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WORK PACKAGE 7: CENTRES OF EXPERTISE (CEs) / QUALITY OF CARE (QoC)

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Keyword List:	Rare Diseases, Centers of Expertise, European Reference Networks, Quality of Care
Abstract:	<p>The aims of the WP were to identify actions which can improve access to higher-quality healthcare in rare diseases (RD). The team has explored various initiatives across the EU Member States (MS) which addressed the Quality of Care (QoC) for RD, with the aim of identifying and sharing good practices in healthcare. Special emphasis was placed on activities relating to Centres of Expertise (CEs). Furthermore, the WP looked at the factors influencing policy decisions pertaining to QoC for RD, and assessed how healthcare systems organize themselves to accommodate RD policies and patients.</p> <p>The main findings of the work carried out refer to the variation between regional and national health systems and different RDs, which are shaping differences in the organization and activities of CEs and their capacity to drive improvements in QoC for RD patients. CEs are having a major impact on several dimensions of QoC for RD patients by enabling access, enhancing the effectiveness of clinical services, developing a committed culture of patient centered care and creating a context of elevated patient safety.</p> <p>However, many issues remain unresolved: adequate support staffing; unevenness of access to timely diagnosis; capturing QoC benefits from Genomics/NGS technologies; use of telemedicine t and CE best practices sharing, among others.</p> <p>The activities and organization of CEs and the strategic vision described in the EUCERD Recommendations are closely aligned. . From a QoC 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 anda high demand for participation in ERNs for RD can be anticipated.</p>

In conclusion, the achievements of CEs for RD in driving improvements in QoC for RD patients are considerable. CEs are a form of organizing diagnosis, treatment and care for RD patients that can provide safe, holistic patient-centered care and be responsive to stakeholder needs. However, as Patient Organization participants point out, timely diagnosis and making expertise even more available are problems that still need to be more solvable.

CEs for RD also face a range of emerging challenges. The availability of resources to meet these challenges is constricted in many settings. It is in this sense that we conclude that CEs for RD have reached something of a crossroads. CEs, as the key health system innovation for delivering healthcare for RDs, should be expected to work to continuously improve QoC. To do this they require continuing support and investment. It is this new wave of investment, including potential organization into ERNS and an associated increase in capacity for interaction and learning, which can drive CEs toward more efficient delivery of health services for RD patients.

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1. Introduction

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 (QoC) 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 QoC improvements. It sought to identify good practices, particularly those systems of practices impacting positively on QoC 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.

2. Presentation of results

2.1. Work package 7: Task, objectives & approach

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

- Opportunities for improving QoC 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 QoC for RD patients;
- Identifying guidelines, strategies and good practices for delivering QoC 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/QoC nexus.

The WP7 approach was built around **three strategies for accessing expert opinion** regarding actions of CEs that lead to improvements in QoC 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 QoC 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?

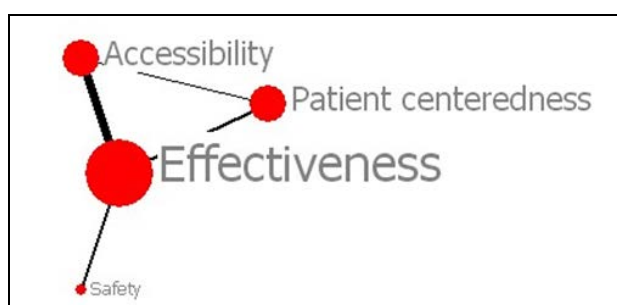
2.2. A framework for understanding Centre of Expertise led improvements in Quality of Care for Rare Diseases

A framework was developed for situating the professional practices making up the work of CEs for RD in relation to improvements in QoC. 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 QoC for RD patients was settled on: **identifiable improvements in patient services and/or patient welfare**.

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



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 QoC**.

The EUCERD Recommendations were then analysed from a QoC perspective, as they provide guidelines for Centres of Expertise for Rare Diseases in four main areas:

- Mission and Scope
- Core competencies
- Role in spreading information and education
- Role in research

The Recommendations are designed to shape the institutionalization of CEs and as such provide *strategic markers for CEs as a health system innovation* focused on rare disease patient care.

The EUCERD Recommendations on Centres of Expertise for Rare Disease include two different references to Quality of Care among the expected capacities of CEs: 1) the production of quality of care indicators; and 2) the introduction and management of externally validated quality assurance processes. The explicit intersection of the EUCERD Recommendations and Quality of Care is thus in terms the capacity to monitor quality of care through information systems. This focuses attention on QoC outcomes.

To understand how the EUCERD Recommendations overall focus on QoC dimensions we analysed each Recommendation individually from a QoC perspective. Our first step was to ask members of the WP7 team to label each of the EUCERD Recommendations according to the Dimension of QoC it appeared to them to be most associated with, using the set of definitions of dimensions shown in Table 1. We also did a keyword analysis, tagging each individual Recommendation according to the relevant dimensions of QoC.

To establish relations between the dominant dimensions of QoC within the Recommendations we then analyzed the pattern of co-presence of these dimensions with the Recommendations focused on Mission and Scope, Core Competencies and Role in Research.

Table 1.

Recommendation	Accessibility	Effectiveness	Patient centeredness	Safety
1. Mission and scope of CEs	3	8	7	3
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		1
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			
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.->25		1	1	
05. CEs contribute to building healthcare pathways from primary care.	1			
06. CEs have links with specialised laboratories and other facilities.	1	1		
07. CEs collaborate with patient organisations to bring in the patients' perspective.			1	
08. CEs contribute to the elaboration of good practice guidelines and to their dissemination.		1		1
09. CEs provide education and training to healthcare professionals from all disciplines, including paramedical specialists and non-healthcare professionals (such as school teachers, personal/homecare facilitators) whenever possible.		1	1	1
10. CEs contribute to and provide accessible information adapted to the specific needs of patients and their families, of health and social professionals, in collaboration with patient organisations and with Orphanet.			1	
11. CEs respond to the needs of patients from different cultures and ethnic groups (i.e. have cultural sensitivity).			1	
12. According to national/international ethical and legal frameworks, CEs should ensure respect of non-discrimination and non-stigmatisation of RD patients across Europe, within their sphere of competencies.			1	
13. CEs contribute to research, to improve the understanding of the disease and to optimise diagnosis, care and treatment, including the clinical evaluation of long-term effects of new treatments.		1		
15. CEs liaise with other CEs at National and European level when relevant.		1		
16. A national directory of formally designated CEs is compiled and made publicly available, including on the Orphanet portal.		1		
2. Criteria for designation of CEs	6	13	5	1
17. Capacity to produce and adhere to good practice guidelines for diagnosis and care. -> 8.		1		1
18. Quality management in place to assure quality of care, including National and European legal provisions, and participation in internal and external quality schemes when applicable.		1		
19. Capacity to propose quality of care indicators in their area and implement outcome measures including patient satisfaction.		1	1	
20. High level of expertise and experience documented, for instance, by the annual volume of referrals and second opinions, and through peer-reviewed publications, grants, positions, teaching and training activities.	1	1		
21. Appropriate capacity to manage RD patients and provide expert advice.		1		
22. Contribution to state-of-the-art research.		1		
23. Capacity to participate in data collection for clinical research and public health purposes.		1		
24. Capacity to participate in clinical trials, if applicable.		1		
25. Demonstration of a multi-disciplinary approach, when appropriate, integrating medical, paramedical, psychological and social needs (e.g. RD board).->4.		1	1	
26. Organisation of collaborations to assure the continuity of care between childhood, adolescence and adulthood, if relevant.	1		1	
27. Organisation of collaborations to assure the continuity of care between all stages of the disease.	1		1	
28. Links and collaboration with other CE at national, European and international level.		1		
29. Links and collaboration with patient organisations where they exist.			1	
30. Appropriate arrangements for referrals within individual Member States and from/to other EU countries if applicable.	1	1		
31. Appropriate arrangements to improve the delivery of care and especially to shorten the time taken to reach a diagnosis.	1	1		
32. Consideration of E-Health solutions (e.g. shared case management systems, expert systems for tele-expertise and shared repository of cases).	1	1		
4. European Dim of CEs	4	3	1	
41. MS with established CEs share their experience and quality indicators with other MS and coordinate their efforts to identify CEs for all RD patients at EU level.	1	1		
42. Networking of CEs is a key element of their contribution to patient diagnosis and care, to ensure that expertise travels rather than patients themselves when appropriate; exchange of data, biological samples, radiological images, other diagnostic materials, and e-tools for tele-expertise are promoted.	1	1		
43. Cross-border healthcare is organised, where appropriate, with designated CEs in neighbouring or other countries, where patients or biological samples can be referred to.	1	1		
44. Member States should provide adequate information to professionals, citizens and patients	1		1	

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

Many avenues to improved QoC 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 QoC 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 QoC; 2) specific activities in which CEs are demonstrably improving QoC; and 3) emerging challenges and existing barriers to further extending the capacity of CEs to deliver improved QoC.

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

CEs and the development of Quality of Care

Centres of Expertise are heterogeneous in their organisation and their resourcing. Pathways to improved QoC 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 QoC appear to unfold in a step-wise manner**, broadening (number of QoC dimensions) and deepening (extent of QoC dimensions) over time.

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

CEs are also increasingly improving practices that can drive the enhancement of QoC 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 QoC. 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 QoC identified by other stakeholders.

The evidence also suggests that as QoC 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.

CE capabilities and activities driving improved Quality of Care

CEs' capacities to drive improvements in QoC 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 QoC, 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 QoC 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 QoC 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 QoC 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.

2.3.2. 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 QoC 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 QoC.

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 QoC. 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 QoC 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 QoC 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 QoC 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 QoC. **CEs identified several areas in which telemedicine innovation would be beneficial** for their operations and for improving the Effectiveness and Patient Safety dimensions of QoC 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 QoC 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 QoC 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.

2.3.3. CEs, Quality of Care and the policy dimension

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 QoC 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.

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 QoC 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 QoC 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 QoC 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 QoC 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 QoC approach could provide a framework for sharing and learning about practices and processes that can drive improvements in specific QoC dimensions. To this extent, **QoC 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 QoC Dimensions across CEs for RD in different MS, to facilitate joint understanding. This could be a foundational step toward developing a shared QoC 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 QoC across MS borders

3. Critical analysis of results

The results obtained in WP7 are focussed on the relationship between the activities conducted by CEs for RD and improvements in QoC for RD patients. The mediating variable in this relationship is the form of organisation of the CE itself. The way CEs use their resources and work to improve the routines and processes that structure the performance of key activities varies from CE to CE. In addition, the national health system in which each CE is embedded and the particular disease or group of diseases that are the focus of CE shape these activities and outcomes in terms of improving QoC.

The results were produced through a focus on process, with very limited attention paid to structure or outcome. This means that the results are not useful for trying to understand what kind of 'outputs' should be expected from CEs in relation to the types and volumes of financial and other resources that are input to the system.

The approach taken in WP7 was to use a stratified sample of CEs to conduct relatively detailed case studies based on qualitative data in the form of expert opinion. This expert opinion was sourced from both within the participating CEs and from linked Patient Organisations and national health authorities. The results paint a picture of the types of activities and strategic foci of CEs working to improve QoC for RD. A clear understanding of existing and emerging challenges was also developed.

The approach taken should not be interpreted as trying to capture an all-encompassing or definitive picture of CEs' efforts to develop QoC. The challenges identified should not be considered a definitive list. Rather, it should be kept in mind that the vast majority of Rare Diseases and many national health systems that did not feature in WP7 at all. This means that the results should be treated with the appropriate caution with regards to their generalizability to other diseases or MS contexts.

Nonetheless, the Recommendations produced by WP7 can be regarded as robust and validated across multiple of the participating CEs. It is likely that the substantive content of these Recommendations would not be surprising to anyone working in the RD field in most MS contexts. With relatively few exceptions it is also likely that the Recommendations would be applicable to the current state-of-play in most CEs across the vast majority of MS.

The major way in which disparities between disease cases and national health systems were overcome in the production of our results was by integrating the activities of CEs into a framework based on dimensions of QoC. Rather than directly comparing what individual CEs were doing, which might involve comparing very different levels of investment in material and human resources, we collated what each CE was doing to try and improve QoC around particular tasks or activities that define the mission of CEs. By integrating the EUCERD Recommendations (as a blueprint for activities and objectives) with dimensions of QoC we constructed a framework that enabled us to organise the information flowing from experts in very different contexts into coherent themes. A range of experiences and challenges could be brought together and certain critical components of CEs efforts to improve quality of care could be identified. A series of emerging challenges could also be defined.

One observation flowing from WP7 is that Quality of Care remains an under-explored framework for sharing experience, learning and methods between MS, that has a relatively low risk of failing due to differences in cultural interpretations or political economic frameworks.

4. Conclusions

This section summarizes the major findings of WP7 regarding CEs and to the transition from CEs to ERNs in relation to the WP7 Objectives. Cooperation activities are also highlighted. Taking into account the main findings and these Conclusions, a set of Recommendations were developed, which have been included in the Results section and are gathered here again for easy browsing:

List 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 QoC 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.

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.

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.

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.

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 QoC Dimensions across CEs for RD in different MS, to facilitate joint understanding. This could be a foundational step toward developing a shared QoC 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 QoC across MS borders.*

The work of WP7 highlights the important role and contribution of CEs in improving Quality of Care for RD patients in different social and cultural context across MS. A logical progression is evident in the step-wise manner in which CEs are improving Quality of Care: starting from enabling Access; to progressively enhancing Effectiveness; to developing Patient-centred Care; and introducing mechanisms to ensure Continuity of Care.

CEs are heterogeneous and distributed in the way they are organised spatially and institutionally. Many CEs span a number of units, organisations, disciplines and their configuration can either be ‘bricks and mortar’ or ‘virtual’. A flexibility of the CE form of organising is, in this way, very beneficial to the development of QoC, as the core functions and roles that CEs play in developing QoC can be institutionalised across the variety of administrative and organisational settings characterising MS health systems.

The evidence suggests that, as a way of organising work for RD patients, CEs can be innovative and responsive to the voices and needs of concerned groups such as Patient Organisations and other stakeholders. As a vehicle for patient-centred care CEs are able to be inclusive and sensitive to patients’ and families’ needs. However, the development of broader interdisciplinarity and the better integrations of clinical and social services remains a challenge identified by many CE Directors.

There is some compelling evidence of the way CEs can bring closer together the frontier of scientific research and the clinical evidence base. The involvement of CE staff in Framework program research consortia, clinical trials, the spinning out of new technologies and the translation of research from lab to clinic shows that CEs are creating vital knowledge spillovers and helping to connect research and commercial practices. At the same time CEs are developing aspects of their research with the support of social stakeholders, particularly Patient Organisations.

The scarcity of RD expertise means that CEs need to become networked and interactive organisations to the greatest extent possible. Sharing knowledge and ideas with clinical colleagues in other Centres, regions and countries is a typical part of CE work. Collaborating with Patient Organisations and social service providers is also acknowledged as vital to CE efforts to improve QoC. More can be done to develop these latter networks. Overall, 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.

The networking capabilities developed by CEs can be further expanded by participation in, or some kind of affiliation to, European Reference Networks (ERNs). Moving to the ERN dimension can potentially provide an extension of the step-wise progression in delivering QoC. This could occur through the trans-national development of standards and protocols, monitoring techniques and processes of evaluation. These developments would likely lead to an Efficiency dividend for CEs in different MS as management and evaluation information systems started to provide greater insight into the conduct of activities – including on a comparative basis. In other words, ERNs will create opportunities for learning and sharing (resources, experiences) that can consolidate understandings of Quality of Care and the development of QoC dimensions for RD across MS

From the viewpoint of prospective ERNs, capturing the networking and other capabilities already institutionalized in consolidated CEs will provide a starting advantage in terms of established pathways for developing QoC. Consolidated CEs will provide ERNs with a base in high level QoC and, more importantly, the potential to expand capacity more rapidly and with greater resource efficiency in places and spaces where QoC needs to be improved. The localized mobilizing of resources and building of networks that CEs have

elaborated in their own contexts to date is time and resource intensive and would be costly to replicate by other organisations. ERN-wide learning about specific elements and relationships that are essential in the efforts to build multi-stakeholder partnerships that can drive improved QoC can be a potentially rich contribution that consolidated CEs can bring to ERNs. Networks that intersect in CEs include formal and informal professional disciplinary and disease-focused networks, family and social stakeholders, and data and technical communities of practice.

The important topic of the grouping of rare diseases is widely discussed by CE Directors and supporting institutions. CE Directors understand and largely support the rationale for grouping diseases. However, many argue that an important part of the challenge shifts from medical to management issues.

The related issue of CE designation criteria requires consistent policy attention. CE Directors are critical of 'self-designation' on the basis of some experience but not overall expertise. However, they also express strong desire for 'balanced' designation criteria and reveal concern that 'black and white' criteria that may be designed to promote a 'too early focus' on Efficiency. Whilst the maximization of outcomes from a given allocation of resources is recognized as a legitimate policy goal, concerns exist regarding the fragility of some hard-won Rare Disease eco-systems that need continued nurturing and development.

There is likely to be a strong demand for ERN membership or some other form of affiliation or access. This is broadly driven by the opportunities to expand capacities and access desired capabilities. More specific demand for ERNs will be driven by a) the possibility of increasing Effectiveness in the delivery of (cross-border) care for RD patients; and b) improving Access for proximate smaller countries with less consolidated CEs and/or networks. In this regard, progress in developing national CE designation processes and the precise definition of available modes of affiliation to ERNs is important.

From a Quality of Care perspective, ERNs for Rare Diseases represent a double opportunity: linking consolidated expert systems AND linking to advance processes of consolidation.

WP7 benefited from considerable cooperation and support from other WPs within the Joint Action. In particular, EURORDIS provided expert advice on interviewing Patient Organizations, including specific guidance on the design of the schedule used in these interviews. The support and advice of the coordinating team from the University of Newcastle was greatly appreciated throughout the work of WP7, as was the collegial atmosphere created by the participating partners who hosted a number well-organized, informative and very useful Workshops in which broad participation from attendees was encouraged.

WP7 was also fortunate to enjoy the cooperation of a number of CE Directors including from the Bulgaria, Czech Republic, Denmark, Finland, France, Germany, Italy, Lithuania, Spain, Poland and the UK, many of whom participated in a Workshop and contributed significantly to a series of topical Roundtables. Valuable support for this participation was provided by the Spanish Ministry of Health, Social Services and Equality. In addition, the participation of health officials from 16 Member States at the WP7 Final Workshop and Presentation of Results was an important opportunity for dissemination and discussion.

SUMMARY OF 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 QoC 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 QoC.
- 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 QoC benefits from these technologies is a key next challenge for CEs.
- Telemedicine retains untapped potential for improving QoC 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 QoC 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 QoC.
- 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 QoC 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, QoC appears to have potential as a framework for the context sensitive diffusion of good/best practices between regions and MS.