Question 1: Agree

Question 2: Agree

Question 3: Agree

Question 4: We agree with the positive assessment of the service provided by the Orphanet database, even though, under certain conditions, the information seems to be presented in a very meagre way. Transnational transfers of knowledge and research results between centres of expertise are already a reality between scientific institutions. However, the aspect of assistance to patients is different, because it is unlikely that a programme of international collaboration can be brought about within a reasonable time frame. At least in Italy, the possibility to undergo treatment abroad exists, although, so far, the main difficulty has not been so much the assistance and treatment (almost always codified) as the availability of diagnostic tests.
It should also be borne in mind that some of the new Member States are probably lagging behind as far as molecular diagnostics is concerned, so a European network of diagnostic services would appear to be more useful than a network of assistance-based services.
Patient mobility is to be discouraged by all means: it would seem a far better idea to have biological samples travelling rather than people. A note could be made about simplifying the procedures for sending biological materials to other laboratories and on the reimbursement of the cost of having tests performed between different countries.

Question 5: The creation of new on-line tools in the field of rare diseases could lead to confusion for both healthcare operators and users. In fact, there already exist many very useful electronic tools and databases on genetic and rare diseases, outside Europe too. It should be stressed that such tools are already operational (e.g. Orphanet) and they need to be developed. Better information is preferable to too much information.

Question 6: The use of EQA (external quality assessment) and proficiency testing should be made mandatory for all the laboratories concerned with diagnosing rare diseases. The existing European-level quality control programme (e.g. EMQN) could be upgraded, perhaps reducing the costs of the associated laboratories and funding analytical and clinical quality control programmes directly.

Question 7: No comment.

Question 8: It is too often forgotten that most rare diseases are genetic diseases, which means that the risk of family transmission, the identification of carriers, the assessment of individuals affected by rare diseases of variable expressiveness and incomplete penetration often remain in the background and are not assessed correctly. However, medical genetics provides for a specific approach to find out how the disease is transmitted, to identify the persons at risk of transmitting it, to diagnose carriers of and those suffering from these diseases, to identify the risks of them occurring and recurring, and to propose prenatal diagnosis.
It is not possible to talk about prevention in relation to a genetic disease without taking into account a correct approach to medical genetics.
For these reasons, encouraging medical geneticists and developing their profession is a vital tool in managing rare diseases, which (let us not forget) develop and recur mainly in the family environment.
Unfortunately, the Communication pays little attention to the work and professionalism of medical geneticists.

Question 8: We agree with the need to release the funding of orphan drugs from local hospital administrations.

Question 9: It would be dangerous to subject diagnostic tests for rare diseases to regulations and standards. Precisely because the frequency of genetic diseases is fairly low, the possibility exists for the rules to be written in fairly general terms (neither could they be specific for all the 6 000-8 000 rare diseases). The risk is therefore that centres of expertise with proven experience would not be able to meet rigid standards (for economic, organisational or other reasons) and might therefore have to stop operating. The outcome could be to standardise the procedure without being able to continue to provide it to consumers.

Question 10: As far as Italy is concerned, before offering therapeutic recreation programmes, the following should be developed: home assistance, organisation of diagnostic services, financial support for families affected, appropriate counselling, the distribution throughout Italy of medical genetics structures.

Question 11: Declaration of non-competence

Question 12: Donations to public research on rare diseases could be made tax-deductible. The involvement of associations of sufferers and their families could then be encouraged, even if all these organisations collaborate, to the best of their abilities, on specific research projects.

Question 13: EU-level recommendations should not be at odds with what has already been done in the Member States, on pain of having to reorganise the national rare diseases networks.

Question 14: A new European Agency could absorb many of the resources available to improve existing programmes. The (relatively recent) work undertaken on rare diseases has been encouraging, and many important results have already been obtained, whilst many other projects are being constantly upgraded. A new Agency could be superimposed on top of the many other effective initiatives (at national and international level) with the risk of slowing the existing ones down. Basically, we have to respond to the patients and their families not only effectively and efficiently but also flexibly, given that scientific knowledge of rare diseases is developing rapidly. The speed at which discoveries are being made about the genes associated with or responsible for genetic diseases (which make up the majority of rare diseases) call for activities in constant change, which is difficult to reconcile with slow-reacting, cumbersome organisations.

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