Nederlandse Phenylketonurie Vereniging

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**Questionnaire completed by:**

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

Yes. The current definition is adequate. It does not ultimately matter what specific figure is mentioned in a definition. The issue is to identify and acknowledge a small group of patients which needs help and (often) does not receive it because its numbers are so small.

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

Yes, we share this view. Under the Government's plans for new health care arrangements, phenylketonuria (PKU) patients in the Netherlands will not receive any contribution towards their own share of health care insurance costs. The reason for this is that PKU patients are not classified as a group. This is a disgrace, and is a state of affairs unworthy of a developed country such as the Netherlands. See also the reply to question 1.

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

If recognition of rare diseases is possible only if a European inventory is set up, then this is a good idea. We take a somewhat different approach. Rather than allocating money to an inventory, the European Union would do better to use it to set up "contact points" for patients.

Specifically: if we, as an association representing PKU patients, were able by means of a European contact point to get the Ministry of Health, Welfare and Sport to recognise PKU as a chronic disorder, this would be a huge step forward.

Why do this at European level? Because of economies of scale (246 000 patients). A small team of medical experts could be set up at European level. There would then be no need to draw up and maintain a list of diseases.

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

The focus must be on the transfer of know-how. At present, this is mainly taking place at national level, and a country with such a small population would derive enormous benefit from collaboration at European level.

Last year, we collaborated with the Australian/New Zealand PKU Association via the European PKU Association. This achieved huge economies of scale.

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

Yes, this is one possibility. The Internet generally increases the accessibility of information. The EU must ensure, however, that the information provided is accurate. There should thus be a focus on "quality", in the sense of "accuracy" and also "readability" for the target group.

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

Regulation at European level. There is a terrific amount of regulatory provisions in Europe. Why does Europe do nothing to safeguard the rights of those with rare diseases? It seems to us that more progress has been made within the European Union than in many of the individual Member States. This justifies a European-level approach.

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

No, this is more a matter to be handled at local level.

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

No comment. We do not consider ourselves sufficiently qualified to answer this question.

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

Yes, definitely. The EU should encourage this.

**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?**

No comment.

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

The establishment of a European-level organisation to set up, maintain and safeguard registers, databases and biobanks in line with guidelines issued to it by the EC. This organisation could also assume the "contact point" function (see question 3).

Finally, this organisation could play a role in funding patients' initiatives at European level (for example, subsidising a European PKU association so that it can perform budgeted activities).

**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?**

The patient is the important thing. Industry has its own interests. At the interface between these interests, "Europe" (i.e. governments) should act to keep in check the natural preponderance of industry (i.e. its financial power).

**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?**

I would first of all want to know the nature of any action plan. In the Netherlands, the Government harps on the fact that it is so important that patients be heard. But if a small organisation representing patients knocks at the door of the Ministry of Health, Welfare and Sport or of a body implementing the heel-prick [= PKU test] programme (the National Institute of Public Health and Environmental Hygiene (*Rijksinstituut voor Volksgezondheid en Milieuhygiëne – RIVM*)), the door is kept firmly shut. Indeed, an additional lock is put on the door so that any pragmatic input from patients will not be heard.

To me, this is good reason not to be very positive about action plans.

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

Definitely. Previous answers (to questions 3 and 11) were given without being up to date on this issue. The idea is a very good one, and a wide range of issues could be covered.