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Enerca: The European Network for Patients with Rare Anaemias

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Summary

ENERCA intends to promote a multidisciplinary approach to care in order to address the complex and diverse conditions implied in the group of rare diseases called Rare Anaemias. Gathering expertise at European level is therefore paramount to ensure equal access to accurate information, appropriate and timely diagnosis and high quality care for patients with a rare anaemia independent from their Country of origin. Moreover, increasing Healthy Life Years by preventing disease and promoting policies that lead to a healthier way of life is important for the well-being of EU citizens and helps to meet the challenges of the Lisbon decision as regards the knowledge society and the sustainability of public finances, which are under pressure from rising health care and social security. In this way, the consolidation of a European Reference Network (ERN) of Experts Centers in Rare Anaemias, is a crucial step to improve the services for clinical management of these diseasesas well as education and social care regardless of their country of origin. This will allow health professionals an easy access to recognized centres of expertise in each RA category and provide new opportunities to undertake innovative and useful actions based on the ERN.

What Are Rare Anaemias?

Rare Anaemias (RA) are a group of rare diseases (RD), in which anaemia are the key clinical manifestation. As all RD the RA have prevalence in Europe of less than 5 cases for 10,000 people. The existence of RA is an exciting tackling exercise for clinical and biological research, and the need of improving the quality of health services for these patients has obliged to mobilise resources that can only be efficient if done in a coordinated European multi centre and multi-disciplinary way. This is due because the RA are a complex group of RD, and according to their mechanism, prevalence and/or relevant clinical and/or social impact in the European population, they have been classified into ten main groups (Table 1). From these, up to 80% are hereditary, and in, the remaining 20%, the underlying cause of RA is acquired, or, remains unknown. Probably, almost one third of RA with unknown origin might be accounted for myelodysplastic syndromes (MDS) or to complex clinical situations with multifactorial mechanisms, in general associated with systemic, non haematological, hereditary or acquired diseases [1].

What Is Enerca?

The European Commission (EC) has been especially sensible to the rare diseases challenge, and, since 2002, has taken an active role by co-financing a large-scale network of experts and specialists working in the field of RA, called EUROPEAN NETWORK FOR RARE AND CONGENITAL ANAEMIAS (ENERCA). ENERCA has been developed through four consecutive phases with a total duration on 15 years. In all these phase developing ENERCA Projects, consecutive achievements have been nourished by all the concepts and initiatives that continuously appear in the discussion platforms; Rare Disease Task Force (RDTF) from 2004 to 2009, and European Committee of Experts in Rare Diseases (EUCERD) from 2010 to 2014. (Figure 1). ENERCA involves associated and collaborating partners from 15 different European countries (Figure 2). and most of they have been working together since 2002, . All ENERCA partners are well known and recognized experts in their respective field.

Table 1: Classification of Rare Anaemias and most significative examples. (Within brackets the expected prevalence is indicated).

GROUP I	Haemoglobin defects (1-2 cases /10,000) Structural Haemoglobinopathies (i.e. Sickle-cell anaemia) Thalassaemias (i.e. Mediterranean anaemia)
GROUP II	Hereditary Haemolytic anaemias (1-5 cases/50,000) Red blood cell Enzymopathies (i.e. Pyruvate kinase deficiency) Red blood cell membranopathies (i.e. Hereditary spherocytosis)
GROUP III	Erythropoietic failure (2-5 cases/1,000,000) Congenital : Diamond-Blackfan anaemia (DBA) and FanconiAnaemia (FA) Acquired : Selective Erytroblastopenia and aplastic anaemia
GROUP IV	Dyserythropoietic anaemias (4-7 cases/1,000,000) Congenital (i.e. CDA type I, type II , type III) Acquired (Myelodysplastic syndromes and other)
GROUP V	Sideroblastic anaemias (3-5 cases/1,000,000) Congenital (Congenital sideroblastic anaemias) Acquired (Secondary sideroblastic anaemia)
GROUP VI	Non sideroblastic anaemias due to hereditary iron defects (unknown) Iron Refractory Iron Deficiency Anaemia (IRIDA) Other defects leading to microcytic anaemias
GROUP VII	Hereditary disorders of folic acid and cobalamin defects (unknown)
GROUP VIII	Paroxysmal nocturnal haemoglobinuria (PNH) (1-5 cases/1,000,000),
GROUP IX	Anaemias due to rare complex mechanisms
GROUP X	Anaemias of unknown origin (AUO)

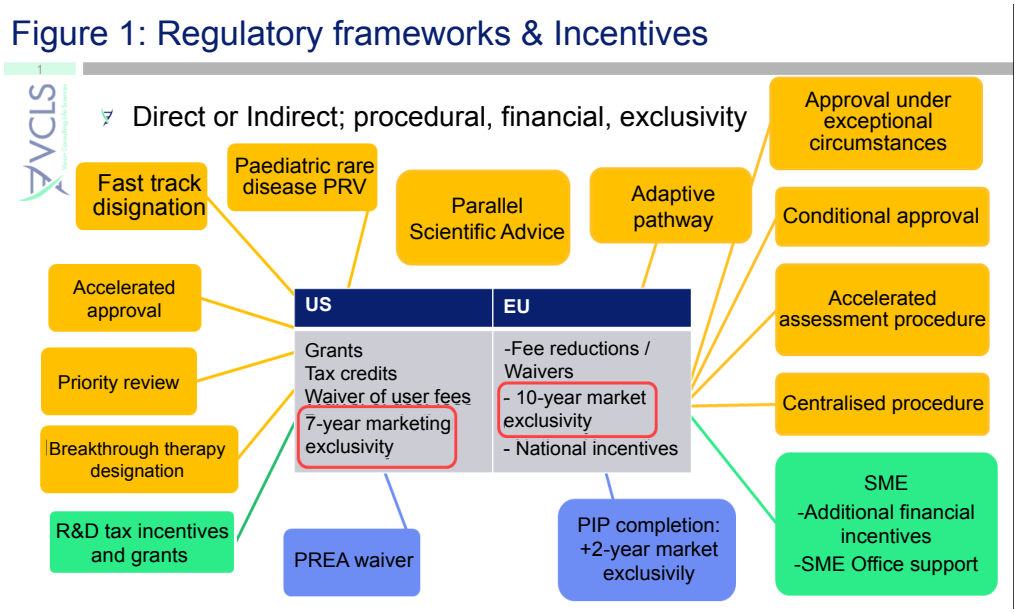

Figure 1

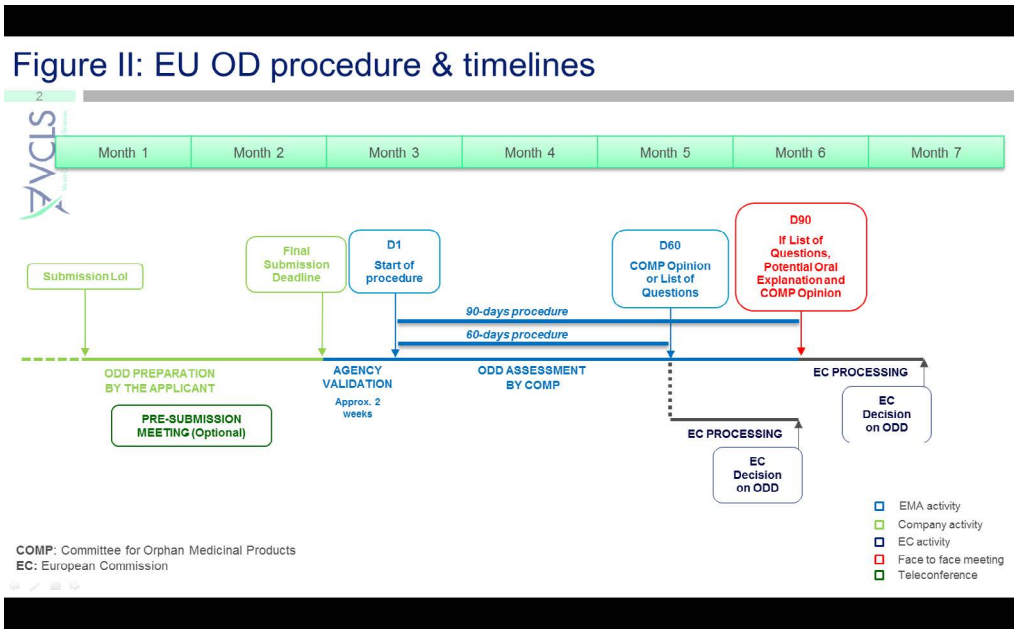

Figure 2

Health Services Provided By Enerca and Policy

During the last 10 years (2002-2012), ENERCA has contributed to promote two pivotal aspects for RA: 1) A specific framework for cross-border healthcare and 2) A European cooperation for providing health services such as diagnostic help, training, information, dissemination and evaluation. All these ENERCA outcomes have had a wide geographical coverage and an efficient impact as a high number of health professionals and patients were implicated. Obviously, ENERCA website (www.enerca.org) has definitely contributed to this success that allows to present, actually, a definitive European added value, as stated in the EU Health Programme 2008-2013 (“common principles in all EU health systems aiming to ensure clarity and confidence with regard to authorities setting and monitoring healthcare standards, have to be implemented”) and proven to be a focal point for the following health services implementation One example of this contribution is the publication of ENERCA White Book [2]

After 2013, the EC has approved co-financing ENERCA Project for a additional tree years (2014-2017), with the aim of developing and implementing the new e-health information and communication technologies (ICT) for assuring the same access to health services in RAs across Europe, independently from the place of residence. This new Project, called e-ENERCA, is based, in part, on previous ENERCA projects achievements, but adapted to the "2011 EU Directive on patients’ rights in cross-border healthcare". e-ENERCA will incorporate the most innovative e-health ICT to create a pan-European interoperable e-health platform for, electronic registry/epidemiological health records (EHR), on-line teaching programes for continuous medical education , and tele-expertise/telediagnosis for medical practice. Accordingly, the and e-learning. e-Health services will be developed through the set-up of three different e-platforms endorsed by ENERCA website (www.enerca.org) : 1) e-Registry, a Pan European registry of RAs for gathering patient’s data necessary to achieve the required sample size for epidemiological surveillance and clinical research 2) e-Learning , a teaching platform for the dissemination of knowledge ,continuous medical education, and best practices awareness and promotion through Internet, and 3) e-Medicine , a platform to provide, at distance, expertise (telexpertise) and diagnostic facilities (telediagnosis) , avoiding , when possible, the need of travelling. The ERN will identify needs and priorities for basic, clinical translational and social research in the fields of the different RA categories: a) Sickle cell disease (SCD), b) Thalassaemia syndromes and d) Very Rare Anaemias (e.g. erythropoietic failure and RBC defects, either hereditary or acquired). This will facilitate ENERCA fostering all these activities and the promotion of interdisciplinary cooperative approaches to the complementarily addressed through national MS and Community programs. One example is sickle cell diseases (SCD).

During the last 30 years, SCD is increasing in Europe due to African immigration, leading to an important impact on health care burden in several countries. Preventive programs, aiming to epidemiological control, and improvement of diagnosis and clinical management of major RA, are crucial for decreasing the

affected birth rate and achieving an efficient balance between morbidity and patient's life expectancy [3]. Accordingly, ENERCA has taken an active role for improving this situation by the following actions : a) the identification of Centres of Expertise on RAs in Europe according to the recommendations of ENERCA White Book b) the promotion of best clinical and laboratory practices and, c) the improving of continuous medical education, by organising topic-specific training courses, workshops and symposia , e) the empowerment of patients, by cooperation with Patient's Associations, and co-organizing a bi-annual European Symposium on RAs with interactive patients-health professionals sessions.

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3 ENERCA: A White Book- Prodrug Multimedia (2014) Recommendations for Centres of Expertise in Rare Anaemias.