Experiences of Rare Diseases: Patients and Families in Scotland

The National Alliance for people with rare diseases & all who support them
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 150 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 each Member State of the EU (including the UK) in June 2009.

The Recommendation calls on Member States to adopt plans or strategies for rare diseases by 2013.

RDUK is campaigning for a plan for integrated service delivery for rare diseases. This would coordinate:

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

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Chair’s Foreword

No family wants to discover that they are at risk from a serious, possibly life limiting disease. For those who do find themselves in this situation there is an urgent need to be able to access information in order to understand what has happened, and to be able to plan for a future which may be radically different. It can be difficult enough to come to terms with the knowledge that there is a relatively common, chronic condition in your family. So if your condition is rare then this can be made much harder because of difficulties in getting an accurate diagnosis and then securing the help and support needed to understand and manage the condition affecting you, your child or a member of your family.

As this report indicates, the NHS is capable of providing a world class service to families with rare diseases, and many patients are very happy with the support they receive. However this high quality is not universal, and too many families struggle to get a diagnosis. A missed or a mis-diagnosis represents a lost opportunity for the family, and can result in avoidable harms to patients as they lose out on treatments that might help, and undergo tests and procedures that may be of little or no use.

For the NHS too, diagnostic delays and errors make planning services more difficult, and can result in inappropriate allocation of resources because the extent of patient need is not fully appreciated. Research may also be inhibited through difficulties of small numbers affected with particular conditions and problems in creating the critical mass of affected families necessary to undertake systematic investigations.

The Scottish Government and NHS Scotland is committed to working with the other nations in the UK to develop a plan for improving services and support to rare disease patients in Scotland by the end of 2013.

As this report shows, there is a basis on which this can be built. But as this report also shows, there is still some way to go before all rare disease patients and families can be confident that they are receiving timely, high quality support. We look forward to a close collaboration with the Government, politicians, clinicians, planners and commissioners to bring this about.

Alastair Kent OBE
Chair, Rare Disease UK
Director, Genetic Alliance UK
Introduction

Between October and November 2012, Rare Disease UK (RDUK) carried out a survey of patients and families living in Scotland affected by rare diseases. The aim of this survey was to find out more about the experiences of people in Scotland living with a rare condition, and to identify some of the common issues and problems they frequently face.

At RDUK, we are too often told of difficulties patients and families affected by a rare disease experience in getting a diagnosis and accessing appropriate service, information and support throughout the progression of their condition. In 2010, RDUK published “Experiences of Rare Diseases: An Insight from Patients and Families” following a survey of rare disease patients across the UK. The survey covered multiple aspects of rare diseases which patients and families had experienced problems with including, diagnosis, coordination of care, awareness and participation in research, access to therapies and services, and access to thorough, reliable information and support.

In 2010, almost 600 patients and families responded to the RDUK Patient Experiences survey, 12% of which were from Scotland. In Scotland, 300,000 people are likely to be affected by a rare disease at some point in their lives and in 2012, RDUK decided to look closer at the experiences of patients and families affected by rare diseases in Scotland.

In October 2012 an online survey, covering multiple aspects of rare diseases, was sent out to all of RDUK’s members, including patient organisations. Many patient organisations forwarded the survey to their own members. Paper copies were available for those who requested them.

A total of 144 responses to the Scotland Patient Experiences survey were received. Of the 144 responses, 64% were from the patients, 33% were from a relative or carer of someone with a rare disease and 3% were from a relative or carer of someone with a rare disease who has passed away.

A total of 89 different rare conditions are represented in the survey. A full list of these conditions can be found in Appendix 1. These conditions ranged from ultra-rare to the more ‘common’ rare diseases and included chromosomal, single gene, multi-factorial and non-genetic conditions, as well as undiagnosed conditions and patients who have multiple different rare diseases. Despite the wide variation in the symptoms, prognoses and medical needs of these conditions, the survey indicated commonalities in experiences in regards to their care, information and support.

Respondents to the survey came from all over Scotland, with 28 out of the 32 Scottish Local Authorities being represented. RDUK are therefore able to suggest that our findings are a good representation of the general population of Scotland.

The results support the need for a strategy for rare diseases in Scotland. RDUK believes that a strategy would reduce the fragmentation of services and ensure that patients of rare conditions are able to access equitable, high quality care, information and support in a timely manner, whilst at the same time making a more efficient use of NHS resources.

Throughout this publication we have included case studies of a number of patients and families affected by rare diseases to provide detail on their experiences and to put some of the findings of the report into context.

RDUK would like to thank all those who took the time to respond to our survey, as well as the patients and families who are featured as case studies.

Summary of Key Findings

Research
1. Patients and families are supportive of research into rare diseases and have a willingness to take part in research into their rare disease.
2. Patients and families are supportive of rare disease registries, but most do not have a registry for their condition or are unaware if a registry exists.
3. Patients and families do not feel that they are given enough information on research into their rare condition.

Diagnosis
1. Patients and families affected by rare diseases are waiting too long for a confirmed diagnosis.
2. A significant number of people with rare diseases receive multiple incorrect diagnoses before they receive their confirmed diagnosis.
3. Patients and families are not receiving sufficient information at the time of diagnosis.
4. The experiences of patients and families during the period of diagnosis can vary significantly.

Information
1. Very few patients feel that their information needs have been met completely.
2. Many patients and families have no person that they can easily approach to answer questions about their condition, care and/or treatment.
3. Many patients and families are forced to search on their own for information on their conditions and for information on further sources of support.

Coordination of Care
1. Patients have multiple health professionals involved in their routine care and treatment and often have their treatment/care administered in multiple locations.
2. Few patients have to travel out with Scotland to access their care or treatment. However, patients and families are generally willing to do so if it means seeing a specialist or accessing treatment not available in Scotland.
3. Very few patients have access to a care coordinator; however, most patients and families believe that having a care coordinator would be beneficial.
4. Where Managed Clinical Networks are available and accessed by rare disease patients, a high standard of service is experienced.
Access to Services

1. Patients and families often experience problems when transitioning from paediatric to adult services for their condition.
2. Patients are experiencing problems accessing medicines for rare diseases in Scotland.
3. Many patients and families experience difficulties accessing additional medical and non-medical services.
4. Patients and families experience difficulties obtaining financial support.
Research

Key Findings

1. Patients and families are supportive of research into rare diseases and have a willingness to take part in research into their rare disease.

2. Patients and families are supportive of rare disease registries, but most do not have a registry for their condition or are unaware if a registry exists.

3. Patients and families do not feel that they are given enough information on research into their rare condition.

Research into rare diseases is vital to enable the development of new therapies, diagnostic tests and preventative measures for patients affected by these conditions.

Key Finding 1: Patients and families are supportive of research into rare diseases and have a willingness to take part in research into their rare disease.

Only 16% of respondents felt that sufficient research was being conducted into the rare disease that they are affected by.

In addition, only 8% of respondents believe that there is enough opportunity to participate in research, with a further third (33%) of respondents being unsure as to whether there are enough opportunities to participate.

However, 61% of respondents have participated in research, with participation in a clinical trial being the most common method of involvement. These figures suggest that if made aware of ways in which to aid research, patients are generally willing to participate.

Research represents hope for many patients and families and as such, patients and families need to be kept up to date on research developments into their condition and need to be made aware of research which may be relevant for them to participate in.

In what ways have you/has the person participated in research?

“I have asked about research, especially genetic research, as there is in my opinion a family history, on several occasions but that is one of the questions the doctor pretends not to hear.”

Mother of a child with idiopathic thrombocytopenic purpura and undiagnosed joint pains and extreme fatigue
“Only reason I am aware of research is either from a consultant carrying out the study/research or through charities who fund the research.”

Patient with Wegener’s granulomatosis

“I, and I suppose others, with my condition would always participate in research.”

Patient with spinocerebellar ataxia type 8

Key Finding 2: Patients and families are supportive of rare disease registries, but most do not have a registry for their condition or are unaware if one exists.

Disease registries can provide essential information for clinical care, planning and service delivery, and are a valuable tool for the initial collection of data on rare disease patients. The research shows that the majority of patients support the use of registries and indicate that most would be willing to join one if it existed and they were made aware of it.

The research shows that only 24% of respondents were aware of a registry that exists for their rare disease. Of those who have a registry for their rare disease, only 16% have not joined. The reasons for not joining were:

- “I haven’t yet made the time to do so.” Patient with cancer
- “It is in the United States.” Patient with transverse myelitis
- “Not joined yet, but I intend to.” Patient with Dravet syndrome

In addition, 92% of respondents for whom no registry currently exists (or who are unaware of such a registry) support the creation of one. The remaining 8% are uncertain.

**Key Finding 3: Patients and families do not feel that they are given enough information on research into their rare condition.**

Only 22% of respondents felt that they were given enough information on research into their rare condition. Despite the very low number of respondents receiving this information, many respondents to the survey cited patient organisations and charities as the primary source of this information. Many other respondents reported that they are reliant on searching the internet for information on research. This highlights the need for more active communication about research projects.

“I only get this information since I joined a charity.”  
Patient with Dravet syndrome

“Information is given through the Behcet’s newsletter.”  
Patient with Behcet’s syndrome

“I get the information from the internet.”  
Patient with a neurological condition

“We have been actively involved through the Niemann-Pick Disease Group (UK), the National Niemann-Pick Disease Foundation (USA) and the International Niemann-Pick Disease Alliance in encouraging research into Niemann-Pick disease.”  
Mother of a patient with Niemann-Pick disease type C

“If i asked my consultant I am sure she would give me a rundown of the research or point me to the right websites”  
Patient with Hodgkin’s Lymphoma
Morag’s Story

Morag who has rare diabetes type MODY HNF4-alpha West of Scotland

“I’m always keen to take part in research, but it can be difficult to find out what’s happening and how to get involved.”

When I was diagnosed with Type 2 diabetes as a slim 26-year-old, it was not as much of a shock to me as it was for the GP who diagnosed me. After all I came from a whole family of unusual cases of “Type 2”, and my Mum was diagnosed at a similar age.

Five years later, I read an article in Diabetes UK’s Balance magazine about a rare genetic form of diabetes called MODY (Maturity Onset Diabetes in the Young), which runs strongly in families, usually appears by age 25, and is not associated with being overweight. In different parts of the country, my Mum and I both saw this and thought, “that sounds just like us!” As the youngest and skinniest family member (and therefore the most improbable case of “Type 2”), I was despatched to my GP with a copy of the article to ask for genetic testing. The tests revealed that we all have a form of MODY called HNF4A.

There are several forms of MODY, associated with mutations in different genes. It’s thought that only 1-2% of all people with diabetes have MODY, but it’s frequently misdiagnosed as Type 1 or Type 2 and most MODY patients have not yet been identified. Getting the correct diagnosis meant that I was able to change to more appropriate medication, giving me better control of my diabetes, and a better chance of avoiding complications later in life (poorly-controlled diabetes can lead to devastating complications like blindness, amputations and heart disease).

Since I have a rare type of a relatively common condition, my experience has been quite different from that of a lot of patients with rare diseases. I’m lucky in that I receive excellent care from the diabetes clinic at my local hospital, and have easy access to a specialist who knows about MODY. It also means that there is a large and active patient organisation (Diabetes UK3), who have been an excellent source of information and support, and without whom my family and I would probably still be misdiagnosed.

There is another side to this though, which is that better-known types often overshadow the rare types of medical conditions. Even the best patient organisations can fall prey to this. Organisations like Diabetes UK need to support the differing needs of large numbers of patients, and resources are limited. It’s inevitable that the issues facing larger groups will take priority over those affecting only a small number of us. Sometimes it feels like we slip through the cracks, all too often, I see the phrase “both Type 1 and Type 2” used to mean all people with diabetes.

Having a rare disease means that I have to know a lot about my own condition; I often need to explain it to medical professionals I meet, since many of them have never heard of MODY. I can do this, as I’ve had access to the education and resources I need, but many patients with rare diseases are not so lucky. I worry what people are supposed to do if they’re not in a position to explain their own condition to doctors.

Better awareness of rare diseases amongst the medical profession would certainly help, but it’s just not possible for all doctors to know about all conditions.

I’m always keen to take part in research, but it can be difficult to find out what’s happening and how to get involved. I think if I wasn’t a member of Diabetes UK and following the right people on Twitter, I simply wouldn’t know about any research at all. Through Twitter I heard about the Scottish Diabetes Research Network (SDRN), and I’ve signed up for a register of patients who are interested in taking part in research.

Last year I took part in a biobank. This was mainly for patients with Type 1 diabetes, but they were also looking for MODY patients to help find easier ways to distinguish between Type 1 and MODY. Once MODY patients have been correctly diagnosed, they are often able to stop injecting insulin, and gain better control over their diabetes from tablets alone, which is life-changing for them. If I can help identify more of these patients by donating samples to a biobank, then of course I’m keen to do so.

My experience with this was mixed, again due to the rarity of the type of diabetes I have. I was given lots of information about what was involved before taking part, and certainly felt that if I’d had any questions the researchers would be happy to answer them. When it came to actually giving blood samples, however, things didn’t go so well. I’m quite squeamish, so even with two separate attempts the nurse was not able to get enough blood from me. I was keen to keep

trying, but the nurse did not seem to grasp how rare MODY actually is, or understand why it was worth getting samples from this one tricky patient when they already had 3,000 patients in the biobank. In the end I was dismissed with the words “you’re not special!” To put this comment into perspective, in 2010 there were 27,910 people in Scotland with Type 1 diabetes, and just 144 MODY patients confirmed by genetic testing. I’m one of those 144 people.

Despite that, I’m still passionate about research, and I would not hesitate to get involved again when the opportunity arises. What my experience shows, I believe, is that educating people at all levels, from clinical researchers to doctors and nurses, about the rarer forms of diseases, is very important. Otherwise opportunities will be missed, and patients will needlessly remain undiagnosed, misdiagnosed, or simply dismissed as attention seeking. Most of all, it highlights the need to listen to patients. I believe that given the right dialogue and opportunities, patients and medical professionals can work in partnership to improve the lot of people with rare conditions.

*Morag asked for her surname and location to remain anonymous.
Diagnosis

Key Findings

1. Patients and families affected by rare diseases are waiting too long for a confirmed diagnosis.
2. A significant number of people with rare diseases receive multiple incorrect diagnoses before they receive their confirmed diagnosis.
3. Patients and families are not receiving sufficient information at the time of diagnosis.
4. The experiences of patients and families during the period of diagnosis can vary significantly.

Timely prevention and diagnosis of rare diseases is essential. Without accurate diagnosis, appropriate screening programmes and targeting of diagnostic tests, patients and families cannot access effective treatment, therapy, or manage their condition appropriately. A delay in diagnosis or misdiagnosis may also involve multiple avoidable appointments with doctors and consultants, incorrect treatments and diagnostic tests and significant distress. Delays in diagnosis can result in missed opportunities for intervention, allowing conditions to become progressively worse and more difficult to treat.4

The research shows that patients and families affected by rare diseases in Scotland often face long waiting times and significant hurdles in securing a final diagnosis.

**Key Finding 1: Patients and families affected by rare diseases are waiting too long for a confirmed diagnosis.**

The research shows that patients and families affected by rare diseases in Scotland can often wait a significant period of time before receiving a correct diagnosis.

Almost half (44%) of respondents had to wait over one year for a confirmed diagnosis. Of this:

- 15% waited over two years;
- 10% waited over five years;
- 7% waited over 10 years.

Worryingly, 4% of patients with rare diseases had to wait over 20 years for an accurate, confirmed diagnosis.

“It was a very difficult time. I had to see a consultant on a private basis to get a diagnosis.”

Patient with chronic mucocutaneous candidiasis

How long did you/the person affected by the rare disease have to wait for a confirmed/probable diagnosis?

Encouragingly, the results also show that almost a third (31%) of patients receive a diagnosis within a three month period. Early diagnosis of a rare disease can often result in earlier access to effective treatment, therapy or management of the condition which, in turn, may result in a gain in life expectancy or quality of life.

“Diagnosis was within three days.”

Patient with myasthenia gravis and post polio syndrome

Out of the 31 respondents who have yet to receive a confirmed or probable diagnosis, more than half (52%) have been waiting for over five years.

“I had experienced the symptoms for probably around 20 years before a diagnosis was given.”

Patient with Behcet’s syndrome

Key Finding 2: A significant number of patients and families receive multiple incorrect diagnoses before they receive their confirmed diagnosis.

An unacceptably high proportion of patients and families affected by rare diseases receive at least one incorrect diagnosis before their final diagnosis is confirmed.

Just under half (49%) of those with a confirmed or probable diagnosis received incorrect diagnoses before receiving the confirmed/probable diagnosis. Of those, more than a third (36%) received more than three incorrect diagnoses.

13% of respondents had received in excess of five incorrect diagnoses before their final diagnosis was confirmed.

Not only is misdiagnosis distressing for the patient and their family, it can lead to a deterioration of the condition as effective treatment, therapy and management of the condition is delayed.

“The continued false diagnosis of this condition nearly cost my daughter the ability to walk.”

Parent of a patient with neurofibromatosis type 1
“An earlier diagnosis could have avoided incorrect medication being given.”

Relative/carer of a patient with Dravet syndrome

“I diagnosed myself after researching on the Internet. When I went to my neurologist with my findings it was like a Eureka moment for him.”

Patient with episodic ataxia

**Key Finding 3: Patients and families are not receiving sufficient information at the time of diagnosis.**

Patients and families are not receiving sufficient information at the time of diagnosis. It is concerning that only 50% of patients and families felt that sufficient information was provided about the condition at the time that the diagnosis was given, or shortly after.

**Who was the main provider of information about the condition at the time the diagnosis was given, or shortly after?**

![Diagram showing the percentage of information provided by different providers.]

For those who did receive sufficient information about the condition at the time of diagnosis, the majority (56%) received the information from their specialist, with a further 7% receiving this information from their Genetic Centre.

“In the course of the first year we were referred to a geneticist who was very helpful.”

Parent of a child with Cri du chat syndrome

“The condition was fully explained and my consultant printed off information for me”

Patient with Takayasu’s arteritis adrenal insufficiency

Findings also indicate that GPs and midwives are not providing sufficient information at the time of diagnosis. Only two respondents received information about their condition from their GP and no respondents had received information from their midwife. Furthermore, only two respondents received information on further sources of information and support from their GP and no respondents received this information from their midwife. These findings indicate that there is a need for better training and sources of information for GPs and midwives to support rare disease patients during the period of diagnosis.

“Primary care not well informed of this condition and a lot more needs to be done with diagnosis and awareness.”

Patient with haemochromatosis
The findings demonstrate the need to promote the vital role of patient organisations for people with rare diseases following diagnosis. They raise awareness and provide information resources for public, patients and families, and medical professionals. However, only 35% of respondents were provided with information about further sources of information and support (e.g. patient support groups).

Many respondents reported having to search for information themselves. 22% of respondents had to search for information on their condition at the time of diagnosis or shortly after. In addition, almost half (47%) of those who did have information from further sources of information and support had to search for it themselves.

“Different specialists gave me different bits of information, it wasn’t very helpful and I had to do most of the work myself.”
Patient with mixed connective tissue disease

There is a need to increase the profile of existing resources, such as Orphanet5, the European portal for rare diseases and orphan drugs. Improved access to good information would lead to empowerment of both professionals and patients resulting in more informed decision making, which may ultimately have cost-saving implications for the NHS as a result of fewer inappropriate tests, treatments or therapies being administered to patients.

**Key Finding 4: The experiences of patients and families during the period of diagnosis can vary significantly for rare disease patients.**

While our research reveals serious problems with the diagnosis of rare diseases in Scotland, our findings also demonstrate that a number of patients and families were experiencing good practice.

It is encouraging to see that 21% of respondents rated the health service as ‘excellent’ during the period of diagnosis, with a further 30% of respondents rating the health service as ‘good’.

“My treatment throughout was excellent!”
Patient with spinocerebellar ataxia type 8

“Everyone concerned was very supportive and helpful.”
Patient with chronic myeloid leukaemia

However, with 29% rating the health service as ‘poor’ or ‘very poor’ and a further 20% reporting ‘average’, it is clear that there is significant room for improvement.

Opportunities for improvement, suggested by respondents, include:

“They could have listened to and taken more seriously what the patient was telling them. Some symptoms were unusual even for the rare disease and were not acknowledged.”
Relative/carer of a patient with transverse myelitis

“GP should have played an advocacy role rather than just flailing around and then providing false reassurances, when all I needed was clarity and adult discussion/information-sharing.”
Patient with phocomelia due to the drug thalidomide

“Before the diagnosis i was made to feel like i was wasting the professional’s time and that there wasn’t anything wrong with me.”
Patient with Behcet’s syndrome

5. www.orpha.net
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<th>Experience</th>
<th>Patient Details</th>
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<tr>
<td>“Better education of GPs re rare diseases.”</td>
<td>Patient with ovarian cancer (germ cell tumour)</td>
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<td>“The first GP I went to see was not helpful, I went in with, what I now know are, diagnostic symptoms and I left with moisturiser. The GP I went to for a second opinion was brilliant, ordered a scan and I was then diagnosed. It would have been helpful if the first doctor was more proactive in trying to help me rather than the wait and see approach she adopted.”</td>
<td>Patient with Hodgkin’s lymphoma</td>
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<td>“If I would have received proper tests such as blood work, scans and X-rays, then we would have come to a diagnosis quicker.”</td>
<td>Patient with cancer</td>
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<td>“Understanding how we were feeling and not being made to feel like a neurotic mother.”</td>
<td>Patient and parent of a child with tuberous sclerosis</td>
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<td>“If the various health specialists had communicated with each other diagnosis may have been made earlier.”</td>
<td>Patient with Behcet’s syndrome</td>
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Lesley’s Story

Lesley Loeliger– Patient with Paroxysmal Nocturnal Haemoglobinura
West of Scotland

“The diagnosis was not great but it was such a relief to finally know what I had. Not knowing was far worse than anything the doctor could have told me.”

I have a rare bone marrow disease called Paroxysmal Nocturnal Haemoglobinura (PNH) and I am one of only 30 people in Scotland with this disease. I am married with 2 children, have a Bachelor of Engineering Honours degree in Electrical & Electronic Engineering and I worked for 11 years in the Semiconductor industry.

It was in 2005 that I started to feel very run down and had constant colds, sore throats and headaches. I ignored them for a few months but eventually I had to go to my GP when I was so tired that I ached all over. My eyes hurt to be open and I had constant, severe headaches.

After a few months of tests, my GP referred me to a consultant haematologist. I soon discovered though that this was the start of a long and difficult journey towards diagnosis. After one and a half years attending the haematologist, undergoing countless tests and receiving several wrong diagnoses, I was no further forward. Each test raised my hopes that I would finally find out what was wrong with me. I would anxiously await the results and feel total desperation with every negative result. Harder though were the times when I was told the tests were positive only to find out a few weeks later that they were wrong and that we would have to start all over again. These months were the hardest and most stressful of any in my illness.

Eventually I was fortunate enough to be seen by a young Registrar who was on his six weeks rotation. He asked me all the usual questions and then said “I think I know what’s wrong with you”. After one and a half years, I didn’t hold out much hope! When I went back for the results he said, “I was right, you have Paroxysmal Nocturnal Haemoglobinuria or PNH”. The Registrar had studied with a doctor who now worked with a world-renowned PNH specialist. I was in the right place at the right time to just happen to meet someone who knew about PNH and who knew where there was an expert. The following week that Registrar moved on.

On my diagnosis day, I was told there wasn’t much known about the condition and I would, on average, get 10 years of life. The diagnosis was not great but it was such a relief to finally know what I had. Not knowing was far worse than anything the doctor could have told me.

During the next year I was surviving on blood transfusions. The transfusions didn’t help with my quality of life and were in fact very tiring. They also didn’t protect me from the risk of a fatal blood clot, they merely stopped my blood levels falling dangerously low. We were told about a drug, called Soliris. It is not a cure for PNH but it can give patients an improved quality of life. The drug is extremely expensive and is not funded in Scotland so I was told I was unlikely to get it. It was heartbreaking knowing there was a drug that could give me my life back but I could not get access to it. Over the next year though, I started to reject my blood transfusions and it may sound strange, but this was very fortunate! As I would not have survived without transfusions, the Scottish Government thankfully agreed to fund me for the drug. I started Soliris in May 2008. Within 6 months I started to feel able to do a lot more and since then I have gradually got stronger. The latest research shows that Soliris not only gives a better quality of life but it restores a patient’s life expectancy to normal.

Once I was strong enough, I was asked to set up a Scottish PNH patient group. I didn’t know where to start but was put in touch with Rare Disease UK (RDUK) who helped me get started and who still work with me to ensure we get the best help for PNH patients in Scotland. We held our first patient group at the start of last year where I got to meet my fellow PNH patients. They told me of their feelings of isolation, their exhaustion, their fights to get funding for the drug and their fears for the future.

The patient group, PNH Scotland, is now a fully registered charity with the aim to raise awareness of PNH help provide information for newly diagnosed patients and ensure drug funding for those deemed to need it.
Information

Key Findings:

1. Very few patients feel that their information needs have been met completely.
2. Many patients and families have no person that they can easily approach to answer questions about their condition, care and/or treatment.
3. Many patients and families are forced to search on their own for information on their conditions and for information on further sources of support.

Too often patients are given a diagnosis but no further information, and are left to research their condition on their own. This frequently results in patients finding unreliable, often alarming information and not being able to discuss this with anyone who understands the condition.

Key Finding 1: Very few patients feel that their information needs have been met completely.

The research shows that only 35% of respondents felt that their information needs have been “met completely” or “fairly well”. Furthermore, 9% felt that their information needs were not met at all.

“If there is a diagnosis then there should be more information and counselling. GPs should be more up to date with their patient’s illness instead of having to troll the internet in front of you.”

Patient with Behcet’s syndrome

“Scottish based support networks would be useful.”

Relative/carer of a patient with hereditary spastic paraplegia

“Information is vital in knowing what you are dealing with and so that you don’t feel as if you are fighting on your own.”

Patient with vasculitis

“Health specialists should have more awareness of charity support.”

Patient with cerebral vasculitis

A patient’s condition may be being managed effectively by the NHS according to best current knowledge of that condition, yet the lack of appropriate people and/or tools to communicate that information and translate into care, together with a patient’s feeling of insecurity and isolation, can lead to a perception of inadequate care. Information is a crucial element of the service a patient receives and should not be viewed in isolation or as something which is optional. Empowering patients through information is a relatively low – cost way of ensuring better management of a condition as well as increasing a patient’s satisfaction with the service that they are receiving.6

As information on rare diseases and their management is often scarce and difficult to find in comparison to information on common diseases, it is of even greater importance that patients with rare diseases are supported to obtain the information that they need.

The areas which respondents indicated that they would like more information on include:

- What to expect from the condition;
- Information for carers and families;
- Education;
- Welfare and benefits;
- Social information e.g. respite and care.

"[More information is needed on] support for siblings of children with rare conditions as they are often forgotten."

Patient and parent of a child with tuberous sclerosis

Key Finding 2: Many patients and families have no person that they can easily approach to answer questions about their condition, care and/or treatment.

A staggering 41% of patients and families do not have someone who they can approach to answer questions about their condition, care and/or treatment.

Of those patients and families who do have someone who they can approach, more than a half (61%) cite their specialist or a specialist nurse as that person. More than a quarter (29%), feel that they can turn to a charity, support worker or care coordinator (employed by either the NHS or a charity), whilst just 10% feel that they can approach their GP.

Who is the main person you can approach that can answer questions about the condition, care and/or treatment?

RDUK believes it is vital that there is someone to support patients with their queries as their disease progresses. There should also be someone available to support families affected by rare diseases, to direct them to appropriate services, to offer them support with their own needs, and to offer support to carers.
Key Finding 3: Many patients and families are forced to search on their own for information on their conditions and for information on further sources of support.

Our survey showed, many patients and families are forced to search online for information on their conditions.

At the time of diagnosis, almost half (45%) of respondents did not receive sufficient information on the condition, care or treatment. A staggering 22% were forced to search for this information by themselves.

“I was given the very basic information. It was written down and suggested that I ‘Google’ it.”

Patient with Wegener’s granulomatosis

“All information since diagnosis seven years ago has been sourced by me.”

Relative/carer of a patient with fibromuscular dysplasia

“I have generally had to seek out information myself. My diagnosis is literally one in a million, the Consultant and GP didn’t have enough to give me.”

Patient with cerebral vasculitis

Furthermore, almost two thirds (61%) of respondents did not receive sufficient information on the further sources of information and support available. Almost half (47%) of respondents were forced to source this information by themselves.

Patient organisations can be a huge source of information for patients and families, and so it is vital that people are made aware of them as soon as possible. Patient organisations are able to give up to date information on the condition, and offer support to newly diagnosed patients. Often they also run a patient forum where patients can discuss queries with others affected by their condition which is something many appreciate. It is not uncommon for patients to have never met someone with the same condition until they come across a dedicated support group. It is vitally important that patients have access to relevant patient organisations following diagnosis.

“Birdshot Support Group was set up after I was diagnosed and there is now lots of good information.”

Patient with birdshot chorioretinopathy and hereditary palmoplantar keratoderma

“If I had not been put in touch with charity (DEBRA) we would have been left in limbo.”

Relative/carer of a patient with dominant epidermolysis bullosa

“As a parent, I would have benefited from a period of intense support post-diagnosis, this wasn't offered, there is a support network for people with [osteogenesis imperfecta] but I didn't know about this until about three years post-diagnosis. I felt isolated, and was diagnosed with post-natal depression, which made life even more complicated for my wee family. I wonder if I was given sufficient support post-diagnosis if this could have been averted.”

Relative/Carer of a Patient with osteogenesis imperfecta and Von Willebrand’s disease
Cherie’s Story

Cherie Colreavey – Mother of Chloe who has cystic fibrosis
West of Scotland

“The overall service of the CF clinics and the CF Trust has been worth its weight in gold”

My pregnancy with Chloe was a completely normal one, but shortly after she was born, Chloe produced a lot of green bile. Our Doctor explained to us that this was meconium ileus and that in 95% of cases, was caused by cystic fibrosis (CF).

We were worried. Chloe was our first baby and we hadn’t really heard of the condition before, or met anyone affected by it. We “Googled” the condition at home, which we now realise was a mistake. The information provided online was devastating and we read many stories about young children spending their lives in hospital and having a very short life expectancy. We were devastated by this, the information online was just very depressing. The Staff Nurse charged with Chloe’s care that day noticed how upset we were and directed us to the cystic fibrosis Trust (CF Trust).

She also explained to us that even just during her career huge improvements had been made in the care of the condition. The CF Trust website confirmed this by showing how life expectancies have increased, that the quality of life for CF children has improved greatly and that there are new medications, treatments, therapies and research being developed all the time. This gave us, not only a better understanding of the condition, but also a much more positive outlook for Chloe.

Chloe had a blockage in her intestine and following surgery for this, and the results of a genetic diagnostic test, we were told that Chloe did have cystic fibrosis. At this time we were directed to the CF Trust for further support and Chloe began accessing the specialised CF service at The Royal Hospital for Sick Kids in Glasgow. The support and advice we received was invaluable and, even though Chloe wasn’t in hospital too long, we got to know the staff well.

We were a little nervous when we were transferred to the new CF Clinic at our local hospital, Wishaw General Hospital – but we needn’t have been, the support given to us has been wonderful. The support we received from the Specialist CF Nurse in the early days was instrumental in us coming to terms with the condition. The service we receive is personalised and rather than sitting in a waiting room with a risk of cross-infection, we are taken to our own private examination room. This is the room where we see all our health professionals, including physiotherapists and dieticians, and we can get all this done in just over an hour. The one downside of this segregation is that we rarely get to see others like us.

It was wonderful meeting Yvonne from the CF Trust for the first time. We met (without Chloe) over coffee and talked about life with CF – Yvonne told me how she has coped with having the condition and she was a real source of support to me. In particular, she helped me deal with the guilt I had been feeling. Chloe’s father and I both had the recessive CF gene and I felt a great deal of responsibility for Chloe having CF – it was extremely helpful to hear that Yvonne didn’t blame her parents and her support has helped me deal with my feelings.

Her help was so valuable that we decided a parent support group would be a great idea. Yvonne got together with the CF nurse at Wishaw General Hospital and there is now a biannual meeting of parents. The meetings are a wonderful opportunity for parents to get together, share their experiences and to hear from experts. At the last meeting, Yvonne had organised for a physiotherapist to offer advice on physiotherapy techniques – this was hugely helpful, reminding us of the importance of physiotherapy and refreshing our minds on certain techniques. The meetings have been hugely beneficial and have gone a long way to help with our ability to manage Chloe’s condition effectively.

Things are certainly not always rosy and we still have our ups and downs, but the support of the CF Trust and the CF Clinic has been incredible in helping us come to terms with Chloe’s condition. The overall service of the CF clinics and the CF Trust has been worth its weight in gold and I know that families receiving a CF diagnosis now can access the much needed support and advice to help them through the very difficult early days.
Anne’s Story

Anne – Mother of Anna who suffered from Cri du Chat Syndrome Scotland

“Without support and information we felt as though we were fighting a battle on our own.”

Anna was born in 1984 with a hole in her heart and Cri du Chat Syndrome. It took a while for diagnosis but we know now that our midwife had suspected Cri du Chat from the moment Anna was born. She had experienced the condition in her own family and recognised the signs. However, as Anna had yet to be clinically checked, she was unable to tell us this.

The night that she was born, the Doctor came to and listed a catalogue of the problems that Anna was presenting. Starting from her head down to her toes, he listed everything that was wrong in a matter of fact way before turning on his heels and heading out the door. Needless to say, I was left hysterical and the nurses left to pick up the pieces. Eventually there was a feeling, like a steel door being shut in my mind, and I knew we would have to fight for Anna.

We pushed for more information on what was wrong with Anna and asked for a diagnosis to be made, but for a long time we received nothing. Most of the health professionals we saw were wonderful, however we did have experiences of being dealt with very insensitively, on one occasion a paediatrician told us that it would be ‘better to put her in an institution and go away and have another child’. On another occasion, when Anna was having a particularly difficult time in hospital, a Doctor announced over his shoulder that it would ‘only be a matter of hours’ as he left the room.

It was somewhere between a month and 6 weeks, after pushing hard for a diagnosis, that a junior Doctor sat us down and told us that Anna had Cri du Chat. We believe now, that health professionals had avoided giving us the name because they didn’t expect Anna to live very long and because the information that was available was out-dated and devastatingly bleak. Other than a few papers from the 60’s and 70’s, there was little information on the condition. With the syndrome being so rare, there was no support group available and one neurologist even told us that he didn’t approve of support groups. Without support and information we felt as though we were fighting a battle on our own.

We did receive some support from the Scottish Association for Children with Heart Disorders and this was very welcome indeed. The lack of information we had received on the specific genetic condition prompted us to start a Cri du Chat Support Group in the late 1980s. We contacted an organisation who held a telephone list of other Cri du Chat families and from that we developed a family support group, holding a well attended first family conference in 1991. Having a support group presented a number of opportunities, for example, clinicians came from around the world to find out more, the University of Leicester produced an up-to-date and more positive information booklet with the group’s help and through dealing with a greater number of Cri du Chat families we became more aware of the wide spectrum of the condition. The value of this information and the value of having someone to talk to has been immeasurable for many families.

Although I am no longer involved with the Cri du Chat Syndrome Support Group, I know that things have changed for families receiving this diagnosis – up to date information is available, support is there if needed, research is being undertaken and the information available presents a much more positive outlook. Now, parents can ‘Google’ the condition name and find sources of information and support and, hopefully, clinicians can use this to support families through initial diagnosis.

Patient groups are a huge source of information and comfort to patients and families with rare conditions and although the diagnosis of a rare condition can still be very difficult, knowing that there is support and help available is very beneficial. In my role as Development Officer for Action for Sick Children, Scotland, I now help other families access the support and information they need. As part of my role, I worked with the National Managed Clinical Network for Children with Exceptional Healthcare Needs on a project – Early support and early expectations when your child has complex healthcare needs. This includes an ‘Early Support’ booklet with information for parents and carers and a workshop for health professionals to assist families when a baby is born with complex healthcare needs or when issues develop in early childhood. This ‘Words Matter’ learning activity workshop for health professionals helps to ensure that when bad news needs to be broken, it is done so in the best possible manner.

*Anne asked for her surname and location to remain anonymous.

9. www.ascscotland.org.uk
Coordination of Care

Most rare diseases affect multiple parts of the body and many different professionals are often involved in the care and treatment, as such there must be good coordination and communication between them.

**Key Finding 1: Patients have multiple health professionals involved in their routine care and treatment and often have their treatment/care administered in multiple locations.**

Almost two thirds (65%) of respondents have at least three health professionals across a range of disciplines, involved in their routine care and treatment.

Almost half (47%) of all respondents reported that their care or treatment was provided in more than three locations, with a staggering 10% having to attend five or more different clinics for their condition.

Attending multiple clinics can result in large disruption to a patient’s and carer’s daily life, and can make regular attendance at school or work very difficult.

The findings of our survey demonstrate that the majority of patients have their care delivered locally, with over half (53%) of respondents having to travel less than one hour to their furthest away clinic or hospital. 13% of respondents continue to have to travel over three hours to reach their furthest clinic.

These figures reflect that often care isn’t, or can’t be, provided in a local hospital so patients need to travel to bigger, regional hospitals for their appointments. Combined with the fact that patients often have to visit multiple clinics, this causes further disruption to normal life, and can have a large financial impact on the patient or the family due to the cost of travel. Improved communication and support between local and regional services would enable more care to be provided locally, reducing the need for patients to travel long distances for their needs to be met.

**Key Finding 2: Few patients have to travel out with Scotland to access their care or treatment. However, patients and families are generally willing to do so if it means seeing a specialist or accessing treatment not available in Scotland.**

The majority (89%) of respondents access their care or treatment in Scotland, with just 11% having to travel outside Scotland. This figure is encouraging as it demonstrates good service provision in Scotland. However, patients did express a willingness to travel in order to access specialist services at both the level of diagnosis and for on-going care and treatment.

One third (33%) of respondents indicated that they would be willing to travel outside Scotland to see specialists in their condition or to have procedures undertaken which are not available in Scotland.

11% of respondents would be willing to travel outside Scotland if it meant minimising the number of trips and seeing all their necessary health professionals in one visit.

For many patients, the cost of travelling outside of Scotland, or the cost of travelling a long distance within Scotland, is a concern. Patients should therefore be assisted in attending specialist clinics when it is necessary for them to do so.
“Health care appointments are difficult enough without adding extensive travelling time and increased stress levels as to how to get there, how to pay for it, etc.”

Relative/carer with osteogenesis imperfect, von Willebrand’s disease

**Are you or would you/the person with a rare disease be willing to travel to access services outside of Scotland? (Please select the statement that describes you best)**

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<th>Statement</th>
<th>Percentage</th>
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<td>25%</td>
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<tr>
<td>Willing to travel outside of Scotland, but only to see specialists in that condition/to have procedures done which cannot be conducted in Scotland</td>
<td>30%</td>
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<tr>
<td>Willing to travel outside of Scotland, but would prefer to minimise the number of trips to see all the necessary health professionals in one visit</td>
<td>5%</td>
</tr>
<tr>
<td>Prefer not to travel outside of Scotland</td>
<td>20%</td>
</tr>
<tr>
<td>Won't travel outside Scotland</td>
<td>15%</td>
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<tr>
<td>Don't know</td>
<td>10%</td>
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“We would have went anywhere to get the right answers.”

Relative/carer of a patient who died from multiple system atrophy

“I would prefer to receive the best possible care from the specialists who have the most available information. I would be willing to travel for this if it potentially meant saving my life.”

Patient with ovarian cancer (germ cell tumour)

“I would be willing to travel to access services if really necessary but I would want financial help for travel.”

Patient with chronic mycotic cutaneous candidiasis
“Health care appointments are difficult enough without adding extensive travelling time and increased stress levels as to how to get there, how to pay for it, etc.”

Relative/Carer of a patient with osteogenesis imperfecta and Von Willebrand’s disease

“Although willing to travel outside Scotland at the moment this will become more difficult/impossible as my son’s condition deteriorates.”

Patient with Niemann-Pick disease type C

“Being close to friends and family kept me strong and travelling is also stressful. It also requires parents to take more time off work and there is cost in doing this.”

Patient with cancer

**Key Finding 3: Very few patients have access to a Care Coordinator, however, most patients and families believe that having a Care Coordinator would be beneficial.**

A Care Coordinator is a general term to describe a trained professional whose role is to ensure that a care plan is in place and acted upon. The Care Coordinator’s role should include being available to talk to the patient about his or her concerns and giving consideration to the needs of the family or carer. The person best equipped to carry out the role of Care Coordinator would vary between conditions but suggestions of appropriate professionals have included specialist nurses working within the appropriate fields, members of the genetics teams or social workers, or a professional employed by a patient organisation.

A Care Coordinator can provide vital support to a patient and their family throughout the progression of their condition to ensure that care is carried out as smoothly as possible.

The research shows that less than a quarter (24%) of respondents had a dedicated care coordinator / care advisor.

“With the advent of a specialist TS clinic in Edinburgh, I finally have someone coordinating my care.”

Patient with tuberous sclerosis

“What coordination of services. I go to my GP about various problems he refers me. I WAIT MONTHS for an appointment. I WAIT MONTHS for tests. Then I WAIT MONTHS for any treatment available. That’s if I have been sent to the correct place by my GP. If not the whole process starts again.”

Patient with transverse myelitis

“The lack of co-ordination or any kind of joined-up thinking or communication is a key issue. It is crazy.”

Patient with phocomelia (due to the drug thalidomide)

“The lack of a coordinated service was exhausting and frustrating.”

Relative/carer of a patient with Hirschsprung’s disease


11. Tuberous sclerosis
“At the moment GP, Renal Consultant & Rheumatologist act independently & report through written communication, which can go astray.”

Patient with Wegener’s granulomatosis and vasculitis

48% of patients and families who currently do not have a designated care coordinator or care advisor, believed that having one would be beneficial.

“Not having a set GP or other care coordinator means that I have to explain my condition to doctors and nurses when I see my GP. And, often, I have to push to have blood work done because they don’t understand the process. If my GP or a nurse just ‘knew my history’ it would make visits to my GP less stressful and frustrating.”

Patient with idiopathic thrombocytopenic purpura

**Key Finding 4: Where Managed Clinical Networks are available and accessed by rare disease patients, a high standard of service is experienced.**

Managed Clinical Networks aim to improve integration of services and coordination between health professionals and organisations from primary, secondary and tertiary care in Scotland. Examples of Managed Clinical Networks in Scotland include, but are not limited to, The Scottish Muscle Network and the Network for Inherited Metabolic Disease (IMD Scotland).

Only ten respondents to our survey were aware of a Managed Clinical Network for their condition and were accessing services through one. Of the ten respondents, nine considered the services to be “very good” or “good”, with no respondents considering the service to be ‘poor’ or ‘very poor’.

“I am well monitored.”

Patient with myasthenia gravis and post polio syndrome

These figures show the need to promote awareness of the available Managed Clinical Networks to ensure rare disease patients have the opportunity to access these services. The opportunity to improve coordination of care exists within Managed Clinical Networks, as they facilitate communication between those involved in a patients care. Many Managed Clinical Networks have also produced clinical and care guidelines, improving the quality of care received by patients.

Gail Currie’s Story

Gail Currie – mother of Neve who has Neurofibromatosis Type 1
Edinburgh, Scotland

“I find it incredible that there is no one person co-ordinating Neve’s condition or care. I find at times that it is a full time job simply maintaining appointments and recording the necessary details of every consultation to relay to the next medical professional we see.”

I’m a wife and Mum of three living in Edinburgh. Our eldest daughter Neve is nine and has Neurofibromatosis Type 1 (NF1), a rare genetic condition.

Neve was born early at 24 weeks and successfully fought for her life. Consultants told us that generally most issues in relation to the prematurity resolve in children by the age of two. We found that by two Neve was still overly clumsy, tripped and fell often, her motor skills were challenged and other little things that when put together added up to a substantial difference from her peers. Regular visits to the GP left us feeling frustrated and it wasn’t until one of her “trademark trips over fresh air” resulted in a reflex anoxic seizure that they took notice and finally relented to refer us to The Royal Hospital for Sick Children in Edinburgh.

Naturally we were relieved that someone was prepared to listen to our concerns about our child, however we quickly realised that this was going to be a mission. Over time we had visited General Medicine, Neurology, Orthopaedics and Physio/Occupational Therapy, every time coming away with more questions than answers not to mention gaining “the porter’s guide” to several NHS establishments! Finally we were invited to take Neve to a Genetics Clinic. With one knowing look from the Genetic Consultant, I knew we were in the right place. In minutes he was almost certain that Neve had NF simply from her demeanour and a brief examination revealing small coffee coloured spots (café au lait marks) which we had assumed were birth marks. Later genetic testing confirmed the diagnosis. Finally we felt that the months of uncertainty were over, someone was going to care about our daughter’s needs and offer us assistance.

The rare and widely varied condition was explained to us and our new journey began. Someone told me once that a worried parent does better research than the FBI; I now know this to be true. The worst thing any parent can do is Google! I was terrified in minutes. I did however stumble upon Confer, a charity supporting those in Scotland with NF and run by parents with children who also have NF. They helped us discover so much about the condition – medically, educationally and socially. We were no longer alone – we had a parent network where people genuinely knew what we were living with and Neve found friends who were just like her. Within two years, I have become the Chairperson of the charity and now assist others exactly in the way we were so invaluably helped.

After diagnosis, we found ourselves living our lives around hospital appointments. Given how rare NF is, it became only too common to have to begin every appointment with an explanation of the condition, how it affected Neve and other clinicians we saw in relation to her care. Groundhog Day was the new normal and it was always a challenge particularly when attended clinics with our other young children! Thankfully I am a lover of detail and I was in a position to offer a chapter and verse history, my diary was my bible and my notes were my lifeline. I am grateful for that skill however I find it incredible that there is no one person co-ordinating Neve’s condition or care. I find at times that it is a full time job simply maintaining appointments and recording the necessary details of every consultation to relay to the next medical professional we see.

I wonder sometimes, why do they have notes? Surely when dealing with something as vitally important as someone’s health, every detail should be recorded and overseen by a doctor charged with the ownership of the patient? What happens to the patients whose parents don’t or can’t record and remember? NF is a condition which can be passed from parent to child – what happens to the kids who have parents with NF too and find processing and recalling details
challenging? The answer is that they slip through the net. Unless there is a key worker or a charity such as Confer to guide and assist, I worry for those like our daughter.

Of course we continue to be grateful for the medical care that Neve receives. She is maturing into a young lady and is very aware of her progressing condition. Our hope for her future is to live as independent a life as possible. In turn we have to hope that the health service will improve its co-ordination of care for patients who see multiple doctors in many different specialities. In the mean time I will continue our “normal life” co-ordinating school runs, hospital appointments, football matches and medical terminology!
Key Finding 1: Patients and families often experience problems when transitioning from paediatric to adult services for their condition.

More than one third (36%) of patients who have been through the transition from paediatric to adult services reported experiencing ‘many problems’ with a further 43% reporting ‘some problems’.

The high number of problems experienced demonstrates the need for the development of resources to ease transition. We suggest this should include condition specific Route Maps \(^{13}\) to lay out the general progression of the condition, and what support is available to patients and healthcare professionals at various stages of the disease. The appointment of a care coordinator would also ensure continuity and communication between all necessary people at transition.

“Not easy at first to leave the care of our excellent paediatrician in Manchester to move to adult services - at age 34! We are getting used to it now.”

Mother of a patient with Niemann-Pick disease type C

“Most of my problems with pain, bodily dysfunction and decreasing ability to cope, worsening mental health have come later in adult life, from age 40 onwards - we seem to have dropped off the scale for being any kind of priority at this age/stage of life.”

Patient with phocomelia (due to the drug thalidomide)

“Many girls fall through the system at this stage. I almost did as Adult services did not meet my needs, partly due to the consultant having no social skills!”

Patient with Turner syndrome

“My son is in the transition from children’s to adults’ services - at the moment we are not too sure who the main person to contact is, this whole transition period is incredibly stressful”

Mother of a child with osteogenesis imperfecta and Von Willebrand’s disease

Key Finding 2: Patients are experiencing problems accessing medicines for rare diseases in Scotland.

The findings of our survey show that many patients are experiencing difficulties accessing drugs for rare diseases in Scotland. 26% of respondents highlighted that they had experienced difficulties, or had been unable, to access medications for their diseases.

For many rare diseases, there are no effective treatments available. For patients who do have an effective treatment available to them, their lives, and those of their family, can be transformed. It is for this reason that experiencing difficulties when accessing drugs for rare diseases can be distressing for the patient and their family.

“Because the drugs for my condition are very expensive, it took over two years from initial diagnosis before I was allowed to have them. Only when I had reached a stage where the deterioration was extreme was I given the full diagnosis allowing the medication. By this time muscle tissue had decreased significantly and I had increased in weight from 55 to 85 kilos - increase in adipose fat/middle-body obesity - leading to various other health issues such as extreme fatigue and heart palpitations and time off work etc. Again because of cost, there is a restriction on how much the GP will prescribe at any one time. This means the amount of time I have to spend ordering and collecting prescriptions is increased.”

Patient with hypopituitarism

**Key Finding 3: Many patients and families experience difficulties accessing additional medical and non-medical services.**

- Our survey shows that patients and families experience difficulties accessing non-medical services.
- Only 13% of respondents found it easy to access respite care.
- 30% of respondents reported being able to access psychological support easily.
- Only 26% of patients and families were able to obtain equipment (for example, wheelchairs) with ease and just 24% were able to obtain home adaptations easily.
- 22% of respondents were able to access support for special education needs.

“Parents requests are not taken into account or listened to properly with regards to education.”

Relative/carer of a patient with global development delay

“My disability is a fairly major one, having very short arms, and I am truly shocked at how reluctant the authorities have been to issue assistance, adaptations and specialist advice/treatments. It took me four years to receive kitchen adaptations, and four years from being assessed as needing them - I am still waiting for bathroom adaptations such as a shower (instead of bath) and close-o-mat toilet (surely that’s essential for someone who cannot reach the lower part of their own body). As for support for my mental/emotional health, I have asked for that many times, in a state of despair and desperation - to be constantly turned away to find my own solutions.”

Patient with phocomelia (due to the drug thalidomide)

“Classroom support for changing and stoma care was always organised last minute and had to be fought for all the way. Continuity of care was difficult, Equipment through occupational therapy was not suited to small child also lack of co-ordination with hospital.”

Relative/carer of a patient with Hirschsprung’s disease

“Any help I had to get I had to find out about the service myself. Especially the equipment and home adaptations I had to do a self referral. Once I accessed these ones the care and support I received were excellent. If it was someone older and they weren’t as clued up as I feel, then I feel that they would be at a distinct disadvantage. I also feel that you have to persist with your consultant to try and gain additional support e.g. physio.”

Patient with vasculitis

“[my] local council OT was helpful in getting adaptations to bathroom but had to wait some time for this to happen - same with health professionals the wait to see a specialist neuro physician was too long.”

Relative/carer of a patient with ataxia telangiectasia
“It is always difficult to get the right information so that you can make decisions about aspects of care... if you don’t know its out there how are you able to access it?.. And when you do find out about a form of treatment, you don’t always get much information about it.”

Relative/carer of a patient with an undiagnosed condition

“Although excellent hospice care is available for children in Scotland, we have been unable to find hospice care for young adults with deteriorating neurological conditions. We have been unable to access provision of an attendant controlled electric wheelchair which seriously curtails our son’s ability to enjoy access to the local environment.”

Mother of a patient with Niemann-Pick disease type C

“I had no idea the NHS could help me with equipment until I’d bought most of what I need myself.”

Patient with connective tissue disease

Key Finding 4: Patients and families experience difficulties obtaining financial support

The survey highlights that many patients and families are not receiving sufficient information on the welfare benefits available and are experiencing difficulties when trying to obtain financial support.

30% of respondents to the survey received no information on welfare benefits, a further 36% of patients and families felt that more information was needed.

“Welfare and benefits are particularly worrying at the moment.”

Relative/carer of a patient with fibromuscular dysplasia

42% of patients and families reported experiencing difficulties in obtaining financial support and/or benefits.

“The harassment and difficulties getting and keeping Employment and Support Allowance and Disability Living Allowance have been so soul destroying at times I have thought of just ending my life. At times without warning I have been left without the means to support myself. If I had not had a relative (daughter) who fed, clothed and housed me I would not be here. Anything I have I have got because I found out about it myself and I made a complete nuisance of myself until I got a satisfactory outcome.”

Patient with transverse myelitis

“Benefit support was hard at first but my specialist nurse came to the house to help. Had more problems with the benefit people on the other end of the phone.”

Patient with cancer

Benefits, such as the Disability Living Allowance, may be available to financially assist patients with rare diseases and their families, but too frequently patients are not aware of or informed about these, and are not given the help they may require to apply for them. Other patients have reported having to battle to access benefits to which they are entitled. This may stem for the relevant authorities lacking awareness of rare diseases and the challenges patients face with these conditions not fitting into boxes easily.

People affected by rare diseases can suffer financially as a result of, for example, travel costs to and from multiple hospital appointments, the need for expensive equipment or home modifications, having to give up employment or take time off work as a result of their conditions, or having to reduce working hours. Financial difficulty adds an extra worry and stress to families and they should be helped to access the social and financial support needed.
Stevie’s Story

Stevie – affected by Thalidomide – Scottish Borders

“If I didn’t have the means to buy the things that are not provided for me, I don’t think I would be able to lead such a full life.”

My story begins in 1960 when I was born with birth defects. My mother, having suffered from morning sickness during pregnancy, had been given the drug Thalidomide. We know now that was a teratogen which caused many different forms of birth defects.

My mother struggled to obtain a diagnosis for me for many years, hampered by the fact that her GP refused to admit to giving her the Thalidomide drug. My mother fought hard to be taken seriously by health professionals, to ensure that I was given the treatment and care I needed and to ensure that I had the best quality of life possible. Despite being told each year that I probably wouldn’t see my next birthday, my mother continued her struggle. By the time my 21st birthday came along, they gave up telling her that I wouldn’t live much longer!

My mother fought to get me into a mainstream school and I am glad she did, I had a great time playing rugby, golf and generally running about. Her commitment to ensuring I led a normal life meant that my parents had to make many difficult decisions.

Indeed, at 13 months old, my parents had to make the very difficult decision to amputate my leg. I received a ‘rocker’ artificial leg which was wooden, heavy, and cumbersome and looked a lot like what Long John Silver would have worn. Then, there wasn’t much choice; you had to take what you were given by the NHS.

Fast forward to now and there is a much greater choice of equipment available for individuals with disabilities such as myself. However, much like when I was younger, it is still a case of having to take what you are given – even if what I am given isn’t suitable. For example, I have been given a wheelchair to assist me. However, it is not suitable for my needs and affects my ability to get out and about. As such, I have had to buy my own power chair to be able to go out shopping and to do the things I need to.

In my experience, local authorities make it extremely difficult to access what you need. There is always a very long wait for home adaptations or for much needed equipment and often, I am given the most basic of equipment. I maintain that this is false economy, why give me cheap, poor quality equipment that has to be frequently replaced when I could have some, perhaps slightly more expensive, that would last me for a number of years and fulfil my needs. It is such a battle to get the things I need that I often, at expense, end up having to buy things for myself. I am lucky that I can do that, but I am sure there will be many people unable to do that and having to settle for insufficient equipment or even no equipment at all.

In addition to accessing equipment, I also have problems obtaining access to buildings! In my town, most residents have the choice of ten hairdressers – I have no choice – I can only go to one as the other nine are inaccessible to me. I don’t get trains anymore as I can’t rely on there being someone at the station to help me off. I am an avid football fan and enjoy travelling to away games but there are still some stadiums that are not accessible to me.

I worry about the financial support too – the changes to the Independent Living Fund and Disability Living Allowance mean it is harder for people like me to get the appropriate care. Often, with a rare condition such as mine, I don’t fit the pigeon holes to allow me to access what I need.

Because I have been able to buy most of the things that I need, I have been able to pursue my interests in football and animals – I campaign for justice for others affected by Thalidomide and offer support to others in my position. If I didn’t have the means to buy the things that are not provided for me, I don’t think I would be able to lead such a full life.
Appendix 1: List of Conditions represented in the survey

Patients and families affected by the following rare conditions responded to the survey:

- Acute intermittent porphyria
- Addison’s disease
- Alkaptonuria
- Alpha-1 antitrypsin deficiency
- Antiphospholipid syndrome
- Atypical haemolytic uraemic syndrome
- Ataxia:  
  - Ataxia SCA 2
  - Ataxia Telangiectasia
  - Episodic ataxia
  - Friedreich’s ataxia
  - Spinocerebellar ataxia type 6
  - Spinocerebellar ataxia type 8
- Behcet’s Disease
- Birdshot chorioretinopathy
- Cancer
- Cerebral vasculitis
- Chiari Malformation and Syringomyelia
- Chronic myeloid leukaemia
- Churg-Strauss syndrome
- Common variable immunodeficiency
- Craniofaryngioma
- Cri Du Chat Syndrome with the possible involvement of other material from another chromosome
- Diabetes type MODY HNF4-alpha
- Dominant epidermolysis bullosa
- Dravet syndrome
- Fabry disease
- Fibromuscular dysplasia
- Fragile X syndrome
- Functional movement disorder
- FXTAS
- Global development delay
- Granulomatosis with polyangitis
- Haemochromatosis
- Hereditary spastic paraplegia
- Hirschsprung’s disease
- Hodgkin’s lymphoma
- Huntington’s disease
- Hypopituitarism
- Hypotonia
- Ichthyosis
- Ichthyosis X linked
- Idiopathic intracranial hypertension
- Idiopathic thrombocytopenic purpura
- Idiopathic thrombocytopenic purpura and undiagnosed joint pain, extreme fatigue
- Immunoglobulin G deficiency
- Langerhan’s cell histiocytosis
- Laurence Moon Bardet Biedl syndrome
- Leukocytoclastic vasculitis
- Limb-girdle muscular dystrophy
- Lupus
- Lymphangiomatosis
- Lymphocytic colitis
- M.E
- Metachromatic leukodystrophy
- Microscopic polyangiitis
- Mixed connective tissue disease
- Multiple system atrophy
- Myasthenia gravis
- Myotonic dystrophy
- Nephrotic syndrome
- Neurofibromatosis Type 1
- Niemann-Pick disease type C
- Osteogenesis imperfecta
- Osteogenesis imperfecta, Von Willebrand’s Disease
- Ovarian cancer (germ cell tumour)
- Phocomelia (due to Thalidomide)
- Pitutary adenoma
- Palomoplantar keratoderma
- Paroxysmal nocturnal haemoglobinuria
- Polychondritis
- Postural orthostatic tachycardia syndrome
- Retinoblastoma
- Ring chromosome 20
- Severe gut motility disorder
- Sleep aponea
- Systemic mastocytosis
- Takayasu’s arteritis
- Tuberous sclerosis
- Turner syndrome
- Undiagnosed condition involving respiratory problems, global development delays, renal problems, dysmorphic features, umbilical hernia and optic nerve problems
- Undiagnosed condition involving variable muscle tone, hip dysplasia, repaired cleft lip and palate
- Undiagnosed condition involving learning disabilities
- Undiagnosed condition involving severe learning difficulties, seizure disorder, macrocephaly, allergic enteropathy, visual impairment, adrenarche and possible metabolic disorder
- Undiagnosed neurological condition
- Urticarial vasculitis
- Vasculitis
- Weaver syndrome
- Wegener’s granulomatosis