

The Report

Rare Diseases

Closing the translation gap

Dr Sam Barrell Finding answers for the people who need them most

Need for speed How do we deliver treatments to patients faster?

Pioneer parents How families are helping drive the rare disease agenda

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Infographic

Rare diseases might be individually uncommon, but collectively they are a huge global health issue



90%
More than nine in ten rare diseases have no approved treatment

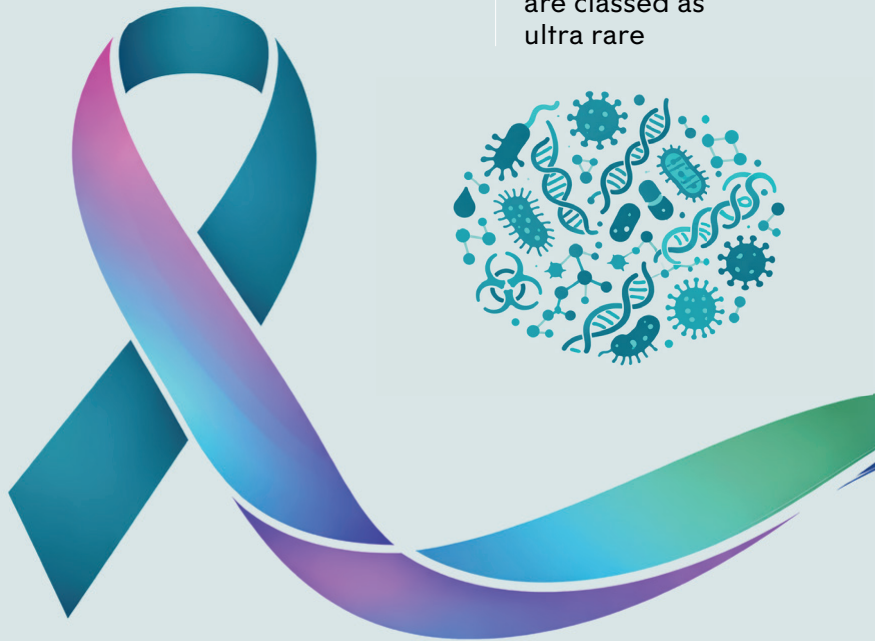
+300m
Over 300 million people are affected by rare conditions globally – more than 3 million in the UK alone

+7,000
More than 7,000 rare diseases have been identified worldwide. These are defined as impacting fewer than one in 2,000 people. 84 per cent are classed as ultra rare



1 in 17

It is estimated that more than 5 per cent of the UK population will be affected by a rare condition



+5 years

One third of people with a rare disease in the UK wait over half a decade for a diagnosis



70%

Seven in ten cases start in childhood, increasing urgency of the diagnostic journey – and the cost of delay



Comment



Dr Sam Barrell
Chief executive officer,
LifeArc

“We need to do more for people living with a rare disease – they don’t have time to wait”

In the UK, more than 3 million people live with a rare disease. Over 90 per cent of these diseases have no approved treatment. Delayed diagnosis and limited treatment options come at an estimated £14.9bn cost to the economy each year.

This should be all the motivation needed to act. But delivering the change needed requires the whole healthcare system – government, NHS, researchers, industry, policymakers and patient groups – to work together.

Throughout my career, whether as a clinician, running an NHS hospital or leading a medical research organisation, I have seen the power of cross-sector coalition to deliver change and transform lives. I have seen the real-world impact of this for families and carers, and I have seen

this in the children and adults who are here today thanks to breakthroughs made possible by collaboration.

It has been at the heart of what we do at LifeArc for over 25 years: the belief that we go further, and faster, together.

It is evident in the story of young Oliver Chu, born with Hunter syndrome, an ultra-rare condition that can be life-threatening and has just one treatment option. He was given an innovative gene therapy by a team at Manchester University Hospitals NHS Foundation Trust in November 2025 and they have been delighted by his progress.

This breakthrough was made possible by organisations like LifeArc joining forces with Great Ormond Street Hospital, the University of Edinburgh and others.

We all know this isn’t yet the norm – but these don’t have to be one-off cases.

Medicine is becoming more personalised and precise. This means some of the scientific and regulatory hurdles slowing progress in rare diseases today could soon be faced by more common conditions. In some cases, they already are.

Improving the development of rare disease treatments and access to them isn’t a niche concern. Overcoming the challenges that hold them back has the potential to pave the way for the future of medicine. We all have a stake in acting now.

That’s why several pharmaceutical companies have made serious, long-term investments in rare disease R&D. That’s why start-ups and biotechs launching in rare diseases can outperform similar companies in more saturated markets.

It’s behind recent announcements by the Medicines and Healthcare products Regulatory Agency (MHRA) on policies making it quicker and easier to get rare disease therapies tested, manufactured and approved in the UK.

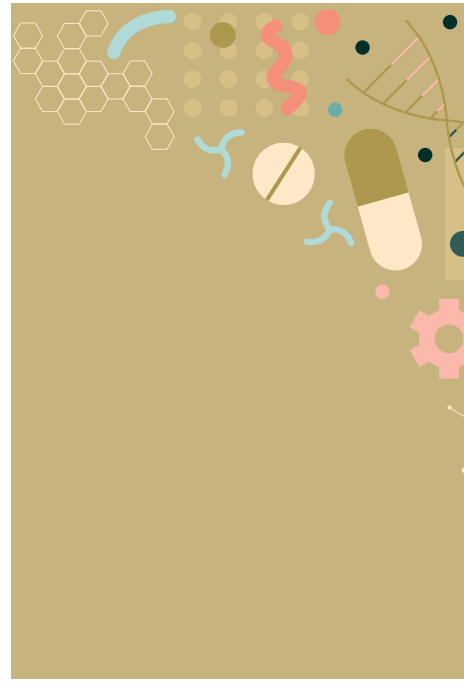
This is all promising, but there is more to be done. We highlighted how we can deliver this change in a report last year – “Accelerating R&D for Rare Disease in the UK”.

It calls for the embedding of rare disease needs into the UK’s health data infrastructure, ensuring new treatments are reviewed and approved in a more joined-up way, with better support for rare disease innovators. This can all be driven forward by appointing a UK-wide Rare Disease Champion.

Rare disease innovations shouldn’t have to defy the odds to succeed. People living with rare diseases deserve a system designed to work for them, not against them. ●

Building better pathways to care

We asked five rare disease experts: how can we get effective treatments to patients faster?



For people living with rare diseases, time is often the most critical and least available resource. Delays in diagnosis, uncertainty about treatment options, and long waits for access to specialist care can shape lives from the earliest years. For many families, progress is measured not in months but in years, as symptoms worsen while effective interventions remain out of reach.

In the UK alone, millions are affected by conditions that individually are uncommon but collectively represent a major public health challenge.

Diagnosis frequently takes several years. During that time, opportunities for early intervention are lost, irreversible damage may occur, and preventable complications accumulate. For children with the most severe conditions, delay can mean the difference between stabilisation and lifelong disability – or survival itself.

Even when promising therapies exist, access is rarely straightforward. Treatments may be available only through limited trials, restricted programmes or overseas centres. Others become trapped in lengthy approval,



commissioning and reimbursement processes. The result is a system in which scientific progress often moves faster than patient access.

These delays carry profound consequences. They affect physical health, educational outcomes, employment prospects and mental wellbeing. They place sustained pressure on families and carers, and long-term costs on health and social care services. Yet many of these impacts remain poorly captured in formal decision-making.

Pressure for change is growing. Families are better informed, clinicians are more aware of emerging options, and new technologies are making personalised medicine increasingly feasible. Expectations have risen accordingly. When treatments exist, or could exist with the right support, slow progress becomes harder to justify.

Against this backdrop, the question of how to accelerate access to effective therapies has become unavoidable. It is not simply about moving faster, but about building pathways that reflect the urgency faced by patients. To that end, we put one simple question to five experts in the field: “How can we get effective treatments to rare disease patients faster?”



Amit Nathwani
Professor of
haemophilia, UCL

Most breakthroughs fail not in the lab but in the funding gap between discovery and the first patient. Early research is often supported by charities and public funders, but as projects move towards first-in-human studies, costs surge and cannot be managed by charities alone. Academia needs a coordinated pathway to move discoveries from lab to patient, recognising that breakthroughs reverberate beyond the rare disease community.

Cancer Research UK showed what’s possible when many small charities unite behind a mission. Rare disease could benefit from a similar co-investment vehicle pooling capital from charities, pension funds and family offices, while de-risking early-stage assets so private investors can confidently participate. This model would allow universities to recycle proceeds into new discoveries. Higher R&D tax credits and improved EIS and VCT incentives would encourage biotech and venture capital to take forward ideas born in academia. Social Impact Bonds could allow government and academia to share in the upside of successful therapies. Finally, a “full stack” IP strategy must support patents from filing to defence, ensuring transformative ideas aren’t abandoned due to legal costs, allowing academic innovation to flourish rather than fade.

The science is ready and UK researchers are already shaping global medicine; our funding system must catch up. Secure the path from campus to clinic, and academic excellence becomes a life-saving reality for millions.



Lawrence Tallon
Chief executive, MHRA

Traditional drug development hasn’t worked well for patients with unique or highly varied genetic diseases. Around 3.5 million people in the UK have a rare disease, yet only a fraction have an approved therapy. Diagnosis can take years; nearly a third of children with the most severe diseases don’t survive past early childhood.

That’s why the MHRA is changing how rare disease therapies are regulated with determination and pace. Our goal is to address barriers that have slowed progress, like small patient numbers and limited evidence, while keeping safety front and centre. We’re exploring how single early approvals could cover both a clinical trial application and marketing authorisation, with careful monitoring once the therapy is in use. We are looking at smarter ways to share evidence, and using real-world evidence and computational modelling to act as control arms.

This shift is now bearing fruit. In January, the first child in the UK received their individualised gene therapy at Great Ormond Street Hospital under an MHRA-approved study. This study uses antisense oligonucleotides developed through a standardised “platform” process, meaning the therapy has the same backbone chemistry but can be quickly customised for different patients. It uses a “master protocol”, allowing multiple related therapies to be tested under one trial. A more flexible, science-led approach will help patients access life-changing treatments more quickly, giving new hope to families who’ve waited too long for a brighter future.



Alice Tuff-Lacey
Programme lead,
Genomics England

For thousands of UK families, the search for answers begins at birth. From that moment, rare conditions can start to take effect, yet a diagnosis takes an average of five years to arrive. Many rare conditions can be treated, and newborn screening has already shown what early detection can achieve, catching conditions early enough to transform outcomes.

But a technological shift is under way. Advances in genomic technology offer a step change in our ability to screen for rare conditions. One sample can be used to generate a full genome sequence that can be screened for hundreds of rare genetic conditions simultaneously, with the flexibility to add conditions as new treatments are developed.

It represents a profound change to how this technology has been used in the UK – as a diagnostic tool for children or adults with existing symptoms.

It also raises questions: does early genomic screening improve health outcomes? How could this be implemented fairly and safely? Is it cost-effective? And can benefits outweigh the potential harms?

The Generation Study, led by Genomics England and funded by the Department of Health and Social Care, was designed to provide the evidence and inform decisions on whether this is the future direction the UK should take. It will screen 100,000 babies' samples and is due to finish recruitment in early 2027.

Its findings may lead to a generational shift in how the NHS begins life with its newest patients.



Dr Raghiv Ali
CEO, Our Future Health

Better access to large data sets. Our Future Health is bringing together up to five million UK volunteers to help researchers discover new ways to prevent, detect and treat diseases, including rarer diseases. This will be the first cohort big enough to better understand the causes of these rarer diseases as well as how to treat and prevent them. Volunteers provide permission to combine information and samples we collect with existing information, including their health records. Taken together, this creates an incredibly detailed picture of the UK's health.

Researchers can apply to study this information and they can now also apply to use our new Clinical Research Recruitment Service. The service is designed to enable faster, more efficient clinical trial and observational study recruitment into clinical trials and observational studies.

So far, over 2.5 million have consented to be invited to take part in additional research. Researchers can work with us to send targeted invitations to selected participants based on information such as diagnosis, genetic, demographic and location data.

The scale and diversity of the cohort means a wide range of diseases and conditions are represented in the programme, helping researchers to find the people they need for their trials or studies, accelerating what is usually a significant delay in the clinical research process. This efficiency can play an important role in getting effective treatments to rare disease patients at a faster rate.



Professor David Jones
Director, Newcastle
Centre for Rare Disease

Faster delivery depends on several interlinked stages: moving discoveries more quickly into development; evaluating safety and efficacy more efficiently; accelerating regulatory and reimbursement decisions; and ensuring timely adoption in clinical practice. Weaknesses at any point can have serious knock-on effects later in the process.

A critical bottleneck is how therapies are assessed. Rare diseases are, by definition, rare, and conventional large-scale trial models are often poorly suited to small patient populations. More efficient trial designs are therefore essential. Platform and adaptive approaches can reduce the size of control groups and allow multiple therapies to be assessed simultaneously, enabling meaningful results with fewer patients.

Delays are also driven by practical barriers. Contracting processes between trial centres remain slow and fragmented, a problem amplified in rare diseases, where more sites are needed to recruit sufficient participants. Streamlining these arrangements would significantly shorten time lines. Targeted recruitment strategies using registries and patient-accessible platforms can help identify participants earlier, democratising access to trials.

Finally, regulators and funders need to take a broader view of acceptable evidence. More flexible evaluation frameworks and better system alignment can reduce unnecessary delays and ensure that promising therapies reach patients faster. ●

Case study

Pioneer parents: fighting for rare disease answers

Families are increasingly helping to shape research, treatment and policy

Andy Kulina's transformation from concerned parent to clinical trial advocate started with a simple Google search. After discovering that a promising nasal insulin treatment for his daughter Olivia's rare condition was unavailable in the UK, he typed in: "How do you conduct a clinical trial?" That query launched a years-long campaign to get all the necessary people and processes in place. This is now poised to culminate in a multimillion-pound trial starting in spring 2027.

Olivia has Phelan-McDermid syndrome, a rare genetic condition caused by changes in the Shank3 gene affecting nerve function. Diagnosed at 18 months, she was one of approximately 500 known cases globally. The condition impacts movement, speech, learning, sleep and mood. Most significantly, Olivia is non-verbal, making it difficult to know when something is wrong.

"You feel helpless when you have a child with a rare disease," Kulina explains. "Especially when there's no cure or treatment. But we were always searching to figure out what we could do to help."

That changed when Kulina heard a podcast featuring a Dutch scientist who had conducted a small trial using nasal insulin for Phelan-McDermid patients. The treatment had been approved for use in some clinics in the Netherlands and Germany, but accessing it meant navigating a complex system.

Unable to import the treatment, Kulina realised the only path was a UK clinical trial that could lead to the drug being approved for use on the NHS. He began fundraising through marathon running and charity auctions, though these raised only thousands at a time. "It doesn't make a dent," he explains. "We wanted to do more."

The breakthrough came at a Cambridge Rare Disease Network conference, where Kulina pitched his idea using a single piece of paper. It featured Olivia's photo, an explanation of the Dutch trial, and a request for researchers or funding.



Professor André Strydom at King's College London agreed to develop and lead the trial, and LifeArc came on board to fund the study. It will recruit around 100 children and young adults aged three to 18, testing whether nasal insulin can improve brain function without affecting blood glucose levels.

Insulin receptors exist not only in the body but also in the brain, where they regulate nerve activity and cognitive function. By delivering insulin nasally, scientists hope to reach brain nerves without impacting glucose levels in the body. Early international studies show promise, and the treatment is already available in some German and Dutch clinics.

The trial exemplifies drug repurposing, an increasingly valuable strategy, given that a large proportion of rare conditions lack approved treatments and using existing drugs can speed up the process. LifeArc CEO Sam Barrell notes that for more than three million people in the UK living with rare diseases, accessing treatments remains hugely challenging. "Innovative trials, including those repurposing drugs, will be crucial," she says.

If successful, the treatment could extend beyond Phelan-McDermid syndrome, with research suggesting potential benefits for Down's syndrome and Parkinson's disease.

For Kulina, the trial represents more than scientific progress. "The most important currency to all families affected by Phelan-McDermid syndrome is hope," he says. ●



LifeArc is dedicated to developing diagnostics and treatments for people with rare diseases and drug-resistant infections.



We fund and invest in pioneering research and promising scientific breakthroughs to address the challenges of rare diseases.



We work with and connect the right partners to make sure that new treatments reach the patients that need them, faster.

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