Visit your doctor…

The doctor does some tests…

You feel ill…

The doctor prescribes iron pills…

...and says: you have anaemia!

You have a Rare Anaemia

ENERCA
EUROPEAN NETWORK FOR RARE AND CONGENITAL ANAEMIAS
WHAT IS A RARE ANAEMIA?

1. ANAEMIA WITH A PREVALENCE < 5 / 10,000 IN EUROPE

2. NOT DUE TO IRON DEFICIENCY

3. CONGENITAL ORIGIN IN MORE THAN 80% OF CASES

4. UNKNOWN AETIOLOGY IN ABOUT 30% OF CASES

5. KEY CLINICAL SIGN OF MORE THAN 62 RARE DISEASES
THE RARE ANAEMIAS IN THE CONTEXT OF RARE DISEASES

• BLOOD TRANSFUSIONS AND IRON CHELATING ARE, IN GENERAL, THE ONLY THERAPEUTIC OPTIONS

• PREVENTIVE ACTIONS TO REDUCE ITS FREQUENCY AND TO ACHIEVE AN EQUILIBRIUM BETWEEN MORBIDITY AND PATIENT’S QUALITY OF LIFE ARE FREQUENTLY REQUIRED (Newborn screening programs)

• MORE THAN 500,000 CHILDREN, WORLDWIDE, BORN WITH A RARE ANAEMIA, MAINLY DUE TO:
  – THALASSAEMIA (Mediterranean Anaemia)
  – SICKLE-CELL DISEASE (Sickle-Cell Anaemia)
THE ENERCA PROJECT
creating an NETWORK since 2002

Co-funded by the Health Programme of the European Union

- First network of experts
- Clear and concise information
- Protocols for diagnosis
- Congenital anaemias only

Phase II (2005 – 2008)
- Network Consolidation
- Haemoglobinopathies database
- Dissemination and awareness
- Congenital and acquired anaemias

Phase III (2009 – 2012)
- Guidelines
- Social awareness
- Patient’s empowerment
- White Book
Online e-health Platforms on rare anaemias
ENERCA nourished by all the concepts and initiatives surrounding rare diseases developed in Europe along its 10 years of life.
ENERCA: A highway to the future

PILOT EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES FINANCED BY THE EUROPEAN COMMISSION

<table>
<thead>
<tr>
<th>Network</th>
<th>Duration</th>
<th>Year</th>
<th>Year</th>
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<td>Dyscerne</td>
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<tr>
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<tr>
<td>PHL</td>
<td>01/08/2008 - 31/07/2011</td>
<td>2007</td>
<td>2008</td>
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<td>2008</td>
<td></td>
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<tr>
<td>Care-NMD</td>
<td>01/05/2010 - 30/04/2013</td>
<td>2007</td>
<td>2008</td>
<td></td>
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<tr>
<td>ENERCA</td>
<td></td>
<td>2007</td>
<td>2008</td>
<td></td>
<td></td>
<td></td>
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</tr>
</tbody>
</table>
Over 90 health professionals from up to 18 European countries also including Eastern countries

STRATEGY: Identification of expert centres in rare anaemias
To increase the **efficacy of diagnosis, treatment and follow up** of patients ... 

To **reduce health inequalities** in the diagnosis and prevention of major rare anaemias

Helping the Doctor = Helping the Patient
How to reduce health inequalities?

Moving forward the creation of a European Reference Network (ERN) in Rare Anaemias (RAs)

Prepared on the basis of three main issues

- Medical & Technical
- Legal & Ethical
- Patients' Expectations

Available soon through ENERCA website
The White Book

Analysis of controversial issues in the three main aspects:

- Legal & Ethical
- Medical & Technical
- Patients’ Expectations

Discussion of results and preparing a report...

Report & Draft recommendations

Final discussion and consensus proposal...

Final Consensus Recommendations Proposal

Directive 2011/24/EU on the application of patients’ rights in cross-border healthcare
Patients’ Expectations

1. Availability of transfusion

Answers received from 415 patients from 14 European countries

2. School teaching/working days lost annually due to the treatment

<table>
<thead>
<tr>
<th>Question</th>
<th>Number of patients</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>None</td>
<td>45</td>
<td>10.84%</td>
</tr>
<tr>
<td>1-5 days</td>
<td>37</td>
<td>8.91%</td>
</tr>
<tr>
<td>6-10 days</td>
<td>20</td>
<td>4.82%</td>
</tr>
<tr>
<td>11-15 days</td>
<td>48</td>
<td>11.57%</td>
</tr>
<tr>
<td>16 or more days</td>
<td>213</td>
<td>51.33%</td>
</tr>
<tr>
<td>No reply</td>
<td>52</td>
<td>12.53%</td>
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</table>
## Patients’ Expectations

### 3. Priorities according to patients

<table>
<thead>
<tr>
<th>Question</th>
<th>% of &quot;Strong support&quot;</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coordinated team with experienced doctor in charge</td>
<td>71.1</td>
</tr>
<tr>
<td>Doctor who understands patients’ needs</td>
<td>67.5</td>
</tr>
<tr>
<td>Discuss treatment plans</td>
<td>62.4</td>
</tr>
<tr>
<td>Experience / technical support for diagnosis and complications</td>
<td>61</td>
</tr>
<tr>
<td>Multidisciplinary care</td>
<td>60</td>
</tr>
<tr>
<td>To follow best practice guidelines</td>
<td>58</td>
</tr>
<tr>
<td>Continuity of care</td>
<td>52.5</td>
</tr>
<tr>
<td>Staff attention to patient concerns</td>
<td>50.6</td>
</tr>
<tr>
<td>Network of expert centres nationally</td>
<td>50.4</td>
</tr>
</tbody>
</table>
ENERCA Professional Platforms

The three professional Platforms are designed, developed and tested within the

**ENERCA Website**

- **Registry** (Cyprus)
- **Learning** (France)
- **e-Health** (Belgium)

Health professionals, Health Authorities and other **Patients**
ENERCA website

www.enerca.org

- Full use of RA-ERN services
- Reliable and updated information
- Training & Educational Material
- News and Agenda (Newsletter)
- Easy access to the new IT tools
Up to 62 Rare diseases with anaemia as key clinical symptom

Information is directly nourished from professionals and patients profiles

What is a rare anaemia?

According to the European Commission a disease is rare when its population frequency is less than 1 case in 10,000 individuals.
Sickle cell anaemia

**Acronym:** SCA  
**Synonym(s):** Drepanocytosis  
**ORPHANET code:** [232](http://www.orpha.net)  
**OMIM code:** [603903](http://www.omim.org)  
**ICD-10 code:** D57.0  
**Include:** Only HbS Homozygosity  
**Group:** Structural haemoglobinopathies  
**Subgroup:** Sickle cell disorders

**Haemoglobin S and sickle cell disorders:** are disorders of the haemoglobin, a major component of the red blood cells. Sickle cell disorders (SCD) are the consequence of the presence of an abnormal haemoglobin called haemoglobin S (Hb S). There are several forms of which the most frequent is due to haemoglobin S homozygosity, while compound heterozygosity lead to a more or less severe form: SC, SD-Punjab, SO-Arab, SB-thalassaemia.

**What causes the disease and how common is it?**  
This is a genetic disease. It is linked to a mutation of the B-globin gene, encoding the β-globin chain, one of the components of haemoglobin (Hb). An individual can be heterozygous for the disorder (Hb AS individuals) when only one of the globin genes is mutated, or homozygous (Hb SS individuals), or compound heterozygotes HbSC, HbSD, HbSO-Arab, HbSB-thalassaemia individuals) when the two beta globin genes are affected. It is a frequent disorder in people originating from Africa, Middle-East, India, and the Mediterranean Basin. This is due to the fact that these areas were or are still infected with malaria and Hb S confers a relative protection against malaria.

**What are the most frequent symptoms if I have the disease?**  
Sometimes red blood cells from patients with sickle cell disorders become sickle-shaped (crescent shaped), have difficulty passing through small blood vessels and are destroyed rapidly. This explains why people with a sickle cell disorder have anaemia (pallor) and jaundice (yellow color of the eyes); when the circulation is blocked by the sickle-shaped red blood.
Find your Centre
Sickle cell anaemia

HAMBURG

GESUNDHEITSZENTRUM MAX-BRAUER-ALLEE
Max-Brauer-Allee 52 - 22765 Hamburg

BERLIN

CHARITÉ - UNIVERSITÄTSMEDIZIN BERLIN; KLINIK FÜR PÄDIATRIE MIT SCHWERPUNKT ONKOLOGIE/HÄMATOLOGIE/KMT
Augustenburger Platz 1 - 13353 Berlin
Stephan Lobitz, Dr.

HEIDELBERG

ZENTRUM FÜR KINDER- UND JUGENDMEDIZIN HEIDELBERG
INF 430 - 69120 Heidelberg
Hermann Heimpel, Professor, Joachim Kunz, Dr.

FREIBURG
Professor Hermann Heimpel

Physician

Center: Zentrum für Kinder- und Jugendmedizin Heidelberg
Address: INF 430 69120 Heidelberg • Germany

Clinical hematology in general, morphology of hematopoiesis, aplastic anemia, MDS, iron metabolism, hemolytic anemias, congenital dyserythropoietic anemias, stem cell transplantation, drug induced blood disorders. Medical education.

Documents

- Unstables Hämoglobin
- Hämoglobin S und Sichelzellanämien
- Hämoglobin Lepore
- Hämoglobin E
- Beta-Thalassämie – leichter Form oder Trägerstatus (Thalassaemia minor)
- Bei der Hämoglobin C-Krankheit liegt
- Hämoglobin M mit Anämie
- Hämoglobin H-Krankheit
Sickle cell anaemia

Members & Centers

Cyprus

NICOSIA

THALASSAEMIA INTERNATIONAL FEDERATION
P.O. Box 28807 - 2007 Strovolos Nicosia

France

STRASBOURG

DORYS
1a place des orphelins - 67200 Strasbourg

Italy

GENOVA

ASSOCIAZIONE LIGURE THALASSEMICI ONLUS
C/I E.O. OSPEDALI GALLIERA VIA VOLTA 8 - 16128 GENOVA
Thalassaemia International Federation (TIF) is a non-profit, non-governmental patient-driven organisation founded in 1986 and working in official relations with the World Health Organization (WHO) since 1996. TIF is an umbrella organisation involving 108 national thalassaemia associations and other members from over 60 countries across the world.

Diseases

- Compound heterozygous sickling disorders
- Haemoglobin C disease
- Haemoglobin D disease
- Haemoglobin E disease
- Haemoglobin M with anaemia
- Sickle cell anaemia
And more...

ENERCA for Patients

Educational material

All About Thalassaemia

The Thalassaemia International Federation (TIF) dedicates this book to all the children in the world born with Thalassaemia. To their indomitable will to survive, their inspiring fight against the disease, their beautiful dreams of a good life and a bright future. TIF is a partner of ENERCA and its educational publications provide concise, up-to-date information on all aspects of thalassaemia, from prevention to clinical management.

ENERCA video about haemoglobinopathies and children

Haemoglobinopathies are inherited blood disorders that reduce the production of haemoglobin, a substance within red blood cells that carries oxygen through all the body. Haemoglobinopathies are inherited illnesses that can pass from parents to their children. Get informed! All you need to do is a haemogram and a haemoglobin study. Ask your family doctor. Even parents who are both carriers of a haemoglobinopathy can have a healthy family together.

The video has been designed by ENERCA professionals to explain the importance of prenatal screenings. It is available also in Spanish, Portuguese, Italian and French at Vimeo.
Video on prevention.....genetic counseling...

English, Spanish, Portuguese, Italian, French and more
Welcome
ENERCA means easy access to high quality information on rare anaemias for patients, citizens, health professionals, stakeholders, authorities and pharmaceutical industry.

For patients
- Find your centre
- Know more about Anaemias
- Do you have Anaemia?
- Create your patient association profile

For professionals
- Flowchart
- MAC
- Find colleagues
- Create your profile

Subscribe to the ENERCA Newsletter
Anaemia Diagnosis

The flowchart provided here intends to be guidance for a first diagnostic approach for rare congenital anaemias in adults. The ENERCA team doesn’t lay any claim to its completeness. Some haematological parameters should be introduced before the flowchart can start. The normal reference values for haematological parameters have been partially obtained from Dacie and Lewis’ PRACTICAL HAEMATOLOGY, 10th Edition 2006, edited by S.M. Lewis, B.J. Bain and I. Bates and are given as follows:

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Women</th>
<th>Man</th>
</tr>
</thead>
<tbody>
<tr>
<td>Haemoglobin</td>
<td>120-150 g/L</td>
<td>130-170 g/L</td>
</tr>
<tr>
<td>Reticulocytes</td>
<td>Woman/Men: 50-100 x 10⁹/L</td>
<td></td>
</tr>
<tr>
<td>Mean cell volume</td>
<td>Woman/Men: 80-100 fl (83-101 fl in Dacie and Lewis' Practical Haematology)</td>
<td></td>
</tr>
</tbody>
</table>

- **Woman**
  - Haemoglobin (Hb) [ ] g/L
  - Reticulocytes (Rets) [ ] x 10⁹/L
  - Mean cell volume (MCV) [ ] fl

- **Man**
  - Haemoglobin (Hb) [ ] g/L
  - Reticulocytes (Rets) [ ] x 10⁹/L
  - Mean cell volume (MCV) [ ] fl
Anaemia Diagnosis

The flowchart provided here intends to be guidance for a first diagnostic approach for rare congenital anaemias in adults. The ENERCA team doesn’t lay any claim to its completeness. Some haematological parameters should be introduced before the flowchart can start. The normal reference values for haematological parameters have been partially obtained from Dacie and Lewis’ PRACTICAL HAEMATOLOGY, 10th Edition 2006, edited by S.M. Lewis, B.J. Bain and I. Bates and are given as follows:

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</tr>
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Result
No anaemia has been identified

- Woman
  - Haemoglobin (Hb): 145 g/L
  - Reticulocytes (Retics): 67 x 10⁶/L
  - Mean cell volume (MCV): 89 fl

- Man

Calculate
Anaemia Diagnosis

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<td></td>
</tr>
<tr>
<td>Mean cell volume</td>
<td>90-100 fl (83-101 fl in Dacie and Lewis' Practical Haematology)</td>
<td></td>
</tr>
</tbody>
</table>

The calculated values are:
- Haemoglobin (Hb): 115 g/L
- Reticulocytes (Retics): 90 x 10⁹/L
- Mean cell volume (MCV): 76 fl

Calculate
Anaemia Diagnosis

The flowchart provided here intends to be guidance for a first diagnostic approach for rare congenital anaemias in adults. The ENERCA team doesn't lay any claim to its completeness. Some haematological parameters should be introduced before the flowchart can start. The normal reference values for haematological parameters have been partially obtained from Dacie and Lewis' PRACTICAL HAEMATOLOGY, 10th Edition 2006, edited by S.M. Lewis, B.J. Bain and I. Bates and are given as follows:

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</tr>
<tr>
<td>Mean cell volume</td>
<td>Woman/Men: 80-100 fL (83-101 fL in Dacie and Lewis' Practical Haematology)</td>
</tr>
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</table>

**Result**

Anemia has been identified

For further investigation of rare anaemias be aware that the more frequent causes of anaemia in clinical practice should be excluded.

These could be drug ingestion, chemotherapy, alcohol intake, haemorrhage of different origin, radiation therapy, chronic diseases (rheumatic arthritis) and any other causes of secondary anaemia.

See the Flowchart
Anaemia Diagnosis

- **Woman**
- Haemoglobin (Hb) 115 g/L
- Reticulocytes (Retics) 90 g/L
- Mean cell volume (MCV) 76 g/L

**Transferrin saturation**

- <15%
  - Low
    - Serum Ferritin
      - Normal/high
        - Clinical or biochemical evidence of inflammation
          - Yes
            - Anaemia of chronic disease
              - Haemoglobin studies
                - Normal
                - Abnormal
                  - Hypochromic red blood cells;
                  - Red cell zinc protoporphyrin;
                  - Serum transferrin receptor;
                  - Bone marrow iron (Perls stain)
          - No
            - Thalassaemia or Hb variants
              - Other studies
        - Normal or low
          - Search for an etiology
ENERCA for Professionals

And more...

Free Webcasting trainings courses
ENERCA for Professionals

and more...

European Symposium on Rare Anaemias in collaboration with patients

Next Edition: The Netherlands, 2015
The new ongoing Project: e-ENERCA

The patient at the centre of the sanitary ecosystem

Our mission is to enhance the communication between healthcare professionals and patients; optimize prevention and monitoring processes and empower the patient. Patients understand they are jointly responsible for their health and wellbeing, at the same time they receive a more personalized care.
The three main reasons for e-health platforms

- Poor implementation of data collection and analysis systems
  >>> e-Registry

- Great variability across Europe to access certified and updated information
  >>> e-Learning

- Deep inequalities among countries for diagnosis, prevention and clinical care of patients
  >>> Telemedicine
Haemoglobinopathies are today the most common genetic disorders in Europe.

Over 330,000 affected children with major haemoglobinopathies are born worldwide each year.

1% of couples are at risk of having a newborn with a severe syndrome.

There are poor data on their precise prevalence, overall burden and trends.
e- ENERCA action for registry

Main targets

- Inventory of **expert centres**
- Electronic record of **health data**
- Epidemiological **surveillance**
- Relevant **clinical information**

Androulla Eleftheriou (TIF)
Michael Angastiniotis (TIF)
(Cyprus)

http://www.sxc.hu/photo/1139316
e-Learning: The challenge of knowledge in rare anaemias

Patient’s global management differs between countries
e-ENERCA action for education

Main targets

- Harmonization of medical CV
- Continuing medical education
- On-line modules for teaching (*)
- e-learning modules:
  - Training courses and workshops
  - Recommendations
  - Self training/self assessment
  - Interaction with experts (link-Telemedicine)

* Complementary to on-site training modules

http://www.sxc.hu/photo/987822

Patricia Aguilar (CHUM) (France)
Telemedicine: The challenge of diagnosis in rare anaemias

- About 30% of rare anaemias remain undiagnosed or misdiagnosed
- Treatment, genetic counselling and/or prognosis are missing or incomplete
- Scarce expertise in diagnosis creates patient’s and family anxiety

http://www.sxc.hu/photo/1078182
e-ENERCA action for tele medicine

Main targets

- Increase communication and sharing of clinical knowledge
- Promote experts to share on-line discussions
- Allow an earlier and more accurate diagnosis and patients follow-up
- Facilitate research by storing large volume of data and images
- Decrease health care costs

Béatrice Gulbis (ERASME) (Belgium)

http://www.sxc.hu/photo/1155570
ENERCA challenge
for ERN long term sustainability (1)

- National recognition of centers of expertise
- RA-ERN recognition by European Commission
- National and European economical support

http://www.sxc.hu/photo/1437890
ENERCA challenge for ERN long term sustainability(2)

Main Targets

- Promote the recognition of Centres of Expertise on RAs and the RA-ERN
- Entry into force of the Directive 2011/24/EU on the application of patients’ rights in cross-border healthcare.

http://www.sxc.hu/photo/469047
e-ENERCA action for sustainability

- Identification of key players in each country:
  - Responsible person of a medical center
  - National Authority involved in the national plans for RD
  - Professional involved in RA diagnosis and management.

- Presentation of the **ENERCA White Book** Recommendations for recognition of Centres of Expertise

Carlos Romeo (UPV/EHU)
Pilar Nicolas (UPV/EHU)

http://www.sxc.hu/photo/469047
NOW:
ENERCA offers a solid platform to develop multidisciplinary IT initiatives for tackling rare anaemias...
Thank You!!!

ENERCA Coordinator Group

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Project Manager - María del Mar Mañú (maria.manu@enerca.org)

Project Assistant - Laura Olaya Costa (laura.olaya@enerca.org)

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Phone: +34 934515950
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