IPPOSI
Irish Platform for Patients' Organisations, Science and Industry

PUBLIC CONSULTATION

RARE DISEASES: EUROPE'S CHALLENGES

The Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) was set up to provide a platform for discussion in Ireland on the development of new therapies and treatments for unmet medical needs.

Background

In November 2007, IPPOSI, together with the Irish Medicines Board facilitated a meeting on Rare Diseases attended by patients' organisations, science, industry and clinicians. The participants agreed that the following steps would be important for Ireland to take:

1. Development of a national plan (Ireland) for orphan medicines, one which would comprehensively encompass development, pricing, access and care issues.

2. Recognition in this national plan of the need to establish one or more Centres of Excellence for rare diseases

3. In developing the national plan, Ireland should examine the activities of other European countries in this area but not necessarily replicate them. In the Irish context access to treatments for rare diseases ought to be driven by societal values rather than by economic considerations.

4. With proper incentives and promotion, orphan medicines could be the gateway to the further development of Ireland's knowledge economy.

5. Patients' organisations are central to the process of promoting awareness of, and research into, rare diseases. They must continue to push for development, and should be supported in their work.

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Irish Response to Consultation

On the 4th of February 2008, IPPOSI facilitated a discussion on the European Commission's Public Consultation document "Rare Diseases: Europe's Challenges" attended by key stakeholders in Ireland including patients' organisations, industry representatives, scientists and clinicians.
The responses to the following questions put by the EC represent a consensus view from this Irish meeting.

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

IPPOSI supports the current EU definition of Rare Disease being extended to the whole of Europe. While this is the IPPOSI position, we acknowledge, at a national level, that different definitions may apply.

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

There is a need for better codification and classification allowing adequate traceability of RD in the health systems.
Ireland currently uses the Hospital In Patient Enquiry System (HIPE) with reference to the ICD 10 (International Classification of Diseases) codes for cost weighting of services. As the WHO is coordinating the revision of ICD10 with a view to increasing codes and classifications of rare diseases, IPPOSI supports this initiative and recommends the adoption of ICD11, when developed with Rare Diseases classification in the EU member states. Better classification of rare diseases and improved appropriate cost weightings would hopefully improve resourcing and recognition of centres of excellence for rare diseases.

IPPOSI also draws attention to the fact that improved coding might impact on access to clinical trials.

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

Yes, there is a need for an accurate, regularly updated inventory of rare diseases together with an inventory of available resources and local and European Centres of Excellence. In Ireland, due to coding issues and the lack of patient registries for Rare Diseases obtaining accurate and reliable information is difficult, aggravating planning and the provision of resources to people with Rare Diseases.

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

IPPOSI supports the EURORDIS submission;

Centres of Expertise and/or Centres of reference (depending on the national policy in the various individual member states) as physical structures for the management and care of rare diseases patients at Member State level - and the European Reference Networks - as the "networking of knowledge and expertise" through either physical or virtual expertise and/or reference centres and teams of experts at the EU level - are fundamental to address the issue of rare diseases at European and national levels.

Transfer of knowledge should be privileged. Patients do not find it pleasant to travel to other countries on a regular basis for treatment. Patients prefer high-quality local care for a variety of reasons (e.g. To avoid language/communication or travel difficulties but also because of family/professional commitments).

However we believe that patient mobility should be facilitated (financially, administratively and logistically) where necessary. Situations where it is necessary to travel to have a second opinion at the moment of diagnosis – to receive important medical interventions (e.g. transplants, surgery) and the definition of therapeutic options or when a treatment is not available in the home country.

In addition to the above, IPPOSI believes that

- Centres of Reference within Europe need to be appropriately licensed with a centralised European authority established to examine each centre.
- IPPOSI agrees with EURORDIS that there are two essential pre-conditions for the establishment of a Centre of Expertise for Rare Diseases:
- Professional qualifications (stemming from clinical and scientific experience) must be documented by publications, grants, pre- existing certification or accreditation;
- There should be a serious commitment to cooperation and information sharing.

- IPPOSI place a further precondition on the establishment of Rare Disease Centres of Expertise and that is to ensure that patients are included in discussions and involved in consultations regarding the establishment of these centres.

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

There is a recognition of the important value of electronic tools and of the systematic use of IT technology particularly in establishing detail of drug usage in the treatment of rare diseases in Ireland.

IPPOSI agree that on-line tools should be implemented. Orphanet and NORD (National Organisation for Rare Diseases) websites are good examples of user-friendly and accessible on-line tools for patients and clinicians. They are important information tools for patients and provide a large range of up to date information on research on any particular Rare Disease. In Ireland evidence is emerging from a REHAB Ireland[1] study which found General Practitioners reporting difficulty locating accurate information on given Rare Diseases to provide their patients with. IPPOSI believe that in Ireland further development in this area is required. There is an urgent need to provide links to websites such as Orphanet through local sites as this would help to ensure access to information for all concerned parties ie patients, families, clinicians, patient organisations and government agencies.

Question 6. What can be done to further improve access to quality testing for RD?
IPPOSI agree on European quality control mechanisms.

Furthermore, if a particular disease is prevalent in any one member state there should be national screening and appropriate testing.

Question 7. Do you see a major need in having an EU level assessment of potential population screening for RD?
IPPOSI contend that recommendations are required from EU to member states on population screening for Rare Diseases.

Population screening policies (for example for treatable inborn errors of metabolism) vary widely between EU member states. Screening policies for each country might be determined by accepted international criteria for screening and different prevalences in each member state.

[1] The Rehab Group is an independent not-for-profit organisation working for social and economic inclusion among people with disabilities and others who are marginalised.

Guidance and direction is required in this instance and in others, from the EU to encourage the establishment of screening systems for Rare Diseases.

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

IPPOSI agree that the EU and member states must work together to provide patients with access to orphan drugs. The Orphan Regulation, based on the principles of societal equity and solidarity is much welcomed however issues around availability of, and degree of access to, treatments for rare diseases across the EU member states continue to arise. Identifying a meaningful and sustainable solution to this issue remains a challenge. Enhancing information and comprehensive understanding of the Orphan System across the EU member states among stakeholders could help to overcome some of these difficulties (particularly with regard to the unique provisions of the Regulation around issues such as (a) evidence that no satisfactory treatment is in existence or (2) that a new treatment is clinically superior to the one in current use).

Because of the rarity of many conditions, current methodologies for conducting HTA have proven to be problematic. It would be beneficial and greatly desirable if decisions taken by experts on the Committee for Orphan Medicinal Products (COMP) and other bodies (eg CHMP) were more widely known and understood by the relevant actors at individual member state level. This is particularly important for rare conditions where normal Health Technology Assessment methodologies are inappropriate. Consideration might be given to facilitating the approval for reimbursement at Member State level by using scientific assessment carried out by COMP prior to market authorization. It would be useful if best practice in the area of reimbursement measures across the EU could be examined. Affordability for patients is a key consideration.

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

There are some obvious advantages to having EU regulation provision in the area of medical devices and diagnostics not least because limited market size for orphan products can work as a disincentive. EU regulation in this area could support companies to invest in niche or non profitable applications which would be beneficial, particularly in the area of diagnostics for niche genetic disorders or for serious rare diseases where early detection could offset the disasterous effect of contracting the disease.

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?

Patient Organisations representing Rare Diseases tend to be small and underfunded. There is little awareness of rare diseases or their treatment within the wider community. There are examples of diseases which affect only one or two individuals in Ireland and as a result there are no networks, counselling facilities or patient groups available to provide support for these people. IPPOSI would ask the EU to recommend that EU member states support umbrella group organisations committed to supporting and advising people with Rare Diseases. FRAMBU in Norway is a good example of one such group providing social supports for patients and their families.

EURORDIS has identified - and is developing via RAPSODY (an EU-funded project) - the following services deemed particularly useful to patients and their care givers: respite care services, help-lines, therapeutic recreation programmes for children and young adults as well as integration at school. Eurordis gives a high priority to the European network of helplines.

Rare Diseases affect patients and their families. We estimate in Ireland that some 140,000[2] people have a Rare Disease and as a result 140,000 families are in need of information, support and counselling services to help them to deal with the impact these diseases have on their lives. A helpline for people with Rare Diseases and their families is a priority for Ireland.

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

The model of governance needs to be European-led and patients should be central to any structure of governance of these resources.

There needs to be a national ethical approval process for the collection of data on patient registries. Currently ethical approval has to be sought from each hospital and every patient has to give written approval to be on a patient registry. The only exception is the National Cancer Registry which is covered by specific legislation.

In the case of patient registries, changes will be required in national policy if the compilation of European registries for Rare Diseases is to be successful.

[2] IPPOSI/IMB meeting on Rare Diseases, Dublin, November 2007

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?

The IPPOSI model was put forward here as a successful model for bringing partners together. The idea for IPPOSI arose from the European Platform for Patients' Organisations, Science and Industry (EPPOSI) who seek to establish a strong European alliance of patients' organisations, academic science, including clinicians and industry jointly working on healthcare policies towards treatment and prevention of serious diseases.

The possibility of industry assisting with the funding of research for rare diseases in collaboration with recognised centres of excellence and universities might be another model that could be considered.

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?

IPPOSI advise that national action plans are vital for the provision of adequate services and information to people with rare diseases and their families. Recognition and appropriate funding of Centres of Excellence will also be important.

European recommendations and guidelines would be very helpful to the acceleration of the process in Ireland.

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

Yes, IPPOSI consider it is necessary to establish an agency and a feasibility study.

Numbers of patients and families affected by individual rare diseases throughout the EU are not as large as those affected by more prevalent diseases. Campaigns on issues of access to treatment and information for rare diseases are often not as successful as those for more prevalent diseases where numbers of patients would be considerably greater. A European solution to a national issue is required and the establishment of a European Agency is deemed to be a step in the right direction.

Additional issues which IPPOSI feel need to be considered as part of this important EU consultation on the area of rare diseases:

Plasma Derived Products.

IPPOSI members feel it important to highlight the area of Plasma Derived Products in this public consultation on rare diseases. IPPOSI members would request the Commission to protect and increase the level of supply of plasma derived therapies which are essential to the health and wellbeing of many patient groups.