Experiences of Rare Diseases: Patients and Families in Wales

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

About Rare Disease UK

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

RDUK was established by Genetic Alliance UK, the national charity of over 150 patient organisations supporting all those affected by genetic conditions, in conjunction with other key stakeholders in November 2008 following the European Commission’s Communication on Rare Diseases: Europe’s Challenges.

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

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

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

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

RDUK is funded by an unrestricted educational grant from its industry members.
**Acknowledgements**

We wish to express our gratitude to the patients and families who took time to respond to the survey and also to those who volunteered their time and knowledge to feature as a case study within the report.

We would also like to thank Stephanie Yin for compiling this report.

*The Rare Disease UK team*
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Chair’s Foreword

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

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

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

The Welsh Government and the NHS in Wales is committed to working with the other nations in the UK to develop a plan for improving services and support to rare disease patients in Wales by the end of 2013. As this report shows, there is a basis on which this can be built. But as this report also shows, there is still some way to go before all rare disease patients and families can be confident that they are receiving timely, high quality support. We look forward to a close collaboration with the Government, politicians, clinicians, planners and commissioners to bring this about.

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

In 2010, Rare Disease UK (RDUK) carried out a survey of patients and families affected by rare diseases in the UK. The aim of the survey was to find out more about their experiences. The findings suggested that some patients’ experiences differed according to the country in which they lived (RDUK, 2010).

In 2012, RDUK decided to conduct country specific surveys in Wales and Scotland. Based on the findings of the 2010 survey, the Welsh version was designed to provide new information in the context of the separate health service of Wales.

The survey was sent electronically to all members of RDUK and also placed on the RDUK website, inviting anyone living in Wales and affected by rare disease to participate. We received a total of 82 responses, representing a small participant pool however this was to be expected. In the 2010 survey, only 6% of participants were from Wales and this was found to be in accordance with UK demographics.

Of the 82 responses, 63.4% were from patients themselves with the remaining 36.6% being from relatives or carers of someone affected with rare disease. All 22 counties had at least one participant representation, with the exception of Anglesey and Merthyr Tydfil, which had no responses.

A total of 59 different rare diseases are represented in the survey. The full list can be found in Appendix 1. Some patients are affected by a single condition, others suffer from multiple conditions whilst some remain undiagnosed.

The findings suggest that there is room for improvement, particularly when it comes to accessing services and information regarding rare diseases. Patients and families are keen to see more development in the area of research, and issues have been identified during the transition from paediatric to adult services.

Throughout the report we have included a few case studies to illustrate and highlight some of the issues patients and families face. RDUK would like to thank the individuals that contributed to the case studies and all those that completed the survey.

Summary of Key Findings

Research

- Patients and families do not feel that enough research is being conducted and want more opportunities to participate.
- Patients are willing to participate in research, even if it requires travelling.
- Patients and families feel they are not kept well informed about research being conducted.

Diagnosis

- Patients and families sometimes wait a very long time to receive a correct diagnosis.
- Patients often receive incorrect diagnoses before arriving to a final correct diagnosis.
- Patients must attend multiple appointments before receiving a final diagnosis.
- The majority of patients were diagnosed in Wales but many are willing to travel for specialised services.
- Ratings for ‘health service received’ show varying degrees of satisfaction.
Information
- Information needs are not well met, particularly for undiagnosed patients.
- Most information comes from specialists, GPs and other health professionals provide little input.
- Even after diagnosis, many patients and families must search for information themselves.
- Patient organisations and charities are a useful and more easily accessible source of further information.

Coordination of care
- Majority of patients have several health professionals involved in their treatment and attend multiple locations to receive care.
- Travel duration can sometimes be lengthy and may even involve travel outside of Wales.
- Only a small percentage of patients have a care coordinator yet the majority feel they would benefit from one.

Access to services
- Access to services, treatments and support remains a major problem area.
- There is difficulty accessing care from allied health professionals\(^1\).
- Patients and families can find the process of applying for help and support problematic, particularly without a diagnosis.
- There appears to be a disparity between paediatric and adult services, with paediatric services being more efficient.

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1. Eg. Physiotherapist, speech and language therapist, occupational therapist
Research

Key Findings:

1. Patients and families do not feel that enough research is being conducted and want more opportunities to participate.
2. Patients are willing to participate in research, even if it requires travelling.
3. Patients and families feel they are not kept well informed about research being conducted.

Key Finding 1: Patients and families do not feel that enough research is being conducted and want more opportunities to participate.

Research is an important component of healthcare. Through research, medical professionals are not only able to make advances in diagnostic tools and treatments, but are able to deepen their knowledge and understanding of diseases. This is particularly important in the context of rare diseases. Rare diseases often require the development of novel interventions which can only be developed through adequately funded research.

For patients and families, research provides hope yet only 17% felt that enough research was being conducted into their condition. Many state that funding is one of the main obstacles to research.

“As we say, my condition doesn’t have the ‘ah’ factor and so obtaining funding for research is difficult.”

Patient with Charcot Marie Tooth disease

“It’s down to us Myasthenics that have to raise the funds for the research, so it’s unlikely they will find a cure in my lifetime.”

Patient with MuSk Seronegative

It is also often the case that patients and families are unaware of how research is conducted and the ways in which to participate. Only 36% were aware of the existence of a registry for their rare disease. Of those for whom a registry does not exist or who are unaware of one, 82% would support the creation of one. The remaining 18% stated “Don’t know” suggesting perhaps a lack of understanding as to what a registry is or could provide in terms of research.

Only 14% of patients and families felt that there were enough opportunities for research.

“When any questions regarding research are put to the neurologists, the corresponding answers are always negative.”

Parent of patient with Ataxia Telangiectasia

2. In the survey, ‘patient registry’ was defined as a database that collects clinical data on patients with a specific disease (or type of disease).
**Key Finding 2: Patients are willing to participate in research, even if it requires travelling.**

Approximately 40% of respondents had already participated in research with biobanking (62%) being the most common form of participation. Other forms of participation included participating in clinical trial e.g. drug trials (21%) and as a patient representative (24%) e.g. on the ethics board for a trial.

“I am currently participating in a clinical research study re. Type 2 diabetes.”

Patient with Guillain-Barré syndrome

“Wales does not have a specialist Duchenne Muscular Dystrophy centre, consequently there are no major research projects going on here e.g. drug trials. When Seth is old enough to consider participation we will have to travel.”

Parent of patient with Duchenne muscular dystrophy

In terms of participation in research, many patients and families are willing to travel if they are able to do so.

“I know there is research going on in the USA and I would like to know if any is being done in the UK I would be happy to travel.”

Parent of patient with Angelman’s syndrome

Patients and families are often very committed and willing to participate in research, even if it will not necessarily improve their own situation.

“I would gladly take part in any studies on KBG Syndrome if it helps a family with a child with the condition and means they don’t have to feel as unsupported as I have.”

Parent of patient with KBG syndrome

**Key Finding 3: Patients and families feel they are not kept well informed about research being conducted.**

One of the biggest barriers patients and families face is that they often find it difficult to access information regarding research. 67% of respondents stated that they weren’t provided with enough information on research in general into their condition.

“My child’s paediatric consultant has limited knowledge regarding research opportunities. I am aware of other research through other parents whom I have met since my child was diagnosed.”

Parent of patient with Nephrotic syndrome

It is often up to the patients and carers themselves to find out about research and research opportunities.

“All the info I have received about my daughter’s condition has been what I have got myself via Facebook or research on the internet.”

Parent of patient with Episodic ataxia type 2

Luckily, some patient organisations provide their members with information however this is not enough considering not all patients and families belong to support groups.

“The only information received is obtained via Ataxia UK publications and conferences.”

Patient with Cerebellar ataxia SCA6
Diagnosis

Key Findings:
1. Patients and families sometimes wait a very long time to receive a correct diagnosis.
2. Patients often receive incorrect diagnoses before arriving to a final correct diagnosis.
3. Patients must attend multiple appointments before receiving a final diagnosis.
4. Majority of patients were diagnosed in Wales but many are willing to travel for specialised services.
5. Ratings for ‘health service received’ show varying degrees of satisfaction.

Key Finding 1: Patients and families sometimes wait a very long time to receive a correct diagnosis.

Encouragingly, the majority of patients received a correct diagnosis within one or two years, with 26% being diagnosed within three months. However, 29% of patients waited over two years, with an alarming majority of them waiting five years or more.

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

“I was told the name of my rare condition at a private appointment when I was 50 years old!!”

Patient with Poland’s syndrome
“Blood tests took an inordinate amount of time to complete. I feel that if results could be obtained somewhat quicker it would be less stressful for all concerned.”  
Parent of patient with Ataxia-Telangiectasia

“It’s very frustrating that in Wales, only one genetic test at a time will be done. I appreciate that the costs involved are high, but it is hard to wait on all these tests to come back.”  
Parent of patient with Episodic ataxia type 2

Waiting for a diagnosis can be a very stressful time for patients and families. The uncertainty and lack of information can be a source of frustration and despair.

“Things improved considerably once the diagnosis was reached; until that point I felt anxious, confused and unsupported.”  
Patient with Fabry’s disease

“Despair is how we feel, we can’t move forward at all until a diagnosis is made. I’m still blaming myself and I’m still finding difficulty getting my head around our unique situation.”  
Parent of four undiagnosed children

“I think the hospital consultants managing my condition didn’t appreciate the time it would take (~8 months) before I was seen by them, or the time it would then take for the genetic testing to be completed. This left me confused about how serious my condition was and whether I would need further treatment, and I was then rushed into making a decision about the type of treatment I would have, and without having receiving the results of the genetic testing.”  
Patient with Familial adenomatour polyposis

Delay in diagnosis can also have dramatic consequences for patients.

“My limbs are paralysed and I need fulltime care. This could have been avoided by prompt diagnosis.”  
Patient with Churg-Strauss syndrome

“Better information on possible diagnosis/treatments while I waited for appointment with the genetics service would have enabled my GP to give me better advice (he underestimated the severity of the condition, and failed to chase up a repeat endoscopy that was delayed by 6 months).”  
Patient with Familial adenomatour polyposis

Some patients remain undiagnosed and have been informed they may never receive a definitive diagnosis. Of those undiagnosed, 85% have been suffering from symptoms for over a year, with a staggering 54% suffering from symptoms for over five years. Lack of diagnosis not only means that patients and families receive less information, it also makes accessing services and support extremely problematic.

“Because he wasn’t diagnosed until just before he was fifteen and because they didn’t know what it was he was pretty much signed off by everyone other than the genetics team who saw him every two years. I felt very unsupported and provided all of his care myself.”  
Parent of patient with KBG syndrome
Key Finding 2: Patients often receive incorrect diagnoses before arriving to a final correct diagnosis.

36% of respondents received incorrect diagnoses with some receiving as many as five or more. This is an alarming statistic as incorrect diagnoses can have serious implications for treatment and cause delays in accessing appropriate care and support.

“I was told it could have been caused by a virus or possibly connected to the mastectomy operation I had for breast cancer a few weeks prior to onset.”

Patient with Guillain-Barré syndrome

“I was previously diagnosed with a bowel condition and was treated for it. But the treatment didn’t help the fault, I was actually diagnosed by accident.”

Patient with Common variable immunodeficiency (CVID)

“I still feel aggrieved that because the ‘usual’ medication for my condition didn’t work for me, it was assumed I didn’t have it. This meant I then had to keep visiting A&E and my GP (who was brilliant) without any help until I went privately.”

Patient with MuSk Seronegative

“It took years before we found out what type of Ichthyosis he had. Also at one time I was told that the problem was my fault but that was not right. This caused me great distress.”

Parent of patient with Lamellar ichthyosis

“I was given an incorrect diagnosis of ‘Sudden Death Syndrome’; this caused considerable stress and anxiety for well over a year.”

Patient with Fabry’s disease

Key Finding 3: Patients must attend multiple appointments before receiving a final diagnosis.

Because of the complexity of rare diseases, it is not uncommon for patients and families to have to attend several appointments with different medical professionals in order to receive a diagnosis. However, the survey revealed that patients sometimes had to attend an excessive amount of appointments before receiving a diagnosis.

<table>
<thead>
<tr>
<th>Appointment Type:</th>
<th>Number of appointments attended before final diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1-4</td>
</tr>
<tr>
<td>GP appointment</td>
<td>67%</td>
</tr>
<tr>
<td>Hospital Outpatient appointment</td>
<td>62%</td>
</tr>
<tr>
<td>Hospital Inpatient appointment</td>
<td>79%</td>
</tr>
</tbody>
</table>

Attending multiple appointments can be stressful for the patients and their families as they often feel frustrated and misunderstood.
“…such an upsetting time. Made worse by having to repeat the same info again and again even though it would be in all medical notes right in front of them.”

Parent of patient with Prader-Willi syndrome

This finding also highlights an ineffective use of resources. Multiple appointments do not only have cost implications for the patients in terms of time, money and energy, but also for the NHS and medical professionals. Patients themselves suggested that if their health professional listened more closely to their concerns they would not have to return to them so often.

“Went to GP, told him I thought I was suffering with this condition as my sister was diagnosed with it. He wasn’t interested!!”

Patient with ataxia

“I had come from abroad with medical records, yet was put through the whole testing procedure once more, just to arrive at the same conclusion after a full year. Using the available prior information and scheduling more frequent appointments should have led to a faster resolution.”

Patient with Albinism and tentative diagnosis of Folliculitis decalvans

Key Finding 4: Majority of patients were diagnosed in Wales but many are willing to travel for specialised services.

Only 23% of respondents had to travel outside of Wales to receive a diagnosis. This figure is encouraging as it demonstrates good service provision in Wales. However, patients did express a willingness to travel in order to access specialised services at both the level of diagnosis and for on-going care and treatment. Specialist services are often few and far between for patients affected by rare diseases. For many patients, particularly in areas close to the border with England, it may be preferable to access a service in England. Patients should therefore be assisted in attending specialist clinics when necessary and where available across the UK. Patients and carers expressed their concern that health services sometimes act as a barrier rather than facilitators in accessing services outside the patients’ locality.

“Only last week I was refused funding to see [a specialist in a specialist centre in London], he has just started a clinic for retroperitoneal fibrosis.”

Patient with an undiagnosed condition

“[The NHS should] ...allow us to travel out of area for a diagnosis (we had to pay privately), and accept that they didn’t know exactly what was wrong.”

Parent of patient with Ehlers-Danlos syndrome type 3

“I have a very good consultant but unfortunately he is often in other parts of the country or in other countries and so he is difficult to contact.”

Patient with Greig cephalopolysyndactyly syndrome

The key is to promote co-ordination of services and communication between different service providers.

“Social services in Ceredigion work very well and closely with NHS and we are grateful for this.”

Parent of an undiagnosed child
Key Finding 5: Ratings for ‘health service received’ show varying degrees of satisfaction.

Overall, how would you rate the service provided by the health service during the period of trying to obtain a diagnosis?

When asked to rate health services received during diagnosis, majority of respondents selected “Good” and “Excellent”, which is an encouraging finding. Nonetheless, with 44% still rating services as “Average” or lower (i.e. “Poor” and “Very Poor”), the results suggest there is still room for improvement.

“I have been treated, if not bullied by the medical profession in lots of areas, who did not know about the condition.”

Patient with Greig cephalopolysyndactyly syndrome

Suggested areas for improvement included:

- Reducing waiting time between appointments
- Providing more information about testing process and diagnosis
- Support and input from specialist health visitor
- Counselling

For many patients improvement of services relies simply on more patience, understanding and openness on the medical professionals’ behalf.

“[Doctors should have] ...been less evasive, and been honest about not being sure what was wrong with me!”

Patient with Fabry’s disease

“Give hope and not as some doctors said, no hope.”

Patient with sarcoidosis

“Be more understanding and listen more.”

Parent of patient with global developmental delay

“Doctors listening to a patient and not deciding that unfamiliar symptoms must be imaginary.”

Patient with Churg-Strauss syndrome
Sadly, some patient’s parents reported very upsetting experiences.

“We also saw the geneticist and the genetic counsellor who was awful if I’d have been a weaker person I would have felt like ending it there and then!”

Parent of patient with Angelman’s syndrome

“It was appalling, given the news in a corridor that our child would be blind within a few years, no further information given at that time.”

Parent of patient with Laurence Moon Bardet Biedl syndrome

It is also important to note that 47% of respondents rated health services received since diagnosis as “Average” or lower. One of the problem areas identified is lack of or poor follow-up.

“I cannot get an annual appointment with a neurologist. I was given just four sessions with a physiotherapist even though my condition is progressive. Orthotics are issued but there are no follow up appointments to see how I’m coping.”

Patient with Charcot Marie Tooth disease

“Lack of sharing of information between professionals seems to be hindering any follow up services.”

Patient with Cerebellar ataxia

“We need a specialist to support the family after diagnosis.”

Carer of a patient with a rare chromosome disorder

Issues with coordination of care are addressed in a later chapter.
Alvina’s story

Alvina Jones, mother of four children suffering from undiagnosed epilepsy syndromes – Carmarthenshire.

People with epilepsy suffer from recurring seizures. There are different types of seizures which can affect people in different ways. The effects can be as unnoticeable as twitching but can also involve loss of consciousness and convulsions.

Alvina is the mother of five children, four of whom suffer from undiagnosed epilepsy syndromes. Along with their complex epilepsy, her children have severe to profound learning difficulties and suffer from physical disabilities such as low muscle tone, bilateral feet and gait issues. Two of her children have been diagnosed with autism, another is undergoing testing for it, and all suffer from behavioural issues, sensory processing problems and speech and language problems. Needless to say, life for the Jones family isn’t easy.

The children’s issues appeared at a young age, with most of them suffering from seizures from as young as nine months old. Alvina recalls a traumatizing incident when they nearly lost their son Joe whose seizure went on for over 90 minutes. “If it hadn’t been for the local GP at the time, Joe wouldn’t be here now.”

Because of the complexity of her children’s condition, as infants they were seen by a neurologist every 3-4 months. Unfortunately, it has got to a stage now where the children’s consultant has informed Alvina that he is unable to provide her with any more answers. “We have no prognosis at all”. Not one of the Jones’ children has a definitive diagnosis which encompasses all of the aspects of their disabilities and this has caused great problems. This has been particularly evident in the areas of education and social care.

Alvina has had to fight, sometimes all the way up to tribunal, in order to arrange provisions for her children. “It’s all about ticking the boxes, you want provision for ASD [Autism Spectrum Disorder], you get that provision and that is it. Joe’s epilepsy can be forgotten, it can be totally forgotten and that’s not my only issue with where Joe is with. Even though it says in his statement that he needs a multidisciplinary team, I don’t see that they’re even working in liaison with each other.”

Unfortunately coming up against barriers doesn’t just occur when dealing with educational services. “Well really it’s the whole lot, the whole lot. Because even in social services terms, the consultant paediatrician requested that we needed more support within the home now and even that has failed.” Social services assess each child individually and do not take into account the family situation as a whole. Alvina and her husband have therefore had to fund all respite and extra care themselves. They are finally moving into a new bungalow without stairs but this is a result of an eight year battle. “Parents have got to be so, so strong and wanting to fight. It’s a daily struggle for me and every day I think: oh what letter’s going to come through the post now. We don’t come off the rollercoaster.”

Fortunately, it’s not all doom and gloom. Alvina describes her GP as very supportive and has a huge admiration for what the consultant has done for them. He has followed her children’s development from when they were babies, which for her second eldest child means 12 years. She has also received a lot of support from several charities who have not only provided her with useful information and advice but also given her an opportunity to share and connect with others. “They have been a lifeline for the last 14 months. They provided me with free anti-suffocation pillows for the children3, they allowed me to speak freely within their forum and that is a huge thing for me, you know, to not be so isolated and being able to speak up.”

It is difficult for Alvina and her family to anticipate the future. With no prognosis and no doctor able to provide her with any information about what to expect, Alvina has many fears regarding her children’s care. She describes the experience of having to be the go-between between health and education as one that makes her melt. “I don’t want this to be a battle, I want them to support us. I want them to see it from my eye view, not from them saying: This fits this box and this fits that box. They need to see it from that view that we do not fit the box. When you’ve got a condition that no one knows about then you do not fit the boxes, so they need to work around the difficulties and be able to give targeted support in a way that that needs to be.”

3. Anti-suffocation pillows provided by Epilepsy Sucks UK.
Key Findings:

1. Information needs are not well met, particularly for undiagnosed patients.
2. Most information comes from specialists, GPs and other health professionals provide little input.
3. Even after diagnosis, many patients and families must search for information themselves.
4. Patient organisations and charities are a useful and more easily accessible source of further information.

Key Finding 1: Information needs are not well met, particularly for undiagnosed patients.

When asked whether sufficient information was provided about the condition at the time of diagnosis or shortly after, 46% responded “No”. Majority of patients (61%) felt they either didn’t receive enough information or were unsure about it.

“As I was only 15 at the time I feel like it could have been of benefit to me if they had informed me better of the tests they were doing and what the outcome of them meant.”

Patient with Panhypopituitarism

Patients expressed that the time around diagnosis can be very stressful and this may hinder their ability to effectively communicate their concerns. It is therefore important that health professionals provide adequate amounts of information without overwhelming the patient and also give them the opportunity to ask questions at a later stage if necessary.

“Provide more written information to take home with us, explaining about the disease and what effect it might have on the person, because when you first receive the diagnosis you are in a state of shock and are unable to think of the right questions to ask.”

Relative/carer of patient with WAGR syndrome

“Additional information is needed sooner after diagnosis, including information on support groups and financial assistance. People have enough to worry about, without having to chase up benefits and support.”

Parent of patient with Wegener’s granulomatosis

When asked whether sufficient information was provided about further sources of information and support at the time of diagnosis or shortly after, only 28% responded “Yes”.

“We should be provided with details of associations and community support.”

Patient with Neurofibromatosis 1

“Any information would be good. As I said I have had no information from the Health Services.”

Patient with Charcot Marie Tooth disease
Please specify how much your/the person with a rare disease’s information needs are met in the following areas:

<table>
<thead>
<tr>
<th>Area of Information</th>
<th>Enough information received</th>
<th>More information needed</th>
<th>No information received</th>
<th>Information not needed/not relevant</th>
<th>Don’t know</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ongoing management of the condition e.g. managing or reducing symptoms</td>
<td>39%</td>
<td>37%</td>
<td>22%</td>
<td>0%</td>
<td>1%</td>
</tr>
<tr>
<td>Treatment</td>
<td>44%</td>
<td>36%</td>
<td>11%</td>
<td>4%</td>
<td>4%</td>
</tr>
<tr>
<td>What to expect from the condition</td>
<td>39%</td>
<td>36%</td>
<td>21%</td>
<td>1%</td>
<td>3%</td>
</tr>
<tr>
<td>Information needed by carers/family</td>
<td>31%</td>
<td>29%</td>
<td>28%</td>
<td>6%</td>
<td>7%</td>
</tr>
<tr>
<td>Education</td>
<td>21%</td>
<td>32%</td>
<td>25%</td>
<td>17%</td>
<td>6%</td>
</tr>
<tr>
<td>Welfare and benefits</td>
<td>15%</td>
<td>31%</td>
<td>35%</td>
<td>18%</td>
<td>1%</td>
</tr>
<tr>
<td>Palliative (end of life) care</td>
<td>3%</td>
<td>8%</td>
<td>19%</td>
<td>53%</td>
<td>17%</td>
</tr>
<tr>
<td>Social information e.g. respite care</td>
<td>6%</td>
<td>18%</td>
<td>29%</td>
<td>38%</td>
<td>10%</td>
</tr>
<tr>
<td>Sources of information and support e.g. charities/patient organisations</td>
<td>28%</td>
<td>33%</td>
<td>29%</td>
<td>4%</td>
<td>6%</td>
</tr>
</tbody>
</table>

“No information has been given on financial issues. My husband has had to give up work, I have found it very difficult to manage working full time, caring for my husband and trying to apply for various benefits.”

Carer of patient with Wegener’s granulomatosis

When looking at whether information needs overall were met since receiving a diagnosis, 42% of respondents stated that their information needs overall were either “not well met” or “not met at all”. Only 33% found their information needs overall met “completely” or “fairly well” with the remaining 25% having “some of their information needs met and others not”.

“Because this syndrome is still very much under study, and so little is known about it, I’m sure there are more answers to come our way. I understand this might be because of the nature of the condition, but it does make it a little hard for us because so much is still so unknown.”

Parent of patient with KBG syndrome

“Any support following diagnosis would have been helpful.”

Parent of patient with muscular dystrophy

Information needs seem to be least well met for those that are undiagnosed or only have a partial diagnosis. Of the eleven respondents that were either undiagnosed or only had a partial diagnosis, less than 20% of respondents selected “Enough information received” in seven of the nine information areas. Majority (over 50%) selected “More information needed” or “No information received”. Only in the areas of “Palliative (end of life) care” and “Social information e.g. respite care” did over 60% select “Information not needed/not relevant”.

4. See above table for details of information areas
Key Finding 2: Most information comes from specialists, GPs and other health professionals provide little input.

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

 Patients and carers expressed difficulty in obtaining information from health professionals. 43% do not have someone they can approach easily to answer questions about their condition, care and treatment. Those who did have someone mainly stated their consultant/specialist. Only 8% selected their GP. This finding is significant because GPs are usually the first point of contact.

“In general I think GP’s and allied health professionals need more training about such conditions and how to spot them early on.”

Parent of patient with muscular dystrophy

“More information about lesser known neurological conditions is needed. There is a lack of training/education of the medical profession at all levels of health care.”

Patient with Guillain-Barré syndrome

“I know I am extremely lucky to be seeing an expert in my condition and I can email her. However, due to the proposed changes in Wales NHS which will affect neurological services, I’m finding the future to be a bit of a grey area at present.”

Patient with MuSk Seronegative

“There should be specialist nurse at the health centre who is there for us and to answer our questions.”

Patient with an undiagnosed condition

Patients and families affected with rare disease can often feel discouraged, particularly when their primary care physician is unable to provide them with any additional information or support. Health care professionals should be encouraged to consult with other professionals and specialists, in order to be able to provide patients with expert information that they could not necessarily obtain on their own.
“A lot of information regarding my son’s condition has been obtained by researching for it myself. My son’s consultant does not profess to be an expert and when I ask him questions which he is unable to answer he consults a nephrologist at Alder Hey Children’s hospital in Liverpool.”

Parent of patient with Nephrotic syndrome

**Key Finding 3: Even after diagnosis, many patients and families must search for information themselves.**

Patients with rare disease must often rely on the internet or even “word of mouth” as sources of information.

“I inherited the illness and was treated like a child after onset of symptoms, although I was 14. Thank god for the invention of google!”

Patient with cerebellar ataxia SCA11

“As condition is rare, it’s hard for professionals to help that much. I have got most of my info from Facebook Support sites.”

Parent of patient with Episodic ataxia type 2

31% of patients had to search for information themselves. This statistic is a slight cause for concern as not all open sources are reliable and accurate. Carers also find the burden of having to search for information themselves very disheartening.

“You have to be very positive and pro-active to get anywhere...can be very upsetting and demoralising. you have to get to a certain state of bravery to even begin to ask for info, help etc...”

Parent of patient with Prader-Willi syndrome

**Key Finding 4: Patient organisations and charities are a useful and more easily accessible source of further information.**

Patient organisations and charities are able to provide patients and families with information and support that other healthcare professionals cannot. Patients and families often feel that GPs lack the knowledge and education required for their particular condition.

“All my information comes from the charity set up to support patients with my condition. I have received no information from the NHS.”

Patient with Charcot Marie Tooth disease

Patient organisations and charities have the advantage of usually focussing on one particular condition or set of conditions, allowing them to amass more specialist and in-depth knowledge about even rarer conditions.

“I am fortunate that I am a Clinic Support & Committee member of LMBBS so have all the necessary information to hand.”

Parent of patient with Laurence Moon Bardet Biedl syndrome

These organisations also often address the non-medical needs of patients and families, which is an important part of treatment.
“I am a Trustee of CMT United Kingdom, as a result I receive information and pass it on to others; the NHS gives no information at all, and is, in the main, poorly informed, although we are trying to educate them!”

**Patient with Charcot Marie Tooth disease**

They are able to inform patients about their rights and help them access the services they are entitled to.

“Information on management and treatment is mostly provided by Ataxia UK and patient support groups. Hospital appointments are so infrequent (and brief!), that no information about the condition is received.”

**Patient with cerebellar ataxia SCA6**

“Professionals should advise patients of support organisations that could help support new patients.”

**Patient with thyroid cancer**
Christine’s Story

Christine Humphreys – Mother of James who is affected by Laurence Moon Bardet Biedl Syndrome (LMBBS) – Newport.

LMBBS is a genetic condition in which the primary symptoms are visual impairment, obesity, extra fingers and/or toes, kidney abnormalities and learning difficulties. There is currently no cure and the visual impairment is inevitable, however some of the symptoms can be effectively treated if addressed early.

Having been foster parents for many years, James came into our lives on 26th January 1982, aged three weeks, wearing plaster casts for talipes\(^5\) and with an extra digit on his left foot.

The next 10 years were full of sleepless nights, tantrums, rapid weight gain, and surgery on both legs, including removal of the extra digit. Decaying milk teeth resulting in extractions, ear and chest infections, and reactions to medication and anaesthetics. An ophthalmologist finally agreed that James had a ‘turn in his eye’ which had been queried since six months, and was extremely short sighted.

Aged 11 years, James’ health began to deteriorate. He was exhibiting ME like symptoms, falling asleep in the classroom, developing very high temperatures and vomiting, and during this period, he would have a weight gain of anything up to 7lbs in 2/3 days. James also began bumping into doors and complaining that he couldn’t see in certain lights, sunlight caused immense problems. We had been discharged from ophthalmology three years previously and a new ophthalmic check-up revealed nothing. James’ education was in main stream school with full support, moving to a special needs unit attached to a comprehensive school, again with full support.

In July 1991, we received a phone call from social services (we had adopted James five years earlier) to say that James had a natural sibling who was believed to have a life threatening illness and that all siblings needed to be examined, particularly their sight. In October my husband was given the news standing in a corridor, having gone for an appointment for the results and told there was no one present to give them. 30 minutes later after giving an ultimatum, a Registrar appeared and said that, yes, James had a hereditary disease and would be blind by the end of his teens; further tests would need to be completed. No additional help was given!

With no more news, I searched the web and contacted the RP Society\(^6\), who put me in touch with the Laurence-Moon-Bardet-Biedl-Society (LMBBS). Initial euphoria that we could have a diagnosis soon led to desolation about how we would all cope with James becoming ‘blind’. Shortly afterwards, as a family we attended our first conference. On our return, I presented all the facts to a psychologist and a paediatrician who agreed that it could be LMBBS and referred us to a geneticist.

Life then changed upon diagnosis, with visits to different hospitals. Great Ormond Street Hospital became our home. James hated it, he didn’t want to be different from his friends.

We had immense support from the LMBBS Society and I became and still am, a member of the committee with my husband as Chairman. The Society has grown over the years, but for James, the annual family conference is like going home and meeting extended family. There he becomes confident, talking to anyone who is prepared to talk with him. Yes his sight has deteriorated, he is registered as blind, but it has been gradual and he has coped and is a credit to himself and to us as a family. He is an ‘adrenalin junkie’ who likes nothing more than hanging upside down on theme park rides or tandem jumping out of aeroplanes to raise much needed funds for his charity.

LMBBS Society, 16 years on has grown, numbers have increased and 2010 saw the start of designated LMBBS Clinics in four hospitals in the UK providing patients with an opportunity to be seen annually by a team of specialists in a morning or afternoon ‘One Bus Stop Clinic’. For further information on LMBBS: www.lmbbs.org or e-mail: chris.humphreys4@ntlworld.com.
Coordination of Care

**Key Findings:**
1. Majority of patients have several health professionals involved in their treatment and attend multiple locations to receive care.
2. Travel duration can sometimes be lengthy and may even involve travel outside of Wales.
3. Only a small percentage of patients have a care coordinator yet the majority feel they would benefit from one.

**Key Finding 1: Majority of patients have several health professionals involved in their treatment and attend multiple locations to receive care.**

67% of respondents stated that they or the person with the rare disease had three or more health professionals involved in their care, with 30% stating five or more.

**How many health professionals across the range of disciplines are involved in your/ the person with a rare disease’s routine care and treatment?**

(For example, GP, community nurse, specialist nurse, hospital consultant(s) and allied health professionals such as physiotherapists, speech and language therapists, dieticians etc).

![Bar chart showing the distribution of the number of health professionals involved in care, with 28.0% respondents stating 1-2 professionals, 30.7% stating 3-4, 18.7% stating 5-6, 8.0% stating 7-8, 4.0% stating 9-10, 5.3% stating more than 10, and 1.3% not knowing.]

52.7% of respondents receive care in three or more different locations. This includes clinics, hospitals and centres.

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7. In the survey, health professionals was defined as including allied health professionals such as physiotherapists, speech and language therapists, dieticians, etc.
In how many different locations (e.g. at home, hospitals, clinics, centres) is care/treatment provided for you/the person with the rare disease?

![Bar chart showing the distribution of locations provided care/treatment]

- 0 locations: 6.7%
- 1 location: 16.0%
- 2 locations: 22.7%
- 3 locations: 30.7%
- 4 locations: 10.7%
- 5 locations: 6.7%
- More than 5 locations: 5.3%
- Don’t know: 1.3%

Having to attend different locations can be very problematic for some patients as they may be affected by mobility issues. The fragmentation of services can also add to the stress of the experience and can interfere with other commitments.

“Would be great to have a one stop shop for Turner Syndrome where all necessary checks are done to save having to take time off work and travel to all the different health professionals.”

Patient with Turner syndrome

**Key Finding 2: Travel duration can sometimes be lengthy and may even involve travel outside of Wales.**

Although the majority of respondents (68%) travel less than two hours to attend appointments, nearly 10% have journeys that last more than five hours.

What is the approximate travel time (round trip) to the furthest clinic/hospital that you/the person affected by a rare disease attendes?

![Line graph showing the distribution of travel times]

- Less than 30 mins: 21.3%
- 30 mins - 1 hour: 8.0%
- 1-2 hours: 5.3%
- 2-3 hours: 9.3%
- 3-4 hours: 4.0%
- 4-5 hours: 2.0%
- More than 5 hours: 1.3%
32% of respondents already travel outside of Wales for elements of their care or treatment. However, over 50% would be willing to travel in order to receive specialist care. This finding suggests that patients and families are realistic in their expectations of the provision of services for rare diseases.

“As with all rare conditions, there are only so many ‘experts’ in the field. Therefore it follows that on occasions it might be necessary to travel to see a particular individual.”

Patient with Fabry’s disease

“I accept that there can’t always be a specialist locally but would like the majority of the care to be done locally even if consultant appointments need to be in London.”

Parent of patient with Ehlers-Danlos syndrome type 3

“I do find it difficult to travel outside Wales but without my consultant in England I would still be waiting for any clues in our condition.”

Patient with Greig cephalopolysyndactyly

Of those unwilling to travel, the majority stated mobility and financial issues as the cause.

“The condition is progressive; I am now 70 and the symptoms are worsening gradually, if an operation should become necessary I should, of course, be prepared to travel outside of Wales, but I would prefer not to.”

Patient with Charcot Marie Tooth disease

But for some, travel can be very problematic and although it means access to specialist treatment, it can also make the experience very unpleasant for patients, particularly children.

“Initially we were referred to Great Ormond Street Hospital which was incredibly difficult as the children don’t like travelling and by the time we arrived at our appointment the children would be agitated and uncooperative.”

Parent of patients with Smith-Lemli-Opitz syndrome

**Key Finding 3: Only a small percentage of patients have a care coordinator yet the majority feel they would benefit from one.**

Only 33% of respondents stated that they have a care coordinator.

“What co-ordination?? Thank goodness I was already very organised!”

Parent of a patient with Prader-Willi syndrome

Of the 54% that do not have a care coordinator, the majority (73%) stated they felt they would benefit from having one.

“I think that someone with a good working knowledge of both health issues and social care issues would be very useful.”

Parent of patient with muscular dystrophy

“Badly coordinated for adults in particular - my son with the condition, a child has much better and more care then I, who have practically non-existent input and have to fight for everything.”

Patient and parent of patient with Ehlers-Danlos syndrome type 3
“Coordination of services is left to us as parents, the children are seen in 3 different counties and very often our GP is not included in correspondence.”

Parent of patients with Smith-Lemli-Opitz syndrome

It is often the case that parents have to become experts in their children’s conditions and must also manage the coordination of services.

“I soon discovered that if I didn’t co-ordinate the 20+ different elements of my sons care, that it simply would not occur and his care would suffer in a major way.”

Parent of patient with Wolf Hirschhorn syndrome
‘Ataxia’ means ‘lack of order’. People with ataxia have problems with movement, balance, and speech. It is a degenerative disease and in my case, the degeneration is very slow. There is currently no cure for this life-limiting neurological condition.

From an early age, I have always walked with a wobble and sounded a bit slurred. Many people just thought “Oh that’s just Alan!” I achieved good qualifications at school and went on to run my own electrical contracting business. But the condition progressed very slowly and lack of co-ordination along with electrical circuitry does not mix!

I managed to carry on, until the day when... making sandwiches for work, I found that buttering bread seemed to be taking more and more concentration. “A simple task, you take for granted”, that’s when I thought about looking into things hoping for a simple answer. How wrong was I?

I went to my GP, who on numerous occasions simply told me to take time off work and rest. This went on for many years. On one visit, my usual GP was not available so I saw a locum. He had recent knowledge of similar conditions/symptoms and asked for neurological tests. He also referred me to a general neurologist where I was diagnosed with cerabellar ataxia. I was told the diagnosis, the probable outcome, that there was nothing that could be done, and “that was that!”

When I returned to my GP, he said I had a rare condition and was unlikely to meet many others with this rare disease. I was told of the national charity Ataxia UK, which was my only point of contact at the time.

So upon returning home, I tried researching ataxia via the internet but at the time, there wasn’t a large amount of consistent information available (along with some good and bad stories).

Since then I have become a Trustee of Ataxia UK, chairman of Ataxia South Wales and also run a global online support network for patients with ataxia8 thus being able to connect with many other sufferers of this neurological condition. I am determined that information should be available to patients with all rare diseases and that support networks are vital so that feelings of isolation do not add to the patients’ worries.

I see general neurologists in a local hospital, but their knowledge of Ataxia is very limited and I often find myself having to tell them about it! My specialist ataxia clinic is at the Royal Hallamshire Hospital in Sheffield. This requires a journey of 257.84 miles, nearly 5 hours by train. Although I am very satisfied knowing that my condition is being monitored, I find that the excellent services provided at this clinic are what is lacking in Wales.

Something I am also keen to see change is the “uniformity” of services available to patients with a neurological condition. I am also aware that many of the services offered to younger patients seem to cease when they transfer into adulthood, which is a very worrying situation.

8. www.livingwithataxia.org
Key Findings:

1. Access to services, treatments and support remains a major problem area.
2. There is difficulty accessing care from allied health professionals.
3. Patients and families can find the process of applying for help and support problematic, particularly without a diagnosis.
4. There appears to be a disparity between paediatric and adult services, with paediatric services being more efficient.

Key Finding 1: Access to services, treatments and support remains a major problem area.

One quarter of patients were either unable to access medication/drugs or experienced difficulty doing so. Only 43% of patients were easily able to access a medical specialist in Wales, with 14% either experiencing a lot of difficulty or being unable to access. 35% experienced difficulty or were not able to access a medical specialist in Wales.

Access to education support is poor with many parents having to fight to organise provisions for their children.

“All the info I have had about IEPs (Individualised Education Programs), Annual Reviews, Statements etc I have had to find out about myself, and I am still not sure of everything. I find the knowledge the schools have (in my experience) about SEN is very limited.”

Parent of patient with Episodic ataxia type 2

Only 18% of patients are able to access benefits/financial support with ease.

“Applied for incapacity benefit but was unsuccessful. I felt that there was no appreciation of the side effects and on-going difficulty with this condition. I felt that the attitude was ‘You had a headache, you had some aspirin and now you must be better’. No sympathy for the physical restrictions of this condition.”

Patient with thyroid cancer

“So far, as my disability has progressed, I have arranged home adaptations myself. This also applies to any walking aids I need. I am fortunate to have a supportive partner who is happy to finance such requirements!”

Patient with cerebellar ataxia SCA6

“Dealing with social services to get respite care has been incredibly difficult, we have litigated twice.”

Parent of patients with Smith-Lemli-Opitz syndrome
Key Finding 2: There is difficulty accessing care from allied health professionals. 42% were unable to access services from allied health professionals or experienced difficulty in doing so.

“I had an excellent O.T (Occupational Therapist) but because of the lack of information on the condition I am no longer able to see them without a new referral.”

Patient with cephalopolysyndactyly syndrome

“I am not aware of any treatment apart from physio(therapy) or hydrotherapy and have been told by the physiotherapy dept at my local hospital that there was no point in them referring me for hydrotherapy as I would need two therapists and this was not possible as there is not enough staff.”

Patient with Guillain-Barré syndrome

Allied health professionals play a vital role in the treatment and care of patients with rare diseases. Many patients suffer from complex symptoms which need to be managed under the care of specialists such as occupational therapists, speech and language therapists, etc.

“Despite problems with slurring speech etc and difficulties with balance, I have only ever been offered different types of immunosuppressant or steroids. The possibility of other therapists such as a speech therapist, has never been offered or spoken about. As it happens, I am not able to take any extra medication due to serious side effects.”

Patient with MuSk Sero Negative

“As far as we know there are no services available apart from speech therapy and physio (therapy) and these are very difficult to be referred to on a regular basis. Treatment is quite sporadic.”

Parent of a patient with Ataxia-Telangiectasia

It is important that allied health services be appropriate and correctly address the needs of the patient. Survey responses seem to demonstrate a lack of adequate provision in Wales.

“I really need physiotherapy as my limbs are so badly affected. When my London hospital suggested physiotherapy I was sent to a group here in Wales. I spent 80% of my time sitting in my wheelchair, very few actual exercises but expected to play elementary word-games (I’m a writer and also proficient in IT). Very boring and depressing, but apparently that’s what’s on offer.”

Patient with Churg-Strauss syndrome

“Wheelchair services are poor. I have been a wheelchair user since 1984 and this year after being assessed by a neurological specialist physiotherapist for the very first time (I did not know they existed), I received the very first manual chair and power chair that actually ‘fitted’ me and are actually comfortable. The previous chair had affected my posture and my left shoulder, pelvis and hip are out of alignment because of this. Sadly after she had sorted me out she transferred to Cardiff!”

Patient with Guillain-Barré syndrome
Key Finding 3: Patients and families can find the process of applying for help and support problematic, particularly without a diagnosis.

Patients and carers encounter many barriers when trying to access services. Many describe the process as a battle and often find the experience confusing and disheartening.

“Nearly everything that I have managed to get for my son has had to be fought for. I really feel that parents who are less articulate or haven’t access to computers etc would have an even more difficult job.”

Parent of patient with Ehlers-Danlos syndrome type 3

“As I do voluntary work with the Occupational Therapy Department, I find it easier than most to access things as I know the ropes. Before I was involved, I found the whole process very confusing and hard to understand.”

Patient with Pseudoachondraplasia

“My local council has just refused to renew my Blue card, even though my condition is progressive and I am worse now than I was three years ago.”

Patient with Charcot Marie Tooth disease

Patients and carers often feel there is a lack of support in dealing with non-medical issues. Though educational and social needs fall outside the remit of health care professionals, many patients feel that the agencies responsible for these lack understanding of their situations. It appears that very little is shared with patients as to what they are entitled to and how to go about arranging for service provision.

“It feels that the levels of support we received in play group were wonderful, but after that we have had to pretty much get on with it on our own where the education and medical services were concerned. The only time people became interested in him was when he was diagnosed with KBG syndrome, then everyone wanted to see him, but to be honest, and this is not disrespect to the genetic service, because I know how hard it was for them to diagnose this condition, but it all came fifteen years too late for us. When I had been in the position of having to put in all the hard work on my own and with no support from the medical services, education services or social services.”

Parent of patient with KBG syndrome

Key Finding 4: There appears to be a disparity between paediatric and adult services, with paediatric services being more efficient.

Only 25% of those that transitioned from paediatric to adult services reported experiencing “no problems”. It appears that many feel a lack of support during the transition period. Carers suggest that health services should recognise that there is a disparity between paediatric and adult services.

“More transition info between children and adult services is essential and given at earlier age, so that families have time to prepare the young person for the changes in services. There are differences in services when you are a child compared to the services available in adult services.”

Carer of patient with centronuclear myopathy
“No information was given to me /parents about my condition and we were not given any information on the severity of our daughter’s condition until last month.”

Patient and parent of patient with Greig cephalopolysyndactyly

“We had no assistance with transition changes.”

Parent of patient with Laurence Moon Bardet Biedl syndrome

“No awareness of the changing needs and how change in situation and circumstance affects patient.”

Relative/carer of an undiagnosed patient

Some parents reported feeling worried about the transition from children to adult services.

“My son is with paediatric services right now, when we move to adult services I have no idea what to expect. His paediatrician coordinates everything at the moment.”

Parent of an undiagnosed patient

For those who had a good experience with children’s services, there are concerns about quality of care dropping once their child transfers to adult services.

“Social services and NHS have been very supportive, but again I don’t know what to expect when we move from children to adult services.”

Parent of an undiagnosed patient

“The paediatric service is much better than the adult one and I am extremely worried as my son is now 14 that he will end up without the care he needs after he goes into adult services.”

Parent of patient with Ehlers-Danlos syndrome type 3

Those that had poor experiences accessing paediatric services are concerned that this will only become more problematic as their child becomes older.

“Children’s health services are adequate, but could be better with more funding available so more staff could be employed. There are long waiting times to access services. Transition from children’s services to adult services could be improved. Currently, the service provided is very poor and not person orientated.”

Parent of patient with centronuclear myopathy
Laura’s Story

Laura Crawford, MBE – Patient with Pseudoachondrplasia – Conwy.

Pseudoachondrplasia is an inherited disorder of bone growth. Individuals affected by the condition are short in stature but have normal size facial features and head size. The may also suffer from joint problems and curvatures of the spine.

I was born with my genetic condition, Pseudoachondrplasia, but it did not manifest itself until I was three years old which is usually the case. My parents were worried because I was always falling a lot, and by the time I was three they noticed that I could not raise my arms properly. They took me to all the relevant agencies where they diagnosed me as having rickets.

In October 1939, I was five years of age and due to start school. The Education Department said I had to attend a special residential school. During my eight years there I got very little academic education. I mainly received treatments such as ultra violet ray lamps, exercises and massage. Cod liver oil was pushed down my throat and as the war was on, there was very little nice food. It wasn’t until I was 11 years old and I was sent into hospital to have one of my legs straightened, that a young doctor there said “This child doesn’t have rickets”. When I was 13, they told my mother that they could do no more for me and I could go home but had to attend a special school. This school also concentrated on health over education. There they also discovered I was riddled with arthritis, in every joint and in my spine. So my medical treatment was increased but nobody ever bothered to search for a proper diagnosis for my genetic condition.

I spent the next few years getting married and having children, who had to delivered by caesarean section. Both my children are free of my genetic condition and have grown up healthy and strong.

It wasn’t until I was in my 60s that I was really concerned about my diagnosis. My niece was taking a course to become a medical secretary and as part of her work, had to do a case study. She asked me if I would be her case study and I agreed but realised that I had never been given a proper diagnosis for my condition. I asked my GP to refer me and wrote to St Mary’s Genetic Department. They did many x-rays and I saw two specialists, but they could not decide what I had. They said it definitely wasn’t Achondrplasia and told me it could even be a condition with no known name. They had a great deal of trouble because all of my bones had deteriorated so much. I had to go back several times. One time, I saw a specialist from Australia and after much deliberation it was decided the nearest they could get was Spondyloepiphyseal dysplasia, known as SED. Another 10 years went by and I got a phone call from St Mary’s. They said they now had tests which could diagnose my condition conclusively.

I have always resented the fact that I was sent away from home in my early childhood. Mostly because I feel I would have achieved much more in life if I had had the education. I was also very institutionalised by the time I went home and found it really difficult to settle into normal life. My early teens were very miserable as I had to learn to ignore the name calling, staring and abuse from other people. The next stage of my life was really wonderful though. I had my daughter, then 14 months later my son. Three years ago I got a Disable Facility Grant with which an annex was built for me on my daughter’s house. That has more or less ensured the rest of my life will be comfortable and happy. I am among my family and see my grand children and great grand children all the time. I am very happy now.

As far as GPs are concerned, I’m afraid they need to listen to us more. I have been a member of the Restricted Growth Association for the past 40 years. I really think that doctors who have a patient with a rare condition
should research as much as they can so that they can advise where specialists are and refer patients as soon as possible. They should also be open to discussing the condition with the patient as often the patient has more knowledge.

I began volunteering with a residents group and local church group but eventually decided to focus my energy on improving things for disabled people. I am part of several committees, monitoring groups and steering groups that campaign for the rights of disabled people. For many years I also assisted with training social workers at Bangor University. I still assist the Occupational Therapy Department with demonstrations of new and updated equipment. In 2009, I was awarded an MBE by Her Majesty the Queen, for my charity work.

Conclusion

The “Experiences of living with a rare disease in Wales” (2012) survey has given RDUK valuable insight into the experience of patients and families living in Wales. It has highlighted some of the key issues they still face and provided suggestions on how to improve the patient experience. Resources need to be used effectively in order to ensure that patients are able to access specialised care whether that clinic or service is available within Wales or outside Wales, which is vital since many GPs have limited knowledge of rare diseases. Improvements need to be made on the diagnostic front and better coordination of care which would save valuable time and money. Access to services such as allied health professionals (e.g. physiotherapists and occupational therapists) and social care professionals also needs to be improved. Patients and families are eager for more funding and information regarding research to be made available. Finally, health services need to address the problem of disparity between paediatric and adult care, and the issues surrounding the transition phase. Patients and their families affected by rare diseases should expect to receive the appropriate care and management of their condition, regardless of where they live.
Appendix 1

Conditions represented in this survey:

- Albinism
- Angelman's syndrome
- Ataxia:
  - Ataxia SCA1
  - Ataxia-Telangiectasia
  - Cerebellar ataxia
  - Cerebellar ataxia SCA6
  - Cerebellar ataxia SCA 11
  - Episodic ataxia, type 2
  - Friedreich's ataxia
- Atypical Haemolytic Uraemic syndrome
- Centronuclear myopathy
- Charcot-Marie-Tooth disease
- Chronic myeloid leukaemia
- Churg-Strauss syndrome
- Common variable immunodeficiency, acquired hypogammaglobulinemia
- Congenital adrenal hyperplasia
- Cystic fibrosis
- Distal spinal muscular atrophy
- Dravet syndrome
- Duchenne muscular dystrophy
- Ehlers Danlos syndrome, type 3
- Epilepsy syndrome
- Fabry's disease
- Familial adenomatous polyposis (FAP)
- Folliculitis decalvans
- Greig cephalopolysyndactyly syndrome
- Guillain-Barré syndrome
- Ichthyosis:
  - Bullous ichthyosis
  - Lamellar ichthyosis
- Idiopathic intracranial hypertension
- Idiopathic thrombocytopenic purpura (ITP)
- KBG syndrome
- Laurence Moon Bardet Biedl syndrome
- Myasthenia gravis, MuSk Seronegative
- Nephritic syndrome
- Nephrotic syndrome
- Neurofibromatosis, type 1
- Panhypopituitarism
- Paraplegia
- Paroxysmal nocturnal haemoglobinuria (PNH)
- Poland’s syndrome
- Pompe acid maltase deficiency
- Pompe’s disease
- Potocki-lupski syndrome
- Prader-willi syndrome
- Primary immunodeficiency
  - Panhypogammaglobulinaemia
- Primary-progressive multiple sclerosis
- Pseudoachondraplasia
- Sarcoidosis
- Smith-Lemli-Opitz syndrome
- Tetralogy of fallot
- Thalidomide
- Thyroid cancer
- Turner syndrome
- Undiagnosed genetic condition
- Variegate Porphyria
- Vasculitis and Central nervous system vasculitis
- WAGR syndrome
- Wegener's granulomatosis
- Wolf hirschhorn syndrome

Experiences of Rare Diseases: Patients and Families in Wales

Rare Disease UK (RDUK)