European Reference Network
for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)

A FAIR compliant registry for the vascular malformation centers within the European Reference Networks.

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www.VASCERN.eu

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Goal

- Combining registries/databases
- Links to biodatabanks and other « resources/databases/research nets
- »big data analyses »

- Multiple Problems:
- Ownership
- Privacy (EU GPRS)
- Compatibility (different databases)

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The FAIR principle

- FAIR
  - Findable (FAIR datapoints, metadata repositories)
  - Accesible (GDPR compliant, after approval by the owner of the data)
  - Interoperable (RDF and Ontologies,SnomedCT)
  - Reusable
- Gaining wide spread acceptance since introduction
  - Sci Data. 2016 Mar 15;3:160018. doi: 10.1038/sdata.2016.18
  - EU research community,
  - G7
  - NIH
- Advantages
  - Not dependant on database program
  - No identical databases needed
  - Data stays locally, full control by the owners
  - In accordance with the new GDPR (General Data Protection Regulation)

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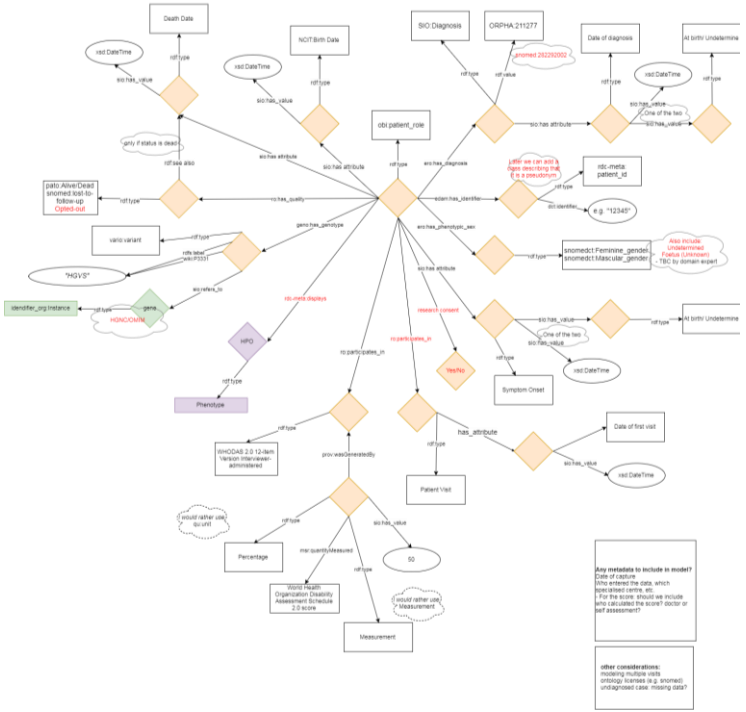
Registry requirements

RDF

Semantic modeling and ontologies

SNOMED, RADLEX,NCIT)



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Common data elements

EUROPEAN COMMISSION
JOINT RESEARCH CENTRE
Directorate F – Health, Consumers and Reference Materials
Unit F.1 – Health in Society

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	• 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	• 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)	
	5.1.	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	
	6.1.	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)	
	7.1.	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
	8.1.	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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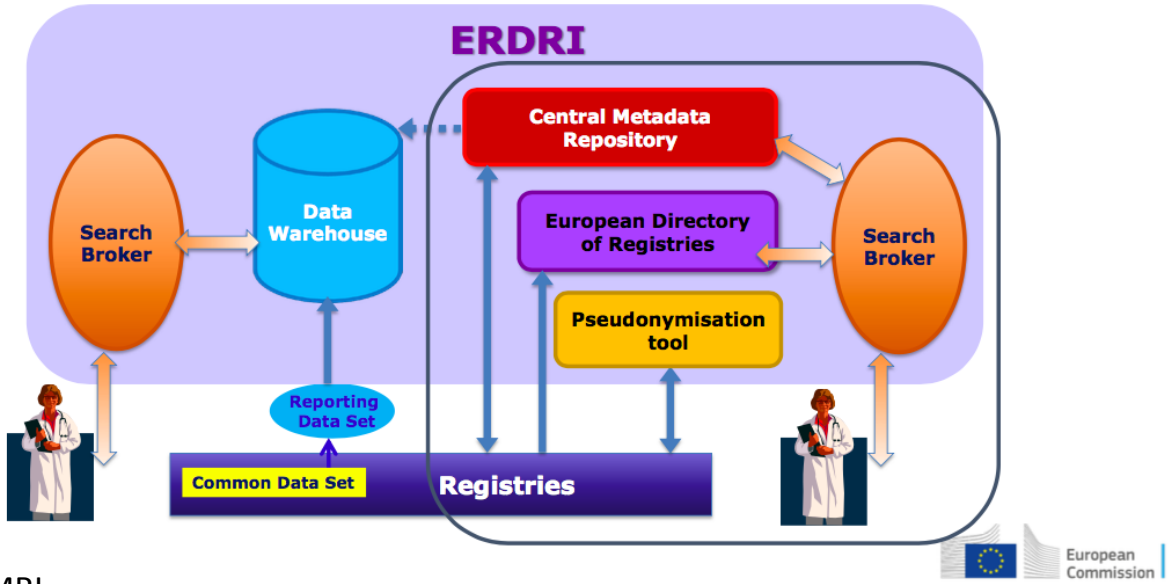
European Platform on Rare Diseases Registration



Global initiatives

- FAIR tools and protocol developers
- FAIR deployment service

Infrastructures & networks

- Research infrastructures: ELIXIR, BBMRI
- European reference networks (ERN)
- European Open Science Cloud

Projects

- ERN registry projects
- RI projects: EXCELERATE, CORBEL, ADOPT
- RD RI projects: RD-Connect, RD-Bridges

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Future

- Registry and the platform are developed and in the final test phase.
- Privacy and owner rights garanteed

- Collaborative research
- Big data analysis