Polka
a project by Eurordis
and its partners

Patients‘
Consensus
on Preferred
Policy Scenarii
for Rare
Diseases

The POLKA Delphi Study

Policy Recommendations on Rare Diseases

Centres of Expertise

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EXECUTIVE SUMMARY

As part of the European process of defining strategies and refining the structural plans for the diagnosis, treatment and management/care of rare diseases, the POLKA project addresses the issue of how to involve patients and their representatives in the most constructive way as partners in this process. POLKA is timely and appropriate as it links the efforts of the European Institutions to member state (MS) initiatives and to the interests of patients. Patients and their representatives participated in the development of "strategies and mechanisms for exchange of information among people affected by rare diseases", as well as in the "support for European Reference Networks for rare diseases in an effort to establish guidelines for best practice on treatment, and to share knowledge on rare diseases, together with evaluation of performance".

In order to do this, based on the results of the RAPSODY project, an innovative mechanism for the exchange and processing of information among patients affected by rare diseases was pursued through the collection, analysis and presentation of relevant experiences and informed opinions on health policy themes from patients and health care professionals, from the personal up to the local centre level through to National and finally to a European shared strategy level.

In order to do this systematically, we used a DELPHI-like methodology whereby we would allow individuals from various backgrounds an equal chance of weighing in with their opinions on what is recognised to be a complex problem – how best to develop policy recommendations for rare disease care.

We instituted a modified Policy DELPHI (non-anonymous). First, participants were asked to fill in an online survey – participants were patients and their representatives and/or families and enablers as well as healthcare professionals, from administration to clinicians, nurses and paramedics (from 15 April 2011 through 15 June 2011). Participants of this survey were from England, Denmark and France, and within the survey were asked if they would be interested in taking part in a follow-up face-to-face meeting to discuss the global results of the survey.

After analysing the survey, we invited participants to attend a face-to-face meeting in order to achieve a site-centric agreement on the same questions (meetings happened between 23 June and 29 July 2011). Through a lively and stimulating series of four-hour plus meetings (even the one that was shortened at the last minute to 2 hours extended to almost 3), we got first-hand accounts and opinions of why certain questions "had to be" answered in just a certain way.

We then worked on synthesising all 7 meetings into answering the same questions in order to revise, amend and update the Declaration on Reference Networks.

The 12 policy areas covered in both the online questionnaire and in the meetings were

- The patient-friendliness and care provided at the Centre
- Easy access to a Centre
- Coordination and cooperation within and outside of the Centre

- The transition from paediatrics to adult care
- Patient autonomy and patient representativity in the Centre
- Provision of information to patients, their families and their enablers
- Raising awareness among external HCPs and non-HCPs
- Patient-oriented and multidisciplinary care
- Social care assistance
- Diagnosis facilities at the Centre
- Performance management (and patient input into that) of the Centre
- Research at the Centre

The data from these two rounds of the POLKA Delphi study were collated and analysed in order to develop the following list of recommendations.

GENERAL RECOMMENDATIONS

One cross-cutting theme is communication. From Centre communication to professionals outside the Centre to clear paths of communication that always include the patient within the centre, communication is a vital tool through which ideas, truth, and understanding of the disease can be better understood and aligned.

ORGANISATION

1. Centres should be set up as separate units providing multidisciplinary care with dedicated staff for patients. The unit should not be 'over-medicalised' and designed such that it takes into account the needs of patients. Referral of a patient to a Centre should be facilitated and rapid.
2. The essential functions of a Centre of Expertise should be able to provide diagnosis, treatment and care for people with the rare diseases it specialises in, as well as train individuals and carry out research.
3. Centres of Expertise should establish themselves as research facilities, include patient organisations in the prioritisation, design and execution of studies and in ensuring appropriate dissemination of research results to all stakeholders.

4. Cross-border agreements between European member states should be set up to allow European patients access, if needed, to other European Centres of Expertise. This interchange would also help in training professionals, especially for those countries with little or no experience in Centres of Expertise.

5. Transition care pathways that ensure smooth transition of patients and patient records from paediatric care to adult care, as well as preparation for any future needs of patients that may arise due to the condition should be clearly mapped out. The transition process should be overseen by a transition coordinator.

PATIENTS AND PATIENT ORGANISATIONS

6. Patients and their families should be regularly provided with information about their health condition using appropriate channels and formats adapted for the target audience, including age, ethnicity and education level. Patient organisations would be particularly useful here as links between health information and the patient/family.
7. Every Centre of Expertise should be linked to a patient-led organisation that is a partner in the centre and has a special emphasis on the social and medical well-being of the patient. These organisations should be funded for being the link between patients and the Centre, for helping orient the patients, and to help the centre communicate effectively with the patients and family/enablers and vice-versa.
8. Systems should be set up at centres to manage performance at Centres of Expertise with representation on committees from patients or patient organisations. For the evaluation of Centres of Expertise in terms of cost-effectiveness, the main issue is that for most rare diseases there is no satisfying health outcome indicator to measure benefit satisfactorily. One way of evaluation would be to ask patients themselves if they are visiting the centre because: 1. There is no other centre and the patient feels he/she has no choice, 2. There is no other centre but the patient feels his/her health is optimised by being treated at the centre, 3. There is a choice between different centres, and the patient feels his/her health is optimised by being treated at the centre he/she chooses.
9. Even if patients are globally satisfied by the quality of care they receive in their Centres, Centres of Expertise need to fulfil other roles than treating patients with best quality care: they need to develop exemplary governance by involving patients’ representatives in their governing bodies or evaluation committees, to conduct research and develop research tools and involve patients’ representatives in these processes, and to communicate with the patients on their global activities.

PERFORMANCE

10. IT systems need to be established to ensure efficient exchange of patient information in the healthcare system, to General Practitioners (GPs), to external specialists, to non-Centre health professionals like ambulance services and Emergency, as well as to the patients themselves.
    a. Systems need to be set up in order to ensure patients are identified and moved appropriately through Emergency Care, always having important information about their condition on hand with immediate access to experts during the emergency.
11. Outreach clinics headed by a Centre should be established to minimise travel for patients. Travel and accommodation services need to be provided for all patients and family and enablers.
12. A dedicated social support worker/welfare officer with knowledge of the condition must be available at Centres of Expertise to help patients navigate the social care system.
13. Centres of Expertise must have access to psychologists to provide counselling to patients, their families and enablers. Complete teams should be composed centre-by-centre on an as needed basis.

TRAINING

14. Training for external HCPs such as GPs and specialists should be mandatory (or offered as extra credit, i.e., CME) to help conceive of something possibly “rare”, and then ensure quicker referral to a specialist when a rare condition is suspected.
15. HCPs (doctors, nurses and allied HCPs) should be institutionally offered diversity training (language, culture, gender) in order to maintain respectful connection with their varied patients and families.
16. Non-HCPs who come in contact with rare disease patients (such as school teachers, carers, employers, etc.) should be offered training. Patient organisations should have a role here.

The final round of POLKA consisted of participants being sent these recommendations and being asked to agree or not with them, using a scale from 1 (very important) to 3 (not important). They were given only 10 days to respond and 100 people did so in a very short turnaround time. Of course, our conclusions are based on a relatively small sample size both in terms of absolute numbers and diseases looked at.

INTRODUCTION

Rare diseases are diseases with a particularly low prevalence. The European Union defines diseases to be rare when they affect not more than 5 per 10,000 persons in the European Union (EU CERD 2011). It is estimated that between 5000 and 8000 distinct rare diseases exist, affecting between 6% and 8% of the population in the course of their lives (EU CERD 2011). Although rare diseases are characterised by a low prevalence for each of them, the total number of people affected by rare diseases in the EU is between 27 and 36 million (EU CERD 2011). Most individual diseases affect one in 100,000 people or less (EU CERD 2011).

Rare diseases are a serious public health concern and are a priority in the EU (European Commission). At the European level, a strategy has clearly been adopted by the Commission, the Council, and the European Parliament, whereas at the Member States level, not all have adopted a robust plan or strategy as of this report. Few European countries have specialised provisions of health services for rare diseases. In 2009, EU health ministers recognised the need for action in the field of rare diseases and agreed on a set of recommendations (Council Recommendations on action in the field of rare diseases 2009). The recommendations concentrate on supporting and strengthening EU countries' plans and strategies for responding to rare diseases, improving recognition and visibility of rare diseases, encouraging more research into rare diseases and forging links between medical centres known as centres of expertise and health professionals in different countries through the creation of European Reference Networks.

A Centre of Expertise[1] may consist of units based at institutions (hospitals or universities) sharing a common idea or vision and an overall and clearly defined set of objectives for clinical care and research. There is no fixed formula for creating a Centre. Centres may differ in size and mode of organisation. The centre must have a well-defined framework for cooperation. They tend to be composed of highly ambitious, original and potentially ground-breaking clinical management and research teams, have excellent basic, applied and translational research. They work integrally with patients and patient organisations and make decisions as a team. They share a physical community and the leaders are outstanding researchers and visionary leaders. They help prepare the next generation, via training and education and have an international profile.

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[1] 2011 Report on the State of the art of Rare Disease activities in Europe of the European Union Committee of Experts on Rare Diseases Part II: European Commission and other European activities – EU-CERD.

A Reference Network[2] is defined as a physical or virtual networking of knowledge and expertise of national Centres of Expertise in more than one European country. The goal of a Reference Network is the improvement in the overall quality and management of care of a single rare disease or group of rare diseases with similar health care needs by complementing, supporting and providing added-value to the existing services and expertise at the national level. Such networking activity between national Centres of Expertise promotes the sharing and mobility of expertise rather than patients themselves, but permits the travelling of patients to cross-border Centres of Expertise when necessary. Patients in every European country can benefit from a Reference Network, although Centres of Expertise that are members of the network are not necessary in every European country.

As part of the EC's commitment to improving healthcare for rare disease patients, in 2008 a 3-year project called POLKA: 'Patients' consensus on preferred policy scenarii for rare diseases' was funded and supported by the European Commission DG Sanco. The main objective of the POLKA project was to facilitate the consultation of the European rare disease community, with the aim of building consensus on preferred public health policy scenarios for rare diseases, including the quality of care available at Centres of Expertise and if that quality level was sufficient.

In order to achieve the POLKA project's objective of gathering expert advice and experience and building consensus on policies for Centres of Expertise on rare diseases, a study called 'The Delphi study' employing a modified Policy Delphi technique was carried out.

This document details the POLKA Delphi study, its findings and the policy recommendations developed from them.

OBJECTIVES

The aims of the POLKA Delphi study were to:

- access the current experiences of patients and their families/enablers and healthcare professionals (HCPs) at selected centres of expertise in Denmark and England;
- identify discussion points within the current services;
- develop policy recommendations based on these findings in order to help improve care for rare disease patients across Europe.

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[2] 2011/24/EU of the European Parliament and of the Council of 9 March 2011 on the application of patients' rights in cross-border healthcare.

METHODOLOGY

DEFINITION OF THE DELPHI TECHNIQUE

The Delphi technique is defined as *"a method for structuring a group communication process so that the process is effective in allowing a group of individuals, as a whole, to deal with a complex problem"*[3]. It is based on the premise that 'pooled intelligences' enhance individual judgment through capturing the collective opinions of experts[4].

According to many authors[5] the Delphi technique is an effective and reliable data collection method that is particularly useful when there is little empirical knowledge and thus uncertainty surrounding the area being investigated. It was originally developed by the RAND Corporation in the 1950s and was applied to assess the direction of long-range trends, with emphasis on science and technology and their effects on society; therefore, it was used principally in forecasting likely inventions, new technologies and the social and economic impacts of technological change[6].

THE DELPHI TECHNIQUE IN HEALTHCARE

The Delphi technique is becoming increasingly popular in health and social research[7] and has been used for various purposes in obtaining a wide variety of outcomes. Its aim[8] has been to identify and prioritise clinical indicators to audit mental health nursing in clinical practice. Several other studies have used the Delphi technique to develop clinical guidelines. One such study[9] used a modified

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[3] Linstone, H. A. & Turoff, M. (1975). *The Delphi Method Techniques and Applications*. Massachusetts, Reading: Addison-Wesley.

[4] Linstone, H. A. & Turoff, M. (1975). *The Delphi Method Techniques and Applications*. Reading, Massachusetts: Addison-Wesley; Moore, C. M. (1987). *Group techniques for idea building*. Thousand Oaks, CA: Sage Publications; Murry, J. W. & Hammons, J. O. (1995). Delphi: a versatile methodology for conducting qualitative research. *Review of Higher Education, 18,* 423-436.

[5] Dalkey, N. C. & dc Helmer, O. (1963). An experimental application of the Delphi method to the use of experts. *Management Science, 9,* 458-467; McKenna, H. P. (1994). The Delphi technique: a worthwhile research approach for nursing? *Journal of Advanced Nursing, 19,* 1221-1225; Crisp, J., Pelletier, D., Duffield, C., Adams, A., & Nagy, S. (1997). The Delphi Method. *Nursing Research, 46,* 116-118.

[6] Adler, M. & Ziglio, E. (1996). *Gazing into the Oracle:The Delphi Method and its Application to Social Policy and Public Health*. London: Jessica Kingsley Publishers.

[7] Hasson, F., Keeney, S., & McKenna, H. (2000). Research guidelines for the Delphi survey technique. *Journal of Advanced Nursing, 32,* 1008-1015.

[8] Hardy, D. J., O'Brien, A. P., Gaskin, C. J., O'Brien, A. J., Morrison-Ngatai, E., Skews, G. et al. (2004). Practical application of the Delphi technique in a bicultural mental health nursing study in New Zealand. *Journal of Advanced Nursing, 46,* 95-109.

[9] van der Linde, H., Hofstad, C. J., van Limbeek, J., Postema, K., & Geertzen, J. H. B. (2004). Use of the Delphi technique for developing national clinical guidelines for prescription of lower-limb prostheses. *Journal of Rehabilitation Research and Development, 42,* 693-704.

Delphi to develop national clinical guidelines for prescription of lower limb prostheses. A "Policy Delphi"[10] was applied to achieve consensus on tobacco policies among legislators. In a recent study, a modified Delphi technique was used to develop international policy options for SARS and SARS-like emerging infectious diseases[11] and its evaluation reported it to be an effective tool in certain Public Health debates[12].

CLASSIFICATION OF THE DELPHI TECHNIQUE

There are several types of Delphi[13]:

- The Classic Delphi
- The Policy Delphi
- The Decision Delphi
- The Group Delphi

The Policy, Decision and Group Delphi are variations of the Classic Delphi. The Policy Delphi is mainly used in social and political issues and is suitable for application in the social sciences. The aim of the Policy Delphi is not to reach stability or consensus, but rather to generate policy alternatives. Here the Delphi is used as an instrument for policy development and promoting participation.

The Policy Delphi is characterised by the following five features:

- Selective anonymity (participants answer questionnaires individually and subsequently exchange views in a group meeting)
- Iteration
- Controlled feedback
- Polarised (opposing) group response
- Structured conflict (policy alternatives generated for an issue).

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[10] Hahn, E. J. & Rayens, M. K. (1999). Consensus for tobacco policy among former state legislators using the policy Delphi method. *Tobacco Control, 8,* 137-140.

[11] Syed AM, Hjarnoe L, Krumkamp R, Reintjes R, Aro AR. Developing policy options for SARS and SARS-like diseases – a Delphi study. *Global Public Health*. 2010. 16:1-13. PMID: 20162483.

[12] Syed AM, Hjarnoe L, & Aro AR, The Delphi Technique In Developing International Health Policies: Experience From The SARS Control Project. *The Internet Journal of Health*. 2009 Volume 8 Number 2.

[13] van Zolingen, S. J. & Klaassen, C. A. (2003). Selection processes in a Delphi study about key qualifications in Senior Secondary Vocational Education. *Technological Forecasting and Social Change, 70,* 317-340.

There are many more variations of Delphi now in existence known as 'modified Delphi'[14], as researchers modify the Classic to suit their needs and few researchers nowadays see the need to use uniform methods. In order to meet the objective of the POLKA Delphi study a modified Policy Delphi technique was employed.

THE POLKA DELPHI TECHNIQUE

In order to meet the objectives of the POLKA Delphi study, a modified Policy Delphi technique was designed and implemented. The Delphi study was carried out in Denmark, England, and partially in France (the questionnaire round only). The Delphi process consisted of 2 rounds – the questionnaire round (Round 1) and face-to-face meeting (Round 2). Figure 1 illustrates the stages of the Delphi study. After recruitment of participants a questionnaire survey was carried out. The results of the questionnaire survey are fed back to participants and used to facilitate discussions at face-to-face meetings. The results of both rounds were collated and used to write the final report and develop policy recommendations. The final report was then shared with individuals for their comments before the final recommendations were delivered to the EC, to the European Union Committee of Experts on Rare Diseases (EU-CERD) and further disseminated to European member states.

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[14] McKenna, H. P. (1994). The Delphi technique: a worthwhile research approach for nursing? *Journal of Advanced Nursing, 19,* 1221-1225.

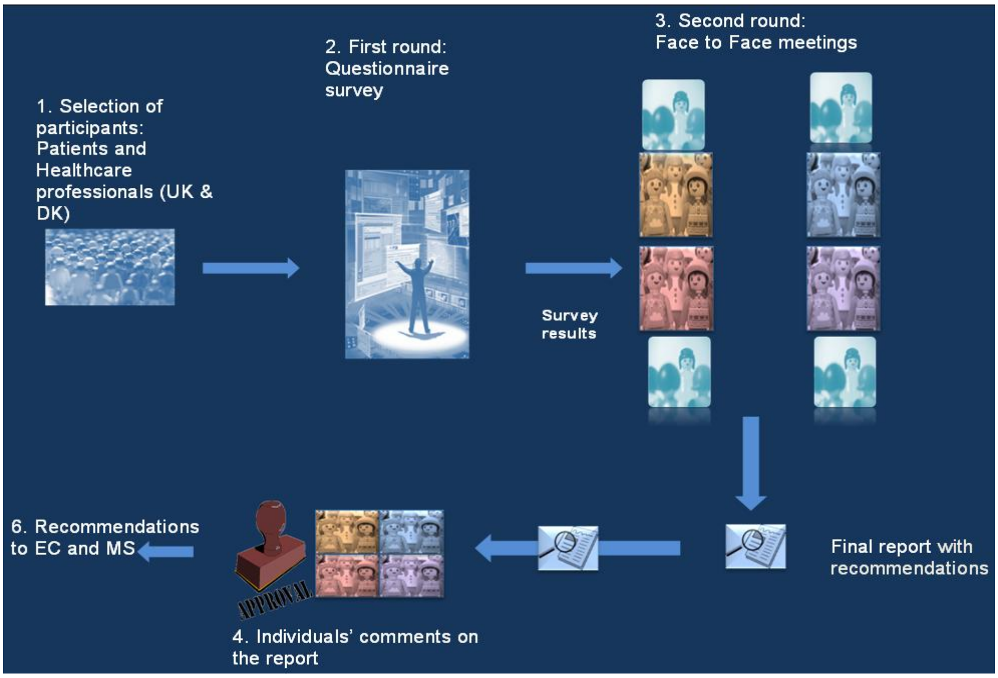

FIGURE 1: THE POLKA DELPHI PROCESS

PARTICIPANTS

Centres of Expertise for rare diseases in Denmark (DK) and England (EN) were contacted to participate in the study. A total of 8 centres, 1 in Denmark and 7 in England, agreed to participate. In each centre, a contact person was established who coordinated the study locally. HCPs and patients/families were identified and invited by the contact person. 52 HCPs and 97 patients from 8 centres of expertise responded to our open invitations to participate in the study (Table 1) – our goal was a total of 140 participants in England and Denmark (56 HCPs and 84 patients).

Originally, France was to participate in the face-to-face meetings as well. The online survey was set up and patients recruited (some 58 French patients filled it in over a three-month period). The medical centres ran into a time squeeze as they were going through a series of internal reviews at exactly the same time. They regretfully were not able to participate although they showed great interest in doing so in the near future. We did include the French patient surveys in the final analysis but no medical centre in France was able to take part.

Country	Health care professionals	Patients
Demark	5	13
England	47	84
France	0	58
Total	**52**	**155**

Table 1: Questionnaire responses by profession and country

ROUND 1: QUESTIONNAIRE SURVEY

Two questionnaire instruments, one for HCPs and one for patients, were developed with the aim of accessing the current experiences of healthcare professionals and patients at the centres of expertise. The questionnaires were built around the Declaration of "Common Principles on Centres of Expertise and European Reference Networks for Rare Diseases"[15] which was developed by the European organisation for Rare Diseases (EURORDIS) based on the outcomes of the RAPSODY project (Rare Diseases Patient Solidarity 2006-2008) and on the results of a survey called 'Eurordiscare3' which gathered data on patients' experience and expectations concerning health services for rare diseases in Europe.

The majority of the questions asked in the HCP and patient questionnaires were similar except for a few which were specific to HCPs or patients. The questionnaire for HCPs consisted of 46 questions and the questionnaire for patients consisted of 39 questions. Questions in both HCP and patients questionnaires were divided into the following 12 policy areas policy areas:

i. The patient-friendliness and care provided at the Centre
ii. Easy access to the Centre
iii. Coordination and cooperation within and outside the Centre
iv. Transition from paediatrics to adult care
v. Patient Autonomy, patient representatives and support groups at the Centre
vi. Provision of rare disease information to patients
vii. Raising awareness and training amongst external healthcare and non-healthcare professionals

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[15] Available at http://download.eurordis.org/europlan/3_EURORDIS_Guidance_Documents_for_the_National_Conference/8_Declaration_Centres_of_Expertise-nov08.pdf

viii. Patient-oriented and multidisciplinary care at the Centre
ix. Social care assistance
x. Diagnostic facilities at the Centre
xi. Performance management of the Centre
xii. Research at the Centre

The questionnaires were distributed by the contact person at each Centre of Expertise. Participants were given a choice of replying to the questionnaire electronically using web-based software on Survey Monkey or by post if they opted for a paper version. For the Danish participants the same questionnaires were translated from English into Danish; and into French for the French.

A total of 155 participants replied to the questionnaire survey from Denmark, England and France with a cut-off date of 9 August (Table 2). All replies were received electronically except 11 replies that were sent by post. A summary of the replies by HCP and patients in each policy area are presented in the following sections. For detailed results, please see Appendix 1 for results from Denmark & Appendix 2 for results from England (note: preliminary results as of 31/08/2011). A separate appendix was compiled to include all the open-ended comments noted.

ROUND 2: FACE-TO-FACE MEETINGS

Following the completion of Round 1 of the POLKA Delphi study, the participants were invited to the face-to-face meetings that were planned separately with each of the 8 Centres of Expertise (Table 2). In the face-to-face meetings, participants were presented with the global results of the Round 1 questionnaire survey and discussions were facilitated by policy area. Participants were asked to give their opinions on the survey results, to highlight what works well and identify areas of concern and suggest and agree on recommendations which would help improve the quality of the services and healthcare for patients.

Centre of Expertise[16]	Rare disease	City, Country	Participants involved	
			HCPs	Patients
The Centre for Rare Diseases, Aarhus Universitets Hospital	Various conditions	Aarhus, Denmark	5	10
Department of Genetic Medicine, Central Manchester University Hospitals	Neurofibromatosis	Manchester, UK	5	2
Birmingham Children's Hospital and Torbay Hospital	Alström Syndrome	Birmingham and Torbay, UK (1 meeting for 2 centres)	3	7
Guy's & St. Thomas' Trust	Epidermolysis Bullosa	London, UK	2	5
Newcastle NCG Mitochondrial Disease Service	Mitochondrial Diseases	Newcastle, UK	7	5
Birmingham Children's Hospital	Epidermolysis Bullosa	Birmingham, UK	7	0
Guy's & St. Thomas' Trust	Xeroderma Pigmentosum	London, UK	6	3
Total			**35**	**32**
Average			**5**	**5**

Table 2: Centres of Expertise and participants of the POLKA Delphi study face-to-face meetings

At the end of each face-to-face meeting, contact details of 2 volunteers (1 HCP and 1 patient or patient representative) were requested with whom the final report could be shared for them to comment on (to be done by September 2011).

DISCUSSIONS

Policy area discussions

Below we outline the 12 policy areas surveyed online and discussed in the meetings. Patient fear was brought up as a possible limiting factor in these discussions (i.e., biting the hand that feeds you), but having external moderators allowed the conversations to be very open and flowing. We would like to raise the caveat that the patients who attended the meetings were possibly more verbose and comfortable being verbose than those who did not attend. Also, FRAMBU (Norway) and Agrenska (Sweden) were mentioned a number of times as model centres of rare disease patient and family centres.

The patient-friendliness and care provided at the Centre

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[16] It should be noted that the Centres of Expertise listed are specialised health services within hospitals.

“The sum of many small details eventually makes a great difference”.

In the online survey, there was a small but steady number of patients who either did not feel welcome, or at best “neutral” at the Centre. When we tried to tease this out face-to-face about what may be some ingredients that make patients uncomfortable, some specifics came up. One was that staff should remember how to listen, in other words, not be impatient or dismissive when listening. “Continuity in care” came up as very important, including a dedicated staff person(s), letting us know that non-continuity may be a weakness. Waiting rooms are generally overcrowded and noisy and hard to be in for long times. Many people need to spend the whole day for their multi-disciplinary visit, so the clinic and waiting room should be as organised and stress-free as possible – “airy, light and modern”, with toys. Food and drink should be provided. When a patient is visiting the centre once a year, re-arrangement of the hospital facilities can be frequent, with different rooms every time. A “conductor” who can take you through the day and help navigate the 5 or 6 visits would be ideal. This could be a staff person or a dedicated patient representative who could also introduce them to others with the same or similar condition. There could be a brochure especially for new patients outlining the services. Ideally, the clinic should be a one-stop shop. Which means that patients and family spend a lot of time there seeing the various HCPs. Respectful staffs are greatly appreciated.

Some patients mentioned that centres need to be accessible to those with special needs and many times are not. There are often no electronic calling systems in waiting areas or appropriately marked (via Braille) doors to clinic rooms. Hospital complexes are generally very big and inadequately sign-posted with expensive parking. Sometimes staff, especially nurses, is not understanding or sensitive enough. *We are “spoken to abruptly”, or we are “not spoken to”, there is “talking amongst consultants as if you are not in the room”.* Some patients have hearing difficulties, and still are called by their names in the waiting room, which can create frustration for both the patient and the health care professional. Throughout this part of the discussion came many positive aspects that need to be underlined. When the staff is extremely understanding and caring, it helps tremendously. A reception team that treats you personally, i.e., by name and not number, is highly appreciated. Some pointed out how important it is to be seen and treated as a whole human being and family instead of a “crooked back and legs”. Staff competence is important, and a “good staff gave (patients) peace of mind”, including being available to answer questions. An understanding nurse helps everything go smoother. Patients think it important for a Centre to provide a diagnosis, follow-up, advice about the present situation and prognosis of the condition.

Possibly because HCPs spend so much time in them, they were willing to design a unit! Of course, patients should be on the design team as well - the ideal environment for a centre would be a unit which is located within a hospital but not “over-medicalised”. This means that the patient is not saturated with information and by systems that are excessively “medical” (i.e., nomenclature used), where they are marginalised and not allowed to feel like an active participant. It in no way implies or should imply a less-medicalisation of the unit, rather simply designed in such a way as to include patients and families in the logic of the unit. It should be designed keeping in mind both the patient and the HCP needs and have all the necessary facilities within or close to it. A purpose-built facility would be ideal but having a section of the outpatient department would be a less expensive and more convenient option which would help in providing multidisciplinary care to rare disease patients with room for family and enablers.

Easy access to a Centre

A red flag, a passport, a special card…

One large issue that everyone agreed on is that funding for travel and accommodation for patients and carers travelling long distances should always be provided: *“Suffering from fatigue I couldn't make the long journey to Newcastle this year”, “I would like more support available nearer to home”, “We need the NCG to pay for travel and accommodation for attendance from afar and not depend on the support group”*; regarding lodging, *“there is a hotel for parents but no provision in the room for our baby, which is very difficult for our daughter who is small. So we sleep on the floor or sitting on a chair next to her.”*

A significant minority of professionals would like to see their patients more frequently (from 19% in England to 40% in DK), depending on the severity of the condition. A large proportion of patients also would like to be seen more or answered that they don’t know if they need to (again, higher in DK than England), which may be considered a level of worry. One widespread policy is the exchange of telephone numbers or direct email addresses so a centre can get in touch and call a patient in as needed (for specialist care, etc.) which gives the centre more opportunity to coordinate individual appointments and can serve more patients. They also can be in easier touch with patients if the patient finds themselves in an Emergency department. Perhaps an updateable booklet on who is available with contact information. It was suggested that a ‘main’ centre of expertise for a disease or a group of diseases should be established with ‘local’ outreach clinics depending on the need. Currently some centres provide outreach services or are in the process of developing them, for example, nurses who travel to homes. It must be noted that having outreach services does not eliminate all travel to the main centre although it should be less. Retreats where both patients, families, enablers and professionals are invited and come are excellent ways of firming up the bonds via a less medicalised environment. This presents an excellent opportunity of sharing information.

Participants, based on their experiences, expressed the importance of having emergency contact details of on-call HCPs at centres in the form of a plastic card accompanied by accessible patient records as there have been many situations when patients need urgent attention at a local Emergency but the doctors on duty do not understand the complexity of the condition or otherwise do not fully listen to them or their carers. Some Centres have already designed and begun distributing such cards, a red flag of some sort (i.e., medAlert bracelets) and handheld patient records (i.e., a wearable USB stick that includes a clearly worded consult letter) to patients. It is considered that a plastic card with ‘do’s and don’ts’, including contact information of clinicians at the Centre, could help emergency staff with limited knowledge of the disease to appropriately triage patients. Ambulance drivers also need to have regular brush up courses on codes of conduct depending on the disease.

Most centres do not have a specialist on site 24/7 but do have one on call. The problem is very few of the patients know that; even some professionals are not sure!

While HCPs agreed that they could see non-national patients, it rarely happens. This may be partly an issue of the mobility of patients. As for referring patients to other Centres cross-border, again most centres agreed they could, but the need is rare – few people move or even go on vacation when they are in need of treatment. Many professionals also agreed that their centre was the best available and were not interested in referring to a suboptimal centre with lower standards. If people could pay (or their governments reimburse), they would have no problem in seeing more non-national patients.

At the same time, it is important to share knowledge and experiences with other centres in Europe and around the world. It is essential for each country to have their own centre of expertise, as it can be difficult for patients to travel abroad on a regular basis. This morphs into a wish or interest in generating international education that would be one or two times a year to bring experts in other places up to speed. Methods of interchanging information with non-national professionals may include Skype, e-Communication, telemedicine, as well as direct hands-on training.

Coordination and cooperation within and outside of the Centre

"The same law needs to be applied the same for everyone everywhere."

HCPs generally think the exchange of information both within the Centre and with other professionals is good to excellent. Patients agreed up to a point but shared many experiences of how it could be better, especially the communication with those outside of the centre. HCPs agreed that this could be and needs to be improved.

Patients agree that healthcare professionals in the centre responsible for their care are both knowledgeable and understanding; while 13-15% said they were only knowledgeable.

The patients say the Centre opens doors in other parts of the health care system as quick diagnosis is important. From their experiences they felt a sense of security after living through a long time of not knowing what was wrong. Some patients found that it took years (up to 8-10 years) before the correct diagnosis was made and adequate information shared.

Patients expressed concerns on the lack of coordination and cooperation of local healthcare facilities with centres of expertise and patients. A frequent story was the importance of referral for gene testing for diagnosis. When people rely on a GP with little to no experience and who may be doubtful, they may wait years before getting the genetic testing. It is not clear that GPs even know of the existence of the Centre, never mind the disease. More communication (via website, etc.) is needed, which can be time-consuming for the Centre. It can be difficult to get GPs to prescribe certain medications or devices due their costs or because the drugs and devices are unlicensed for the condition they are prescribed, or again, due to a lack of intimate knowledge of the condition.

Some centres overcome this by supplying them directly if funding allows them to do so. The Centre needs to be the central axis in all this to standardize care via a National or International Protocol. This protocol would include flow charts of who to call in case of doubt or contrary opinions. Once again, a memory stick maintained by the patient would help.

Some health care professionals receive calls from external HCPs daily. This represents a significant amount of time and may become a difficulty.

Results of testing or interdisciplinary collaboration sometimes can also take “a long while” in the patient’s eyes, and whoever’s responsibility that is, the timeline is not clearly outlined from the beginning. Patients do not know who to call to get clarification when these things happen. Even email communication would be appreciated.

Coordination letter with other HCPs (records and updates) are a problem as they involve many specialities. *“Patients should automatically be sent appointments instead of having to chase them up all the time”, (frequent) “delays in getting coordination letters posted”, “local appointments [the]y can be sporadic and (it) also ends up (with) the patient/mother chasing and ensure/trying to ensure that notes are passed on to a range of other care professionals”.* Simplified coordination letter with other HCPs should be used. Most centres do send coordination letter with other HCPs to GPs in paper format. Although that often functions well, at times it causes delays (more than 2 weeks). The letters need to be written in understandable language for the patient, which would make it more accessible to the GPs and other external consultants as well. Setting up IT systems designed so data can be shared instantly with healthcare facilities across the country was suggested by many participants. Another option for timely data exchange could be the setting up of web-based databases holding patient records which could be accessed by HCPs across the country. This would also save money on duplication of laboratory tests and save patients distress and time. Of course, this needs to be a secure system that follows the Good Practice guidelines. Some centres are linking up with local specialists caring for patients.

The transition from paediatrics to adult care

“I’m blind. If I have to stay the night, the layout of the room should make sense.”

Transition of care from childhood to adulthood was generally an unknown. And the more one knows about it, the less satisfactory it is. HCPs were generally more negative than patients, who maybe have not been through it yet, so were more likely to express an “I don’t know” answer. The transfer of records seemed even less smooth than the human transfer. Again, this may be a question of data transfer and incompatibility of systems, but this should be very adaptable.

Some centres of expertise have a transition care pathway which works well. Dedicated adult and paediatric nurses can carry out the transition on multiple home visits. There were suggestions of having a dedicated transition coordinator with knowledge of the disease who can act as a link to the community. At one centre community nurses are available for family as well. Patient organisations can help maintain consistency, even while maintaining confidential the coordination letter with other HCPs, etc., during the transition, and queries to or from GPs. The transition care pathway should be designed such that it includes developing skills the patients might need in the future due to the progression of the condition, such as learning sign language before the onset of deafness, or braille before becoming blind, etc. A clinical psychologist is needed for psychometrics and psychotherapy for the family as well as the patient, who also accompanies the patient through the transition. Patient groups can work with HCPs and GPs on appropriate methods of communication.

In some centres, the transition starts being discussed early, when the child is 11-12, and involves the parents. The decision to leave the paediatric unit for the adult one should not be arbitrary according to a birthdate, but depending on whether or not the patient feels ready for it.

Patient autonomy and patient representativity in the Centre

“As a carer of John for all his life, I will not be told to sit down over there and wait as if I am not part of the solution.”

Almost all of the HCPs said they always or almost always took patients’ opinions into account when deciding their treatment. Patients felt similarly. *“Leaving (the hospital) I gave a sample to a medical analysis laboratory. Result: the doctor denied that the infection was real, and I actually had a double infection, fungal and bacterial! This added to the contradictions between words and actions and the* *denial of much of what I experienced does not help my belief or hope for an improvement in the medical care of my problems."*

While 80% of HCPs said the centre supports patient representatives, a large amount of patients had no idea (between 45% and 62%). All participants agreed that contact with the patient organisation at or through the Centre is important since from there you get good medical information, as well as about social help and about experiences of other patients. *"Increased patient involvement is very important, facilitating patient support group is necessary".* Patient organisations play an important role in providing patients information at the correct education level and age level in the correct formats (social media for teenagers), helping with social care applications, having a social care 'connector', meeting with first-time or newer patients, working with the family as well as enablers, speaking with HCPs on behalf of patients as well as in some cases funding research, hence it is crucial to have a patient organisation functioning in every centre as a partner. If/when necessary, there can even be confidentiality agreements with the PO in order to safeguard patient data or Centre autonomy.

Families with fewer resources need extra help to get the right treatment and services. Patient organisations help in the orientation period.

Provision of information to patients, their families and their enablers

"As a patient who has been here for many years, I would gladly be in a phonebook to help any new patients get accustomed."

74-100% of HCPs said they always provide patients with information on the condition upon diagnosis. 67-69% of patients agreed. This speaks to the issue of when is the right time to give the information, and in what format, by whom and to whom? *"At first we need to digest the news of the disease. We are not able to assimilate more information right then. It takes a second interview later to be able to ask the right questions and receive answers."*

Participants agreed that concrete and reliable information needs to be shared (on diagnosis and prognosis) when the patient and family are prepared, which may be a question of how much, when, how often, by whom [outreach professionals (i.e., at home)], in what formats, all in order to help avoid misunderstandings or even denial. And if HCPs do not have the answer, they should impart that – knowing that there is a lack of accumulated knowledge on this rare disease would be helpful to the patient and family. And patients and their families need to know and understand that they can ask questions when they are ready (by telephone, email, etc.). Patient organisation websites and newsletters provide information on the condition and the contact information of the centre for patients and family. These can be introduced by the outreach worker or the sharer of the diagnostic information, etc. Or they can be part of the diagnosis (being present). They can set up meetings with other patients or parents. For young people, a community youth worker to act as a link may work. There is a lack of appropriate information resources for adolescents/teenagers; these need to be developed with teenagers. Most patients would like to meet similar patients to share experiences, and this happens usually via the patient organisation. Centres can help arrange this if patients feel it is helpful, and a more informal communication between patients via Facebook, etc. Again, availability of follow-up by psychologists would be useful.

The "patient not being ready to accept information", "Patient/parents may not speak English", "Blindness/Deafness" is a concern, there is "no accessible written information for children or people with learning difficulties/ Literacy" are typical problems in conveying information. A course on communication strategies would be helpful for all professionals. Patients also need to learn how to communicate more effectively with the health professionals, including those who can teach professionals on issues that may be considered more social. HCPs showed great interest in these types of patients and POs.

Barriers due to cultural issues are a problem. Barriers due to cultural issues can be a problem. It was agreed that staff should receive repeated diversity training to maintain a sense of connection with patients. Interpreters need to be used when needed.

Information days are very successful. Also, care days outside the centre (summer meetings in a picnic setting, etc.) are opportunities for teaching as well as social bonding. The professionals also get to see patients in a more real-life setting. Summer camps/getaways may be a congenial place for people to assimilate and understand certain information on a more profound level. Oftentimes, the patient and family may feel overwhelmed by Information at the centre, and then return home to a relative vacuum.

Raising awareness among HCPs and non-HCPs

“For the good of the patient, we cannot continue to work in silos.”

HCPs said they receive rare disease enquiries frequently (weekly & daily, some only monthly) from other healthcare professionals as well as from non-HCPs like schoolteachers or employers.

Most all of the HCPs said their centre provides training on rare diseases it specialises in for other healthcare professionals (including European and other international HCPs). It may be worthwhile considering non-HCPs also. Some centres have specialist nurses and community nurses to do local or national education at schools for example.

All of the HCPs said their centre develops and disseminates guidelines on care to individuals and organisations. GPs should be provided with information in the form of care guidelines, websites and Centre of Expertise contact details. Some centres had training days for community nurses, hospital nurses, school nurses, dieticians and physiotherapists.

Most patients reported that it was difficult to get a referral to the centre since it was thought (by GPs and/or specialists at other hospitals) that nothing was wrong or that it was not necessary to be forwarded to the centre. Many times the local level of the health care system feels capable of performing the treatment, even without all the correct competences. It is important to revisit the speciality training curriculum for GPs, and other specialists in order to help in early detection and diagnosis. The need for GPs to understand the importance of referring patients with no diagnosis for genetic testing was also highlighted. High risk populations, such as Asians, need to be screened appropriately. Some patients do have quick diagnosis and referral to the centre, especially if the symptoms/handicap of the patient were visible. This raises concerns on the knowledge of GPs about rare diseases.

Patient-oriented and multidisciplinary care

“A conductor in partnership with the patient organisation can be a lifeline.”

HCPs said their centre provides care that balances all the needs of the patient and their families always, almost always or sometimes. A large amount of patients agreed. The patients say the centre is very good in terms of taking care of the patients' family needs in a comprehensive way. However, there are issues where often the family may not recognise their own needs. Patient groups again can be partners to help offer greater and more efficient social support. *"I'm not being followed in a specialist centre, but I would like to be followed … where a service is dedicated to my illness. I find the procedures now complex, with long waiting times, which costs a lot (I am a student) and is not practical because the rheumatologist is not familiar with the disease, and I need to be followed by different specialists (dermatologist, a psychologist ...) I would rather go to a place where everyone comes together and consults."*

HCPs said their centre provides care and treatment to respond to the multidisciplinary needs of patients generally. The vast majority of patients agreed with a notable "sometimes to almost never" group.

Both HCPs and patients highlight the importance of having a psychologist as part of the service for patients as well as carers and family, making home visits or having a consult outside the framework of the centre, in an autonomous locale. A psychological service needs to be integrated and funded for issues like pain, disfigurement, sexuality, body image, etc. Psychology, podiatry and occupational therapy are typically understaffed areas.

Social care assistance at the Centre

"There are few quick fixes. An on-site social worker is one of them."

Dedicated social workers in general are not available at centres. Adequate access to them is minimal. Although sometimes there may be "links" to social workers, for the most part patients (up to 92%) did not know if there was access within the centre or how to go about finding one.

A social care counsellor who knew the rules at the centre would be very helpful. Many cases of social work breaking down due to inadequate understanding of the disease were thought to be resolvable by having a social contactor on site. Most HCPs agreed that it is high on the wish list to get a social care counsellor or manager for the centre, not only for the patients but also for the staff who need professional knowledge about issues in social care support for rare diseases. Often doctors and nurses spend time helping patients by contacting social services and providing supporting documents (which are often ignored). It was also thought that social care counsellors of the municipalities would benefit from contact to a fellow social counsellor with special knowledge about rare diseases to process applications. This could avoid frustration as applications are rejected often because the civil servants who processed them did not have an understanding of their conditions. Patients can often secure social care via patient organisations.

Diagnosis facilities at the Centre

"As long as we have access to one, it does not have to be in the hospital".

Most of the centres had the necessary laboratory facilities to provide diagnosis for rare diseases they specialise in. If there are no facilities on site, they have the possibility of sending the test out including to international laboratories. Some centres have quarterly or regular video conferences with laboratories.

Performance management (and patient input into that) of the Centre

"There are many unknowns we don't know about."

Overall, there are no comments box at centres, or at very few. Most patients did not have known the answer to this question.

A satisfaction survey is carried out at some centres, but not all. Again, many patients had no idea.
Few if any patient representatives are included in the centre's governing body. Few professionals know, almost no patients know. Again, beyond whatever the answer is, if people do not know, it cannot be of use.

Although patients do not need to fill out forms as part of the evaluation process, they can speak up if there is a need for something; it must be emphasized that criticisms or even needs are often easier to generate anonymously via a public suggestion box. Patients are encouraged to give feedback, although many may feel reticent to do so. Dialogue in the consultation is essential and meetings such as the Delphi face-to-face meeting are considered important. The Centre works for patients, they need to be considered a partner.

It is difficult to measure satisfaction of patients via surveys or suggestion boxes. Patient organisations again can play an important role in sharing patients' opinions to help improve patient experience and care. The Centres have close links with the patient organisations and have regular meetings to share information and discuss issues. The Centre can arrange regular consultation meetings to get feedback in a non-threatening environment, i.e., an electronic monitor to answer 20 questions, for example, for which patients receive reimbursement for their time, moderated by an outsider.

Research at the Centre

"Neurologists, dermatologists, geneticists, dentists, physiotherapists, opthamologists, psychologists, all at your fingertips."

While most all professionals answered that their Centre carried out some sort of research, only a fraction said that the information garnered was shared with other stakeholders beyond journals. Anywhere from only a quarter to up to two-thirds of patients were able to answer about clinical research, but again very few knew if information was shared.

Some HCPs say their funding agency does not encourage research while others say it is encouraged. All think research is necessary and intrinsic and should be a formal part of the clinic. Patient organisations often provide information on research studies to patients and fund some of the research done by the centre. They can help in instigating research, forming CABs and sharing results. One reason information seems not to be shared is that patients do not understand the time required to complete a research study. All research issues, including timeline on when information will be shared, needs to be clarified with patients from the start (in the Informed Consent, etc).

SPECIFIC RECOMMENDATIONS TO BE IMPLEMENTED:

- Waiting rooms should be as organised and stress-free as possible – "airy, light and modern", with toys when appropriate. Food and drink should be provided.
- A patient conductor can help the patient go through the day.
- Information, help and signs should be available in Braille and for people with hearing problems.
- Staff should maintain an understanding attitude.
- There need to be travel and accommodation stipends.
- Outreach services (visiting nurses) and outreach centres (to save on travel) should be employed.
- A card, or passport, or memory stick bracelet – can help with lack of coordination.
- Patient group retreats, with Centre staff invited, for bonding and education.
- International expert education - Skype, e-Communication, telemedicine, as well as direct hands-on training.
- A clear explanation of who is on call and when and the appropriate phone numbers/emails.
- GP education, including flow charts, of who people can be referred to and when (a National Protocol devised by Centres of Expertise).
- Coordination letters with other HCPs easily and quickly available.
- Transition care pathways need to be established (nurses, psychologists, etc.). POs can help.
- Other specialists need to be involved (autonomous psychological services for family and enablers also). Podiatry and occupational therapy are other typically understaffed areas.
- Social service workers in the centre who know the disease and the system. POs can help.
- Information not only has to be given, but has to be given in the right format at the right time in the right way. POs can help with website and patient-derived material. Information days at the centre and outside.
- Trainings for non-specialists and for specialists, local and international.
- Patients need to be part of the governing body and the evaluation process (satisfaction measures via a third party) and they have to know about it.
- Research is vital for rare diseases and should be encouraged. Patients need to be involved as advisors.

DELPHI QUESTIONNAIRE IN FRANCE

In France, patients and their representatives only participated in the first and third rounds of the process (the online questionnaires). Those who expressed the wish to comment on the results were not invited to share their comments in meetings (see explanation below), unlike in England or Denmark.

Together with the French Alliance, we identified 17 centres and patients' organisations that were more likely to participate. For each organisation, a contact person was identified. The role of this contact person was to involve its organisation both in the questionnaire ('membership), and with the organisation of a meeting with their health care professionals.

REASONS WHY NO MEETINGS COULD BE ORGANISED IN FRANCE

- No intervention from a representative of the Ministry of Health or HAS to propose the initiative
- Centres of Expertise already participate in 4 types of evaluation: from the hospital, from the HAS/Comité de Labellisation, from the European Reference Network, from the CNAM… and all those take place from March to July. This would have been a fifth evaluation exercise and it did not seem to be the right moment. Nevertheless, some leaders expressed the desire to work with us in the future, maybe merging some questions from other evaluations with ours.
- Understaffed and sick leave left the head of centre with no resources to add more work
- Shyness of patients' representatives vis-à-vis their health care professionals
- No preparation or early contact with centres by POLKA coordinator

PATIENTS' GROUPS CONTACTED

Association Française des Syndromes d'Ehlers-Danlos
Association Française du syndrome de Marfan
Association Française du syndrome d'Ondine
Association contre les Maladies Mitochondriales
Association des patients de la Maladie de Fabry
Association Française de l'Ostéogénèse Imparfaite
Association Française du Syndrome de Rett
Association Keratocône
Association pour l'Information et la Recherche sur le Syndrome de Sapho
Génération 22
Hypophosphatasie Europe
HTAP France
Association Surrénales
Association Française pour le Syndrome de Gilles de la Tourette
Association PSP France
Association des POIC
Association Française ataxie de Friedreich

QUESTIONNAIRE: 58 RESPONSES RECEIVED

Diseases	Number of responses
Sapho syndrome	6
Marfan syndrome	6
Gilles de la Tourette	2
Ondine syndrome	2
Congenital adrenal hyperplasia	12
Primary pulmonary hypertension	7
Fabry disease	4
Mitochondrial diseases	2
22q11	2
Progressive supranuclear palsy	2
Addison, Cushing	4
Friedreich’s Ataxia	1
Corticotroph pituitary deficiency	3
Craniopharyngioma	0
Pheochromocytoma	1
Blank/other	4
Total	58

RESULTS

Some responses from France differed slightly from the other countries:

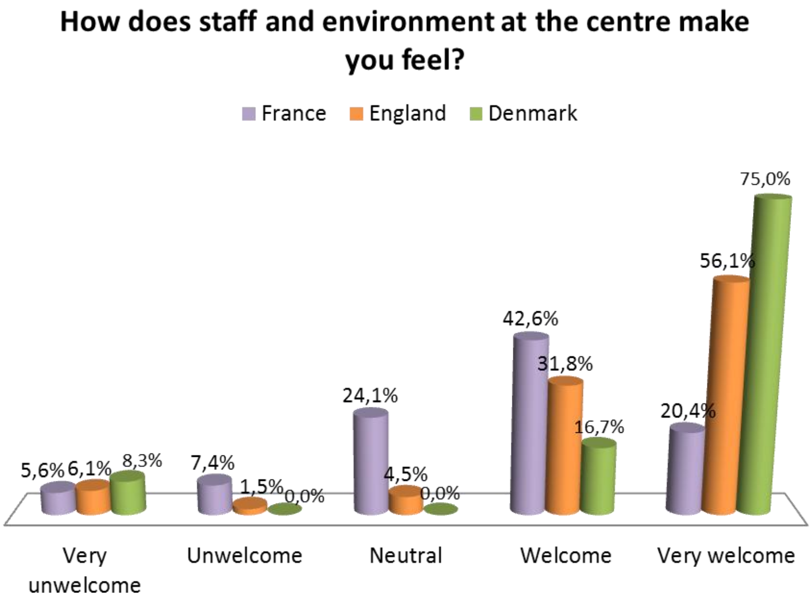

FIGURE 2

Based on a total of 134 responses, the 58 French respondents less often replied they felt very welcome than in England or Denmark. In general they felt welcome (42.6%) (Figure 2).

Similarly, French respondents declared more often difficulties in seeing their health care professionals as often as they would have wished to: one third responded “no” (Figure 3).

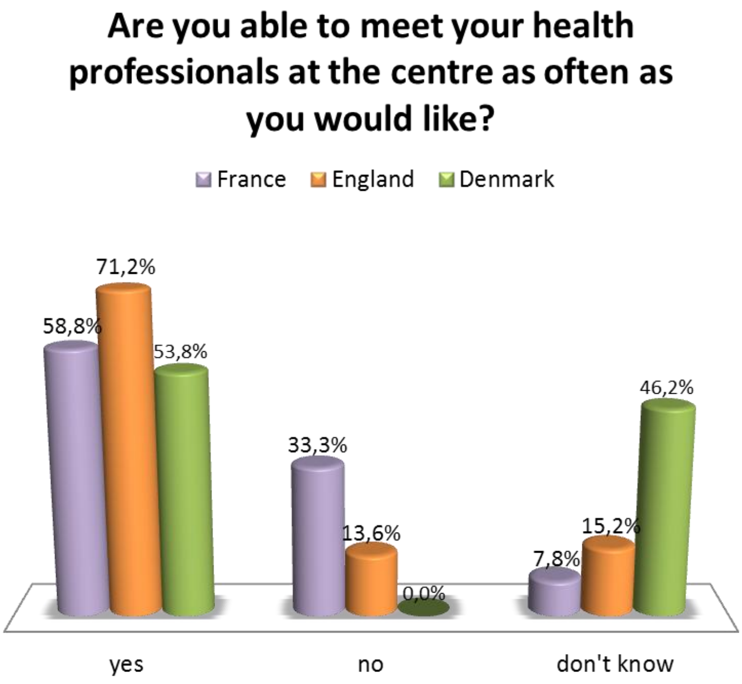

FIGURE 3

About emergency care, French respondents seemed to be more aware of the existence of an emergency care unit in their centre (Figure 4):

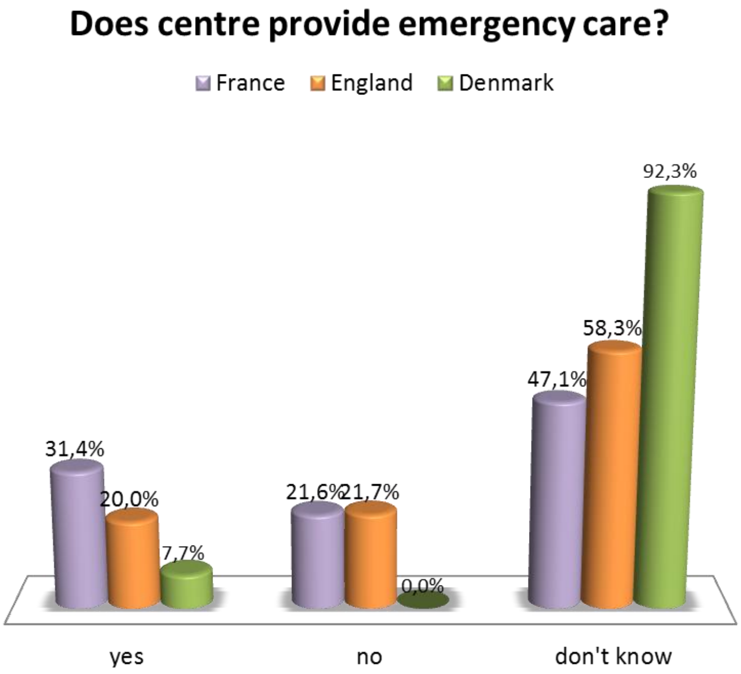

FIGURE 4

Regarding cooperation and collaboration with other HCPs both in the centre and outside the centre, French respondents were less satisfied than in England or Denmark (Figures 5 and 6):

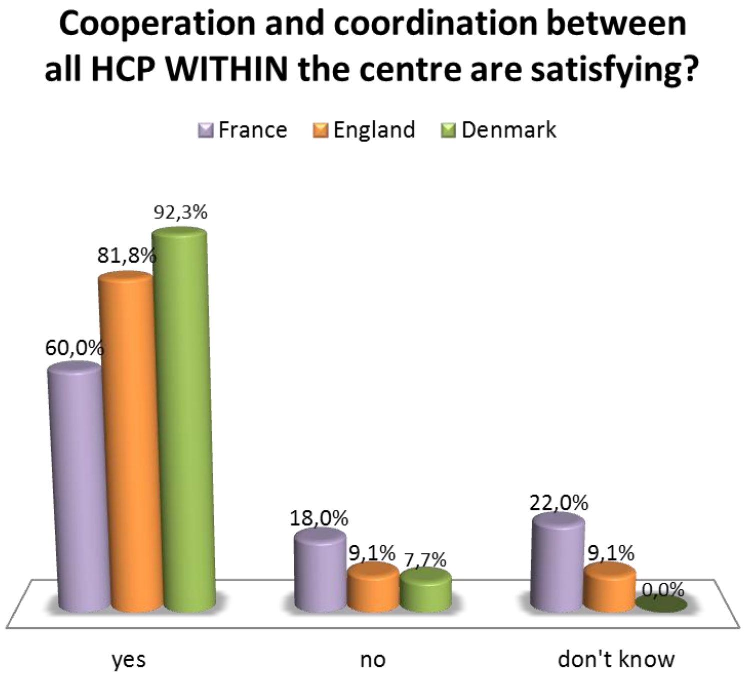

FIGURE 5

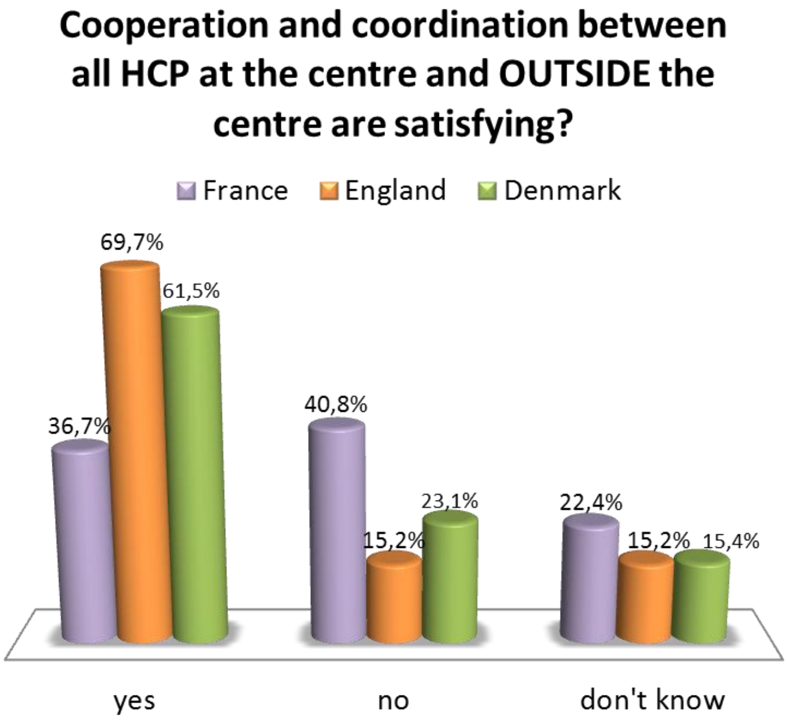

FIGURE 6

When asked whether their opinion is considered when deciding on treatment, Frnech respondents were again less satisfied, with 38% reporting their opinion to be rarely considered (sometimes, almost never or never) (Figure 7).

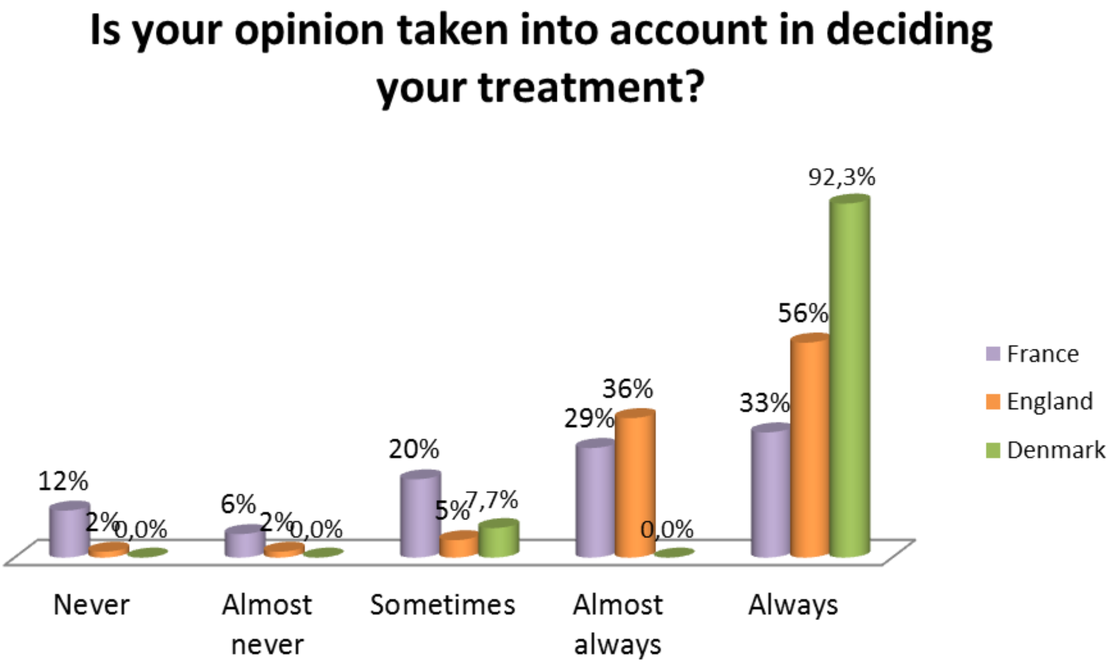

FIGURE 7

Otherwise, for all other questions, responses from France were in line with those from England and Denmark.

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APPENDICES

APPENDIX 1 ROUND 1 RESULTS: DENMARK (PRELIMINARY RESULTS)

A summary of the Round 1 replies by HCPs and patients from Denmark by policy area.

i. *Patients and the Care Environment at the Centre of Excellence*

HCPs

- All of the HCPs felt the staff and environment at their CoEs make patients feel welcome or very welcome.

Patients

- 92% of the patients felt welcomed by the staff and environment at their centres while 8% felt staff made them feel unwelcome or very unwelcome.

ii. *Access to the Centre of Excellence*

HCPs

- 3/5 of the HCPs said they were able to see their patients as often as they wanted while 40% said they would like to see them more often.
- All of the HCPs said there was no specialist available on site 24/7. 2/5 said a specialist was available on call 24/7 while 3/5 said no specialist was available on call 24/7 .
- All HCPs said they never or almost never provided treatment and care to other (non-Danish) European patients.
- 2/5 HCPs said they were able to almost always or always refer patients to other specialist centres in Europe. 2/5 said they were able to refer sometimes while 1 said never or almost never.

Patients

- 54% of the patients said they were seen as often as they wanted to be seen by HCPs. 46% replied they do not know if they are seen as often as they would like to be seen.
- 93% and 77% patients said they did not know if a specialist was available 24/7 on site or 24/7 on call, respectively, while 8% and 23% said a specialist was available 24/7 on site or 24/7 on call, respectively.

iii. *Coordination and cooperation within and outside the Centre of Expertise*

HCPs

- All of the HCPs said the cooperation and coordination between healthcare professionals within the centre is satisfactory.

- All of the HCPs said the cooperation and coordination between healthcare professionals outside the centre is satisfactory.
- 3/5 of the HCPs said they think patient information is exchanged effectively between healthcare professionals within the centre. 40% said it was neither effective nor ineffective (neutral).
- 2/5 of HCPs said that they think patient information was exchanged neither effectively nor ineffectively (neutral) with HCPs outside the centre. 1 said it was effective while 1 other said it was ineffective.

Patients

- 93% of patients said they thought the cooperation and coordination between healthcare professionals within the centre is satisfactory. 8% said it is not satisfactory.
- 62% of patients said they thought the cooperation and coordination between healthcare professionals outside the centre is satisfactory. 23% said it was not satisfactory while 15% said they did not know.
- 75% of patients said patient information is exchanged effectively or very effectively between healthcare professionals within the centre. 17% were neutral while 8% said it was very ineffective.
- 46% of patients said patient information was exchanged neither effectively nor very effectively (neutral) HCPs outside the centre. 46% said it was effective or very effective while 8% patients said it was ineffective or very ineffective.
- 83% of patients said that healthcare professionals in the centre responsible for their care were both knowledgeable and understanding, while 15% said they were only knowledgeable.

iv. *Transition of care*

HCPs

- 4/5 of the HCPs said the transition of care from childhood to adulthood was not satisfactory while 1 said it was satisfactory for patients. 1 said it was satisfactory for HCPs, 3 said it was not satisfactory and 1 said they did not know. 3/5 HCPs felt transition of health records was satisfactory, 2 said it was not.

Patients

- The majority of patients said transition of care from paediatric care to adult care was satisfactory for patients and healthcare professionals. 41%, 38% and 41% said it was satisfactory for patients, HCPs and hospital records, respectively. While 52%, 57% and 57% did not know for patients, HCPs and hospital records, respectively.

At the meetings, it occurred that most of the families had small children and therefore no experience with the transition from childhood to adulthood.

v. *Patient Autonomy, Patient representatives and support groups at the Centre*

HCPs

- All of the HCPs said they always or almost always took patients' opinions into account when deciding their treatment.
- 4/5 of the HCPs said the centre supports the patient representatives. 1 said the centre does not support the patient representatives.

Patients

- 93% of patients said their opinions were always or almost always taken into account when deciding treatment. 8% said their opinions were neither taken into account nor disregarded (neutral).
- 31% of the patients said the centre supported activities of patient representatives while 8% said the no it does not support activates of patients. 62% said they did not know.

vi. Provision of rare disease information to patients

HCPs

- All HCPs said they always provided patients with information on the condition upon diagnosis.

Patients

- 69% of patients said they were provided with information about their condition upon diagnosis. 8% said they were not provided with any information while another 5% said they do not know. 8% replied the question is not applicable.

vii. Raising awareness and training amongst healthcare and non-healthcare professionals

HCPs

- 3/4 of the HCPs said they receive rare disease enquiries frequently (weekly & daily) from other healthcare professionals. 1 said they received queries monthly.
- 2/5 of the HCPs said they receive rare disease enquiries monthly and another 2 said they received enquiries weekly from non-healthcare professionals. 1 said they receive enquiries annually.
- All of the HCPs said their centre provides training on rare diseases it specialises in for other healthcare professionals.
- All of the HCPs said their centre develops and disseminates guidelines on care to individuals and organisations.

viii. Patient-oriented and multidisciplinary care at the Centre of Expertise

HCPs

- 3/5 of the HCPs said their centre provides care that balances all the needs of the patient and their families always or almost always; 1 said sometimes and 1 said never.
- 3/5 of the HCPs said their centre provides care and treatment to respond to the multidisciplinary needs of patients almost always, 1 said sometimes and 1 said never.

Patients

- 83% of patients said their centre provides care that balances all their families' needs always or almost always, 8% said sometimes while 8% said almost never.
- 68% of patients said their centre always or almost always provides care and treatment which responds to the multidisciplinary needs of patients, 25% said sometimes while 16% said never or almost never.

ix. *Social care assistance at the Centre of Expertise*

HCPs

- All HCPs said a dedicated social worker did not exist at the centre.
- 1/5 said there was adequate access to a social worker at the centre while 4 said there was not adequate access.
- 4/5 of the HCPs said there were adequate links between social workers and HCPs while 1 said there was no adequate access.

Patients

- 66% of patients said they did not know if a dedicated social worker existed at the centres and 33% said no social worker existed.
- 92% of patients said they did not know if there was adequate access to a dedicated social worker and 8% said there was not.
- 68% of patients said they do not know if there were adequate links between social workers and HCPs, 17% said yes there were while 17% said there were not.

x. *Facilities for diagnosis of rare disease at the Centre of Expertise*

HCPs

- 88% XX of HCPs said the centre had facilities to provide accurate diagnosis for some or all of the rare diseases they specialise in.

xi. *Performance management of the Centre of Expertise*

HCPs

- All HCPs said there was no comments box at the centre.

- 1/4 of the HCPs said a satisfaction survey was carried out at the centre, 2 said it was not carried out and 1 said they do not know.
- All HCPs said patient representatives were not included in the centre's governing body.
- All HCPs said a patient representative was not included in the centre's evaluation committee.

Patients

- All patients said they did not know if there was a comments box at the centre.
- 15% of patients said the centre carried out a satisfaction survey while 84% said they do not know.
- 100% of patients said they did not know if patient representatives were included in the centre's governing body.
- 100% of patients said they did not know if patient representatives were included in the centre's evaluation committee.

xii. Research at the Centre of Expertise

HCPs

- 4/5 of the HCPs said the centre carries out basic research, 1 said no.
- All HCPs said the centre carried out clinical research.
- 4/5 HCPs said the centre carried out clinical trials of medicinal products, while 1 said they did not know.
- All HCPs said the centre carried out translational research.
- All HCPs said the centre has a research infrastructure.
- 1/5 of the HCPs said research carried out at the centre was always shared with other stakeholders beyond journal publications. 4 said it was shared sometimes.

Patients

- 23% of patients said the centre carried out clinical research and 77% said they do not know.
- 23% of patients said the centre carried out clinical research and 77% said they do not know.
- 15% of patients said the centre carried out clinical trials of medicinal products and 85% said they do not know.
- 15% of patients said the centre carried out translational research and 85% said they do not know.
- 23% of patients said the centre has a research infrastructure and 77% said they do not know.
- 92% of patients said they do not know if research carried out at the centre was shared with other stakeholders beyond journal publications. 8% said it was shared sometimes.

APPENDIX 2. ROUND 1 RESULTS: ENGLAND (PRELIMINARY RESULTS)

A summary of the Round 1 replies by HCPs and patients from England by policy area:

i. *Patient and Care Environment at the Centre of Expertise*

HCPs

- 89% of HCPs felt the staff and environment at their centres make patients feel welcome or very welcome while some (11%) were neutral. 9% said staff and/or the environment makes patients feel unwelcome. XX

Patients

- 87% of patients felt welcome by the staff and environment while some (4%) were neutral. 8% said staff and/or the environment makes them feel unwelcome or very unwelcome.

ii. *Access to the Centre of Expertise*

HCPs

- 81% of HCPs said they were able to see their patients as often as they wanted, while some (16%) said they would like to see them more often.
- 56% of HCPs said there was no specialist available on site 24/7, however 54% said a specialist was available on call 24/7. 28% and 22% of HCPs did not know if a specialist was available 24/7 on site or 24/7 on call, respectively. 24% of HCPs did not know if a specialist was available on call, while only 17% said a specialist was available 24/7 on site.
- 54% of HCPs said they provided treatment and care to other (non-UK) European patients almost always, while 35% said sometimes and 11% said never or almost never.
- 41% of HCPs said they were able to almost always or always refer patients to other specialist centres in Europe. The same proportion said they were able to refer sometimes, while 19% said never or almost never.

Patients

- 72% of the patients said they were seen as often as they liked by HCPs; however some (13%) said they would like to be seen more often.
- 58% and 42% of patients said they did not know if a specialist was available 24/7 on site or 24/7 on call respectively. 19% and 40% said a specialist was available 24/7 on site or 24/7 on call, respectively. 23% and 18% patients said no specialist was available 24/7 on site or 24/7 on call, respectively.

iii. *Coordination and cooperation within and outside the Centre of Expertise*

HCP

- 93% of HCPs said they thought the cooperation and coordination between healthcare professionals within the centre is satisfactory. 5% and 2% said it was not or they do not know, respectively.
- 65% of HCPs said they thought the cooperation and coordination between healthcare professionals outside the centre is satisfactory. 21% said not while 14% said they do not know.
- 89% of HCPs said they think patient information is exchanged effectively or very effectively between healthcare professionals within the centre. 9% were said it was neither effective nor ineffective (neutral) while 2% said it was very ineffective.
- 81% of HCPs said that they think patient information was exchanged effectively or very effectively with HCPs outside the centre.

Patients

- 82% of patients said they thought cooperation and coordination between healthcare professionals within the centre is satisfactory. 9% said it was not satisfactory and another 9% said they did not know.
- 69% of patients said they thought the cooperation and coordination between healthcare professionals outside the centre is satisfactory. 16% said no, it was not satisfactory while 15% said they didn't know.
- 75% of patients said patient information is exchanged effectively or very effectively between healthcare professionals within the centre. 21% said it was neither effective or ineffective (neutral) while 4% said it was ineffective or very ineffective.
- 34% of patients said patient information was exchanged neither effectively nor ineffectively (neutral) to HCPs outside the centre while 51% said it was effective or very effective. 14% said it was ineffective or very infective.
- 85% of patients said the HCPs at the centre responsible for their care were both knowledgeable and understanding while 13% said they were only knowledgeable and 1% said they were only understanding.

iv. *Transition of care*

HCPs

- 86%, 84% and 63% of HCPs said it was satisfactory for patients, HCPs and hospital records, respectively. 2%, 2% and 12% replied transition was not satisfactory for patients, HCPs and hospital records, respectively. 12%, 14% and 26% replied do not know if it is satisfactory for patients, HCPs and hospital records, respectively.

Patients

- 41%, 38% and 41% of patients said it was satisfactory for patients, HCPs and hospital records, respectively. 52%, 57% and 57% replied that they do not know if it is satisfactory for patients, HCPs and hospital records, respectively.

v. *Patient Autonomy, Patient representatives and Support Groups at Centre*

HCPs

- 98% of HCPs said their opinion was taken into account always or almost always when deciding on treatment. 3% said they neither take nor disregard patients' opinions.
- 81% of HCPs said the centre supported the patient representatives. 5% said not while 14% said they did not know.

Patients

- 94% of patients said their opinion was always or almost always taken into account when deciding their treatment. 4% said they neither take nor disregard patients' opinions. 2% said their opinion was never or almost never taken into account.
- 48% of patients said the centre supported activities of patient representatives while 7% said no and 45% said they did not know.

vi. *Provision of rare disease information to patients*

HCPs

- 74% of HCPs said they always provided patients with information of the condition upon diagnosis. 9% said they never provided patients with information while another 9% said sometimes. 7% said they did not know.

Patients

- 67% said they were provided with information of the condition upon diagnosis. 6% said they were not provided with information while another 6% said do not know. 21% replied the question is not applicable.

vii. *Raising awareness and training amongst healthcare and non-healthcare professionals*

HCPs

- 49% of HCPs said they receive rare disease enquiries frequently (weekly & daily) from other healthcare professionals. 33% said they received queries monthly and 19% yearly.
- 30% of HCPs said they receive rare disease enquiries monthly, 26% weekly and another 26% said they received enquiries yearly from non-healthcare professionals. 2% receive them daily while 16% said they do not receive them.
- 67% of HCPs said their centre provides training on rare diseases it specialises in for other healthcare professionals. 14% said no and 19% said do not know.
- 91% of HCPs said their centre develops and disseminates guidelines on care to individuals and organisations, 2% said they do not and 7% said they do not know.

viii. *Patient oriented and multidisciplinary care at Centres of Expertise*

HCPs

- 86% of HCPs said their centre always or almost always provides care that balances all the needs of the patient and their families while 14% said sometimes.

- 84% of HCPs said their centre provides care and treatment to respond to the multidisciplinary needs of patients and 14% said sometimes.

Patients

- 84% of patients said their centre always or almost always provides care that balances all their families' needs, 12% said sometimes while 4% said never or almost never.
- 84% of patients said their centre provides care and treatment to respond to the multidisciplinary needs of patients, 12% said sometimes while 4% said never or almost never.

ix. *Social care assistance at Centres of Expertise*

HCPs

- 36% of HCPs said a dedicated social worker existed at the centre while 40% said a social worker did not exist at the centre and 24% said they do not know.
- 37% said there was adequate access to a social worker at the centre, 34% said there is not adequate access to a social worker and 29% said they did not know.
- 53% of HCPs said there were adequate links between social workers and HCPs while 23% said there were not and 23% said they did not know.

Patients

- 47% of patients said they did not know if a dedicated social worker existed at the centres, 30% said one did exist and 23% said no one did not exist.
- 53% of patients said they do not know if there was adequate access to a dedicated social worker, 24% said yes there was adequate access to a dedicated social worker while 24% said no.
- 49% of patients said they do not know if there were adequate links between social workers and HCPs.

x. *Facilities for diagnosis of rare disease at Centres of Expertise*

HCPs

- 88% of HCPs said the centre had facilities to provide accurate diagnosis for some or all of the rare diseases they specialise in.

xi. *Performance management of the Centre of Expertise*

HCPs

- 44% of HCPs said there was a comments box at the centre, 39% said there was not and 17% said that they did not know.
- 86% of HCPs said a satisfaction survey was carried out at the centre, 5% said not and 9% said they do not know.
- 23% of HCPs said a patient representative was included in the centre's governing body, 26% said they were not included and 51% said they do not know.
- 28% of HCPs said a patient was included in the centre's evaluation committee, 21% said they were not and 51% said they did not know.

Patients

- 27% of patients said there was a comments box at the centre, 24% said there was no comments box and 45% said they did not know.
- 54% of patients said the centre carried out a satisfaction survey, 13% said they did not and 33% said they do not know.
- 21% of patients said a patient representative was included in the centre's governing body, 16% said they were not and 63% said they do not know.
- 13% of patients said a patient representative was included in the centre's evaluation committee, 16% said they were not while 71% said they did not know.

xii. *Research at the Centre of Expertise*

HCPs

- 67% of HCPs said the centre carries out clinical research, 5% said it does not and 5% said they do not know.
- 93% of HCPs said the centre carried out clinical research, 2% said not and 5% said they do not know.
- 73% of HCPs said the centre carried out clinical trials of medicinal products, 12% said it does not and 14% said they do not know.
- 48% of HCPs said the centre carried out translational research, 9% said it did not and 58% said they do not know.
- 59% of HCPs said the centre has a research infrastructure, 12% said it does not and 44% said they do not know.

- 50% of HCPs said research carried out at the centre was always shared with other stakeholders beyond journal publications, 36% said sometimes, 2% said never and 12% said they do not know.

Patients

- 63% of patients said the centre carried out clinical research and 37% said they do not know.
- 66% of patients said the centre carried out clinical research, 2% said it did not and 32% said they do not know.
- 59% of patients said the centre carried out clinical trials of medicinal products, 2% said it did not and 39% said they do not know.
- 18% of patients said the centre carried out translational research, 2% said it did not and 80% said they do not know.
- 31% of patients said the centre has a research infrastructure, 5% said it does not and 65% said they do not know.
- 19% patients said they do not know if research carried out at the centre was shared with other stakeholders beyond journal publications. 63% said they do not know and 18% said sometimes.

APPENDIX 3. ROUND 2 RESULTS: DENMARK

i. *Patient and Care Environment at CoE*

- Overall, the patients felt the centre functions well. They were happy with the way the reception functions.
- Some patients pointed out how important it is to be seen and treated as a whole human being and family instead of crooked back and legs.
- Patients also said how important staff competence was to them. They said having good staff gave them peace of mind. The competence of the centre staff was rated good. Patients also said staff should “know what we talk about”; it is also good that they arrange annual consultations; email replies would be/are appreciated.
- Patients think it important for a Centre to provide a diagnosis, follow-up, advice about the present situation and prognosis of the condition. Continuity in physician and nurse care is important.

ii. *Access to CoE*

- Patients said it is good to have nurses as gatekeepers at the centre. HCPs said all patients give a contact number to the nurses and the number can be shared with other professionals, e.g., a physiotherapist if needed - there are no fixed calling hours since that would mean serving fewer patients.
- The centre is very helpful in case of urgent treatment at A&E departments.

iii. *Coordination and cooperation within and outside centre of expertise*

- The patients say the centre pushes you forward when need and opens doors in other parts of the health care system.
- Some patients say quick diagnosis is important. From their experiences they felt a sense of security after living through a long time of not knowing what was wrong. Some patients found that it took years (up to 8-10 years) before the correct diagnosis was made and all information needed found by parents.
- The importance of referral for gene testing for diagnosis was highlighted. One family had a negative experience as they were not referred to a gene test, due to expense. The patient refused to take a cheaper test and thus has to wait a year before they got approval for the expensive test and referral to the centre.
- It was questioned if GPs were aware of the centre’s existence. HCPs say the centre has a website and tries to make their existence known through various sources. It was thought that paediatricians in general are aware of the centre.

- Patients said sometimes it takes a long while to get test results, which is not necessarily the centre's fault, it is usually due to other specialists. Interdisciplinary collaboration does not always function within and outside the centre, even if the centre sends reminders. Patients say it would be nice to know if there is a delay in test results and if they can call and get someone to answer or leave a message and be called back.

iv. Transition of care

This policy area was not discussed

v. Patient Autonomy, Patient representatives and support groups at the Centre of Expertise

- Contact to the patient organization is important since from there you get good information, as well as about social help and about experiences of other patients.

vi. Provision of rare disease information to patients

- Patients thought the most important thing was to get good information about the disease; the timing of information and examinations could be improved.
- Information about relevant patient organisations was seen as very important, and was received from the centre when possible.
- Patients said there is a need for concrete information about the prognosis of the disease. Currently there is no information on diagnosis in Danish. HCPs feel that some diseases are so rare that there is not much accumulated knowledge about either the diagnosis or the prognosis – but just letting patients know this would be helpful for patients.

vii. Raising awareness and training amongst healthcare and non-healthcare professionals

- Most patients reported that it was difficult to get a referral to the centre since it was thought (by GPs and/or specialists at the university hospital) that nothing was wrong or that it was not necessary to be sent on to the centre. Daycare or acute hospitals (the patient needs to visit) are better at referring patients to the centre. However, daycare cannot refer patients – they can contact the GP or another medical person and point out the necessity of referral. In some cases, the local level of the health care system feels capable of performing the treatment, even without all the correct competences. On the other hand, a few patients had experiences of quick diagnosis and referral to the centre, especially if the symptoms/handicap of the patient were visible. This raises concerns on the knowledge of GPs about rare diseases.

viii. Patient oriented and multidisciplinary care at the centre of expertise

- The patients said the centre has been very good in terms of taking care of the patients' family needs in a comprehensive way. However, there are issues where often the family does not recognize their own needs.

ix. Social care assistance at the centre of expertise

- HCPs give material and inform patients about special needs and how they should contact the municipality about these and there are many contacts from the municipality's social counsellors after the diagnosis has been made. Patient representatives said that whenever statements/letters from the centre were needed in support of social care applications, they were provided. Advice provided to contact he municipality (council) was considered good by patients; however they felt it would be really good to have a social care counsellor who knew the rules at the centre. Many patients reported about problems they had with the municipality (for years) due to people there not knowing the diagnosis or the chronic character of the disease. The HCP said that it is high on the wish list to get a social care counsellor for the centre, not only for the patients but also for the staff who need professional knowledge about issues in receiving social care support for rare diseases. It was also thought that social care counsellors of the municipalities would benefit from contact to a fellow social counsellor with special knowledge about rare diseases to process applications.

x. Facilities for diagnosis of rare disease at CoE

This policy area was not discussed

xi. Performance management of CoE

- HCPs pointed out that families with less resources need extra help to get the right treatment and services; it is the "resource-strong" parents who are at the meeting and that might mean that we give a one-sided picture. Some HCPs point out that patients do not need to fill out forms as part of the evaluation process, they can say aloud if there is a need for something. HCPs encourage them to give feedback and said that dialogue in the consultation is essential and meetings such as the Delphi face-to-face meeting are really important.

xii. Research at CoE

This policy area was not discussed

APPENDIX 4. ROUND 2 RESULTS: ENGLAND

i. Patient and Care Environment at the centre of expertise

- Most of the patients said they were satisfied with the overall facilities of the centre. However, participants pointed out that in general, healthcare facilities in England are inadequately designed to cater for patients with special needs. This causes difficulties and results in rare disease patients facing hardships during their visits to such facilities. Examples are hospital wards which are not set up with the needs of patients in mind. In hospital departments and GP surgeries visited by patients with special needs such as those who are visually impaired, there are no electronic calling systems in waiting areas or appropriately marked (via Braille) doors to clinic rooms.
- Hospital complexes are generally very big and inadequately sign-posted with expensive parking. Waiting rooms are generally overcrowded and noisy. HCPs were of the opinion that the ideal environment for a centre would be a unit which is located within a hospital but not "over-medicalised". It should be designed keeping in mind both the patient and HCPs needs and have all the necessary facilities within the unit or close to it. A purpose-built facility would be ideal but having a section of the outpatient department would be a cheaper and more convenient option which would help in providing multidisciplinary care to rare disease patients.
- Generally, patients say they are very satisfied with the staff at centres of expertise and find them to be extremely understanding and caring. However, patients also frequently come across HCPs, especially nursing staff outside the centres, who do not understand the illness and needs and sometimes behave insensitively.
- Patients are content with the way clinics are organised even though they have to wait sometimes - they prefer to have multidisciplinary clinics lasting one or two days rather than multiple visits. However, they did say that the centres should provide funding for travel and accommodation for patients and carers travelling long distances - at the moment it is sometimes provided by some patient organisations but otherwise the patient has to bear the cost. HCPs said they take into account each person's needs when planning the visit to make it as convenient as possible, taking into account things like travel or other needs of the patient or those accompanying them. Sometimes it is inevitable for the visit to run less smoothly than planned due to matters that may arise during the visit or due to patients changing appointments, etc.
- Patients like having dedicated staff for their visits.

ii. Access to centres of expertise

- HCPs say frequency of visits depends on severity of condition. Patients were satisfied with the frequency of their visits to the centres. For the most part, they do not feel the need to be seen more often. HCPs were generally satisfied with how often they see their patients and did not feel constraint of time or resources. They also added that if an urgent need arises they can almost always accommodate an appointment. It was suggested that a 'main' centre of expertise for a disease or a group of diseases should be established with 'local' outreach clinics depending on the need. Currently some centres provide outreach services or are in the process of developing them. It must be noted that having outreach services does not eliminate all travel to the main centre although it should be less.

- HCPs were happy to see European patients as long as their treatment is funded (either by their government or privately) and does not impact on resources for English patients. HCPs also said that they do not need to refer patients to other centres in Europe as the services in England are of the highest standards, but they do think it is important to share knowledge and experiences with other similar centres in Europe and around the world. If the number of cases is high in a country, it is essential for each country to have their own centre of expertise, as it can be difficult for patients to travel aboard on a regular basis. This morphs into a wish or interest in generating international education that would be one or two times a year to bring experts in other places up to par.
- Participants, based on their experiences, expressed the importance of having emergency contact details of on call HCPs at centres in the form of a plastic card accompanied by accessible patient records as there have been many situations when patients need urgent attention at a local A&E department but the A&E doctors do not understand the condition or listen to them or their carers. Some CoEs have already designed and begun distributing such cards and handheld patient records (i.e., a wearable USB stick) to patients. It is considered that a plastic card with 'do's and don'ts', including contact information of clinicians at the CoE, could help emergency HCPs as well as A&E staff with limited knowledge of the disease to appropriately triage patients .

iii. Coordination and cooperation within and outside centre of expertise

- Coordination and cooperation within the centres is satisfactory. Patients expressed concerns on the lack of coordination and cooperation of local healthcare facilities with centres of expertise and patients. One reason for this may be limited knowledge of rare diseases amongst HCPs outside the centre. Communication with GPs consumes a lot of consultants' time.
- HCPs said sometimes it can be difficult to get GPs to prescribe certain medications or dressing material due their costs or because the drugs and devices are unlicensed for the condition they are prescribed, or again, due to a lack of intimate knowledge of the condition. Some centres overcome this by supplying them directly if funding allows them to do so.
- Coordination letter with other HCPs are a problem as they involve many specialities. Simplified coordination letter with other HCPs should be used. Most centres do send coordination letter with other HCPs to GPs in paper format. Although that often functions well, at times it causes delays (more than 2 weeks). Setting up IT systems designed so data can be shared instantly with healthcare facilities across the country was suggested by many participants. Another option for timely data exchange could be the setting up of web-based databases holding patient records which could be accessed by HCPs across the country. This would also save money on duplication of laboratory tests and save patients distress and time.
- Some centres are linking up with local specialists caring for patients.

iv. *Transition of care*

- Many centres of expertise have a transition care pathway which works well. Some CoEs have dedicated nurses to carry out the transition sometimes done together with adult and paediatric nurses on multiple home visits. There were suggestions of having a dedicated transition coordinator with knowledge of the disease who can act as a link to the community. At one centre community nurses are available for family as well. Patient organisations can help maintain consistency, even with maintaining confidential coordination letter with other HCPs etc during the transition, or queries to or from GPs
- The transition care pathway should be designed such that it includes developing skills the patients might need in the future due to the progress of the condition, such as learning sign language before the onset of deafness, or braille before becoming blind, etc.

v. *Patient Autonomy, Patient representatives and support groups at centres of expertise*

- Patients said they were almost always given autonomy in deciding their treatment. HCPs say that all that is possible is done to take patients' needs and opinions into account when deciding treatment.
- The centres are reviewed and monitored by the National Commissioning Group (NCG) which funds the services.
- Most centres do not have a comments box but carry out a satisfaction survey annually as it is an NGC requirement.
- Patient organisations play an important role in providing patients information at the correct education level and age level in the correct formats (social media for teenagers), helping with social care applications, having a social care 'connector', meeting with first-time or newer patients, working with the family as well as enablers, speaking with HCPs on behalf of patients as well as in some cases funding research, hence it is crucial to have a patient organisation functioning in every centre.

vi. *Provision of rare disease information to patients*

- It is important to give information to patients when they are prepared for it. This is best done by not providing all information at once but giving it slowly over time, e.g., by outreach nurses, or possibly repeatedly and in differing formats. Patients need the opportunity to contact HCPs via email or phone. Nurses are usually the ones contacted.
- Information should be provided regularly. Patient organisation websites and newsletters provide information on the condition and the contact information of the centre for patients and family.
- Barriers due to cultural issues are a problem. Involving a community youth worker to act as a link may work. It was agreed that it would be helpful for staff to receive diversity training multiple times.
- Interpreters need to be used when needed.
- Some centres hold information days which are very successful. Also, care days outside the centre (summer meetings in a picnic, etc) are opportunities for teaching as well as social bonding. The professionals also get to see patients in a more real-life setting.
- Patient organisations were considered by patients and HCPs to help patients with meeting other patients, appropriate and understandable information, etc. These need to be patient-led exercises.

- There is a lack of appropriate information resources for adolescents/teenagers; these need to be developed with teenagers.
- Patients would like to meet similar patients to share experiences, this happens usually via the patient organisation. Centres would like to help arrange this if patients feel it is helpful.
- Informal communication between patients via Facebook, etc.

vii. Raising awareness and training amongst healthcare and non-healthcare professionals

- Participants stated that HCPs and non-HCPs (such as teachers, carers, employers, etc.) should be identified and provided with appropriate training. The group of individuals who are a priority are those who come in contact with rare disease patients. Some centres have specialist nurses and community nurses who educate those identified as important to have a basic knowledge of the condition. Some centres provide organised trainings to those who are interested which includes European and occasionally other international HCPs; there can be language barriers.
- In a few cases GPs can fail to recognize a patient suffering from a rare disease despite referring the patient to a number of different specialities but returning without a diagnosis. This flags up concerns over the diagnosis taking too long to be made. It is important to revisit the speciality training curriculum for GPs, and other specialities who are likely to come across rare disease patients in order to help in early detection and diagnosis. The need for GPs to understand the importance of referring patients with no diagnosis for genetic testing was also highlighted. High risk populations, such as Asians, need to be screened appropriately.
- HCP contacts made by other HCPs or non-HCPs outside the CoE are generally for a specific reason or a specific patient.
- GPs should be provided with information in the form of care guidelines, websites and CoE contact details.
- Some centres had training days for community nurses, hospital nurses, school nurses, dieticians and physiotherapists.

viii. Patient oriented and multidisciplinary care at centres of expertise

- Both HCPs and patients highlight the importance of having a psychologist as part of the service for patients as well as carers and family making home visits or having a consult outside the framework of the centre. A psychological service needs to be integrated and funded for issues like pain, disfigurement, sexuality, body image, etc.

ix. Social care assistance at the centre of expertise

- Patients experienced frequent difficulties in filling out the paper work necessary to receive benefits. Sometimes they were not aware of their entitlements. They also expressed frustration as their applications were rejected mainly because the civil servants who processed them did not have an understanding of their conditions. Where patient organisations existed patients were helped secure social care by patient organisations. Some CoEs had a dedicated support or welfare worker to help with social care. Often doctors and nurses spend time helping patients by contacting social services and providing supporting documents (which were many times ignored).

x. Facilities for diagnosis of rare disease at centres of expertise

- Most of the centres had the necessary laboratory facilities to provide diagnosis for rare diseases they treated. If there are no facilities on site, they have the possibility of sending the test out including to international laboratories.

xi. Performance management of centres of expertise

- In hospital PALS XX exist and therefore most centres do not have an evaluation committee.
- For conditions where patient organisations are established they play an important role in sharing patients' opinions with HCPs to help improve patient experience and care. The centres have close links with the patient organisations and have regular meetings to share information and discuss issues.
- HCPs say it is difficult to measure satisfaction of patients. The centre aims to improve the quality of life of patients therefore it is important to see patients at the centre regardless of the severity of their condition.

xii. Research at centres of expertise

- Some HCPs said their funding agency does not encourage research while others say it is encouraged.
- Patient organisations often provide information on research studies to patients and fund some of the research done by the centre.
- Patients would like research results shared and this is often not done. HCPs said the reason for this is that patients do not understand the time required to complete a research study. All research issues, including timeline on when information will be shared, needs to be clarified with patients from the start (in the Informed Consent, etc).

APPENDIX 5: ONLINE QUESTIONNAIRE – ALL OPEN-ENDED COMMENTS ENGLAND

Please list up to 3 factors that you think make patients feel welcome at the centre

3. HCPs

Staff: Small team so patients see familiar faces, friendly approachable staff, organisation/ information pre arrival, Easy phone and email contact between staff and patients, multi-disciplinary clinical staff, being available to answer questions, outreach nurse team visits patients at home, Enthusiastic staff with understanding of disease, designated specialist nurse, so long term relationship built, staff that are aware of the complex needs of the patients.

Environment/surroundings: New hospital building, Pleasant reception area, Availability of toys for young children, take into account additional needs of patients, Comfortable, clean and pleasant waiting area.

Visit: organised clinics, close involvement, no rushing/ lots of time for patients for appointments/ You are treated as a 'real' person not a 'number', Open access - no barriers to obtaining appts, Opportunity to speak with different members of the team, Coordinated visits / investigations, being prepared-read notes, Coordination letters are copied to patients, being informed about exactly what is going on, food and drink are provided, patient support group representative on hand in clinics.

4. Patients

Staff: Friendly, welcoming, know you personally and your condition/name etc., caring, approachable, knowledgeable about patients condition, punctual, dedication, caring, respectful, designated staff, make patient feel special.

Environment/surroundings: airy and light waiting area, modern surroundings. Child friendly, clean, lunch and drinks area, comfortable surroundings (seatings as well).

Visit: efficient organisation of visit or treatment, being seen on time, being given enough time with medics, extensive feedback, there are other patients with the same syndrome at the clinic at the same time, patients views not dismissed, take into account additional needs of patients, given choice.

Please list up to 3 factors that you think make patients feel unwelcome at the centre

5. HCPs

Staff: Rude members of staff, Other departments in the hospital have less understanding of the condition, no eye contact, stressed staff, unhelpful staff.

Environment/surroundings: Confusion over finding right place, cramped accommodation, Finding hospital and parking, Noisy areas, large hospital complex.

Visit: No permanent clinic, long waits, Having to travel from afar, Too many observers in the consultation, Can be an excess of form filling, no room ready, Being intimidated by a multidisciplinary examination, untidy, chaotic clinics, Not having a self-contained unit, some don’t like groups, no records.

6. Patients

Staff: contact with HCP who are not knowledgeable about illness, dismissive treatment by doctors, unfriendly unsympathetic staff, spoken to abruptly, not being spoken to, different staff, lack of communication between medics, Talking amongst consultants as if you are not in the room, unhelpful nurses.

Environment/surroundings: overcrowded clinics, noisy waiting rooms, no parking, no calling system for deaf, ticket style system, lack of personal space or privacy, Occasional use of substandard facilities and or necessary equipment unavailable, The clinic sometimes being situated in a ward that is unhelpful and uncomfortable to staff and patients alike, expensive canteen.

Visit: being made to wait, lack of introductions to other patients attending 6 hour multi-disciplinary clinics, lack of preparation, moving clinics, rushed through appointment, Lack of privacy and lack of light in ophthalmology examination rooms, No structured appointments, Conflict of information, distance, delay in getting coordination letters posted, lack of information, lack of choice, the cost of getting to the hospital, patients files are not computerised so when you bring the children to the hospital the doctors not familiar with them or those that know nothing of the condition could just go on computer to get the info they need.

32. What format of disease information do you think would be (or would have been) most helpful?

Patients

- Expert to email
- Face to face session with healthcare professional
- DVD on the condition explaining how to do self care
- Extensive lay term literature
- Large print
- Information brochure
- Online / Facebook page to exchange tips with other patients
- Internet
- Helpline (with patient organisation)
- Online patient community
- Contact details for the health care professional so that questions can be answered after the consultation
- Link to other similar patients

33. In your opinion, which activities aimed at raising awareness and knowledge of rare diseases for healthcare professionals (i.e., trainings, publications etc.) are most effective? (Patients)

- Publications
- Face to face education sessions
- Training
- Attending a clinic, hands on experience
- Conferences
- Meeting 'real' patients and their families
- Group sessions with consultants and specialist nurses
- Involving patient reps and charities in training sessions
- Good communication between all services
- Training for the relevant healthcare professionals that are likely to come across the key symptoms first, research
- A Medical handbook or written information describing the basics about the disease and professional that can be contacted for more information, internet

34. Barriers in providing information to patients (HCPs)

- Patient not being ready to accept information

- Patient/parents may not speak English

- Blindness/Deafness

- Potential poor sources of information

- Compliance with Trust standards - Approval takes a long time, issues of confidentiality

- No accessible written information for children or people with learning difficulties/ Literacy
- When diagnosis is not 100% certain

39. Based on your current experience, which activities aimed at raising awareness and knowledge of rare diseases for healthcare professionals (i.e. trainings, publications etc.) are most effective? (HCP)

- Training for those involved with a specific patient
- Personal visits to school etc to talk re condition
- Presentations at national and international meetings
- Visiting other healthcare professionals
- Information leaflets
- Inclusion in courses for specialist doctors in training
- information through specialist organisations and at their meetings
- Brief summary guidelines
- National meeting presentations
- Public media including papers, TV
- Lectures
- Publications
- Study days
- Medical handbook
- Study days for all HCPs
- personal mail in response to questions
- Other units visited by specialist nurses
- Care guidelines

Patient representation

47e. HCPs:

- Feedback is given after the Adult's clinic sessions where problems are discussed
- Very close relationship between Alstroms UK and International and specialists at this centre
- Focus groups and invited suggestions at 'open days'. Looking into patient representative at present

48e. Pts:

•At the end of each clinic problems are discussed and a summary of the day

•ASUK also evaluate the clinic with patients

53. Additional suggestions or comments towards improving the quality of care and treatment of patients with rare diseases in your centre?

Professionals:

•Would like permanent clinic site with all facilities, eg treatment rooms, bathroom, kitchen, lounge for families to relax etc. A dream!

•I would like to provide a service to patients with other rare skin disorders as good as the one we provide for EB. I would like to extend our informal "diagnostic opinion" service and put it on a more formal secure basis

•We need the NCG to pay for travel and accommodation for attendance from afar and not depend on the support group

•better education of management and finance to give more support

•One complaint from families in the far north of the country is the distance they have to travel. They would like another centre in the north of the country. It would be good if we could offer all families funding to get to their appointments but this is not possible. The nursing team are often stretched and although expected to participate find it hard to manage extra activities such as research and development.

Patients:

•Broader knowledge of rare diseases amongst the wider population of health professionals; greater recognition that often the most knowledgeable people, outside of the health specialists, are the parents/carers.

•I am very happy with the care I receive.

•Listen to the voice of the patients and their families. They are the ones who become experts in the condition. Respect between them and health care professionals will strengthen these clinics and hopefully will improve the health and well being of people with rare conditions.

•More nursing staff are required who are adequately trained and aware of the needs of the patients with EB. The specialist medical team are superb in their care of the patients but are overstretched. On some occasions the centre finds it difficult to accommodate the needs of our patients, particularly if they are admitted for any length of time and the need for nursing care is intense. It can also on occasion be difficult to obtain supplies of dressings. We have struggled for some time with poor facilities in which to hold our monthly MDT clinic, however strong protests and representation from the patient community were instrumental in getting a move to more suitable facilities.

53/54: Additional suggestions or comments towards improving the quality of care and treatment of patients with rare diseases in your centre?

Professionals:

•1. Facilitating patient support group 2. Social worker/ advise re financial assistance available 3. Use of Public forums-including media - this was brought up by our group of patients', any publicity is better than none' 4. Greater collaboration across the different mitochondrial research groups through out the world

•I can only speak about NF2 service, major quality of life survey in progress aiming to improve care for all patients, dedicated team aiming to improve care

•Increased patient involvement is very important. We are looking at the best ways to facilitate this. We are publishing guidelines to enable patients and health professionals to improve the care provided. Patient 'logbooks' and 'mito cards' allow rapid and clear information about a rare condition to be passed to other professionals. It also encourages patients involvement in their own care plans. We now have 24 hour on call access to a specialist opinion in mitochondrial disease. We are arranging outreach clinics (e.g. Glasgow and Manchester) so that patients do not have to travel as far for an expert opinion - and to help guide local practice.

Patients:

•Centre is 120 miles from where I live! Would be nice if was closer to my home. Otherwise the specialist nurses and Consultant are very helpful.

•Just to confirm that I feel that more needs to be done to make GP's and employers for aware of MELAS and that it is a disease. Also, contact details for our centre on a card or something would be helpful so that we can contact members of the team direct when we need to, and also days and times that they are available for us to contact (e.g. 24 hours)?

•It would be great to have a centre closer to where we live

•Yes**:** **-** consultations via Skype. Suffering from fatigue I couldn't make the long journey to Newcastle this year. A phone consultation was satisfactory however I think it would be worthwhile introducing Skype as they do in the States. - More communications such as a newsletter from the clinic to patients. Co-ordination of a patients group / forum both on and off line. - A detailed guide for GPs/doctors explaining Mito and its effects including which drugs/supplements to use etc which patients can hand out as when appropriate. - A mito nurse who could respond to ad-hoc mito queries similar to the role of a diabetes nurse.

•I would like more support available nearer to home. Perhaps a specialised nurse. We can e mail both the staff at the RVI at Newcastle and the Western General Hospital in Edinburgh if we need advice concerning MELAS. It would be very helpful to patients if knowledge of and recognition of the complications arising from MELAS could be more widely known by medical staff not just specialists. Treatment to improve symptoms would be great.

•overall me and my family are happy with the care and support received.

•patient should be given email address for all professionals involved in care, proper communication support needs to be provided for deaf hard of hearing patients, patients should automatically be sent appointments instead of having to chase them up all the time, I am supposed to be reviewed in the specialist NF2 clinic every 6 months. it has now been 7 months and I haven’t heard anything.

•Always have found the clinic and staff within this very efficient helpful and the day at the clinic very informative. The day at the clinic for my child provides me with a comprehensive report and ability to discuss with health professionals spanning all areas of the conditions problems a chance to talk ask questions etc... With local appointments they can be sporadic and also ends up the patient/mother! chasing and ensure/trying to ensure that notes are passed on to a range of other care professionals. At the clinic with the specialist areas for this day this is all done. Obviously the problem of outside care passing notes in is not always there but for understandable reasons. Find the clinic a very valuable needed service for patients and families. Hope that they will continue and may also offer more support with maybe an extra than one yearly appointment as my child grows in age and concerns/problems may heighten.

Appendix - Questionnaire for HCPs

Appendix - Questionnaire for Patients