Response to the public consultation on RARE DISEASES: EUROPE'S CHALLENGES by **EFAPH** (**European Federation of Association of Patients with Haemochromatosis)**: Jean Rialland (President EFAPH), Dr Françoise Courtois (Medical Councillor EFAPH), Pr. Pierre Brissot (Hepatology at University Rennes, Vice-President EFAPH), Dr Barbara Butzeck (EFAPH members states Representative and President German Haemochromatosis Association), Pr. Graça Porto (Hematology Consultant at Santo António General Hospital, Porto, Portugal; Scientific Committee of APH (Portuguese Hemochromatosis Association)

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Question 1: Is the current EU definition of a rare disease satisfactory?

Although we agree with the concept of a broad definition like the one proposed, we would like to stress that a definition of RD should not be used to restrict the scope of action on a particular disease just because there are conflicting data about its prevalence. A good example is Hereditary Hemochromatosis. Although considered by some as one of the most prevalent genetic conditions in European populations (3 homozygotes per 1.000 citizens), it is becoming evident that the clinical penetrance of the disease is quite low (<10% of homozygotes develop the disease that means less 3 per 10.000 citizens)...

Comment Pr Graça Porto (Portugal): *Hereditary Hemochromatosis is an example of genetic disease in which premature morbidity and mortality are avoidable, i.e., although potentially lethal, it is compatible with a normal life if diagnosed and treated (with a simple treatment) at early stages. In order to enhance case detection high indexes of clinical suspicion are fundamental since symptoms are non-specific. Concerted actions are therefore needed to increase awareness, provide information and support knowledge about the disease among clinicians*.

Question 2: Do you agree that there is a pressing need to improve coding and classification in this area?

**Yes, it is a necessity**. Taking again the example of Hemochromatosis, the existing ICD classifies Hemochromatosis as one entity (E83.1) and does not discriminate the rare forms of Hereditary Hemochromatosis, such as Juvenile Hemochromatosis, ferroportin disease, etc, or HFE-linked hemochromatosis from other common acquired iron overload syndromes.

Question 3: Can a European inventory of rare disease help your national/regional system to better deal with RD?
**Yes**. To improve prevention, diagnosis and care of patients with RD, we think that **the priority is to develop national and regional centres of reference** (for specialised care, research and experts, organising healthcare); these centres of reference compulsatory should include patient organisations for an active collaboration (e.g. Pr Pierre Brissot University of Rennes and FFAMH* and AHO**).
The other actions must also set to work (dissemination of appropriate information and support to information networks).

Question 4: Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?

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Although in many cases mobility of patients is important, we think that transfer of knowledge is more crucial to help the development of best practices that will help more and more patients in their own countries. The development of national/regional centres of reference that will be linked to EU reference networks is a major aim. Although, of course, MS are responsible for the identification and support of their expert centres, the "pressure" and support from EU to those programs is critical.
"*A good example was the development of a National Plan for Rare Diseases in Portugal, announced at the Eurordis meeting in Lisbon, 2007, when the country was committed with the presidency of the EC. We hope to have this program supported financially to achieve our goals*"(Dr Graça Porto- Portugal).

Question 5: Should on-line and electronic tools be implemented in the area?
**Yes**, these electronic tools are essential for the patients communities (education, informations, exchanges of experiences, announcement of new care centres, meetings, late scientific results, guidelines and recommendations).

Question 6: What can be done to further improve access to quality testing for RD?

Efforts should be spent to attract more participants of the several MS on EU programmes on the quality of genetic testing (Eurogentest, EMQN schemes, etc.)

Question 7: Do you see a major need in having an EU level assessment of potential population screening for RD?

It may be important for some diseases as proved for PKU and congenital hypothyroidism. However, great care must be taken with new policies that must be taken with rigorous assessment criteria. There are some concerns about its relevance because of the heterogeneity of the European populations in terms of its genetic diversity.

Question 8: Do you envisage the solution to the orphan drugs accessibility problem on a national scale or on an EU scale?

EFAPH thinks that an EU scale would be better than a national scale.

Question 9: Should the EU have an orphan regulation on medical devices and diagnostics?
**Yes**.

Question 10: What kind of specialised social and educational services for RD patients and their families should be recommended at EU level and at national level?

Respite care services, therapeutic recreation programmes for children and young adults and psychological support should be recommended at a national level. Information services and help lines and financial support should be recommended at EU level.

Particularly for genetic haemochromatosis campaigns for a family education are necessary and helpfull on a national level, in order to prevent the outcome of the disease as early as possible.

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Question 11: What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?

National networks supported by research and public health services and European funded networks to link international (European) to national/regional databases would be appropriate. These databases demand a large confidentiality to protect personality of patients.

Question 12: How do you see the role of partners (industry and charities) in an EU action on RD?
Yes of course a financial support is obligatory to empower patients, but in case of haemochromatosis without any pharmaceutical treatment it is nearly impossible to find partners in industry.

Question 13: Do you agree with the idea of having action plans? If yes should it be at national or regional level in your country?

We agree about actions plans at different levels (mundial, european and national).
It would be ideal if there is a national plan, but unfortunately because of several reasons such as: language, culture, policy,...one national plan is often impossible and in this case we recommend to start regional plans to gather them in the future (if possible).

Question 14: Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?
**Why not?** But which links between Eurordis and the new agency will exist?

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