Commission Expert Group on Rare Diseases

RECOMMENDATIONS

TO SUPPORT THE INCORPORATION OF RARE DISEASES INTO SOCIAL SERVICES AND POLICIES

April 2016

1. BACKGROUND TO THE RECOMMENDATIONS

Providing holistic care to people living with Rare Diseases (RD) is particularly challenging because:

- Expertise and knowledge on RD and their consequences are scarce and difficult to access;
- RD are often chronic, highly complex, severely disabling and frequently affect life expectancy[1];
- No specific treatment is currently available for most RD and existing treatments are not always able to minimise all the complex impairments generated by the disease;
- A high percentage of people with a RD are affected by motor, sensorineural or intellectual impairments, which can occur simultaneously[2];
- Even when they are not associated with a disability, RD will in many cases influence the person's health condition and impact on daily life in a disabling way[3].

This recommendation focuses specifically on those RD that generate complex impairments. The combination of rarity, complexity and lack of effective treatment creates huge obstacles to the provision of holistic care and in many cases significant medical, psychological and social needs remain unmet.

People with a RD often need follow up care and support from different categories of health professionals, often from several different medical specialities, as well as by social workers and other social and local service providers[4] which requires a level of coordination not easy to organise in most health care systems.

Professionals lack knowledge of the condition and the appropriate routines[5] as the standard of care is not established for most diseases. Additionally, service-providers within a range of sectors seem to be reluctant to get involved in managing diseases that are unknown to them[6].

Studies have demonstrated that the quality of life of people with a rare chronic disease compares unfavourably to that of people affected by more prevalent chronic disorders, both physically and psychosocially[7] and that RD patients have a more negative experience in terms of medical care and of loss of social-economic activities[8].

Failure to meet the serious unmet social needs of people with a RD and their families affects people's dignity, autonomy and other fundamental human rights expressed in the Universal Declaration of Human Rights and in the United Nations Convention on the Rights of Persons with Disabilities[9].

The results of the EURORDIS Care Survey[10] clearly demonstrate some of the social challenges that patients' and families' face: 1/3 of patients and an additional 1/3 of the families interviewed had to reduce or stop their professional activity due to the disease; 1/3 of patients/families required assistance from a social worker in the year preceding the survey; 1/3 of families experienced difficulties in accessing a social worker or did not meet one at all; 1/5 of families had to move house to relocate to one better adapted to their health needs.

The EUROPLAN final report based on 15 EUROPLAN National Conferences (2010-2011) recommends that «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 and educational development» and recognises that «social services are instrumental to the empowerment of people with a RD and to improve their well-being and health»[11].

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[1] A. Schieppati et al., 2008.
[2] Guillem P. et al., 2008 - France, 1739 children with RD: a RD was at the origin of 26% of cases of severe neuromuscular impairment; proportion of impairments due to a RD: 3.3% severe psychiatric disorders; 16.0% intellectual impairment; 37.2% hearing impairment; 41.2% neuromuscular, skeletal, movement impairment; 81.1% visual impairment; Tozzi A, et al., 2013 - Italy, 516 parents of children with a RD: nearly 70% of children had a disability (49.2% motor; 33.3% intellectual; 22.4% both).
[3] Grut L, Kvam MH, 2013.
[4] EURORDIS, 2009 – average patient required more than nine different medical services, over the two-year period preceding the survey.
[5] Kemper, A.R et al., 2006 quoted in Berglund, B. et al., 2010.
[6] Grut L, Kvam MH, 2013 & EURORDIS, 2009 - a majority of patients reported a reluctance of professionals to treat them due to the complexity of their disease.
[7] Nispen RMA van et al., 2003 quoted in van Weely S, Leufkens HGM, 2008.
[8] Nispen RMA van et al., 2003 quoted in A. Schieppati, et al., 2008.
[9] Universal Declaration of Human Rights, articles 22, 23, 24, 25, 26, 27; United Nations Convention on the Rights of Persons with Disabilities, Preamble e), n), r) and x) and Articles 16, 17, 18, 19, 24, 25, 26, 27, 28, 30.
[10] EURORDIS, 2009.
[11] EUROPLAN final report recommendations based on the 15 EUROPLAN National Conferences, 2010-2011.

In addition, it is important to address, remove and prevent barriers in the environment that hinder the participation of people with disabilities generated by RD in society, on an equal basis with other citizens, notably in respect of education, employment, daily life activities, accessibility and mobility.

Effectively coordinated integrated care and support services, involving health, social and local services as well as the community at large, are essential to overcome the particular challenges of RD and to ensure that people affected by a RD can secure the assistance they require from mainstream social and local services.

2. KEY PRINCIPLES ON WHICH THESE RECOMMENDATIONS ARE BASED

- Health is a state of complete physical, mental and social well-being[12];
- Social dimension, and with it, social services contribute to core values and objectives of the EU Member States and of the European Union, such as achieving a high level of employment, social protection, health protection, gender equality, and economic, social and territorial cohesion[13];
- Social services aim at improving citizens' quality of life and at providing social protection. They assist vulnerable individuals and persons who have a range of special needs and risks, such as those needing long term care or facing disability, poverty and those at risk of social exclusion[14];
- Social care consists of helping people to participate fully in all aspects of life, particularly those who require extra help. Social care workers contribute to help individuals maintain their independence, which increases their quality of life and helps them lead fuller more enjoyable lives[15];
- Social services are often closely interlinked with health services. Experts consider that there is a need for a chain of actors and providers to take care of individual needs and to provide solutions in an integrated and coordinated manner[16].

3. SCOPE OF THE RECOMMENDATIONS

These recommendations mainly focus on empowering health services' attempt to facilitate integrated care provision to enable them to play the role they need to play in supporting the incorporation of RD specificities into mainstream social and support services, within a holistic and person-centred approach and a human rights perspective.

More comprehensive information on social support in other environments, such as education, employment and leisure can be consulted on the RD-Action paper on "Social Care and Inclusion for People Living with a Rare Disease".

4. METHODOLOGY FOR THE ELABORATION OF THE RECOMMENDATIONS

These recommendations were developed within the European Union Committee of Experts on Rare Diseases (EUCERD) Joint Action (N° 20112201) and are based on the outputs of several key publications and multi-stakeholder consultations, including:

Policy background

- United Nations Convention on the Rights of Persons with Disabilities (2006);
- Communication from the Commission on Rare Diseases: Europe's Challenges (2008)[17];
- Council Recommendation on an Action in the Field of Rare Diseases (2009)[18];

Previous positions of the EUCERD/Commission Expert Group on Rare Diseases

- EUCERD recommendations on Quality Criteria for Centres of Expertise on Rare Diseases (2011);
- EUCERD recommendations on European Reference Networks for Rare Diseases (2013);
- Rare Disease European Reference Networks: Addendum to EUCERD Recommendations of January 2013 (2015);

[12] Preamble to the Constitution of the World Health Organization,1946.
[13] Huber M. et al., 2006.
[14] *ibid.*
[15] National Institute for Social Work, United Kingdom.
[16] *ibid.*
[17] 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, 11 November 2008, COM(2008) 679.
[18] Council Recommendation of 8 June 2009 on an action in the field of Rare Diseases (2009/C 151/02).

EUCERD Joint Action Documents

- EUROPLAN National Conferences reports (2012-2015) and National Plans/Strategies for RD[14];
- 'Rare Diseases: Addressing the Need for Specialised social services and Social Policies' (2012);
- 'Guiding Principles for Specialised Social Services' (2013);
- 'Guiding Principles on Training for Social Services Providers' (2014)[19];
- EUCERD Joint Action workshop on 'Guiding Principles for Social Care in Rare Diseases' (2014);

European studies on the needs of people living with a RD and their families

- EURORDIS Care Survey – The Voice of 12000 patients[20] (2009);

Other EU funded projects

- EUROPLAN Report on the 15 National Conferences (2010-2011)[21].

The draft recommendations underwent a consultation process involving members of the Commission Expert Group on Rare Diseases, partners of the EUCERD Joint Action, members of the EURORDIS Social Policy Advisory Group and other relevant stakeholders in the field of health and social care.

5. TARGET GROUPS FOR THESE RECOMMENDATIONS

These recommendations aim at advising Member States and the European Commission on issues that should be considered when organising the holistic care of RD patients within national health and social care systems.

EU funding programmes should be mobilised to support the implementation of these recommendations.

The recommendations mainly focus on rare and very complex conditions which, as explained in the background, have specific needs.

[19] Documents may be downloaded from the EUCERD website (http://www.eucerd.eu/).
[20] EURORDIS, 2009.
[21] Documents may be downloaded from the EUROPLAN website (http://www.europlanproject.eu/).

RECOMMENDATIONS TO THE EUROPEAN COMMISSION AND MEMBER STATES

1. **The incorporation of RD specificities into mainstream social services and policies is a necessary element to be considered in future National Plans and Strategies (NP/NS) for RD and should be incorporated when existing NP/NS are evaluated and revised. In particular:**
    - Training of professionals should be promoted;
    - High quality information should be made available.

2. **Centres of Expertise have a key role in facilitating integrated care provision in line with the EUCERD recommendations on Quality Criteria for Centres of Expertise on Rare Diseases22 (4, 9, 10):**
    - Centres of Expertise (CEs) bring together, or coordinate, within the specialised healthcare sector multidisciplinary competences/skills, including paramedical skills and social services;
    - CEs provide education and training to (…) non-healthcare professionals (such as school teachers, personal/homecare facilitators);
    - CEs contribute to and provide accessible information adapted to the specific needs of patients and their families, of health and social professionals.

3. **European Reference Networks for RD have a key role in facilitating integrated care provision in line with the EUCERD recommendations on European Reference Networks for Rare Diseases (10)**23 **and the Directive on patients' rights in cross-border healthcare (Article 12, 4-ii)**24**:**
    - Rare Disease European Reference Networks (RD ERNs) need to collaborate with each other, as well as with patient groups, health and social care providers;
    - RD ERNs follow a multi-disciplinary approach;
    - RD ERNs could function as a platform to share experiences and promote cooperation between MS, to develop precise descriptions of the services required and elaborate common guidelines.

4. MS should promote measures that facilitate multidisciplinary, holistic, continuous, person-centred and participative care provision to people living with rare diseases, supporting them in the full realisation of their fundamental human rights. In particular:
    - MS should ensure that people living with a RD are afforded the same standards of care and support as the ones available to other citizens with similar requirements;
    - MS should recognise the particular challenges posed by rare and complex conditions.

5. MS should promote measures that support patients/families affected by RD to participate in decisions regarding their care plan and their life project:
    - MS should develop information and training tools for patients and families affected by a RD which empower them and increase their capacity to undertake a participative role in care provision;
    - Care providers should be prepared to give non-directive assistance and support patients and families to express their wishes, set priorities, take decisions and direct their own services if they wish to do so.

[22] EUCERD recommendations on Quality Criteria for Centres of Expertise on Rare Diseases,2011.
[23] EUCERD recommendations on European Reference Networks for Rare Diseases,2013.
[24] Directive on patients' rights in cross-border healthcare, 9 March 2011(2011/24/EU).

6. Transfer of information between care providers, within the limits of data protection legal frameworks, should be promoted to support holistic care provision.

7. MS should promote coordination and networking between all parties involved in the care provision of persons affected by RD, including public, private and civil society organisations as well as between providers and patient/disability organisations.

8. RD specificities should be integrated into national systems assessing a person's level of functioning, in line with the United Nations Convention on the Rights of Persons with Disabilities.

9. The elaboration and dissemination of good practices for social care in RD should be encouraged.

10. Socio-economic research in the field of RD care provision/organisation should be supported both at MS level and at European Union level. Support should be provided for research on the following topics:

- Socio-economic burden of RD;
- Accessibility and appropriateness of healthcare services, including social services, for people living with a RD and their families;
- Effectiveness and cost-effectiveness of social services and support, as well as rehabilitation and assistive technologies for people with a RD;
- Innovative care practices in health and social services and their impact on the quality of life of people living with RD.

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Communication from the Commission on European Disability Strategy 2010-2020: A Renewed Commitment to a Barrier-Free Europe: http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=COM:2010:0636:FIN:EN:PDF

EUCERD/Commission Expert Group on Rare Diseases Documents

EUCERD recommendations on Quality Criteria for Centres of Expertise on Rare Diseases (2011)

EUCERD recommendations on European Reference Networks for Rare Diseases (2013)

Rare Disease European Reference Networks: Addendum to EUCERD Recommendations of January 2013 (2015)

EUCERD Joint Action Documents

Paper "Rare Diseases: Addressing the Need for Specialised social services and Integration into Social Policies", written in the context of EUCERD Joint Action, Work Package 6, November 2012: http://www.eurordis.org/sites/default/files/paper-social-policies-services-eja-wp6.pdf

Workshop Report: 'Guiding Principles for Social Care in Rare Diseases' (Norway, 2014): http://www.eurordis.org/sites/default/files/eja-wp6-workshop-report-guiding-principles-social-care.pdf

Workshop Report: 'Guiding Principles for Specialised social services' (Denmark, 2013): http://www.eurordis.org/sites/default/files/EJA-WP6-Workshop-Report-Guiding-Principles.pdf

Workshop Report: 'Training for Social Services Providers' (Romania, 2012): http://www.eurordis.org/sites/default/files/eja-wp6-workshop-report-training-social-services-providers.pdf

Document 'Guiding Principles for Specialised social services': http://www.eurordis.org/sites/default/files/EJA-WP6-Guiding-Principles-Specialised-Social-Services.pdf

Document 'Guiding Principles on Training for Social Services Providers': http://www.eurordis.org/sites/default/files/training-for-social-services-providers.pdf

Map and information on Specialised social services: http://www.eurordis.org/specialised-social-services

Case Studies and Fact sheets on Social Services

Therapeutic Recreation Programmes: http://www.eurordis.org/content/therapeutic-recreation-fact-sheet-case-studies

Respite Care Services: http://www.eurordis.org/content/respite-care-fact-sheet-case-studies

Adapted Housing Services: http://www.eurordis.org/content/adapted-housing-fact-sheet-case-studies

Resource Centres for RD: http://www.eurordis.org/content/resource-centres-fact-sheet-case-studies

National Plans and National Conferences

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