

**Slovenian PKU Association**

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

Ljubljana, 17<sup>th</sup> February, 2008

**Public Consultation on Rare Diseases**

Dear Sir,

As Slovene Association 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 and for that reason we answer to the Public Consultation on Rare Diseases, which is a necessary Europe's challenge

After consultation with University Childrens Hospital in Ljubljana, particularly with Ms. Mojca Žerjat Tanšek, MD, PhD, pediatrician and endocrinologist at University Childrens Hospital in Ljubljana, Slovenia, Department of Endocrinology, the below answers are given on behalf of the entire population of pku patients in Slovenia on questions from "**Public Consultation on Rare Diseases**".

Question 1.

I agree with the definition but I notice an important difference in practice. The incidence of rare diseases is very diverse. The prevalence 4 per 10 000 enables a group of patients who are able to make the requests to the health policy of the states and they create the support's groups, but the prevalence 1 per 1 million or less represents a much different situation for the patients and for their recognition. These small groups need additional attention and maybe also a sub-definition.

Question 2.

We surely need a specific coding and classification system. Slovenian health service has a medical insurance system which has the main diagnosis as a clue to the payment for the medical treatment. The hospitals are not able to get the real costs for the patients with rare diseases

Question 9

There should be an orphan regulation on medical devices and diagnostics.

Question 10

The special professional teams (physician, dietitian, nurse-educator, social worker, psychologist) for the follow up of patients with RD should be based in every center at national level. The support of parents groups, youngsters groups, "mothers" groups (to help the pregnant women with RD over the pregnancy) and educational camps should be encouraged.

Question 11.

All medical centers in MS should have their own registries, databases and biobanks but the regulations, infrastructures and financial support should be advised from EU central resource. This scheme has to allow the communications between the different sources and centers.

Question 12

Small states and centers do not have sufficient resources for the research. They should be implemented in the programs, studies and financial schemes of the bigger projects in EU. The parent's societies (from the field of RD) are applying for the state's money and charity in Slovenia, but they are always overdriven with the bigger groups of patients. The unified RD patients would have much more successful access to these sources on the EU level.

Question 13

Every MS has different medical and financial situation. The responsible medical centers on national level should prepare the action plans. Afterwards the data from the plans should be integrated in a basic EU document. The recommendations from the EU action plan should be the golden standard for all MS.

Question 14

European Agency on RD should be a main coordinator in all steps of the suggested policy. We support also a feasibility study.

The patients and their physicians have waited this kind of programme for many years.

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

With kind regards,

Natalija Stošicki  
president of the Slovenian PKU Association

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