Rare disease research in the 10 Year Health Plan – a workshop summary

Background

In January 2025 the Association of Medical Research Charities and Genetic Alliance UK organised a workshop on rare disease research in the 10 Year Plan for Health.

The workshop focused on the opportunities and challenges of the Government's '3 shifts' – analogue to digital, sickness to prevention, and hospital to community. We also discussed a vision for rare disease research and what a successful 10 Year Plan for Health would mean in this area. Insights from the workshop were shared directly with the Department for Health and Social Care to support the development of the 10 Year Plan for Health.

We brought together spokespeople from 14 health charities and sector organisations who represent patients living rare disease or work in the rare disease research sector.

Action Medical Research
Ataxia UK
Brittle Bone Society
CureGRIN Foundation
DEBRA UK
Department of Health and Social Care
Duchenne UK
Fight for Sight
Great Ormond Street Hospital Charity
Muscular Dystrophy UK
Scleroderma & Raynaud's UK
Tuberous Sclerosis Association
UK Rare Epilepsies Together
Unique

We chose this topic because:

Rare diseases affect large numbers of people in the UK

- Over 3.5 million people in the UK are living with a rare disease
- There are around 7000 rare genetic diseases, with scientific research continually identifying new diseases
- 1 in 17 people are affected by a rare disease at some point in their lives
- 7 in 10 rare diseases affect children, with more than 3 out of 10 children with a rare disease dying before their fifth birthday

Patients with rare diseases collectively have significant unmet health needs.

Many rare diseases remain untreatable. There are no treatment options for many people living with rare diseases, and getting new treatments to patients is extremely challenging. Of the 3000 potential treatments for rare diseases that have made it past Committee for

Medicinal Products, only 300 have ever had that market authorisation. This means 90% of promising treatments never get to patients. The treatments that have been authorised only cover 132 of the 7000 rare diseases. In addition, the treatments that are authorised for rare disease are often high cost and the burden of treatment is high for the patient.

Research to find new treatments is not equitable. New treatments are often the best hope for patients with rare diseases to improve their health outcomes. Yet there is significant inequality in research across the range of rare diseases, with only 23 of 7000 rare diseases attracting the majority of research focus. For most rare diseases there is little to no research conducted, giving patients no hope that their health will improve in future.

Diagnosis of rare diseases is extremely challenging. Most clinicians will see very few patients with rare diseases during their whole career, which hampers identification. In addition, not all rare diseases have been identified yet, so patients may not be able to get a diagnosis. This may mean living their life with a serious disease that clinicians have no idea how to manage or treat and with no knowledge about disease progression.

Research can help improve diagnosis, treatment and care options for people with rare diseases, improving lives, and reducing the impact on healthcare and society. Progress in this area would improve lives and reduce healthcare and societal costs. Taking part in research gives patients access to specialist healthcare and early access to innovative rare disease treatments.

A vision for rare disease research

We asked attendees what three words will describe a successful 10 Year Plan for rare disease research. They said:

Equity: across research into different rare diseases, and between common and rare diseases.
Access: to research nationally and internationally, which would allow the best care and early access to treatments.
Joined-up: speeding up the diagnostic process to allow patients to access research earlier.

Another popular word from attendees was **transferable:** sharing learning from treatments and approaches for one rare disease to another.

We also asked attendees what a successful 10 Year Plan would mean for rare disease research, focusing on what would be the same and what would be different.

What will be the same?

Diagnosis of rare diseases will continue to improve. After patients fail to get a clinical diagnosis for a common disease, they will have genomic testing for rare diseases. Data collected during the genome sequencing process will be stored at the National Genomic Reference Library, allowing rare diseases to be identified. Where someone has a disease that has not yet been formally recognised by science or medicine, the person's data can be safely used to help identify the disease in future.

What will be different?

Research findings will provide an evidence base for the NHS to innovate to improve the health of patients with rare diseases. Patients with rare diseases across the whole of the UK will have timely and equitable access to cross-disciplinary research which will lead to better diagnosis, care and treatment. Rare disease research will be an attractive career option for clinicians and researchers.

We will be smarter about finding new treatments for rare diseases. This will include:

- Repurposing existing medicines for rare diseases;
- Knowledge sharing between types of rare diseases.

This will involve:

- Looking across rare disease types to find commonalities which may be treatable, instead of only thinking of each disease separately;
- Regulators will work with patients to better understand what a good outcome measure for new therapeutics in areas of unmet need is;
- We will find new ways to test treatments, recognising that for some rare diseases it may not be possible to get a big enough sample for a phase 2 clinical trial;
- A radical rethink about funding for research into rare diseases will invigorate the landscape. This could involve new models for working with industry to encourage investment into innovation where there is a limited market.

We will be working more closely with rare disease communities to better understand their needs, preferences and priorities for research and treatment. Understanding how those with rare diseases want to frame their conditions and their priorities for treatment and support will mean that research reaps the greatest rewards.

The 3 shifts

In this part of the workshop we focused on the Government's '3 shifts' – analogue to digital, hospital to community and sickness to prevention. We used questions provided by the Government as prompts for the discussion, and below we outline a summary of participant views.

Analogue to digital

When you think about how we could use technology in the NHS, what are your hopes?

Currently we have shockingly little information about how many people in the UK are living with rare diseases and their health outcomes. Even for rare diseases that are better known, such as Muscular Dystrophy, we do not know how many people are living with the condition in the UK right now. There are not disease registries for all conditions, and where there is rare disease data available it is often poor quality and rarely interoperable. This hinders those commissioning care and researchers alike.

The move from analogue to digital has the potential to address these challenges. **Having access to accurate, high-quality patient data would accelerate research into rare diseases to transform treatment and care.**

A unified patient record: Having a fully comprehensive, universal electronic health record for everyone in the UK would provide a much better understanding of the prevalence of rare diseases, their comorbidities and health outcomes. The electronic health record could be combined with a unified genomic record for each patient and patient reported outcomes and experiences.

Patients with rare diseases often see many different healthcare professionals in different settings at different times of their life. Trying to connect-up their experiences at the moment is virtually impossible. Capturing patient experiences on their electronic records, implemented in a meaningful and comprehensive way, would make a big difference to both the patients themselves and research that could be conducted to understand their experiences.

Data collection should be mandatory across healthcare, providing data to understand trends and needs and to accelerate research. We have already seen how this reporting can improve understanding and research for Cystic Fibrosis where the Cystic Fibrosis Trust's Registry has been collecting data for 20 years.

Recording patient data in this way could allow researchers to spot trends and inform commissioning, and ensure patients are contactable to receive innovative new treatments for rare disease as they are discovered.

Using patient records and the NHS app to give access to research: Patients with rare diseases could find out about research studies via the NHS app. This would allow researchers to maximise participation in trials, which is particularly important for rare diseases numbers of patients affected are much smaller.

Having new ways to hear about and take part in research would help level the playing field across the UK. Currently patients with rare diseases that have a centre of excellence nearby are the most likely to be able to participate in research and get access to the most innovative treatments and care. All patients should get access to research regardless of where they happen to live.

Access to international research studies: Identifying those living with rare diseases in the UK by using health data, will help researchers collaborate more easily with other countries. This can allow more UK patients early access to innovative treatments and care. This is particularly important for rare diseases where the pool of patients is too small to run studies exclusively in the UK.

When you think about how we could use technology in the NHS, what are your fears?

We are fearful that we will not see enough change in this area. This would include not investing enough in interoperable digital technologies and data infrastructure and capability to improve our understanding of rare diseases. Data about rare diseases in the UK is often incomplete, inaccessible and poorly coded. This is significantly hampering UK health research, and without efforts to improve, we could lose the opportunities described here.

Many people with rare diseases, particularly younger populations, are likely to want more ambitious use of technologies such as apps to take part in research and report treatment outcomes, and this is in stark contrast with the NHS that is dominated by outdated technology. However, we need to be mindful to avoid digital exclusion.

What technologies do you think the NHS should prioritise and why?

The way rare disease patient data is recorded and coded urgently needs to be standardised. As new diseases are identified all the time, diagnostic manuals need to be updated regularly, to allow new diseases to be included in records and coded correctly. Patients that have rare diseases that are as yet not identified must be included in data reporting so they can also take part in research.

Findings from research projects should also be integrated into electronic health records to inform care. For example, during a research project based in Manchester, using data from newborn babies to understand pharmacogenomics, researchers ensured the findings were recorded on the patient electronic record to help inform future healthcare.

AI and machine learning may accelerate the development- and repurposing of- drugs by helping to select candidates that are most likely to benefit from them. This could be a particular help for rare diseases where the funding is limited and patient numbers are small. These approaches might bring the cost of drug discovery and development down in the longer term, but will depend on high-quality data.

What technologies are you worried about and why?

The nature of having a rare disease means there will be very few others with the same disease in the UK. Extra care will be needed to anonymise patient data to retain patient confidence.

Like any area of medicine, there will need to be robust processes in place to ensure that patients are in control of their data and can understand how their data is used. This means providing clear and accessible information about storage and use of patient data, and engaging with patients about research.

Hospital to community

What difference – good or bad – would this make to you?

Opportunities:

Bringing care and research closer to home: The nature of rare diseases means that patients are often treated in a small number of centres of excellence, which may not be close to their home. Travel, especially for those with disability, can be inconvenient, costly and time consuming. The same is true for taking part in research.

Patients want and need access to this highly specialised expertise, but standard procedures such as taking blood and measuring blood pressure could be carried out in the community. These approaches could free up time for specialists in the centres of excellence to focus on care and research, with routine research monitoring taking place in community settings.

Research conducted in the community could also widen participation, giving access to a more diverse patient population. Some research projects will also be more suited to a community setting, to see if interventions such as exercise therapy or diet can improve quality of life.

Challenges:

The need to build research capacity and capability and rare disease knowledge in community settings: A move to community for patients with rare diseases would involve significant logistical and cultural changes. To ensure this didn't cause disruption to patient care and research, a carefully planned and considered approach would be needed.

To make the transition there would be a need to build research capacity and capability in community settings, such as more research-active GP practices and pharmacies.

Community clinicians would also need to build the necessary expertise and training to properly support rare disease patients. Research activities would need to be coordinated between the community and centres of excellence, and data and information would need to flow seamlessly between the different settings.

Patients are likely to need reassurance that their quality of care and their ability to take part in research trials wouldn't reduce as the care moved into the community. We know of examples where patients feel a community setting is not catering for their needs which has had a negative impact on their health.

Sickness to prevention

What difference – good or bad – would this make to you?

Prevention isn't an option for most rare diseases. But in lieu of an effective treatment for many of these diseases, research is vital to **help people to keep as healthy as possible, slow disease progression, and prevent the accumulation of co-occurring conditions.** The sooner that patients are diagnosed with a rare disease, the sooner they can get expert advice to empower people to live more healthily with their condition. Research is key to finding new and more effective ways to identify and diagnose patients with rare diseases earlier.

Newborn screening for more rare genetic diseases should be championed and embraced, rather than waiting until symptom onset to diagnose. Research such as the Generation Study can inform the best way to carry out screening to have the maximum benefit for patients and their families.

It can be difficult to make the economic case for screening for rare diseases, but for individual patients who are diagnosed many years earlier it can have disproportionately positive impact on their life. Research can help quantify the impact of early diagnosis and secondary prevention to make the case for these measures when effective.

What forms of prevention do you think the NHS should prioritise and why?

We need to look beyond 'preventable diseases' when we think about improving UK health. We need to find better ways to help people who are diagnosed with rare diseases to live as well and healthily as they can. This approach can also prevent progression of diseases, and comorbidities, even if there isn't a treatment.

Research in this area will be most effective if it is coproduced with affected communities to better understand their preferences and needs.

For more information please email policy@amrc.org.uk