



EUCERD Joint Action WP7 Workshop

From Centres of Expertise for Rare Diseases
to European Reference Networks

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WP7 General Overview and Outcomes

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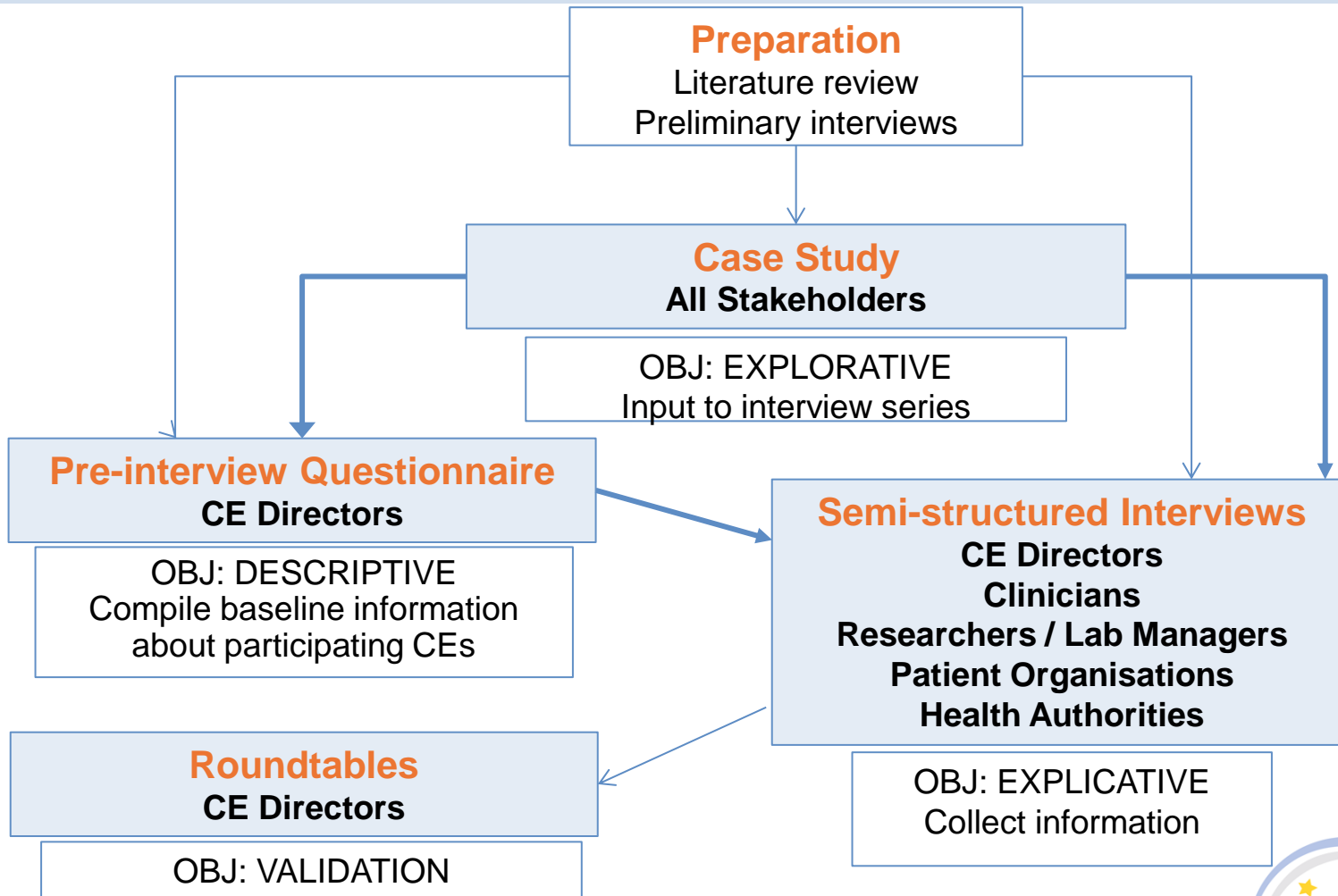
Aims & Objectives of WP7

- Understand how the activities of Centres of Expertise (CEs) are contributing to an improvement in Quality of Care (QoC) for Rare Disease (RD) patients
- Identify CEs' major dimensions of achievement in relation to QoC and the existing and emerging challenges
- Understand the potential of CEs, as health system innovation, to contribute to the future of RD care and policy

WP7 Contribution to the EJA

- Inform the EJA about the practice and knowledge dynamics of CEs engaged in improving QoC for RD patients
- Provide evidence regarding CEs as a health system innovation for addressing the challenges of working for RD in Europe
- Provide insight into the degree of alignment of top-down processes (conceptual vision, institutional strategy) and bottom-up activities (existing practices, pragmatic approach)
- Highlight the potential for shared learning to drive both *CE development* and *QoC improvement*, including across regional and national borders
- Identify emerging challenges for CE policy

WP7 Approach



Case Study Outcomes

- General: **built a knowledge base** regarding CEs for Rare Diseases
 - Core competences; Principal activities; Key stakeholders and relationships
- Methodological: directly informed the design and testing of the **interview series**

Four framing expectations:

- CEs are **complex settings** for improving practices
- **Practices are always contextualised** – different diseases and health systems will shape practice improvements in specific ways
- Management and coordination of CEs are **integral** to the development and diffusion of practices to improve QoC
- **Networks are essential** for building on core competences and extending capabilities to improve practices

CE Selection: National Profiles

Country	Size / No. of inhabitants#	NP/NS	Type of country
France	Large / 65,397,912	Adopted	A
Spain	Large / 46,196,277	Adopted	A
Germany	Large / 81,843,809	Not adopted	B
Italy	Large / 60,850,782	Not adopted	B
Poland	Large / 38,208,618	Not adopted	B
UK	Large / 62,989,550	Not adopted	B
Bulgaria	Small / 7,600,000	Adopted	C
Czech Republic	Small / 10,500,000	Adopted	C
Denmark	Small / 5,580,516	Not adopted	D
Finland	Small / 5,501,267	Not adopted	D
Lithuania	Small / 3,199,771	Not adopted	D

*Countries with 10 to 30 million inhabitants have been excluded

#European Union January 1, 2012 (http://europa.eu/about-eu/countries/index_en.htm)

These 11 countries from the EU-27 represent 395,179,750 million people, 78.5% of the total EU population (503 million).

CE Selection: Disease Profiles

Diseases	Estimated prevalence (/10,000) [Orphanet]
Rare Diseases ($\geq 1/10,000$)	
Single disease	
Cystic fibrosis	1.26
Monosomy 22q11	2.0
Group of diseases	
• <i>Inherited metabolic diseases</i>	
Congenital sucrase-isomaltase deficiency	2.0
Gaucher disease	0.2
• <i>Congenital rare anaemias</i>	
Sickle cell anaemia	1.5
Very Rare Diseases ($\leq 0.6/10,000$)	
Single disease	
Rett syndrome	0.4
Lowe (oculocerebrorenal) syndrome	0.02
Group of diseases	
• <i>Connective tissue diseases</i>	
Osteogenesis imperfecta	0.66
• <i>Immunodeficiencies</i>	
Common variable immunodeficiency	0.4
Severe combined immunodeficiency - ADA	0.02

CEs as health system innovation

- Centres of Expertise as key actors driving change
 - What are actually existing 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 organization being transformed; what factors are driving changes?
 - How is CE management & strategy evolving and influencing change?
- The comparative dimension: improving Quality of Care
 - Impact of country profile & profile of rare disease: importance of **context**
 - Goals of stakeholders: importance of **perspective** (professionals, families, health authorities, patient organisations, etc.)

The EUCERD Recommendations

- Strategic markers for CEs as health system innovation focused on RD patient care
- 4 main areas: Mission & scope of CEs; Designation criteria for CEs; Designation process; European dimension
- Mission & Scope
 - Patient Focus
 - Core competencies
 - Role in spreading information & education
 - Role in research
- Designation criteria
 - Leadership & credibility
 - Multidisciplinarity & inclusiveness
 - Capacity
 - Links & collaborations
 - *Mechanisms for measuring performance & for evaluation*
- European Dimension
 - Sharing experience & indicators
 - Cross-border considerations
 - Networking

Guidance on organisation and activities of CEs

A Quality of Care framework

- Widely diverging (cultural) understandings and applications of QoC between, and within, EU Member States (Vollaard et al. 2013)
 - limits the current usefulness of best practice methodologies;
 - not all dimensions of QoC can be assessed using best practice;
 - additional complementary approaches desirable
- European Joint Action on Quality of Care: toward a shared conception built around:
 - patient safety;
 - cooperation on practical issues;
 - information sharing;
 - learning

Quality of Care

- Quality of care is the kind of care which is expected to maximize an inclusive measure of patient welfare, after one has taken account of the balance of expected gains and losses that attend the process of care in all its parts (Donabedian 1980)
- Quality of care is the degree to which health services for individuals and populations increase the likelihood of desired health outcomes and are consistent with current professional knowledge (IOM 1990)
- Quality of care is the level of attainment of health systems' intrinsic goals for health improvement and responsiveness to legitimate expectations of the population (WHO 2000)

WP7 working definition: identified improvements in patient services and/or patient welfare

➡ How do CEs drive such improvements for RD patients?

Dimensions of Quality of Care

- System markers for the provision of health services:
 - **Accessibility** - Equity - Equitable
 - **Effectiveness** - Safety - Competence - Appropriateness
 - **Efficiency**
 - **Patient-centeredness** - Continuity - Timeliness
 - **Safety**
 - Acceptability Responsiveness Satisfaction Transparency +

Guidance on how practice improvements in Centres of Expertise impact on Quality of Care for Rare Disease patients

EUCERD Recs on CEs & QoC Dimensions

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 healthcare providers					
07. CEs collaborate with patient organisations to bring patient perspective.				2	



Improving Practices Example: Transition to adulthood

- ✓ CEs are innovating to manage patients' transition from childhood to adulthood
 - Clinicians working in adult medicine start attending sessions with paediatricians during adolescence
 - Adult specialists build familiarity inside CE before they move to adult clinic
 - Training of new adult RD specialists within the CE
 - Expansion of clinical team to include adult specialists
 - CE paediatricians attend adult clinic/hospital for first year out of CE
 - CEs collaborating with other hospital units to build adult patient unit
 - CEs retaining patients after they turn 18 years of age
 - Linking to policy processes, including National Plan development
 - Collaborating with social workers

*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...*

QoC Dimensions: CONTINUITY OF CARE; ACCESSIBILITY; SAFETY
EUCERD Recs 10, 13, 26, 27: CORE COMPETENCIES; TRAINING; PATIENT FOCUS

WP7 Outcomes 1: CE activities

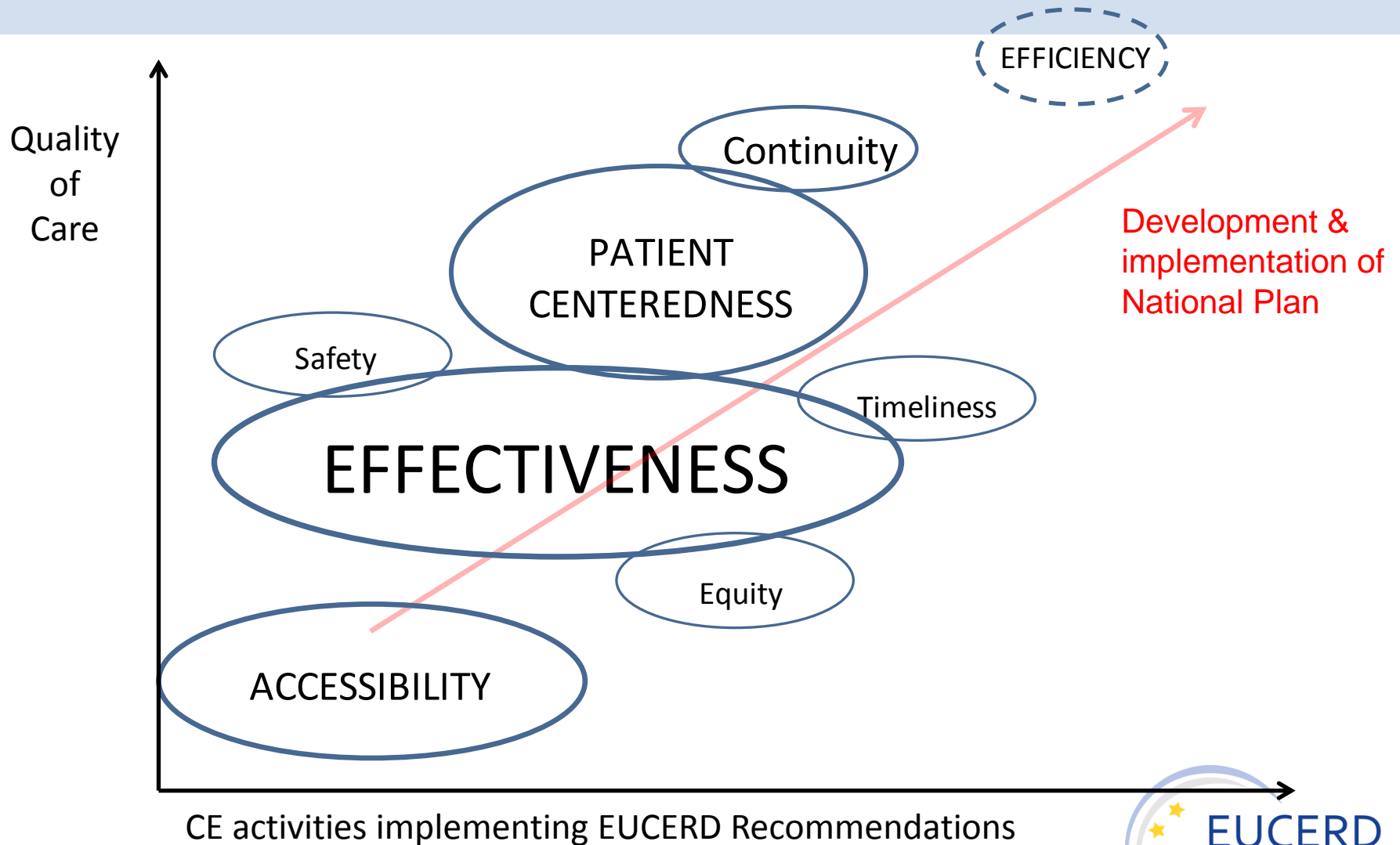
Centres of Expertise are:

- Engaging in a **full spectrum** of clinical activities
- Establishing and coordinating a **multidisciplinary** medical capability – strong focus on regular clinical (and management) meetings
- Conducting basic, clinical and translational **research**
- In some cases, developing broader multidisciplinary capabilities **extending into social services** and social care
- Developing **patient-centred** case management routines – strong focus on new patient circuits
- Building & benefiting from well-developed **networks**
- Performing both **bridging and integrating** functions linking diverse knowledge, actors and stakeholders
- Playing increasingly important roles as **information hubs and knowledge brokers**

WP7 Outcomes 2: Quality of Care

- The **degree of consolidation** of systems of practices, organizations and networks in RD contexts across MS is **highly heterogeneous**
- From a **Quality of Care perspective**, pragmatic CE activities and the EUCERD Recommendations are closely aligned
 - QoC advances along more dimensions in the context of national planning
- CE practice improvements are impacting most in dimensions of **accessibility, effectiveness** and **patient-centred care**
- Developing a **diversified networking capability** has QoC benefits
- CE Directors & Clinicians value very highly the capacity to provide **holistic and continuous patient-centred care** – drives motivation & work satisfaction
- Patient Orgs value very highly the ‘professionalization’ of CEs and retaining a strong focus on **access to timely diagnosis**
- QoC has potential as a framework for **cross-border** diffusion

WP7 Outcomes 3



Conclusions: Centres of Expertise

- From a **Quality of Care perspective** a logical progression is evident in CEs working for Rare Disease: from enabling Access, to enhancing Effectiveness, to developing Patient-centred and Continuity of Care, etc.
- **CEs are heterogeneous and distributed in their organisation** – spanning a number of units, organisations, disciplines, relatively more or less ‘virtual’
- The evidence suggests that, as a way of organising work for RD patients, **CEs can be innovative and responsive**
- CEs can bring **closer together the frontier** of scientific research, the clinical evidence base and the demands and objectives of social stakeholders
- As a vehicle for patient-centred care CEs are able to be **inclusive and sensitive** to patients’ and families’ needs
- **CEs are networked and 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

Conclusions: from CEs to ERNs

- Moving to the ERN dimension can potentially provide an extension of the step-wise progression in delivering QoC through an Efficiency dividend (standards, monitoring, evaluation)
- Can 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
- ERNs that can capture the capabilities institutionalized in consolidated CEs are likely to have a starting advantage and the potential to expand capacity more rapidly and with greater resource efficiency
 - The localized mobilizing of resources and building of networks that CEs have elaborated to date is time and resource intensive and would be costly to replicate

Conclusions: Moving on to ERNs

- CEs bring existing well-developed Networking capabilities:
 - Direct links & collaborations with a diverse range of stakeholders
 - Formal and informal professional disciplinary and disease-focused networks
 - Emerging data/technical communities of practice (NGS, bioinformatics)
 - Local, regional, national, international levels
- Grouping diseases
 - CE Directors understand & largely support the rationale for grouping diseases;
 - BUT argue an important part of the challenge shifts from medical to management issues
- Designation criteria
 - CE Directors are critical of 'self-designation' on the basis of some experience but not overall expertise
 - BUT express strong desire for 'balanced' designation criteria
 - Concern about 'black and white' criteria and a 'too early focus' (on Efficiency)
 - Recognition of the fragility of some hard-won Rare Disease eco-systems

Conclusions: Moving on to ERNS 2

- In terms of acquiring capabilities and capacities, there is high potential demand for ERN membership and access
- The need for ERNs here is driven by a) 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
 - Importance of national CE designation processes and available modes of affiliation to ERNs
- From a **QoC perspective**, need to consider carefully how move forward to develop the dimension of Efficiency as context remains critical and stakeholders are wary of a 'one size fits all' approach
- ERNs for Rare Disease represent a double opportunity: linking consolidated expert systems AND linking to advance processes of consolidation

Thank you

