



Dutch Steering Committee on Orphan Drugs  
PO Box 93245  
2509 AE The Hague  
The Netherlands

European Commission  
Health and Consumer Protection Directorate-General  
Rare Diseases Consultation  
HTC 01/198  
11, Rue Eugène Ruppert  
L-2557 Luxembourg  
sanco-rarediseases-consultation@ec.europa.eu

Re: Reaction on the Public consultation 'Rare diseases: Europe's challenges'

### **General remarks**

The Dutch Steering Committee on Orphan Drugs supports the initiative of DG Sanco to propose a Commission Communication and a Council recommendation on rare diseases. Although attention has grown for (some of the) rare diseases in the last decade these life-threatening or chronically debilitating diseases need more special and combined efforts on national level, European level and even on a higher international level in order to improve the situation of patients with a rare disease.

The steering committee focuses its reaction on some of the questions in the public consultation and adds some remarks to it. The steering committee would also like to add some issues in addition to the consultation paper.

### **Reaction on questions in the consultation paper**

#### *1. Is the current EU definition of a rare disease satisfactory?*

The definition of a rare disease is always arbitrary. The EU definition of a rare disease is felt as too broad by some countries and is not always followed by national reimbursement bodies. The steering committee would like to draw the attention to the fact that a rare disease, next to criteria like prevalence and seriousness of the disease, is actually also defined by so-called orphanised conditions: e.g. it may take a long period of time before the disease is diagnosed (when that is possible); symptoms of the disease may occur at early age; no expertise on management of the rare disease may be present.

Looking at the list of products with an orphan designation it is clear that there is much attention for some rare disorders with a prevalence of less than 5 per 10,000 residents in the European Union, whereas for other rare disorders no orphan medicinal product is in development. The committee would like to plea that in the communication also actions will be proposed to give attention to rare diseases with more orphanised conditions for which no treatment is available until so far. Suggestions may be to incorporate budget for research for these diseases in work programmes of the Seventh Framework programme (2007-2013) and in the second programme of Community action in the field of health (2008-2013) and to implement additional incentives for sponsors to encourage development of orphan medicinal products for these rare disorders.

*2. Do you agree that there is pressing need to improve coding and classification in the area of rare diseases?*

The steering committee is very much in favour of improving the coding and classification of rare diseases and to include more rare diseases in the ICD-11. It is a way to get more insight in the prevalence of the diseases. The steering committee works on the issue of the ICD-11 close together with Cineas in the Working Group on Coding and Classification of the Rare Diseases Task Force.

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

On-line and electronic tools may be of great help in several issues in the area of rare diseases. However, it is important that these kind of tools will be funded in such a way that these tools are sustainable. These on-line and electronic tools may be part of the proposed European information desk for care or European Agency of rare diseases (see our answer to question 8).

*7. Do you see a major need in having a EU level assessment of potential population screening for rare diseases?*

It is important that existing knowledge concerning issues of population screening is exchanged as much as possible between those stakeholders that are involved in population screening, including neonatal screening. These issues include technical points, and organisational and ethic considerations. Furthermore, more knowledge on population screening for other rare diseases on EU level should be collected for which screening could be a good instrument.

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

As reimbursement has to be decided on national level, solutions to the accessibility problem has to be found on a national scale. However, there could be some facilitation on European scale.

One of the factors that influence the accessibility is that many small and medium enterprises do not have experience with the reimbursement procedures in the different member states. Hiring of expertise in all countries may be too expensive for these companies. As a consequence they will start in some (bigger) member states and will extend the number of member states in the following years. A European information desk for care may be set up to facilitate information on reimbursement issues and research on rare diseases. In the Netherlands a similar initiative will start to improve innovation in care for prevalent and non-prevalent diseases. A European information desk for care for rare diseases may be part of the proposed European Agency of rare diseases.

Reimbursement bodies in each member state have to analyse the dossier before deciding on reimbursement. The young initiative of the Medicine Evaluation Committee (MEDEV) of the European Social Insurance Platform to analyse the dossier by several member states and give an informal advice to the other member states may help to improve accessibility.

More European initiatives of this kind may be encouraged to improve accessibility of orphan drugs.

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

It may be worthwhile to explore whether a European Regulation on medical devices and/or on diagnostics should be made. As medical devices for rare diseases or conditions are included in the US Orphan Drug Act the exploration should also involve the expectations and results in the USA.

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

Government of (web-based) registries, databases and biobanks is an important issue. A model of governance could be that all stakeholders involved in building registries/databases/biobanks have to give permission to use data or materials. The government model has to be decided in a very early stage of building registries/databases/biobanks.

The possibility to get long-term funding is important to be able to build a sustainable registry/database/biobank. In this long-term funding period evaluation reports should be incorporated to be able to stop the grant when no progress has been made. A grant of three or four years is not the appropriate funding method for building operable registries/databases and biobanks that may take years in case of some rare diseases.

*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?*

There are good examples of public-private partnerships e.g. to boost research on neglected diseases. Recently also the European Commission has started the Innovative Medicines Initiative, a public private partnership with the European Federation of Pharmaceutical Industries and Associations. Thus, it should be possible to use the common interest of public parties, charities and industry and build public-private partnerships in rare diseases.

*13. Do you agree with the idea of having action plans? If yes should it be at national or regional level in your country?*

The steering committee agrees with the idea of having action plans in each member state. As a matter of fact, the establishment of the steering committee is part of a Dutch action plan. These action plans will be different in each member state, as different activities will be needed due to the cultural, social and economic climate in each member state. Therefore the steering committee is not in favour of a 'one size fits all' European action plan. As The Netherlands is a relatively small country with 16 million residents the action plan should be at national level.

*14. Do you consider it necessary to establish a new European Agency on rare diseases and to launch a feasibility study in 2009?*

It is important that there will be long-term implementation of rare diseases policies at Community level and long-term interest for those issues that are still not solved or that will arise in the next years. A new European Agency on rare diseases could be a good instrument for the long-term implementation and interest. To be sure that implementation and coordination will also take place on a national level the steering committee proposes that in addition to such a European Agency a national contact/coordination desk will be created in each member state and/or maintained when there is already such a contact point.

**Additional issues**

1. The Dutch Steering Committee on Orphan drugs would like to emphasise that the EU may have a significant added value in the development of guidelines for management and treatment for rare diseases by collecting the (restricted) knowledge and expertise from each member state. This opportunity to combine the existing knowledge at EU level should be encouraged.

2. The steering committee supports the view that Community actions in the field of rare diseases are needed. However, it would also like to draw the attention to initiatives that are not set up specifically for rare diseases, but that may be of benefit for rare diseases. An example is the Innovative Medicines Initiative that aims to support the faster discovery and development of better medicines for patients and enhance Europe's competitiveness by ensuring that its biopharmaceutical sector remains a dynamic high-technology sector. As the objective of the communication is to sum up the necessary elements for an efficient policy addressing the important issue of rare diseases in Europe, the steering committee would like to suggest that rare diseases could also benefit significantly from these kinds of initiatives. According to the steering committee the link between themes in the area of public health and themes in the area of orphan drug development, like the activities of the Committee on Orphan Medicinal Products and new Regulations like the Regulation on Advanced Therapies and the Regulation on Paediatric Medicines may be elaborated more explicitly. Although treatment will never be available for a part of the large group of rare diseases, innovation may result in new treatment modalities for another part of this group. For the development of some new treatment modalities like gene therapy rare diseases have even been frontrunner.

Thus, next to specific actions on rare diseases, also opportunities that are not specifically designed for rare diseases should also be used to improve the quality of life of patients with a rare disease.

3. The steering committee would also like to draw your attention to the EC Directive on clinical trials on medicinal products for human use. In case of investigating new therapeutics for rare diseases multinational multicenter clinical trials are required to include a sufficient number of patients. A substantial number of these trials will be run by investigators working in university medical centres in EU member states or by small biotech companies. Unfortunately, complex EU legislation and extensive bureaucracy hinder multicenter trials with medicinal products across EU member states. The steering committee therefore proposes improvements in the implementation of the Clinical Trial Directive (2001/20/EC), in particular on issues that may facilitate performing clinical trials specific for rare diseases. Examples of facilitation may be simplification of the various EU forms, procedures, insurances, sponsorship & responsibility and more support in ICT-tools for electronic submission of clinical trial applications.