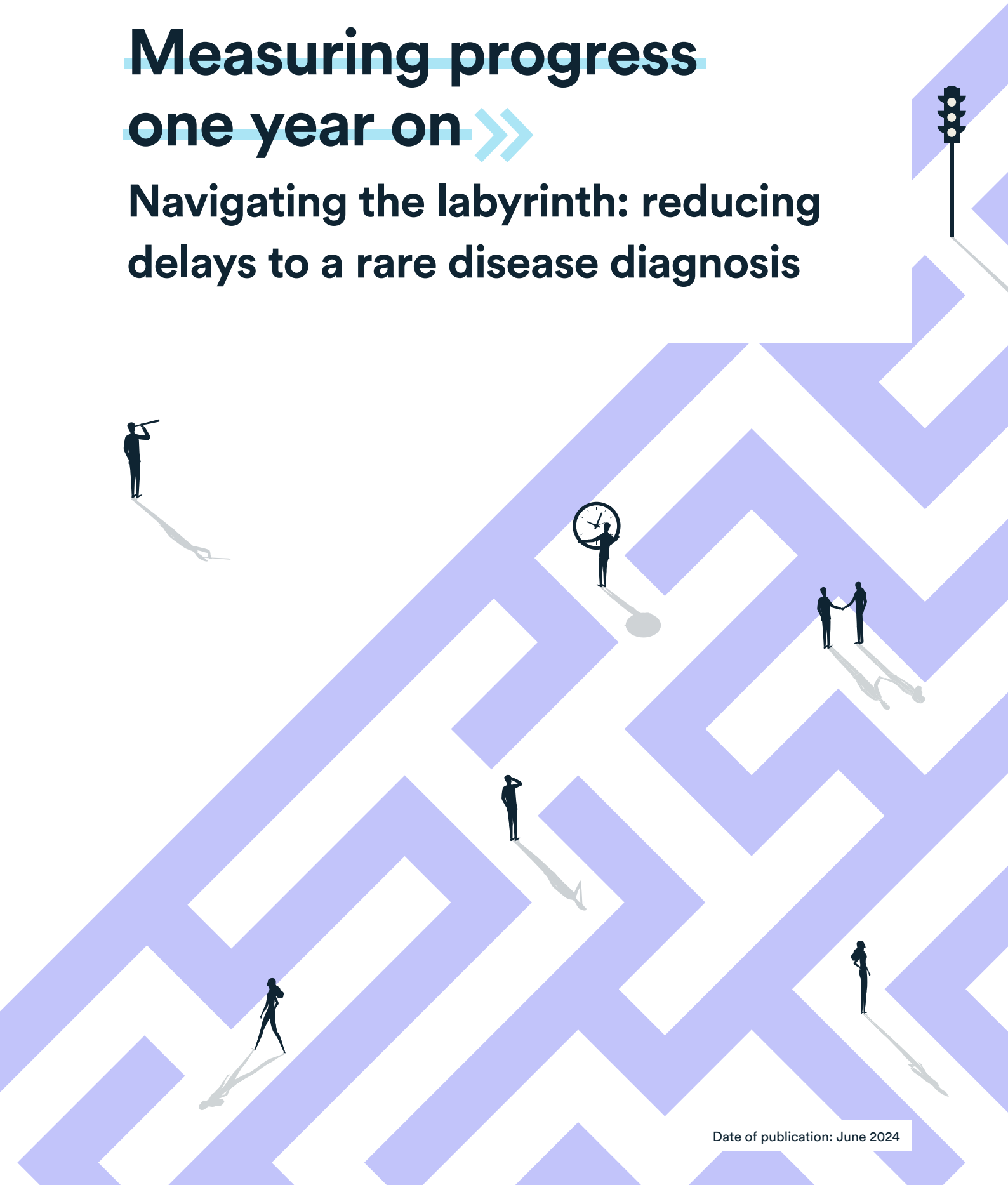
Specialised Healthcare Alliance
FOR EVERYONE WITH RARE AND COMPLEX CONDITIONS

Measuring progress one year on

Navigating the labyrinth: reducing delays to a rare disease diagnosis



Date of publication: June 2024

Introduction

A diagnosis of a lifelong – and sometimes life-limiting – rare or complex 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 earlier as they navigate the diagnostic labyrinth, filled with twists, turns and dead ends.

In June 2023, the Specialised Healthcare Alliance (SHCA) published our report, *Navigating the labyrinth: reducing delays to a rare disease diagnosis*, which explored the barriers to a diagnosis and the opportunities presented by the policy landscape to transform the diagnostic process for the better. A year on, this briefing examines the degree of progress made against the five key opportunities we identified:

Click here to read our original report

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**

The wider health landscape in 2024

The NHS is facing significant pressures across the whole system, not limited to rare diseases. Capacity continues to struggle to keep up with demand across both primary and specialist care, with an understaffed workforce having to work overtime to keep services afloat. Measures to increase NHS productivity in the last year include the expansion of Community Diagnostic Centres (CDCs) to speed up diagnosis across 155 sites in England,[1] and expanding the prescribing powers of pharmacies to free up primary care capacity.[2] However, recovery efforts from the effects of the COVID-19 pandemic are still falling short, with people waiting longer than ever in England to access care.[3]

Measuring progress

Since the publication of our *Navigating the labyrinth* report in June 2023, governments in England, Wales and Northern Ireland have published updated Rare Disease Action Plans to monitor progress against their key commitments. Each national Action Plan is published against the backdrop of the *UK Rare Diseases Framework*, which set out overarching ambitions over a five-year period in 2021. These Action Plan updates are as follows:

England: *Rare Diseases Action Plan 2024* (February 2024)

Wales: *Refresh of the Rare Disease Action Plan 2022 to 2026* (December 2023)

Northern Ireland: *Rare Diseases Action Plan Progress Report – Year 1* (September 2023)

Scotland: No update to the 2022 *Rare Disease Action Plan* published yet – expected in 2024

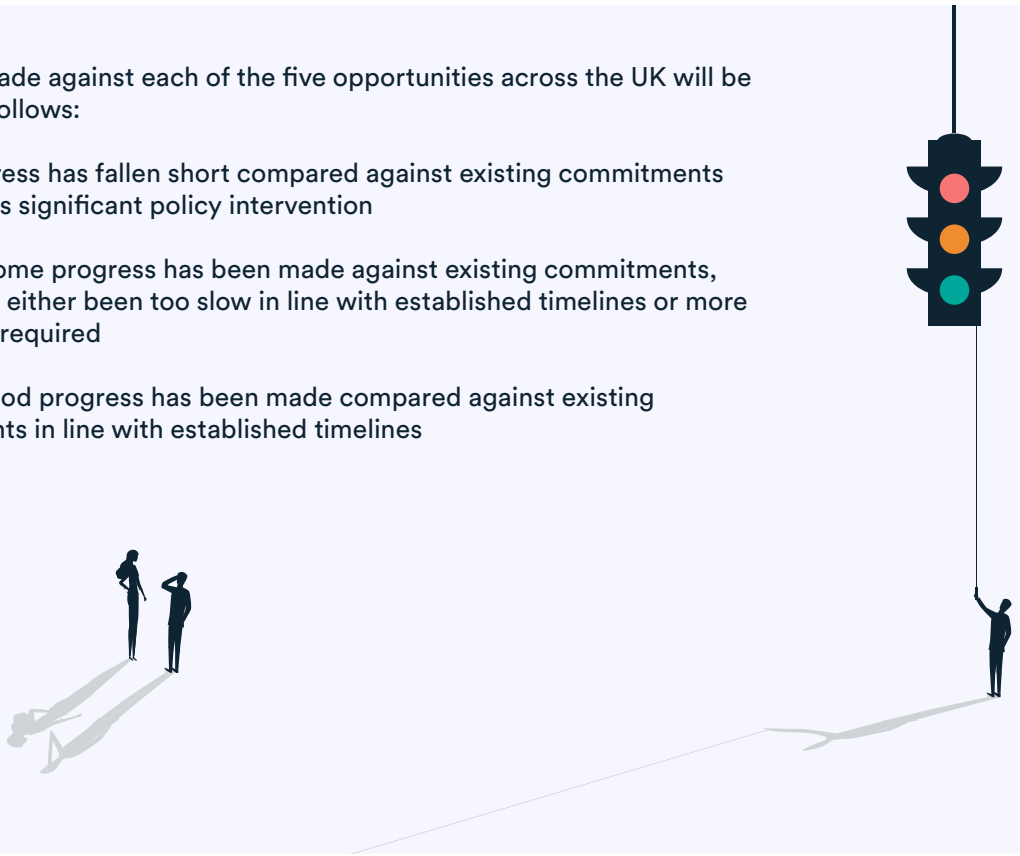
One of the four main priorities of the *UK Rare Diseases Framework* is to help patients get a diagnosis faster. This briefing will assess the progress made by each of the devolved nations against this priority as set out in the Action Plan updates.

Progress made against each of the five opportunities across the UK will be graded as follows:

Red: progress has fallen short compared against existing commitments and requires significant policy intervention

Amber: some progress has been made against existing commitments, but this has either been too slow in line with established timelines or more ambition is required

Green: good progress has been made compared against existing commitments in line with established timelines



Testing for rare diseases

Progress rating: Amber

Advancements in testing mean that diagnosing a rare disease should be quicker and more straightforward than ever. Innovations in newborn screening and whole genome sequencing offer real hope for people with a suspected rare disease, but the UK has been slow in adoption. It is important that testing is widely and equitably accessible to ensure people receive a timely diagnosis.

Recommendation 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

Recommendation 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

Progress made	Progress delayed or not made
In England, key progress made in newborn screening includes the positive recommendation of several new conditions for screening by the UK National Screening Committee (NSC), as well as the publication of a paper comparing the principles of newborn screening in the UK and Europe to bring us into greater alignment. **Progress has also been made in whole genome sequencing as part of the Generation Study,** which is aiming to sequence the genomes of over 100,000 babies and gather evidence to consider whether whole genome sequencing could be rolled out as part of a future newborn screening programme. All milestones have been met to date, with an initial list of over 200 rare diseases caused by variations in over 500 genes to be included when the study begins. It is positive that Genomics England is committed to sharing findings with the charity sector and partners as the study progresses. **With regard to data, NHSE published Genomic Testing Service data for the first time and will continue to do so on a quarterly basis,** with the remit expanded to include a greater focus on turnaround times and access to testing within different communities. CDCs have now delivered over 7 million tests across 160 sites in England as part of wider diagnostic activity. In Wales, the collection of rare disease data at a national and local level is being considered. In Northern Ireland, a review of care pathways has been undertaken focusing on time to diagnosis. In Scotland, the Scottish Strategic Network for Genomic Medicine (SSNGM) has been established to provide a streamlined genomics service.	**However, there have been several significant delays for key actions in England.** Work to improve the evidence base to support the UK NSC make robust decisions about newborn screening for rare diseases has been delayed due to resourcing constraints, meaning some action-specific monitoring and evaluation targets will be carried forward into this year. Furthermore, the commitment to commission research on how best to measure the diagnostic odyssey (Action 17) has faced delays; a contract for the research has only recently been agreed and the project is expected to run to 2026. Additionally, although CDCs are delivering large numbers of tests across England, without specific data published on the number of rare disease diagnoses made as a result, it is unclear to what extent people living with rare diseases are benefitting. In Northern Ireland, the review of care pathways has been limited to specific conditions and the outcome of the review remains unclear. 

Support for undiagnosed patients

Progress rating: Green

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 – especially given that more than 20% of rare diseases do not have a known genetic cause. Syndrome without a name (SWAN) clinics can provide much-needed co-ordination and oversight of a patient's diagnostic journey and should be rolled out across the UK.

Recommendation 3: The NHS in England and the devolved nations should open further 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

Recommendation 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

Progress made	Progress delayed or not made
In Wales, the UK's first SWAN clinic has successfully assessed 105 referrals from across Wales as of January 2024, with its funding extended to March 2024. Progress has been made in developing the clinic, including specialist nurses supporting patients and their families around appointments, creating individualised care plans, partnering with local charities, and planning debriefing appointments for patients with genetic counsellors. **The clinic has also designated two Clinical Nurse Specialists (CNSs) as care co-ordinators for patients.** Wales has also committed to an additional action supporting diagnosis of non-genomic conditions in recognition that advancements in genomic testing will not help everyone. **In England, a service model has been agreed and funding secured to pilot two SWAN clinics – one for children and one for adults.** NHSE has utilised learnings from Wales in designing the pilot, with sites to be selected in 2024.	As the pilot period for the Wales SWAN clinic comes to an end – originally designed to be two years – **Government commitment is vital for long-term funding and staffing to continue the work of the clinic and assess learnings to date.** For England, **it is important that progress is made at a good pace in establishing the clinics**, with further communication with those behind the Welsh clinic to share learnings. NHSE should share public updates on developments and timeframes to ensure transparency. Scotland and Northern Ireland should consider the establishment of a SWAN clinic for their populations. 

Awareness among healthcare professionals

Progress rating: Amber

Healthcare professionals cannot be expected to have knowledge of every rare disease. It is therefore vital that they have access to information and educational resources to help recognise symptoms, decide when to make referral to a specialist, and navigate difficult conversations with patients sensitively. Some patients report feeling confused or not believed during their interactions with the health system, especially those who wait many years for a diagnosis, so it is important that healthcare professionals are equipped with accessible information to support patients and signpost them to other services.

Recommendation 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

Recommendation 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

Recommendation 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

Progress made	Progress delayed or not made
In England, progress has been made in the development of the GeNotes online tool designed to provide healthcare professionals with information and education about rare diseases. GeNotes covers both genetic and non-genetic conditions and has been launched in seven specialities, with five more to follow. GeNotes is being further embedded into primary care by guiding GPs on when to consider genomic testing. Work on the Rare Disease Education Hub, which includes information on non-genetic rare diseases, has also continued. **As part of efforts to improve medical education**, the National Genomics Education Programme (NGE) has published a genomics toolkit for nurses, with a similar toolkit for midwifery to follow, as well as launching a massive open online course (MOOC) with patient and expert interviews. In Wales, actions include surveying qualified healthcare professionals and undergraduates on their learning needs, and incorporating a rare diseases module in the undergraduate curriculum for medical students. In Northern Ireland, there is also ongoing work to include rare disease content in the medical curriculum on a long-term basis, as well as webinars and workshops to enhance understanding. In Scotland, a survey of healthcare professionals has been conducted by Genetic Alliance UK with support from the Office for Rare Conditions, to provide insight into perceptions and knowledge of rare conditions. Work has also been undertaken with the NHS to develop a public-facing information page on rare conditions to signpost people to relevant charities.	Progress has clearly been made in this area, but **there is more work to be done to ensure that healthcare professionals across the UK receive training on rare diseases as standard,** permanently built into the medical curriculum, rather than needing to seek it out. This education should specifically cover the diagnostic odyssey and the process of making referrals to specialists. Training should be extended to all relevant healthcare professionals beyond doctors or GPs, including nurses and midwives. **There is also a need for greater collaboration between the NHS and the charity sector** to ensure that people are signposted to helpful resources or services during the diagnostic process. 

Mental health support during the diagnostic journey

Progress rating: Red

Mental health support is an unmet need for people navigating a rare disease diagnosis. In 2024, the SHCA surveyed our charity members on mental health and published the findings in a report with policy recommendations. Most notably, 72% of respondents said that mental health support is poorly integrated into wider care pathways for people living with a rare disease, whilst 91% said the charity sector is under significant strain in having to offer mental health support which should be provided by the NHS.

Click here to read our mental health report

Recommendation 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

Recommendation 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

Progress made

In England, the most significant development is that **all new and revised service specifications for rare diseases published by NHSE are now required to consider people's psychosocial needs and ensure co-ordinated pathways for access to mental health support.** As part of this process, NHSE's Genomics Unit will work with mental health commissioners to explore options to increase access to clinical psychology for genomics services over a four-year period. NHSE has also committed to encouraging healthcare professionals working in genomics services to look for points across the patient journey when mental health input is required. These actions signify a promising step forward in recognising the importance of integrating mental health support into wider care, from the point of diagnosis.

Some progress has also been made in Northern Ireland in aligning actions with the national mental health strategy, including the implementation of a Regional Mental Health Service and a review of how mental health impacts people diagnosed with a rare disease using data from the UK Biobank.

Progress delayed or not made

Further progress is being hindered by slow timelines and is falling short of full integration of mental health support. Action 20 for England concerns commissioning research to improve co-ordination of care in the NHS. It has now been confirmed that this review will specifically address integrating mental health support into rare disease clinical care; however, the contract for the research is still being agreed and the project is expected to run to 2026. The outcomes of the research when published will "support decisions on how best to implement cost-effective care co-ordination in the NHS", but this is not a firm commitment.

Additionally, NHSE has written to Integrated Care Boards and providers to emphasise that "mental health services should be offered based on need and should not exclude anyone" because of their diagnosis. **However, efforts to mitigate exclusion of people with rare diseases is not equal to proactively offering mental health support during the diagnostic process.**

Similarly in Northern Ireland, the Progress Report is clear that their commitment to mental health "does not mean the provision of dedicated mental health resources within physical health services, but rather the creation of effective pathways to allow individuals to access specialist support", which **falls short of proper integration.**

A blueprint for the future of rare disease diagnosis

Progress rating: Amber

Our report identified NHSE's Inherited White Matter Disorders (IWMDS) Diagnostic and Management Service as a potential blueprint for other rare diseases. The service, based in London but also offering virtual support, was established in 2023 and aims to fast-track patients with a range of rare diseases (known as leukodystrophies) to a diagnosis. The service provides access to genetic testing, specialist multi-disciplinary teams, and a clinical registry for healthcare professionals.

Recommendation 10: NHS England should carry out an assessment of learnings from the implementation of the 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

Progress made	Progress delayed or not made
The IWMDS service has been successfully established, with 400 patients in touch with the service at any given time. Notably, the service was developed in collaboration with patients and charities, which is vital in creating a patient-centred model of care. The service is staffed by a range of specialists including neuropsychologists, nurses, and consultants in neurology, metabolics, clinical genetics and neuroradiology; it offers holistic support beyond treatment, including advice on genetics, condition management, and life planning.[4] **As a result of the success of the service, new and revised service specifications for rare disease published by NHSE have built on the success of the IWMDS service model in promoting care co-ordination.** This includes a requirement for specialist hubs to advise and support local services. NHSE has also published guidance for similar clinics providing virtual support patients with rare or complex multi-system disorder.[5]	**NHS England has not yet published a specific assessment on the learnings from the IWMDS,** but it is promising to see the model already being rolled out to other rare diseases. NHSE describes the service as "a pioneering [...] future-facing model of care", signifying ambitions for applying the successes even more widely.[6] **The devolved nations should also look to adopt this model in their health systems.** 

Where do we go from here?

Our progress report shows that reducing delays to a rare disease diagnosis is possible through dedicated policy effort. Some milestones have been met across the UK in the past year against existing commitments. However, this report also highlights how much more there is still to be done to help people navigate the diagnostic labyrinth. Progress has been too slow or hindered altogether in places as a result of limited funding and resources. Although budgetary challenges are significant for the NHS, diagnosing rare diseases earlier can reduce the strain on the wider health system and free up both capacity and resources, particularly in primary care, as people are put onto the right pathway.

From advancements in genomics to new models of patient-centred services, there are clear opportunities for the next government to capitalise on. The next government must recommit to prioritising rare diseases and accelerate progress on existing commitments, as well as setting out their vision for the future. **For the 1 in 17 people living with a rare disease across the UK,[7] action to reduce delays to diagnosis cannot wait.**

References

1 UK Government (2024). *Community diagnostic centres deliver more than 7 million checks*.
2 NHS England (2024). *Over 10,000 NHS pharmacies begin treating people for common conditions*.
3 The Guardian (2024). *Almost 10 million people in England could be on NHS waiting list*.
4 UCLH Hospitals (2024). *UCLH and UCL improving care for patients with rare 'white matter' disorders*.
5 NHS England (2023). *Virtual clinics in Highly Specialised Services (HSS): guidance for services supporting patients with rare and complex and multi-system disorders*.
6 NHS England (2024). Rare Disease Day 2024: *harnessing genomic medicine and cutting-edge treatments to improve outcomes for patients with rare conditions*.
7 UK Government (2021). *The UK Rare Diseases Framework*.

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About the SHCA: The Specialised Healthcare Alliance (SHCA) is a coalition of over 130 patient organisations and charities, who support people across the UK living with rare and complex conditions. Our work is funded by 11 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.