



# RARE DISEASES: ADDRESSING THE NEED FOR SPECIALISED SOCIAL SERVICES AND INTEGRATION INTO SOCIAL POLICIES

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*“Placing one foot in front of the other, I've climbed to higher lengths. Reaching beyond my own limitations, to show my inner strength. No obstacle too hard, for this warrior to overcome. I'm just a man on a mission, to prove my disability hasn't won.”*

Robert Michael Hensel, Born with Spina Bifida  
Guinness World Records holder for the longest non-stop wheelie in a wheelchair (6.178 miles)

**«When you have a rare disease it feels like you are so alone and no one cares»**

Janet, mid 50's, living with Alkaptonuria, USA



**«My wife, Svitlana, Yuliya's mother is a wonderful woman! Her work deserves other publication – "how mothers fight with a Rare Disease at home". There is no professional help at home – for our category, a staff is not stipulated, there are no volunteers»**

Vitaly Matyushenko, father of Yuliya, 5 years old, living with Spinal Muscular Atrophy, Ukraine

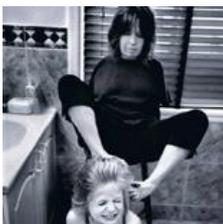
**«I hope that there will be a good and nice place for her when she leaves home, that there will be nice people around her who will take good care of her, especially when we – her parents – are not here anymore. That some nice people will bring her birthday presents, bring her home for Christmas, and take her to see places that will bring her joy»**



Jane Villemoes, mother of Cecilie, 14 years old, living with Angelman Syndrome, Denmark

**«MP has so many medical appointments, and therapy sessions each week that I had to stop working. I have only 4 hours free to come back home, do the cleaning, cook, go to supermarket, deal with the infinite bureaucratic processes that we have to deal with to get a special school, special social wealth assistance and ask for national families budget support. Then, I pick him up again, come back home and accompany him in all the exercises his therapist has given him. I go to bed exhausted and I don't get a lot of help at home. I'm an architect, I loved my work and I miss it a lot! At this moment, it is impossible for me to find a job»**

Sandra, mother of MP, 3 years old, living with Congenital Disorder of Glycosylation, Spain



**«Our home looks like any other but if you pay attention, we have special tools. In the kitchen for example, our cupboards have a button instead of a knob and with a simple push, the shelves come down. My wife has a special chair that can go high and enable her to reach things with her feet and to close the house, we have an electronic remote control»**

Björn Håkansson, , 52 years old, living with Thalidomide, Sweden

**«Services provided seem to differ from area to area. It is not possible to get a 'check list' of all the people you need to talk with. Also, service providers differ in the amount of interest they show»**



Denis Ryan, husband of Anne, living with Huntington Disease, Ireland

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## EXECUTIVE SUMMARY

The following paper has been written in the context of the European Union Committee of Experts on Rare Diseases (EUCERD) Joint Action Working for Rare Diseases (2012-2015) Work Package 6, focused on “Specialised Social Services and Integration of Rare Diseases into Social Policies and Services”. For this reason, the approach to the issues described in the paper is limited to the scope of the work performed within the Joint-Action, not always comprising all the social services and social policies needed for People Living with Rare Diseases (PLWRD).

The confrontation with a Rare Disease (RD) is a life changing and often devastating event, both for PLWRD and for their relatives, generating additional social support needs. Due to different constraints, in various countries, PLWRD and their families might find themselves without an appropriate and inclusive social support framework or without access to the existing social benefits.

This paper derives from real testimonies of PLWRD about their social conditions and difficulties, encouraging reflection on these issues. It also outlines the need for Specialised Social Services that can support PLWRD, as well as the need to integrate RDs into social policies so to address social needs.

On behalf of PLWRD in Europe, EURORDIS urges decision-makers to facilitate the access of PLWRD to Specialised Social Services and to promote social policies addressing their needs. The focus of these reflections and actions shall not necessarily be the development of new Specialised Social Services specific to RDs, but rather the access of PLWRD to existing Specialised Social Services where they can be integrated – such as, for example, services for people with disabilities. However, reflection on Specialised Social Services specifically addressing RDs is also encouraged, within the framework of the development of Centres of Expertise, providing multidisciplinary care to PLWRD and gathering expertise that should be available to local medical and social care providers.

Equally essential is the need to ensure that services themselves can be provided with access to guidelines and good practices facilitating the work of the social services providers when integrating PLWRD, providing valuable tools to existing Specialised Social Services as well as to new services.

The need for social policies goes beyond the present review. PLWRD and patient advocates have now constantly been reminding policy makers that there is a need for social policies that promote the autonomy and the quality of life of PLWRD such as the recognition of the diagnosis and its consequent incapacities influencing access to school, employment, housing and financial support.

The urgency to address the social challenges faced by PLWRD increases as diagnosed people face longer life expectancies and greater life expectations, due to the advances in the therapeutic and medical fields. Nowadays PLWRD face longer life spans, different expectations towards an autonomous and fulfilling life. Families, also face challenges for longer years, leading to a crescent need of social support and benefits.

The current global economic situation and its impact on the different Member States (MS) generate additional social challenges for PLWRD, as many of these are not able to find comfortable professional/occupational situations or provide for themselves, increasing the economic overall burden of the supporting family.

The need to integrate RDs into social policies shall be reflected on, considering the several baseline documents and data collections made so far, in which European and national authorities, patient groups, PLWRD and families have manifested the need for such policies to be generated and to become a reality of everyday life.

This document will provide an insight on the need to address Specialised Social Services and to advocate for the integration of RDs into social policies at EU level, which can then be reinforced at MS level. This insight will be based on a literature review of different documents, surveys and studies, developed at EU level, at patient representatives level and at academic level. From these documents, special attention will be dedicated to the following:

- **Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's Challenges (November 2008);**
- **Council Recommendation on an Action in the Field of Rare Diseases (June 2009);**
- **Communication from the Commission: European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010);**
- **EURORDISCare Survey Programme (2002 - 2008);**
- **EUROPLAN final report recommendations based on the 15 EUROPLAN National Conferences (2010-2011);**
- **Federación Española de Enfermedades Raras' study on the Situation of Social-sanitary Needs of PLWRD – ENSERio (2009);**
- **"An investigation into the social support needs of families who experience rare disorders on the island of Ireland" performed by RehabCare (2008);**
- **Caller Profile Analysis performed by the European Network of Rare Disease Help Lines (November 2011).**

The document will also provide a definition of different types of Specialised Social Services currently available which can integrate PLWRD and their relatives:

- **Respite Care Services:** short term care for PLWRD so that family members and carers can experience temporary relief from the stress of care giving while making it possible for PLWRD to live according to their usual daily routine;
- **Therapeutic Recreation Services:** any organised recreation activity (summer camp, *ad hoc* trip) giving PLWRD the possibility to take a break from thinking about diseases and treatments and focus on fun and leisure;
- **Adapted Housing:** group homes aiming to help PLWRD to enjoy the highest possible level of autonomy, in their own home, where they are isolated and are supported by specialised staff;
- **Resource Centres:** a combination of information provision, social and medical services, often under partnership or cooperation with Centres of Reference/Expertise. These centres are specialised in handling rare complex cases and provide services such as training courses, information and guiding, information about social services, documentation and research, daily support therapies, medical and psychological consultations;

- **Complex Case Managers:** an element that can ensure that there is a better coordination in the care pathway of PLWRD, functioning as a link between the medical and the social needs of the patient, particularly in situations most complex due to the course of care and the need for the intervention of multiple structures and professionals;
- **Other habilitation services** supporting PLWRD in their daily life or in their complementary therapeutic procedures, increasing PLWRD's autonomy and quality of life.

This document will then address EURORDIS' role concerning Specialised Social Services and social policies, namely in the context of the EUCERD Joint Action Working for Rare Diseases:

- **Mapping of Specialised Social Services:** identification and mapping of existing services and identification of good practices; organisation of visits to services in different MS ; development of case studies based on best quality existing services; online publishing of collected information, making it accessible to decision makers, patient advocates, families and PLWRD;
- **Assessing Training Needs of Social Services Providers:** description of training needs for social services providers and identification of good practices in this field; sharing of resources and information collected with Specialised Social Services, decision makers and patient advocates;
- **Integration of RDs into Social Policies:** identification of existing social guidelines and dissemination of methods, tools and good practices' validation processes;
- **Elaboration of the EUCERD report on guiding principles for social care in RDs and draft of EUCERD Recommendations in the social field.**

The goals of the EUCERD Joint Action "Working for Rare Diseases" concerning Specialised Social Services and social policies will be attained by involving EUCERD members, MS decision makers, the Council of National Alliances for Rare Diseases (CNA), the Council of European Federations for Rare Diseases (CEF) and other patient groups and advocates.

Active involvement of all stakeholders will be promoted through different activities, including presentations, country visits and workshops.

EURORDIS encourages decision-makers, patient representatives, PLWRD and families to engage into the reflection on how access to Specialised Social Services and Integration of RDs into social policies can be achieved in each MS, in cooperation with national authorities, decision makers, patient representatives/advocates, families and PLWRD.

## ADDRESSING THE NEED FOR SPECIALISED SOCIAL SERVICES AND INTEGRATION INTO SOCIAL POLICIES

### 1) THE EXPRESSED NEED FOR SPECIALISED SOCIAL SERVICES AND SOCIAL POLICIES

Over the last years, attention around RDs has been mostly focused on diagnosis, access to treatment, access to proper health care, research on orphan drugs and new treatment options, concentrating on the urgent medical issues that surround PLWRD. However, while attention was being focused on these important matters, other issues have risen, as PLWRD face longer life expectancies and greater life expectations.

Social issues as a whole have been constantly reported as a main concern among PLWRD and families, therefore becoming a concern of patient organisations and decision makers.

This Paper, built upon findings and reflections carried out over recent years, provides a robust analysis of the need for PLWRD to access Specialised Social Services and of the need to integrate RDs into social policies at EU and national levels.

To achieve this analysis it is required to revisit some of the main documents and data findings that state the need for the integration of RDs into social policies and the need for Specialised Social Services. This insight will be based on a literature review of different documents, surveys and studies, developed at EU, patient representatives and academic levels.

#### 1.1 Literature Review – Social Policies

One of the findings of the EURORDISCare Survey Programme was that «social security systems are usually designed around common diseases and are not flexible enough to take into consideration unprecedented health needs» (EURORDIS, 2009). Similar conclusions have been found in various studies performed by several sources, targeting diverse populations and using different methodologies.

In the framework of the Network of Public Health Institutions on Rare Diseases, a European project, funded by the European Commission, 302 questionnaires were completed by patients and caregivers in France, Italy, Romania, Spain, Turkey and United Kingdom during 2004-05. «In general, respondents thought that health care accessibility was worse than quality and that social care and legal provisions were worse than health care, with some differences among countries» (Kodra, *et al.*, 2007).

The ERES Study which asked over 700 PLWRD in Spain to analyse the socio-sanitary impact of RDs in the country, also stated that «patients were more satisfied with doctor's care than social worker's (47% and 2.4% respectively), considering doctors more accessible than social workers (32.4% and 13.1% respectively)» (Avellaneda, *et al.*, 2007).

Finally, a smaller scale study entitled "You have to sit and explain it all, and explain yourself" performed in the United Kingdom, together with mothers of adults with Angelman, Cornelia de Lange, or Cri du Chat also brought some interesting conclusions into the debate of the importance of integrating RDs into social policies. Some themes emerged from the analysis of the interviews

performed: including uneven medical and social care service provision, and the inertia of social care services. These mothers also reported that accessing appropriate social care services was a lengthy and complex process (Griffith, *et al.*, 2011).

Researchers performing another study in Spain, on the “Needs in rare diseases during paediatric age” have indeed come to an important conclusion, by stating that «social needs are becoming more relevant in developed countries where health care services, even with limitations, have greater availability than social services. Furthermore, it seems that health care and social services for persons with rare diseases need to be improved to address the patients' needs and to provide better support to families» (Gaité, *et al.*, 2008).

Other six sources deserve particular attention, due to their relevance and dimension, and to their specific findings, allowing further comprehension of PLWRD reported social needs at EU and MS levels:

- Council Recommendation on an Action in the Field of Rare Diseases (June 2008);
- Communication from the Commission: European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010);
- EURORDISCare Survey Programme (2002-2008);
- Federación Española de Enfermedades Raras' study on the Situation of Social-sanitary needs of PLWRD – ENSERio (2009);
- “An investigation into the social support needs of families who experience rare disorders on the island of Ireland” performed by RehabCare (2008);
- Caller Profile Analysis performed by the European Network of Rare Disease Help Lines (November 2011).

### **1.1.1 Council Recommendation on an Action in the Field of Rare Diseases (June 2009)**

In 2009, the European Council adopted the Recommendation on an Action in the Field of Rare Diseases, defining the EU's strategy in the field of RDs.

In this Council Recommendation, social care is referred to under the recommendation «I. Plans and Strategies in the Field of Rare Diseases», section a): «elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems».

Social care is again referred to in the recommendation «V. Gathering the Expertise on Rare Diseases at European Level», section 17): «Gather national expertise on rare diseases and support the pooling of that expertise with European counterparts in order to support: (a) the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases».

### **1.1.2. Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions, European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010)**

Considering the number of RDs that generate some mental or physical disability, it is of utmost importance to mention this document and some of its findings. Among other issues, this Communication from the Commission stated that:

- For people with disabilities the rate of poverty is 70% higher than the average, partly due to limited access to employment;
- The rate of employment for people with disabilities is only around 50%;
- Policy and regulatory frameworks do not reflect the needs of people with disabilities adequately, neither do product and service development. Many goods and services, as well as much of the built environment, are still not accessible enough;
- In the 16-19 age group the rate of non-participation in education is 37% for considerably restricted people, and 25% for those restricted to some extent, against 17% for those not restricted. Access to mainstream education for children with severe disabilities is difficult and sometimes segregated.

In this document, the Commission has identified eight main areas for action, concerning the European Disability Strategy: Accessibility, Participation, Equality, Employment, Education and Training, Social Protection, Health, and External Action. The Commission also brought attention to the following needs:

- To ensure accessibility to goods, services including public services and assistive devices for people with disabilities;
- To improve the accessibility of sports, leisure, cultural and recreational organisations, activities, events, venues, goods and services providing quality community-based services, including access to personal assistance.

### **1.1.3 EURORDISCare Survey Programme (2002 - 2008)**

The EURORDISCare Survey Programme was performed in three different stages, in 23 countries, addressing 18 RDs: EURORDISCare1 from November 2002 to April 2003; EURORDISCare2 from September 2003 to June 2006; and EURORDISCare3 from May 2006 to April 2008. This large international survey, with over 20.000 copies distributed, involving 130 patient organisations provided a massive data collection on PLWRD health and social conditions (EURORDIS, 2009).

The survey shows that not only Specialised Social Services and social policies have become a priority at EU policy level but, mostly, they have been a priority for PLWRD and families for quite some years already.

Namely, the survey concluded that:

- Every year, 28% of the PLWRD needed the assistance of a social worker. For about one-quarter of these, access to this assistance was difficult: difficult access (18%), very difficult (9%) or even impossible (4%);
- 92% of PLWRD consider that «informing patients about their rights and guiding them towards social services, schools, leisure activities or vocational guidance» is essential (55%) or useful (37%);
- Globally, social assistance services respond inadequately to the expectations and needs of PLWRD (only 37% are satisfied), especially when the demands are specific to the disease. This inadequacy of the social assistance is more severe for the low income PLWRD (only 26% satisfied);
- On average, 16% of PLWRD (up to 24% for the low income group) were forced to move house because of their disease;
- 90% of PLWRD consider that «creating material for teachers, employers, social services, insurance companies and the general public to inform them about patients' needs and improve social integration of patients» is useful or essential;
- 92% of PLWRD consider that «training local professionals to respond to the specific needs of patients» is useful or essential;
- PLWRD expect that specialised centres which integrate medical care and specific social services linked to the rarity of the disease will provide training for local professionals as well as information material about their disease and guide them in order to improve social integration.

#### **1.1.4. Federación Española de Enfermedades Raras' study on the Situation of Social-sanitary needs of PLWRD – ENSERio (2009)**

This pioneering study has been released by the Spanish Rare Disease Patient Federation, FEDER. This study goes deeper into the reality of those affected by RDs in Spain. The concerns expressed by patients surveyed in this study, reinforce once again the need to integrate RDs into social policies.

A sample of the conclusions from this study is presented below:

- The services that patients and families enquired mostly use are in fact the general health services (75%). However, a fair amount of PLWRD also feels the need to attend general social services (46%) and leisure/recreational services (20%);
- Patients enquired mentioned that they generally need support for their daily basic and advanced activities:
  - o In domestic life, 46% of the patients enquired;
  - o In their transport mobility, 42%;
  - o In their personal mobility and posture, 40%;
  - o In their leisure and free time activities 37%;
  - o In their educational or professional activities 39%;
  - o And in their self-care 32%;
- Only 1 in 10 patients stated that he/she would not need any sort of assistance (Garcia, *et al.*, 2009).

### **1.1.5 RehabCare's Study, An investigation into the social support needs of families who experience rare disorders on the island of Ireland (2008)**

RehabCare's investigation included an extensive literature review on these issues and a study performed in Ireland, surveying patients at a national level. From the literature review it is important to highlight the following:

- «Van Nispen *et al* (2002), for example, in their study with rare disease patients in the Netherlands, found that approximately 40% of their sample of people with rare diseases wanted to get more information on the treatment or on the financial aspects of their illness. The authors stated that there was an indication that people with rare diseases were far more likely to face difficulties with housing and finance» than people with more prevalent diseases (quoted by McGarvey and Hart, 2009);
- «Financial pressures are seen amongst families experiencing all disabilities. Redmond *et al* (2000) in their study of parents of children with disabilities found that 15 out of the 17 cases reported a financial pressure on families due to the loss or diminution of the mother's income» (quoted by McGarvey and Hart, 2009);

The investigation performed by RehabCare with families and patients in Ireland brought other elements into evidence and discussion. The study conclusions are stated below:

- There is often a loss of income in families where a child has a RD. RDs by their nature can be 'chronically debilitating' and some participants in this research stated that they were forced to cease fulltime employment to care for their child (McGarvey and Hart, 2009);
- More than half of the participants stated that they experienced financial difficulties due to having a child with a disability. Owing to the high level of support needed by many children with RDs, in many cases one or more parents had to give up work to take care of their children. (...) For an individual with a RD, financial loss was reported as almost always a reality (McGarvey and Hart, 2009);
- Due to the lack of information and support for people with rare disorders many of the participants initially had great difficulty getting information on their entitlements (McGarvey and Hart, 2009);
- Some participants felt that the carer's allowance was insufficient to replace the loss of a fulltime income and that they were struggling to survive (McGarvey and Hart, 2009).

The authors then concluded that «people need practical support on financial issues and that the provision of help and advice on financial issues is essential» (McGarvey and Hart, 2009).

### **1.1.6 Caller Profile Analysis performed by the European Network of Rare Disease Help Lines (November 2011)**

Another important data collection, deriving directly from the field work in different MS is the Caller Profile Analysis (CPA) performed under the activities of the European Network of Rare Disease Help Lines (ENRDHL). This CPA included 11 help lines from 7 EU countries plus 1 EU candidate country. The information was gathered by each help line over the month of November 2011, accounting for a total of 1739 enquiries.

In this CPA, 9.3% of the requests made by families and PLWRD concerned “Social Care”, being the third biggest subject enquired about, after “Information on Disease” (31%) and “Centres of Expertise/Experts” (16.4%). PLWRD are therefore actively contacting local help lines to get support in accessing social care and social services, expressing this need as one of their current main worries.

## **1.2 Literature Review – Social Services**

Hennepe (1999), in his study “Rare diseases need structured care: the diagnosis is just the start” stated that «a multi-disciplinary approach is needed for this population [PLWRD and families]. Many family participants in this research felt that people with RDs are often unsuitably placed within generic services. Some participants felt that there was an expectation for them to fit into these more generic services and that this often did more harm than good in terms of their progress. The majority of the family participants in this research felt that there was a huge need for all services to respond to the unique and complex needs of rare disorder patients» (quoted by McGarvey and Hart, 2009).

A study performed by the Istituto Superiore di Sanità (National Centre for Rare Diseases), in 2007, entitled “Accessibility and quality to health social services in Italy for the patients with rare diseases: the opinion of associations” evidenced that there was a high level of dissatisfaction with availability, quality and integration of health and social services in Italy (Agazio, *et al.*, 2005).

In Ireland, participants in the study performed by RehabCare reported that «services for rare disorders were too scattered and felt that a one-stop shop approach to social support and therapy provision was needed» (McGarvey and Hart, 2009). The authors also mention that «some of the participants felt that they had to constantly chase up services and experienced frustration at the fragmentation of services for people with rare disorders. The large number of people involved in the process of acquiring a service was also frustrating» (McGarvey and Hart, 2009).

The inequity of access to these services and their disparities in quality were also mentioned by the participants of this study, referring to «the location of services as being a barrier to access». Some patients «felt that there were better services in urban areas and that they were forced to travel for these services» (McGarvey and Hart, 2009).

Research from RehabCare also suggested that «flexibility and person-centred approaches which fit the service to the individual’s specific needs should be adopted by all service providers, as the very nature of rare disorders means that often they will not blend effectively with generic disability services» (McGarvey and Hart, 2009).

Other documents also deserve special attention, concerning their findings on the need for Specialised Social Services which can integrate PLWRD:

- Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's Challenges (November 2008);
- EUROPLAN Final Report Recommendations based on the 15 EUROPLAN National Conferences, >2200 participants (2010-2011) ;
- Federación Española de Enfermedades Raras’ study on the Situation of Social-sanitary needs PLWRD – ENSERio (2009).

### **1.2.1 Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's Challenges (November 2008)**

In 2008, the European Commission adopted the communication on RDs entitled "Rare Diseases: Europe's Challenges". Section 5.2 of this communication specifically addresses the «Access to Specialised Social Services» mentioning Centres of Expertise «essential role in developing or facilitating Specialised Social Services which will provide the quality of life of people living with a RD».

The communication further defines that Respite Care Services and Therapeutic Recreation Programmes «need to be sustainable to pursue their goals: awareness-raising, exchange of best practices and standards, pooling resources using Health Programme and Disability Action Plans».

### **1.2.2 EUROPLAN Final Report Recommendations based on the 15 EUROPLAN National Conferences, >2200 participants (2010-2011)**

As stated in the guidance document developed by the EUROPLAN's partners and experts, «Specialised Social Services are instrumental to the empowerment of people living with RDs and improve well-being and health. For people living with a rare, chronic and debilitating disease, care should not only be restricted to medical and paramedical aspects, but should also take into account social inclusion and psychological or educational development. Therapeutic Recreational Programmes encourage personal development. Respite Care Services give family members and carers downtime opportunities».

The final report of EUROPLAN's National Conferences (NC) clearly states that «mechanisms need to be devised to recognise and integrate PLWRD into existing social services (rehabilitation, integration into school and workplaces, recreation and respite services), while recognising their specificities and providing quality services in response to their needs».

Within the EUROPLAN's recommendations R6.5, R6.7, R6.9 are specifically addressing Specialised Social Services and social policies:

- «R6.5 - Specialised Social Services are a support for people living with a chronically debilitating rare disease and their family carers». This recommendation states that «mechanisms need to be devised to recognise and integrate PLWRD into existing social services (rehabilitation, integration into school and workplaces, recreation and respite services), while recognising their specificities and providing quality services in response to their needs».
- The same recommendation also states that «it is necessary that the importance of specialised services for PLWRD and families is recognised. It is also demanded that their support through public money becomes a priority for the whole community. Evaluation of the services provided should be carried, quality systems should be adopted as well as guidelines, and staff should receive adequate professional training. Guidelines and best practices developed at the EU level (...) should be used and supported further»;
- R6.7 suggests that «a directory of centres providing Specialised Social Services, including those offered by patients' associations, is compiled, kept updated and communicated to national, regional and patients' websites», focusing on the need to spread awareness and information about existing Specialised Social Services which can integrate PLWRD and families;

- R6.9 raises awareness about the need for developing information and education materials for specific professional groups dealing with PLWRD (i.e. teachers, social workers, etc.), keeping in mind that social services providers need training as well in order to be able to provide better care and resources to PLWRD and families.

### **1.2.3 Federación Española de Enfermedades Raras' study on the Situation of Social-sanitary needs of PLWRD – ENSERio (2009)**

The main conclusions of this study regarding Specialised Social Services are the following ones:

- A fair amount of the PLWRD enquired felt the need to attend general social services (46%) and leisure/recreational services (20%);
- 24% of the PLWRD/families enquired reported that their housing facilities were not adjusted to the patient's needs and comfort. In fact housing adaptation was stated as the sixth main investment made by patients/families affected by RDs;
- PLWRD felt discriminated when accessing leisure and recreational programmes (32%). This perception then reinforces the need to invest in the integration of RD patients into Therapeutic Recreation Programmes.

## **2) SPECIALISED SOCIAL SERVICES**

When referring to Specialised Social Services, it is important to remember the different types of services that have been identified at EU level.

The services can be listed as:

- Respite Care Services (RCS)
- Therapeutic Recreation Programmes (TRP)
- Adapted Housing (AH)
- Resource Centres (RC)
- Other habilitation services supporting PLWRD in their daily life or in their complementary therapeutic procedures, increasing PLWRD's autonomy and quality of life.

### **2.1. Respite Care Services**

Respite care is provided, on a short term basis, for PLWRD who either live at home or attend a respite centre, so that their carers can have a short relief from care giving. Many of the PLWRD involved might otherwise require permanent placement in a facility outside their home.

One of the important purposes of respite is to give family members and carers temporary relief from the stress they experience daily while providing care for a family member living with a RD. Respite care enables the caregivers to maintain the ability to continue care giving. The benefits to carers

described in the literature also fall into these two broad categories: stress reduction and self-esteem increase; and improved family functioning (Merriman and Canavan, 2007).

A second purpose of respite is to make it possible for PLWRD to live according to their usual daily routine and to provide a place to experience and perform recreational and meaningful activities away from their parents/other caregivers. Benefits to service users mentioned in the literature are socialisation, and enjoyment of experiences outside the home (Merriman and Canavan, 2007).

Respite exists all over Europe under different names and programmes. However many countries are not familiar with the concept and as a result services are not available to PLWRD in these countries.

Respite Care Services (RCS) can be offered in various ways:

- Residential respite: the person living with the RD goes away to an adapted centre to be looked after by someone else, a “respite care family”, for a while;
- Domiciliary care: some services offer a caregiver who comes to the family home, and take over care giving duties for a while so that the regular carer can have a break from the daily routine of care giving;
- Day care respite: day care centres, nursing homes, institutions or respite care group homes with assisted living facilities (no overnight facilities);
- Emergency respite: services that give access to respite on a short notice in the event of an unexpected emergency.

Respite is one of the services most often requested by family caregivers, yet it is in critically short supply, inaccessible, or unaffordable regardless of the age or disability of the individual needing assistance. While the focus has been on making sure families have the option of providing care at home, little attention has been paid to the needs of the family caregivers.

Without respite, not only can families suffer economically and emotionally, caregivers themselves may face serious health and social risks as a result of stress and exhaustion associated with continuous care giving. Three fifths of family caregivers aged 19-64 surveyed recently by the Commonwealth Fund<sup>3</sup> reported fair or poor health, one or more chronic conditions, or a disability, compared with only one-third of non-caregivers.

EURORDIS believes that every person living with a RD and every carer have the right to respite. The long term implications of providing respite care involve benefits for carers and PLWRD. The families will become better carers because of the relief respite provides and due to the exchange of experience with respite care providers while PLWRD will increase their life quality. Respite often prepares PLWRD to live more independently in the present but also as grown-ups. In this way, the quality of the overall care provided will improve.

## **2.2. Therapeutic Recreation Programmes**

Therapeutic Recreation Programmes (TRP) for PLWRD are any organised recreation activity (summer camp, *ad hoc trip*) which gives PLWRD the possibility to take a break from focusing on their disease and treatment to concentrating on fun and leisure.

There are three main formats that TRP may follow:

- 'Single illness' TRP: that integrate people living with a specific condition, or for siblings of patients with a particular condition;
- 'Mixed illness' TRP: that include people living with a variety of chronic conditions, or siblings/spouses of patients with different conditions;
- 'Mainstream or integrated' TRP: that involve people living with chronic conditions, siblings/spouses of patients with chronic conditions, and people not affected by any illness.

TRP tend to be residential, with some services conducting one session per year, and others conducting sessions on a year round basis. TRP sessions are relatively short in duration, typically lasting between 7 and 14 days. However, TRP length can range from half a day to 3 months.

The scope of individual TRP varies in a number of respects. While camping programmes provide recreational activities, the range of recreational activities offered differs, being influenced, for example, by factors such as the TRP's geographic location, financial considerations and the age range of participants. However, common activities include arts and crafts, canoeing, adventure, camping and horse-riding.

Leisure and recreational activities will help PLWRD gain self-confidence: opening new fields of activities and new horizons in their lives. Ultimately, PLWRD and their close ones need time off. 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 PLWRD and families. TRP allow PLWRD and their families to meet these needs in a safe, friendly and well-adapted environment.

The study "Outcomes associated with participation in a therapeutic recreation camping programme for children from 15 European countries: Data from the Barretstown Studies" mentions that «benefits were noted in their [patients'] experience of physical symptoms, affect pertaining to physiological hyperarousal and quality of life in the short and longer term. Positive changes were also noted in relation to self-esteem as it pertains to global self-worth and physical attractiveness» (Kiernan, Gormley and MacLachlan, 2004).

The study then concluded that «these findings clarify previous research and suggest that camping programmes have an important role to play as a complementary intervention in facilitating adjustment to chronic illness» (Kiernan, Gormley and MacLachlan, 2004).

TRP extreme importance for siblings has also been already reported. Siblings tend to share common difficulties which include a lack of understanding and knowledge about their brother or sister's disorder, also the feeling of being left out and potential embarrassment at their brother or sister looking and behaving differently (McGarvey and Hart, 2009).

Perrin (1999), in a study carried out in the Republic of Ireland, described how siblings who were involved in age-appropriate 'sibshops' where they could meet other children who have siblings with a disability were encouraged to share experiences, ask questions and learn coping strategies. The author concluded that the children involved in the groups were extremely well-adjusted; they exhibited feelings of happiness, appreciation, consideration and love for their family and brother or sister (quoted by McGarvey and Hart, 2009).

TRP can then become a tool to not only benefit patients but also their relatives, if involved in proper shops were they can meet peers and share difficulties and strategies to cope with these. More literature can be found on the need for peer support among parents and family members consulting the same study by McGarvey and Hart (2009).

### **2.3. Adapted Housing**

Adapted housing and related services represent a particular type of service, often associated to multi-handicaps. Sometimes called “therapeutic apartments”, these services allow PLWRD to develop and enjoy some level of autonomy within the comfort of their own home, alone or with some peers, under needed supervision of supportive staff, rather than being placed in an institution.

Adapted housing might also be a specific local/regional grant awarded to the patient’s family in order to pay for any house adaptation work, so to keep families from moving into other facilities or in order to adjust regular buildings to certain specific needs (wheelchair, small size, hearing disabilities, autistic spectrum disorders, etc.).

Adapted Housing services make it possible for PLWRD to live as independently as possible, being monitored by supportive staff habilitated to provide assistance for any daily routine activities that cannot be performed independently. PLWRD can thus enjoy a high level of independence and autonomy, still integrated in a community with some peers and not jeopardising their safety or their clinical and therapeutic needs.

### **2.4. Resource Centres**

A new type of service has been developed in several countries: a combination of information, social and medical services, generally defined as Resource Centre for RDs. Due to its main mission, this resource represents a Specialised Social Service.

Actions performed by Resource Centres for RDs more specifically target PLWRD, often under partnership or cooperation with Centres of Reference/Expertise or themselves being a Centre of Reference/ Expertise.

Resource Centres’ services include training courses, information and guidance services, provision of information about social services, documentation and research. Daily support therapies, medical and psychological consultations are often also provided by these centres.

These centres commonly create a bridge between PLWRD/families and all the stakeholders involved in patient care, such as medical services, rehabilitation and therapeutic services, social services and social support authorities, education professionals and other professionals directly working with PLWRD.

On the social level, Resource Centres provide guidance and support to PLWRD in accessing their rights and offer them social empowerment on different levels. Additionally, these centres provide training, guidance and information to different carers, including not only the PLWRD’ relatives but also social services providers and adapted education teachers. By serving all these groups, the Resource Centres have an essential role in the improvement of the global social care of PLWRD.

Resource Centres can then be seen as a service specialised in handling rare complex cases. These services can also coordinate with other complex case managers located at other regional or national services, assuring proximity support to PLWRD and families.

These services can as well be considered complementary to medical services, helping to fulfil the multidisciplinary mission of Centres of Expertise, as suggested by the Communication from the Commission and by the Council Recommendation on an Action in the Field of Rare Diseases.

These services could fit into the description of the «one-stop shop style of service for rare disorders» referred to by the participants in the RehabCare Study “An investigation into the social support needs of families who experience rare disorders on the island of Ireland”, being able to provide «flexibility and person-centred approaches which fit the service around the individual’s specific needs» (McGarvey and Hart, 2009).

The Nordic models, such as the Norwegian model in particular, can serve of great inspiration with respect to resource centres.

## **2.5. Complex Case Managers**

A number of PLWRD require special individualised social support due to the specificities of the diagnosis and its consequences.

The Complex Case Manager concept has been emerging in some countries, France being one of the examples. The section A-5-1 of the II French National Plan for RD is dedicated to “Complex Case Managers or Insertion Technicians”.

Complex Case Managers are defined in the French National Plan as the element that can ensure that there is a better coordination in the care pathway of PLWRD, functioning as a link between the medical and the social needs of the patient, particularly in situations most complex due to the course of care and the need for the intervention of multiple structures and professionals.

Complex Case Managers are promoted as a solution for PWLRD that require a strong link between medical professionals, supporting health professionals (nurses, occupational therapists, dieticians, etc.), medico-social services (psychologists, social workers, carers) and different other institutions responsible for social support or benefits (social security, employment institutions, local authorities, schools).

These professionals will then work as facilitators of the coordination of the care pathways of PLWRD guaranteeing the best articulation between the several services which support the person living with a RD in the course of their life.

The French Association for Myopathies (AFM) has started a project of such kind, employing over 150 Complex Case Managers, with the mission of: informing PLWRD about the disease and its consequences; making the link between the family and the diverse specialists that are involved in the management of the diagnosed person; identifying the needs of the family and facilitating the search of solutions for these needs.

## **3) EURORDIS’ ROLE IN ADDRESSING SPECIALISED SOCIAL SERVICES AND INTEGRATING RARE DISEASES INTO SOCIAL POLICIES**

Rare Diseases are a priority area for action in the European Commission’s Public Health Programme (2008-2013). The European Union Committee of Experts on Rare Diseases (EUCERD), formally established via the European Commission Decision, 30 November 2009, is mandated to assist the European Commission (EC) in formulating and implementing the Community's activities in the RD field, and to foster exchanges of relevant experience, policies and practices between the MS and stakeholders.

EUCERD has now launched a Joint Action “Working for Rare Diseases” (EJA), funded by DG Sanco, engaging one main partner and eight associated partners (all being EU Member States, except EURORDIS as Patient Organisation), composed of 8 Work Packages addressing different issues and current challenges around RDs.

EURORDIS is responsible for the Work Package 6 (WP6), dedicated to “Specialised Social Services and Integration of RDs into Social Policies”.

The EJA will specifically address the following priority areas:

- Enhancing visibility and recognition of RDs;
- Contributing to the development and dissemination of knowledge on RDs from specialised research, to the support of healthcare professionals and the empowerment of PLWRD;
- Contributing to improvements in access to quality services and care, from diagnosis, to care and social support and innovative therapies.

To achieve its aims, the EJA is constructed in such a way as to add value to the many investments which have already taken place at EU level, by building on the achievements of projects that have received EU funding in the past years. The EJA will comprise five main areas of work:

- The implementation of plans and strategies for RDs at national level;
- The standardisation of the RDs nomenclature at international level;
- The mapping of the provision of Specialised Social Services and integration of RDs into mainstream social policies and services;
- The leveraging of the value of EU networking for improving the quality of care for PLWRD;
- The integration of RDs initiatives across thematic areas and across MS.

The EJA work will be supported by a variety of methodologies, including literature reviews, questionnaires or Delphi procedures, dissemination of information and web based exchange platforms, consensus workshops and generation/validation of specific recommendations.

The expected outcome is an integrated strategy for the implementation of RD policies through the exchange of experience between national health authorities already involved in RD policy definition/implementation and via a series of recommendations from the EUCERD, clearly communicated to national policy makers, patient organisations and learned societies.

EURORDIS’ role will then be to support the EUCERD in this EJA, contributing on the issue of Specialised Social Services and Integration of RDs into social policies and Services, through the specific work package described below.

### **3.1. EUCERD Joint Action “Working for Rare Diseases” Work Package led by EURORDIS**

The WP6, “Specialised Social Services and Integration of RDs into Social Policies and Services”, aims to address PLWRD’ and families’ “real-life” social challenges, through working within the following framework:

### **3.1.1. Specialised Social Services**

- Respite Care Services: mapping; facilitating online access to information on existing services (online shared tool), sharing good practices and guidelines;
- Therapeutic Recreational Programmes: mapping; facilitating online access to information on existing services (online shared tool), sharing good practices and guidelines;
- Other rehabilitation services, including adapted housing: mapping; facilitating online access to information on existing services (online shared tool), sharing good practices and guidelines

### **3.1.2. Integration of RDs into Social Policies and Services**

- Training: describing training needs for social services providers and identifying/sharing existing practices in this field;
- Social guidelines: identification of existing social guidelines; disseminate methods, tools, and validate processes of good practices.

### **3.1.3. Work Package's Main Activities**

- Collection of contacts and research on existing Specialised Social Services;
- Update of online information on Specialised Social Services;
- Presentations at the European Conference for RDs 2012 Brussels and at CNA and CEF meetings;
- Country visits to familiarise with existing Specialised Social Services and assess good practices;
- 3 workshops on Specialised Social Services, Training of Social Services Providers and Social Polices, in December 2012, November 2013 and November 2014, respectively;
- Elaboration of the EUCERD report on guiding principles for social care in RDs / draft of EUCERD Recommendations in the social field.

### **3.1.4. Work Package's Outcomes**

- Dissemination of information on different types of Specialised Social Services and their benefits for the RD community and for polyhandicap community;
- Increased awareness for the need to Specialised Social Services;
- Increased visibility of existing Specialised Social Services at international and national levels, among key policy makers, PLWRD, researchers and physicians;
- Dissemination of information on good practices and guidelines for implementing and running Specialised Social Services;
- Provision of information on existing services which constitute case studies;
- Increased awareness of the need to train Specialised Social Services providers and provision of guidelines in order to perform such training;
- Integration of RDs into social policies and services across MS.

#### **4) HOW TO MAKE SPECIALISED SOCIAL SERVICES AND INTEGRATION OF RARE DISEASES INTO SOCIAL POLICIES A REALITY**

The need for Specialised Social Services and for the Integration of RDs into social policies and Services has been repeatedly identified and expressed by different stakeholders - PLWRD and families, EU authorities, health and social care professionals, academic researchers -, as stated in different surveys and data collections presented in this current Paper.

Furthermore, there is absolute evidence that efforts are being made at different regional, national and international levels, in order to build up structures, systems and services which can indeed satisfy these PLWRD' and families' needs.

Such systems, services and structures demand enormous coordination of efforts between PLWRD/families, patient representatives, national authorities and EU. EJA's WP6 is the tip of an iceberg for which EURORDIS assumes responsibility: for the primary mapping, collection of guidelines and recommendations. However, such identification of services and guidelines will require the participation and active involvement of PLWRD /families, patient groups, representatives and advocates, National Alliances, European Federations, services providers themselves and other advisers in order to actually turn into reality this integration of RDs in social policies and services.

EURORDIS therefore encourages all of these entities to actively join this on-going exercise which is part of the implementation of its mission and aims at providing PLWRD and families with better long life care, essential support and hope.

National Alliances shall feel free to use the present literature review to advocate for the sustainability or creation of Specialised Social Services and social policies at national level.

In the context of the current EUROPLAN II activities and the conferences planned for the coming year, an effort shall also be made to include the thematic of Specialised Social Services and social policies into the outline of the conference programmes, so to incentive discussions around the subject, preferably involving the national, regional and local competent authorities in the discussions.

Furthermore, as National Plans are being drafted, commented and approved, there is a higher chance of advocating for the inclusion of social policies and Specialised Social Services into the National Plan. EURORDIS therefore advises all advocates to strive for the inclusion of a representative from authorities competent for social policies and services in the National Plan work group, as a starting point. The drafting group shall also be encouraged to guarantee that social policies and Specialised Social Services are not only mentioned in the National Plan but also assigned with a corresponding budget allowing the development and sustainability of the proposed measures.

It is expected as well that the current project within the EUCERD Joint Action can serve as an exchange of experiences among countries, allowing the sharing of information on good practices, quality services and different sustainability and governance methods, increasing the chances of success of existing and future Specialised Social Services and social policies for PLWRD.

EURORDIS further advocates for the promotion of other initiatives, grants and projects that reinforce the promotion of Specialised Social Services and integration of RD into Social Policies at EU and national levels.

## 5) REAL LIFE TESTIMONY ON THE BENEFITS OF A SPECIALISED SOCIAL SERVICE



**«We were relieved to know that professionals were taking care of him in a place adapted to his needs. If we leave him with family, only for a few hours, they don't always know what to do and their homes are not adapted for his wheelchair or his grabbing objects, etc. We knew that the respite centre was a safe place for him to freely move around. While he was gone, we realised that the 200 sq. meters activity room is probably the biggest place in the world that Pierre has to move around and crawl around freely - wherever he wants to go! How wonderful for him! The smiles he had across his face, discovering, helped us know that he was in the right place.**

As all parents with children who go away to camp, we had more time to ourselves. We were able to take time for breakfast, without having to rush and get Pierre ready. We were able to think about planning time to see friends or even travel close by without worrying about all of the constraints related to travelling with a wheelchair and a child who never enters a room without being noticed. We're not easy guests to invite over or to travel with...

**And, although I knew that I would be able to physically rest, without having to lift him and dress him and change him, I underestimated the psychological and mental rest it would also provide... I didn't have to worry constantly about where he was, what he could grab, how he could hurt himself or hurt others, what he needed, which medication to give him, organise, dress, clean... take care of him... He was being taken care of by someone else for the first time in 6 years overnight.**

We will definitely use the service again. We continue to communicate with them in order to help them better care for him. They are able to tell us the problems and we try to offer solutions from our experience with him. Since Pierre cannot communicate verbally, we try to exchange our ideas about what he is trying to express and what he is telling them he wants and needs to do.

**I would highly recommend respite care to other parents. It is hard to let go and trust others to take care of our children, but we also need it. We're allowed to need it and to want it. It gives us another view about our child's needs from a team of professionals as well as allows us to take a step back at how we care for him. It helps to rejuvenate us to helping him to develop. We are so physically and mentally exhausted from just responding to his day-to-day needs that we don't always have the energy to think about how we could help him more... With this rest we are able to think about ourselves, feel better about ourselves and therefore become better parents for Pierre.»**

**Lara Chappell, Mother of Pierre, 6 years old, living with Angelman Syndrome, France**

## 6) TABLE OF ACRONYMS

ACRONYM	Expanded Meaning
CPA	Caller Profile Analysis
CEF	Council of European Federations
CNA	Council of National Alliances for Rare Diseases
DG Sanco	Direction Générale de la Santé et des Consommateurs
EC	European Commission
EJA	EUCERD Joint Action: Working for Rare Diseases
ENRDHL	European Network of Rare Disease Help Lines
ENSERio	Estudio sobre situación de Necesidades Sociosanitarias de las personas con Enfermedades Raras en España
EU	European Union
EUCERD	European Union Committee of Experts on Rare Diseases
EUROPLAN	European Project for Rare Diseases National Plans Development
FEDER	Federación Española de Enfermedades Raras
MS	Member State(s)
NP	National Plan
RCS	Respite Care Services
RDs	Rare Disease(s)
TRP	Therapeutic Recreation Programmes

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