M/UK/UNB-U/0552
July 2026

ALEXION®
AstraZeneca Rare Disease

Beyond the prompt: How people living with rare diseases are using AI in the gaps between care



How people living with rare diseases are using AI in the gaps between care

As AI becomes increasingly embedded in healthcare, understanding real-world experiences will be critical to ensuring these technologies are safe, trusted and genuinely useful for the rare disease community.

Through our patients insights forum we learnt that people living with rare diseases aren't using AI to replace their doctors. They're using it in the four-year wait for a diagnosis (the average for the rare disease community), copying parts of their medical records to get clarity on test results, and in the questions they don't want to ask out loud.

They told us that they have already made AI part of their care, not because they trust it, but because gaps in care pathways have left them with no one else to ask, or they felt their question didn't warrant time from stretched healthcare professionals. That has implications for the NHS, industry, and every patient group that provides support for people in the rare disease community.

7000+
rare diseases affect an estimated **3.5 million** people in the UK[1]



4-6 years
the **average** wait for a rare disease diagnosis[2]



1 in 7
people in the UK have used **AI instead of seeing a GP**[3]



About the patient insights forum:

The Alexion patient insights forum brought together people living with rare diseases and carers to explore their experiences of using AI tools, where they found value, where they had concerns, and what they believe responsible AI in healthcare should look like. The insights presented here reflect their lived experiences and priorities.



> **That period between the consultant saying 'I am concerned' and eventually receiving a diagnosis is just a complete emotional rollercoaster. You want something you can rely on. Because everyone is telling you something different.**

Parent of a child with a rare disease

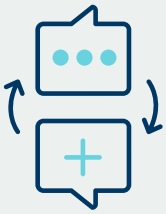
AI is a workaround, not a replacement

People living with a rare disease are often asking AI to translate, prepare, and join the dots for them, not necessarily to provide a diagnosis.

Five use cases were identified:

01.

Translating complex medical language into something they can understand.



02.

Preparing for appointments and knowing which questions are worth asking.

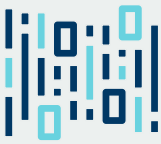


> **I use ChatGPT like Google, but I say: tell me where everything has come from. Then I can get the bullet-pointed ideas... see it has come from somewhere I trust, and then follow that link.**

Person living with a rare disease

03.

Decoding test results that arrive in a portal before a clinician can explain them or asking for further details after a consultation.



04.

Joining the dots between symptoms that specialists are looking at in isolation.



> **There is something in that vulnerability of AI not being someone you know. Your deepest, darkest fears. You want to voice them to a stranger.**

Parent of a child with a rare disease

05.

Using the anonymity it brings to ask personal questions, that might otherwise be left unasked, such as around fertility, dating, and other sensitive topics that are uncomfortable in-person.



What shapes AI use?



The single biggest factor shaping AI use is whether you have easy access to a specialist team. People with named consultants and responsive nurses turn to AI less, while people with less contact with healthcare teams rely on it most.

> **Someone showed me CoPilot at work and it was like a light bulb... but I would not use it for lupus information.**

Person living with a rare disease

The moments that matter most are the moments AI gets wrong

Participants were consistent in their scepticism about the quality of AI-generated information for rare and ultra-rare diseases.

Not a general distrust of technology, but participants flagged that AI can only reflect the data it has been trained on, and for rare diseases that data is often sparse, outdated, or skewed towards worst-case presentations.

> **Even my consultants do not know enough about my condition. So how can AI know? I have got an ultra-rare condition, and they are still trying to learn about it.**
>
> **Person living with an ultra-rare disease**

Comfort with AI at work doesn't mean comfort with **AI for health.**

Three risks of AI use for people living with rare diseases:



01.

Pre-diagnosis, when people are most frightened and least supported, and most likely to meet a worst-case answer first.



02.

The tendency to catastrophise was a specific frustration for forum participants. AI tools, drawing on population-level data, tend to surface worst-case presentations regardless of individual prognosis.



03.

People now receive raw clinical data through NHS apps, including complex radiology reports, before a clinician has had the opportunity to contextualise it for them. This leaves some people reaching for AI in the gap.

> **I know all of it and it still terrifies me when I look. It will still say your life expectancy is lower, this is going to go wrong.** And it is not necessarily true.

Person living with two rare diseases

> AI mixes **good and bad advice** in ways that are hard to tell apart.[4]

A 2026 Nature Medicine study found patients using AI didn't make better medical decisions than those using ordinary online searches, because AI mixes good and bad advice in ways that are hard to tell apart.[4] But it does have its positive use cases that should be explored further, including in pattern recognition and as a diagnostic imaging tool.

> **One of the things AI doesn't do is have** pre-formed judgements or dismiss you.

Person living with a rare disease

Build it with us in mind

Across the forum, the clear ask was for AI grounded in trusted, community-curated information, designed with the people who'll use it.

> AI is making existing gaps in rare disease care **more visible**.

The rare disease community identified five ingredients they believe are essential for trustworthy AI tools:

01.

Built on trusted data: curated by charities, specialists and people living with a rare condition.



02.

Transparent about sources: every answer comes with its receipts.



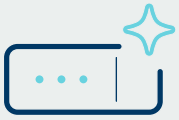
03.

Calibrated for pre-diagnosis: helps people ask the right questions, not diagnose.



04.

Equipped with good prompts, because no one writes their best question at 3am.



05.

Honest about its limits and knows when to hand over to a human.



What we can do:

01.



Industry:

Co-design with the community.

Invest in condition-specific data infrastructure as pre-competitive, not proprietary.

02.



NHS:

Standard practice should be human first, portal second.

Close the rare disease gap on NHS digital platforms. Develop clear, condition-specific guidance on AI for people living with rare diseases together with patient organisations.

03.



Charities:

Publish trusted prompt guides.

Reach newly diagnosed people before any harmful patterns set in, and where possible, consider building or endorsing condition-specific AI tools.

References

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