

Millions of people with rare diseases to benefit from faster diagnosis and better access to treatment

- New technology and digital tools will support faster diagnosis, and improvements to virtual consultations will make it easier for patients to see multiple specialists at once
- Action plan developed with the NHS and rare disease community

Millions of people with rare diseases will benefit from faster diagnosis and new treatments following the launch today of England's first Rare Diseases Action Plan by Health and Social Care Secretary Sajid Javid.

There are more than 7,000 rare diseases, affecting an estimated 3.5 million people in the UK. Their complex nature means it is difficult for healthcare professionals to receive training on every condition or for patients to access the relevant specialist.

People living with rare diseases, such as muscular dystrophies or Huntington's disease, can go through multiple appointments and referrals before a diagnosis is made due to the complexity of conditions, making it difficult for individuals and their families to coordinate their care.

The action plan, published today, includes 16 commitments to further improve care and has been developed in partnership with NHS England and NHS Improvement, the National Institute for Health and Care Excellence, Health Education England, Genomics England, the National Institute for Health Research, NHS Digital and the Medical Research Council.

Drawing on the UK's strengths as a global leader in science, the Action Plan includes commitments on research, including an announcement of £40 million of new funding to the National Institute for Health Research (NIHR) BioResource, to further their work in characterising and understanding rare diseases.

The Action Plan will also help increase the ability to spot genetic conditions during the screening of newborn babies.

Health and Social Care Secretary Sajid Javid said:

"This action plan will speed up diagnoses and care and allow our fantastic workforce to better support patients, by drawing upon the UK's world-leading science and technology.

"I am committed to levelling up our health system so that everyone regardless of their condition can receive treatment that is tailored to their needs."

Actions include:

- Improvements to newborn screening so diagnoses can be made earlier,

including a new research pilot using whole genome sequencing to screen for rare genetic conditions in healthy newborns, and improvements to the way the UK National Screening Committee makes decisions on rare diseases

- A new digital tool called 'GeNotes', which will allow healthcare professionals to quickly access information on rare diseases to improve diagnosis so they can provide the right care for their patients
- Developing a toolkit for virtual consultations to increase the effectiveness of videoconference and telephone clinic calls, making it easier for patients to coordinate care between multiple specialists without the need to travel long distances
- Supporting access to new treatments through new programmes like the Innovative Medicines Fund, while continuing to work with NICE on new treatments being assessed.
- Monitoring uptake of drugs for patients with rare diseases to, by measuring the number of people accessing a drug and comparing with the number expected to access it, to ensure equal access to treatment across the country
- Pilot new approaches to care for patients with undiagnosed rare conditions. Following consultation with rare disease patients and their families, these pilots are currently under design, but examples could include a holistic one-stop paediatric clinic or a more targeted adult neurology clinic, or the use of virtual expert multidisciplinary teams.

The government will also continue investing in the development of nucleic acid therapies, for example, through the world-class Gene Therapy Innovation Hubs and the Nucleic Acid Therapy Accelerator.

Minister for Patient Safety and Primary Care, Maria Caulfield said:

"People with rare diseases deserve the best care and treatment. Marking Rare Disease Day 2022 by publishing England's first Rare Diseases Action Plan is a significant step in supporting people with rare diseases to access even better coordinated care and treatment.

"We have listened carefully to people living with rare diseases to make sure their needs and priorities are placed at the heart of this plan. We will continue to work closely with the rare disease community over the coming year to develop this even further."

Prof. Lucy Chappell, Chief Executive of the National Institute for Health Research said:

"Around one in 17 people will develop a rare disease at some point in their lives, so while they are individually rare, cumulatively these diseases affect a substantial proportion of the population. The impacts on these individuals and their families are wide-ranging. Our research needs to continue to address early diagnosis, effective treatments and supporting them to live well with their conditions.

"The work of the NIHR's BioResource has already helped produce some truly ground-breaking discoveries, such as those made through the 100,000 Genomes Project, and today's significant new investment will ensure it remains at the

cutting edge of the fight to understand rare diseases, and help the people who have them.”

Nick Meade, Director of Policy of Genetic Alliance UK said:

“Rare Disease Day is great timing for this step forward. The delivery of this plan despite the challenges of the current environment shows the commitment of the delivery partners to make meaningful progress.

“Our community’s voice has been heard more than ever in the development of this plan. This collaboration has helped us see how well these priorities of diagnosis, awareness, coordination and care can combine to have an impact greater than the sum of their parts. We are excited to move into the implementation phase and to see real improvements for people living with rare conditions.”

Professor Dame Sue Hill, Chief Scientific Officer for England, said:

“With genetics playing a role in over 80% of all rare diseases, genomics can be vital in delivering faster and more accurate diagnoses, as well as more effective treatments. The NHS Genomic Medicine Service is therefore key to helping more patients get the right treatment quicker and supporting this new action plan.

“The NHS already tests for more than 360 rare and inherited signs of illness covering around 3,200 rare diseases and 203 cancers through our National Genomic Test Directory, and despite the pandemic, has continued to secure new, innovative and life-changing medicines to help people with rare and genetic conditions, including one-shot gene therapies, transforming their lives.”

This follows the UK Rare Diseases Framework announced last year which set out priorities for all four nations to speed up diagnosis, raise awareness and improve treatment and care.

The devolved administrations will publish their own action plans by the end of 2022.