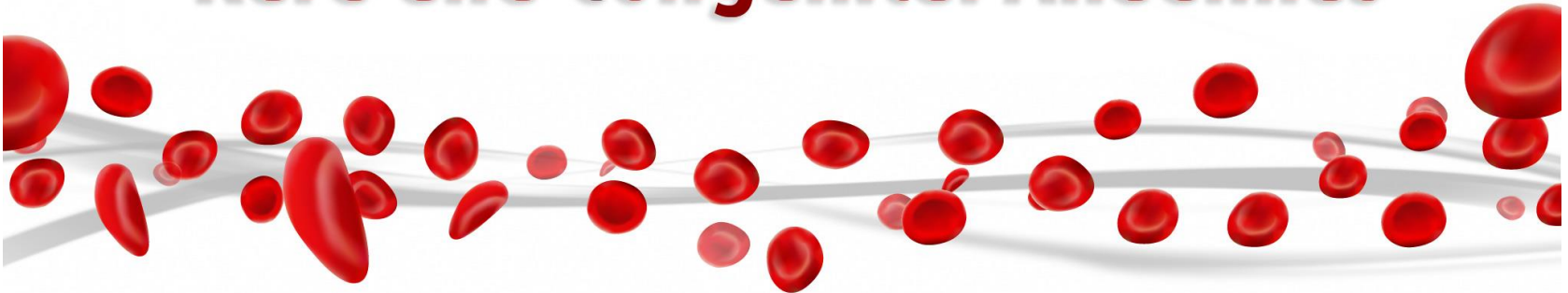


# European Network for Rare and Congenital Anaemias



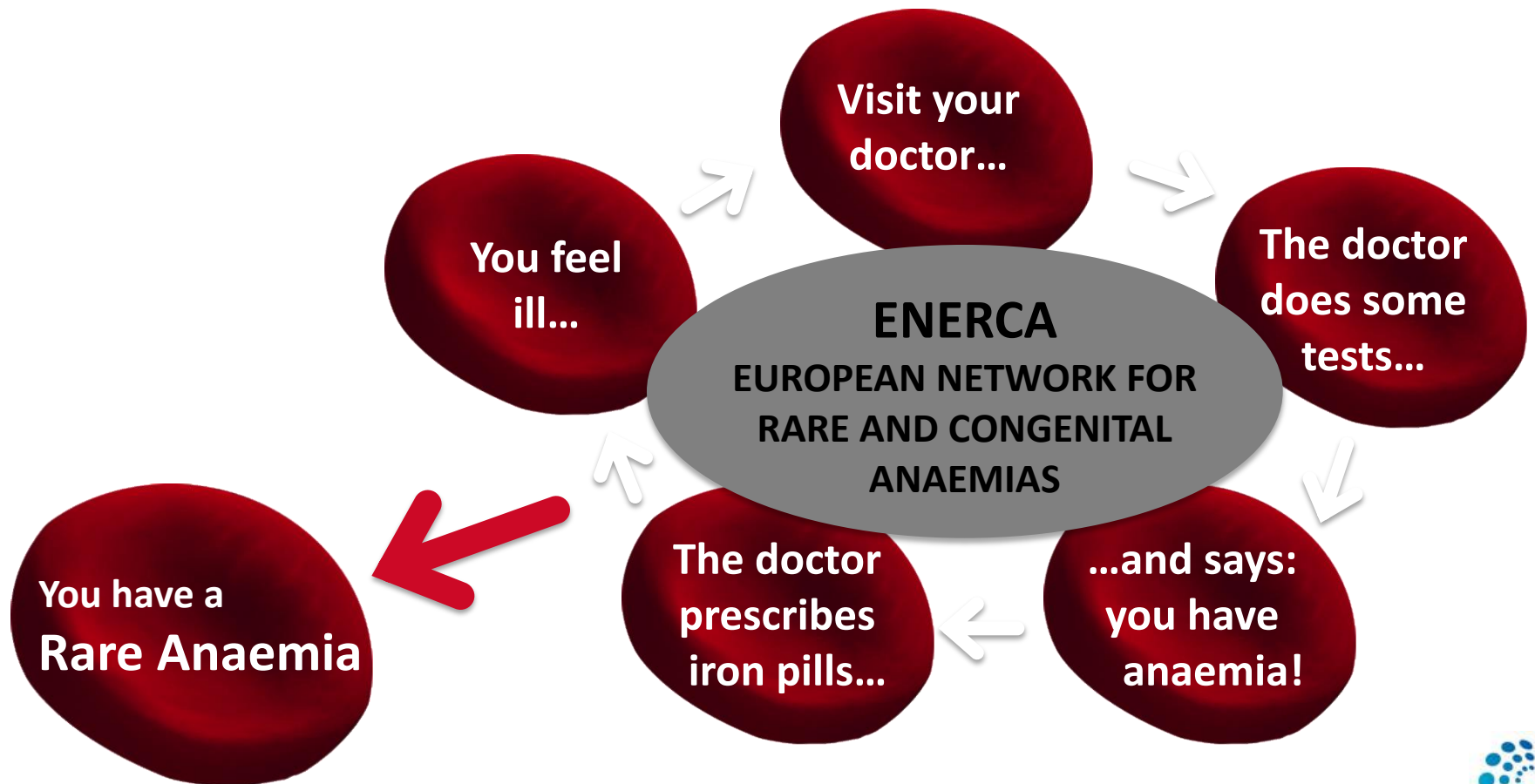
**How can ENERCA become a European Reference Network?**

Joan-Lluís Vives Corrons  
Lisbon, 9 October 2015

enerca

# The clinical challenge

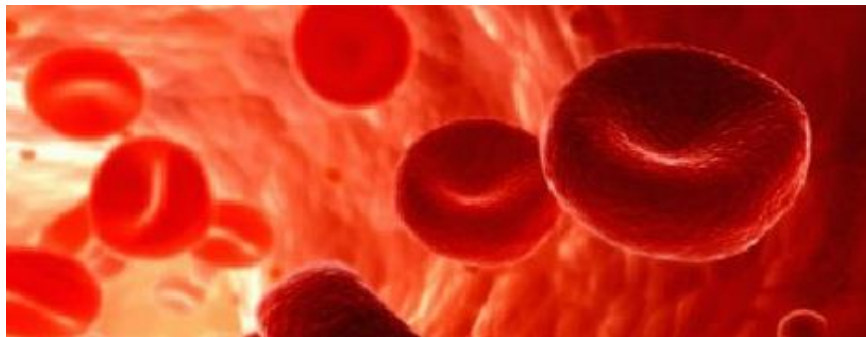
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# WHAT IS A RARE ANAEMIA ?

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1. **KEY CLINICAL SIGN** IN MORE THAN 62 RARE DISEASES
2. **HEREDITARY ORIGIN** IN MORE THAN 90% OF PATIENTS
3. **UNKNOWN ORIGIN** IN MORE THAN 10% OF PATIENTS



# THE RARE ANAEMIAS IN THE CONTEXT OF RARE DISEASES

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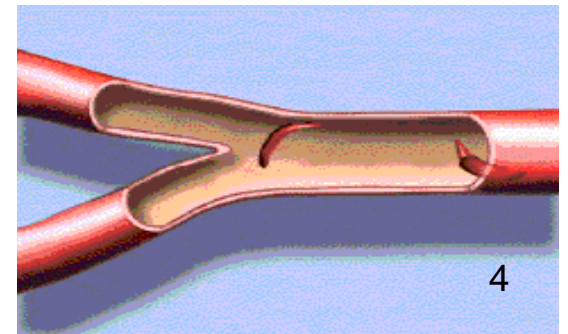
**MORE THAN 500.000 CHILDREN BORN ,EVERY YEAR , WITH A RARE ANAEMIA**

THE MOST FREQUENT ARE :

1. Thalassaemia (Mediterranean Anaemia)
2. Sickle Cell Disorders (Migration Impact)

## MAIN CLINICAL IMPLICATIONS

- **TREATMENT (PALIATIVE) → To reduce the severity of anaemia**  
Blood Transfusions and Iron chelating
- **PREVENTION → To reduce mortality and morbidity**  
Newborn screening



# RARE ANAEMIAS IN THE CONTEXT OF HAEMATOLOGICAL DISEASES

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1. ALMOST ALL HAEMATOLOGICAL DISEASES ARE RARE

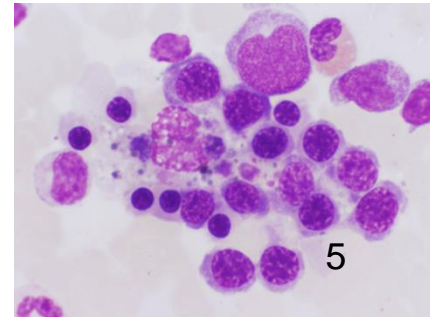
2. THE TARGET ORGAN IS THE HAEMATOPOIETIC SYSTEM

- Leukaemias, lymphomas and related neoplasms
- Bleeding & Coagulation disorders
- Anaemias and erythropoietic related disorders
- White blood cell disorders leading to immune dysfunction

3. SECONDARY RARE ANAEMIAS

**Anaemias present as key clinical manifestation in non-haematological rare diseases:**

- Atypical Haemolytic Uremic syndrome (aHUS)
- Chronic kidney anaemia (CKA)
- Chronic liver disease (CLD)
- Hypothyroidism
- Inflammatory anaemia
- Other



# RARE ANAEMIAS IN THE CONTEXT OF HAEMATOLOGICAL DISEASES

## THE ACTION PLAN FOR ERN

- 1. RARE ANAEMIAS AND RED BLOOD CELL DISORDERS**
- 2. RARE BLEEDING & COAGULATION DISORDERS** belong to EUHANET
- 3. RARE LEUKOCYTE DISORDERS** belong to Rare Immunological and Autoinflammatory diseases
- 4. RARE MALIGNANT HAEMATOLOGICAL DISORDERS** belong to oncological ERNs

A list of rare diseases included in the [ORPHA code for Rare Haematological Diseases](#) has been already analysed and split into different categories to be covered by the ERN. Accordingly, two different ERNs in Rare Haematological Disorders are suggested:

**1. ERN in Rare Anaemias and Red Blood Cell disorders (non-oncological)**

Rare Anemias (ORPHA108997)

Poliglobulia (ORPHA98427)

**2. ERN in Rare Bleeding and Coagulation diseases (non-oncological )**

Rare coagulation disorder (ORPHA98429)

Twin to twin transfusion syndrome (ORPHA95431)

Congenital analbuminemia (ORPHA86816)

# RARE ANAEMIAS IN THE CONTEXT OF HAEMATOLOGICAL DISEASES

## THE ACTION PLAN FOR ERN

### PATIENTS INVOLVEMENT

Patients Associations will participate through the **Thalassaemia International Federation (TIF)**, one of the main ENERCA partners.

TIF is already part of ENERCA executive committee since 2008

Other Rare Anaemias and Related disorders patient's Associations will join in the next future

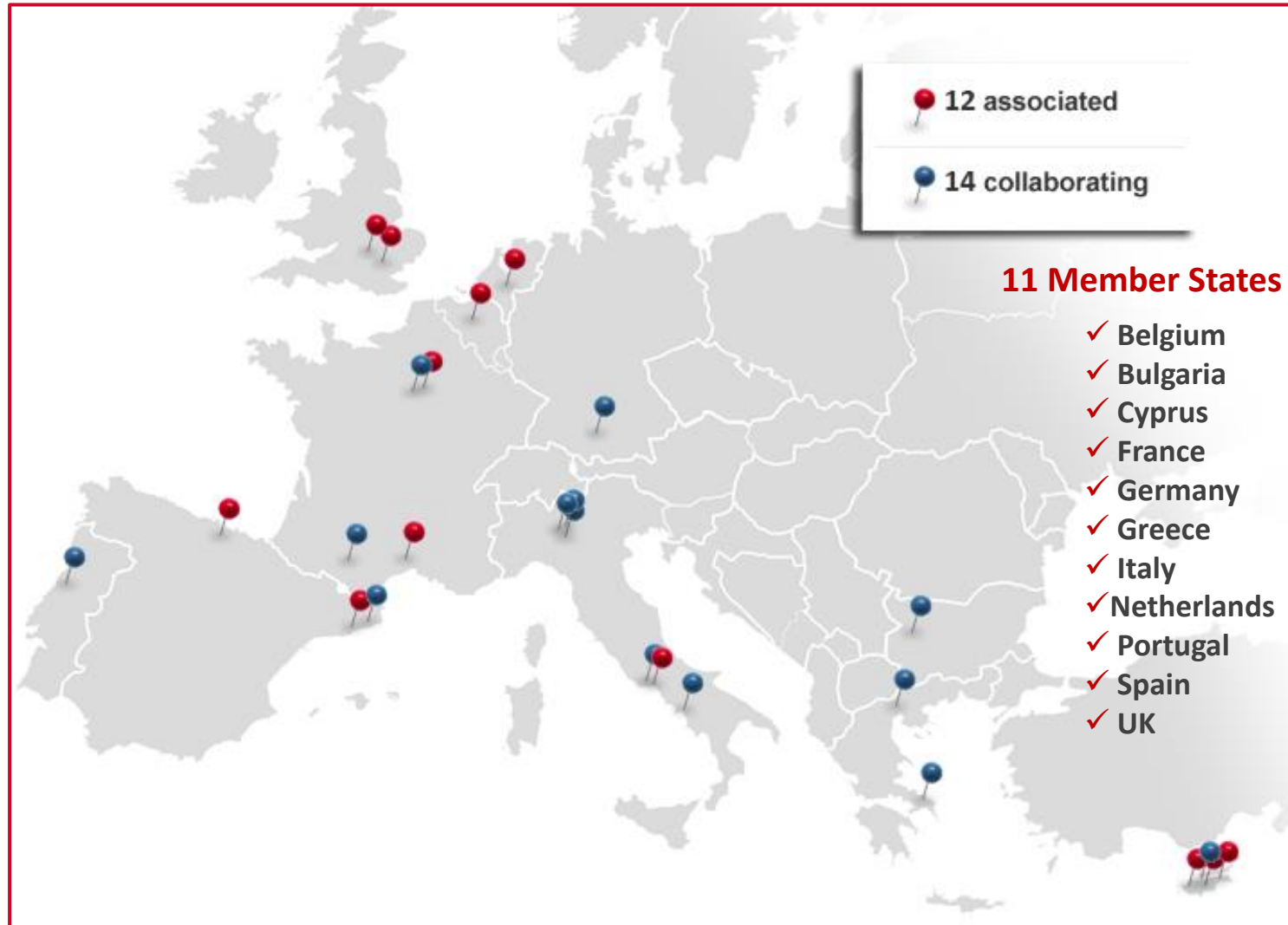
### EUROPEAN HAEMATOLOGY ASSOCIATION (EHA)

Collaboration between **EHA** and ENERCA for the ERN consolidation is currently analysed

# EUROPEAN NETWORK FOR RARE AND CONGENITAL ANAEMIAS



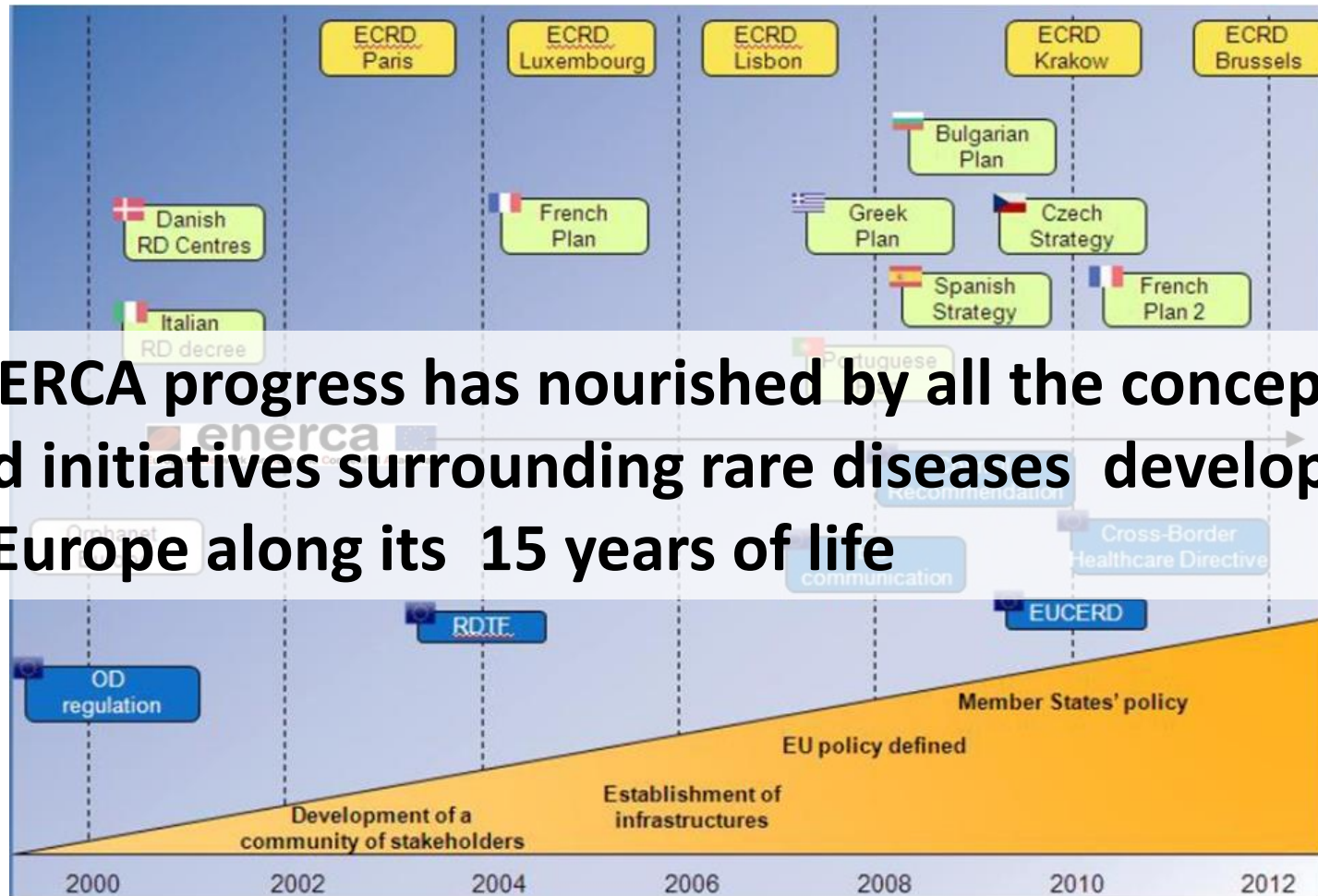
2013-2016





# THE BASIS OF ENERCA DEVELOPMENT (2002-2016)

ENERCA progress has nourished by all the concepts and initiatives surrounding rare diseases developed in Europe along its 15 years of life



2016



# THE FUNDAMENTAL OF ENERCA PROGRESS (FOUR PHASES OR INDIVIDUAL PROJECTS)

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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 & acquired anaemias**

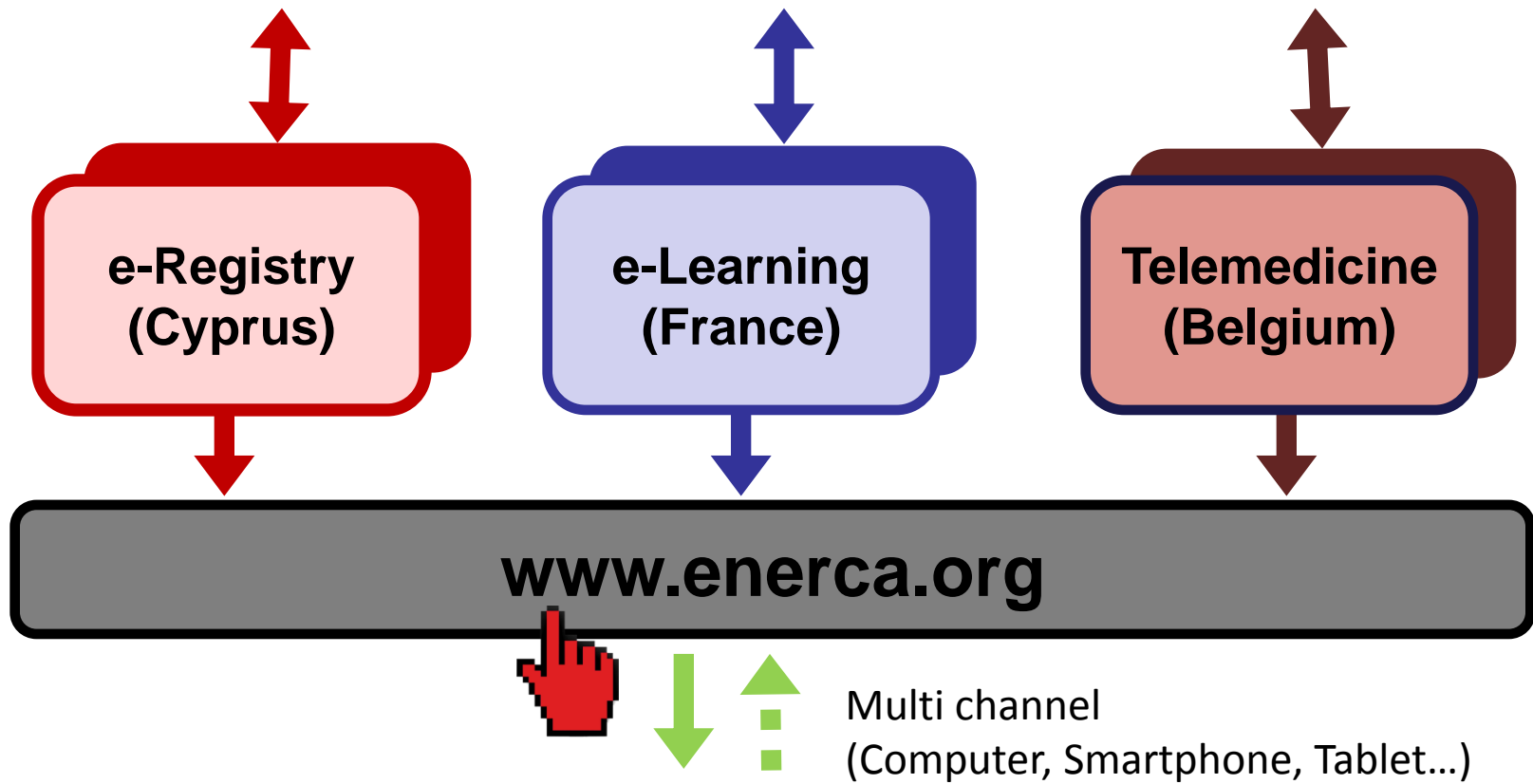
## Phase III (2009 – 2012)

- @ Guidelines
- @ Social awareness
- @ **Patient's empowerment**
- @ **White Book**

# PHASE IV (2013-2016)

## ICT Platforms development (@-ENERCA)

### Consortium & members



Health professionals, Health authorities .... **Patients**

# THE CHALLENGES OF ENERCA

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This is not  
an easy job!

@ **Improve** the existing poor implementation of comprehensive data collection for rare anaemias

→ e-Registry

@ **Harmonise** the existing variability in diagnosis practices, clinical care and prevention of rare anaemias in the EU

→ e-Learning

@ **Decrease** the existing inequalities for the access to high quality health care in rare anaemias between the different European Countries

→ Telemedicine

# THE NEXT ENERCA CHALLENGE

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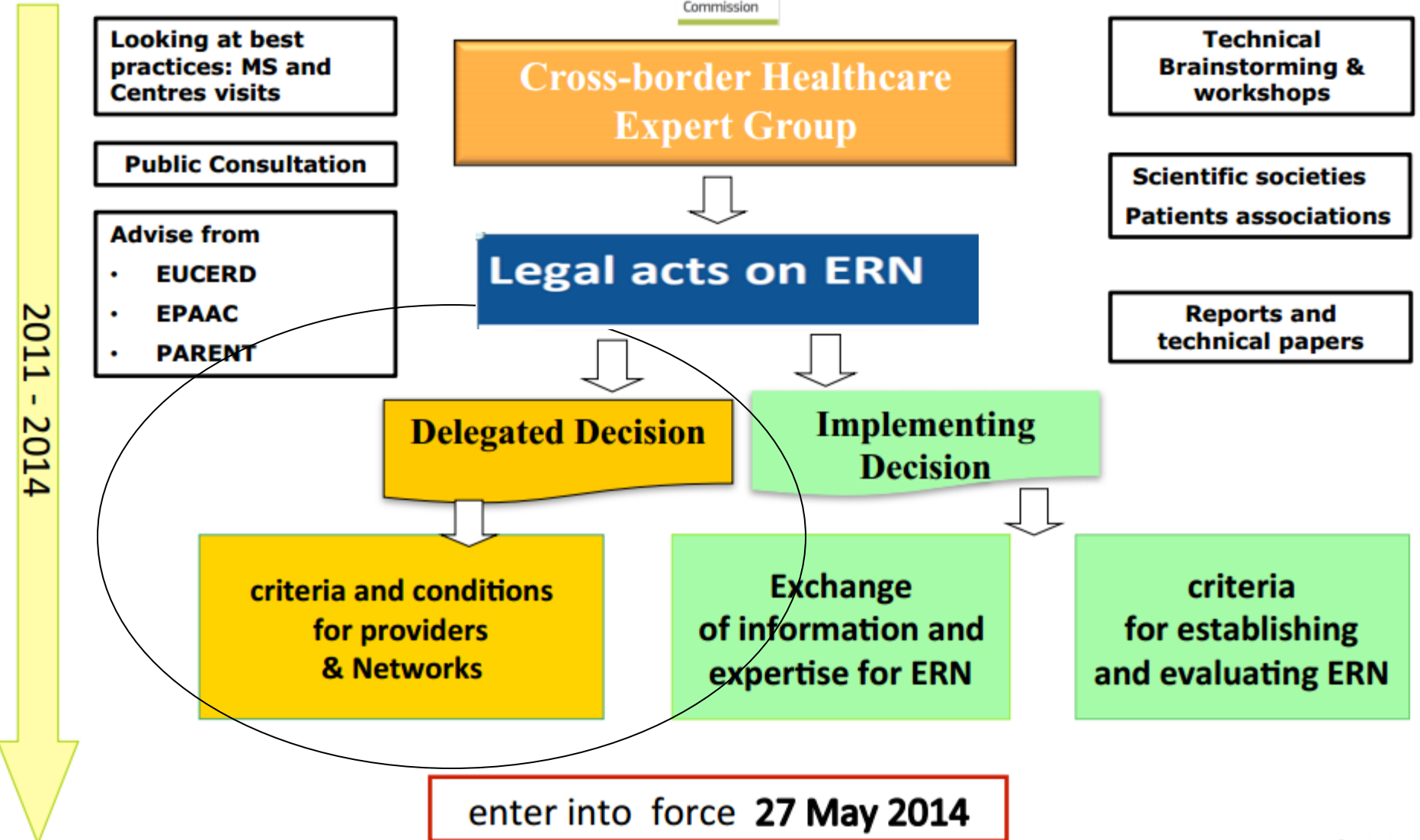
To become an **European Reference Network (ERN)** on Rare Anaemias

Analyse the **impact of the entry into force of the Directive 2011/24/EU on the application of patients' rights in cross-border healthcare.**



**SUSTAINABILITY**

**European Reference Networks (ERN): aim of Article 12:  
(Directive Patient's Rights to Cross border Healthcare )**



[http://ec.europa.eu/health/ern/docs/ern\\_delegateddecision\\_20140310\\_en.pdf](http://ec.europa.eu/health/ern/docs/ern_delegateddecision_20140310_en.pdf)

[http://ec.europa.eu/health/ern/docs/ern\\_implementingdecision\\_20140310\\_en.pdf](http://ec.europa.eu/health/ern/docs/ern_implementingdecision_20140310_en.pdf)

## BASIC REQUIREMENTS

### ➤ 10 Members in at least 8 Countries of the European Union

**MEMBER** : Health care provider or CENTRE providing diagnosis, prevention and treatment

**MEMBERSHIP APPLICATION** : Individual CENTRE application

Requires a written statement from the **Member State (MS)** certifying that the centre accomplishes with the *eligibility criteria for centres of expertise prepared by their respective National Plan or Strategy for rare diseases, as part of MS National Legislation*

**MEMBERSHIP CERTIFICATION** : This is a *certificate provided by each MS* for MEMBERS  
If a MS cannot designate MEMBERS in its own Country, associated and/or collaborative national centres have to be designated and encourage them to cooperate with the ERN

**Associated National Centres** . Healthcare providers who do not fulfil the required criteria

**Collaborative National Centres**. University and Research Centres providing knowledge and quality care tools

**In Spain**: To be part of a **ERN in rare diseases** , the CENTRE has to be previously certified by the National “Ministerio de Sanidad, Seguridad Social e Igualdad (MSSSI) as National Centre of Expertise called **CSUR** (Centres, services and Units of Reference) – On October 29, 2015, there is a meeting at the MSSSI where considerations will be established for the recognition of rare anaemias as rare haematological disorders.

## BASIC REQUIREMENTS

### ➤ 3 Objectives (at least), out of 8 Objectives are required

- 1) To facilitate **EUROPEAN COOPERATION** on highly specialised healthcare systems
- 2) To contribute to the **POOLING OF KNOWLEDGE**
- 3) To facilitate **CONCENTRATION OF EXPERTISE** for providing high-quality healthcare
- 4) To maximise **COST-EFFECTIVENESS** by concentrating resources where appropriate
- 5) To reinforce **RESEARCH, EPIDEMIOLOGICAL SURVEILLANCE AND TRAINING**
- 6) To facilitate **MOBILITY OF EXPERTISE** to develop and share knowledge and best practices
- 7) To encourage the **DEVELOPMENT OF QUALITY AND SAFETY BENCHMARKS**
- 8) To provide **ACCESS TO HEALTH SERVICES** for all patients with RD wherever they live



## EXAMPLE: TWO BASIC REQUIREMENTS TAKEN FROM ENERCA

### ➤ Objective 4: MAXIMISE THE COST-EFFECTIVE USE OF RESOURCES

- a) e-ENERCA WP4  
**e-registry** platform of patients for shaping policies addressing **specific needs** to concentrate resources
- b) ENERCA WITE BOOK  
**Centers of Expertise (CoE)** recommendations on technical, ethical and patient's expectations



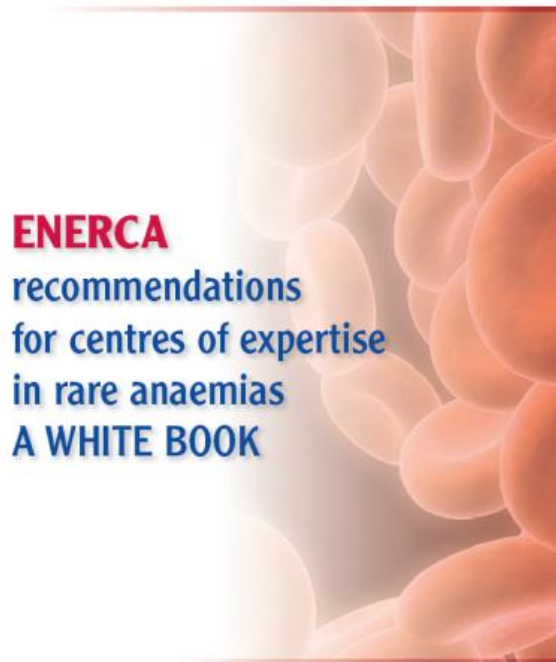
### ➤ Objective 6 : FACILITATE MOBILITY OF EXPERTISE

- a) ENERCA 3 :  
**Good Practices (GP)** recommendations on diagnosis, prevention and clinical management
- b) e-ENERCA WP5:  
**e-learning platform** for continuous medical education – Courses and Symposia
- c) e-ENERCA WP6:  
**Telemedicine platform** for sharing knowledge ,expertise, and clinical management



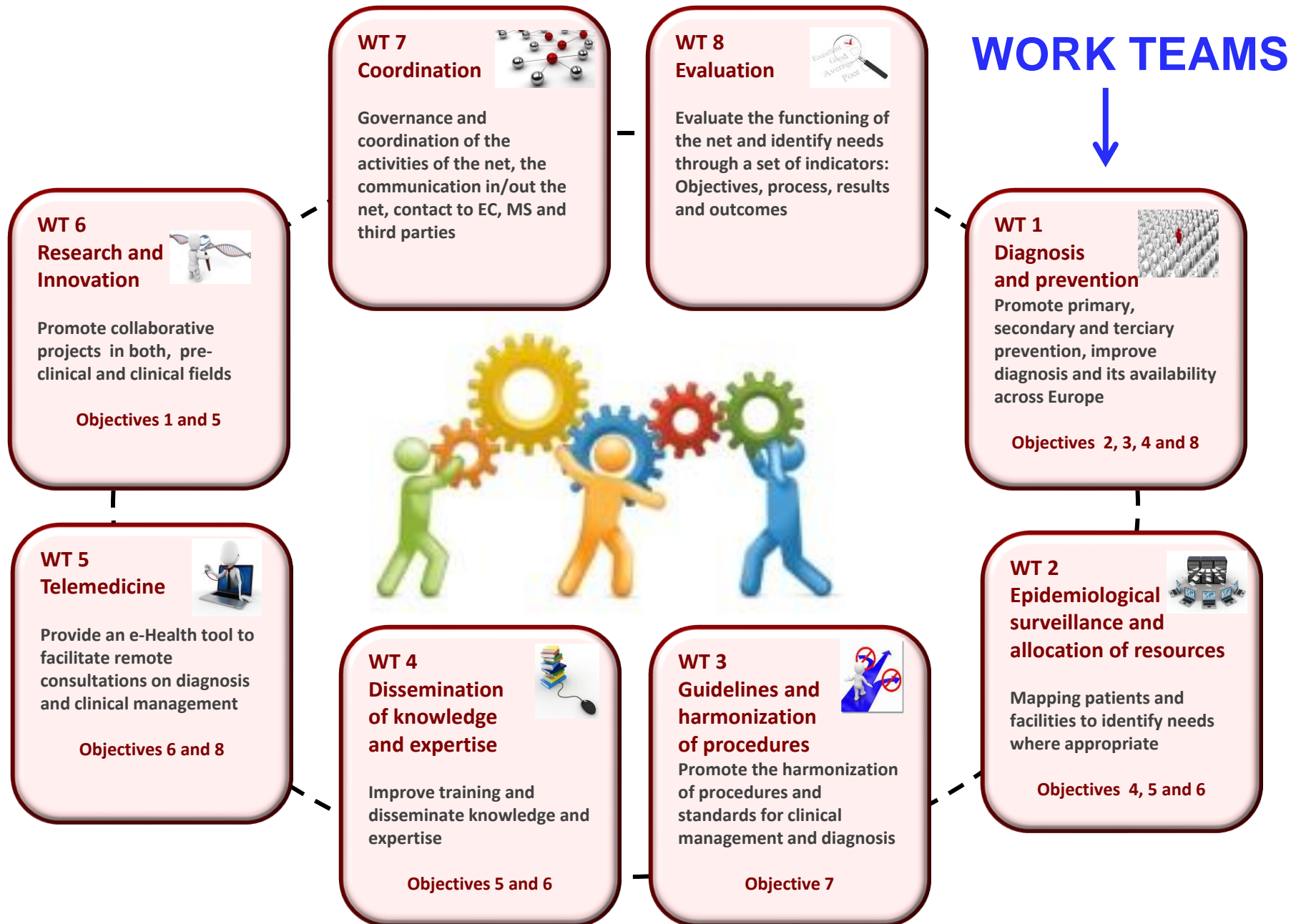


# ENERCA WHITE BOOK

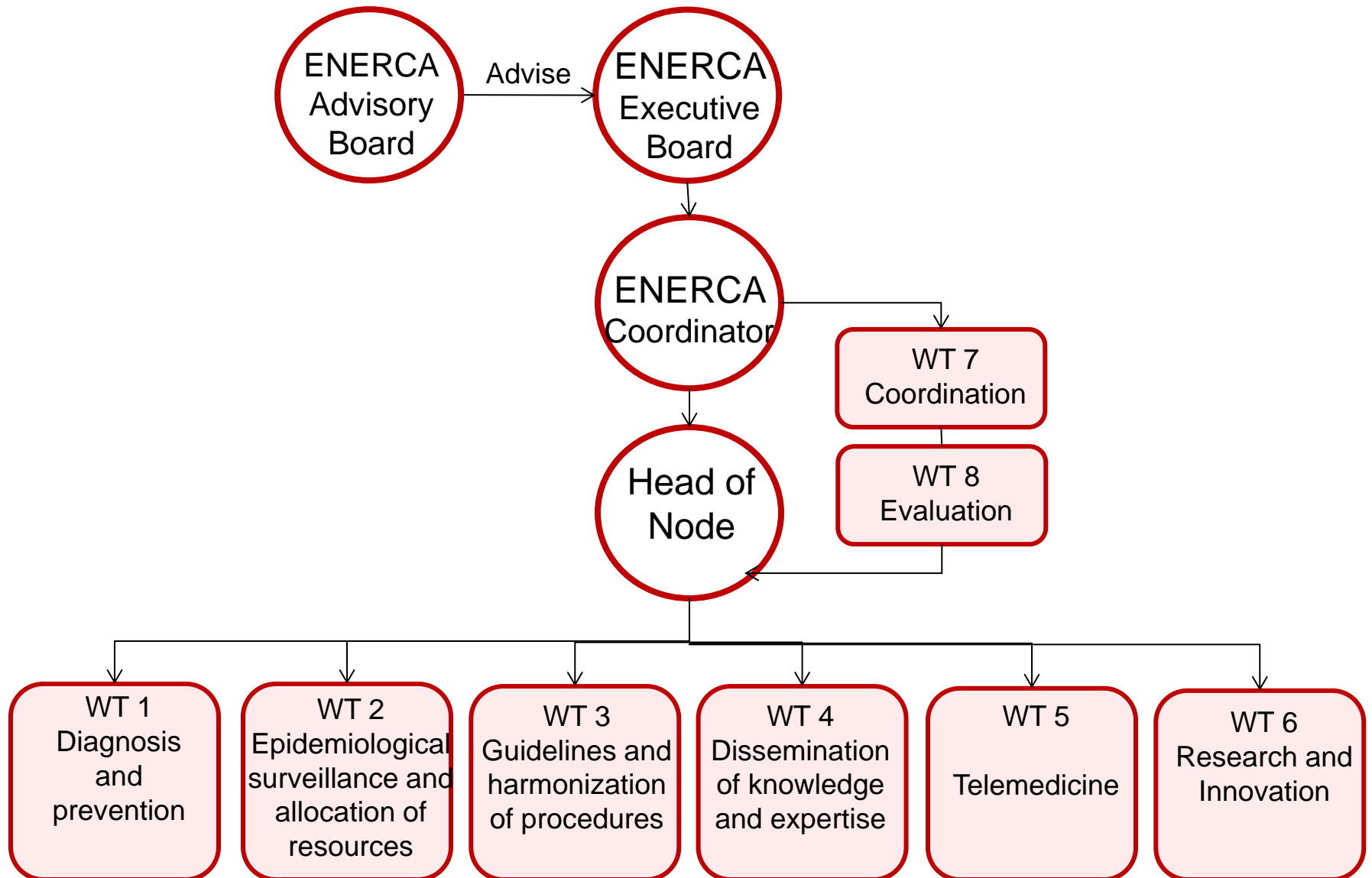


- A **position paper** and the most important ENERCA outcome after 10 years of experience for the creation of a **European Reference Network (ERN)** in rare anaemias
- It contains the **essential specific criteria** to be considered in the process of identification and recognition of **Centres of Expertise** as healthcare providers.
- Centres of Expertise will be the **nodes** of the future ERN, that will act as a **focal point** for information, medical training, research, and dissemination of rare anaemias

# ERN FOR RARE ANAEMIAS - ORGANISATION CHART



# ENERCA ERN Governance



# ENERCA Coordination Team

Head of Project – Joan-Lluís Vives Corrons ([jl.vives@clinic.ub.es.org](mailto:jl.vives@clinic.ub.es.org))  
Project Manager - María del Mar Mañú Pereira ([maria.manu@enerca.org](mailto:maria.manu@enerca.org))  
Project Web manager- Victoria Gutiérrez ([victoria.gutierrez@enerca.org](mailto:victoria.gutierrez@enerca.org))  
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