



EURORDIS Position Paper on “Centres of Expertise and European Reference Networks for Rare Diseases”

EURORDIS - the European Organisation for Rare Diseases – represents 310 rare disease organisations from 34 different countries, 23 of which are EU member states, and thereby reflects the voice of an estimated 30 million patients affected by rare diseases in the European Union.

In response to the Commission Public Consultation “Rare Diseases: Europe’s challenges”, EURORDIS has developed a Position Paper on Centres of Expertise and European Reference Networks. EURORDIS has launched its reflection process on this issue at the Annual Membership Meeting in Berlin (April 2006, 180 participants from 20 countries). This reflection has been enriched and deepened through different steps in the last couple of years:

- through the achievements of the relevant Work packages (WP5 – WP8) of the Rare Disease Patient Solidarity Project (RAPSODY), funded by DG SANCO; 270 patient representatives, health care professionals and decision makers have participated in one-day national workshops in 11 EU Member states following the same methodology and agenda; 80 representatives from 11 countries participated in a two-days European Workshop of synthesis in Prague in July 2007; the final synthesis was presented at the European Conference on Rare Diseases 2007, in Lisbon;
- through EURORDIS participation in and dialogue with the EU High Level Group on Health Services and Medical Care - Working Group on European Reference Network, in 2006 and 2007;
- through the expertise built, shared and disseminated at the DG SANCO Rare Diseases Task Force;
- through the internal consultation process on which EURORDIS has based its contribution regarding Community action on health services, in November 2006.

This process has led EURORDIS to express the **utter need for Centres of Expertise and European Reference Networks for Rare Diseases**, given the



overall lack of expertise and the necessity of gathering the scarce knowledge for the benefit of a maximal number of rare disease patients. It is utopia to believe that each of the 5 to 8 thousands rare diseases could benefit from a specific Centre of Expertise in every EU Member State. The establishment of European Reference Networks is the appropriate way to exploit limited human, medical and scientific resources through a consistent and efficient approach that would equitably benefit EU citizens wherever they live.

Therefore, both the Centres of Expertise and/or Centres of reference (according to the national policy set up in the concerned Member States) - physical structures for the management and care of rare diseases patients at Member states 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.

It also has to be underlined that the Community added-value of establishing European Reference Networks is particularly high for rare diseases, given the direct consequences of rarity, which implies both limited number of patients and scarcity of expertise at national level, thus maximising the cost-effectiveness of rational quality healthcare provision and of common European research infrastructures such as registries, databases and biobanks.

The RAPSODY EU-funded project launched a dialogue between patients, health care professionals and both national and European policy makers, with the aim at addressing the needs and expectations primarily of patients and families, but also of the other stakeholders on Centres of Expertise and European Reference Networks for rare diseases. The present document is based on the main outcomes of the RAPSODY project and aims at pointing out the following thirteen elements as being fundamental in the current reflection on Centres of Expertise and European Reference Networks.

1. Pre-conditions:

Two essential pre-conditions are required for a Rare Disease Centre of Expertise:

- **Professional qualifications** - stemming from both clinical and scientific experience - that have to be documented by publications, grants, pre- existing certification or accreditation;



- Serious **commitment to cooperate** and share information.

The general “atmosphere and attitude” has also been underlined as an important element of success for a Centre of Expertise: trust - rather than competition - among experts is deemed necessary to ensure effective cooperation.

2. Multidisciplinarity:

Different stakeholders do recognise that only a **multidisciplinary approach** can be effective in providing adequate care to rare diseases patients. It is important to underline that “**care**” **includes both medical and social aspects** for the management of rare diseases patients. Rare diseases are complex and involve different medical specialties, as well as a wide range of paramedical healthcare professionals, working in close collaboration with social workers. Patients and families expect that Centres and Networks have to successfully meet the challenge of organising care through a multidisciplinary approach.

3. Different levels of coordination:

The importance of a good coordination between professionals has been stressed repeatedly, in particular:

- Coordination within and between centres of expertise, within European Reference Networks, and between centres of expertise and primary care centres;
- Coordination between care and research activities;
- Coordination with the aim at circulating information and organising continuum of activities by placing the patient at the centre of the system and making better use of existing expertise and resources;
- Coordination between various services in order to improve quality of care by reducing the psychological burden of patients (feeling lost in the system, lack of support, language barriers, administrative obstacles, etc).

4. Importance of a global and comprehensive approach:

There is a **need to integrate medical and social aspects, at all levels** (primary care centres, centres of expertise and European Reference Networks). It has been



recognised that social support is often underestimated and that there is a need to develop a common European approach to social services when they are specific for rare disease patients. Among the specific administrative tasks of the European networks, there must be the support in favour of patient mobility for cross-border care when necessary, especially concerning the reimbursement issue, which may prove insurmountable for some patients and families.

5. Capacity to pool patients:

A **critical mass of patients** is a necessary condition for improving scientific and medical knowledge on a disease: there has to be enough patients enrolled in clinical trials and this can only be possible, in the case of rare diseases, through European Reference Networks given the scarce number of patients within a single country.

6. Main expectations:

- The European Reference Networks will have a strategic role in the **harmonisation of care and the improvement of quality treatment** for all patients throughout the European Union: within the Reference Networks, the level of knowledge and expertise will be shared in different Centres. If needed at specific moments of the development of the disease, it will be considered as **“normal and fair” to travel from one Centre to another** within the same Network for confirming a diagnosis, seeking a second opinion or for important medical intervention (surgical operation, transplantation and other invasive medical interventions). It should not be an administrative, legal and medical fight for a patient to travel abroad for unwished medical reasons.
- The EU Networks will have a major impact on the development of **best practices recommendations, standards and guidelines** for diagnosis, treatment, care and social support of rare disease patients at international level. In fact, specific expertise will be further developed and identified, thus allowing confirmed recommendations to be based on “demonstrated experiences” within the Networks;
- The dissemination of European **reference diagnostic and therapeutic protocols, ensuring equity** at EU level by reducing the impact of the “post code lottery” and therefore increasing trust in local services;
- The provision of expert opinion, confirmation of diagnostic and therapeutic options.

7. Research activities at European and International level:



All interested parties consider that Centres of Expertise and European Reference Networks do **facilitate international research** by performing the following tasks:

- To link excellence of care with excellence of research, in the same “place” where patients are gathered and where multidisciplinary expertise on the disease can be found;
- To allow multi-centre clinical studies as well as partnership with pharmaceutical companies;
- To provide **shared research resources**: databases, biological resources (DNA, RNA, tissues, cells), registries (harmonisation of standard operating procedures), international epidemiological surveillance and pharmacovigilance;
- To facilitate participation in **EU-funded research projects**.

8. Perform education and training:

Centres of Expertise and European Reference Networks are expected to be instrumental in promoting education and training activities such as:

- **Information and communication** outreach activities towards the public, but also the primary health care professionals in order to improve referrals and follow up;
- **Training activities for health professionals**, including staff exchanges, meetings and conferences to exchange best practices, harmonise processes and disseminate standards and guidelines

9. Empowerment of patients:

Empowering activities for patients and their representatives have to be performed at different levels through **information, education and training**. They will help patients and families building their capacity to manage the medical and social aspects of their disease, enhance their autonomy, increase their compliance and generally improve their quality of life.

10. Collaboration with patient organisations:

Centres and European Reference Networks must cooperate closely with patients' representatives in the following ways:



- Patient organisations must be **actively involved in the management and evaluation** of both Centres of Expertise and European Reference Networks as experts for the production of information documents, guidelines for diagnostic and care, the choice of the research tools and clinical trials to be performed within the networks;
- Centres and EU Networks must facilitate the creation of patient groups when they do not exist;
- They shall improve relations and exchanges between healthcare professionals and scientists on the one hand, and patients on the other hand;
- Broad links between European Reference Networks, research networks, information networks and patient organisations should be fostered.

11. Evaluation:

European Reference Networks should be initially evaluated at EU level via an **agreed set of criteria** (minimum set of standardised criteria and objectives) and then regularly assessed on common indicators through both soft values and hard values. European Reference Networks could therefore play an active role in the evaluation of national centres of expertise and/or reference (according to the national policy set up in the concerned Member States). There is also a need to **develop methods and tools for European reference networks to perform regular self-evaluation**.

Proposed set of soft values for the evaluation of Centres of Expertise and /or Reference

- Cooperation with patient organisations
- Patient-oriented approach e.g. coordination, information to patients
- Improved outcomes
- Improved atmosphere
- Improved quality of life
- Avoiding unnecessary complications
- Awareness and knowledge dissemination
- Information provision to local centres

Proposed set of hard values for the evaluation of Centres of Expertise and/or Reference

- Time to get the diagnose



- Waiting time for medical consultations and tests
- Genetic consultation
- Multidisciplinary approach
- Cooperation with other centres and EU networks
- Guidelines and recommendations
- Quality control
- International and national networking
- Economic assessment

12. Funding, sustainability and governance:

European Reference Networks are perceived to be cost-effective. They need proper funding for their specific European and international activities. The funding should be a **long-term public funding in order to ensure their sustainability**. EU Networks should be encouraged to establish good governance structures (leadership, regulation, steering committee) and share coordination practice. They should be able to disclose their procedures and outcomes.

13. Flexibility:

It has been underlined that flexibility is required in relation to the geographical coverage of the Networks, as well as to the selection of type of centres belonging to the Networks. There should not be any obligation for a Network to have centres in all member states. The European Reference Networks should be able to identify the Centres of Expertise together with the national health authorities and the patients groups.

Conclusions:

- EURORDIS firmly believes that the establishment of Centres of Expertise and European Reference Networks will play a **key role for improving the lives of people living with a rare disease**, by facilitating the communication between various health care professionals involved in daily care management, improving and harmonising the organisation and provision of high quality care, reducing the time to obtain accurate diagnosis, agreeing treatment and care guidelines, providing expert advice and ensuring that patients have access to the best adequate technologies and treatment at specialised centres and/or through European Reference Networks. “*Expertise*



should travel rather than patients” who should travel in another Member State only when they consider it necessary.

- EURORDIS encourages the **mobility of knowledge**, which includes mobility of health professionals, as well as data, samples and expertise. Travelling abroad is not enjoyable for patients and their care-givers, often family members. **Patient mobility has to be supported and facilitated even if limited to key moments** of the development of the disease when patients and families mostly feel the need for a second opinion before important decisions and major therapeutic choices.
- EURORDIS also wishes to express its **appreciation for the inclusive process** that has been established by the European Commission for the EU-wide reflection on Centres of Expertise and European Reference Networks, with all relevant stakeholders.