

# GENE PEOPLE

the genetic conditions support network

## Gene People's Response to the Clinical Genomics Service Specification Consultation

October 2022

### Introduction

Gene People responded to NHS England's consultation on the Clinical genomics service specification document.

The following text is the responses submitted online via the consultation website. Where the question was a selection on a scale, the answer given is stated.

### Document 1: Clinical Genomics Service Specification

#### **Q11. To what extent do you think the service specification achieves its key priorities to:**

1. Embed clinical genomics into mainstream clinical pathways? To some extent
2. Provide care for patients and their families in a coordinated way? To some extent
3. Ensure all those that would benefit from the clinical genomic service and/or genomic testing are able to access it? To some extent
4. Improve outcomes for patients and their families? Unsure/Neutral
5. Improve access to and the provision of services by aligning clinical genomic services to the NHS Genomic Medicine Service national network? To some extent
6. Ensure the workforce has capacity to implement the key priorities arising from the Service specification and meet increasing demand for services? Unsure/Neutral

**Q12. Please provide any further information relevant to how well you think the Service specification achieves its key priorities.**

The service outlined in the specification is absolutely one that Gene People supports. We also feel that the genetic condition and rare disease communities' voices have been heard and their views incorporated. However, even with allowing for implementation to take time, we still have some concerns about delivery of the service specification in practice and how this might impact on patients.

- **Embed clinical genomics into mainstream clinical pathways**

How prepared are other specialties to engage with mainstreaming? It will be a notable increase in workload for them. How much do other specialties appreciate the utility of genomics and the types of patient outcomes that are traditionally sought in clinical genetics, such as, patient empowerment and adjustment? How will a genetic diagnosis and related psychosocial support for patients be given clinical utility and measured in the work of other specialties? In our experience at Gene People, even when a genetic situation is relatively straightforward, e.g., Carrier testing for CF, there are psychosocial issues and confusion that can arise for patients. To date the mainstream setting does not seem adequately equipped to respond, even though CF carrier testing has been taking place in this setting for some time.

It is not clear how GCs based in mainstream services will interact with clinical genetic services and develop pathways. We feel these links need to be strengthened and formalised so that everyone understands their role and the boundaries.

Despite the training and support available for mainstream clinicians with implementation of genomics into their practice, we still have concerns about the workforce in the mainstream setting having adequate skills and time to enable accurate genetic test result interpretation and communication of results to patients.

- **Provide care for patients and their families in a coordinated way**

Co-ordination of care is a crucially important element of the Rare Disease Framework and a real concern for families. In our experience these needs are more likely to be met in parts of England where there are dedicated rare disease centres, such as, Birmingham. There is an infrastructure in place

which allows more effective development of MDT working. In other areas the infrastructure is much more challenging. Setting up joint working and MDTs is not solely the responsibility of clinical genetics, which is a small specialty within the NHS. Is there reciprocal understanding with other specialties?

There is much within the Service Specification that relies on time and commitment from other specialties and services. How willing and able they are to take this on makes a significant difference to the success, or not, of achieving the service as it is outlined. We feel this needs to be acknowledged.

- **Management of patients by clinical genetics**

Many patients and families would love to have management from clinical genetics, and we would support this happening. Over the past decade it seems to us that management and follow-up from genetics has generally been decreasing with families reporting to us that once the genetic diagnosis has been made by the genetic team they are then discharged back to the care of others, e.g., paediatricians. This has included those with complex, multi-system conditions. Are clinical genetic services willing and able to provide long-term management for those that need it?

The specification appears to expect a substantial advisory role of clinical genetic service clinicians. We are concerned about how realistic this is. Will there be capacity to provide advice on the scale that it is likely to be needed? A related concern is that if a large proportion of clinician time is shifted to providing advice to other professionals rather than face-to-face patient contact, will this risk losing skilled clinicians, and detrimentally impact the attraction of clinical genetic roles to skilled people who want to do patient facing work.

- **Psychological and mental health needs**

We welcome integration of mental health services for people with genetic conditions. How much of the psychological needs of patients impacted by genetic conditions should be provided within clinical genetics and when should patients be referred on is unclear. When referral to specialist psychological services is needed, there are concerns about the capacity of these services, waiting times and their eligibility criteria. Also, how familiar

practitioners may be with the specific needs and experiences of people affected by genetic and rare conditions.

Gene People was involved in responding to the recent Mental Health and Wellbeing consultation, available here:

<https://genepeople.org.uk/about-us/national-policy/>.

- **Patient outcome measures**

These seem to be focused on evidence of documentation in the clinical notes. It is also vitally important that results and decisions are communicated to patients evidenced in letters to patients.

We appreciate the considerable effort and thought that has been put into developing this service specification and we fully support its aspirational vision, with the hope that it will become reality.