**To:** **European Commission**
**Directorate General for Health and Consumer Affairs.**

**From:** **Cystic Fibrosis Worldwide (CFW)**
**www.cfww.org**

**RE: PUBLIC CONSULTATION. RARE DISEASES: EUROPES CHALLENGES**

**Question 1**: The CFW Board is in favor of supporting the current EU definition for the whole of Europe, since it is the one used in all the European institutions and documents, as well as in most Member states. A more restrictive definition would endanger the reimbursement of Orphan Drugs for some of the "less rare" rare diseases (e.g. Cystic Fibrosis).

**Question 2:** Cystic Fibrosis (CF) is well defined by ICD E 84. An exact definition does not generally apply to all rare diseases. Clear and explicit classification and adequate codification are a necessary precondition for being traceable in health information systems. Thus the EU should undertake the necessary efforts for a better codification of rare diseases as described in the current consultation

**Question 3:** A European inventory of RD as suggested by the current EU consultation and which is being up-dated regularly is necessary to raise the national health authorities' awareness for these diseases and their special needs of medical care, treatment and research. Additionally it would offer patient and support groups a sound foundation on which to develop their requests for improvements of care and treatment.

In all European member states there is some knowledge about the treatment of CF-at least individual experts can be found. Expertise could be disseminated in a relatively uncomplicated way by educational programmes for doctors and allied health professionals and at European or worldwide conferences. There is a good structure for the exchange of information/expertise in the field of Cystic Fibrosis The problem, however, is that not all countries health authorities perceive the treatment of CF-patients by a team of medical and allied health CF-specialists as necessary. In several countries health care systems do not provide appropriate access to drugs (with CF e.g. enzymes or oral antibiotics) and medical devices (with CF: high-performance nebulizers and Pep-masks). In many countries the structural quality (number of doctors and allied health experts, medical-technical equipment etc.) of their CF centres does not conform to the European Standard of CF care (1).[i]

**Question 4**: The CFW Board supports the development of national/regional centers of reference and the establishment of European Reference Networks.

The issue of centers of reference is of primary importance for rare diseases patients. This issue was discussed at the European Workshop on Centers of Expertise and Reference Networks for rare Diseases in Prague 12-13 July 2007. The CFW Board supports the idea of not only creating but also funding European Reference Networks. These should have a capacity to provide expert advice, to produce and adhere to best practice guidelines, to implement outcome measures and quality control. They should also be involved in creating and funding epidemiological surveillance, such as patient registries. Databases and registries are very important tools to develop clinical research and improve care. The CFW board believes that EU has a major role in supporting and maintaining registers of patients with rare disorders. Many of these groups are so small that they have to be maintained at a European level. The registries should include information on disease severity and treatment. These registries could play an important role for the evaluation of novel and old therapies, as it would often be difficult to enroll sufficient number of patients in each country. Thus these registers must have long-term funding and cannot be subjected to uncertain funding cycles. Registries can also play a critical role in auditing the performance at different care centers, serve to promote best practice and help to identify poorly performing centers in need of assistance.

**Question 5:** The most important task for the EU is to promote patients' access to competent care and treatment of RD Europe wide. To promote E-health in connection with rare diseases is therefore of secondary importance. Electronic services and E-technologies cannot replace direct treatment of competent doctors and allied health professionals. They can only support and improve the field in which treatment is situated like research, exchange of experience and knowledge and the establishment of EU accepted standards.

**Question 8 and 9**: The CFW board supports the idea of the Commission to propose necessary legislative modifications in order to guarantee equitable access not only to orphan drugs, but **all necessary drugs** throughout the EU. Very expensive orphan drugs and other necessary drugs have to be funded at a system level rather than the local hospital or the local community. Medical devices can often play an equally important role as many medicines in health outcome, e.g. for administering the drugs, for medical diagnosis or other treatments. The CFW board supports the idea of EU having an orphan regulation on medical devices and diagnostics.

**Question 10**: The CFW board is in favor of specialized social and educational services for patients and families. The burden of treating Cystic Fibrosis is high. Best practice treatment is time consuming and expensive. The treatment has to be done on a daily bases in the home and sometimes in the hospital (many oral drugs, inhalation therapy, chest physiotherapy, intravenous antibiotic treatment and more).The parents/patients need support and ongoing education to perform the treatment correctly and cost effectively. Respite care service and financial support to families for unpaid caring and other extra costs related to the

treatment is also needed. Perhaps there is a possibility for the EU to coordinate the development of basic principles on which services should be provided in all member states. In this case the benchmark must be the highest level of social services offered; an adaptation towards the lowest level must be avoided.

**Question 11**: The EU should support the building and exploitation of such registries and even fund them, at least initially. Principally the question of ownership has to be answered. The pharmaceutical industry is interested in having access to these types of registries.The CFW board thinks it is important that patients and/or patient representatives involvement is critical to the management of patient registries and databases. Patients are more than just material providers and they have the right to participate in decisions that concern them.

**Question 12:** The CFW board wants to see a priority for research on rare diseases. Biomarkers as a tool for the following of disease progression are of course important, but many of the rare diseases are severe and the patients are more in need of basic research into the fundamental causes and pathophysiology of their disease. This is well exemplified in Cystic Fibrosis where the pipeline of novel therapies did not take a step forward until the last decade, despite the basic defect being discovered in 1989. Despite a great deal of research it was not until recently that understanding of the disease has reached the stage where novel therapies can begin to be envisioned and developed

The major problems for the rare diseases are the lack of interest from Big Pharma in the development of novel therapies. This means that the effort has to be undertaken by academia and, in some cases, health consumer based research organizations. However, as the diseases are rare, it is difficult to obtain sufficient funding in each European country. Therefore the EU should take a major step towards funding basic research and the development of novel therapies for rare diseases. This can be considered an important role for EU as these patients otherwise will be left without the important help they need. These efforts must be organized in frame work programs where the best scientists come together. This would also require a proactive approach to bring them together.

As the Big Pharma lack interest in orphan drugs, it is very important that drug development and clinical trials can be supported by EU. Part of that work could be a joint effort with small biotech companies, something that will also contribute to the development of new industries.

The CFW board and its European members consider it important that the EU is proactive in the development of novel therapies in Europe. The current trend is that all these efforts take place in the USA. The European patients will of course eventually benefit from these developments, but it is still an inferior approach to Europe taking a more active role. That should improve competition and lead to faster development -something European patients are eagerly waiting for.

**Question 13:** The CFW board finds it disappointing that nothing is mentioned about financial or other support to Patient Organizations. Because of the fact that a rare disorder is rare, it is often difficult to develop and sustain patient representative organizations within a state or even within a region. Empowerment is necessary to improve health outcomes and Patient Organizations play an important role in this.

We support the suggestion to establish action plans at the national level of member states. Member states should decide if they need regional action plans also. These action plans should take all aspects of RD into consideration: that is research, therapy, care and medical treatment, psychosocial services, access to therapeutic measures, to medical devices and drugs. These action plans should on the one hand address aspects affecting **all rare diseases and** on the other hand consider particular issues of single rare diseases. National patient organisations must be resourced to enable them to be involved in developing these action plans. The EU could support activities to establish national action plans by

- Inviting member states to start activities
- coordinating these activities
- coordinate the development of European wide guidelines for the elaboration of action plans for RD
- strongly recommending member states to adopt these guidelines

Consequently we support the EU approach in this issue as presented in the current public consultation.

**Question 14**: The CFW board supports the idea of creating an EU Agency for Rare Diseases. That body would ensure long-term funding and sustainability of essential activities in the field of rare diseases. The support otherwise depends on calls for proposals and attribution of funding that has to be regularly secured with a maximum of 3 to 5 years per project.

An adequate representation and involvement of patient organisations at national and/or regional levels must be guaranteed. If need be patient groups representing a single disease or very rare diseases may conjoin nationally to be represented as a group within the agency

---

[i] 1. Kerem, E., S. Conway, et al. (2005). "Standards of care for patients with cystic fibrosis: a European consensus." J Cyst Fibrosis **4**(1): 7-26.