Centres of Excellence for Rare Diseases

The National Alliance for people with rare diseases & all who support them

www.raredisease.org.uk
About Rare Disease UK

Rare Disease UK (RDUK) is the national alliance for people with rare diseases and all who support them. Our membership is open to all and includes patient organisations, clinicians, researchers, academics, industry and individuals with an interest in rare diseases.

RDUK was established by Genetic Alliance UK, the national charity of over 160 patient organisations supporting all those affected by genetic conditions, in conjunction with other key stakeholders in November 2008 following the European Commission’s ‘Communication on Rare Diseases: Europe’s Challenges’.

Subsequently RDUK successfully campaigned for the adoption of the Council of the European Union’s Recommendation on an action in the field of rare diseases. The Recommendation was adopted unanimously by EU Member States (including the UK) in June 2009 and calls on Member States to adopt plans or strategies for rare diseases by 2013.

We are now weeks away from the publication of the first UK wide plan for rare diseases and RDUK is working with the health departments in each of the four home nations to ensure integrated service delivery for rare diseases. This would coordinate:

- Research;
- Prevention, diagnosis and screening;
- Treatment;
- Care and support;
- Information;
- Commissioning and planning.

RDUK is funded by an unrestricted educational grant from its industry members. Details of whom can be found at www.raredisease.org.uk.

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## Terminology

In this report we will be referring to ‘Centre of Excellence’ for consistency, as this is the term used by the Department of Health in England. This is the equivalent to all other names used and these are explained in the introduction.

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Appendices are available at: www.raredisease.org.uk/documents/appendices.pdf
Chair’s foreword

While everyone who has a rare disease hopes that they will be treated by expert clinicians working in state of the art facilities supported by well trained professionals from a range of other medical and other professionals, there has been little consensus to date as to what criteria have to be met in order to justify the term “Centre of Excellence”.

The European Union Committee of Experts on Rare Diseases (EUCERD) has laid down what it deems to be the minimum standards, and these are an important baseline. However, as this report demonstrates, a Centre of Excellence is not just about the delivery of high quality clinical care. A Centre of Excellence is also about providing the opportunity to seek out information and explore what it means for you and your family, confident that it is reliable and trustworthy. It is about a place to meet others in the same situation as yourself for mutual peer-to-peer support. It is somewhere that all the family should be able to come to so everyone can be in the picture about what the future holds. It is also about a place where patients and professionals work together to create new knowledge, and develop better understanding that will result in better care, not just within the Centre of Excellence itself, but also by outreach into local hospitals, primary care and the community.

When this happens, patients experience the reality of the philosophy that inspired the creation of the NHS sixty five years ago. This report demonstrates that this is possible, but not yet universal. We hope it will support those who are currently providing world class care for rare disease patients, and serve as a stimulus to those who aspire to this.

We wish to express our gratitude to everyone who took the time to respond to our surveys, all those who were interviewed and those who volunteered their time to feature as a case study.

Alastair Kent OBE
Chair, Rare Disease UK
Director, Genetic Alliance UK
Executive summary

This report sets out a broader understanding of what a Centre of Excellence should be and identifies criteria by which a clinic can be identified as ‘excellent’. In this report we refer to ‘Centre of Excellence’ for consistency, as this is the term used by the Department of Health in England. We take it as axiomatic that high quality clinical care for patients and families is a prerequisite for any Centre of Excellence.

We collected our data through two surveys, one for patients with rare conditions and their carers, and one for a selection of clinicians, academic researchers, patient organisations and industry representatives; and through interviews with clinicians, patients, industry representatives, patient organisation representatives and commissioners. Following data collection we held a workshop with a range of stakeholders to capture their opinions on how Centres of Excellence can be developed and sustained in future years.

Research and registries

Members of every community of stakeholders recognised the important role that Centres of Excellence can play in research into rare conditions. Our respondents valued research into diagnosis and treatment just as much as they valued the higher profile research to develop cures that is often associated with rare conditions.

Information and support

Providing information and support is crucial to helping patients to manage their own condition. Centres of Excellence should provide patients with access to high quality information about their condition. A named care coordinator can provide much needed support to patients and their families.

This report provides evidence that supports the existing Department of Health in England proposal for a list of key characteristics that every Centre of Excellence should have:

- Coordinated care;
- Adequate caseload for expertise;
- Not dependent on a single clinician;
- Arrangements for transition from children’s to adults’ services;
- Engaged with people with rare conditions;
- Research active.¹

We propose the addition of two further criteria:

- Education and training for medical professionals.
- Membership of international networks of excellence.

We believe that NHS national commissioning bodies are the appropriate organisations to designate and monitor Centres of Excellence in the UK.

Sustainability of Centres of Excellence

Education and training for healthcare professionals was identified as a key component of the sustainability of Centres of Excellence. Professionals based in Centres of Excellence are ideally placed to play a role in undergraduate medical education, specialist training and in continued professional development. All of the Centres of Excellence we have spoken to promote education and training.

Collaboration and communication

Our findings confirmed the importance of collaboration and communication between Centres of Excellence and other Centres of Excellence nationally and internationally, local healthcare providers, and patient organisations. We found that there is room for improvement in all of these areas of collaboration and communication.
Introduction

‘What is a Centre of Excellence?’ At RDUK, this is a question we are hearing more often. Our results show that many people are unsure whether the centre they attend is a Centre of Excellence, and are unaware of what such centres are and what types of services they provide. This confusion and lack of clarity is exacerbated not only by the lack of a uniform definition of these centres in the UK and across Europe, but also by the range of official terms for a centre, such as ‘Centre of Reference’, ‘Centre of Expertise’, ‘Reference Centre’ and ‘Expert Centre’. In this report we refer to ‘Centre of Excellence’ for consistency, as this is the term used by the Department of Health in England. A centre can be ‘virtual’ with a network of experts across different hospitals or ‘physical’ with their own building.

“I am not aware of what the Centres of Excellence do”.

Parent/carer of a child living with Trismus Pseudocamptodactyly Syndrome & Arthrogryposis Multiplex Congenita.

Across Europe over 30 million people are affected by a rare condition. As a result a number of activities have been taking place at European government level, focusing on rare diseases and looking at Centres of Expertise as one of the ways of improving the care and treatment patients across Europe can access.

In 2009, the Council of the European Union called for each Member State to adopt a rare disease plan or strategy by the end of 2013, which had to include an emphasis on Centres of Excellence.

The EU Committee of Experts on Rare Diseases (EUCERD), established in 2009, advises the European Commission on the preparation and implementation of Community activities in rare diseases. In January 2013, the 51-member EUCERD unanimously adopted recommendations on European Reference Networks (ERNs) for Rare Diseases, which cover a range of issues including governance and funding. The overall vision is that ERNs will provide the framework for healthcare pathways for rare disease patients through a high level of integrated expertise. The areas covered in this report support the relevant EUCERD recommendations.

Following the adoption of the Council of the European Union Recommendation in 2009, the departments of health in the four UK nations have been working together to develop a UK Rare Disease Plan. The Consultation on the UK Plan for Rare Diseases was launched in February 2012, and a summary of the responses was published in November 2012, which recognised the need for Centres of Excellence and outlined a list of key characteristics that these centres should have, including:

- Coordinated care;
- Adequate caseload for expertise;
- Not dependent on a single clinician;
- Arrangements for transition from children’s to adults’ services;
- Engaged with people with rare conditions;
- Research active.

While these characteristics are all essential for a Centre of Excellence, RDUK believes that a definitive list of criteria should be determined with further input from people who live with a rare condition and all those who support them. We take it as axiomatic that high quality clinical care for patients and families is a prerequisite for any Centre of Excellence. The main aim of this report is to develop a broader understanding of what a Centre of Excellence should be and establish criteria under which a centre can be classed as a Centre of Excellence.
Methodology

In order to capture the wide range of views from the rare disease community RDUK conducted two online surveys and carried out a number of interviews with those involved with Centres of Excellence. The first survey was for patients and carers and the second survey was for multiple stakeholders (clinicians, academic researchers, patient organisations and industry representatives). The surveys were sent out in April 2013 to all RDUK members for a period of six weeks. Paper copies were available on request.

Additionally, a series of interviews were undertaken with clinicians, patients, industry representatives, patient organisation representatives and commissioners. The questions asked can be found in appendix 2, 3 and 4 (available online at www.raredisease.org.uk/documents/appendices.pdf). Five centres were approached and clinicians and patients were interviewed from each centre to gain their perspective of Centres of Excellence. A broad range of rare conditions affecting both adults and children were looked at. A list of conditions and centres we spoke to can be found in appendix 5 (available online at www.raredisease.org.uk/documents/appendices.pdf).

RDUK also held a workshop with a range of stakeholders to capture their opinions on how Centres of Excellence can be developed and sustained in future years. A list of attendees can be found in appendix 6 (available online at www.raredisease.org.uk/documents/appendices.pdf).

Respondents

We received a total of 436 completed responses to the patients and carers survey. Of these, three had to be discounted: two came from respondents living outside the UK and one because the individual did not state whether they had a rare condition.

Of the 433 valid responses to the patients and carers survey, 60% were from patients, 28% were from a carer/parent of a child living with a rare condition, 10% were from a carer/parent to an adult living with a rare condition and the remaining 2% were from a parent/carer of a person who had died in the past two years from a rare condition. Almost all (98%) of patients and their carers who replied to the survey have had their condition diagnosed. Just 27% of these people attend a Centre of Excellence for their rare condition. It should be noted that those who stated that they attend a Centre of Excellence have done so based upon their personal definition of what a Centre of Excellence is.

The multiple stakeholder survey received 61 completed responses. 46% of these were patient organisation representatives, 30% clinicians, 13% industry representatives and 11% academic researchers. Survey questions can be found in appendix 1 (available online at www.raredisease.org.uk/documents/appendices.pdf).
Lorna Brown’s Story

Lorna Brown, mother of Stephen and Russell living with Fabry disease – Kirkcaldy, Scotland

The Cardiomyopathy Association explains that: ‘Fabry disease is a rare disease that can affect the heart as well as many other organs. Fabry disease is caused by mutations (spelling mistakes) in the genetic code that determines the make-up and function of cells. The mutations in Fabry disease result in a deficiency of a chemical called a-galactosidase A. This chemical is an enzyme that normally digests fatty material inside cells throughout the body. In its absence, fatty material is not broken down and as a consequence it accumulates leading to swelling of the cells which then stop working properly’.⁶

Lorna’s sons Stephen and Russell, who are now 39 and 37 years old, were diagnosed with Fabry disease at the ages of 13 and 11. Both Stephen and Russell had been attending the Royal Free Hospital’s Centre of Excellence for Lysosomal Storage Disorders (LSDs) since their diagnosis. However, for the last two years Stephen has been monitored in the Royal Victoria Hospital, Belfast.

Before receiving a diagnosis and subsequent treatment from the specialists at the Centre of Excellence, Stephen and Russell’s local doctor and local consultant found it difficult to manage their pain. The ongoing issue of pain (Stephen and Russell lived with background pain in their hands and feet) affected their education and social development. For Lorna and her family, attending this Centre of Excellence has not only significantly improved both her son’s quality of life but has provided the family with emotional support;

“Fabry is such a rare condition, so knowing advice and reassurance from the team at the Royal Free was only a phone call away considerably eased the burden of anxiety I felt as a mother. The treatment allowed Stephen and Russell to participate more fully in normal physical and social activities”.

Their local consultant took on board any concerns the family had. However deeply interested as he was, he did not have the level of expertise available at the LSDs Centre of Excellence at the Royal Free Hospital. “Our consultant was absolutely wonderful, meticulous and anything we said that appeared strange he would immediately look into it and see if there was any way he could deal with it. He was excellent, consulting regularly with the Royal Free, and the team at the Royal Free Hospital said if they could have that level of notes from all consultants it would make their job much easier. He really was a great doctor but he didn’t have all the knowledge at his fingertips as the professionals at the Centre of Excellence did and who continually care for patients with Fabry disease”.

In Lorna’s eyes, having the chance to attend the Royal Free Hospital’s Centre of Excellence for LSDs has saved her children’s lives. “Our boys are now 39 and 37 and they are getting to the age where they could have been facing vital organ failure and even coming to the end of their lives. The outlook was not good but now if you look at them you would think they are perfectly healthy. They are limited in some ways but not like they used to be and they don’t have the pain they used to have”.

Lorna’s son Russell

Stephen with his wife Elieen

Excellence has not only significantly improved both her son’s quality of life but has provided the family with emotional support;
Findings

Key attributes and facilities for a Centre of Excellence

In both surveys, respondents were asked to select the attributes they felt a Centre of Excellence should have or provide. The table below shows the three most popular choices in both surveys. Full questions can be found in appendix 1 (available online at www.raredisease.org.uk/documents/appendices.pdf).

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<thead>
<tr>
<th>Patients and Carers</th>
<th>Respondents</th>
<th>Range of Stakeholders</th>
<th>Respondents</th>
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<tr>
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<tr>
<td>Links with local healthcare providers</td>
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<td>Links with local healthcare providers</td>
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<tr>
<td>Links with patient organisations</td>
<td>152</td>
<td>Registries</td>
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Respondents were also asked to rank the five facilities they felt a Centre of Excellence should have. The table below shows the three most popular choices in both surveys.

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<th>Patients and Carers</th>
<th>Respondents</th>
<th>Range of Stakeholders</th>
<th>Respondents</th>
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<tbody>
<tr>
<td>Free parking</td>
<td>104</td>
<td>Facilities to stay overnight</td>
<td>14</td>
</tr>
<tr>
<td>Information room</td>
<td>90</td>
<td>Private family room</td>
<td>13</td>
</tr>
<tr>
<td>Facilities to stay overnight</td>
<td>84</td>
<td>Child friendly waiting and treatment rooms</td>
<td>12</td>
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We are aware that some of the figures above, especially among stakeholders, are low. However, it should be noted that these include the views of a majority of patient organisations.
Sustainability of Centres of Excellence

Key findings

1. In both of our surveys, education and training for medical professionals was identified as one of the most important functions that a Centre of Excellence must undertake.
2. Centres of Excellence should not be dependent on a single clinician.
3. Centres of Excellence need access to a range of funding sources in order to provide all of their services sustainably and successfully.

Key finding 1: In both of our surveys, education and training for medical professionals was identified as one of the most important functions that a Centre of Excellence must undertake. It has long been recognised that there is little training in rare conditions in the UK medical curricula. It is encouraging to find that many Centres of Excellence are providing education and training to students. All of the Centres of Excellence we have spoken with promote education and training and some have their own research facilities which involve undergraduate and postgraduate research students.

58% of patients and carers and 49% of respondents to the multiple stakeholder survey see education and training as the key aspect of the work of a Centre of Excellence.

“A main ethos of this clinic is to inspire the next generation of researchers and clinicians”.

Dr Suvankar Pal – Anne Rowling Clinic.

Respondents identified some practical ways in which professional interest in rare conditions can be further developed. Students should be invited to spend time in Centres of Excellence to attend clinics and participate in research. Professionals based in Centres of Excellence are ideally placed to educate both trainees and current healthcare providers about rare conditions and the role Centres of Excellence can play in care provision.

The National Genetics Education and Development Centre has developed a number of models to deliver specialist training. This resource should be used by Centres of Excellence to deliver training, which will ensure best practice and make sure health professionals are fully engaged with rare conditions.

Key finding 2: Centres of Excellence should not be dependent on a single clinician. Respondents felt that, in order to preserve the quality of service if a team member is not available, it is important that a Centre of Excellence has sufficient staffing for service expertise and that a centre is not dependent on a single professional.

“What do you do if somebody goes off on long term sick [leave]?”,

Professor Bishop, Sheffield Children’s Hospital.

Key finding 3: Centres of Excellence need access to a range of funding sources in order to provide all of their services sustainably and successfully. Participants proposed that budgets for funding specialised services should be protected and allocated by the relevant specialised services commissioner. Our Improving Lives, Optimising Resources’ report recommended that the value of specialised services be recognised and that resources should be safeguarded to fund these services.

Our findings show that the NHS does not provide sufficient funding to sustain a Centre of Excellence. Interviewees identified a number of different funding streams from different aspects of their work. For example, clinical research into rare diseases is funded by a variety of funding sources which include industry, patient organisations, the National Institute for Health Research and the Medical Research Council.
“In our Centre of Excellence, the main part of our funding for medical staff is actually via the University in the salaries for the clinical professors and the training grade doctors who come into the group on research money. So although all these people deliver a vital clinical role within the NHS, it is the strong link with the research side that provides continuity of funding and the ability to deliver care and maintain our role as a top performing centre”.

Professor Kate Bushby, MRC Neuromuscular Centre, Newcastle.

Recommendations

- Centres of Excellence should play a role in undergraduate medical education, specialist training and in continued professional development.
- Workforce planning must ensure continuity of services.

Sustainability should be supported through a range of designated funding streams to provide clinical care and research facilities, as well as other aspects of a Centre of Excellence’s role.

Collaboration and communication

Key findings

1. Communication between Centres of Excellence and local healthcare providers needs to be improved.
2. Communication between Centres of Excellence across the UK and globally is patchy.
3. Collaboration between Centres of Excellence and patient organisations plays a crucial role in informing and supporting patients.

Key finding 1: Communication between local healthcare providers and Centres of Excellence needs to be improved. Encouragingly, over half of respondents (57%) stated that communication between their Centre of Excellence and general practitioner is either ‘excellent’ or works well ‘most of the time’. However, 32% stated that such communication ‘needs improving’ or is ‘non-existent’.

In comparison, just 28% of respondents reported that communication between their Centre of Excellence and local hospital is either ‘excellent’ or ‘works well’. Almost 40% of people who completed our survey described this communication as ‘non-existent’ or ‘needs improving.’ This figure may actually be greater as over 10% of respondents replied that they ‘didn’t know’ how to describe this relationship.

Our study also found that nearly half (46%) of respondents who attend a Centre of Excellence only receive some or do not receive any copies of letters sent from their centre to their local healthcare providers. This could mean that at times letters may not be sent to local healthcare providers, which can result in the patient having to provide updates and explain their condition at every appointment.

“Communication and collaboration is perfect, if only you could bottle it and spread it around. It does not work like that in every authority unfortunately”.

Elaine Healey, Chair of Brittle Bone Society and parent carer of two children with Osteogenesis Imperfecta who attend Sheffield Children’s Hospital.
When respondents were asked to select in order of importance the five most important (see page 9 for top survey results) attributes they felt a Centre of Excellence should have or provide; good links with local healthcare services was chosen as the second most important element of Centres of Excellence in both our surveys. It was the second most popular choice among 44% of patients and carers and 36% of clinicians, academic researchers, patient organisation and industry representatives.

An overwhelming majority (84%) of respondents to the Consultation on the UK Plan for Rare Diseases agreed that each expert centre must know its network of local hospitals and that local hospital must know the pathway to the expert centre which will offer help, support, advice and assistance.5

**Key finding 2: Communication between Centres of Excellence across the UK and globally is patchy.** Centres of Excellence should work with local healthcare services to manage a patient’s condition to the highest standard and ensure that their day to day life is disrupted as little as possible. Every patient should have access to the best possible care regardless of their location.

Many respondents noted that it would be beneficial for medical professionals to liaise across the UK and globally, so they are informed of new treatments and upcoming clinical trials. In addition, Centres of Excellence should share their knowledge and expertise with local healthcare providers, which would raise awareness on a much broader scale.

> “Worldwide research is ongoing and it would be good to have healthcare professionals up to date with what is currently being trialled and up to date treatments. It would also be beneficial for them to coordinate globally”.

A parent/carer of an adult living with Lebers Hereditary Optic Neuropathy.

Although collaboration across the UK and internationally currently takes place for some conditions, this needs to become accepted good practice for all rare conditions. Networking allows for research opportunities to be identified and developed. Some conditions are so rare that just a few patients have them in the UK. Progress in research into these conditions greatly facilitates the formation of networks across Europe and the world to share data and knowledge, support research and develop best practice.

> “We are fortunate to have an outstanding Centre of Excellence. Other areas which might benefit from improvement in the future are: networking between this Centre of Excellence and other similar ones elsewhere in the world...”

Parent/carer of a child living with Barth Syndrome.

**Key finding 3: Collaboration between Centres of Excellence and patient organisations plays a crucial role in informing and supporting patients.** When patients and carers in our survey were asked to select in order of importance the five most important attributes they felt a Centre of Excellence should have or provide, almost 40% rated links with patient organisation as the third most important aspect of a Centre of Excellence (see page 9 for top survey results).

Patient organisations play a key role in providing additional support which is complementary to the health service.
“Patients are consulted by the Trent Regional Immunology & Allergy Consortium within individual centres, for example in the production of patient leaflets. Individual patients can be very engaged...”

Clinician’s response in survey.

In order for the Centre of Excellence to meet patient needs, the patient voice needs to be heard. This is best done by working in partnership with patient organisations, where they exist.

“It is very important clearly with any Centre of Excellence that services are developed in partnership with the patient groups. This is so that patients have an active role in how the service gets developed so these services meet the patient needs”.

Dr Timothy Barrett, Birmingham Children’s Hospital.

Patient organisations have a great deal of expertise and play a crucial role in the development of Centres of Excellence.

“They [patient organisations] are the real experts”.

Professor Atul B. Mehta, Royal Free Hospital.

**Recommendations**

- Centres of Excellence should ensure communication to local healthcare providers is of a high standard.
- Existing best practice in collaboration between Centres of Excellence in the UK and the rest of the world should be adopted by all centres in the UK.
- Resources should be allocated to support the valuable role of patient organisations in supporting and representing patients.

**Research and registries**

**Key findings**

1. Members of every community of stakeholder recognised the important role that Centres of Excellence can play in research into rare conditions.
2. Our respondents identified the collection of information held at Centres of Excellence in registries as an important tool for research into rare conditions.

**Key finding 1:** Members of every community of stakeholder recognised the important role that Centres of Excellence can play in research into rare conditions. It has long been recognised that research into rare diseases is vital to improve diagnosis, care provision and to enable the development of new treatments. In gathering patients and expertise, Centres of Excellence provide the ideal site to base research into rare conditions. This point was repeatedly made by stakeholders from all sides.
“It is about critical mass, critical mass of patients, critical mass of doctors, critical mass of physiotherapists, clinical trial managers and so forth. They are essential to this clinical research that we do”.

Professor Kate Bushby, MRC Neuromuscular Centre, Newcastle.

“A key goal for all of us is to develop new treatments for rare diseases and hopefully one day a cure for some of them”.

Dr Timothy Barrett, Birmingham Children’s Hospital.

“Without research my daughter probably would not be alive”.

Mother of daughter living with Osteogenesis Imperfecta Type 3.

Many Centres of Excellence deliver the full pathway of care for conditions. The relatively high volume of patients that they see allows the development of best practice in diagnosis and treatment. Our respondents valued research into diagnosis and treatment just as much as they valued the higher profile research to develop cures that is often associated with rare conditions.

**Key finding 2: Our respondents identified the collection of information held at Centres of Excellence in registries as an important tool for research into rare conditions.** In the multiple stakeholder survey respondents placed registries in the top three important attributes for a Centre of Excellence (see page 9 for top survey results). This echoes the recommendation of the Council of the European Union which identified patient registries and databases as key tools in developing clinical research in the area of rare diseases and in improving patient care and healthcare planning.9

Respondents stated that registries are an important tool in developing an understanding of the natural history of a rare condition. Collating information will enable the identification of common symptoms that are associated with a rare condition and help increase awareness among healthcare professionals in the future and facilitate the development of good practice in managing rare diseases.

“A registry defines the natural history of the disease and phenotype. It then ensures every patient is equitably cared for and is a basis for research”.

Dr Graham Lipkin, Queen Elizabeth Hospital, Birmingham.

Our findings corroborate the response to the Consultation on the UK Plan for Rare Diseases which asked ‘Do you agree that registers are an important tool in rare disease and could be a core component of the service specification of an expert centre?’ This question received the highest number of positive responses with 90% of replies agreeing.1

**Recommendations**

- Research at Centres of Excellence should develop all aspects of the patient pathway including diagnosis and treatment.
- All Centres of Excellence should either maintain registries for the diseases they treat or contribute to national and international registries as necessary.
Information and support

Key findings

1. Patients expect to be able to access high quality information at a Centre of Excellence.
2. Direct access to a named care coordinator is crucial in helping patients manage their own care effectively.
3. Patients who attend a Centre of Excellence for their rare condition value the opportunity to meet with others who are going through the same experience.
4. The cost of attending a Centre of Excellence can be a deterrent to patients to bring their family members to their appointment.

Key finding 1: Patients expect to be able to access high quality information at a Centre of Excellence. Our survey showed that the majority (60%) of respondents who attend a Centre of Excellence said that there is either no facility for them to access information on their condition or that they are unaware whether or not facilities to access information in their centre exist.

An information room with a computer and access to the internet was a popular choice of facility for patients in a Centre of Excellence. This corresponds with our report Experiences of Rare Diseases: An Insight from Patients and Families, in which over half (52%) of respondents felt that they had not been given enough information on their condition following diagnosis.

"People with rare conditions need information at the time of diagnosis. We did not get this and as a result we did not know how bad the condition was, and thought it was just an easy problem to put right. [...] You end up going online and frightening yourself and the family".

Patient response to the Consultation of the UK Plan for Rare Diseases.

"Much of the information that is given, whilst it is free, is not necessarily given unless the correct question is asked. More information with regard to expected treatment path, predicted outcomes would be useful to aid planning and to prepare the whole family for what may lie ahead".

Parent/carer of child living with Primary Female Epispadias.

Route Maps, which describe routes to access care for individual conditions would be a means to ensure better quality information is available for patients. Doctors and patients can work together to follow the steps needed to achieve this, as outlined in Genetic Alliance UK’s ‘Route Maps for Rare Conditions Toolkit’ launched in 2013. The toolkit is an evolving guide which signposts the way to health and social care for patients and their families in order to improve access to information, leading to earlier diagnosis, and improved care and support.
Key finding 2: Direct access to a named care coordinator is crucial in helping patients manage their own care effectively. It was encouraging to find that 87% of patients and carers who responded to our survey and attend a Centre of Excellence for their condition have access to a specialist health professional who they can contact remotely by phone and email. However, only 56% of respondents who attend a Centre of Excellence stated that they have access to a support worker or a named care coordinator who can assist in ensuring they receive coordinated care.

Non-specialist healthcare professionals in hospitals should also have access to a specialist in rare conditions as needed. Healthcare professionals need to be able to coordinate the delivery of routine or emergency care with the requirements of the patient’s rare condition.

“A emergency contact number for when we become very ill. Where they completely understand our condition and can properly help us”.

Patient living with Panhypopituitarism, when asked if there was anything they would change in the centre.

A report launched by RDUK in 2013 outlined the importance of every rare disease patient having access to a named care coordinator. Care coordinators can help their patients have a positive experience of the care they receive, by ensuring the effective coordination of services providing a vital source of information and support, and assisting effective self-management.12 This report highlights that care coordinators represent significant value for money and that they can save the NHS money across many different areas.

Key finding 3: Patients who attend a Centre of Excellence for their rare condition value the opportunity to meet with others who are going through the same experience. As outlined earlier in the report (page 9), the majority of people who replied to our survey do not attend a Centre of Excellence. Of these respondents, just over 66% said they would benefit from attending a Centre of Excellence, while almost 30% ‘didn’t know’ if they would benefit. Clear criteria are needed for what a Centre of Excellence is and performance monitoring is needed so that patients are aware of the benefits that these centres could bring.

Interviewees as well as patient and carers who responded to our survey felt the benefits of attending a Centre of Excellence were increased knowledge, experience and understanding from medical professionals, whilst others wished to meet people in the same position who understand their fears and anxiety.

“This is the opinion of many of our families: they want a team of specialists that understand this condition”.

Kerry Leeson-Beevers, Alstrom Syndrome UK.

“Access to information regarding new medicines, coping strategies or [clinical] trials would be great for our son”.

Parent/carer of a child living with Hypo-pituitarism Agenesis of the Corpus Callosum

The outcome of a workshop that RDUK held in July 2013, also indicated strongly that social interaction with other patients living with a rare condition is essential as they can support each other and share experiences. Attending a Centre of Excellence often gives patients the extra support they need to manage their condition more effectively.

Key finding 4: The cost of attending a Centre of Excellence can be a deterrent to patients to bring their family members to their appointment. Over 80% of patients either only take one person or go on their own when attending their Centre of Excellence. Our findings suggest that this is due to either travel and accommodation costs or lack of space and facilities in Centres of Excellence to accommodate the patient and their family.
Of those who responded, for 25% an outpatient appointment has required them to stay overnight; 45% stay with friends or family or in a hotel; 35% of these respondents either pay for this stay themselves or are funded by their parent or carer.

“Quite often I go to my Centre of Excellence on my own because I have to travel to London from Cheshire, so unless I feel it is important for my husband to be there, due to travel costs, [I go on my own]”.  
Patient living with Scleroderma.

In the survey for multiple stakeholders, 35% of respondents selected facilities to stay overnight as the most important facilities that a Centre of Excellence should have. This result is supported by 29% of patients and carers who also chose facilities to stay overnight as one of their top three (see page 9 for top survey results) facilities a Centre of Excellence should have.

“[We would like] accommodation for more than one parent to be able to stay”.  
Parent/carer of a child living with Bladder Exstrophy.

Parking was seen by patients as a significant cost. Many patients spend the whole day or a number of days at a Centre of Excellence and therefore paying for parking can become expensive. Free parking remains a popular issue for patients.

**Recommendations**

- Centres of Excellence must ensure patients are fully informed about their condition and provide facilities for patients to access information on their condition.
- Every patient with a rare condition should have a named care coordinator within a Centre of Excellence to ensure they can access information and support as needed.
- Centres of Excellence should facilitate peer support for people living with a rare condition.
- Centres of Excellence should consider means by which they can lower the cost of a family attending an appointment.
Niamh MacConnell’s Story

Niamh MacConnell, mother of Lucy who has Osteogenesis Imperfecta Type 3 – Galway, Republic of Ireland

The Brittle Bone Society explains that: ‘Osteogenesis Imperfecta (OI) is a genetic disorder of collagen, a protein which forms the framework for the bone structure. In OI the collagen may be of poor quality, or there may just not be enough to support the mineral structure of the bones. This makes the bones weak and fragile and results in the bones being liable to fracture at anytime even without trauma. Type 3 is a severe form of the condition’.

Niamh’s daughter, Lucy, who is now six years old, was diagnosed with OI Type 3 in 2007. They have been attending the Sheffield Children’s Hospital Centre of Excellence for OI since Lucy was just four weeks old.

Attending the Sheffield Centre of Excellence for OI has not only been crucial to Lucy’s survival but Niamh and her family would be lost without it. The Centre of Excellence has also worked with Lucy’s local consultants to help the family manage the condition. “This Centre of Excellence has made all the difference in the world. I cannot even imagine where we would be without it, firstly I don’t even know if Lucy would be alive and secondly I think she would be much more disabled”.

When Lucy first attended the centre, Niamh would go with her daughter and either her husband or Lucy’s grandma would accompany them as it was too difficult for just one adult to go with Lucy and help her on transport as well as carrying all the equipment she needs. Lucy would be admitted at the hospital for three or four nights and she would have her treatment and see the multi-disciplinary team. However, Lucy now receives all her treatment in Ireland with the guidance of clinicians at the Sheffield centre. Clinicians at Sheffield direct Lucy’s local paediatrician in Ireland in relation to management of the condition such as doses and blood monitoring. Every six months, they still visit Sheffield but now this only requires one overnight stay and a clinic appointment where they meet the whole team. “In the beginning it was such a relief for us to visit a centre like Sheffield, where the team is so experienced in managing children with a rare condition like OI. However, with time and the benefits of the knowledge, confidence and skills that the Sheffield team gave to us, we felt ready to move on to shared care between Sheffield and our local team. This transition allowed for less disruption to family life as there was a reduced need for costly and time-consuming trips away from home and from our other child”.

As the family became more familiar with OI and with the support of the Centre of Excellence Niamh pushed to have all of Lucy’s treatment in Ireland. For almost 4 years Lucy was having alternate treatments in Sheffield and Ireland. When they were at the centre in Sheffield they would spend three days on the ward which would involve Lucy taking time off pre-school and both Niamh and her husband taking time off work. “I just found that it was a lot of time and I really felt it was a waste of their precious resources having us there at that point for those long admissions”.

Niamh thought it would be a much more efficient use of time to have Lucy’s admissions back in Ireland because their local paediatric hospital, University Hospital Galway, facilitates this at the weekends and they can take Lucy the short distance from home to have her treatment. Niamh highlights the benefits this brings to the whole family.
“This is brilliant because we can bring Lucy home in between her two treatment infusions on the Saturday night. All we have to do is go into hospital for four hours on a Saturday and four hours on a Sunday and it really does not interfere much with our lives which is lovely for everyone in the family at this stage”.

Lucy no longer misses as much school and the arrangement has enabled the family to coincide visits to Sheffield with her school holidays. Niamh and her husband also do not have to take as much time off work or miss out on their other child’s activities. “To have her treatment locally is wonderful. It makes a massive difference to our lives because we can just continue with our normal life. We continue to need and very much value the expert advice and guidance from the specialist team at Sheffield Children’s Hospital. OI is a rare condition so the necessary level of expertise is not available for children like Lucy in Ireland. We are lucky to have the benefits of specialist care at Sheffield Children’s Hospital and local support at University Hospital Galway; it is a perfect combination for Lucy’s evolving needs”.

Conclusions

Having examined the concept of a Centre of Excellence, it is apparent that the absence of a concrete definition of a Centre of Excellence in the UK limits the value that the rare disease community and health services can gain from exploiting the concept. We believe it is vital that a definition be agreed. The sensible choice for a designating body is the commissioner.

The criteria from the Department of Health in England make a useful starting point for this definition. These are listed here:

- Coordinated care;
- Adequate caseload for expertise;
- Not dependent on a single clinician;
- Arrangements for transition from children’s to adults’ services;
- Engaged with people with rare conditions;
- Research active.

Our findings have supported these criteria. Following our work, we elaborate on these and propose some additional criteria:

- **Coordinated care.** The provision of a named care coordinator to every patient with a rare condition is an important component of coordinated care. This should be augmented with good quality communication and coordination with patients’ local healthcare providers.
- **Adequate caseload for expertise.** This is a key requirement for the sustainability of a Centre of Excellence, and goes hand-in-hand with the following criterion. This critical mass of patients not only supports the sustainability of care provision, but also facilitates other activities of a Centre of Excellence such as research.
- **Not dependent on a single clinician.** This criterion was supported by our findings, and is vital for the sustainability of Centres of Excellence.
- **Arrangements for transition from children’s to adults’ services.** This is a key issue for those affected by childhood onset rare conditions. This is an issue that affects all care provision, within and without Centres of Excellence, and we have purposely left discussion of this for our next report, which will cover transition both between paediatric services and adult services and between adult services and geriatric services in the UK.
- **Engaged with people with rare conditions.** This criterion needs to cover our three recommendations regarding the use of a Centre of Excellence as an information hub, a location for peer interaction and that Centres of Excellence should work with patient organisations.
- **Research active.** The Department of Health in England’s final criterion is supported by our work. Specifically the benefits of locating research in a Centre of Excellence were recognised and the importance of recording and sharing information collected either through their own registry or through participation in a national registry.
Our additional criteria are:

- **Education and training for medical professionals.** Education and training is an important component of the sustainability of a Centre of Excellence. Strong links need to be made in both directions between education providers and Centres of Excellence.

- **Membership of international networks of excellence.** Where applicable it is vital that Centres of Excellence do not work in isolation. Exchange of best practice leads directly to high quality care and high quality research with benefits for those affected by rare conditions and health services.

In addition, as stated in our introduction, we take excellent healthcare provision to be a prerequisite of a Centre of Excellence for rare conditions.

Once this definition of a Centre of Excellence is agreed upon, it will be necessary to appoint a body to designate candidate centres as Centres of Excellence. It is clear to us that the most appropriate body for this task for rare conditions is the relevant NHS national commissioner.

Status of Centres must then be monitored under a framework that allows designation of new centres and policing of standards. The creation and existence of Centres of Excellence is an evolutionary process, and it will be necessary for commissioners to create an environment that allows newly founded centres to develop into a position where they may attain the status of a Centre of Excellence.
End Notes

1. Consultation on the UK Plan for Rare Diseases’, Department of Health, February 2012
2. European Reference Networks in the field of Rare Diseases: State of the Art and Future Directions’, Rare Diseases Task Force, Third Report, July 2008
4. ‘The European Union Committee of Experts on Rare Diseases.’
5. ‘EUCERD Recommendations on European Reference Networks for Rare Diseases.’
6. The Cardiomyopathy Association
7. ‘Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy’, Rare Disease UK, February 2011
8. The National Genetics Education and Development Centre
9. European Commission:
10. ‘Experiences of Rare Diseases: An Insight from Patients and Families’, Rare Disease UK, December 2010
11. ‘Route Maps for Rare Conditions Toolkit’, Genetic Alliance UK, January 2013
12. ‘Rare Disease Care Coordination: Delivering Value, Improving Services’, Rare Disease UK, February 2013
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