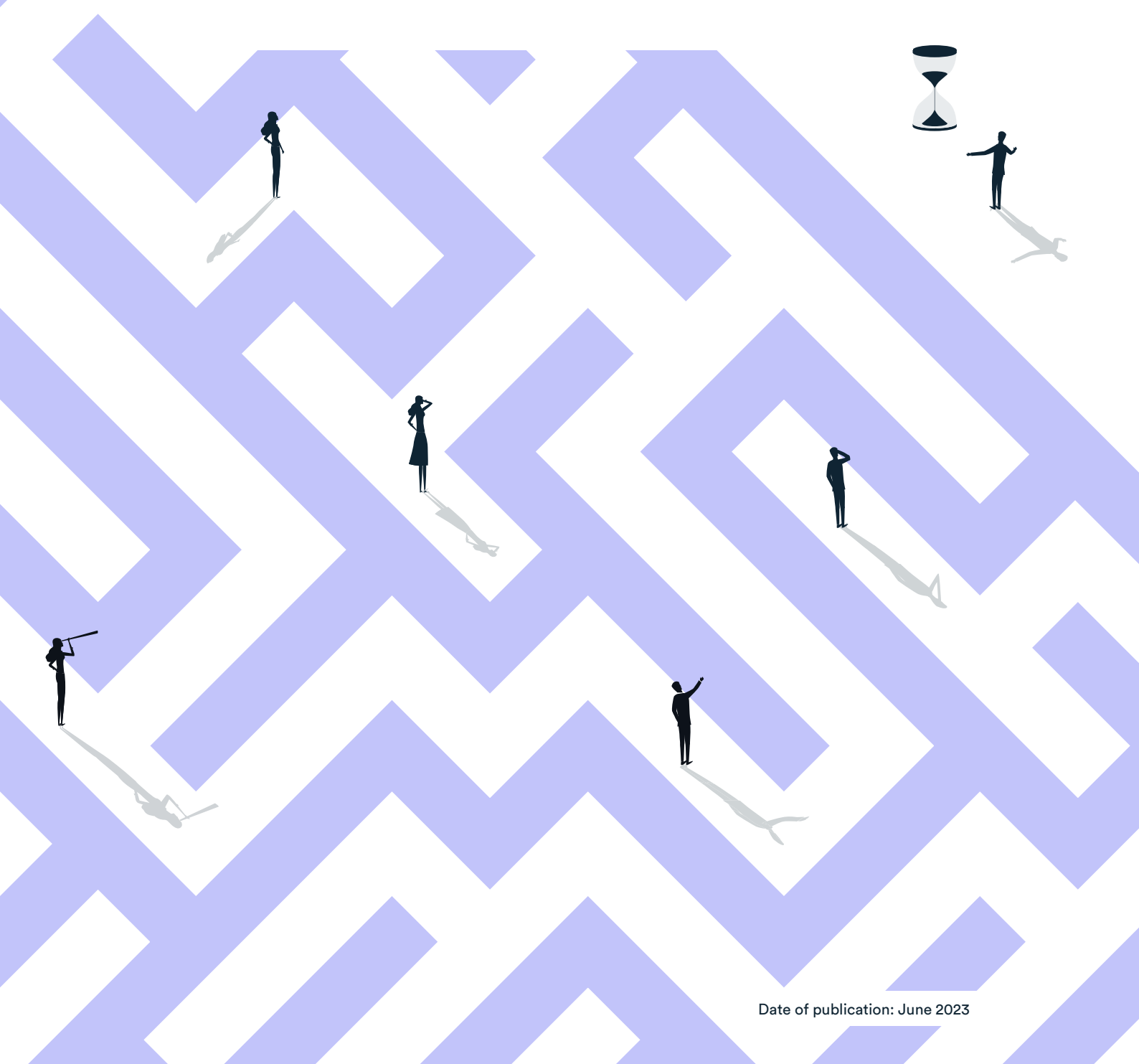


Navigating the labyrinth: reducing delays to a rare disease diagnosis



Recommendations

A diagnosis of a lifelong – and sometimes life-limiting – condition can be difficult to accept, but it can also open up possibilities of treatment, specialist care and community support. However, receiving a diagnosis is not the moment that a person’s rare disease journey begins. For many, their journey will have started years before.

The title of this report is inspired by the experiences of one member of the Specialised Healthcare Alliance (SHCA), who believes that their personal journey to their rare disease diagnosis is best described as a *labyrinth*: a complex maze filled with wrong turns and dead ends which is challenging to navigate. This report explores the barriers to a diagnosis that patients face when navigating the labyrinth, and the opportunities presented by the policy landscape to transform the diagnostic process for the better.

Testing for rare diseases

- 1** The NHS in England and the devolved nations should commit to nationally rolling out successful pilot schemes for rare disease testing, including rapid whole genome sequencing services and newborn screening, with new targets for adoption
- 2** Data should be collected on delays to diagnosis for rare disease patients, including through collecting and publishing data to better understand how community diagnostic centres (CDCs) are meeting the needs of rare disease patients in England

Support for undiagnosed patients

- 3** The NHS in England and the devolved nations should open further *syndrome without a name* (SWAN) clinics across the UK for patients living without a diagnosis over the next five years, following a successful pilot scheme in Wales and work to establish the first clinic in England. Government funding for these further SWAN clinics should be confirmed immediately
- 4** SWAN clinics should designate a diagnostic co-ordinator to patients, who can liaise between the healthcare professionals involved in a patient’s diagnostic journey and help them navigate the pathway

Awareness among healthcare professionals

- 5 The NHS in England and the devolved nations should create a digital tool for healthcare professionals specifically for rare disease diagnosis – one place where they can access information, receive answers to questions from other clinicians, and find and utilise e-learning resources created by patient organisations
- 6 As part of ongoing reviews into rare disease modules in medical curricula, the NHS in England and the devolved nations, and relevant medical schools, should ensure that healthcare professionals are trained not only in how to recognise rare disease symptoms, but how to engage with patients and families with sensitivity
- 7 NHS England should update the [online directory](#) for medical conditions to include links to registered charities for each rare disease, to help patients reach the correct support and enable healthcare professionals – especially in primary care – to signpost patients to the right place

Mental health support during the diagnostic journey

- 8 As part of the integration of mental health support into rare disease care pathways, the NHS in England and the devolved nations should ensure that mental health support is offered to patients and their families whilst seeking a diagnosis, not just from the point of diagnosis onwards
- 9 The NHS in England and the devolved nations should collect and publish data to determine whether mental health support for patients seeking a rare disease diagnosis is available equitably across the UK

A blueprint for the future of rare disease diagnosis

- 10 NHS England should carry out an assessment of learnings from the implementation of the *Inherited White Matter Disorders (IWMDs) Diagnostic and Management Service*, with a view to identifying whether the service could be developed into a model for use in other rare diseases

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1. Introduction

A diagnosis of a lifelong – and sometimes life-limiting – condition can be difficult to accept, but it can also open up possibilities of treatment, specialist care and community support. However, receiving a diagnosis is not the moment that a person’s rare disease journey begins. For many, their journey will have started years before.

The Specialised Healthcare Alliance (SHCA) has developed this report to explore the barriers to receiving a timely and accurate rare disease diagnosis, and the impact that a long diagnostic journey has on patients and their families. This report has been informed and guided by the insights of our member charities, who together represent millions of people living with a rare disease in the UK.

Although the diagnostic process can be lengthy and burdensome, this report identifies several key opportunities for change, where new commitments from policymakers have the potential to transform the experiences of patients and their families moving forward. Improving the diagnostic journey has rightfully been placed at the centre of The UK Rare Diseases Framework, and accompanying national Rare Disease Action Plans. The SHCA acknowledges the commitments and progress made so far by policymakers to shorten the diagnostic journey. In alignment with and in addition to the commitments contained in these publications, we have developed recommendations under five themes:

- 1 Testing for rare diseases
- 2 Support for undiagnosed patients
- 3 Awareness among healthcare professionals
- 4 Mental health support during the diagnostic journey
- 5 A blueprint for the future of rare disease diagnosis

We hope that our recommendations will be considered by policymakers as part of ongoing work to develop future Rare Disease Action Plans across England and the devolved nations. Throughout the production of this report, our SHCA charity members have affirmed their commitment to helping implement the Rare Disease Action Plans; we therefore call on policymakers to ensure the patient community are included in their efforts to shape the diagnostic journey for the better.

Rare diseases in numbers

A rare disease is a condition which affects less than

1 in 2000 people¹

There are over

7000

recognised rare diseases, which can be life-limiting and life-threatening¹



New rare diseases are still being discovered, with

60

new conditions found through a study this year²



80%

of rare diseases have a genetic basis¹



1 in 17

people in the UK will be affected by a rare condition during their lifetime; this equates to 3.5 million people in our population¹



75%

of rare diseases affect children and more than 30% of children with a rare disease die before their 5th birthday¹



2. The diagnostic labyrinth

Some people living with a rare disease will be diagnosed in early life, some in later life, and some never at all. The delay in receiving a correct rare disease diagnosis is often referred to as the '*diagnostic odyssey*'. The phrase is used to describe what can be a long and burdensome process from a person's first symptoms all the way to a final, accurate diagnosis. Patients must navigate the system through primary care, specialist care and testing to reach their diagnosis before they can begin the next phase of treatment and condition management.



The diagnostic odyssey is well-documented and the phrase resonates with many in the rare disease community. Indeed, the UK Government is currently commissioning research on how best to measure the diagnostic odyssey, with input from the rare disease community, signalling the widespread recognition of the patient experience.⁴

However, one SHCA member reflects from personal experience that seeking a rare disease diagnosis may be more accurately described as a *labyrinth*: a complex maze filled with wrong turns and dead ends which is challenging to navigate. An odyssey is a story with a beginning, middle and end. For many people living with a rare disease, their journey to a diagnosis is not so linear and may never reach its conclusion.

Patients report numerous barriers and dead ends as they navigate the diagnostic labyrinth, including:

Not being understood by healthcare professionals: our SHCA member charities report that this is one of the most significant causes of delays to diagnosis. Patients report that some healthcare professionals can be sceptical about their experiences, or may dismiss their symptoms as anxiety or other mental health issues. Healthcare professionals in primary care, especially GPs, provide the gateway into the system and are often the first stepping stone in a patient's journey to their diagnosis. As there are over 7000 rare diseases, healthcare professionals cannot be expected to have knowledge of each and every one; this makes listening to patients and trusting when they say something is not right all the more important.

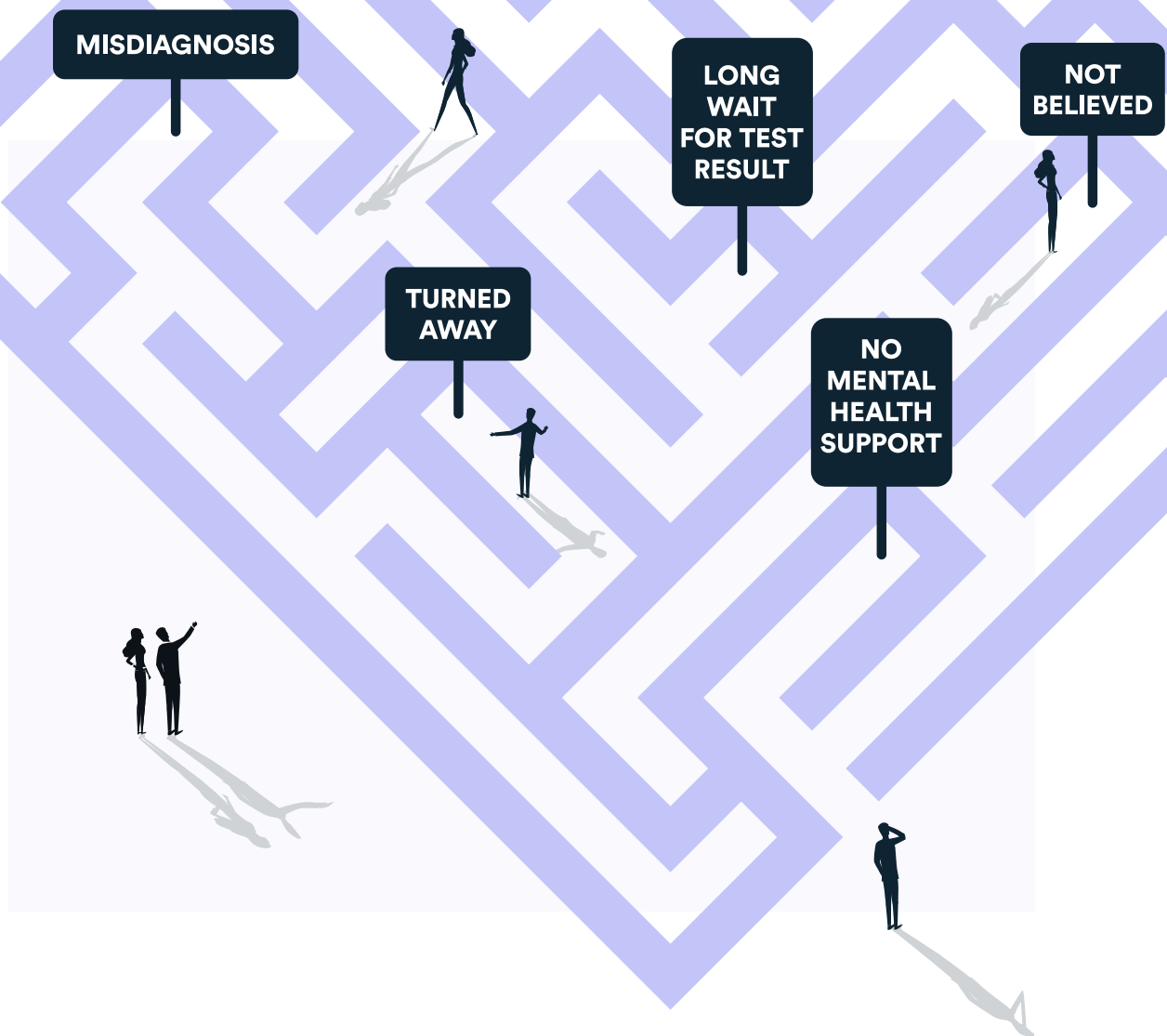
"I spent eight months trying to be diagnosed by my GPs under the NHS, but none of them seemed to look at my symptoms as a whole to find the cause, they only treated individual symptoms. Unfortunately, due to it being a rare condition, the GPs didn't believe me, and thought I was a paranoid female. It was only until I found a private [specialist] who finally listened that I was diagnosed." (Quote from an SHCA charity member)

Limited knowledge of rare diseases and their symptoms amongst GPs and other healthcare professionals: satisfaction with the information that patients are given by healthcare professionals is at its lowest when patients are waiting for a diagnosis, with less than a third of patients satisfied with what they were told.⁵ Patients can spend years stuck in limbo when their symptoms are not recognised or researched, which delays the referral process to a rare disease specialist. Wider system pressures on the NHS can limit the time and resources that healthcare professionals in primary care can dedicate to investigating unknown or unusual symptoms.

"I have been under investigation for [a rare disease] for around two years now, but it seems the various consultants I've seen so far lack enough familiarity or experience with the condition to help me. My life has been turned upside down with no finish line in sight." (Quote from an SHCA charity member)

Delays in accessing testing: when patients are either not understood by a healthcare professional or their symptoms are not recognised, this can delay access to testing. Testing is a crucial part of many people's diagnostic journey, especially for genetic rare diseases, even if this means ruling out certain conditions rather than arriving at a final diagnosis. Patients report struggling to get referred for testing in a timely fashion and feeling frustrated when the timeline for testing is not communicated with them effectively.⁵

"We became concerned about [our child] in the middle of the pandemic and everything felt really slow. We didn't see anybody in person to start with and when we eventually did, the paediatrician said she wanted to 'hold off' on genetic testing as there was no reason to worry us at this point. It became increasingly apparent, though, that things were not right... He is our second child and we just knew there was something seriously wrong. When we eventually had genetic testing, [he] was almost 4 and I felt I could not go on when they told us he had [a terminal rare disease], he was going to die... We lost around 10 months when the paediatrician wanted to 'watch and wait' which I don't think I will every forgive her for." (Quote from an SHCA charity member)



Lack of support while waiting for a diagnosis: patients may feel forgotten or lost in the system while they wait, especially those who spend many years seeking their diagnosis or never receive one at all. Knowing that something is wrong with your health, but not getting answers about what the cause could be, can be very distressing. Patients report that the diagnostic process can be disjointed and fragmented, without coordination or oversight by designated healthcare professionals to guide them through the next steps.⁵ This includes a lack of support for symptom management and mental health.

*“It was just before [our child’s] 4th birthday when we were told it was a terminal condition and our world fell apart. **The emotional rollercoaster of knowing something is wrong but waiting to find out what is so hard and we kept holding on to hope. It is hard to have hope with this diagnosis, but we are very grateful that there is a treatment.**” (Quote from an SHCA charity member)*

CASE STUDY

One SHCA member shared their story of their rare disease diagnosis. As a former GP, they were aware of the possible significance of their symptoms. They rang a specialist centre directly, who validated their concerns and told them to ask their GP to refer them. A patient without a medical background would have been most unlikely to be able to do this, highlighting how difficult it can be for patients and their families to access a correct diagnosis when the condition is very rare.



The cost of waiting

Each and every day spent waiting for a rare disease diagnosis matters. A long delay for the correct rare disease diagnosis comes at a significant cost – to the individual, their loved ones, and the NHS.

Physical health: delays to a rare disease diagnosis means a delay in a patient beginning treatment and accessing the right support to manage their condition. Whilst waiting for a diagnosis, a patient's condition may worsen, or they may be living in significant pain. Receiving a diagnosis in a timely fashion opens doors to treatment, allows patients to manage their symptoms, and ultimately improves outcomes.⁵ For some rare and complex conditions, treatment is only successful if the condition is recognised at an early stage and the patient can begin therapy in a fixed window. In some cases, accelerating the diagnostic process is therefore a matter of life or death.



Emotional wellbeing: waiting for a diagnosis also has an impact on the mental health of patients and their families. Patients report extremely high levels of anxiety as a result of their rare disease, which can result in a misdiagnosis of a mental health condition instead of the correct rare disease.⁶ In one survey, **83%** of patients reported that the process of getting a diagnosis had a negative impact on their mental health; **80%** said that not being believed by healthcare professionals was detrimental to their wellbeing.⁶ Waiting for a rare disease diagnosis, often while a patient's condition is deteriorating, means that patients and their families can be stuck in a limbo-state, where they know something is wrong but are not able to access the right care.



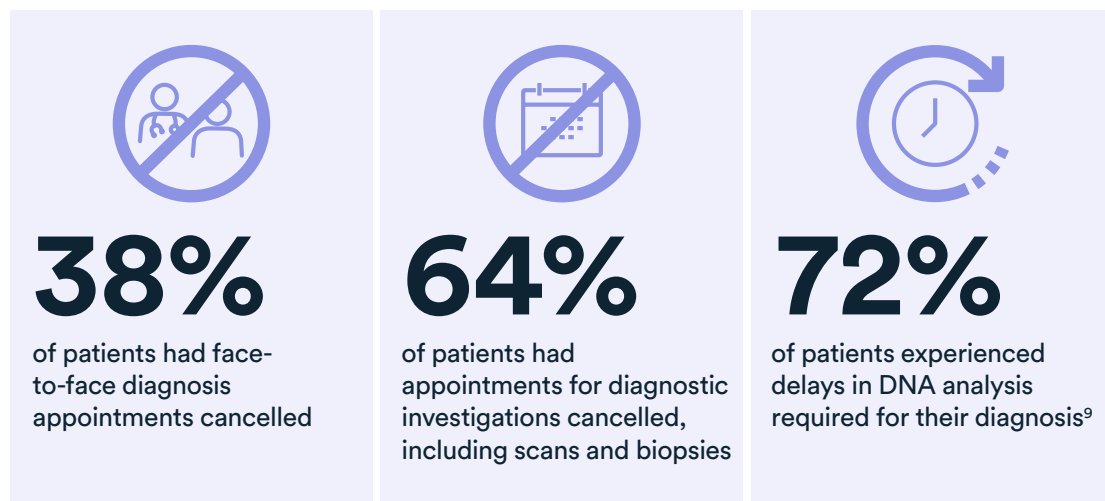
Economic: research shows that the long diagnostic process for rare diseases has cost NHS England over **£3.4billion** over a 10-year period.⁷ This figure encompasses the cost of hospital activity, including admissions, outpatient appointments and emergency care attendances, which is significantly higher than comparable patient populations who also require hospital visits. However, the figure excludes primary care, such as GP appointments, which would likely increase the cost significantly given how long many patients spend in this first setting. Shortening the diagnostic process would benefit not only patients, but also the wider health system.



3. The impact of the pandemic

The COVID-19 pandemic has exacerbated delays to diagnosis for rare disease patients.⁸ System-wide pressures, from workforce capacity to waiting lists, has left no area of the NHS untouched. Disruption across the entire diagnostic pathway – including primary and specialist care, along with testing – has meant that patients are waiting longer for their rare disease diagnosis today than they were before the pandemic.

A survey of patients living with one type of rare disease found that at the height of the pandemic:



Another study found a **36%** reduction in the number of diagnoses during the pandemic, compared to the year before.¹⁰ Patient organisations have also reported a decline in the number of newly diagnosed families requesting support during the pandemic, in one case by over **50%**, indicating a significant drop in diagnosis rates.¹⁰

This drop in diagnosis does not mean there are fewer patients living with a rare disease; rather, there are more patients still waiting for a diagnosis. These missing patients have not disappeared. Three years on from the height of the pandemic, many will still be navigating the diagnostic labyrinth with years potentially added on to their journey. The NHS is still catching up with these missing patients and trying to tackle further delays to a diagnostic pathway where waiting is already the norm.

The pandemic has also had a lasting effect on how patients receive a rare disease diagnosis. An increase in virtual appointments has brought benefits for rare disease patients in some ways, for example by removing the need to travel to primary care settings or hospital appointments where unnecessary.⁵ However, it is important that virtual appointments are not the default. If denied the opportunity to meet with a GP or other healthcare professional face-to-face, patients can find it challenging to communicate their symptoms, be afraid to ask questions, and may be less likely to be understood. Face-to-face contact is as important today as it was before the pandemic for patients seeking and receiving their diagnosis.

CASE STUDY

One SHCA member shared how the COVID-19 pandemic has affected both the patients they support and their work as a rare disease charity.

Since the pandemic, their charity helpline has been receiving significantly more calls from people seeking medical advice about their rare disease, which the charity is not able to provide. Delays in speaking to a GP or a specialist mean that people are turning to the charity sector for medical help, as they feel they have nowhere else to go. This is an indication that the system is not providing the support necessary to help patients navigate the diagnostic labyrinth.

The charity is struggling with the burden of providing support for people who are not able to access appropriate care from the NHS. Staff are receiving challenging call training from helpline specialists given the “traumatic” nature of some of the calls they receive. The pressure is unsustainable.

“The biggest impact [of the pandemic] for us is that patients are turning to a charity for medical information which we simply cannot give them... They are turning to us in their droves because they cannot get that help anywhere else.” (Quote from an SHCA charity member)



4. Opportunities to reduce delays to diagnosis

Context: The UK Rare Diseases Framework and national Rare Disease Action Plans

In January 2021, the Government published The UK Rare Diseases Framework which aims, in part, to improve the timeliness of diagnosis for rare diseases.¹ The Framework is guided by 4 interlinking priorities:

- Priority 1:** helping patients get a final diagnosis faster
- Priority 2:** increasing awareness of rare diseases among healthcare professionals
- Priority 3:** better coordination of care
- Priority 4:** improving access to specialist care, treatments and drugs

Priority 1 directly addresses delays to diagnosis. Central to the Framework is the acknowledgement that many patients face a long journey to a diagnosis, often spanning many years. Getting the right diagnosis in a timely fashion must be improved if patients are to benefit from other pathway improvements further along the line, such as access to specialist care and treatment. Improving delays to diagnosis is therefore a necessary stepping stone to improving outcomes, in line with the broader goals of the Framework.

Priority 2 is integral to helping patients get a final diagnosis faster. The Framework acknowledges what this report has also highlighted: limited awareness of rare diseases among healthcare professionals is a barrier to securing a diagnosis. Healthcare professionals cannot be expected to have knowledge of each and every rare disease. However, given that the first step in a person's journey to a rare disease diagnosis is usually primary care, it is important that GPs and other healthcare professionals are broadly aware of rare diseases and are provided with the education and resources to recognise unusual or unexplained symptoms in patients.

The Framework set out a five-year plan to improve the lives of people with a rare disease. The Framework is being implemented across the UK, with individual Rare Disease Action Plans for England, Scotland, Wales and Northern Ireland setting out specific commitments against each of the 4 priorities.^{11,12,13,14} The next section will explore how these UK-wide and national commitments have the potential to improve the journey to a rare disease diagnosis for patients, and where policymakers could go further.

“Our vision is for rare disease patients across the UK to get a final diagnosis faster and for research into previously unrecognised conditions to identify new rare diseases and provide new diagnoses”
(The UK Rare Diseases Framework)

“Our vision is for healthcare professionals to have an increased awareness of rare diseases and use of genomic testing and digital tools to support quicker diagnosis and better patient care”
(The UK Rare Diseases Framework)

Key opportunities

Testing for rare diseases

Whole genome sequencing

Testing a person's genes is vital in diagnosing a genetic rare disease, which make up **80%** of known rare diseases.¹ In contrast to other kinds of genetic tests, whole genome sequencing (WGS) tests all of a person's genes at one time.

The UK Rare Diseases Framework sets out the benefits of whole genome sequencing: a national testing programme has the potential to transform lives by enabling quicker and more accurate diagnoses for patients.¹ Expanding access to whole genome sequencing for people with a suspected rare disease is a key action in all four of the national Rare Disease Action Plans. For example, the Scottish Government has committed to considering increasing capacity for whole genome sequencing as part of the *Genomics in Scotland* strategy.¹⁵

Notably, NHS England is the first national health system in the world to provide whole genome sequencing as part of routine care, through the NHS *Genomic Medicine Service* (GMS). The GMS was launched in 2018 and rolled out across England from April 2020, with the aim of standardising and integrating whole genome sequencing by 2025. Under the GMS, seriously ill children, children with cancer and adults with certain rare conditions are offered whole genome sequencing. The GMS aims to sequence 500,000 whole genomes by 2023/24, building on the success of the *100,000 Genomes Project* which delivered rare disease diagnoses for previously undiagnosed patients.¹⁶

Whilst the GMS has returned over 1,000 rare disease diagnoses via the Genomics England Clinical Research Interface,⁴ the service has been experiencing worsening backlogs, with reports of patients and their families waiting for over a year to receive their results.¹⁷ There is concern among the patient community that the 500,000 target is unrealistic and the service will require additional funding to tackle these long waits for results.

CASE STUDY

In 2020, NHS Wales became the first country in the UK to introduce rapid whole genome sequencing to diagnose rare diseases in critically ill babies and children.¹ *The Wales Infants' and Children's Genome Service* (WINGS) conducts whole genome sequencing when there is no single genetic test available, or previous diagnostic tests have not led to a diagnosis.¹⁸ Testing can be done in under 48 hours and deliver a rapid diagnosis for families at a time of worry and uncertainty.

Work across the UK to expand whole genome sequencing is essential for reducing delays to diagnosis and ensuring that the first diagnosis a person receives is correct. **Policymakers should be ambitious in rolling out whole genome sequencing to a wider proportion of people with a suspected rare disease – not only seriously ill children – to accelerate diagnosis for all, and ensure that sequencing services are adequately funded to prevent backlogs.**

Newborn screening

Today, newborn blood-spot screening is offered to all babies in the UK at 5 days old. The screening process tests for **9** rare diseases, including cystic fibrosis and sickle cell disease.¹⁹ Screening at this stage is especially important given the prevalence and high mortality rate of rare diseases in young children. Newborn screening is a key tool in reducing delays to diagnosis, leading to earlier and more choice of treatment, and ultimately better outcomes.

However, the UK currently falls significantly behind comparable nations in the number of rare diseases we screen for; many countries in Europe screen for more than **20** conditions, including Italy, Poland and Austria.²⁰ The UK Rare Diseases Framework sets out the need to change this.

The *UK National Screening Committee* (UK NSC), which provides recommendations to the Government and the NHS, is considering the expansion of the UK's newborn blood-spot screening programme. So far, the UK NSC has recommended screening for tyrosinaemia, a very rare genetic condition, and is evaluating the addition of severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA). However, screening for rare diseases can and should only be approved if diagnosis at such an early age would do more good than harm. The value of a rare disease diagnosis where there is either no treatment option or low likelihood of survival carries a huge emotional burden and must be considered sensitively.

Additionally, a research study to be conducted by NHS England and Genomics England could see participation across the devolved nations; the pilot will carry out whole genome sequencing of 100,000 newborns, screening for up to 200 treatable rare diseases where early diagnosis has the potential to transform outcomes.⁴

England, Scotland and Northern Ireland have all committed to expanding newborn screening for rare diseases. **By improving how decisions are made on newborn screening, expanding the number of conditions screened for and learning from successes in other countries, diagnoses can be made earlier where appropriate.**

CASE STUDY

There are many stories of where an expanded newborn screening programme could have made a difference. One such story is three-year-old Nala, who is now terminally ill with metachromatic leukodystrophy (MLD).²¹ A gene therapy for MLD is now available through NHS England, but Nala was diagnosed too late to be treated. Her little sister Teddi was also diagnosed with MLD, and has been able to receive treatment.

MLD is not one of the rare diseases screened for at birth in the UK. However, newborn screening pilots in other countries have identified MLD in newborns, and there are calls to include MLD in the NHS England and Genomics England research pilot. This is just one example of how early diagnosis through newborn screening could grant faster access to treatment and potentially save lives.

Community Diagnostic Centres

The rollout of new community diagnostic centres (CDCs) across England – and similar initiatives in the devolved nations – aims to make diagnostic services faster and more accessible. This presents an opportunity to improve access to diagnostics for people with rare diseases.

The creation of CDCs was recommended following a review in 2020 of NHS diagnostic capacity, in order to meet current and future demands of the NHS.²² CDCs offer diagnostic tests in local communities where services can be accessed conveniently in places such as high streets and shopping centres. A GP, pharmacist or hospital can refer a patient with unexplained symptoms to a CDC, where patients can access a range of diagnostic tests including X-Rays and MRI or CT scans, which can conduct preliminary testing in the rare disease diagnostic process.

CDCs were initially designed to separate diagnostic testing from hospital settings to prevent the spread of COVID-19; they have now been rolled out across England to create a more accessible, robust diagnostic pathway. CDCs have carried out over **3 million** tests across England since their creation, making up **5%** of all diagnostic activity.²³ By 2025, it is hoped that **9 million** tests will be performed each year.²³

Elsewhere in the UK, 3 Rapid Cancer Diagnostic Services (RCDSs) have been established in Scotland,²⁴ whilst Wales is served by the Rapid Diagnosis Clinics (RDC) Programme.²⁵ Both services aim to speed up the diagnostic process for patients with vague symptoms which may be cancer. Most patients referred to the services will be ruled out as having cancer, and may go on to be diagnosed with a rare disease, or alternatively may be found to have a rare form of cancer.

CASE STUDY

A new Community Diagnostic Centre in Welwyn Garden City can test patients for liver fibrosis, which includes a rare condition called congenital hepatic fibrosis (CHF).²⁶ The liver FibroScan, a machine which can painlessly diagnose significant liver damage in minutes, has been installed at the QEII Hospital CDC. This specialist technology, available within a local community, could help to diagnose the rare liver disease.

For rare disease patients, CDCs and other similar initiatives should provide a quicker and more accessible avenue for testing. Access to testing in local communities can reduce the distance people have to travel, and also benefit the wider system by freeing up hospital capacity elsewhere.

To better understand how CDCs are meeting the needs of rare disease patients, there is an opportunity to collect and publish more data about whether community testing is translating into faster and more accurate diagnoses. This includes information on how many people with a suspected rare disease are being referred to CDCs for testing and how many diagnoses have been delivered. **CDCs and other testing initiatives have the potential to speed up rare disease diagnosis, including by ruling out other conditions, but more data is needed to understand their direct impact so far.**

Support for undiagnosed patients

Whilst advancements in testing can deliver a quicker and more accurate diagnosis for many patients, it is important that people who spend many years waiting for a diagnosis, or are never diagnosed, are not forgotten.

The England Rare Diseases Action Plan for 2022 acknowledges the importance of supporting people living with undiagnosed rare diseases and signposting them to appropriate services.¹¹ Action 5 of the Plan is dedicated to piloting new approaches for undiagnosed rare diseases, through outpatient appointments, inpatient stays and assessment by multiple clinicians. Bringing together healthcare professionals across the pathway, from testing to mental health support, can ensure that people awaiting their diagnosis are supported by the system.

In the England Rare Diseases Action Plan for 2023, the Government confirmed that a proposal for a pilot *syndrome without a name* (SWAN) programme has been developed.⁴ The pilot aims to reduce delays to diagnosis for patients with undiagnosed rare diseases, and support those for whom a diagnosis may never come. The SWAN pilot will be implemented in England in 2023 following funding agreement.

SWAN clinics can provide much-needed co-ordination and oversight of a patient's diagnostic journey. The vast majority of people living with a rare disease do not have an appointed care co-ordinator who can help them navigate the system, leaving many patients feeling lost and missing opportunities for appointments or testing.^{3,5} SWAN clinics can give patients and families the co-ordinated support and management of their diagnosis that they need to take some of the burden away.

CASE STUDY

The UK's first specialist SWAN clinic opened at the University Hospital of Wales in 2022 as a two-year pilot.²⁷ Designed to be a 'one-stop shop' for diagnosis, the clinic aims to reduce delays to diagnosis and improve pathways for people living with rare, undiagnosed conditions. The SWAN clinic is supported by rare disease charities and is already offering hope to patients and their families across Wales.²⁸

If SWAN pilots in England and Wales are successful, similar schemes should be adopted more widely across the UK to ensure that undiagnosed patients are able to access the care and support they need. SWAN clinics must be Government-funded and receive adequate support to operate effectively. Partnerships with charities, who have years of expertise in supporting undiagnosed patients, can further bolster their success. **Pilot SWAN clinics present a real opportunity to reduce the burden of waiting for many years for a diagnosis or living with an undiagnosed rare disease.**

Awareness among healthcare professionals

Wider awareness of rare diseases among healthcare professionals is crucial in improving the speed and accuracy of diagnoses. As a key priority in The UK Rare Diseases Framework, the theme of awareness is embedded in each national Rare Disease Action Plan.

As healthcare professionals and GPs cannot be expected to have knowledge of each and every rare disease, it is important that they are aware of rare diseases more broadly and recognise when it is appropriate to make a referral to specialist, especially in primary care. The opportunities to improve awareness of rare diseases among healthcare professionals, and therefore reduce delays to diagnosis, fall under four branches:

- **Education:** awareness can be improved by embedding training on rare diseases within the medical school curriculum. This is particularly important for primary care as the first stepping stone in the pathway to a rare disease diagnosis. Each Rare Disease Action Plan contains specific commitments on identifying current training gaps and improving education to promote a better understanding of rare diseases. For example, Health Education England (HEE) is reviewing current curricula and will support the creation of new training resources.¹¹ Upskilling the workforce – including patients' primary points of contact in GPs, midwives and nurses – to identify a wider range of rare disease symptoms across the care pathway is crucial for enabling earlier diagnosis.
- **Information:** it can be challenging for healthcare professionals to navigate the breadth of information about rare diseases online. Healthcare professionals should therefore be equipped with the tools to help them find the relevant information they need. One such example is GeNotes, a digital resource created by HEE to provide healthcare professionals with concise clinical information about genomics at each stage of the rare disease pathway.¹¹ There is a need for a similar tool, designed specifically for diagnosis, to help healthcare professionals recognise symptoms and decide when to make referrals. This tool should house accurate information on diagnosis in one place, including e-learning resources - many of which already exist and have been designed by patient organisations specifically for this purpose.
- **Communication:** our SHCA member charities have reflected that poor communication from healthcare professionals can cause delays in patients accessing charity support. In some cases, patients are not being told enough information about their rare disease and are not being signposted to the correct place to receive charity support; for example, where a condition is so rare that it falls under an umbrella term, some patients have not been informed what this broader category is, and have not been able to find the relevant patient organisation. By providing healthcare professionals with up-to-date lists of registered rare disease charities and information about the kind of support on offer, they can signpost patients to the crucial non-medical support offered by the charity sector.
- **Belief:** efforts to heighten awareness of rare diseases amongst healthcare professionals will only be meaningful if coupled with a change in how patients are treated when they are seeking a diagnosis. Healthcare professionals should receive training on the importance of listening sensitively to patients and their families, and how the physical and emotional impact of navigating the diagnostic labyrinth may affect people differently.

Policymakers must ensure that efforts to improve awareness of rare diseases among healthcare professionals is holistic, encompassing not only how to recognise symptoms, but also how to communicate with and listen to patients and their families.

Mental health support during the diagnostic journey

There is a growing understanding that mental health support for people living with a rare disease is fundamental to their care. Specialist mental health support for patients and their families while they navigate the diagnostic journey should be integrated into their care, not seen as an additional service. However, many patients and families are not able to access this support when they need it, and the support offered is not always helpful.⁶

Our SHCA member charities have reflected that mental health provision is missing from the diagnostic pathway, especially for progressive conditions when a patient's health may deteriorate rapidly while they are still seeking their diagnosis. Our members also observed that patients are subject to an inequitable postcode lottery in the quality and accessibility of mental health care. Access to high-quality, specialist mental health support should not depend on where someone happens to live.

There are opportunities to improve mental health provision for people seeking a rare disease diagnosis. In each of the national Rare Disease Action Plans, mental health provision is identified as an area for improvement, where support should be integrated within the wider rare disease pathway. However, the actions in the Rare Disease Action Plans are primarily focused on mental health support for diagnosed patients, who are able to access treatment and care pathways. It is important that mental health support is also available during the diagnostic journey.

Seeking a rare disease diagnosis is a complex process with many challenging emotions attached. Psychological support for patients and their families navigating this process should be offered, rather than needing to be sought out; the support should also be delivered by mental health professionals who understand the impact of living with a rare disease on a person's emotional wellbeing. Access should be equitable across the UK to remove the postcode lottery which allows some patients to fall through the cracks. **Efforts to integrate mental health support into rare disease care pathways are welcome, but policymakers must ensure that this support is available to patients and their families whilst seeking a diagnosis, not just from the point of diagnosis onwards.**

A blueprint for the future of rare disease diagnosis

Decisive and rapid action by policymakers is needed across all four areas – testing, support for undiagnosed patients, awareness among healthcare professionals and mental health support – for maximum impact. The recently announced *Inherited White Matter Disorders (IWMDs) Diagnostic and Management Service* could potentially provide a blueprint for the future of rare disease diagnosis.

The new IWMDs service was announced by NHS England in March 2023 to fast-track patients with a range of rare diseases (known as leukodystrophies) to diagnosis.²⁹ The new service will review more than **300** patients each year and aims to speed up diagnosis and therefore ensure the best possible quality of life for patients living with often degenerative and life-limiting conditions.

The new service will provide access to the following initiatives:

- **Specialist multi-disciplinary teams:** the service will grant patients rapid access to a specialist multi-disciplinary team, who can guide patients through the diagnosis process and support with symptom management. Patients will be offered virtual support where appropriate to reduce unnecessary travel to face-to-face appointments, and will be able to access local support from nearby clinics.
- **Genetic testing:** to enable faster diagnosis, the service will carry out genetic testing through new national genetics laboratories, with specific expertise in sequencing IWMDs.
- **Clinical registry:** the service will register patients with a new IWMD clinical registry, which will create opportunities for clinicians to learn more about the conditions and identify patients for clinical trials.

Should the service be successful in this specific disease area, it could provide a blueprint for transformation across the whole rare diseases landscape. The new service directly addresses some of the barriers to rapid and accurate diagnosis that this report has set out.

The service can harness opportunities for change by treating diagnosis as a part of the rare disease pathway, where professional support is required not from the point of diagnosis onwards, but much earlier from symptom onset. The NHS should explore whether the IWMDs service can be developed into a model for use in other rare diseases across the UK.

5. Our recommendations

The journey to a rare disease diagnosis can be described in many ways: an odyssey, a labyrinth, a burden, a battle. When we listen to the patient community, their experiences reflect the length of time and significant effort it takes to secure a final, accurate diagnosis.

There are many opportunities to reduce delays to diagnosis in the UK, which this report has outlined. The UK Rare Diseases Framework and the accompanying national Rare Disease Action Plans rightfully place diagnosis at the heart of what needs to change to improve lives. In alignment with, and in addition to, these commitments, we recommend the following steps should be taken by policymakers to reduce current delays to an accurate rare disease diagnosis. These recommendations should be considered as part of ongoing work to develop future action plans across England and the devolved nations.

Testing for rare diseases

Advancements in testing mean that diagnosing a rare disease should be quicker and more straightforward than ever. Across the UK, we must harness these opportunities in genetic and community testing to reduce delays to diagnosis.

- 1 The NHS in England and the devolved nations should commit to nationally rolling out successful pilot schemes for rare disease testing, including rapid whole genome sequencing services and newborn screening, with new targets for adoption
- 2 Data should be collected on delays to diagnosis for rare disease patients, including through collecting and publishing data to better understand how community diagnostic centres (CDCs) are meeting the needs of rare disease patients in England

Support for undiagnosed patients

It is important that people who spend many years waiting for a diagnosis, or are never diagnosed, are not forgotten.

- 3 The NHS in England and the devolved nations should open further *syndrome without a name* (SWAN) clinics across the UK for patients living without a diagnosis over the next five years, following a successful pilot scheme in Wales and work to establish the first clinic in England. Government funding for these further SWAN clinics should be confirmed immediately
- 4 SWAN clinics should designate a diagnostic co-ordinator to patients, who can liaise between the healthcare professionals involved in a patient's diagnostic journey and help them navigate the pathway

Awareness among healthcare professionals

As there are over 7,000 rare diseases, healthcare professionals cannot have knowledge of each and every one; this means that information about rare diseases and their symptoms must be easy to find.

- 5 The NHS in England and the devolved nations should create a digital tool for healthcare professionals specifically for rare disease diagnosis – one place where they can access information, receive answers to questions from other clinicians, and find and utilise e-learning resources created by patient organisations
- 6 As part of ongoing reviews into rare disease modules in medical curricula, the NHS in England and the devolved nations, and relevant medical schools, should ensure that healthcare professionals are trained not only in how to recognise rare disease symptoms, but how to engage with patients and families with sensitivity
- 7 NHS England should update the online directory for medical conditions to include links to registered charities for each rare disease, to help patients reach the correct support and enable healthcare professionals – especially in primary care – to signpost patients to the right place

Mental health support during the diagnostic journey

Specialist mental health support for patients and their families while they navigate the diagnostic journey should be integrated into their care, not seen as a separate service.

- 8 As part of the integration of mental health support into rare disease care pathways, the NHS in England and the devolved nations should ensure that mental health support is offered to patients and their families whilst seeking a diagnosis, not just from the point of diagnosis onwards
- 9 The NHS in England and the devolved nations should collect and publish data to determine whether mental health support for patients seeking a rare disease diagnosis is available equitably across the UK

A blueprint for the future of rare disease diagnosis

NHS England's *Inherited White Matter Disorders (IWMDs) Diagnostic and Management Service* could potentially provide a blueprint for the future of rare disease diagnosis, through direct access to specialist multi-disciplinary teams and genetic testing.

- 10 NHS England should carry out an assessment of learnings from the implementation of the *Inherited White Matter Disorders (IWMDs) Diagnostic and Management Service*, with a view to identifying whether the service could be developed into a model for use in other rare diseases

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Specialised Healthcare Alliance

FOR EVERYONE WITH RARE AND COMPLEX CONDITIONS

About the SHCA: The Specialised Healthcare Alliance (SHCA) is a coalition of over 130 patient groups organisations and charities, who support people across the UK living with rare and complex conditions. Our work is funded by 9 corporate supporters. We advocate for better services for patients, keeping rare diseases at the forefront of the national conversation. The SHCA focuses exclusively on overarching policy and does not address individual therapeutic issues. The SHCA has no political affiliation and seeks to work across party lines.

For more information about the work of the SHCA, please visit www.shca.info or email SHCA@incisivehealth.com