EUROCAT (European Surveillance of Congenital Anomalies) Response to

Public Consultation regarding European Action in the Field of Rare Diseases.

February 2008.

These comments represent the views of the Steering Committee of EUROCAT (European Surveillance of Congenital Anomalies).

We feel the document is clear, comprehensive and helpful. We are pleased to see scope for action on the prevention of congenital anomalies, and recognition of the importance of environmental as well as genetic factors.

Q1. Is the current EU definition of a rare disease satisfactory? The current definition is problematic in a number of ways. Firstly, the definition of "disease" is not clear – there needs to be guidance around the grouping and splitting of disease entities and subentities. Secondly, for prevention of chronic rare diseases, including congenital anomalies and genetic diseases, an incidence-based definition (yearly rate of newly diagnosed cases) is more appropriate and practically easier to estimate in the European population. We do not feel that Action on Rare Diseases should be delayed or complicated by definitional issues, but the document could mention these issues, and recommend a sensible approach should be taken when assessing whether any one action qualifies as relating to a "rare disease".

Q2. Do you agree that there is a pressing need to improve coding and classification in this area? Yes, and EUROCAT is working with the Rare Diseases Task Force on improving coding and classification systems.

Q3. Can a European inventory of rare diseases help your national/regional system to better deal with RD? This is most appropriate for treatment/care issues rather than prevention. An inventory is not needed for congenital anomalies.

Q4. Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How? This must depend on the details of the disease – its level of rarity, whether diagnosis or treatment is the main problem needing expertise, what methods are needed for diagnosis and/or treatment, how long rehabilitation is and which professionals are involved etc. It is preferable for patients to stay in their country, and with their own language, but the system should aim at the highest quality of continuing care, and this may sometimes require mobility of patients.

Q5. Should online and electronic tools be implemented in the RD area? Yes. Purpose-oriented use of appropriate online and electronic tools should be part of the design of Actions. Collaboration with e-Health initiatves at EU level may be productive and provide a larger budget.

Q6.What can be done to further improve access to quality testing for RD? We support a system of quality marking of laboratories, so that samples can be sent across borders with confidence.

Q7. Do you see a major need in having an EU assessment of potential population screening for RD? EUROCAT has recently conducted a survey of prenatal screening policies across Europe and their impact. This survey shows how important cultural factors are in framing and implementing policies, and we feel that screening policy is most appropriately dealt with on a Member State level. At European level however, it is useful to have information on screening policies and programmes in different countries and their evaluation, to inform national policy development.

Q9. Should the EU have an orphan regulation on medical devices and diagnostics? Yes, for congenital anomalies these are often more important than drugs, and incentives are needed to improve devices for the treatment of cardiac defects, hydrocephalus, skeletal dysplasias and diaphragmatic hernias for example.

Q10. What kind of specialised social and educational services for RD patients and their families should be recommended at EU level and at national level?. The issue is not just the existence of services but their quality. The EU could have a role in assessing quality.

Q 11. What model of governance and of funding scheme would be appropriate for registries, databases and biobanks? Public Health uses must be distinguished from Research Uses, and given a special funding stream. There is huge European Added Value to be gained from the Public Health uses, but funding at a European level must be long term, while allowing flexibility in priorities to recognise new developments. There must be quality control, examination of confidentiality arrangements, and long term sustainability in the infrastructure. Funding should be public, not industry, and prioritise public health action results rather than intellectual property.

Q12. How do you see the role of partners (industry and charities) in an EU action on rare diseases? What model would be most appropriate? All stakeholders need to be involved in designing actions. Industry funding should however be arms length where possible e.g. channelling multi-industry funding through an EU fund to be spent on Actions.

Q13. Do you agree with the idea of having action plans? If yes, should it be at a national or regional level in your country? Yes. Action plans should include prevention. They should be national, concerning national implementation of EU recommendations. However, there could also be some specific task forces at an EU level to co-ordinate or evaluate the implementation of specific parts of action plans. One such task force could consider national plans regarding folic acid supplementation and fortification.

Q14. Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?

We need sustainable long term co-ordination and funding of action in the area of Rare Diseases. We do not specifically need a European Agency - we already have EMEA and networks such as EUROCAT, EURORDIS, Orphanet and other RD networks with specific remits. An agency would create bureaucracy that would swallow up too much budget. There is more to be gained from European Committees and Task Forces which are flexible to need and include the appropriate expertise. One such Task Force that we suggest concerns the investigation of clusters of congenital anomalies and cancers. Clusters often create a level of public concern where an independent European Task Force would be seen as more independent than a national response.