

EUCERD JOINT ACTION WORKSHOP

GUIDING PRINCIPLES FOR SOCIAL CARE IN RARE DISEASES



Oslo, 9-10 October 2014

INTRODUCTION

The workshop was organised in the context of the ongoing activities of the [European Union Committee of Experts on Rare Diseases \(EUCERD\) Joint Action \(N° 2011 22 01\) \(EJA\)](#), Work Package 6 (WP6): [‘Provision of Specialised social services and Integration of Rare Diseases into Social Policies and Services’](#).

The workshop was dedicated to ‘Guiding Principles for Social Care in Rare Diseases’ and was composed of several background presentations followed by group and plenary discussions ([Appendix I – Agenda](#)).

There were 20 participants, from 13 countries, including members of the [Commission Expert Group on Rare Diseases](#) (CEGRD), representatives of national health/social authorities, patient representatives, advisors of the [EUROPLAN](#) project, health care professionals, social services, social workers and social scientists ([Appendix II – List of Participants](#)).

Context: why guiding principles for social care in RDs?

- It is estimated 4.000 to 5.000 RDs have no treatment. Social services/policies are therefore instrumental to empower patients/families and to improve their well-being;
- Rarity, complexity and lack of treatment lead to a scarcity of knowledge, expertise, resources and good practices, creating huge obstacles to the provision of holistic care;
- Care pathways remain very complex in most Member States (MS) and there is a need to compile guiding principles for social care in RDs based both on patients/families’ unmet social needs as well as on the experience of MS and on National Plans/Strategies.

Main Objectives

- To present examples of provision of social care in RDs in different MS;
- To stimulate the debate on social care provision for RD patients/families;
- To build consensus on a list of priority issues to be included in the CEGRD Draft Recommendations on Social Care for Rare Diseases.

Specific Issues Addressed

- Goals of the EJA and the specific goals of EJA WP6;
- The need for guiding principles for social care in RDs;
- Unmet social needs of RD patients and their families;
- Current models of organisation of social care for RDs in MS;
- Measures extracted from adopted National Plans and from reports of the EUROPLAN National Conferences which can be useful to address patients/families’ social needs;
- The role of centres of expertise (CEs) in social care.

Background Reading Materials

Two documents were prepared and sent to the participants in advance, in order to support the preparation for the workshop and for the group discussions:

- 1) Document on RD patients/families' unmet social needs, based on the review of:
 - The [Communication from the Commission on Rare Diseases: Europe's Challenges](#) (2008);
 - The [Council Recommendation on an Action in the Field of Rare Diseases](#) (2009);
 - The [EUROPLAN Report on the 15 National Conferences \(2010-2011\)](#);
 - The policy background/literature review EJA document '[Rare Diseases: Addressing the Need for Specialised social services and Social Policies](#);
 - [EURORDISCare Survey](#), as well as other important European surveys and studies.
- 2) Document containing transcriptions referring to social measures, extracted from adopted National Plans and/or reports of EUROPLAN National Conferences:
 - National Plans included: Belgium, Bulgaria, Czech Republic, France, Germany, Hungary, Ireland, Portugal, Romania, Slovakia, Slovenia, Spain and United Kingdom;
 - Reports of EUROPLAN National Conferences included: Cyprus, France, Greece, Hungary, Lithuania, Luxembourg, Romania and Slovakia.

Format of the Workshop and Methodology of Group Discussions

The workshop started by introducing the [EJA](#) and EJA WP6: '[Provision of Specialised social services and Integration of Rare Diseases into Social Policies and Services](#)'.

This introduction was followed by a set of comprehensive presentations on social care pathways in France, Norway, Italy and Sweden.

Finally, the workshop included group and plenary discussions on main issues regarding social care provision for people living with RDs. The discussions were preceded by a presentation based on the materials sent to participants in advance, containing the list of patients/families' unmet social needs linked to the measures proposed in the adopted National Plans and in the reports of the EUROPLAN National Conferences.

The discussion groups were composed of diverse profiles and countries of origin, in order to enhance the diversity and quality of the discussions. The groups were encouraged to:

- Comment on patients/families' unmet social needs and on the measures proposed in National Plans and reports of EUROPLAN National Conferences;
- Consider which measures currently proposed in MS could be of interest to be shared amongst MS;
- Discuss the role of CEs in relation to provision of social care;
- Indicate if the issues presented are indeed important and if they should be incorporated in the CEGRD Draft Recommendations on Social Care for Rare Diseases.

The recollection of the comments from the group and plenary discussions is presented in the section [Discussions](#).

Next Steps

A draft document for the CEGRD Draft Recommendations on Social Care for Rare Diseases will be developed based on this workshop report. This draft will undergo a revision and consultation process involving the workshop group rapporteurs, members of the CEGRD, partners of the EJA, members and volunteers of EURORDIS and other relevant stakeholders.

The draft document will then be provided to the CEGRD for further consultation and revision.

PRESENTATIONS

EUCERD Joint-Action, by Victoria Hedley (VH)

VH's presentation laid the groundwork for the understanding of the context of the workshop by providing the wider context on the EJA, its goals, partners and work packages.

VH presented the general goals of the EJA as a whole – ultimately, the implementation and realisation of the [Communication from the Commission on Rare Diseases: Europe's Challenges](#) and of the [Council Recommendation on an Action in the Field of Rare Diseases](#).

Up to now, three key sets of EUCERD Recommendations have been generated by the EJA, dedicated to [European Reference Networks \(ERNs\) for RD](#), [RD Registration and Data Collection](#), and [Core Indicators for RD National Plans](#). Key tools and resources have been elaborated, including three annual editions of the ['State of the Art'](#) Report, summarising RD activity across Europe.

20 out of the 24 EUROPLAN National Conferences have been organised, following a common set of [content guidelines](#), in the format of multi-stakeholder events gathering all parties to reinforce dialogue and discuss the content of National Plans/National Strategies and its implementation. The analysis of [Conference Reports](#) already published demonstrates some recurring needs and problems across countries: not all areas are covered in the National Plans, most actions are without funding allocation, many policy measures are difficult to implement and some disease areas are left uncovered. 16 [National Plans/National Strategies](#) have been adopted so far.

Significant progress has been made in the area of coding and classification of RDs, through the harmonisation of terminologies and the integration of RDs into the forthcoming ICD 11, as well as with the drafting of the CEGRD ['Draft Recommendation on Ways to Improve Codification for Rare Diseases in Health Information Systems'](#).

Research on quality of CEs has been conducted together with 16 CEs, based on an ethnographic study, integrating dimensions of quality of care (QoC) and the EUCERD [recommendations and good practices regarding the quality of CEs](#). An inventory of good practices for improving QoC for RD patients is currently being compiled.

Significant emphasis continues to be placed on integrating and harmonising RD efforts across such key topics as European Reference Networks, registries and genetic testing, to ensure the sustainability of RD initiatives. [Download the presentation](#).

Specialised social services and Integration of RDs into Social Policies: State of the Art, by Raquel Castro (RC)

RC started by introducing the problematic of RDs and EJA WP6: ['Provision of Specialised social services and Integration of Rare Diseases into Social Policies and Services'](#).

The main activities of EJA WP6 were presented: identification and mapping of specialised social services; training of social services providers; and integration of RDs into social policies and services.

RC also shared the definitions of specialised social services i.e. Therapeutic Recreation Programmes (TRP), Respite Care Services (RCS), Adapted Housing (AH) and Resource Centres (RC).

RC then presented the progress and outcomes of EJA WP6:

- Paper '[Rare Diseases: Addressing the Need for Specialised social services and Integration into Social Policies](#)';
- [Mapping of Specialised social services](#) (84 services, from 22 countries) and publication of information on [TRP](#), [RCS](#), [AH](#) and [RC](#) on EURORDIS and [EUCERD](#) websites;
- Elaboration of factsheets on [TRP](#), [RCS](#), [AH](#), [RC](#);
- Country visits: Agrenska RC (Sweden, 2012); Frambu RC (Norway, 2012); Bátor Tábor, TRP (Hungary, 2013); Group Homes AH for Prader-Willi Syndrome (Denmark, 2013); CREER RC (Spain, 2014) and Hendaye Hospital RCS (France, 2014);
- [Workshop on 'Guiding Principles for Specialised social services'](#) (Romania, 2012);
- Document on '[Guiding Principles for Specialised social services](#)';
- [Workshop on 'Training for Social Services Providers'](#) (Denmark, 2013);
- Document on '[Guiding Principles on Training for Social Services Providers](#)';
- Workshops at [EURORDIS Membership Meeting 2013 Dubrovnik](#) ;
- Presentations and distribution of Guiding Principles documents at [ECRD 2014 Berlin](#) and poster award for '[Integration of Rare Diseases into Social Services](#)'.

The next steps were also presented, with specific emphasis given to the elaboration of the CEGRD Draft Recommendations on Social Care for Rare Diseases, to be delivered at the end of EJA (2015) and which will be based on the outcomes of this workshop as well as on future consultation with MS and other stakeholders.

Finally, RC presented the communication actions which have been carried out so far via [EURORDIS](#) and [EUCERD](#) websites, partners' newsletters, EURORDIS [eNews](#), [Facebook](#) and [Twitter](#), highlighting that these online communications have reached over 252.000 views. [Download the presentation.](#)

Introduction to the Workshop: context, theme, purpose, methodology and expected outcomes, by Dorica Dan, Project Leader, EURORDIS

DD gave an introduction to the workshop, presenting its context, objectives, methodology and expected outcomes.

DD then presented the list of unmet social needs identified through a literature review of important EU policy documents and surveys. These social unmet needs were then linked to measures extracted from adopted National Plans and/or reports of EUROPLAN National Conferences which could possibly help to address the unmet social needs.

This detailed presentation of both needs and policy measures supported the workshop group and plenary discussions. [Download the presentation.](#)

State of the Art of Social Care Pathways: France, by Christel Nourissier (CN)

CN started by giving an overview of RD patients' unmet social needs, highlighting that patients in France expect to have:

- A comprehensive evaluation of their complex needs by well-trained professionals;
- Individual and regularly updated plans according to their needs and aspirations regarding for example schooling, employment, day or full time care, etc.;
- Special attention to transition periods to prevent breakdowns in care provision;
- And simplified paperwork as well as coordinated answers to their needs.

CN presented the legal framework for social care provision in France, namely the 'law of equal rights and opportunities for people with disabilities' as well as the role of the National Solidarity Fund for Autonomy (CNSA) and of the local services for people with disabilities.

Further analysis was presented on the challenges of the current care provision model:

- Information directories on social services indicate if services are adapted to mental, sensorial or motor disabilities, but rarely mention RDs;
- People with RDs often end up at the bottom of waiting lists to access services;
- Patients do not know how the system works and they need to fill in a lot of paperwork every year, which is discouraging, particularly for the most vulnerable;
- Getting a comprehensive evaluation of incapacities is difficult and the process has long delays. Additionally, the legal evaluation guidelines exclude many limitations which can occur in rare diseases;
- The links between social services are unsatisfactory and 48% of patients are not satisfied with the coordination of social professionals;
- Respite Care Services, personal assistants and day care centres are available but social workers are not trained to address the specific needs of RDs and access to these services is difficult, due to waiting lists.

The measures on access to social policies and social services included in the French 1st and 2nd National Plans for RDs were also presented:

- Mission of CEs, including information and training of health and social professionals, training to patients/families and coordination with provision of primary care, medical and social care. CN highlighted that pilot networks in the field of social care have been launched by CEs in two French regions;
- Identification of 23 reference networks for diagnostics and care which will support the development of good practices in coordination with local disability services;
- Measures to improve the link between medical and social services: case managers (ongoing); [Orphanet Disability Project](#) (ongoing), which improves scientific knowledge on the disabling consequences of RDs and develops RD disability core sets using a tool derived from and compatible with the [International Classification of Functioning, Disability and Health \(ICF\)](#).

CN then presented the national French Strategy on rare disabilities and how it links to the 2nd National Plan for RDs as well as the most recent efforts of the National Solidarity Fund for Autonomy in coordinating and engaging all stakeholders involved in care provision, via the establishment of complex case management teams responsible for:

- Identifying resources;

- Evaluating people with rare disabilities;
- Supporting the integration and empowerment of patients and families.

This will be achieved by working both with medical care and social care providers, including local services (schools, employers, assistants at home, etc.); all this in cooperation with the regional health agencies and the local disability services. [Download the presentation.](#)

State of the Art of Social Care Pathways: Norway, by Ragnhild Øverland Arnesen (ROA)

ROA started by presenting patients' reports, namely the survey conducted by SINTEF (Norway), in 2009, entitled '[Facing Ignorance: people with rare disorders and their experiences with public health and welfare services](#)':

- Social care services are not very informed about rare diseases;
- Patients/families reported a common experience of having to fight for their rights;
- Transitions between different life periods are difficult;
- Non visible disabilities are neglected;
- Resource Centres are important to provide support which increases quality of life. These centres also help patients to feel in a stronger position when meeting with local services and provide knowledge to patients/families on their rights with an attitude of support and empathy;
- While at kindergarten there are positive experiences; but at school it is often difficult to get support adapted to individual needs;
- At work, patients can't follow their dreams but need to take what they can get;
- Independence and empowerment are important to provide patients with the opportunity to keep their self-esteem and to improve their quality of life.

ROA then presented two cases of patients with Prader-Willi Syndrome living in Norway: a 3 year old boy and a young lady, 36 years old. While access to support at different levels might be obtained, there are often differences in the kind of services that patients/families receive, even within the same diagnosis.

ROA concluded by giving an overview of the situation of people with disabilities nowadays, referring that this new generation has grown up going to normal school like other children and has expectations of a normal life, however:

- Adapted work opportunities are insufficient and employers are not willing to hire disabled people for 'normal' jobs;
- Municipal group home apartments are scarce and families need to find private solutions, with the support of the State Housing Bank;
- The municipalities, responsible for providing services, are trying to reduce costs;
- Care givers and employers lack knowledge and need to be trained.

ROA ended her presentation by acknowledging that life for patients is getting better at several levels and by launching the question of what will happen to patients if politicians reduce social care resources. [Download the presentation.](#)

State of the Art of Social Care Pathways: Norway, by Stein Are Aksnes (SAA)

SAA started by presenting the Norwegian welfare system, funded through taxes and based on universal rights, working like a security net that is meant to catch all citizens who, for different reasons, cannot provide for themselves.

Answering to the question of 'do we need special care for people with rare disorders?', SAA concluded that multidisciplinary, long term, individual and coordinated services are needed for rare diseases as for other non-rare disorders. But, for rare disorders there are particular challenges generated by the lack of knowledge and understanding, isolation and complexity of needs, which extend beyond medical needs to educational, economic, social and psychological needs.

SAA then presented the Norwegian National Advisory Unit (NKSD) for RD, composed of 9 centres that build and spread knowledge and know-how on around 350 RDs via their free-of-charge multidisciplinary services to patients and families.

These centres are not primarily responsible for diagnosis, treatment and follow up but are complementary services providing multi-professional, life-long, individually based services including referral, consultations, training, counselling, information, support to local services and guidance on coping strategies in transitional phases.

The right to an individual plan and coordinator established within the Norwegian welfare system was the next focus of SAA's presentation. In Norway, anyone who needs long-term, coordinated services is entitled to an 'individual plan', tailor made to his/her individual needs as well as the right to have one professional responsible for following up and coordinating this plan at all times.

Families are also entitled to a training allowance and refund of travel expenses in the event that they attend courses provided by the centres from the NKSD, and support may be available to cover the costs of taking part in courses abroad for diagnoses which have no courses available in Norway.

SAA finalised his presentation with some suggestions on what can be done to improve social care in RDs: continue to raise awareness for social issues; include social challenges in National Plans/Strategies; voice the needs of people with RDs in the development of legislation and regulations; continue to make expertise accessible and to spread knowledge on RDs, in close cooperation with other services. The vision of the NKSD is to make RDs more known through knowledge and collaboration. [Download the presentation.](#)

State of the Art of Social Care Pathways: Italy, by Simona Bellagambi (SB)

SB started by presenting the systems in Italy to explain why dedicated systems for RDs are usually not mentioned: people living with a RD are recognised with a specificity in the process of care within the National Health System, namely via the centres of reference and other regulations establishing the national network for RDs; the social and socio-health system addresses social issues from the care needs and people with RDs fall into this path due to their needs related to loss of autonomy, without there being defined dedicated paths.

Social-health systems differ between the 20 regions of Italy due to the national regulatory framework which gives a lot of responsibility to the regions and allows regions to define

their own rules for certain areas of the integrated system of social interventions and services. While Essential Levels of Assistance have been approved for the healthcare system, the equivalent for social services - Essential Levels of Social Benefits - has not yet been approved, and therefore there is a disparity in services provided in different regions and sometimes within the same region, in different municipalities. Consequently, there are different rules and criteria for obtaining certain benefits, different methods of planning and monitoring social services, and patients with similar conditions from different regions end up having access to different opportunities.

Social Services in Italy entail everything dealing with integration and reduction of isolation, such as school integration, job placement, leisure activities, day care, housing, home care, mobility and independent living schemes.

A study called "Diaspro Rosso" performed in Italy has demonstrated that having a person with a rare disease significantly alters the daily routine of the family, future plans and individual projects as well as social life. The study further indicated that the level in which families are affected is higher in groups of patients with little or almost no autonomy and that RD patients face difficulties regarding education and employment.

The survey also revealed that the main carer is more frequently a woman (60% of cases), usually a parent or spouse of the patient. Out of these women, only 47.4% work full time and 32,6% don't work at all.

The vision of UNIAMO, the Italian national alliance of RD patient organisations, on how to address the specific needs of RD patients is focused on the role that CEs can have in linking social and social health service providers, as established in the project on quality of CEs. Nonetheless, a recent experimental evaluation model for CEs which analysed 5 CEs for Thalassemia has shown that social and health care integration (continuity of care) is among the services less performed in these CEs.

Meanwhile, several issues remain open and to be handled regarding both general issues and social and health care service provision as well as specific issues concerning RD patients. [Download the presentation.](#)

State of the Art of Social Care Pathways: Sweden, by Maria Montefusco (MM)

MM started by presenting the fundamentals of the Swedish care model, based on the welfare system and on the [UN Convention on the Rights of Persons with Disabilities](#) and then presented more concretely the Swedish model listing all relevant legislation to address medical needs - aiming at better health - and to address social needs - aiming at independence and participation into society.

MM then explained the responsibilities of each of the administrative levels: the state-level responsible for regulations, finance, some health care and some social support; the regional-level responsible for most of the health care, including habilitation; and the municipal-level where most social services and support for people with disabilities are provided, as well as education and some health care services.

More detail was provided on the Social Service Law, which lays out a range of services and support to people with disabilities of all ages, aiming at independent living and reasonable

living conditions. The needs of the beneficiaries are assessed by social workers according to methods derived from the [International Classification of Functioning, Disability and Health \(ICF\)](#) and municipalities also have a role in providing different kinds of support to family members. The Law of Service and Support aims at ensuring good living conditions for certain groups of people with disabilities, entitling them to: counselling; personal assistance; escort services; assistance of contact person; relief services at home; short-term stays away from home; short-term supervision for school children over 12 years old; accommodation in foster or residential care activities for children and young adults who need to live outside their home; adapted housing for adults; and daily activities for people in active working age who are not employed or at school.

The education system in Sweden is shaped around children's and students' individual needs. Therefore, people with a RD might benefit from special assistance adapted to their needs aiming at supporting them to achieve their goals. As far as employment is concerned, people have the right to reasonable accommodation and assisting technology and sheltered/protected work including day time activities. The Swedish system comprises social cooperation and entrepreneurship, support to employees, support to employers and an anti-discrimination Act on reasonable accommodation at work.

One of the key issues of the Swedish model is coordination, which is achieved through: a permanent coordination contact in the health care system (coordination of actors, treatments, services on different levels); and in the case of complex needs or certain groups of people with disabilities, the establishment of individual plans (with an overview of needs and coordination of actors, treatments and services).

This system is not yet completely functional. The National Board of Health and Welfare recently acknowledged that the coordination contact is not known among health care specialists and therefore this possibility is often not presented to patients. On the other hand, social workers are not motivated to put the measure in practice. Few municipalities and counties actually have routines in place for individual planning and only 60% of them coordinate support according to the Law of Service and Support. The situation in schools and regarding employment is also not yet compliant with the legal framework.

Challenges remain on getting the systems and actors of society to work together and more efficiently to provide services that address the complex needs in a person centred perspective and on insisting in more patient/user-centred evaluation so that care provision can be steered accordingly.

The National Agency for RD recently launched an [information film](#) to raise awareness for the need for coordination. The model of cooperation coordinated by the National Agency and involving CEs, non-medical services and patient organisations opens doors to a more positive future. [Download the presentation.](#)

DISCUSSIONS

The workshop discussions were preceded by a presentation based on the materials sent to participants in advance, containing the list of patients/families' unmet social needs linked to measures proposed in adopted National Plans and in the EUROPLAN National Conferences.

The following points summarise the key issues and concerns debated during this extremely rich brainstorm and debate exercise, organised within overarching subjects.

Social Care and National Strategies and National Plans for Rare Diseases

- People living with RDs should not be submitted to lower nor higher standards of social care than other citizens;
- The RD community needs to stimulate the existing social care system to streamline resources in order to address the needs of RD patients;
- In order to promote the improvement of social care for people with rare diseases, National Plans should strive to include measures on social care in coordination with improved medical care, and should incentivise the adoption of good practices, such as the 'Guiding principles' compiled within EJA WP6;
- Monitoring the implementation of social measures might be difficult due to several challenges faced with assessing and quantifying the impact of social measures;
- Policies need to promote stability and continuity of care, regardless of changes occurring at political and governance levels;
- Many social duties are currently being partially or totally fulfilled by patient organisations, which is not ideal and non-inclusive of undiagnosed patients or patients who don't have an organisation dedicated to their disease.

Ideas from National Plans and National Conferences for RDs

Grants for research projects in Human and Social Sciences (FR)

Tenders to promote social research on rare diseases (HU)

The patient in the centre of care: care pathways & individual care plans

- Patients and families need to drive their life car. The road is irregular and the way is complex. Patients might need special tools to be able to drive as well as guidance from a navigation system, providing them with information, directions and alternative routes to support and empower them to successfully reach their destinations. In 2014, the tools are still being put in place and patients often remain dependent on the good will of people they find by the road;
- The terminology of care itself leads to the impression that patients are taken care of instead of being able to take care of themselves. There should be more investment in informing and empowering patients to have an active role in their own care;
- Patients should get all information needed according to individual needs and type of diagnosis. This information would allow them to make informed decisions and to contribute actively to the design of a personalised plan and care pathway together with the necessary/relevant care providers;

- Training is one of the most important and effective tools to empower patients. There should be systems to support patients to attend training courses and National Plans and Strategies for RDs should strive to include this issue;
- There needs to be further reflection on how to reconcile the need for personalised services and the other care system scenarios;
- Therapeutic care pathways should be one tool to level the care and the services provided (diagnostics, therapeutic, health and social care). They must be flexible to integrate into the regional or local context in which care is provided;
- The concept of individual care plan, when existing, varies from country to country. While in some countries it works as a check list where specialists can tick boxes of services that the patient will need, in other countries the plan goes as far as presenting patients' expectations and vision for their personal and professional future and is used as a tool to support patients overcoming various challenges. The training of the professionals responsible for designing the care plans also influences their quality;
- Every patient should be entitled to an individual care plan designed following an individual assessment of needs, abilities and disabilities. Care plans would then vary from one person to another – even in patients with the same condition – according to the severity of the disease and the consequent limitations in the performance of activities;
- Individual care plans should be simple and holistic. These plans have a particularly important role in ensuring smooth transition periods for patients
- The individual plan should not just include treatments and care provision but also include a vision for the patient's life, including at a professional level;
- Doctors, therapists, parents, social workers, and other care providers should get together to implement the patient-centred individual care plan at a local level;
- It is necessary to have coordinators of the individual care plans, who are responsible for establishing the needed contacts and ensuring follow up and monitoring.

Ideas from National Plans and National Conferences for RDs

Give patients information about their condition so that they can develop a personalised care path plan with their clinical and social care team (UK)

Care intervention plans (UK, RO)

Establishment of networks of health care and social assistance clustered around CoE (RO)

Complex care protocols and models that specify the care and referral processes, as well as the health services and social services (ES)

Specialist clinical centres bring together multidisciplinary teams of health and social care professionals (UK)

Specialist clinical centres must have protocols in place to share expertise with local services (UK)

Care coordinator in the CEs (BE) and case managers (FR, HU, ES)

Bodies to coordinate health and social care services (ES)

Therapeutic education for patients and families (HU)

Information campaigns (HU)

The patient in the centre of care: case management to facilitate personalised care

- Case managers are one solution to adapt the system to patients' individual needs;
- Case managers can be seen more as facilitators than as coordinators;

- The link between case managers and the other experts comes from patients/families' needs. The case manager can make patients and experts aware of available resources and care needs respectively and facilitate communication between patient and carers;
- The case manager should remain the same for as long as possible, as this will ensure stability which is particularly important in the transition periods;
- Case managers could ideally be located at a more local level (in connection to authorities responsible for the assessment of disability or to autonomy policies and support services) but they could also be less close to patients as long as the actual care is provided locally;
- The role of the case manager is not to provide medical care but to make all the services work together and to implement an individual holistic care plan;
- Case managers should create a network of needed health and social service providers;
- They need to encourage service providers to be engaged, motivated and responsible for their role within the network of services to patients/families;
- Case managers could be trained by CEs;
- Case managers could also be organised in teams, or linking teams, connected to CEs. This would facilitate coordination of health and social care according to patients' individual needs within an approach of holistic and continuous care;
- Case managers should motivate, encourage and train the other key professionals to use the coordinating services and to fulfil their part of the puzzle according to the personal needs-based network of service and support.

Ideas from National Plans and National Conferences for RDs

Care coordinator in the CEs (BE)

Bodies to coordinate health and social care services (ES)

Case managers (FR, HU, ES)

Centralisation-decentralisation of knowledge and expertise

- There is too much inconsistency in care provision between regions and countries;
- Due to the nature of RDs, there should be experts in central places at national and international level who gather and contribute to the constant evolvement of information and knowledge on RDs. It is important to centralising expertise sufficiently so that care professionals can get in touch with people who know a lot about RDs;
- Given that the provision of social services is and should be performed locally whenever possible, it is necessary that the information and competence gathered centrally are constantly accessible at local level. And local experts/carers do not need to be at the same level of knowledge as the expert in central structures.

Ideas from National Plans and National Conferences for RDs

National competence centres (NO)

National Institute for Rare Diseases (HU)

Training social service providers & training and education programmes (RO, HU)

Training programmes that enable health and social care professionals to better identify rare diseases to help deliver faster diagnosis and access to treatment (UK)

Creation of a central information portal for RD (DE)

Organising information campaigns (HU)

Disability factsheets (FR)

Creation of a central information portal for RDs (DE)

Coordination & networking

- MS need to promote better coordination. There should be a system to bring all parties together: a working group on RDs should be a requisite for each MS;
- Difficulties also arise from the fact that the databases of local service providers are generally not cross referenced with databases from CEs or from the health care system at large and therefore it is currently hard to have a comprehensive picture of all the services and care provided to patients;
- Holistic care should include medical and social care in connection with other services provided on a daily basis at local level;
- The concept of multidisciplinary teams should encompass health and social care professionals as well as patient representatives who should be included in a meaningful way, all throughout the care process;
- Professionals from different regions and services should know how to contact directly experts in other services;
- Networking has a central role in order to ensure holistic and continuous care;
- There should be funding allocated for networking activities. Local authorities and services should consider having a specific budget for networking. If networking becomes part of every service's duty, it will be included in its budget;
- CEs are funded by the healthcare system and their services are meant to include social issues as well. But in reality these two types of services are normally coordinated by different Ministries and have distinct budgets. There should be further reflection regarding the need for funding dedicated to networking activities between CEs and local care providers;
- Should there be a shared methodology for working in multidisciplinary teams?
- In countries where there are Resource Centres for RDs (i.e. Sweden, Romania, Spain, Norway, France, etc.) these can be associated to CEs as part of their network;
- ERNs can be a way to promote international networking in particular for Resource Centres for RDs which support patients beyond medical issues.

Ideas from National Plans and National Conferences for RDs

Working group on RDs (several MS)

Bodies to coordinate health and social care services (ES)

Organise all existing structures in one single network (GR)

Establishment of networks of health care and social assistance clustered around CEs (RO)

Complex care protocols and models that specify the care and referral processes, as well as the health services and social services (ES)

Specialist clinical centres must have protocols in place to share their expertise with local services (UK)

Specialist clinical centres bring together multidisciplinary teams of health and social care professionals (UK)

Other ideas

In Norway, the Ministry of Work has a certain budget to invest on the social situation of people in order to allow them to get back to a professional activity. The social security and the Ministry of work keep a centralised and universal system of welfare. The Ministry of Work funds units in all municipalities for this work. This system has a top structure and goes all the way to local communities. The system has been installed 10 years ago (www.nav.no).

Information and data sharing

- Interoperability is key to the success of holistic care provision;

- Some structures will always be more centralised (e.g. CEs, regional hospitals). To overcome the distance between these structures and local care providers there is a need to develop the systems used to share information and transfer knowledge;
- MS should strive to promote data compatibility and compatible IT systems that allow the safe sharing of relevant information between different authorities, and service providers within the terms of the data protection regulations of each MS;
- MS might want to consider developing a complete and up-to-date national RD information hub (via Orphanet for example) that contains information on the CEs, research, therapies and social services available in that particular country;
- Directories of specialised services could be helpful as a one-stop-shop for information;
- Disability fact sheets per disease can be very useful for professionals from the social sector to know more about patients' conditions;
- MS could consider translating the disability fact sheets elaborated by Orphanet into their languages;
- The existing help lines for RDs can also be of support in this transmission of knowledge and expertise as they gather a lot of information and contacts;
- New technologies such as teleconferencing and telemedicine are extremely useful to facilitate safe communication and sharing information.

Ideas from National Plans and National Conferences for RDs

Creation of a central information portal for RD (DE)

Creation of a central information portal for RDs (DE)

Data collection: the need for proof that social support works

- At the moment it is not always possible to follow a patient's care pathway, which might be divided between different systems and care providers who are not necessarily in contact with each other;
- There is a lack of studies on social and daily life needs of patients/families;
- There is an increasing demand for data that demonstrates the dimension of the need for social care in RDs and that provides evidence of the efficacy and cost-effectiveness of social support. Equally relevant is the need to obtain more data on the costs generated due to the absence of provision of adequate health and social care;
- This data is essential to support MS in making informed decisions regarding the provision of health and social care;
- Collecting data on patients' care at local social services is a big challenge and solutions need to be found in order to maximise the input of information and minimise the use of resources in this task: can RD registries include some key information on social issues?; should CEs collect this kind of information?; should care coordinators have this task?; Could patients report this information themselves?;
- Further reflection is also required, regarding what needs to be - and what can be - assessed concerning patients/families social and living conditions, on what type of data should be collected and on what type of methodology should be used in order to demonstrate clearly the actual impact of social care provision.

Ideas from National Plans and National Conferences for RDs

Grants for research projects in Human and Social Sciences (FR)

Tenders to promote social research on rare diseases (HU)

Training professionals and patients/families

- Training for professionals is key;
- The EUCERD Joint Action (WP6) compiled [Guiding Principles on Training for Social Services Providers](#) as well as [examples of training programmes](#) that can serve as inspiration for MS and training providers when implementing and designing training programmes on RDs for social care providers;
- The development of training courses should be a cooperative multidisciplinary process involving CEs, patients/families and other relevant stakeholders and professionals;
- E-learning methods will enable training programmes to be more cost-effective and to include more professionals from different geographic areas;
- Local authorities could have a specific budget for working with good practices, participating in trainings or develop their own training programmes. This would work as an incentive to improve their practices and to do the key things for RD;
- Informing and training patients are key in order to empower them and to increase their autonomy.

Ideas from National Plans and National Conferences for RDs

Training social service providers & training and education programmes (RO, HU)

Training programmes that enable health and social care professionals to better identify rare diseases to help deliver faster diagnosis and access to treatment (UK)

Educational guides for teaching staff (ES)

Therapeutic education for patients and families (HU)

Other ideas

EUCERD Joint Action document on [Guiding Principles on Training for Social Services Providers](#)

EUCERD Joint Action document on [examples of training programmes for social services providers](#)

The need to improve disability (and ability) evaluation systems

- Although it is important to evaluate the degree of disability/incapacity to sustain the process of allocation of compensation measures, it is also important to evaluate ability/capacity which enables the maximisation of patients' potential allowing them to attain a higher level of autonomy and societal participation;
- It is necessary to have a system that recognises what patients are able to do and which brings them the feeling of value, supporting them to be more active. On the other hand, there is the fear that a system focused on evaluating abilities might lead to a loss of social support, particularly in countries very affected by the economic crisis;
- Systems need to be flexible to patients' needs and be able to identify their incapacities (even the least "visible") but the system also needs to guarantee that the evaluation is fair from case to case and that the customization to patients' needs does not lead to inequalities;
- Existing evaluation systems do not take into account that the addition of several disabilities linked to various RDs has a much higher impact on a patient's life than one particular disability or the simple sum of various disabilities;
- The degree of disability/ability of people with a rare disease should be assessed within a coordinated approach;

- Reorganising evaluation systems can represent a high investment of resources and time for MS and should therefore be a carefully planned process;
- The [International Classification of Functioning, Disability and Health](#) (ICF) can serve of inspiration to re-structure the methods for evaluation of incapacity;
- 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;
- It's good to have the ICF as a global framework, but in practice it is not possible to use this theoretic classification. The World Health Organisation recommends that countries build up tools based on the ICF, which would require some adaptation to be used in practical terms and for RDs;
- ICF is often inadequate for RD and there needs to be additional work to make it more valuable for RDs. But it does not make sense that each MS does this individually, as that will lead to a loss of transferability of knowledge between experts in different countries (which is so well achieved with the ICF);
- The [Orphanet Disability Project](#), involving experts and patients from 43 countries, develops RD disability core sets derived from and compatible with the ICF and is an important tool that can support the evaluation of (in)capacity in RDs;
- In some MS the evaluation forms are filled in by physicians who know patients' clinical data but don't necessarily know all their daily life challenges;
- Patient files can be an efficient way to increase the scope and the quality of the assessment of (in)capacities: physicians can introduce the clinical and medical data whereas patients can self-report particular issues and daily life challenges;
- IT compatibility within the system of evaluation of incapacity is essential: it allows for communication between professionals/departments and supports patients' mobility avoiding the re-starting of processes whenever patients change region.

Ideas from National Plans and National Conferences for RDs

Implementing a new system of assessing disability and functioning (CY) & improvement of system of evaluation of incapacity supported on ICF and in RD core sets developed by Orphanet in consultation with various stakeholders (FR)

Professionals must be properly informed about the ICF system and its application (CZ)

Disability factsheets (FR)

Proposal to reorganise the structure and protocols followed when evaluating RD patients and their level of disability (GR)

Proposal to make it mandatory to all Medical Bodies as well as the Central Board of Health to contribute to the process of Disability Severity Assessment and Disability Certification Centres (GR)

Proposal to widen the specialisation of the personnel on the committees of the Disability Certification Centres (GR)

Good practices

- The EUCERD Joint Action (WP6) compiled [Guiding Principles for Specialised social services](#) and case study documents which contain many good practices on different Specialised Social Services that can serve of inspiration to MS and to care providers to improve quality of social care provided to RD patients/families;
- Common protocols in general are a good practice but they have limits in regards to social care provision which is much more personalised than other care areas. For this

reason, patients should be entitled to individual assessment of their specific needs in coordination between different experts and departments;

- It would be useful to have general/generic protocols at national or EU level defining basic criteria and procedures serving as broad guidelines and methodologies;
- The key point is to develop common tools and core guiding principles for caring needs, which not being too detailed can be applicable in several countries;
- Existing [Resource centres](#) for RDs can serve as a platform to collect and disseminate best practices for social care;
- [Orphanet's disability fact sheets](#) are also useful tools for social care and MS should be encouraged to translate these fact sheets.

Ideas from National Plans and National Conferences for RDs

Defining the tools and interventions necessary for the improvement of social care (HU)

Defining the development objectives of social and rehabilitation services (HU)

To propose special forms of support and the inclusion of children and young people with rare diseases in the school system (SI)

Occupational reports and guides aimed at the adaptation of work stations to disabilities (ES)

Educational guides for teaching staff (ES)

Other ideas

EUCERD Joint Action document on [Guiding Principles for Specialised Social Services](#)

Funding

- Having common budgets for all the multidisciplinary work required to follow up patients can be a burdensome process;
- But there might be the need to allocate funding to some of the important elements which support the team work across areas and departments (e.g. networking);
- Local authorities might want to consider making available specific budgets for local social services to be able to network;
- There is a need for funding dedicated to networking activities between CEs and local care providers. CEs should also consider that a part of their funding might need to be allocated to these networking activities as it is not feasible in most MS to fund these activities via the social package, which often depends on a different Ministry;
- Funding for training is essential both in CEs as well as in social services, as an incentive to continuous learning and improvement of social care provision;
- There should be financial support for patients attending training courses and funding allocated to the development of training courses for patients and families.

The role of CEs, according to the EUCERD recommendations

- The [EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States](#) are quite inclusive regarding the role of CEs in social care, namely through recommendations 3, 4, 5, 8, 9 and 10.
- Questions remain as to whether these recommendations are already being put in practice at all centres in all MS, knowing that working in these matters is essential to increase the quality of care provided by CEs;

- Example of Italy: a recent survey showed that even at the most experienced and well managed CEs the social aspects were the least performed;
- CEs should know what is available and be a first stop for patients offering them guidance and establishing the link with the various social care providers, according to patients' needs;
- Assuming that not all MS have or will have national Resource Centres for RDs, the responsibility for guiding RD patients towards adequate social care providers be the responsibility of the CEs;
- The concept of 'multidisciplinary' care/teams referred in the EUCERD recommendations needs to be comprehensive and habilitation/rehabilitation multidisciplinary teams can have a huge role in following up patients' social integration. That is the case in Norway, for example;
- CEs need to have staff members in the multidisciplinary team who are responsible for creating the link with social care provision. CEs can't rely on patient organisations to assume this role even though patient organisations and volunteers can be a valuable support in this matter;
- It could be useful to develop a list of non-medical issues that should be addressed by CEs in coordination between experts and patients, as is being done in Sweden.

EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States

3. The combined scope of all CEs within a MS covers all RD patients' needs (...)
4. CEs bring together, or coordinate, within the specialised healthcare sector multidisciplinary competences/skills, including paramedical skills and social services (...)
5. CEs contribute to building healthcare pathways from primary care.
8. CEs contribute to the elaboration of good practice guidelines and to their dissemination.
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.

Source: EUCERD Recommendations on Quality Criteria for Centres of Expertise

Other ideas

Platforms of coordination between CEs and local services providers in France:

[Vivre avec une Anomalie du Développement en Languedoc Roussillon](#)

[PRIOR Plateforme Régionale d'Information et d'Orientation des Maladies Rares](#)

APPENDIX

I) AGENDA

DAY 1: Thursday 9 October

08:45 Welcome to all participants *by Dorica Dan, EURORDIS and Kjetil Ørbeck, Frambu (15')*

09:00 EUCERD Joint Action *by Victoria Hedley, Newcastle University (20')*

09:20 Specialised social services and integration of rare diseases into social policies: state of the art *by Raquel Castro, EURORDIS (20')*

09:40 Introduction to the workshop: context, theme, purpose, methodology, and expected outcomes *by Dorica Dan, EURORDIS and Raquel Castro, EURORDIS (20')*

10:00 Coffee Break (15')

10:15 Visit to Frambu Resource Centre (30')

10:45 State of the Art of Social Care Pathways (40' presentations, 20' discussion)

France *by Christel Nourissier, Alliance Maladies Rares, EUROPLAN advisor and member of the Regional Committee for Rights and Autonomy of People Living with Disabilities*

Norway *by Ragnhild Øverland Arnesen, Prader-willi Association Norway; Stein Are Aksnes, general director of the Norwegian National Advisory Unit on Rare Disorders*

12:45 Lunch (1h)

13:45 State of the Art of Social Care Pathways (Cont.) (40' presentations, 20' discussion)

Italy *by Simona Belagambi, UNIAMO, EUROPLAN advisor*

Sweden *by Maria Montefusco, EUROPLAN advisor, project manager at the Council of Nordic Cooperation on Disability, Nordic Centre for Welfare and Social Issues*

15:45 Coffee Break (15')

16:00 Group Discussions (2h30')

19:30 Networking Dinner

DAY 2: Friday 10 October

08:45 Recap from Day 1 *by Raquel Castro, EURORDIS (15')*

09:00 Discussions in plenary *moderated by Raquel Castro and Dorica Dan, EURORDIS (1h30')*

10:30 Coffee Break (15')

10:45 Conclusions *moderated by Raquel Castro and Dorica Dan, EURORDIS (2h)*

12:45 Lunch (1h)

14:00 End of Workshop

II) LIST OF PARTICIPANTS

Country	Name	Organisation
BELGIUM	Ingrid Jageneau	RaDiOrg - Rare Diseases Organisation Belgium
DENMARK	Stephanie Jøker Nielsen	Rare Diseases Denmark
FINLAND	Päivi Vataja	Finnish Network for Rare Diseases
FRANCE /PT	Raquel Castro	EURORDIS
FRANCE	Myriam de Chalendar	Orphanet
FRANCE	Christel Nourissier	Alliance Maladies Rares
HUNGARY	Gábor Pogany	HUFERDIS - Hungarian Federation of People with Rare and Congenital Diseases
ITALY	Simona Belagambi	UNIAMO - Federazione Italiana Malattie Rare
MALTA	Francis Agius	Ministry of Health
NORWAY	Lisen Mohr	Frambu
NORWAY	Kjetil Ørbeck	Frambu
NORWAY	David Bergsaker	Frambu
NORWAY	Ragnhild Arnesen	Prader-Willi Association
NORWAY	Stein Are Aksnes	National Advisory Unit on Rare Disorders
ROMANIA	Dorica Dan	EURORDIS/NA/CEGRD
SPAIN	Marta Fonfría	National Institute Social Services
SWEDEN	Maria Montefusco	Nordic Centre for Welfare and Social Issues
SWEDEN	Anders Olauson	Ågrenska
UK	Ian Johnston	International Federation Social Workers
UK	Victoria Hedley	Newcastle University

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Other Relevant Documents

[Map and information on Specialised social services](#)

[‘Guiding Principles for Specialised social services’](#)

[‘Guiding Principles on Training for Social Services Providers’](#)

[Workshop Report: ‘Guiding Principles for Specialised social services’](#)

[Workshop Report: ‘Training for Social Services Providers’](#)

Case Studies and Fact sheets on Social Services

[Therapeutic Recreation Programmes](#)

[Respite Care Services](#)

[Adapted Housing Services](#)

[Resource Centres](#)