EU Consultation

Response to the consultation on Rare disorders: Dr Alison Kerr

Here are my comments on the numbered questions raised-
2. It will be useful to encourage health professional to determine the diagnoses and carry out assessments of the needs of people with rare diseases and to record a very simple outline without identifying individuals, so that figures from the different European states can be compared, with a view to directing support where it is needed.

3. Medical textbooks and journals already provide a wealth of information on diagnosis and treatment. Some is on the internet but a lot of that is misinformation or advertising. Specialists in each rare disease might usefully be selected and funded to recommend and keep up to date collections of information and reference lists that can be placed on the internet as a readily available guide for health professionals and parents. Best practice can also be indicated in this way. I do not think that a new list of rare diseases would be particularly helpful. Such lists do already exist.

5. The summary of information for each disorder might include details such as where to find help with diagnostic testing. Standards for testing already exist in the UK and I suspect in most western European countries. I suspect that these standards are already stated on the internet. They might be referenced in the disease summaries.

6 &7. The value of screening and possibilities for prevention disease differ for each disease and each case would have to be decided individually, taking advice from the expert group of clinicians, researchers and parents involved with that disease. European funding might be provided directly for projects specifically directed to achieve this.

8. Issues regarding medication inevitably rest with the health services in each state however the internet information on each disease should make clear which medications if any have been proven to be safe and effective and should not recommend any that have not been thoroughly evaluated.

9. Disease registries are of value provided that the clinical evaluations on which they are based are correct and the questions are relevant, simple and not duplicated. Researchers in this area might be encouraged by direct funding and the provision of statistical support. In this way prevalence, outcomes and interventions can be determined for specific rare diseases. Such data can be published and copies accessed through the internet.

12. If key health professionals, parents or charitable organisations are undertaking specific projects to improve standards in diagnosis, assessment and recording of

rare diseases they should be able to apply for direct funding for a proportion of that work.

14. A coordinating European office might comprise a small staff capable of directing enquiries appropriately, entering data on computer and analysing it statistically, with senior epidemiological guidance (perhaps 3 people full time and a part-time clinical epidemiologist). However there is a real danger that all the available money could be spent on such an office which should only exist to facilitate the work in the field. Funding should mainly go to specific practical projects.

I hope the above is helpful:

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