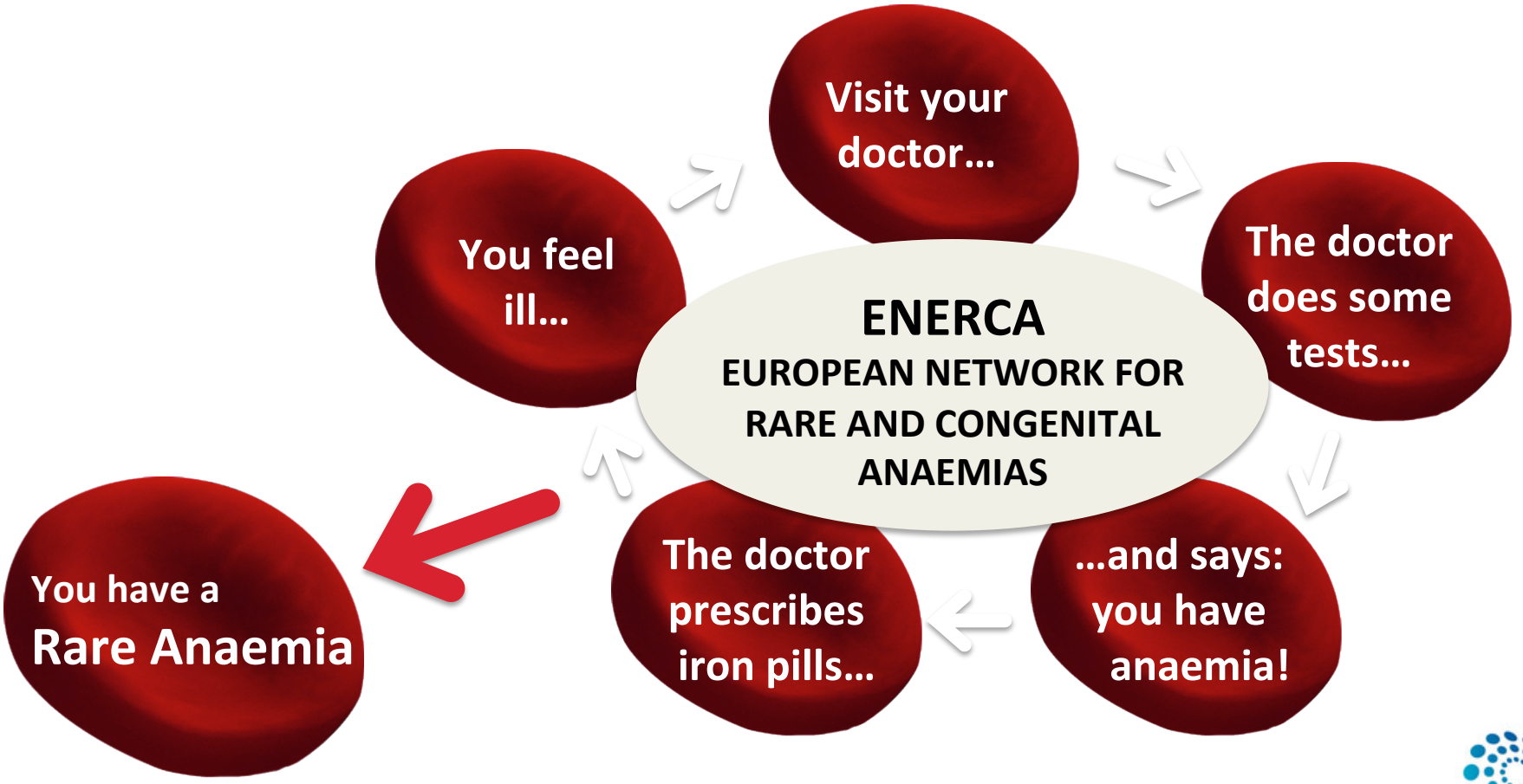
enerca
European Network for Rare and Congenital Anaemias

The Rare Anaemias

a clinical challenge



WHAT IS A RARE ANAEMIA ?

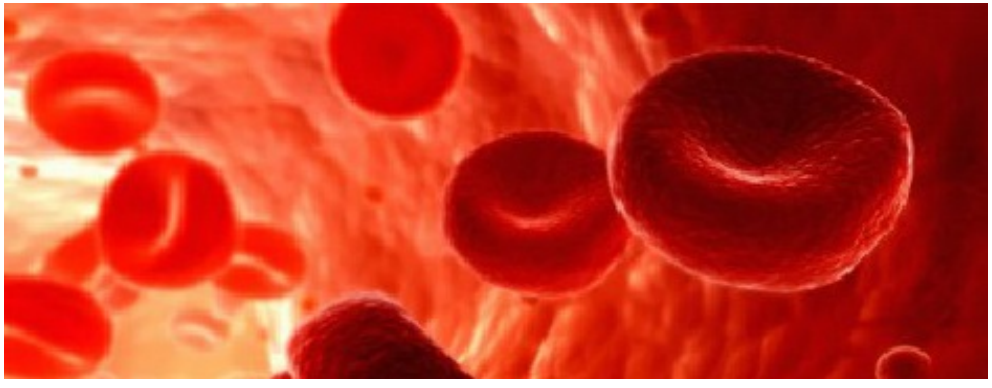
1. ANAEMIA WITH A PREVALENCE < 5 / 10.000 IN EUROPE

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 :

  - THALASAEMIA (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

Phase I (2002 – 2004)

- 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**

Phase IV (2013-2016) : ICT tools implementarion

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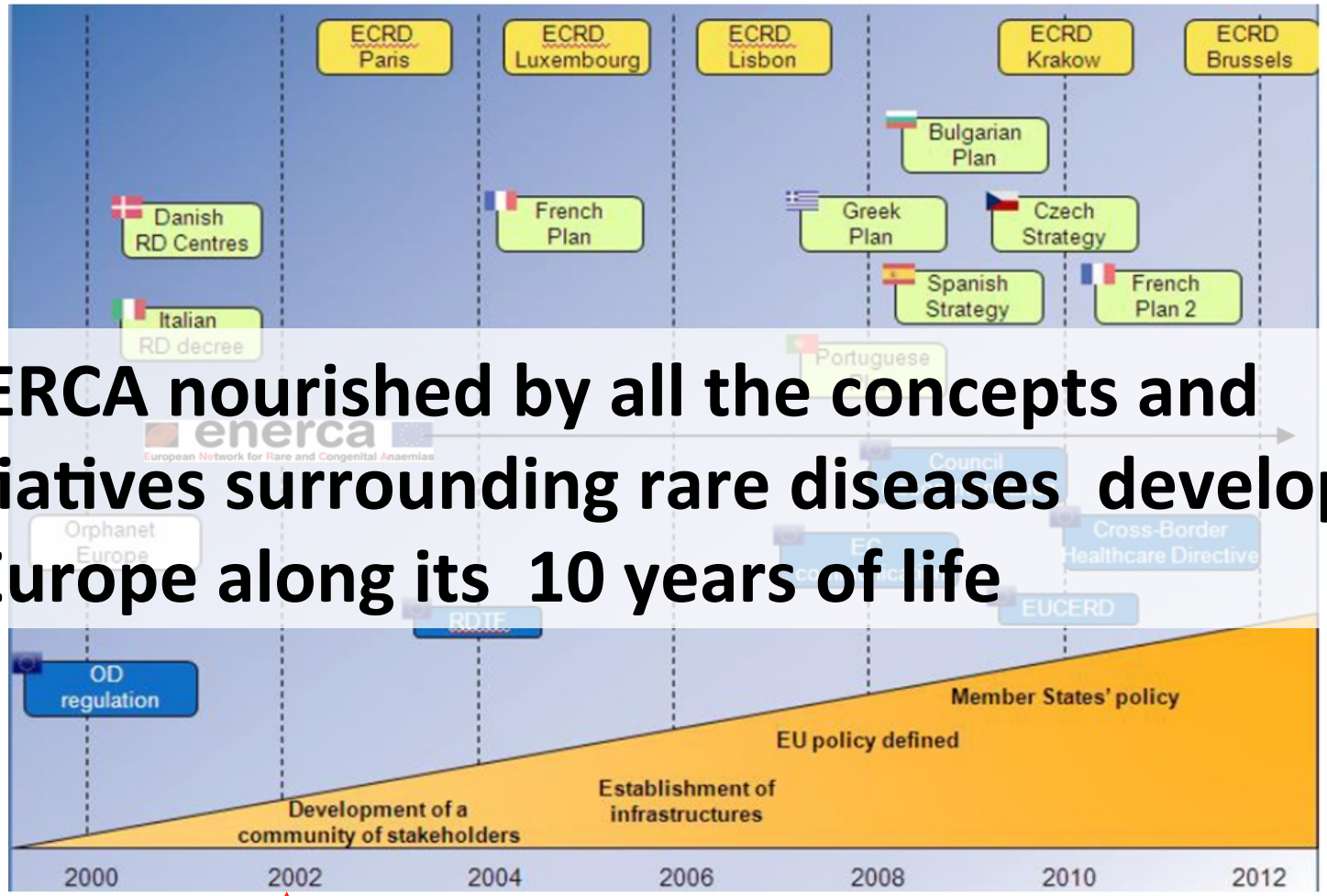
Online e-health Platforms on rare anaemias

Co-funded by the Health Programme of the European Union

www.enerca.org

THE ENERCA PROJECT (2002-2012)



ENERCA nourished by all the concepts and initiatives surrounding rare diseases developed in Europe along its 10 years of life

European Network for Rare and Congenital Anaemias

ENERCA :

A highway to the future

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

	2007	2008	2009	2010	2011	2012	2013
ECORN CF	01/04/2007 - 31/03/2010						
Dyscerne	April 2007 - March 2010						
PAAIR	April 2007 - March 2010						
EPNET	01/04/2007 - 31/03/2010						
EN-RBD	April 2007 - March 2010						
PHL		01/08/2008 - 31/07/2011					
Neuroped		24/04/2008 -23/04/2011					
Euro Histio Net		01/09/2008-31/08/2011					
TAG			01/12/2008 - 30/11/2011				
Care-NMD				01/05/2010 - 30/04/2013			
ENERCA							

ENERCA PROJECT

12 Associated Partners
14 Collaborating Partners
> 30 Affiliated Members

Over **90 health professionals**
from up to **18 European countries**
also including **Eastern countries**

STRATEGY : **Identification of expert centres in rare anaemias**

Executive Consortium

DESCA
The Simplified FP7 Model Consortium Agreement
www.DESCA-FP7.eu
Version 3.0 March 2011

H.CLINIC
Spain
WP1/2/3

→ **Coordination**

e-Health

H. ERASME
Belgium
WP6

Patients

THALASSAEMIA INTERNATIONAL FEDERATION

Etical & Legal issues

UPV
Spain
WP7

TIF
Cyprus
WP4

CHUM
France
WP5

Registry

Learning

ENERCA MAIN POLICY

To increase the **efficacy of diagnosis, treatment and follow up** of patients …

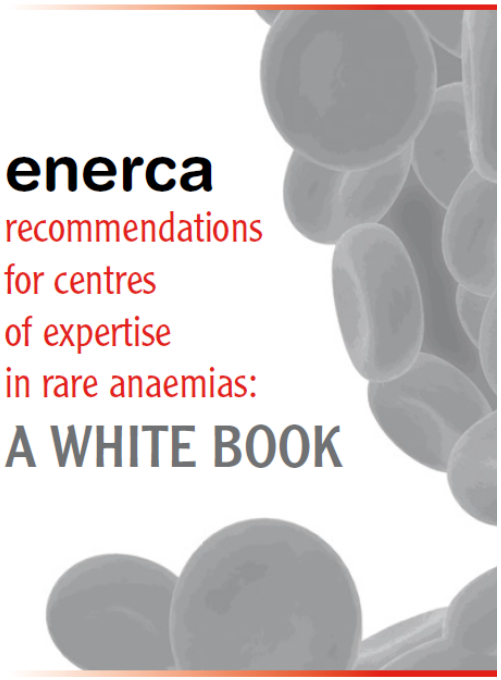
To **reduce health inequalities** in the diagnosis and prevention of major rare anaemias

↓

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

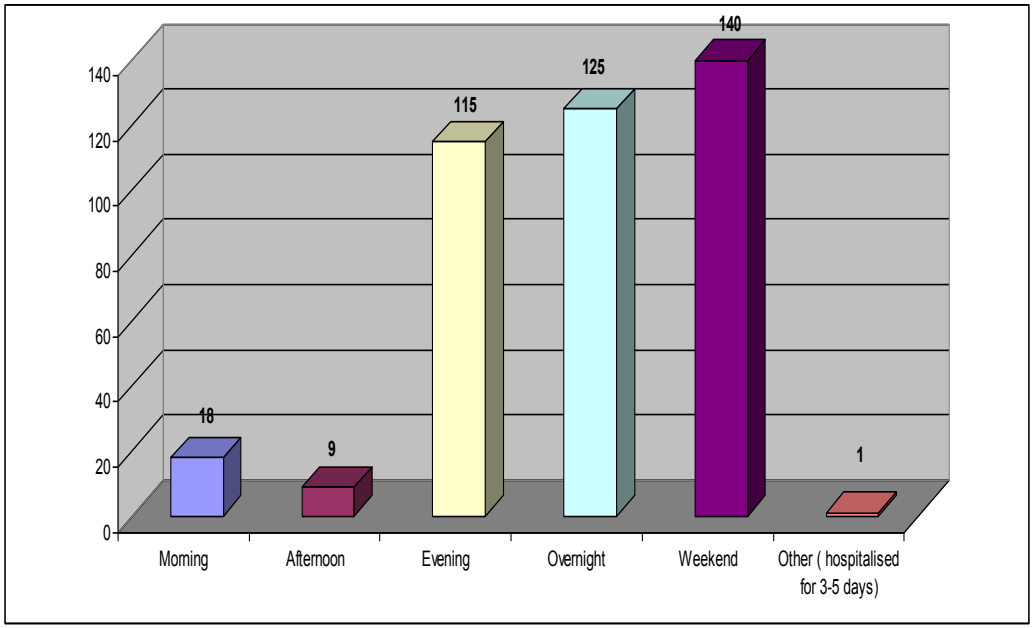
Analysis of controversial issues in the three main aspects:

Discussion of results and preparing a report…

Final discussion and consensus proposal…

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

Question	Number of patients	Percentage
None	45	10.84%
1-5 days	37	8.91%
6-10 days	20	4.82%
11-15 days	48	11.57%
16 or more days	213	51.33%
No reply	52	12.53%

Patients' Expectations

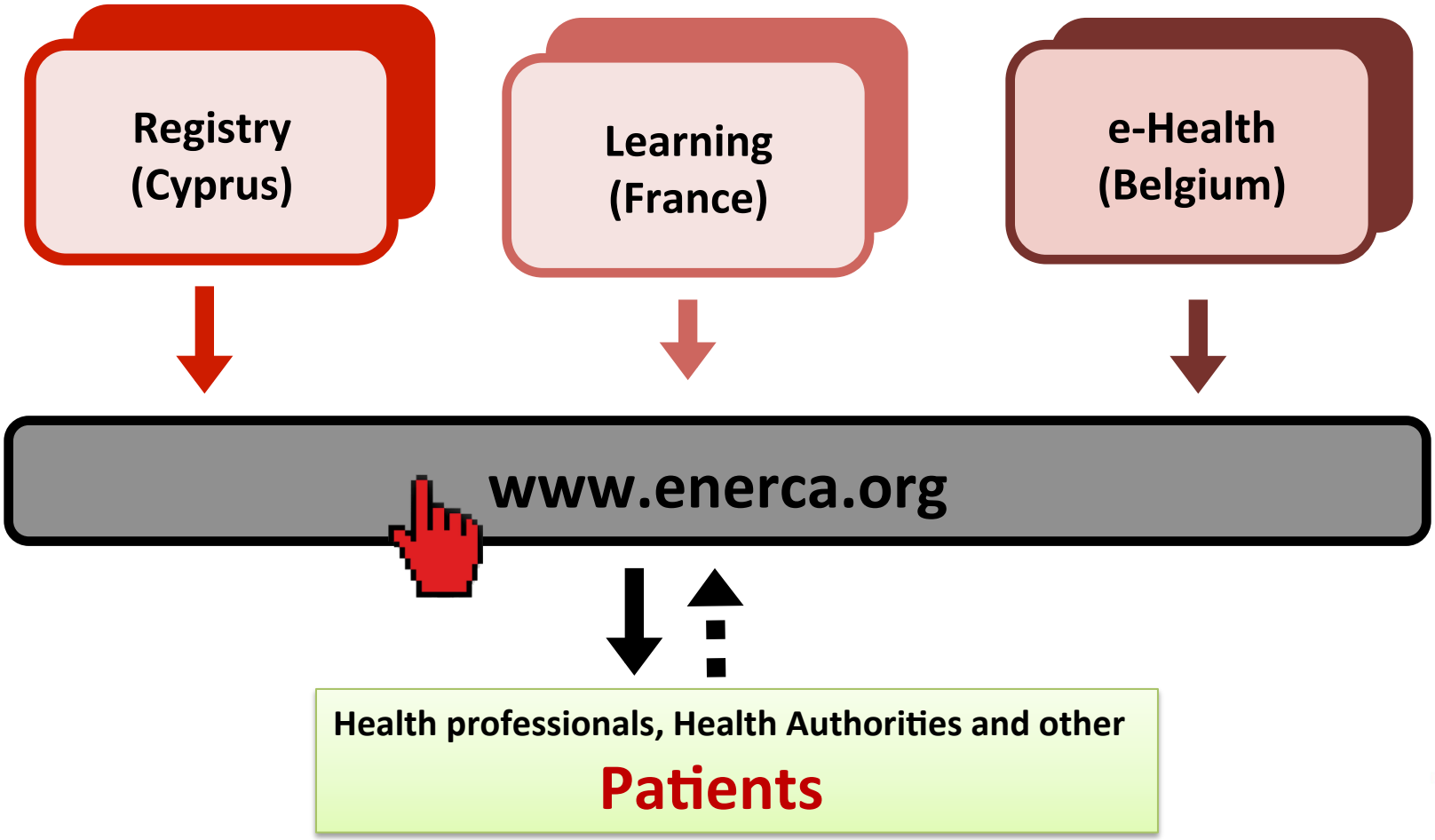
3. Priorities according to patients

Question	% of "Strong support"
Coordinated team with experienced doctor in charge	71,1
Doctor who understands patients' needs	67,5
Discuss treatment plans	62,4
Experience / technical support for diagnosis and complications	61
Multidisciplinary care	60
To follow best practice guidelines	58
Continuity of care	52,5
Staff attention to patient concerns	50,6
Network of expert centres nationally	50,4

ENERCA Professional Platforms

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

ENERCA Website



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

Contact Intranet Translate

Search

About Enerca | Anaemias | Members & Centers | Patient Associations | Activities | News & Agenda | For Professionals

Home > Anaemias

Anaemias

Know r

- Aceruloplasminemia
- Adenosine deaminase increased activity -ADA-
- Adenylate kinase deficiency
- Aldolase deficiency
- Alpha-thalassaemia - trait or carrier
- Atransferrinemia
- Autosomal dominant sideroblastic anaemia
- Autosomal recessive sideroblastic anaemia
- Beta-thalassaemia - trait or carrier
- Beta-thalassaemia major (and intermedia)
- CDA with thrombocytopenia (GATA I mutation)
- Compound heterozygous sickling disorders
- Congenital acanthocytosis
- Congenital dyserythropoietic anaemia type I
- Congenital dyserythropoietic anaemia type II
- Congenital dyserythropoietic anaemia type III
- Delta Beta-thalassaemia
- Diamond- Blackfan-Anemia
- DMT1- deficiency anaemia
- Familial hypoplastic anaemia
- Fanconi anaemia
- Gamma-glutamyl-cysteine synthetase deficiency
- GLRX5-related Sideroblastic anaemia
- Glucose phosphate isomerase deficiency
- Glucose-6-phosphate dehydrogenase deficiency
- Glutathione reductase deficiency
- Glutathione synthetase deficiency
- Haemoglobin C disease
- Haemoglobin D disease
- Haemoglobin E disease
- Haemoglobin H disease
- Haemoglobin Lepore
- Haemoglobin M with anaemia
- Hereditary Elliptocytosis
- Hereditary persistance of fetal haemoglobin
- Hereditary Spherocytosis
- Hereditary Stomatocytosis
- Hexokinase deficiency
- Hydrops fetalis
- Imerslund-Gräsbeck-Syndrom
- Iron - refractory iron deficiency Anemia
- Kearns-Sayre syndrome

Do you have Anaemia?

What is a

Anaemia
haemoglo
and the s
haematoc
lower than
but not in
types of a
also a dec
number of

What is a rare anaemia?

According to the European Commission a disease is rare when its population frequency is less than 5 cases in 10,000 individuals.

Up to 62 Rare diseases with anaemia as key clinical simptom

Information is directly nourished from professionals and patients profiles

Sickle cell anaemia

Acronym: SCA
Synonym(s): Drepanocytosis
ORPHANET code: 232
OMIM code: 603903
ICD-10 code: D57.0
Include: Only HbS Homozygozity
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 calle 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 ß-globin chain, one of the components of haemoglobin (Hb). An individual can be heterozygous for the disorder (Hb AS individuals) when only one o 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 originatin from Africa, Middle-East, India, and the Mediterranean Basin. This is due to the fact that these areas were or are still infect with malaria and Hb S confers a relative protection against malaria.

What are the most frequent symptoms if I have the disease?

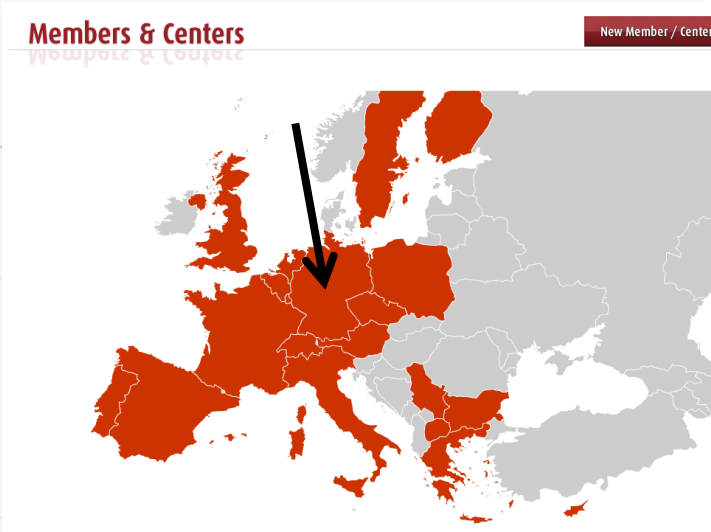
Sometimes red blood cells from patients with sickle cell disorders become sickle-shaped (crescent shaped), have difficu passing through small blood vessels and are destroyed rapidly. This explains why people with a sickle cell disorder have anaemia (pallor) and jaundice (yellow color of the eyes); when the circulation is blocked by the sickle-shaped red blood

Find your Centre

Sickle cell anaemia

General info | Members & centers | Patient Associations | Enerca Documents | Scientific Publications | Useful links

Germany



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.

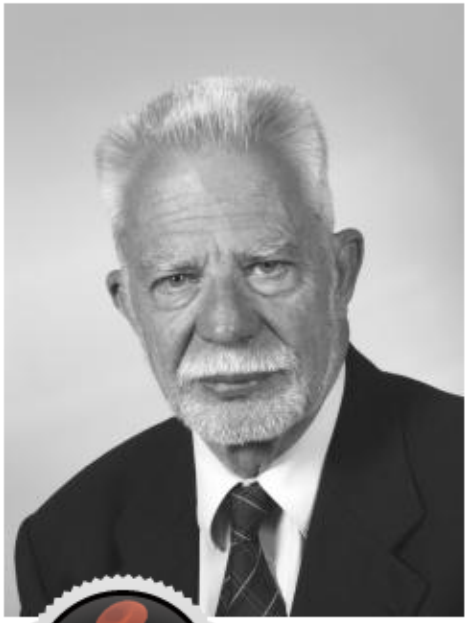
HEIDELBERG

ZENTRUM FÜR KINDER- UND JUGENDMEDIZIN HEIDELBERG
INF 430 - 69120 Heidelberg

Hermann Heimpel, Professor Joachim Kunz, Dr.

FREIBURG

Members & Centers list



enerca partner

Professor Hermann Heimpel

Physician

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

Clinical hematoloy 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

- Congenital Dyserythropoietic Anemias: "New observations, and practical aspects of diagnosis and therapy" (2007)
- Unstabiles Hämoglobin
- Hämoglobin S und Sichelzellkrankheiten
- Hämoglobin Lepore
- Hämoglobin E
- Beta-Thalassämie – leichte Form oder Trägerstatus (Thalassaemia minor)
- Bei der Hämoglobin C-Krankheit liegt
- Hämoglobin M mit Anämie
- Hämoglobin H-Krankheit

Sickle cell anaemia

General info | Members & centers | Patient Associations | Enerca Documents | Scientific Publications | Useful links

Cyprus

NICOSIA

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

France

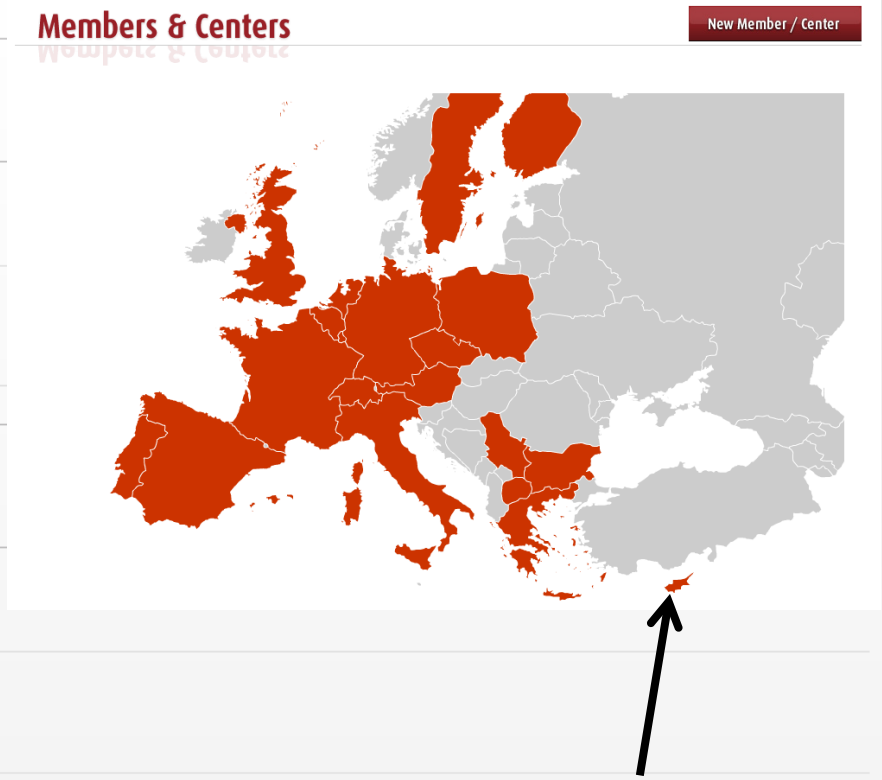
STRASBOURG

DORYS
1a place des orphelins - 67200 Strasbourg

Italy

GENOVA

ASSOCIAZIONE LIGURE THALASSEMICI ONLUS
C/O E.O. OSPEDALI GALLIERA VIA VOLTA 8 - 16128 GENOVA



Patient Associations

Associations list

Thalassaemia International Federation

enerca partner

Thalassaemia International Federation

E-mail: thalassaemia@cytanet.com.cy
Phone: +357-22-319-129
Fax: +357-22-314-552
Website: http://www.thalassaemia.org.cy
Adress: P.O. Box 28807. 2007 Strovolos Nicosia • CYPRUS
Contact Person: Dr. Androulla Eleftheriou Executive Director

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

ENERCA for Patients

And more…

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European Network for Rare and Congenital Anaemias

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About Enerca | Anaemias | Members & Centers | Patient Associations | Activities | News & Agenda | For Professionals

Home > Activities > Educational material

Educational material

- Rare anaemias definitions
- Collaboration
- Education material
- Training
- Newsletters

All About Thalasaemia

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 oxigen 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 haemoglobins 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.

Educational Material

Video on prevention…..genetic counseling…


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Welcome
ENERCA means easy access to a high quality information on rare anaemias for patients, citizens, health professionals, stakeholders, authorities and pharmaceutical industry.
News
Agenda
January 22nd 2013
Summary report of the 6th Meeting of the EUCERD
The two-day meeting was the opportunity to tackle a number of priority subjects, including the elaboration of the EUCERD Recommendations on European Reference Networks for Rare Diseases, and the proposal from the European Commission to create an EU platform for rare disease registration. ENERCA participated in the sessions presenting its 10 years of experience.
January 17th 2013
Become a Friend of the Rare Disease Day
On 28 February, the entire rare disease community worldwide will converge to raise awareness for rare diseases and the millions of people affected. ENERCA has already signed up as a "Friend of Rare Disease Day". This initiative has been created to give visibility to those who want to do something to raise awareness for rare diseases. The whole campaign is coordinated by EURORDIS at the international level and the National Alliances of Patient Organisations at the National level.
For patients
Find your center
Know more about Anaemias
Do you have Anaemia?
Create your patient Association Profile
For professionals
Flowchart
MAC
Find colleagues
Create your profile
Subscribe the ENERCA Newsletter
Sign Up »
Previous Newsletters

Flowchart

ENERCA for Professionals

Diagnosis Flowchart

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:

Haemoglobin:	Woman:	120-150 g/L
	Man:	130-170 g/L
Reticulocytes:	Woman/Man:	50-100 x 10^9/L
Mean cell volume:	Woman/Man:	80-100 fL (83-101 fL in Dacie and Lewis' *Practical Haematology*)

Woman ○

Man ○

Haemoglobin (Hb)		g/L
Reticulocytes (Retics)		x10^9/L
Mean cell volume (MCV)		fL

Calculate

ENERCA for Professionals

Anaemia Diagnosis

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Woman ○

Man ◉

Haemoglobin (Hb)	145	g/L
Reticulocytes (Retics)	67	x10^9/L
Mean cell volume (MCV)	89	fL

Calculate

Result

No anaemia has been identified

ENERCA for Professionals

Diagnosis Flowchart

Anaemia Diagnosis

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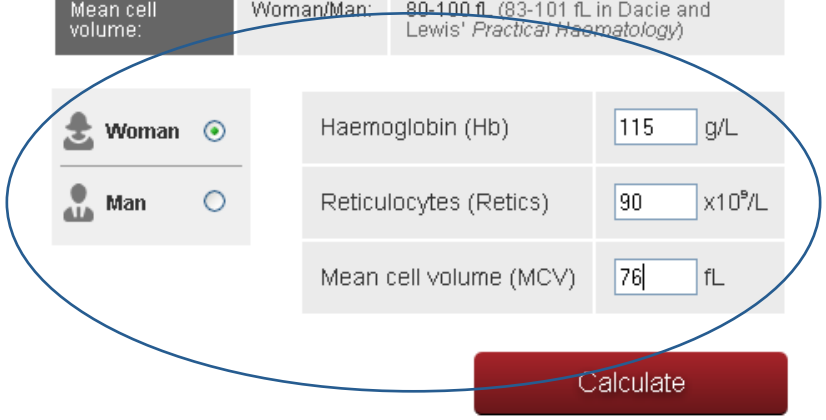

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Diagnosis Flowchart

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Mean cell volume:	Woman/Man:	80-100 fL (83-101 fL in Dacie and Lewis' *Practical Haematology*)

Woman ◉

Man ○

Haemoglobin (Hb)	115	g/L
Reticulocytes (Retics)	90	x10^9/L
Mean cell volume (MCV)	76	fL

Calculate

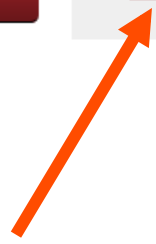
Result

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



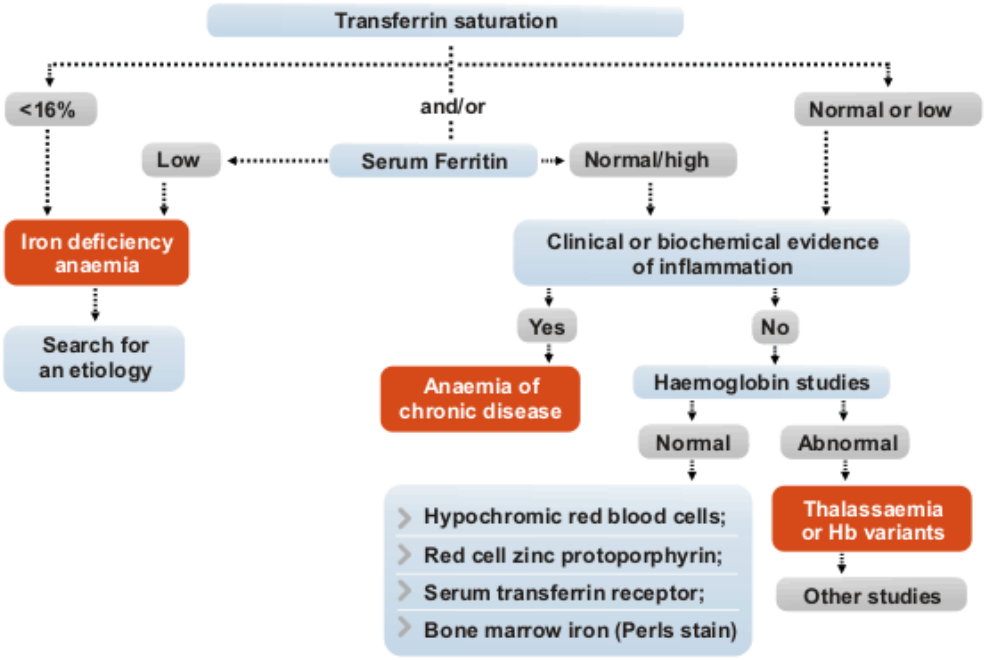
ENERCA for Professionals

Diagnosis Flowchart

Anaemia Diagnosis

Anaemia Diagnosis

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



ENERCA for Professionals

And more…

Free Webcasting trainings courses

ENERCA for Professionals

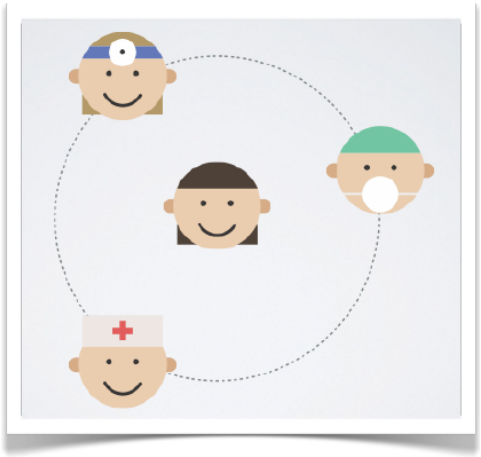
and more…

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



Information & Communication TOOLS

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.

PATIENT | HEALTHCARE PROFESSIONAL | INDUSTRY | PUBLIC AND PRIVATE HEALTH SYSTEM

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**

e-Registry: The challenge of epidemiology in rare anaemias

Some rare anaemias are not so rare: Haemoglobinopathies

- **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.

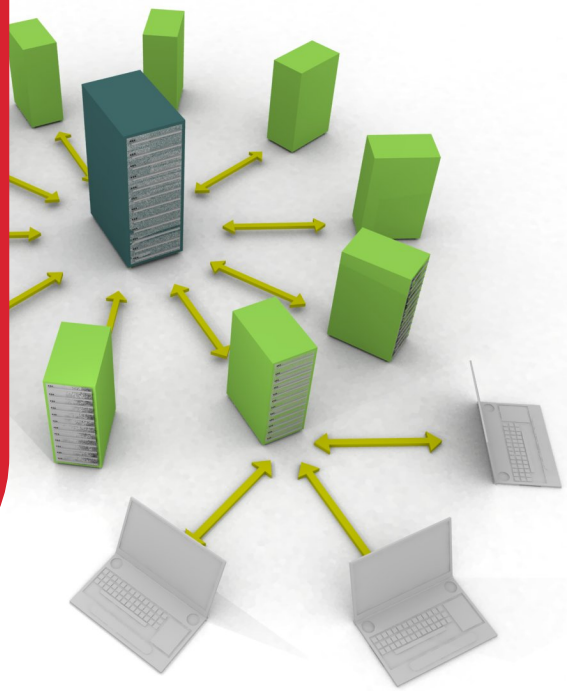
http://www.sxc.hu/photo/1380779

e- ENERCA action for registry

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

Main targets

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



http://www.sxc.hu/photo/1139316

e-Learning: The challenge of knowledge in rare anaemias

e-ENERCA action for education

Patricia Aguilar (CHUM) (France)

Main targets

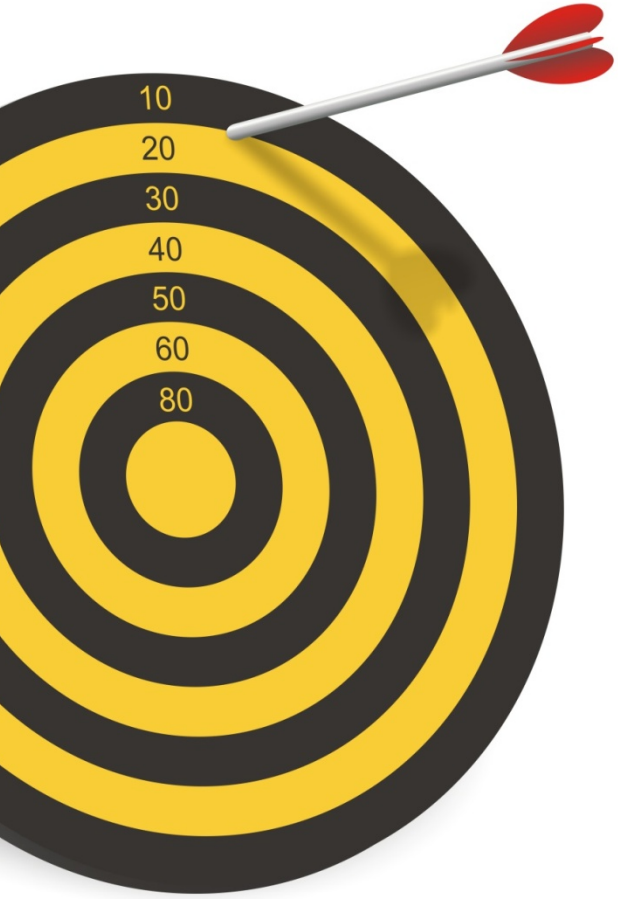
- Harmonization of **medical CV**
- Continuing **medical education**
- On-line modules for **teaching (*)**
- **e-learning** modules:

  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

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

Béatrice Gulbis (ERASME) (Belgium)

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**

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 Comission
- 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

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

- 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

http://www.sxc.hu/photo/469047

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Thank You!!!

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