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The Commission´s public consultation regarding European Action in the Field of Rare Diseases

European Commission
Health and Consumer Protection Directorate-General
Rare Diseases consultation
HTC 01/198
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Comments from the Swedish Association of Rare Disorders on a proposal circulated for consideration: Rare Diseases: Europe´s challenges

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The Swedish Association of Rare Disorders is a disability organization acting to improve the conditions for people suffering from a rare disease (RD) with complex disorders. At present we represent about 50 different distinct RD and have circa 7 500 members. The diagnoses vary from one to the next but they have several problems in common like multiple disabilities (polyhandicap), and they are congenital, most of them genetic, and chronic. The Swedish Association of Rare Disorders is the only Swedish disability organization addressing RD, with rarity as such as a starting-point.

Introductory remarks
It is gratifying for us, the Swedish Association of Rare Disorders, to note the focus on the particular conditions for RD and the increasing interest in these issues. The work being done within the EU is important. We hope that the fact that RD again are prioritized in the Public Health Programme and in the Commission´s programme of Community action in the field of health (2008-2013) will result in improvements for persons with RD.

We hope there will be a political urge and enough resources in Sweden to make the necessary changes and efforts. But these actions must be taken in close collaboration with us, the disability organization representing the group aimed at. This is also emphasized by the Commission.

We also want to point out that there might be a risk that health care providers and professionals to a great extent would wish to influence the work. It is not the easiest thing to represent somebody else. Unintentionally you might contribute to developing activities with other aims than those promoting the patient.

Not just a health-care problem
Furthermore we want to emphasize that it is important that all professionals involved know that RD exist. It is essential that all professional groups concerned are informed about the difficulties, in all aspects of life, that people with RD have to deal with. The questions answered here below are almost entirely focusing on the health-care, which may result in RD being regarded exclusively as a health-care problem. But that is not the case.

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Question 1
Is the current EU definition of a rare disease satisfactory?
We don´t agree at all to the EU definition. We would like to keep the Swedish definition and also try to influence other states towards the same point of view. If common diagnoses are included, which the definition 5 per 10 000 persons imply, we believe there is an obvious risk that the definition rare will be undermined. The understanding of the true and special circumstances related to the Swedish definition will no longer be evident and awareness of the complexity of RD will decrease. On EU-level ultra rare is being used, as a new definition, since the EU definition for rare is too wide.

We also want to call attention to the fact that our definition refers to RD which lead to chronic functional disorders. We do not include curable or acute diseases.

Question 2
Do you agree that there is a pressing need to improve the coding and classification in this area?
Yes. We believe that a better classification would be of great value and necessary to create conditions needed to improve health-care for and knowledge of RD.

Question 3
Can a European inventory of rare diseases help your national/regional system to better deal with RD?
A list of RD is of course valuable. It can improve the chances of care on equal terms and collaboration at EU level on principles for diagnosis and treatment strategies. But we must stress that it is important to supplement the list with information and guidelines to health care providers at all levels to make a full use of it.

Question 4
Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?
The transfer of knowledge is important. It is required when developing and evaluating treatments and contributes with new ideas to research. This is vital both on the national and international level. But the information is clearly not altogether available when it is given in a foreign language.

To improve knowledge we need to create national medical centres (centres of expertise) with nationwide coverage. They should be responsible for the co-ordination of treatments and also have the total responsibility for acute as well as elective therapies. The responsibility should also include supporting rehabilitation in relation to the RD. The different professionals required for different RD must be represented at the centre and they must develop a teamwork. The transfer of knowledge from the centres to local health care providers is important to ensure continuous care for the patient at home. Furthermore, the national medical centres should be obliged to take care of "their" RD during the patient´s lifetime to see how it is affected in different periods. The centres should also promote research and development on each RD, transfer the results and develop networks and co-operate with centres abroad.

It is important that the patient has access to a centre when needed and the distance is not crucial. It is better to travel to get accurate care, rather than seeing a local health care provider lacking qualifications about the RD.

Additionally, we believe that some kind of centralized control is required to secure that the centres cover the different RD and the patients´ needs. Otherwise there might be a risk the centre is based on the interest the professionals have for one specific RD. In the document from the Commission it says that it is essential to identify centres of expertise and give them adequate funding. But we also think it necessary to have an action plan for those RD that lack support from expertise.

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Collaboration on the national level is also of great importance when building the centres, to guarantee equivalent development and quality.

Question 5
Should on-line and electronic tools be implemented in this area?
It is appropriate to implement and use electronic tools as a means to develop good and accurate care.

Question 6
What can be done to further improve access to quality testing for RD?
The suggestions in the document are satisfactory. But it is important to be aware of, and prepared for, the ethical problems and scenarios that might occur in different test situations. The health system must be trained, and educated to handle this. In these areas we also see a risk in industrial profit interest.

Question 7
Do you see a major need in having an EU level assessment of potential population screening for RD?
Screening is of great importance when it is carried out for the reason to find adequate treatment to prevent or reduce disability or illness. But an ethical problem might occur if it is carried out with other aims in view, for instance the choice to keep or not to keep a child who might have a disability. The care giving staff has not the knowledge or the means to give the advice or support needed. Therefore it is vital that the assessments concerning screening are standardized and uniform on the EU level.

Question 8
Do you envisage the solution to the Orphan Drugs accessibility problem on a national scale or on an EU level?
It is of considerable value if access to Orphan Drugs is controlled on a EU level, partly to cover the costs for nations with small populations and partly to equalize access to Orphan Drugs. The chances to develop new costly drugs would improve and the costs for some expensive RD treatments would be reduced. This would give way to more humanism and less economy.

Question 9
Should the EU have an orphan regulation on medical devices and diagnostics?
To have medical devices and diagnostics within EU is a good idea, it guarantees a supply.

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?
Persons with RD do not have equal access to care services they are entitled to, which persons with more common disabilities get more easily. This is due to lack of knowledge by those professionals who decide what measures should be taken and also decide over economical and other subsidies. To overcome these problems, massive efforts must be made to inform those working with RD and those in educational programs to improve their knowledge. The societies representing different RD groups work hard for their members, which was confirmed in the member investigation presented by the Swedish Association of Rare Disorders in 2004. The support from the societies is momentous. They inform persons with RD about their diagnose, their rights and what they can do to get accurate treatment or any kind of support. The RD societies make it possible to meet others in the same situation and experience a feeling of togetherness and consequently to improve quality of life.

But the societies have to deal with the major problem of not getting any subsidies for their work. In Sweden, societies with a nationwide coverage are not eligible for any economical grants. The societies are expected to work on a municipality or county council level and to get their funding

from local or regional authorities. This is impossible for the RD societies that cover the whole nation. This results in big variations in activity and a strong dependence on the real enthusiasts to survive. The RD societies´ difficulties in financing their activities must be taken into consideration when new plans are made for persons with RD.

Generally there are all too few educations about RD. The educations are held by the rehabilitation centres in Sweden and therefore they are unequally distributed and insufficient. The Family program at Ågrenska in Gothenburg is highly estimated by our members. But many of them indicate that it is not available to them since their county councils are reluctant to pay for the costs. Furthermore, the educations, which last five days, are offered very seldom for each specific RD, which might be once in a life time and that is not satisfactory.

Conclusion: A lot needs to be done, in the first place on a national level, to improve the situation for people with RD.

Question 11
What model of governance and funding scheme would be appropriate for registries, databases and biobanks?
These systems should be controlled by a Government authority and not private businesses.

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?
Our concern is what will happen to government funding if benefit societies take over. The different partners´ roles must be taken into consideration. Our view is that the industry should pay to different research funds, and researchers within the RD field should be able to apply for means from the funds. We are positive to collaboration with the industry and benefit societies, on the condition that clear agreements for “the rules of the game” are made.

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?
We believe that a national activity plan for RD must be developed as soon as possible in Sweden. The plan must include all aspects that are important to help persons with RD to improve quality of life and guarantee fundamental rights. That means significant improvements in health care, care, education, work and leisure time. The conditions for the RD societies, that work on a non-profit basis for their members, must improve as well.

Question 14
Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?
Yes. The establishment of such a EU Agency may help the EU to support and influence MS to take steps that would benefit RD. Consequently this would lead to improvements and equal rights in the EU member states, which we welcome.

Yours sincerely

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