

**Workshop: Creating a Sustainable Environment for Holistic & Innovative Care for Rare Diseases & Complex Conditions**  
**Frambu Resource Centre for Rare Diseases, Oslo, 12-13 April 2018**  
**Breakout Sessions Recap**

**Overview of breakout sessions**

	<b>Chair &amp; Rapporteur</b>	<b>Topics Discussed</b>	<b>Photos</b>
<b>1</b>	Chair: Edmund Jessop, former member of the Commission Expert Group on Rare Diseases; INNOVCare Advisory Group  Rapporteur: Teresinha Evangelista, EURO-NMD ERN	<b>Health &amp; Social Care: structural coordination &amp; cooperation to ensure person-centred holistic care pathways</b>	
<b>2</b>	Chair: Raquel Castro, INNOVCare  Rapporteur: Rebecca Tvedt Skarberg, EURORDIS European Patient Advocacy Group (ePAG)	<b>Patient empowerment &amp; engagement: strategies to ensure person-centred care &amp; meaningful patient engagement in care design</b>	
<b>3</b>	Chair: Biruté Tumiene, Board of Member States for ERNs  Rapporteur: Victoria Hedley, RD-Action	<b>Knowledge sharing: effectively promoting the exchange of good practices/expertise/information to support holistic care</b>	
<b>4</b>	Chair: Chair: Ursula Holtgrewe, INNOVCare  Rapporteur: Ildiko Vajda, EURORDIS Social Policy Advisory Group	<b>Professionalization of the coordination of care: who coordinates care? What is the role of case managers? How to train them?</b>	
<b>5</b>	Chair: Michelle Battye, EuroGen ERN  Rapporteur: Valentina Rupel, INNOVCare	<b>Data collection: demonstrating the impact of the services on patients' QoL/public health &amp; the cost-efficiency of integrated care</b>	

*Themes based on outcomes of the INNOVCare project regarding important issues for the implementation of holistic care for RD and on inputs from the RD-Action, regarding RD policy and the implementation of Centres of Expertise and European Reference Networks.*

**Guiding questions**

- **Can you think of any good practices relating to this topic, which work well in your experience?**
- **What needs to happen within each of these topics, to improve the status quo for rare diseases?**
- **What might ERNs specifically do to make an impact?**

**Breakout sessions outline** (5 groups of 12-13 participants)

**DAY 2: Friday, 13 April**

10:00 Introduction to the exercise (10') *Raquel Castro, EURORDIS*

10:10 Discussions in parallel sessions ('120) *Themes & chairs as above; coffee in rooms*

12:10 Recap of breakout sessions (40') *Poster session*

## S1. Health & Social Care: structural coordination & cooperation to ensure person-centred holistic care pathways

Chair: Edmund Jessop, former member of the Commission Expert Group on Rare Diseases; INNOVCare Advisory Group

Rapporteur: Teresinha Evangelista, EURO-NMD

First Name	Last Name	Organisation	Profile	Country
Anders	Olauson	Ågrenska Resource Centre	Social Service	Sweden
Beatriz	Martinez-Lozano	Regional Ministry of Health-Region of Murcia	Public body/Regional	Spain
Edmund	Jessop	National commissioning Group, National Health Service	Public body/National	UK
Isabel	Fernandez	Federación Española de Enfermedades Raras (FEDER)	Patient Representative	Spain
Graham	Slater	Federation of esophageal atresia and tracheo-esophageal fistula support groups e.V.; ERNICA (inherited and congenital anomalies)	Patient Representative	Germany
Joanna	Das	University of Newcastle	Academic	UK
Mandy	Andrew	NHS Lanarkshire	Public body/Regional	UK
Mariangela	Pellegrini	AP-HP Hôpital Saint-Louis; ERN EuroBloodNet (Haematology)	European Reference Network	France
Marta	DeSantis	Istituto Superiore di Sanità	Academic	Italy
Rasmus	Bruun	National Board of Social Services	Public body/National	Denmark
Stein	Are Aksnes	Norwegian National Advisory Unit on Rare Disorders	Public body/National	Norway
Teresinha	Evangelista	University of Newcastle; ERN EURO-NMD (Neuromuscular)	European Reference Network	UK

### Challenges addressed:

- People living with a rare disease (RD) and their carers face a significant care burden, which seriously affects their daily life - [see here the results of the 1<sup>st</sup> European survey on the social impact of RD](#); e.g. 65% have to visit different health, social and local services in a short period of time; 67% find that these services communicate badly with each other;
- People with a RD often struggle to access the various teams/experts needed to provide multidisciplinary care *within* a [Centre of Expertise](#), let alone to navigate the health and social care systems at local, regional and national level. Often, hospitals and tertiary care do not liaise well with social support providers;
- Local and social support services are often closer to the patient, but typically they are the furthest from the specialised knowledge/expertise on RD;
- Structural lack of coordination/collaboration between public bodies and between care providers has been repeatedly pointed out as a key bottleneck, which limits the provision of integrated care for RD.

### Guiding questions:

- Can you think of any good practices relating to this topic, which work well in your experience?
- What needs to happen within this topic to improve the status quo for rare diseases? What needs to happen to achieve structural coordination and cooperation?
- What might ERNs do to make an impact?

### Sources provided in advance to inspired the discussion – Index, details in respective sections below

- [Recommendations from the Commission Expert Group on Rare Diseases](#)
- [Challenges and strategies emerging from INNOVCare's interviews at national level](#)
- [Outcomes of breakout session from previous INNOVCare Workshop: "Advancing Holistic and Innovative Care for Rare Diseases & Complex Conditions" \(2017\)](#)
- [Reflections on the possible role of European Reference Networks](#)

### Recommendations from the Commission Expert Group on Rare Diseases

[Commission Expert Group on Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#), adopted unanimously by all MS in 2016:

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 Diseases](#) (4, 9, 10):

- 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) and the [Directive on patients' rights in cross-border healthcare](#) (Article 12, 4-ii):

- 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 (recommendation 4).

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.

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

### Challenges and strategies emerging from INNOVCare's interviews at national level

*The interviews were conducted mostly with public bodies and healthcare providers, in a selection of EU countries. The issues listed are therefore not a reflection of the perspectives of all stakeholders/national contexts.*

#### ✓ Collaboration between public bodies

Challenges mentioned in interviews:	Strategies mentioned in interviews:
<ul style="list-style-type: none"> <li>• Federalism/regionalism hinders the exchange of knowledge and the collaboration between services;</li> <li>• RD fall into the responsibility of the social ministry, the health ministry and the ministry of education. Despite national plans and advisory boards these issues are not part of a common agenda of political bodies;</li> <li>• Legal frameworks and funding schemes hinder a stronger collaboration of the administrations of health, social and educational services. "We have different ministries and different funding and accreditation schemes" (Regional policy maker, Romania).</li> </ul>	<ul style="list-style-type: none"> <li>• Stronger involvement of local administration in the organisation of care, as they are the closest to the patients/families (regional administration, Austria);</li> <li>• Coordinated budget of the political bodies responsible for social and health care provision;</li> <li>• Budget dedicated to the coordination of different political bodies. "It has to be clearly stated in the legislation that collaboration is welcome and be provided with special funds" (regional Policy maker, Romania);</li> <li>• Building on the positive experiences of collaboration between political bodies can contribute to making collaboration across political bodies not the exception, but the rule;</li> <li>• Qualified staff responsible for communication/mediation between different institutions (regional policy maker, Romania).</li> </ul>

✓ **Collaboration between different care professionals**

<b>Challenges mentioned in interviews:</b>	<b>Strategies mentioned in interviews:</b>
<ul style="list-style-type: none"> <li>• Doctors are not used to collaborate with other care professionals: “They need to learn how to work collaboratively and to become experts of networking, involving all care professionals” (regional social policy maker, Spain);</li> <li>• Due to a lack of coordination and networking between professionals, continuity of care cannot be guaranteed for patients with complex needs; this negatively affects the quality of care patients receive (patient representative, Spain).</li> </ul>	<ul style="list-style-type: none"> <li>• Public procurement to foster collaboration between different services: “The public administration can foster cooperation by setting new standards when commissioning different services” (social policy maker, Spain);</li> <li>• Training of professionals on RD: involving patients, their relatives and different care professionals to help all parties understand the complexities of the situation of the person with a RD;</li> <li>• Protocol of treatments or intervention plans that inform on the treatments the patient has received so far;</li> <li>• Mapping of services to ensure that patients and professionals know where to find the experts;</li> <li>• Sharing competences between professions: “Different care professionals should have equivalent positions to ensure lateral transfer of expertise between professions e.g. social workers, nurses, doctors.” (health care expert, Austria)</li> <li>• “Professionals have to accept, that they cannot be ‘experts for everything’ but they need to become ‘experts in personal care’ and to learn how to share information (social care administration, Spain);</li> <li>• Case managers: “You need somebody who makes sure that everybody does what they agreed on in the intervention plan, set up after assessing the primary needs of the patients. This would be the role of the case manager.” (health policy maker, Spain).</li> </ul>

**Outcomes of breakout session from previous INNOVCare Workshop: “Advancing Holistic and Innovative Care for Rare Diseases & Complex Conditions” (2017)**

✓ **Important issues for the implementation of integrated care for rare diseases:**

- **Focus on specifics of integrated care necessary for RD;**
- **Difference between RD and other diseases:**
  - **Rarity** (difficulty to meet other with the same disease);
  - **Lack of knowledge** about the diseases (difficulty to find information);
- For these reasons, it is **necessary to use multilevel strategies** in the integrated care implementation;
- **Top-down and bottom-up** approaches:
  - **National or regional level: top down** approach is most appropriate; the rights of the patients need to be defined as well as outcomes and training programs for resource centres;
  - **Local level:** where the patients reside, a **bottom up** approach will provide innovative methods, best practices and inspiring models of care;
  - **Intermediate level: rarity and lack of knowledge require this to connect the local and national level;** offer experience, knowledge and organizational support;
- **Incentives/tools needed** to promote integrated care for rare diseases:
  - **National level: financial or non-financial**, while experience, support and training can be transferred throughout community using technology, remote care, ICT record, platforms, etc.;
  - **Intermediate level: coordination and patient training, advocacy and active hearing;**
  - **Local level: provision of services uses case management**, supported by pathways; care is provided in **multidisciplinary teams**; leadership is shared.
- Discussion on **funding:**
  - **Value based commissioning does not seem appropriate** for RD;
  - **User led personal budgets that transfer responsibility for care to patients do not seem appropriate;**
  - **Resource centres for RD need to be financed as part of the system**, publicly. Their payment shall be according to the outcomes and goal achievement.

## Reflections on the possible role of European Reference Networks

### Could ERNs perhaps...?:

- **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?
- **Create personalised health and social care plans for people with rare diseases**, possibly both those receiving virtual referrals and the patients visiting constituent HCPs?

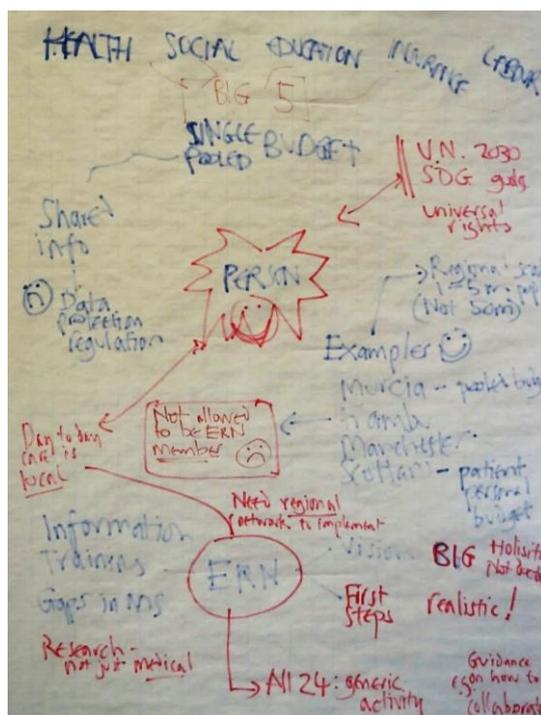
The Annex II of the [Commission Delegated Decision \(2014/286/EU\) of 10<sup>th</sup> March 2014](#) stipulates a particular duty of HCPs/CEs wishing to join an ERN:

1. (b) "With regard to organisation, management and business continuity, applicant providers must: (iv) ensure coordination with and easy access of the provider to other resources or specific units or services necessary for managing patients".

- **Are these other resources, units or services always strictly medical?**
- **Also consider the potential roles of ERNs and CEs recommended by the EUCERD** and how they might play a role in bridging the gaps between health and social care for RD (see pages 2 and 3).

## Outcomes of the discussion

Poster developed during the session:



### Examples of good practices:

- Spain – the region of **Murcia approved a plan for integrated care for RD**, including health and social care; a **12 million € budget** was assigned to the plan, for **3 years** of implementation; the **budget is provided by the different administrations**; the plan includes the use of **case management for RD**, using an interconnection case manager in the hospital and 9 case managers in the community; the region has implemented data sharing between health and social administrations;
- Manchester – pilot project, with a **single pulled budget for health and social**;
- Scotland – the **patient is in charge of their own budget** and they can swap it between needs
- England – people with disabilities are entitled to have a personal budget;
- Norway – you have the right to a CM if you have a long life disability.

**Points raised during the discussion:**

- There is a need for **cross-budgetary system**, as it is not ideal that the health budget pays everything. In RD the health cost is one of the less demanding ones; education, work etc. are much more demanding;
- **Day to day care should be given locally;**
- It would be useful to know the countries that have **instituted universal rights at the national level;**
- **United Nations agenda** – we should take in consideration the united Nations (SDG) UN Sustainable Development Goals;

**The role that ERNs can undertake:**

- Offer the model of holistic care; it was proposed that ERNs should consider **the 5 aspects of holistic care** from the beginning (health, social care, education, insurance, work); it also would be useful to separate the concepts of multidisciplinary and holistic care;
- **Produce information on RD** e.g. leaflets, training courses, research on social and educational aspects; they could also create e-learning platforms; to produce this information, ERNs can use information also produced by the European Network of Resource Centres for RD – [RareResourceNet](#);
- **Support patient empowerment** and alertness by providing information and materials e.g. webinars;
- **Report GAPS of knowledge in member states;** and **provide guidance on how to collaborate on an administrative level;** ERNs should have a regional network to implement at a local level the guidance emanating from the ERN;
- There is a need to evaluate the impact of ERNs, as they are evolving over time; the Network maturity matrix could be used for this.

## S2. Patient empowerment & engagement: strategies to ensure person-centred care & meaningful patient engagement in care design

Chair: Raquel Castro, INNOVCare partner; Rapporteur: Rebecca Tvedt Skarberg, European Patient Advocacy Group (ePAG)

First Name	Last	Organisation	Profile	Country
Carolin	Engelhorn	Kindness for Kids	Social Service	Germany
Gunilla	Jaeger	Ågrenska Resource Centre	Social Service	Sweden
Judit	Germuska	Great Ormond Street Hospital London; ERN EpiCARE (Epilepsies)	European Reference Network	UK
Judit	Váradiné Csapó	Muscle Dystrophy Association; ERN EURO NMD (ERN on neuromuscular diseases)	Patient Representative	Hungary
Lenja	Wiehe	EURORDIS - Rare Diseases Europe	Patient Representative	France
Lisen	Julie Mohr	Frambu Resource Centre for Rare Disorders	Social Service	Norway
Monica	Solomie	Ministry of Work, Family, Social Protection and Elderly	Public body/National	Romania
Oda Marie	Jordal	Dermatology, Oxford University Hospitals; ERN Skin	European Reference Network	Denmark
Raquel	Castro	EURORDIS - Rare Diseases Europe	Patient Representative	France
Rebecca	Tvedt Skarberg	Norwegian National Advisory Unit on RD; ERN BOND (Bone disorders)	Patient Representative	Norway
Tiina	Stelmach	Estonian Ågrenska Foundation Resource Centre	Social Service	Estonia
Vibeke	Sparring	Karolinska Institutet	Academic	Sweden

### Challenges addressed:

- Patient engagement: involvement of patient experts as equal partners in the design and delivery of care services and policies;
- Person-centred, holistic and participative care: guaranteeing that care is provided according to the individual needs and wishes of the patient/carer;
- Patient empowerment: building the capacity of patients to manage their daily life, aiming at increasing quality of life, self-management and prevention.

### Guiding questions:

- Can you think of any good practices relating to this topic, which work well in your experience?
- What needs to happen within this topic, to improve the status quo for rare diseases?
- What might ERNs specifically do to make an impact?

### Sources provided in advance to inspired the discussion – Index, details in respective sections below

- **Recommendations from the Commission Expert Group on Rare Diseases**
- **Challenges and strategies emerging from INNOVCare's interviews at national level**
- **Reflections on the possible role of European Reference Networks**

### Recommendations from the Commission Expert Group on Rare Diseases

Extracts of [Commission Expert Group on Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#), adopted unanimously by all MS in 2016:

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: 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 CEs on Rare Diseases](#) (4, 9, 10): 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 ERNs for Rare Diseases](#) (10) and the [Directive on patients' rights in cross-border healthcare](#) (Article 12, 4-ii): RD ERNs need to **collaborate with each other, as well as with patient groups**, health and social care providers;

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.

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.

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**.

### Challenges and strategies emerging from INNOVCare’s interviews at national level

*The interviews were conducted mostly with public bodies and healthcare providers, in a selection of EU countries. The issues listed are therefore not a reflection of the perspectives of all stakeholders/national contexts.*

#### ✓ **Communication between doctors and patients**

Challenges mentioned in interviews:	Strategies mentioned in the interviews:
<ul style="list-style-type: none"> <li>• Lack of time of doctors: “Doctors spend a lot of time on things which are not their core competences e.g. administration. This time is lacking for the treatment” (health service, Romania); “Doctors have little time to listen to the patients”(health services, Austria);</li> <li>• Complex cases are considered time-consuming, but are not remunerated adequately. “The treatment of patients with complex needs is time consuming. Yet this is not taken into account by the insurance scheme and not refunded adequately. Hence primary care doctors prefer to have less complex cases to deal with.” (Health care administration, Austria).</li> </ul>	<ul style="list-style-type: none"> <li>• Communication training for doctors (Health care expert, Austria);</li> <li>• Case management: to provide orientation and information (case manager, Spain);</li> <li>• Care plan that doctors, patients and their relatives decide on together (health care expert, Austria);</li> <li>• Doctors share competences with other care professionals; e.g. family/community-nurses can guarantee a low threshold access to care, answering to the patients’ and relatives’ needs (health care expert, Austria);</li> <li>• Shared trainings for patients, doctors and social care professionals: “Paediatricians who took part in the training program, involving patients and their relatives, reported that they got deeper knowledge of the diagnosis and its complexity as well as increased knowledge on the everyday life of people with RD. They said that the trainings were very helpful for their professional life. (Care provider, Sweden).</li> </ul>

#### ✓ **Lack of trust in the competences of patients; Patients and carers lack information on the disease**

Challenges mentioned in the interviews:	Strategies mentioned in the interviews:
<ul style="list-style-type: none"> <li>• The competences of patients in organising their care are not taken into account appropriately. The patients themselves, the doctors and the political bodies lack trust in these competences;</li> <li>• The relation between doctors and patients is rather hierarchical. Patients ask the doctor how their diseases can be cured; They have rather low health competences, lack information on their disease and do not consider themselves to have high agency in making relevant decisions concerning their care (Health services, Austria);</li> <li>• “Doctors are not used to take patients expertise into account. The health competences and abilities of patients are considered rather low. These hierarchies negatively influence the way doctors communicate with patients (Health care administration, Austria); “Integrated care means to actively involve the patients in the organization of care. They have to be empowered to care for themselves”(Health Services, Austria);</li> <li>• Political bodies do not trust the competences of patients and their relatives to organise care: “Case managers could solve the issue that families who are poor often use the money for daily life and not for the disability of their child. Case managers should therefore coordinate and check if the money is used for the patients’ needs and not for other family problems” (Romania, social care administration).</li> </ul>	<ul style="list-style-type: none"> <li>• Self-reporting systems for patients: “We have patient reported outcomes. They are reading the outcomes and can do self-reporting. That is very nice. The system used to be quite paternalistic. But this culture is changing now.” (Health services, Romania);</li> <li>• Trainings for patients and their relatives;</li> <li>• Guidelines for patients;</li> <li>• Help-lines for patients and their relatives informing on services.</li> </ul>

✓ **Low recognition of the volunteer work of self-help groups**

<p><b>Challenges mentioned in the interviews:</b></p> <ul style="list-style-type: none"> <li>• A lot of work needs to be done to inform and empower patients and to advocate for RD patients is done by patient organisations. However, they work mainly voluntarily;</li> <li>• Patient organisations cannot be solely responsible for the empowerment of patients, but it also needs to be put on the agenda of political bodies.</li> </ul>	<p><b>Strategies mentioned in the interviews:</b></p> <ul style="list-style-type: none"> <li>• Financial support for self-help groups.</li> </ul>
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**Reflections on the possible role of European Reference Networks**

Annex II of the [Commission Delegated Decision \(2014/286/EU\) of 10<sup>th</sup> March 2014](#) states as follows:

<p>ANNEX II</p> <p><b>CRITERIA AND CONDITIONS FOR APPLICANTS FOR MEMBERSHIP OF A NETWORK</b></p> <p><b>1. General criteria and conditions for all applicant healthcare providers</b></p> <p>All applicants wishing to join a Network shall comply with the following criteria and conditions:</p> <p>(a) as regards patient empowerment and patient-centred care, applicant providers must:</p> <p>(i) have put strategies in place to ensure that care is patient-centred, that patients' rights (such as the right to informed consent; the right to information concerning their own health; the right to access to their medical records; the right to privacy; the right to complain and the right to obtain compensation, the right to be empowered and to participate (for example, through customer relations management strategies, patient education strategies and active engagement strategies for patients and families throughout the healthcare institution)) are respected;</p>
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The [Addendum](#) to the original EUCERD Recommendations contains quite specific proposals regarding patient engagement in the Networks:

***“Necessity of Patient Involvement in RD ERNs:** Patients and patient representatives should play an integral role in the decision and opinion making process in RD ERNs and be involved in structural and clinical network activities. It is recommended that RD ERNs demonstrate meaningful patient involvement, patient-centeredness and empowerment through recognition of the role of patients, as experts by experience and co-producers of knowledge, in RD ERN structural and clinical activities and therefore demonstrate meeting the legal requirements in the Delegated Acts.*

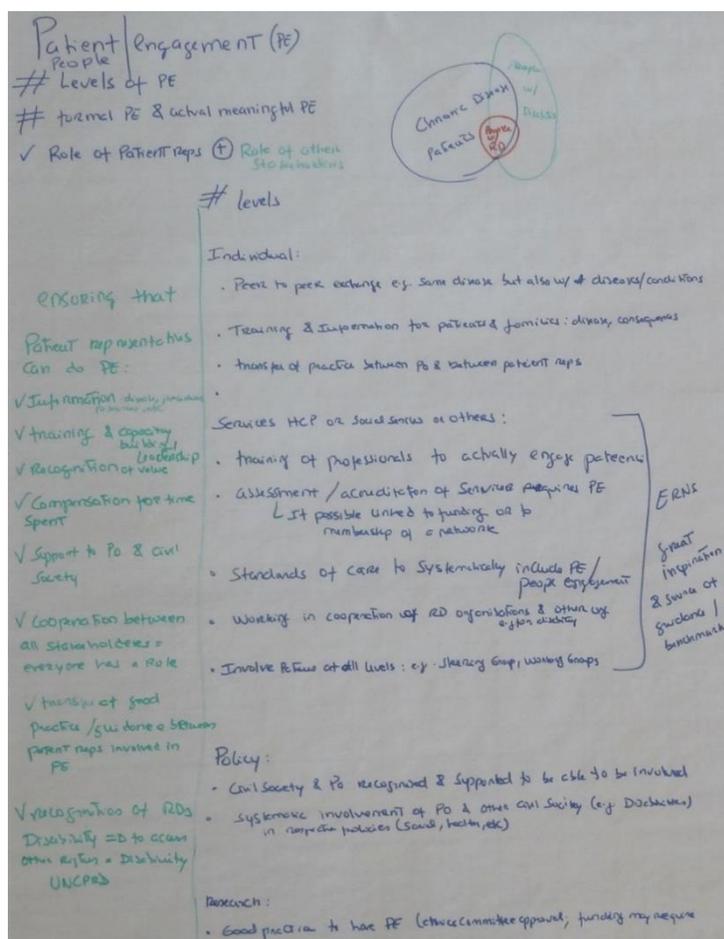
*Patients' involvement depends on the scope of RD ERNs in the following dimensions:*

- *To advise on planning, assessment and evaluation of Centres of Expertise and European Reference Networks based on their experience, with a consistent approach*
- *To ensure transparency to quality of care, safety standards, clinical outcomes and treatment options*
- *To promote and encourage a patient-centric approach in both delivery of clinical care, service improvement and strategic development and decision-making*
- *To ensure all ethical issues and concerns for patients are addressed, balancing patients' and clinical needs appropriately*
- *To ensure care is patient-centred and respects patients' rights and choice*
- *To ensure the application of personal data protection rules, compliance of informed consent and management of complaints*
- *To ensure feedback on patient experience and the active evaluation of patient experience”*

- **What role could ePAGs (European Patient Advisory Groups) play here, in encouraging HCPs to meet the requirements of Annex II above?**
- **What does patient empowerment really mean in the context of ERNs?**
- **Also consider the potential roles of ERNs and CEs recommended by the EUCERD, and how those might relate to patient empowerment and patient engagement (see pages 6 and 7).**

Outcomes of the discussion

Poster developed during the session:



Word cloud elaborated on site: **What do we mean by patient engagement?**



**General considerations:**

- It is important to define what do we mean by patient engagement;
- There are different levels of patient/beneficiary engagement;
- Formal level patient/beneficiary engagement does not always mean actual meaningful engagement;
- All stakeholders have a role to ensure patient/beneficiary engagement;
- Various policies to ensure patient engagement are in place. But, how can you make sure people with a RD who have certain disabilities are actually targeted in relevant policy/service development?
- Many RD do not have a patient organisation or only have a small one; this makes representation difficult;
- Various factors influence the degree of patient engagement e.g. level of employment for people with disabilities, level of integration into disability laws, accessibility, level of independence etc.;
- Resources are an issue when it comes to ensuring patient representation. Little or no funding is available for patient engagement, which is based on volunteer work. Many patient representatives have little time and resources to participate. This needs to be recognised and solved to make patient engagement sustainable. For ERNs to be sustainable, the ePAGs work needs to be better recognised;
- On the other hand, we need to beware to not “professionalise” the patient representative role;
- Civil organisations also need funding, employed staff and recognition in order to support patients to participate in patient engagement. Can we learn from non-RD organizations on this?
- Some experiences in RD are the same as common disorders; but some are different. What is different can be difficult to see.

**Key issues to ensure that patient/beneficiaries can be engaged in the development of services/policies:**

- Information available to patients e.g. on the disease, on procedures, available resources etc.
- Training and capacity building, including leadership training;
- Transfer of good practice/guidance between patient representatives;
- Recognition of the expertise of the patient representative and of its value, including adequate time-spent financial compensation, on equal footing with other experts (although it is important that patient representatives are able to bring forward the grass roots experience);
- Patient organisations and civil society should be supported to help provide information and training to the representatives;
- Regarding the social and daily life support, it is important that the disabilities generated by RD are recognised so that people living with RD can be involved in relevant policies/services.

**Suggestions for strategies of patient/beneficiary engagement at different levels:**

Individual level:

- Peer to peer support within same disease and with patients with other diseases;
- Training and information for patients and families;
- Transfer of practice between patient organisations and between patient representatives;

At the level of HCP and Social Services -> ERNs are paving the way in these aspects and a source of benchmark and inspiration for other services beyond HCP:

- Involvement of patients/beneficiaries should happen at all levels e.g. management boards, steering groups, working groups;
- Assessment/accreditation process of services should require that patient/beneficiaries engagement mechanisms are in place; if possible, this should be linked to their opportunity to access funding or to be members of a specific network (in ERNs for e.g.);
- Training of professional to meaningful engage patients/beneficiaries with a RD;
- Standards of care to systematically include patient/beneficiary engagement;
- Rare disease organisations to work closely with organisations for people living with disability, to ensure the involvement of people living with a RD in services designed for people living with disabilities;

Policy level:

- Patient organisations and other civil society organisations (e.g. disability) should be recognised as important partner in the development of policy and supported to provide their contribution;
- There should be systematic involvement of representatives of people living with a rare disease in the development of policies that affect their care and daily life;

Research level:

- It is a good practice to involve patients in research and the review of ethics committee as well as the funding mechanisms themselves have been having a role in incentivising this (e.g. from UK was shared);

**Specific considerations on the role and ERNs regarding patient engagement?**

- ERNs are paving the way in these aspects and are a source of benchmarking and inspiration for other services beyond HCP;
- In ERNs, we find patient engagement on a person-centred level. In some ERNs, patients have helped shape the network from the beginning. Clinical leads have worked with patients to establish the network and will be measured by the level of patient engagement from the EU;
- Today ERNs are very health care centred rather than social care centred. However, since RD are incurable we should focus more on the living with the RD, and less on "fixing it". Coping strategies, advocacy and QoL issues are more important to patients with RD and should be more important to ERNs. Maybe we need to move away from a strict medical approach.
- What is the role of the patient in a network? There is danger of lack of recognition of patients being on the same level as HCP teams. It can be difficult to become equal partners if patient engagement is seen as not possible, too difficult, topics not relevant.

### S3. Knowledge sharing: effectively promoting the exchange of good practices/expertise/information to support integrated, holistic care

Chair: Victoria Hedley, RD-Action; Rapporteur: Joanna Das, RD-Action

First Name	Last Name	Organisation	Profile	Country
Biruté	Tumiene	Board of Member States for ERNs	Public Body/National	Lithuania
Bronwyn	Kerr	Genomic Medicine, Manchester; ERN ITHACA (Congenital malformations and rare intellectual disability)	European Reference Network	UK
Emilia	Severin	University of Medicine and Pharmacy	Public body/Regional	Romania
Helena	Kääriäinen	National Institute for Health and Welfare Helsinki	Public body/National	Finland
Hélène	Dollfus	Hôpitaux Universitaires de Strasbourg; ERN EYE (Eye diseases)	European Reference Network	France
Isabella	Brambilla	Dravet Italia Onlus; Dravet Syndrome European Federation;	Patient Representative	Italy
Juan Manuel	Torres Canizales	Madrid Health Service; ERN TransplantChild (Transplantation in Children)	European Reference Network	Spain
Maria Beatrice	Michelis	Istituto Giannina Gaslini; ERN BOND (Bone disorders)	European Reference Network	Italy
Sara	Perez	Asociación nacional Síndrome de Apert y otras craneosinostosis sindrómicas; ERN CRANIO (craniofacial	Patient Representative	Spain
Silvia	Manea	Veneto Region	Healthcare Provider	Italy
Trine	Tangeraas	Horst-Schmidt-Kliniken; MetabERN (Hereditary metabolic)	European Reference Network	Norway
Victoria	Hedley	University of Newcastle	Academic	UK
Vlasta	Zmazek	Debra Croatia, Croatian Alliance for Rare Diseases	Patient Representative/Social Service	Croatia

#### Challenges addressed:

- Quality information and expertise on RD and their consequences is scarce and difficult to access;
- 65% of people living with a RD have to visit different health, social and local services in a short period of time; 67% find that these services communicate badly with each other;
- Social and support services are often closer to the patient and able to support them on a daily-life basis, however, they are typically the furthest from the specialised knowledge/expertise on RD;
- Transfer of expertise/knowledge on RD - via cooperation and transfer of information between services, elaboration/dissemination of good practices, training of professionals and patients – is recurrently mentioned as one of the most important factors to support holistic care for people living with a RD.

#### Guiding questions:

- Can you think of any good practices relating to this topic, which work well in your experience?
- What needs to happen within this topic, to improve the status quo for rare diseases? What needs to happen to ensure that knowledge, information and expertise on RD are:
  - Generated/updated (as applicable), by parties with relevant expertise, and is quality-assured?
  - Effectively and efficiently shared between care providers, in a format/language that allows them to understand the implications of the RD in their care areas? Shared with patient organisations/individual patients, in a lay language and with a view to empowering them?;
- What might ERNs do to make an impact? What might Centres of Expertise/HCPs do to make an impact?

#### Sources provided in advance to inspired the discussion – Index, details in respective sections below

- **Recommendations from the Commission Expert Group on Rare Diseases**
- **Challenges and strategies emerging from INNOVCare's interviews at national level**
- **Reflections on the possible role of European Reference Networks**

#### Recommendations from the Commission Expert Group on Rare Diseases

[Commission Expert Group on Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#), adopted unanimously by all MS in 2016:

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) (...). 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 Diseases](#) (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) and the [Directive on patients' rights in cross-border healthcare](#) (Article 12, 4-ii):

- 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 (...).

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.

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

#### Challenges and strategies emerging from INNOVCare's interviews at national level

*The interviews were conducted mostly with public bodies and healthcare providers, in a selection of EU countries. The issues listed are therefore not a reflection of the perspectives of all stakeholders/national contexts.*

#### ✓ Knowledge of RD professionals on appropriate services

Challenges mentioned in interviews:	Strategies mentioned in the interviews:
<ul style="list-style-type: none"> <li>• Doctors in primary care often do not know where to find specialized offers and therefore cannot inform their patients properly. (health care provider, Austria);</li> <li>• Doctors are not aware of existing programs as they are not properly informed by their professional interest group (health care provider, Austria).</li> </ul>	<ul style="list-style-type: none"> <li>• Regional care plans on RD;</li> <li>• Databases on services : "Public and private providers can build together a shared database on people with complex needs/Rare diseases" (regional social care administration, Romania);</li> <li>• Helplines for patients, their relatives and care givers: Knowledge of RD professionals on appropriate services;</li> <li>• Case managers can establish new networks between health and social care professionals (patient representative, Austria).</li> </ul>

#### ✓ Mainstream knowledge on RD

Challenges mentioned in interviews:	Strategies mentioned in the interviews:
<ul style="list-style-type: none"> <li>• RD patients are a heterogonous, small group. Due to a lack of visibility of RD, doctors (especially in primary care) often lack the expertise to correctly diagnose RD;</li> <li>• "Existing guides to doctors on RD are too complex" (health care provider, Austria);</li> <li>• RD patients often have to wait long for a diagnosis and can be mistreated;</li> <li>• Awareness raising and networking is currently mainly performed by patient's organisations. Contact to political key-figures is essential to be successful.</li> </ul>	<ul style="list-style-type: none"> <li>• Trainings for primary care doctors: "Paediatricians who took part in the training program, involving patients and their relatives, reported that they got deeper knowledge of the diagnosis and its complexity as well as increased knowledge on the everyday life of people with RD. They said that the trainings were very helpful for their professional life. (care provider, Sweden).</li> <li>• Easy understandable guidelines for primary care professionals on RD: "We need good, comprehensive and understandable guidelines primary care centres can use. They have to comprise information facilitating networking and communication. (health care administration, Austria);</li> <li>• Educational protocols for carers and teachers: "We have an education protocol which helps teachers, social workers at school and paediatrics to decide on how to act if they have pupils with RD" (patient representative, Spain).</li> </ul>

✓ **Build Knowledge on RD**

Challenges mentioned in interviews:	Strategies mentioned in the interviews:
<ul style="list-style-type: none"> <li>Centres of expertise contribute to the generation and dissemination of knowledge on rare diseases. A dedicated budget for centres which are not attached to hospitals would ensure sustainability and foster independency of these centres from investors (i.e. pharma industries) (patients representative, Austria);</li> <li>Social care professionals often lack knowledge on the progressive course of rare diseases. Hence, handicaps of people affected are not assessed properly. (health care administration, Spain).</li> </ul>	<ul style="list-style-type: none"> <li>Databases on RD/ Orphanet: “For specialised services it is very important to be part of the European Reference Networks, for knowledge exchange. Therefore, to speed up the designation of competence centres is very important. (patient representative, Austria);</li> <li>Integration of RD into Curricular of specialised doctors: “RD are an important part in trainings of genetics” (health administration, Romania).</li> </ul>

**Reflections on the possible role of European Reference Networks**

**How feasible is it that ERNs could...?:**

- 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)
- 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)

The [Commission Delegated Decision \(2014/286/EU\) of 10<sup>th</sup> March 2014](#) states as follows:

(1) Article 12 of Directive 2011/24/EU provides that the Commission is to support the Member States in the development of European Reference Networks ('Networks') between healthcare providers and centres of expertise in the Member States, in particular in the area of rare diseases (?). For the purposes of this, the Commission shall adopt a list of specific criteria and conditions that must be fulfilled by European Reference Networks and healthcare providers wishing to join and become a Member of a Network ('Member'). The Networks should improve access to diagnosis, treatment and the provision of high-quality healthcare to patients who have conditions requiring a particular concentration of resources or expertise, and could also be focal points for medical training and research, information dissemination and evaluation, especially for rare diseases.

- What types of 'information dissemination' do you feel ERNs/ their HCPs could address?**

The RD-ACTION document, [Recommended Practices for Standardising Data in the Context of the Operation of ERNs](#) (generated with the ERN Coordinators in 2017) proposed that ERNs could play an important role in generating and curating data concerning rare conditions, particularly through the Orphanet database:

3. Each ERN should, at some stage, consider reviewing the existing Orphanet nomenclature relative to its thematic grouping (i.e. disease or procedural area) – ideally, ERNs should envisage:

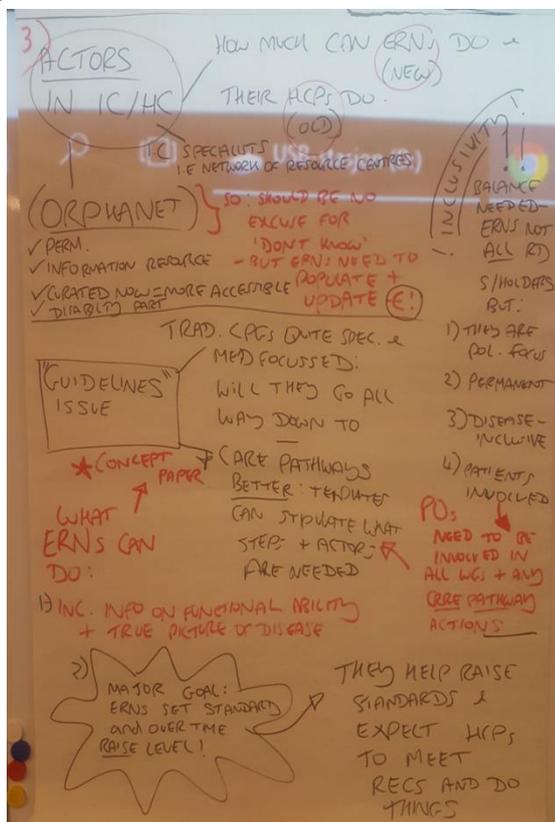
- establishing Working Groups/ Transversal groups on coding
- establishing guidelines on how to code diseases under the heading/subdomain
- contributing to the improvement and curation of the nomenclature, particularly through the Orphanet Knowledge Management System<sup>12</sup>

- What other sorts of information could ERNs/their HCPs generate, to address the gaps experienced by various sets of stakeholders (e.g. patients, primary care physicians, social care workers, educational professionals)?**
- Consider the potential roles of ERNs and CEs recommended by the EUCERD, and how those might support the knowledge sharing (see pages 12 and 13).**

### Outcomes of the discussion

#### What needs to happen within this topic, to improve the status quo for rare diseases?

A key conclusion is that in fact, Centres of Expertise/HCPs should be leading the way here – it is necessary to find a way to implement the existing Recommendations, to encourage CE/HCPs to actually deliver on their role of building bridges between medical and social arenas at the local level. Ultimately, there is an important role perhaps for MS authorities, to ensure their designated CEs (HCPs in the framework of ERNs) are forging links to paramedical and social providers.



#### Who is involved/should be involved?

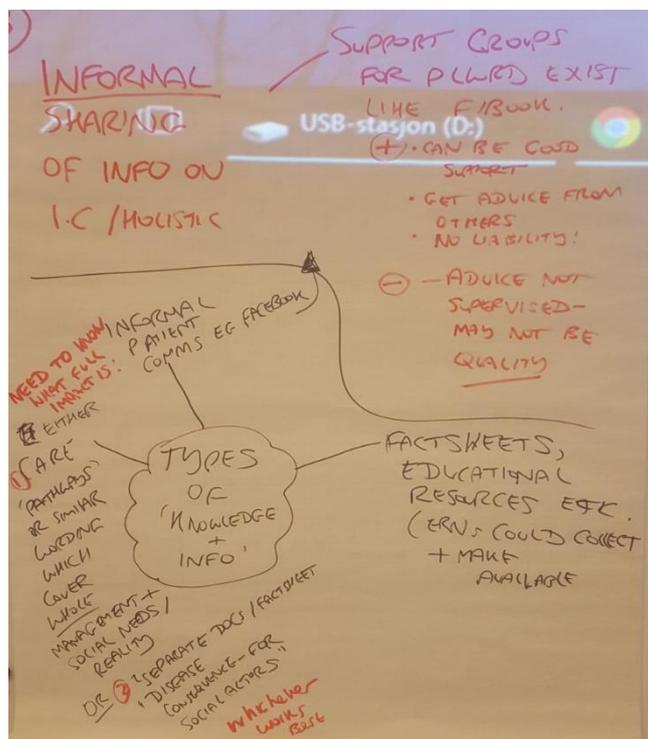
It is necessary to consider the wide range of actors involved in the provision of (ideal) integrated, holistic care for RD. The group mainly considered the following categories here: integrated care specialists (e.g. Resource Centres) and organisations which unite these (such as [RareResourceNet](#)); ERNs (which are essentially a conglomeration of HCPs or CEs); and Orphanet. The role Orphanet can play in supporting the identification of 'what works' for patients and illuminating the true needs, to support many different stakeholders, was discussed at length. Orphanet seems sustainable, and has a global outreach. It is not logical to create multiple overlapping repositories of high quality information on rare diseases. The problem is, Orphanet now seeks to cover a broad range of activities, and information on the diseases and their impact must be kept up-to-date and needs to be curated. The new Orphanet platform for curation should offer better opportunities for experts to curate the information. Orphanet has medically-oriented articles on the diseases but also, for some conditions, includes information on disability, which seek to explain the broader impact of these rare conditions. It would be good to increase the number of these disability pages, to use this global information repository as a place to find the high quality information which is needed by actors outside the medical community (e.g. educators, social care workers, families etc.)

#### Need to ensure links between formal ERN actors and everyone else

There was a strong emphasis on being realistic about the role ERNs themselves can play, but also a plea for the Networks to be open and inclusive. ERNs offer unprecedented opportunities to improve the availability of integrated, holistic care for PLWRD (e.g. they are comprehensive in disease coverages, theoretically covering all RD in time; they are designed to ensure robust and meaningful patient involvement; they are permanent, as opposed to time-bound projects; and they have significant visibility and political support (are often viewed as the 'next big thing' for RD field). However, not all centres with expertise in RD will be formally part of an ERN, and many actors engaged in integrated holistic care will also never be formally part (i.e. it is not logical to think of creating bureaucratic structures to make all such players full partners or 'affiliated' centres, for instance). Therefore, it is crucial to make best use of the ERNs and what they can bring, but allow those working in the Networks to collaborate effectively with actors outside of the Networks on this sort of work.

### What types of knowledge/information are important here?

There is a need to define the types of information and knowledge resources which fall under the category of supporting integrated, holistic care, e.g. one category would be informal patient-to-patient advice, which is very valuable for patients and families and often these days is found via social media. There is of course a question of quality here, as these sites are unregulated (no clinician involvement, traditionally –then again, the fact that these sites are *not* official means nobody fears legal reprisals for any advice provided). Another type of knowledge are Guidelines (or at least Consensus Statements/ Care Pathways – see below); then there is perhaps a third category of Disease management resources -factsheets or equivalent- for social/educational/employment actors, which may never be part of a traditional clinical practice guideline but are nonetheless crucial for this subject.



### What might ERNs do to make an impact? What might Centres of Expertise/HCPs do to make an impact?

ERNs could take a leading role in populating Orphan/curating the content relating to true impact of rare diseases, such as the disability articles mentioned above (just as [RD-ACTION recommended](#) that ERNs might be the best-placed entities to oversee improvement of the Orphacode nomenclature and phenotypic terms relevant for their ERN disease areas). However, doing this work takes time, and resource. And it will be important for the actors from ERNs to do this very much in partnership with resource centres and patient organisations etc. who are not formally inside the ERN (as for this topic, one needs many types of expertise).

Some good examples of how ERNs can add value in this area emerged in the survey and discussions on Day 1. E.g. ERNs can be the *authority* –by uniting the relevant experts outside the Networks too- to set the standard, to really discover and then elucidate what a particular condition will mean for patients in the broader sense (considering medically-oriented symptoms of course and prognosis, but also psychosocial consequences, specific concerns in terms of behaviour, less-obvious abilities and disabilities the patient is likely to experience) and to specify the components of the ‘dream team’ of multidisciplinary specialists who *should* be involved in a patients care. Although ERNs cannot describe local pathways, they *can* advise that patients with condition X should have access to the following specialists and social actors, and create/agree/highlight the sorts of information and resources that could be used by those outside the HCP/CE sphere.

ERNs can encourage the HCPs under their membership (which are –or *should* be, in the vision for ERNs-Centres of Expertise) to actually BE real CEs, that is, to fulfil the criteria established and agreed by the MS in the EUCERD Recommendations. ERNs cannot force HCPs to upskill and, for do as the Recommendations ask, i.e.: “bring together, or coordinate, within the specialised healthcare sector multidisciplinary competences/skills, including paramedical skills and social services”; “provide education and training to (...) non-healthcare professionals (such as school teachers, personal/homecare facilitators” etc.

But they CAN raise awareness amongst their constituent HCPs that such activities are important for all centres claiming to have expertise in RD, especially those which are part of an ERN, and thus actually help to set that standard in the disease area/ sub-domains of the ERN.

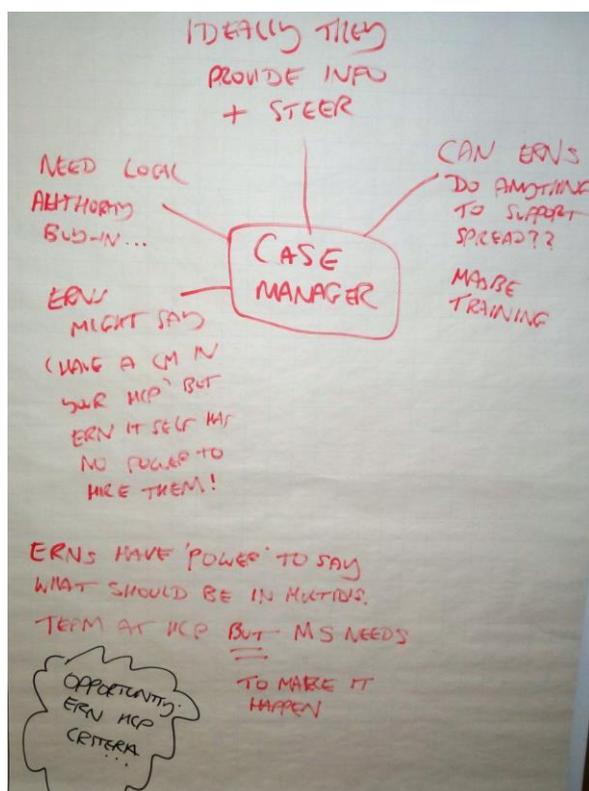
ERNs could perhaps work to create shared resources for particular diseases which can be used across the board, regardless of the national health and social care situation

- e.g. a resource on education for children with a rare eye disease could be translated –this is a key issue in fact, the need for translation of outputs- and used by teachers in countries other than France (or wherever it was created);
- a leaflet on the daily life impact of Prader-Willi Syndrome could be used in any country, as the information is simply establishing and stating the NEEDs (it is then up to the national system to agree in which buildings or which type of institution or through which budget those needs are met).

The group proposed that the following activities were therefore indeed feasible for ERNs to do (resourcing is, as always, an issue of course, and ERN funding is very scarce):

- **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)
- **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)

Related to these points, it is important to distinguish between Clinical Practice Guidelines (which are typically clinically oriented )and the sorts of Guidance needed here to impact on integrated, holistic care. One can term such Guidance as ‘Clinical Care Recommendations’ or Consensus Statements (where there is a lack of evidence base) etc. but this would not typically include advice for paramedical and social sphere. Thus it will be necessary to think of whether a different sort of resource is needed, when it comes to clinical care recommendations (which would include the various medical and paramedical -e.g. dietician- actors necessary in the care of a patient) but also putting together a ‘package’ to elucidate what the condition means for a patient’s broader societal life, in terms of key information for social care workers, educators, employers etc. Advice on ALL these areas is needed, and ERNs are in theory a perfect entity to gather that consensus across Europe and ‘speak’ with an authoritative voice to set the standards: but whether this advice could be encapsulated in one ‘Guidelines’ statement or in more than one resource, needs to be clarified.



#### S4. Professionalization of the coordination of care: who coordinates care? What is the role of case managers? How to train them?

Chair: Ursula Holtgrewe, INNOVCare; Rapporteur: Ildiko Vajda, EURORDIS Social Policy Advisory Group

First Name	Last Name	Organisation	Profile	Country
Anja	Frankenberger	Kindness for Kids	Social Service	Germany
Christine	de Kalbermatten	MaRaVal	Patient Representative/Social Service	Switzerland
David	Bergsaker	Frambu Resource Centre for Rare Disorders	Social Service	Norway
Dorica	Dan	Romanian Prader Willi Association - NoRo Resource Center	Patient Representative/Social Service	Romania
Eduardo	Tizzano	Hospital Universitari Valle Hebron	Healthcare Provider	Spain
Ildiko	Vajda	VSOP (Dutch Genetic Alliance)	Patient Representative	Netherlands
Jan	Lubinski	Pomeranian Medical University Szczecin; ERN GENTURIS (Genetic tumour risk syndromes)	European Reference Network	Poland
Marta	Fonfria	CREER (National Reference Center PLWRD and their families)	Public body/Social Service	Spain
Sebastian	Ardelean	Salaj County Council	Public body/Regional	Romania
Ursula	Holtgrewe	ZSI - Centre for Social Innovation	Academic	Austria
Veronica	Wingstedt de Flon	National Agency for Rare Diseases	Public body/National	Sweden
Zsuzsa	Almasi	Romanian Prader Willi Association - NoRo Resource Centre	Social Service	Romania

#### Challenges addressed:

- Information and expertise on RD and their consequences is scarce and difficult to access;
- 65% of people with a RD have to visit different health, social and local services; 67% find that these services communicate badly with each other ([see results of European survey on the social impact of RD](#));
- Social and support services are often closer to the patient, but typically they are the furthest from the specialised knowledge/expertise on RD;
- Transfer of expertise/knowledge on RD - via cooperation and transfer of information between services, elaboration/dissemination of good practices, training of professionals and patients – is recurrently mentioned as one of the most important factors to support holistic care for people living with a RD.

#### Guiding questions:

- Can you think of any good practices relating to this topic, which work well in your experience? What needs to happen within this topic to improve the status quo for rare diseases? How can coordination of care be ensured? What is the role of a case manager or care coordination?
- What might ERNs specifically do to make an impact? Assuming that no additional resources are available for ERNs: can you name 2-3 things they/their HCPs can do with little resources to effectively support progress in this area?

#### Sources provided in advance to inspired the discussion – Index, details in respective sections below

- **Recommendations from the Commission Expert Group on Rare Diseases**
- **Role of case manager – Outcome of INNOVCare discussions**
- **Training of case managers – Outcome of INNOVCare's discussions**
- **Skills of Case Managers – outcome of INNOVCare's discussions**
- **Challenges and strategies emerging from INNOVCare's interviews at national level**
- **Reflections on the possible role of European Reference Networks**

#### Recommendations from the Commission Expert Group on Rare Diseases

[Commission Expert Group on Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#), adopted unanimously by all MS in 2016:

3. **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 Diseases](#) (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;

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) and the [Directive on patients' rights in cross-border healthcare](#) (Article 12, 4-ii):
- 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**;
4. **MS should promote measures that facilitate multidisciplinary, holistic, continuous, person-centred and participative care provision** to people living with rare diseases (...).
5. **MS should promote measures that support patients/families affected by RD to participate in decisions regarding their care plan** and their life project (...)
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.

### Role of case manager – Outcome of INNOVCare discussions

*(Outcome of Workshop on Improving Integrated Care for People Living with Rare Diseases and Complex Conditions, Sweden, 2016)*

- Single and stable point of contact
- Listen, inform, support and empower patients and families
  - Provide knowledge/tools for patients to be able to acknowledge their personal situation
  - Inform about existing resources (services, rights, compensations)
  - Encourage health literacy and compliance to treatments
  - Develop working methods that support empowerment of patients and families
- Assess/monitor needs & be an observatory of those needs
- Holistic and patient-centred care planning and care co-ordination:
  - Identify resources available in the territory - services and support
  - Refer to available resources and speed up access to services and support
  - Simplify care pathways & support navigation for patients/families and professionals
  - Support transition periods & deliver continuous structured support to prevent care ruptures
  - Facilitate work of care providers as a multidisciplinary team: create the link/facilitate coordination between services & professionals; mobilise them to build networks
- Hub of information and knowledge
- Inform, support and empower professionals involved in care provision
- Facilitating networks of service providers
- Prevention of avoidable high risk situations
  - Identify most vulnerable cases at risk of serious situations and possible prevention measures
  - Follow up patient to avoid over-use of health services for lack of information

### Training of case managers – Outcome of INNOVCare's discussions

- |  |  |
|--|--|
| <p>1. Introduction</p> <ul style="list-style-type: none"><li>• Project presentation</li><li>• Vision, Definition of case management</li><li>• Skills, qualification and abilities for case managers</li><li>• Case studies – other national experiences</li></ul> <p>2. Rare diseases - general information</p> <ul style="list-style-type: none"><li>• Access to diagnosis in RD</li><li>• Assessment of the degree of disability</li><li>• Access to education</li><li>• Employment</li><li>• Social inclusion</li></ul> <p>3. Case management and communication</p> <ul style="list-style-type: none"><li>• Communication</li><li>• Efficient communication</li><li>• Developing communication skills</li><li>• Conflict solving &amp; Networking</li></ul> | <p>4. Resilience</p> <ul style="list-style-type: none"><li>• General information on family resilience</li><li>• Empowering patients and families</li><li>• The announcement of diagnosis and its impact on the family</li><li>• Personal development and self-esteem</li></ul> <p>5. Development and coordination</p> <ul style="list-style-type: none"><li>• Description, definition and possibilities</li><li>• Legislation - rights and obligations</li><li>• Finding the right information and accessible</li><li>• Available Services</li></ul> <p>6. Work methodology</p> <ul style="list-style-type: none"><li>• Methodology of case management for people with rare diseases and their families</li><li>• Working Tools used in case management of people affected by rare diseases and their families</li></ul> |
|--|--|

### Skills of Case Managers – outcome of INNOVCare's discussions

- Positive approach;
- Effective communication;
- Negotiation skills;
- Knowledge of contractual and risk arrangements, the importance of obtaining consent, confidentiality and client privacy;
- Ability to perform ongoing evaluation;
- Critical thinking and analysis;
- Ability to plan and organize;
- Promote client autonomy and self-determination;
- Knowledge of health, education and care services.

### Challenges and strategies emerging from INNOVCare's interviews at national level

*The interviews were conducted mostly with public bodies and healthcare providers, in a selection of EU countries. The issues listed are therefore not a reflection of the perspectives of all stakeholders/national contexts.*

#### ✓ Challenges identified

- **Reservations against Case Management (CM)** e.g. not seen as needed for all RDs; restricted to a few patients because of lack of resources; they need to adjust to existing structures and this may restrict their work; perception that CM may be needed only temporarily;
- **Training and profession of case managers** e.g. lack of training; the lack of definition on who should take the role of CM; the clear need for training and for networks in the community.

#### ✓ Strategies suggested

- Training on CM can be integrated in the curriculum of nurses;
- CM can be envisioned as a new social profession: CMs are trained for e.g. at the university;
- Supervision for CM;
- Information on the services available to support networking;
- Apps helping patients with the coordination of care can be an additional tool supporting the coordination done by case managers and helping the patients to organise care themselves.

### Reflections on the possible role of European Reference Networks

The [Commission Delegated Decision \(2014/286/EU\) of 10<sup>th</sup> March 2014](#) outlines the following role for every ERN:

- (2) To fulfil the requirement set out in point (i) of Article 12(4)(a) of Directive 2011/24/EU ('have knowledge and expertise to diagnose, follow up and manage patients with evidence of good outcomes'), the Networks must:
- (a) promote good quality and safe care to patients suffering from certain diseases and conditions by fostering proper diagnosis, treatment, follow-up and management of patients across the Network;
  - (b) empower and involve patients in order to improve the safety and good quality of the care they receive.

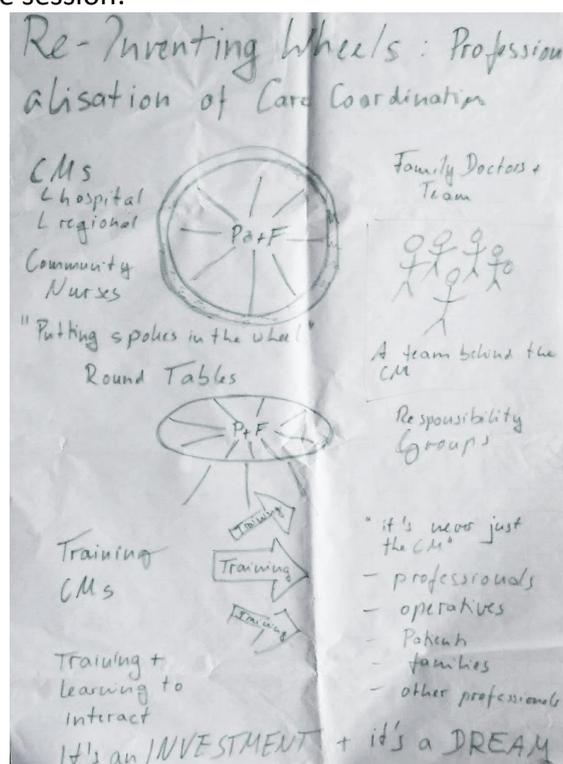
**2(a) specifies follow-up and management of patient across the Network. What about between the Networks, and outside of the Networks?**

Could ERNs...?:

- Create personalised health and social care plans for people with rare diseases, possibly both those receiving virtual referrals and the patients visiting constituent HCPs?
- Could they help to define the role of a Case Manager in their constituent Centres?
- How much could be common to all HCPs/local situations?

### Outcomes of the discussion

Poster developed during the session:



#### Main strategies discussed:

- Re-inventing the wheels: professionalisation of care coordination;
- Patients and families are at the centre;
- The Case Manager (CM) is putting spokes in the wheels – he is the engine driving the wheel;
- A CM should have different functions: keeping an eye on the patient, bridging the gap between the patient, hospital, school etc.
- CM cannot solve all problems alone. Combining resources is important;
- There should be CM in the hospital AND at local level e.g. regional level, community nurses;
- There should be a team around the CM or the CM should be part of a team (around a care provider) e.g. medical team at the hospital, family doctors, etc.; it's never just the CM there are other professionals, operational issues and the patients and families involved in the organisation of care; The patient has to know that he/she does not depend on one person;
- There should be a responsibility group for each patient (see more about this RG in the following page, under "Norway"). We imagine the activities of the team as a rolling wheel which has to keep rolling on;
- CM training is key essential;
- CM is an INVESTMENT and is a DREAM.

#### Training, skills and qualifications of case managers

- NoRo (Romania) is working on training curricula (see page 19); between 90-120 of training are needed;
- In Switzerland there is a 16-day training course for CM delivered in French; 15 people were trained in the first round;
- In Norway (Frambu) there are also courses for professionals, face to face or via video-training. There are courses for specific diagnosis or for a handicap;
- Some beneficiaries with a RD have 50-60 providers. Every day a different person helps them with daily activities. This is not a good situation, because knowledge about the patient cannot be accumulated;
- It is important that the CM is aware of the RD. The CM does not have to know all details of the RD, but needs to know the needs of the patients and the major challenges;
- CM should have a global vision about different subjects related to RD and should access training modules which are relevant for them to be able to support the patients and families; CM should be proactive and to know how and where to find resources;
- CM who work in hospitals will receive hospital-practice-oriented training modules, whilst those who work in a local care setting will receive modules that are relevant for them;
- Cooperation with a genetic counsellor is need, so that the CM is able to explain the major genetic aspects to the family.

### Examples of from different European countries:

#### Germany:

- There are social paediatrics centres by law and there are standards for how to organise these centres. Only in some cases it works well; the implementation depends on the hospital and on decision makers;
- In Munich, there are 3-4 centres, which work well: there have staff to provide e.g. medical and psychological care, physiotherapy, dietary advice etc. and legal advice to support patients and families;
- There is no national coordination for Centres of Expertise for RD and there are no standards for the centres, so the number is growing rapidly;
- There is no legal basis for the national plan for RD (NP). There are huge regional differences in care organisation. The ministry of health worked on the NP, but its implementation and the coordination of the actions downstream into the regional health care system is missing.
- Some hospitals have a Case Manager (CM), but not all. There is no extra budget from the state for a CM. Hospitals receive funding for the paediatric centre, but not for a CM.

#### Norway

- There is at least one hospital in each of the 20 Norwegian counties. There are 20 rehabilitation units for children and adults with various care providers. There is no NP for RD and everything is integrated in the national health care system;
- At the community level, there is a so-called a Responsibility Group (RG), for anyone who needs extra care and assistance and thus not only for RD. Such a group consists of care providers with different specialities e.g. GP, nurses, physiotherapists etc. (the GP is paid to be part of the RG). In each community there is a person responsible for the coordination; this has been implemented for 18 years already and works quite well; the
- Each beneficiary has a broad individual care plan including e.g. information about the diseases, future-possibilities, housing, self-care etc.;
- Empowerment of patients and families means that they are able to face the problems and know their rights. At Frambu, parents are also educated. Employer get financial compensation when parents are in Frambu, because they cannot work that week. The motto is "put resources into the system and you will have self-confident, independent individuals, who may be able to live on their own".

#### Sweden

- In Sweden, there are 290 municipalities. The CM are supposed to cooperate and to write individual care plans, but it does not always work. There is similar legislation as in Norway.

#### Switzerland

- The landscape is scattered: there are 5 or 6 university hospitals in which care is organised differently, because they belong to different municipalities. There is no NP but there is a concept of the plan, which describes what should be done in theory. There is no legal frame to make networks for RD.

#### Poland

- There is a national programme aimed at CM (only in relation to hereditary cancers). The case managers are members of the team of the GP of the family;
- The CM tasks have naturally evolved. The CM has to be pro-active and have to be able to convince patients to engage in surveillance activities. The major challenge is not to lose contact with families.

#### Spain

- There is a team of geneticists in the hospital. They identify a family at risk and a CM contacts the family. The genetic counsellor is working together with the CM;
- There is an individual plan for the family. Connection with primary service is very important. Diagnosis of all needs of the patient is important too.

## S5. Data collection: demonstrating the impact of services on patients' QoL/public health & the cost-efficiency of integrated care

Chair: Michelle Battye, EuroGen ERN; Rapporteur: Valentina Rupel, INNOVCare

First Name	Last Name	Organisation	Profile	Country
André	Gubbels	Federal Public Service for Social Security - SPF Sécurité Sociale	Public body/National	Belgium
Anica	Ježić	Ministry of Social Policy and Youth	Public body/National	Croatia
Désirée	Gavhed	Karolinska University Hospital; ERN RITA (Immunodeficiency, autoinflammatory, autoimmune diseases)	European Reference Network	Sweden
Juliet	Tschank	Zentrum für Soziale Innovation (Centre for Social Innovation)	Academic	Austria
Maria João	Freitas	Raríssimas; Casa dos Marcos Resource Centre	Patient Representative/Social Service	Portugal
Mary	Kearney	Irish College of General Practitioners; ERN-RND (Neurological)	Patient Representative/ERN	Ireland
Maud	Le Graët	Finovatis	Academic	France
Michelle	Battye	Sheffield Teaching Hospitals NHS Foundation Trust; ERN eUROGEN (ERN on urogenital diseases and conditions)	European Reference Network	UK
Peter	Lindgren	Karolinska Institutet	Academic	Sweden
Till	Voigtländer	Board of Member States for ERNs	Health Care Provider/ Public Body/National	Austria
Valentina	Rupel	Institute for Economic Research	Academic	Slovenia

### Challenges addressed:

- Lack of data on the full socio-economic impact of RD for patients and carers, including direct clinical and treatment issues (for which some degree of natural history data is needed) but also beyond;
- Lack of data on the impact of the provision of services such as integrated care, case management, etc. both on the QoL of people living with a rare disease as well as the impact for the system e.g. efficiency gains, economic impact.

### Guiding questions:

*Given that this a very broad topic, the breakout session was rather exploratory and took into account the potential roles of ERNs discussed Day 1 of the workshop, during Session 2.*

- Can you think of any good practices relating to this topic, which work well in your experience?
- What needs to happen within each of this topic to improve the status quo for rare diseases? How can data be systematically collected?
- What might ERNs specifically do to make an impact?

### Sources provided in advance to inspired the discussion – Index, details in respective sections below

- **Recommendations from the Commission Expert Group on Rare Diseases**
- **INNOVCare's evaluation model**
- **List of elements to consider when planning an evaluation model – Outcome of INNOVCare's discussions**
- **Reflections on the possible role of European Reference Networks**

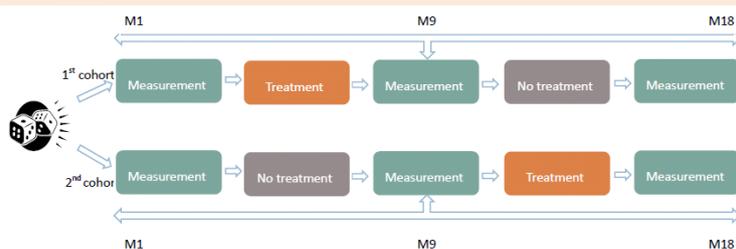
### Recommendations from the Commission Expert Group on Rare Diseases

[Commission Expert Group on Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#), adopted unanimously by all MS in 2016:

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.

### INNOVCare's evaluation model



#### Evaluation design:

- Basic two-condition repeated measures design, also known as rotation design;
- **Sampling:** random proportionate stratified sampling; stratifying variables: type of patient (NoRo or external), age group (nine levels) and sex;
- **Randomisation:** stratified random assignment (random allocation of participants into the 1st and 2nd cohorts) based on the variables: type of patient (NoRo or external), age group (three levels), sex and location (urban or rural) as well as simple random allocation of participants to the four case managers;
- **Ex-post assessment of the randomisation procedure:** checking balance of the two cohorts based on the following variables -age, age group, type of patient, sex, location, disease cluster, degree of disability;
- **3 points of measurement:** March 2017, November 2017, July 2018.

#### Questionnaires:

- **Soft items** based on the 8 main goals of the intervention as defined in the logic model of intervention;
- **Hard items** based on demographical information of household & use of healthcare resources;
- Also include existing question batteries/questionnaires e.g. DISABKids, EQ-5D, Zarit Burden Interview (ZBI).

### List of elements to consider when planning an evaluation model – Outcome of INNOVCare's discussions

*(Outcomes of breakout session from INNOVCare Workshop: "Advancing Holistic and Innovative Care for RD & Complex Conditions", 2017)*

- Take into account **previous studies and questionnaires** developed for other complex conditions;
- **Evaluation and service design need to be aligned** to take into account e.g. lines of intervention addressing the needs of the person with a RD; services already provided; competing services;
- Important to **evaluate the true benefits for the person**;
- **Evaluation is contextual:** services are contextual **to patients'/families' individual needs:** evaluation instruments taking into account the **complexity** are key; there may be added challenges for undiagnosed; Evaluation to be based on **country's context** to take into account available assets and expectations;
- **Time frame** for evaluation: tackle the time accessing case management; **Assessing long-term social and economic impact is key**;
- **Control group** to allow comparison;
- Remember to address **ethical issues**;
- Ensure **data protection** by providing data in aggregated form; ensure an adequate management of data;
- In case management assessment: interesting to **compare impact of case managers with different profiles** e.g. social worker, lawyer, nurse; case managers can also have **negative impacts**; measuring the case manager's effort when supporting 30 people with different complexity: **tracking frequency of contact with families and social services.**

#### Notes on economic evaluation of costs and benefits:

- Clarify what will be measured: the **hypothesis is that the intervention might be more costly but will provide better quality of life and societal benefits**;
- Measure **economic savings in health care**;
- **Other costs to be considered** e.g. pharmacy, respite care or other social services;
- It would be useful to **analyse the social network** from an economic impact point of view;
- Consider **services also represent job creation** for municipalities/regions/care providers;
- For some services the budget is pre-defined: make sure to **assess the cost of actual services provided**;
- **Salary of case management** to be evaluated in comparison to the national average wage.

#### Example of evaluation instruments:

- **Quality of life impact:** [Zarit](#) to measure burden of caregivers; [Barthel](#) for cases where physical impairment is crucial; instrument to **measure the complexity** of disease and care pathway is important; **Consider social evaluation** – difficult to find instruments that measure the right outcomes; **Specific instruments to measure QoL in children with cognitive impairment**;
- **Economic impact:** [MAFEIP](#) "Monitoring and Assessment Framework for the European Innovation Partnership on Active and Healthy Ageing"; Model from [Canada \(PRISMA\)](#).

## Reflections on the possible role of European Reference Networks

This is a very broad topic, potentially, depending on how one defines it! As per the Workshop Outline, how could ERNs:

- **Contribute to the collection and integration of data, to improve knowledge and understanding of rare diseases and their impact on patients and wider society?**

More specifically, how might the ERN/the HCPs which are part of ERNs contribute (eventually) to the collection of **several categories of data**:

- One the one hand, we can think of simply evaluating patient satisfaction with their ERN referral, and indeed this was already incorporated already to the core indicators table (see below);
- The next level might be to think about questionnaires etc. which member HCPs could eventually routinely disseminate to all RD patients they see, in order to capture comparable data on experience of care in centres of expertise for rare diseases. This falls into the debate of what is a HCP responsibility and what an ERN can influence or not – and many HCPs already do this, in their own way, so we need to think carefully about what one would collect and WHY;
- More detailed and ambitious level - several coordinators have been vocal in a desire to explore how patients might contribute information on their disease symptoms and QoL experiences via registries, surveys, apps etc. In other words, ERNs -or at least the Centres of which they are composed- could play a powerful role in increasing our knowledge base on the real impact of rare conditions on patients and families, and it might be interesting to think about how this can be done.

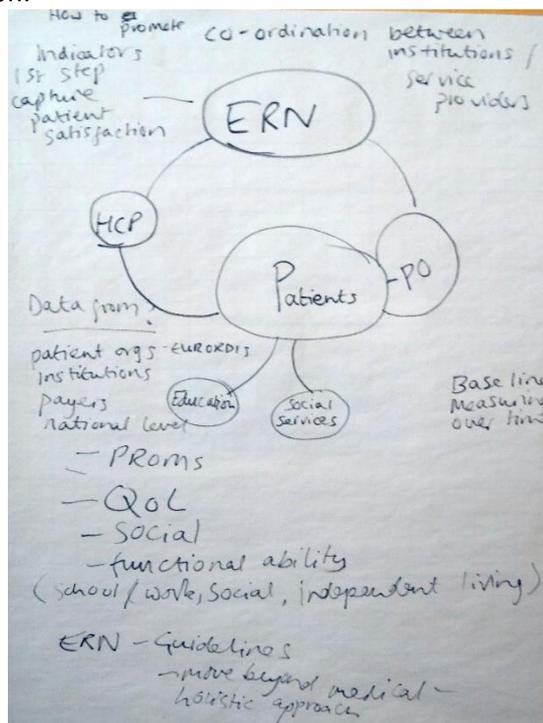
**In March 2018, a document on ‘Continuous Monitoring of ERNs’ was adopted by the Board of MS of ERNs. This document contains a table of proposed core indicators which all ERNs will collect, “to allow a closer follow up of the activities performed by the networks. This system should help to build a quality improvement system, to define appropriate outcomes of the ERNs, to identify areas of success and potential pitfalls and to demonstrate the value of the ERNs, ultimately learning from the experience.”** The document was initially drafted by DG SANTE, was refined during an RD-ACTION workshop, and then further revised and finalised by the dedicated **Working Group on Monitoring and Assessment of the ERNs. The issue of how to collect/asses patient satisfaction was highlighted for future, longer-term attention as follows:**

- **How might we move beyond this first step, to in-time collect more meaningful data on the impact of Centres of Expertise/ERNs, and on the impact of integrated care for people with rare diseases/complex conditions?**

Long-term indicators and/or indicators which need further development	
Indicator name	Examples of questions to be discussed
Level of patient satisfaction	<p>Patient satisfaction (of patients who have been entered in CPMS, had a panel review discussion and for whom an outcome report has been produced) could be measured using the Rare Barometer Voices, which is a tool used to carry out EURORDIS quantitative surveys on issues affecting people living with a rare disease. The Rare Barometer Voices survey software enables high-quality, secure data collection and analysis. It is recommended that as a first step, adults are asked the following questions:</p> <ul style="list-style-type: none"> <li>• Did you give your consent for your data to be shared for: the Clinical Patient Management System; registries; research? (Yes or No).</li> <li>• How satisfied were you with the information given to you about the ERN? (Not satisfied; Quite satisfied; Very satisfied).</li> <li>• Was the outcome report giving the ERN advice or recommendations shared with you? Yes or No.</li> <li>• How satisfied are you with the ERN advice or recommendations? (Not satisfied; Quite satisfied; Very satisfied).</li> <li>• How satisfied are you that your treating clinician involved you in discussions about how the ERN contributed to your case? (Not satisfied; Quite satisfied; Very satisfied)</li> <li>• Do you have any comments or suggestions about the activities of the ERN?</li> </ul> <p>The process of the measurement of patient satisfaction needs to be defined as an integrated process of monitoring the ERNs. The adult patient, and in case of children, parents should complete the question outside the hospital environment, following provision of information of where and how to complete the online-survey, by the clinician. Funding will be needed to make any necessary adaptations to the Rare Barometer Voices and to translate the questions into different languages.</p> <p><u>Comment:</u> The assessment is not regarded as research and therefore not considered to need the same requirements. It is a proposed first step. Research with regard to measurement of patient satisfaction would require different considerations such as a robust and patient-centred methodology of the development and validation of a cross-cultural instrument. Such an instrument is adjusted to the specific population, on which it aims to measure and need to follow standards for translation/cultural adaption of questions.</p>

### Outcomes of the discussion

Poster developed during the session:



Group discussion focused on the role of ERNs in assisting data collection.

#### The role of ERNs

- The primary role of ERNs is clinical guidance at EU level in a sense of «expertise networks of influence»;
- Being first entities for cross border structural cooperation between clinicians and patients, ERNs have power to be trusted when defining clinical indicators and guidelines and shining light on best practices;
- However, ERNs for now are focused on clinical/medical data. The indicators for clinical excellence are clearer and attainable;
- The main question is: do we go beyond and is there a role for ERNs to play in holistic care for patients?
- The working group agree that the wider indicators are important, meaning Quality of Life (QoL) and social functioning (education/work, independent living, housing); ERNs' role is in the definition of indicators/of the best outcomes set, of indicators measuring coordination and quality; and of indicators to assess the level of holistic care;
- It is unknown whether the indicators of holistic care are possible to define at EU level as the system, stakeholders, institutions, funding etc. vary across EU MS.

#### What data to collect?

- The group agree that both types of data can be collected by ERNs: clinical data as well as experience and evaluation data;
- Currently, the data depends on the indicators set by ERNs;
- The role of the patient organisations in ERNs is not yet clear (although the situation seems to vary across ERNs); Do or should patients have a role in Clinical Patient Management System (CPMS), for instance? Do or should case managers have a role in CPMS?
- The data beyond clinical that is especially missing is related to the socio-economic data and to the impact of treatments received by the patients (PROMs, QoL, patient satisfaction). These data is sometimes collected, but is mostly project-based and not structural/registry type. There are also exceptions like patient organisation for haemophilia which has collected data at European level;
- The goals of integrative care are clear, but it is not clear how to measure and how to assess to what degree those goals were achieved.

### Who collects the data?

- Due to the differences in the systems, the data needs to be collected always at national levels;
- Still, thinking in terms of cross-sectorial data, who should be responsible for gathering data on the impact on patients generated by various actions across sectors e.g. health, education, housing, social life, treatments?
- The public administration is organised in silos so, this type of data collection is highly improbable there.
- Other suggestions of institutions who can collect data:
  - Institutions like payers of care, who want the care to be effective and want to provide incentives to support integrative care – in turn they need to evaluate it and thus collect data;
  - Patient organisations who have access to the patients and can collect data from them. Patient organizations in that case should have a goal to promote and stand for integrated care – consequently, they must be empowered to stand for this goal. The interest to move towards integrated care must be expressed by the patients through patient organisations;
  - Health care providers who can pass the links to surveys to patients, which is rather limited option, but can be used for patient satisfaction with advices put forward by ERNs. If satisfaction with ERN advices is measured, it is important to see whether the advices were implemented.

### When to collect the data?

- The data (indicators) beyond the clinical sphere are to be defined after the collection of clinical data becomes a clear-cut process and people have experience and ERNs are fully operational;
- The data is needed in order to define and adapt the evolution of patients' holistic care;
- Currently, there is a need to map the data in registries to see what exists and what is needed additionally. The data beyond clinical needs to be longitudinal, otherwise it will not make sense.