ERN-EuroBloodNet, ENROL Registry and data driven projects

Corrina McMahon

British Blood Transfusion Society (BBTS) red cell special interest group meeting

17th September

European Reference Networks (ERNs)

- Due to the small patient number and the often limited knowledge, rare diseases, affecting less than 1 in 2000 individuals, are the area in public health in which joint efforts among MS is most justified and crucial.

- **European Reference Networks (ERNs) involve healthcare providers across Europe**. They aim to facilitate discussion on complex or rare diseases and conditions that require highly specialised treatment, concentrated knowledge and resources.

- The first **24 ERNs** covering 24 different medical specialities were officially approved by the EC in December 2016 and started their activity in March 2017, one of them being **ERN-EuroBloodNet**.

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

European Commission | Health

ERN-EuroBloodNet

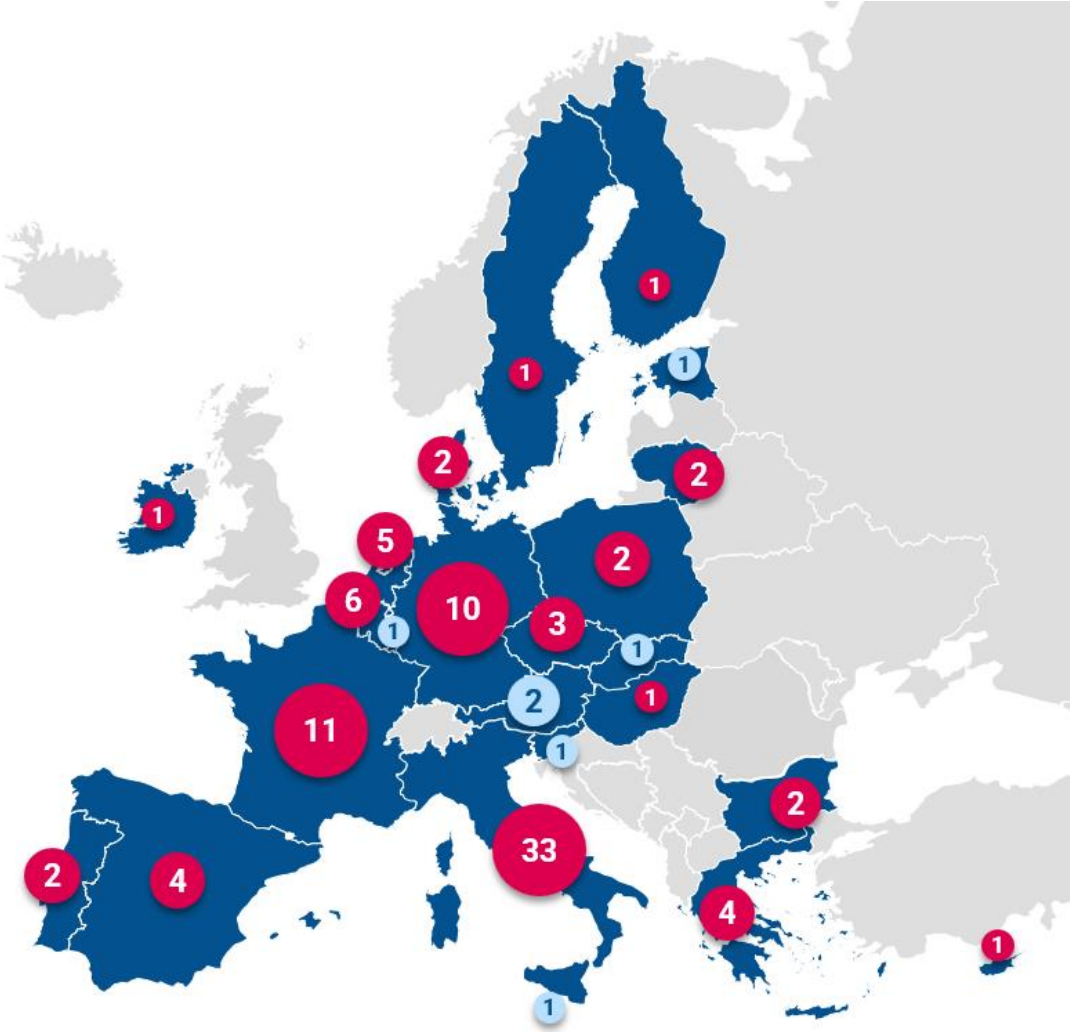
Collaborative network that brings together individuals and institutions committed to improving healthcare services in >450 Rare Hematological Diseases

Oncological Hub

- ☐ Myeloid Malignancies
- ☐ Lymphoid Malignancies

Non-Oncological Hub

- ☐ Rare red blood cell defects
- ☐ Bone marrow failure and haematopoietic disorders
- ☐ Rare bleeding-coagulation disorders and related diseases
- ☐ Haemochromatosis and other rare genetic disorders of iron metabolism and heme synthesis



97 Healthcare providers in
24 EU Member States

90 Members in 18 Member States

7 Affiliated Partners in 6 Member States

11 EPAGs

EURORDIS
RARE DISEASES EUROPE

ERN-EuroBloodNet Transversal Fields of Action

Cross Border Health

Enhancing equal access to highly specialized outcome-based healthcare and cross-border health for RHD in EU

Best practices

Promoting best practice in prevention, diagnosis and clinical care across EU by the development and update of ERN Clinical Practice Guidelines and other Clinical Decision Making tools

Continuing Medical Education

Spreading cutting-edge knowledge and facilitate continuing medical education in the field of RHDs

Telemedicine

Providing inter-professional consultation of RHD complex cases by sharing of expertise and safe exchange of clinical information through the Clinical Patient Management System (CPMS)

Clinical trials and research

Strengthening the use and re-use of RHD health data for the provision of best healthcare and for the promotion of research and innovation.

With high impact on patients' holistic approach, as for SCD

SCD

Centres of expertise

52 Members
3 Affiliated Partners
19 Member States

Best practices

ERN-EuroBloodNet on SCD
Comparative analysis of
National CGP & recommendations

Highly specialized procedures

EU–MS access to
bone marrow transplant &
transcranial Doppler

Training

International neurovascular
training course on SCD

EDU-Blood Academy
Topic on focus

Webinar programs for
health professionals
& patients

Patients, education & empowerment

4 editions of
SCD Patient Sessions

Preventive programmes

ERN-EuroBloodNet endorsed
recommendations on
newborn screening for SCD in Europe

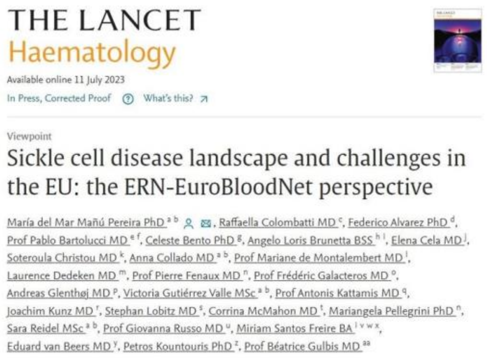
Patients' registries

Collaborative platform
on RBC & COVID-19

RADeep

Research

3 EU-funded Research Projects



THE LANCET
Haematology

Available online 11 July 2023

In Press, Corrected Proof

Viewpoint

Sickle cell disease landscape and challenges in the EU: the ERN-EuroBloodNet perspective

María del Mar Mañú Pereira PhD [a b], Raffaella Colombatti MD [c], Federico Alvarez PhD [d], Prof Pablo Bartolucci MD [e f], Celeste Bento PhD [g], Angelo Loris Brunetta BSS [h i], Elena Cela MD [j], Soteroula Christou MD [k], Anna Collado MD [a b], Prof Mariane de Montalembert MD [l], Laurence Dedeken MD [m], Prof Pierre Fenaux MD [n], Prof Frédéric Galacteros MD [o], Andreas Glenthøj MD [p], Victoria Gutiérrez Valle MSc [a b], Prof Antonis Kattamis MD [q], Joachim Kunz MD [r], Stephan Lobitz MD [s], Corrina McMahon MD [t], Mariangela Pellegrini PhD [n], Sara Reidel MSc [a b], Prof Giovanna Russo MD [u], Miriam Santos Freire BA [i v w x], Eduard van Beers MD [y], Petros Kountouris PhD [z], Prof Béatrice Gulbis MD [aa]

ENROL Registry & Data Driven research

EU Strategy on Rare Diseases and Patients registries

EU 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

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

SPIDER

European Commission

Joint Research Centre

European Reference Networks registries

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES
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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

EUROPEAN JOINT PROGRAMME RARE DISEASES

ERDERA
EUROPEAN RARE DISEASES RESEARCH ALLIANCE

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

24 ERNs Central Registries:

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

European Rare Blood Disorders Platform - ENROL

ERN-EuroBloodNet umbrella for both new and already existing registries on rare hematological disorders (RHD)

Aims to avoid fragmentation of data by promoting the standards for patients registries' interoperability in line with the EU-RD-Platform

ENROL's principle is to maximize public benefit from data on RHD with the only restriction needed to guarantee patient rights and confidentiality, in agreement with EU regulations for cross-border sharing of personal data

enrol
European Rare Blood
Disorders Platform

- ➡ possibility to share and pool data
- ➡ reach critical numbers
- ➡ analyse KPIs, perform clinical trials & research projects
- ➡ knowledge generation (evidence)
- ➡ **better healthcare for RHDs patients**

ENROL Consortium

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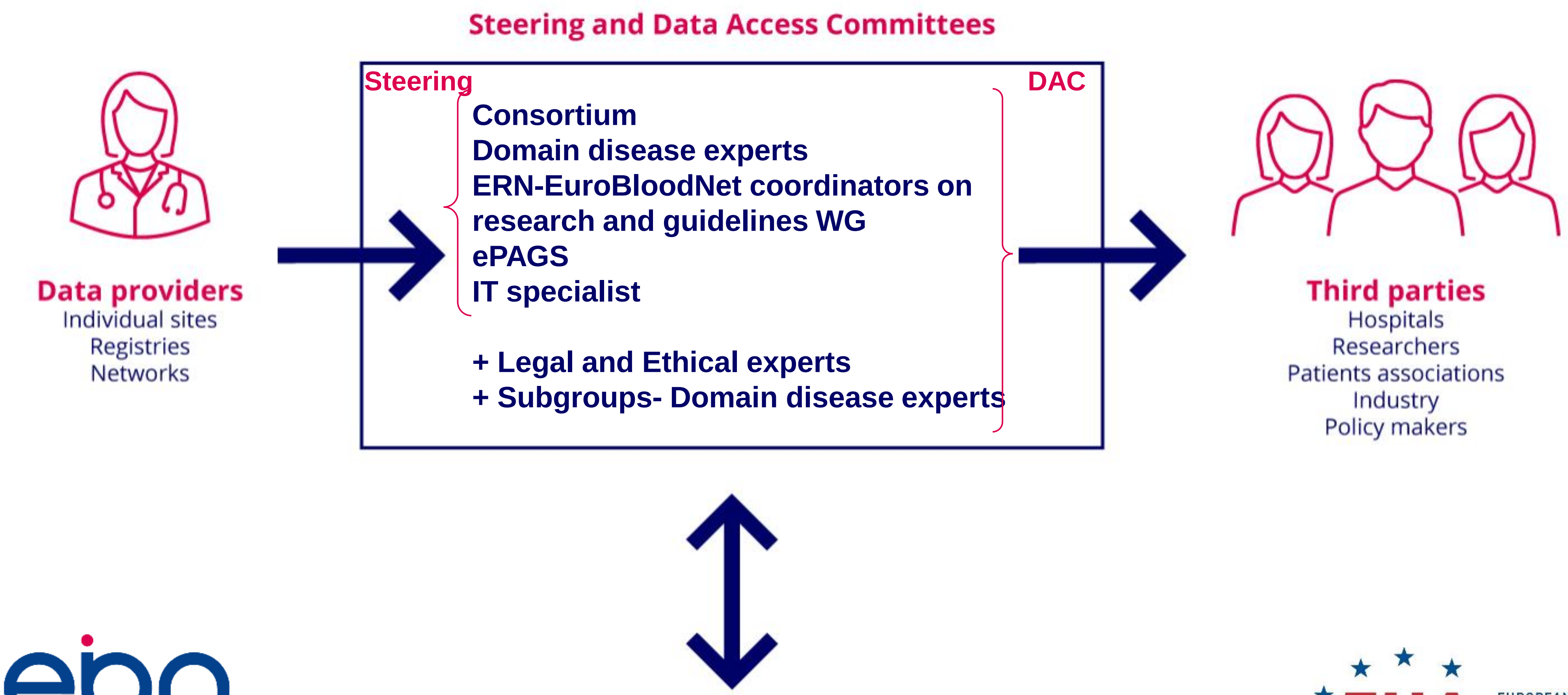
ENROL is endorsed by the European Hematology Association

Governance



Steering and Data Access Committees

Steering

DAC

Consortium
Domain disease experts
ERN-EuroBloodNet coordinators on research and guidelines WG
ePAGS
IT specialist

+ Legal and Ethical experts
+ Subgroups- Domain disease experts

Data providers
Individual sites
Registries
Networks

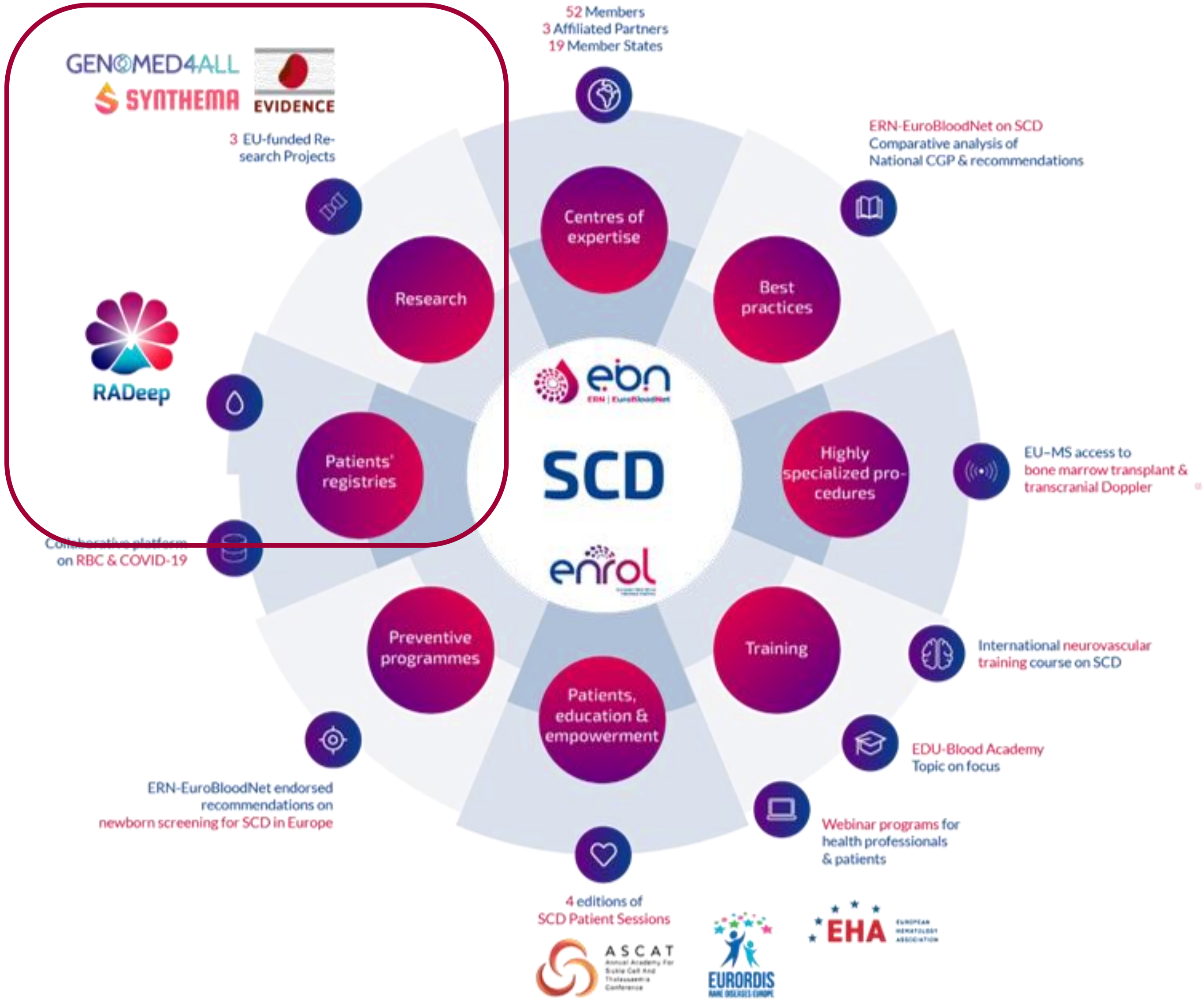
Third parties
Hospitals
Researchers
Patients associations
Industry
Policy makers

Involvement of stakeholders for Long-term Sustainability

ERN | EuroBloodNet

EUROPEAN HEMATOLOGY ASSOCIATION

Sickle cell Disease – ERN actions on patients registries and advancing research



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GENOMED4ALL
SYNTHEMA
EVIDENCE

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RADeep

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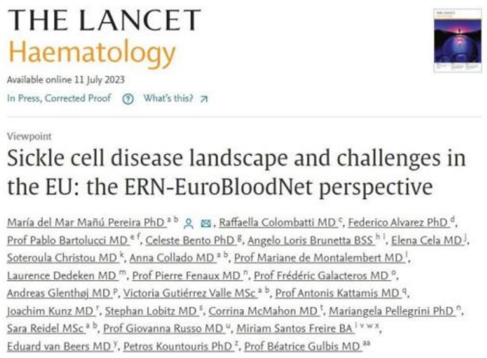

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In Press, Corrected Proof ⓘ What's this?

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Rare Anaemia Disorders European Epidemiological Platform (RADeep)

- To enable epidemiological and health burden surveillance of RADs in the EU to improve healthcare planning
- To enable translational and clinical research by collecting enough amount of high quality real world data to generate real world evidence for identification of reliable biomarkers for:
  - Disease progression
  - Prognosis
  - Response to treatments

➡ possibility to share and pool data

➡ reach critical numbers

➡ perform clinical trials, research projects

➡ knowledge generation (evidence)

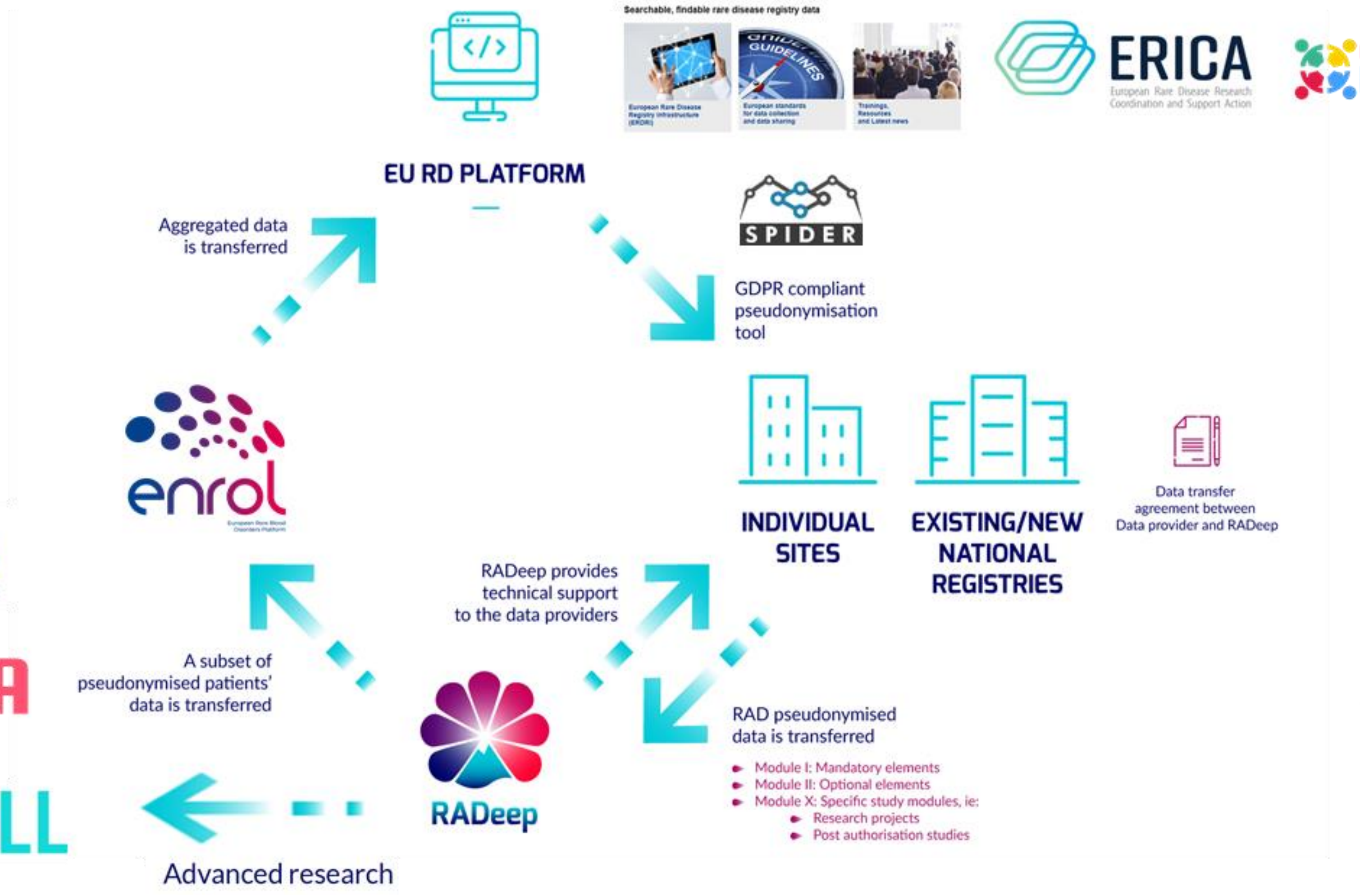
➡ **better healthcare for RADs patients**

RADs involve heterogeneous conditions grouped into the following groups:

- **Sickle cell disease and related diseases**
- **Thalassaemia and related diseases**
- **Pyruvate Kinase Deficiency and other red blood cell enzyme defects**
- **Red blood cell membrane defects**
- **Constitutional dyserythropoietic anaemias and rare constitutional aplastic anaemias**
- **Sideroblastic anaemias and constitutional anaemias due to iron metabolism disorders**

Data driven research

- Linking RWD sources: clinical (EHRs), research (Genomics, Metabolomics, functional studies) and patient self-reported data (PROMs, Health forms, socio-economic) to advance research in SCD, THAL and other RADs.



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ERICA

European Rare Disease Research Coordination and Support Action

EUROPEAN JOINT PROGRAMME RARE DISEASES

EU RD PLATFORM

SPIDER

Aggregated data is transferred

GDPR compliant pseudonymisation tool

INDIVIDUAL SITES

EXISTING/NEW NATIONAL REGISTRIES

Data transfer agreement between Data provider and RADeep

RADeep provides technical support to the data providers

A subset of pseudonymised patients' data is transferred

RAD pseudonymised data is transferred

- Module I: Mandatory elements
- Module II: Optional elements
- Module X: Specific study modules, ie:
    - Research projects
    - Post authorisation studies

enrol

RADeep

Advanced research

Data driven AI models for personalized medicine and synthetic data generation in hematology

ERDERA

EUROPEAN RARE DISEASES RESEARCH ALLIANCE

SYNTHEMA

GENOMED4ALL

ebn

ERN | EuroBloodNet

Take home messages

- On the basis of the strategy of the European Commission and EU Member States on rare diseases, the ERN-EuroBloodNet, ENROL, and RADeep registries have been able to implement the useful and necessary tools to map expertise and best practices in SCD, support and train experts, educate and empower patients, and make registries interoperable while safeguarding patient rights.

- ERN-EuroBloodNet has undoubtedly transformed SCD into a prioritized disease in the EU. By establishing a framework for action, disparities in healthcare in the EU have been reduced.

- Better integration of these actions into national health care systems is a crucial next step. It is addressed through the Joint Action on Integration of ERNs into National Healthcare Systems (JARDIN), which aims to integrate ERNs at the national level and, for research and innovation, through to European Rare Diseases Research Alliance (ERDERA).

THANK YOU!

ERN | EuroBloodNet

www.eurobloodnet.eu

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eurobloodnet-european-reference-network-on-rare-hematological-diseases

Eurobloodnet - European Reference Network on Rare Hematological Diseases

ERN-EuroBloodNet's EDUcational Youtube channel

Co-funded by the European Union

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