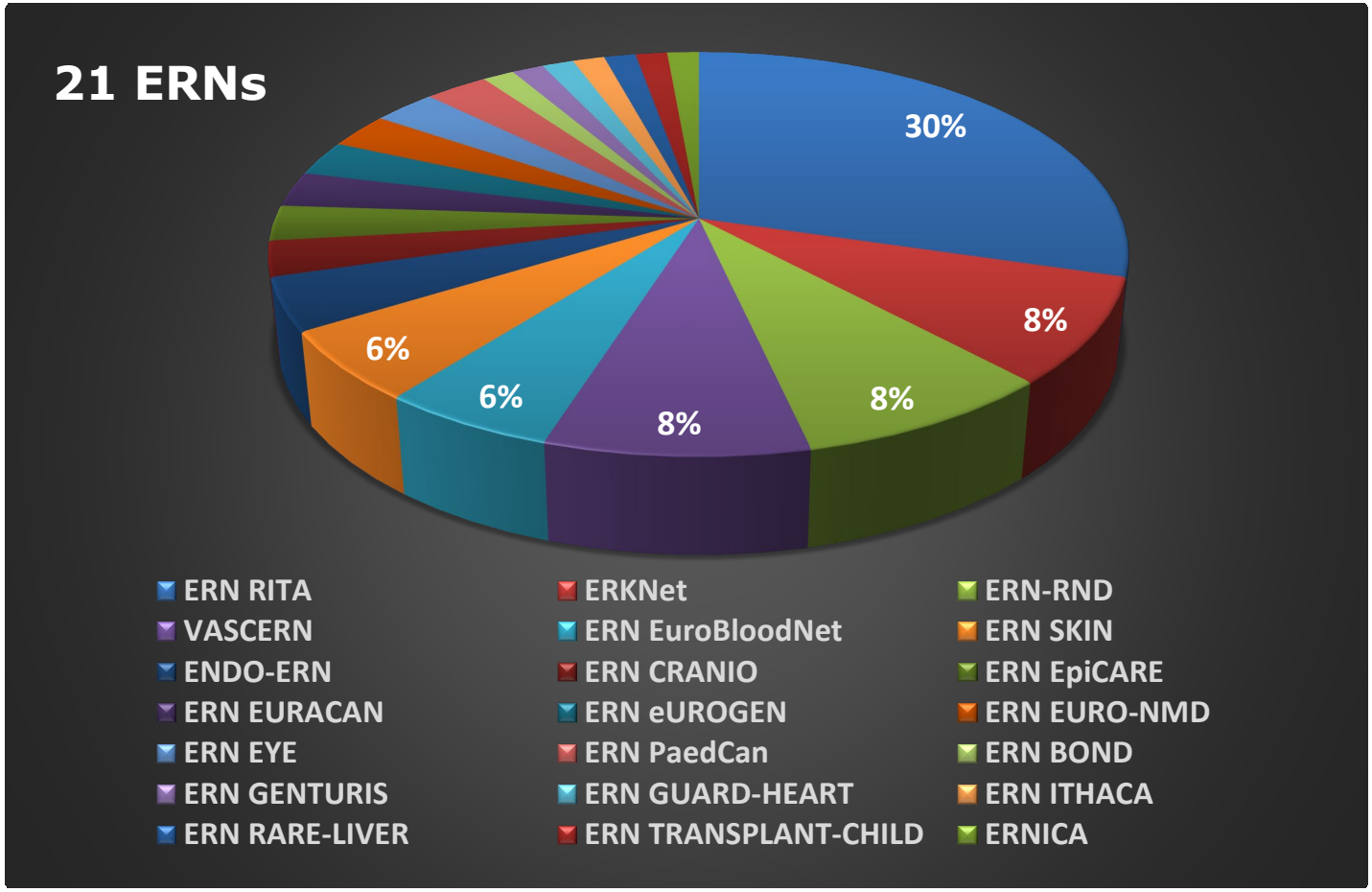
- Giving **VISIBILITY** to registries. Registries remain owners of their data and decide about the use of the data.

Opportunities for better integration EU RD Platform with HMA/EMA catalogues

- European Directory of Registries aligned with metadata list of HMA/EMA catalogues
- Data Elements Repository to enrich HMA/EMA catalogues
- Search tools that could be combined
- Pseudonymisation/data linkage/data transfer (SPIDER)
- Standards for data collection:

  Set of common data elements for RD registration - explore integration with EMA Data Quality Framework in the context of common data model

ERN representation in the EU RD Platform



European Commission

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

European Rare Disease Registry Infrastructure (ERDRI)

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

ERDRI Search broker (ERDRI.sebro)

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

European Commission

EU RD Platform

DATA availability and use

for various purposes, advancing knowledge on RD:

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

→ reach critical numbers

→ perform studies, research

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)	

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/