



## **EURORDIS Position Paper on Specialised Services for People Living with 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 Specialised Services aimed at improving the daily lives of people living with a rare disease. This document is largely based on the outcomes of the Rare Disease Patient Solidarity project (RAPSODY) a major project led by Eurordis, involving 10 partners. The project - co-funded by the European Commission, the Baxter International Foundation and Sigma Tau Pharmaceuticals - aims at empowering the rare disease community, covering various aspects of the urgent need to improve quality and access to essential services for rare disease patients at European level. In particular, this paper takes on board the latest developments of the RAPSODY project about Help lines, Therapeutic Recreation Programmes and Respite Care Services, as they have been recently addressed during the European Conference on Rare Diseases held in Lisbon on 27-28 November 2007.

This document was finalised through consultation with the partners and networks’ members of the RAPSODY project, as well as the EURORDIS European Public Affairs Committee, representing a broad range of rare diseases and EU Member States.

Improving the quality of care, information and social services is instrumental to the empowerment of people living with rare diseases. Specialised Services in the field of rare diseases must be an important component of national strategies to be incorporated in National Plans for Rare Diseases.

1. **Information services and help lines** increase the opportunities for patients and carers to access and exchange relevant information on the disease they live with and manage daily.

At national level, existing patient-run help line services for rare diseases should be consolidated and long-term plans ensuring their sustainability should be put in place.



A unique EU-wide number for social services, a 116 number, should be established with the support of Member States and the European Commission.

2. **Online communities of patients** are a privileged means to create and maintain contacts among extremely isolated patients before they can be connected with the network. Among the most used tools are mailing lists or new tools which use the potential of the e-technologies.

Tools available to online patient communities in the field of rare diseases, such as e-health tools, should be implemented with the financial support of the European Commission and Member States.

3. **Therapeutic Recreational Programmes** give children the possibility to stop thinking about disease and treatment and to focus on fun and leisure, thus allowing personal development to thrive.

Member States should support Therapeutic Recreation Programmes specially adapted to the needs of children living with a rare disease and foster the creation of new Programmes. Exchanges between Programmes should be encouraged and staff should receive appropriate training.

4. **Respite Care Services** are provided on a short-term basis for disabled people who usually live at home. It gives family members and carers time and temporarily relief, prevents burn out.

Respite care services should be provided as part of a combination of services (centre-based, home-based, etc.), tailored to the needs of each patient. Awareness should be raised on the importance of such services and a cost/benefit analysis performed to demonstrate their positive impact on quality of life and health outcomes.

The European Networks of –respectively- Help Lines, Therapeutic Recreation Programmes, created thanks to the EU-funded RAPSODY project, should be supported beyond the lifetime of the project, to pursue their goals: awareness-raising, exchange of best practices and standards, pooling resources.

## 1. Specialised Services for Rare Diseases

**Improving the quality of care, information and social services is instrumental to the empowerment of people living with rare diseases. Specialised Services are an important part of national strategies for rare diseases.** This should be reflected in the European guidelines for the elaboration of **National Plans on Rare Diseases**, and in each one of the National Plans.



## 1.1. Rare Disease Help Lines

**Information services and help lines increase the opportunities for patients and carers to access and exchange relevant information on the disease they live with and manage daily.** The production of knowledge through this process is of invaluable importance in the field of rare diseases.

Patients wish to develop a European Network of Rare Disease help lines and information services run by professionals and volunteers. The quality of personalised advice will greatly benefit from a unique EU wide number for services of social value, the exchange of best practices and common tools, such as harmonised answer forms, the creation of a knowledge database, and training for individuals. Consequently, patients would be addressed to the most appropriate centre of expertise, eventually informed about the ongoing clinical trials relating to their disease. Emergency situations and mobility issues would be better handled. The services would also contribute to establishing contact between very isolated patients.

Actions to be undertaken:

- **Member States should develop or consolidate existing patient-run help line services for rare diseases to achieve a consistent level of service at EU level, and commit to long-term plans ensuring their sustainability.**
- **The European Network of Rare Disease Help Lines, set up thanks to the RAPSODY project, should continue to be supported beyond the lifetime of the project,** to pursue its goals: awareness-raising, exchange of best practices and standards.
- Help lines should develop common tools, a common regulation for validation of information and patient confidentiality. Existing resources should be made available in all European languages.
- **Member States and the European Commission should support the establishment of a unique EU-wide number for social services – a 116 number.** Once the number has been reserved at EU level, national governments should make sure that local help lines are financially supported.
- Funds should be released at both national and EU level for training and financial support of help line operators, and for training of doctors and other professionals, to improve knowledge of rare diseases.
- Finally, continued support should be provided to create and develop connections between European rare disease patients without diagnosis and without association, as they are indeed in the weakest and most exposed situation.

## 1.2. Online patient communities and e-health in the field of rare diseases



For many rare diseases, the number of patients in each Member State is very small. **Virtual communities** are thus a **privileged - if not the only - means to create and maintain contacts among extremely isolated patients before they can be connected with the network.** Among the most used tools are **mailing lists** or discussion lists, an email communication method for a group of people involved with a particular disease. Under the aegis of EURORDIS, 18 of such mailing lists have been created.

Patients' discussion lists aim to provide information, help in the everyday management of the disease, provide support to patients, their families and carers, break their isolation, share experiences, and possibly support their public interventions (media, scientific and medical circles, public organisations).

Beyond mailing lists, **new tools are nowadays available to patient communities** which use the potential of the e-technologies and the opportunities offered by the web: wikis, blogs, social networking tools... These instruments are promising insofar as they can expand the functions of mailing lists, reinforce online communities by creating stronger clusters of patients, promote the exchange of information and tailored services to patients. In other words, new tools **allow for a greater empowerment of patients.**

EURORDIS therefore supports the Commission's view that "e-Health tools are very efficient and should be a strong part of the EU strategy on RD".

Actions to be undertaken:

- **E-Health in the field of rare diseases (on-line and electronic tools) should be implemented** (Question 5 of the Public Consultation).
- The **European Commission should provide financial support for these activities** through the Public Health Programme and the Framework Programme for Research. **Such measures should also be supported at Member States level.**
- Existing resources should be made available in all EU languages with specific funding.
- An analysis should assess the comparative efficiency of the new e-Health tools on the basis of the real benefit for patient communities.

### **1.3. Therapeutic Recreation Programmes**

Most rare diseases affect children: about 50% of all people affected by rare diseases are less than 19 years old. Starting life with a condition that will impact quality of life, life expectancy, social relations, ability to move, to learn, to accomplish daily life activities is a hurdle, even more unbearable when invasive and complex medical interventions are required.



Personal development, education and learning cannot fully thrive if the life of a child is centred around a disease: children need to enjoy other activities. Children need to play, develop artistic skills, and they need to have fun with other children with or without the same condition.

Siblings also need attention. Leisure and recreational activities will help children gain self-confidence: it opens new fields of activities and new horizons in their lives.

Ultimately, children need a break. They need to spend some days in an environment where they can stop thinking about their disease, where they can meet, socialise and play with other children.

**Therapeutic Recreational Programmes have been created to give children the possibility to stop thinking about diseases and treatment and to focus on fun and leisure.**

Action to be undertaken:

- **Member States should support Therapeutic Recreation Programmes, specially adapted to the needs of children living with a rare disease. The creation of new Programmes should also be fostered.**
- The coordination of the Programmes' activities should be facilitated by European funds. In particular, the **European Network of Therapeutic Recreation Programmes, created thanks to the RAPSODY project, should be supported beyond the lifetime of the project**, to pursue its goals: awareness-raising on existing Programmes, exchange of best practices and standards, pooling resources.
- Exchanges between Programmes should be encouraged, so that children benefit from a larger offer of activities and locations, thus favouring cultural and language exchanges; for this purpose the movement of patients across European borders should be facilitated.
- Staff should benefit from training programmes supported both at national and European level to accommodate and to educate children and young adults from different age groups (age ranging from 6 to 25 years old), and also to take care of their medical conditions.

#### **1.4. Respite Care Services for Rare Diseases**

Living with a rare disease or caring for a child with a rare disease affects its immediate family, social network and relatives. Facing everyday life challenges causes considerable concern, and strain. The rarity of the disease brings additional stress, as families often have to face the lack of knowledge amongst professionals in crucial areas such as medical, social and pedagogical.



**Respite care is provided on a short term basis for disabled people who usually live at home. It gives family members and carers time and temporarily relief, prevents burn out.** Another important purpose is to reveal new abilities of the person living with the disease/disability, allowing him/her to try and perform recreational and meaningful activities.

Respite care services can be offered in various ways, notably: residential respite, i.e. staying with a “respite care family” for a while; domiciliary care; day care centres, nursing homes, institutions or respite care group homes with assisted living facilities; emergency respite services accessible on short notice when unexpected emergency occurs.

Action to be undertaken:

- **Provide respite care services as part of a combination of services (centre-based, home-based, etc.), tailored to the needs of each patient;**
- Raise awareness about the importance of respite care services among decision-makers and professionals at both national and European level;
- Carry out a cost/benefit analysis to demonstrate the utility of such services and their positive impact on quality of life and health outcomes;
- Make a larger range of services available, such as resource centres, sheltered workshops, supported accommodation, residential services, services for people with autistic behaviours...;
- **Support the European Network of Respite Care Services, created thanks to the RAPSODY project, beyond the lifetime of the project,** to pursue its goals: awareness-raising on existing services, exchanges and collaboration among centres, identification of best practices.