Specialised Healthcare Alliance
FOR EVERYONE WITH RARE AND COMPLEX CONDITIONS

GENETIC ALLIANCE UK

Manifesto for rare diseases

A rare disease is a condition which affects less than **1 in 2000** people[1]

There are over **7000** recognised rare diseases, which can be life-limiting and life-threatening[1]

1 in 17 people in the UK will be affected by a rare condition during their lifetime; this equates to 3.5 million people in our population[1]

75% of rare diseases affect children and more than 30% of children with a rare disease die before their 5th birthday[1]

Rare and complex diseases can be difficult to diagnose, challenging to treat and complicated to care for. In recent years, scientific developments have deepened understanding of rare diseases and led to advancements that offer hope of improved outcomes. Exciting innovations such as AI-driven diagnostics, genomic testing, cell and gene therapies, and digital care support could help to address the inequalities that people living with rare diseases face.

However, this will only be possible through genuine partnership between the government, NHS, patients, charities, and innovators, to unblock the barriers that are holding back progress. Strong political leadership is required to set clear expectations, ensure that all parts of the system work together, and provide the resources needed to deliver change.

The next government must take steps to build on the progress that has been made to date through initiatives such as the UK Rare Diseases Framework. Delivering on the ambitions set out in the Framework could improve the lives of millions of people by delivering faster diagnosis, better coordinated care, more awareness among healthcare professionals, and improved access to specialist care and treatment.

We would also like to see the next government set new ambitions. This manifesto sets out the shared perspective of the Specialised Healthcare Alliance and Genetic Alliance UK on priorities for the next five years.

1 Helping patients get a timely diagnosis

- Support faster access to testing and ensure a timely diagnosis once testing is complete, with clear targets for how long the whole diagnostic process should take
- Open further *syndrome without a name* (SWAN) clinics across the UK over the next five years to support individuals who do not receive a diagnosis after genetic testing, providing clinical support and opportunities for re-analysis in future
- Improve early diagnosis before symptom onset by expanding existing UK newborn bloodspot screening for rare conditions in line with international best practice, as well as learning from research studies focused on genomic newborn screening

2 Increasing awareness among healthcare professionals

- Ensure rare disease modules are included in medical curricula, including training on not only how to recognise rare disease symptoms, but how to engage with patients and families with sensitivity
- Create a digital tool for healthcare professionals specifically for rare disease diagnosis – one place where they can access information, receive answers to questions from other clinicians, and utilise and find e-learning resources created by patient organisations
- Promote the inclusion of rare disease training as part of continuing professional development courses, with a particular focus on primary care

3 Better coordination of care

- Introduce new roles to support people with rare diseases to navigate services, including named care-coordinators
- Ensure that mental health support is offered to patients and their families both whilst seeking a diagnosis and from the point of diagnosis onwards
- Enable access to digital technologies that can increase access to care outside of traditional settings, including through incorporating support for accessing specialist rare disease services remotely into the NHS App

4 Improving access to specialised care, treatment and drugs

- Publish a strategy aimed at accelerating access to breakthrough innovations for rare diseases, including AI-driven diagnostics, and cell and gene therapies
- Provide clearer signposting to NHS specialist services for people living with rare conditions, and enhance understanding of UK data about who has rare conditions and which rare conditions they have, so the NHS can provide targeted services and support
- Ensure the Innovative Medicines Fund works as intended to support access to new rare disease medicines

[1] UK Government (2021). *The UK Rare Diseases Framework*.