



12th June 2026 - EHA Congress



Workforce development & capacity building for rare diseases

# ERN infrastructure on education for patients and professionals & best practices

**Domenico GIRELLI**

*Azienda Ospedaliera Universitaria Integrata (AOUI) Verona, Italy*

**Béatrice GULBIS**

*Hôpital Universitaire de Bruxelles – ULB, Brussels, Belgium*



European  
Reference  
Network

Hematological Diseases  
(ERN EuroBloodNet)

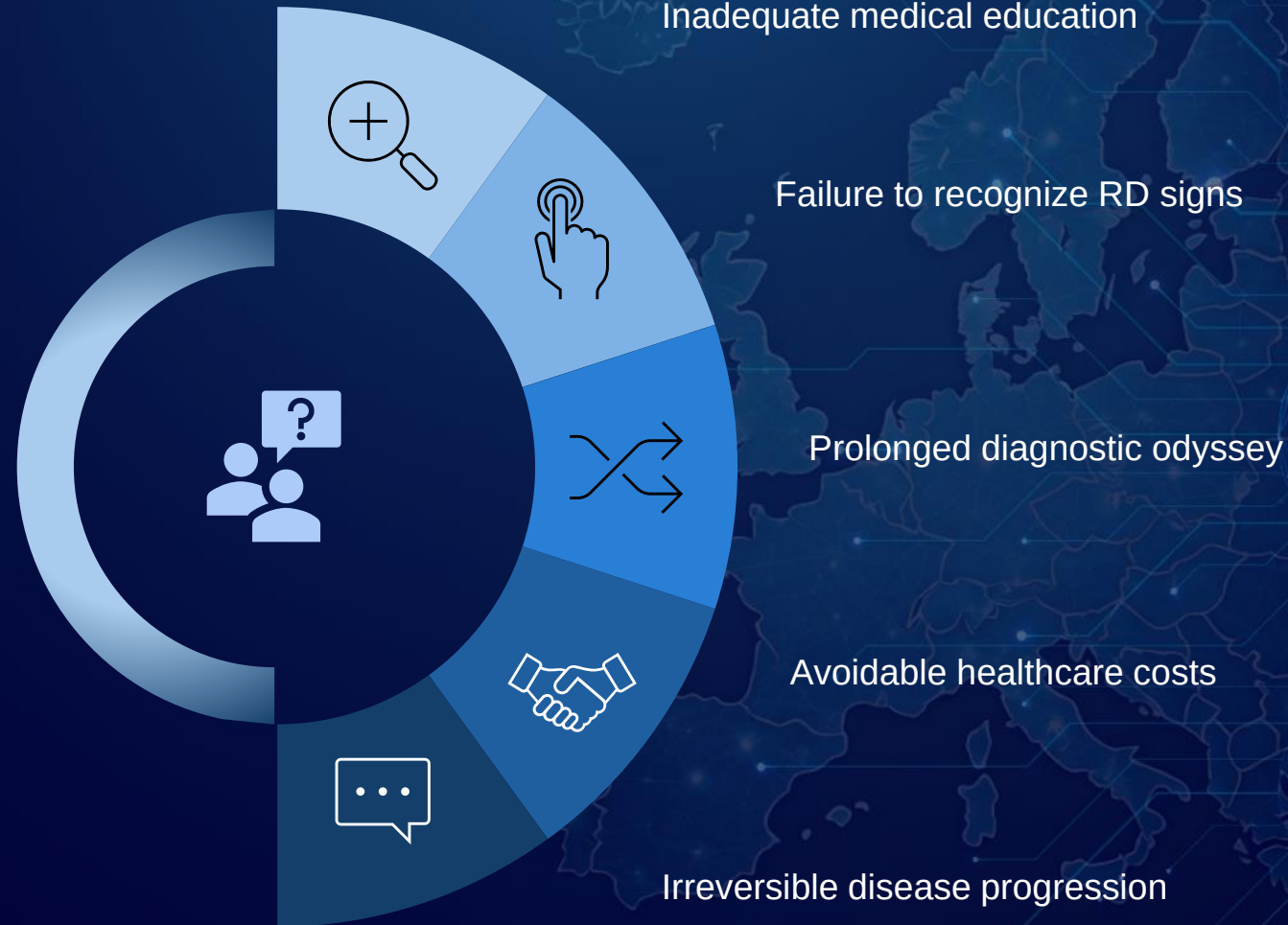


Funded by  
the European Union

# WHY EDUCATION MATTERS

If education reduces average diagnostic delay by just one year, the potential savings are massive, given 7.6× cost differentials across 30 million EU patients.

This reduction is achievable through systematic curricular integration, without additional training years.



# THE RARE DISEASES REALITY IN EUROPE

**65.9%** of medical students rate their rare disease knowledge as 'Poor' or 'Very Poor'.

**Only 6%** of known rare conditions have approved treatments.

*Sources: EURORDIS Rare Barometer 2024; Öztürk et al. 2025; EURORDIS/Draghi Report 2024*





# SESSION 1: EU ACTION PLAN FOR RARE DISEASES

## PRIORITY ACTION 2: WORKFORCE DEVELOPMENT

From European Policy to ERN-EuroBloodNet Action



### EUROPEAN PRIORITY FOR RARE DISEASES

**HLM4RARE WG2** : Strengthen workforce development and capacity building for rare diseases and formalise the ERN Academy



### CHALLENGES

- Rare disease **expertise remains unevenly distributed across Europe**, limiting equitable access to specialized knowledge and care.
- **Rare diseases** are not yet systematically embedded in medical, nursing, scientific and allied **health curricula**.
- Structured training for professionals, patients and families, **including digital and AI skills**, remains insufficiently recognized at EU level.



### ERN-EUROBLOODNET RESPONSE



#### Continuing Medical Education



#### Best Practices

#### KEY INITIATIVES

ITN-Marie Curie, Preceptorships  
EDIT-SCD, Edu programs in GENOMED4ALL & SYNTHEMA

## EU ACTION PLAN FOR RARE DISEASES



Governance  
& Action Plan



Workforce  
Development



Diagnostics &  
Early treatment



Clinical Trials



National  
Infrastructure



Real-World  
Evidence



Equitable access  
to therapies



Funding &  
Sustainability





# SESSION 1: EU ACTION PLAN FOR RARE DISEASES

## PRIORITY ACTION 2: WORKFORCE DEVELOPMENT

From European Policy to ERN-EuroBloodNet Action



### KEY TAKE-AWAYS from HLM Rare 2025

- **Skills gaps drive delayed diagnosis**, uneven care, weaker registries, fewer trials and slower research transaction
- ERN education already exists but is fragmented. This is why the **ERN Academy is needed** to unify and certify it
- **Talent retention** needs clear career paths and support
- **GPs** need rare-disease awareness and “red flags” **training**, as well as clear pathways
- **Digital and data competencies** are now essential, but clinicians must be appropriately trained guide, validate and implement these tools

## EU ACTION PLAN FOR RARE DISEASES





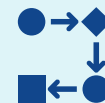
# SESSION 1: EU ACTION PLAN FOR RARE DISEASES

## PRIORITY ACTION 2: WORKFORCE DEVELOPMENT



### KEY TAKE-AWAYS from HLM Rare 2025

- **Skills gaps drive delayed diagnosis**, uneven care, weaker registries, fewer trials and slower research transaction
- ERN education already exists but is fragmented. This is why the **ERN Academy is needed** to unify and certify it
- **Talent retention** needs clear career paths and support
- **GPs** need rare-disease awareness and “red flags” **training**, as well as clear pathways
- **Digital and data competencies** are now essential, but clinicians must be appropriately trained guide, validate and implement these tools



### ACTION PLAN TOPICS Translated for ERN-EuroBloodNet

- **What role can we further play in reducing disparities in rare diseases expertise?**
- How can we attract and retain the next generation of specialists in rare haematological diseases?
- How can we better support the upskilling of general practitioners to improve early recognition and referral of people with rare hematological diseases?
- Digital and data competencies are now essential: what is currently needed to improve rare-disease specialists’ and patients’ skills and to better support their needs?

## EU ACTION PLAN FOR RARE DISEASES





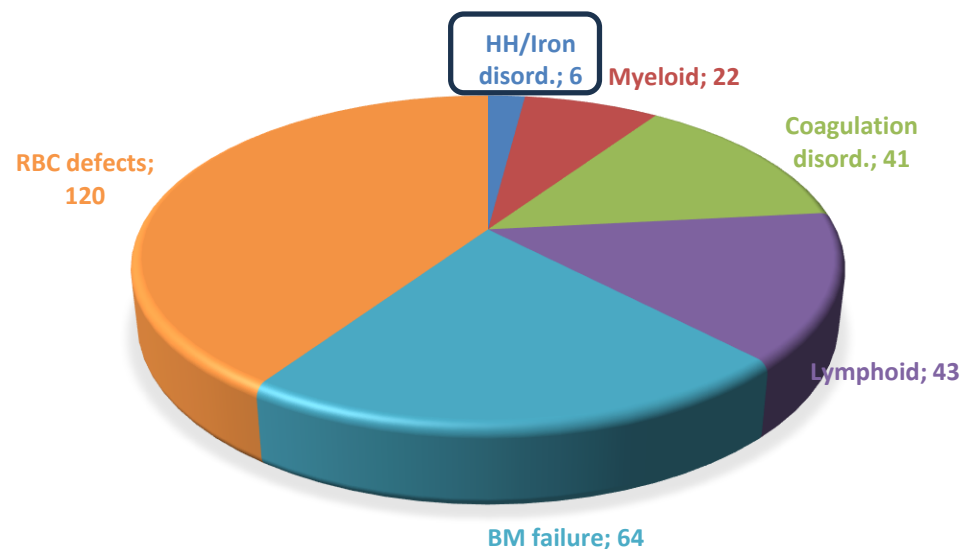


# Educational activities Oct 2023-Dec 2025

*Connecting expertise. Advancing knowledge. Improving lives across Europe.*



**CONTINUING  
MEDICAL EDUCATION**



+ For all rare haematological disorders; 41

**What strategies can be used to promote and harmonize education across all rare hematological disease subnetworks?**

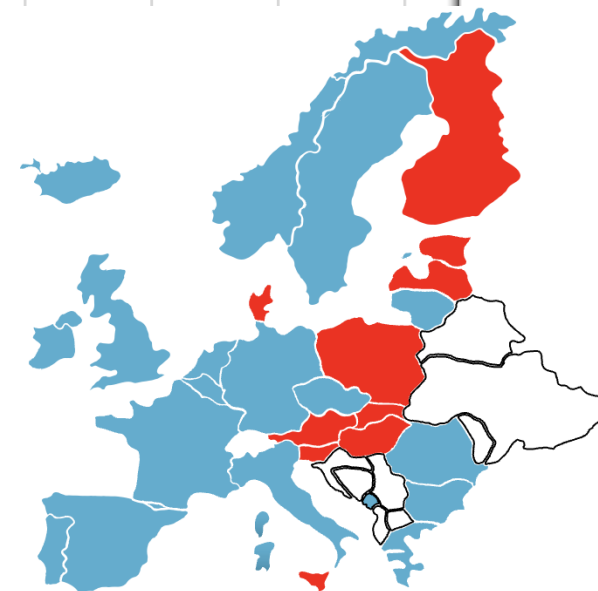
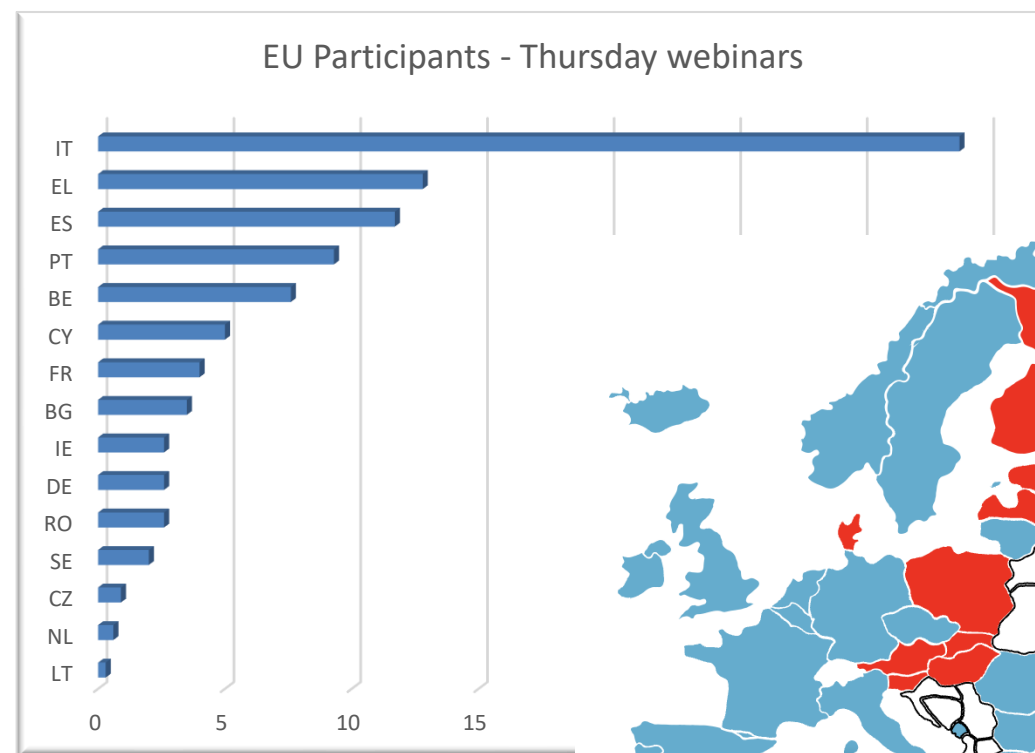
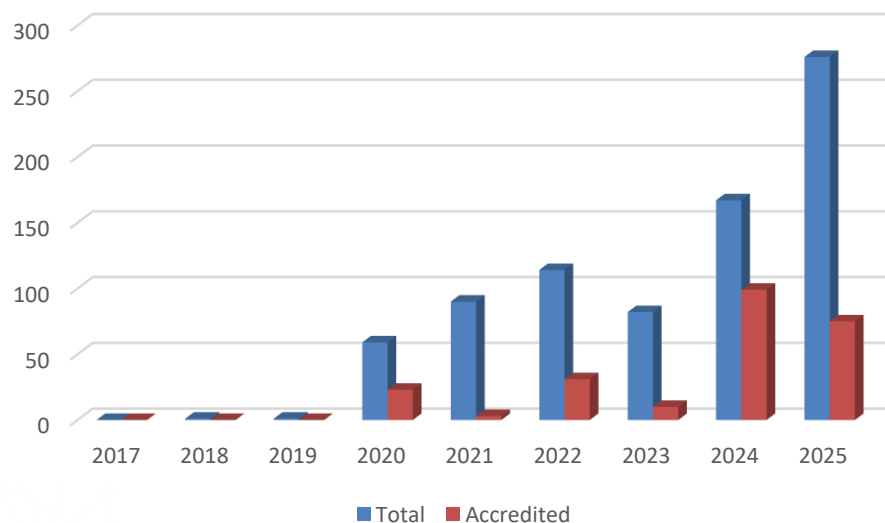


# Educational activities Oct 2023-Dec 2025

Connecting expertise. Advancing knowledge. Improving lives across Europe.



## CONTINUING MEDICAL EDUCATION



How to reach health care providers in East and North EU member states?





# Best practices 2017 - 2025

Connecting expertise. Advancing knowledge. Improving lives across Europe.



BEST PRACTICES



## Bridging Gaps In Sickle Cell Disease Care: ERN-EuroBloodNet's Comparative Analysis of National Recommendations Across Europe



Minke A.E. Rab<sup>1,2</sup>, Anna Collado Gimbert<sup>3,4</sup>, Alba M. Albert<sup>5</sup>, Amina Nardo Marino<sup>6</sup>, Andreas Glenthoj<sup>7</sup>, Anita Rijnveld<sup>8</sup>, Anna Vanderfaetle<sup>9</sup>, Habibi Anoshah<sup>10</sup>, Antonis Kattamis<sup>11</sup>, Bart Biemond<sup>12</sup>, Daniela Cuzzubbo<sup>13</sup>, Eduard J. van Beers<sup>14</sup>, Elena Cela<sup>15</sup>, Erfan Nur<sup>16</sup>, Giovanna Russo<sup>17</sup>, Hoiger Carlo<sup>18</sup>, Jaroslav Cermak<sup>19</sup>, Joachim Kunz<sup>20</sup>, Laura Tagliaferri<sup>21</sup>, Luca Malcovati<sup>22</sup>, Maddalena Casale<sup>23</sup>, Mariane de Montalembert<sup>24</sup>, Marta Morado<sup>25</sup>, Martin Colard<sup>26</sup>, Raffaella Colombatti<sup>27</sup>, Stephan Lobitz<sup>28</sup>, Maria del Mar Mañó Pereira<sup>29</sup>, Beatrice Gubie<sup>30</sup>  
<sup>1</sup>Erasmus University Medical Center Rotterdam, The Netherlands; <sup>2</sup>Hospital Universitario Vall d'Hebron, Spain; <sup>3</sup>Vall d'Hebron Institut de Recerca, Spain; <sup>4</sup>Copenhagen University Hospital - Rigshospitalet, Denmark; <sup>5</sup>CHU Saint-Pierre, Belgium; <sup>6</sup>Assistance Publique - Hôpitaux de Paris - Hôpital Henri Mondor, France; <sup>7</sup>Aghia Sophia Children's Hospital, Greece; <sup>8</sup>Amsterdam UMC, University of Amsterdam, The Netherlands; <sup>9</sup>Azienda Ospedaliero Universitaria Policlinico "G. Rodolico - San Marco", Italy; <sup>10</sup>University Medical Center Utrecht, The Netherlands; <sup>11</sup>Hospital General Universitario Gregorio Marañón, Spain; <sup>12</sup>Ulm University Medical Center, Germany; <sup>13</sup>Institute of Hematology and Blood Transfusion, Prague, Czechia; <sup>14</sup>Heidelberg University Hospital, Germany; <sup>15</sup>Foundation IRCCS Policlinico San Matteo, Italy; <sup>16</sup>Università degli Studi della Campania "Luigi Vanvitelli", Italy; <sup>17</sup>Assistance Publique - Hôpitaux de Paris - Hôpital Necker Enfants Malades - Université Paris-Cité, France; <sup>18</sup>Hospital Universitario La Paz, Spain; <sup>19</sup>Hôpital Erasme H.U.B., ULB Belgium; <sup>20</sup>Azienda Ospedale - Università di Padova, Padova, Italy; <sup>21</sup>Gemeinschaftsklinikum Mittelrhein gGmbH, Germany

## Pediatric Blood & Cancer

RESEARCH ARTICLE | [Open Access](#) |

### Limited access to transcranial Doppler screening and stroke prevention for children with sickle cell disease in Europe: Results of a multinational EuroBloodNet survey

[Vincenzo Voi](#), [Victoria Gutierrez-Valle](#), [Daniela Cuzzubbo](#), [Corrina McMahon](#), [Maddalena Casale](#), [Maria Del Mar Mañó Pereira](#), [Mirco D'Agnolo](#), [Baba P. D. Inusa](#), [Mariane de Montalembert](#), [Raffaella Colombatti](#) ✉ ... See fewer authors ^

First published: 10 July 2024 | <https://doi.org/10.1002/pbc.31190> | [VIEW METRICS](#)



European  
Reference  
Network

Hematological  
Diseases (ERN EuroBloodNet)



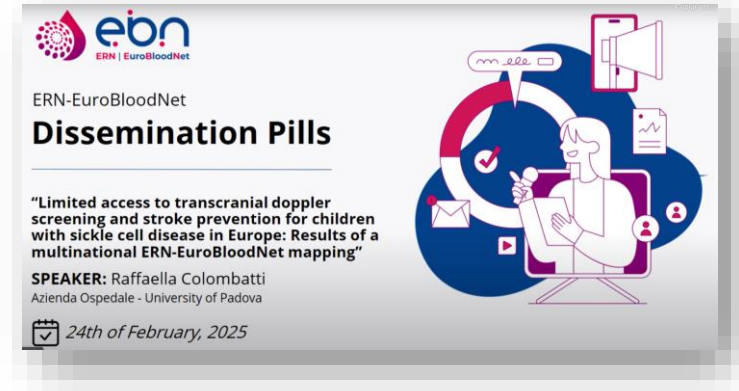
Funded by  
the European Union



# Best practices & education 2017 - 2025

Connecting expertise. Advancing knowledge. Improving lives across Europe.

Translate guidelines into education activities



Translate guidelines into clinical practice



European  
Reference  
Network

Hematological  
Diseases (ERN EuroBloodNet)



Funded by  
the European Union



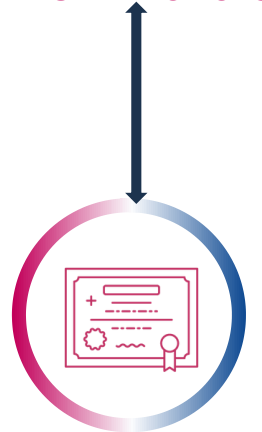
# Best practices 2017 - 2025

Connecting expertise. Advancing knowledge. Improving lives across Europe.

Collaborations +++



BEST PRACTICES



CONTINUING  
MEDICAL EDUCATION



Hematological  
Diseases (ERN EuroBloodNet)



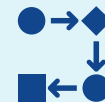
Funded by  
the European Union



# SESSION 1: EU ACTION PLAN FOR RARE DISEASES

## PRIORITY ACTION 2: WORKFORCE DEVELOPMENT

From European Policy to ERN-EuroBloodNet Action

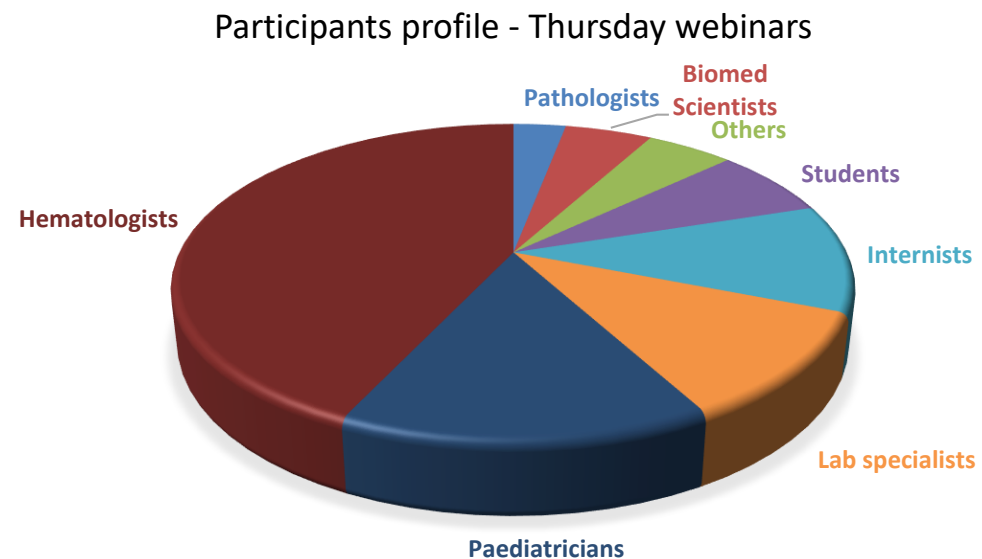
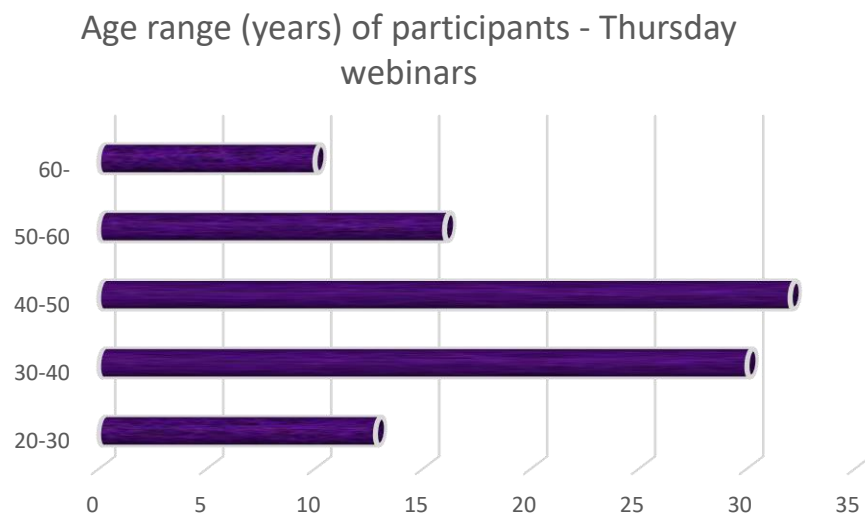
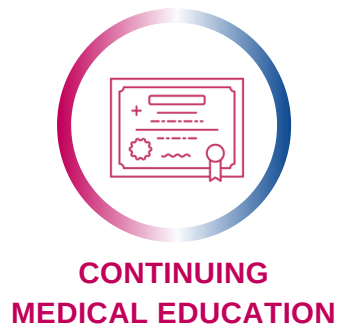


### ACTION PLAN TOPICS Translated for ERN-EuroBloodNet

- What role can we further play in reducing disparities in rare diseases expertise?
- **How can we attract and retain the next generation of specialists in rare haematological diseases?**
- How can we better support the upskilling of general practitioners to improve early recognition and referral of people with rare hematological diseases?
- Digital and data competencies are now essential: what is currently needed to improve rare-disease specialists' and patients' skills and to better support their needs?

## EU ACTION PLAN FOR RARE DISEASES





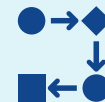
**How can the next generation of healthcare providers be effectively attracted and engaged?**



# SESSION 1: EU ACTION PLAN FOR RARE DISEASES

## PRIORITY ACTION 2: WORKFORCE DEVELOPMENT

From European Policy to ERN-EuroBloodNet Action



### ACTION PLAN TOPICS Translated for ERN-EuroBloodNet

- What role can we further play in reducing disparities in rare diseases expertise?
- How can we attract and retain the next generation of specialists in rare haematological diseases?
- **How can we better support the upskilling of general practitioners to improve early recognition and referral of people with rare hematological diseases?**
- Digital and data competencies are now essential: what is currently needed to improve rare-disease specialists' and patients' skills and to better support their needs?

## EU ACTION PLAN FOR RARE DISEASES







## Patients and HCPs collaboration and partnership



- ✓ The future ERN Academy cannot be built only for professionals.
- ✓ Most successful educational initiatives are those developed *with* patients, not only *for* patients:
  - Patients' sessions during meetings
  - Regular surveys listening to the patients' perspective





## Actions to promote more effective educational activities

- ✓ To engage GP: webinars or other resources in **local languages**; certification of attendance officially recognized as valid for Continuing Medical Education **(CME) credits**.
- ✓ A similar policy might help engage hospital doctors more widely in countries where expert centers for a given disease group are too few or absent (e.g., disorders of iron metabolism).
- ✓ Structured collaboration with other ERNs for overlapping disease groups (e.g. iron disorders and ERN-RARE-LIVER/ ERN-RND – rare neurological diseases).



# From successful projects to sustainable infrastructure

## WHAT SHOULD EUROPE BUILD NEXT?

1



### Formal recognition of the ERN Academy

Recognize education and training  
delivered within ERNs  
as a European standard of excellence.

2



### European certification pathways for rare disease expertise

Create harmonized, transparent  
and portable pathways to recognize  
rare disease expertise across Europe.

3



### Dedicated and protected funding for education and capacity building

Ensure stable, long-term investment  
in education and training within ERNs,  
linked to the **MFF 2028–2034**.

# Best practices & Education

*Connecting expertise. Advancing knowledge. Improving lives across Europe.*

## How can we improve communication and engagement with general practitioners?



### BEST PRACTICES

Topic on Focus: EU Health Policy for patients organizations



**FITHAD**<sup>®</sup>  
FONDAZIONE ITALIANA TALASSEMIA E DREPANOCITOSI  
LEONARDO GIAMBRONE



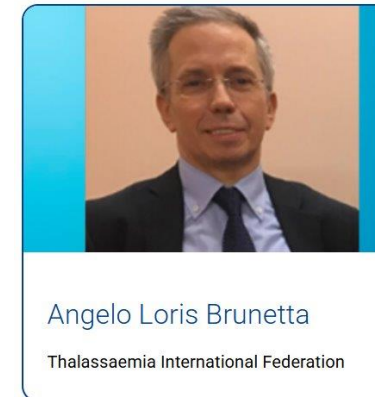
European  
Reference  
Network  
for rare or low prevalence  
complex diseases  
Network  
Hematological  
Diseases (ERN EuroBloodNet)

1. Spazio Europeo dei Dati Sanitari (EHDS)  
→ Registri, dati dei pazienti, aspetti etico-legali della governance  
Relatore: Iñigo de Miguel (esperto legale, collaboratore ERN-EuroBloodNet)
2. Regolamento sulle Sostanze di Origine Umana (SoHO)  
→ Sangue, plasma, sistemi di emovigilanza, raccolta e distribuzione  
Relatrice: Gabriella Peluso Cassese
- 3....



### CONTINUING MEDICAL EDUCATION

## ePAG



Upcoming: same webinar series in English  
for a wider audience



# ERN-EuroBloodNet actions on SCD awareness day

Every 19 June marks **World Sickle Cell Disease Day**, and ERN-EuroBloodNet complementary

De overgang van pediatrische naar volwassenenzorg bij...  
ERN-EuroBloodNet's EDU

Transition des soins pédiatriques aux soins pour...  
ERN-EuroBloodNet's EDU

Transição do cuidado pediátrico para o cuidado...  
ERN-EuroBloodNet's EDU

Transición del cuidado pediátrico al cuidado de...  
ERN-EuroBloodNet's EDU

Transizione dalle cure pediatriche alle cure per adult...  
ERN-EuroBloodNet's EDU

Der Übergang zur Betreuung Erwachsener mit...  
ERN-EuroBloodNet's EDU

Przejsięcie do opieki nad dorosłymi z chorobą...  
ERN-EuroBloodNet's EDU

Transitioning from Pediatric to Adult Care with Sickle Cell...  
ERN-EuroBloodNet's EDU

**2025.** Animated video co-created with patients about transitioning from pediatric to adult care (in 8 languages)

HOW TO IMPROVE OUTCOMES IN SICKLE CELL DISEASE

This survey should take no more than 10 minutes to complete!

THE LANCET Haematology

ASCAT

ANSWER THE SURVEY!

NB: Available translations can be found in the "Translations" section, right after the "Welcome!" page, by clicking "Next" at the bottom.

ebn

ebn  
ERN | EuroBloodNet

WORLD SICKLE CELL DISEASE DAY  
JUNE 19th

THE LANCET Haematology

ASCAT

Survey on how to improve outcomes in Sickle Cell Disease.

**2024.** Launch of a global survey to gather the voices of people living with SCD.

European Reference Network  
for rare or low prevalence complex diseases

Network Hematological Diseases (ERN EuroBloodNet)

**WORLD SICKLE CELL Awareness Day 2023**

COVERING MORE THAN 400 RARE AND COMPLEX HEMATOLOGICAL DISEASES, ERN-EUROBLOODNET OFFERS ITS ONLINE FACILITIES, SERVICES AND EXPERTISE TO SUPPORT ORPHAN BLOOD DISORDERS

ERN-EuroBloodNet's multi level actions on SCD aims at wide distribution of knowledge & cross border patients support

- 1 EUROPEAN PATIENTS REGISTRIES**
  - **ENROL** - European Rare Blood Disorders Platform
  - **BADep** - The Rare Anemia Disorders European Platform
  - **Collaborative Platform on RBODs and COVID-19 patients**
- 2 RESEARCH**
  - **GenoMed4All** - Genomics and Personalized Medicine for all through Artificial Intelligence in Hematological Diseases
  - **EVIDENCE** - Erythrocytes Properties and Viability in Dependence of Flow and Extra Cellular Environment
  - **INHERENT** - International Hemoglobinopathy Research Network
  - **EDITSCD** - Assessing efficacy and safety of genome EDITing approaches for SCD
  - **SYNTHEMA** - Synthetic generation of HEMAtological data over federated computing frameworks
- 3 CONTINUING MEDICAL EDUCATION**
  - **Webinars**
  - **Preceptorships**
  - **Training Course on Transcranial Doppler**
- 4 PATIENTS EMPOWERMENT**

**2023.** Poster gathering ERN-EuroBloodNet's multi-level actions in the field of Sickle Cell Disease (SCD)

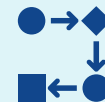




# SESSION 1: EU ACTION PLAN FOR RARE DISEASES

## PRIORITY ACTION 2: WORKFORCE DEVELOPMENT

From European Policy to ERN-EuroBloodNet Action



### ACTION PLAN TOPICS Translated for ERN-EuroBloodNet

- What role can we further play in reducing disparities in rare diseases expertise?
- How can we attract and retain the next generation of specialists in rare haematological diseases?
- How can we better support the upskilling of general practitioners to improve early recognition and referral of people with rare hematological diseases?
- **Digital and data competencies are now essential: what is currently needed to improve rare-disease specialists' and patients' skills and to better support their needs?**

## EU ACTION PLAN FOR RARE DISEASES



# AI: TRANSFORMING RARE DISEASE EDUCATION

## Adaptive learning

AI-personalized training platforms for rare disease curricula

## Diagnostic decision support

DDSS reducing income losses through earlier correct diagnosis



## Genomic analysis

ML/DL for variant interpretation and phenotype-genotype correlation

## Facial phenotyping

AI tools analyzing facial features to suggest genetic diagnoses

03

## NLP on Health Records

Identifying undiagnosed patients from clinical text in EHR systems





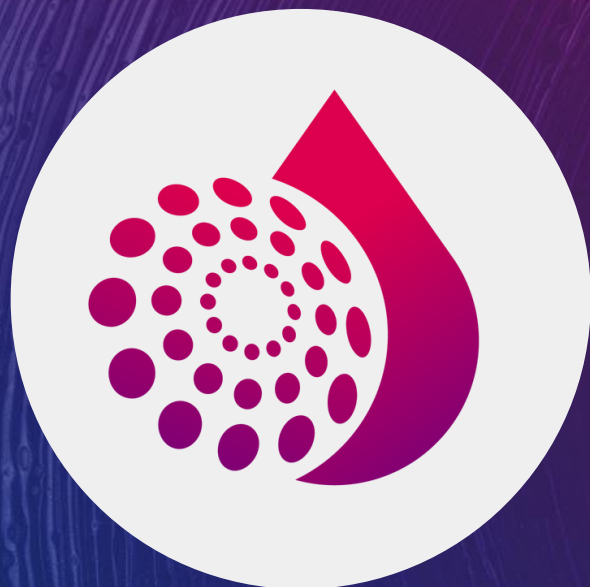
## Best practices & education

### Observations

- ◆ A robust infrastructure has been established.
- ◆ Education, a mechanism for translating knowledge (*best practices, research findings, and evidence-based approaches*) into practice.

### Perspectives

- ◆ Reducing disparities in expertise
  - Translate existing models to all rare hematological subnetworks
  - Pursue/expand collaborations
- ◆ Tackle next generations
- ◆ Include multidisciplinary team and patients
  - Focus on language barrier and national health care pathways
- ◆ AI
  - Use modern tools for training
  - Include AI into education programmes and update of guidelines



# THANK YOU!

Best practices & Education team



European  
Reference  
Network

Hematological  
Diseases (ERN EuroBloodNet)



Funded by  
the European Union

