



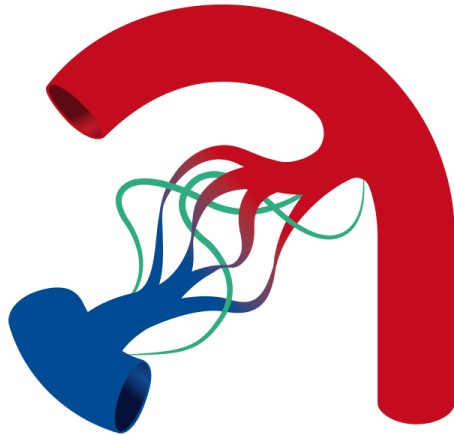
**European  
Reference  
Network**

for rare or low prevalence  
complex diseases



**Network**

Vascular Diseases  
(VASCERN)



# Registry Working Group session



# Whats done

- Last meeting vasca was selected as the WG were the pilot with the FAIR registry would start with focus on the common data set.
- Common dataset was the set selected by the EU (8 items) which had to be part of any registry.
- Part of the EJP-RD grant (Nijmegen 700k)
  - Health train expert t'Hoen appointed as chair
- Working group of FAIR experts
- Vascern part of the Go-Fair network
- Data steward at Radboud
- Data steward at the VASCERN office (job description finished)
- *Collaboration between ERN ENDO- ERN BOND and VASCERN*



## 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"> <li>String</li> </ul>   | 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"> <li>Date (dd/mm/yyyy)</li> </ul>  |   |
|                         | 2.2.       | Sex                                   | Patient's sex at birth                        | <ul style="list-style-type: none"> <li>Female</li> <li>Male</li> <li>Undetermined</li> <li>Foetus (Unknown)</li> </ul> |   |
| 3. Patient Status       | 3.1.       | Patient's status                      | Patient alive or dead                         | <ul style="list-style-type: none"> <li>Alive</li> <li>Dead</li> <li>Lost in follow-up</li> <li>Opted-out</li> </ul>    | If dead then answer question 3.2  |
|                         | 3.2.       | Date of death                         | Patient's date of death                       | <ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>  |   |
| 4. Care pathway         | 4.1.       | First contact with specialised centre | Date of first contact with specialised centre | <ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>  |   |

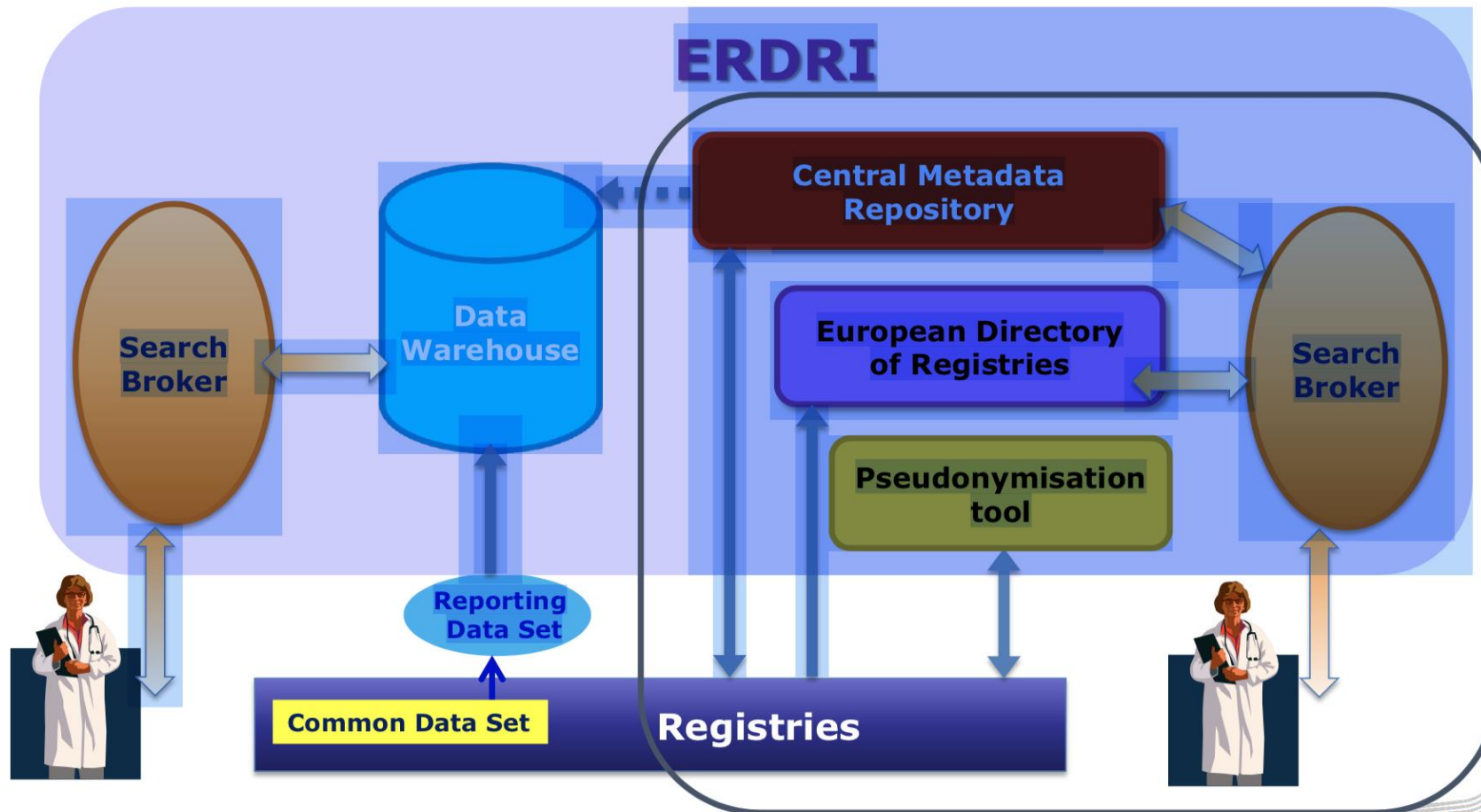
|                    |      |   |  |  |   |
|--------------------|------|---|--|--|---|
| 5. Disease history | 5.1. | Age at onset                                    | Age at which symptoms/signs first appeared   | <ul style="list-style-type: none"> <li>Antenatal</li> <li>At birth</li> <li>Date (dd/mm/yyyy)</li> <li>Undetermined</li> </ul> |   |
|                    | 5.2. | Age at diagnosis                                | Age at which diagnosis was made  | <ul style="list-style-type: none"> <li>Antenatal</li> <li>At birth</li> <li>Date (dd/mm/yyyy)</li> <li>Undetermined</li> </ul> |   |
| 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                             | <a href="http://www.orphadata.org/cgi-bin/inc/product1.inc.php">http://www.orphadata.org/cgi-bin/inc/product1.inc.php</a> |
|                    | 6.2. | Genetic diagnosis                               | Genetic diagnosis retained by the specialised centre   | International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code                          | <a href="http://www.hgvs.org">http://www.hgvs.org</a>   |
|                    | 6.3. | Undiagnosed case                                | How the undiagnosed case is defined  | <ul style="list-style-type: none"> <li>Phenotype (HPO)</li> <li>Genotype (HGVS)</li> </ul>                                     |   |
| 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"> <li>YES</li> <li>NO</li> </ul>  |   |
|                    | 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"> <li>YES</li> <li>NO</li> </ul>  |   |
|                    | 7.3. | Biological sample                               | Patient's biological sample available for research   | <ul style="list-style-type: none"> <li>YES</li> <li>NO</li> </ul>  | If YES answer question 7.4  |
|                    | 7.4. | Link to a biobank                               | Biological sample stored in a biobank  | <ul style="list-style-type: none"> <li>YES (if appropriate use link)</li> <li>NO</li> </ul>                                    | <a href="https://directory.bbmri-eric.eu">https://directory.bbmri-eric.eu</a>   |
| 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"> <li>Disability profile / Score</li> </ul>   | <a href="http://www.who.int/classifications/icf/whodasii/en/">http://www.who.int/classifications/icf/whodasii/en/</a>     |



# Whats Done

- Common data set (semantic modeling and machine language) is now ready. First evaluation by VASCA is done.
- Start with the registry asap after approval by the WG.
- Adding of WG specific items is possible
  
- Will be use-case for the further implementation of the FAIR principle for other ERN's
  
- Developments within the ERDiRI (european rare disease registration infrastructure)

# European Platform on Rare Diseases Registration





# Next steps

- Support team formed.
- Use-case for the all the other ERN's,
- Implementation of the FAIR data set in the registries of the other WG
  - Discussion in each WG (please add to the agenda!)
    - **Yes or no for implementation**
      - Yes HTAD, HHT, PPL, Vasca.
      - No MSA?
    - **Any additional data elements**

Further discussion of plans in the friday WG meeting



# Next steps

- Identical proces like vasca except that common data elements are ready
- Responsible person within the other WG. Mile stone: up and running aug 2019
- Set up a core team. (data stewards Paris, Nijmegen, ? Other ERN's)
  - Team will help WG and HCP with set up of the registries
- Costs: server cloud services, registry program. (proposal prepared by the datasteward team)
- NB: local data input has to be taken care of the local HCP