

The Genetic Interest Group (GIG) is an umbrella group for patient organisations supporting patients of inheritable conditions and their families. The majority of the conditions that these patient organisations represent are without cure or treatment; they therefore look toward innovative biomedical research with hope and expectation for the delivery of cures and/or treatments.

GIG welcomes the draft Communication on Rare Diseases as an important strategic document that will help in the development of high quality and equitable national services for patients with rare diseases.

In addition to the questions posed in the Consultation document, GIG urges the EC to continue and enhance its activities supporting patient organisations and their networking at national and European level. For conditions which are rare in individual Member States, only by sharing the knowledge and experience of patients across Europe who are faced with complex and serious medical conditions can their needs be recognised and appropriate strategies and actions be put in place.

We address the individual questions posed in the Consultation below:

**Question 1. Is the current EU definition of a rare disease satisfactory?**

GIG is in favour of supporting the current EU definition for the whole of Europe because it is already used in European institutions and documents as well as in most Member States. It would be confusing to change the definition in this Communication, and a more restrictive definition would endanger reimbursement of orphan drugs for some of the ‘less rare’ diseases.

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

It is essential that rare diseases are accurately coded for traceability in healthcare systems. Most rare diseases are genetic, and appropriate management of families (including risk assessment, counselling, offering of genetic tests where available and other interventions) depends on access to accurate information about diagnoses of affected family members, for example on death certificates and hospital discharge systems.

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

While good related information sources exist (such as commercially available dysmorphology databases; publicly available web-based overviews of the molecular basis and clinical management of genetic conditions; and in the UK a web-based inventory of clinical molecular genetics laboratories and the genetic tests they offer), a single source pulling together scientific and clinical information for European clinicians would be a key tool in enhancing recognition and care of rare diseases, and to support research.

**Question 4. Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?**

GIG supports the conclusions laid out in the report of the EURORDIS workshop (“European Workshop on Centres of Expertise and Reference Networks for Rare Diseases”, Prague, July 2007) which state that only multidisciplinary approaches are effective in providing adequate care for people with rare diseases. Rare diseases are often complex, requiring care from different medical specialities and coordinated social care. Close communication, i.e. the transfer of knowledge, is critical, within and between centres and within Networks. Without effective knowledge transfer there is a much higher chance of incorrect diagnosis

taking place at non-specialist centres where patients first present, and the sharing of knowledge internationally is required to develop consensus guidelines on how best to treat patients and to coordinate research.

Problems such as language barriers, difficulties of travelling and family and work commitments, mean that patients should not be expected to travel outside their own countries for treatment. However, GIG supports the EURORDIS position that where necessary (e.g. for a second opinion at diagnosis, or where a treatment is not available in the home country) patients' travel should be facilitated, financially and administratively.

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

These are key tools to underpin the transfer of knowledge discussed under question 4, to link research groups and share data, and to give access to good information for patients and families. Several EU funded projects, including those in which GIG has been involved, have demonstrated the importance of on-line and electronic tools.

For patients and families, information technology not only opens up sources of information and advice, but can reduce isolation by connecting them with others affected by the same conditions in other countries.

**Question 6. What can be done to further improve access to quality testing for RD?**

The Consultation document provides an important outline of measures required to improve access to quality testing. A further area that will speed up the development of a comprehensive service across Europe, and ensure rapid transfer of new technologies into practice, is a Europe-wide mechanism to identify gaps in provision across Member States and to encourage development of laboratory services accordingly. GIG urges the Directorate to give this full consideration as a matter of priority.

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

GIG supports the aim outlined in the Consultation document to encourage cooperation in this area to generate evidence on which decisions at Member State level should be based.

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

The EU and Member States must cooperate and coordinate their activities in this area. The EURORDIS proposal (that the scientific assessment of therapeutic added value, and the estimation of the reference price for each orphan drug, be carried out at EU level, and that the Committee for Orphan Medicinal Products (COMP) (involving all Member States and patient representatives at the EMEA) be in charge of this assessment) is an interesting model, and recognises the need for scarce expertise to be shared. However, there is as yet no consensus on the methods that should be used in such assessments, and this is a key area requiring further work.

Crucially, accessibility will require national commitment to implementation, for example: to allow licensing appraisal processes to be tailored to rare diseases rather than applying standard methods where cost-effectiveness drives the outcome and may not be representative of the value of the treatment; to ensure the UK NHS supports prescribing orphan drugs through an effective commissioning process; and to ensure all prescribing physicians are made aware when innovative products become available so that all patients will be treated with the right medicine at the right time. Both resources and expertise will be needed to secure this commitment at national level.

**Question 9. Should the EU have an orphan regulation on medical devices and diagnostics?**

Current devices regulation focuses on quality and safety, but does not include assessment of clinical utility (unlike regulation of medicines). The IVD directive will need to be revisited to ensure clinical utility is properly taken into account.

In addition, the regulatory approach should be proportionate to the risks of the procedure and to its impact on the individual. Genetic tests for example generally require only a low-risk procedure (a blood test), but the results can have far reaching implications for the individual and their family.

Regulation of mixed devices (i.e. those associated with a pharmaceutical product) will require careful work to ensure clarity and a framework that is proportionate with the level of risk presented.

Many companies operating in the field of medical devices and diagnostics for rare diseases are small, and therefore the burden of regulation should be kept to a minimum, with a single European application

**Question 10. 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 key role for the EU must be to develop mechanisms to disseminate good practice in social and educational service provision across Europe and to support trans-national professional training. These frameworks should be underpinned with robust mechanisms for translating knowledge, expertise and standards right through to the professionals dealing directly with patients and their families.

Particularly helpful areas that the RAPSODY project has identified include respite care services; help-lines; therapeutic recreation programmes for children and young adults; integration at school.

**Question 11. What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?**

It is essential that patients should be associated with the governance and management of these resources. As well as providing data and samples, patients are the ultimate intended beneficiaries of these initiatives, and as such have a right to participate in decisions that concern them directly.

Common frameworks of governance should be developed that can be applied across rare diseases and countries. The international P3G Consortium ([www.p3gconsortium.org](http://www.p3gconsortium.org)) is developing models of governance and protocols for data sharing to maximise interoperability while allowing national flexibility.

Funding for registries, databases and biobanks should be sustainable to reap maximum benefit from the establishment of such frameworks and patients need to be reassured that commercial interests do not 'appropriate' the governance and funding for these resources.

**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?**

It is clear that both sectors should be heavily involved, so that industry develops good market knowledge and can be incentivised to invest, and so that patient organisations – representing those whom an action will aim to benefit – have a voice from the start.

Patients are the direct beneficiaries of work towards the development of new treatments for currently untreatable or incurable conditions. It follows that patients, as a group, are best placed to state what the priorities should be in the development of new treatments. In this

process of development, Patient Organisations should be consulted first, to ascertain what patients most need in a new treatment.

This is especially important for conditions with multiple symptoms. It is often not the primary symptom that patients most want a solution for. Evidence from unpublished research into clinical depression showed that patients' highest priority was a solution to the more practical problem of difficulties with concentration, rather than the central issue of happiness.

Patient input needs to continue throughout any developmental process, be it towards a new treatment, or towards incentives to deliver treatments; patients are the ultimate judge of the success or failure of this initiative. It is important that comprehension of patients' views and needs is not taken for granted at any stage; no assumptions should be made at any stage about patients' wishes.

**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?**

GIG strongly supports developing action plans to impact directly on services for patients and families with rare diseases, and on research and therapeutic development. Adoption of such national plans should be recommended by the EU. Within the UK, devolution of health services means action plans need to be developed at regional level (England, Wales, Scotland, Northern Ireland).

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

GIG supports EURORDIS in advocating the creation of such an Agency to ensure sustainable activity and funding in the essential field of rare diseases. Patients and families affected by rare diseases will continue to suffer inequitable access to healthcare and the products of innovation if investment in rare diseases relies on repeatedly securing awards for 3 to 5 year projects.