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European Commission
Rare Diseases consultation

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Ihr Zeichen | Ihre Nachricht vom | Mein Zeichen

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I am responding to the public consultation in my capacity as

- Orphanet coordinator for Germany
- EuroGentest participant and unit leader
- Director of a Department of Human Genetics at a German University Clinic with about 10 years experience in translational research

In my view the current document already describes the situation in the field of RD adequately, and I strongly support the proposals made therein for further development. I have some suggestions in detail.

1. Section 3 does not explicitly mention Orphanet although this has been and still is the major practical activity in the field of RD in Europe and worldwide.
2. Q 1: The public health impact of a disease is marked by both prevalence and incidence (both measures being connected through disease duration); incidence figures should not be neglected.
3. Q 2: An ICD nomenclature for RD is absolutely necessary. Orphanet is optimally suited for developing such a nomenclature, already having established catalogues in several languages. Orphanet activities in this direction should be funded through the Commission.
4. Q 4: Further support of Orphanet, as the key element for information collection and distribution in the field of RD, is absolutely necessary. Additional support is needed for translation into other European languages. – Depending on country and disease, the establishment of reference centres AND patient mobility should be supported. A careful needs assessment across EU member states is necessary.
5. Q 5: The usage of the Internet in every day primary care practice is still underdeveloped even in the big member states, and efforts are necessary to improve the situation. Funds should be provided for researching such improvements. On the other hand, traditional help lines (by phone) should not be neglected. National reference centres as well as European institutions (such as Orphanet central and the national Orphanet offices) should be funded to provide this kind of support.
6. Q 6: As already mentioned in the text, EuroGentest is the central European network to harmonize genetic laboratory practice and assess the clinical validity and utility of genetic testing. The answer to Q 6 is to sustain EuroGentest beyond its current funding period ending in 2009.
7. Q 7: EuroGentest is currently assessing genetic screening practices across Europe. A report will be issued in 2008. What already emerges from this activity is a lack of coherence of screening practices. The issue of personal choice, just briefly mentioned in the text, is central.

Personal choice requires adequate informed consent and counseling which, in their traditional forms, are likely to be the biggest cost factor in screening programmes. Research should be funded to explore the development of products that could serve to enhance efficiency.

8. Q 9: The vast majority of RD genetic testing is done by assays developed in-house (“home-brews”). These testing activities should continue to be under professional self-regulation, but they require a common European roof; sustaining EuroGentest would be the answer.
9. Q 13: From the perspective of Germany and other countries, national action plans, similar to the French plan, are urgently needed. A key element in improving the reality of patients with RD is their identification in primary care. Thus, among other things, training of health professionals in RD is of prime importance. European guidelines for developing national action plans would be helpful.
10. Q 14: A European Agency for RD would be the optimal common roof for the continuation of Orphanet, as the major data platform for RD, and EuroGentest, as the major network of experts and service providers in this area.

In summary, I strongly support the aims of this consultation. In my view, further funding and sustainment of the activities of Orphanet and EuroGentest are central to improving care of patients and their families with RD.

Jörg Schmidtke