Experiences of Rare Diseases:
An Insight from Patients and Families

By Lauren Limb, Stephen Nutt and Alev Sen - December 2010

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

RDUK was established by Genetic Alliance UK, the national charity of over 130 patient organisations supporting all those affected by genetic conditions, in conjunction with other key stakeholders in November 2008 following the European Commission’s Communication on Rare Diseases: Europe’s Challenges.

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

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

- Research
- Prevention and diagnosis
- Treatment and care
- Information
- Commissioning and planning

into one cohesive strategy for all patients affected by rare disease in the UK. As well as securing better outcomes for patients, a strategy would enable the most effective use of NHS resources.
Chair’s Foreword

Advances in scientific research over the last two decades have highlighted the emerging possibility of addressing the needs of patients and families with rare genetic disorders in ways that would have been dismissed as science fiction only a few years ago. This gives rise to great optimism about the future. However, future possibilities must not blind us to the situation of patients and families struggling with the daily reality of life with a rare disease today.

This survey, reporting the experiences of nearly 600 families with rare diseases across the UK, is one of the largest that has ever been carried out. The picture it paints is a bleak one, with significant numbers reporting delays in diagnosis. Wrong diagnoses are common (sometimes happening two, three or more times). Access to specialist knowledge and expertise is too often fraught with difficulties and unnecessary institutional obstacles get in the way for patients and families seeking the help and support they need and should be able to expect.

The evidence reported in the pages that follow highlight the need to act now to bring services and support together in a strategic, coherent plan to utilise knowledge and resources effectively and in so doing, produce the best possible benefits for patients and families given current scientific understanding of good clinical practice, delivered in a timely, user-friendly way.

Alastair Kent
Chair of Rare Disease UK
Director of Genetic Alliance UK
Introduction

Over the summer of 2010, Rare Disease UK (RDUK) carried out a survey of patients and families affected by rare diseases. The aim of this 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 and problems they frequently face.

At RDUK, we are too often told of the difficulties patients with rare diseases experience in getting a diagnosis and accessing appropriate services, information and support throughout the progression of their condition. We therefore felt that it was important to conduct a survey to gain a better understanding of these issues and to better understand the scale of the problem.

The survey covered multiple aspects of rare diseases which our members have informed us they experience problems with including, diagnosis, coordination of care, awareness and participation in research, access to drugs and services, and access to thorough, reliable information and support.

The online survey was sent out to all of RDUK’s members, including patient organisations, many of which forwarded it to their own members. Paper copies were available for those who requested them.

We were delighted to receive a total of 597 responses to our survey. Of these, 27 responses had to be discounted as they came from patients or families living outside the UK.

Of the 570 valid responses, 47% were from the patient themselves, 49% were from a carer or family member of someone with a rare disease, and the remaining 4% were from other interested parties which included healthcare professionals responding on behalf of a patient, and carriers of a rare disease.

A total of 119 different rare conditions are represented in the survey. A full list of these conditions can be seen in Annex 1. These conditions ranged from ultra-rare to the more “common” rare diseases and included chromosomal, single gene, multifactorial and non-genetic conditions, as well as undiagnosed conditions, and patients who suffered from multiple different rare diseases. Despite the wide variation in the symptoms, prognoses and medical needs of these conditions, the survey indicated commonalities in experiences in regards to their care, information and support. The survey also highlights inequalities in the services received by patients with different rare diseases and even between those affected by the same rare disease in different parts of the country.

78% of respondents live in England, 12% in Scotland, 6% in Wales and 3% in Northern Ireland. The remaining 1% of responses were patients living in the Channel Islands. This is broadly representative of the population of the UK as a whole and as such demonstrates that we are able to suggest that our findings are a good representation of the general situation in the UK.

The results support the need for a national strategy for rare diseases in the UK and complements RDUK’s work in investigating what should form the basis of a strategy. We believe that a national strategy would reduce the fragmentation of services and ensure that patients of rare conditions are able to access equitable, high quality care, information and support in a timely manner, whilst at the same time making a more efficient use of NHS resources.

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

RDUK would like to thank everyone who took the time to respond to our survey, as well as the patients and families who are featured as case studies.
Summary of Key Findings

Research
1. Patients and families generally show an appreciation of the importance of research and a willingness to be involved in research.
2. Patients are not well informed of research into their condition. This can result in low patient participation in research projects.
3. Patient organisations act as a vital source of information on research into rare diseases.

Patient Care, Information and Support
1. Patients and families are not provided with enough information on all aspects of their condition, both at first diagnosis and subsequently.
2. Information to patients and families must be provided in a range of formats and at various levels of medical and scientific detail to ensure full understanding and informed decision making.
3. Patient organisations are often the main or only source of information for rare disease patients.
4. There is a lack of support for rare disease patients with their medical and non-medical issues.

Coordination of Care
1. The majority of patients’ care is poorly coordinated.
2. Patients have to attend multiple clinics for different aspects of their condition, often at a long distance from where they live.
3. The majority of patients do not have access to a specialist centre for their condition.
4. Patients frequently experience problems with medical, psychological, financial, social and other issues at transition periods.

Diagnosis
1. Patients and families affected by rare diseases wait far too long for a correct diagnosis.
2. A worrying number of people with rare diseases receive incorrect diagnoses before their final diagnosis is made.
3. Patients and families worry about the level of awareness of rare diseases among healthcare professionals.
4. Patient organisations play an important role in the diagnosis of rare diseases.
5. The experiences of patients and families of diagnosis can vary greatly raising concerns about equality of access and fair treatment in different parts of the country.

Access to Treatment
1. Trying to obtain medicines can be distressing for some patients and families.
2. There is no licensed treatment available for most patients with rare diseases.
3. Some patients are informed of off-label or unlicensed medicines but often patients and families have to inform their doctor.
4. Patients and families experience inconsistencies in access to medicines.
Research

Key Findings

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2. Patients are not well informed of research into their condition. This can result in low patient participation in research projects.
3. Patient organisations act as a vital source of information on research into rare diseases.

Research into rare diseases is vital to enable the development of new therapies, diagnostic tests and preventative measures for patients affected by these conditions. However, there are often obstacles that hinder research into rare diseases, including the reluctance of funders to support these projects, the lack of a network to aid research, and the low numbers of available patients for trials due to the rarity of these diseases.

Involvement in research need not be an onerous burden for patients; joining a registry, for example, generally requires little effort on behalf of the patient. But patients need to know of the existence of research projects so that they can participate and the tools can then be used to their full potential, which is especially crucial for some rare diseases with small patient numbers. These figures show that the large majority of patients support the use of registries and indicate that most would be willing to join one if it existed and they were made aware of it.

Our survey showed that only 25% of patients or families were aware of a registry for their condition. Of those that did know that one existed, 67% had joined the relevant registry. This figure may actually be as high as 85%, as a further 18% were unsure as to whether they had joined it or not.

Of those that did not know of a registry for their condition, only 2% said they would not welcome the creation of one.

These figures suggest that if made aware of ways in which to aid research, patients are generally very willing to participate.

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Awareness and Information

Key Finding 2: Patients are not well informed of research into their condition.

Only one third (35%) of respondents said that they are informed of clinical trials into their condition.

“We only find out because we are a member of a charity that funds research. If we were not a member of this we wouldn’t know about trials etc.”

Relative of a patient with cerebellar ataxia

“Clinical trials? What clinical trials? Diagnosis and treatment is just try and see!”

Mother of a child who died from haemophagocytic lymphohistiocytosis

“We found out about the clinical trials ourselves after the death via the internet, [the] trial was being run in [the] hospital [where our son was being treated] but they didn’t tell us about it.”

Relative of patient (deceased) who had haemophagocytic lymphohistiocytosis

1 A registry was defined in the RDUK patient survey as a database that collects clinical information from all patients with a particular condition (or type of condition).

2 Clinical trials were defined in the patient survey as a particular type of research that tests a new treatment or medical device on a large number of participants to determine its safety and effectiveness.
57% of respondents said that the patient had not participated in any research into the condition. Of those that had taken part in research, biobanking and participation in clinical trials were the most common ways in which they had been involved. Respondents also said they had filled out research surveys, undergone brain scans, been an aid for medical training, taken part in behaviour and observational studies and undergone genetic testing in aid of research into rare diseases.

We speculate that participation in clinical trials and other research is lower than desired due to the lack of information available. The results suggest that if patients were made more aware of relevant research projects being planned or carried out they may be more willing to participate in them.

“We would be very willing to participate in trials or research.”

Patient with Langerhan’s cell histiocytosis

“[I] would be willing to provide information to aid research.”

Patient with hypopituitarism

Less than a quarter of respondents feel they are given enough information on clinical trials (24%) and only one third feel they are given enough information on research in general (33%).

Research represents hope for many patients and families, if not for themselves, then the hope that the disease may be cured or prevented in future. Patients and families need to be kept up to date on research into their condition so that they are empowered with information on potential new therapies that they can then discuss with their specialists. They also need to be aware of research which may be relevant for them to participate in - if they don’t know it exists, they can’t take part.

**Key Finding 3: Patient organisations act as a vital source of information on research into rare diseases.**

Many respondents told us about the role of patient organisations in informing them of research into their conditions. Often we were told that this is the only way in which patients get any news on research, and that if it wasn’t for patient organisations, they wouldn’t hear anything at all about research being conducted. Many others responded that they are reliant on searching the internet for information on research. This highlights the need for more active communication about research projects.

“I only feel I get up to date information because of my involvement with a patient group, otherwise, I would not hear about any ongoing research.”

Patient, acute intermittent porphyria

“The only information about research and trials that I have gained is through the Myasthenia Gravis Association UK, which I only found out about by accident when my researching my condition myself on the internet.”

Patient, ocular myasthenia gravis

“We recently started a children’s branch of the MGA [Myasthenia Gravis Association] - we held a weekend in June and invited two of the research teams to update us on their research. It was absolutely fascinating/wonderful to hear about their work and how it affected our children.”

Parent of child with congenital myasthenic syndrome

“Patient support groups are an ideal vehicle for the publication of trials but it is extremely rare that information is received from healthcare professionals or pharma industry.”

Relative/carer of a person with Gorlin syndrome
Carl Tilson’s story

Carl Tilson who has Duchenne muscular dystrophy - Manchester, England

I have been living with Duchenne muscular dystrophy for 23 years having been diagnosed at 5 years old. To start with the local doctor suggested my mum was an over-anxious mother having seen me fall. My muscle biopsy proved he was wrong.

Having this condition sometimes feels like I have been sentenced to a life of misery only to die, however I enjoy my life through campaigning for my cause, which is sometimes as demanding mentally as it is physically. Sometimes I think it would be good to receive emotional support and advice on benefits, leisure activities and perhaps even dating.

I have found that the lack of a multi-disciplined team of specialists in each region is impacting on the lives of young children and young adults every day. I have experienced having to fight for everything from equipment to medicine. You are made to change from children services into adult services at 18 years old and immediately you find out you’re now left with no direction, and left to your own devices with no specialist support, for example, adult services don’t continue checking your heart and breathing function until you ask for it. Another experience, which does leave a bitter taste, was once I became quite poorly with a really bad chest infection and had to stay in hospital with a weak immune system and I was put into a ward with mostly old men with bad coughs and chest problems.

Since I first went to Newcastle where there is a multidisciplinary team, I have benefited quite hugely by receiving an annual check-up, learning about the things available and talking to Professor Kate Bushby who helped me understand about using steroids which quite frankly saved my life. I do feel though that updates on current trials are not widely available which leaves me and my family feeling isolated with frustration.

Also, the problem is that companies behind getting these treatments are mainly private with no government body overseeing their process, so things can be long-winded and delays can happen! Muscle wasting won’t wait, it’ll keep eating away, so living with Duchenne time is not on our side and any time wasted or delays can be fatal for us.

Carl is actively involved in Action Duchenne: www.actionduchenne.org

“I have found that the lack of a multi-disciplined team of specialists in each region is impacting on the lives of young children and young adults every day”
Diagnosis

Key Findings

1. Patients and families affected by rare diseases wait far too long for a correct diagnosis.
2. A worrying number of people with rare diseases receive incorrect diagnoses before their final diagnosis is made.
3. Patients and families worry about the level of awareness of rare diseases among healthcare professionals.
4. Patient organisations play an important role in the diagnosis of rare diseases
5. The experiences of patients and families of diagnosis can vary greatly raising concerns about equality of access and fair treatment in different parts of the country.

Timely prevention and diagnosis of rare diseases is essential. Without accurate diagnosis, appropriate screening programmes and targeting of diagnostic tests, patients and families cannot access effective treatment, therapy, or manage their condition appropriately. A delay in diagnosis or misdiagnosis may also involve multiple avoidable appointments with doctors and consultants, incorrect treatments and diagnostic tests and significant distress.

Our research shows that patients and families affected by rare diseases in the UK often face significant hurdles in securing a final diagnosis. This has serious implications for their life expectancy and quality of life and results in an inefficient use of NHS resources.

“Where I had a tumour which is rare, doctors kept sending me home saying it was migraines or the flu, oh and yuppie flu. Then when they didn’t have any more ideas they told me it was in my imagination.”

A patient with craniopharyngioma

Chart 1: “How long did you/your family member have to wait for a final diagnosis following the onset of disease symptoms?”

% by length of time waiting for a diagnosis

<table>
<thead>
<tr>
<th>Duration</th>
<th>Percentage</th>
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<tbody>
<tr>
<td>less than 3 months</td>
<td>26%</td>
</tr>
<tr>
<td>3 months to 1 year</td>
<td>7%</td>
</tr>
<tr>
<td>6-9 months</td>
<td>6%</td>
</tr>
<tr>
<td>3-6 months</td>
<td>11%</td>
</tr>
<tr>
<td>1-2 years</td>
<td>10%</td>
</tr>
<tr>
<td>2-5 years</td>
<td>17%</td>
</tr>
<tr>
<td>5-10 years</td>
<td>8%</td>
</tr>
<tr>
<td>10-20 years</td>
<td>7%</td>
</tr>
<tr>
<td>Over 20 years</td>
<td>4%</td>
</tr>
</tbody>
</table>

Base: 481 respondents, UK, 2010
Source: Rare Disease UK survey on patients and family experiences of rare diseases
Difficulties in Accessing Diagnosis

Key Finding 1: Patients and families affected by rare diseases wait far too long for a final diagnosis.

Our research shows that patients and families affected by rare diseases can face lengthy delays in accessing a correct diagnosis.

Almost half (46%) of patients with rare diseases had to wait over one year for a final diagnosis following the onset of disease symptoms.

Of this:

One in five (20%) waited over five years.

Over one in 10 (12%) waited over 10 years.

Out of the 19 respondents yet to receive a confirmed diagnosis, 16 have been waiting over a year. Of which, ten have waited five years or more.

“Varying diagnoses and incorrect information given. It was only after the death of our youngest daughter at the age of 6 months that diagnosis was received.”

Mother of a patient with Gorlin syndrome

“I have a genetic disease but was not diagnosed until I was 42 although I had attended a chest consultant/s regularly since the age of seven.”

Patient with primary ciliary dyskinesia

These findings are a major concern. If patients are not diagnosed early enough the delay in accessing effective treatment, therapy or management of the condition can result in life expectancy and quality of life being seriously compromised.

The results also reveal a striking variation – over a quarter (26%) of respondents received a diagnosis within 3 months of the onset of symptoms on one hand, whereas over a third of respondents (36%) waited over 2 years on the other.

Delays in diagnosis and multiple visits to doctors is a drain on NHS resources which could be more efficiently used if the diagnostic pathway for rare diseases was improved. It can also be very stressful for patients and families having to be passed from doctor to doctor, without anyone being able to confirm a diagnosis.

Chart 2: “How many doctors did you/your family member see before the final diagnosis was made?”

% by number of doctors seen before diagnosis

<table>
<thead>
<tr>
<th>Number of Doctors</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 to 2</td>
<td>32%</td>
</tr>
<tr>
<td>3 to 5</td>
<td>46%</td>
</tr>
<tr>
<td>Over 15, 4%</td>
<td></td>
</tr>
<tr>
<td>11 to 15</td>
<td>3%</td>
</tr>
<tr>
<td>6 to 10</td>
<td>15%</td>
</tr>
</tbody>
</table>

Base: 495 respondents, UK, 2010
Source: Rare Disease UK survey on patients and family experiences of rare diseases
Key Finding 2: A worrying number of people with rare diseases receive incorrect diagnoses before their final diagnosis is made.

The results of the survey suggest that an unacceptably high proportion of patients and families affected by rare diseases receive incorrect diagnoses before their final diagnosis is made.

Close to half (46%) of patients were given incorrect diagnoses before receiving their final diagnosis.

Almost one third (30%) had received three or more misdiagnoses.

Not only is misdiagnosis distressing for the patient and their family, but it can lead to a deterioration of the condition as effective treatment, therapy and management of the condition is delayed. This can sometimes result in more expensive interventions being required and, in the worst cases, death.

“Several [incorrect diagnoses], including making up the symptoms and one said that it was terminal cancer!”

Patient with myasthenia gravis, neuromyotonia, and Morvan’s syndrome

“My [son] was not tested to see if he had [the] same gene mutation as my daughter … at age 4 he became ill [and] was misdiagnosed. We were told on three separate occasions “this is not the same disease as your daughter” even though some symptoms were very similar. He was diagnosed with HLH (XLP) five days before his death last year.”

Parent of a patient with haemophagocytic lymphohistiocytosis

Information, Awareness and Diagnosis

Key Finding 3: Patients and families worry about the level of awareness of rare diseases among healthcare professionals.

Patients and families responding to our survey expressed serious concerns about levels of knowledge and awareness of rare diseases among healthcare professionals. The poor practice of some GPs, which included instances of not believing patients’ symptoms and not referring on to specialists, were highlighted as especially distressing.

“GP thought he was attention seeking suffering middle child syndrome.”

Parent of a child with Langerhan’s cell histiocytosis

“Many incorrect diagnoses - but the worst diagnosis doctors (especially GPs) give is there is nothing wrong with your child - perhaps the problem is with you (the parent).”

Mother of a child who died from haemophagocytic lymphohistiocytosis

“Many doctors had no idea and some even said the symptoms were in my head.”

Patient, myasthenia gravis
“Being medically qualified I diagnosed my daughter’s NFI [neurofibromatosis type 1] at birth, based on axillary freckling. The neonatal paediatrician falsely reassured me that there was nothing wrong. The diagnosis was confirmed by other paediatricians at age 4.”

Mother of a daughter with neurofibromatosis and hydrocephalus

It would be impossible for doctors to know about each and every rare condition – there are over 6000 recognised at present. What our findings do indicate is a clear need for better training, sources of information and systems for referral to specialists to support doctors in diagnosing rare diseases.

“I get tingles on my feet and sometimes I fall over. The local district hospital community paediatrician admitted that she didn’t know what was wrong, that it was “probably genetic”, but insisted that it didn’t need further investigation - and refused to see [our son] again. If we had believed her we would have lost our son...I do not expect [doctors] to know about every rare condition - that would be impossible. But I do expect them to have the humility to admit when they are out of their depth and refer on - not just ignore.”

Mother of a child with congenital myasthenic syndrome

Key Finding 4: Patient organisations play an important role in the diagnosis of rare diseases.

Our research demonstrates the need to promote the vital role of patient organisations for people with rare diseases in improving diagnosis. They raise awareness and provide information and resources for the public, patients and families, and medical professionals. Patients and families were supported with information and resources helping them ask the right questions of doctors and in seeking appropriate treatment, therapy or management of the condition if diagnosed.

“All support is via [the] Haemophilia Society, or located by myself online. My son was diagnosed in the USA and there was much more information regarding research supplied there… Haemophilia is congenital, i.e. present at birth and for life. My son visited his GP on 5 occasions regarding excessive bruising symptoms.”

Parent of a child with severe haemophilia A

“Equally striking was that in a number of cases, the patient or family member diagnosed their condition by doing research themselves after doctors repeatedly failed to diagnose them.

“We had to push for diagnosis and do a lot of research ourselves and guide the Geneticist in what testing to perform.”

Relative of a patient with a rare chromosome deletion

Inequality of Experience

Key Finding 5: The experiences of patients and families of diagnosis in the UK can vary greatly.

While our research reveals serious problems with the diagnosis of rare diseases in the UK, there were also clear examples of good practice resulting in early diagnosis, improved quality of life, and efficient use of NHS resources.

“We were referred to the Royal Alexandra Hospital, Paisley, by my family doctor as soon as symptoms occurred. I was given a series of tests very shortly afterwards and the condition [was] diagnosed very quickly. (I know from fellow members of the Association that I was fortunate in the doctors who treated me.)”

Patient with ocular myasthenia gravis

“We were referred to a consultant neurologist as soon as symptoms occurred. I was given a series of tests very shortly afterwards and the condition [was] diagnosed very quickly. (I know from fellow members of the Association that I was fortunate in the doctors who treated me.)”

Relative of a patient with progressive supranuclear palsy

We believe these instances of good practice should be the norm across the UK. There are inequalities in the experience of those with the same condition in different parts of the country and wild variations between service received by patients and families with different rare diseases. Avoidable variations are unfair and unjust.

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Relative of a patient with progressive supranuclear palsy
Jayne Hughes’ story

Jayne Hughes, mother of Amy who has Cockayne syndrome – Merseyside, England

Amy went through the first 15 years of her life with no diagnosis. Various syndromes were mentioned to our family and then dismissed. At no point were we given any support groups to contact. We felt very isolated and alone and struggled to come to terms that we had no diagnosis and no care plan at all. After researching on the internet for some years we found what we thought Amy had - Cockayne Syndrome (CS).

We struggled to get basic treatment and therapies for Amy and eventually, following many visits to many hospitals, we decided to raise funds to take Amy to Boston Children’s Hospital, USA, to see a geneticist who specialised in CS. This doctor took one look at Amy and said ‘she has it without a shadow of a doubt’; music to our ears despite the awfulness of the illness. During this visit we met children for the first time that looked like our child!

Upon our return from Boston in 2006, our family set up a support group for CS called Amy and Friends and in 2007 we became a registered charity. We wrote to genetic departments, paediatric departments and hospices all over the UK trying to find other CS sufferers and their families and to date have found over 60. We became the first support group for CS in the UK and Europe and have families from several other countries from as far and wide as South Africa to Australia.

Several months ago, Amy and I were visiting our local supermarket when I did a double take! There in a trolley sat a little girl who looked just like some of our other CS children and I knew, beyond doubt, that she had CS. Did I rock this family’s world or leave them in oblivion? I remembered my time before Amy was diagnosed of scouring medical books, useless hospital visits and frustration beyond belief and knew that I had to say something...

I approached little Jessie and her mum Jo. I saw Jessie’s little tremor, noticed her thick glasses, thin hair, pedro boots and said how sweet she looked. Her mum told me she was aged 4 and as she was roughly the size of an 18 month child, I knew that she had severe growth problems including microcephaly. She had the telltale high pitched squeaky voice too. Her mum told me that she was unable to walk unaided and finally that she was undiagnosed. I told Jo that I was certain I knew what Jessie suffered from and gave her our website details - completely conscious that I was about to change their lives forever. I gave Jo my contact details and then left them. I felt very uneasy and unsure if I had done the right thing.

I waited to hear and lay awake every night worrying about this family. Four months later I received an email from Jessie’s Dad. Jessie had originally been told that she had a duplication on chromosome 22 but following our meeting, Jo had visited her paediatrician and told them of our conversation. They organised an MRI immediately.

Jo and Jess had then gone to see their geneticist who, ironically, was the same geneticist that Amy had seen for the first 12 years of her life. She mentioned our meeting and conversation and was told ‘I know who that lady is and I take my hat off to her as I think she is right.’ Testing followed.

Whilst waiting for the results, Jessie came to visit Amy and our family. She lives 30 seconds away from us and we are totally unrelated. My 9 year old son met her and said, ‘hey, she has CS1 you know...where did you find her mum?!’

Jessie has now been formally diagnosed with CS1 and since her diagnosis we have learned of two other children with CS in the Merseyside area.

CS is often misdiagnosed or completely missed but hopefully we can change this and help these children take part in the trials that are beginning. It is hoped that the treatment given will slow down the ageing process and in the meantime we can help with support and understanding.

Jayne Hughes set up a support group for Cockayne syndrome, Amy and Friends, www.amyandfriends.org
Anna Pickering’s story

Anna Pickering – who has Ehlers-Danlos syndrome, postural orthostatic tachycardia syndrome and Raynaud’s syndrome – Suffolk, England

At the age of 13, my parents noticed I wasn’t using one of my fingers at all and were very concerned about it. They thought it might have been an injury caused by a skiing accident so took me to the GP for investigation. From then on I had numerous tests and was at a hospital appointment roughly every other month for 3 years. The tests I underwent included nerve conduction studies, MRIs, EEGs, lumbar punctures and lots of others that I can’t remember right now. The appointments were either in Ipswich, Cambridge or London. I had to go to see so many different consultants because, although they all could see there was something wrong, nobody could quite work out what it was. I feel now that I was receiving reasonably good care at that point, although I wasn’t communicated to very well.

I remember a time, I would have been aged 16, when I was sent out of the doctor’s office so he could tell my mum that I had MS. The doctor didn’t say anything about it to me and expected my parents to be able to answer any questions I had. I don’t really know what happened for that diagnosis to be reversed, although I feel that my next London appointment had something to do with it.

After that appointment, I returned to the GP for test results and the letter from the consultant said he thought I was being difficult because I wasn’t giving him the answers to all of his questions. I was most upset about this as I hadn’t been given the chance to talk freely; I was only answering a series of set questions he had prepared. There were also quite a few students in the room and I felt quite overwhelmed by that. At the time I didn’t know it wasn’t normal to be in pain with most movements, I thought everybody felt the same, which was why I never told anyone. I didn’t want to be seen complaining. This was one of my last appointments as a child and I believe this could be one of the reasons why I wasn’t taken seriously as an adult.

Once I turned 18 everything changed. I didn’t hear anything back about my test results and there were no follow up appointments made. Frankly, nobody was interested.

This was very upsetting for me as I was in so much pain and I felt I wasn’t being taken seriously anymore. I managed to get a referral to a rheumatologist but he was very dismissive. He couldn’t understand why I felt so much pain and he said that there couldn’t be anything wrong with me. I felt like I was wasting his time and my confidence was at an all time low.

When I was 22, I was referred to a physiotherapist because of my back pain. She happened to mention that I was hypermobile but I didn’t really think much about it.

A year later I was referred to a hand therapist as the problems I had at school with writing had not got any better, in fact I think they’d got a lot worse. This therapist also mentioned hypermobility so when I got home, I Googled this word which had been mentioned to me a few times.

On a follow up physio appointment I saw a different person and she told me I needed to see a psychologist because she couldn’t understand why I feel so much pain all the time. She was very dismissive and upset me a great deal.

About a year later I managed to get a referral to a Professor who specialises in hypermobility syndrome/Ehlers-Danlos syndrome (HMS/EDS) and got my diagnosis. The elation I felt at getting answers and being taken seriously cannot be put into words!
Jo Grey’s story

Jo Grey who has multiple endocrine neoplasia (MEN) 2a. Her mother, Janet and son Cameron, also have the condition – Kent, England

“Do you believe you can fly?” asked the psychiatrist. If I didn’t have my head over a bucket puking up the nothing I had eaten for weeks, I would have made HER fly – right out the window – for asking such a stupid question. Wearily, I shook my head. Couldn’t she see that I was ILL?

For weeks now I had been retching and vomiting, unable to keep even water down. I had been suffering episodes of severe ‘migraine attacks’, palpitations and breathlessness off and on for three years since the birth of my first child, but now they were significantly worse. My GP was insistent, despite my protests, that the attacks were due to postnatal depression and had prescribed a cocktail of drugs to no avail. I was desperate.

“Do you want to harm yourself?” the psychiatrist persisted. A sensible question at last! All I could manage was to nod pathetically into my bucket. Suddenly, urgent phone calls were made: to my husband to check if I had health insurance; to my GP; to a psychiatric hospital.

“They can take you tonight, it’s all arranged” she said. I could not have cared less anymore.

Alan, my husband and my mother, carried me almost unconscious into The Priory near Bromley in Kent. This quiet unassuming place was to be my saviour. After 3 years of pain, it was now a matter of days before they had diagnosed a large adrenal tumour (NOT postnatal depression). The tumour (a phaeochromocytoma) had been causing my blood pressure to reach immeasurable heights and had put me at serious risk of stroke and heart failure. In fact, the surgeon who finally removed it hadn’t seen anything quite like it for size and activity (probably because he doesn’t perform post-mortems).

Once again, things began to move fast and within the space of 3 years I had been genetically diagnosed with multiple endocrine neoplasia type 2a (MEN2a), along with my mother and my then 2 year old son, had both tumour-containing adrenal glands removed along with my cancerous thyroid, as did my mother, and my son had a prophylactic removal of his thyroid to avoid developing thyroid cancer. We do relatively well now, on a different cocktail of drugs, and are regularly monitored for recurrence (which resulted in the replacement of one of my shoulders in 2007 due to a thyroid cancer bone metastasis).

As you might expect, primary healthcare is one area of concern for me for patients with rare diseases like MEN; particularly regarding diagnosis. The specialist NHS hospital team were fantastic throughout, confirming the need for rare diseases like MEN to be managed in an expert multidisciplinary setting.

I had so very nearly died and this would have left 2 very young children motherless. The thought of what could have happened still haunts me 10 years on.

Jo Grey is Chief Executive Officer of AMEND – the Association for Multiple Endocrine Neoplasia Disorders – www.amend.org.uk
Patient Care, Information and Support

Scarce Information

Key Finding 1: Patients are not provided with enough information on all aspects of their condition.

Over half (52%) of the respondents to our survey felt they hadn’t been given enough information on their condition following diagnosis.

“We simply received the name of the condition, then Googled it.”

Relative/carer of a person with progressive supranuclear palsy

The majority of patients (65%) were given initial information by their specialist. Other people were provided initial information by the internet, patient organisations, GPs and other sources. Some told us that shockingly they were given no information at all when first diagnosed with their condition. These patients are left to their own initiative to find information on their condition (despite the fact that good-quality information is not always easy to find without guidance), or they may come across information only by chance.

Key Findings

1. Patients and families are not provided with enough information on all aspects of their condition, both at first diagnosis and subsequently.
2. Information to patients and families must be provided in a range of formats and at various levels of medical and scientific detail to ensure full understanding and informed decision making.
3. Patient organisations are often the main or only source of information for rare disease patients.
4. There is a lack of support for rare disease patients with their medical and non-medical issues.

“When we heard about our condition we were left to deal with it. I saw an article in paper about a lady who had the same condition as me and there was a number to ring which was Ataxia UK. I can’t thank them enough for bringing us out of the woods.”

Patient, familial hemiplegic migraine type2 with cerebellar ataxia

The fact that most patients are first given information by their specialist shows that there is a need for healthcare professionals to have access to reliable, up-to-date sources of information on rare diseases, so that they are able to pass it on to the patient and their family, particularly as the specialists themselves may not know a great deal about the condition.

Nearly two-thirds (64%) of patients were not given details of the relevant patient support groups at the time of diagnosis.

“Support groups, helplines and newsletters from the Pituitary Foundation have been invaluable for information before my operation and after diagnosis and treatment.”

Patient with craniopharyngioma
Unfortunately, there are no support groups for many rare diseases meaning that patients and families may never get the information and guidance they need.

“Any information would be great, I’ve not had one leaflet. My baby has no skin or skull, and I leave hospital, without even a health visitor’s home appointment, support group, nothing!”

Mother of child with multiple complex rare conditions

Two thirds (66%) of patients and family members say that they now feel they have sufficient knowledge of their condition. Whilst this finding is encouraging, it also suggests a significant amount of patients for whom there is a lack of available information on rare conditions, too few opportunities available to discuss their condition with experts, or a lack of knowledge of rare diseases amongst professionals.

The areas which respondents indicated they would like more information on include clinical management of the condition, progression of the disease and the long-term prognosis, the cause of the condition, current research projects, treatment options, and advice on what to tell schools, A&E and family members. Many people also replied that although they did feel they have a sufficient knowledge of their condition, they always welcome the opportunity to learn more, and like to be kept up-to-date on recent developments.

“You can never have too much information.”

Relative of a patient with haemophilia A

“I think we could always want more, and as research progresses we should get told everything!”

A patient with single system Langerhan’s cell histiocytosis

Key Finding 2: Information should be accessible in a range of formats and at various levels of medical and scientific knowledge to ensure full understanding and informed decision making.

The most common format for information to be provided in was leaflets, with almost half our respondents being given leaflets on their condition. Over a quarter were directed to websites, with many also being given information in an ‘other’ format, which was mainly verbal information from their specialist, but also included print outs from websites, photocopies from books and information in a letter or over the phone. A large number of respondents reported being given no information at all when they were first diagnosed, just the name of the condition which they then had to go and research for themselves.

“[We received a] phone call for diagnosis on New Year’s Eve with the comment from GP ‘I don’t know anything about it go on the computer and look it up’. We didn’t have a computer!!!”

Relative of a patient with myasthenia gravis

“Was told the bare minimum in a five minute chat most of which turned out to be incorrect.”

Relative of a patient with bilateral and chronic uveitis

When asked what their preferred method of being given information on their condition was, the most popular were, to be directed to websites (69%), and to receive leaflets (66%). These are simple ways in which to give information and directing patients and families to the information that exists is of little cost to the NHS, but at the same time, makes a significant difference to patients and families. The preference for information from websites again highlights the need for healthcare professionals to have access to a stock of reliable online resources which they can then simply direct patients to.

39% of respondents like to attend discussion groups and a third like to get information from books (34%) or academic journals (34%). However, over a quarter (28%) of respondents did not understand all the information they were given about their condition.

These findings indicate a need for information to be accessible at a range of different levels of understanding. Some patients will prefer to have very thorough, technical information on the condition, whereas others like to have simple, basic knowledge of their disease meaning information must be signposted according to individual preference. This will ensure that the patient or family will be able to understand and use the content to support effective and appropriate decision making.

“I understood some of the contents, but most of the information would have been understood by someone with a medical background.”

A patient with Langerhan’s cell histiocytosis

Key Finding 3: Patient organisations are often the main source of information for rare disease patients.

For many patients (52%) the main source of information is the relevant patient organisation. Many patients and families rely on these groups to inform them of the condition, what to expect and what support exists/they are entitled to. This again highlights the need for patients to be informed of these organisations (where they exist) when they are first diagnosed with a rare condition.
“All support given comes via TSA [Tuberous Sclerosis Association] - clinic, online sites, research, written info, family liaison workers etc.”

Relative of a patient with tuberous sclerosis

“We were Advised to contact P.S.P Association which has proved invaluable.”

Relative of a patient with progressive supranuclear palsy

“Information was only forthcoming (leaflets, DVDs) after I became involved with Ataxia UK.”

Patient with cerebellar ataxia

“Our families, friends and members of Myeloma UK and the support group have been wonderful. My husband’s employers have been supportive. The actual nurses are very good. The doctors have been at times barely adequate. The public bodies dealing with drug funding and benefits have rarely been helpful and have at times been obstructive and occasionally very offensive.”

Wife of patient with myeloma

Insufficient Support

Key Finding 4: There is a lack of support for rare disease patients with their medical and non-medical issues.

Over one third (37%) of patients do not have someone who they can approach with questions on their condition.

We believe it is vital that there is someone who they can talk to to ensure that they are caring for the patient as best they can. There should also be someone available to direct them to appropriate services, to offer them support with their own needs, as caring for a loved one can be a very difficult job and the impact of this on the carer must be recognised. This ties in to the issue of the lack of a Care Coordinator discussed in “Delivering Coordinated Care”.

“I have never been given an opportunity to talk about my condition with a professional apart from when seeing my consultant in a teaching hospital (usually with 5 + students).”

A patient with palmoplantar keratoderma areata

Our survey shows that support for patients’ and families’ non-medical concerns is greatly lacking.

- Only one third (33%) of survey respondents feel that they receive sufficient support with their social needs.
- Only 29% feel they receive sufficient psychological support.
- Less than one quarter (24%) feel they receive adequate support with financial concerns.

Many of those who said that support in these areas is now being provided reported having to repeatedly ask or fight for access to obtain that support.

These are shocking statistics. Not only do patients and families with rare diseases frequently face a battle to get a diagnosis, they then have to battle to find out the medical impact and how to manage their condition as well as having to cope with day-to-day life without adequate support.

“Support in all areas has been poor. Benefits are now coming in because of constant pressure of me and my parents.”

Patient with Becker muscular dystrophy

“Would desperately like more financial help - I would like to work less hours but receive benefits and do not know how to work out what cutting my hours will mean to me but there seems to be no where I can turn for help.”

Patient with centronuclear myopathy

“We are proactive parents, asking questions at every opportunity. Not sure that all TS carers would consider they had access to either information or professional support.”

Parent of a child with tuberous sclerosis

Of those respondents that are carers for someone with a rare disease, 61% said that their role as carer affected their ability to hold paid employment. Coupled with a lack of information on available benefits and financial assistance, this may result in further financial difficulties for families affected by rare diseases.

“I had to give up my full time job and work part time to fit in with my caring commitments.”

Relative of a patient with juvenile Batten disease

“Frequent illness of child and complex care needs make paid work impossible for me.”

Relative of a patient with an undiagnosed condition

“I would find it impossible to work.”

Relative of a patient with central core myopathy
Anne Begg MP’s story

Anne Begg, MP for Aberdeen South who has Gaucher’s disease – Scotland

I was in my late thirties before I even spoke to someone else who had the same disease as myself. It was a very emotional experience and suddenly lots of things in my life fell into place.

Thirty years before, my mother had insisted that an eight year old shouldn’t look nine months pregnant and something was wrong. Our GP said it was just a ‘baby tummy’, my mother insisted that at eight it couldn’t be. I bruised easily, always looked slightly jaundiced and had been in hospital the year before with a mysterious ‘sore tummy’. One feel of my stomach by a consultant told him I had a huge spleen, but I would need a stay in hospital and a bone marrow biopsy to discover what had caused it.

So the Gaucher’s diagnosis was eventually established and all it said in the medical dictionary my mother looked up was that it was “an inborn error of metabolism”. This was the sum knowledge of my condition - it was very rare, it was more common in the Jewish community and there were two forms (actually there are three), the child one which meant death before the age of 5 and the adult one. I was still alive so we deduced that I had the adult form!

And that was that. Computers hadn’t been invented, far less the internet or Google so there was simply no information about what it meant to have this condition nor any way of tracing fellow sufferers.

Nor was there access to a specialist, not even a haematologist, and the doctors who looked after me for the next thirty years could only deal with the effects of my condition. So a general surgeon took out my spleen when I was 11, followed by the surgeons who had to correct the hiatus hernia, then the obstructed bowel which resulted from the splenectomy. After that, it was the orthopaedic surgeon who dealt with my multiple fractures and painful joints.

I had adjusted my life to living with a degenerative disease and didn’t even think about a treatment or cure. There wasn’t one so there was no use fretting.

Then out of the blue a letter arrived saying there was a new treatment for Gaucher’s called Enzyme Replacement Therapy and was I interested. If I wanted more information there was a Gaucher’s Association and I could phone the Secretary, Susan Lewis. I did and spoke for the first time in my life to someone who also had Gaucher’s. It was a long conversation!

“...I was in my late thirties before I even spoke to someone else who had the same disease as myself. It was a very emotional experience and suddenly lots of things in my life fell into place.”
Rhya Homewood’s story

Rhya Homewood who has hereditary neuropathy with liability to pressure palsies – Kent, England

I became a regular face at my doctor’s surgery when I began experiencing some very bizarre problems including pain in my hands and feet, pain in my shoulder and limbs, and numbness in one side of my mouth including half my lips just like I’d been to the dentist. I was exhausted and felt frail. After being a single parent of four very active boys and taking them hiking, doing loads of sports and being a dance instructor, I could barely get out of bed because I was in so much pain. I couldn’t dress myself or even cut up my own food at meal times.

We were all baffled and all my doctors could do was dish out the painkillers which didn’t help much and try referring me to various consultants to see what they made of things. Eventually I saw a consultant neurologist who listened to my tale of woe. He decided to run a few tests, telling me that apart from having carpal tunnel syndrome (which I had heard of), I fit the profile of a very rare condition called hereditary neuropathy with liability to pressure palsies (HNPP). My neurologist had only ever encountered four other people with the condition and treated two of them, but had never diagnosed someone with it as it’s so rare.

Finally the results came through - a conclusive positive result for HNPP. I was informed that my nerves are demyelinising and are actually deleting, so the muscles no longer get enough nerve signals from the brain and begin to atrophy from loss of use. This condition is incurable and degenerative.

My GP was relieved at the HNPP diagnosis saying to me, ‘I knew there was something wrong and now this proves that you weren’t imagining all these weird symptoms’.

People don’t understand about HNPP, so I tell them I have M.S. just to give them a point of reference. Having a rare condition is an awful position to be in. I had always been fast, strong, able and capable. Now I often end up feeling helpless and useless and hopeless because no one knows how to help you.

Finding Rare Disease UK has been a great experience because I met people with very diverse problems and somehow I didn’t feel quite so alone anymore.

I educate everyone I meet, telling them about HNPP and what it does. I hope that I can continue to spread the word to influential ears in the hope that services for people like me are more widely available and training for medical practitioners in managing patients with a rare disease is improved.

I want to raise awareness as much as I can and keep the plight of people who have rare conditions in the limelight because we have a right to make the most of our lives and with the right support, who knows perhaps the job of foreign secretary is open...? One can dream still.

“I often end up feeling helpless and useless and hopeless because no one knows how to help you.”
Fiona Fisher’s story

Fiona Fisher, mother of Jonathan who has Lowe syndrome – Fife, Scotland

We are the Fisher family living in Fife and looking after our son Jonathan aged 16, with Lowe syndrome, a rare, life-limiting, life-threatening genetic condition that affects only boys. Jonathan has multiple and profound physical, learning and sensory disabilities and complex medical needs, including epilepsy and a renal disorder. He needs 24/7 care and supervision. We also have two healthy daughters, aged 17 and 9.

Living with Lowe syndrome has given us a unique insight to how the UK supports families like ours, from health, education and social services to benefits, leisure and wider society in general. We’ve had to fight long and hard all his life for all manner of services and support, as a child and now he’s legally an adult in Scotland, we’re currently pursuing legal welfare and financial guardianship under the Adults with Incapacity Act Scotland (2000). We have to work hard to cultivate positive working relationships and continually promote our knowledge of Lowe syndrome to professionals.

Crucial to our family has been the support of the Lowe Syndrome Association (LSA) based in the USA. It is a non-profit, patient advocacy charity run by parent volunteers that fosters information, friendship and research of the condition worldwide. Without the LSA, I firmly believe that Jonathan would not still be with us as we gained crucial knowledge about his renal function at a medical conference they hosted in 1998 which subsequently saved his life twice.

Over the years, caring for Jonathan and being grateful for outside support has encouraged me to be involved in a number of UK groups, including Contact a Family, Genetic Alliance UK, PAMIS, InControl Scotland as well as chair of his school parent council. In the same period, I’ve become increasingly politicised about how the UK supports parent carers like me.

Before I had Jonathan, I was a graduate biochemist with 3 years experience in pathology laboratories. If I had been able to stay at work, I estimate I could have reasonably earned £250,000 over the past 16 years. Instead I have been paid Carer’s Allowance to the tune of around £33,000. That’s not per year, that’s for a total of 16 years work, the equivalent of 20 something pence per hour.

On top of that I have no right to a day off, emergency or sickness cover, health and safety training including moving and handling, nor an occupational pension. And because my husband works, I have no entitlement to free prescriptions or dental care, even though I earn less than a Job Seeker. If I had given Jonathan over to state care, he would have easily cost a million and a half to look after so far. ■

“...We’ve had to fight long and hard all his life for all manner of services and support...”
Coordination of Care

Fragmented Care

Key Finding 1: The majority of patients’ care is poorly coordinated.

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

75% of respondents told us that the patient does not have a designated Care Coordinator or Care Advisor.

A Care Coordinator is a trained professional responsible for seeing that a care plan is in place and carried out, and they can greatly assist in ensuring continuity of care at transition times. They are also available to talk to the patient about their concerns and to support the needs of the family or carer. A Care Coordinator can provide vital support to a patient and their family throughout the progression of their condition to ensure that care is carried out as smoothly as possible.

Not having a Care Coordinator can aggravate a number of issues for patients and their families. Some of the problems experienced by our respondents included:

- Each professional the patient comes into contact with looks at specific elements of the condition, but no one being concerned with the condition as a whole.
- Patients or families having to repeatedly tell their story to all professionals involved in their care.
- Feelings of being lost in the healthcare system.
- Patient notes being lost.
- Patients and families not knowing who to go to with queries on their condition.
- A lack of continuity in those involved in the care of the patient.

“Many meetings with medical professionals become us informing them about the condition!”

Wife of a patient with multiple myeloma

“My husband’s care is split between two hospitals... One never requests records from the other so tests are duplicated and delays blamed on each other. No one takes overall responsibility and there is no one person to refer to for even simple requests let alone really important ones. Repeated requests for information go unanswered.”

Parent of son with tuberous sclerosis

“There are] too many new people coming into our son’s life and ours – [this is] really difficult when everyone changes particularly when the family has had to have an open door to all involved.”

Patient with primary ciliary dyskinesia

“Many meetings with medical professionals become us informing them about the condition!”

A relative/carer of a person with non bullous erythrodermic ichthyosis

Key Findings

1. The majority of patients’ care is poorly coordinated.
2. Patients have to attend multiple clinics for different aspects of their condition, often at a long distance from where they live.
3. The majority of patients do not have access to a specialist centre for their condition.
4. Patients frequently experience problems with medical, psychological, financial, social and other issues at transition periods.
The lack of a Care Coordinator often means that there is no-one to take central responsibility for all the different aspects of care received by the patient. As well as the negative impacts already mentioned, this can result, in the worst case scenario, in serious mismanagement of care leading to increased hospital admissions. It is therefore shocking that so few patients have access to someone to fulfil this role.

Respondents were asked who they would like to fulfil the role of Care Coordinator. One fifth (21%) said they think it should be a specialist nurse, but the lack of an overwhelming preference suggests that as long as there is someone available to centrally coordinate care, patients and families don’t mind who it is, provided they have sufficient knowledge of the condition and the situation. This is backed up by a number of comments received from family members:

“If anyone would coordinate my daughter’s care it would be wonderful as I’ve been doing it for years.”

Parent of a patient with 1q21.1 microdeletion

“So long as it is one person - perhaps a specialist nurse if there was one!”

Mother of a child who died from haemophagocytic lymphohistiocytosis

Some patients/carers are happy to coordinate their own care, especially as they often have become the experts in their condition. When this is the case, patients or carers must be empowered to do so.

“As a mother, I know best but would wish specialists to take more notice of what I know is needed”

Parent of child with Prader-Willi syndrome

Key Finding 2: Patients have to attend multiple clinics for different aspects of their condition, often at a long distance from where they live.

A quarter (25%) of all patients attend either 3 or 4 different clinics, with over one in ten (12%) patients having to attend more than five different clinics for their condition.

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

“With so many hospital appointments to attend, I find great difficulty in trying to hold down part-time employment.”

Parent of two children with X-linked agammaglobulinemia

“Although no problem with my employer there are lots of hospital appointments where I feel bad about taking time out”

Relative of a patient with panhypopituitarism

Two thirds (66%) of patients that attend clinics need to travel for over an hour to get to their furthest clinic, with one in three (32%) having to travel for over two hours. More than a tenth (15%) of all patients travel for over three hours to reach their furthest clinic.

These figures reflect that often care isn’t, or can’t be, provided in a local hospital so patients need to travel to bigger regional hospitals for their appointments. Combined with the fact that patients often have to visit multiple clinics, this causes further disruption to normal life, and can have a large financial impact on the patient or their family due to the cost of travel. Improved communication and support between local and regional services would enable more care to be provided locally, reducing the need for patients to travel long distances for their needs to be met.
Key Finding 3: The majority of patients do not have access to a specialist centre for their condition.

Less than half (45%) of respondents said that they are aware of a specialist centre for their condition.

Less than a third (32%) of respondents knowingly access a specialist service for their condition. 31% of respondents were not sure if there is a specialist centre for their condition which may be indicative of a lack of information about available services.

The majority of those who named specialised centres, named services that are commissioned at a national level for those conditions by NHS Specialised Services in England. However, the diseases these services cover are a very small proportion of the total number of rare diseases.

One issue highlighted was the difficulty patients sometimes experience in accessing experts in their condition who are based in a different home nation to themselves.

“[We can’t] afford £225 for rail fare to be in London for a 10am appointment [which takes] 3-4hrs... so we drive down and stay overnight in a [hotel]. [It] takes 4x longer, but is half the cost.”

Mother of a child with Proteus syndrome

“The majority of patients do not have access to a specialist centre for their condition. The centre will be made up of a team of multidisciplinary specialists, as well as scientists and researchers. Specialist centres support patients across the UK, not just in their local area.

A number of respondents highlighted that the inconvenience of having to attend multiple appointments is aggravated by other practical issues, most notably, problems in parking at hospital car parks.

Patients and families are generally happier to travel if it means all their appointments can be carried out in a one day multidisciplinary clinic where they can meet all the specialists in one place. This highlights the value of ‘one-stop shop’ clinics which cause less disruption to a family, and supports the need to further develop these for more conditions where possible.

There are also good examples highlighted of professionals travelling to patients:

“All professionals visit the school so I don’t have to visit them all (brilliant idea).”

Mother of child with X-linked congenital ichthyosis and multiple other conditions

Expertise on rare diseases is often hard to come by, so patients should be supported to access specialists where they do exist without being unnecessarily restricted by territorial boundaries between the NHS in each of the four home nations.

Transition Troubles

Key Finding 4: Patients frequently experience problems with medical, psychological, financial, social and other issues at transition periods.

Almost 30% of patients reported experiencing problems in the transition from paediatric to adult services. This may actually be higher as a significant number of respondents replied that they ‘didn’t know’ whether problems had been experienced.

“I was ‘forgotten’ about by medics when I turned 18”

Patient with Ehlers-Danlos syndrome, postural orthostatic tachycardia syndrome, Raynaud’s syndrome

“We are in the process of transition. As the support line, I have listened to parents who have gone through this and who were desperate for an adult centre. Only now do I fully understand their desperation”

Relative of a patient with Proteus syndrome

3 In the survey, we defined a specialist centre as a centre that is able to provide expert advice on diagnosis, assessment and treatment of a particular condition. The centre will be made up of a team of multidisciplinary specialists, as well as scientists and researchers. Specialist centres support patients across the UK, not just in their local area.
Problems were experienced in all areas of care – medical, psychological, financial and social.

Medical problems experienced in transition included:

- Lack of communication between specialists, or between specialists and the patient/family.
- Patient or family not being given a discharge pack/handover/transitional arrangement.
- Lack of knowledge of the condition within the adult service.
- No one to take responsibility for the patient.
- No holistic approach to care.
- Conflicting information received.
- No consultant available to take on care.
- No more clinics offered/no specialised adult service.

Psychological problems included:

- No support offered to patient or family.
- Feelings of isolation and of being ‘cut off’ by a team the patient has come to know.
- Difficulties arising due to the patient being unable to deal with change.
- Families not being offered any counselling even when there is a great need for it.
- Psychological support of adults services not being as good as that in paediatric services.
- Patient now being treated as an adult despite having the understanding of a child.

Financial problems experienced:

- Lack of information given on the changing financial support available for patients.
- Patient needing to attend more clinics as an adult, resulting in increased costs of travel.
- Patients no longer being able to claim benefits despite having been able to get them as a ‘young person’.

“Trying to secure appropriate long-term benefits in adulthood was a bureaucratic nightmare.”

Mother of a daughter with 18q minus

Social problems experienced in transition:

- No (or very limited) social support offered.
- Not being provided with any information on available social clubs or groups, or how to access those that do exist.
- New social teams have different approaches to old teams.
- Social workers lack experience or knowledge of the condition.

“Huge change for the family - new teams - no introduction - difference in approach.”

Mother of a child with X-gammaglobulinemia

The high number of problems experienced demonstrates the need for the development of resources to ease transition. We suggest this should include disease routemaps to lay out the general progression of the condition, and what support is available to patients and healthcare professionals at various stages of the disease; as well as patient-held records to give the patient or family control of their medical notes which they can then pass to the relevant professionals to prevent them having to continuously repeat their stories and medical history. The appointment of a Care Coordinator would also ensure continuity and communication between all necessary people at transition.

“Needed counselling, but [the] waiting list was so long my daughter attempted suicide.”

Mother of a daughter with 18q minus
Kay Parkinson’s story

Kay Parkinson, mother of Charlotte and Matthew who had Alström Syndrome – Devon, England

We first heard the words Alström Syndrome in 1996, when our two children were then 18 and 15. Soon after birth, they had both been extremely sensitive to bright lights and their eyes seemed to wobble up and down. My daughter also collapsed at 11 weeks in heart failure due to dilated cardiomyopathy but we were told that there was no connection between any of the symptoms they were experiencing and that my daughter’s heart condition was probably due to a virus. They were both registered blind by the time they were 5.

Charlotte also developed hearing problems at age 7. We had been given various different diagnoses during their lifetime which seemed to change as different eye specialists came and went. We really became disinterested in the diagnosis (which only related to their eye conditions anyway) and concentrated on giving our children as happy a life as we could.

When our son was 16, he collapsed - also with heart failure due to dilated cardiomyopathy and for the first time the medical professionals started to think that maybe there was a connection between all the problems that my children were experiencing.

Another new diagnosis was made – Leber’s amaurosis. We were then asked if we would be willing to go to Great Ormond Street Hospital as a consultant there was looking into this condition. This consultant said their condition was not Leber’s amaurosis but she did know what it was, it was called Alström Syndrome and she had written a paper on 22 people with the condition. She also said that my children would probably also be diabetic (they were) and to get them checked for this.

In 2003 Matthew’s heart started to deteriorate and he was referred for a heart transplant at Papworth Hospital. He received his first offer of a heart just four hours after going on the list; unfortunately that heart could not be used. Just two days later he received a second offer of a heart and this time the operation was able to go ahead. Sadly, Matthew only lived for one week after the transplant.

In 2007, Charlotte went into kidney failure and went onto dialysis. This put an enormous strain on her already weak heart and she was referred for a combined heart and kidney transplant at Queen Elizabeth Hospital in Birmingham. Charlotte waited over a year on the list and finally got the call that the organs were available in April 2009. Charlotte never awoke from the surgery.

Had Matthew and Charlotte been diagnosed sooner they would not have needed transplants at such a young age. Both had un-treated diabetes for many years before being diagnosed, this is now a standard test at all our Alström clinics. Matthew should have been screened for dilated cardiomyopathy when his sister collapsed at 11 weeks and we always screen for this annually now at the clinics. Medication can help slow the progression as do regular exercise and healthy diets. Matthew collapsing in heart failure, which is the worst possible state to have been left in, meant he struggled from then on.

Matthew and Charlotte are a classic, tragic example that a failure to treat is not cost effective on very many levels.

Kay Parkinson set up Alström Syndrome UK Support Group for people affected by Alström syndrome. She now helps to run the nationally commissioned clinics to ensure all Alström patients have access to the services they require. www.alstrom.org.uk
Craig and Gemma Mitchell’s story

Craig and Gemma Mitchell, parents of Ella Mitchell who has a 6p25 Deletion – Kent, England

Our elder daughter Ella was born in 2003 and after a tough start suffering with infantile spasms, a serious form of epilepsy was diagnosed with a rare chromosome disorder at nine months old. She has part of chromosome 6 missing (a ‘6p25 deletion’), a very rare condition about which doctors and other involved professionals have little knowledge. Ella has severe learning and communication difficulties, hypermobile joints, and a pronated right foot/ankle and can tire easily. She wears strong glasses and has regular hearing tests. She also has a skin condition called morphoea and is on strong medication for this which requires regular blood tests. Naturally, she hates having them done so they are very stressful for her and everyone else involved.

Ella requires lots of additional care and has at least 2 hospital appointments per month, often at London hospitals, which means lots of travel from our home in Kent. We have regularly come into contact with doctors and other professionals who are seeing Ella for the first time and the rarity of her chromosomal disorder means they often have little knowledge or understanding of it. We have lost count of the number of times a doctor or other professional has asked us to “begin at the beginning”, as they try to understand Ella’s condition and the health issues associated with it. We were even once asked “so.... why are you here?” by a doctor! This just adds to the stress of living with a disabled child and coping with all the issues that result.

Parents and carers of disabled children are often tired to the point of exhaustion and almost never get a break as children like Ella require constant care and attention. There is very little public awareness or understanding of what it really means to raise a disabled child, particularly one with a rare disorder.

Having a care management pathway or someone to act as a care coordinator right from the time of initial diagnosis would mean not having to answer the same questions over and over again as each ‘new’ person we see tries to understand Ella’s disorder. Parents of disabled children often have to fight for even the most basic services and it seems that those with rare diseases have to struggle even more. Deliveries of nappies for Ella recently stopped pending a ‘reassessment’, but nobody actually told us this in advance. Simply finding out who we needed to speak to about this became a struggle in itself!

Awareness of conditions such as Down’s syndrome, cerebral palsy and autism is currently high but parents of children with rare disorders constantly have to explain their child’s condition and justify why they need services. This is often very time-consuming and extremely frustrating and upsetting.

Craig works for Unique, a source of information and support to families with a child with a rare chromosome disorder. Please visit www.rarechromo.org
When I was 16, I fell out of bed and broke my leg. The doctor in our local hospital wanted to put me in a plaster cast but my Mum knew that this would only encourage muscle wastage. She asked Hammersmith to write to my doctor to explain that a plaster cast was not suitable. The doctor came storming over to my bed with fax in hand and said “tell your Mum that if she wants Hammersmith to treat you we’ll send you up in an ambulance and they can deal with you instead”. I realised at that moment that doctors don’t know it all and I have to stand up for my needs. I haven’t let SMA stop me from living life to the full - I did well at school and then went to University. After graduating I moved back to London and have had a successful career, including at the BBC where I now work. We would drive up to Hammersmith Hospital every year to the SMA clinic where the top neurological doctors and physiotherapists were. It’s very easy to look at disability from a medical standpoint; to be quite black and white about what’s causing the disability and to fix things with medical intervention. I think doctors should also consider the emotional and psychological side of life. There were no egos at Hammersmith and the medical professionals worked with me and my parents rather than just dish out prescriptions and remedies.

In March this year, I became very ill and ended up in intensive care. There, my kidneys, liver and stomach stopped working and my heart was playing up as well. The doctors decided to put me in an induced coma and I was there for two months. I don’t have any memory of my stay in hospital from the time I was in A&E until the day I woke up. I had to have chest physiotherapy three times a day, occupational therapy to strengthen my arms and hands, several blood tests and doctor visits.

Thanks to the care that I received, from both the hospital staff and my personal care assistants, I have been able to get back to work and my normal way of life. Although I have SMA, with the appropriate support, I have found it is possible to lead an active and fulfilling life.

“Thanks to the care that I received, from both the hospital staff and my personal care assistants, I have been able to get back to work and my normal way of life.”
Charles and Miranda’s story

‘Charles’ husband of ‘Miranda’ - Somerset, England Confirmed diagnoses - Wegner’s granulomatosis, autoimmune hypophysitis, severe premenstrual syndrome. Suspected diagnoses - chronic fatigue syndrome, sleep apnoea

"Your condition does not match anything in my medical reference books and I am not sure what more we can do, other than if you found somewhere you thought could help, I would be more than happy to arrange a referral". It was not for the want of skill or effort on the part of my wife Miranda’s GP that led to her being told this at one of her frequent visits to Dr Clarkson. Since she had become ill in 1991 after the birth of our first child, her GP had been most supportive and spent many hours of his own time trying to track down what could have caused her to become so disabled.

Her symptoms of insomnia, fatigue, sweating and menstrual irregularities had defied all efforts to control and bring in relief for her. Some of her symptoms were non-specific and although they all started simultaneously, the unknown was if this was caused by one or more undiagnosed conditions. Dr Clarkson had an excellent rapport with many consultants at both the general district hospital and the regional tertiary hospital and for the past nine years he had not hesitated to refer Miranda for further investigations by a number of different specialities.

All of these investigations drew a blank. Miranda’s condition was not responding to any treatment. Steadily, she was finding it increasing difficult to manage. Had it not been for me being present when her GP said he was not sure what else he could do, Miranda said she would not have coped. She had started to tell herself that she must be making it up as no one could put a name to her condition or help her in anyway. She started to get depressed and was referred to a psychiatrist. Miranda was started on anti-depressants, but these only made her symptoms worse. After being seen by several different psychiatrists, she was advised to start lithium treatment. Thankfully, in 2001 she was referred to The Maudsley Hospital. They categorically stated that primary depression was not the cause of her condition, although they could not suggest what was.

Miranda and I took a decision to sell our family business, to enable me to spend more time in caring for her and our two children. This also allowed me to start researching doctors and hospitals that could possibly help my wife. I spent hours calling secretaries of consultants to ask if they had ever seen or could help such a case as Miranda’s. This resulted in a number of NHS and private referrals, some different treatments, but still no nearer a much needed diagnosis. One evening an NHS consultant retuned my call and although he did not feel he should be involved in Miranda’s care, suggested that the Mayo Clinic in Rochester USA may be worth considering. Miranda’s GP agreed and within a week she was admitted for extensive tests, including her first ever MRI scan. This revealed a swollen pituitary stalk and an abnormal auto-antibody result, although even Mayo admitted her condition was very very complex. This trip cost our family $20,000 and exhausted all of our savings.

On returning to the UK with the Mayo Clinic’s recommendations, the local PCT were concerned about escalating costs of investigations without a positive outcome and the family GP had to fight long and hard to get funding for the referrals the Mayo Clinic had advised.

Over the next 10 years, Miranda was seen by several different departments in several London teaching hospitals. During this time, it was largely down to my efforts that correspondence and test results were circulated between hospitals and it was only after being finally diagnosed earlier this year that I was able to persuade one consultant to telephone another hospital to discuss her case!

Meanwhile, the strain of 20 years has taken its toll on our family. I ‘burnt out’ trying to look after them, manage my wife’s care and hold down a job. The two children require psychological support and we are currently being helped to find suitable support that we need on a daily basis.

‘Charles’ and ‘Miranda’ wanted to remain anonymous.
**Key Findings**

1. Trying to obtain medicines can be distressing for some patients and families.
2. There is no licensed treatment available for most patients with rare diseases.
3. Some patients are informed of off-label or unlicensed medicines but often patients and families have to inform their doctor.
4. Patients and families experience inconsistencies in access to medicines.

**Treatment and Therapies**

There are no effective treatments or therapies available for most patients with rare diseases, however, effective treatment can transform the lives of patients and their families and many live in hope that a treatment will be developed.

“Eventually we arrived at a tertiary neuromuscular centre...and gained an accurate diagnosis and appropriate drugs. My son’s life has been transformed by the diagnosis and drug regime. Without this he would have a very poor quality of life - and he could have died.”

Mother of child with Congenital Myasthenia

**Access to Medicines**

**Key finding 1: Trying to obtain medicines can be distressing for some patients and their family.**

A number of patients and family members chose to highlight the distressing experience of gaining access to medicines. This was due to:

- Feeling like they had to battle doctors or PCTs to access medicines
- Delays in access
- Failure to access

“We were informed in Yorkshire that the drug was a red drug and as such could only be prescribed by a hospital consultant. This resulted in my husband knowing there was a drug which would help him but having to liaise between a consultant and GP, a very upsetting experience.”

Wife of a patient with myasthenia gravis

“GP’s want to protect their budgets. [The] drugs the patient needs [are] very expensive, so [the] GP passes the buck and the NHS will only prescribe the drugs once condition has become so bad that patient [is] almost blind. I wasn’t willing to let the condition get to that point so paid to be seen privately. Total disgrace.”

Relative of a patient with chronic and bilateral uveitis

“Growth hormone treatment is routinely prescribed in the US, but families in the UK have a huge battle to get this as part of their children’s treatment.”

Relative of a patient with 18q minus syndrome

“Guidelines say it should be prescribed but the consultant will not do so, in limbo at the moment.”

Patient with ataxia and irreversible B12 neurological damage

**Key finding 2: There is no licensed treatment available for most patients with rare diseases.**

Only around one third (35%) of respondents said that there is a licensed treatment for the rare disease they are affected by. Of this 89% receive the licensed treatment.

Of the remainder, a third (34%) responded that there isn’t a licensed treatment available whilst a further third (31%) do not know whether there is a licensed treatment for their condition available or not. This could possibly reflect a lack of understanding about whether the treatment is licensed or not, or it could be reflective of the lack of information given to patients about their condition and treatment options. Many respondents elaborated they take treatments to ameliorate the effects of the condition but there is no treatment for the condition itself.
Key finding 3: Some patients are informed of off-label or unlicensed medicines but patients and families often have to inform their doctor.

One in five respondents (18%) have been informed of off-label or unlicensed medicines for their condition, although it was apparent from many of the responses that the original information came not from the GP or consultant but from external sources. Patient organisations play a role in disseminating this information.

“I have to give my members details to take to their GP or consultant to enable him to write a script.”

Patient with systemic mastocytosis

“I have had to find information for myself, my doctor still doesn’t have a clue”

Patient with hypermobility syndrome (EDS III)

Key finding 4: Patients and families experience inconsistencies in access to medicines

80% of patients who have been informed about off-label or unlicensed medicines for their condition have been able to access these medicines. When asked how easy it was to access these medicines the results showed a wide disparity between those who found it unproblematic on the one hand, to those having to fund it themselves on the other:

- 50% of respondents indicated that it was not problematic
- But
- 14% said that they had to appeal to their PCT. Appeals can be a lengthy process which cause significant anxiety and distress for patients and their family and can have other knock on implications.

“The delay in accessing [the drug] caused a 5 month delay in my husband’s return to work. Now we are fighting for benefits as his pay has run out which wouldn’t have happened without the delay.”

Wife of patient with multiple myeloma

It isn’t just appeal processes that cause difficulties in access. Many patients have to persuade their clinician, who may know little about the condition to prescribe the medicine.

“It took over a year of negotiation by our GP and ourselves before our son’s neurologist would agree to prescribe [the drug].”

Parent/carer of patient with Niemann Pick Type C

Other Issues

Some patients raised the issue of continuation of funding when an unlicensed medicine which they are using is given a license and the price to the NHS increases substantially.

“The [drug] really works for me so I hope and pray the PCT will let me continue taking it.”

Patient with congenital myasthenia whose doctor is currently liaising with the PCT regarding continuation of treatment

Other answers reflected the importance of research to develop new therapies, but highlighted concerns regarding funding:

“About one third of GPs will prescribe [the medicine].”

Relative/carer of a patient with progressive supranuclear palsy who has to fund medicine privately

“A third (34%) had obtained the medicine by other means, most notably, having to fund it privately or by participating in research

50% of patients/their family members who had not been able to access off-label or unlicensed medicine responded that they know of people with the condition in other areas that can access the medicine. Whilst the number of people responding to this question was small, it does allude to the post-code lottery people with certain conditions face in accessing medicine.

A relative/carer of a person with alkaptonuria ochronosis

“There is no known cure, treatment or medication for alkaptonuria - although we have created the AKU Society and research is being carried out. Funding is of course difficult.”

“Rare diseases attract less funding for research, therefore less is known, particularly about treatment.”

Mother of daughter with neurofibromatosis and hydrocephalus

Other Issues

In the patient survey RDUK used the following definitions of unlicensed and off label: An unlicensed medication is a medication without a UK product licence. These include medicines undergoing clinical trial, those awaiting UK authorisation and those that are licensed in, and imported from, another country. An off-label medication is an approved medication that is prescribed outside the terms of the product licence. This may be in relation to the approved age, indication, dose of frequency, route of administration or formulation of the medication.
Nick was diagnosed with multiple myeloma at Easter 2009. This “bone marrow cancer” accounts for 1% of all diagnosed cases of cancer. The few GPs who encounter it will probably see only one case in their working lives. He had been diagnosed in the summer of 2008 with arthritis (“well, you are an ex rugby player”). When he couldn’t lift the shopping out of the car for me we demanded more tests.

His spine has collapsed causing him to lose 7 inches in height. He has lost 6 stone in weight.

Nick was due to have a stem cell transplant in November 2009 but in October he failed to produce enough stem cells and needed the specific drug to treat this condition. There is NO alternative drug or treatment available for patients with multiple myeloma. This drug was available on the NHS in parts of England at the time. It was not available in Wales, where we live. In England, he could have paid for it privately. In Wales we could not do so. After an Individual Patient Appeal it was administered in April 2010.

The delay of 5 months has had repercussions. Had it gone ahead his Hickman line would have been removed after the transplant; instead he had weekly visits for the extra months from a district nurse to have it flushed. In January, it became infected and he spent three weeks in hospital with intravenous antibiotics and intensive nursing. Since an incorrect line was fitted in its place he had a total of six unnecessary surgical procedures. His paraprotein levels began to rise after October 2009 indicating that his cancer was active so he also had to have two further bone marrow biopsies. He was prescribed a further course of another drug to control the cancer.

In January 2010, Nick was diagnosed with a hernia. This would have required a local anaesthetic and a simple keyhole repair as an outpatient. As I write this in October 2010 he needs a general anaesthetic, a full operation and three nights in hospital to repair the damage which has occurred during the delay.

Had the original drug been available when it was needed none of this additional expense to the NHS would have been incurred. I cannot describe our emotions.

Nick had intended to return to work in July 2010. This will now not happen until January 2011. His sick pay has ended. Jobcentreplus consider that he is not seriously ill (“well you haven’t had a tumour have you? It isn’t like you have had an organ transplant”) and we have battled for every penny of the benefit we receive. No provision has been made for his Pension contributions, putting him at further risk when the cancer returns. Nick is a Deputy Headteacher. Instead of claiming benefits for these months he should have been contributing to society.
Annex 1: List of conditions represented in the survey

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

**Single conditions**
- 18q minus
- Acromegaly
- Acute intermittent porphyria
- Addison’s disease
- Adrenoleukodystrophy (ALD)
- Adrenomyeloneuropathy
- Agammaglobulinemia
- Alagille syndrome
- Alkaptonuria
- Alpha-1 antitrypsin deficiency
- Ataxia telangiectasia
- Bardet Biedl syndrome
- Becker muscular dystrophy
- Behcet’s syndrome
- Beta keto thialase defecit
- Birdshot chorioretinopathy
- Bullous ichthyosis
- CD 40 Ligand PID
- Centronuclear myopathy
- Cerebral vasculitis
- Cerebellar ataxia
- Cerebral palsy
- Charcot-Marie-Tooth syndrome
- Children’s interstitial lung disease (chILD)
- Chromosome 4q deletion (13.1-21.2)
- Chronic inflammatory demyelinating polyneuropathy
- Common variable immunodeficiency
- Congenital myasthenia
- Congential hypopituitarism
- Congential lung surfactant deficiency
- Cushing’s disease
- Cyclical cushing’s syndrome
- Cystinosis
- Degos disease
- Distal spinal muscular atrophy (adult onset)
- Duchenne muscular dystrophy
- Ehlers-Danlos syndrome
- Epidermolytic hyperkeratosis
- Erdheim-Chester disease
- Erythropoietic protoporphyria
- Fragile X syndrome
- Gorlin syndrome
- Haemophagocytic lymphohistiocytosis
- Haemophilia
- Harlequin ichthyosis
- Hashimoto’s encephalitis
- Hereditary angioedema
- Hirschsprung’s disease
- Histiocytosis
- Huntington’s disease
- Hyper parathyroid jaw tumour syndrome (HPT-JTS)
- Hypermobility syndrome
- Hypogonadotrophic hypogonadism
- Hypopituitarism
- Ichthyosis bullosea of siemens
- Ichthyosis lamellar
- Ichthyosis vulgaris
- Ichthyosis-Sjogren (Larsson Syndrome)
In this section, we list a variety of rare diseases and conditions, each accompanied by an Insight from Patients and Families. Here is the list of rare diseases:

- Infantile Batten’s disease
- Interstitial lung disease
- Juvenile Batten’s disease
- K.I.D. Syndrome
- Kabuki syndrome
- Lambert-Eaton myasthenic syndrome (LEMS)
- Lamellar ichthyosis
- Langerhan’s cell histiocytosis
- Laurence Moon Bardet Biedl syndrome
- Lennox Gasteau Syndrome
- Lowe syndrome
- Macrocephaly capillary malformation (M-CM)
- Metaphyseal chondrodysplasia
- Migrating partial epilepsy of infancy (MPEI or MMPSI)
- MPS III A (Sanfilippo syndrome)
- Myasthenia gravis
- Netherton syndrome
- Neurofibromatosis and hydrocephalus
- Niemann-Pick disease type B
- Niemann-Pick disease type C
- Non-bullous erythrokeratodermia ichthyosis
- Noonan syndrome
- Obliterative bronchiolitis
- Ocular myasthenia gravis
- Olliers disease/Maffucci syndrome
- Panhypopituitarism
- Paroxysmal nocturnal haemoglobinuria (PNH)
- Peroneal muscular atrophy
- Perthes disease
- Pitt-Hopkins syndrome
- Pompe disease
- Prader-Willi syndrome
- Primary ciliary dyskinesia
- Primary immune deficiency
- Primary lung cancer with secondaries
- Progressive supranuclear palsy (PSP)
- Proteus syndrome
- Rare chromosome deletion
- Relapsing polychondritis
- Rett syndrome
- Severe haemophilia A
- Sheehan’s syndrome
- Sickle cell anaemia
- Simpson Golabi Behmel syndrome
- Single system LCH
- Spinal muscular atrophy
- Stickler syndrome
- Systemic mastocytosis
- Tar syndrome
- Thyroid cancer
- Trichorhinophalangeal syndrome type I
- Tuberous sclerosis
- Turner syndrome
- Uveitis chronic and bilateral
- Von Hippel-Lindau disease
- Von Willibrand’s disease
- West syndrome
- Williams syndrome
- Wilson’s disease
- Worster-Drought Syndrome
- X linked ichthyosis.

### Multiple conditions

- 18 p deletion/Monosomy 18
- 1q21.1 micro-deletion which has caused autism, epilepsy, severe learning disabilities & severe challenging behaviour
- Adams Oliver syndrome, apnea, cutis aplasia, skull defect, 6mm ASD, wet lungs, pulmonary arterial stenosis, abnormal feet/hands, abundant toes, webbed toes
- Addison’s; neutropenia; autonomic disfunction
- Behcet’s Disease, Fibromyalgia, Osteoarthritis, degenerative disc disease, IBS, gastritis
- Bilateral perisylvian polymicrogyria, epilepsy, global developmental delay, talipes
- Bladder extrophy & epispadias
- Born with thyroid not working, caused epilepsy, deafness and ataxia at different stages of life
- Central core myopathy, cleidocranial dysostosis.
- Craniopharyngioma brain tumour, diabetes insipidus, hypopituitarism, fibromyalgia, adult growth hormone deficiency
- Connective tissue disease, secondary adrenal insufficiency, arachnoid cyst near pituitary gland
- Craniopharyngioma resulting in panhypopituitarism
- Craniopharyngioma, diabetes insipidus, hypopituitarism, hypothyroidism
- Ehlers-Danlos syndrome, postural orthostatic tachycardia syndrome, Raynaud’s syndrome
- Erythromelalgia and undiagnosed condition(s)
- Familial hemephlegic migraine type2 with cerebellar ataxia
- Familial partial lipodystrophy (Dunnigan-Kobberling), Lamin A/C Mutation/PPARG
- Heart defect, repeated sinus, ear & chest infection, coughing
- Hypermobility syndrome and Raynaud’s and possible fibromyalgia
- Infantile spasms, polymicrogyria ocular motor apraxia global development delay scoliosis
- Kartagener’s syndrome and primary ciliary dyskinesia
- Lymphocytic hypothyroiditis and Guillain Barre syndrome
- Myasthenia gravis, neuromyotonia, Morvan’s syndrome
- Myasthenia gravis and chronic fatigue syndrome
- Nail patella syndrome and hypermobility syndrome
- Narcolepsy with Cataplexy
- Neurofibromatosis and hydrocephalus
- Oculopalatal tremor and cerebellar ataxia
- Ollier’s disease and Maffucci syndrome
- Papillary thyroid cancer, follicular variant and either ‘side effects’ or other undiagnosed condition
- Paraneoplastic limbic encephelitis with small cell lung cancer suspected
- Pierre Robin syndrome, cleft palate, bi-lateral talipes, multiple epiphyseal dysplasia, hypermobile dislocating joints
- Pituitary adenoma, adrenal insufficiency, hypothyroidism
- Pituitary tumour, craniopharyngioma,
- Prader-Willi syndrome, tourette syndrome
- Proxysmal tonic upgaze and episodic idiopathic vertical nystagmus
- RAS, ADHD, sleep disorder
- Relapsing polychondritis, lupus, sjogrens syndrome, osteoporosis, vasculitis, depression, anorexia
- Stiff Man syndrome, type 1 diabetes, hypothyroidism
- Systemic sclerosis with associated microstomia, severe Raynaud’s
- Terminal disease destroying brain and nerve functions
- Tetrology of Fallots & Di-George syndrome
- Townes Brocks syndrome and atrial septal defect
- Townes Brocks syndrome, chronic renal failure, gut motility disorder, ASD
- Tuberous sclerosis and epilepsy
- Undiagnosed disease of genetic origins - specific gene not yet singled out. History of 3 other genetic cancers over last 15 years.
- Williams syndrome, autism, ADHD
- RAS, ADHD, sleep disorder
- Relapsing polychondritis, lupus, sjogrens syndrome, osteoporosis, vasculitis, depression, anorexia
- Stiff Man syndrome, type 1 diabetes, hypothyroidism
- Systemic sclerosis with associated microstomia, severe Raynaud’s
- Terminal disease destroying brain and nerve functions
- Tetrology of Fallots & Di-George syndrome
- Townes Brocks syndrome and atrial septal defect
- Townes Brocks syndrome, chronic renal failure, gut motility disorder, ASD
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