



Sickle Cell Disease within the ERN-EuroBloodNet. Patients' Focus

Mariangela Pellegrini

7e Journée des associations de patients, 6 May 2024



The ERN-EuroBloodNet

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

Oncological Hub

- Myeloid Malignancies
- Lymphoid Malignancies



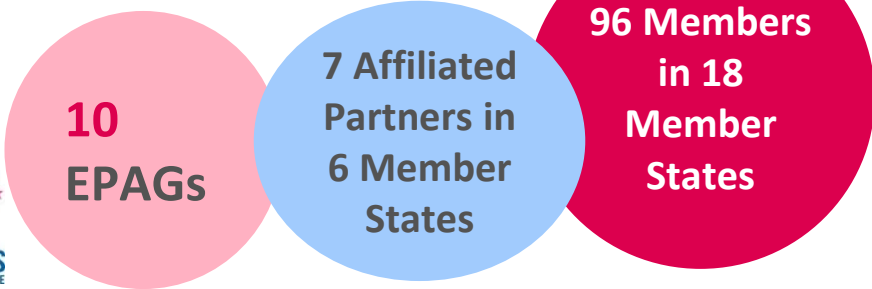
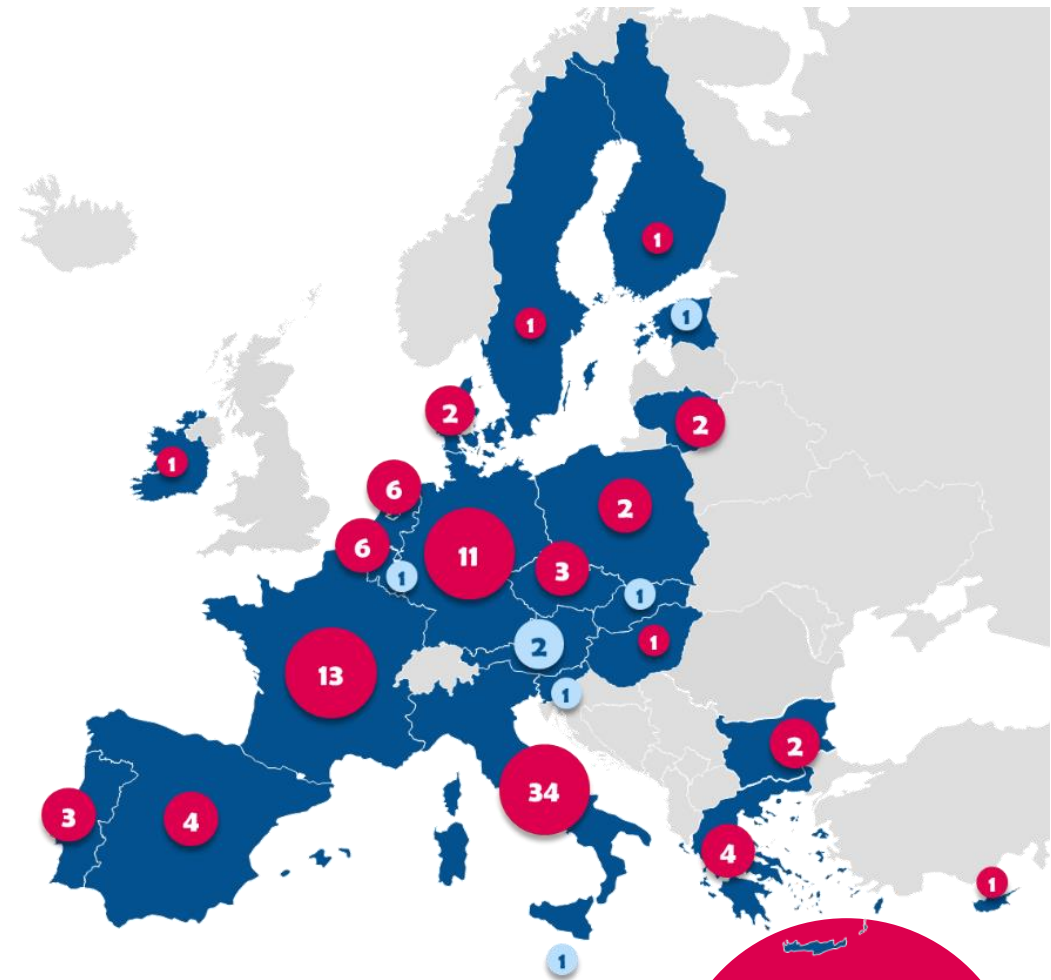
Pierre Fenaux



Béatrice Gulbis

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





Objectives & Transversal Fields of Action

ERN-EuroBloodNet concentrates multidisciplinary teams and highly specialized procedures across EU with the main goal to promote excellence for best health care in RHDs at the EU level by:



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.



European Patients Advocacy Groups (ePAGs) in the core of the ERN-EuroBloodNet

**ERN A
PATIENT-CENTERED
ENVIRONMENT**



ePAGs in the
GOVERNANCE:

- Participation to SSB, BON, Progress:
 - As member
 - As speaker
 - As chair & co-chair of a session



ERNs play a crucial role in benefiting patients by facilitating collaboration and information exchange among healthcare professionals across Europe.

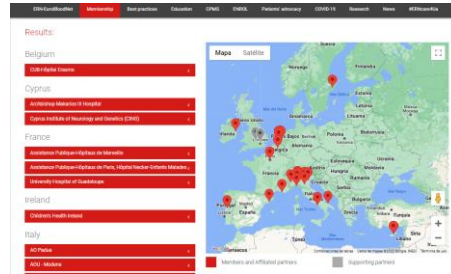
- ◆ **Access to Expertise**
- ◆ **Cross-Border Collaboration**
- ◆ **Standardized Care**
- ◆ **Data Sharing and Research**
- ◆ **Patient Empowerment**
- ◆ **Training & Education**
- ◆ **Cross-Specialty Collaboration**
- ◆ **Disease Cards: Expertise, activities and materials developed by the ERN searchable by disease groups**



SOME CHALLENGES: SICKLE CELL DISEASE

Challenge: Scattered expertise

REPOSITORY searchable by disease!



Challenge: Transcranial Doppler screening for stroke prevention

MAPPING: <30% patients followed as Guidelines standards



Challenge: Bone marrow transplant

CROSS BORDER driving force among 2 ERN members to have the highly specialized procedure in place



Challenge: Health professionals' education & Best practices

WEBINARS for health professionals

SCD New treatments revision

07/10/2021 17:00

Speakers: Mariane de Montalembert

Professionals

Subnetworks: Red blood cell defects

Disease Groups: Sickle cell disease and related diseases

Thursday Webinar - Sickle Cell Disease new treatments

Thursdays Webinars

Sickle Cell Disease New Treatments revision

Mariane de Montalembert

AP-HP Pitié-Salpêtrière

ERN EuroBloodNet Subnetwork Red Blood Cell

Paris, France

October 2021

Ver en YouTube



Challenge: Patients' education & Advocacy

EDUCATIONAL SESSIONS at international Congresses



WEBINARS for patients and associations

ERN-EuroBloodNet & EURORDIS' model supporting the establishment of EU Network of Patients Org.



Challenge: Registries & Research

RADeep European registry pre-dating ERN aligned with ENROL



Genomed4all: AI and OMICs led by ERN-EuroBloodNet members



1) Repository of CPGs and other CDMTs

116 International documents compiled - [69 searchable by Quality Domains*](#):

- Scope and purpose: Prevention, Diagnosis, Treatment
- Patients' involvement
- Rigour of development

*** 23 Documents compiled for RBC!**

Search guidelines

Disease group
Pyruvate Kinase Deficiency ▾

Scope and Purpose
|
All
1. Prevention
2. Diagnosis ✓
3. Treatment
4. Prevention and diagnosis
5. Diagnosis and treatment
6. Prevention, diagnosis, treatment

Rigour of development
B - Consensus based ▾

SEARCH

Guidelines by subnetworks

Bone marrow failure →

Myeloid malignancies →

Prevention and management of SCD patients: Recommendation endorsed by ERN-EuroBloodNet



Research Paper

Newborn screening for sickle cell disease in Europe: recommendations from a Pan-European Consensus Conference

Stephan Lobitz , Paul Telfer, Elena Cela, Bichr Allaf, Michael Angastiniotis, Carolina Backman Johansson, Catherine Badens, Celeste Bento, Marelle J. Bouva, Duran Canatan, Matthew Charlton, Cathy Coppinger, Yvonne Daniel, Marianne de Montalembert, Patrick Ducoroy, Elena Dulin, Ralph Fingerhut, Claudia Frömmel, Marina García-Morin, Béatrice Gulbis, Ute Holtkamp, Baba Inusa, John James, Marina Kleanthous, Jeannette Klein, Joachim B. Kunz, Lisa Langabeer, Claudine Lapouméroulie, Ana Marcao, José L. Marín Soria, Corrina McMahon, Kwaku Ohene-Frempong, Jean-Marc Périni, Frédéric B. Piel, Giovanna Russo, Laura Sainati, Markus Schmugge, Allison Streetly, Leon Tshilolo, Charles Turner, Donatella Venturelli, Laura Vilarinho, Rachel Yahyaoui, Jacques Elion, Raffaella Colombatti, with the endorsement of EuroBloodNet, the European Reference Network in Rare Haematological Diseases ... [See fewer authors](#) ^

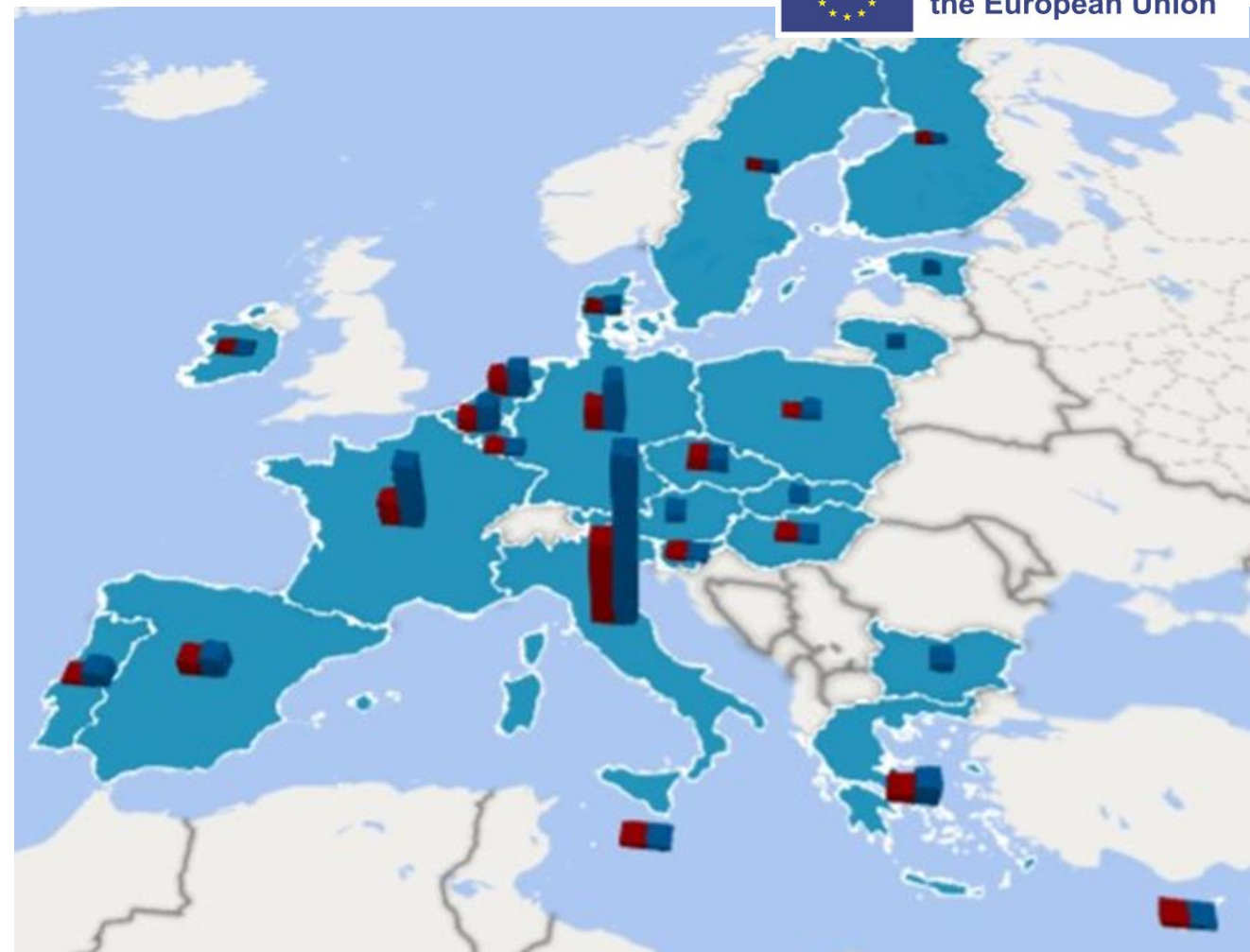
Next step: Assessment of the implementation, policy reports at national level



RED BLOOD CELL DEFECTS SUBNETWORK GEOGRAPHICAL COVERAGE

103 Members in 24 MSs
Croatia, Latvia,
Romania and Norway not
covered yet

53 Members in 19 MSs with
RBC defects expertise



- ERN Members
- RBC Members



SCD REPOSITORY OF EXPERTS



Home EuroBloodNet **Membership** Best practices Educati

Research

- Membership criteria
- Members and representatives
- Affiliated Partners
- Supporting Partners
- Search experts**
- Cross border health
- Supporting Partners

Search experts:

By disease

Type one disease *

SICKLE CELL DISEASE AND RELATED DISEASES

By subnetwork

<https://eurobloodnet.eu/members/expert/search>

Results:

Belgium

CUB-Hôpital Erasme

- Alina Ferster
- Benghiat Fleur Samantha
- Béatrice Gulbis

Cyprus

Archbishop Makarios III Hospital

France

Assistance Publique-Hôpitaux de Marseille

Assistance Publique-Hôpitaux de Paris, Hôpital Henri-Mondor

Assistance Publique-Hôpitaux de Paris, Hôpital Necker-Enfants Malades

CHU de Pointe-à-Pitre/Abymes

Germany

Charité Universitätsmedizin Berlin

Ulm University Medical Center (UUMC)

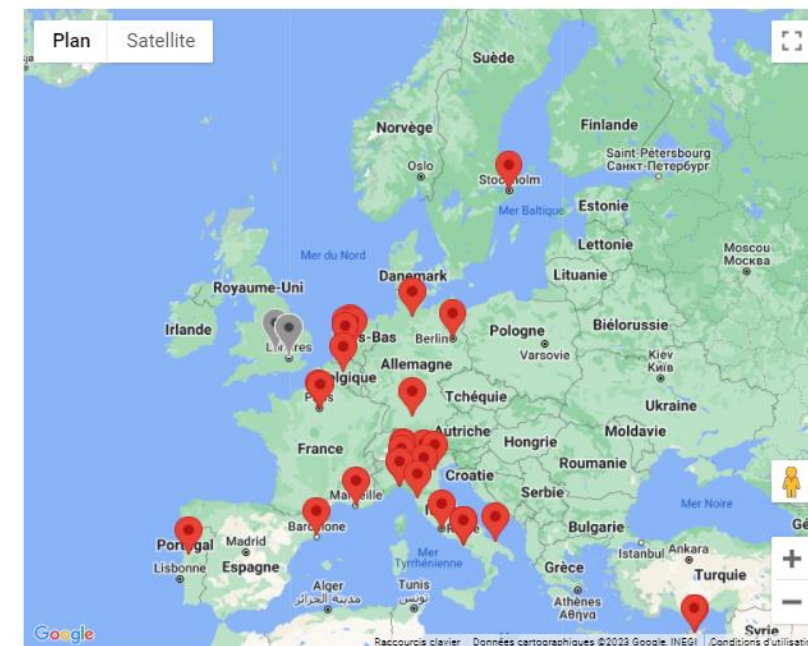
Universitätsklinikum Hamburg-Eppendorf

Italy

AO Padua

AORN A Cardarelli

AOU Consorziata polyclinic - Bari



Legend: Red pin = Members and Affiliated partners; Grey pin = Supporting partners

Diseases groups

Sickle cell disease and related diseases (SCD)

Repository of educational material produced in the ERN-EuroBloodNet focused on this disease group.

Reference Centers & Experts



Patients Organizations supporting group



Guidelines & other CDMTs



Education Health Professionals



Education Patients



Research & Publications



Ongoing projects



Diseases covered



External useful links



Education Patients

PAST WEBINARS

UPCOMING WEBINARS

VIDEOS

ONSITE TRAININGS

Past webinars

Webinar SCD and COVID-19

🕒 14/12/2020 16:00

Speakers: *Mariane de Montalembert, Noemi Roy*

Target: Patients

Subnetworks: Red blood cell defects

Disease Groups: Sickle cell disease and related diseases

New therapies for Sickle Cell Disease

🕒 31/01/2022 17:00

Speakers: *Malika Benkerrou*

Target: Patients

Subnetworks: Red blood cell defects

Disease Groups: Sickle cell disease and related diseases



FOCUS PATIENTS



ASCAT 2022

Patients representing different countries: France metropole and Overseas, Italy, Spain, Portugal, Ireland, Sweden, Germany, UK, Australia, Netherlands, Zambia



Facilitating the establishment of EU NET SCD

ERN-EuroBloodNet &
EURORDIS' **model** for
supporting the establishment
of a European Network of
Patient Organizations
for under-represented diseases





Facilitating the establishment of EU NET SCD



Co-funded by
the European Union

- ✓ A bottom-up umbrella network of national and local organizations of SCD patients' advocates
- ✓ Each Country is represented by two spokespersons. A spokesperson is a national representative that advocate on behalf of all the national SCD patients' organizations. The role is the mediation among the European Network of SCD Patients' Organisations and the national patients support groups.
- ✓ Among all spokespersons, a representative one will become ePAG for the ERN-EuroBloodNet.





Facilitating the establishment of EU NET SCD



Co-funded by
the European Union

First National Meeting in Milan, the 29th of March 2019



Patients organisations:

- 2 federal associations
- 4 local associations
- 1 forthcoming associations

3 EuroBloodNet representatives

- Raffaella Colombatti,
physician
- Loris Brunetta, ePAG
- Mariangela Pellegrini, ERN
Coordination Team





Facilitating the establishment of EU NET SCD



Co-funded by
the European Union

Second National Meeting in Bruxelles, the 19th of June 2019



Patients organisations :

- Federal associations
- Local associations
- Forthcoming association

2 représentants

d'EuroBloodNet :

- Béatrice Gulbis, physician
- Mariangela Pellegrini,
ERN Coordination Team





Facilitating the establishment of EU NET SCD



Co-funded by
the European Union

Third National Meeting, in Paris the 29th of June 2019



Patients :

- 1 Federal associations
- 14 associations
- 1 National RBC Network
- Eurordis
- ePAG ERN-NDM

4 représentants d'EuroBloodNet :

- Maryse Etienne Julan,
physician
- Mariangela Pellegrini, ERN
Coordination Team
- Ariane Weinman, Eurordis
- Sonia Pavan, filière MCGRE





Facilitating the establishment of EU NET SCD



Co-funded by
the European Union

Fourth National Meeting, in Madrid the 18th of February 2020 (Spain and Portugal)



Patients :

- 1 Spanish association
- 1 Portuguese association

4 représentants d'EuroBloodNet :

- Victoria Gutierrez, ERN Scientific Manager
- Mariangela Pellegrini, ERN Coordination Team
- Mar Manu Pereira, ERN-EuroBloodNet Scientific Director

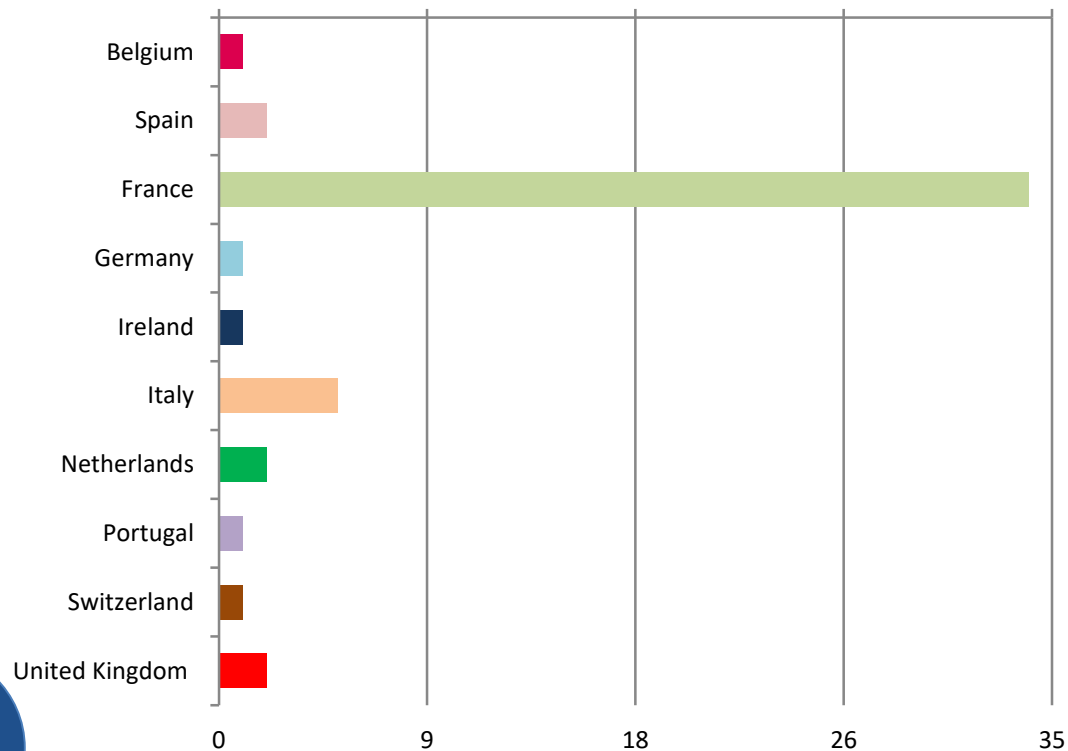




Facilitating the establishment of EU NET SCD

- A bottom-up umbrella network of national and local organizations of patients' advocates. Each Country is represented by two spokespersons.

Patients Organizations reached



12
European
countries
reached

50
Patients
organisations
reached

Number of POs per country reached by ERN-EuroBloodNet

✓ **Europe**
SCD Patients representatives reached are from: Spain, Portugal, Italy, France (France metropolitan and France Overseas), Cyprus, Belgium, Germany, Ireland, UK, Austria, Netherlands, Switzerland.

✓ **Outside Europe**
GASCDO, the Global Alliance of SCD organisations, Australian Sickle Cell Advocacy, Safiyya Sickle Cell Foundation in Nigeria, Sickle Cell Parents Association, Nagpur, India, Sickle Cell Disease Association of Canada, PRDPF, USA. And Ghana, Kenya, Abuja.





Final goal: find **an ePAG on SCD** for the ERN-EuroBloodNet

France: Dianaba Ba & Constant Vodohue & Nell Tsang-San-Moï & Danitzia Logis

Belgium: Liza Wema & A. Musongo

Italy: Nkongne Gyslène Michel & Costanza Musci

Portugual: Miriam Freire & Décio Temporário

Spain: Antonio Arenas García & Alexis Rodríguez Gallego

RBC ePAG: Loris Brunetta & ????????

Procedure for electing the ePAGs
for the ERN-EuroBloodNet is
done co-joint with EURORDIS





Participate in the global campaign to raise awareness of sustainable development

2020



Patients Videos Testimonies WSCD2020

Spreading patients' needs and expectation during the World Sickle Cell Disease Awareness Day 2020 (WSCD)

2021

World Sickle Cell Awareness Day 19th June 2021



Hematological Diseases (ERN EuroBloodNet)

ERN-EuroBloodNet's multi level actions in the field of Sickle Cell Disease (SCD) aims at wide distribution of knowledge and cross border patients support using different approaches:

Webinars for health professionals and patients



Online [educational courses](#) targeting SCD. Visit EuroBloodNet's EDU channel on [YouTube](#) to watch videos.

Preceptorships for health professionals



Short stays on SCD in expert Health Care Providers, members of the ERN-EuroBloodNet, accredited by European Board of Accreditation in Hematology (EBAH) Continuing Medical Education points.

SCD patient priority blog



A [blog](#), written by people living with SCD.

Training sessions for patients and caregivers



SCD Educational Sessions for people living with SCD at ASCAT [2019](#) and [2020](#).

Repository of Educational Material for Health professionals and patients



[International existing documents](#), for best practices, patient therapeutic education and development of (unique) international guidelines on clinical management.

European patients' registries



[ENROL](#) - European Rare Blood Disorders Platform

[RADeep](#) - The Rare Anemia Disorders European Platform

Visit www.eurobloodnet.eu and www.radeepnetwork.eu

Supporting the Establishment of the European Network of SCD Patients Organizations



A [bottom-up umbrella network](#) of national and local organizations of [SCD patients advocates](#).

coordination@eurobloodnet.eu
www.eurobloodnet.eu
www.eurobloodnet.eu/newsletter/



Covering more than 400 rare and complex hematological diseases, ERN-EuroBloodNet offers its online facilities, services and expertise to support orphan blood disorders.



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

2022

For World Sickle Cell Day 2022:

EuroBloodNet
WSCD2022

Sickle cell gave me a voice

I learned the power of communication, sharing information and experiences, standing up for my rights and difficulties as a patient, and speaking up for SCD locally and globally.

Support, sharing and raising awareness are essential.

Miriam, Portugal/UK

Together we are stronger!

#SickleCellDisease #WSCD2022 #appdh #escf #EuroBloodNet #EURORDIS

2023



Participating writing articles: SCD BLOG and ERN-EuroBloodNet Newsletter

Sickle Cell Disorder- let's get talking!

[Home](#) [Contact](#)

Why be a patient advocate?



<https://streetlight.ufhealth.org/disease-specific-support/sickle-cell-population/>

Why a patient advocate? Early days

I was diagnosed with sickle cell disease when I was 18 months old. Average life expectancy for a child with SCD was around 6 years. Living in Portugal well into my adult years there was little knowledge of sickle cell even in the healthcare community, apart from haematology teams. As an overprotected child, the natural and easy attitude was to let my mother speak on all occasions and take care of everything: appointments, medication, medical tests, treatments, the lot. One day my haematologist consultant



Contact us |

[ERN-EuroBloodNet](#) [Membership](#) [Best practices](#) [Education](#) [CPMS](#) [ENROL](#) [Patients' advocacy](#) [COVID-19](#) [Research](#) [News](#) [#ERNcare4Ua](#)

Newborn screening for sickle cell disease in Belgium

Newborn screening for sickle cell disease in Belgium

🕒 2021-03-28



A request from Belgian SCD patients advocates' group

Sickle cell disorders (SCD) are rare hematologic genetic disorders that in their severe forms can lead to disability or even death. SCD is mostly prevalent in Africa sub-Saharan regions, but due to the augmented immigration flows, its incidence in Europe has increased and the patient's number will continue to grow. A set of interventions is required at global public health level, as for instance **newborn screening**. Indeed, the early detection of SCD reduces childhood' mortality and morbidity and it improves the prognosis of the diseases.

To tackle SCD patients' needs in Europe, the **ERN-EuroBloodNet together with EURORDIS is establishing the European Network of Sickle Cell Disease Patients' Organisations** aiming to provide a centralized European point of contact for patients and caregivers. It will be a bottom-up umbrella network of national and local organizations of SCD patients' advocates.

The ERN-EuroBloodNet decided to act at the local level inviting patients support group and/or motivated patients to EuroBloodNet National Meetings dedicated to the promotion of the project. National Meeting' objectives are: training in patient advocacy, listening to SCD patients' needs and expectations, encourage, if not existent yet, to form a national association/federation/group.



Patient webinar on CDA



Patient webinar on SCD



YouTube

Topic on Focus on SCD for patients and their families

Once per month, every Monday 5 pm – 6 pm (Paris CET)

1. **January** “New therapies for SCD
2. **February** “Adult Patients Quality of Life”
3. **March** “Hydroxyurea and fertility”
4. **April** “Neurological Damages in SCD”
5. **May** “Neonatal Screening”
6. **June** “Gestational Risk”,
7. **July** “SCD and immune disease”
8. **September** “Genetic Counselling and embryo selection
9. **October** “BMT: survival infertility and other complications,
10. **November** “Polyuria and Enuresis: kidney damages”
11. **December** “Priapism”

Topics identified via Surveys on patients' expectations regarding the educational topics to be addressed



PATIENT EDUCATION

- Teaching objectives defined by People living with SCD
- Joint program of international organisations
- Program addresses educational gaps
- Worldwide free open access both for live session and recorded e-learning module



ERN-EuroBloodNet&ASCAT
SCD Patients Educational Session
at ASCAT 2022

- RACISM
- MENTAL HEALTH
- BONE MARROW TRANSPLANT
- PREGNANCY

January 26-28, 2022 Virtual Global Conference

European Reference Network for rare or low prevalence complex diseases

SCD Patients' Educational Session at ASCAT January 2022: Spotlight on Mental Health

ERN-EuroBloodNet's EDU

358 subscribers

Analytics Edit video

0 0 Share Download Clip Save

Webinars

Topic on Focus EuroBloodNet

Adult Patients Quality of Life

EuroBloodNet Topic on Focus on SCD for Patients ...

ERN-EuroBloodNet's EDU

Public

6 videos Last updated on Aug 11, 2022

Play all Shuffle

ERN-EuroBloodNet Topic on Focus on Sickle Cell Disease (SCD) for patients and their families aims to disseminate very important topics related to SCD to the patient community and their families: the latest advances in treatment and diagnosis, improving the quality of life in the field of this rare red blood cell disease.

The 11 Topics of this program have been chosen by people living from SCD answering the ERN-EuroBloodNet Survey on Prioritization of Educational Needs.

PROGRAM

New therapies for SCD
Speaker: Malika Benkerrou (AP-HP Hospital Saint-Louis, Paris)

Adult Patients Quality of Life
Date: 14th February 2022

Sort

- EuroBloodNet Topic on Focus on SCD for Patients & their families: Adult Patients Quality of Life
ERN-EuroBloodNet's EDU
1:02:00
- EuroBloodNet Topic on Focus on SCD for Patients & their families: Hydroxyurea and fertility
ERN-EuroBloodNet's EDU
44:10
- EuroBloodNet Topic on Focus on SCD for Patients & their families: Sickle Cell Disease (SCD) for patients and their families: the latest advances in treatment and diagnosis, improving the quality of life in the field of this rare red blood cell disease.
ERN-EuroBloodNet's EDU
1:17:17
- EuroBloodNet Topic on Focus on SCD for Patients & their families: Sickle Cell Disease (SCD) for patients and their families: the latest advances in treatment and diagnosis, improving the quality of life in the field of this rare red blood cell disease.
ERN-EuroBloodNet's EDU
54:48
- EuroBloodNet Topic on Focus on SCD for Patients & their families: Sickle Cell Disease (SCD) for patients and their families: the latest advances in treatment and diagnosis, improving the quality of life in the field of this rare red blood cell disease.
ERN-EuroBloodNet's EDU
39:52
- EuroBloodNet Topic on Focus on SCD for Patients & their families: Sickle Cell Disease (SCD) for patients and their families: the latest advances in treatment and diagnosis, improving the quality of life in the field of this rare red blood cell disease.
ERN-EuroBloodNet's EDU
51:20



YouTube

SCD Patients' Educational Session at ASCAT 2020

ERN-EuroBloodNet's EDU

Public

9 videos Last updated on Apr 14, 2021

Play all Shuffle

Sickle Cell Disease patients and parents' patients Educational Session at the 15th Annual Sickle Cell and Thalassaemia Conference 2020 (ASCAT) is a joint project of ERN-EuroBloodNet, ASCAT and BSH.

This project has provided for the second year the perfect opportunity for people living with SCD to experience how important is to participating in Research's Agenda, while demonstrated that the medical profession is willing to change and listen to the patient voice.

49 people living with SCD have registered to the online educational session.

According to the ASCAT 2020 program, each session of the program were moderated by one healthcare

Available on EuroBloodNet's EDU YouTube channel and on the e-learning environment on ERN-EuroBloodNet website



PATIENT EDUCATION at ASCAT

- Physicians and patients working together during the ASCAT Conference on Patients Education
- Patients present the outcomes of the session at the plenary closing conference
- Capacity to improve information, build collaborations, influence research and impact on care models

SINCE 2019

WITH THE ERN-EUROBLOODNET, PATIENTS
ARE AT THE CORE OF ASCAT CONGRESS





Gathering patients needs conducting worldwide surveys and presenting the results at scientific congresses



European Reference Network
for rare or low prevalence complex diseases

Network Hematological Diseases (ERN EuroBloodNet)



ASCAT

THE LANCET
Haematology

Research - Education - Policy



Hematological Diseases (ERN EuroBloodNet)



Co-funded by the European Union



OJRD-D-23-00156R1

Sickle cell disease: embedding patient participation into an international conference can transform the role of lived experience

Mariangela Pellegrini; Subarna Chakravorty; Maria del Mar Manu Pereira; Béatrice Gulbis; Catriona Gilmour-Hamilton; Sandy Hayes; Mariane de Montalembert; Baba Psalm Duniya Inusa; Raffaella Colombatti; Noemi Roy Orphanet Journal of Rare Diseases



Thank you!