

ILLUMINATING THE RARE REALITY



Help illuminate the rare reality

There are over
6,000
rare diseases

It takes on average
4 YEARS
5 DOCTORS
3 MISDIAGNOSES
before you get a correct diagnosis

It's estimated that
3.5m
people in the UK will be affected by a rare disease at some point in their lives

Rare Disease Day
28
FEB

Despite the vast scale of people affected, the realities of a rare disease aren't always visible or well understood. However there is hope. We want to shine a light on stories of joy, happiness and love of life. Help us illuminate the rare reality.

Add your star to help illuminate the rare reality of living with rare diseases.

raredisease.org.uk/reality



RARE DISEASE UK

ABOUT RARE DISEASE UK



Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working with the rare disease community and the UK's health departments to effectively implement the UK Strategy for Rare Diseases.



Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 200 patient organisations. Genetic Alliance UK provides the Secretariat for the APPG on Rare, Genetic and Undiagnosed Conditions.



SWAN UK (syndromes without a name) is a patient and family support service run by Genetic Alliance UK. SWAN UK offers support and information to families of children with undiagnosed genetic conditions.

Published by

Genetic Alliance UK
CAN Mezzanine
49-51 East Road
London
N1 6AH

Telephone:

+44 (0)20 7831 0833

Website:

raredisease.org.uk

Registered charity numbers:

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Lead author:

Sarah Gilbert

Photo accreditation:

Michael Hall ©

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RECOMMENDATIONS

Recommendation one: Review and refresh the UK Strategy for Rare Diseases before the end of 2020. The strategy has become obsolete as technologies, policies and systems have moved on. Its commitments urgently need updating to ensure rare disease patients receive high quality services, treatment and support regardless of where they live in the UK.

Recommendation two: Improve diagnosis and early intervention. Make the most of data and technologies for faster and more accurate diagnosis of rare diseases. Realise the value of the UK's investment in genomics to ensure that rare and genetic conditions are identified as soon as possible, improving health outcomes and saving money for the NHS.

Recommendation three: Modernise rare disease care and treatment. Introduce a comprehensive culture of best practice in the NHS, delivering evidence-based and coordinated care.

Recommendation four: Enable people living with rare diseases to access accurate information. The NHS should provide information about rare conditions and signpost to support groups.

Recommendation five: Protect and enhance the UK's role in rare disease research. Ensure capacity for, and commitment to, research into rare diseases is not diminished and that the UK continues to attract investment.

LIVING WITH A RARE CONDITION

A rare disease is defined by the European Union as one that affects less than 5 in 10,000 of the general population. There are between 6,000 and 8,000 known rare diseases. Collectively rare conditions are not rare.

One in 17 people will be affected by a rare condition at some point in their lives. This equates to approximately 3.5 million people in the UK. Approximately 80% of rare diseases are believed to have a genetic component¹. Often rare diseases are chronic and life-threatening. Rare diseases can be single gene, multifactorial, chromosomal or non-genetic.

Those living with a rare condition can face significant challenges in getting a diagnosis, accessing treatment and receiving coordinated care, as well as challenges with employment, education, social life and mental health.

Caring for a child with a rare disease can impact parents' and carers' emotional wellbeing. They also worry about the impact on siblings. Brothers and sisters care a great deal about their siblings' wellbeing, and show increased emotional maturity, empathy and compassion².

Our survey examining the impact that living with a rare condition has on mental health found that 88% of individuals feel emotionally exhausted because of their rare disease³. Patients often describe the experience of not being believed when presenting symptoms to healthcare professionals – particularly when there is no simple explanation. This has a significant impact on the time it takes to reach a diagnosis and can lead to additional anxiety and low mood or depression.

In 2018, we asked young people directly about their experience of living with a rare disease but some found it difficult to explain as they did not see themselves as different from their friends. For young people affected by rare diseases it is important to note that their condition does not form an integral part of their identity². However, as they reach adolescence and gain more understanding about their condition, living with a rare disease can become more challenging. During a focus group, adolescents told the Rare Disease UK team they felt there was a lack of support from healthcare professionals on the emotional impact of being a teenager with a rare condition. A major concern for many teenagers was the impact their health has on their education.

‘The whole process of diagnosis was one which I never wish to repeat. I was labelled a “neurotic” parent by a paediatrician and not taken seriously from my first concerns.’

Parent of a child with Noonan syndrome⁴

DIAGNOSIS

On average, rare disease patients...



receive three misdiagnoses



consult with five doctors



wait four years

before receiving a correct diagnosis.

On average, rare disease patients wait four years to receive a diagnosis with some waiting over 20 years⁴. The term ‘diagnostic odyssey’ refers to the time taken between a patient first developing symptoms and receiving a correct medical diagnosis. For people living with a rare condition this is often a long and eventful journey, frequently with several misdiagnoses until a final, correct diagnosis is reached. Without a diagnosis it can be impossible to access appropriate care and treatment, and without intervention a patient’s health can significantly deteriorate.

Value of a diagnosis

The primary value of a diagnosis might be in supporting the medical management of the condition, by empowering healthcare professionals to make treatment-related decisions that are more effective, but a diagnosis can offer much more too. A diagnosis offers improved access to care and support services. A diagnosis can bring answers to long-standing questions, providing a better understanding of what the future may hold, such as how the condition may progress. A molecular diagnosis allows couples to exercise reproductive choices if they wish to. Receiving a diagnosis may also offer hope, as it can often be the first step towards a treatment or, in some cases, a cure.

Individuals receiving a rare diagnosis may find new information emotionally challenging.

One may discover that the condition is life-limiting, or information about carrier-status or other family members that may be at risk. Receiving a diagnosis can lead to feelings of isolation and increase further uncertainty, particularly where conditions are life-limiting. Some may feel the diagnosis has come too late. Despite the importance families place on receiving a diagnosis, it may not be the final hurdle. In some instances a diagnosis will not lead to treatment options or further information⁵.

Newborn screening

The heel-prick test and other forms of newborn screening have the potential to identify babies at risk of developing rare or genetic conditions, allowing diagnosis before symptoms develop. Many rare conditions of early childhood are progressive and irreversible. The opportunity to provide stabilising treatment before the deterioration in health or development of a child occurs, allows us to prevent the most serious effects of these conditions, which can include severe disability and death.

Currently within the UK, between five and nine conditions are screened for (nine in England, Scotland and Wales, five in Northern Ireland). This figure compares unfavourably to newborn screening programmes in most other high income countries, who routinely screen at birth for over 20 conditions⁶.

Genome sequencing

In 2018 NHS England launched the new Genomic Medicine Service (GMS), with full implementation expected by 2020. As this service rolls out, it will become possible to offer earlier diagnosis and to identify many rare diseases more systematically in clinical practice.

People living with a rare or genetic condition want as much information as possible about their health, but when it comes to genome sequencing, obtaining a diagnosis is a priority above other information that may arise from a genome sequence⁵. Given that receiving a diagnosis can also be an emotional experience, it is essential that parents, patients and families are well supported during this process. Understanding a diagnosis and what this means can be harder when the findings are complex or not clear cut, as can be the case with genetic testing and whole genome sequencing. Expectations around what a test may return and how long this process might take must therefore be carefully managed. Support from a specially trained healthcare professional, such as a genetic counsellor, should be provided throughout the process as this can help individuals and families adjust to the new information a diagnosis can bring.

Undiagnosed conditions

The rarest genetic conditions raise particular challenges to diagnosis, and it is important to recognise that some patients may never receive a diagnosis. **It is estimated that around 6,000 children are born in the UK each year with a syndrome without a name.** Historically around half of these children have remained undiagnosed.

Those families that do receive a diagnosis may wait for many years. A condition may be undiagnosed for several reasons – a patient may not have been referred to a clinician with the appropriate expertise, they may have an unusual presentation of a known condition making diagnosis more challenging, or the condition may be so rare it is not yet studied (a syndrome without a name).

For families affected by undiagnosed conditions there are a number of worries and concerns regarding:⁷



The future of the affected child



The short and long term development of a condition



How the condition will be managed and treated



How other and future children might be affected



Uncertainty in the day-to-day and future planning



Repeated negative and inconclusive results

People with undiagnosed conditions often have complex and significant health needs which are challenging to identify and address. In our 2016 patient experience survey, we found that **73% of people with an undiagnosed condition felt their lack of diagnosis was a barrier to treatment⁴.** An absence of information about the underlying cause can mean healthcare professionals are unsure which treatments may be successful and how the condition may progress. Families fight to be heard and struggle to gain recognition and understanding from professionals. In some instances, support may not be received until a crisis point is reached⁷.

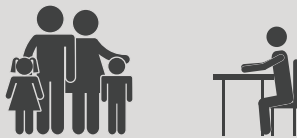
Recommendation: Improve diagnosis and early intervention. Make the most of data and technologies for faster and more accurate diagnosis of rare diseases. Realise the value of the UK's investment in genomics to ensure that rare and genetic conditions are identified as soon as possible, improving health outcomes and saving money for the NHS.

RARE DISEASE CARE AND TREATMENT

Many rare diseases affect multiple systems of the body, which means several different professionals need to be involved to deliver effective care and treatment. The quality of overall care received is often affected by the quality of coordination between these healthcare professionals.

Only 13% of rare disease patients had access to someone to fulfil the care coordinator role.⁸

What is a care coordinator?



A care coordinator is a trained professional responsible for ensuring a care plan is in place and carried out. Their role can assist in ensuring the care provided to patients is as smooth as possible.

Why are care coordinators needed?

The average rare disease patient...

travels one to two hours



attends at least three clinics



at least quarterly



to attend appointments related to their condition.

Without properly coordinated care, people affected by rare diseases often have to devote significant periods of time juggling and attending numerous appointments at different hospitals, sometimes travelling long distances to access the care they need. In addition to caring and medical tasks, time is spent making telephone calls to various providers and services, organising meetings, working with support groups, and researching information. When a patient has no care coordinator, management of a rare condition becomes a full-time role, taking up all a patient or carer's personal time, causing many people to decide to leave employment⁹.

For many, not having access to a coordinator means a lack of information and support – or even feeling lost within the healthcare system. Uncoordinated care can lead to significant financial costs such as expensive travel, petrol, parking, time off work, childcare for siblings, accommodation and food costs⁹. For some, this may be a regular occurrence⁹. Efficient coordination of appointments, particularly where substantial travel is required, would go some way to lowering these financial costs.

Integration with mental health services

Poor or no coordination can also have significant psychosocial impacts on employment, relationships and emotional wellbeing. Yet mental health services for rare disease patients are not routinely coordinated with other aspects of care. Mental health provision should be an integral part of rare disease care plans and considered as important as physical health.

Care coordinators

Care coordinators can help patients to have a positive experience of the care they receive, providing support and information, facilitating effective self-management and preventing hospital admissions. Provision of care coordinators is not just beneficial for patients and families but the wider health and social care system due to their cost effective nature. Care coordinators save GPs' and consultants' time, help prevent unplanned hospital admissions and reduce the length of hospital stays. Therefore, there is a significant need for funding of care coordinators, and for this funding to be sustainable.

Access to treatment

The day-to-day challenges of those living with a rare condition is often exacerbated by the absence of any effective treatment. The existence of proven and effective treatments and medicines does not guarantee access due to our current approaches to making funding decisions across the UK. The UK is slower to make a decision, and more likely to say no, than Spain, Italy, Germany and France on access to rare disease medicines.

Currently only half of licensed orphan medicinal products are available on the NHS. Genetic Alliance UK, the charity that runs the Rare Disease UK campaign, is developing proposals to improve decision making for rare disease medicines with a focus on improving access.

Centres of Excellence

A Centre of Excellence is a specialist clinic where expert healthcare professionals come together to provide the very best care and treatment for rare disease patients. Our survey on rare disease care found that over half of people who had access to a specialist centre rated their care as 'good' or 'excellent'. For those respondents who had none of their care provided at a specialist centre less than a quarter rated their care as 'good' or 'excellent'¹⁰. Rare Disease UK supporters identified three key attributes Centres of Excellence should be able to provide: education and training, links with local healthcare providers, and links with patient organisations¹¹. Access to high quality, evidence based care, such as the treatment offered at specialist clinics, should

be accessible to all rare disease patients in the UK. Collaboration and sharing of this knowledge across the UK is vital to encourage best practice and improve the experiences of care for those affected by rare conditions.

Transition from child to adult services

Transitioning from child services to adult services can be a challenging experience for many rare disease patients. It can come as a shock to many, with the process seeming too sudden. Parents also often feel anxious and unsupported once this change takes place. Effective communication and care coordination are essential in supporting patients and families living with rare conditions during transition to adult services.

After the transition to adult services, many young people find themselves managing their own care for the first time. Young people living with rare conditions want to be more involved in decisions about their care; they wish to be supported to begin to take this role, rather than experiencing a sudden shift in approach².

Young people report that they feel 'in limbo' between adult and child services, and as though they do not fit in either place². Many feel they would benefit from dedicated teenage or young adult services that would offer a phased entry into adult services.

Recommendation: Modernise rare disease care and treatment. Introduce a comprehensive culture of best practice in the NHS, delivering evidence-based and coordinated care.

'I noticed how different it was because when you're a child they're a lot more thorough with you and take the time to explain things and they're more smiley but then on my first appointment in adult care I thought "Oh dear" and I was being treated like an adult.'

Patient affected by a rare condition¹²

INFORMATION AND SUPPORT

Sources of support are essential for patients on their rare disease journey. Potentially valuable sources of support might include patient organisations and networks, online communities on social networks, online resource collections, local disability support groups, and local government or NHS provided support services. Living with, or caring for someone with, a rare condition has a significant emotional impact, and many families benefit from support from those who share similar experiences. Unfortunately, a support group for every single rare disease does not exist and families may face challenges in finding appropriate and relevant support and information. Our research has found that families receive different levels of support after a diagnosis¹³. There is a need for support to be provided to families throughout their lives and throughout their journey with their condition – from being undiagnosed through to participating in research or accessing treatment opportunities. It is important that healthcare professionals are able to direct patients and carers to sources of support.

The role of patient organisations

Charities and support groups are highly valued by those who use their services or participate in their networks, though some patients are unaware of their relevant group and the support they can provide. Connecting with others affected by the same rare disease can empower individuals and families to understand more about the condition and support one another. Some families may wish to be put in contact with those in a similar situation in order to relieve feelings of isolation.

The possibility of connecting with another family may offer the hope of gaining further understanding about the condition.

As well as providing a supporting role, patient organisations are a valuable source of information for families. Information can be an important form of support in itself, it can be vital in order to help families come to terms with their diagnosis by providing them with sensitively presented information. Up-to-date and reliable information is necessary for patients to make informed choices. **We found that up to 70% of respondents did not feel they were given enough information after their diagnosis⁴ and 45% had to research information themselves.**



45% had to research information about their condition for themselves.

Patients and families actively seek information related to their rare condition and how it may develop in the future. Areas of interest for families include information on the cause, underlying genetics, research opportunities, prognosis, treatments available and the effects of treatment⁴. When equipped with up-to-date and reliable information patients are often empowered to become experts in their condition and can make informed decisions regarding care and treatment.

Recommendation: Enable people living with rare diseases to access accurate information. The NHS should provide information about rare conditions and signpost to support groups.

‘The one thing that we desperately wanted to know was more about the other two children with the same gene change, and that bit the geneticists weren’t able to share. Because if we can connect with other families and learn about the problems they have, it might help signpost and give a clearer prognosis for Georgia.’

Parent of a child with an undiagnosed condition¹³

RARE DISEASE RESEARCH

Research is fundamental to improving our understanding of rare conditions. It can result in a host of benefits such as: uncovering new information about a condition, developing a treatment, improving diagnosis and raising awareness of a condition. For families affected by rare conditions, research offers the potential of a brighter future and improved prognosis. Participating in research can be empowering for rare disease patients. Many patients are keen to participate in research, such as clinical trials, in the hope of developing a therapy to cure or alter the effects of a condition.



80%

of rare disease patients said they would be interested in research participation.

However, only

25%

felt they were given enough information about research.



Patient organisations play an important part in sharing research with their communities and as such researchers should seek to engage with patient organisations where possible. For many rare disease patients, both the internet and patient organisations are a key way of finding out about research opportunities.

Genomic research

Genomic medicine promises to improve diagnosis, the accuracy and timeliness of diagnosis and to support the development of new treatments. The 100,000 Genomes Project reached completion in 2018 and as a result of its success, whole genome sequencing will now be integrated into the NHS as the new Genomic Medicine Service (GMS). Alongside the launch of the GMS, the Government committed to sequence one million whole genomes through the NHS and UK Biobank. These initiatives will provide a growing resource for research into genetic conditions.

Registries

Registries offer the opportunity to progress research by collating information about those affected by rare conditions. The National Congenital Anomalies and Rare Disease Registration Service (NCARDRS) records those

people with congenital abnormalities and rare diseases across the whole of England. This information can improve understanding of congenital anomalies and rare diseases, and facilitate research about prevention, causes and treatment. The data collected can also be used to inform the planning, commissioning and improvement of NHS services.

European Reference Networks

European Reference Networks (ERNs) are virtual networks connecting patients and clinicians across Europe. There are currently 24 thematic ERNs facilitating research by providing access to a large enough cohort of patients to support clinical trials, in addition to linking researchers to share knowledge and develop new treatments. ERNs have the potential to transform rare disease care, and raise standards and equity in rare disease care across the EU. Prevention of rare diseases is also a major aim of ERNs. Given the possible ejection of the UK from participation in ERNs it is vital that research in rare conditions is encouraged and funded.

Recommendation: Protect and enhance the UK's capacity for, and commitment to, research into rare diseases, ensuring that funding is not diminished and that the UK continues to attract investment.

KEEPING THE STRATEGY UP TO DATE

In 2013 the Department of Health and Social Care published the UK Strategy for Rare Diseases, a policy document that sought to ensure rare disease patients receive high quality services, treatment and support regardless of where they live in the UK.

The UK Strategy for Rare Disease is comprised of 51 commitments spread across five interlocking themes:

- **Empowering patients, families and carers affected by rare disease**
- **Identifying and preventing rare diseases**
- **Diagnosis and early intervention**
- **Coordination of care**
- **Research**

Implementation plans were published in Scotland, Wales and Northern Ireland by 2015. In 2017, the All Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions held an inquiry into the lack of plan for England¹⁴. This report indicated that the absence of a plan in England – where the majority of the patient population, the majority of treatment centres, and the majority of research are located – was holding not just England back, but the other three home nations too. Coordination between nations, a crucial element of the EU's flagship ERNs for rare disease, was found to be particularly difficult with poor understanding of plans within England.

In 2018 the Department of Health and Social Care and NHS England finally published implementation plans that indicated their approach to implementing the strategy, dividing their duties between the commitments falling within their responsibility.

Despite the missed opportunities, there has been significant progress in relation to the UK Strategy for Rare Diseases. These include: the development of the National Institute for Health Research's BioResource for Translational Research in Common and Rare Diseases which opens up routes for researchers to study rare diseases; and the establishment of the National Congenital and Rare Disease Registration Service, which will create a much clearer picture of the incidence of rare diseases in England, facilitating planning, research and clinical practice¹⁴.

However, there are limitations to the strategy and challenges to overcome. The UK Strategy for Rare Diseases, published in 2013, is now out of date. Some aspects of the strategy are no longer in existence, such as the UK Genetic Testing Network.

There are also several examples of progress that have taken place alongside the strategy. This includes the launch, and completion of, the 100,000 Genomes Project; which is now being integrated into the NHS in England. Working with the Department of Health and Social Care, NHS England supported the development of ERNs – allowing for the exchange of expertise, faster diagnosis, improved clinical care and improved collaboration of research. These are great leaps forward, but they are not built into the strategy, leaving missed opportunities for a strategic approach.

There has been a significant change in the policy landscape with Brexit endangering the UK's continued participation in ERNs and risking significant change to the research and treatment landscape. Consequences of Brexit are likely to be most keenly felt among people affected by rarer conditions as cross-border collaboration and information sharing is most important where patients, knowledge and clinical expertise is scarce.

With just under a year left until the strategy reaches its deadline, Rare Disease UK is calling for a review and refresh of the strategy. There is a continued need for the strategy with many patients still experiencing difficulties in relation to care, treatment, support and reaching a diagnosis. The strategy must continue beyond 2020, with better consolidated commitments that take account of the changes that have occurred over the last six years. This will support further developments in research, expertise and knowledge of rare conditions, to ensure that no one gets left behind just because they have a rare disease.

Recommendation: Review and refresh the UK Strategy for Rare Diseases before the end of 2020. The strategy has become obsolete as technologies and systems have moved on. Its vision and recommendations urgently need updating to ensure rare disease patients receive high quality services, treatment and support regardless of where they live in the UK.

LOOKING TO THE FUTURE

This report is a compilation of Genetic Alliance UK and Rare Disease UK's findings to highlight areas of importance for rare disease patients, and underpins our call for a refresh and review of the UK Strategy for Rare Diseases. Despite some progress in the implementation of the strategy it is clear that rare disease patients still face fundamental challenges in relation to care, mental health, the diagnostic odyssey, research and access to information – and that further action can be taken to continue to improve the lives of rare disease patients across the UK.

A refresh and review of the UK Strategy for Rare Diseases would provide an avenue for this action to take place in a measurable and impactful way, in addition to raising awareness of rare conditions as a health priority to decision-makers.

We have identified several areas where there is a substantial need for continued research and renewed policy focus. These include:

- The journey to diagnosis. Using new developments to improve identification and aid faster diagnosis, while recognising that families need to be prepared for the impact of receiving a diagnosis, and the consequential positive and negative implications that may follow. Families should also be aware that the condition may remain undiagnosed, and be supported to understand what this may mean for their care and treatment.
- The care that rare disease patients experience. It is important that all rare disease patients have a care pathway that takes in to account their needs as their condition develops, that is evidenced based and implements best practice, and that is coordinated to gain efficiencies and deliver the best experience possible to patients and families. Focus should also be placed on the transition between adult and child services in order to empower young people to feel involved and make decisions about their own care.
- Access to reliable and up-to-date information and support. For many rare conditions there may not yet be a patient organisation available, and finding reliable up to date information may be difficult. Where possible, patients should be provided with information about their condition or pointed in the direction of where to find it, and should have support throughout the entirety of their diagnosis journey, including after receiving a diagnosis.
- Research is of key importance to people living with a rare condition as it provides solutions to unmet health needs. Incentivising research in specific rare condition remains a top priority for most patient organisations due to the possible benefits such as developing a treatment, a cure or altering the path of a condition. Further investment and communication of research will be vital for rare disease patients.

This report will be delivered to MPs at the 2019 Rare Disease Day reception, and to the Minister responsible for rare diseases, Baroness Nicola Blackwood. This will accompany an open letter from Genetic Alliance UK members and Rare Disease UK supporters in calling for a refresh and review of the UK Strategy for Rare Diseases.

As major opportunities and challenges for rare disease care draw near, a renewed strategic approach will be crucial to prevent fragmentation and ensure equity for people living with rare conditions. To beat rare diseases it is vital that the UK is joined up nationally and internationally as we experience the significant changes to come.

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