



Workshop Report

on

Centres of Expertise for Rare Diseases

Improving Practices and Networks

Madrid, 31 March - 1 April 2014



Co-funded by
the Health Programme
of the European Union



Introduction

The workshop was organized by CIBERER, within the scope of Work Package 7 (Quality of Care) of the EUCERD Joint Action (EJA): Working for Rare Diseases. It was held at the Spanish Ministry of Health, Social Services and Equality in Madrid on 31st March- 1st April 2014. It was attended by over thirty experts in the field of rare disease, representing various stakeholders (see appendix 1).

Specific questions to be addressed

- a) Presentation of the EJA and specific objectives of Workpackage 7 (WP7)
- b) Presentations of preliminary WP7 results on Centers of Expertise (CEs) and Quality of Care (QoC)
- c) Discussion of the results among stakeholders
- d) Raising awareness of the Commission-supported initiatives on rare diseases (RD) in Europe (EU)

Agenda

Day 1

- 15:00-15:15 Welcome. Representative from the *Spanish Ministry of Health, Social Services and Equality*.
- 15:15-15:30 Brief introduction to the EUCERD Joint Action Project. Stephen Lynn, *University of Newcastle, UK*.
- 15:30-16:00 Overview of WP7. Francesc Palau, *CIBERER, Spain*.
- 16:00-16:45 WP7 Methodology & Preliminary Results. África Villanueva/Richard Woolley, *INGENIO, Spain*.
- 16:45-17:15 Coffee break
- 17:15-19:00 Open Discussion: Implementing EUCERD Recommendations for CEs.

Day 2

- 9:30-10:00 Presentation of the Centre of Expertise on Gaucher Disease – Lysosomal Storage Disorders. Pilar Giraldo, *Miguel Servet University Hospital, Zaragoza*.
- 10:00-11:00 Round table. Informal networks of external collaboration of CE's and European Networks of Reference.
- 11:00-11:20 Presentation of one participant CE. Hilary Longhurst, *Barts and the London NHS Trust, UK*.
- 11:20-11:35 Coffee break
- 11:35-11:55 Presentation of one participant CE. Pavel Drevinek, *Cystic Fibrosis Centre. Motol University Hospital - 2nd Medical School Charles University in Prague, Czech Republic*.
- 11:55-12:15 Presentation of one participant CE. Bruno Bembi, *Regional Coordinator Centre for Rare Diseases, Academic Hospital "Santa Maria della Misericordia" di Udine, Italy*.
- 12:15-13:15 Round table. Relevant challenges for CEs.
- 14:15-15:15 Round Table. The natural history of CEs. Where CEs come from?
- 15:15-15:45 Closing remarks. Francesc Palau, *CIBERER, Spain*

Brief Description of Each Presentation and Round Table

DAY 1

Presentation 1: *Brief introduction to the EJA Project* (Stephen Lynn)

In this presentation, Stephen Lynn, EJA Project Manager, introduced the EUCERD Joint Action project, covering all aspects: framework, aims, associated and collaborating partners and workpackages' description and specific objectives.

Presentation 2: *Overview of WP7* (Francesc Palau)

In this presentation, Francesc Palau, leader of WP7, summarized the main objective of workpackage 7, which is to identify:

- Actions which could improve access to higher-quality healthcare in rare diseases (RD), enhancing patients' quality of life.
- Initiatives across the EU Member States (MS) which aim to address the quality of care for rare diseases, with the objective of identifying and sharing good practices in healthcare.
- How healthcare systems organize themselves to accommodate RD policies and deliver good quality care to patients.

Dr. Palau emphasized that the work of WP7 also aims to support and advise the European Commission Expert Group on Rare Diseases by providing evidence and key insights regarding the actual designation and operation of CEs for RD across different national realities and disease settings.

A brief summary of the methodology used -based on ethnographic research, an online questionnaire and semi-structured interviews with expert RD stakeholders- was also presented.

Finally, outcomes to date and future activities were briefly outlined to introduce the following presentations made by representatives of the *Instituto de Gestión de la Innovación y del Conocimiento*, INGENIO (CSIC-UPV), an Institute subcontracted by CIBERER to develop part of the work.

Presentation 3: WP7 Methodology & Preliminary Results (Richard Woolley)

In this presentation, Richard Woolley, INGENIO's researcher collaborating with CIBERER, explained in detail the approach and methodology followed by the team and presented the work conducted to date (see full presentation in Annex 2):

- Objectives and outcomes of the ethnography carried out at the Reference Unit selected.
- Objectives, framework and outcomes of the interviews carried out at different CEs throughout the EU:
 - o Good practices identified and analyzed:
 - Laboratory testing
 - Regular clinical meetings
 - Patient management
 - Continuity of care
 - Networking
 - o Some preliminary conclusions about CEs
 - o Contribution to the work of the EJA

Round table / Open Discussion 1: Implementing EUCERD Recommendations for CEs

Francesc Palau, leader of WP7, started with a short presentation of the EUCERD recommendations for CEs. A lively debate ensued with contributions from several workshop participants. Some participants, especially CE members, expressed their satisfaction with this initiative, as they feel the experience of the existing CEs is not always considered. They highlighted the importance of having clear quality criteria for CEs in the form of EUCERD recommendations, as in their opinion self-designated CEs are pervasive in the European context and often do not fulfill minimum quality criteria. Another aspect concerning all recommendations was the importance of building a “culture” around rare diseases. Participants feel that some crucial RD stakeholders, such as health authorities or hospital directors, do not always understand the specificity of rare diseases and that sometimes this creates difficulties for professionals and patients. As one participant put it, “making the agenda more visible would help”.

Participants also expressed their concerns about specific issues related to CEs:

- Continuity of care:

According to the participants there is a substantial amount of work to be done in order to establish fruitful collaborations between departments and assure the continuity of care between childhood, adolescence and adulthood. This seems to be a quite recent problem: as

one of the participants put it *“now they arrive to adult age and we have to organize this aspect, not to lose and abandon them”*. Participants also valued some organizational solutions for this problem as more convenient than others; for example, they feel that the existence of different hospitals for adults and children makes this transition more difficult. Another issue signaled was the necessity of psychological support for adolescents in the transition phase.

- Multidisciplinarity and holistic care:

Several participants mentioned the need to provide holistic care to RD patients, delivering not only diagnostic services and treatment but also psychological and social support. Participants feel the pediatric approach to rare diseases management is more holistic than the approach often offered in adult services, where the care provided is aimed at treating acute problems, especially in hospital settings. Some of the participants pointed to the work of internists as a possible solution for the coordination of care, especially in multi-organ diseases.

Another specific issue discussed was the multidisciplinary competences/skills needed in CEs (e.g. psychological help, peer support groups, specialized social services..).

- CEs design:

The issue of single vs. multiple diseases center was discussed. All participants agreed on the importance of prioritizing multiple diseases centers. A participant proposed a model of evolving CE: CEs could start dealing with one disease and then move towards other related diseases. It was pointed out by several participants that organizing centers by groups of diseases would be cost effective as “shared resources” could be used. In many cases social workers, psychiatrists, nurses and other healthcare providers can deal -and are dealing- with different diseases. Participants also feel that dealing with more than one disease is the way to maximize the use of resources.

- Education:

The role of CEs in education at several levels was emphasized. Regarding specialized university training, it was mentioned that a curriculum design could include sessions taught by medical professors and patients, offering the students different perspectives on rare diseases. Education of healthcare professionals outside the field of rare diseases was also mentioned and identified as a priority.

- Human Resources:

Several conversations dealt with different aspects of human resources available in CEs'. A participant raised the question of the succession of the head of CEs. Many CEs seem to be very dependent on their head of department and their very existence is linked to the career of these individuals.

Other conversations focused on the need to have psychologists, social workers, secretaries, information technology persons and trial coordinators as an integral part of the staff of the CEs. At the same time participants stressed the difficulty of having proper funding for their work. In many health systems, reimbursement is only contemplated for diagnosis and therapy, which makes more difficult the financing of the aforementioned positions associated with continuous care. One participant highlighted the importance of having Information Technology professionals. These professionals are not contemplated in the current organization of the

existing CEs. Information Technology professionals are going to be even more important once telemedicine becomes widely available in the future. Several participants agreed with this and highlighted that in many cases other professionals (such as secretaries) have to teach themselves to manage IT systems.

Finally, the importance of integration and education of healthcare professionals working outside the CE was signaled. Some key problems were raised, such as the fact that these people may not be familiarized with the specificities of rare disease patients and as such they will either avoid dealing with these problems or risk to make a wrong decision; for example, social workers who are not familiar with rare diseases are sometimes “*too scared*” and “*afraid to take the risk*”, but they “*do not understand the risk and damage of not allowing the pediatric patient to carry on with normal social activities*”. It was stressed that CEs should support regular case conferences and make use of telemedicine tools to help those professionals.

Another participant linked some of these human resources problems with the origin of CEs: as many CEs come from “*University settings*”, they are not prepared to work as a “*formal [healthcare] institution*”. A related aspect brought up was the professionalization of patient support groups or “*peer supporters*”. This support work is, in many occasions, carried out by volunteers belonging to patient associations and due to its importance the improvement of their work conditions would have a considerable impact on quality of care.

- Financial sustainability:

Worries regarding CEs financial sustainability were expressed by several participants at the workshop.

It was stated that there is a need for a clear view regarding the real cost per patient of the different available treatments. In particular, one participant feels that there is a general misconception that treatments are very expensive because what comes to mind is enzyme replacement therapy, but there are many inexpensive treatments for many RDs.

Another participant, referring to the results of the interviews, commented that in some countries, CEs are providing care to RD patients without any realistic prospect, or in a context in which long running negotiations, for example, might be taking place as to how best to fund or pay for the services they already provide. So there is a strain on centers or hospitals providing these services and there is as yet no a clear way to pay for some of the services they are providing.

Some participants agreed that financial support by Governments and the EU should be conditional on the outcome of periodical evaluation of the CEs activity and good practices.

- EUCERD Recommendations for Centers of Expertise:

A comment was made concerning the findings of the interviews related to the low visibility of EUCERD’s agenda. Making this more visible would facilitate the activities of the CEs within the Institutions they belong to.

Also, the feeling is that there is a lack of information regarding the implementation of the Recommendations. Finding out how closely Recommendations align with the reality of European CEs is one of the aims of this work package.

One participant suggested that the work being done by WP7 and the discussions generated in this workshop could have added value for the EUCERD Recommendations.

- Other issues raised:

- In most countries, the health system is not organized in a way that facilitates the creation of CEs.
- There is a need to potentiate collaboration with Patients' support groups.
- Importance of professional communities maintaining informal or even quasi-formal relationships when constituting the future European Reference Networks (ERNs).
- Need for specialized social services.
- Communication problems between regional centers, national centers and foreign centers.
- Centers' self-designation versus officially designated CEs.
- Laboratories listed as expert laboratories but are not designated as such.
- Collaboration between Centers should be enabled, avoiding competition between them, especially at national level.
- Need to provide criteria for the evaluation of networks and CEs across countries.
- Periodical evaluation of CEs is necessary but criteria must be pertinent. If a country sets out quality standards for CEs, evaluation is straightforward because it can be done requesting information according to those standards.

Having guidelines facilitates evaluation on behalf of health authorities.

- A participant commented that EURORDIS is doing some research work regarding access to social services, the connection between medical and social services, and training of non-medical professionals in order to elaborate a future recommendation for guiding principles for social care. Collaboration between CEs and social services was also raised.

DAY 2

Presentation 4: *Centre of Expertise on Gaucher Disease – Lysosomal Storage Disorders* (Pilar Giraldo)

This presentation by Dr. Giraldo focused on the Center of Expertise on Gaucher Disease and other lysosomal storage disorders, covering its origins, organizational structure of clinical assistance and patient management, and activities such as participation in clinical trials, research projects, training and dissemination of information.

Round table / Open Discussion 2: *Informal networks of external collaboration of CE's and European Reference Networks* (Africa Villanueva)

This round table was moderated by Africa Villanueva, research fellow at INGENIO. A short slide presentation included a chart of a Centre of Expertise (CE) network illustrating network characteristics and highlighting the complexity and challenges that their management represents for CEs. It was also highlighted, as part of the preliminary results of WP7, that a significant proportion of CEs networks are characterized by informal/personal relationships among members, including links with other CEs.

The presentation intended to initiate the debate regarding CEs informal networks and the role of future European Reference Networks (ERN). A lively debate followed with contributions from many of the participants. There was consensus on the existence of informality within the current networks, but the need for some formalization was also expressed. The final discussion focused on the difficulties/needs and benefits of formalization:

- Difficulties/needs of formalizing informal networks:
 - Participants noted that CE networks operate at different levels ranging from local/regional to European or even international levels, involving diverse types of stakeholders. They agreed that networks should be formalized at all levels, even at the local team level, in the form of ERNs. The difficulty of identifying all network members and shared activities performed at the different levels was pointed out. For this reason, it was emphasized that a crucial role of a network coordinator is to describe the system accurately and try to avoid possible rigidities that might appear among members. Moreover, it was stated that there is a need for sustainable funding to create and coordinate these networks.
 - It was argued that formalization is a complicated process that may generate some bureaucratic burdens and a considerable waiting time until the network is officially recognized. A participant noted that these difficulties may be critical and may constitute an important barrier for small CEs to formally join these networks, due to the scarcity of time and other resources to allocate for networking purposes.

- Benefits of formalizing informal networks:
 - Participants agreed that at the beginning of a collaborative work a network operates informally, often at this stage being too dependent on people and not necessarily on institutions. The formalization of these linkages provides awareness to the rest of society but also enhances the sustainability of these structures of expertise. For example, it was commented that the improved formalization could significantly contribute to greater assistance for patients during situations of emergency, as the rest of the medical system could have easier access to the names and locations of medical experts.
 - The composition of future ERNs for RD was debated. Among the different opinions, some participants suggested the importance of including not only other CEs, but also healthcare providers, medical societies and patients' organizations; they proposed that ERNs should be designed to unite all stakeholders under one umbrella to avoid parallel and redundant structures.
- The ways in which formal networks in the form of ERNs could positively impact on enhancing key activities, such as access to diagnosis and treatment for all European citizens, was also discussed. Moreover, desirable key roles for ERNs were proposed, including improving registries, generating guidelines and protocols for emergency treatment, and contributing to the approval of new treatments. There was consensus that a formal and official ERN legitimates participants and practices, and might provide better access to funding for conducting such shared activities.
- There was also a consensus on the importance of ERNs for political lobbying in order to achieve equal access to high-cost drugs for all European citizens. A participant suggested that ERNs could become the main actor to persuade the EU to fund these treatments at the European level and guarantee equal access to orphan drugs to patients from less wealthy countries.

Presentation 5: *The experience of one participant CE (Hilary Longhurst)*

In this presentation, Dr. Longhurst introduced the Primary Immunodeficiency Clinic, Department of Immunology at the Barts Hospital and the London NHS Trust, detailing its origins, structure, working staff and activity. Dr. Longhurst also pointed out those aspects that could be improved, such as bureaucracy, logistics, capacity or infrastructure.

Presentation 6: *The experience of one participant CE (Pavel Drevinek)*

In this presentation, Dr. Drevinek introduced the situation of Centers of Expertise and Standards of Care for Cystic Fibrosis: Best practice guidelines, CF Center's framework and quality management. He focused on the experience of the Cystic Fibrosis Centre at the Motol

University Hospital - Charles University Medical School in Prague (Czech Republic). Dr. Drevinek provided information regarding the CE's origins, specialized staff, quality standards, achievements and challenges.

Presentation 7: *The experience of one participant CE* (Bruno Bembi)

This presentation was focused on the experience of the Regional Coordinator Centre of the Rare Diseases Network of Friuli Venezia Giulia Region. Dr. Bembi centered his presentation on the specific needs of RDs, which fully justify the necessity to constitute networks integrated by Italian centers located at different regions. Specific functions and the role of the Regional Coordinator Centre were approached, as well as its organizational chart, network composition, disease distribution, clinical activity, laboratory activity, diagnostic capabilities, research programs in which the Centre participates, ongoing clinical studies, partners, scientific production and dissemination activities.

Note: The following round tables originally scheduled were substituted by a round table entitled: ***ERNs: design and opportunities*** since both the organizers and assistants felt that the subject of the two round tables proposed had already been addressed during previous discussions.

Round table: Relevant challenges for CEs.

Round Table: The natural history of CEs. Where CEs come from?

Round table / Open Discussion 3: *ERNs: design and opportunities*

The final round table, moderated by Francesc Palau, WP7 leader, was devoted to the future design and implementation of European Reference Networks (ERN). Participants showed an uneven knowledge regarding existing pilot ERNs and European policies related to future ERNs. A lively debate ensued with contributions from several workshop participants. Key questions to be addressed by future ERNs were highlighted:

- It seems that there isn't a pre-conceived ERN model. Most probably, the Commission is expecting to receive ERN proposals with different models through a specific Horizon 2020 Call. This Call would be launched to explore and validate the options for modeling ERNs, and could entail actual networking activity or the development of a network to be implemented through this Call.
- The EUCERD recommendations on ERNs, although lacking definition of an "ERN model" offer guidance about the organizational aspects and flow of knowledge and expertise. These Recommendations should be taken into account in the preparation of the future ERNs; but at

the same time, we now have the opportunity to go back to them with the knowledge generated within this Joint Action and other initiatives and provide input to revise them.

- Belonging to an ERN will be voluntary but in order to be part of it, a CE or healthcare provider must be officially recognized as such at a national level. ERNs will be recognized by a special logotype. The problem many participants foresee at this point is the fact that criteria to be nationally recognized differ from one country to another. It is important that ERNs are institutionalized in order to be able to deliver better care to patients with less effort, avoiding the need to provide care in the form of personal relationships.

- The issue of whether ERNs should target single or multiple diseases was raised again, along with comments regarding the way grouping should be done or the difficulty of covering all RDs and setting up priorities for RDs and ERNs. The question of whether priorities should be set in terms of needs and not so much in terms of capacities and quality of the centers was also raised.

- A good model could be the Italian system of organizing Centers to be coordinated by another Center. If this model could be replicated all over Europe, “Networks of Networks” could be created.

- What selection criteria should be taken into consideration for a CE to become part of an ERN?

- Some participants argued that priority aspects to be covered by an ERN should be diagnosis and treatment. Other participants believe that diagnosis doesn’t represent a big challenge with the emergence of new technologies and that it can be outsourced. However, others argued that interpretation of all the genetic data generated requires a lot of expertise. In answer to this concern, some participants replied that one of the aims of ERNs is to include Centers or expert groups to provide this service to the rest of nodes within the network, as well as having the capacity to produce Good Practice guidelines and implement outcome measures and Quality Control (QC). Thus, the network could also be in charge of QC of Next Generation Sequencing (NGS) data generated by private labs.

- Regarding other possible ERN functions, some participants understand that besides diagnosis and treatment, emphasis should be placed on ability to provide/organize education and training activities at all levels.

- There was a consensus about the importance of IT technologies for ERNs, and participants referred to the first round-table, when the need for IT professionals in CEs was highlighted. Questions regarding the use of telemedicine or e-Health to offer better healthcare and specialized education and training were also brought up.

A participant mentioned that the need to address the issue of having a suitable IT platform for ERNs is still in discussion within the Commission.

- Participants also felt that, in some cases, patients (and not expertise) would need to travel from their home country to seek high quality healthcare. In those cases, participants proposed that the ERNs developed a joint decision-making procedure to decide which patients would need to travel. Some participants expressed their countries’ authorities’ fears related to RD patients’ ‘massive migration’ looking for expert centers outside their countries. In some

countries (e.g. Italy), looking for medical assistance externally is allowed in the event that care cannot be provided nationally.

- Another concern expressed was the fact that the concept of ‘Standards of Care’ varies from one country to another so it would be important to ensure that care provided in different countries is harmonized.

- Minimum requirements for healthcare providers to be part of ERNs were discussed as well. It was emphasized that there should be several levels of expertise within an ERN, where members with less expertise can benefit from knowledge generated by other ERN members. At this point, it was highlighted that some national healthcare systems already include several levels of expertise in their RD healthcare models, and thus can be used as inspiration for designing future ERNs.

Related to this issue, participants agree that it is not necessary to have experts in each country for a given disease or group of diseases but it is important to rely on physicians being able to recognize the disease or at least to know in which group of rare diseases it is included, in order to seek expert resources in other Centers /Countries.

On the contrary, social services must be provided by all countries so that they are easily accessible to all RD patients.

- Participants agree on the importance of Learned Societies at the European level, which can constitute the basis of ERNs. Many professionals know each other by means of these associations. ERNs will have to formalize what now are numerous, diverse and informal networks of practice around RD built mainly around professional associations.

- Other issues:

- Specific Horizon 2020 Call.
- Coordination of ERNs with European Regulatory Agencies to study the possibility of overseeing some of the clinical trials to ensure that some RD subgroups are not excluded from studies. Need for an international database of available resources.

Closing Remarks (Francesc Palau)

The workshop was closed by Francesc Palau with a ‘thank you’ from the organizers to all participants for a fruitful workshop. He also summarized the most relevant points raised during the two day event and mentioned WP7 future activities. Briefly, this WP will continue to conduct interviews with other stakeholders, including patients’ associations representatives and healthcare authorities (mainly Expert Group members), to achieve a full set of responses. The results of the ethnographic and qualitative research will then be analyzed in depth, drawing out conclusions regarding the real-life operations and networking of CEs for RDs, whilst identifying key challenges and good practices/models. The analysis of results will entail completion of the Report on Healthcare system actions in RD.

Annexes

Annex 1. List of participants

	Last name, name	Country	Organization
1	Palau, Francesc	Spain	CIBERER / WP7
2	Gomez, Beatriz	Spain	CIBERER / WP7
3	Corrochano, Virginia	Spain	CIBERER / WP7
4	Molas, Jordi	Spain	Ingenio-CSIC-UPV / WP7
5	Barberá, José David	Spain	Ingenio-CSIC-UPV / WP7
6	Villanueva, Africa	Spain	Ingenio-CSIC-UPV / WP7
7	Woolley, Richard	Spain	Ingenio-CSIC-UPV / WP7
8	Palanca, Inés	Spain	Ministry of Health, Social Services and Equality
9	Soler, Pilar	Spain	Ministry of Health, Social Services and Equality
10	Cortés, Marta	Spain	Ministry of Health, Social Services and Equality
11	Casado, Paloma	Spain	Ministry of Health, Social Services and Equality
12	Arias, Carmen	Spain	Ministry of Health, Social Services and Equality
13	Saiz, Isabel	Spain	Ministry of Health, Social Services and Equality
14	Peña-Rey, Isabel	Spain	Ministry of Health, Social Services and Equality
15	Giraldo, Pilar	Spain	Hospital Miguel Servet
16	Belmatoug, Nadia	France	Hôpital Beaujon
17	Vives, Joan Lluís	Spain	Hospital Clínic de Barcelona
18	Longhurst, Hilary	United Kingdom	Barts and the London NHS Trust
19	Drevinek, Pavel	Czech Republic	Motol University Hospital
20	Lund, Allan M.	Denmark	Centre of Reference for Rare Disabilities – KSH
21	Bembi, Bruno	Italy	Regional Coordinator Centre for Rare Diseases. Academic Hospital "Santa Maria della Misericordia", Udine
22	Lynn, Stephen	United Kingdom	University of Newcastle
23	Dan, Dorica	Romania	Eurordis
24	Utkus, Algirdas	Lithuania	Center for Medical Genetics
25	Graliński, Jacek	Poland	Former EUCERD Member
26	Posada, Manuel	Spain	ISCIII
27	Cruz, Jordi	Spain	FEDER
28	Evangelista, Teresinha	United Kingdom	University of Newcastle

	Last name, name	Country	Organization
29	Castro, Raquel	France	Eurordis
30	Mengel, Karl-Eugen	Germany	Villa Metabolica
31	Rodwell, Charlotte	France	INSERM
32	Hiort, Olaf	Germany	Lübeck University
33	Jessop, Edmund	United Kingdom	NHS England
34	Nafría, Begoña	Spain	Hospital Sant Joan de Deu
35	Arellanesova, Anna	Czech Republic	Czech Association for Rare Diseases
36	Weinman, Ariane	France	Eurordis
37	Cardellach, Francesc	Spain	IDIBAPS
38	Frazzica, Rosa Giuseppa	Italy	Istituto Superiore di Sanità
39	Laricchiuta, Paola	Italy	Istituto Superiore di Sanità
40	Ancochea, Alba	Spain	FEDER

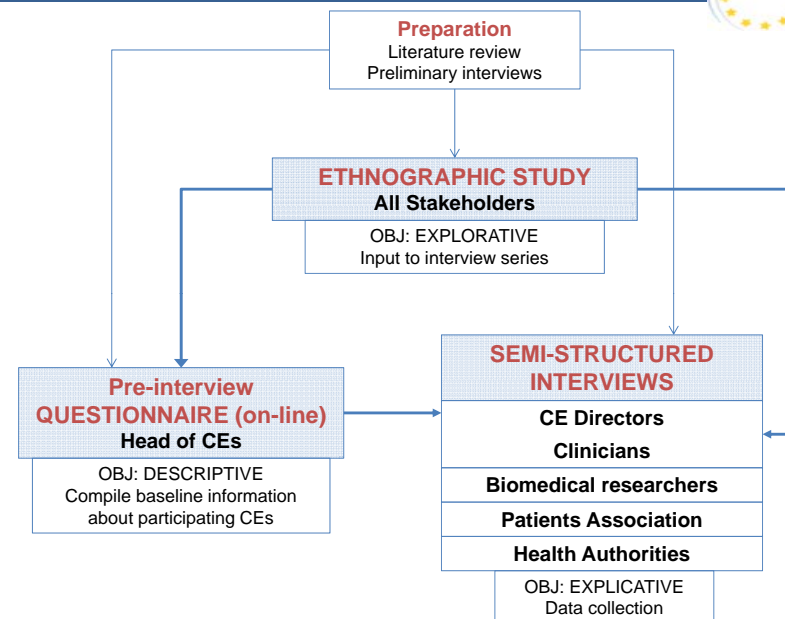
EUCERD JOINT ACTION WORKSHOP WP7 METHODOLOGY AND PRELIMINARY RESULTS

Ministry of Health, Social Services and Equality
Madrid, Spain
March 31st- April 1st, 2014

África Villanueva & Richard Woolley



Approach & methodology



Site of the ethnography

Reference Unit for Gaucher Disease & other Lysosomal Diseases

Location: Hospital Universitario Miguel Servet, Zaragoza

Founded: 1993

Access: Aragon Regional Health Authority

Director: Dr. Pilar Giraldo Castellano

Principal Activities:

1. **Clinics** for lysosomal diseases (diagnosis, follow-up of patients, treatment administration, referral to clinical trials, development of clinical guidelines)
2. **Basic, translational and clinical research** in the field of lysosomal diseases (enzymatic and genetic research, development of pharmacological chaperones, clinical trials)
3. **Training & awareness** (internal training, courses for specialists)

Outcomes of the ethnography

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

Four framing expectations:

- CEs are highly complex settings for developing good practices
- The management and coordination of CEs are integral to the development and diffusion of good practices
- Practices are always contextualised – different diseases and health systems will shape good practices in specific ways
- Networks are essential for building on core competences and extending capabilities to enhance good practices

Developing the interview framework



Objectives:

- Analyse how CEs' activities align with the EUCERD recommendations and how these activities contribute to improving Quality of Care
- Compile an inventory of good practices
- Identify future challenges for CEs

Our Framework:

- The EUCERD Recommendations for CEs
- Dimensions of Quality of Care
- Base unit of analysis: Groups or systems of practices on which the activities performed by CEs are based

5

The EUCERD Recommendations



- Strategic markers for CEs as health system innovation focused on Rare Disease 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

Guidance on organisation and activities of CEs

6

Quality of Care (QoC)



- 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)

- **Dimensions of Quality of Care:** system markers for the provision of health services

Effectiveness Efficiency Accessibility Safety Equity
Appropriateness Timeliness Acceptability Responsiveness
Satisfaction Continuity Transparency +

- Structures (inputs) – Processes – Outcomes

Guidance on practice improvements

7

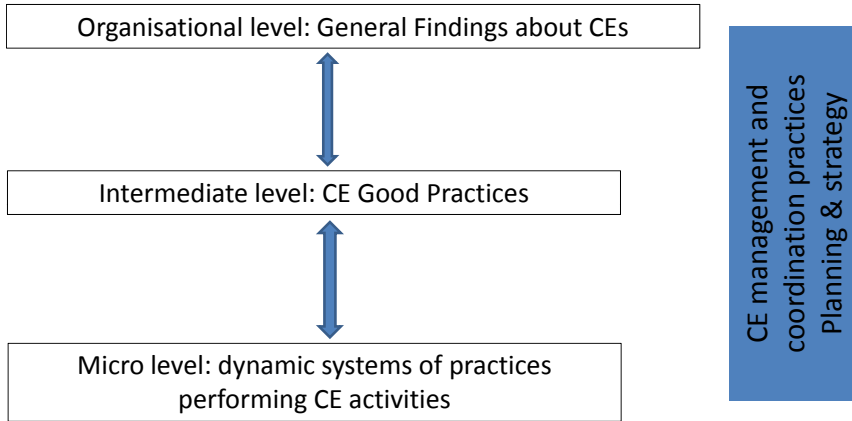
Developing the interview framework (cont.)



- The comparative dimension: improving QoC
 - Impact of country profile & profile of rare disease: importance of **context**
 - Goals of stakeholders: importance of **perspective** (professionals, families, health authorities, patient organisations, etc.)
 - Centres of Expertise
 - **What are 'actually existing' CEs doing, or wanting to do, across 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?**
- What good practices can be identified and diffused?
- How to contribute to EJA objectives?
 - Highlight the alignment of top-down (conceptual, institutional strategy) and bottom-up (existing practices, pragmatic strategy)
 - Opportunities for further development and improvement

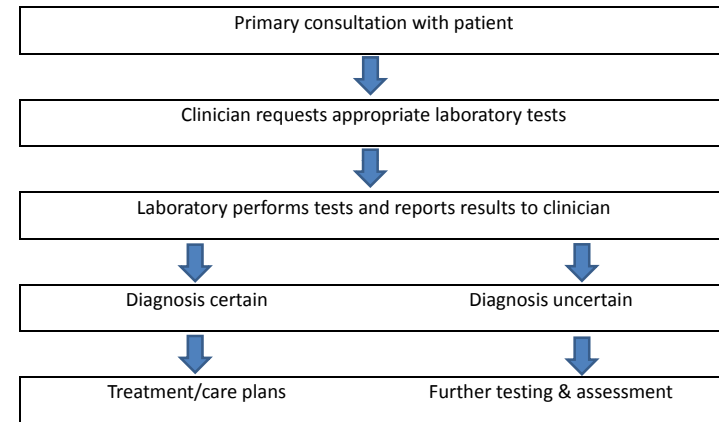
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Analysing CE practices

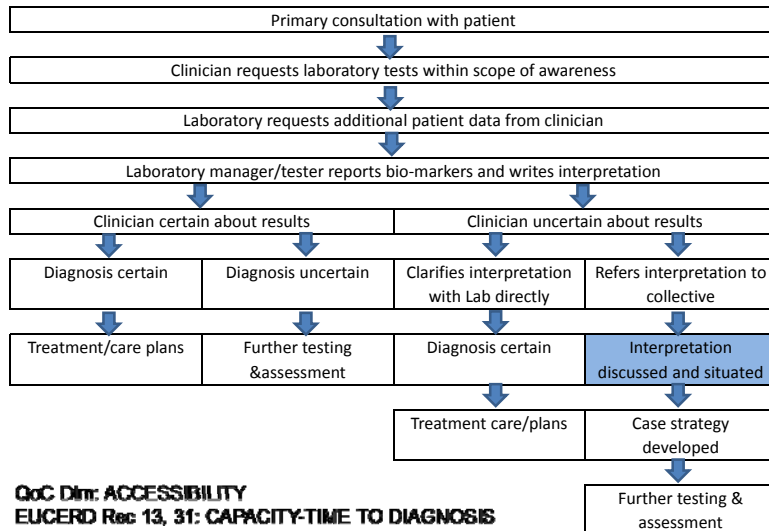


Laboratory testing: A simple model

QoC Dim: ACCESSIBILITY
EUCERD Rec 13, 31: CAPACITY-TIME TO DIAGNOSIS



Laboratory testing: empirical synthesis



QoC Dim: ACCESSIBILITY
EUCERD Rec 13, 31: CAPACITY-TIME TO DIAGNOSIS

Laboratory testing: potential for practice improvements

Laboratory Manager	Clinicians do not include sufficient clinical detail in the case data that accompanies genetic testing request <i>Reduces quality of the interpretation that can be written</i>
Genetic testers	Clinicians sometimes lack a clear understanding of what tests they can request and the significance of the results that they could obtain <i>Existing testing capabilities are being underutilised</i>
Laboratory	Capabilities required to fully exploited NGS technology lacking <i>Capacity to expand the number of genes/conditions being tested is being underutilised</i>
Clinicians	Interpretations written by the laboratory are of poor quality or seem to be unclear or lacking decisiveness <i>Informal or formal process to acquire additional information or opinion is required</i>

QoC Dim: ACCESSIBILITY
EUCERD Rec 13, 31: CAPACITY-TIME TO DIAGNOSIS

Laboratory testing: Transformation of practices



QoC Dim: ACCESSIBILITY
EUCERD Rec 13, 31: CAPACITY-TIME TO DIAGNOSIS

Laboratory staff	Institutional course on writing genetic test result interpretations
EUCERD Recs:	TRAINING; TRANSLATIONAL RESEARCH;
Genetic testers	Visit or extended stay in collaborating laboratory (preferably using same NGS technology)
	TRAINING; NETWORKING; RESEARCH STATE-OF-ART;
Clinicians	Lab Manager introduces and leads discussion of tests in CE meetings <i>Vendor provided in-house training on NGS technology?</i>
	TRAINING;
CE	In-house development, adoption and promotion of guidelines for clinical data to accompany testing requests
	DEVELOPMENT OF GUIDELINES; GOOD PRACTICE DIFFUSION;

Outcome: An improved system of practices around the process of genetic testing for new CE patients; significant potential to reduce delays in diagnosis.

CE good practice: Laboratory testing



- ✓ CEs are fine-tuning processes associated with laboratory testing to reduce time to diagnosis and to incorporate new technology and tests
 - Providing increasingly detailed clinical data with testing requests
 - Diffusing knowledge about tests and testing capabilities within the CE
 - Undertaking relevant training, including absorbing tacit knowledge
 - Adapting to new forms of data, including working with bio-informaticians
 - Adapting to technological change
 - Producing new guidelines
 - Building new networks and relationships with relevant experts
 - Consolidating informal relationships and interactions

*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.*

*Of course we also have had to learn how to write this, **how to make it clear...** of course sometimes the clinicians call us one hundred times because they don't get the idea!*

QoC Dim: EFFECTIVENESS; EFFICIENCY; CONTINUITY
EUCERD Rec: CAPACITY-TIME TO DIAGNOSIS; MULTIDISCIPLINARITY & INCLUSIVENESS; PATIENT FOCUS

CE good practice: Regular clinical meetings



- ✓ CE meetings are facilitating multidisciplinary exchanges of knowledge and expertise
- ✓ Good practice example: **Interpreting laboratory test results**
 - The interpretation is circulated and can be annotated.
 - Laboratory staff can explain technical aspects.
 - Alternative interpretations can be exchanged and discussed based in multiple disciplinary knowledges and accumulated experience.
 - A conjoint interpretation is negotiated and implications discussed.
 - A case strategy can be developed based in shared understandings and grounding specific requirements or expectations of CE members.
 - Future coordination of case strategy can be outlined around the common basis for understanding the patient.
 - Builds on direct and informal interactions.

*In general, with the personal contact, that's one point. And with some - with clinical meetings on specific cases, with discussion, with the problem discussion. 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.*

QoC Dim: EFFECTIVENESS; EFFICIENCY; CONTINUITY
EUCERD Rec: CAPACITY-TIME TO DIAGNOSIS; MULTIDISCIPLINARITY & INCLUSIVENESS; PATIENT FOCUS

CE good practice: Regular meetings



- ✓ Regular face-to-face interaction integrating knowledge and expertise
 - Director; Clinical Specialists - Neurology, cardio-vascular, pulmonary, etc; CE Clinicians - Paediatricians, general; Testing Laboratory - Manager, technician; Nurses; Case manager; Researchers; Social worker; Other specialists – physio, nutrition, etc.
- ✓ Clinical meetings
 - Overview of case management
 - Specific diagnosis and therapeutic issues
 - Process management
- ✓ Management and future planning meetings
 - General functioning of the CE
 - Liaison and collaboration with stakeholders
 - Human resource issues
- ✓ Specific case management meetings

QoC Dim: EFFECTIVENESS; EFFICIENCY; CONTINUITY
EUCERD Rec: CAPACITY-TIME TO DIAGNOSIS; MULTIDISCIPLINARITY & INCLUSIVENESS; PATIENT FOCUS

CE good practices: Patient management



✓ CEs are working to organise the best possible new patient circuit

- Revision of documentation prior to visit, selection of clinical protocol
- Dedicated specialised nursing staff in charge of coordination, programming
- Utilising E-systems such as intranets for scheduling evaluations, tests,
- Enhancing baseline assessment grounding ongoing case management
- Fostering direct & informal communication with experts
- Providing assistance with accommodation and/or travel
- Establishing reception areas
- Providing information for families

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...

QoC Dim: ACCESSIBILITY; EQUITY; SAFETY
EUCERD Rec 4, 10, 11, 25, 31, 32: CAPACITY-TIME TO DIAGNOSIS; MULTIDISCIPLINARITY; PATIENT FOCUS; SENSITIVITY

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CE good practices: Patient management II



✓ CEs try to improve management of patient care in local and/or home contexts

- Assigning of physical and other therapies to local/regional services
- Detailed letters and instructions for localised care by GP or specialist
- Direct liaison with GPs and community care services
- Regular follow-up visits including monitoring of proposed treatment & care
- Coordination with social workers
- Interaction with Patient Associations

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.

QoC Dim: CONTINUITY; ACCESSIBILITY; SAFETY
EUCERD Rec 4, 5, 10, 11, 29, 31: INCLUSIVENESS; PATIENT FOCUS; SENSITIVITY

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CE Good practices: Continuity of Care



✓ 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 Dim: CONTINUITY; ACCESSIBILITY; SAFETY
EUCERD Rec 10, 13, 26, 27: CORE COMPETENCIES; TRAINING; PATIENT FOCUS

CE good practices: Networking



✓ CEs are linking, and actively facilitating, relationships between stakeholders that contribute (directly and/or indirectly) to improving quality of care

- CEs are integrating transversal collaborations between medical disciplines
- CEs seek assistance with diagnosis and care from other CEs or medical experts nationally and internationally as required
- CEs are providing a focus for connections and interactions between university and clinical researchers
- Emerging stakeholder concerns, and relevant information, are being disseminated among stakeholders through CEs
- CEs are interacting/cooperating/collaborating with Patient Assoc. on range of activities
- CEs are playing a coordinating role in multi-stakeholder activities such as clinical trials, information days, etc.

*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.*

QoC Dim: EFFECTIVENESS; CONTINUITY
EUCERD Rec 4, 6, 7, 26, 27, 28, 29: LINKS & COLLABORATIONS; MULTIDISCIPLINARITY; INCLUSIVENESS

Some good practice analyses in progress



- Research practices & the clinical interface
 - Clinical research
 - Research networks
 - Translational research
- Working with Patient Associations
 - Interviews currently underway with PA representatives
- Information and awareness
 - Information about diagnosis and referral for physicians
 - Information relevant to diagnosis and treatment for families, patients
 - Information about CE services for physicians and patients
 - Information about social services and social work support
 - Building awareness of RDs in medical communities, organisations
- Planning and management
 - CE strategies and opportunities

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Preliminary general findings about CEs



- ✓ CEs are heterogeneous in their organisation
- ✓ CE Directors and clinical staff value 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
- ✓ Many CEs are in 'survival mode' due to funding problems linked to financial crisis
- ✓ Next-generation sequencing (NGS) technology is central to many CE futures and is transforming practices
- ✓ CEs can be the vehicle for important knowledge spill-overs between clinical practice, clinical research, basic research and clinical trials
- ✓ CEs are playing increasingly important roles as information hubs and knowledge brokers
- ✓ Many CEs provide a multi-disciplinary capability and the capacity to coordinate effectively between disciplines

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Some apparent big challenges for CEs



- Human resources – developing and sustaining personnel including transferring knowledge to next generation of specialists, the availability of specialised support staff, training adult specialists
- Telemedicine or e-services – building pathways from primary care and facilitating routine consultations for remote patients, sharing securely share patient information and clinical research data within European and global networks
- Technical change – finding cost effective ways to take advantage of the possibilities of new technologies, data sharing potential
- Continuity of care – transition of from paediatric contexts of relatively holistic care to (emergent) adult contexts
- Information & awareness – CEs are increasingly becoming hubs for the diffusion of information and raising of awareness about RDs, among the population, medical specialists, general physicians, etc.

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Some firm Conclusions about CEs



- CEs are heterogeneous and distributed in their organisation – spanning a number of units, organisations, disciplines, etc.
- The evidence suggests that, as a way of organising work for RD patients, CEs can be innovative and responsive
- CEs can integrate multiple dynamics and drivers of change in pursuit of the objective of improvement in Quality of Care
- CEs can bring together the frontier of the scientific 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

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- Inform the EJA about the core practices and knowledge dynamics of CEs engaged in improving QoC for RD patients
- Provide evidence and support for CEs as a system innovation for addressing the particular challenges of working for RD in Europe
- Provide key insights regarding processes of alignment of different national and disease settings and Criteria for CE designation (& for ERN membership).
- Contribute to the Review of EUCERD Recommendations for CEs

Thank you for your attention.

Questions or comments?



Annex 3 Documents circulated to participants

EUCERD Recommendations. Quality Criteria for Centres of Expertise for Rare Diseases in Member States (24 October 2011)

http://www.eucerd.eu/?post_type=document&p=1224