



IDENTIFYING SOCIAL CHALLENGES FOR PEOPLE LIVING  
WITH RDs:  
Discussion of the EURORDIS position paper



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*“Rare Diseases: understanding this Public Health Priority” - 11/2005*

***Social consequences: living with a rare disease has implications in all areas of life, whether school, choice of future work, leisure time with friends, or affective life. It may lead to stigmatisation, isolation, exclusion from social community, discrimination for insurance subscription (health insurance, travel insurance, mortgage), and often reduced professional opportunities (when at all relevant);***

The **social aspects and implications of rare diseases also have to be kept in mind:** the territorial and financial services to support families and patients have to be organised and developed on a local basis, such as day care services, respite centres, emergency units, socialisation and rehabilitation centres, summer camps, education services and professional training. The problems related to the “after us” - when the life-long carer and/or parent of a rare disease patient disappears - have to be tackled and brought to the attention of national and European decision-makers.

The current experiences in this field have to be evaluated and valid organisational and managerial models have to be defined. It has to be underlined that the challenges and problems in relation to the social services do last for the whole life of a rare disease patient and become so important that medical aspects of the disease can be given second line priority.

*“Rare Diseases: understanding this  
Public Health Priority” - 11/2005*

*Families and health care workers frequently complain about the extreme difficulty in taking the necessary **administrative steps required to receive social benefits**.*

*Major and arbitrary disparities exist between countries - and even between regions within the same country - in the allocation of financial aid, income support and reimbursement of medical costs. In most cases, a significant proportion of these expenses is born exclusively by the families, thereby generating an **additional inequality**.*

*It is also important to underline that, in a family where a child has a rare disease, most often one of the parents – usually the mother – either completely stops or significantly reduces work remunerated outside home. As a consequence, **while expenses increase dramatically, incomes is considerably reduced**. In the case of an adult rare disease patient who is well enough to be able to work, the work hours must be adapted to allow for medical visits and appropriate care. In terms of logistics, much remains to be done to ensure real equality between a disabled and a healthy citizen. It is well accepted that impairment leads to a disability if the environment and regulations do not take into account the special needs of people with impairment to participate in society. The impairment is a part of our being. The **disability comes from outside by disabling factors**.*

*“Rare Diseases: understanding this Public Health Priority” - 11/2005*

*EURORDIS Position Paper on Specialised Services for People Living with Rare Diseases - 02/2008*

*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.*



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*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.*

*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 ehealth tools, should be implemented with the financial support of the European Commission and Member States.*

*“Rare Diseases: understanding this Public Health Priority” - 11/2005*

*EURORDIS Position Paper on Specialised Services for People Living with Rare Diseases - 02/2008*

*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.*

*“Rare Diseases: understanding this Public Health Priority” - 11/2005*

*EURORDIS Position Paper on Specialised Services for People Living with Rare Diseases - 02/2008*

*EURORDIS Position Paper on Integratio of RD into Social Policies - 2014*

*Rare disease patient solidarity project - RAPSODY*

*EUCERD JOINT-ACTION, WP 6 – Provision of Specialised Social Services and Integration of RD into Social Policies and Services*

*FUTURE PROJECTS - searching for funding and patnership opportunities*



*“Rare Diseases: understanding this Public Health Priority” - 11/2005*

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*FUTURE PROJECTS - searching for funding and patnership opportunities*

Issues have been identified :

- With EURORDIS board of directors
- With EURORDIS European Public Affairs Committee (EPAC)
- Via the Joint Action work package 6

And are sustained by data and quotations from:

- EU documents
- EUROPLAN I Final Report
- EURORDISCare Surveys
- Joint Action/EUCERD Documents
- Several published studies compiled in the [literature review paper](#) on social policies

1. Lack of **long term, funded and sustainable policies and structures at national levels and in the national strategies** for the integration of people living with RD into social services and policies
2. Weak **coordination between health and psychosocial complementary care, between central and regional/local infrastructures, and between sectors and between Ministries**, leading to a consequent lack of multidisciplinary holistic approach
3. Lack of systems to accurately **evaluate patients' disability degree and consequent lack of adequate compensation measures**
4. Lack of **information and understanding of disabilities by professionals and structures** resulting from the medical conditions and **implications on daily life** of patients and families
5. **Insufficient compensation measures leading to, among other things, financial difficulties**
6. **Geographic disparities** in access to social care and compensation measures both between and within countries

7. Lack of **information and support to patients** concerning their rights and the administrative/bureaucratic issues to handle in order to access their rights and proper compensation
8. Insufficient sharing of **best practices**
9. **Lack of training of social sector professionals** to deal with rare, complex cases resulting in unprepared services and structures
10. **Scarcity** of social services and social policies/benefits and **difficult access** to those
11. **Insufficient quality** of services provided
12. Difficulties in **accessing and keeping a professional activity**
13. Difficulties **accessing education**

14. Lack of **Case Managers** guiding patients/families (like a GPS navigation system) to access the different types of care and structures needed to achieve their autonomy and to follow their life plan
15. Lack of **personalised/flexible measures** and policies **helping patients' and families to pursue their life project**
16. Lack of **measures to remove some burden from family in daily care** (personal assistants, day time structures etc.)
17. Lack of systems to deal with **transition from childhood to adulthood** and **ageing** of adult populations, considering the psychosocial consequences for patients and families
18. **Discrimination** in access to services



# SOCIAL CHALLENGES IDENTIFICATION EXERCISE: ISSUES IN DETAIL

# CHALLENGES:

## 1. Lack of long term, funded and sustainable policies

- «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.



## Final Report



# 1. Lack of long term, funded and sustainable policies

- «social security systems are usually designed around common diseases and **are not flexible enough** to take into consideration unprecedented health needs» **(EURORDIS, 2009). EURORDISCare Survey Programme**



- «*Plans and Strategies in the Field of Rare Diseases*», 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**». Council Recommendation on an Action in the Field of Rare Diseases (June 2009)
- «*Policy and regulatory frameworks do not reflect the needs of people with disabilities*» Communication from the Commission, European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010)



# 1. Lack of long term, funded and sustainable policies

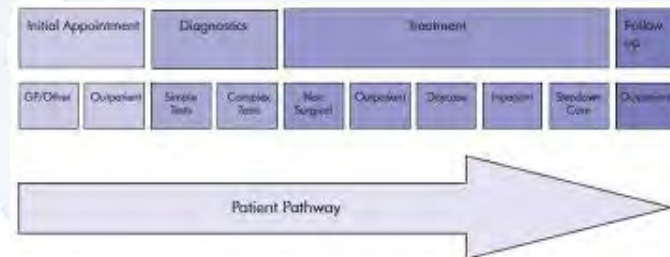
- «*Respite Care Services and Therapeutic Recreation Programmes need to be sustainable to pursue their goals: awareness-raising, exchange of best practices and standards, pooling resources using Health Programme and Disability Action Plans*». European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's Challenges



## 2. Weak coordination between health and psychosocial care, between central and regional/local infrastructures → lack of multidisciplinary holistic approach

- «a multi-disciplinary approach is needed for this population [PLWRD and families]» Hennepe (1999) quoted by McGarvey and Hart (2009)
- «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.»

- «high level of dissatisfaction with availability, quality and integration of health and social services in Italy»
- Istituto Superiore di Sanità (2007), study “Accessibility and quality to health social services in Italy for the patients with rare diseases: the opinion of associations”





### 3. Lack of systems to accurately **evaluate patients' disability degree** → **lack of adequate compensation measures**

- In recent years, there has been an increased emphasis on development of the scientific basis of functional capacity evaluation (FCE). *Ex. The Orphanet Disability Project: Indexing the functional consequences of rare diseases with the International Classification of Functioning, Disability and Health (ICF).*
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- Functional limitations are the effect of the patient's impairment on his or her ability to perform meaningful tasks.
  - The term functional connotes performance of a purposeful, meaningful, or useful task that has a beginning and an end with a result that can be measured.
  - Functional limitations are the proximal cause of disability.

## 4. Lack of information and understanding of disabilities by professionals and structures resulting from the medical conditions and implications on daily life of patients and families

- *«Patients enquired mentioned that they generally need support for their daily basic and advanced activities:*
  - *In domestic life, 46% of the patients enquired; 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) ENSERio*
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- *In their transport mobility, 42%;*
  - *In their personal mobility and posture, 40%;*
  - *In their leisure and free time activities 37%;*
  - *In their educational or professional activities 39%;*
- *«On average, 16% of PLWRD (up to 24% for the low income group) were forced to move house because of their disease» EURORDISCare Survey Programme*

## 5. Insufficient compensation measures leading to, among other things, financial difficulties

- «For people with disabilities the rate of poverty is 70% higher than the average, partly due to limited access to employment» Communication from the Commission, European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010)
  - «people with rare diseases were far more likely to face difficulties with housing and finance» Van Nispen *et al* (2002), study with rare disease patients in the Netherlands
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- «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**» Redmond *et al* (2000) quoted by McGarvey and Hart (2009)
  - «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) RehabCare's Study, An investigation into the social support needs of families who experience rare disorders on the island of Ireland (2008)

## 6. Geographic disparities in access to social care and compensation measures

- «patients «felt that there were better services in urban areas and that they were forced to travel for these services» (McGarvey and Hart, 2009) RehabCare.





## 7. Lack of information and support to patients

- «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%)»  
EURORDISCare Survey Programme

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



## 8. Insufficient sharing of **best practices**

- *«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».*  
**Council Recommendation on an Action in the Field of Rare Diseases (June 2009)**

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## 9. Lack of training of social sector professionals

- *«92% of PLWRD consider that «training local professionals to respond to the specific needs of patients» is useful or essential»* **EURORDISCare Survey**
- *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)*  
**EURORDISCare Survey Programme**

## 9. Lack of training of social sector professionals to deal with RD

- *«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.» EURORDISCare Survey*
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- *«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.» EUROPLAN Final Report*

## 10. Scarcity of social services and social policies/benefits and difficult access to those

- «Every year, 28% of the PLWRD needed the assistance of a social worker. For about 1/4 of these, access to this assistance was difficult: difficult access (18%), very difficult (9%) or even impossible (4%)» EURORDISCare Survey Programme
  - «Accessing appropriate social care services is lengthy and complex process» (Griffith, et al., 2011) "You have to sit and explain it all, and explain yourself", study in the UK with RD families
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- «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) RehabCare's Study, An investigation into the social support needs of families who experience rare disorders on the island of Ireland (2008)

## 11. Insufficient quality of services provided

- *«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»* (Gaite, et al., 2008).



## 12. Difficulties in accessing and keeping employment

- *«The rate of employment for people with disabilities is only around 50%»*  
Communication from the Commission, European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010)
- *«Some participants in this research stated that they were forced to cease fulltime employment to care for their child»* (McGarvey and Hart, 2009)

RehabCare



## 13. Difficulties accessing education

- «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.» Communication from the Commission, European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010)

## 14. Lack of Case Managers guiding patients/families

- «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). RehabCare



## 15. Lack of **personalised/flexible measures and policies** helping patients' and families to pursue their unique life project

- «*Many goods and services, as well as much of the built environment, are still not accessible enough*» Communication from the Commission, European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe (2010)
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- «*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). RehabCare

## 16. Lack of measures to remove some burden from family in daily care (personal assistants, day time structures etc.)

- There are only some measures for adapting the criteria for handicap also to rare conditions in order to better consider the benefits for the disabled person and the family. Nordic countries might be an example in this area (Norway, Finland, Denmark) and also the measures from French National Plan & Orphanet Disability projects;

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## 17. Lack of systems to deal with transition from childhood to adulthood and ageing of adult populations, considering the psychosocial consequences for patients and families

- Transfer arrangements to adult facilities, which vary considerably between clinics and countries, are usually needed due to regulations governing access to pediatric services. Transition from childhood to adulthood in Duchenne muscular dystrophy (DMD); Sunil Rodger<sup>1\*</sup>, Birgit F Steffensen<sup>2</sup> and Hanns Lochmüller<sup>1</sup>

## 18. Discrimination in access to services

- «*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.*» **ENSERio (2009)**
  - *There is no disease so rare that it does not deserve our attention!*  
Orphanet

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- **Genetic discrimination** occurs when people are treated differently, for example by their employer or insurance company, because they have or are perceived to have a gene mutation that causes or increases the risk of an inherited disorder. It may also refer to any and all discrimination based on the genotype of a person rather than their individual merits. **Wikipedia, the free encyclopedia.**





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