

GOOD DIAGNOSIS

Improving the experiences of diagnosis
for people with rare conditions



**GENETIC
ALLIANCE** UK

ABOUT GENETIC ALLIANCE UK



Genetic Alliance UK is the largest alliance of organisations supporting people with genetic, rare and undiagnosed conditions in the UK. Our 200+ members and the people they support are at the heart of everything we do.

We advocate for fast and accurate diagnosis, good quality care and access to the best treatments. We actively support progress in research and engage with decision makers and the public about the challenges faced by our community.

We run two long standing projects:



Rare Disease UK, a campaign focused on making sure the new UK Rare Diseases Framework is as successful as possible, and to ensure that people and families living with rare conditions have access to a final diagnosis, coordinated care and specialist care and treatment.



SWAN UK (syndromes without a name), the only dedicated support network in the UK for families that have a child or young adult with an undiagnosed genetic condition.

Thank you to all those who participated in this project and generously shared their diagnosis experience. Without your contribution, this report would not have been possible. This report includes anonymised quotes contributed by people who participated in our Good Diagnosis workshops, Rare Resources (Scotland) Project and the Rare Experience 2020 Project.

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FOREWORD



Diagnosis is universally important in a rare disease journey. It might come following a diagnostic odyssey, it

might come soon after birth through newborn screening, and for some members of our SWAN UK (syndromes without a name community) it will never come. But, for all affected by a rare condition, a diagnosis – or its absence – is a crucial element of their experience.

The UK has made a great deal of progress in building infrastructure that will accelerate diagnosis, our genomic medicine programmes have the potential to shorten the diagnostic odyssey for many.

However, there are gaps that are still to be filled before we can claim to have timely and accurate diagnosis for all. Traditional newborn screening technology is under-used here compared with the majority of our neighbours in the European Union, and we still need to work on diagnostic pathways that cater to the 28% of rare conditions that have not been linked to a genetic cause.

One of the bitter-sweet consequences of the pandemic has been a change towards online meeting with our membership – the positive side is that we have had more regular contact, and more frequent discussions on issues that matter to our members. This report is the fruit of that increased contact. In the autumn of 2021, the concept of a ‘good diagnosis’ came out of a weekly meeting.

The report has been a fascinating – and sobering – opportunity to examine individuals’ experiences of receiving (or not receiving) a diagnosis of a rare condition. People go on a diagnostic odyssey because they are looking for answers, for treatment, for the next necessary step to maintain their health. Unfortunately for too many the end of the diagnostic odyssey is not the launchpad they were looking for. We can’t ignore that these

negative experiences will often be so much worse for people from marginalised communities.

Our findings are important: we should work together between nations to share and generate information; we need to take account of people’s needs while they are on their diagnostic odyssey and when it ends; and we should work towards a set of standards for what comprises a good diagnosis.

Alongside the clear messages from people living with rare conditions, we are delighted to have contributions from Breaking Down Barriers, the CONCORD research team, Medics4RareDiseases, and Unique. Their pieces show the complexity of this issue, and how many dimensions of the problem need to be examined and addressed.

It is reassuring to see that the four priorities of the UK Rare Diseases Framework appear well chosen when considered in the scope of this work: to improve the experience of diagnosis (priority 1), we need to coordinate people’s care (priority 3) and healthcare professionals need to be able to provide information (priority 2), and if we can get this right, we will be able to deliver treatments (priority 4).

We hope this report and its findings set the tone for future work on evolving and implementing the Action Plans of the UK. We want to move towards greater collaboration, more shared platforms and broader consultation with everyone living with a rare condition.

Handwritten signature of Nick Meade in black ink.

Nick Meade

Joint Interim Chief Executive and Director of Policy, Genetic Alliance UK

Handwritten signature of Lauren Roberts in black ink.

Lauren Roberts

Joint Interim Chief Executive and Director of Support and Engagement, Genetic Alliance UK

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INTRODUCTION

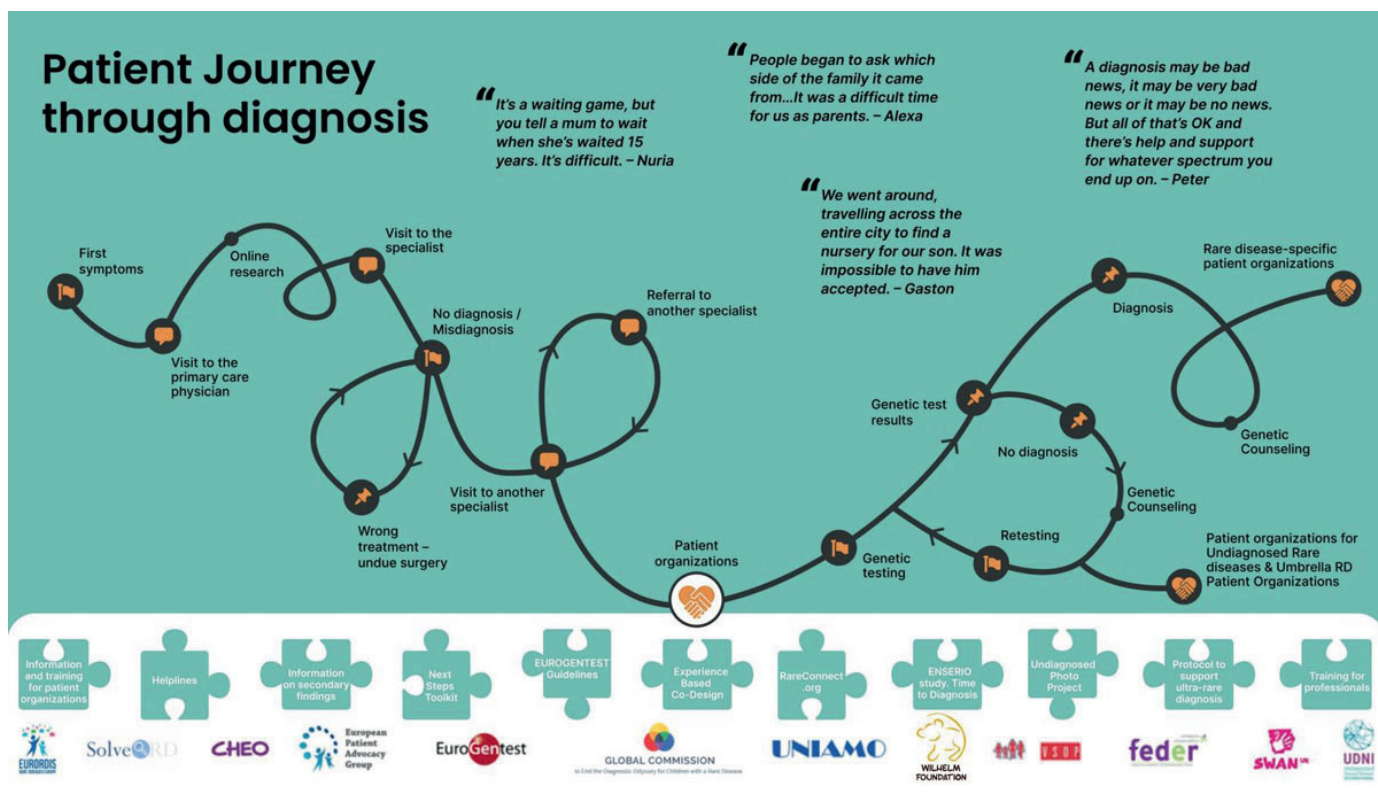
In the UK, 1 in 17 people will be affected by a rare condition during their lifetime. Unfortunately many will face significant challenges in getting a diagnosis, with more than a third of people with a rare condition having to wait more than five years. This ‘diagnostic odyssey’, a term used to describe the time between first symptoms and receiving a final diagnosis, can have a significant impact on a person’s quality of life and wellbeing.

Stakeholders in rare conditions must strive to ensure people with rare conditions receive a fast and accurate diagnosis. Diagnosis can lead to improved medical management and can open doors to accessing care, treatment and support services. It can bring answers to long-standing questions, provide a better understanding of what the future may hold and can lead to improved experience and outcomes for people with rare conditions.

The value of diagnosis has been recognised by the UK Rare Diseases Framework which has rightly prioritised helping people with rare conditions to get a diagnosis faster. The Framework expresses commitments to investment and improvement of genomic, diagnostic and screening services and acknowledges the important role of healthcare professionals in recognising and diagnosing rare conditions.

But speed of diagnosis is only part of the picture. How a person is supported on their journey to diagnosis is equally important and all too often people with rare conditions will report feeling unsatisfied with the experience and with the level of information and support that they are provided.

In December 2021, Genetic Alliance UK set out to better understand people’s experience of diagnosis and to identify what matters most to people with a rare condition when they are



Infographic by Solve-RD Community Engagement Task Force, led by EURORDIS solve-rd.eu/community-engagement-task-force/

on their diagnosis journey. Through a series of workshops, we asked people to reflect on their experiences at three key stages of their diagnosis journey: their search for a diagnosis, receiving their diagnosis, and following diagnosis. This work built on the findings of our Rare Resources (Scotland) and Rare Experience 2020 projects, and featured three interactive workshops designed to identify the principles of good diagnosis.

It was clear from our findings, that a person's experience of diagnosis was significantly influenced by the healthcare professionals involved in their care. To improve the speed and experience of diagnosis, we must urgently address how healthcare professionals are equipped to support people living with rare conditions.

It is important to note that this report is not a criticism of those who work for the NHS and provide care to people with rare conditions. Rather it is an acknowledgement that the systems and structures are not necessarily in place to support healthcare professionals to deliver the best possible diagnosis experience for people living with rare conditions.

This report will outline what is needed to make a diagnosis 'good'. Of course, this is not an easy task. The experience of diagnosis is, at its essence, a very personal and individual experience. There is no one right way to pursue and receive a diagnosis, what works for one person might not be appropriate for another. However, there are some guiding principles identified by our work which have been presented in this report.

GOOD DIAGNOSIS IS...



GOOD DIAGNOSIS IS ACCURATE AND TIMELY

People with rare conditions want access to an accurate diagnosis faster

Over a third of people living with a rare condition will wait for more than five years to obtain a definitive diagnosis, often receiving a number of misdiagnoses along the way.

88%

had a diagnosis (but 35% of these waited more than 5 years)
(Genetic Alliance UK, 2020)

8.8%

were undiagnosed (almost two thirds of these had been undiagnosed for more than 5 years)
(Genetic Alliance UK, 2020)

33%

with a definitive diagnosis had 2 or more misdiagnoses prior to their diagnosis
(Genetic Alliance UK, 2020)

56%

who did not have a definitive diagnosis stated that they had been misdiagnosed in the past.
(Genetic Alliance UK, 2020)

A fast and accurate diagnosis can enable access to appropriate care and opportunities for interventions that may improve a person's health. Fast diagnosis can mean greater treatment choice and support informed decision-making, leading to people with rare conditions being able to better manage their condition.

Delays in diagnosis can also have a significant impact on a person's mental wellbeing. Our workshop participants described feelings of anxiety while waiting for answers and stress and worry

about what their future may hold. In addition, juggling multiple healthcare appointments, referrals and tests get in the way of everyday life, and the emotional ups and downs experienced throughout the journey can have a significant impact on mental wellbeing.

Benefits of accurate and timely diagnosis extend beyond direct impact on the person with a rare condition. Shortening the journey to diagnosis and avoiding often costly unnecessary referrals, tests and treatments will benefit NHS Services.

It is important to acknowledge that challenges around diagnosis for rare conditions are well known and recognised by those responsible for delivering rare condition policy and services in the UK. Recent years have seen significant investment and advances in genomics and diagnostic services which will go some way to reducing the diagnostic odyssey for people living with rare conditions. We must continue to make the most of data and technologies for faster and more accurate diagnosis of rare conditions and to realise the value of the UK's investment in genomics to ensure that rare and genetic conditions are identified as soon as possible.

Delays in diagnosis could be avoided if healthcare professionals had improved training and access to information about rare conditions

Beyond investment in screening and diagnostic services, workshop participants indicated that raising awareness of rare conditions among healthcare professionals and improving training on how to recognise and diagnose a rare condition was integral to receiving a timely diagnosis. Participants shared that the length of their diagnostic journey could be affected by a number of factors including:

- Not being listened to or having their symptoms/concerns taken seriously
- Healthcare professionals having limited knowledge of rare conditions
- Delays in making referrals or ordering tests
- Long time periods between appointments and receiving test results
- Poor coordination between healthcare professionals

Workshop participants also commented on a tendency amongst healthcare professionals to suspect that unusual symptoms in individuals are often the result of an unusual presentation of a common condition. With greater awareness and access to better information, clinicians are more likely to be able to identify when this is not the case and an individual in fact has a rare condition.

Measures are needed to improve awareness of rare conditions among healthcare professionals to increase the likelihood of a rare condition being considered among the differential diagnoses at an early consultation.

Whilst there is acceptance that it is not possible for healthcare professionals to know about every rare condition, people with rare conditions expect healthcare professionals to:

- Be open to exploring rare conditions as a possible diagnosis
- Acknowledge the limitations in their knowledge and understanding of rare conditions
- Be willing to undertake research and training to better understand rare conditions
- Facilitate speedy referrals to specialist services for diagnosis

Increasing healthcare professionals' knowledge and awareness of rare conditions is essential to improving both the time taken to receive an accurate diagnosis and to ensure a satisfactory experience of diagnosis.

Improving access to reliable information and training materials for healthcare professionals will help to achieve this.

People with rare conditions want to be believed when they present with symptoms

A common experience shared during our Good Diagnosis workshops was that many people with rare conditions felt that they were routinely dismissed, not listened to or not believed by their healthcare professionals on their journey to diagnosis, leading to avoidable delays in obtaining a diagnosis.

'In over 20 years, and after living in 3 different towns, I have only met 3 doctors who did not dismiss me or even laugh in my face.'
[Participant, Rare Experience 2020 Survey]

Participants expressed frustration at the time wasted attending multiple appointments to repeat symptoms but to have no action taken. Language such as 'fight', 'battle' or 'struggle' is often used to describe how they had to approach their appointments. This, when a person is unwell and in need of care and support, is detrimental to both a person's physical and mental wellbeing.

There was discussion during workshops that the likelihood of having an experience of feeling dismissed or disbelieved was more common amongst women, either presenting with their own symptoms, or seeking help for their child or loved one, suggesting a perception of prejudice within the health service. Concerns were also raised by the group that gender may not be the only factor in determining the likelihood of whether a person may be believed – it was noted that ethnicity, social class and age may determine how a person is treated when first presenting symptoms.

'It's that 'young and female' thing again – it's a common factor in so many stories of late diagnosis and poor care. It's bad for all women but it's particularly bad when you are younger'
[Participant, Good Diagnosis Workshop]

Ensuring a good diagnosis for all



People who are from diverse and marginalised communities and affected by a rare/genetic condition, often experience further disadvantage. There are

many contributing factors and action needs to be taken to ensure everyone receives equitable access to healthcare, services, and support.

A rapid evidence review recently published by The NHS Race and Health Observatory reveals major ethnic inequalities in healthcare. Although not specific to rare conditions, the report focuses on areas pertinent to the rare disease community including ethnic inequalities in:

- Mental Health Services
- Maternal and Neonatal Healthcare
- Digital Inclusion and Access to Healthcare Services
- Genetic Testing and Genomic Medicine Studies
- The NHS workforce

The review includes a set of recommendations

for practice and policy and for further research in each of these areas. One of these recommendations' states:

'Research is required to understand at what points in the care and referral pathway in genetic testing and counselling services, ethnic inequalities are apparent, and what the nature of these inequalities are.'

Breaking Down Barriers actively involves people with lived experience to listen, learn, and understand more about the impact of rare/genetic conditions and health inequalities. We know from speaking directly with those affected by rare/genetic conditions that people often encounter barriers in terms of language and access to inclusive and accessible services. We continue to see information, resources, and a workforce that is not representative of our diverse population or reflective of different cultures and religious beliefs.

Seeking and receiving a diagnosis of a rare/genetic condition can have a devastating impact on individuals and families. That agony is even further compounded when parents are made to feel blame for their child's genetic condition based on their cultural practices, such as marriages between close blood relatives. We also hear the devastating consequences of healthcare professionals having pre-conceived ideas and making assumptions based on a person's ethnicity or religion. Is it any wonder that we continue to hear that people from diverse and marginalised communities often have a lack of trust in healthcare providers?

Families not only need accessible information and support at the point of diagnosis, but they also need access to a care pathway that supports them and their families throughout their journey. There are some positive examples and models of care where healthcare professionals have actively worked to improve their own cultural competence, to design services and support around what their community say they need, rather than what service providers have assumed they need. We need to learn from these examples of good practice and stop thinking of communities as 'hard to reach'. Unfortunately, services and support can all too often be hard to access.

Kerry Leeson-Beevers, Chief Executive, Alström Syndrome UK & Breaking Down Barriers

Participants also reported receiving a misdiagnosis of mental health conditions on their journey to diagnosis, believing that this occurred because of healthcare professionals' unwillingness to acknowledge the possibility of a rare condition. These findings support those identified in the Genetic Alliance UK Rare Experiences 2020 Report which highlighted that:

'There appears to be an issue with patients with rare conditions being misdiagnosed with mental health conditions, as some respondents indicated that their misdiagnosis was sometimes due to physical symptoms being treated as psychological symptoms. This kind of misdiagnosis is the most worrying to find, as this results from a failure of healthcare professionals to properly engage with ill people seeking help. Some respondents mentioned they have been labelled as "drug-seekers" or "nuisances"'

[Participant, Rare Experience 2020 Survey]

Workshop participants who had experienced this shared that long delays to diagnosis resulting from not being believed, or having a misdiagnosis of mental health condition, ultimately severely impacted their mental health, with some participants noting symptoms of depression and anxiety being experienced as a result.

'Doctors should not assume patients are lying just because their knowledge or ability to diagnose is limited. The lack of belief or rudeness and dismissal by doctors who did not have knowledge to diagnose was the worst part of the journey.'

[Participant, Rare Experience 2020 Survey]

Once again, these experiences highlight why it is important to improve awareness of rare conditions among healthcare professionals; to increase the likelihood of a rare condition being considered and to help ensure people with rare conditions are listened to and believed.



I AM SOMEONE



RARE

GOOD DIAGNOSIS IS INFORMED AND SUPPORTED

People with a rare condition want to be supported throughout their diagnosis journey

A diagnosis journey can be eventful and difficult. Juggling multiple appointments, processing sometimes difficult and complex information, all while unwell, can leave people with rare conditions feeling alone and overwhelmed.

Having a source of professional support during this time can be extremely beneficial. Workshop participants highlighted the benefits of having someone to turn to to discuss their conditions, their worries and to answer their questions.

Support organisations play an integral role in providing high quality support and information to people throughout their diagnostic journey

People with rare conditions need to be made aware of available reliable sources of information that they may want to use to research their condition or access support and information. These sources should include the relevant support organisations where they exist. These organisations often have forums and helplines where people with a rare condition are able to discuss their concerns with others who have experienced similar situations. This can be a simple, effective way to address some of the support needs that people with rare conditions may have.

Workshop participants highlighted the importance of being given the information of relevant support organisations as early in the diagnosis journey as possible as this can help people to source information in their own time, when they feel that they are ready.

Healthcare professionals may need help to do this as they may not be aware of support organisations, may not know how to find them, and in some cases there may not be an organisation at all. Rare condition umbrella organisations, such as Genetic Alliance UK can support healthcare professionals to identify sources of information and support.

Informing Diagnosis



The needs of families are at the heart of Unique's mission, and this includes the desire to provide families worldwide with the support and information they need immediately following diagnosis

and throughout their child's life. Unique's guide writing team, consisting of two scientific information officers, has produced almost 300 guides for chromosome and single gene disorders (SGDs), and over 400 translated guides in 18 different languages.

Healthcare professionals and families alike must be able to trust that the information we publish is informed, accurate and up-to-date. In turn, while much of the information available from medical sources can be written using stark language that may be particularly distressing for those who may have just received a diagnosis, we strive to present the information we provide using easily-accessible, family-friendly formats and language. All our guides are freely available to families and professionals through our website.

Unique guides are of use not only to family members directly affected by a rare genetic condition, but also help explain more to extended family members and friends, together with medical and education professionals who are involved with the care of a child but do not have much knowledge of their specific genetic condition.

We believe that the insights and experiences of families are key to the value of the information we provide. Life-time data is collected from Unique members, together with concerns, advice and reassurance on topics such as behaviours, education and social issues. It is these minute details, which only those living with a rare genetic condition experience and share, that is invaluable to newly diagnosed families.

All of our guides are reviewed, most are translated, and some are written by clinical geneticists or medical/scientific experts who kindly volunteer their time to help share their knowledge. The very professionals who make use of our guides also help ensure that Unique's guide production continues to go from strength to strength.

Sarah Wynn PhD, Chief Executive Officer, Unique

People with rare conditions need access to high quality information throughout their diagnosis journey

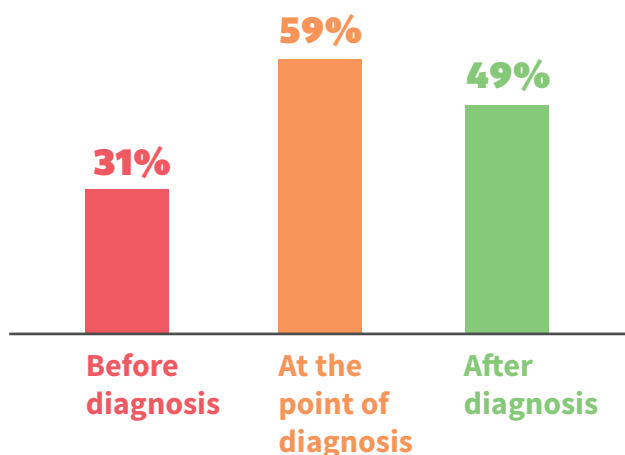
People with rare conditions are not provided with sufficient reliable information throughout their diagnosis journey.

'Anything would be an improvement. Diagnosis given in three line letter from NHS trust hospital, no context support or information.'
[Participant, Rare Experience 2020 Survey]

Workshop participants reported that their information needs can change over time and in response to the different stages of their diagnosis journey. They recognised that the type and level of information a person may need will depend on the individual. For example, one workshop respondent spoke of finding comfort in reading any and all information relating to their child's condition that they could find, while another explained that after their child's diagnosis, they felt so overwhelmed by the diagnosis itself that they did not feel able to take in any more information.

Although each person has their own individual information needs and preferences, what they do have in common is that they can find it difficult to source good quality, reliable information on their medical, psychological and other needs. A lack of information can lead to feelings of isolation, uninformed decision making, poor management of their condition and delays in access to appropriate care, treatment and services.

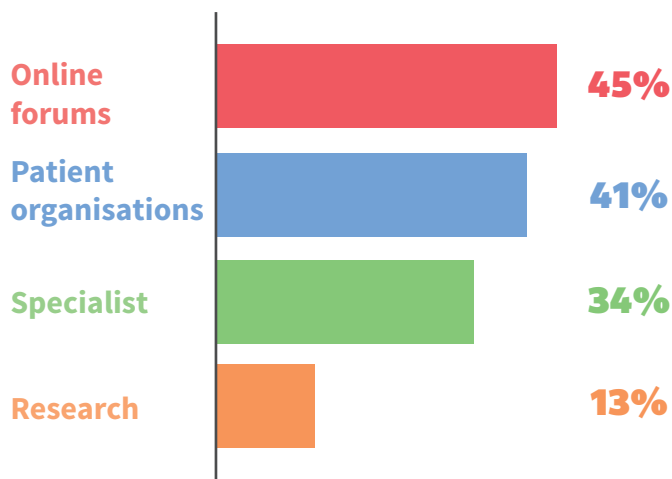
Percentage of respondents satisfied with information provided by healthcare professionals



(Genetic Alliance UK, 2020)

As information on rare conditions and their management can often be hard to find, it is of vital importance that people with rare conditions are supported by their healthcare professionals and/or the NHS to source and access the information they need.

Who have been the main sources of support and information?



Online forums (a source for 45% of participants) and support organisations (41%) were the most frequently used sources of information. Around a third (34%) of respondents said a specialist was their main source of support and information. Many respondents (13%) looked for information themselves (eg by looking at journal articles, books and/or contacting researchers). (Genetic Alliance UK, 2020)

Too often, people are left to search for information themselves and this can risk people finding inaccurate, unreliable or sometimes upsetting information.

'Access to up-to-date, accurate information is absolutely essential when a prenatal diagnosis of a rare condition is given – this is necessary for parents to make fully informed decisions. We've seen parents finding information with outdated information on prognosis which could have led them to making a decision which they would not have done had they had access to the correct, most recent information and the support group.'
[Participant, Rare Resources Workshop]

Where there is limited accessible information available, healthcare professionals should help to source any available information, even if very complex – and offer to discuss with the person.

Of course, it is important to recognise that it can also be a challenge for healthcare professionals to access information too. Although there is a considerable wealth of information on rare conditions available, there remains some gaps, particularly for the extremely rare conditions. NHS websites such as NHS Choices (England and Wales) and NHS Inform (Scotland) do provide some information on a small number of rare conditions, but this is typically limited and for the more well known rare conditions. Specific online resources for rare conditions, such as orpha.net are not widely known about or user friendly.

‘Help was available in the hospital but the doctor did not refer me when he gave the diagnosis. Also there is a local charity that He did not know about.’

[Participant, Good Diagnosis Workshop]

Workshop participants shared many examples of good quality information, support organisations and websites, but expressed frustration at the lack of a central repository for rare conditions information. Having a NHS branded, recognisable and well known source of rare conditions information and resources would be beneficial to both people with rare conditions and healthcare professionals.

Helping healthcare professionals deliver a good diagnosis



When a disease is rare it results in a paucity of data and first-hand experience. This in turn leads to a paucity of information for both the patient and their healthcare

professionals. The impact of this can be felt throughout a patient’s journey to diagnosis.

However a lot is known about rare disease as a collective group of conditions and this knowledge can be used to equip medical professionals to manage their undiagnosed and diagnosed rare disease patients. Medics4RareDiseases (M4RD) provides access to essential information and training on Rare Disease as a collective group of conditions, including pragmatic tips and practical

tools. Working closely alongside people with lived experience of rare diseases, advocates, clinicians and other experts, M4RD has developed ‘Rare Disease 101’, an online training resource that is freely available to all medical professionals.

We understand the limitations doctors face in being empowered to effectively and efficiently manage rare disease in their clinical practice. A lack of rare disease education, heavy workloads and ongoing training commitments present a challenge to trainees and doctors in the NHS supporting people with rare diseases, and with over 7,000 rare diseases, it would be unrealistic for them to become an expert in every one.

With these challenges in mind, M4RD designed Rare Disease 101 to be flexible and pragmatic. It addresses the basics of rare diseases, key practical information, websites, knowledge sources and invaluable tools that are readily available but the average healthcare professional is unlikely to know of. With stories, examples from the rare disease community, it highlights the common challenges that patients face in accessing healthcare.

The patient-doctor relationship is at the heart of everything we do. By providing healthcare professionals with practical tools and advice on managing both diagnosed and undiagnosed patients, with rare conditions, we hope to reduce the diagnostic odyssey and drive tangible improvements in care.

We also want to improve the experience of patients. For many people with a rare condition, the journey to a diagnosis can be challenging, overwhelming and distressing. Using appropriate language, acknowledging limitations in knowledge and delivering news in a way that balances realistic expectations with hope, can make a big difference to the way in which a person views the quality of their care.

People with rare conditions talk about diagnosis being an ongoing process that continues after receiving the actual diagnosis and the impact of having a diagnosis can be felt in lesser and greater magnitude at different points in life. Healthcare professionals need to provide ongoing support in order to help them live their lives to the fullest – and M4RD can support health professionals to do that.

Lucy McKay, Chief Executive Officer, M4RD

People with a rare condition would benefit from having the opportunity to access support for their mental and emotional wellbeing throughout their diagnosis journey

The diagnosis journey can also have a significant impact on mental and emotional wellbeing, yet mental health services for people with rare conditions are rarely available, or offered.

Good Diagnosis workshop participants highlighted the importance of having the opportunity to access mental health support throughout the diagnosis journey. There are three key stages where mental health support may be valuable:

The journey to diagnosis – mental health support should be offered to people seeking a diagnosis for their rare condition in recognition of the impact searching for a diagnosis can have on a person’s mental wellbeing. The journey to diagnosis can be frustratingly long, and it is not uncommon for people to have to fight to be listened to and have their symptoms taken seriously. This can have a profound effect on a person’s emotional and mental health.

At the time of diagnosis – the delivery of a diagnosis can be emotionally challenging and overwhelming and how a diagnosis is delivered can itself have a significant impact on the person receiving it. Diagnosis must be delivered with compassion and recognition of the significance to the person receiving. Ensuring people have the opportunity to access mental health support during this time is necessary.

Patients and carers are rarely asked about their mental health by healthcare professionals: around

50%

**are never asked
(Rare Disease UK, 2018)**

Post-diagnosis – people with rare conditions can often experience anxiety, depression and other mental health responses in response to their diagnosis. This can occur for a long period of time

following the delivery of diagnosis and may come and go in the years post diagnosis. Healthcare professionals must recognise the long-term impact of a diagnosis on a person’s mental health and ensure there is ongoing opportunity to access mental health support.

Healthcare professionals must also recognise the impact of a diagnosis of a rare condition on the whole family. A diagnosis of a rare condition may mean families members have to come to terms with news that that a person they love has a life limiting or life shortening diseases, they may have to come to terms that their expectations for their child will not be realised, they may have to suddenly change their lives to provide care and support. It is therefore of great importance that consideration is given to how the family unit can be supported throughout the diagnosis journey.

Participants in our Good Diagnosis workshop indicated that they had not been offered access to support services for their mental and emotional wellbeing, and some participants who had requested it had either not received it, or experienced significant waiting times to access services.

Participants shared personal experiences of how the diagnosis journey impacted mental health and wellbeing – including the impact it had on social life, relationships with family and friends, ability to work and financially provide for oneself.

‘Follow up for psychological support would definitely have been beneficial – it’s possibly expected that GPs will deal with that but mine were out of their depth and not wanting to educate themselves.’

[Participant, Good Diagnosis Workshop]

Healthcare professionals should consider how to integrate mental health support in the care plan of any person on the journey to diagnosis. Some workshop participants indicated that the healthcare professionals involved in their care weren’t very aware or understanding of the emotional and mental implications of the diagnostic odyssey. Healthcare professionals must be provided with training, information and resources to support them to help their patients access mental health support.

Healthcare rights must be clearly communicated

Many people who have negative experiences of obtaining/trying to obtain a diagnosis speak of how they have had to ‘fight’, ‘battle’ and be their own advocate, all whilst feeling unwell. This can lead to breakdown in relationships with their GP or other healthcare professionals and can have a significant impact on their emotional and mental wellbeing. People report not knowing where to seek support, how to complain or where to get advice on how to get a second opinion.

When a person begins their journey to diagnosis, there is little information and guidance on what to expect. This can mean that people with rare conditions may not have a clear understanding of the challenges to get a diagnosis or the possibility of there being a long journey to diagnosis.

As many people with rare conditions can often be mislabelled as a ‘neurotic patient’ or as having health anxiety, there are people with rare conditions experiencing delays in getting referrals for diagnostic tests or assessments because they are not being taken seriously by their principal healthcare professional. Workshop participants described being frustrated at poor communication of timelines for tests and referrals. Others described

their ‘battles’ and ‘struggles’ to get referred for tests, or for assessment at specialist centres.

‘Patients cannot exert their rights unless they know what they are.’

(Participant, Good Diagnosis Workshop)

A clear message that emerged from both the Rare Resources (Scotland) Project and the Good Diagnosis workshops was that, when empowered with knowledge of healthcare rights, people with rare conditions felt more confident attending appointments, challenging decisions about their care that they were not happy with and advocating for their own care. However, it was also clear that many did not know what their healthcare rights were or how to enforce them.

Workshop participants clearly indicated that it would be beneficial for people with rare conditions to know and understand what they can expect from health services, what is expected of them and what can be done when a person is not satisfied with the care they are receiving. The development of a Rare Conditions Charter for Diagnosis would be a helpful tool to support people with rare conditions to better understand their rights and responsibilities.



GOOD DIAGNOSIS IS COLLABORATIVE AND COORDINATED

People value a considered approach to receiving a diagnosis, based on their individual needs

Receiving a diagnosis of a rare condition will never be easy. Discovering that you, your child or loved one has a health condition that might be life limiting or changing is likely to be an emotionally challenging experience. How a person responds to this information will depend on the individual and the circumstances they find themselves in – much as there is no one way to respond to a diagnosis, there is no one way to deliver one.

However, how a diagnosis is delivered can be as important as the diagnosis itself. Workshop participants identified a number of things healthcare professionals should consider when deciding how to deliver a diagnosis. Factors such as location, timing, level of information shared, support services that should be recommended and decisions on what should happen next must all be considered and tailored to the individual.

‘I think being told directly is needed, personally I’d prefer face-to-face or video call – not finding out when a GP mentions it has been in my file for years (as happened, although not with a rare condition). I’ve mostly known when a diagnosis would possibly be given in an appointment though, because I’ve known it’s been set up to discuss the results of tests.’

[Participant, Good Diagnosis Workshop]

Whilst there is not a one-size-fits-all approach to delivering a diagnosis, our workshop participants did offer some recommendations for healthcare professionals.

- Diagnosis should be delivered by someone who has an understanding of the rare condition or rare conditions in general. It is important for the person delivering the diagnosis to be able to explain what the condition means for the person. If information

is limited, it is important this is acknowledged and explained.

- Diagnosis should be delivered face-to-face wherever possible.
- Prior to the appointment, information on what will happen at the appointment and what to expect can be helpful. Information should be factual, but reassuring.
- Careful attention to language is needed. People value a balanced honesty – they do not appreciate being presented with only the worst case scenario.
- Diagnosis appointments must not be rushed – there must be adequate time for questions, emotions and discussion.
- No one should leave an appointment where they receive a diagnosis without information on the condition, information for relevant support groups and contact details for follow up support.
- People should be offered mental and emotional wellbeing support.
- People should be offered a follow up appointment at a time of their choosing.
- People should be provided with sufficient follow up support.
- People do not wish to be ‘discharged’ following diagnosis, but rather have the opportunity for follow up appointments and support, with discharging being mutually agreed when the person is ready.

Unfortunately, during our Good Diagnosis workshop we heard from some participants that their experience of diagnosis was extremely negative as a result of a lack of consideration of their needs.

Whilst there was an acceptance that no healthcare professional would deliberately set out to deliver a diagnosis in a manner that would cause distress, there was also an acknowledgement that some healthcare professionals may not have experience of delivering diagnosis of rare conditions and as such may not appreciate the impact such a diagnosis can have.

‘I wasn’t warned that I was going to receive her diagnosis at a routine appointment. I was by myself and after 5 years of searching I was understandably emotional.’

[Participant, Rare Experience 2020 Survey]

‘It was an emotional time with lots of suggested conditions that were tested for (which were negative) before finding the right diagnosis. Some doctors acted as if we should be elated at receiving a diagnosis not recognising the enormity and detestation of having your baby diagnosed with life long multiple complex health problems. It wasn’t a relief to get a diagnosis, it was devastating.’

[Participant, Rare Experience 2020 Survey]

Ensuring healthcare professionals have access to good quality information and materials to support the delivery of the diagnosis may be helpful. Training resources, featuring people with lived experience of rare conditions sharing their recommendations for effective diagnosis should be developed.

People with a rare condition want to be treated as a partner in healthcare decision making throughout their journey to diagnosis

“Patient is expert” sounds great, but is often an empty phrase – hierarchy of healthcare professionals/ patients real obstacle in reality?

[Participant, Good Diagnosis Workshop]

Many workshop participants report an imbalance in the relationship they have with their healthcare professionals, for example people with rare conditions can be acknowledged by their healthcare professionals as being an ‘expert’ and more knowledgeable about their condition, but when it comes to decision making or requesting a particular action, they do not feel listened to. This can be frustrating for the person with the rare condition and can lead to delays in accessing referrals to specialist services, appropriate tests, care or treatment.

People want to feel that they are an active and valued partner in their care – this requires that the person have access to information about their condition, about their care and to know and feel

confident in asserting their rights. However, this also means asking a person with a rare condition who may often feel very unwell, to be their own advocate.

A diagnosis care plan could be a helpful tool in ensuring individuals are informed, consulted and able to participate in the decision making around their diagnosis. This diagnosis plan could include information such as a description of symptoms and details of how the symptoms will be managed, what steps will be taken to achieve a diagnosis (for example, what tests will be undertaken, referrals made, how often will there be a review), what third sector organisations can be referred to and details of how the person’s mental and emotional wellbeing will be supported.

Such a care plan should be shared and flagged to all healthcare professionals involved in the person’s care.

People want all healthcare professionals involved in their care to work collaboratively and share information

For many people, the complexities of their rare condition mean that their care is delivered by different healthcare professionals in different locations. Coordinating appointments and managing the communication between specialties can often become the job of the person with the rare condition, making them coordinators of their own care. People report being frustrated by the lack of communication between parties involved in their care – for example, late referrals, poorly updated notes, IT systems that do not talk to each other resulting in long waits whilst letters are written, sent and received. People also express frustration at having to chase up their own appointments, test results and having to regularly repeat themselves to the same clinicians. The above can lead to feelings of wasted appointments, lack of confidence in their care and missed opportunities to access the correct care or treatment.

Where possible, people with rare conditions would like to see greater effort from the healthcare professionals involved in their care to assist in making appointments more manageable and efficient. People value the opportunity to attend multidisciplinary appointments where possible and for telehealth to be used to bring together clinicians from different locations where appropriate.

‘My local optician, the eye specialist team at [my local hospital], my GP and local pharmacy have worked really well together to deal with recurrent episodes of uveitis’

[Participant, Good Diagnosis Workshop]

Indeed, the rapid expansion of telehealth services necessitated by the Covid-19 pandemic has brought many opportunities for people with rare conditions to have their care delivered in more efficient ways. The use of telehealth can bring benefits such as removing the need for lengthy travels to expert centres, or enabling a support worker or counsellor to join the appointment. However, participants in our workshops were keen to stress that telehealth is an important tool, but should not be the default way in which care is delivered and will not be appropriate in all instances. For example, telehealth appointments would not be suitable for physical examinations or the delivery of a diagnosis.

Decisions on whether telehealth tools are used should rest with the person with the rare condition. Telehealth appointments should not be the default because it is most convenient for healthcare professionals, but rather when its use benefits the person living with the rare condition.

People with a rare condition want to have the opportunity to access specialist services, even if this means travelling to another nation within the UK

Some of the participants in our Good Diagnosis workshop were referred to a specialist centre for their rare condition, either to receive a diagnosis when that rare condition was suspected, or to receive care following a diagnosis being made.

Those who had had the opportunity to do this were, for the most part, very pleased with the standard of care that they received and valued the opportunity to access these services.

Unfortunately, for some people, getting a referral to a specialist service can be another example of a ‘fight’ or ‘struggle’ and there can often be significant delays between a person asking for a referral and a referral being made. When asked why they felt this was the case, workshop participants suggested that this could be because of one of the following reasons relating to their principle healthcare professional:

- They did not want to accept that they did not have sufficient expertise to diagnose/manage the person’s rare condition
- They were intrigued by the possibility of a rare diagnosis and did not want to ‘give up’ the person
- They did not accept the person’s suggestion of what the diagnosis may be and was not prepared to make a referral
- They did not agree with the diagnosis that had been suggested/given by another healthcare professional
- They were unsure how to make a referral (particularly when referring a person to a service in a different part of the UK/different healthcare system)

Healthcare professionals must be supported to make referrals to appropriate specialist services, and would benefit from having access to a source of information containing the details of specialist services for rare conditions in the UK, including contact details and referral criteria.

More must also be done to ensure that there is improved knowledge sharing between specialist centres and local services as the diagnosis of a rare condition can often depend on the individual having access to a specialist centre for that condition. Clinicians in local services must be able to recognise the limitations of their own knowledge and when they need to actively seek help and guidance from specialist centres and experts.

When people living with rare conditions do have to access services outside of their health board or home nations, whether for diagnosis or for routine care they must be financially supported to do so. People report not being offered information on how to access travel cost support – and in some cases the reimbursement system is not adequate when a person does not have the means to pay upfront for their travel.

People value a dedicated care coordinator to support the management of their care throughout their diagnosis journey

Most rare conditions affect multiple body systems, meaning that many professionals from different specialties and disciplines need to be involved in the care and treatment of the individual. As such, it is vital that there are formal links between all those involved to ensure that the person with the rare

conditions is receiving the best possible care and that it is optimally coordinated.

Fragmented care can result in individuals feeling 'lost in the system', which leads to a lack of confidence in the care that they receive. Poor coordination and communication make it difficult for the individual to keep track of when they should be visiting which professional, resulting in missed opportunities for receiving vital care and support. It also makes it difficult for a person with a rare condition to identify who to go to for help with a specific problem or question and where to access the care they need. Often it has resulted in individuals having different professionals looking at specific elements of an individual's condition, but no-one being concerned with the condition as a whole. Fragmented care can be costly for the NHS too, resulting in services being used inefficiently and not making the best of professionals' and the individual's time. Poorly coordinated care makes forward planning more difficult and can result in missed opportunities for interventions, sometimes leading to costly alternative treatments further down the line or avoidable emergency hospital admissions.

People with rare conditions should be offered a designated care coordinator to liaise between themselves and the services they use, and to ensure that the right services are brought together at the right time. The care coordinator should assist in liaising between the different professionals involved in the care of a person with a rare condition, making sure that services are used more effectively and the person with the rare conditions has as smooth an experience of receiving care as possible.

Most people with rare conditions do not have, but would value, access to a dedicated care coordinator. People reported coordinating their own care and information – leading their own search for a diagnosis, being responsible for collating evidence, and finding/organising their own condition specific support after a diagnosis.

People would value the support of someone before, during and after a diagnosis. For example, having a coordinator who is an advocate for families looking for a diagnosis (e.g. chasing referrals, appointments, tests, ensuring all those involved in a person's care are up to speed), being present at key appointments (e.g. when a diagnosis is given), and being a point of contact particularly after receiving a diagnosis –

addressing immediate questions and facilitating the appropriate follow up support.

The care coordinator should be a trained professional whose role is to ensure that a care plan is in place and acted upon. The care coordinator's role should include being available to talk to the person with the rare conditions about his or her concerns and giving consideration to the needs of the family or carer. The person best equipped to carry out the role of care coordinator would vary between conditions but suggestions of appropriate professionals have included specialist nurses working within the appropriate fields, members of the genetics team or social workers. Where the role of care coordinator is additional to a professional's existing role, it should be included in their job description and specific time should be allocated to carrying out this role. Care coordinators should be able to make appointments for individuals and schedule multiple appointments for the same day. There should be a system that gives priority to booking these coordinated appointments, thereby reducing the need for a person with a rare condition to travel to hospital on multiple occasions.

Coordinating Care



The findings of the CoOrdINated Care Of Rare Diseases (CONCORD) study have improved our understanding of how care is currently coordinated for patients with rare conditions, and are informing how care should be coordinated in the future.

Findings from the study indicated that many patients affected by rare conditions do not experience coordinated care – for example, they do not have access to a care coordinator, care plan or specialist centre. Participants shared their personal experiences of uncoordinated care and the wide range of impacts it had on them, including impacts on their physical health, finances and wellbeing.

Many different ways to coordinate care for patients with rare conditions were identified, showing that one size does not fit all. However, the study made a number of recommendations for future practice including improving access to care coordinators and planning and developing appropriate models of care coordination which

suit different needs and which can be tailored towards different services and individuals.

The CONCORD study found that having the support of a professional to coordinate care may help patients to access the right care in a timely way and reduce the burden associated with coordinating their own care. It may be that the benefits of having a care coordinator are more significant for particular patients such as those establishing a care plan, going through a period of transition or for those with the most complex needs.

Improving coordination of care is key to improving the diagnosis experience for patients

and carers. The findings of the CONCORD study should be considered by those designing and delivering care to those looking for a diagnosis as well as people needing care and support following a diagnosis.

Contribution provided by Dr Amy Simpson, Senior Researcher, Genetic Alliance UK

The CONCORD study was conducted by researchers from University of Cambridge, University College London and Genetic Alliance UK, in collaboration with patient representatives. The study ran from 2018 to 2021 and was funded by the National Institute for Health Research (NIHR).



EXTRACTOR

PAIN

Muscle

GOOD DIAGNOSIS IS ACKNOWLEDGED AND RESPECTED

Diagnosis is not the end of the journey

Even when a diagnosis is delivered in the best way possible, with lots of time given for questions and discussion at the appointment, it is still common for people with rare conditions to have lots of questions about their diagnosis in the days, weeks and months that follow.

Workshop participants described mixed experiences when asked about whether they had been offered follow up support at the time of diagnosis. There were some positive experiences of people being able to either access the specialist clinician, or a specialist nurse to discuss their diagnosis, but for most, no follow up care was provided. Some perceived the lack of follow up appointments being as a result of limited workforce capacity (i.e. no available staff to take appointments) while others suspected that they hadn't been offered an appointment because their healthcare professional did not know how to help them.

'A diagnosis shouldn't stop at the name.'

[Participant, Good Diagnosis Workshop]

The impact of not having follow up care can be very significant. It can lead families to feel overwhelmed and stressed when questions cannot be answered, it could delay a person accessing the services, treatment and medicines that could improve their lives or stop further progression of the condition.

'The appointment with the genetics team where we received the diagnosis was overwhelming. It would have been useful to have a follow up appointment a few months later after we had time to process the diagnosis and think of questions. Left the hospital baffled, no plan in place for treatment, no reassurance.'

[Participant, Rare Experience 2020 Survey]

A diagnosis care plan should be established with every person seeking a diagnosis of a rare condition for themselves or their child. The plan

should outline what they should expect from the diagnostic odyssey including; how they will be supported to get a diagnosis, how long it may take, what NHS services they may expect to access. The care plan should be updated with details of how the person will be supported at the time of diagnosis. This should include the information that they will be provided with, the proposed care plan, details of where mental health support can be accessed, details of relevant support organisations and details of post-diagnosis follow up support will be offered and provided.

Healthcare professionals must acknowledge the impact of receiving a diagnosis of a rare condition on a person's mental and emotional wellbeing

A diagnosis of a rare condition can be life changing and many workshop participants explained that even though they anticipated that they may have a rare condition, finally receiving a diagnosis was an emotional and overwhelming experience.

Despite the impact a diagnosis can have, opportunities to access support for mental and emotional wellbeing are not routinely shared or offered by healthcare professionals.

82%

agree or strongly agree that being asked more frequently about their wellbeing and mental health by their healthcare professions would improve it
(Rare Disease UK, 2018)

Those who had experienced a difficult journey to diagnosis, often defined by having to fight to be listened to or believed, described the profound effect receiving a diagnosis could have on their mental wellbeing. Some participants described their feelings of vindication being outweighed by symptoms of/akin to post-traumatic stress disorder.

'I finally fought for a diagnosis at 24 but the damage of having been labelled a lazy liar since childhood despite my severe chronic pain, still leaves psychological scars. I doubt myself to this day due to negative and dismissive comments from these doctors and other adults. They let me down.'

[Participant, Rare Experience 2020 Survey]

Once more, we heard from workshop participants that they perceived the lack of opportunity to access mental health support to be down to healthcare professionals not having sufficient understanding of rare conditions to recognise the impact of diagnosis and the diagnostic odyssey.

Training resources, developed in collaboration with people living with rare conditions that demonstrate the emotional toll of the diagnostic odyssey and how healthcare professionals can support their patients on their journey, should be created.

Healthcare professionals must accept a diagnosis of a rare condition made by a clinical expert and take the necessary steps to ensure the person with the rare condition receives the most appropriate care

45%

of diagnosed respondents were diagnosed by a doctor who specialises in a specific rare condition.

(Genetic Alliance UK, 2020)

Some workshop participants reported that, when a diagnosis of a rare condition is given by a specialist, it is not always acknowledged or respected by other healthcare professionals involved in their care. Examples include:

- A healthcare professional receiving the results of a diagnostic test and not communicating this result to the person with the rare condition because they did not deem it important
- GPs continuing to make referrals for tests as if to try and disprove a diagnosis

- GPs openly stating that they did not believe a diagnosis that had been given
- Healthcare professionals refusing to make referrals to appropriate services or start appropriate treatments because they disagree with the diagnosis

Workshop participants expressed frustration that their routine medical care can be significantly affected when their GP or regular healthcare professional does not adequately respect their rare condition diagnosis. Some report that it means their GP or regular healthcare professional will attribute all new symptoms to the rare condition without exploration, and others report that their GP or regular healthcare professional continue to assess on a symptom by symptom basis, without consideration of whether it may be as a result of or part of the rare condition. In both cases, this indicates that the lack of knowledge and expertise of rare conditions is affecting the routine care of people living with a rare condition.

Some workshop participants also believed that they had experienced delays in accessing specialist services throughout their diagnosis journey because their GP or regular healthcare professional was somewhat proprietorial over their healthcare – as if they felt a sense of ownership and were afraid of losing control over the person's care. This demonstrates the importance of coordinated care and good communication between local and specialist services.

People with rare conditions want non-medical services to acknowledge and respect their rare condition

Some individuals may have a range of non-medical needs as a result of their rare condition – this can include, but is not limited to:

- Financial or social security support
- Assistance with reasonable adaptations or additional support in the workplace or place of education
- Adaptations in the home
- Need for carers to support everyday living
- Psychological support

Workshop participants spoke of the challenges in accessing services or support for these matters when there is only a suspected diagnosis, or indeed when a diagnosis of a rare condition has been made.

'It's important to have the diagnosis in writing too as it's needed to access the appropriate support and benefits.'

[Participant, Good Diagnosis Workshop]

Again, people with rare conditions report feeling that their condition is not taken seriously or that they are not believed when seeking to access social care services. Lack of awareness of rare conditions outside the medical profession, across social care and education for example, can mean people with rare conditions are regularly subjected to long drawn out needs assessments, refusal of services or having to 'fight' and 'battle' for their needs to be met. This once again places the onus on the person with the rare condition to have to advocate for themselves, often at a time when their physical and emotional health is low.

During our Good Diagnosis workshops there was consideration of the disconnect between rights to services and access to services. It was noted that in policy and legislation, there is a narrative that support is provided based on the individual's needs, regardless of diagnosis. In reality, the experience is that, with or without a diagnosis for a rare condition, those making decisions about whether or not support will be offered are less likely to understand the impact of the condition on the person.

This can be addressed through:

- Developing a diagnosis care plan which can outline the person's symptoms, medical and non-medical needs, the possible diagnoses and what steps are being taken to achieve a diagnosis. This can then be used as a tool to inform discussions with other services.
- Providing people who either have received a new diagnosis, or who are on their journey to diagnosis, the opportunity to access a care coordinator who can clearly communicate the person's needs on their behalf and can navigate the social care system.
- Raising the profile of rare conditions within social care and providing access to good quality information and training materials on rare conditions.

The Undiagnosed Community



This report shows that an accurate, timely and well-informed diagnosis can be extremely valuable to individuals affected by a rare condition. However, it is important

to acknowledge that not everyone will receive a diagnosis. Some people may not be able to access the tests necessary to identify their condition, while others may be affected by a condition that is so rare it is yet to be discovered. These ultra rare conditions may never be identified, even using whole genome sequencing, with diagnostic rates currently sitting at around 25% (Genomics England 2021).

Living without a diagnosis can have a huge impact on an individual and their family. Without a diagnosis it can be unclear what treatments and therapies would be best to manage a person's condition and ensure they have the best possible quality of life. Even when additional support such as respite care or portage is afforded to a family based on their child's needs, a diagnosis is often required to fill in the necessary paperwork to qualify to access these services. No diagnosis also means no prognosis, leaving individuals and their families with little to no idea how their condition will progress and even whether or not the condition will be life-limiting.

Challenges such as not being taken seriously by healthcare professionals and struggling to access accurate information that some individuals with rare conditions experience on their diagnostic journey, can be experienced by an undiagnosed person or their parent or carer throughout their life. All of this can have an extremely detrimental impact on individuals' and families' mental health.

We must ensure that people living without a diagnosis have access to the support that they need. Improving awareness of rare and undiagnosed conditions is a necessary step to ensuring healthcare professionals are equipped to support families.

Isabel Rundle, Engagement and Support Manager, SWAN UK

MAIN MESSAGES AND RECOMMENDATIONS

Our Good Diagnosis project has underlined the importance of diagnosis for people living with rare conditions. Fast and accurate diagnosis can lead to improved medical management, access to care, treatment and support services. It is therefore encouraging that the UK Rare Disease Framework has prioritised helping people with a rare condition to get a diagnosis faster.

But a fast diagnosis is not enough to ensure a good diagnosis. Our findings demonstrate the significant emotional and mental health impact of the diagnostic odyssey on people with rare conditions and their families and whilst speeding up the journey will certainly help to improve the experience, a good diagnosis can only be achieved if people with rare conditions have access to good information and feel supported from the beginning of their journey, to the point of their diagnosis and beyond.

Increasing awareness of rare conditions among healthcare professionals

Our findings show that central to a good diagnosis are healthcare professionals who recognise and are aware of rare conditions. Of course no healthcare professional can be aware of all rare conditions individually, but ignorance of rare conditions generally leads to slower referral, slower diagnosis, misdiagnoses and slower access to appropriate specialised care. We must urgently take steps to raise awareness of rare conditions among healthcare professionals and to provide them with the information and resources required to support their people with rare conditions.

RECOMMENDATION ONE: The UK Rare Disease Framework Delivery Partners should consider developing a central repository (such as an online portal) of information on rare conditions for healthcare professionals. This should include:

- Information on specific rare conditions
- Information on specialist services available throughout the UK including contact details and referral criteria

- Information on available support organisations
- Training materials and resources

Ensuring people with rare conditions receive information and support throughout their diagnosis journey

Participants in our workshops reported having insufficient information to meet their medical and non-medical needs throughout their diagnosis journey. This can contribute to feelings of isolation, poor management of their condition or uninformed decision making.

Too often people are left to research their own symptoms, or diagnosis and this can lead to people finding unreliable, potentially alarming information which they have to digest without the support of a person who understands the condition.

Throughout their diagnosis journey, a person's information and support needs can change and so it is important that their needs are frequently reviewed. Ensuring that people have access to the information they need, at the right time, can contribute to them better managing their care.

As information on rare conditions and their management is often more scarce and difficult to find than information on common diseases, it is of even greater importance that people with rare conditions are supported by the NHS with the information they need. Throughout the diagnosis journey, individuals and families need to be made aware of available reliable sources of information that they may want to use to research their condition or access support and information. These sources should include the relevant support organisations where they exist.

The individual's emotional and mental wellbeing needs should also be addressed and this should be included as an integral part of their care plan and services should be readily accessible if and when they are needed. Support for the needs of the individuals' wider families and support network should be considered too, as the diagnosis journey can also be overwhelming for those caring for or living with a person with a rare condition.

RECOMMENDATION TWO: People with rare conditions should be given a diagnosis care plan when they begin their journey to diagnosis. This should include:

- Description of symptoms and how they will be managed
- What steps will be taken to try to reach a diagnosis, for example, what tests will be ordered, referrals made or how often there will be a review
- Details of third sector organisations that can provide support
- Details of how a person’s mental and emotional wellbeing will be supported through the journey to diagnosis
- Details of how the person’s family will be supported throughout the diagnosis journey

The diagnosis journey can be difficult to navigate. Multiple appointments, lots of tests, referrals to specialist services are just examples of the complexity the healthcare demands of people with rare conditions. Trying to coordinate this care, and manage regular daily tasks, when unwell can be very challenging.

These challenges can be made easier with the involvement of a professional care coordinator who can provide an important role in ensuring that the right services are brought together at the right time. A care coordinator can liaise between the different professionals involved in the individual’s care, make appointments and chase up tests and referrals, helping to reduce the burden on the individuals and ensure that services are used efficiently.

A care coordinator can ensure that a care plan for the diagnosis journey is in place and acted upon and that the individual has a person available to talk about their concerns to, giving consideration to the needs of the family or carer.

RECOMMENDATION THREE: People living with rare conditions in the UK should be offered access to a care coordinator throughout their journey to diagnosis.

Good Diagnosis: Patient Rights Charter

When a person begins their journey to diagnosis, there is often little information and guidance on what to expect.

Workshop participants described feeling frustrated at poor communication of timelines for tests and referrals. Others described their ‘battles’ and ‘struggles’ to get referred for tests, or for assessment at specialist centres.

Even once a diagnosis is made, it may not be the end of the struggle. When an individual has a diagnosis of a rare condition that their principal healthcare professional does not know a lot about, they may continue to experience delays in being started on the correct care or treatment pathway, they may experience delays in accessing appropriate services, medicines or research.

Workshop participants explain that fighting for the right care or treatment can be extremely challenging, particularly when unwell. The fight can be harder when it is not clear what rights they have, or who can support them.

It is vital that people with rare conditions are aware of what to expect from the diagnosis journey, aware of their rights and how they can challenge decisions.

RECOMMENDATION FOUR: A Rare Conditions Good Diagnosis Patient Rights Charter should be developed and included in each national action plan. The Charter should clearly communicate the standard of care people with rare conditions should expect to receive. The Charter should be based on the identified Principles of Good Diagnosis as defined by this report.

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