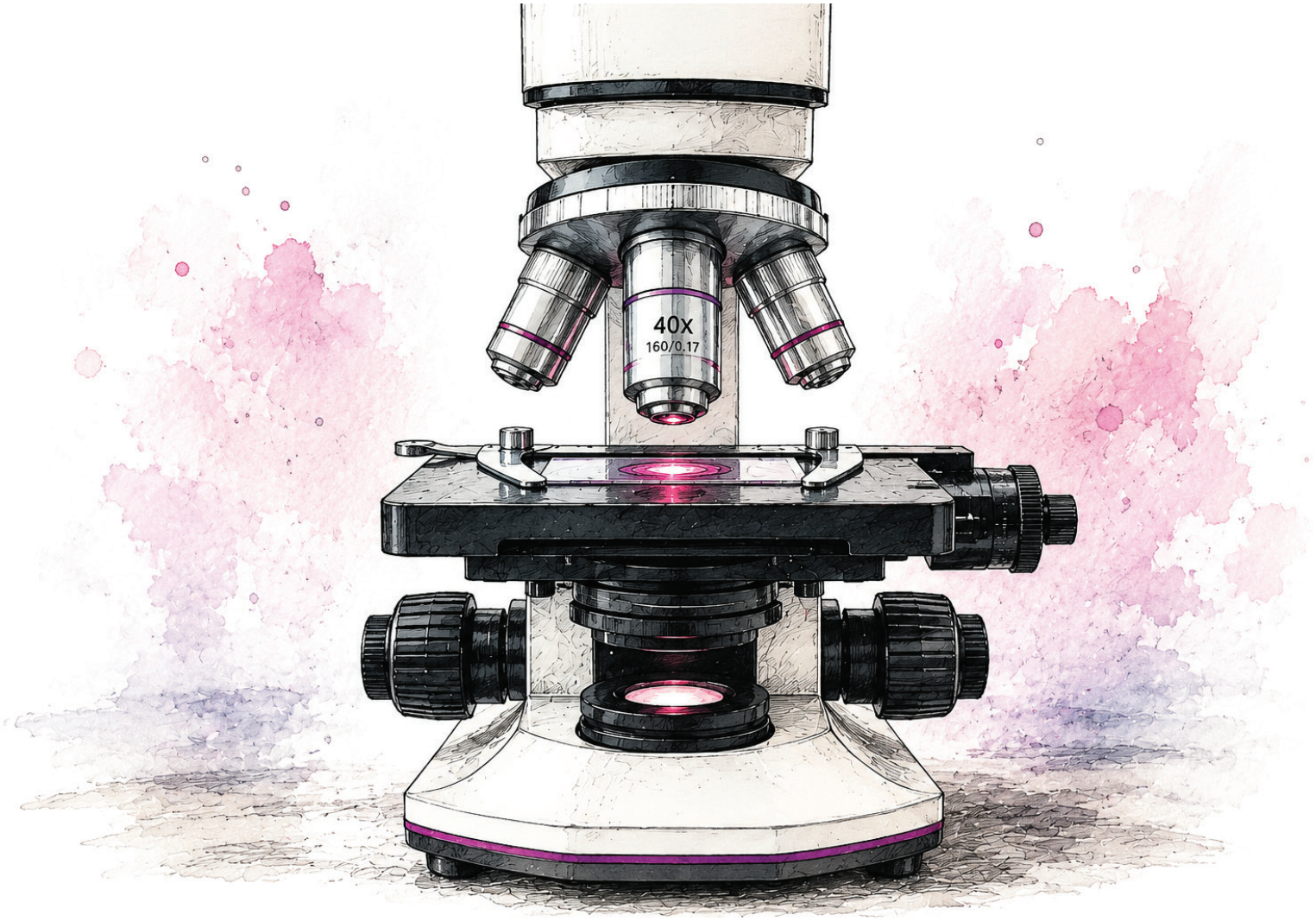
Re:State



From rare to revolutionary: how science can reach every patient

Laxshia Ganesharatnam

August 2026

From rare to revolutionary: how science can reach every patient

Around 3.5 million people in the UK are living with a rare disease, and more than 7,000 rare diseases affect patients and families across the country. A rare disease is defined as a condition that affects fewer than 1 in 2,000 people. Although each rare disease may affect only a small number of people, rare diseases are far from rare when considered together.

As population health initiatives often dominate the policy conversation, the needs of patients with the rarest conditions risk being overlooked. Despite significant scientific advances such as novel gene therapies and improved diagnostics, the NHS continues to struggle to deliver access, coordination and scale. Fewer than 5 per cent of rare diseases currently have approved treatments and many patients require complex, lifelong care.

Re:State was pleased to host a high-level roundtable to explore the gap between scientific progress and service delivery in rare diseases, and to discuss how progress in science can be matched by changes in service delivery, regulation, data and workforce planning so that more patients can benefit from emerging innovation. The event brought together a diverse group of industry and public sector leaders and experts and was kindly supported by UCB. The discussion was introduced by Louise Fish, Chair of the Rare Diseases Advisory Group, NHS England; Colin Wilson, Deputy Director of Research Infrastructure, Office for Life Sciences, Department of Health and Social Care; and Lesley Perkin, Patient Engagement & Policy Lead for Rare Diseases and Epilepsy, UCB, UK&I.

A system built for rare cases, not rare diseases

Despite growing evidence on rare diseases and how to treat them, the NHS has struggled to keep pace with developments in research and treatment. This gap between discovery and delivery set the context for much of the discussion.

Participants discussed how NHS England centrally commissions a small number of highly specialised services for groups of patients with the rarest and most complex conditions. These services are well-regarded where available, but the model is difficult to scale, especially as it depends on a small number of centres and substantial goodwill from clinicians.

There are currently 28 Rare Disease Collaborative Networks (RDCNs), which bring together expert providers to share expertise, support research and improve patient experience, while acting as a forum for discussing complex cases and coordinating care. However, participants stressed that these networks are largely underfunded and depend on voluntary contributions, raising concerns about long-term sustainability.

In practice, access to care can vary depending on where patients live, which professionals are available, and whether a local service has the capacity to support people with rare diseases. The result is a system which has pockets of excellence, rather than a consistently reliable model for rare disease care.

A key concern within this is that diagnosis does not consistently lead to a clear or coordinated pathway. Rather, it can leave patients and families without a clear point of responsibility or contact for ongoing care, even when specialist knowledge of services exists somewhere in the system.

Practical insight: patients and families should be supported with a clear single point of contact to coordinate information from experts and practitioners.

Science is moving faster than services

The discussion highlighted the rapid pace of scientific development, particularly in areas such as gene therapy and CRISPR-based treatments. The UK has real strengths in discovery science and translational research, and recent policy activity, including reform in the regulatory space and greater attention to speeding up clinical trials, create an opportunity to accelerate access to innovation.

However, participants were clear that scientific progress alone is not enough. Advances in a specific area will not necessarily make a difference in others (e.g. diseases which are not genetic in nature). Meanwhile, the health system still struggles with the barriers that prevent therapies from moving from research to routine care. These include fragmented data, delays in approvals, difficulty designing and recruiting for trials, and uncertainty around reimbursement. Therefore, scientific advancements often don't translate into routine care at the pace of discovery, limiting their timely impact on patient experience.

Practical insight: the system needs to be redesigned so that regulatory, data and reimbursement pathways evolve at the same pace as scientific innovation.

The missing link: care coordination

A lack of care coordination also emerged as a central theme throughout the discussion, as this gap affects the entire patient journey – from diagnosis through to treatment and ongoing support. Many patients experience a gap in support between diagnosis and a clear care pathway, with no single point of responsibility for managing their condition. In some cases, a rare disease diagnosis can feel like a dead end or the end of the road, because clinicians lack the knowledge, treatments or institutional practices needed to move care forward. This was contrasted with the experience of more established disease pathways, like cancer. One participant compared the process to cancer diagnosis, saying: "it would be unthinkable to leave the room without a clear set of next steps", whereas that is often the case with rare diseases.

Participants emphasised that care coordination is not simply administrative but a prerequisite for better outcomes. It can help patients navigate services, support clinicians to understand a patient's history and improve the quality of data collected over time. Without it, patients and families are often left to do much of the work themselves, creating avoidable strain and variation in care and continuity across the system.

Practical insight: care coordination should be treated as a core tenet for rare disease care, not an optional add-on.

Changing our understanding of rare diseases

One of the most important themes from the discussion was the case for thinking about rare diseases in groups rather than as isolated conditions. Participants argued that although rare diseases are diverse, many share common characteristics, diagnostic approaches, and in some cases, treatments. This creates the possibility of building models of care and research around similarity, rather than commissioning and studying each condition entirely separately.

Rare diseases are clearly not the same, nor should they be treated the same, but finding ways to group them constructively could make research, expertise and service delivery easier to organise at scale. Several participants discussed whether this approach could be particularly valuable in conditions such as cystic fibrosis, highlighting how existing specialist expertise could provide a foundation for supporting related diseases.

Practical insight: grouping rare diseases on similar characteristics, diagnostic approaches, or treatments could enable more efficient use of specialist expertise and research capacity.

Barriers in research, regulation, and access

Participants discussed the barriers that slow down rare disease research and treatment development. Clinical trials were described as especially difficult, both because patient populations are small and because the administrative burden on researchers remains high. Even studies that are not especially complex can still be delayed by lengthy approval processes and red tape, which in turn makes it harder to maintain momentum and recruit patients.

Regulation and reimbursement were also identified as critical issues. The roundtable heard that the MHRA's ongoing work on rare diseases and new therapies is welcome, but the broader system still needs to adapt if the UK is to become a better environment for rare disease innovation. Participants noted that industry needs confidence in the regulatory and market landscape if it is to invest in developing treatments for very small patient populations and that reimbursement can be particularly difficult when there are smaller numbers of cases.

Health economics was another recurring theme. When rare diseases are considered individually, it can be more difficult to make the investment case on an individual basis, even though their collective impact is undeniably significant. That is why grouping rare diseases and recognising their collective impact is vital as it can provide a more complete picture.

Practical insight: reimbursement and evaluation frameworks need to be fit for purpose for small patient populations and collective disease burden.

Workforce and the future NHS

The roundtable also touched on workforce capacity and the practical realities of delivering care. Participants described a system in which specialist nurses, research nurses, and clinical academics are under pressure, and where responsibility for rare diseases can fall unevenly across teams. There were concerns that the NHS does not always have the right skills in the right places, or the infrastructure needed to support emerging models of care and research.

This linked back to wider questions about the future of the NHS. Participants stressed that rare diseases need to be considered in service planning, not left to fit into models designed for more common conditions. In practice, that means ensuring patients can move between hospitals, community services and specialist centres with clear continuity of care and trial opportunities.

Practical insight: rare disease workforce planning should take into account the skills and infrastructure that need to be in place to support emerging models of care and research.

Conclusion

The UK has many of the ingredients needed to improve rare disease care and research: strong science, institutional expertise, and policy momentum. But participants felt that these ingredients were not yet well aligned. Better data, better coordination, a more workable research environment, and a more strategic approach to grouping diseases are all crucial to making progress.

Above all, the discussion returned repeatedly to the patient. Scientific breakthroughs will only matter if they can be translated into timely diagnosis, clearer pathways and more reliable care. They need to be matched by a system that can deliver them.

Re:State


@restate_thinks


@re-state.bsky.social


www.re-state.co.uk