ERN | EuroBloodNet

RADeep

13th June 2025 - EHA Congress

Role of ERN Registries in advance research in hematology

EPIDEMIOLOGY AND DATA DRIVEN

Speaker: Sara Reidel

European Reference Network | Hematological Diseases (ERN EuroBloodNet)

Funded by the European Union

EU Strategy on Rare Diseases and Patients registries

EU Platform on Rare Disease Registration (EU RD Platform)

Copes with the fragmentation of RD patients data contained in hundreds of registries across Europe by releasing standards for interoperability:

- ✓ Common data elements (16)
- ✓ Pseudonymization tool
- ✓ European Directory of Registries/ Central metadata repository

European Commission Joint Research Centre

SPIDER

ERDRI

European Reference Networks registries

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES
Share. Care. Cure.

European Reference Networks

European Commission | Health

24 ERNs Central Registries following the standards defined by the EU RD Platform:

- ✓ Build
- ✓ Upgrade
- ✓ Link

ERICA & European Joint Program on Rare Diseases (EJP_RD)

ERICA
European Rare Disease Research Coordination and Support Action

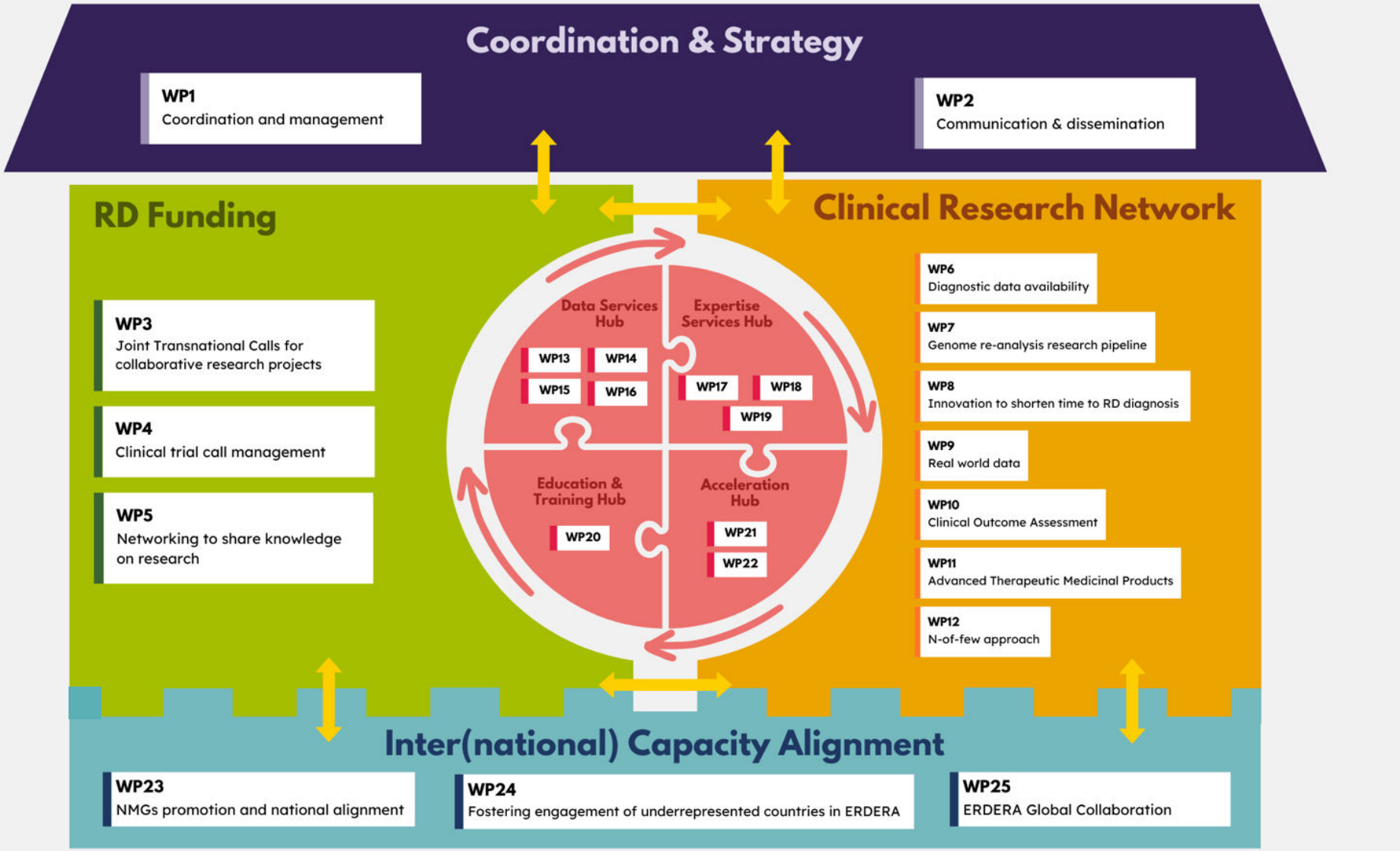
ERDERA
European Rare Diseases Research Alliance

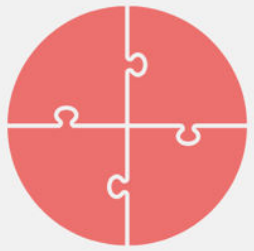
24 ERNs Central Registries:

- ✓ Domain specific Common Data Elements
- ✓ Legal and Ethics issues & Informed consent
- ✓ FAIR Principles

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

ERDERA
European Rare Diseases Research Alliance





WP13
Rare Diseases-Virtual Platform (RD-VP): Finding and accessing the data ecosystem

WP14
Data readiness services

WP15
Data sharing and analysis services

WP16
Knowledge bases and ontologies for RD research

WP17
Mentoring and consultancy

WP18
Regulatory support service

WP19
Methodological Support

WP20
Education and training in rare diseases research

WP21
Technology accelerator

WP22
Public-Private Collaboration Accelerator

ERDERA
European Rare Diseases Research Alliance

CLINICAL RESEARCH NETWORK _ Outcome research

Real World Data

- **Task 9.1 –** Use of primary healthcare data (EHRs) for RD outcome research
- **Task 9.2 –** Use of population-based data for RD outcome research
- **Task 9.3 –** Integrating patient cohorts for natural history / standard-of-care reference studies
- **Task 9.4 –** Development of a blueprint and inventory of regulatory-grade natural history cohort data
- **Task 9.5 –** Disease progression modelling and prognostic biomarker research
- **Task 9.6 –** Development of a regulatory grade clinical trial simulation platform for rare diseases

T9.1: eUROGEN; EURO-NMD; CRANIO, EpiCare, EuroBloodNet; T9.2: EpiCare; ERNICA; MetabERN; T9.3: ITHACA, ERKNet, ERNs ENDO-BOND; T9.4: RND, EuroBloodNet, ERKNet, EYE; T9.5: EURO-NMD; T9.6: ERKnet; DDF

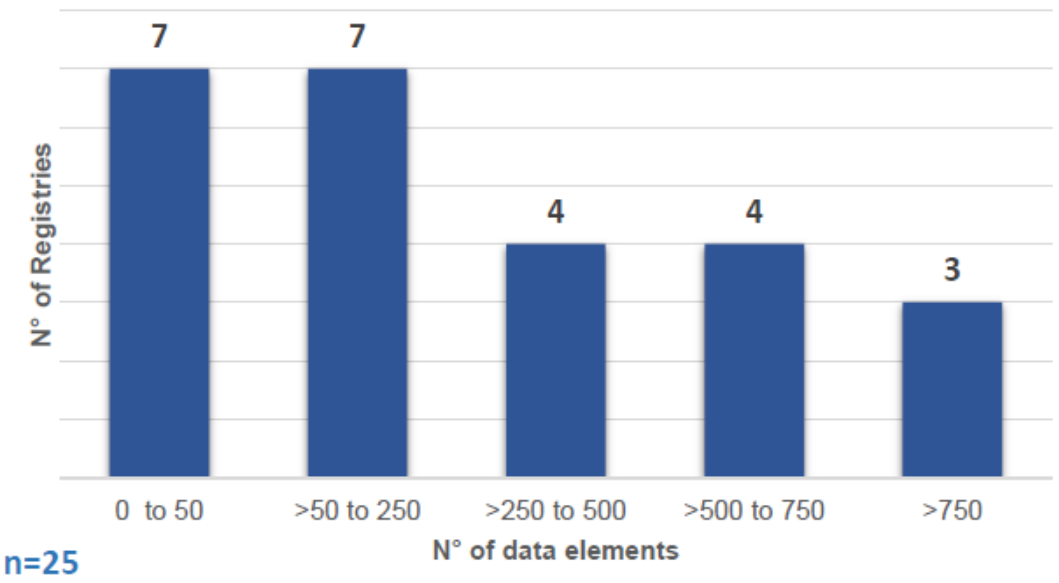
Clinical Outcome Assessment

- **Task 10.1** – Platform for regulatory-grade patient-centred COA development and validation
- **Task 10.2** – Development and Implementation of Clinical Outcome Assessment Tools
- **Task 10.3** – Unveiling the Hidden Burden: Estimating the Socioeconomic Impact of Rare Diseases for Informed Decision Making and Resource Allocation

T10.1: RND; ITHACA; mito-InterERN workgroup (EURO-NMD, RND, MetabERN, Eye, EpiCare); EuroBloodNet; CRANIO;
T10.2: EpiCare; EURO-NMD; ERKNet; EuroBloodNet; ERN-RND

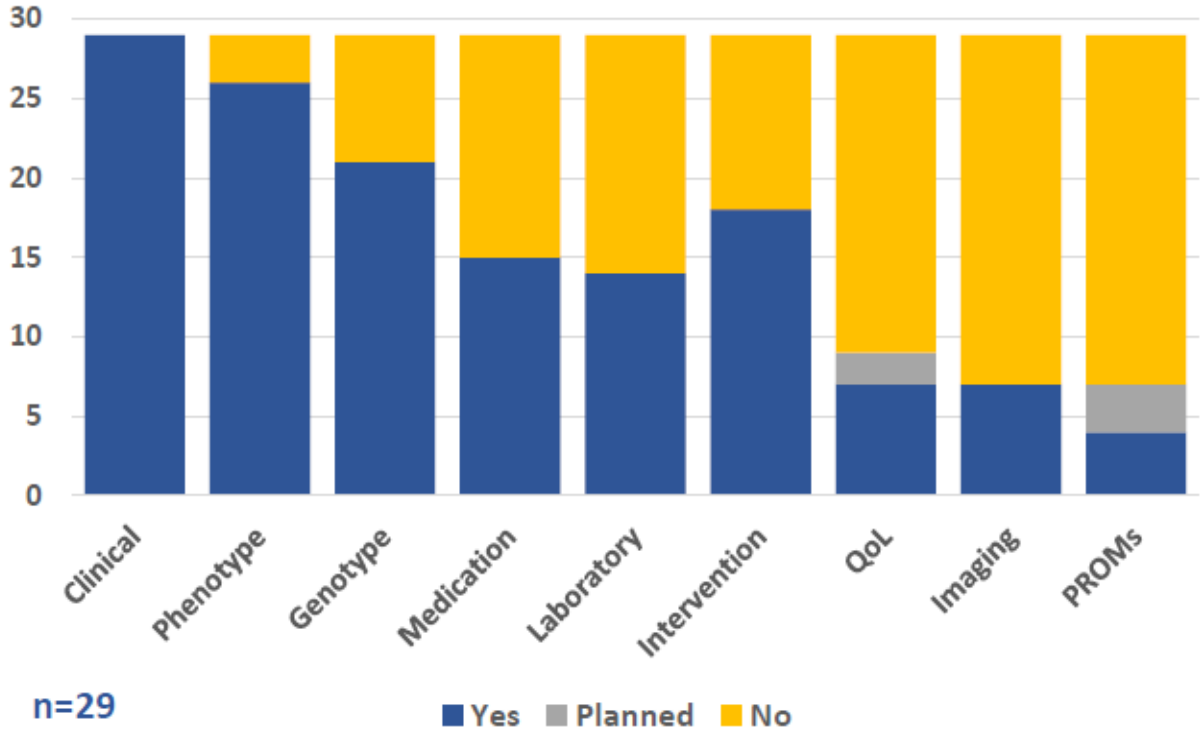
Information Collected by the 29 ERN Registries

Number of Data Elements



n=25

Type of Data Elements

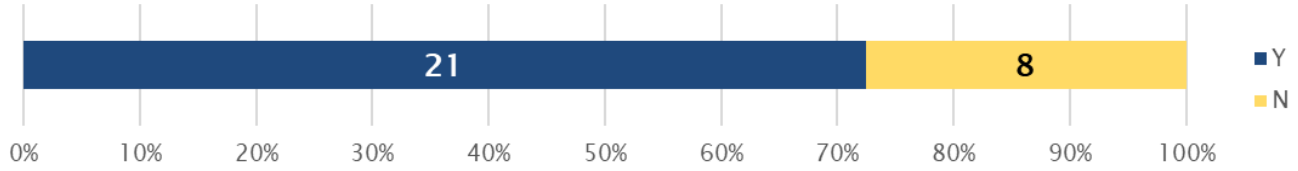


n=29

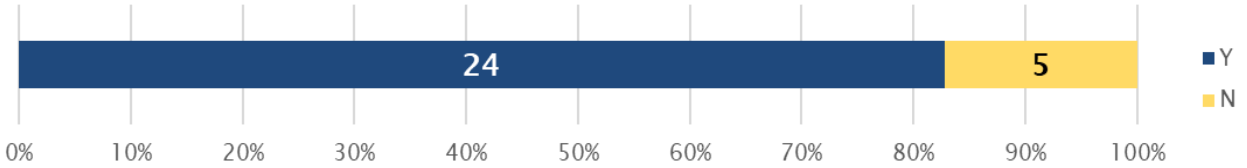
Slide provided by Franz Schaefer (ERNs Coordinators Group)

Quality Control Measures

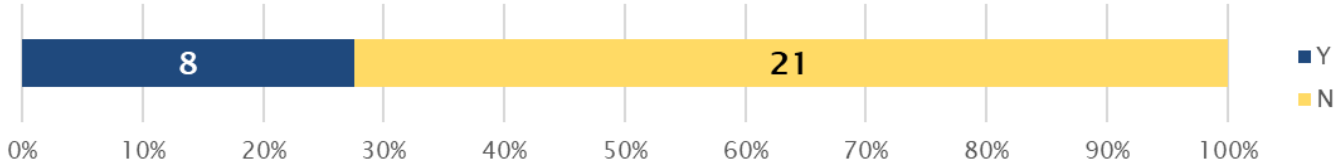
Online plausibility checks at data collection



Periodic offline data consistency checks, query system (5 six-monthly; 3 annually, 16 TBD)



Periodic trainings for staff in charge of data entry



Slide provided by Franz Schaefer (ERNs Coordinators Group)

European Rare Blood Disorders Platform - ENROL

ENROL Registry is conceived in the frame of the ERN-EuroBloodNet as an umbrella for both new and already existing registries on rare hematological disorders (RHD)

ENROL Registry avoids fragmentation of data by promoting the standards for patients registries' interoperability in line with the EU-RD-Platform for 4 main objectives:

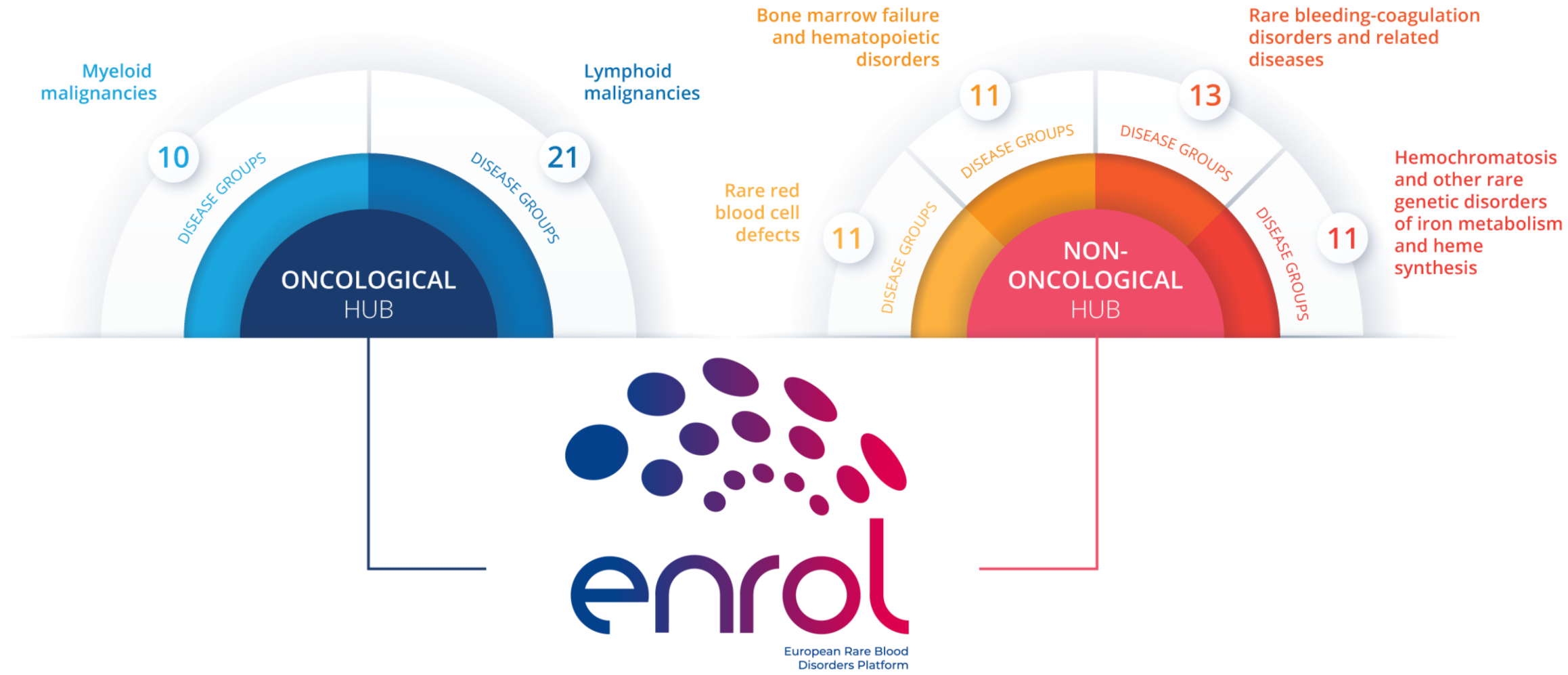
Facilitate epidemiological surveillance

Enhance health planning

Enable the identification of patients' cohorts

Promote research & innovative therapies

ENROL Registry



ENROL Collection of Pseudonymised Patient-level Data

European Platform on Rare Disease Registration (EU RD Platform)

Collection of Rare Diseases Common Data Set (RD-CDS) - 16 elements

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)	

EU RD PLATFORM

EUROPEAN COMMISSION
JOINT RESEARCH CENTRE
Directorate F – Health and Food
Unit F.1 – Disease Prevention

ENROL Collection of Pseudonymised Patient-level Data

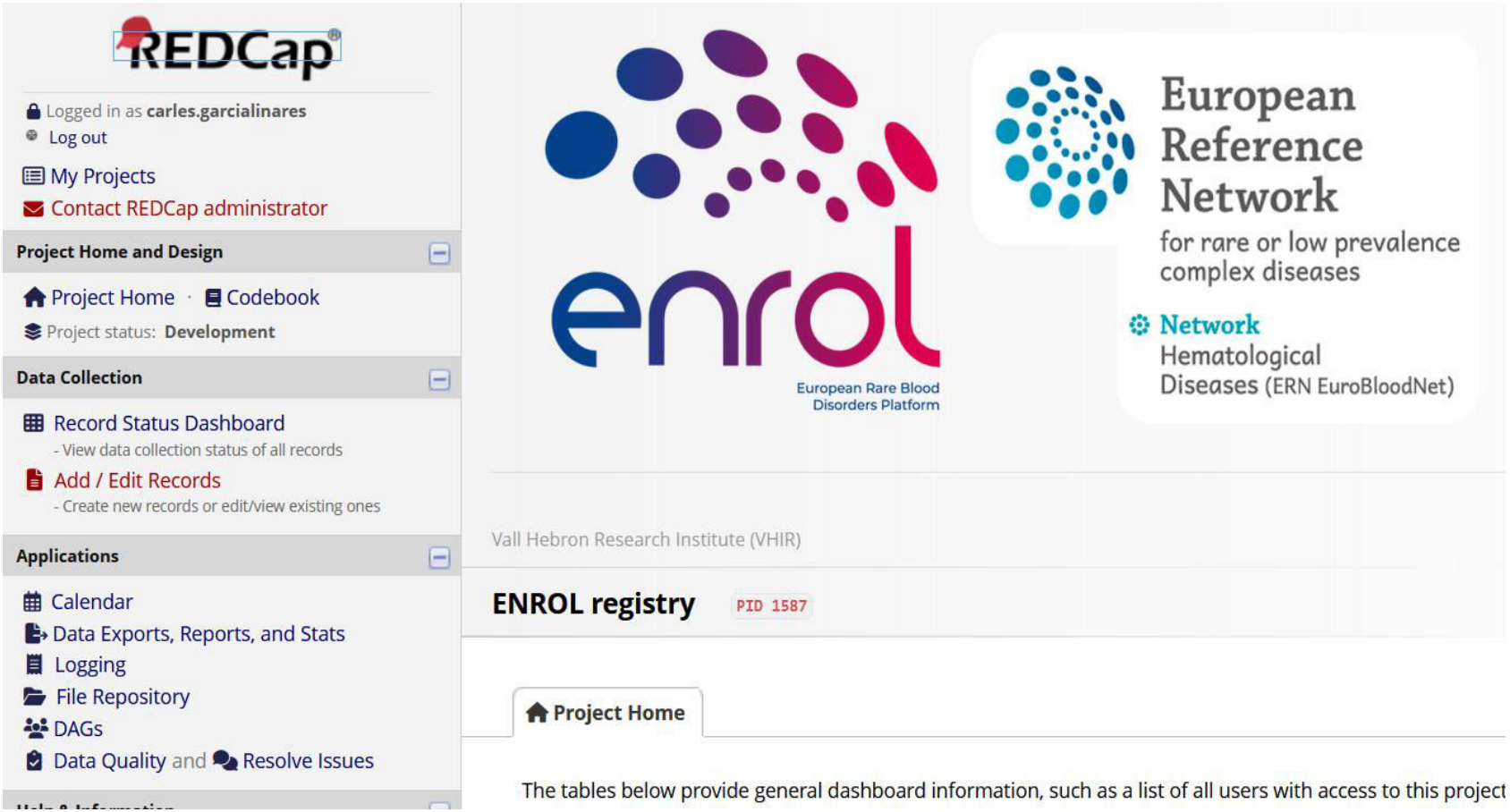
5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
	5.2.	Age at diagnosis	Age at which diagnosis was made	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
6 Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis 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/

EU RD PLATFORM

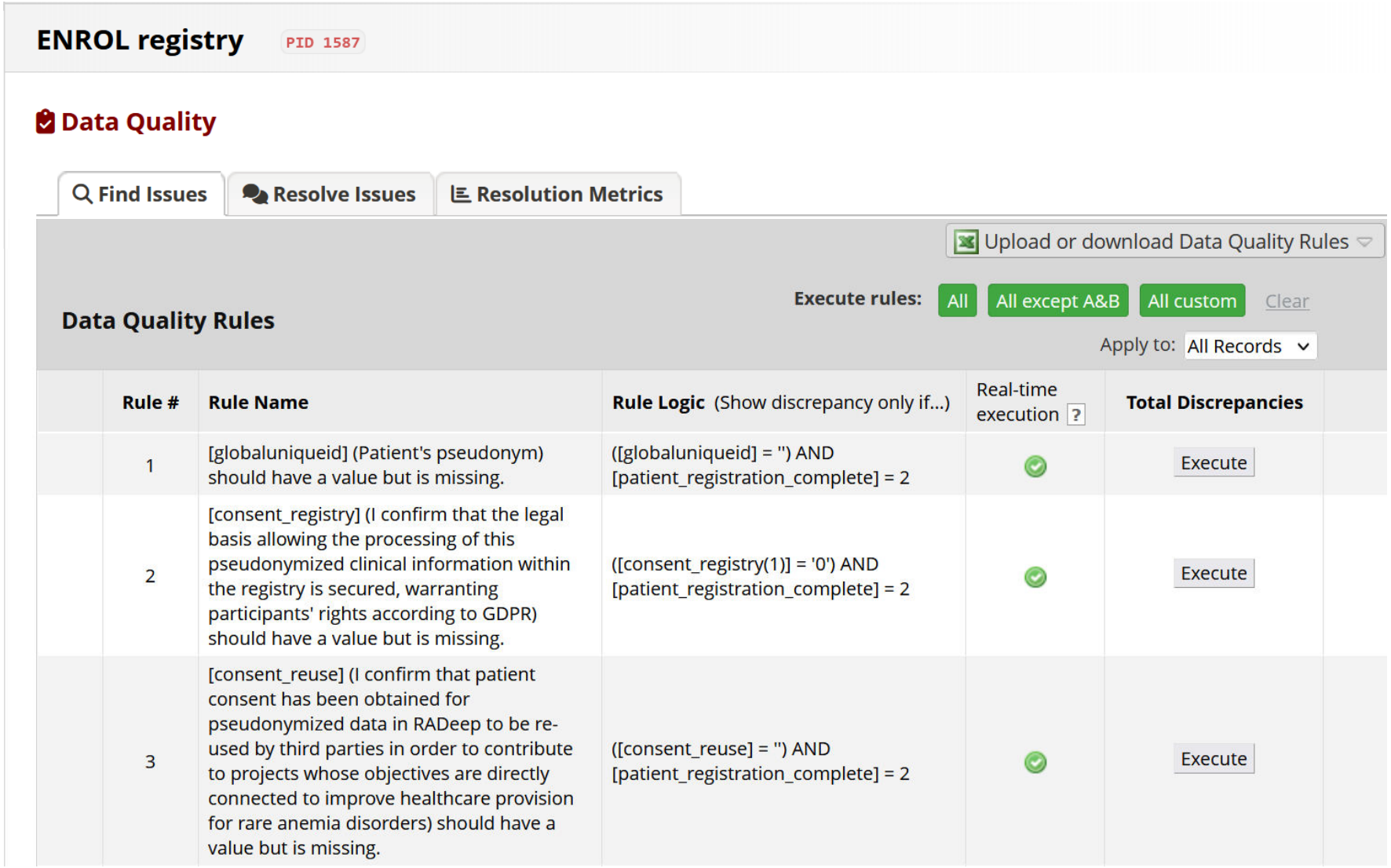
EUROPEAN COMMISSION
JOINT RESEARCH CENTRE
Directorate F – Health and Food
Unit F.1 – Disease Prevention

ENROL REDCap

- Secure web application for building and managing databases, supporting regulatory compliance (21 CFR Part 11, FISMA, HIPAA, GDPR).
- It enables rapid development/implementation of changes, with a user-friendly interface for data collection and analysis.
- It ensures data integrity and confidentiality through validation tools and role-based access control.



ENROL REDCap – Data Quality Module

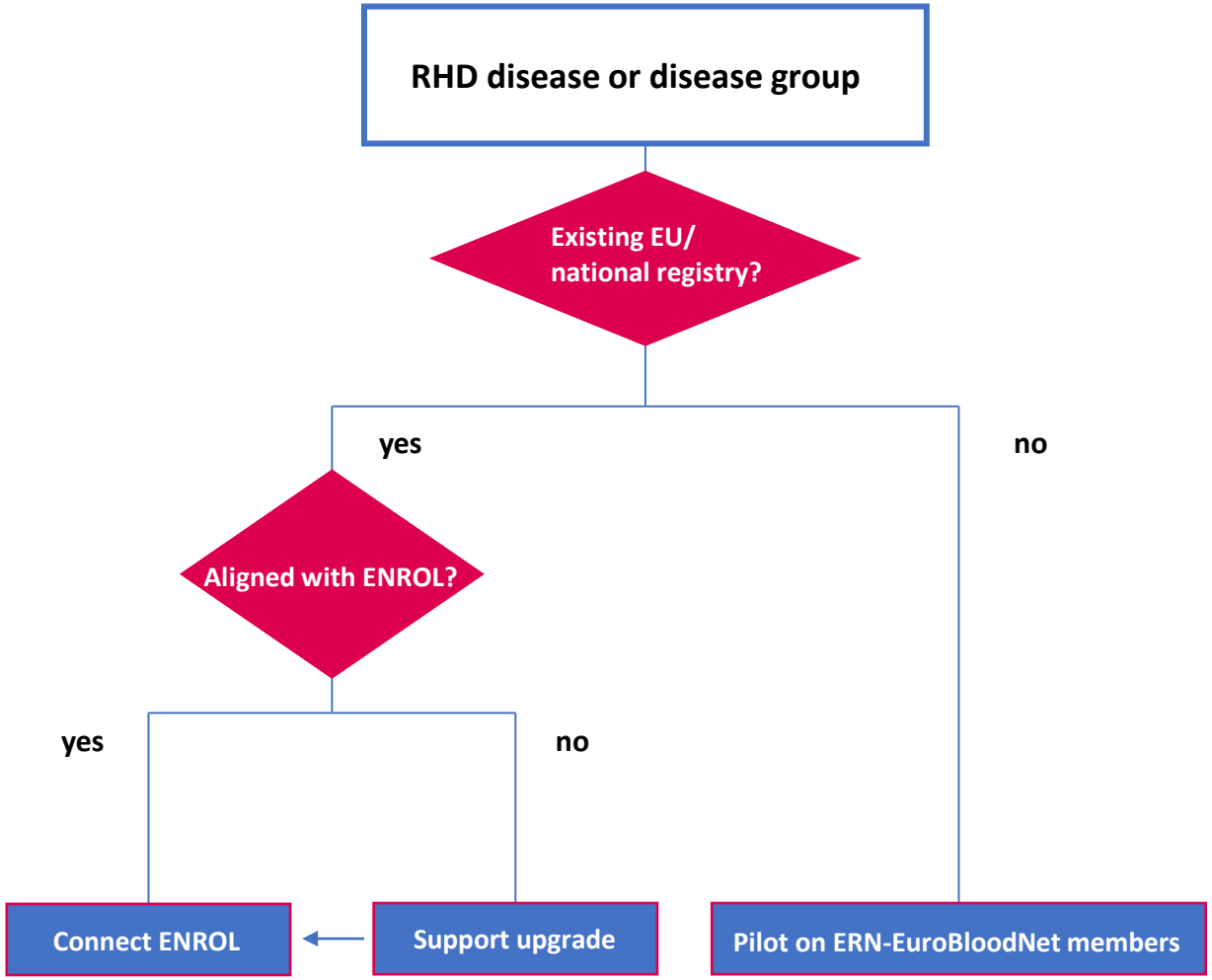


ENROL strategy for data collection

Strategy for data collection includes combination of data sources:

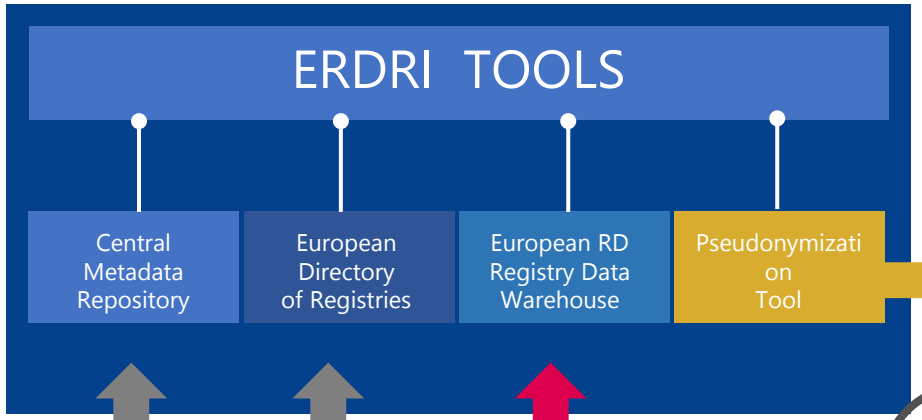
a) Individual sites:
   a) HCPs ERN Members
   b) HCPs non Members

b) Existing/New National/EU registries



ENROL Strategy for data collection

European Platform on Rare Diseases Registration



SPIDER

PSEUDONYMISATION TOOL GDPR COMPLIANT

Data providers generate the pseudonym.

Keep locally the link between pseudonym and personal data.

Aggregated Data
No personal Data

enrol
European Rare Blood Disorders Platform

DATA PROVIDER:

- **Individual sites**
- **Existing/New National/EU registries, eg:**

RADeep EUblast **TWIST**

Aggregated level data

Pseudonymised individual patient level data

Data transfer agreement

ENROL Data Transfer from RADeep

Country	Nº of patients
Belgium	887
Cyprus	166
Denmark	123
France	932
Greece	117
Italy	1100
Spain	987
The Netherlands	373
Total	**4.685**

ENROL Strategy for data collection

EU RD PLATFORM

PSEUDONYMISATION TOOL GDPR COMPLIANT

Data providers generate the pseudonym.

Keep locally the link between pseudonym and personal data.

enrol European Rare Blood Disorders Platform

enrol EpiBlood

Aggregated level data

Pseudonymised individual patient level data

Data transfer agreement

DATA PROVIDER:

- Individual sites
- Existing/New National/EU registries, eg:

RADeep EUblast TWIST

- Re-use and linkage of clinical data
- AI and data driven solutions

- Re-use and linkage of clinical data
- Patient Referral system for CTs

GENOMED4ALL

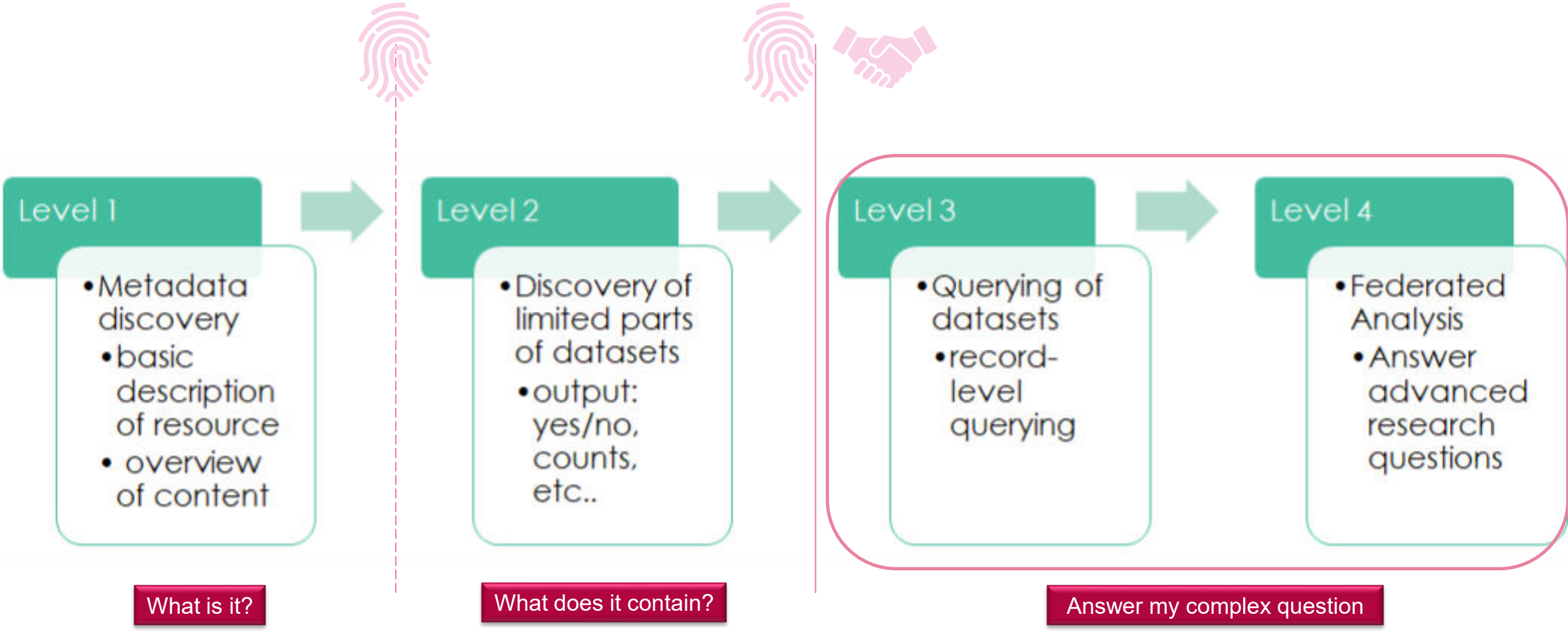
SYNTHEMA

IMPACT - AML
European project on Relapsed/Refractory Acute Myeloid Leukemia

Realise D
compRehensive mEthodological and operational Approach to cLinical trialS in ultra-rarE Diseases

The Four Levels of Connection to the Virtual Platform

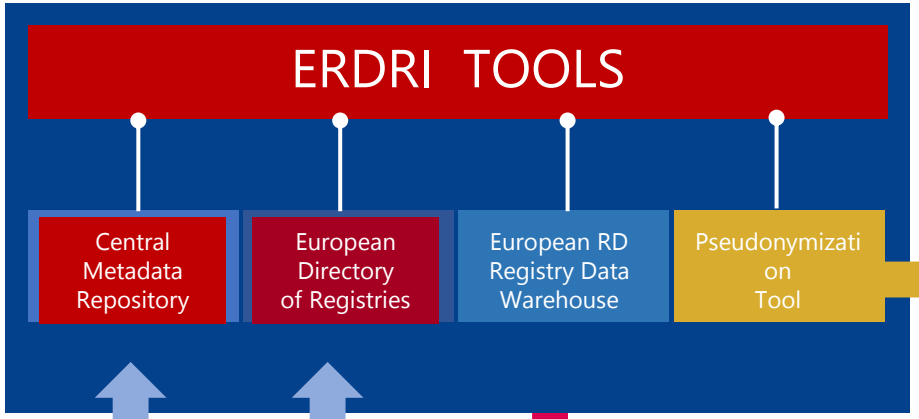
ERDERA
European Rare Diseases Research Alliance



Slide provided by Franz Schaefer (ERNs Coordinators Group)

ENROL Strategy for data collection

European Platform on Rare Diseases Registration



SPIDER

PSEUDONYMISATION TOOL GDPR COMPLIANT

Data providers generate the pseudonym. Keep locally the link between pseudonym and personal data.

DATA PROVIDER:

- **Individual sites**
- **Existing/New National/EU registries, eg:**

RADeep EUblast TWIST

Aggregated Data No personal Data

enrol EpiBlood

Aggregated level data

Pseudonymised individual patient level data

Data transfer agreement

enrol European Rare Blood Disorders Platform

ERN & ENROL Manuscript under development!
Thank you very much for your contributions

FINDABLE ACCESSIBLE INTER-OPERABLE REUSABLE

enrol

European Rare Blood Disorders Platform

Thank You