Rare2030

Foresight in Rare Disease Policy

A knowledge-base summary:

INTEGRATED, SOCIAL AND HOLISTIC CARE FOR RARE DISEASES

https://www.rare2030.eu/our-work

TABLE OF CONTENTS

1. INTRODUCTION TO THE TOPIC

'Holistic care' covers the 360° spectrum of the health, social and everyday needs of people living with a rare disease and their families. Holistic care involves, for example:

- The provision of timely, high-quality, integrated care according to the unmet needs;
- Breaking down barriers in access to care, treatment, education, employment, leisure, psychological support and all aspects of social inclusion;
- Enabling people to fully enjoy their fundamental human rights, on equal footing with other citizens.

This topic is thus very broad: it encompasses everything from ensuring the coordination between health and social care, to paramedical support (such as dietary, psychological support etc.), to the social care sphere (e.g. adapted habitation, respite care, resource centres), to a person's inclusion in broader societal life (e.g. education, employment, relationships, etc.). Please note that 'Integrated Care' as a concept is also further explored in the Knowledge Base Summary for Sub-Group 8, 'Accessing Healthcare' (see below, p 10).

A significant challenge for patients, professionals, and health and social systems in Europe is the absence of streamlined, integrated pathways to allow people living with RD to navigate health and social care systems. This is particularly problematic in view of the **complexity** of many of these 8000 rare conditions, and the **lack of awareness and understanding in all sectors of society regarding the full impact of the conditions** (e.g. how they manifest in patients and the myriad ways in which different aspects of daily life can be adversely affected). Very often there is poor communication and collaboration between even the different 'medical' actors delivering specialised care: the disjoint becomes yet more notable when attempting to integrate paramedical and social care professionals.

The need for a holistic approach to person-centred care is particularly important in the rare disease field, where only ca. 5% of conditions have a dedicated therapy of any kind; in such cases, the integration of paramedical and social disciplines alongside the classical 'medical' approach to treatment and management is hugely valued *and* valuable. Evidence from the first European survey on the everyday impact of rare diseases - 'Juggling care and daily life: The balancing act of the rare disease community'' - confirms that the consequences of living with a rare disease are far-reaching, beyond the health niche. 85% of the respondents declared that the rare disease impacts upon several aspects of their health and everyday life. 7 in 10 people living with a rare disease or caring for an affected relative have to reduce or stop their professional activity and 69% also face an income decrease.

THE POLICY PERSPECTIVE

The importance of the subject for rare and specialised conditions has long been acknowledged in European Policy and so-called 'soft law' documents. The Commission Communication of 2008, entitled *Rare Diseases: Europe's Challenges* dedicates a Section (5.2) to these issues:

"Access to specialised social services Centres of expertise may also have an essential role in developing or facilitating specialised social services which will improve the quality of life of people living with a rare disease. Help Lines, Respite care services and Therapeutic Recreation Programmes, have been supported and need to be sustainable to pursue their goals: awareness-raising, exchange of best practices and standards, pooling resources using Health Programme and the Disability Action Plans."

The **Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02** addresses this topic in several ways (the **emphasis** is our own, added here for clarity):

- Member States (MS) were asked to elaborate and adopt NP/NS to guide and structure "relevant actions in the field of rare diseases within the framework of their **health and social** systems"
- MS were asked to "Identify needs and priorities for basic, clinical, translational **and social research** in the field of rare diseases and modes of fostering them, and promote interdisciplinary cooperative approaches to be complementarily addressed through national and Community programmes
- MS were asked to **gather** national expertise on rare diseases and **support the pooling** of that expertise with European counterparts in order to support [amongst other things] the sharing of best practices on diagnostic tools and medical care **as well as education and social care in the field of rare diseases**

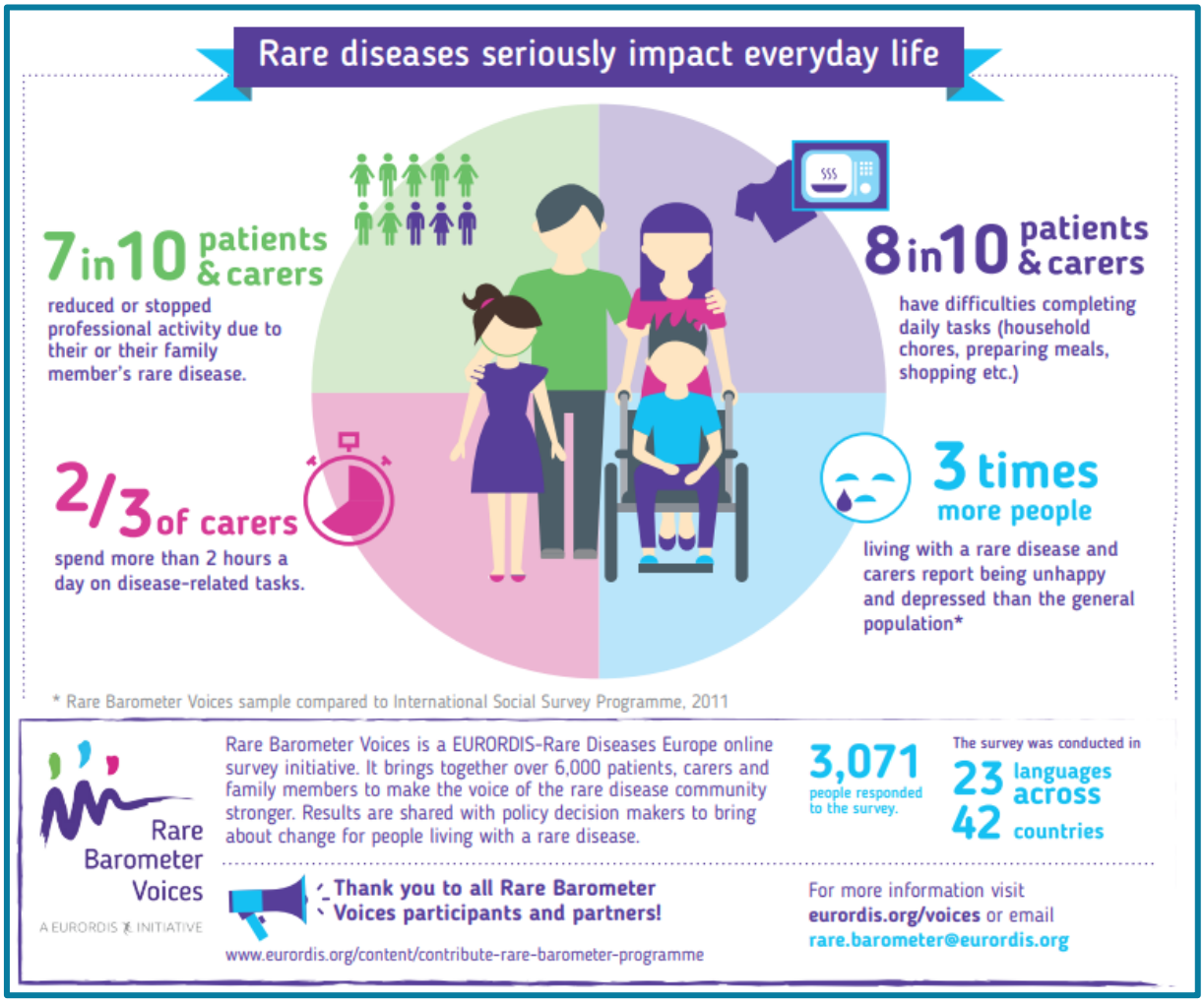
An important policy document, specific to rare diseases, is the set of *Recommendations to support the incorporation of rare diseases to social policies and services* drafted under the rare disease Joint Actions and adopted by the Commission Expert Group on Rare Diseases in 2016 (see below for more details)

2. THE IMPACT OF RARE DISEASES ON EVERYDAY LIFE: RARE BAROMETER VOICES SURVEY

In 2017, a survey entitled 'Juggling care and daily life: The balancing act of the rare disease community' was conducted by EURORDIS-Rare Disease Europe via Rare Barometer Voices, in the scope of the EU-funded project INNOVCare. The purpose of the survey was to assess the impact of rare diseases on oft-overlooked areas of life, including mental, social and physical functions, household budget, employment and job careers, family life and well-being. 3071 people responded to this survey. The full report can be accessed here .

The main findings included the following:

- Rare diseases have a serious impact on Activities of Daily Living
- People living with a rare disease and their carers spend significant time managing the disease and the care pathway
- The disease generates a strong impact on employment and work-life balance as well as important economic burden
- Care pathways are complex and hard to manage
- People living with a rare disease and their carers lack information and feel that social services are badly prepared to support them
- There is a serious impact on the mental health of people living with a rare disease and their carers



3. RELEVANT INITIATIVES, RESOURCES, AND RECOMMENDATIONS

3.1. Table of Initiatives

Several initiatives have answered the call of the Commission Communication, the Council Recommendation, and other major policy and legislative documents, and have attempted to better understand the realities and needs of patients and create resources to address these. The following table showcases a number of past and ongoing initiatives and organisations.

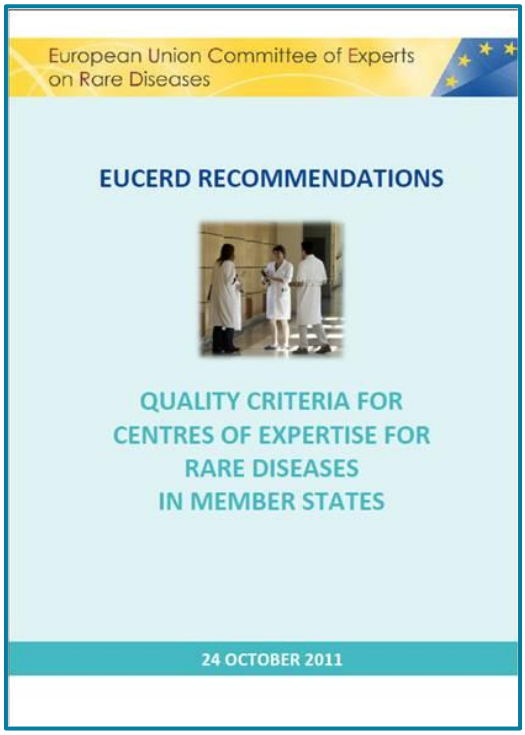
Initiative/Body	Achievement and Outputs to advance this cause
Commission Expert Group on RD (mandate expired)	2016 Recommendations aimed towards the Member States and the European Commission, focusing on "empowering health services' attempts to facilitate integrated care provision to enable them to play 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." **Recommendations to support the incorporation of rare diseases to social policies and services**
INNOVCare project (no longer funded)	✓ *INNOVCare* published a set of Recommendations in 2018 to support the implementation of integrated care and integrated service delivery, coordinated between health, social and community services: **INNOVCare Recommendations** ✓ A pilot of case management for RD was implemented and evaluated within INNOVCare, with various positive outcomes for people living with a RD: increase in the level of information about their disease, their rights and available services as well as in their capacity to manage their own care; reduction in burden faced by caregivers; improvement in coordination between care providers: ✓ The project also produced sample training curricula for case managers for RD: **Training Curricula for Case Managers for RD** A full list of resources can be found here: **https://innovcare.eu/resources/**

EURORDIS	EURORDIS-Rare Diseases Europe and its over 800 member organisations launched a position paper calling for the provision of holistic care for the 30 million Europeans living with a rare disease and their families, by 2030. EURORDIS proposed strategy to achieve holistic care by 2030 is based on 3 pillars: ✓ Pillar 1: Quality and adequate social services and policies; ✓ Pillar 2: Integrated care: bridging health and social care; ✓ Pillar 3: Equity of rights and opportunities. EURORDIS and its members call upon the EU, all European countries and all stakeholders within the health and social sector, to take action based on its ten recommendations (listed on page 9). **Position Paper 'Achieving holistic, person-centred care to leave no-one behind' (2019)**
RD-ACTION (no longer funded)	✓ **Breakout session summary on Creating a Sustainable Environment for Holistic & Innovative Care for RD & Complex Conditions** (including specific opportunities for ERNs and their constituent HCPs to add value in this topic. This was the result of a joint workshop organised by RD-ACTION and INNOVCare in April 2018) ✓ **RD-ACTION Policy Brief 'Integrated Care'**
RareResourceNet	RareResourceNet – the European Network of Resource Centres for Rare Diseases – aims to accelerate the development and the implementation of holistic high quality care pathways for people living with a rare disease across Europe, to contribute to raise standards of care and support.
EUCERD Joint Action (no longer funded)	**Papers and factsheets on the types and functioning of specialised social services for rare diseases:** ✓ **Guiding Principles for Specialised Social Services** ✓ **Guiding Principles on Training for Social Services Providers**

3.2. European Recommendations

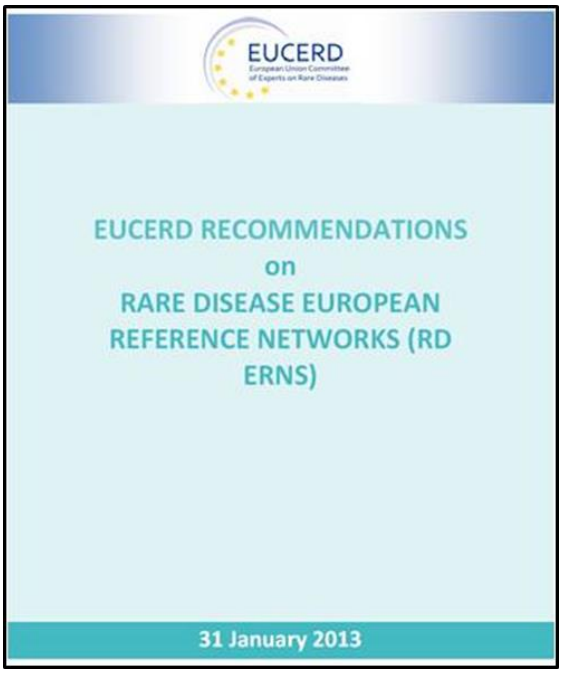
At EU level, several sets of the Recommendations adopted by the EUCERD (European Union Committee of Experts on Rare Diseases) and the Commission Expert Group on Rare Diseases (CEGRD) have a bearing on this topic. **Firstly**, the ***EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases***. Adopted in 2011, this document includes consensual criteria for designating Centres of Expertise (CEs) in European countries (some baseline parity in terms of national perceptions of a CE was essential,

particularly as CEs were envisaged to form the 'core' of -then future- ERNs, although ultimately the term HCP (Health Care Provider) was used in the ERN-related legislation.) The most pertinent criteria for *this* topic are as follows:



- ✓ (4) CEs bring together, or coordinate, within the specialised healthcare sector multidisciplinary competences/skills, including paramedical skills and social services, in order to serve the specific medical, rehabilitation and palliative needs of rare diseases patients.
- ✓ (9) CEs provide education and training to healthcare professionals from all disciplines, including paramedical specialists and non-healthcare professionals (such as school teachers, personal/homecare facilitators) whenever possible.
- ✓ (10) CEs contribute to and provide accessible information adapted to the specific needs of patients and their families, of health and social professionals, in collaboration with patient organisations and with Orphanet.
- ✓ (25) Demonstration of a multi-disciplinary approach, when appropriate, integrating medical, paramedical, psychological and social needs (e.g. RD board).
- ✓ (26) Organisation of collaborations to assure the Continuity of care between childhood, adolescence and adulthood, if relevant.
- ✓ (27) Organisation of collaborations to assure the continuity of care between all stages of the disease.

There is no official data to illustrate how many CEs -or indeed ERN HCPs- in fact comply with the criteria above, and thus with the European vision of a *true* centre of expertise for rare diseases.



Secondly, the ***EUCERD Recommendations on Rare Disease ERNs*** reinforce the importance of CEs for rare diseases -and by extension, ERN HCPs- functioning in a truly multidisciplinary manner, complying with the aforementioned 2011 Recommendations.

Thirdly, of course, the policy resource most explicitly and powerfully linked to rare diseases is the set of ***Recommendations to support the incorporation of rare diseases to social policies and services*** adopted unanimously by the Commission Expert Group on Rare Diseases in 2016. These **ten recommendations** mainly focus on empowering health services' attempts to facilitate integrated care provision, to enable them to play the role they need to play in supporting the incorporation of rare diseases

specificities into mainstream social and support services, within a holistic and person-centred approach and a human rights perspective:

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.

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.

3.3. EURORDIS Position Paper Of 2019

In May of 2019, EURORDIS issued a Position Paper to raise awareness of the needs of people living with a rare disease and their families in this broad area.

The document highlights complementary policy-based approaches to this topic, incorporating the **UN Convention on the Rights of Persons with Disabilities** and the **UN Sustainable Development Goals** relating to health, non-discrimination, and inclusivity.

(Image from https://sustainabledevelopment.un.org/sdgs)

10 high-level Recommendations were agreed in the EURORDIS position paper 'Achieving Holistic Person-Centred Care to Leave No One Behind', each accompanied by more specific points and advocacy approaches.

1. **Making full use of EU instruments and European networks to implement holistic care for rare diseases**
2. **Creating a supportive political environment at national level for holistic care for rare diseases**
3. **Gathering and disseminating knowledge and good practices, to ensure that the needs of people living with a rare disease and their carers are adequately addressed by specialised and mainstream services**
4. **Implementing specific mechanisms that ensure integrated care provision to rare diseases**
5. **Guaranteeing meaningful engagement of organisations and representatives of people living with a rare disease in the design and implementation of policies and services**
6. **Implementing specific measures that ensure access of people living with a rare disease and their carers to adequate social services and social protection**
7. **Ensuring the recognition and adequate compensation of the disabilities experienced by people living with a rare disease**
8. **Creating the conditions for people living with a rare disease and their carers to access adapted and sustainable employment**
9. **Implementing specific mechanisms that empower people living with a rare disease and their carers, in co-creation and co-delivery with organisations representing people living with a rare disease**
10. **Eliminating all types of discrimination, ensuring that people living with a rare disease have access to social, labour, educational, leisure inclusion on equal footing with other citizens**

4. HOW MIGHT ERNS SUPPORT THE PROVISION OF MORE INTEGRATED AND HOLISTIC CARE FOR PEOPLE WITH RARE DISEASES?

ERNs, at first glance, have a less clear responsibility to act in the integrated & holistic care sphere (compared to, for instance, the duty to support cross-border virtual consultations for complex cases) – at least when 'integrated' is defined as connecting medical, paramedical and social actors. Nonetheless, the *EUCERD Recommendations on Rare Disease European Reference Networks* defined several responsibilities in this area, which were reinforced in the *Recommendations to support the incorporation of rare diseases to social policies and services*, as can be seen above, page 9. The first large-scale workshop on this topic, organised by RD-ACTION and INNOVCare initiatives in 2018, demonstrated that there are in fact several areas in which ERNs -or at least, the HCPs of which they are composed- can make a significant difference to patients living with a rare disease.

- ✓ Spread understanding of the benefits of joined-up, holistic care pathways for patients (encompassing less strictly medical professionals, such as physiotherapists, psychological therapists, and social support appropriate to the specific needs of people with rare diseases and their families)
- ✓ Support and propel the drive to identify how best to provide care for patients with rare and complex conditions and define patient pathways (e.g. ERNs may help to define best practices and support their inclusion to comprehensive clinical practice guidelines or care guidelines)
- ✓ Create personalised health and social care plans for people with rare diseases, possibly both those receiving virtual referrals and the patients visiting constituent HCPs
- ✓ Engage in tertiary prevention activities, including the creation of dedicated guidance from the ERN for patients and families and for local health and social actors (some activities may of course sit more logically with the actual Centres of Expertise i.e. the HCPs here)
- ✓ Embed good practices to support integrated care for patients in their constituent HCPs (and eventually 'affiliated' partners), and in time help to diffuse good practices to broader heath systems
- ✓ Contribute to the collection and integration of data, to improve knowledge and understanding of rare diseases and the impact of patients and wider society

NB: the role of ERNs in supporting the delivery of integrated care at a national level will be addressed in the Knowledge Base Summary for the topic 'Accessing Healthcare'. In June 2019, the ERN Board of Member States adopted a statement on ***Integration of the ERNs to the healthcare systems of Member States***. This important document provides guidance around 5 topics: national rare disease plans/strategies and legal framework for ERN integration; patient care pathways; referral systems to the ERNs; support by Member States to ERN Coordinators, full members and affiliated partners; and information on ERNs provided at Member States level. Although several of these titles have a definite relevance for '*holistic* care', and for the issues explored in this Summary document, the focus of the BoMS Statement is primarily on the healthcare domain.

5. NEGLECTED AREAS OF STUDY

5.1. Research on The Socio-Economic Burden Posed by Rare Diseases

Few projects to-date have sought to estimate the full socio-economic burden of rare diseases. Individual disease communities may have conducted research in this area: some seeking to demonstrate the benefits of truly multidisciplinary care approaches, as delivered by genuine expert centres able to unite all necessary specialists across not only medical but also psychological, social, and educational actors. However, research on the full impact of rare diseases to society at large seems scarce and fragmented: the field is missing broad studies assessing, for instance, the costs of disjointed medical and social care for patients and health systems, and the economic impact (to patients and families and to society at large) of patients/family members being forced to abandon or reduce employment due to affliction with the disease or the need to act as -potentially unpaid- carers.

A 2010-2013 project, BURQOL-RD, was funded by the 2nd Public Health Programme. The project set out to conduct the first comprehensive analysis on this scale in the rare disease field, by employing a single methodology to assess both direct costs and indirect costs of rare diseases across numerous health systems. The team assessed the socio-economic burden for 10 different rare diseases, using what they termed the *BURQOL-Metre*, and also proposed a methodological framework to measure the health-related quality of life (HRQOL) of patients and their caregivers (see http://burqol-rd.eu/pag/publications.html for publications).

However, there has been limited activity in this sphere since the end of this project, despite the fact that the CEGRD Recommendations explicitly call for a renewed focus:

"Recommendation 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".*

5.2. Quality of Life Data

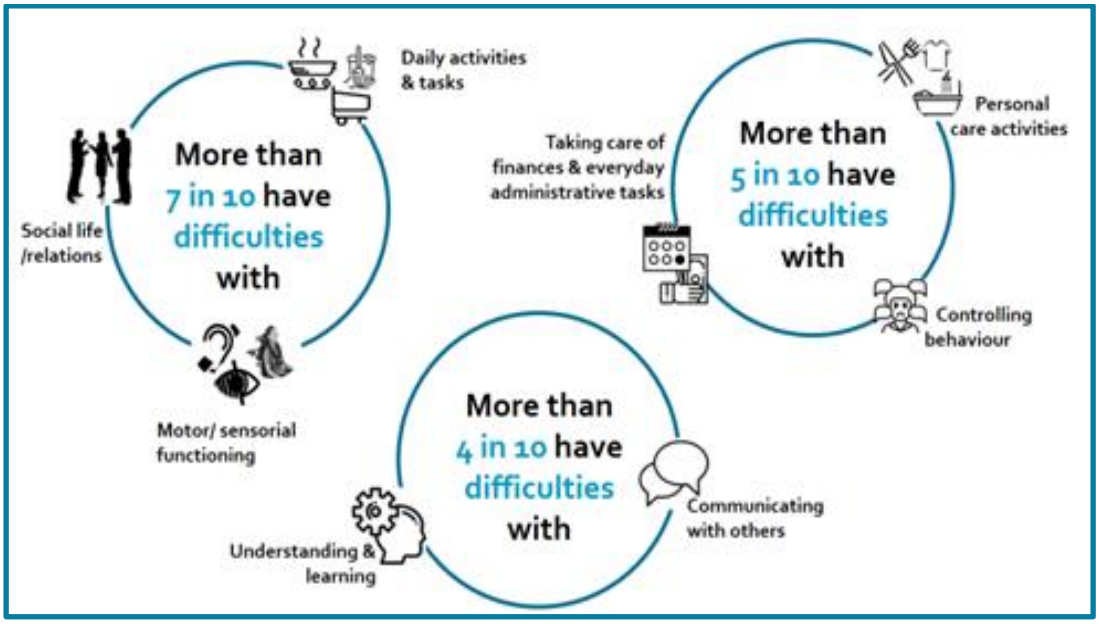
One solution to better understand how complex and often multisystemic conditions (whether rare diseases or otherwise) affect patients is to explore quality of life using approved tools and scales. The issue of how best to capture Health related quality of life (HRQoL) for those living with a rare disease or requiring highly specialised procedures and interventions, is challenging. On the one hand, particularly when assessing health technology (for instance via clinical trials), decision-makers seek comparable data to determine the relative effectiveness of medicinal products/aids/devices etc. At the same time, however, generic HRQoL measures such as EQ-5D often omit the sorts of specificities and dimensions which really matter to patients.

More appropriate, relevant, and *standardised* QoL measures would provide a broader base for the selection and measurement of Patient-Centred Outcomes.

5.3. Rare Diseases and Disability

A high percentage of people with a rare disease are affected by motor, sensorineural or intellectual impairments, which can occur simultaneously. 72% of people living with a rare disease involved in EURORDIS' recent European survey on the impact of rare diseases on daily life, reported having difficulties with motor or sensorial functioning.

According to the same survey, people living with a rare disease face serious limitations in their Activities of Daily Living (ADLs):



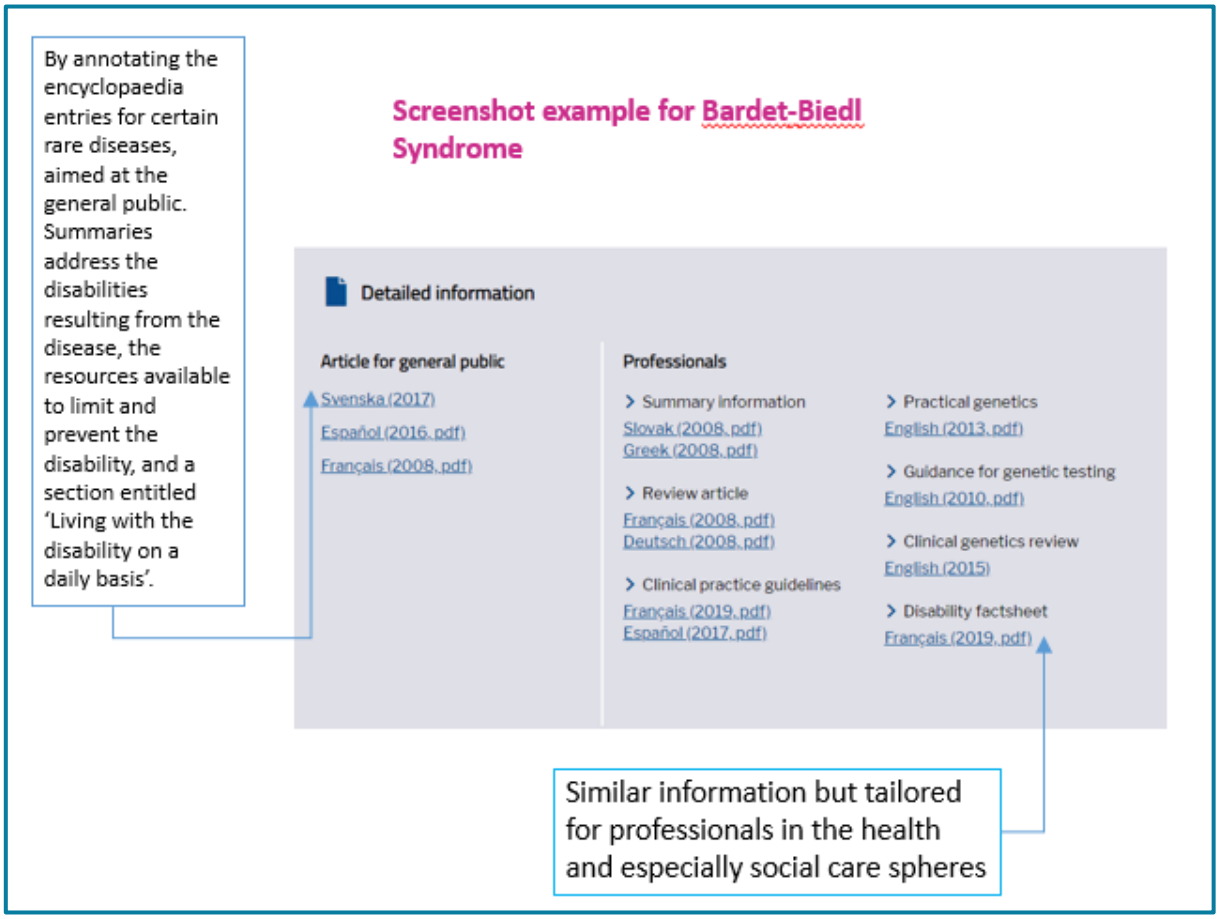
A major challenge for many rare conditions is the complexity of the phenotype and the lack of awareness by social (and often more generalist medical) professionals of the diverse and often hidden ways in which a condition can impact on a patient's life. Disabilities due to rare diseases are typically poorly understood by all but the most specialised professionals, which makes it difficult to find up-to-date reliable information on the manifestation of a disease, not purely in terms of *medical* problems but also considering how the condition could affect eating, sleeping, working, studying, behaviour, etc.

Indeed, the recognition of their disability is the main challenge for people living with a rare disease:

- 34% of the EURORDIS survey respondents who had undergone a disability assessment felt that the percentage of disability assigned to them was too low;
- 19% of the survey respondents had not been enrolled to any sort of disability assessment, despite feeling this would be warranted.

The Commission Expert Group on Rare Diseases recommends to Member States that rare diseases specificities should be integrated into national systems when assessing a person's level of functioning, in line with the United Nations Convention on the Rights of Persons with Disabilities.

An important avenue to address some of these issues is the creation of robust and RD-sensitive systems to categorise disability (and also ability, respecting what patients are able to do and the areas in which they can engage in ordinary societal activities, perhaps with a little additional support). The ICF is a good baseline in the sense it is an international framework that allows for exchange between MS but also between different actors such doctors, social workers etc. in a single country. AS it stands, however, ICF is often inadequate for rare diseases and requires adaptations. **Orphanet** is attempting to address this situation, in several ways, through the information resources available when searching for given conditions in the encyclopaedia (see image below).



The Orphanet Disability Project, involving experts and patients from across Europe and beyond, is developing RD disability core data sets **derived from and compatible with** the ICF-CY (the International Classification of Functioning, Disability and Health-Children & Youth version). The goal is to map activity limitation/restrictions by disease, using the Orphanet Functioning Thesaurus. The information is gathered via a questionnaire sent to medical experts, disability specialists and patient organisations.

Users can find information relating to activity limitations and participation restrictions; frequency in the patient population (i.e. what proportion of the patient community will be affected by each of these limitations or restrictions – are they common to all patients or only a small subset?); whether the disabilities are permanent or transient; the severity of the limitations and restrictions; whether they relate to delay in the development of abilities or to *loss* of abilities, etc. The data is analysed and standardised to constitute the Orphanet Functioning Database.

Over 1 000 RDs have been assessed so far, with the support of the French *Caisse Nationale de Solidarité pour l'Autonomie*. However, there are many thousands of rare diseases, and often the resources generated are only available in certain languages. An example of the data set provided for annotated diseases is shown below.

ORPHA:110 Bardet-Biedl syndrome

:: Activity limitation/participation restriction is described according to the Orphanet Functioning Thesaurus, derived and adapted from the International Classification of Functioning, Disability and Health – Children and Youth (ICF-CY, WHO 2007). The provided information is assessed from the whole patients' population affected by the disease, receiving standard care and management (specific and/or symptomatic management, prevention and prophylaxis, devices and aids, care and support). Functional consequences are organized by their frequency in the patients' population. This general information may not apply to specific cases. Some difficulties reported here can occur with a different temporality or severity degree, and others that are not listed can nevertheless arise.

✓ Loss of an ability

Very frequent

		Temporality	Severity
Seeing/watching	✓	Permanent limitation	Severe
Driving	✓	Permanent limitation	Severe

Frequent

		Temporality	Severity
Acquiring language		Acquisition delay	Moderate
Being aware of space		Permanent limitation	Moderate
Receiving messages in sign language	✓	Permanent limitation	Severe
Receiving nonverbal messages		Permanent limitation	Severe
Speaking		Acquisition delay	Moderate
Producing messages in sign language		Permanent limitation	Severe
Walking long distances		Permanent limitation	Moderate
Performing vigorous activities (climbing, running, jumping, swimming,...)		Permanent limitation	Moderate

6. RESULTS OF THE LITERATURE REVIEW*

**The earlier sections of this document were elaborated via research, partner expertise, and data stemming from the Resource on the State of the Art of Rare Disease activities in Europe. This final section is a summary of the results of a literature review performed by INSERM Orphanet, and is designed to highlight peer-reviewed publications which may suggest trends in this broad topic.*

Rare diseases are often incapacitating and life-limiting diseases which have a tremendous impact on the patient's life, as well as the lives of caregivers and families. Whilst traditional healthcare concentrates efforts in treating the disease *per se*, patients often require supplementary support in order to improve their life experience and face the many obstacles to be surmounted in relation to their disease. Nonetheless, often this **aid and support is not provided by the healthcare system or else is not accessible** (8; 9; 12). Hence, a whole range of unmet needs are waiting to be filled in order to alleviate the difficulties and suffering faced by patients. By extension, **caregivers and families of patients also see their life very deeply affected by the condition** and express as well a need for support which is rarely filled (8; 9; 12; 13).

Consequently, the trends show that **patients and caregivers tend to feel the socioeconomic burden of the disease very heavily**. Most patients need assistance in many areas of their life such as domestic life, transport/mobility, personal mobility/posture, leisure activities, educational or professional activities and self-care (10). A majority of patients are **barred from employment prospects and many caregivers or family members are obliged to either reduce their working hours or cease their activity altogether.** This results in patients living well beneath the poverty line, with them and their caregivers having to grapple with economic difficulties (9). Indeed, many are forced to incur substantial out-of-pocket costs, which greatly affects their lifestyle.

Caregivers, patients and families also suffer from **psychosocial effects**. In addition to the psychosocial effects and mental health issues arising from the patient's condition, individuals report **exclusion, discrimination and moral suffering** (1; 6). Parents for instance report feeling **socially isolated and desperately lonely**. They often express emotions of anxiety, fear, anger, frustration and uncertainty and **share common unmet needs regardless of what disease their child has** (13). Moreover, **very little social and economic support is offered for caregivers** who are most of the time not even recognised as such (12; 14).

Furthermore, the **difficulty in accessing care and the labyrinthine structure of the healthcare system complicates the tasks of caregivers who are forced to put much of their energy into accessing the right service and care available**. The **lack of information** from which patients and caregivers alike suffer leads them to **adopt multiple roles** placing them in complex and highly demanding situations (1; 2).

Nevertheless, in spite of these trends which seem to indicate that there is still much to be done in order to offer holistic care integrating all aspects of the condition, some efforts and changes can be distinguished in the support services developed and in the healthcare frameworks. **Social inclusion, psychological and educational considerations are gradually being integrated in national health programmes and frameworks** (6) and more specialised and support services are available (8). Services thought to greatly benefit rare disease patients and their surroundings include the following: therapeutic recreation programmes (8); one-stop-shop services such as resource centres offering family programmes; respite care; summer camps; familial support (6; 8); and the intervention of case managers who ensure the coordination between team members and the user, fill information gaps, and provide expertise in navigating the healthcare system (6; 10). The **use of internet support groups** has also been evaluated as helpful for emotional support, finding medical information and psychological support (5).

The Commission Expert Group on Rare Diseases **Recommendations to Support the Incorporation of Rare Diseases into Social Policies and Services** also points towards the right direction, recommending that Member States should ensure that people living with a rare disease are afforded the same standards of care and support as anyone else, and that the specific challenges posed by rare and complex conditions need to be recognised (6; 7). This document also **promotes the development of holistic and integrated care pathways for rare diseases** and requests Member States to include special measures in their national plans and strategies.

Finally, the time-limiting nature of many rare diseases makes **palliative care** a central component of the care management of patients: emphasis is placed on the need to focus on ways to improve this critical discipline. Such attention is key for **the establishment of a complete holistic healthcare system as regards to rare diseases**. Therefore, some researchers emphasise some elements which are crucial in order to fully accompany and ease the suffering of the patients, their family and their caregivers on this challenging path. Examples of such efforts for holistic end-of-life experiences include (1): improving **education on palliative care** approaches; identifying and responding to the **unmet needs of caregivers and families** who are affected by the evolution of the disease, including lack of information, emotional distress, feelings of uncertainty; attention on **ways of communicating, and facilitating decision-making**; consideration of **ethnic and cultural differences** (4); and **integration of the transitions experienced by the patients** during the last stages of the evolution of their disease.

REFERENCES FROM THE RARE DISEASE LITERATURE REVIEW

FULL LIST OF ARTICLES / PUBLICATIONS FOUND IN THE LITERATURE REVIEW:

https://docs.google.com/spreadsheets/d/1SRXASsFiD9sdQz286SVo860XdTpGaOIncyjIhGphULI/edit#gid=364400914 =364400914

SELECTED BIBLIOGRAPHY OR KEY PUBLICATIONS

1. Adams, L.S., Miller, J.L., Grady, P.A., 2016. The Spectrum of Caregiving in Palliative Care for Serious, Advanced, Rare Diseases: Key Issues and Research Directions. Journal of Palliative Medicine 19, 698–705. https://doi.org/10.1089/jpm.2015.0464
2. Baumbusch, J., Mayer, S., Sloan-Yip, I., 2019. Alone in a Crowd? Parents of Children with Rare Diseases' Experiences of Navigating the Healthcare System. J Genet Couns 28, 80–90. https://doi.org/10.1007/s10897-018-0294-9
3. Baynam, G., Bowman, F., Lister, K., Walker, C.E., Pachter, N., Goldblatt, J., Boycott, K.M., Gahl, W.A., Kosaki, K., Adachi, T., Ishii, K., Mahede, T., McKenzie, F., Townshend, S., Slee, J., Kiraly-Borri, C., Vasudevan, A., Hawkins, A., Broley, S., Schofield, L., Verhoef, H., Groza, T., Zankl, A., Robinson, P.N., Haendel, M., Brudno, M., Mattick, J.S., Dinger, M.E., Roscioli, T., Cowley, M.J., Olry, A., Hanauer, M., Alkuraya, F.S., Taruscio, D., Posada de la Paz, M., Lochmüller, H., Bushby, K., Thompson, R., Hedley, V., Lasko, P., Mina, K., Beilby, J., Tifft, C., Davis, M., Laing, N.G., Julkowska, D., Le Cam, Y., Terry, S.F., Kaufmann, P., Eerola, I., Norstedt, I., Rath, A., Suematsu, M., Groft, S.C., Austin, C.P., Draghia-Akli, R., Weeramanthri, T.S., Molster, C., Dawkins, H.J.S., 2017a. Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework, in: Posada de la Paz, M., Taruscio, D., Groft, S.C. (Eds.), Rare Diseases Epidemiology: Update and Overview. Springer International Publishing, Cham, pp. 55–94. https://doi.org/10.1007/978-3-319-67144-4_4
4. Baynam, G., Molster, C., Bauskis, A., Kowal, E., Savarirayan, R., Kelaher, M., Easteal, S., Massey, L., Garvey, G., Goldblatt, J., Pachter, N., Weeramanthri, T.S., Dawkins, H.J.S., 2017b. Indigenous Genetics and Rare Diseases: Harmony, Diversity and Equity, in: Posada de la Paz, M., Taruscio, D., Groft, S.C. (Eds.), Rare Diseases Epidemiology: Update and Overview. Springer International Publishing, Cham, pp. 511–520. https://doi.org/10.1007/978-3-319-67144-4_27
5. Cacioppo, C.N., Conway, L.J., Mehta, D., Krantz, I.D., Noon, S.E., 2016. Attitudes about the use of internet support groups and the impact among parents of children with Cornelia de Lange syndrome. Am. J. Med. Genet. 172, 229–236. https://doi.org/10.1002/ajmg.c.31504
6. Castro, R., Senecat, J., de Chalendar, M., Vajda, I., Dan, D., Boncz, B., 2017. Bridging the Gap between Health and Social Care for Rare Diseases: Key Issues and Innovative Solutions, in: Posada de la Paz, M., Taruscio, D., Groft, S.C. (Eds.), Rare Diseases Epidemiology: Update and Overview. Springer International Publishing, Cham, pp. 605–627. https://doi.org/10.1007/978-3-319-67144-4_32
7. Commission Expert Group on Rare Diseases, 2016. Recommendations to support the incorporation of rare diseases into social services and policies.

http://ec.europa.eu/health/rare_diseases/docs/recommendations_socialservices_policies_en.pdf. Accessed 19/06/2019.

8. EUCERD, 2012. Addressing the need for specialised social services. https://www.eurordis.org/sites/default/files/paper-social-policies-services-eja-wp6.pdf Accessed 19/06/2019.
9. EURORDIS,2017. Juggling care and daily life: the balancing act of the rare disease community. Survey performed via EURORDIS survey initiative Rare Barometer Voices. http://download2.eurordis.org.s3.amazonaws.com/rbv/2017_05_09_Social%20survey%20leaflet%20final.pdf Accessed 19/06/2019.
10. FEDER, 2009. Federación Española de Enfermedades Raras. Study on the situation of social-sanitary needs of people with rare diseases in Spain–ENSERio
11. INNOVCare, Bridging the gaps between health, social and local services to improve care for people living with rare and complex conditions, https://innovcare.eu/wp-content/uploads/2018/09/INNOVCare-project-Recommendations.pdf. Accessed 19/06/2019.
12. Métral, A., 2016. ÊTRE AIDANT-E Entre solidarité choisie et précarité subie. Pour la solidarité. http://www.pourlasolidarite.eu/sites/default/files/publications/files/na-2017-aidants-proches.pdf Accessed 19/06/2019.
13. Pelentsov, L.J., Fielder, A.L., Laws, T.A., Esterman, A.J., 2016. The supportive care needs of parents with a child with a rare disease: results of an online survey. BMC Fam Pract 17, 88. https://doi.org/10.1186/s12875-016-0488-x
14. Rice, D.B., Cañedo-Ayala, M., Carboni-Jiménez, A., Carrier, M.-E., Cumin, J., Malcarne, V.L., Hagedoorn, M., Thombs, B.D., Scleroderma Caregiver Advisory Team, 2019. Challenges and support service preferences of informal caregivers of people with systemic sclerosis: a cross-sectional survey. Disability and Rehabilitation 1–7. https://doi.org/10.1080/09638288.2018.1557268

Rare2030

Foresight in Rare Disease Policy

The health of 30 million people living with a rare disease in Europe should not be left to luck or chance. The Rare 2030 foresight study prepares a better future for people living with a rare disease in Europe by gathering the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations.

Since the adoption of the Council Recommendation on European Action in the field of Rare Diseases in 2009, the European Union has fostered tremendous progress to improve the lives of people living with rare diseases. Rare2030 will guide a reflection on rare disease policy in Europe through the next ten years and beyond.

PARTNERS

EURORDIS RARE DISEASES EUROPE

orphanet Inserm

Newcastle University

FONDAZIONE Telethon

Innovation for sustainability isINNOVA

PRESIDIO OSPEDALIERO UNIVERSITARIO Santa Maria della Misericordia di Udine

European Reference Network | MetabERN European Reference Network for Hereditary Metabolic Disorders

SERVIZIO SANITARIO REGIONALE EMILIA - ROMAGNA

European Reference Network | Bone disorders (ERN BOND)

Imperial College London

The Rare2030 project is co-funded by the European Union Pilot Projects and Preparatory Actions Programme (2014-2020). This leaflet is part of the pilot project PP-1-2-2018-Rare 2030. The content represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission or any other body of the European Union.

This document was elaborated by Rare 2030 WP4, under the leadership of Newcastle University and with input from all partners. The document should be referenced as follows: Hedley, V., Castro, R., Rodwell, C., and Simon, F. (2019) ***Rare 2030 Knowledge Base Summary on Integrated, Social and Holistic Care for Rare Diseases*** **https://www.rare2030.eu/our-work/**