RESPONSE

10 Year Health Plan for England

December 2024

Introduction to this document

Genetic Alliance UK's response to the 10-Year Health Plan was developed following a workshop with our members on 12 November 2024. Thank you to everyone who attended and contributed their views. The response incorporates views shared in the workshop as well as views gathered via engagement in Genetic Alliance UK's wider work.

As discussed in our workshop , this consultation is the first of multiple opportunities to feed into this process. Genetic Alliance UK has secured a position on the Research, Life Sciences and Innovation Working Group, one of seven 'enabling' working groups that sit alongside four 'vision' working groups which will feed directly into the development of the 10-Year Health Plan.

If you have content, data or experiences that you would like us to feed into this working group, please share them with policygroup@geneticalliance.org.uk.

Question 1: What does your organisation want to see included in the 10-Year Health Plan and why?

The challenges

Although rare conditions are individually rare they are collectively common, affecting 3.5 million people in the UK. This means that millions of people living with different rare conditions are experiencing the same difficulties. Delivering timely diagnosis, better coordinated care, more awareness among health professionals, and improved access to treatment and care – the ambitions set out in the UK Rare Disease Framework – could improve the lives of millions. It is currently estimated that there are over 7,000 rare conditions, with new conditions regularly identified through scientific progress. Eight out of 10 rare conditions are caused by a change to someone's genetic code. Some conditions are so rare that there may only be one or two families in the UK affected by them. Other conditions are so new to science that they affect just one person, which means their condition remains undiagnosed and we can't give it a name.

Rare diseases can be both life-limiting and life-threatening, and disproportionately affect children. 75% of rare conditions affect children and more than 30% of children with a rare condition die before their fifth birthday. People with rare conditions and their families can face a lifetime of complex care and living with a rare condition can also have a huge impact on someone's education, financial stability, mobility and mental health.

The opportunities

The 10-Year Health Plan has the potential to build on many activities already underway to support people living with genetic, rare and undiagnosed communities. Genetic Alliance UK's response to this

consultation describes the key needs of the three and half million people affected by genetic, rare and undiagnosed conditions living in the UK, highlights ongoing progress and activity and specifies key next steps to expand on this progress and make the outcomes more equitable.

With so many key initiatives underway, it is crucial that momentum is not lost, and that the 10-year health plan focuses on enabling, facilitating and accelerating work that is already beginning to deliver change for some people with genetic, rare and undiagnosed conditions. With continued coordination from the renewal of the UK Rare Diseases Framework, increased ambition and more financial support, these promising early benefits can be spread to more people affected by genetic, rare and undiagnosed conditions.

Question 2: What does your organisation see as the biggest challenges and enablers to move more care from hospitals to communities? (Shift 1)

The challenges

Many rare conditions are lifelong and complex. As a result, people affected by genetic, rare and undiagnosed conditions often need support and expertise from a wide range of healthcare professionals such as GPs, specialist hospital consultants, specialist nurses, physiotherapists, occupational therapists, speech and language therapists, and learning disability nurses. This can mean having multiple appointments across different settings and on different dates. Effective coordination of care is essential to help people living with genetic, rare and undiagnosed conditions and their families minimise the impact of these appointments on their busy lives and help healthcare professionals to work together effectively to provide high-quality and joined-up care.

For people living with genetic and rare conditions, their care is already distributed between multiple hospital settings ranging from specialist centres to local trusts. For those with the most significant need, their care cannot be shifted from hospitals to a community setting, but a similar effect can be achieved by making their care more efficient. Hospital time can be freed up by working to make sure people with genetic, rare and undiagnosed conditions spend less time in hospital. Care coordination, or coordinating between an individuals' healthcare providers can achieve this. Sharing best practice will also make healthcare more efficient, networking between healthcare professionals working in the same discipline can achieve this.

Genetic Alliance UK published a report in 2023 - Care Coordination - geneticalliance.org.uk/wp-content/uploads/2024/01/Coordinating-Care-Report-2023.pdf which contains case studies on existing successful care coordination approaches in the NHS, and examples of people who need improvements to their care.

What's already happening? What are the next steps?

The UK Rare Diseases Framework's 3rd priority is coordination of care. Under this priority there have been three actions and multiple focus areas delivering change for people living with rare conditions. It is crucial that these steps forward are built upon with progress to deliver the 10 year plan.

Action 10: develop a toolkit for virtual consultations - improving use of videoconference and telephone clinic calls in services for patients with complex, multi-system rare diseases - owner NHS

England - complete.
Next step: Virtual consultations for people with complex multi-system rare conditions will mean less travel and more efficient use of hospital time - how much is this tool being used? Can it be expanded?

Action 20: commission research to provide the evidence needed to operationalise better co-ordination of care in the NHS - owner DHSC - extended. Funded by NIHR, a research consortium led by Professor Stephen Morris, RAND Professor of Health Services Research at Cambridge Centre for Health Services Research, University of Cambridge is underway, with work completing in May 2026.
Next step: It is crucial that the findings of this study are implemented rapidly.

Action 21: include the definition of co-ordination of care in all new and revised services specifications for patients with rare diseases, and ensure the priorities of the UK Rare Diseases Framework are embedded across NHS England highly specialised services - owner NHS England - extended.
Next step: NHS England's Highly Specialised Services are a great place to use new approaches to improve outcomes and efficiencies. It is crucial that these are then disseminated more widely into specialised commissioning, including that which is delegated to Integrated Care Boards.

The **NICE quality standard on transition from children's to adult services** was updated in December 2023 to include the experiences of people living with rare conditions. This will address the potential for organised paediatric rare disease care to become chaotic when people with genetic, rare or undiagnosed conditions transition to multiple disciplines in the adult environment. Chaotic care is dangerous and frustrating for the individual, but also an inefficient use of hospital time.
Next step: the UK Rare Diseases Forum has established an independent advisory group to work with NICE to develop a NICE quality standard which could sit across all care for people with rare conditions. This can be used to drive more efficient coordinated care.

Networking between clinicians
NHS England's Highly Specialised Commissioning team have established Rare Disease Collaborative Networks, overseen by NHS England's Rare Disease Advisory Group. The 20 RDCNs bring together motivated, expert clinicians to harmonise care provision and share best practice. This is an unfunded programme.
Next step: The UK's Rare Disease International Mirror Group hosted by Newcastle University is working to examine the impact of the UK's challenges in engaging with European Reference Networks (ERNs) for rare conditions. RDCNs are conceived 'bottom-up' led by clinicians and are not driven by population need. ERNs divide the field of rare conditions into 24 intersecting divisions, this means everyone with a rare condition should have at least one ERN which can drive progress in care and research. Could RDCNs expand to become more equitable and be funded to drive excellence?

Mental health
There are many aspects of genetic, rare and undiagnosed conditions that contribute to poor mental health. These include chronic health challenges, an unpredictable future, anxiety about personal or family member's health, the impact of progressive conditions and of course grief. It is important to acknowledge also that poor care coordination for people living with rare conditions can directly cause negative mental health outcomes. (BMC Health Serv Res. 2022 May 14;22:648. doi: 10.1186/s12913-022-08060-9 Mental health care for rare disease in the UK – recommendations from a quantitative survey and multi-stakeholder workshop.)

As per the UK Rare Disease England Action Plan (2024), in 2023 NHS England emphasised to integrated care boards and providers, 'that mental health services should be offered based on need

and should not exclude anyone because of a particular physical health or neurological diagnosis'. 'All new and revised NHS England service specifications for patients with rare diseases are now required to consider users' psychosocial needs and ensure co-ordinated pathways for access to mental health support.'
Next step: In the context of NHS capacity challenges, a significant burden of funding mental health support for families affected by genetic, rare and undiagnosed communities is falling to individual support organisations. While this point could equally be made as part of the shift from treatment to prevention, investment in mental health services for this community will reduce more serious mental health challenges for the future.

Question 3: What does your organisation see as the biggest challenges and enablers to making better use of technology in health and care? (Shift 2)

The challenges

We know surprisingly little about the 3.5 million people in the UK who are estimated to be affected by rare conditions. Which conditions affect them? What is the prevalence of these conditions? What causes these conditions? What services and treatments are available for people living with these conditions now, and how can we build on and improve the care that they receive in future?

Genetic Alliance UK's 2024 report, the Stats Behind the Stories argues that we need to segment and better understand UK data about who has rare conditions, and which rare conditions they have, so that the NHS can provide the right services and support. As our member, Hereditary Brain Aneurysm Support says: "It's often said in the rare disease world that if you can't be counted, then you don't count. And of course, counting data is counting people. Each data point has a person and a powerful story behind it." Our report found opportunities to:

- improve diagnosis of non-genetic rare conditions by examining healthcare records more systematically
- use incidence of more common indications to better understand the prevalence of rarer conditions
- examine the equity of care provision for rare conditions
- act on inequity by defining populations in with an inclusive approach

It is well understood that collecting data about an individual's rare conditions can benefit their own healthcare, can benefit research, and can improve the quality of commissioning decisions on a condition by condition basis. The Stats Behind the Stories argues that overarching planning and commissioning decisions on a population level will benefit from using the same data, and that this will drive equity, including between conditions.

A key finding of our report was that it was challenging to answer relatively simple questions about commissioning and diagnostic arrangements on a condition by condition basis, which demonstrates gaps in the UK's dataset for people living with genetic, rare and undiagnosed conditions. To fill this gap would provide a tremendously valuable foundation on which we can build equitable innovation in care and research. Our report concluded that working with the National Disease Registration Service, incorporating the National Congenital Anomaly and Rare Disease Registration Service, would be crucial for success in this area.

What's already happening? What are the next steps?

The **National Congenital Anomaly and Rare Disease Registration Service** collects data on rare conditions via the NHS and via research activity within the NHS. A minimum data set has been defined, and NHS England Highly Specialised Services contribute data.

Next step: This approach needs to be better funded and more routine. It is only through systematic expansion of the registration of rare conditions that this data set can match up against the impact of the National Cancer Registration Service.

The **National Genomics Research Library** is a database that (with consent) collects the genomes from people accessing diagnoses through genome sequencing and from participants in research studies and enables approved researchers to work with the de-identified genomic data alongside their health data. **DECIPHER** (DatabasE of Chromosomal Imbalance and Phenotype in Humans using Ensembl Resources), is a database of genomic variation initiated in the UK and now funded by an international research consortium. This is a powerful tool that presents genomic data for a patient registry in a dynamic up-to-date searchable interface. The **NHS Genomic Medicine Service** and the **Generation Study** contribute to and use these tools.

Next step: As well as the vital use of these data sources and resources to drive innovation in the UK, they also need to be examined for their potential use in examining the quality of care provision for people with genetic, rare and undiagnosed conditions.

Question 4: What does your organisation see as the biggest challenges and enablers to spotting illnesses earlier and tackling the causes of ill health? (Shift 3)

The challenges

Eight out of ten rare conditions are genetic in origin, and the vast majority of all genetic, rare and undiagnosed conditions cannot be prevented. However, early identification and diagnosis of genetic, rare and undiagnosed conditions can massively improve outcomes and potentially significantly reduce ill-health.

The NHS genomic medicine service has delivered a powerful genomic model for diagnosis, which brings tremendous potential for shortening the diagnostic odyssey for people and families affected by genetic, rare and undiagnosed conditions. However there are two significant gaps in the diagnosis environment in England that need to be addressed: screening, or presymptomatic identification of people who will develop genetic, rare or undiagnosed conditions; and the diagnosis of non-genetic rare conditions.

Screening The UK National Screening Committee (UK NSC) is the body that advises ministers and the NHS on which conditions to screen the population for. Currently, in England , the heel-prick test for newborns screens for nine conditions, and in 2022 the UK NSC recommended that tyrosinaemia type 1 be added to this list (it is not yet screened for). The UK is clearly out of step with the majority of developed countries in screening policy development and decision-making, as there are more than 20 European countries that screen for 20 or more conditions using the same technology. The failure in

joined up policy-making is most clear for families with children affected by spinal muscular atrophy (SMA).

NICE first granted access to an innovative disease-modifying treatment for SMA in 2019. There are now three treatments that have been approved. All of these treatments would be best delivered to babies before they develop SMA, as this would mean the treatment would permanently cure them of the condition. Sadly, because the UK does not screen for SMA, most children must go through a diagnostic journey before they can access the treatment, which is often delivered after they have permanently lost the ability to walk. Four years after the first medicine was granted access by NICE, UK NSC in June 2023 approved the beginning of a planning process for an in-service evaluation (ISE) of screening for SMA in real world NHS services. Around 300 babies with SMA would have been born during this four year period.

Non-genetic conditions The valid emphasis on genomics in the diagnostic pathway for rare conditions poses the risk that conditions that are not genetic in origin are not detected and people with these conditions get 'stuck' in a cycle of genomic testing. To reduce the diagnostic odyssey for all people living with genetic, rare and undiagnosed conditions, it is important to build non-genetic diagnostic tools and pathways into genomic diagnostic pathways.

What's already happening? What are the next steps?

The Generation Study The UK has an opportunity to take a quantum leap forward in newborn screening with the Generation Study, run by Genomics England, which is the most ambitious research project to examine the potential of newborn screening using whole genome sequencing in the world. The study is examining the power of the technology to screen for 100s of conditions at birth.
Next step: without urgent action to assess the UK NSC's capability to make decisions about newborn screening in the context of the results of this study, there is a real risk that Genomics England will publish the results of its study, and other countries will be able to act quicker to implement its findings. England stands to watch as its key decision-maker in newborn screening lacks both the methodology and the capacity to assess the opportunities that the generation study may deliver.

Blood-spot task group The 2022 England Rare Disease Action Plan launched a Blood-spot task group to develop approaches to evidence gathering and development that can support good decision making on rare condition newborn screening.
Next step: with newborn screening recommendations, such as UK NSC's 2022 decision to recommend screening for tyrosinaemia type 1, unimplemented, it is clear that there are wider issues affecting England's progress towards an environment where newborn screening can be a more effective tool to identify conditions early. Workforce and capacity issues appear to be holding back implementation of the 2022 recommendation as we approach 2025.

Non-genetic conditions The DHSC organised a working-group on non-genetic conditions in 2023. The report identified strands of existing work that could specifically help to address this challenge, such as specialised networks, educational tools and a need to focus on collecting data. The importance of supporting the referral from primary care that starts a diagnostic odyssey was also identified as a key area that deserves further attention.
Next step: if we are to have a comprehensive and equitable pathway for diagnosis it is important that the recommendations from the non-genetic conditions working group are taken forward.

Question 5: Please use this box to share specific policy ideas for

change. Please include how you would prioritise these and what timeframe you would expect to see this delivered in, for example:

- Quick to do, that is in the next year or so
- In the middle, that is in the next 2 to 5 years
- Long term change, that will take more than 5 years

Priority in the next year

The benefits of the next steps discussed so far will amplify each other. Better diagnosis will improve data collection, and better data collection will improve decision-making about commissioning diagnostic pathways. Care can be more easily coordinated with early diagnosis and better data collection. More organised care can lead to more efficient data collection.

Genetic Alliance UK's response identifies ongoing activity that is gradually improving the environment; the majority of these activities have come from the UK Rare Diseases Framework, which comes to an end in 2026. All of these activities will benefit both from increased capacity within the NHS and the agencies responsible for delivering and overseeing this progress (particularly UK National Screening Committee, screening infrastructure and teams within the NHS, the Genomic Medicine Service, and the National Disease Registration Service), and from continued coordination and oversight that will be facilitated by a policy successor to the UK Rare Diseases Framework. Many of the ongoing activities relating to that policy will continue after its term comes to an end, including studies on newborn screening (the Generation Study) and on care coordination (CONCORD 2).

Most importantly the UK Rare Diseases Framework has an established and sophisticated infrastructure for consulting and engaging people living with genetic, rare and undiagnosed conditions that has facilitated the progress to date. It is crucially important that this momentum and community are not lost.

Medium term priorities

The Generation Study will deliver the results of the most powerful study ever on the value of genome sequencing for newborn screening soon. If urgent changes are not made to the UK's capacity and approach to decision making on screening for rare conditions, then England risks publishing groundbreaking results that we are not ready to react to.

Overarching priority

We do not know how many people live with genetic, rare and undiagnosed conditions in England. We do not know who individuals with specific conditions are and how to reach them. We do not know how many people might be living with treatable conditions but no diagnosis. We can only begin to solve these problems if we make sure that England's health data infrastructure is systematically recording everyone living with a genetic, rare or undiagnosed condition. The National Disease Registration Service needs to be empowered to do this.