



Centres of Expertise and Quality of Care for Rare Diseases

Report from the EUCERD Joint Action WP7

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Executive Summary

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KEY POINTS

Stakeholder participation

- 15 Centres of Expertise in 11 Member States plus Patient Organisations and health authorities.

Methodology

- In-depth three month case-study; 63 interviews, 3 Expert Roundtables
- Focus on the development of systems of practices to drive improved Quality of Care for Rare Disease (RD) patients in Centres of Expertise.

Main Findings

- Variation between regional and national health systems and different Rare Diseases are shaping differences in the organisation and activities of CEs and their capacity to drive improvements in QC for Rare Disease patients.
- CEs are having major impact on several dimensions of Quality of Care for RD patients:
 - Enabling ACCESS for current and prospective RD patients;
 - Enhancing the EFFECTIVENESS of clinical services for RD patients;
 - Developing a committed culture of PATIENT-CENTRED CARE for RD patients and their families.
 - A context of elevated PATIENT SAFETY is being created.
- CEs are having increasing impact on further dimensions of Quality of Care for RD patients:
 - Processes and structures are being put in place to enhance CONTINUITY OF CARE, particularly with regard to the transition from childhood to adulthood.
 - Efforts are being made on multiple fronts to improve the TIMELINESS of care, including building pathways from primary care and improving the design and management of new patient circuits.
- Adequate support staffing, particularly specialist nursing and case management skills, is central to CEs capacity to drive improvements in QC.
- Patient Organisations continue to focus on the unevenness of access to timely diagnosis which remains a fundamental challenge for CEs, ERNs and MS.

- Genomics/NGS technology is central to many CE futures and is already transforming practices; capturing QC benefits from these technologies is a key next challenge for CEs.
- Telemedicine retains untapped potential for improving QC for RD patients – more needs to be known about current telemedicine practices, opportunities and user needs.
- Sharing CE best practices in producing and diffusing information for patients and professionals, and experiences in brokering knowledge among stakeholders, could drive improvements in QC across all disease and health system contexts.
- Improving links and coordinating services with social care providers and social services is a challenge recognised by many CEs as an avenue to significantly improve QC.
- The activities and organisation of CEs and the strategic vision described in the EUCERD Recommendations are closely aligned.
- A high demand for participation in ERNs for RD can be anticipated to a) access capabilities; and b) facilitate cross-border patient access to established CEs.
- From a QC perspective, ERNs appear to have the potential to drive improvements in both the Effectiveness and the Efficiency of delivery of services for RD patients in MS.
- From a learning perspective, QC appears to have potential as a framework for the context sensitive diffusion of good/best practices between regions and MS.

In participants' own words

When we have, for example, some **specific problems to be solved that needs different expert collaboration**, we **put together the experts and we discuss together**. (CE Clinician)

We are very dependent on our clinical nurse specialist that coordinates many of these programs where we have conditions referred for further evaluation. [It's] a really big job **trying to coordinate all the evaluations with the other specialists and then with the families**. That's really very big work... (CE Director)

It becomes more and more obvious that a lot of the clinics around Europe and also probably in the US, have this problem that we started out originally as part of a children's department, many of us are paediatricians by training, but as the patients get older and they stay alive, I think **many of the Clinics are trying to figure out a solution that is good for the patient** to still have this care that they need... (CE Clinician)

Some of the new technologies, what we are capable of doing using **new technologies are sometimes pretty difficult to understand**. Like what results we can expect using this or that technology. So, we -- again -- try to introduce that during our meetings like reintroducing the new test; we have this new technology. We have the instrument here; we introduced this new test. (CE Clinician & Lab Manager)

A benefit of being one part in these trials is the **sharing of standards, procedures and quality**, that kind of thing. We can take on some new ways but also we learn the way some things are done internationally where we don't have much experience. (CE Director)

So, **the social worker in particular is important...** as contact with the local regions. I mean to get them reimbursement and to get them help with home physiotherapies and things like that. **There's a lot of communication there to get the correct help to the families**. (CE Director)

It's like a cycle with a social worker, a doctor, and they go through certain treatments and they advise them how to do things from how to - basically how to proceed the day with [the condition]... **It's really hard from a psychological point of view**. It only takes a week, but you cannot really realize or get to the realization that your child is not well ... It is hard for the families. **That's where the [patient] association is closely cooperating with the Centre of Expertise**. We arrange for meetings with other diagnosed families so they can talk about stuff and just feel better about everything, if you can feel better about it. (Head of Patient Organisation)

Introduction

The EUCERD Joint Action: Working for Rare Diseases started on 1 March 2012 and will support for a three-year duration the activities and mandate of the Committee. From 2014 it supports the activities of the European Commission Expert Group on Rare Diseases (CEGoRD) which replaces EUCERD. This Joint Action is led by Prof. Kate Bushby, former Vice-Chair of EUCERD and member of CEGoRD.

EUCERD was mandated to assist the EC in formulating and implementing the Community's activities in the field of rare diseases, to foster exchanges of relevant experience, policies and practices between the Member States and stakeholders. Rare diseases are a priority area for action in the Public Health Programme (2008-2013). These activities have been defined in the Communication of the European Commission, entitled "Rare Diseases: Europe's challenge" (11 November 2008) and the Council Recommendation on an action in the field of rare diseases (8 June 2009). Specifically, this Joint Action addresses the following priority areas of the Council Recommendation:

- Enhancing the visibility and recognition of RD.
- Contributing to the development and dissemination of knowledge on RD, from specialised research, through to the support of the healthcare professionals and the empowerment of patients.
- Contributing to improvements in access to quality services and care, from diagnosis, through to care and social support and innovative therapies.
- To achieve its aims, the Joint Action builds on the achievements of previous European initiatives in the field, such as the EC Rare Disease Task Force, Orphanet, the Europlan project, and the outputs of and the several rare disease networks that have received EU funding over the past years.

This Joint Action comprises five main areas of work:

- the implementation of plans and strategies for rare diseases at national level;
- the standardisation of rare disease nomenclature at international level;
- mapping the provision of specialised social services and integration of rare diseases into mainstream social policies and services;
- the leveraging of the value of EU networking for improving the quality of care for rare diseases; and
- the integration of RD initiatives across thematic areas and across Member States.

The expected outcome is an integrated strategy for the implementation of rare disease policies through the exchange of experience between Member State health authorities already involved in rare disease policy definition and implementation and via a series of recommendations from the EUCERD and clear communication of these recommendations to national policy makers, patient organisations and learned societies.

Work package 7: Task, objectives & approach

In the context of the EUCERD Joint Action, work package (WP7) aimed to identify actions of Centres of Expertise (CEs) for rare diseases (RD) leading to improved Quality of Care (QC) for RD patients within the healthcare systems of EU Member States (MS). Healthcare here is defined very broadly and includes the entire continuum of services, from diagnosis to care, rehabilitation and social services. WP7 explored organisational and professional practices in CEs that can drive QC improvements. It sought to identify good practices, particularly those systems of practices impacting positively on QC for RD patients. The major objective was thus to **identify actions that could improve quality of care in rare diseases** and this was addressed in the context of three dimensions that vary between (and within) MS:

- The relevance of the country profile and the disease profile for the actions and policies adopted at national and European levels, by a range of interested stakeholders;
- Healthcare systems policies and preparations for working for RD; and
- MS decision-making regarding CEs and their role in healthcare system innovation for RD.

WP7 was designed to **identify specific avenues to improve Quality of Care for RD patients** in MS, including:

- Opportunities for improving QC by linking national dedicated structures (i.e. CEs) with European Reference Networks (ERNs) for RD;
- Identifying new or emergent questions or challenges, or neglected aspects, that impact on the delivery of the best possible QC for RD patients;
- Identifying guidelines, strategies and good practices for delivering QC for RD patients, as well as additional areas where these are required; and
- Calling attention to any major issues that appear to have been overlooked by the RD community in relation to the CE/QC nexus.

The WP7 approach was built around **three strategies for accessing expert opinion** regarding actions of CEs that lead to improvements in QC for RD patients.

- First, an **in-depth case study** was conducted in one CE over a period of three months.
- Second, a **structured interview series was conducted involving fifteen CEs in ten MS**. This interview series included CE Directors and staff, linked Patient Organisations (PO) and national or regional health administrators/policymakers.
- Third, a **two-day Workshop** was held involving a range of stakeholders. The Workshop included opportunities for presentations from participating CEs, Roundtables on specific issues and open discussion.

Information gathering regarding CEs focused on **process and organisation at the level of professional practices**.

- What are CEs doing, or wanting to do, across the entire continuum of services, to improve QC for RD patients?

- 'Practices are dynamic': How are activities and their organisation being transformed? What factors are driving changes?
- How are CE management and strategy evolving and influencing change?

1. A framework for understanding Centre of Expertise led improvements in Quality of Care for Rare Diseases

A framework was developed for situating the professional practices making up the work of CEs for RD in relation to improvements in QC. The **unit of analysis** was those groups or systems of practices on which the activities of CEs are based.

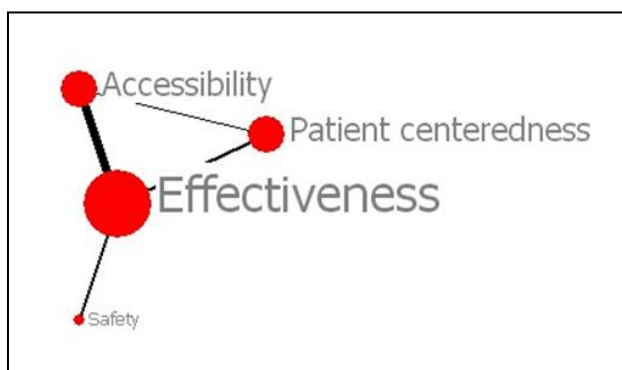
Guidance on the organisation and primary activities of CEs was provided by the [EUCERD Recommendations for CEs](#). The Recommendations served as strategic markers for the implementation and development of CEs as a health system innovation. WP7 focused on Recommendations relating to the Mission and Scope of CEs, Designation Criteria and the European Dimension.

Prominent **definitions of Quality of Care** were reviewed including Donabedian (1980), IOM (1990) and WHO (2000). A simple working definition of improvements in QC for RD patients was settled on: **identifiable improvements in patient services and/or patient welfare**.

The EUCERD Recommendations were analysed from a QC perspective (excerpt shown).

MISSION & SCOPE	Accessibility	Effectiveness	Patient Centredness	Safety	Efficiency
01. CEs tackle diseases or conditions requiring specific care due to the difficulty in establishing a diagnosis, to prevent complications and/or to set up treatments.		1		2	
02. CEs are expert structures for the management and care of RD patients in a defined catchment area, preferably national, and at international level if necessary.	1		2		
03. The combined scope of all CEs within a MS covers all RD patients' needs, even if they cannot provide a full range of services with the same level of expertise for each RD.			1		
04. CEs bring together, or coordinate, within the specialised healthcare sector multidisciplinary competences/skills, including paramedical skills and social services, in order to serve the specific medical, rehabilitation and palliative needs of rare diseases patients.		1	2		
05. CEs contribute to building healthcare pathways from primary care.	1	2			
06. CEs have links with specialised laboratories and other facilities.		1			
07. CEs collaborate with patient organisations to bring in the patients' perspective.		1	3	2	

The Recommendations are strongly focused on three **dimensions of QC**:



Information on the actions of CEs to improve professional practices and their organisation was gathered to highlight the extent to which CE activities 'on the ground' are a) **aligned with the vision of the EUCERD Recommendations**, and b) **driving improvements in various dimensions of QC**.

2. Main findings about CEs and improving Quality of Care for Rare Disease patients

Many avenues to improved QC for RD patients were identified in the actions of CEs, CE staff and associated organisations. At the level of everyday practices, these avenues are too numerous to document and acknowledge in this summary document. More detail on these activities is available in the WP7 Final Report.

This section summarises the major findings of WP7 in relation to the work of CEs to improve QC for RD patients. The evidence supporting these findings is drawn from the case study, interviews and expert roundtables conducted. The section is divided into three sub-sections dealing with 1) general findings about CEs efforts to deliver improved QC; 2) specific activities in which CEs are demonstrably improving QC; and 3) emerging challenges and existing barriers to further extending the capacity of CEs to deliver improved QC.

3.1. Centres of Expertise and Quality of Care for Rare Disease patients

3.1.1. CEs and the development of Quality of Care

Centres of Expertise are heterogeneous in their organisation and their resourcing. Pathways to improved QC for RD patients in CEs are therefore also likely to be heterogeneous to some extent.

From a Quality of Care perspective, a logical progression is evident in CEs working for RD. **Advances in QC appear to unfold in a step-wise manner**, broadening (number of QC dimensions) and deepening (extent of QC dimensions) over time.

CE practice improvements driving the enhancement of QC for RD patients are impacting most to enable Access, enhance Effectiveness and develop Patient-Centred Care. Ensuring patient Safety is another primary QC concern of CEs, and one that is transversal to these other dimensions.

CEs are also increasingly improving practices that can drive the enhancement of QC for rare disease in relation to **ensuring Continuity** of care, particularly in the transition from childhood to adulthood. Efforts continue across a range of activities to improve the **Timeliness** of care.

As a way of organising work for RD patients, CEs can be innovative in the pursuit of the objective of improving QC. In particular, CEs integrate multiple dynamics and drivers of change in pursuit of this objective. There is also some evidence that CEs can be responsive to shortfalls in QC identified by other stakeholders.

The evidence also suggests that as QC deepens, particularly with regards to institutionalising continuity and patient-centred care, CEs are able to be more inclusive and sensitive to patients' and families' needs.

3.1.2. CE capabilities and activities driving improved Quality of Care

CEs' capacities to drive improvements in QC depend on the activities in which they are currently engaged.

- CEs are engaged in a **full spectrum of clinical activities** including testing, diagnosis, treatment and care. The extent of this engagement is uneven in some cases, partly due to historical arrangements and uncertainty about resources.
- CEs are **characterised by a multi-disciplinary capability** and coordinate effectively between disciplines and medical specializations, particularly through regular meetings, informal interactions, intermediary support by case managers or nursing specialists, and use of communications/information systems. Physical proximity of specialists' everyday workplaces contributes to enhanced opportunities for both formal and informal interactions.
- CEs are **conducting basic, clinical and translational research**. Many are involved in research collaborations, consortia and clinical trials. Some CEs are conducting or coordinating these types of multi-partner activities.
- Many CEs are increasingly **developing activities in terms of social services and social care**. These CEs cite an identifiable need to do more in this area and express a willingness to do so.

Many CEs exploit their capacity to share knowledge between medical disciplines effectively. The capacity of many CEs to integrate disciplinary specialists within a CE-based case management framework brings direct benefits to QC, including reducing time to diagnosis, opening up alternate trouble-shooting perspectives and problem solutions, and increasing patient safety.

The multiple and diverse character of CE activities appears to contribute to QC benefits. As an organisational innovation, **CEs integrate, and facilitate connections and synergies between, a variety of different types of activity**. This promotes the circulation of tacit and new knowledge that may otherwise be more difficult to transfer and apply where it is needed.

- CEs can be the vehicle for important knowledge spill-overs between clinical practice, clinical research, basic research and clinical trials.
- Knowledge spill-overs are also created when CE professionals inform other parts of the medical community about a RD, or train families or other support groups in care techniques.
- Quality of Care is enhanced at the clinical interface when translation practices lead to improvements in processes or products available to RD patients.

From a professional perspective, **work motivation and satisfaction among CE personnel are profoundly linked to deepening and extending the quality of care** provided to RD patients. CE Directors and clinicians have a strong belief in the effectiveness of patient-centred care for RD patients.

- CE Directors and clinical staff report valuing very highly the capacity to provide a 'holistic' experience of patient-centred care.
- CE Directors and clinical staff value very highly the capacity to provide continuity of individual patient care – preferably over the life-course.

- CEs' self-assessments of the QC benefits of holistic patient-centred care were supported by the linked Patient Organisations consulted.

CEs acquire significant **benefits from well-developed networks**. In the field of RD, networks are vital for acquiring capabilities and accessing resources that are relatively scarce. Networks are also vital for sharing effective practices and adopting common standards and protocols across a range of activities.

- A number of different CE network dimensions are identifiable including medical care/specialist networks; research networks; networks between CEs; participating in training networks; and linking to social services and civil society organisations, including Patient Organisations. Some linkages with primary care also exist.
- CEs acquire specific competences through their network activities including participation in clinical trials, mobilizing extended multi-disciplinary and medical specialization resources, developing shared clinical guidelines or protocols, collaborating to provide continuity of care, and various modes of liaison and interaction with Patient Organisations.

CEs linking and networking with different types of actors creates **transversal relationships across primary, specialised clinical services and social care**. This can lead to benefits for Quality of Care, including more clearly defined RD patient pathways, and the integrated management of care (patient-centredness).

Improvement in QC for RD patients is linked in multiple ways to flows of knowledge and information that can be inclusive of a wide range of professional and social stakeholders. **CEs are playing increasingly important roles as information hubs and knowledge brokers**, supporting the efforts of Patient Organisations to make RD more visible and to provide information resources to families, GPs and medical specialists.

3.2. Future challenges and opportunities to improve Quality of Care for Rare Disease patients in Centres of Expertise

Adequate resourcing of CEs remains a challenge. Slowdown or stagnation in the effort to improve Quality of Care for RD patients is possible without appropriate funding and other resources required to maintain and, ideally, to expand CE activities.

- Currently, some CEs describe operating in 'survival mode' due to funding problems linked to recent financial crises that impacted MS.

Recommendation 1. Attention must continue to be paid to ensuring resource sustainability, as an essential requirement for the maintenance and development of CE activities driving improvements in Quality of Care for RD patients.

Human resources are a defining challenge for CEs wanting to improve QC for RD, in two broad senses. First, in determining which services can be provided to RD patients through the CE. Second, in determining to what extent efforts to improve service delivery can drive improvements in related dimensions of QC.

A number of specific human resource challenges for CEs for RD can be identified. Several of these challenges are quite generic, but some also derive significantly from the specific characteristics of RD healthcare. These challenges include:

- developing and sustaining key clinical personnel
- transferring knowledge to the next generation of specialists
- the availability of specialised support staff (nursing, social work, IT, etc.)
- training adult specialists
- accessing state-of-the-art training to up-skill current staff, including in relation to emerging technologies.

Recommendation 2. Designated CEs should include, or include access to, a level of support staffing sufficient to cover three essential functions: case management; patient and family interaction; liaison and coordination with social care providers, social services, Patient Organisations and other stakeholders.

The availability of key support staff is essential for driving improvements in QC. All CE Directors emphasised in the strongest terms **the vital importance of specialised nursing staff**. Specialised nurses are valued for their clinical expertise, their contribution to safe and comfortable patient interactions, their coordination and case-managing capabilities, and their skills in liaison and informal interactions with a range of stakeholders. These capabilities deliver tangible and intangible benefits to CEs. In particular, the case manager role was singled out by several CE Directors. Where specialised nurses are working in CEs, they are integral to explicit and implicit efforts to improve Quality of Care in all dimensions.

Recommendation 3. Designated CEs should include a dedicated specialist nurse or access to sufficient specialised nursing capabilities.

A major emerging challenge to the patient-centred care dimension of QC is the **transition from paediatric context of relatively holistic care to uncertain contexts for adult care**. CEs are using a number of strategies to address this challenge, including:

- Training adult specialists
- Integrating specialist from future adult contexts into current CE case management processes
- Provisionally extending the eligible age for remaining in paediatric care.

Recommendation 4. CEs and health authorities develop a targeted, coordinated and multi-faceted strategy for improving continuity of QC standards in the transition from childhood to adulthood for RD patients.

Historically, improvement in Quality of Care for Rare Disease patients has been very substantially driven by Patient Organisations. This continues to be the case. However, Patient Organisation representatives are generally positive and supportive regarding the potential of CEs for organising service provision for Rare Diseases.

Patient Organisations value very highly the professionalization of CEs. This refers, first, to the institutionalisation of **processes of self-governance of the CE** that contribute to improvements in strategic and operational planning. Second, processes to communicate openly regarding operation and strategic issues are considered highly desirable.

Patient Organisations were able to describe tangible QC improvements linked to planning and cooperation involving themselves and CEs. A level of frustration exists where an absence of **clearly designated and mutually understood pathways** restricts more extensive development of these activities.

Recommendation 5. Consideration be given to assessing the advantages and disadvantages associated with formalizing an inclusive, participative governance process for CEs, such as an Advisory Board, to include Patient Organisations and other relevant stakeholders such as social service providers, health authorities, research managers, etc. (See also 3.3.2 The European dimension.)

CEs are not currently exploiting the potential of telemedicine to transform activities that could improve QC. **CEs identified several areas in which telemedicine innovation would be beneficial** for their operations and for improving the Effectiveness and Patient Safety dimensions of QC in particular.

- Building pathways from primary care by connecting general practitioners/patients with CE specialists.
- Facilitating routine consultations for remote patients of CEs.
- Sharing patient information and clinical research data within regional, national, European and global networks.

Recommendation 6. A study be undertaken of the actual use of telemedicine in CEs for Rare Disease, with a view to understanding current best practices.

Recommendation 7. An assessment be made of the capacities of CEs, including in terms of IT equipment and specialist skills, to incorporate an increased telemedicine dimension into their current activities.

CEs face an important immediate challenge to **make optimal use of technical change relevant to RD healthcare**. In particular the emergence of next generation sequencing (NGS) technologies and a range of genomics specializations offers potentially significant benefits for CEs. Indeed, genomics/NGS technology is central to many CE futures and is already transforming practices.

From a QC perspective, NGS offers the important potential to improve both Effectiveness and Efficiency.

- Identify new conditions and reduce the time to diagnosis for many known conditions.
- Enhance the sharing and comparing of case data.

These opportunities also bring challenges. The division of labour between clinicians and laboratory testing facilities is changing, with the emergent importance of bioinformatics expertise. Training and/or informing staff in relation to new tests, forms of data and the writing and understanding of interpretations of test results, for example, will increase the

benefits derived from these new technologies by increasing the absorptive capacity of the current workforce. (There is also an education/formation dimension to this challenge.)

What is vitally important is that CEs have access to **cost effective ways** to take advantage of the possibilities of new technologies.

Recommendation 8. Investment in new genetic testing and analysis technologies be coordinated such that the maximum number of CEs can access state-of-the-art facilities, but without unnecessary duplication of investments across MS or their regions.

Recommendation 9. Assessment be made of the demand for training and/or information within CEs regarding relevant aspects of genomic testing and data potential for RD.

There is an increased **expectation among stakeholders that CEs play a greater role as an information hub**. Many CEs already play an important role in the diffusion of information and awareness-raising regarding RDs to the general population, medical specialists, general practitioners, social services, etc. CEs with strong networking capabilities also increasingly function as knowledge brokers between multiple stakeholders and organisations. However, the extent to which this occurs varies between CEs.

Recommendation 10. A specific study could assess the expectations CEs face in terms of providing information. This would include assessing demand for different types of information, modes of information diffusion and access, and the profiles of user groups. Current information-sharing and knowledge-brokering activities being performed by CEs could also be documented, with a view to identifying effective models.

An opportunity exists for CEs to be part of an innovative multi-stakeholder model linking medical and social services that can provide significant Quality of Care benefits. However, in practice, developments remain uneven. A pressing challenge remains in relation to the **organisation and coordination of social care and other social services for RD patients and families**. Various models exist, apparently depending significantly on the availability of (a) social worker(s), their affiliation and their role. Patient Organisations often provide necessary assistance to bridge gaps, including utilising volunteer labour. From the QC perspective, a more sustainable system is required to address this challenge, which is connected to both the human resource/support staff and management/governance challenges identified.

Recommendation 11. Designated CEs should include, or include access to, a level of support staffing (e.g. trained social worker) sufficient to cover liaison and linked activities involving social care providers, social services, Patient Organisations and other external stakeholders, coordinated through CE management and governance processes.

The diverse activities of CEs and the complex professional and knowledge dynamics that drive change in these activities present increasing **management challenges**. Three pressing challenges are:

- The management and coordination of multiple network relations and participations, including those associated with the provision of social care and social services;

- Management of increasingly diverse Rare Diseases within a CE due to processes of grouping of diseases;
- CE governance, strategy and planning.

Recommendation 12. The specific management and governance challenges faced by Centres of Expertise be more fully considered in future policy and planning.

3.3. CEs, Quality of Care and the policy dimension

3.3.1. CEs, Quality of Care and the work of the Expert Group

The pragmatic activities and key organisational features of CEs appear closely aligned with the strategic vision for Rare Disease CEs contained in the EUCERD Recommendations.

CEs in which Quality of Care for RD patients is relatively highly developed tend to be those in which implementation of activities and organisational features delineated in the EUCERD Recommendations are most advanced.

CEs operating in a context where National Plans are established (and in some cases already being implemented) are likely to be relatively well-advanced in Quality of Care terms.

A relative lack of awareness of the EUCERD Recommendations exists within RD practitioner circles, including CEs. The apparent alignment between the Recommendations and the ‘reality on the ground’ in CEs appears due to the sound design and continuing relevance of the Recommendations, rather than being a reflection of explicit recognition or deliberate implementation of the Recommendations on the part of CEs.

Recommendation 13. Consideration be given to methods to further disseminate the EUCERD Recommendations, and information regarding the work of the Expert Group more broadly, with the aim of expanding the scope of awareness within the RD community in MS.

CE Directors understand and largely support the rationale for gradual **grouping of diseases** to deliver critical mass and avoid unnecessary duplication of investments. However, they argue that an important challenge is the shift from medical questions to management issues associated with grouping diseases and the associated increase in the diversity of disease expertise and approaches to diagnosis, treatment and care.

CE Directors are highly critical of any CE designation, including ‘self-designation’, made on the basis of a very small number of cases of a particular rare disease – but where overall expertise in relation to the diagnosis, treatment and care of rare disease patients is lacking. A **strong preference for ‘balanced’ designation criteria** was expressed. Concern exists regarding criteria that establish thresholds for designation that may be out of reach for developing CEs. From a QC perspective, this can be interpreted as a preference to delay focus on Efficiency where CEs have not yet fully consolidated their activities and organisation or reached their maximum capacity in terms of services and cases.

3.3.2 The European dimension

The **degree of consolidation** of systems of practices, organizations and networks in particular RD contexts across MS is highly heterogeneous.

- From a QC perspective, the kinds of variables, indicators and criteria that shape processes of inclusion in ERNs are thus very important – **a significant design challenge exists to capture the value embedded in existing CEs and their networks** at an appropriate level.
- ERNs for Rare Disease represent a double opportunity: linking consolidated expert systems AND linking to advance the process of consolidation.

ERNs that can capture the capabilities institutionalized in CEs will have a **starting advantage and the potential to improve QC with greater resource efficiency**.

- The localized mobilizing of resources and building of networks that existing CEs have been elaborating is time (and resource) intensive. It would be very costly to replicate this work.

From a QC perspective, **ERNs represent a potential opportunity to improve Efficiency**, but context remains critical and stakeholders are wary of a ‘one size fits all’ approach.

ERN membership or affiliation processes may be enhanced by providing an opportunity for applicant entities to demonstrate:

- Quality of Care capabilities and their future QC strategies; and
- their networking competences (clinical, specialist, research, social) and dimensions (local, regional, national, cross-border) in the field of Rare Diseases.

These capabilities and competences could be assessed relative to the type of ERN membership or affiliation under consideration.

CEs are networked and, increasingly, interactive organisations. There appears to be a high potential for formal networks of CEs to define new, effective spaces and flows of knowledge, expertise and care for RD patients in Europe. This opens up the **potential for enhanced processes of shared learning, knowledge diffusion and adopting improved practices**. Quality of Care offers a possible framework for driving shared improvements in healthcare for RD patients – whilst retaining the appropriate flexibility to accommodate the cultural diversity that exists between, and within, MS. A shared QC approach could provide a framework for sharing and learning about practices and processes that can drive improvements in specific QC dimensions. To this extent, **QC represents a potentially effective and sensitive model for the adoption of good practices for RD healthcare in Europe**.

Recommendation 14. Consideration be given to harmonizing concepts of Quality of Care and relevant QC Dimensions across CEs for RD in different MS, to facilitate joint understanding. This could be a foundational step toward developing a shared QC framework to underpin future knowledge sharing and good practice diffusion.

Recommendation 15. Consideration be given to developing appropriate governance processes for CEs with regard to managing the sharing of knowledge, the coordination of patient care and the improving of QC across MS borders

Summary of WP7 Recommendations

Recommendation 1. Attention must continue to be paid to ensuring resource sustainability, as an essential requirement for the maintenance and development of CE activities driving improvements in Quality of Care for RD patients.

Recommendation 2. Designated CEs should include, or include access to, a level of support staffing sufficient to cover three essential functions: case management; patient and family reception; liaison and coordination with social care providers, social services, Patient Organisations and other stakeholders.

Recommendation 3. Designated CEs should include a dedicated specialist nurse or access to sufficient specialised nursing capabilities.

Recommendation 4. CEs and health authorities develop a targeted, coordinated and multi-faceted strategy for improving continuity of QC standards in the transition from childhood to adulthood for RD patients.

Recommendation 5. Consideration be given to assessing the advantages and disadvantages associated with formalizing an inclusive, participative governance process for CEs, such as an Advisory Board, to include Patient Organisations and other relevant stakeholders such as social service providers, health authorities, research managers, etc.

Recommendation 6. A study be undertaken of the actual use of telemedicine in CEs for Rare Disease, with a view to understanding current best practices.

Recommendation 7. An assessment be made of the capacities of CEs, including in terms of IT equipment and specialist skills, to incorporate an increased telemedicine dimension into their current activities.

Recommendation 8. Investment in new genetic testing and analysis technologies be coordinated such that the maximum number of CEs can access state-of-the-art facilities, but without unnecessary duplication of investments across MS or their regions.

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Recommendation 12. The specific management and governance challenges faced by Centres of Expertise be more fully considered in future policy and planning.

Recommendation 13. Consideration be given to methods to further disseminate the EUCERD Recommendations, and information regarding the work of the Expert Group more broadly, with the aim of expanding the scope of awareness within the RD community in MS.

Recommendation 14. Consideration be given to harmonizing concepts of Quality of Care and relevant QC Dimensions across CEs for RD in different MS, to facilitate joint understanding. This could be a foundational step toward developing a shared QC framework to underpin future knowledge sharing and good practice diffusion.

Recommendation 15. Consideration be given to developing appropriate governance processes for CEs with regard to managing the sharing of knowledge, the coordination of patient care and the improving of QC across MS borders.