European Society for Phenylketonuria (E.S.PKU)
and Allied Disorders Treated as Phenylketonuria

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Hiddenhausen, Germany
February 14th, 2008

European Commission
Health and Consumer Protection
Directorate-General
Rare Diseases Consultation
HTC 01/198
11, Rue Eugene Ruppert
L-2557 Luxembourg

Public Consultation on Rare Diseases

Dear Sir,

Thank you for your invitation to answer the Public Consultation on Rare Diseases, which really is one of Europe's challenges. As the only Europe-wide-working umbrella-association of 27 national patients-organisations for Phenylketonuria (PKU) and allied inherited disorders, we easily can underline the importance to collect all kinds of experiences and to improve diagnostic and treatment strategies, as well as to assure quality-controlled high levels of healthcare in the field of rare disorders.

After consultation with our Scientific Advisory Committee, represented by its chairman Francjan J. van Spronsen, MD, PhD, Associate professor pediatrics at University Medical Centre Groningen (NL), Department of Paediatrics, at Beatrix Children's Hospital, the answers below are given on behalf of the entire executive board of our association, which was founded in 1987.

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

The definition is clear, and E.S.PKU supports it should be accepted by all Member States of EU.

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

In the sense of clear lining of any disease to available medical knowledge there is pressing need to improve coding and classification. Following the advice of our SAC, we have to recognise an increasing number of new diseases defined year by year. Metabolic disorders like PKU and allied disorders are multi-systemic, and difficult to trace in the register of the database. Having a special search-tool for metabolic disorders would be an improvement.

Question 3: Can a European inventory of rare diseases help your national/regional system to better deal with RD?

Having a complete inventory of all RD known would be helpful for patients and patient's organisations to insist in being recognized as afflicted by a rare disease. Patients should be ensured not to be forced to proof their suffering on an RD to receive adequate treatment and care.

On the other hand it must also be ensured that all rare diseases are listed in this inventory at once, and the inventory is under an ongoing update, as we have to recognize that by modern screening methods still RD's are detected that have never been described before.

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

Following our experiences, treatment of RD should absolutely be centralized. Patients should be grouped, centralized and treated by a centre with specific expertise in the specific RD's. By this, transfer of knowledge between centres is supported as well as a high-quality standard of treatment. Therefore centres should be recognised officially only when fulfilling specific service standards, which one must be networking with each other.

At the same time it is a must to ensure the mobility of patients to avoid refusal of treatment by patients themselves. Centres must be allocated in a reachable distance.

Question 5: Should on-line and electronic tools be implemented in this area?

For helping scientific professionals to reach an evidence-based level of knowledge not only for diagnosis but also for management of patients, any useful tool should be implemented and supported. Online and electronic tools are the most adaptable and useful instruments in these times. Well designed, thy can be useful for professionals, centres and patients and patients-organisations.

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

Within our association it is of daily discussion why screening strategies differ form country to country. Two aspects of screening must be fulfilled on a world-wide level:

- Screening for inherited disorders must be regular standard in every country for every child
- Screening methods and strategies must be comparable and follow common international guidelines, which must give safety to prevent any handicap, injury or discrimination as much as possible.

Therefore, discussion on European level is highly appreciated.

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

Screening is realistic when treatment is available. Therefore, we see a major need in having an European wide high level of treatment, followed by reliable screening programmes.

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

Comparing the reports of our national member associations, we also recognize different national accessibility to the required products, which sometimes aren't even orphan drugs, but e.g. dietary food for special medical purposes. Still we recognize the difficulties to convince national authorities and producers of the necessity of international availability of these products. Therefore we would appreciate higher engagement of EU to raise the pressure on those administrations who to not fulfil their legal and moral obligations.

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

To ensure a high-levelled and quality-controlled treatment of both, children and adults, a specifically experienced multi-discipline health-care-team for any medical services should be common standard for all EU MC. Therefore we would appreciate an orphan regulation on medical devices and diagnostics.

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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?

Beside the economical need for guaranteed reimbursement for the dietary products for special medical purposes, such as Amino-Acid-Mixtures and / or special low-protein food for treatment of PKU-patients at every age, these patients need a specific experienced multi-disciplined team of health carers:

- laboratory services
- dieticians and nutritional services
- adult physician and paediatrician services
- neurologists (pediatric)
- psychologists and neurophysiologists as well as
- social workers

Other even more rare disorders may need services from other disciplines in addition, such as physio-therapeutic services or others.

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

Some decades ago, Prof. Fölling, Prof. Guthrie and Prof. Bickel set the first milestones and proved the influence of nutrition in the outcome of some RD's. After a period of fast exploring nowadays we have to recognise a slow-down in fundamental research activities, e.g. how to overcome the problems of barriers, especially the blood-brain-barrier. This knowledge would most likely help to treat the patients in the most optimal way. Therefore it is necessary to combine research efforts by industry and university networks.

Question 12: How do you see the role of partners (industries and charities) in an EU action on rare diseases? What model would be the most appropriate?

Because the variety of activities to improve the quality of treatment is so high, many steps have to be taken:

- Progress in scientific research
- implementation and support of patient's activities
- ongoing industrial product development
- and many more.

It will for sure not gain any remarkable result for better treatment if one of these issues alone is promoted. We appreciate EU playing the role of a pacemaker and coordinator of these different approaches to ensure economically reasonable and targeted action and to avoid waste of resources and capacities by uncoordinated "single" research.

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Scientists and health-carers have to link their knowledge; Patients and patient's organisations should work as "transponders" between national authorities, scientists and patients themselves, in close cooperation with EU, on a European level.

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?

Even in larger European countries, knowledge on national level may hardly be sufficient to increase the knowledge of specific rare diseases. Combining efforts on an international level and collecting knowledge is of high importance on international, European level for both, scientific centres and patient organisations. These European centres and patient organisation may share and lead to coordinated national action plans to provide the care at a level as high as possible. Therefore a real EU budget on databases of rare disorders is of large importance.

Question 14: Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?

E.S.PKU supports the establishment of such a European Agency on RD for the following reasons:

- Screening programmes in newborns still aren't on an appropriate level in all European countries, even if their need is proofed by the results of those countries that already have screening programmes established on high levels.
- Treatment of RD's in adult patients is facing a lack of experience as long as many of the diseases are still first- or second generation diseases, which need special care beside evidence based medicine
- There are a lot of new metabolic disorders which are not described so far in any registries. Registering may identify the need of research and organisational needs in these specific diseases.

We appreciate your initiative and are highly interested to share our experiences with you.

With kind regards
on behalf of the Executive Board of E.S.PKU
and in accordance to the Chairman of E.S.PKU Scientific Advisory Committee

Tobias S. Hagedorn, Secretary of E.S.PKU

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