A member of:

OUR POSITION ON THE DOCUMENT "Consultation regarding European action in the field of Rare Diseases".

Dear Sirs,

We have given our full attention to the lengthy document*" **Consultation regarding European action in the field of Rare Diseases".***

We find this document really important for those of us who suffer from a low-prevalence disease like ataxias, but we wish to make some observations on the document which we hope will be taken into account, given that they stem from the experience of living with a rare disease.

1. We believe that the term "rare disease" should be retained and no distinction made between *"Rare Diseases"* and *"Extremely Rare Diseases",* in that this would only lead to terminological errors and we do not believe that it is really significant for anyone. This subject was already discussed at the meeting held in Brussels on 13 September, and the majority at that Conference supported keeping a single term for all low-prevalence diseases, i.e. "*rare diseases".*

2. We found it especially appropriate to make the distinction between those rare diseases which permit one to carry on a more or less normal life and those which cause a serious disability and which, in our view, should

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attract special protection and greater investment in research, given that they entail extraordinary suffering for those struck by them and for their families, barring them access to work, which has repercussions on their economies at household level as well as on Member States' economies. Investing in research into these pathologies will bring sizeable savings in the medium term for the Member States and a better quality of life for many people suffering from a multiple disability.

3. We take the view that the conditions on which orphan drugs are sold should be improved. It can easily take 10 years from a pharmacist undertaking research to drugs going on sale and being readily accessible to all patients, which by any standards is too long for this kind of very serious disease which puts sufferers' lives at risk.

4. We believe that, from a policy point of view and given the huge heterogeneity of rare diseases, it may be useful to group them and to create generalist networks, but we totally disagree with this approach, in that the various rare diseases may have some points in common, as mentioned in the text of the consultation, but every one of them is of course unique and specific, and the information on each one is always different from that on the others. It may be that the problem of dispersion would be obviated if patients' groups for each disease were represented in the information networks. Every disease has its characteristics and peculiarities and information and research has to be specific to each disease; there should be no attempt to lump them together to keep costs down.

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5. As for research into rare diseases under the Seventh Framework Research Programme, the first and second calls for expressions of interest excluded rare neurodegenerative diseases and, in spite of our multiple proposals, we have to date been given no grounds for this.

6. We think that the creation of networks of reference centres for rare diseases is an excellent initiative, provided that, in the end, innovations in research and treatment are accessible to all patients and not just to citizens of certain countries whereas they are barred to others with less economic and research capacity.

Our responses to the questions set out in the document are as follows:

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

- *We find it entirely satisfactory.*

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

- *We agree entirely with the response of the document.*

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

- *We wholly agree with the document.*

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

- *We take the view that Orphanet and Internet are useful tools for disseminating knowledge, but we find it hard to agree that lives can be saved in this way. Having access to sufficient information is important, but it will never replace the role of an expert physician. Furthermore, most doctors are not familiar with Orphanet, and it is not a priority for them. I believe it is much more useful to create networks of experts in every rare disease which can put their knowledge at the service of doctors in related disciplines. That could save lives.*
- *Patient mobility could be useful only in special cases. Patients with seriously incapacitating diseases would find it difficult to travel from their own countries to receive treatment in another, and they might be able to do so once or twice but would never be able to travel for prolonged treatment. We believe that this can only prove useful in conjunction with information networks made up of researchers and experts in specific pathologies.*
- *For us the model involves creating networks of excellence in each rare disease with scientists and expert physicians pooling their knowledge and research and having regular meetings to update their knowledge and to take stock of the state of research. This model is the most useful for patients. Patient mobility can only be an option in extraordinary cases, if the reference country has no doctors or researchers or any infrastructure*

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for the treatment of the diseases. Financing should be made available for a combination of country-level and European-level research.

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

- *We see this as the fastest means of communication. We agree with the case made in the document.*

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

- *We do not agree to carrying out genetic analysis on the general population to detect rare diseases. Given the numbers involved, we believe that is not economically viable, and in any case those resources could be used in research into a cure for diseases. Furthermore, if it were decided to undertake screening for rare diseases, it would never be possible to do this for all of them and there would never be agreement on which diseases should be screened for and which should not, particularly when any screening of the population or of newborn children is not going to prevent them developing a disease.*

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

- *As stated in our response to the preceding question, only in those diseases in which it could be useful would population screening be advisable, but it makes no sense in the case of most rare diseases. For ataxias, in*

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particular, it would be absurd to carry out large-scale genetic analysis and this would only prove useful in the event of a family history, especially in the preconception stage.

- *We agree entirely with the comment on access to orphan drugs or drugs at the experimental stage, where access can depend on the country of residence. One solution entails speeding up the procedures for putting drugs on sale and making it obligatory for drugs companies to sell their products in every country.*

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

- *EU scale is the only one we can envisage. It is hard for any one country to contribute to a solution for access to orphan drugs.*

Question 9: Should the EU have an orphan regulation on medical devices and diagnostics? *We agree entirely with the views set out in the document on a coordinated compassionate use programme and support this, as it tallies with our approach. Compassionate use of experimental drugs is on many occasions the only way of gaining access to drugs and certainly often the only one, given the time it takes for these to come on the market. We support this approach, support extending it to hospitals and its being financed not by every hospital but by external financing. Sometimes physicians cannot supply drugs by this means in the face of opposition from hospital managers because it brings considerable increases in their spending on medication. The subject of* ***financing for compassionate use*** *should be studied so that the costs are not met by the hospitals but by the national health services.*

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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?

- *Social services are fundamental in the context of RD. An added difficulty in the Spanish case is that the country is divided into 17 Autonomous Communities and that each region has responsibility for social, health and educational services, which makes for considerable differences not just with our neighbouring countries but even at the level of the States themselves, according to where patients live.*
- *Notwithstanding the above, it is fundamental in the case of RD that the social and educational services look at whether the evaluation systems they use for their records are appropriate for the patients' health and disability characteristics.*

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

- *We believe the funding model should be a combination of national and European Union schemes within specific programmes.*
- *We agree entirely with what the document sets out under the headings: **"Data protection", "Networks of research for RD", "Coordination between MS funding agencies" and "Intensifying Research"***

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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?

- *No response*

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 agree entirely with having action plans, and believe they should be at national and also at European level. EU Member States should be obliged to comply with them.*
- *We believe it is very important that an Advisory Committee on rare diseases be set up, with representation of the different European associations on some rare diseases - or at least on some types of rare diseases: neurological, etc.*
- *We also agree entirely on the statement on funding for* rare diseases from the EU budget.

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Question14: Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?

- *We also believe it is fundamental to* ***set up a Community agency on rare diseases*** *because it would guarantee sustainability and long-term financing, and we agree with the approach taken in the document.*

We believe it is essential to highlight the need for greater investment on research into rare diseases. Any research into these pathologies will go some way towards curing these diseases, or at least improving the quality of life of patients and their families and permitting patients to be integrated into society so that they can live full, normal lives.

Our greatest plea is for more resolute promotion of research into rare diseases and promotion of research into and the development of orphan medicinal products.

The Management Board of the Galician Ataxias Association.

Galicia, January 2008

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