New e-Health Services for ERN on RAs

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ENERCA nourished by all the concepts and initiatives surrounding rare diseases developed in Europe along its 10 years of life.
Rare Anaemias: How many?

Up to 62 Rare Anaemias:

- ORPHA NUMBER
- ICD-10 CODE
- OMIM

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 SCD, the most frequent is sickle cell anaemia (Hb SS), which is due to haemoglobin S homozygosity. Compound heterozygosity for SCD leads to a more or less severe form: SC, SC-Punjab, SC-Arab, SC-thalassaemia.

What causes the disease and how common is it?

This is a genetic disease. It is linked to a mutation of the α-globin gene, encoding the β-globin chain, one of the components of haemoglobin (Hb). An individual can be heterozygous for the disorder (Hb α-β- individuals) when only one of the globin genes is mutated, or homozygous (Hb SS individuals), or compound heterozygotes HbSβ0, HbSββ0, HbSββα- (thalassaemia individuals) when the two β-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 infested with malaria and Hb S confers a relative protection against malaria.
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 Health Professional**

**Helping the Patient**

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

Recommendations for the recognition of Experts Centres

2013
e-ENERCA: A new challenge

¿Why no create an Interactive Network for the permanent exchange of knowledge and experience?

Second opinión / Interprofessional support

Case Coordinador

Diagnóstic Orientation

Experts information

Diagnosis

Experts

Co-funded by the Health Programme of the European Union

enerca
The e-ENERCA PROJECT: Main Objectives

- Epidemiological surveillance (e-registry)
- Continuing Medical Education (e-learning)
- Telemedicine & Telediagnosis (e-health)
- Networking (CEs and ERN)
- Communication with third parties
- Sustainability
Over 90 health professionals from up to 18 European countries also including Eastern countries

STRATEGY: Identification of expert centres in rare anaemias
e-ENERCA Consortium

HOSPITAL CLINIC DE BARCELONA (Main Beneficiary)
HÔPITAL ERASME
UNIVERSIDAD DEL PAIS VASCO (UPV)
CHU MONTPELLIER (CHUM)
THALASSAEMIA INTERNATIONAL FEDERATION (TIF)

Co-funded by the Health Programme of the European Union
WP1 – Project Coordination
HOSPITAL CLINIC – Joan-Lluis Vives Corrons

WP2 – Project Dissemination
HOSPITAL CLINIC – Joan-Lluis Vives Corrons

WP3 – Project Evaluation
HOSPITAL CLINIC – Joan-Lluis Vives Corrons

WP4 – Epidemiological surveillance for major rare anaemias and e-registry
THALASSEASMA INTERNATIONAL FEDERATION (TIF) - Androulla Eleftheriou and Michael Angastinoitis

WP5 - Continuing medical education (CME) in rare anaemias and e-learning
CENTRE HOSPITALIER UNIVERSITAIRE DE MONTPELLIER (CHUM) - Patricia Aguilar–Martínez

WP6- ENERCA tele-expertise for rare anaemias: tele-medicine & tele-diagnostic
HÔPITAL ERASME - Béatrice Gulbis

WP7 – European Reference Network (ERN) in Rare anaemias sustainability
UNIVERSIDAD DEL PAIS VASCO (UPV) – Carlos Romeo and Pilar Nicolás
Assessment of the entry into force of the Directive 2011/24/EU that recognize the important value of the networking in rare diseases
Expertise share & Research promotion

Tele-diagnosis & Tele-expertise

Research

Clinical Consultations

By Joan Lluís Vives Corrons - Published: 05/22/2012
DIAMOND BLACKFAN ANAEMIA

Comments (0)

By Joan Lluís Vives Corrons - Published: 05/19/2012
ERYTHROCYTE ATP AND HAEMOLYTIC ANAEMIA

Comments (0)

By Silvia Arias Galbis - Published: 04/23/2012
Undiagnosed anaemia

Co-funded by the Health Programme of the European Union
The e-ENERCA background for starting up

- **WHITE BOOK** for the recognition of **Expert Centres** and **ERN** for Rare Anaemias
- **CLASSIFICATION OF RARE ANAEMIAS** (ICD-11-Orphanet-WHO)
- **ENERCA recommendations** on diagnosis and/or therapeutic procedures in Rare Anaemias
- **EXTERNAL QUALITY ASSESSMENT SCHEMAS (EQAS)** for RAs diagnostic tests
- **EUROPEAN REGISTRIES** for Rare Anaemias (Haemoglobinopathies)
- **CONTINUING MEDICAL EDUCATION (CME)**
  - **European Symposia on Rare Anaemias** (Barcelona, Cyprus, Madrid, Sophia, Ferrara)
  - **National Training Courses** (Portugal, Belgium, France)
- **CONTRIBUTION TO HEALTH POLICIES**: **Policy report** on Perspectives of health and migration on haemoglobinopathies
- **ENERCA WEB (Dissemination and ICT incorporation)**
  - Catalogue of centres, health professionals and patients associations of RAs
  - Rare Anaemias list (62) including codification (ORPHA, MIM and ICD)
  - Educational material: Video on haemoglobinopathies & comics
  - New Information Communication Technologies (TIC) implementation (**e-Health**)
THE EUROPEAN SYMPOSIA ON RARE ANAEMIAS
A policy report on Health and Migration - Haemoglobinopathies -

A policy overview of haemoglobinopathies across ten EU member states (the EU, Belgium, Cyprus, France, Germany, Greece, Italy, The Netherlands, Spain, Sweden and the United Kingdom) and underlines the existing challenges, best practices and policies facing patients with haemoglobin disorders.

To draw attention to the lack of awareness of haemoglobinopathies among the public and healthcare practitioners, highlights the importance of consistent and comparable data and emphasises that both traditional and more recent migration and mobility flows are posing increasing challenges to European healthcare systems that are tackling the prevention, diagnosis and treatment of these diseases and patients’ needs.

Developed by a group of experts working with the European Network for Rare and Congenital Anaemias (ENERCA) and the Thalassaemia International Federation (TIF), in collaboration with the International Organization for Migration (IOM), Migration Health Division, Regional Office Brussels.

Co-funded by the Health Programme of the European Union.
OFFICIAL PRESENTATION of the Policy Report on Health and Migration –Haemoglobinopathies-
European Parliament, Brussels. Wednesday, 26 June 2013
The ENERCA Web

Welcome

ENERCA means easy access to high-quality information on rare anemias for patients, citizens, health professionals, stakeholders, authorities and pharmaceutical industry.

www.enerca.org

E-Health Implementation

- To endorse the new e-health platforms
  - Cross links between ENERCA and the new platforms
  - Interoperability, expandability and remote access

- To promote Smart Phones applications

- To connect with social networks: Facebook, Twitter...

ENERCA for Patients

- To promote activities with Patient’s Associations
- ENERCA Help-line for second consultations
- Publish interactive patients-professionals meetings

ENERCA for Professionals

- Help to diagnosis by anaemia flowcharts
- Provide information through ENERCA Help-line
- Facilitate experts interaction (Diagnosis & Research)

Co-funded by the Health Programme of the European Union
Communication and interaction with third parties

European Union Committee of Experts on Rare Diseases (EUCERD)

- Stakeholder target groups
  - ICD classification for RA
  - WEBSITE crosslinks

- Joint programmes on:
  - Genetics
  - Newborn screening

- Patients’ Associations links
- Educational material

- Help-line

- Research

Co-funded by the Health Programme of the European Union
ENERCA contribution to Research

ENERCA is collaborating with the following research projects:

1. **RD-CONNECT**: An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. FP7 HEALTH.2012.2.1.1-1-C: Coordinator: Hanns Lochmüller

2. **RARE-BEST PRACTICES**: Best practice and knowledge sharing in the clinical management of rare diseases. FP7-HEALTH.2012.2.4.4-3-INNOVATION-1 Coordinator: Domenica Taruscio

3. **SPAIN-RDR**: Spanish Rare Disease Registries Research Network. A collaborative research joint project proposals on RD Patients´ Registries, serving as core the Institute for Rare Diseases Research (IIER) and the cooperation of the Units of the Autonomous Communities of Spain Coordinator: Manuel Posada de la Paz
Combined Molecular Microscopy for Therapy and Personalised Medication in Rare Anaemia Treatments

CoMMiTMenT

HEALTH.2013.1.2-1: Development of imaging technologies for therapeutic interventions in rare diseases
FP7-HEALTH-2013-INNOVATION-1

Coordinator: Lars Kaestner

CoMMiTMenT will allow for the functional identification of channelopathies on a molecular level, i.e. the identification of malfunctioning ion channels or transporters that cause the disease or the associated symptoms

Kick-off meeting
09.10.2013

WP5- Treatment concepts for personalized medicine
Dr. Joan Lluis Vives Corrons
IDIBAPS
e-ENERCA added value

✓ Consolidation of the European Reference Network (ERN) in Rare Anaemias (ERN-RA) with effective implementation of the objectives of the Directive 2011/24/EU

✓ Development of the new Information Communication Technologies (ICT) endorsed in the ENERCA website to facilitate the communities to share their expertise without physically travelling (tele-expertise) and to implement best clinical and laboratory practices (e-health tools)

✓ Official recognition of ERN in RAs by the European Commission