

GP 10 Year Health Plan response

Name of organisation Gene People

Q1. What does your organisation want to see included in the 10-Year Health Plan and why?

Gene People

Gene People is a registered charity working in the UK. We provide support and information to those living with a genetic condition, their families and carers. We strengthen the resilience of the condition-specific support groups that work with individual communities. We advocate and collaborate with all stakeholders to achieve change so that the lives of our communities are improved.

In this response, we are focusing on the experiences of those living with a rare genetic condition rather than the common conditions as we know that responses are being submitted by large patient organisations for the common genetic conditions, whereas rare and ultra rare genetic condition-specific support groups are usually unable to participate in consultations.

Gene People is also a participant in several coalitions that will be making submissions to this consultation. We endorse the responses from the Specialised Healthcare Alliance, Prescription Charges Coalition, Genetic Alliance UK and the Disabled Children's Partnership.

The rare disease context

There are approximately 3.5 million people affected by rare conditions in the UK (<https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework>), which means many more people from their wider families are involved in caring for and supporting them.

Estimates of the numbers of rare diseases have, until recently, suggested that there are around 7,000 conditions. Rare-X have published a report that posits that the number of rare diseases exceeds 10,000 (<https://rare-x.org/blog/2022/06/07/rare-x-releases-new-report-that-uncovers-large-number-of-previously-uncounted-rare-diseases/>), suggesting a considerable amount of mis- and under-diagnosis. Approximately 80% of rare conditions have a genetic cause. Very few conditions are screened for currently, although projects like the 100,000 Genomes Project have been seeking to advance diagnosis for patients.

Most rare conditions begin to present symptoms in childhood, with a significant proportion of those conditions (more than 30% [<https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework>]) resulting in the death of a child before their fifth birthday. There are treatments for approximately 5% of rare conditions and access is defined by local arrangements.

Many rare diseases need high levels of health and social care interventions and can have a high impact on family life, including increased financial pressures and poor mental health(<https://bmchealthservres.biomedcentral.com/articles/10.1186/s12913-022-08060-9>, <https://contact.org.uk/help-for-families/campaigns-and-research/counting-the-costs/> , O’Dwyer et al. <https://doi.org/10.1016/j.comppsycho.2021.152261>, [https://www.thelancet.com/journals/eclinm/article/PIIS2589-5370\(21\)00514-9/fulltext](https://www.thelancet.com/journals/eclinm/article/PIIS2589-5370(21)00514-9/fulltext)).

In many cases there may be biological (as well as psychological and social) drivers of emotional and behavioural symptoms. For example, there may be direct brain and behaviour interactions in relation to the genetic and neurodevelopmental under-pinning of rare diseases. Children with intellectual disability – which is common in the rare disease group - are also known to have greater need for help with emotional and behavioural symptoms and have higher rates of mental ill-health.

Having a family member with a rare condition can cause several social determinants of health inequalities, such as poverty, issues with employment, educational attainment, and housing (https://www.health.org.uk/publications/reports/the-marmot-review-10-years-on?gclid=CjwKCAjwqauVBhBGEiwAXOepkaON3KNp93I5Se7nFcBvdE4blLa8aBEprm9_1QO4d7r75tqaMdOh7BoCwRoQAvD_BwE). Having a rare disease in a family is itself a health inequality, given the lack of services and understanding for these conditions.

Despite the prevalence of rare and genetic conditions as a whole, they are largely overlooked. As these conditions can individually impact specific organs or be totally systemic and many people and families require extensive social care support, our community experiences the entirety of the health and social care system. Mostly, there is a sense that this community is let down by these services because of the complexity and diversity of the conditions and the sometimes small numbers of affected people. In our view, if the NHS can work for this community it will be able to work for everyone, therefore, the experience of the rare and genetic conditions’ community should be viewed as the litmus test for the NHS and its performance.

How NHS rare services work

NHS services for rare conditions are not uniform in their delivery; for very rare conditions services may be nationally commissioned with very few specialist clinicians, other rare conditions may have local or regional specialist centres with expert clinicians, but many families rely on the care and support locally available, struggling to get a timely diagnosis and access relevant specialist knowledge.

What we would like to see in the Plan

Gene People and our community welcome the commitment to publish a Rare Disease Action Plan in 2025, however, there is much that the 10 Year Health Plan could include to improve the health outcomes and lives of people with a rare disease:

- The diversity of the UK rare disease population should be acknowledged in service design, clinical trial offerings and the treatments made available. This diversity is multi-dimensional, including, for example, the differences between conditions, cultural, linguistic, ethnic and geographical factors
- The importance of properly integrating health with social care is magnified for our communities as the majority do not have access to disease modifying treatments
- There needs to be specific mental health provision for families living with a rare disease. Gene People led a consortium that responded to the Mental Health and Wellbeing Plan consultation, which can be found on our website (<https://genepeople.org.uk/about-us/national-policy/>)
- A systems approach is required to ensure that the different parts of the life sciences system work together without duplication of effort to achieve fast access to innovative, effective and safe treatments for rare disease patients where these exist.
- For those with no prospect of a treatment or who choose not to take a drug, the system should work with them not against the individual and their carers with care properly co-ordinated and seamless access to multi-disciplinary support as needed
- The recommendations from the O'Shaughnessy Review on Clinical trials should be implemented at pace
- Siloes should not exist in a 21st-century healthcare system; every clinician should be able to access information about a patient without needing them to re-explain their situation each time
- In all things the patient should truly be at the centre of decisions, not the convenience of the system
- There is a disparity between the screening programmes offered to newborns in Europe and the UK. The efficacy of many treatments for rare disease hinge on early diagnosis, which is hampered by healthcare professionals lack of knowledge of rare conditions. Screening is a key tool to unlock diagnosis and the health benefits of swift treatment for the patient and healthcare system. We support the Generation Study, believing that the possibility it has created for early diagnosis of over 200 rare conditions where early detection creates the opportunity for disease modifying interventions will be rolled out nationwide following the completion of the pilot phase.
- The UK is beginning to falter in the number of innovative technologies available for rare disease patients as it is being viewed as increasingly uncompetitive in

reimbursement and difficult to operate in. UK patients should have the same opportunity of accessing a treatment as any other person in Europe.

Q2. What does your organisation see as the biggest challenges and enablers to move more care from hospitals to communities?

For the rare disease community this shift is more about fewer hospital admissions and a shorter diagnostic odyssey rather than moving out of hospital care.

Challenges include:

- The diagnostic odyssey for rare conditions is famously protracted and can take several years. During this time, the patient will undergo multiple tests, sometimes in different departments with different specialist clinicians. In some cases, a late or mis-diagnosis can result in harm being done to the patient with unnecessary or inappropriate treatments.
- There have already been advances of homecare arrangements for some rare patients. There can be an increase in burden on the patient and/or care giver to coordinate care across departments and suppliers, which is an unwelcome side effect of at home treatment. There can be issues about safe storage of equipment and the ability to maintain sterile environments. Moving the care from hospital to the home would need to be fully supported with additional staffing and expert care givers and not expect the family to become the main medical support.
- Lack of understanding of the range and complexity of rare conditions by healthcare professionals with a focus on individual symptoms rather than a holistic approach to a person's health

Enablers include:

- A way to reduce reliance on hospital to manage symptoms is to shorten the diagnostic process by encouraging clinicians to consider that a condition might be rare sooner, for example after a certain number of return appointments with the GP with one symptom possibly alleviated but other symptoms remaining.
- A crucial way to reduce hospitalisations is to invest in disease modifying or symptom alleviating treatments. Whether the NICE processes are supporting timely access to such treatments is debatable. It should be noted that the patient voice in any drug development and appraisal is critical as listening to what really matters to patients will result in higher take up of treatment thereby more improved health outcomes.
- Innovation in treatments such as gene therapies hold great promise but may not be curative. Incremental change in a disease may be sufficient to significantly impact the lives of patients and reduce hospital admissions. This will vary from disease to disease and the efficacy of the treatment. These incremental

improvements must be rewarded and encouraged within the reimbursement system because of the benefits to the patient and health care system.

Q3. What does your organisation see as the biggest challenges and enablers to making better use of technology in health and care?

We assume for that this shift is referring to everyday technologies not technologies such as gene therapies.

The biggest **challenges** are:

- Lack of digital infrastructure within the NHS – we hear about some Trusts using faxes and paper records
- Lack of investment in technology – the disbanding of NHS Digital sent the wrong message about the prioritisation of this within the service
- Inconsistent levels of digital literacy within the workforce
- Digital exclusion of some parts of the population leading to their not being able to participate with digital health care systems. NB this is not purely to do with age and should not be treated as such
- All parts of the NHS not speaking to each other digitally causing issues for all patients but especially rare disease patients who sometimes carry their own ‘passports’ in order to get the care they need.

The biggest **enablers** to resolve the system challenges would be investment and focus.

Q4. What does your organisation see as the biggest challenges and enablers to spotting illnesses earlier and tackling the causes of ill health?

Causes of ill health in genomics have to be sensitively handled as we are not proposing or endorsing eugenics at all. We are proposing that families are given the information, advice and tools to make informed decisions about the lives of their loved ones who have conditions.

Challenges include:

- The number of diseases tested for at birth is behind the number tested for in Europe. Increasing the number of diseases tested for could dramatically improve health outcomes
- Healthcare professionals lack awareness of rare diseases and are reportedly taught to only think about common conditions. There is a lack of understanding about what genetic tests GPs can order elongating the diagnostic odyssey. There is a perception that ‘treatment drives diagnosis’ meaning if a treatment does not exist then diagnosing the disease is not beneficial which leads to underreporting

of disease, thereby hampering research efforts, and not allowing families to access other forms of support or to make informed decisions about their lives

- There is a low level of public health literacy around genetic testing and screening, that particularly disadvantages those from minority and stigmatised groups.
- Siloed data – there is little cross-referencing of data which could lead to identification of new conditions and with that the ability to provide support to those families and seek treatments.
- An acute shortage of clinical geneticists, genetic counsellors and laboratory scientists able to test, diagnose and counsel at risk families enabling them to make informed reproductive choices and, if they wish, avoid the birth of a child with a life limiting genetic condition.

Enablers include:

- Planning and service delivery undertaken on a national level so as to not recreate differences between conditions. The majority of the calls to the Gene People helpline are about screening, especially from wider family members who cannot currently access testing for many conditions
- The Generation Study screening for treatable rare genetic conditions needs to be fully evaluated on completion, including input from participants and patient organisations, and recommendations followed for implementation if the pilot has been successful
- Investing in the development of a skilled workforce with the future in mind, that is, not planning for today's needs but using the well-developed NHS horizon scanning capability to determine what roles will be needed and when
- Utilising the computational skills and expertise that the UK has to unlock data from siloes to serve patients better, with proper data governance and security in place
- Improving public health literacy through reviewing the school curriculum and public campaigns aligned to the prevention agenda

Q5. 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**

Begin to actively plan for the evaluation of the current Rare Disease Framework and the development of the next one

Work with ICBs to ensure geographical equity across diseases for patients and families

Start to address the cumbersome processes of the National Screening Committee for adding new conditions to the neonatal heel prick. This is particularly important for non-genetic rare conditions

Raise the awareness and visibility of the rare and genetic conditions community within the NHS by the appointment of a Rare Disease Tsar. The appointment of Dame Lesley Regan as Women's Health Ambassador has improved the focus on women's health immensely. We consider that rare and genetic conditions have such a significant impact on the NHS and are sufficiently complex to warrant a tsar. Gene People would be able to assist with nominations to this post for announcement on Rare Disease Day 2025.

Ensure that the 10 Year Health Plan does not cause any unintended consequences or overlap of effort or responsibility with the other plans and strategies currently in progress. This would be easily and transparently demonstrated by a publicly available document, table or spreadsheet that outlines each plan and strategy and who is responsible for delivering each action. The plans and strategies that impact the rare and genetic community most are: Rare Disease Framework, O'Shaughnessy Review, Genomics Strategy, Workforce Plan, Life Sciences Strategy and the Nursing and Midwifery Genomics Framework.

Bring together the Deans of Medical Schools to agree a common approach to introducing appropriate awareness of rare diseases into the undergraduate medical curriculum

Look at the sustainability of specialist provision for rare disease families and instigate succession planning arrangements

Collaborate effectively across the four nations of the UK in the delivery of equitable and timely specialist care for rare disease patients and families.

Strengthen opportunities to engage with patients and families in priority setting for service provision

Reinstate clinical genetics as a clinical speciality within the Research Delivery Network

Develop plans to systematically address inequalities in access to NHS services and support and address barriers to access to participation in R&D.

• In the middle, that is in the next 2 to 5 years

Recognise and support the contribution the Patient organisations make to the delivery of expert care to rare disease patients and families through the provision

of funds and the development of capacity building programmes that will increase their effectiveness and secure better integration with statutory sector agencies.

Expand the numbers of laboratory scientists, genetic counsellors and clinical geneticists to increase capacity to ensure that families where a newborn child with a rare condition is identified through Whole Genome Sequencing can be seen and given appropriate information and support quickly.

Invest in the training of laboratory scientists, genetic counsellors and clinical geneticists to increase capacity to match demand.

Develop training programmes for community midwives to enable them to explain the screening programme to pregnant women and their partners when the Generation Study is rolled out nationwide.

Review the effectiveness of processes used by NICE, ILAP, EAMS etc to determine whether these are appropriate and effective in the light of emerging technologies such as gene therapy.

Plan for the adoption of new ways to integrate and coordinate care through emerging technologies such as AI and remote monitoring.

Boost mental health services and develop the ability of mental health practitioners to work with patients for whom mental health issues are an aspect of their syndrome as well as supporting the wider family to enjoy as normal as possible a family life.

Address the reasons why the number of clinical trials in the UK is falling.

Remove barriers to collaboration with colleagues in the EU and make it possible for UK clinicians and scientists to participate in ERNs and pan-European research and development programmes.

Develop and fund placements for early career doctors in rare disease patient support organisations and research funding charities.

- **Long term change, that will take more than 5 years**

Integrate health and social care for rare disease patients and families

Support prompt and comprehensive delivery of Education Health and Care plans so children with rare diseases are able to fulfil their potential to the fullest extent possible

Boost funding for rare disease research and development through NIHR, MRC, LifeArc etc.

Create a comprehensive digital NHS record keeping system that actually enables accurate, relevant and comprehensive patient data (including that entered by patients and families) to be accessed promptly by those with a legitimate need to do so.

Develop partnerships with industry such that relevant novel technologies can be anticipated and their introduction to the NHS planned for.