Expert services for rare anaemias across Europe

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Introduction

New challenges and priorities are given in the EU Health programme 2007-2013. The objectives of the programme are to improve citizens' health security, to promote health to improve prosperity and solidarity, and to generate and disseminate health knowledge. If challenges and priorities have been defined globally for rare diseases by the European Commission, persons involved in rare anaemias have taken the opportunity to contribute to the empowerment of patients with rare anaemias. One of the ENERCA partners objectives was the mapping of existing centres that take care of patients with rare anaemias in Europe. Another goal was to obtain a directory of facilities available per centre for patients with rare anaemias. We thought that with those results it could realistically help to define a consensus regarding the criteria to be recognised as a centre of expertise for haemoglobinopathies and very rare anaemias.

Methodology

In various European countries, a questionnaire was distributed via ENERCA partners to colleagues or national and local scientific societies. It is certain that all centres have probably not been informed of this initiative. For France and United Kingdom, there is already recognition of expertise centres in the field of haemoglobinopathies and that is why the questionnaire was not distributed.

The main sections of the questionnaire were a general overview of the centre and its activity *i.e.* centre of expertise or general centre, type of patients followed, number of patients followed annually, if a laboratory, average number of samples tested annually, the expertise covered in the centre *i.e.* diagnosis and prevention, follow-up/case management such as allocated services and staff for management of acute and

chronic events, criteria (Proof) of expertise *i.e.* availability of specialised services, specific treatments, patients services, decision supports and registries, a link with research and finally existence of publications, grant, teaching and training activities.

Results and Discussion

Ninety centres answered the questionnaire

For rare and very rare anaemias, availability of specialized equipments and treatments are satisfactory. But our results show that for laboratories as well as for clinical centres necessary tools to give a diagnosis, to follow and manage the patients are not always available. If it exists, the dedicated and specialized teams are not always implemented.

It is obvious that with a rare disease, few patients and sometimes geographic isolation, it is very complex to provide all the expected services.

Our results show also that, even in centres of expertise, a registry is not always implemented. Nevertheless, collection of a core data set will support the continuous improvement of clinical care. Decision supports for the health workers and for the patients are also missing in several centres. In view to improve those points, difficulties to create and collect data should be investigated.

Accessibility to grants is not frequent even in centres of expertise. A proposal should be that more grants and funds could be dedicated to rare diseases.

Finally, in view to share tools and all aspects in the management of patients with rare or very rare anaemias as well as to improve the knowledge of these diseases at all levels, networking should be encouraged.

Conclusions

General recommendations, recommendations for laboratory diagnosis of rare and very rare anaemias or for genetic counselling were proposed by the ENERCA partners. They are presented, will be edited in the *WhiteBook for the creation of a European Reference Centre for Rare Anaemias* and will be available on the ENERCA website.

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Thalassemia Reports 2013; 3(s1):e30
doi:10.4081/thal.2013.s1.e30

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*Parts of this work were presented at the
"3rd Pan-European Conference on Haemoglobinopathies and Rare Anaemias",
Limassol (Cyprus), 24-26 October 2012.*