



the genetic conditions support network

Gene People response to the Genomic Medicines Service Specification August 2025

Who this response is from

Gene People is a national charity working with the many thousands of people affected by genetic conditions – not just those who have symptoms, but those around them who may need testing, advice and help. We aim to improve the lives of anyone affected by a genetic condition by providing the information and support they need at the time they need it.

Gene People spans frontline services to influencing national policy by:

- Providing information and support via our free genetic counsellor-led helpline
- Creating web-based resources to inform families and healthcare professionals
- Acting as a focal point for the many small condition-specific charities and groups with free training sessions
- Acting as a resource for the community by administering a free membership scheme for genetic condition-specific support groups
- Advocating on behalf of the genetic community and inputting to national policy initiatives and consultations.

Our helpline takes 150-200 enquiries each year with almost half of these being regarding conditions we will be contacted about only once. Our partnership network of condition-specific support groups numbers over 160, many of which are patient- or parent-led.

Our Response

Gene People welcomes the ambitions for caring for the rare disease community as set out on page 57 'Rare disease', especially the ambition to reduce the diagnostic odyssey. This will reduce stress and anxiety for families and reduce cost for the NHS.

Gene People also welcomes the commitment to embedding patient and public voice (PPV) within the Genomic Medicines Service as set out in the Principles and in section 22 on page 14 referring to the PPV roles that are required in each regional service. We would like more detail on how this will work in practice so that the experience and voice of people living with rare disease will be represented and heard particularly when many organisations in this field are very small and under-resourced.

In addition, we are unclear as to whether giving the responsibility for PPV to the communications function is the correct place for this to sit if the emphasis is on achieving co-production of changes to service design and delivery of services.

Throughout the specification there are three distinct health areas: Rare, Cancer and Population Health. We are concerned that each area is siloed from the other. We would welcome sight of the internal structures and frameworks that will enable advances in one to be advances in all and to reflect the patient experience. For example, the diagram on page 11 does not show arrows between the clinical areas – how will learnings and efficiencies be shared across the three areas regionally and nationally?

Our expectation is that technology will continue to evolve and therefore ask that the most effective technology available is being used to give the highest diagnostic yield for patients, and that the technology and testing used is clearly and consistently communicated in the lab reports to clinicians and patients. Having flexibility when thinking about rare disease is crucial, making section 46 on page 33 ‘Whole exome sequencing’, a concern.

We are confused by the section relating to Genomic Point-of-Care and Near-Patient Testing (p37, sections 64-67). It is our understanding that health technologies are now to be assessed for cost-effectiveness by NICE whereas this section implies that cost-effectiveness will be assessed by the NHS. It would be useful to clarify this matter. This section also suggests that the deployment of Point-of-Care and Near-Patient Testing will be led locally, which could create inequity of access across England and potentially to specific communities. The expertise of those making decisions locally is not made explicit in the Specification, and it is unclear if these specialist decisions will be made by people with the necessary specialist expertise. Patients and the public would, we believe, expect that those taking such important decisions would have the requisite expertise so as to safeguard their health.

There is a word missing in section 73 on page 39.

We noted the reference to cascade testing on page 47 in section 97 for population health. We request that a similar commitment to deliver cascade testing for family members for rare diseases be included.

Gene People welcomes the clear emphasis on data and system interoperability (p76). This could have very positive implications for our community, both those with rare and population health conditions. We do ask that the registries owned, curated and funded by patient organisations are included in considerations as they are a valuable and under-utilised resource.

We welcome the role of Rare Disease Scientific Lead (page 154) to provide focus on this complex area. We would suggest that there is a need for this role to also collaborate with peer counterparts in the other devolved nations.

We welcome the inclusion of working with patient organisations in the role description for the Rare Disease Head of Programme and in the Research and Innovation Head of Programme (pages 160 and 174). We would appreciate sight of the work programme and deliverables for these items in due course. Gene People would be happy to share our thoughts on what should be included.

Gene People recognises that this is a clinical specification, however, the document itself refers to working in partnership with other parts of the healthcare system. We, therefore, ask that as mainstreaming of genomics continues that the Specification includes the need for reports to be clear and consistent as it will be increasingly that these are used by non-genetic health professionals and/or patients and families.

The availability of the geneticists to assist clinicians and healthcare professionals also needs to be made clearer. For example, can questions be answered only in writing or also by phone? If so, this availability needs to be made known to those who would need that service.

We would suggest that how this Specification intersects with educating and informing healthcare professionals about how and when to refer to the Genomic Medicines Service is made clearer, as we know from our own helpline that currently clinicians can be confused as to when they can or cannot do so. The drive to 'think genomics' is welcome and there are several patient organisations that could help with that endeavour. It should also be recognised that not all rare diseases are genetic in causation.

We would also suggest that, on finalisation of the Specification, a lay summary is produced for patients and the public to understand what is available from their Genomic Medicines Service locally and nationally and what is not.

We look forward to the publication and consultation on the 10 Year Workforce Plan and the specific workforce plans under the remit of the NHS Genomic Medicines Services leadership's control. The need for sufficient high-quality staffing across England is fundamental to achieving the ambition set out in the Specification.