CPME Answer to the Public consultation regarding a European Action in the Field of Rare Diseases (RD)

CPME welcomes this public consultation regarding a European Action in the field of Rare Diseases (RD) as these tend to be neglected in a market and profit driven economic environment. It is in this field that the legitimacy of Community Action is clearly given as coordinating and pooling of resources and knowledge will clearly improve the European Citizens' health status.

We do think that it would be useful to go further than pooling currently available resources on this topic and to create incentives, including financial ones, on the level of research, coordination, information as well as towards PPPs (public private partnerships).

This initiative is closely linked to the efforts made on a European level to promote access to validated and reliable information and to increase patient mobility across borders. The targeted creation of centres of excellence or reference is part of the general outlook on European healthcare in the future.

If a new agency were to be installed it should closely collaborate with national initiatives in the different MS and coordinate with other existing ones such as for instance the ECDC or EMEA.

A more differentiated approach towards "rare", "very rare" and "extremely rare" diseases should be promoted. Each one of these categories proves to be a different challenge and calls for different levels of coordinated efforts or knowledge concentration.

Finally if patient empowerment is encouraged and part of the coordination effort towards improved information pathways, the scientific community and physician organisations should be involved from the start in planning and conceptual initiatives.

4.1 To improve identification and knowledge of RD

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

The current definition of 5 per 10.000 inhabitants is a very large one, including diseases with a rather wide distribution and well recognised in the medical community. The 100 or so "rare diseases" near this threshold represent a rather large number of patients who generally do not have major difficulties finding information about their disease or access to highly qualified specialised care.

The next category of “very rare diseases” with a larger number of illnesses but a smaller number of patients is more in need of coordinated information and research policy than the “rare disease” category. It may be difficult to guarantee equal access to high quality care for all patients if they are restricted to regional or national level.

The last category of “extremely rare diseases” is the one which might benefit the most from coordinated action on EU level. Pooling of research and funds will lead to the creation of cross border centres of excellence. Information networks and resources about these diseases should extend across EU borders in order to pool into worldwide databases and research results.

These three categories reflect different needs and call for different approaches and should not be given the same priority.

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

To tackle this problem through coding issues means approaching the problem from the wrong side. Coding is a controversial instrument in medicine as it implies several underlying issues.

One of these issues is the lack of certainty of a diagnosis and the lack of providing this accuracy factor in the different coding schemes. Another issue is that very often the person coding the disease on death certificates or hospital discharges is not the same one that has established or has tentatively established a diagnosis, thus increasing the error margin.

Finally, a disease is rarely isolated and it is not always guaranteed that the so-called “associate diseases” are coded correctly and according to the correct priority. Some interactions between diseases are rather complex and not well rendered by any coding system (be it ICD 11 or whichever other known codification). All this is true for the “common diseases” but as you can easily imagine in the case of “rare, very rare or extremely rare diseases” all these issues grow exponentially and the error margins tend to become enormous.

It will be very important however to proceed further towards unification of denomination of diseases across acronyms, cultures and languages. This will not automatically lead to an accurate picture of prevalence through coding because of the above-mentioned issues which have to be tackled on a more generic scale in the medical community.

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

CPME as a European association can not answer on national issues

4.2 To improve prevention, diagnosis and care of patient with RD

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

This is one of the issues we would like to be facilitated through an eventual directive on health care services. Reference centres are usually established through reputation and do not benefit from any publicity. Even the regional or national health care community is not always aware of the relevant pathways for patients with RD.

We would favour an approach from the individual MS to identify relevant centres in their countries and to link up with comparable ones in other MS. To which extent European regulation should determine the prevalence and relevance of these centres to guarantee a reasonable geographical distribution throughout the EU should be determined among MS through perhaps some European RD institute comparable to the ECDC.

Every effort should be made to make information about these reference or centres of expertise, once identified and validated, available to the patient and health care community alike. The information should be accurate, transparent, adapted to the patient's needs and language independent.

Knowledge sharing between these centres should be encouraged on national and European levels by all means available.

Patient mobility is one of the key components in an eventual future health services directive and should be facilitated wherever necessary and requested. May we remind about the fact that although patients prefer to be treated close to their home and in their familiar cultural and linguistic environment, they should be able to access better care in the EU, be it by necessity or choice[1].

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

The importance of e-health tools for information dissemination and research is evident and has been stressed by CPME on numerous occasions. E-learning could become an important dissemination tool for Continuing Professional Development (CPD) on RD for physicians.

As RD are rare or very rare and as they are very often genetically linked it is of utmost importance to protect the confidentiality and the privacy of patient data. As one of the goals will be to create European data bases a common legal framework should guarantee the highest standards of data protection regardless of the MS.

We recommend a much larger involvement of patient and physician representations in this field as this is currently the case. To leave this issue exposed solely to industrial ICT developments would not be in the interest of patients.

[1] See CPME Answers to Health Services Consultation of 26 January 2007: http://cpme.dyndns.org:591/Adopted/2007/CPME_AD_EC_260107_010_EN.pdf

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

We would be very satisfied if the goals described such as transborder flow of lab tests, bridging of regulatory differences among MS in confidentiality practices, reimbursement, sample transport and storage could be achieved.

A European institute for RD should be able, much in the same approach as towards identification of centres of competence or excellence, to encourage and classify reference laboratories.

It should be avoided though in both cases, laboratories and centres of excellence, to create de facto monopolies and thus eliminating competitiveness.

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

This question cannot be answered in this context. This involves potentially highly controversial ethical considerations, apart from scientific argumentation.

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

EU-scale

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

If we have EU regulation or incentives for RD and orphan drugs it makes sense to initiate the same for medical devices.

Question 10: What kind of specialised social and educational services for RD patients and their families should be recommended at EU level and national level.

CPME being a European organisation, we can not comment on national initiatives. Please refer to answers given by MS organisations.

To unify or standardise social services in the EU constitutes a major challenge, by far more complex than anything planned through the draft health services directive. Help lines and information services could be of course coordinated through European portals but they should build on existing national initiatives. Incentives to create necessary social services should be put in place by European action programs but this should be imbedded in a general approach, not necessarily limited to RD.

4.3 to accelerate research and developments in the filed of RD and OD

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

The European effort should concentrate on the linking and knowledge transfer of existing data-bases in the different MS. Common standards for establishment and maintenance of these data banks should be established and most importantly access rules and confidentiality should be enforced by the highest standards.

No new structures should be put in place. A European institute for RD could monitor these developments through existing EU initiatives. Funding of databases, registries and biobanks should be supported through MS and the coordinative efforts, standardisation commissions and quality standardisation should be addressed through European funding.

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?

n.a.

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?

European action plans on this level should prove useful. They should mainly concentrate on giving templates for national initiatives and coordinate efforts to avoid duplication and to stay comprehensive. Particular attention should be put towards knowledge transfer and exchange. Incentives for PPP (public private partnerships) should be given.

As a European organisation we have to refrain from answering the second part of the question

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

As we have mentioned several times in our comments a European agency would prove useful if it is modelled on for instance the ECDC. The main goal would be standardisation, coordination, creating incentives, defining priorities and linking with other European agencies working directly or indirectly on the same topic. This agency should be staffed by national representatives in those countries where action plans are in force. This would imply that a feasibility study should be launched.