

Statement

Concerning the announcement RARE DISEASES of the European Commission

E-mail from EURORDIS of 25 November 2007

Dear members of the European Commission,
Dear Sirs,

PHA Europe is an umbrella organization for national patient associations in the field of pulmonary hypertension. The organization, founded in 2003, comments the single questions as follows.

Question 1: Definition „Rare Diseases“

We suggest accepting the current definition for the entire European area. Most of the member countries and also the EMEA already use this definition, which leads to a largely standardised comprehension. Confusion could be caused by introducing a new definition.

Question 2: Codification and Classification

If inconsistent codification and classification exists, we believe it should be improved as far as possible.

Question 3: Register of Rare Diseases

Yes, a correct register of rare diseases should be compiled. Concerning pulmonary hypertension there are none or only a few register, allowing a statement about prevalence and etiology.

Question 4: European Reference Networks

European reference networks should grant priority to knowledge transfer as well as to mobility of patients. Regarding pulmonary hypertension it can fortunately be noticed that a knowledge transfer between the European centres for treatment of pulmonary hypertension has developed within the last years. Continuous conferences, in particular in line with the scientific project “Pulmotension”, guarantee the knowledge transfer.

Besides this, however, in individual cases the mobility of patients should be granted priority, e.g. if a therapy option, for example lung or heart-lung-transplantation, can be carried out better in an other European centre.

After European reference networks, national and regional reference networks are urgently to be followed. The reference networks are to be nominated pursuant to standardised and realistic manageable criteria, e. g. professional qualification, interdisciplinary teams, documentation of qualifications by means of publications, obligation to cooperate, information and continuing education, combination of research and medical care, integrating medical and social aspects in the treatment of patients, attendance at research, cooperation with self-support organizations of patients, documentation of medical care quality by participating at the compilation of a register.

Question 5: Online and Electronic Appliances

These are basically suggestive. However they can only be effective if they are steadily serviced and updated. The availability of a data base which collects all European research and study projects of the single diseases would certainly be helpful. Patients would be permitted to have access to studies and the latest research activities and also physicians in private practice could receive information on current developments.

Question 6: Tests for Rare Diseases

The diagnosis of pulmonary hypertension is very difficult due to the non-specific symptoms of the disease.

Question 7: Population Screening

At the moment the right-heart cardiography is the most important examination method referring to a suspected diagnosis pulmonary hypertension. A method for early diagnosis is to be developed. Would it be assured today, however, that a right-heart cardiography can be carried out with each left-heart cardiography, the diagnosis of pulmonary hypertension could be improved.

Question 8: Access to Medicines for Rare Diseases – Orphan Drugs –

In order to guarantee the necessary expertise, a solution on EU level is the only possibility. The equal access to all therapy options in all member countries of the European Union is a preferential aim of self-support associations.

Question 9: Medical Devices and Diagnostics

Yes, in our opinion such a regulation would be reasonable. This would have to include the inhalation devices as well as the infusion technics.

In respect of the oxygen care the need for regulations is considerable concerning the technics demand in order to improve the quality of life and mobility of patients with oxygen care.

Question 10: Social and Educational Services

Coordinated programme for “Compassionate Use”

This is where we see considerable need for action. Our experiences, especially at the beginning of the new era – medicamentous therapy options for pulmonary hypertension – showed the complexity and input which was necessary to receive indispensable to life medicines before their approval by the EMEA.

Patients with pulmonary hypertension who do not fit in the approved indication have still problems to receive the medicines which are indispensable to life for them.

The mentioned specialised social services are all on EU level and can also be recommended for the single countries.

Patients with pulmonary hypertension need psychological support in order to cope with the limitation of quality of life and life time. Also therapeutic leisure time programmes are necessary to develop new future prospects (education, profession, family planning) and to improve quality of life especially for children and juveniles.

Question 11: Data Base, Register, Archive

For pulmonary hypertension there are different organised data bases in Europe. There is a national register in France, one in Switzerland, a closed register in Germany, register which became necessary in connection with the approval of medicines by the EMEA in order to control the side-effects of medicines. An independent register to control the quality of treatment and survival and to learn about epidemiological data would be required.

It is especially important to involve patient associations in the support of the data base.

Question 12: Function of Partners

In our opinion all persons concerned with the health-care system should be involved in the research for rare diseases. The way this can occur depends on the requirements of each disease. It is indispensable to intensify the research in the field of rare diseases.

Question 13: National Action Plans

We consider the compilation of national action plans for rare diseases as urgently necessary!

Question 14: European Agency

Yes, we believe a European agency for rare diseases should be established in order to guarantee long-term finance and actionability in the field of rare diseases.

In conclusion we may suggest including the following items in the announcement:

1. Top priority should be granted the research in the field of rare diseases, as rare is not that rare as it seems.
2. The financial and organisational support of patient associations on EU level is an indispensable measure in order to maintain or ensure the high-grade quality of the activities of patients associations.

Please do not hesitate to contact us if you have any questions.

Bruno Kopp

President

PHA Europe (Pulmonary Hypertension Association Europe)

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