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Sent: Thursday, February 07, 2008 1:55 PM
To: SANCO RAREDISEASES CONSULTATION
Cc: anja.helm@eurordis.org
Subject: Public Consultation regarding a European Action in the Field of Rare Diseases

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European Commission
Health and Consumer Protection Directorate-General
Rare Diseases consultation
HTC 01/198
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The **Thalassaemia International Federation (TIF)** is a non-profit, non-governmental patient-driven organisation, founded in 1986 and working in official relation with the World Health Organization (WHO) since 1996. It is, today, an "umbrella" organization with 98 National, European and International thalassaemia and other disease oriented associations, medical and research centres, health professionals in the field, industries and other interested organisations and individuals participating from over 60 countries of the world. Thalassaemia International Federation **mission** is 'equal access to quality healthcare for every patient with thalassaemia across the world' and is **prime objective** is to support the development and implementation of effective control programmes, including prevention and clinical management of thalassaemia across the world.

Thalassaemia International Federation, would like to express its great admiration for the tremendous amount of work put mainly by the European Commission and Eurordis into developing the document 'Public Consultation Rare Diseases: Europe's Challenges'. More importantly for the great work done in the course of the very short period of time towards the promotion of rare disease- an almost "dark field some year ago, in Europe. We take this opportunity to make some comments on the specific questions identified in this document. Kindly find TIF's comments below:

Question 4: Should the European Reference Networks privilege the transfer of knowledge? The
mobility of patients? Both? How?
Answer: Recognized Reference Centres should outreach both to primary care services and network with other centres. Tools like ITHANET (an EU electronic infrastructure funded project- http://portal.ithanet.eu) should facilitate their pathway and transfer of information and knowledge promoted.

Question 6: What can be done to further improve access to quality testing for RD?
Answer: Accredited Reference Laboratories, related to the reference centers must be established. Networking is necessary so that labs can specialise in specific investigations.

Question 7: Do you see a major need in having an EU level assessment of potential population screening for RD?
Answer: Yes, both for early identification leading to treatment and for prevention in selected Rare Diseases. The EU should set standards, including ethical practices for prevention programmes throughout the European Union.

Question 8: Do you envisage the solution to the orphan drugs accessibility problem on a national scale or on an EU scale?
Answer: On both scales. Since the orphan drugs must reach the target (patients), whenever they are located considering both equity and patient mobility.

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?
Answer: Health care workers at all levels need re-education in chronic care support as a first step. Time for doctors/patients communication must be allowed in health planning. Information to the public concerning Rare Diseases must be intensified and taking into account religion, cultural moral and languages values.

Question 12: How do you see the role of partners (industry and charities) in an EU action on rare diseases? What model would be the most appropriate?
Answer: Industry /patients /academics should collaborate to develop networks of experts centres, with electronic connection (such as GRIDS- part of ITHANET project), to facilitate multicentre projects. This is especially important in Rare Diseases where pooling of data is essential.

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?
Answer: National level because of special national needs and characteristics.

We look forward to receiving the final document with the official position of the European Commission.

Kindest regards,

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