Clinical Genomic Services: Revised Service Specification Public Consultation

Consultation Guide

Contents

Purpose of this document

NHS England is committed to working with a wide range of patients, patient groups and other stakeholders in the development of its commissioning of services. A public consultation is an opportunity to check whether proposals are right and supported, the public understand their impact, and identify any alternatives before decisions are made.

NHS England has launched this consultation to seek views on proposed changes to the Clinical Genomic Service Specification.

Clinical Genomics (previously called Clinical Genetics) are services where doctors (typically Clinical Geneticists) and Genetic Counsellors work with other health professionals to diagnose genetic conditions and/or assess the risk a patient will inherit or develop a genetic condition. They provide and co-ordinate treatment for patients with genetic conditions, and counselling and emotional support to patients and their families.

The consultation will run for 30 days from **1 September to 30 September 2022**. This document sets out:

- How care is currently provided.
- How the revised service specification will change care and the way that services are delivered, and the reasons for these changes.
- How the proposed changes will be implemented.

The document also has information about how you can share your views with NHS England. At the end of the consultation period, all feedback will be considered before the revised Service Specification is published. We will also consider feedback to inform how the revised Service Specification is implemented in the future, with conversations taking place with each service to determine the best way to deliver care and service transformation at a local level.

Feedback from patients, their families, and the public

NHS England wants to make sure that you can use this consultation as an opportunity to tell us what matters to you.

Therefore, we would like you to feedback on the things that were important to you if you used genomic services in the past, and would be important to you if you used them in the future.

To complete this questionnaire, you do not need to review all the documents included in the Consultation.

We recommend that you read this Consultation Guide, in particular the section that provides a 'Summary of changes for patients and their families'.

You may wish to review the revised Service Specification, but it is not essential for you to be able to provide you feedback in the consultation questionnaire.

You are welcome to review and provide feedback on any or all documents included in this consultation should you wish to. You will be given the option to do this at the end of 'Feedback from patients, families and the public' section of the consultation questionnaire.

The questions in the online consultation questionnaire are included below (page 17).

Feedback from health care professionals, professional bodies and other organisations

NHS England is seeking your feedback to identify the extent to which the work undertaken so far, and documents produced, are complete and set out the correct priorities, impacts and risks.

We recommend that you read this consultation guide alongside the other documents published as part of the consultation. While this single consultation guide has been produced to summarise the proposals, the other documents provide additional detail.

Documents included in this consultation

- **Clinical Genomic Services: Draft Service Specification** – The Service Specification is a contractual document that describes the clinical service and sets out appropriate standards and quality measures that provider organisations must satisfy.
- **Equality and Health Inequalities Impact Assessment** (EHIA) – This document assesses the potential impact of the revised Service Specification on population groups that may be disproportionately affected by changes to the Service Specification and make appropriate recommendations to mitigate any inequity.
- **Stakeholder Testing Engagement Report** – Details the stakeholder testing already undertaken seeking views on the Service Specification and EHIA.
- **Integrated Impact Assessment** – Sets out the potential impact of the proposed changes to the Service Specification on both patients and services.

Background

Genetic counselling has been provided as part of healthcare since 1946 when the first clinic in Europe was started at Great Ormond Street, London, to support families affected by rare, inherited diseases. These foundations strengthened in the 1960's when the first genetic laboratory services were set up. Genetic testing knowledge and techniques have improved greatly since these early days over 70 years ago, providing patients with diagnoses and treatment options they would not previously have had.

With this "*long and proud history of embracing technology and innovation to deliver faster diagnoses and more effective treatments for patients, the NHS continues to lead the world in harnessing the power and potential of genomics – the study of genes in our DNA and their function"* – Professor Dame Sue Hill, December 2020.

Technology has developed quickly in recent years; the first genome took an international team 13 years to sequence but this can now be done on a desktop machine in a little over 24 hours.

The NHS harnessed this technology and took genomics on the next huge step forward through the 100,000 Genomes Project. This provided the evidence to demonstrate the benefits to patients of whole genome sequencing technology and highlighted areas of transformation required to embed genomics across the entire health care system. As a result, NHS England has worked with partners to develop a genomics infrastructure to support this work; the NHS Genomics Medicine Service (NHS GMS). The NHS GMS includes:

- Seventeen NHS Clinical Genomic Services (NHS CGSs) that deliver a comprehensive clinical genomic and counselling service that directs the diagnosis, risk assessment and lifelong clinical management of patients of all ages and their families who have, or are at risk of having, a rare genetic or genomic condition including inherited cancer.
- A national network of seven NHS Genomic Laboratory Hubs (NHS GLHs) who are responsible for the delivery and performance of the genomic testing within their geographical region.
- Seven NHS GMS Alliances, aligned to the NHS GLH geographical footprints, that oversee and co-ordinate the embedding of genomics into routine care locally and across the whole population the NHS serves in England. The NHS GMS Alliances bring together the vital multi-disciplinary clinical leadership and expertise, along with the operational and digital functions, that are necessary to make this happen.

A diagram to demonstrate how the NHS GMS aligns to the wider healthcare system can be found in Appendix 1.

The NHS GMS plays a key role in delivering the NHS Long Term Plan (NHS England, 2019) commitments that will enable the NHS to harness the power of genomic technology and science to improve the health of our population. These commitments include:

- To be the first national health care system to offer *whole genome sequencing* as part of routine care, including for all children with cancer and children who are seriously ill with a likely genetic disorder.
- Extended access to *molecular diagnostics* and offer *genomic testing* routinely to all people with cancer.
- Early detection and treatment of high-risk conditions including expanding genomic testing for *Familial Hypercholesterolaemia*.
- Linking and correlating genomic data to help provide new treatments, diagnostic approaches and help people make informed decisions about their care.

The NHS GMS also contributes to the Government's ambition to create the most advanced genomic healthcare system in the world, underpinned by the latest

scientific advances, to deliver better health outcomes at lower cost as set out in Genome UK: The future of healthcare (2020). The strategy sets out the vision for the future, focussed on three key areas:

- **Diagnosis and personalised medicine** incorporating the latest genomic advances into routine healthcare to improve diagnosis, stratification, and treatment of illness.
- **Prevention** through the enabling of predictive and preventative care to improve public health and wellness.
- **Research**, supporting fundamental and translational research and ensuring a seamless interface between research and healthcare delivery.

The NHS GMS will support the delivery of the commitments set out in the NHS Long Term Plan and Genome UK: The future of healthcare through its provision of:

- Consistent and equitable care to the country's 55 million people.
- Common national standards, specifications, and protocols.
- A single National Genomic Testing Directory offering the use of all technologies from single genes to whole genome sequencing.
- Opportunities for all patients to participate in research for their individual benefit but also to inform future care of other patients.
- A national genomic knowledge base to provide real world data to inform academic and industry research and development.

Clinical Genomic Service Specification: the case for change

This consultation considers the 'NHS Clinical Genomic Service' element of the NHS GMS.

The existing NHS CGS specification was published in 2013 and included both clinical genetics and genomic testing services. Since its publication, the NHS GMS has been established, leading to the separation of the clinical genomic and laboratory functions through the formation of NHS GLHs in 2018. It is important that the revised specification reflects the infrastructure changes brought about by the formation of the NHS GMS. It also needs to reflect developments within the NHS GMS, including:

- The delivery of NHS CGSs as a network, working in tandem with the NHS GLHs supported by the NHS GMS Alliances.
- Highlighting new ways of working through virtual (online and telephone) and face to face clinics, *multi-disciplinary teams* (MDTs), *genomic test advisory boards* (GTABs), and multidisciplinary clinics (MDCs).

- Recognition of the roles of all the health care professional groups within clinical genomics to ensure the best use is made of peoples' skills.
- Adaptation of the workforce to reflect the rapid advances in genomic testing technology, increased provision of testing including *rapid exome sequencing*, specialist genetic testing, increased range of cancer genomic testing, and whole genome sequencing.
- To reinforce the need to ensure equity of access and provision of genomic testing to all groups based on clinical need; and
- The increased role of mainstream medicine in the delivery of genomics and how NHS CGSs will support colleagues to facilitate this provision for patient benefit.

This Consultation Guide sets out how the revised Service Specification reflects the NHS CGS contribution to the NHS GMS and how the service will support the delivery of the national commitments for delivering genomics within the healthcare system.

Service Specification Revision: Our work to date

The Service Specification was revised between July 2020 and March 2021 by the Service Specification Working Group, made up of a subsection of *Genomics Clinical Reference Group* members.

Stakeholder testing of the revised Service Specification was undertaken in March 2021, following which some refinements were made to make sure that the network and commissioning governance arrangements were clear and to ensure we used consistent terminology. Stakeholder feedback also provided valuable insight into other factors that required consideration as part of the Integrated Impact Assessment and will also inform the development of the *Commissioning Implementation Plan*. The Engagement Report has been included as part of the suite of documents in this consultation.

A subsequent review and revision of the Service Specification was undertaken in March 2022 following feedback from the NHS GMS People and Communities Forum and publication of a new specification template and associated guidance by NHS England. The changes were reviewed by the Service Specification Working Group in May 2022 and it was agreed that, whilst the document layout, language and content had changed, the key priorities and services requirements were correctly articulated.

The Integrated Impact Assessment was undertaken and validated by a Confirm and Challenge Workshop held with representatives from the Service Specification Working Group in December 2021. The potential impacts and risks highlighted by this work will further inform the development of the Commissioning Implementation Plan and are highlighted within this Consultation Guide.

How is care currently provided?

What we know about Clinical Genomic Services

- ✓ Clinical Genomics is a clinical specialty that supports the diagnosis and care of patients and families with suspected or confirmed genetic and genomic conditions.
- ✓ Patients that benefit from care can have complex and rare diseases the often remain undiagnosed for a long time. Others may have cancers with a genetic predisposition or have a significantly increased risk of developing certain tumour types for genetic reasons.
- ✓ Rare diseases impact on the lives of approximately 6 – 8% of the population, and at least 80% are genetic.
- ✓ 5 – 10% of cancers arise in individuals with a genetic predisposition.
- ✓ Many people with rare diseases experience what is referred to as a 'diagnostic odyssey'. During this time, they are likely to have many diagnostic tests before a diagnosis can be made or may not receive a diagnosis at all. During this time their quality of life can be poor due to their symptoms, with doctors prescribing lots of treatments to manage those symptoms the best they can.
- ✓ Some treatments used to manage certain conditions, particularly cancers, can vary in their effectiveness depending on a patient's genes. In some cases, a patients' genes can cause treatments to be harmful.
- ✓ Genomic testing can help diagnose genetic conditions, ensure appropriate treatment is given and give patients a better chance of getting the support they need. It can also tell patients if they have an increased risk of developing a condition later in life so that they can be monitored and make informed choices about how they can minimise risks and their health care options in the future.
- ✓ Genomic testing can lead to life changing diagnoses, so it is crucial that patients and their families receive the right support to cope with their results.

There are seventeen NHS CGSs across England (Please see Map of NHS CGSs in Appendix 2). Each NHS CGS is responsible for a defined geographical area, accountable to its host NHS Trust and, through them, to NHS England.

Outpatient care has historically been provided through a 'hub and spoke' model, with clinics provided from outpatient facilities within the host NHS Trust and via outreach clinical in district general hospitals and other suitable stings across the geographical catchment area of the service. However, the COVID-19 pandemic has led to a reduction in the number of sites from which services are provided. Typically, there are now fewer locations from which services are provided but an increased number of patients that access services via video consultation or over the telephone.

In the outpatient clinic (or via video consultation or over the telephone), patients and/or their family are assessed by the most appropriate clinician. The clinician may review patients to establish a diagnosis or consider which genetic test(s) would help to diagnose an existing condition. They may also assess the risk of people inheriting or developing a genetic condition. The clinicians are usually *Clinical Geneticists* or *Genetic Counsellors*, but some services also have specialist nurses and family history co-ordinators who will also see patients. If required the clinician will organise genetic testing, which will be done by one of the seven NHS GLHs. Follow-up appointments are provided to discuss the results of the genomic testing.

The NHS CGSs don't only organise genomic testing, they also provide long term management and support for patients and their family. This might include regular monitoring and/or counselling. Some patients remain under the care of the service for many years. Others might be discharged and return to the service later in life when they need more support with their health and care choices.

NHS CGSs work very closely with other clinical specialties in hospitals. Some of those specialties have a good understanding of genomics and how to use genomic testing to inform the care of their patients. There are other specialties where the use of genomics has only been introduced recently because new tests have been developed that can help with the diagnosis and care of their patients. The NHS CGS provides expertise to support clinical specialties and advice where complex clinical management or the interpretation of genomic testing results is needed.

What changes are being proposed and why?

NHS England is proposing to publish a new Service Specification for NHS CGSs to reflect:

- The separation of NHS CGS and laboratory functions since the formation of NHS GLHs in 2018.
- The advances made in genomics since the last specification was developed, bringing the NHS CGSs in line with current practices such as the inclusion of cancer genomics.

NHS England is proposing six key priorities that will arise from the implementation of the new Service Specification, which are:

1. The **embedding of genomics into mainstream clinical pathways** (**referred to as '*Mainstreaming*' Genomics**) to increase the confidence of clinicians in clinical specialties and primary care to directly request appropriate genomic testing in line with the National Genomic Test Directory and understand how results can impact on patient care.
2. **Care for patients and their families will be provided in a co-ordinated way (referred to as Co-ordinated Care)** with the NHS CGSs working closely with other parts of the health and care system to enable involvement of everyone contributing to care, avoiding duplication and achieving shared outcomes.

3. **All those that would benefit from the NHS CGS and/or genomic testing are able to access it (referred to as Equity of Access)** in line with their clinical need and the National Genomic Test Directory. As such, NHS CGSs, in partnership with their local NHS GMS Alliance, will improve engagement with communities that have historically not been reached to reduce barriers to accessing services.

4. **Improving outcomes for patients and their families** by supporting the reduction of the 'diagnostic odyssey', helping patients to get a diagnosis sooner and enabling the delivery of personalised care, informed by accurate genomic data, to reduce symptoms and improve quality of life.

5. **NHS CGSs will form networks aligned to the NHS GLH and NHS GMS Alliance geographical footprints (referred to as Networked Care Provision)** to optimise access to and provision of services. These seven regional networks will also work as part of the NHS GMS national network to share best clinical practice, support workforce development, participate in research, develop guidance, set direction of the NHS GMS as a whole and ensure equitable access for patients and their families.

6. **Workforce planning and development** to ensure that services can function effectively, implement the key priorities arising from the revised specification, contribute to the wider NHS GMS and contribute to building the future workforce needed to meet increasing demand for services.

How does the revised specification change care and what is the impact on services?

Summary of changes for patients and their families

- ✓ More doctors and nurses in lots of different health care services will understand genomics, how genomic testing can be used to help to diagnose patients and how the results can be used to provide the best care possible for patients and families.
- ✓ This means patients and their families might not have to be referred by their doctor to an NHS CGS and can receive genomic testing, diagnosis, treatment and the support they need faster through the doctor they typically see and already knows their medical history.
- ✓ Patients and their families will be able to trust that the specialist clinical genomic staff will support their usual doctors and nurses to understand genomics and be available to provide expert support if those doctors and nurses need it.
- ✓ If patients and families don't feel they aren't able to access NHS CGSs but feel that they would benefit from them will be able to refer themselves for an assessment. Each NHS CGS will be asked to ensure that information for how patients can refer themselves is publicly available.
- ✓ When patients and their families have complicated symptoms and/or need a lot of support, their doctors and nurses will be able to refer them to the NHS CGS for specialist care.
- ✓ Sometimes genomic testing and receiving what can be a life changing diagnosis is distressing for patients and their families. The NHS CGSs will continue to provide emotional support but, if patients and families need it, they will be referred to specialist clinical psychologists who can help them to cope and adjust. This specialist support has not always been available to patients and their families in the past.
- ✓ NHS CGSs will work in a co-ordinated way with other clinical services that are involved in providing care and treatment for the patient to ensure that everyone knows what is going on and the outcomes they are working towards.
- ✓ Patients and their families will be able to receive NHS CGSs in places that are convenient for them and without having to travel long distances. This might be by appointments that take place on the telephone or video call. Services will regularly seek feedback from patients to make sure that they are happy with how their care has been provided and make changes if needed.
- ✓ We hope that the changes to how genomics services are provided will make sure that patients receive diagnosis of their conditions and the treatment they need to improve their health and/or emotional well-being faster. Patients and their families will also be able to understand their risk of developing conditions in the future sooner so that they can make decisions about their own health and future medical care.

1. Mainstreaming Genomics

Mainstreaming genomics supports all clinicians within the healthcare system to embed genomic medicine into their own clinical practice and patient pathways. To achieve this, it will be essential for the NHS CGSs, in partnership with their local NHS GLH and NHS GMS Alliance, to train and educate clinicians in other clinical specialties so that they are confident and understand the genomic testing that might benefit their patients, and how to understand the results to decide how to manage the care of their patients. There are many clinical specialties that already have this confidence, but for others the use of genomics in their patient care may be relatively new.

This will mean that some patients who might previously have been referred to the NHS CGS, particularly those with common genetic conditions, may remain under the care of their mainstream specialty.

To ensure that patients can be cared for by the right service, the NHS CGSs will work alongside the NHS GMS Alliances and mainstream specialties to develop pathways where they don't already exist, and maintain/further develop pathways where they are already established, to embed genomic testing and/or advice for all service users with genetic conditions and where genomic testing is provided as part of cancer care.

To support those pathways, clinical geneticists and genetic counsellors may work jointly in clinics with mainstream clinicians. In addition, NHS CGSs will support through regular multidisciplinary team meetings and ensure that jointly developed referral pathways are in place to ensure patients with complex needs are referred to NHS CGSs and/or other appropriate clinical services. Where required, there will also be support, in real-time if appropriate, for mainstream care of patients with less complex needs through the provision of expert advice and guidance by telephone, video link or email. This way of working has been successful with those mainstream specialties where the use of genomics is already well established.

Mainstreaming genomics is expected to change the demand placed on NHS CGSs. Through the mainstreaming of genomics into other clinical specialties, the number of services users referred to NHS CGSs to access testing and diagnosis is expected to reduce as the specialty clinicians become confident to take on this role.

However, the increase in genomic awareness and testing in mainstream specialties is expected to increase the number of patients diagnosed with a rare disease, or cancer/risk of cancer with a genetic predisposition. This, along with the identification of new genetic conditions and treatments as more technology is developed, is expected to lead to a higher number of patients with complex disorders referred to NHS CGSs. The complexity of these patients' conditions is likely to require long term management and increased clinical and psychosocial input.

2. Co-ordinated Care

Many people living with rare diseases need care and support from lots of services in the health and care system because their condition is both chronic and complex. Research has shown that care for people with rare conditions is often not co-ordinated, resulting in them attending multiple appointments on different days, with

different professionals in different locations (Walton et al, 2022). Patients with rare conditions, and/or their families, seldomly have a designated care co-ordinator so often undertake tasks like chasing and organising appointments, chasing test results, passing information between different healthcare professionals and telling their story over and over again to different clinicians. This creates additional burden for the patient and/or their family and can have a negative impact on the physical, psychosocial, and financial health.

NHS CGSs will play a key role in ensuring that the care is co-ordinated in a family-centred way, including medical, psychosocial, educational and vocational needs irrespective of diagnosis, service user circumstances and geographical location. Therefore, NHS CGSs are being asked to co-ordinate care, in a family-centred way, with other parts of the health and care system so that care is provided as smoothly as possible. This will link to the work done to develop pathways with mainstream clinical specialties, including cancer services, as described in the section above but also ensure that co-ordinated pathways are in place that provide access to specialised/ highly specialised care, treatment, drugs, social care, mental health and special educational support.

NHS CGSs will also participate in the thirteen Rare Disease Collaborative Networks, each made up of providers who have an interest in a particular disease and are committed to working together to progress research, increase knowledge and improve patient experience and outcomes. These networks are based on the principle that, where practical, 'the knowledge moves rather than the patient', and coordinate care by operating national virtual MDT meetings and in person clinics.

3. Equity of Access

NHS England aspires to ensure that all patients that would benefit from genomic testing, and the care and support provided by clinicians that are confident with all aspects of how genomics can inform clinical decision making, are able to access it.

The patients that can access NHS CGSs currently differs across some services in England. **Therefore, NHS England will work with services to develop consistent and agreed criteria to ensure that all eligible people can access services regardless of where they live**.

The number of eligible people may vary across NHS CGSs depending on what type of population they have in their area. For example, rare diseases are more commonly diagnosed in childhood and some conditions that have a genomic predisposition can be more common in people from certain ethnicities.

The Service Specification requires each NHS CGS, in partnership with its NHS GMS Alliance, to prioritise equity of access and the provision of genomic care for all eligible patients, with particular attention given to addressing the unmet needs in communities that have historically not been reached.

Where not already in place, NHS CGSs will identify champions to improve their engagement with seldom heard communities and actively recruit into patient participation groups so that they can influence service development and improvement.

To inform the development and ongoing delivery of services, NHS CGSs will undertake and support patient and public participation activities and act on feedback received from patients and users of services. They will work to **reduce barriers to accessing the NHS CGSs, genomic testing and research and improve engagement with communities that have historically not been reached**, increasing awareness of the NHS CGS across other healthcare providers and the local population.

NHS England proposes that all services should minimise barriers that would stop people accessing services. Therefore, services should be provided in a way that is sensitive to the needs of each individual and have provision for any person within a *protected characteristic group* or groups that can face *health inequalities*. Further detail of how NHS England has considered these groups of people can be found in the Equality and Health Inequalities Impact Assessment.

Where access to the NHS CGS is not achieved through standard routes such as via a General Practitioner or mainstream service, national criteria for accepting patient generated self-referrals will be defined. This mechanism is expected to reduce barriers to access services.

Equity of access can be affected by the location from which the NHS CGSs are provided. NHS England does not propose to change the number or location of the main seventeen NHS CGS providers. However, it will be important to ensure that this does not present a barrier to patients that live some distance from the main provider. Historically services were provided from satellite locations across the geographical area covered by the main service. However, COVID-19 led to reduction in the number of sites in which clinics were available but an increase in the number of patients able to access services via video consultation or over the telephone ('virtual' consultations). **As the COVID-19 restrictions have eased, it will be important to review the number of satellite clinics required in view of the increased use of virtual consultations, whilst balancing the impact on patient experience and equity of access.**

NHS CGSs will need to monitor their services to ensure that no patient group is discriminated against and that the service is available to all eligible individuals across its entire geography.

Some clinical geneticists and genetic counsellors have areas of specific expertise that is not available across all services. Therefore, this **expertise will be made available on a national and/or regional level so patients in all geographical locations can benefit.**

The revised Service Specification emphases the importance of access to clinical psychology to provide essential care to people affected by genomic conditions that require more complex or sustained psychological support. One NHS CGS in England currently employs a clinical psychologist to provide this care, nine services can refer their patients directly to clinical psychologists within local mental health services and five services has no direct access to clinical psychology for their patients. **Therefore, NHS England will work with services to ensure that pathways to clinical psychology are available to all patients if clinically required.** This may be through the recruitment of clinical psychologists into NHS

CGSs or by working with local mental health services to ensure that referral pathways are in place to sufficiently skilled clinical psychologists in local mental health services, depending on the expected local need.

4. Improving Outcomes

Through the mainstreaming of genomics and improving equity of access NHS England aspires to underpin patient care with the latest scientific advances and deliver better health outcomes at a lower cost to the health system.

Wider implementation of genomics in patient care is expected to support earlier detection and faster diagnoses of rare diseases, **reducing the diagnostic odyssey experienced by many patients.** The average diagnostic odyssey lasts for eight years, during which time patients can face multiple referrals, inconclusive tests and sometime incorrect diagnoses before a final diagnosis is reached. During this time, the patient's condition may deteriorate and the uncertain journey through services can have a negative impact on their mental health. Also, the delay in reaching a diagnosis can mean missing the window of opportunity for patients to benefit from treatment and result in poorer outcomes.

The use of genomics can help to target treatments and improve outcomes for many patients. NHS CGSs will be required to work alongside other clinical specialties to offer new and/or improved care pathways to manage and treat genetic conditions as new genomic tests and therapies become available.

Sometimes genomic testing will identify a life changing or life limiting disease for which treatments are not available and the outcome for the patient may unfortunately not be changed. The aim of the NHS CGS in this circumstance is to improve psychological resilience and empower patients and families to make informed decisions about their or their child(ren)'s future. During the first year following publication of this revised Service Specification, all NHS CGSs will contribute to the development of a nationally consistent methodology to measure the impact that the service has on outcomes for patients and their families. Monitoring against the agreed methodology will commence in the subsequent year.

Genomics can also support the improvement of people's health through targeted screening programmes that improve disease prevention. This will include **personalised screening and testing for family members of cancer patients, where clinically indicated, to identify if they are at an increased risk of cancer.**

A multi-disciplinary and networked approach to the management of patients' care will ensure that a wide range of clinical expertise is brought together to interpret the outcomes of genomic testing and translate the results into safe and effective clinical treatment and care.

5. Networked Care Provision

The seventeen NHS CGSs are each responsible for a defined geographical area but **will form networks across the seven NHS GLH and NHS GMS Alliance geographical footprints to optimise access and provision.** These seven regional networks will also **work as part of the NHS GMS national network** to

share best clinical practice, support workforce development education and training, research, develop guidance, set direction for the NHS GMS as a whole and ensure equitable access for patients and their families.

The NHS GMS national network, working closely with other stakeholders, will undertake research and quality improvement schemes to determine the evidence for changes to clinical practice, thus ensuring optimal patient care and experience and as part of the NHS Genomic Medicine Service Research Collaborative. Alongside this, services will support recruitment to clinical trials and the translation of research into clinical practice, supporting the adoption of cutting-edge technologies into health care.

Each NHS CGS will undertake joint consultations with other clinical specialties to improve clinical pathways, reduce time to diagnosis, facilitate ongoing management and reduce the burden on patients and their families. In addition, each service will provide clinical expertise into MDTs to support both NHS GLH and clinical specialty colleagues. Similarly, cancer genetics teams working as an integral part of each NHS CGS, will work with cancer specialties to implement pathways for patients into inherited cancer MDTs.

6. Workforce planning and development

NHS England aspires to NHS CGSs that have enough suitably skilled staff to ensure that they can provide a high-quality service to their eligible population without limiting their activity within available resources and/or developing lengthy waiting lists. NHS CGSs should also have enough experienced staff to develop new pathways of care to achieve mainstreaming of genomics and, in partnership with the NHS GMS Alliances and Health Education England, provide the education, training and support needed to develop the competence and confidence required in mainstream services.

In addition to the expected increase in direct patient care, demand for NHS CGSs is also anticipated to increase in other ways as genomics is being embedded into mainstream specialties, such as:

- Education and training provided to mainstream services.
- Time spent participating in Multi-disciplinary Team meetings.
- Time spent providing expert advice and guidance to mainstream services and primary care.
- The contribution made by NHS CGS teams to NHS GMS Alliances and NHS GMS transformation and improvement programmes.

Therefore, sufficient capacity should be planned for the workforce to support this additional burden likely to be placed on NHS CGSs.

NHS England is committed to patients receiving care from the most appropriate clinician, which may be a Clinical Geneticist, Genetic Counsellor and/or suitably trained and experienced mainstream clinician.

The Integrated Impact Assessment that has been undertaken indicates that NHS CGSs may not currently have enough staff in post to either provide care to all eligible patients or achieve mainstreaming of genomics.

NHS England will work with each provider to plan the workforce, including suitable skills and experience, that is required to deliver the requirements of the Service Specification. **This will help to reduce any existing waiting lists so that patients can receive their care in a timely manner**.

There is also scope for NHS CGSs to consider introducing or increasing other roles within their teams, such as genomic associates, genomic practitioners, genomic assistants and family history co-ordinators to free up Clinical Geneticist and Genetic Counsellor time for both patient facing and pathway development activities.

NHS England will support NHS CGSs to work jointly with Health Education England, NHS GMS Alliance, professional bodies, and local Deaneries (organisations responsible for post-graduate medical training) to create, maintain and provide workforce development plans that reflect the requirements to meet ongoing local demand, population needs and the future aspirations of the NHS GMS.

How will the proposed changes be implemented?

NHS England is committed to supporting NHS CGSs to introduce the revised Service Specifications in a gradual way so that they can achieve the priorities set out within it. It will be important that NHS England works with each of the services to develop a Commissioning Implementation Plan that ensures a national approach to introducing the Service Specification in a way that is sensitive to each service's population and regional needs.

NHS England expects that the Commissioning Implementation Plan will include an agreed approach to:

- Establishing a single, national reporting framework for activity undertaken by the services, cost of service delivery and for monitoring the Quality Standards detailed in Section 6.2 of the Service Specification.
- Development of a tool that will be used to measure the impact that the service has on patient outcomes.
- Consistent operating procedures that the service will adopt, including guidance for intra-specialty collaboration to achieve mainstreaming and consistent eligibility criteria.
- A service delivery model that provides equity of access across England, including minimum requirements for providing services in locations across each geographical area and achieving equity of service provision.
- Developing optimum, efficient and cost effective workforce structures with sufficient staff to manage care for the eligible population, a skill mix that is capable of developing new care pathways and providing training and support for mainstream clinicians and makes effective use of additional roles such as

genomic assistants, genomic associates, genomic practitioners and family history co-ordinators.

- Joint working with Health Education England to ensure that systems are in place to increase the available staff to meet future demand for services.
- Developing a consistent financial model that supports the required capacity and service delivery model, achieving equity of access and supporting transformational requirements within the NHS GMS.

NHS England proposes to have a phased approach to introducing the new Service Specification to ensure that all commissioning decisions are based on robust information and effective service planning. Therefore, the Commissioning Implementation is planned to be delivered across **three phases**. The plan outline is expected to be:

1. Development and implementation of robust activity and service cost monitoring frameworks, with retrospective data submission from October 2022. Development of a nationally consistent methodology to measure the impact the service has on patient outcomes. This phase will also include a full-service review undertaken jointly by NHS England and NHS CGSs, and the development of standard operating procedures and consistent eligibility criteria – **Year 1 (22/23)**
2. Interpretation of Year 1 reported activity and financial data jointly by NHS England and NHS CGSs. Development of a workforce plan informed by both the activity reported by NHS CGSs for 22/23 and the joint work undertaken to understand the population needs of each service. NHS England will also work with NHS CGSs to develop of a national financial model that reflects local service specialisms/expertise and population needs – **Year 2 (23/24)**
3. Shadow monitoring of new financial model, activity and workforce to ensure robust, making refinements if required – **Year 3 (24/25)**

What do you think of our proposed changes?

NHS England would like to hear what people think about the proposed changes to the NHS CGS in England.

Feedback from patients, families, and the public

We are asking patients, families and the public about the things that were important to them if they used genomic services in that past, and what would be important to them if they use them in the future.

Specifically, we are asking:

1. For people or their families that have used genomic services in the past, we are asking:
    a. What was good about the care that you and/or your family member experienced?

b. How could you and/or your family member's experience of services have been improved?

2. All patients, family members and the public how they think the proposed changes to the NHS CGSs might impact on the experience people will have of services if they need care in the future.
3. If there are any changes and/or improvements that could be made that we have not considered.

Feedback on all Consultation Documents

We are asking health care professionals, representatives from other professional bodies, and members of the public should they wish to, to provide feedback on all consultation documents.

Specifically, we are asking:

1. To what extent do you think the Service Specification achieves its key priorities to:
    a. Embed clinical genomics into mainstream clinical pathways?
    b. Provide care for patients and their families in a co-ordinated way?
    c. Ensure all those that would benefit from the NHS CGS and/or genomic testing are able to access it?
    d. Improve clinical outcomes for patients and their families?
    e. Improve access to and the provision of services by aligning NHS CGSs to the NHS GMS national network
    f. Ensure the workforce has capacity to implement the key priorities arising from the Service Specification and meet increasing demand for services?
2. Do you think the revised Service Specification will have a positive impact on patient care, outcomes and experience?
3. To what extent do you think the Integrated Impact Assessment identifies the potential impact and risks associated with:
    a. Patients and patient pathways?
    b. The delivery of Clinical Genomic Services?
    c. Capacity and workforce planning?
    d. Financial risk and cost pressure?
4. To what extent do you think this Consultation Guide and the Integrated Impact Assessment has identified the actions that require consideration as part of the Commissioning Implementation Plan?

5. To what extent do you think the Equality and Health Inequalities Impact Assessment identifies that impact of the revised Service Specification on:
    a. Protected characteristic Groups?
    b. People who experience health inequalities?
6. To what extent do you think the Stakeholder Engagement has:
    a. Involved the correct stakeholders?
    b. Used engagement methods that have provided opportunities for stakeholders to be involved?
    c. Considered stakeholder feedback to inform the development of the Service Specification, Integrated Impact Assessment and the actions that will be included in the Commissioning Implementation Plan?
7. Are there any changes or additions that you think need to be made to the Service Specification?
8. Are there any changes or additions that you think need to be made to the Integrated Impact Assessment?
9. Are there any additional actions that you think need to be considered when developing the Commissioning Implementation Plan?

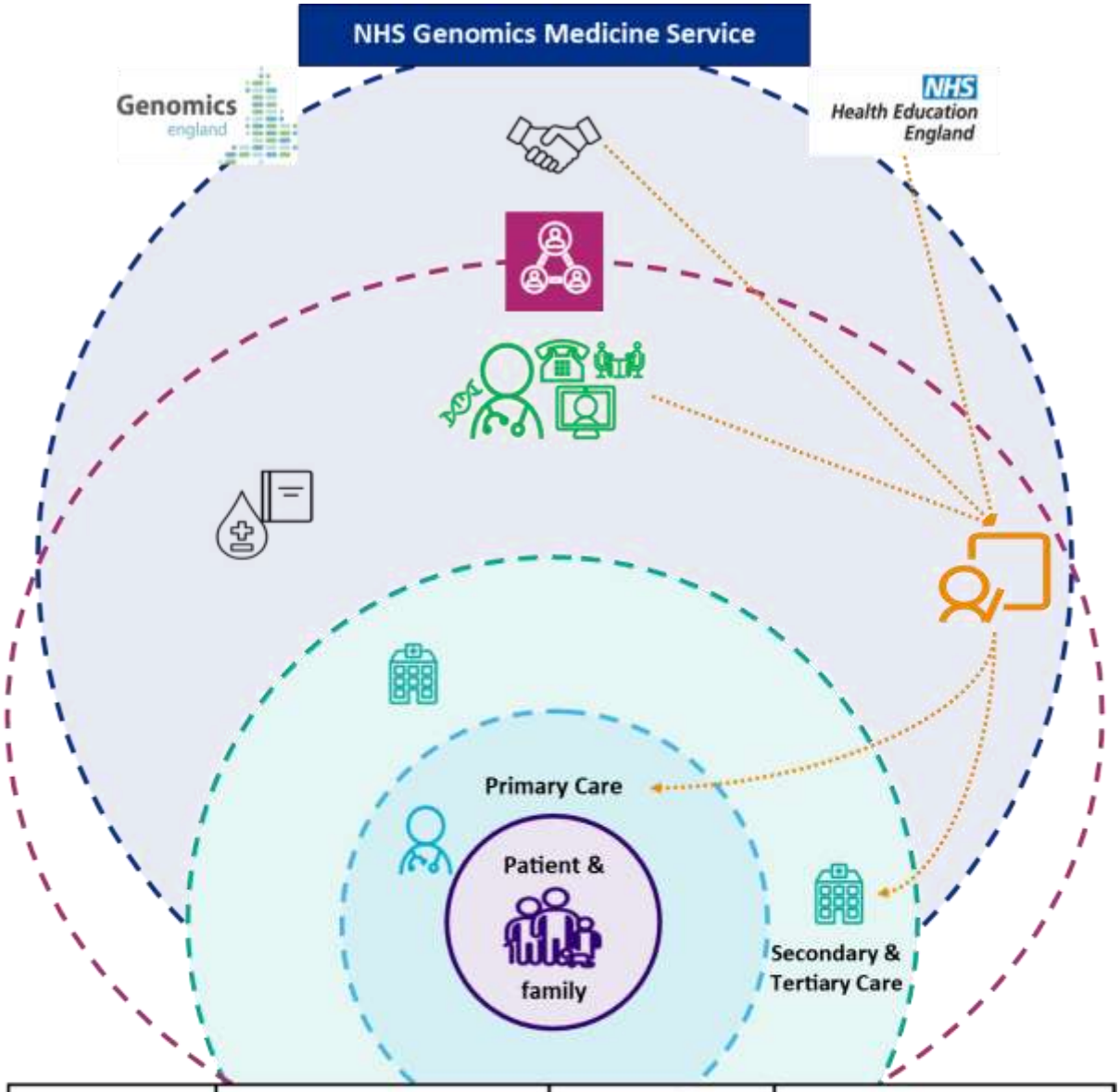
The consultation is set to run from **1 September to 30 September**.

You can provide your views with NHS England by completing the online survey which can be accessed here: https://www.engage.england.nhs.uk/specialised-commissioning/clinical-genomics-service-specification-survey.

You can also attend one of our Webinars and Engagement Events which you can register to by following the link above.

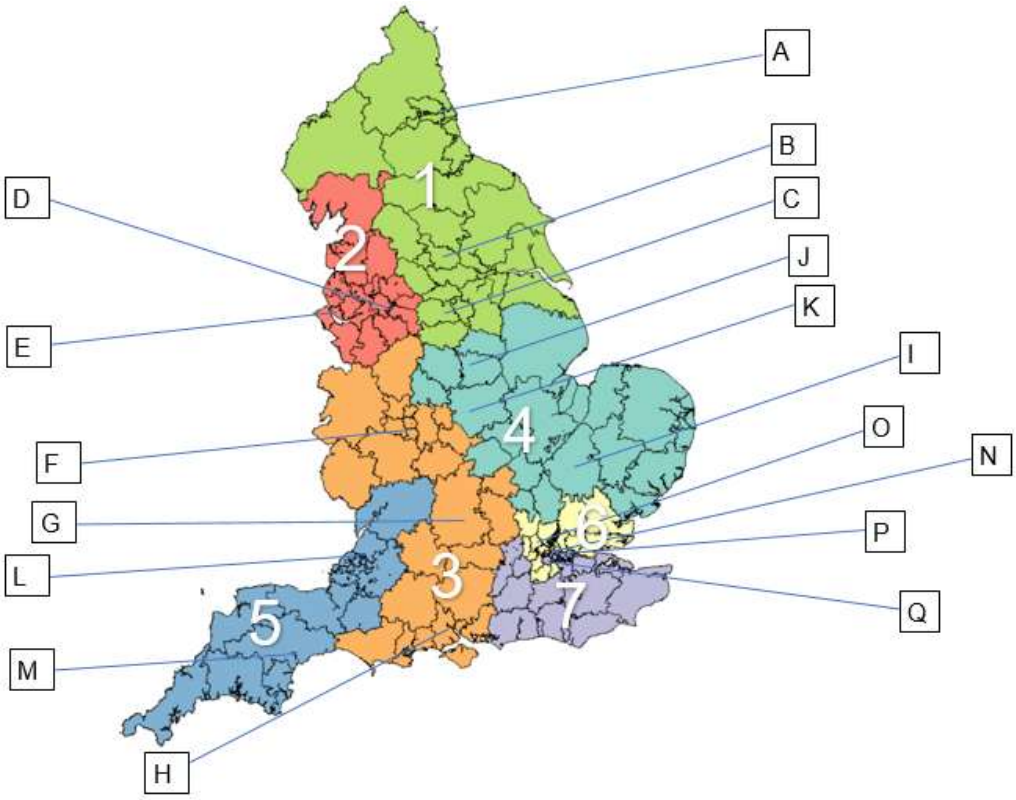
Your views will help NHS England to further shape and refine plans for Clinical Genomic Services and will help us to make sure that we are delivering safe and high-quality care that meets the needs of patients and families.

Appendix 1: NHS Genomic Medicine Service



	Patients and family members of all ages		17 x Clinical Genomics Services
	NHS Genomics Medicine Service	7 x NHS Genomic Medicine Service Alliances	Clinical Geneticist/ Genetic Counsellor
	Telephone appointments	Face to face appointments	Online video Appointments
	7 x NHS Genomics Laboratory Hubs	National Genomics Test Directory	Training and education
	Elements of care provision involved in Multi-Disciplinary Collaborative Care	Primary Care/General Practitioners	Secondary and Tertiary Care – Hospital Specialities
	Multi-disciplinary Collaborative Care via Multi-Disciplinary Team Meetings	General Practitioner	Acute Hospital

Appendix 2: Genomic Medicine Service Geographies



Geography Key	Genomic Medicine Service Alliance Geography	Genomic Laboratory Hub Geography	Clinical Genomic Services Key	National Clinical Genomic Service comprised of the individual Regional Clinical Genomic Services
1	North East and Yorkshire	North East and Yorkshire	A	Northern Genetics Service
			B	Yorkshire Regional Genetics Service
			C	Sheffield Clinical Genetics Service
2	North West	North West	D	Manchester Centre for Genomic Medicine
			E	Liverpool Centre for Genomic Medicine
3	Central and South	Central and South	F	West Midlands Regional Genetics Service
			G	Oxford Centre for Genomic Medicine
			H	Wessex Clinical Genetics Service
4	East	East	I	East Anglian Medical Genetics Service
			J	Nottingham Clinical Genetics Service
			K	Leicestershire Clinical Genetics Service
5	South West	South West	L	Bristol Clinical Genetics Service
			M	Peninsula Clinical Genetics Service
6	North Thames	North Thames	N	North East Thames Regional Genetics Service
			O	North West Thames Regional Genetics Service
7	South East	South East	P	South East Thames Regional Genetics Service
			Q	South West Thames Regional Genetics Service

Glossary of Terms

Whole Genome Sequencing is the analysis of the entire genomic DNA sequence of an individual (e.g. through a blood sample) or a tissue (e.g. cancer) to diagnose a rare condition or direct optimum treatment.

Molecular Diagnostics is the detection of genomic variants to help detect and diagnose disease and monitor response to treatment.

Genomic Testing are lots of different technologies used to find changes in genes that can cause health problems.

Familial Hypercholesterolaemia is an inherited condition causes high cholesterol that's passed down through families and is caused by one or more faulty genes. It's caused by a genetic variant that means your liver is unable to remove excess 'bad' cholesterol and can lead to an increased risk of heart attacks and/or strokes.

Multi-disciplinary Teams are groups of people that bring together the expertise and skills of different professions to assess, plan and manage care jointly. A genomics multi-disciplinary team may involve a range of professionals including specialist clinicians such as cardiologists, oncologists, specialist nurses, clinical geneticists, genetic counsellors, clinical scientists, and any other health care professional involved in the patient's care.

Genomic Test Advisory Boards act in an advisory capacity, reviewing and providing interpretation of complex genomic testing results. Genomic Test Advisory Boards feed into the clinical multi-disciplinary teams for final decisions to be made in conjunction with the clinical information.

Rapid Exome Sequencing is a genomic technique used to gather specific information whilst allowing tests to be completed and interpreted far more quickly, e.g. for a very sick baby. As many disease-causing variants are found in the exome, this approach can provide the information needed for diagnoses.

Mainstreaming is a term used to describe the process to move genomics from the narrow confines of Clinical Genomic Services to multiple clinics and specialities across the health system from general practitioners to specialist hospitals.

Genomics Clinical Reference Group provides impartial, evidence based clinical and scientific advice and leadership to inform the ongoing development of the NHS Genomic Medicine Service.

Commissioning Implementation Plan sets out the actions that must be taken and timescales required to introduce the new Service Specification.

13Q Assessment sets out the approach we have taken to meet our statutory duty under section 13Q of the National Health Service Act 2006 (as amended) to make arrangements to involve the public in commissioning services for NHS Patients.

Clinical Geneticists are doctors with a primary Medical Qualification, membership/fellowship of a relevant Royal College or equivalent, and entry onto the GMC Specialist Register for Clinical Genetics. Clinical Genetics is a medical specialty affiliated to the Royal College of Physicians.

Genetic Counsellors are health professionals demonstrating professional competence in genetic counselling at Masters level through either accreditation by the Genetic Counselling Registration Board (GCRB) for inclusion on the Academy for Healthcare Science (AHCS) register; or as a clinical scientist (genomic counselling specialty) with the Health and Care Professions Council (HCPC).

Protected characteristic groups are protected from discrimination by law. Protected characteristics include age, disability, gender reassignment, marriage and civil partnership, pregnancy and maternity, race and ethnicity, gender, and sexual orientation.

Health inequalities can be experienced many groups of people including looked after and accommodated children, carers, homeless people, people involved in the criminal justice system, people with addictions and substance misuse problems, people on low incomes, people who have poor literacy, people living in deprived areas, those living in remote, rural and island locations and any others in groups that face health inequalities such as veterans.

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