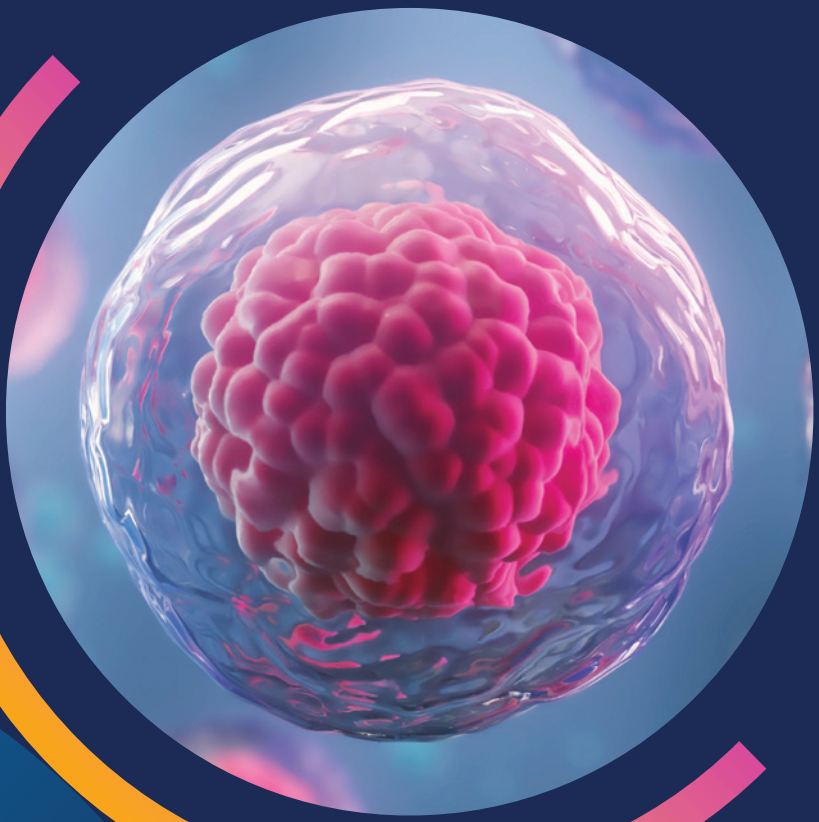
Accelerating R&D for rare disease in the UK:

An opportunity to change millions of lives

The impact for people living with rare diseases



LifeArc

WITH THE SUPPORT OF GENETIC ALLIANCE UK

Our recommendations

Diagnosis and data

01. Make sure rare diseases are included in plans for the Health Data Research Service, which will aim to transform how NHS data is used.

02. Set clear standards for rare disease data collection and storage – and adequately support existing registries to maintain, streamline and unify their data.

03. Make whole genome sequencing easier to access and improve medical training, so that more doctors can spot rare diseases and understand the value of a diagnosis.

Market access: making drugs available

04. Health departments should invest in generating evidence that shows the real costs of rare diseases to all areas of society.

05. Regulatory and assessment bodies, responsible for approving treatments, must align their evidence requirements and tailor them to rare diseases.

Support for innovators developing drugs

06. Create a support service that guides researchers through the complex drug development and approval process.

07. The next UK Rare Disease Framework must come with funding to map the full journey of rare disease research, so that researchers can plan efficiently and get treatments to patients, faster.

Crucial drivers of change

- A **UK-wide champion and coordinator for rare diseases** is appointed to lead and coordinate efforts across the UK.
- Ministers from all four UK nations commit to **a refresh of the 2021 UK Rare Diseases Framework,** reflecting today's challenges and opportunities.
- There is **a review of the methods** that Government could use to encourage more research into rare diseases in the UK.

Contents

This project was jointly convened by LifeArc and Genetic Alliance UK. It was overseen by a Taskforce with a diverse range of expertise. A full list of Taskforce members can be found at the end of this report. Contributions by the Taskforce were made purely in an advisory capacity. The members of the Taskforce participated in an individual capacity and not as representatives of their organisations. Consultation on this project ended April 2025.

“The challenges we face are clear – but so is the potential for meaningful change.

We can no longer wait for tomorrow. We must act today to demand better, to work together and to transform the system for the millions of people living with rare diseases.”

Dr Sam Barrell, CEO of LifeArc

An opportunity

For the millions of people living with rare diseases, time is critical.

We know families are facing immense pressure, daily challenges and a long journey to get answers. Many wait years for a diagnosis and even when they get one, there is little hope of an effective treatment.

Pioneering science has led to incredible breakthroughs, and the potential for new treatments already exists. But right now, huge hurdles sit between a researcher with a promising idea and a treatment that is approved for use.

We believe these hurdles can be overcome. LifeArc joined forces with Genetic Alliance UK to bring together a taskforce of experts from across the rare disease community, including patient advocates. We challenged them to come up with actionable solutions that could change the system and change lives. We are grateful to the many individuals and organisations who contributed their expertise and insight.

LifeArc is committed to improving the lives of people with rare diseases. Since 2019, we have committed more than £170 million to research in the field. Through our scientific expertise, facilities and partnerships, our aim is to make sure promising research turns into tests and treatments, quickly.

This report shares recommendations that we believe will help people with rare conditions get diagnosed faster and have better access to treatments. It's written for anyone who wants to understand the challenges and be part of the solution. Many recommendations are aimed at policymakers, but there is a role for us all.

Together, we have an opportunity to change millions of lives.

1 in 17 people

live with a rare disease

Prof Amit Nathwani, Taskforce chair and Professor of Haematology, UCL

Dr Sam Barrell, CEO, LifeArc

What needs to change?

Despite progress, rare disease research lags behind that of more common diseases. People living with these conditions continue to fall through the cracks. Our expert taskforce of people from across the rare disease sector has helped us identify three critical areas for action.

> “We want to make the UK a great place to do rare disease research, so that more treatments can get to more people, as fast as possible.”
>
> **Professor Amit Nathwani**

95%
of rare diseases lack an approved treatment

Diagnosis and data

If we

improve access to whole genome sequencing, better train doctors to spot symptoms and establish a clear plan for expanding and unifying rare disease registries, with robust national data standards...

We could

make sure people are diagnosed sooner and unlock the life-changing discoveries hidden within genetic data. By improving how we capture and handle all rare disease data, we also increase the chances of those innovations being approved to treat the people who need them.

Market access: making drugs available

If we

know the true costs of rare diseases to society and align regulatory requirements...

We could

show that many rare disease drugs do offer value for money, helping more of them get approved.

Support for innovators developing drugs

If we

make the process of developing and approving treatments simpler and more affordable...

We could

grow the number of innovators working in the UK, attract investment into rare disease research and get more people the treatments they need, sooner.

If we work together to deliver the recommendations in the following sections, we can create this future. Not only will people with rare diseases have a better diagnosis and treatment journey, but we will also make the UK the go-to place for rare disease research.

Diagnosis and data

The current situation

Data needs to work harder for people with rare diseases. Currently, it is split up, recorded differently, and hard to bring together. Registries that capture information on rare diseases do exist, but they are undervalued and disconnected, making it harder to use that data to drive change.

A diagnosis can be life-changing for people and their families. It also unlocks progress: new research, better testing, targeted treatments, and an understanding and record of how many people are affected. Yet too many people struggle to get answers.

Many rare diseases are caused by genetic changes, so whole genome sequencing – reading someone's entire genetic code – could provide a diagnosis for many. But challenges, including a lack of capacity and staff, are causing delays and restricting access. Too many people are still waiting.

For non-genetic conditions, spotting symptoms is vital for diagnosis and treatment.

Our recommendations

To improve diagnosis and data:

01.

Make sure rare diseases are included in plans for the Health Data Research Service, which will aim to transform how NHS data is used.

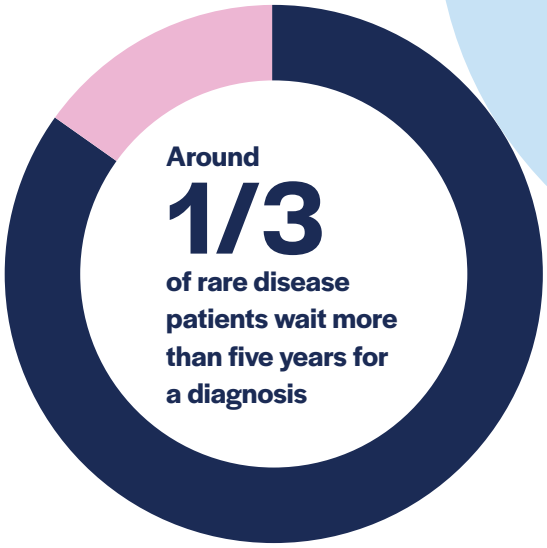
02.

Set clear standards for rare disease data collection and storage – and adequately support existing registries to maintain, streamline and unify their data.

03.

Make whole genome sequencing easier to access and improve medical training, so that more doctors can spot rare diseases and understand the value of a diagnosis.

> “Improving diagnosis will not only help patients, it will open up a world of new data to drive even more innovation.”
>
> **Professor Amit Nathwani**



Market access: making drugs available

The current situation

Developing new medicines is expensive and, when that cost is only spread across a small number of people, prices are high. But high costs can sometimes mean treatments aren't approved for use. What's often missing in these decisions is the full picture, and the true cost to society. Rare diseases don't just affect the NHS, they impact families, carers, education, work, mental health and more.

The UK must fully understand the true cost of rare diseases and these wider considerations should be part of the knowledge we use to assess the value of new treatments. Time and money could also be saved if we made the regulation and approval process easier. This highlights the need for regulators and decision-makers to align their processes and tailor them to account for the full impact of rare diseases.

Our recommendations

To improve market access:

04.

Health departments should invest in generating evidence that shows the real costs of rare diseases to all areas of society.

05.

Regulatory and assessment bodies, responsible for approving treatments, must align their evidence requirements and tailor them to rare diseases.

Rare diseases in the UK are likely costing the NHS

tens of billions

of pounds

> “Understanding the full cost of rare diseases to the UK would show the true value of treatments, making it clearer how they would benefit not just the individuals, but the whole of society.”
>
> **Professor Amit Nathwani**

Support for innovators developing drugs

The current situation

The UK needs to support the people developing new treatments. Right now, researchers face a process that is complex and costly. The UK must make it easier for them to turn their scientific discoveries into treatments that reach patients, quickly.

Our recommendations

To better support innovators:

06.

Create a support service that guides researchers through the complex drug development and approval process.

07.

The next UK Rare Disease Framework – produced by the Government to address ongoing challenges faced by people with rare conditions – must come with funding to map the full journey of rare disease research. Specifically, it needs to pinpoint the most expensive and time-consuming steps, so that researchers can plan efficiently and get treatments to patients faster.

The European Medicines Agency has identified more than

3,000

potential rare disease therapies as 'promising'. Just 268 of them have ever received market authorisation.

"The path from discovery to treatment is complex, expensive and confusing, meaning some promising innovations never make it to the clinic. This needs to change."

Professor Amit Nathwani

Crucial drivers of change

Progress in the areas outlined in this report will be easier to achieve if:

A UK-wide champion and coordinator for rare diseases is appointed to lead and coordinate efforts across the UK.

Ministers from all four UK nations commit to **a refresh of the 2021 UK Rare Diseases Framework,** reflecting today's challenges and opportunities.

There is a **review of the methods** that Government could use to encourage more research into rare diseases in the UK.

How you can help

The UK has a unique opportunity to lead the world in rare disease research and development, and change millions of lives in the process. But if we don't act now, patients will keep feeling the impact of our inaction.

There is a role for us all, and you can help make change happen. Please share this report, talk about it, and call on others to get involved. We also welcome you to work with us to help turn these recommendations into reality. If we do, the UK's world-leading life science sector can become life changing for people with rare conditions.

Read the in-depth report
lifearc.org/rd-report

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Charity numbers
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