



RARE DISEASE | UK

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



The Rare Reality – an insight into the patient and family experience of rare disease

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. Membership is open to all and includes patients and family members living with a rare disease, patient organisations, clinicians, researchers, academics, and industry. RDUK provides a unified voice for the rare disease community, capturing the experiences of patients and families and raising the profile of rare diseases across the UK.

RDUK was established in 2008 following the European Commission's Communication on Rare Diseases: Europe's Challenges. Following this Communication, RDUK successfully campaigned for the adoption of the Council of the European Union's Recommendation on an action in the field of rare diseases (June 2009). The Recommendation outlines how EU member states should develop and implement plans or strategies for rare diseases by the end of 2013.

Since the adoption of the Council of the European Union's Recommendation, RDUK until November 2013 worked to ensure that the UK's health departments acted on their obligation to develop a UK Strategy for Rare Diseases, and worked to engage the rare disease community to shape its content. We are now focused on ensuring the implementation of the UK Strategy is effective and accountable.

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Disclaimer

The views and opinions expressed in the rare disease case studies, and the quotes from patients, are the respondents' own, and do not necessarily reflect the views of RDUK.

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Chair's Foreword

For many years 'rare' was synonymous with 'not many affected, not much can be done, so not very important'.

Rare diseases are higher on the healthcare agenda today than they have ever been. Researchers and clinicians are more aware of the impact of rare diseases, and more interested in patients and families with these conditions than ever. At least 6000 different rare diseases have been identified, and as research progresses more are identified, and the list grows on an almost daily basis. NHS England's budget for directly commissioned services, many of which are exclusively focussed on patients with rare or very rare conditions, is in excess of £10 billion a year. Scotland, Wales and Northern Ireland devote a similar proportion of their healthcare budgets to rare disease patients and their families. The International Rare Disease Research Consortium is stimulating a global effort to develop a diagnostic for every rare disease by 2020.



With all this attention you might think that the experience of patients and families, looking to the NHS, would be improving in leaps and bounds. Five years ago Rare Disease UK surveyed over 600 families. The results painted a mixed picture. While parts of the NHS were 'quite excellent' the experience for many was that of delayed diagnosis, misdiagnosis, poorly coordinated care and difficulty accessing appropriate information. This year, in a much larger survey, we find that this mixed bag is still the reality for many. Delayed diagnosis, access to expert help and poorly coordinated care are still the experience for too many rare disease patients and their families.

Of course, things are not all gloomy. There are more therapies licensed than there were five years ago. The advent of Genomics England and the development of Genomic Medicine Centres and Genomic Clinical Interpretation Partnerships, as well as the establishment by Public Health England of the National Congenital Anomaly and Rare Disease Registration Service bode well for the future. The National Institute for Health Research's Rare Disease Translational Research Collaboration (NIHR RD-TRC) is delivering promising outputs which should result in better understanding and new therapies.

All these, however, will take time, effort and a sustained commitment to realising the fundamental principles underpinning the NHS, and delivering to everyone a service that meets their needs in ways that reflect current scientific understanding and best clinical practice.

What this survey reveals is that, despite progress in some areas, further improvement is needed and more attention must be paid to developing processes that will ensure awareness of rare diseases permeates every corner of the health service – like letters running through a stick of rock.

The framework to make this ambition a reality already exists. The UK Strategy for Rare Diseases is a six year vision incorporating 51 commitments that must be achieved by 2020. Systematically implemented across the four nations of the UK it can deliver high quality healthcare to patients and families with rare diseases. As this report clearly indicates, there is much that needs to be done if the advances experienced by some are to become the experience of all.

Rare disease patients are realistic. They know that things take time and that progress does not happen uniformly across all disease areas. However, their patience is not infinite, and their tolerance for the inefficiencies and the waste of scarce resources that is too often part of their daily experience of healthcare cannot be taken for granted. Concerted, sustained efforts to deliver the commitments in the Strategy by all four national health services across the UK should be a priority if the good things highlighted in this report are to become the norm. And if the inefficiencies and inequalities highlighted all too often in its pages are to be rooted out and become a thing of the past.

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

Introduction

In the UK, 1 in 17 people will be affected by a rare disease at some point in their life; this amounts to approximately 3.5 million people. Collectively, rare diseases are not uncommon. There are over 6,000 known rare conditions. A single rare disease may affect up to 30,000 people in the UK. Others may affect just one.

In November 2013, RDUK welcomed the UK Strategy for Rare Diseases. It was the first time since the establishment of the NHS that patients and families affected by rare conditions have had a clear and strong commitment from government that their healthcare needs will be met. The UK Strategy for Rare Diseases is signed by the health ministers of each country in the UK, with cross-party support, and contains 51 distinct commitments that are designed to improve health and social care for rare disease patients. Effective implementation of the Strategy will ensure people living with a rare disease have access to the best evidence-based care and treatment that health and social care services, working with charities, researchers and industry can provide.

Over the summer of 2010, three years before the UK Strategy for Rare Diseases was published, Rare Disease UK carried out a survey of patients and families affected by rare diseases. The aim of the survey was to find out more about the experiences of people living with a rare condition in the UK, and to identify some of the common issues they frequently face. The problem areas highlighted from this survey in the report 'Experiences of Rare Diseases: An insight from Patient and Families' were around diagnosis, research, information, care coordination and access to treatments.

Just over five years on, and following the publication of the Strategy, which addresses these issues in policy, there is a need for an up to date picture of the patient experience of rare disease. We therefore carried out a second survey to determine if the situation for rare disease patients in the UK has changed in the last five years, and if the recommendations from the Strategy have started to be implemented and positively affect patients' care and treatment.

A survey of 85 questions was conducted over the summer of 2015. The survey was open to anyone affected by a rare condition. It was shared extensively across our networks and by many other rare disease patient organisations. In total 1203 eligible responses were received and analysed (non-UK residents were excluded from analysis). Over 450 different rare conditions are represented in the survey. The survey results are supported by ten in-depth case studies of parents and patients affected by rare conditions. For further information on methodology please refer to annex 1.

82% of respondents live in England, 9% in Scotland, 5% in Wales and 3% in Northern Ireland. The remaining 1% indicated their residence as 'other UK'. This is broadly representative of the population of the UK as a whole and as such we are able to suggest that our findings are a reasonable representation of the general situation in the UK.

We hope this report will provide a useful comparison point for a further survey in 2020, the deadline given in the Strategy for its 'vision to be reality'. Four chapters correspond to both the structure of the Strategy and the 2010 report: patient empowerment, diagnosis, coordination of care and research. A further chapter attempts to address issues around access to treatment raised in our 2010 report. We also dedicate a chapter to undiagnosed conditions, which are often extremely rare conditions that haven't been identified yet. Undiagnosed patients are frequently overlooked yet they face similar barriers to those that are encountered by rare disease patients.

The report that follows presents an updated insight into the patient and family experience of rare disease. We hope it will promote effective implementation of the UK Strategy for Rare Diseases across the UK and help to ensure that no one gets left behind just because they have a rare disease.

Summary of key findings

Very little has changed for rare disease patients in the last five years. Patients are still experiencing difficulties in diagnosis, accessing information about their condition, receiving appropriate coordinated care, accessing treatments and finding out about research.

Patient Empowerment

1. Patients and families are given very little information about their condition, both at, and after, their first diagnosis.
2. Patients are frequently left to research their condition (with very little help or direction), unless they connect with a patient organisation, who are often a vital source of both information and support.
3. Patients who become an expert in their own condition are often left to inform and educate the medical professionals they encounter.
4. Rare disease affects many aspects of an individual's life including their social, educational and employment opportunities. Increased awareness and understanding, including adjustments where needed, can help to reduce this impact.

Diagnosis

1. Patients face significant delays on their journey to secure a diagnosis.
2. The majority of patients receive at least one incorrect diagnosis, and visit numerous doctors, before they receive a final diagnosis.
3. Patients can experience issues persuading medical professionals to believe their symptoms and describe how their condition is initially written off as 'psychological' or, parents are described as 'neurotic'.

Undiagnosed

1. Patients without a diagnosis do not feel they are given enough information and support on their journey to get a diagnosis.
2. Not having a diagnosis is a significant barrier to accessing coordinated care and appropriate treatment.
3. Many undiagnosed patients and families never stop hoping for a diagnosis.

Coordination of Care

1. Information is not shared effectively between hospital trusts, or between specialist and local services.
2. Patients frequently have to attend multiple clinics and travel significant distances to them.
3. Most patients do not receive coordinated care and feel they are left to coordinate their own care.
4. Many patients do not know if there is a specialist centre for their condition.

Access to Treatment

1. For many patients the day-to-day challenges of managing their condition are made worse by the absence of an effective treatment.
2. In the absence of effective licensed medicines, unlicensed or off label medicines are an important route of access to treatment for rare disease patients.
3. Where a treatment has been licensed for a rare condition, patients may struggle to access it due to the complexities of appraisal and commissioning processes.

Research

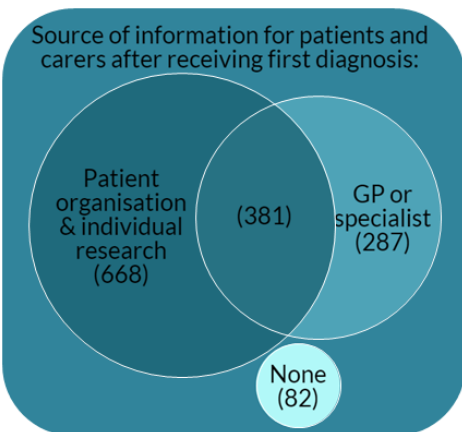
1. Most patients would participate in research if given the opportunity.
2. Patients highly value research into their condition but do not feel like they are given enough information about it.

Patient empowerment

Information

Accurate, reliable and up-to-date information is vital for patients to be able to make informed choices about their condition. People with long term conditions are likely to spend less than 1% of their time in contact with health professionals^[1]. The rest of the time they, their carers and their families, are left to manage on their own. The UK Strategy for Rare Diseases, and the respective implementation plans that followed in Scotland, Wales and Northern Ireland, as well as NHS England's Five Year Forward View, all acknowledge the importance of improving access to information about their condition and their personal medical history. Sadly, our survey indicates that most patients and families continually find themselves being given very little information about their condition, both at the time of, and after, their first diagnosis.

Nearly 70% of respondents did not feel they were provided with sufficient information on their condition following diagnosis. In addition, 35% of respondents indicated that they did not understand all the information they were given.



65% of respondents were provided with information by a specialist and 45% indicated that they had to research the information themselves. Nearly 10% of respondents said that they did not receive any information at all.

"When we were told by [the] neonatal doctor about [the] diagnosis, he simply read it off a page and walked away without any discussion." – Parent of a child with trisomy 13.

"More information about the actual diagnosis would have been good! We were told over the telephone and later received a print out from Google through the post on the condition." – Parent of a child with Coffin-Siris syndrome.

"I am under a brilliant neurologist... he was very honest right from the start and has always given me any information regarding my condition or treatment even if it was written for doctors. He also listens and will do what I feel is the best for me. This is important as you need to feel in control of something that you have very little control of." – Patient, chronic inflammatory demyelinating polyneuropathy (CIDP).

"If it wasn't for the internet I would know nothing about my condition." – Patient, cavernous hemangioma.

Key Finding: Patients and families are given very little information about their condition, both at the time of, and after, their first diagnosis.

65% of respondents felt like they have sufficient knowledge of their condition. Many patients are experts in their conditions, and spend a lot of time on research. There are however some concerns around access to correct, reliable information.

"Everything I know about my condition is due to my own research on the internet. I am unaware how much of this is correct or what I'm missing." – Patient, Morquio syndrome.

"I would love more information from the doctors and not have to rely on what I have researched online as I may have things wrong or misunderstood." – Patient, hereditary neuropathy with liability to pressure palsy (HNPP).

"[My knowledge is] sufficient as I have 'googled' for information. I would like to have some clear and easy to understand information that I could pass to others to read rather than me trying to explain what I struggle to understand myself." – Parent of a child with C3 glomerulonephritis (C3GN).

Patients need to be given information in a format that they understand. We asked patients what format they received information in and what format they might like to receive information in if they had the choice. Chart 1 compares the preferred format of information to what is received. It is clear that although websites and leaflets are the most sought after, and the most frequently given out, demand is not being met.

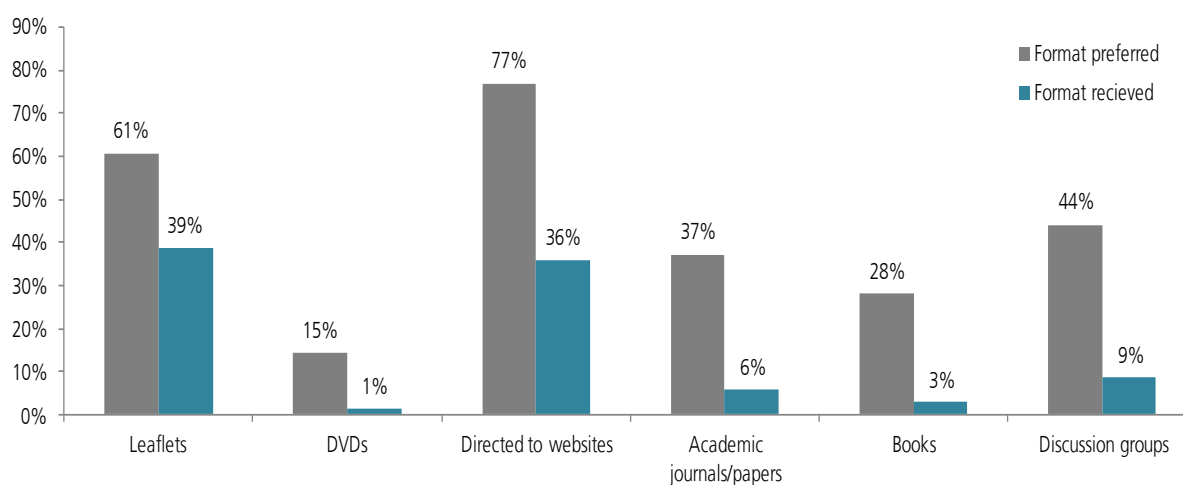


Chart 1: Information types. Comparison of format preferred vs format received by respondent

When asked, respondents suggested that they would like more holistic information on a number of areas relating to their condition. This included information around the cause, underlying genetics, treatment options, the effects of treatment, research, the prognosis (including what the future might hold) and simply 'everything'.

"[I would like to know] why she's got these conditions, what are the long term issues, are there any complementary therapies that might help and what exactly is expected as an outcome from each operation?" – Parent of a child with idiopathic intracranial hypertension.

"Because it's so rare and the treatment I received so new, I don't know how likely my condition is to relapse, I have no real prognosis, and no idea of how long I might live. I also still do not fully understand how the disease works." – Patient, Wegener's granulomatosis.

"I would like to know how it will affect my children. I believe it is genetic but am waiting to see the consultant to discuss this and the implications for my children." – Patient, multiple conditions.

Ongoing information and support

Encouragingly, 44% of respondents felt like they had a specific person they could go to, such as a specialist or a member of a patient organisation, with questions about the condition. However, this still leaves 50% of patients with no one to turn to and 6% who did not know.

Information is a vital tool in empowering patients. Patients need a reasonable understanding of their condition to make informed decisions about care and treatment. Empowering patients to become their own expert is vital to ensuring that they continue to receive the correct care and appropriate ongoing support. Patient groups play a crucial role in providing information, support and reducing feelings of isolation. However, 4 in 5 respondents were not given information on patient support groups by a specialist at the time of diagnosis.

Due to the large numbers of rare diseases, a support group for every rare disease cannot exist, this means that if patients and families do not get information from a specialist they may never get the information they need.

"It is the most isolating part [that there are] no groups, no one to share the illness with." – Patient, mesenteric panniculitis.

When individual research and research carried out online is excluded, patient organisations, specialists and other families were indicated to be the main source of information for respondents.

"[I needed] more information at initial diagnosis and an appointment with a specialist nurse to help access support. Patients need to have a voice in order for care and support to be relevant. If they're not involved, the available support is a 'supposed need' instead of an 'actual need'." – Patient, autoimmune hepatitis.

Key Finding: Patients are frequently left to research their condition (with very little help or direction), unless they connect with a patient organisation, who are often a vital source of both information and support.

Educating professionals

Patient experts often find themselves educating the medical professionals they encounter about their condition:

"On many occasions I end up explaining and 'educating' professionals about my condition as they have little knowledge and/or have not seen a case like me before. I am also apparently not a 'typical' Cushings patient so I tend to defy what is expected and confuse doctors! My questions are often unanswered, or I am told honestly that they do not know the answer." – Patient, pituitary apoplexy and Cushings disease.

"I like being able to talk about my condition to medical professionals to increase their awareness. My consultant made me feel like I am doing something positive by him using me to teach others about the condition." – Patient, multiple cylindromas.

"Every time I go to a new hospital I have to educate professionals about myasthenia." – Patient, myasthenia gravis.

"You are the expert but you are rarely heard... patient led organisations need more power to educate." – Parent of a child with fetal valproate syndrome.

The importance of the patient voice should be acknowledged and the detailed knowledge of patients respected. Patients are experts on the day-to-day effects of their condition. The UK Strategy for Rare Diseases sets out appropriate education programmes and as awareness increases, patients should feel more encouraged and comfortable in their knowledge of their rare condition.

Key Finding: Patients who become an expert in their own condition are often left to inform and educate the medical professionals they encounter.

Planning of services

There has been a positive move towards patient involvement in the planning of services as the 'patient voice' is being recognised as an important element. The UK Strategy for Rare Diseases committed to 'make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services'.

A small percentage (8%) of respondents indicated that they had been involved in the planning of services at some level. This is an encouragingly high proportion, given that engagement is never going to be comprehensive, and will be based on a representative approach. Examples of how they were involved include: representation on NICE committees, through patient groups and/or charities, as a representative in local authorities and through Healthwatch.

Nearly half (48%) of respondents indicated that they would be interested in being involved in the planning of services. 38% of respondents were unsure and 14% uninterested.

As set out in the Statement of Intent from NHS England and the national plans for rare diseases from the other nations, all four countries in the UK have patient representatives on the relevant planning and monitoring groups. For example NHS England has patient representatives on their Rare Disease Advisory Group and NHS Wales involves patients in the Welsh Implementation Group for Rare Diseases.

What has changed in the last five years?

Only 1 in 3 of the eligible patients felt like the information and support they had received in the last five years has improved.

When asked if information provision had improved in the last five years respondents who indicated it had, stated that this was often due to self-research, or more information available via the internet, often referencing the importance of online support groups.

“There is a little more information available from official health organisations than there used to be, but overall, the provision of information and support is still incredibly poor.” – Patient, Sweet’s syndrome.

“My condition now has a Facebook group where people with the condition and their families talk about what helps them, this wasn’t around when I first was diagnosed so this has been a great advancement.” – Patient, erythropoietic protoporphyria (EPP).

“Yes, but I have searched that information out for myself - partly by finding the right people and talking to them. I am very rarely handed the information (in fact, I am usually the person giving it to others).” – Patient, MODY (maturity onset diabetes of the young caused by changes in HNF4A).

The wider impact of rare conditions

Both patients and carers who responded to the survey highlighted the huge impact a rare condition can have on all aspects of their life. 2 in 3 respondents (66%) said their rare disease, or their caring responsibilities, affected their ability to hold paid employment. Similarly 56% of respondents indicated that their rare condition affected their education.

The wider impact of rare conditions raised by respondents included the social impact of a rare disease. Many respondents described feeling isolated, particularly when trying to explain their condition and how it affects them. Only one fifth of respondents (21%) felt they received sufficient social support.

Difficulties making long-term plans or maintaining employment due to uncertainty and the unpredictable nature of a condition were frequently described. A recurring theme was that numerous medical appointments get in the way of everyday life and make working, or education, extremely difficult.

Whilst some respondents explained they took early retirement for ill-health, many respondents described feeling forced into early retirement (or reduced hours) by their employer or because they felt ‘unreliable’.

“I don’t feel employers realise how debilitating these conditions are – I am currently on a first written warning for being absent from work due to my condition after first diagnosis whilst in hospital. I am on a phased return at present but feel I am being pressured to go part time.” – Patient, Wegener’s granulomatosis.

“I have to take leave to attend all of the multiple appointments my son has to attend. In addition I am currently signed off sick due to the stress associated with caring for my son with a rare condition.” – Parent of a child with alternating hemiplegia of childhood.

“Because MG is rare and most people I meet have never heard of it, it can be difficult finding the right words to explain the condition in a work environment.” – Patient, myasthenia gravis.

“I resigned from a full time position because I was about to go down a route of disciplinary action and forced job change on the basis that I took planned unpaid leave for a lumbar puncture and [my employers] felt that I was no longer capable of carrying out my duties when they became aware of my condition.” – Patient, idiopathic intracranial hypertension.

More education and awareness for employers (and in academia) should be available to enable them to make adjustments to ensure people with a rare condition can continue working if they want to. There were a number of examples where adjustments had been made successfully.

“I work flexi time so have no set start time which helps enormously when I am having a bad morning (which is most mornings as I have other chronic conditions as well). I also have disability sickness absence as well as normal sickness absence so that helps. I wouldn’t be able to hold down a full time job otherwise.” – Patient, Cushing’s disease.

“Care and support plans can be a great help. My university has a really good plan in place to support me.” – Patient, Ehlers-Danlos syndrome (type 3).

The survey responses highlight the pressures having a rare disease can put on paid employment and unsurprisingly many patients have financial concerns. Sadly, only an eighth of respondents (13%) felt they received sufficient support with these concerns.

Having a rare disease can be difficult and impact on a person’s psychological state in many ways, this should not be overlooked.

“This whole experience of being diagnosed has had a massive impact on my mind and confidence.” – Patient, acromegaly.

“Having to search for all the information myself has led me to various levels of misinformation which has only added to my stress.” – Parent of a child with Chiari malformation, idiopathic intracranial hypertension.

It is not uncommon to hear patients who develop mental health problems alongside their rare condition yet only 1 in 7 respondents felt they received sufficient psychological support. For serious genetic conditions, access to a genetic counsellor is vital and, as many respondents highlighted, increasing general awareness and understanding of rare conditions is extremely important.

Key Finding: Rare disease affects many aspects of an individual’s life including their social, educational and employment opportunities. Increased awareness and understanding, including adjustments where needed, can help to reduce this impact.

Dee, 57 – experience of information and support

Dee has neutropenia, suspected early-stage lupus, and fibromyalgia (all autoimmune conditions). Neutropenia means patients have low levels of the white blood cells neutrophils. These cells fight infections. People with neutropenia are more vulnerable to infections and in some people infections can spread quickly through the body and lead to a life-threatening condition called sepsis.



A few years ago I told my GP that I wasn't happy taking so many tablets. I'd had thirteen courses of antibiotics in just six months. Throughout my life I was frequently ill with chest infections, skin infections, eye infections etc. They were treated with multiple doses of antibiotics – one dose was rarely enough – but the symptoms would often return. I knew there had to be a reason why I was so susceptible to infections.

My GP sent me for a blood test. The results showed that my blood levels were a little lower than what is deemed 'normal' and I was referred to a haematologist. I was eventually told that I was suffering from a mild, and unexplained (idiopathic), form of neutropenia. I was advised that there were no treatments available and that I would be monitored through quarterly blood tests. At this time I was still getting infections and I was told very little about what was happening. My own research into the condition was very worrying: the information I came across frequently referred to leukaemia. I had a bone marrow biopsy but wasn't told why, and, according to my doctor, the results appeared fine.

I did not know what was happening, why, or even what the future might look like. I joined a US Facebook group called Neutropenia Support and from this I learnt a lot about the condition including that, in the US, neutropenia patients were being treated. I began to feel a little angry about why I wasn't. I obtained my medical notes for the past ten years – I wanted to know more about my condition and find an explanation for my history of ill health.

I approached the Severe Chronic Neutropenia International Registry. They gave me the contact details of a UK professional who was a member of their advisory panel. I contacted her, we then had a phone consultation and I shared my medical notes. Subsequently she wrote to me and advised that I might benefit from a treatment called GCSF (granulocyte-colony stimulating factor). GCSF stimulates the bone marrow to produce neutrophils and would hopefully enable my levels to fall within the normal range.

I took her letter, and my notes, to my next haematology appointment (which was with a different person). He agreed that we should trial the GCSF treatment for six months. I responded well. My blood levels improved, I had no infections, and generally felt a lot better. I have now been on this medication for nearly four years and have significantly improved. I do still get the odd infection which requires antibiotics but no more than other people – perhaps less.

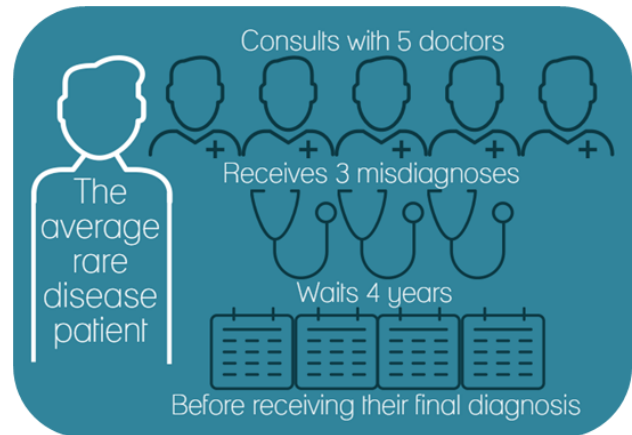
It shocked me that there was no support network in the UK. In the US Facebook group, I had met and regularly contacted UK residents who, like me, had experienced problems and misunderstanding and had encountered a lack of knowledge about the condition. They too felt isolated and worried. So, in 2013 I started a new Facebook group called the UK Neutropenia Support Group. This allowed people in the UK to come together to support each other and share experiences. The group has grown to almost 250 members including patients, parents, friends and family. Although none of us are medical specialists, between us we have a lot of experience.

Diagnosis

The long and difficult journey many patients have to go through to secure a rare disease diagnosis has been widely acknowledged across the healthcare sector (and dubbed the ‘diagnostic odyssey’). The UK Strategy for Rare Diseases committed to ‘work to achieve reduced times for diagnosis of rare diseases’. Delayed diagnosis can have a significant impact on a patient’s health, treatment options and even life expectancy. Early diagnosis and intervention can enable patients to access the most effective treatments in a timely manner, and importantly, help them find answers about their condition.

Our 2010 report highlighted that patients and families affected by rare conditions in the UK often face significant delays in securing a final diagnosis, and sadly it appears that little has changed.

Following the onset of symptoms nearly half (45%) of all respondents waited over one year to receive a diagnosis and 1 in 4 patients had to wait over five years for a diagnosis (a larger percentage than we found in 2010). Rare disease patients wait an average of four years for their diagnosis. This could be attributed to a number of factors including an increase in the number of babies surviving into childhood (thanks to advances in medicine, technology and care) as well as the increased number of respondents to the survey.



“A lot of time was wasted at the beginning by my GP who didn’t believe my symptoms were real - even though physically she could see that my muscles were wasting. It took 12 months for a new doctor to finally believe me and send me to see a consultant. My doctor at King’s College Hospital has said that if they had caught my condition earlier I might not have been so bad.” – Patient, polymyositis/scleroderma overlap, U3RNP positive, telangiectasia and Raynaud’s phenomenon.

“The worst thing is not knowing what is wrong with you. I waited 18 years for a diagnosis.” - Patient, hypermobile Ehlers-Danlos syndrome and postural orthostatic tachycardia syndrome (PoTS).

Due to the large number and the complex nature of rare conditions it would be impossible for primary care staff (such as GPs) to know and recognise every single rare condition. Many patients acknowledged this but felt that more awareness of rare diseases was needed.

“Any rare disease with a range of symptoms is difficult to diagnose. My condition was only recognised by a consultant who had seen a similar case 13 years earlier.” – Patient, Whipple’s disease.

“As my condition is very rare, it is not surprising that it was not correctly identified. However, the delay did not help me health wise - the disease had more time to damage my body and organs.” – Patient with a number of rare conditions including Raynaud’s syndrome and polymyalgia rheumatica.

“[it is] not always easy to reach a correct diagnosis if symptoms overlap with other conditions.” – Patient, mal de débarquement syndrome.

Awareness raising, training and education initiatives can have a positive impact on the ‘diagnostic odyssey’^[2]. There has been a concerted effort in some areas to improve diagnostic pathways for rare diseases but sadly many patients on the ground are still waiting for this change.

Key Finding: Patients face significant delays on their journey to secure a diagnosis.

Our research shows that most patients have to see numerous doctors on their search for a diagnosis. 71% of patients had to see over 3 doctors and 1 in 10 patients stated that they had to see more than 10 doctors before getting a final diagnosis – an average rare disease patient consults with 5 doctors. The high number of patient-doctor interactions currently required to secure a diagnosis puts unnecessary strain on NHS resources. With improved diagnostic pathways this could be reduced.

A number of respondents highlighted delays in getting the correct referral, or delays in receiving specialist treatment. This could be an important step in reducing the diagnostic odyssey.

“My GP diagnosed me, gave me a letter and sent me to hospital. The letter was ignored which delayed treatment. Tests were done for other things first.” – Patient, Guillain Barre syndrome.

“I struggled to get a referral to a neurologist from my GP. My GP was adamant I didn't have dystonia because I could walk around his office. We had to demand a referral. He didn't accept it until I received my diagnosis.” – Patient, dystonia (DYT1).

“I was not referred to [the specialist centre] by my consultant for a number of years and I could have been receiving specialist care much earlier than I did.” – Patient, paroxysmal nocturnal haemoglobinuria.

Over half (52%) of people said they had been given an incorrect diagnosis before receiving their final diagnosis. The average number of misdiagnoses was three. Misdiagnosis can be extremely stressful for patients and family members; it can prevent access to effective treatments, lead to incorrect, potentially damaging treatments, being prescribed and even cause unnecessary deterioration in the condition. The percentage of individuals reporting a misdiagnosis is higher than was previously indicated in 2010. This could be due to the increased numbers of patients surveyed or a change in diagnostic practice but further investigation would be needed to determine if this is the case. 37% of patients had received three or more incorrect diagnoses before getting a final diagnosis.

Key Finding: The majority of patients receive at least one incorrect diagnosis, and visit numerous doctors, before they receive a final diagnosis.

A worrying number of respondents expressed the difficulties they had in being believed about their condition/symptoms when there was no simple explanation apparent. Many respondents reported being accused of being a ‘hypochondriac’ or a ‘neurotic parent’.

“The whole process of diagnosis was one which I never wish to repeat. I was labelled a ‘neurotic’ parent by a paediatrician and not taken seriously from my first concerns. I saw numerous health and child professionals and had it not been for my then new partner, I would have given up.” – Parent of a child with Noonan syndrome.

“I was told that it was psychological, which meant that the diagnosis was further delayed as then I had been ‘labelled’ and was taken even less seriously.” – Patient, polymyositis.

Working to increase awareness and education of rare diseases in line with the UK Strategy for Rare Diseases will, it is hoped, ensure patients’ symptoms are not written off. Increased awareness for primary care and specialist professionals will ensure that when there is no obvious explanation, lesser known, rare conditions will be considered, and patients referred to the appropriate place, instead of patients being doubted and told ‘it’s all in your head’.

Key Finding: Patients can experience issues persuading medical professionals to believe their symptoms and describe how their condition is initially written off as ‘psychological’ or, parents are described as ‘neurotic’.

We did hear about some positive diagnosis experiences from patients, however, many of these patients said that they felt ‘lucky’ or explained that they had to pay privately to get their diagnosis. These instances of good practice show that early diagnosis is possible and that it can result in both improved patient outcomes and better use of NHS resources.

“I was extremely lucky to see a GP who had just been on a neuro placement. She recognised the signs/symptoms of myasthenia gravis immediately.” – Patient, myasthenia gravis.

“It was a long process. I was very ill and doctors didn't seem to take me seriously as they didn't recognise what was wrong. I was eventually told it must be psychological. I went to a private rheumatologist who realised what I had and immediately referred me back to a rheumatologist in the NHS with his findings.” – Patient, relapsing polychondritis.

“My GP told me there was no medical reason for worsening migraines and that sometimes they just happen; suggested opticians as a last resort - optician spotted papilloedema and sent me to A&E – idiopathic intracranial hypertension was diagnosed the next day!” Patient, idiopathic intracranial hypertension.

The advances in medical technology around genetics and genomics in recent years should lead to improved patient diagnosis. We need to ensure that these advances are taken up by the NHS.

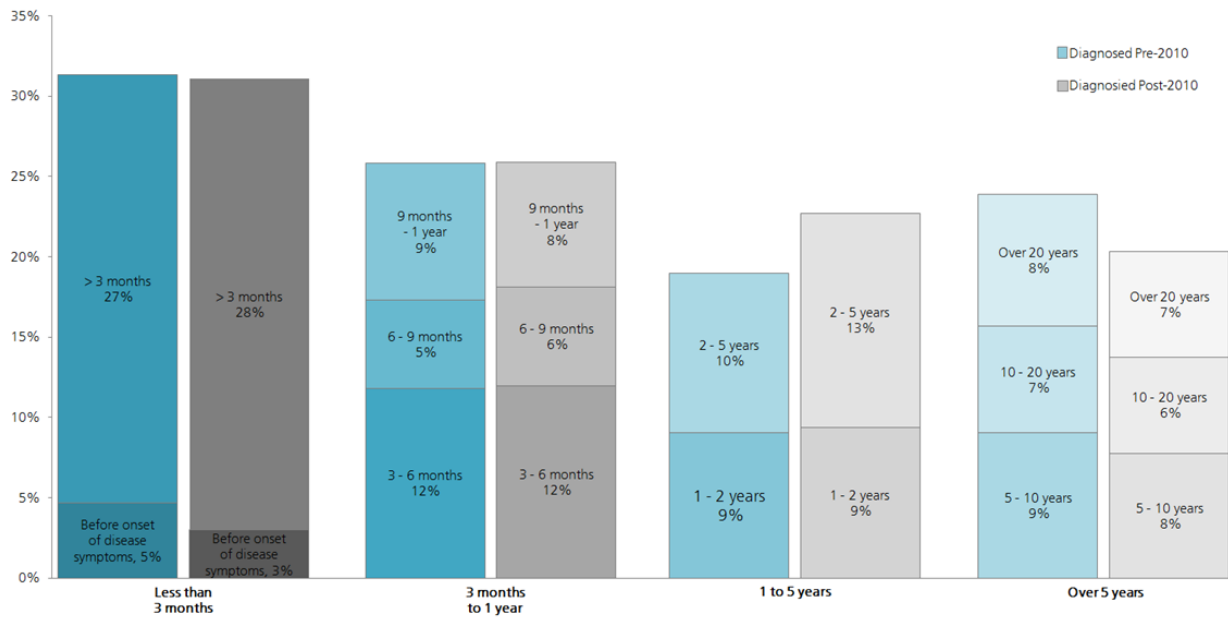


Chart 2: Length of time spent waiting for diagnosis. Percentage wait for respondents diagnosed pre-2010 and post-2010.

What has changed in the last five years?

When comparing patients diagnosed after 2010 to those diagnosed before 2010, there is limited evidence of improvement. 1 in 3 patients still have to wait over 2 years to achieve a final diagnosis.

Half of patients (50%) diagnosed before 2010 had at least one misdiagnosis and the same is true for patients diagnosed after 2010.

Of patients diagnosed before 2010, 67% of patients felt they did not receive enough information upon diagnosis; this was 63% for patients diagnosed after 2010.

There are a number of positive commitments in the UK Strategy for Rare Diseases which aim to improve diagnosis (including high quality training, computerised prompts, care pathways and genetic testing). As these commitments are implemented we would expect the wait for diagnosis, the number of misdiagnoses and the information given to patients to improve.

Stacey, mother of Imogen, 4 – experience of diagnosis

Imogen has Myhre syndrome, a genetic condition that affects many functions of the body. The gene that is mutated, SMAD4, is part of an important cell-signalling pathway, which allows cells in the body to communicate with each other. When the communication (signalling) is abnormal, as is the case with the faulty SMAD4 gene in Myhre syndrome, it affects the development of many body systems – which explains why Imogen has many varied symptoms and features.



At my 20-week scan, I was told by the doctor at our local hospital that Imogen had a twisted spine, and I was referred to a more specialist hospital. I had scans at 24 and 26 weeks and at both of these I was told that her spine was normal. It wasn't an easy birth: I ended up having a c-section, and they didn't check Imogen's spine. It wasn't until she got pneumonia at eighteen months and had her lungs x-rayed that they noticed she had scoliosis of the spine. This, combined with her not putting on weight, having heart problems and being jaundiced at birth, meant the doctors suspected she had Alagille syndrome, a type of liver disease. This, however, turned out not to be the case.

Imogen was diagnosed on 14 February 2014, when she was two and a half. The week before we had been contacted by our geneticist's secretary who invited us to the hospital for a meeting. We assumed this was a general catch-up appointment. However, when I walked into the consultation room, I knew it was something more because Imogen's geneticist was accompanied by a second geneticist, as well as a paediatrician and a counsellor. The team explained that when the test for Alagille syndrome came back negative, they had done more research into Imogen's features and had tested for another syndrome. This syndrome was called Myhre syndrome and the test came back positive. When I found out that we finally had a diagnosis for Imogen, I broke down in tears of relief. The geneticist explained the syndrome, stating that it was extremely rare and that Imogen was the youngest person ever to be diagnosed with it, the only child in the UK with it and number 33 in the world.

As the syndrome is so rare, the geneticists were very vague about Myhre and they themselves had to research the syndrome before our discussion. They could only tell us about the literature they had read online. The geneticist handed me some documents she had printed from the internet and offered us counselling. Later, when we Googled Myhre, the pages that came up were the same as they'd given us.

Myhre syndrome affects the development of many body systems and explains why Imogen has many varied symptoms and features. Imogen has many medical professionals involved in her care and each area of her condition is managed separately by the relevant specialist or consultant: physiotherapist, radiologist, otorhinolaryngologist and orthopaediatrician. Imogen wears a DMO (Dynamic Movement orthosis) suit for her scoliosis, leg splints, hearing aids, glasses (when she will tolerate them) and has regular medication. We're left to bring it all together and often find ourselves having to explain Myhre because no one knows what it is. It can be really hard. Myhre syndrome affects Imogen's heart and we have to be really careful. When Imogen isn't well and we take her to the doctors it's almost as though because they don't understand the condition, and because they've not heard of it, they assume we just want an excuse to take her into hospital. The reality is that we just want to check and make sure that her heart is OK. We know it can change in a matter of seconds: waiting 24 hours could be life-threatening.



Despite all this, Imogen is an extremely happy little girl and is full of energy. She enjoys going out with her friends and has a good sense of humour. She is outgoing, determined and truly independent. She makes us smile, and we are proud of her every second of every day.

Lucy*, 24 – experience of diagnosis

Lucy has hypopituitarism (an underactive pituitary gland) and postural orthostatic tachycardia syndrome (PoTS). PoTS is a form of dysautonomia where the body doesn't regulate heart rate, temperature or blood pressure correctly. Even standing up can be a challenge for patients with PoTS as their body is unable to adjust to changes in blood pressure.

My condition appeared out of the blue around two years ago. I was feeling generally unwell and had fainted a number of times so my GP referred me to a series of specialists. Within four months an endocrinologist had diagnosed me with hypopituitarism (an underactive pituitary gland) but I kept collapsing and I sensed something else wasn't right. My concerns were initially dismissed: I was told I was dehydrated, that 'women faint' or that I had a virus.

Nine months after my hypopituitarism diagnosis a specialist suspected that I also had PoTS. There was a lot of trial and error before I was diagnosed. When I went for autonomic tests I was feeling really unwell. The clinician told me I clearly had PoTS and that I did not need a particular test. Yet when I went to see my consultant again she said the diagnosis couldn't be confirmed until I had that test. My diagnosis was confirmed about four months ago after all the tests were repeated.

My main source of information has been Facebook support groups. When my endocrinologist diagnosed me she didn't tell me much – she didn't know much herself. In the groups I talk online to other people who have PoTS and get advice on what works for them, including medications, professionals and clinics. For example, I've been prescribed a number of different drugs and have been able to ask other patients how the drugs made them feel, and if my experiences are unusual or a cause for concern.

The specialists have tried me on three or four different types of medication, alongside general advice to increase my fluid and salt intake. The first couple of drugs, which my endocrinologist initially wanted me to try, were licensed, but the most recent drug I'm taking is off-label. My endocrinologist explained that the drug isn't predominantly used to treat this condition so it's not licensed for PoTS. However I was keen to try it because I'd heard it had significantly helped other patients with their PoTS symptoms. My endocrinologist appealed to the local health board to get it for me. It appears to be helping me so far – keeping my blood pressure up which in turn helps prevent me from collapsing.

I haven't seen any medical professionals who really know about PoTS yet. When I'm ill I have to explain the condition and request fluid therapy (one of the most effective treatments). It makes me feel uncomfortable to tell doctors what PoTS is and how to treat it when they're the ones that are supposed to know what to do!

My doctor has also referred me to a specialist clinic in London – the Autonomic and Neurovascular Medicines Unit. I am hopeful that the specialist there will know more about the condition and if there is anything else that can be done, or different treatments I can try.

I don't know why I have these two conditions, or what's caused it. It could be linked to genetics, an underlying condition or a virus. None of the doctors I have seen have tried to explain this to me, but I'd like to find out.

**name has been changed*

Undiagnosed

Patients and families alike have told us that a diagnosis is often the key to unlocking access to effective medical care and treatment. Some patients and families might live for a long time with an undiagnosed condition. This might be because they have not been referred to a clinician with the appropriate expertise. Alternatively they may have an unusual presentation of a condition which makes diagnosis more challenging than usual. Finally, they might have a condition that is so rare, clinicians have not studied it yet; we call these conditions syndromes without a name. This section highlights those answers from respondents who stated that their condition is undiagnosed.

Obtaining a diagnosis can be a long and difficult journey. When a condition is consistently difficult to identify it is most likely an extremely rare condition. Understandably, clinicians have difficulties identifying these rare conditions. Nearly half of the respondents who identified as 'undiagnosed' have been waiting over 5 years.

"I have found that as soon as other conditions are ruled out the wait to get a diagnosis is painfully slow. With anything that requires uncommon tests or treatment the funding is not there and so we are pushed to one side and made to wait a horrendously long time to get answers. In the meantime treatment and services are impossible to access as we have no clear diagnosis. So we are just left waiting for answers." – Patient with an undiagnosed condition.

"A diagnosis is vital in getting all the support that is required for quality of life." – Parent of a child with an undiagnosed condition.

"Just because 'it' does not have a name doesn't mean it doesn't exist." – Parent of a child with an undiagnosed condition.

There is a huge emotional impact to not having a diagnosis. Patients and parents do not feel in control of their situation and describe feeling isolated and uncertain about what the future holds. In fact 7 out of 8 undiagnosed respondents (87%) felt that they have not been provided with enough information and support throughout the diagnosis process.

Key Finding: Patients without a diagnosis do not feel they are given enough information and support on their journey to get a diagnosis.

1 in 4 undiagnosed respondents have seen more than 10 doctors in their search for a diagnosis. There is notable disparity when you consider that only 1 in 10 of patients who have a diagnosis reported seeing over 10 doctors. The increased number of doctors seen by undiagnosed patients could be positive if they are searching for the right specialist who can help them understand or even diagnose their condition, but negative if they are being pushed from pillar to post not receiving the care and treatment they need. It would be interesting to investigate this further in future work.

Barrier to care and treatment

Our research has highlighted that even with a diagnosis patients can still struggle to access appropriate care and support – this is even more apparent when it comes to undiagnosed conditions. 73% of respondents who did not have a diagnosis felt it had been a barrier to accessing treatment.

"Health professionals seem unsure how to help me as they seem reluctant to prescribe things when they are unsure of the underlying condition. I have a local physiotherapist but had to wait 6 months for this service. I have had to fund my wheelchair and mobility needs myself and research my own condition." – Patient with an undiagnosed condition.

"Can't access support services at all as my 5 children don't fit a correct box for their needs." – Parent of a child with an undiagnosed condition.

Patients with undiagnosed conditions often have significant and complex health needs. It is not always easy to identify and address these needs without a diagnosis. Little can be understood about the appropriateness of treatments and therapies, or how the condition is likely to progress. 4 in 5 respondents indicated that being undiagnosed had been a barrier to receiving appropriate coordinated care. Fragmented and poorly coordinated care not only has a huge impact on the

patient and their family but it can cost the NHS through the seemingly endless rounds of doctor's appointments, tests and even in inappropriate treatments.

"I've had no support at all." – Patient, undiagnosed condition.

"Without a diagnosis it seems the medical profession do not care....I gave up trying to find out what is wrong with me years ago and now live my life with a load of meds that to a degree help some of my symptoms." – Patient, undiagnosed condition.

Key Finding: Not having a diagnosis is a significant barrier to accessing coordinated care and appropriate treatment.

The NHS needs to take advantage of the clinical benefits arising from advances in genetics and genomics to help overcome the unique challenges faced by families affected by undiagnosed conditions. Through Genetic Alliance UK's support service SWAN UK (Syndromes Without A Name) we know that many patients and families never stop hoping for a diagnosis. A diagnosis can provide an explanation, bring new treatment opportunities, increase understanding of the condition – it could help them get an answer to that all important question - 'why?'

"Our reason for needing a diagnosis is for there to be proper investigative care into any associated conditions of a syndrome. This is often misinterpreted as needing a diagnosis to describe our child." – Parent of a child with an undiagnosed condition.

Key Finding: Many patients and families never stop hoping for a diagnosis.

Lisa, mother of Amelia, 7 – the impact of an undiagnosed condition

Lisa's daughter Amelia has a genetic condition that doctors have so far been unable to identify. This is most likely because it is an extremely rare condition and it is possible that it has never been studied enough to get a name: a syndrome without a name. Amelia's undiagnosed condition affects all her muscles, causing globalised weakness and fatigue. She has been given many individual names of the symptoms that affect various parts of her body but has no unifying diagnosis.



There was no indication during my pregnancy that there was anything wrong with Amelia. After she was born she had a lot of trouble maintaining her body temperature. I struggled to feed her and my gut was telling me – even at that early stage – that something wasn't quite right.

She was a difficult baby. She constantly looked uncomfortable and was struggling to gain weight. I raised my concerns with the health visitor and my GP but it wasn't until my nine-week check that we were pulled to one side and asked lots of questions by lots of people who then suggested conditions and carried out tests.

We were referred to Great Ormond Street Hospital, and the doctors there were fantastic. Amelia had lots of tests (some of which were horrific to see performed on a tiny baby), and saw many different doctors from various specialities, including respiratory and neuromuscular. This was great in the sense that we felt they were trying to get a complete picture, but the jump from laid-back local services to a rush of people everywhere was really scary.

We expected to be told exactly what was wrong with Amelia and find out the best way to treat her. But that didn't happen. She had more tests, but these came back inconclusive. We were left in a limbo, still facing the unknown. The doctors suggested she might have congenital myasthenia, but they didn't want to start treatment which could make the situation worse without knowing the gene affected. We received conflicting advice from our local physiotherapy team and the Great Ormond Street Hospital team about potential therapies – I felt a power play between the two teams. Conflicting information and a professional lack of understanding is extremely difficult for us as parents. As time has gone on Amelia has been referred to more and more specialists. Tests always throw up more questions than answers.

The doctors have now stopped looking for any of the congenital myasthenia genes; she's been tested for about six different genes. Some doctors say she doesn't have it but other members of the team say she does. A physiotherapist at Great Ormond Street Hospital saw her collapse and said he felt Amelia should be referred to the myasthenia team because of how fatigued her muscles were but the neuromuscular specialist dismissed this. It's difficult because effectively the neuromuscular team are her main 'case consults', the guiding team responsible for her care, and yet they're the ones who don't listen to us. It's made us feel isolated.



I've learnt that the best doctors are the ones who can simply admit that Amelia's medical picture is odd. The ones who accept that we have this bit of a pattern, we have that bit of a pattern, but we don't have the full picture and we need to keep an open mind. The best doctors are those who concede that we need to be grateful for and encourage the things that she's doing really well with, and that we need to support the things that she's struggling with.

One time when we walked into a hospital ward for some tests, Amelia looked at me and said, 'Mummy, when will they start hurting me?' It is hard to explain that we're not doing these things because we're being awful, but because we want to get some answers for her. It has crossed our minds to consider at what point we should stop and say enough is enough. But then in the back of our minds we think what if we're not giving her the right treatment? What if there's something better out there? What if there's more that we can do?

As Amelia grows up it's becoming harder to answer certain things. She's reached a point now where she wants to know why her muscles work one minute and don't the next, or why she can run around with her friends one day but not the next. We try and make it into a positive thing – I'll show her pictures of other children who have feeding tubes to make it normal for her. It's frustrating when I can't answer the questions that she has; I can't say to her, 'Well, you know, this has happened because of this,' or, 'We know that at some point this might happen.' We just can't do any of that.

I've been a member of SWAN UK (Syndromes Without A Name) for three years now. I was able to share our experience of Amelia and I just felt really involved and supported. Our children's issues are very different – there are children who are seriously medically complex, children who are affected emotionally or intellectually. Yet despite the differences, there's a unique bond that holds us all together.



Coordination of care

Rare diseases are often life-long and serious, affecting multiple systems of the body. Many of them are progressive, meaning that the health and quality of life for affected individuals will continue to deteriorate throughout their lives. We know that many patients have numerous professionals involved in their care, and as such it is essential that there is coordination and communication between them all. As noted in the UK Strategy for Rare Diseases patients visiting different departments at the same hospital on different days, or multiple hospitals, is not the best use of time or resources.

Our survey found that 1 in 3 respondents have to attend three or more different clinics, with a further 12% attending more than five different clinics for their condition. 23% of respondents indicated that they attend clinics at least monthly, 32% at least every 6-8 weeks, 55% at least quarterly, and 92% at least once a year. For the average rare disease patient this means attending no less than three clinics, at least, during every quarter.

"I see a neurosurgeon, neurologist, multiple sclerosis specialist, physiotherapist, endocrinologists, rheumatologist, occupational therapist...and I'm told different things which doesn't help." – Patient, cavernoma T9-T10, multiple sclerosis, osteoporosis, pituitary failure.

"I find myself explaining everything and passing on test results and procedures to every doctor I see because the communication is so unreliable and patchy." – Patient, Barakat (HDR) syndrome.

Many respondents highlighted that information was not shared between different hospital trusts, or, between specialist and local services. Information sharing is essential for patients to be treated effectively. The UK Strategy for Rare Diseases states that 'good communication between patients, their families and professionals is essential to ensure that the primary care plan is agreed and the care team has information and appropriate specialist support'.

"I'm currently seeing 8 different specialists in 3 different hospital trusts who are not set up for sharing information. It's a part-time job for me just tracking and organising this and making sure that I pass information across. Often it would make sense for a set of appointments or tests to happen in a particular order, but bringing this about is virtually impossible so we end up with delays and repeat visits." – Patient, Addison's disease.

"The coordination of care, under 5 hospitals and 19 clinics, is done by us the parents as there is no one else to do it. We also have to inform each clinic on any findings as they do not copy each other into any reports and we have to do all the updates for them." – Parent of a child with 22q deletion syndrome and cerebral palsy.

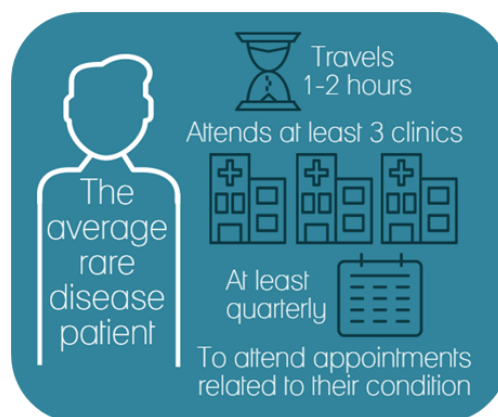
Key Finding: Information is not shared effectively between hospital trusts, or between specialist and local services.

Not only do patients have to frequently visit multiple clinics, nearly half of respondents reported that they travelled over an hour to get to their furthest clinic, and 11% had to travel for more than 3 hours.

"Care is not joined up; my GP has little to no understanding of my illness, so anything in relation to it is immediately referred back to my neurosurgeon in London. It's much further to travel to, especially when I'm feeling really sick." – Patient, idiopathic intracranial hypertension.

"My care has been very fragmented. The GP, neurologist, psychologist, and physiotherapist have all told me different things about my condition and refer me to different people." – Patient, idiopathic intracranial hypertension and fibromyalgia.

Key Finding: Patients frequently have to attend multiple clinics and travel significant distances to them.



81% of patients do not have a care coordinator or advisor, with a further 8% unsure of whether or not they do.

“We have one wonderful consultant who has given us his email address. Any concerns we get in touch and he replies within half an hour...that support is invaluable to us.” – Parent of a child with a chromosome deletion.

“When one person takes control it really works. When I was first diagnosed my consultant arranged all the different specialties I needed to see. As soon as he was gone my care just fell apart and I was lost in the system.” – Patient, relapsing polychondritis.

“I make a point of linking up my doctors who are involved in my care. I currently see a consultant in transplant clinic and a psychiatrist and they are both good at asking about each other’s care and treatment plan so they can treat me holistically.” – Patient, autosomal recessive polycystic kidney disease (ARPKD).

“Without the regular support of a specialist to coordinate my support and treatment, I am in a position where I have to act as a care coordinator to 15 specialist services!” – Patient, Ehlers-Danlos syndrome (EDS).

“We have a very good paediatrician who coordinates care – she listens and takes advice from us, and so far we have had no issues accessing things we need.” – Parent of a child with macrocephaly-capillary malformation (M-CM).

Key Finding: Most patients do not receive coordinated care and feel they are left to coordinate their own care.

Of the 12% who have a care coordinator the role was most frequently fulfilled by a specialist nurse, a doctor or another specialist. When asked who would be the ideal care coordinator, respondents indicated doctor/specialist (29%), specialist nurse (19%), patient themselves (16%) and no preference (16%).

A number of respondents commented on their preferred skills for a care coordinator:

“I would like a specialist nurse to work in partnership with me in between visiting consultants.” – Patient, vasculitis.

“They need to be easily and regularly contactable. Then to actually follow through!” – Patient, Ehlers-Danlos syndrome.

“A well informed person in a coordinating role would be very reassuring.” – Relative/Carer, adrenomyeloneuropathy (AMN).

Several respondents also indicated that, whilst a care coordinator would be helpful, they would still want to be involved in their own care:

“Although it would be ideal to have a single person (or preferably team) coordinating care for continuity I would still like to be involved myself to some degree so that I can feel I have some control over my disease and treatment.” – Patient, scleroderma.

“I would like the specialist and patient to work together.” – Patient, limited cutaneous systemic sclerosis.

Being the parent of a child with a rare disease can be difficult because they are often shouldered with the dual responsibility of being a parent and juggling the child’s care.

“I would LOVE someone to be our coordinator. Having to do everything myself is so stressful on top of everything else. To be able to ‘just’ be a mum rather than a case manager would be so welcome!” – Parent of a child with Chiari malformation and idiopathic intracranial hypertension.

The UK Strategy for Rare Diseases states that ‘responsibility for coordination will depend on the case and the circumstances’, responses to our survey indicate that this is a sensible approach to take as long as the overall aim of having an individual responsible for care coordination is achieved.

Transition

The UK Strategy for Rare Diseases committed to the development of seamless pathways for transition. It suggests that arranging coordinated transition from children's to adults' services should be a minimum standard for specialist centres.

Of respondents that had transitioned from the paediatric team to adult services 16% experienced problems in transition to adult services (this is less than what was reported in our 2010 report).

“Coordinated care diminishes when adulthood is reached.” – Parent of a child with Noonan syndrome.

“My son is 18 and in transition. He needs support in terms of becoming more independent and coordinating his own care going forward, but he missed out on a transition service now in place. We are waiting for a social care assessment to be undertaken in order to see how he might benefit from intervention to support independent travel and living in the future, as at the moment he is solely dependent on his parents to care for his needs.” – Parent of a child with ring chromosome 20 syndrome.

A few respondents highlighted good practice, among these, the importance of patient support groups, charities and social media for families going through transition was emphasised.

“She transitioned at 17, they suggested she should go up early rather than start surgeries and things under paediatrics and have to change to adult care half-way through.” – Parent of a child with idiopathic intracranial hypertension.

“I set up a parents support group on Facebook for parents like me, going through transition. It has really helped to talk to other families going through the same thing.” – Parent of a child with partial octasomy of chromosome 15q.

Our 2014 Patient Experiences of Transition Between Care Providers report found that transition is often better for those affected by more common conditions. The report also outlined the problems faced by rare disease patients going through the transition process and made recommendations to facilitate successful transition. The recommendations include: good coordination and communication, age-appropriate services and consideration of patients' individual circumstances.

Specialist centres

Many patients with a rare condition need input and expertise from numerous specialists. Coordinated, multi-disciplinary care is extremely important to patient well-being. Specialist centres can be an effective and cost efficient way of ensuring patient care is handled effectively and efficiently. Clinical centres bring together multi-disciplinary teams of health and social care professionals.

40% of respondents don't know if there is a specialist centre for their condition. Of the 30% of respondents who were aware of a specialist centre for their condition, 66% access it.

Key Finding: Many patients do not know if there is a specialist centre for their condition.

The UK Strategy for Rare Diseases outlines what specialist centres should do as a minimum standard. This includes having a sufficient caseload to build expertise, more than one clinical 'expert' who knows about the condition, coordinating care and transition, involving patients themselves, supporting research and ensuring their expertise is available to families and their healthcare teams (sharing expert knowledge with local professionals).

We asked patients and their families what their specialist centre does, and what they would like them to do. The majority of respondents (over 80%) indicated that they would like a specialist centre for their condition to have a number of patients with the same diagnosis and more than one clinical expert with knowledge of the condition. Of those respondents who attended specialist centres, over 80% indicated that their centre fulfilled these criteria. In addition 60% of patients thought coordination of care was an important role for specialist centres but only 10% of patients reported that their centre did it.

“The John Radcliffe Hospital team is fantastic. They keep you informed and provide expert care. It’s like a family.” – Patient, common variable immune deficiency (CVID).

“The Walton Centre is everything you could wish for; they are world leaders in this very debilitating disease.” – Patient, neuromyelitis optica (Devic’s disease).

“It’s such a huge relief to attend a hospital appointment and not have to be in ‘self defence mode’, to not have to fight to be believed or be afraid of humiliation. I can be honest about my symptoms without fear of the reaction.” – Patient, Ehlers-Danlos syndrome.

“Since being referred to the specialist centre I have access to a specialist nurse and consultants who are specialised in my condition.” – Patient, paroxysmal nocturnal haemoglobinuria.

Many patients have to travel a long way to access a specialist centre, and whilst some are more than happy to do this, others felt frustrated or found it difficult.

“Would rather travel and pay out a few hundred pounds for the visit for the security of being seen by the right person.” – Patient, Cowden syndrome.

“The location of specialist centres may be an issue for some but the thought of having a team of experts in one place (where my son could see more than one specialist on the same day) is appealing.” – Parent of a child with macrocephaly capillary malformation.

“Travelling to a specialist centre is going to be so difficult due to the nature of my condition.” – Patient, Behçet’s syndrome.

The importance of specialist centres working together with local care teams cannot be understated. In most cases the majority of care is provided locally by GPs and in local hospitals; sharing expertise will help to improve care and have a positive impact on patient health. Coordination with local healthcare teams also enables a balance to be achieved with the need to travel for specialist care.

“Working closely with local providers will ensure patients and carers only have to travel a minimal amount.” – Former carer, progressive supranuclear palsy.

80% of patients thought sharing expert knowledge with local professionals is an important role for specialist centres but only 30% of patients reported that their centre did it. The UK Strategy for Rare Diseases states that ‘centres must have protocols in place to share their expertise with local services’ whilst we know some specialist centres do this successfully some respondents highlighted problems with communication breaking down.

“The problem stems from the GP not listening to the specialist centre.” – Patient, Sheehan’s syndrome.

“Specialists should liaise with local hospitals when the patient is to be treated locally, for any condition, not just the rare disease.” – Patient, congenital myasthenic syndrome.

“At the moment there is a lack of communication between the specialist centre and local professionals. This is due to a lack of admin help, I think, rather than a lack of will on the part of the doctors. Ideally notes should be emailed back to the patient’s own clinicians within a week of a consultation so that everyone is aware of what is going on. When the specialist centre reduced my medication they didn’t inform my GP and so I had to explain why.” – Patient, spontaneous coronary artery dissection.

Those who did not know of a specialist centre for their condition, or do not access it, expressed how useful it would be:

“We would love it to all be in one place and one appointment to take time off work and school for.” – Parent of a child with Prader-Willi syndrome.

“It would be fantastic and a vision to have a specialist centre for treating r(20) patients. At present, due to the rarity of the condition patients feel isolated and their medical teams are often in the dark as to treatment options and understanding the condition, as it is often the only patient they have with r(20). There appears to be no collaboration.” – Parent of a child with ring chromosome 20 syndrome.

“It would be ideal to have a specialist centre that knows about rare diseases. Knowledge is too scarce about rare diseases.” – Patient, mast cell activation disorder.

“Patients should travel to specialist centres where possible but these centres should provide outreach services where necessary. They should also provide a summary of the condition to patients and carers along with any advice.” – Patient, pheochromocytoma and retroperitoneal fibrosis.

What has changed in the last five years?

When asked if the coordination of their care had improved in the last five years, respondents presented a mixed picture with 25% feeling their care is better, and 35% feeling it is less coordinated now than in 2010. This variation could be explained by the differences in care provided to patients with different rare diseases, in different regions and under different clinicians (who have varying knowledge and experience) as well as the increased number of respondents to the survey.

“A nurse specialist was employed in 2013 and since then care has improved.” – Patient, idiopathic intracranial hypertension.

“My old endocrinologist used to coordinate things even though he didn't have too, he was a lovely guy who cared about his patients, after he retired the new ones often talked about coordination but didn't actually do it.” – Patient, Ehlers-Danlos syndrome, postural orthostatic tachycardia syndrome, Addison's Disease and osteoporosis.

“I have always had to coordinate all of it, including correcting admin errors (like having my phone number recorded incorrectly) and information in my medical record (which is largely inaccurate). I have a good GP (at long last) but she is restricted in what she can do for me.” – Patient, mal de débarquement syndrome, dysautonomia, PoTS and convergence insufficiency.

“[My care is] still completely uncoordinated.” – Patient, Ehlers-Danlos syndrome type 3.

There was also variation in respondents reporting change in the number of clinics attended. 66% of patients feel like they are attending the same amount, or more clinics now than they were in 2010. Encouragingly, 30% of respondents reported that specialist centres that they attend were established in the last 5 years. We are hopeful that as the Strategy is implemented the number of patients who receive their care in a specialist centre will increase and that coordination of clinic visits will be improved.

“Since all my care has been centred at one hospital it's easier.” – Patient, idiopathic intracranial hypertension.

“I think my specialist centre is excellent and feel confident that if I had extra needs I could go to them for help.” – Patient, vasculitis.

Shelly, mother of Katie, 21 – experience of care coordination

Shelly's daughter Katie is affected by idiopathic intracranial hypertension (IIH), a neurological condition of unknown cause defined by increased intracranial pressure around the brain without the presence of tumour or disease. The space around the brain is filled with cerebrospinal fluid (CSF). If, due to a variety of factors, the pressure around the brain rises then the space containing the fluid cannot expand. It is this excessively high CSF pressure that produces the symptoms of IIH. The condition can cause blindness and some patients present symptoms similar to those of a brain tumour.



We were given no information at all when Katie was diagnosed at the age of thirteen. I asked the doctor, who said he didn't have any and that I should Google it when I got home. It was terrifying: there was simply nothing available then. Fear of the unknown is horrendous. I did find a forum for IIH patients and families, which has since become a registered charity, IIH UK, for which I am treasurer. Being able to speak to other people, other parents who were going through the same experience as us, was a tremendous help.

When Katie was diagnosed our local hospital liaised with the paediatric neurology department in Newcastle but as a family we felt very left out; we didn't have much opportunity to have a say in her treatment. When she transitioned into adult services at seventeen, and as I became more informed about IIH (more informed than some of the doctors), I made sure that Katie's voice was heard during appointments. Katie has autism, she is high-functioning, knows her own mind and now feels more in control, of both the medical side, and the psychological side of living with a rare condition.

Although Katie doesn't have a care coordinator as such, we now have direct access to a CSF specialist nurse. She's lovely and we have her direct mobile number. If Katie is having problems I can just give her a call. We discuss the situation and if she feels it's needed, she just says, 'Come up to the ward and we'll see what's going on.' The CSF specialist nurse is our main point of contact, along with our ophthalmologist, who, if Katie's having any visual problems, will ensure she's seen pretty quickly – sometimes even the next day.

Last time I rang up, Katie was having extensive abdominal pain – not head pain, abdominal pain. I rang the CSF nurse and she told us to bring Katie up to the ward at 11am. They took blood, and checked for infection, because Katie has a VP shunt that drains the CSF from her skull into her abdomen. The bloods were clear, so Katie was sent for an x-ray to check everything was connected and working as it should be, which it was. So they decided that the end of the tubing was probably a little stuck on the inside, preventing the CSF from flowing, and that once she was active and moving around it would become unstuck and work again. To be certain, they scheduled Katie for an ultrasound three weeks later and everything was essentially normal.

This coordination has made an amazing difference to the quality of Katie's care. Previously when she was having problems I didn't know who I could ring. I'd go along to the GP who would spend a week trying to contact somebody at the hospital. Then the hospital would have to get back to us. Everything was always dragged out. This doesn't happen now. We've got direct access, and we feel very privileged to have it.

Although Katie has to go to a number of different clinics and travel a considerable distance, the appointments are well scheduled and organised. When she has an appointment the next one is booked there and then, so we know well in advance when the appointment is going to be. She's also had a number of medications, and although there is no drug designed to specifically treat IIH, they address the symptoms. An off-label medication, acetazolamide, reduces pressure in her head and prevents Katie from going blind. She also has melatonin to help her sleep.

Everything's running smoothly now and it's how I wanted it to be. If I could change one thing it would be that the information and support which is available now would have been available to us in the early days when we found out she had IIH. There was nothing, no information, no care plans, no treatment plans or anything. Even now, doctors will just treat their patients however they think they should be treated. We're lucky Katie has better coordination and the treatment that's right for her but there's no care plan even today – and we're working to change that.

Richard, 60 – experience of a specialist centre

Behçet's disease (also known as Behçet's syndrome) is a rare chronic auto-inflammatory multisystem disorder of unknown cause, typically characterised by recurrent mouth ulcers, genital ulcers, eye inflammation, joint pain and skin lesions. It can cause blindness and can lead to life-threatening complications.

I was diagnosed with Behçet's disease in 1995. In a way I was lucky as I had quite a few symptoms simultaneously which meant clinicians were able to join up the dots. Still, I had to go backwards and forwards to my local hospital and GP. On one occasion a nurse, who was dressing the ulcers on my leg, mentioned my diagnosis – a diagnosis I hadn't yet been told about. She said I had Behçet's disease but couldn't tell me what it was. I rushed to the library and looked it up (the internet didn't exist back then). I thought my whole world had come to an end – Gray's Anatomy listed all the complications of the condition, stated that any organ could be affected, and that patients can go blind. I was devastated to think that I might lose my sight. I looked up the medication I was on and was horrified to find out it was mainly used for leprosy – did they think I had leprosy too?

Thankfully, I contacted the Behçet's Syndrome Society who were extremely knowledgeable and advised me to see a Behçet's specialist. Tests with the specialist confirmed my diagnosis, and through trial and error we found the right medication for me which cleared up the ulcers and alleviated the debilitating headaches I'd had for the previous three years.

I joined the UK Behçet's Syndrome Society. The founder of the charity was calling for the creation of a Behçet's specialist centre and after hearing about others' desperate situations, I knew he had the right idea. A group of us found out about some central specialist funding that we could apply for from AGNSS (Advisory Group for National Specialised Services – now replaced by NHS England specialised commissioning). We got the ball rolling by approaching Behçet's clinicians, and three of them supported a joint application.

The first year we applied, AGNSS deferred our application by a year in order to understand more about the medication budget. The director of the Behçet's Society carried out health economics to prove the financial case for the specialist centre. The most important point he made was that going blind from Behçet's creates a huge burden on the state in both health and social terms. A drugs pathway was produced, outlining the different levels (and expenses) of drugs patients could receive on a scale (the most expensive drugs would be given on clinical need). The application was supported by a membership survey which highlighted the impact Behçet's had on patients, their employment and their finances. After this second application we were awarded the money – £20 million over five years – and Centres of Excellence were set up in London, Birmingham and Liverpool.

The centres have been running since April 2012; I go to the fantastic Behçet's centre in London. Patients are able to see all the clinicians they need to see in one day. If you need medication you know you're not going to lose out to a 'postcode lottery' and when you're very unwell the doctors at the centres know what the situation is with Behçets and they have an in-depth knowledge of it now. You don't have to go through and explain everything, see a different doctor and explain everything again. They know exactly what it's like to have Behçet's so you have confidence in them. The centre provides coordinated care and their 'hub and spoke' approach allows them to share their expert knowledge with local professionals. The centre also has a psychologist who helps patients cope with the psychological aspects of the condition, and support workers to help deal with non-medical aspects. The database of patients creates research opportunities that just weren't possible previously, such as the search for a diagnostic test and even, in the long term, a cure. The centres are truly transforming the care of Behçet's patients.



The team at the London Behçet's Centre of Excellence

Access to treatment

The findings of this report illustrate the day-to-day challenges faced by those living with a rare disease. These challenges are made worse by the absence of any effective treatment. As evidenced in the chapter on research, patients, families and carers look towards research and innovation to develop therapies and treatments that may alter the course of their condition or improve the quality and length of their lives.

Access to licensed medication

A high percentage of respondents (45%) indicated that they know of a licensed medication for their/their family member's condition. Of those who know of a licensed treatment 87% indicated that they have access to it. This may not be an accurate portrayal of the current landscape. For instance, we are aware that there are only around 100 medicines for rare diseases with a marketing authorisation in Europe. Of these 100 medicines, EURORDIS, the European organisation for rare diseases, states that 'A third of patients do not have access to the orphan medicine that they need and another third only have access after waiting years'^[3].

Potential reasons why the percentage of respondents indicating they know of a licensed treatment for their condition was higher than expected include a lack of knowledge around what licensing means, whether the treatment is licensed or not, and it could be reflective of the lack of information given to patients about their condition and treatment options. Additionally many patients will be given treatments to alleviate their symptoms e.g. antiepileptic drugs for epilepsy, which are not licensed for a specific rare disease. Many respondents did in fact explain this in their response:

"There is no medicine for this condition. Only drugs to try alleviate various symptoms." – Relative/carer, hereditary hemorrhagic telangiectasia.

Key Finding: For many patients the day-to-day challenges of managing their condition are made worse by the absence of an effective treatment.

There are eight formal routes through which licensed medicines for rare conditions can be appraised and/or commissioned in order for them to be made available to patients through the NHS in England^[4]. The number of licensed medications available is unknown but it is extremely unlikely that 45% of patients have access.

"Until very recently there were no treatments. This week a drug was licensed in the EU for treatment of newly diagnosed patients, even this is only effective in about 1 in 4 patients. Research is proceeding on gene therapy." – Patient, Leber's hereditary optic neuropathy.

Many patients highlighted their experience of accessing licensed medications:

"One was routinely prescribed. Another medication I receive is licensed but not routinely prescribed and special permission was required. This took some time until the consultant applied for it but once he did it was approved very quickly." – Patient, acromegaly.

"My GP refuses to prescribe due to cost, so medication is provided by the hospital." – Parent of a child with neuromyelitis optica.

Access to unlicensed medication

Unlicensed or off-label medicines are an important route of access to treatment for rare disease patients. 1 in 5 respondents felt they had been informed of off-label/unlicensed medications. Of these, 55% had accessed the medication. Just under 40% indicated it was routinely prescribed, but over half of respondents did not know how easy it was to access.

"Had to obtain exceptional funding for some medication." – Patient, relapsing polychondritis.

"The medication was too expensive so [my doctor] will not fund it as there is a cheaper alternative available." – Patient, panhypopituitarism.

"The consultant is unwilling to even consider it." – Patient, Addison's disease.

“I had to wait a year for a specialist appointment, then 10 months for the follow up, then argue with my GP about following the consultant’s instructions to prescribe off-licence medication.” – Patient, Ehlers-Danlos syndrome.

“Doctors surgery refused to repeat prescribe. Have to go to Birmingham Children’s Hospital each time.” – Relative/ carer, congenital myasthenia.

Key Finding: In the absence of effective licensed medicines, unlicensed or off label medicines are an important route of access to treatment for rare disease patients.

What has changed in the last five years?

Little appears to have changed in the last 5 years to improve access to treatment for rare disease patients. Reflecting on the results in our 2010 report, a similar proportion of patients are struggling to access medication in 2015. Additionally the 2010 report highlighted the lack of information given to patients about their treatment options and this confusion still appears to be an issue.

Patient organisations and support groups play a vital role in helping to inform patients about what treatments could help them through both information provision and peer-support.

68% of respondents did not know if access to licensed medicines had become easier. 1 in 8 (13%) felt it had, and 1 in 5 (19%) felt it had not. Over half felt that they did not know if it had become easier to access off-label/unlicensed medication. 25% felt that it had not and only 11% felt it had improved.

“Some treatments have become almost standard despite being off label.” – Patient, Addison’s disease and Ehlers-Danlos syndrome.

“More off label medications are available but not many specialist consultants who are knowledgeable about them or willing to prescribe.” – Patient, idiopathic intracranial hypertension.

The huge variety of responses to improvements in the last five years illustrates the disparity and inequalities in the system. A patient’s access to a medicine appears to be affected by a huge variety of factors both national (such as whether it is licensed and whether a health technology appraisal body has approved it) and local (if it is known to the patients doctor, if the doctor considers the medicine beneficial to the patient from the evidence available and if it’s cost-effective enough for their local budget).

“There is no appropriate specialist consultant for my condition in Scotland but it seems impossible to obtain a referral ‘out of country’. It is extremely frustrating to see people elsewhere in the UK getting excellent, and fairly inexpensive, treatment which allows them to have work and have a good quality of life.” – Patient, hypoparathyroidism.

There are clear inequalities in access to medicine across the UK. These inequalities have been highlighted in Genetic Alliance UK’s patient charters: ‘a multiplicity of approaches and pathways, [means] the risk of making inconsistent decisions that result in inequitable access to medicines for patients with rare conditions is high^[5] and ‘inequity of access to specialised therapies for patients living in Wales has led to a number of families being forced to take drastic action, relocating their family and moving across the border to gain access to the therapy^[6]. This was supported by respondents, 57% of whom indicated that they knew of other patients in different areas who could access a drug they could not, or had to appeal to access.

Key Finding: Where a treatment has been licensed for a rare condition, patients may struggle to access it due to the complexities of appraisal and commissioning processes.

Patients have a right to access medicines that are deemed to be clinically effective and cost-effective. The NHS Constitution supports this right. The constitution additionally states that the NHS should provide a comprehensive service for all and promote equality by paying ‘particular attention to groups or sections of society where improvements in health and life expectancy are not keeping pace with the rest of the population^[7].

For patients fortunate enough to have a treatment (be that licensed or unlicensed) for their rare condition, there are still a number of barriers that must be overcome for them to benefit and have access to the treatment. This includes ensuring that there are appropriate frameworks in place for licensing medicines for rare conditions, determining whether or not they are cost-effective enough to be made available on the NHS and ensuring that medications are consistently prescribed off-label to patients who would benefit.

Fiona, 42 – experience of diagnosis and treatment

Fiona has scleroderma, also known as systemic sclerosis, a multisystem autoimmune disease which causes widespread vascular damage and fibrosis. She also has the related conditions Raynaud's phenomenon, rheumatoid arthritis and underlying lupus.



I underwent hundreds of tests and saw countless different doctors before I was diagnosed. At four, when my mum first took me to see the doctor, he said that my hands turned blue in the cold due to bad blood circulation and that I'd grow out of it. At the age of eight I started getting very crusty lumps underneath my nails. I was diagnosed with scleroderma when I was nine. It's extremely rare for children to get scleroderma. My parents were told that I was one of just five in Europe, and the only one with the related conditions.

I have struggled to get information about my condition from when I was first diagnosed, and my parents weren't told much, right through to the present day. For example, my mum was not told that these conditions could make me lose my fingers. We attended a demonstration of heated gloves at the hospital and a woman there had the tops of two fingers missing. The demonstrator asked, 'When did you have your fingers amputated?' She replied, 'I didn't, they eroded through scleroderma.' I was ten years old, newly diagnosed, and devastated to hear this. My mum and I looked at each other and willed up. Why had we not been told this?

Most of the information I have about my illness I found on the internet, in medical books, or by asking doctors. I've probably learnt more from the internet than I have from my doctors. I feel that I have been let down because I've not had enough information about the impact of my condition. I know what I go through everyday but not necessarily the reasons why. You can get lots of information about other illnesses just by walking into hospital and picking up a leaflet. I've had to find out information about the condition, and even about treatments, for myself.

A few years ago my brother gave me a newspaper clipping about a scleroderma patient who had taken sildenafil (a form of Viagra). Sildenafil dilates blood vessels, which in theory allows more blood into your fingers and toes. The author of the article explained how successful it was for her: she didn't feel the cold as badly as before and could even go outside when it snowed. I was puzzled as to why I hadn't been offered this. So, I showed the article to my consultant who, to my surprise, already knew about the treatment but had never offered it to me. I asked him why he hadn't offered this treatment to me when he knew it had been successful for similar patients. He couldn't really answer my question but let me try the drug. Unfortunately it's not been as successful for me as for the woman in the article. However, my tissue viability nurse recently mentioned another drug that has been used to treat patients like me. I am hopeful that the consultant will know about this drug too, and I intend to raise it with him.

It can feel like I am simply left to suffer. I've waited eight weeks for urgent operations which are meant to be done within four weeks. This feeling of isolation has led me to want to share what I know with other patients. Until now I haven't really got involved with any patient groups, but over the last year I've been quite unwell, and have spent a significant amount of time at home on my own. I found a few relevant patient support groups on Facebook and have set up my own group specifically for patients with scleroderma. Previously I'd never met anyone with the condition I've got but now I have spoken to quite a few people. The group has nearly 40 members now, both patients and people who know someone with the condition. I don't feel so alone – I can talk online to one or two of them and know that they'll understand what I'm talking about.

Neil, 45 – experience of diagnosis and research

Kallmann syndrome, or congenital hypogonadotropic hypogonadism (CHH), is a congenital hormonal condition, which results in failure to go through puberty or failure to reach puberty fully. It causes infertility and is associated with reduced or complete loss of the sense of smell.



I was diagnosed at 23. Before that I was dismissed as a ‘late bloomer’ and told to wait, that puberty would happen eventually – even though I had a number of related symptoms. I’d been dismissed by a number of medical professionals and it wasn’t until I started my first job at the Royal Free Hospital in London that I found out about Kallmann syndrome (KS) from an expert endocrinologist who was also working there. I was standing in the corridor chatting to him one day, when he asked me a question no doctor had asked me before: ‘Can you smell?’ – and it all went from there.

I’ve been on and off testosterone treatment in different forms since I was 23. There isn’t a ‘full treatment’ or cure. Testosterone treatment aims to ensure that hormones circulate at the normal level. It induces most aspects of puberty but doesn’t treat the infertility. Endocrinologists who have never met or treated a patient with KS before might not know about the longer-lasting, higher-dose testosterone injection which is more effective than the short-lived treatment used for primary hypogonadism. There are not many KS specialists in the UK. It’s difficult to get the appropriate testosterone treatment or hear about alternative treatment options; it’s even more difficult to get fertility treatment.

I have had fertility treatment twice as part of a clinical trial. Because I was based at the Royal Free Hospital while I was actually working there and my doctor was a KS specialist, I was able to go on a clinical trial straight away. Fertility treatment is expensive and for men with KS it’s a fairly long and drawn-out process as it can take about two years to be effective. It’s slightly quicker for women. If you’re not on a trial, fertility treatment can be difficult to get hold of because it’s so expensive: GPs are reluctant to prescribe it due to budget restraints – you have to be in the right place at the right time.

I was on a different clinical trial last year for a brand new fertility treatment which was supposed to act more quickly than previous treatments; the whole trial was designed for men with KS. It normally takes about eighteen months but this new treatment is supposed to work within six to twelve months. It was a worldwide trial and there were only about four patients from each participating country. It worked quite well. As it’s a new developmental drug there will be another phased trial, but it sounds promising.

Most of the information about KS at the moment is on Facebook groups and websites – it’s generated by me and other patients – there’s very little information from the doctors. Most patients have never met anyone else with the condition and there are elements of the treatment that doctors don’t have time to explain properly (even if they know enough to be able to). The Facebook groups let you talk to other patients who’ve been through the same thing. We discuss treatments, diagnosis, coping, relationships – sometimes younger patients have problems with relationships because they look so immature physically that, emotionally they doubt it can happen. When they talk to the older patients who have been there and done that, who are married with children, it gives them more confidence.

Once you’ve got the treatment living with KS is fairly straightforward, especially if you don’t need fertility treatment. It’s primarily a case of maintaining hormone levels to ensure normal function is kept and the risk of osteoporosis and type 2 diabetes is reduced – I only have one appointment a year to check testosterone levels. Getting the diagnosis isn’t as easy, and even though the condition isn’t life-threatening – like some other rare diseases – quality of life is affected. I think it took me about ten years of treatment to ‘catch up’. I still don’t look quite my age, but when I was in my mid-twenties and looked around fifteen it wasn’t always easy. KS patients can get very depressed if they’re not diagnosed or can’t get on with treatment. There is a huge psychological impact of not growing up, and not experiencing adolescence and puberty at the same time as your peers.

Research

For many patients and families research is their only hope for a more effective treatment or cure for their condition. Many are keen to participate in clinical trials that may eventually offer a cure or improve diagnosis, if not for themselves then for future generations. Many simply hope to understand what is going on in their own bodies. These hopes are not unfounded. Research is fundamental to improving the understanding of rare conditions. It can result in a variety of benefits, including: increasing awareness of a disease, developing a treatment, improving diagnosis opportunities, uncovering information that will increase medical knowledge and even help improve the health of the general population.

“I would be happy to take part in any research if it means future generations do not suffer.” – Patient, Ehlers-Danlos syndrome.

“Research is an important part of understanding a condition and helping to find a cure. I am very pleased I was able to take part in a trial.” – Patient, idiopathic intracranial hypertension.

“I would like to be more involved in research where possible. More data can lead to better understanding.” – Patient, polymyositis with interstitial lung disease.

Patient involvement in research

It has long been acknowledged that there are a number of barriers to rare disease research and to involving patients in the research. Technological developments, however, are beginning to breakdown many of these barriers. For example, the internet and social media enable patients to link up, share information and facilitate recruitment.

Patients and patient groups are essential partners to knowledge sharing, the identification and prioritisation of research topics and of course, actively involving patients in clinical trials. They also play an important role in funding research^[7]. The UK Strategy for Rare Diseases acknowledges the importance of engaging and involving patients in research stating that their input ‘can improve both the quality and effectiveness of research’ and ‘their involvement should be encouraged at all stages of the research process’. However, involving patients in rare disease research can be challenging due to the small numbers of patients and their diverse nature. Patients and patient groups are often left to find out about research on their own – in fact, over half (53%) of respondents indicated that they felt uninformed about research into their condition.

“Medical staff (when I asked) were either dismissive or were not aware of current research - I knew more than them and this was very unhelpful.” – Patient, mal de débarquement syndrome.

“I would say that the only reason that I am well informed about research into my condition is because of my own profession, and because of the patient support group. I have never been informed of any research by the doctors whose care I have come under.” – Patient, cyclical vomiting syndrome.

Only 25% of respondents felt they were given enough information on research (this is less than was indicated in our 2010 survey). The internet and patient organisations were highlighted as the main routes to finding out about research. How can patients be involved in research if they are not given, or do not know where to find, information about it?

“To find details about research you have to know where to look. Promotion and interpretation of the research relies on a few individuals and organisations.” – Patient, hereditary spastic paraplegia.

“There still isn’t enough being done to let people know there is a research project going on. I would not have known about the research projects related to my conditions if I had not been intensely proactive looking for it.” – Patient, spontaneous coronary artery dissection.

“[Being involved in research] can feel like a postcode lottery, in that one has to be in the right place at the right time.” – Patient, osteogenesis imperfecta (type 3).

Despite the lack of information given to patients, it is encouraging that respondents to our survey are interested in research and clearly understand the potential benefits. 80% of respondents expressed interest in research participation, with 15% unsure and only 4% of respondents said they would not be interested in participating in research.

“If anything came along that she was suitable for, she’d jump at the chance to take part.” – Parent of a child with idiopathic intracranial hypertension.

“I am glad to be asked to participate in anything which can help both me and other patients with the condition. I am grateful research is going on!” – Patient, birdshot uveitis.

Key Finding: Most patients would participate in research if given the opportunity.

The UK Strategy for Rare Diseases acknowledges patient registries as an important method of involving patients in research. It can be an important first step in collecting a pool of data which can be used for research. However, only 18% of respondents were aware of a patient registry for their condition, and of these, 71% had joined the registry. Of those who were not aware of a registry 76% indicated that they would join (and 22% were unsure).

“As there doesn't seem to be a patient registry for EDS-3 most patients do not know about research that may be taking place.” – Patient, Ehlers-Danlos syndrome type 3 (hypermobility).

“It would be nice to be included in a database of patients and kept up to date with research. Especially as it's potentially a progressive condition.” – Parent of a child with tethered spinal cord syndrome.

Public Health England's National Congenital Anomaly and Rare Disease Registration Service (NCARDRS), which will collect data on all patients with a rare disease, will go some way to ensuring that the benefits of information collection are realised. It should help improve general understanding of rare diseases. NCARDRS states that it will 'support all research into congenital anomalies, rare diseases and precision medicine including basic science, cause, prevention, diagnostics, treatment and management'^[9]. The potential for this service to act as that important first step for rare diseases that do not have their own registry, or previous research, is exciting.

'Commitment to innovation and to the promotion, conduct and use of research to improve the current and future health and care of the population' forms part of the third principle of the NHS in aspiring to 'the highest standards of excellence and professionalism'^[10]. Yet only 29% of respondents had participated in some form of research. Providing samples to a biobank or participating in clinical trials were the most common forms of research respondents were involved in. Anecdotally we know that where there are limited treatment options, as is the case with rare conditions, patients are even more inclined to participate in research.

“I have not been approached by the NHS but I have had to approach them...The hope of new treatment is life-changing and I could have missed out on the chance to try this [medicine].” – Patient, erythropoietic protoporphyria.

“Medical staff seem very suspicious if asked about research and very dismissive... This isn't very supportive or promising in terms of change for future sufferers. Extremely disappointing!” – Patient, Gullian-Barré syndrome.

“Every patient who has a rare disorder should be given the opportunity, as part of their care plan, to take part in clinical trials.” – Parent of a child with tuberous sclerosis complex.

Not only can patients directly participate in research, such as clinical trials, they can also provide researchers with insights into life with a rare condition. These insights are fundamental to adding value to research and ensuring it is relevant to the needs of patients. Patients and family members can offer alternative views to researchers and health professionals, they have in-depth understanding as an 'expert' in themselves and their condition. Patients may have different priorities for outcomes and suggestions that researchers may not otherwise consider.

“Research methodologies need to ensure that patient defined outcomes and meaningful patient experience is part of clinical research.” – Patient, common variable immune deficiency.

“[I] look forward to having patients/carers involved in designing research.” – Former carer, progressive supranuclear palsy.

Sharing information about research

Patient organisations were highlighted as an important avenue for relaying information about research, and research outcomes, to patients. However, researchers should make an effort to engage with relevant patient groups to discuss recent results and relaying the information in non-specialised language. This is even more important when patients have been directly involved in research – as they want to know their involvement has had an impact. Sadly, many patients who had been involved in research expressed frustration at not being updated on progress or finding out the research results.

“We have been asked several times to take part in research. Photos, blood samples and family history have been taken every time and we are told we will be kept informed but no one ever does. When you try to find out yourself no-one seem to know anything!” – Parent of a child with Alagille syndrome.

“There is a distinct lack of inclusion by the research department leaving you wondering what’s going on. We know it’s not going to happen overnight but as I have children and grandchildren I would like updates on progress however small.” – Patient, scleroderma and related conditions.

Many respondents felt that the results of research should be made available to patients participating in research, free of charge and in an accessible, understandable way. Projects to develop scientific reports into non-specialised language, information booklets, workshops and so forth would be welcomed by the wider patient community (not just those directly involved in the research). Some patients called for open access to research relevant to their condition:

“A version [of research results] needs to be written so that people without a medical degree can read it.” – Patient, idiopathic intracranial hypertension.

“Patients and their carers do not routinely have access to full journal articles, so these should be made available to those who want them if they have a link with the condition (i.e. they or a family member has the condition in question).” – Parent of a child with neurofibromatosis type 1.

“Actively support researchers to share relevant information via the media/directly to charities.” – Relative, ataxia telangiectasia.

In the long-term, these efforts to communicate science would enable closer links to be established between researchers, patients, patient groups and policy makers. These links could help progress future research and shared knowledge will help empower patients in their own health and care.

Key Finding: Patients highly value research into their condition but do not feel like they are given enough information about it.

The frustration at the lack of available information about research was accompanied by a general feeling amongst respondents that there is not enough money or investment in rare disease research. This is a theme we previously identified in our Funding Support for Rare Disease Research: Raising Awareness and Increasing Transparency report which highlighted that there is a ‘perception amongst the rare disease community that public and major charitable funding bodies do not support research into rare diseases very well, in particular that there is not enough funding awarded’ and that ‘there is a low level of awareness amongst the rare disease community of the opportunities for research, and feedback was that funding opportunities are not promoted effectively’^[11]. We know that research efforts are being made through initiatives such as the 100,000 Genomes Project but it is important that there be more publicity, information and awareness of research that is already taking place.

What has changed in the last five years?

When asked if awareness and involvement in research had changed in the last five years the response was mixed. Responses indicate that whilst awareness has improved for nearly half (48%) of respondents, only 22% of respondents felt more involved in research.

A noteworthy number of respondents feel less involved. There are a number of reasons why this might be, including that the clinical trial or other research project they were participating in has ended.

Our survey identified an increase in the awareness of research. The key factors attributed to this increase include: the internet, social media and patient groups. Patient groups were praised for the important role they play in facilitating research engagement.

“I think the internet age we live in gives all patients and carers alike the opportunity to access far more information [about research].” – Patient, aggressive fibromatosis (desmoid tumour).

“[engagement in research has improved] through the work of charities spreading the word.” – Patient, idiopathic intracranial hypertension.

“Social media is being used as a tool for recruitment, and also through the internet there is opportunity for people to be proactive and find research in which they would be eligible to participate.” – Patient, Cowden’s syndrome.

“A lot of the research into rare conditions is being driven by patient groups.” – Patient, mal de débarquement syndrome.

A number of respondents highlighted that patient involvement in research, and recognition of patients and patient groups as equal partners in research, has been improving. After all, patient involvement in research includes not only direct participation (e.g. through clinical trials or surveys) but also involvement in the research process (such as in research prioritisation, research design and analysis).

“Patient voices are being heard a bit more.” – Patient, systemic mastocytosis.

“We [the patient organisation] were lucky to be contacted by a pharma company working in the field. We were involved from the beginning. They are hoping to manufacture a prophylactic treatment...” – Relative, neonatal alloimmune thrombocytopenia (NAIT).

For many patients and families living with rare conditions research offers them the potential for a brighter future and an improved prognosis. They hope that the continued interest in ‘their’ condition from the research community will lead to greater understanding, treatment options and even a cure.

Tony, 58 – experience of research

Tony has alkaptonuria (AKU), also known as black bone disease or black urine disease. AKU causes severe, early-onset osteoarthritis. Bones turn brittle and black, and the disease causes severe pain and osteoarthritis as life progresses. It is caused by the lack of an enzyme called homogentisate 1,2-dioxygenase.



I wasn't diagnosed until 2011. I could have been diagnosed as a child but it wasn't until I was in my fifties that I was told I had alkaptonuria (AKU). A rheumatologist saw my bluish ears and the brown spots in my eyes and asked me whether I had dark urine, then joined up the dots saying he was 99 per cent certain I had the condition. A sample of my urine was sent off for testing, which confirmed the diagnosis.

After my diagnosis a doctor I saw in Cardiff mentioned the AKU Society to me, so I got in contact with them. I found out that most patients from England and Scotland went to the Robert Gregory National Alkaptonuria Centre in Liverpool where they are given a small dose of the drug nitisinone, off-label, which appears to slow down progression of the condition. In Wales the Welsh Health Specialised Services Committee (WHSSC) wouldn't agree for AKU patients to go to the centre or have the drug because it wasn't licensed for AKU even though this wasn't an issue for English or Scottish patients.

Eventually, after appealing, we were allowed to go to the centre for a thorough assessment with specialist tests and the end result was that patients from Wales found out exactly what was wrong with them. That was quite scary because I realised that I had far more wrong with me than I initially thought. Usually patients at the centre have annual checkups, but in Wales we had to reapply for funding which could be tricky.

In the meantime we knew that there was a clinical trial into nitisinone for AKU coming up, called SONIA 2. It was highly unlikely that we'd get nitisinone off-label in Wales but at least with the clinical trial we could have a 50/50 chance of getting the drug. The other advantage of the trial is that they carry out most of the tests which are done at the specialist centre – it would guarantee that for the four years whilst we were on the trial we'd get the tests we needed done and we'd also have access to the specialist for advice.

I made a decision that I would take a chance; after all if the trial is successful, nitisinone could be licensed and it would be easier for all AKU patients to access it. Another patient from Wales also decided to go on the trial. By doing this, we felt like we were doing something for the condition as well. As it happened both of us were randomly assigned the control group – we didn't get the drug. Yet we still feel like we'll get something positive out of it. Of course you hope that you're the one who will be given the drug, but the control group is important too – if it wasn't there the trial would be irrelevant.

Being in the control group does mean that you stand a greater chance of deteriorating, and I am classed as being in the advanced stages. I know since I've been on the trial that my condition has deteriorated. I know from the way my joints feel and from the scans. In fact at my last check up the doctor wouldn't show me the latest scan because he didn't want me going away feeling depressed by looking at the deterioration from the previous year.

I met someone from Spain on the trial, who was on nitisinone and at 65 he'd had hip replacements, stents in his heart and before starting the trial could hardly walk or open his hands. His daughter said to me: 'My dad used to have a really wicked sense of humour and he lost it, but since he's been on the drug he's started to get his life back.' It's nice seeing people who are on the drug and how they are responding but it is worrying to think how much more I might deteriorate in the next two years.

Two years in, the trial findings are quite positive, and it looks like nitisinone will be approved, so hopefully when it's licensed it will be prescribed for us in Wales, especially as it's not an expensive drug. They keep saying, 'Two more years and you should get the drug,' but in the meantime at least us patients on the trial are doing our bit for AKU.

Since 2010: The last five years

Sadly very little appears to have changed for rare disease patients in the last five years. They still experience difficulties in diagnosis, accessing information about their condition, receiving appropriate coordinated care, accessing treatments and finding out about research.

The UK Strategy for Rare Diseases was only published in 2013 and set out 51 distinct commitments that are designed to improve health and social care for rare disease patients. Effective implementation of the Strategy will ensure people living with a rare disease have access to the best evidence-based care and treatment that health and social care services, working with charities, researchers and industry can provide. The UK Strategy for Rare Diseases states that its vision must become a reality by 2020, therefore we are optimistic that we will see an improvement in the patient experience of rare disease in the next five years but a strategy is only as good as its implementation.

Ongoing developments in genetic medicine accompanied by a host of policy initiatives including the Accelerated Access Review and the amendment of the Human Fertilisation and Embryology Act in early 2015 to allow mitochondrial donation, mean that progress in healthcare for those affected by rare conditions is being made. We look forward to seeing improvements in patient experience to match these policy promises. The issues faced by patients affected by rare conditions, outlined in this report, are becoming ever more relevant and pressing. They cannot be ignored. In fact, as this report highlights, the genomics revolution means there is an increased movement towards personalised, individualised medicines – some clinicians are predicting that soon all conditions will be 'rare'.

Some activity within certain teams in the four national health services and in research infrastructure has been occurring; and progress has been made in terms of internal policy and direction. Progress includes the commitments in the UK Strategy for Rare Diseases, and the respective implementation plans that followed in Scotland, Wales and Northern Ireland, as well as some progress in England to implement the commitments. This seems to be the result of Rare Disease UK's campaigning but, as this report indicates, has yet to have a huge impact on patients on the ground. Additionally, the progress does not appear to be uniform across the countries and regions, or between different rare conditions. We need to ensure that all patients have fair access to appropriate care and treatment for their rare disease – no one should be left behind because they are 'rare'. The UK Rare Disease Forum will be presenting a progress report to the health ministers of all four home countries in February 2016 and every two years thereafter.

The challenge now is to maintain progress where it has been made and encourage progress where it has been slower, to ensure the promise of the Strategy becomes reality in the years ahead.

Jo, 46 – how the patient experience has changed

Dermatomyositis is an autoimmune disease: the body's immune system attacks the muscles, skin, blood vessels and connective tissue creating inflammation, destroying muscles and causing progressive weakness. If dermatomyositis affects the heart, lungs, or ability to swallow it can be life-threatening. Myositis is a general term encompassing a group of rare diseases known as the inflammatory myopathies: dermatomyositis, juvenile dermatomyositis, polymyositis, and inclusion body myositis.



When I was first diagnosed, 21 years ago, all I knew was the name dermatomyositis, which I could not even pronounce. There were no leaflets available. Two years later I was given a rather archaic Arthritis Research leaflet, which had the contact details of a myositis charity on the back. So I rang Myositis UK and was able to talk to someone about the disease for the first time. I subsequently became a trustee for this charity and I now know that social media has greatly helped patients to find information and connect with each other. Even today though, some patients are still told by their doctors, 'Don't Google it, it's scary.' They're left very isolated and at times they're not getting the appropriate treatment.

The charity has worked over many years with a network of doctors and researchers in the UK and worldwide. The first International Myositis Conference for doctors and researchers was held in Stockholm in 2015. There's also what are regarded as specialist centres for myositis in various parts of England; I travel to the one in London from Norfolk. I relapsed three years ago with sudden breathing difficulties, but at that point nothing was showing up in my blood, which can happen from time to time. It was only because I had a doctor in place who really understands the disease that I was given the appropriate treatment – a dramatic contrast to my first hospital admission. I put part of this down to my own efforts – when my doctor retired I asked if I could be taken on by one of the doctors that I knew through the charity and my GP referred me. I was monitored by the specialist clinic for several years, and then I had a major relapse. If I'd been in the same position in a local hospital, getting weaker and with nothing showing up in my blood, I don't know what the outcome would have been – I don't think it would have been very favourable.

There's still a huge lack of understanding from many professionals who've never even heard of the disease let alone its implications. If you refer to dermatomyositis some guess that it's a skin disease, and that's all. I've even been in contact with doctors and professionals who have dismissed it as 'just a skin disease'. In fact, at my recent surgery I was in the operating room waiting to be anaesthetised, and I was surprised to hear the doctor saying, 'Oh, it's a skin disease,' and of course I had to correct him. I was about to be put under and even though I was very nervous about having the surgery, I gave him a quick run-down of what dermatomyositis is. He asked me if I was in the medical field. I told him, 'No, I'm a patient.'

I've had battles in the past with my local GP surgery over medications and monitoring bloods. Some GPs won't prescribe some of the medications, and I believe it's down to budget. I've had some medications not prescribed even though I've had them the previous year. I'm on various immunosuppressants, anti-inflammatories, painkillers, antimalarial medication, and every eight weeks I have intravenous immunoglobulin treatment and stay in London for five intensive days. It's instrumental in keeping me fairly well because other treatments in the past have failed to keep me stable.

When you have a rare disease, you sometimes become a 'pet patient' for a while because you're a novelty, and it's interesting for doctors. But I know from experience that this interest can wane, and then you can be left in limbo.

Awareness of rare diseases is the key for patients to be diagnosed. The charity promotes myositis to rheumatology, neurology and dermatology conferences, so that doctors and other professionals will learn to recognise it. Education about this disease is critical. Huge strides have been made but patients are still lucky to get diagnosed quickly and onto the right treatment path. There are still too many stories of people struggling to get diagnosed or not receiving appropriate treatment.

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Annex 1 - Methodology

A survey of 85 questions was conducted over the summer of 2015. The survey was open to anyone affected by a rare condition. It was shared extensively across our networks and by many other rare disease patient organisations. In total 1203 eligible responses were received and analysed (non-UK residents were excluded from analysis). Over 450 different rare conditions are represented in the survey. The survey results are supported by ten in-depth case studies of parents and patients affected by rare conditions. Further methodology:

- Unless stated otherwise results exclude respondents that selected 'don't know'.
- The estimated average was calculated for results linked to diagnosis and care coordination to create a snapshot of the patient experience.
- Three key methods were used to analyse the change perceived by patients in the last five years:
 1. Quantitative analysis of questions which asked patients whether or not they thought change (positive or negative) had occurred.
 2. Qualitative responses to the survey which were relevant to change (or lack of) were analysed thematically.
 3. Results were filtered by date of diagnosis and respondents who were diagnosed before 2010 were compared to those who were diagnosed after 2010. Respondents who did not provide the year they were diagnosed, or who have not yet been diagnosed, were excluded from this form of analysis.

