ORDEM DOS MÉDICOS
Conselho Directivo
COLÉGIO DE GENÉTICA MÉDICA

Comments from the Board of the

College of Medical Genetics of the Portuguese Medical Association (Ordem dos Médicos)

on "Rare diseases: Europe's challenge"

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

We support the current EU definition of rare disease of less than 5 per 10,000, for the whole of Europe, even if it is broader than others also used. One problem with such a numerical definition, however, is that prevalence is obviously not documented for many of the rare diseases, and particularly the rarest ones. Otherwise, the very rare disorders, with unknown prevalence, would still risk receiving the least attention.

Thus, other alternative and/or supplementary criteria should also be used. Also, diseases that are rare in the general population may be frequent in certain population subgroups. We thus support the definition of diseases with an overall population prevalence below 5:10,000 are considered rare.

This number, nevertheless, should be referred also to any specific population, given that some conditions common in some areas or countries may be rare in other (or underdiagnosed), as is often the case with so many genetic diseases. Thus, population specific prevalence values should be used, in order not to exclude some populations or population groups.

On the other hand, population prevalence should not be the sole criterion, as ignoring other factors (such as age, gender or ethnicity) may cause underestimation and overlooking of the needs of some (already more vulnerable) population groups.

Question2: Do you agree that there is a pressing need to improve coding and classification in this area?

Yes. Adequate coding and classification are essential for definition of health policies and health care plans, as well as for individual health care provision. We support that, as mentioned in the draft, a "working group on classification and codification of rare diseases", advising the WHO in the review process of ICD, as well as an "active cooperation of the EU Statistical Programme", are very

much needed to ensure the use of the new codes to cover so far neglected rare diseases in future ICD versions.

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

Though certainly difficult to achieve, a European inventory of rare diseases would be extremely valuable and useful for our national health system and, particularly, for national registries of rare diseases, and to improve awareness and provide support for research in rare diseases. As mentioned, this should be regularly updated, and should be classified by medical specialty, aetiology and prevalence.

The European Commission should provide financial and other support for this activity, and guarantee its sustainability. It would be of great importance for national plans and to organize national health care for rare diseases.

This inventory should be available to all Member States and, possibly, through Orphanet, for clinicians, other professionals, the media and the public at large, as a means of recognition of rare diseases and to foster its quality health care in Europe, as well as supporting research.

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

Both, depending on certain factors, namely the overall and regional prevalence of each rare disease.

The establishment of European networks of reference centres is perhaps the most important issue to improve knowledge and care in rare diseases. It would be important to recognize officially in some way, some centres of expertise which, as mentioned, are often solely established by national or international reputation.

The transfer of knowledge and of experience within European networks would thus be of primary importance, particularly for the more frequent among the rare diseases. Mobility of patients should be used only in certain instances, as with very specialized treatments.

For the very rare diseases, however, this may be different, as concentrating services in small number of reference centres (within each country or even across Europe) will be the only way these can acquire the experience needed to provide them with enough quality.

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

Yes. IT tools are indeed essential to share information and overcome isolation of patients and families, as well as of professionals dealing with rare diseases, and the EC should continue promoting and financing the already existing resources, such as Orphanet and other EuroGentest databases (as the quality lab database).

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

EuroGentest and the EMQN have had so far an important role in Europe to try to promote and harmonize quality testing and counselling in rare genetic diseases. The sustainability of these efforts, and, in particular, of the EuroGentest quality lab network should be supported by the EU.

Fostering the creation of national and European reference centres and networks, particularly for the more rare disorders, and assuring its sustainability, supporting the creation of new external quality assessment schemes, where unavailable, and promoting the exchange of information and the diffusion of Orphanet to all European countries and language groups would be important additional measures for the EC to take, in order to improve access to quality testing, particularly in the field of the more rare genetic diseases, where transborder flow of samples has a great impact, and harmonization of quality testing and counselling, and of recommendations and practices, is known to be a significant problem.

The rather demanding development and certification of certified reference materials for genetic testing of certain rare diseases is certainly an additional area where the support of the EC would be crucial for improving and harmonizing quality testing throughout Europe, in the line of previous and current EU funded projects, such as CRMGEN, EMQN and EuroGentest.

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

Yes. Only through a European initiative can enough evidence be generated and gathered, to support decision making both at the EU and the MS level. The WHO criteria (Wilson and Jungner, 1968) are still important for assessing the need and opportunity to introduce new screening programmes. The need to evaluate analytic validity, clinical validity and clinical utility for any given test, and in any particular context, including population screening, but also its ethical, legal, and social aspects is common to all European countries.

Evaluation systems such as the Gene Dossiers, by the UK Genetic Testing Network, or the German initiative should be harmonized and extended to the whole of Europe, as is being done through EuroGentest. The EC should continue supporting these efforts and ensure its long-term sustainability.

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

Regulatory issues as those mentioned in the draft document (delays, marketing, access, reimbursement, prices, etc.) can only be dealt with properly at the European level. To promote equal access to orphan drugs throughout the EU, the EC should also envisage the necessary legislative modifications and encourage its introduction in the Member States, namely providing evidence on health technology assessment, promoting the harmonization of regulation and approval of new treatments, and the reimbursement of costs at the national level, particularly for the very expensive drug and other treatments.

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

The regulatory model used for orphan drugs was very successful and should be extended to diagnostics, namely to support and provide incentives for the development of orphan medical devices and certified reference materials for quality testing of rare diseases.

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?

Patients and professionals should be able to easily separate reliable from non-reliable information. Some sort of review system, as is done by Orphanet, and official recognition should be in place. Reference centres should share among them any available educational resources and develop and make available specific information tools and educational materials on the rare diseases, particularly the rarest ones, in a clear and understandable writing, and on a clear language accessible to all their patients and families.

Reliable information on rare diseases should be available both in electronic and paper support. The Patient Leaflets developed by EuroGentest (and accessible through its website) are an example of general information materials (mostly about services) already available in many European languages, but specific information materials are still needed for some of the rarest diseases.

The creation and support of existing patient associations for rare diseases and of independent umbrella associations and European federations should be encouraged by the EC. The EC should also put in place a system to recognize these associations and networks, and to evaluate the quality of the information they provide and their reliability, including any potential conflicts of interest they may have (e.g., with the pharmaceutical industry or with private or other professional groups).

Of all the social activities needed, as mentioned, creation and development of respite care services deserves a special mention, as an action that the EC should encourage and support.

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

An expert group at OECD is currently developing principles and best practices regarding the governance of human genetic research biobanks and genetic databases, and it would be important that these are considered, even while at their drafting stage, when proposing models for European and MS resources.

Public funding is perhaps the best solution for rare diseases databases/biobanks, given their rarity. This links, of course, with the need for orphan regulation for research and development of medical devices.

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?

All sectors should be involved, including research institutions and universities, as well as MS charities and the industry. Public-private partnerships supported and followed closely by the EC might be appropriate for some actions.

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?

National action plans for rare diseases must definitely be set up (rather than at the regional level), but European coordination of the national plans would be more than desirable.

In Portugal, a national plan is being developed and we see no need for plans at the regional level.

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

According to the definition provided of a Community Agency, the creation of a European Agency dedicated to rare diseases would certainly be the right instrument to promote and coordinate at EU level initiatives such as registries for rare diseases, biobanks and research databases, clinical trials, information databases, networks of centres of reference, consensus clinical care recommendations and promotion of quality of services on rare diseases and its assessment.

We believe that a feasibility study would be very appropriate, before launching such an initiative.

Overall, this board finds the document very well elaborated, and fully supports its substance and contents. We believe it will be crucial to advance our (and other) national action programmes on rare diseases.

We thank the Commission and DG SANCO for this important initiative and for the opportunity for this Board commenting on it.

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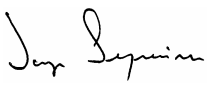

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