

Awake just worrying



The importance of patient voice in understanding rare disease, and the need for a stronger evidence base.

**Patient
Experience
Library**

December 2023

1. Burden of disease

“I was awake from 4am this morning just worrying,” says Lara in a report on the Covid experiences of people with “underlying conditions” including rare diseases.¹

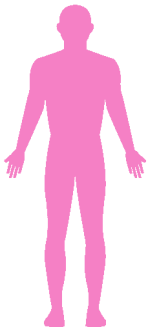
People with rare disease have plenty to worry about: long waits for diagnosis and treatment, baffled looks on the faces of health professionals, doubts about the effectiveness of treatments.

And there are very many people like Lara. Individual rare diseases affect small numbers of people, but overall experience of rare disease is recognised by the Department of Health and Social Care as “collectively common”.

The government’s Rare Disease Framework states the importance of “patient voice” to help improve understanding of rare diseases. But how does patient experience show up in the research?

We went looking for the literature. We found that lived experience of rare disease is under-researched compared to other parts of the patient experience evidence base. There are gaps in knowledge, and a likelihood that health charities and patient groups are bearing an unfair share of the research burden.

We look at the implications for national NHS bodies and for research funders, and suggest ways to get better learning. And if you want to explore the evidence base for yourself, you can skip straight to our [interactive map](#) to see what it looks like.



2. In it together

Collective experience

1 in 17 people will be affected by a rare disease at some point in their lives, according to Rare Disease UK. This, they say, amounts to 3.5 million people in the UK.²

The government's Rare Diseases Framework agrees. It says that "Although rare diseases may be individually rare, they are collectively common".³

Rare diseases are long term conditions: Genetic Alliance UK states that "around 70% begin in childhood and are life-long".⁴

Here too, the Rare Diseases Framework agrees: "Rare disease patients and their families can face a lifetime of complex care".⁵

Collective voice

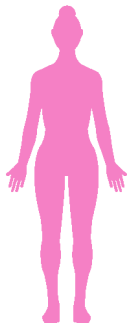
Government recognises the importance of hearing from patients and families. In the five underpinning themes to the Rare Diseases Framework, "patient voice" comes first.⁶

This raises an important question: how do we hear the collective voice of people with rare conditions?

One answer might be that an NHS aiming to be both "patient-centred" and "evidence-based" would develop a research literature to help it understand the experiences of those 3.5 million people.

But does such a body of evidence exist? We decided to find out.

We mapped the evidence base on lived experience of rare disease, aiming to find out who is listening to patients. We wanted to know who researchers are talking to, what they are talking about, and where the findings are being published.



3. What we found

The numbers

We ran a series of searches in the Patient Experience Library⁷ which acts as the national evidence base for patient experience and involvement. We set a five-year time frame for the search – starting in 2018 and running through to 2023.

Within that period, we found 68 reports touching on the healthcare experiences of people with rare conditions.

The number is striking. We recently ran a similar evidence-mapping exercise, looking for literature on people's experiences of digital healthcare.⁸ There, over a five-year period, we found 157 reports – over twice as many. Another recent analysis of patient experience through the Covid pandemic found 696 reports in just two years.⁹

The government's Rare Disease Framework describes rare diseases as "collectively common". But when compared with other parts of the patient experience evidence base, the literature on rare disease seems sparse.

Saturation

We looked at the main topics covered by the 68 reports that we had found. Many reports covered more than one aspect of people's experiences, so there were 123 topics in all.

The three largest topics were "communication" (28%), "service access" (24%) and "research" (23%).

This makes perfect sense. People with rare conditions need good communications to help them understand diagnoses, medications and self-care. Access to services is a big issue, given problems with long waits for diagnosis and referral. And patients and families are often keen to get involved in research, to help improve clinical understanding and responses.

So there seems to be a good proportion of the evidence base dedicated to people's experiences of these well-known issues. That is good news – but it perhaps raises the question as to whether the research focus should start to move elsewhere.



Gaps

Experiences of rare disease appear to be under-researched compared to other aspects of patient experience. That creates the potential for gaps in knowledge.

But gaps can be hard to identify – how, after all, can we “know what we don’t know”?

A clue might lie in the Rare Diseases Framework. Here, government commits itself to “ensure that the needs of rare disease patients are appropriately reflected across wider government policy, including mental health and social care”.¹⁰ It also promises “representation from those whose voices can often go unheard, including patients from Black and Minority Ethnic (BAME) or disadvantaged backgrounds”.¹¹

But we were unable to find studies on experiences in mental health and social care services for people with rare conditions. And our searches for studies having a primary focus on Black or disadvantaged groups were similarly unrewarding. Other than four reports on sickle cell disease, we found just one paper, looking at “underserved” groups.¹²

A further clue to gaps in the evidence base comes from analysis of the sources of the research.

In other evidence-mapping exercises (such as digital and Covid, as above), we have found that professional journals and the local Healthwatch network tend to be the biggest sources of published studies.

Unusually, that was not the case for rare disease, where open access journals accounted for just 11 papers (16% of the literature we found), and Healthwatch a mere 4 reports (6%).

Is it possible that these sources see the rare disease community as too small or too hard to reach? We cannot know for sure. But what we can see is that in research on patient experience of rare disease, health charities and patient networks are having to make much more of the running than elsewhere.



4. Implications

For this exercise in mapping the evidence base on patient experience of rare disease, our searches were exclusively within the Patient Experience Library¹³. The Library only collects open access literature, so it is possible that further evidence lies behind paywalls put up by journals and other research databases. Even so, there are some useful learning points:

1. A unique archive on patient experience of rare disease is now freely available.

Researchers, policymakers and campaigners: Use our [open access evidence map](#) to see to see how we have visualised the evidence base.

2. Gaps in the evidence base seem likely.

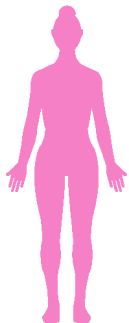
DHSC's Rare Diseases framework puts "patient voice" at the top of a list of underpinning themes. But our analysis suggests that Black and disadvantaged voices are largely absent from the evidence base, and that there is little, if any, evidence of rare disease patients' experience in mental health and social care.

Department for Health and Social Care and National Institute for Health Research: Use our evidence map as a basis for understanding and remedying the unevenness in the evidence – particularly with a view to addressing "patient voice" commitments in the Rare Diseases Framework.

3. There is an unfair research burden.

Other [evidence mapping exercises](#) have shown that professional journals and local Healthwatch tend to be the biggest sources of evidence on patient experience. But for rare diseases, charities and patient groups are having to do the heavy lifting.

National Institute for Health Research and NICE: Use our findings as a basis for mobilising research funding and effort towards the unheard voices of rare disease.



Finally...

This report is part of a wider evidence mapping project for patient experience and engagement.

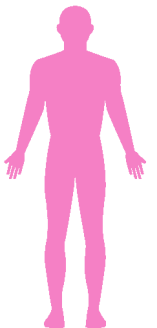
While medical research has rigorous prioritisation processes, evidence gathering on patient experience is, essentially, a free-for-all. One consequence is extensive duplication and waste. Another is big gaps in the evidence base. These are very often in relation to so-called “hard to reach” communities – the very people whose voices really need to be heard.

With funding from the Health Foundation’s Q Community, we have undertaken the first ever exercise in mapping key areas of the evidence base on patient experience. Our results are being disseminated via a series of reports like this one, and through a set of interactive online evidence maps.

We are laying the foundations for better research prioritisation in patient experience – to steer time and money more effectively, and to help the NHS ensure that its promises of patient-centred care are soundly evidence-based.

Our evidence maps can help research funders to see how to get better value for money, help researchers to see how to avoid time-wasting and duplication, and help patient advocates to see who is – and is not – getting heard in patient experience evidence-gathering.

For more on the project, visit the [evidence maps page](#) on our website. And if you want to partner with us to produce more maps, please get in touch: info@patientlibrary.net



Acknowledgements

We would like to thank:

Dr Sebastian Stevens and [Apollo Health Innovations](#) for research assistance.

The Health Foundation's [Q Community](#) for funding via the Q Exchange programme.

Q Community members for backing this project, and supporting our vision.

Sorcha McPhillips and the Huntington's Disease Association Northern Ireland for practical support and advice.

Endnotes

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- 2 <https://www.raredisease.org.uk/what-is-a-rare-disease/>
- 3 Department of Health and Social Care, 2021. UK rare diseases framework. Page 3.
- 4 Genetic Alliance UK, 2020. Rare Experience 2020: The lived experiences of people affected by genetic, rare and undiagnosed conditions. P4.
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- 10 Department of Health and Social Care, 2021. UK rare diseases framework. Page 3.
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- 12 Witham, M. D. et al, 2020. Developing a roadmap to improve trial delivery for under-served groups, results from a UK multi-stakeholder process. *Trials* 21:694 <https://doi.org/10.1186/s13063-020-04613-7>
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