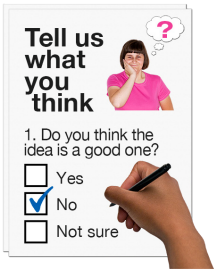
Clinical Genomic Services: Revised Service Specification Public Consultation

Consultation Guide

Easy read

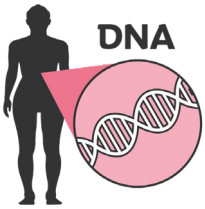
About this Guide

NHS England works with patients, patient groups and others to develop its services.



This consultation helps us check whether our ideas for change are right, that they are clear and that we look at all options before we decide.

It helps tell us what *you* think about our suggested changes to the Clinical Genomic Service Specification.



Clinical Genomics are services where doctors and Genetic Counsellors work with other professionals to diagnose genetic conditions and look at the risks whether a patient will inherit a condition.

They offer treatment for patients with genetic conditions and support to them and their families.

This easy read guide tells you:

- How care is offered at the moment
- How a new service specification will change the way services are delivered
- How the changes will happen
- how you can tell NHS England *your* views.

We will look at everybody's views before the new Service Specification is decided.

Documents in this consultation

- Clinical Genomic Services: Draft Service Specification
  This describes the clinical service and sets out standards providers must meet.

- Equality and Health Inequalities Impact Assessment
  This assesses the impact of the new Service Specification on groups that may be affected by changes to the Specification.

- Stakeholder Testing Engagement Report
  This tells you about our testing. It asks for your views on the Service Specification and Impact Assessment.

- Integrated Impact Assessment
  This tells you about the possible impact on patients and services of the changes to the Specification.

List of difficult words

Whole Genome Sequencing

Analysing the whole genomic DNA sequence to diagnose a rare condition or identify a genetic change in a cancer that may be important in treatment decisions.

Molecular Diagnostics

Finding out genomic changes to diagnose disease and check how well treatment is working.

Genomic Testing

Using different technologies to find changes in genes that can cause health problems.

Multi-disciplinary Teams

Groups that bring together skills of different professions working together to manage care.

Teams may include oncologists, specialist nurses, clinical geneticists, genetic counsellors and others.

Genomic Test Advisory Boards

These review complex genomic test results. Boards feed into clinical teams to help make final decisions.

Rapid Exome Sequencing

This gathers genomic information for diagnosis whilst allowing tests to be done quickly, say, for a sick baby.

Mainstreaming

Moving genomics to clinics and specialities across the health system - which includes all services from GPs through to Specialist Hospitals.

Genomics Clinical Reference Group

This group provides clinical and scientific advice to develop the Genomic Medicine Service.

Commissioning Implementation Plan

The actions needed to bring in the new Service Specification.

Genetic Counsellors

These are professionals trained in genetic counselling.

Protected characteristic groups

These 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.

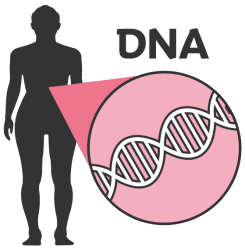
Health inequalities are experienced by many:

- looked after children
- carers
- homeless people
- people involved in the criminal justice system
- people with substance misuse problems
- people on low incomes
- people who have poor literacy
- people living in deprived areas
- those living in remote, rural and island areas
- any others in groups that face health inequalities.

Background

Genetic testing offers patients new diagnoses and treatment choices.

The NHS set up the 100,000 Genomes Project. This showed the benefits of whole genome sequencing technology.



The Project helped us understand the changes that we need to make to how genomic medicine is delivered in the health care system.

NHS England has worked with partners to develop this: the NHS Genomics Medicine Service.

This includes:

- Seventeen NHS Clinical Genomic Services that deliver a genomic and counselling service.

  This directs the diagnosis, risk assessment and clinical management of patients and their families affected by a rare genetic or genomic condition.

- A national network of NHS Genomic Laboratory Hubs who deliver the genomic testing in each region.

- Seven NHS Genomics Medicine Service Alliances that embed genomics into care locally and for everyone in England.

  The Alliances bring together multi-disciplinary expertise to make this happen.

The NHS Genomics Medicine Service helps deliver the 2019 NHS Long Term Plan commitments that helps us to use genomics to improve everyone's health.

These commitments include:

- To be the first health care system to offer whole genome sequencing as part of routine care. This includes all children with cancer and children who are seriously ill with a genetic disorder
- Extended access to molecular diagnostics and offering genomic testing to people with cancer
- Early detection and treatment of high-risk conditions
- Linking genomic data to provide new diagnoses, treatments and helping people make decisions about their care

The NHS Genomics Medicine Service uses the latest science to deliver better healthcare at lower cost. This is set out in Genome UK: The future of healthcare (2020).

This vision for the future focusses on three areas:

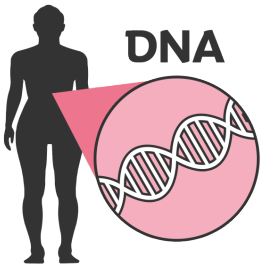
- Diagnosis and personalised medicine and bringing genomics into routine healthcare for better diagnosis and treatment.
- Preventative care to improve health.
- Research, supporting research and linking research and healthcare delivery.

The NHS Genomics Medicine Service helps deliver the NHS Long Term Plan and Genome UK: The future of healthcare through providing:

- Consistent and equitable care
- Common national standards
- A single Testing Directory using all technologies from single genes to whole genome sequencing
- The chance for all patients to take part in research to make care better for them and future patients
- A genomic knowledge base.

Changing the Clinical Genomic Service Specification

This consultation looks at the 'NHS Clinical Genomic Service' part of the NHS Genomics Medicine Service.



The new specification will reflect developments in the NHS Genomics Medicine Service, such as:

- Delivering NHS Clinical Genomic Services as a network, working with the NHS Genomic Laboratory Hubs supported by the NHS Genomics Medicine Service Alliances.
- New ways of working through online, phone and face-to-face clinics, multi-disciplinary teams and genomic test advisory boards.
- Recognising the roles of all health care professionals within clinical genomics to best use people's skills.

- Helping the workforce reflect advances in genomic testing technology, rapid exome sequencing, specialist genetic testing, increased range of cancer genomic testing, and whole genome sequencing.
- The need to provide genomic testing to all groups based on clinical need.
- A bigger role for mainstream medicine for delivering genomics.

This Consultation Guide sets out how the new Service Specification will help our commitments for delivering genomics within healthcare.

Updating the Service Specification

The Specification was updated between July 2020 and March 2021. We asked lots of people what they thought of the updated specification as we developed it and used their feedback to improve our work.

A review of the Specification was done in March 2022 following feedback, and publication of a new specification template and guidance by NHS England.

From this, changes were made, and these were checked in May 2022.

We have worked to understand the impact of the new service specification on patients, staff and the health care system. The impacts and any risks identified will be used to develop the Commissioning Implementation Plan.

How is care provided at the moment?

- Clinical Genomics supports patients and families with genetic and genomic conditions.
- Patients with rare diseases are often not diagnosed for a long time. Others may have cancers with a genetic predisposition or are more likely to develop certain tumour types.
- People with rare diseases often have many tests before a diagnosis can be made. So their quality of life can be poor, with treatments prescribed to manage symptoms.
- Genomic testing can diagnose genetic conditions,

helping patients get the treatment they need.

It can tell patients if they have a bigger risk of developing a condition so they can be monitored and make choices about their health care options.

- Genomic testing can mean life-changing diagnoses. Patients and families need the right support to cope with their results.

There are seventeen NHS Clinical Genomic Services across England. Each Service covers a particular area.

The COVID-19 pandemic meant that there are fewer locations from which face-to-face services are provided. However, more patients now use services over video or phone.

The clinician may review patients to reach a diagnosis or decide which tests would help diagnosis.

Clinicians are usually Clinical Geneticists or Genetic Counsellors, but some services also have specialist nurses and family history co-ordinators.

The clinician may organise genetic testing, which will be done by one of the NHS Genomic Laboratory Hubs. There will be follow-up appointments to discuss test results.

The NHS Clinical Genomic Services provide long term support for patients and their family. This might mean regular monitoring and counselling. Some patients stay under the care of the service for years. Others are discharged and return to the service later when they need more support.

NHS Clinical Genomic Services work closely with other clinical specialties in hospitals.

The Services provides expertise to clinical specialties and advice where complex clinical management or interpreting genomic results is needed.

What changes are suggested?

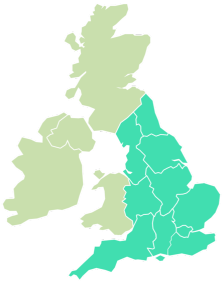
NHS England is publishing a new Service Specification for NHS Clinical Genomic Services.

We suggest five priorities arising from introducing the new Service Specification:

1. Embedding genomics into mainstream clinical pathways to increase the confidence of clinicians to ask for the right genomic testing and understand how results can affect patient care.
2. Care for patients and families will be co-ordinated. Health agencies will work together to achieve shared outcomes.

3. Everyone who would benefit from the NHS Clinical Genomic Service and/or genomic testing is able to access it in line with their clinical need and the National Genomic Test Directory.

4. Better outcomes for patients and their families, helping patients to get a diagnosis sooner and delivering personalised care.
5. NHS Clinical Genomic Services will form networks matched to the NHS Genomic Laboratory Hub and NHS Genomics Medicine Service Alliance areas. These seven regional networks:
    - will share best clinical practice
    - support workforce development



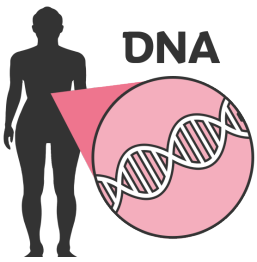
- do research and develop guidance
- set direction of the NHS Genomics Medicine Service
- make sure patients get equal access.

How might the new specification affect care and services?

1. Mainstreaming Genomics

This supports all healthcare clinicians to make genomic medicine part of their work.

NHS Clinical Genomic Services, with their NHS Genomic Laboratory Hub and NHS Genomics Medicine Service Alliance, will support and train those in other specialties to understand genomic testing and understand patients' results.



The NHS Clinical Genomic Services will develop pathways to embed genomic testing, offer advice for patients with genetic conditions and offer genomic testing as part of cancer care.

Clinical geneticists and genetic counsellors will work jointly with mainstream clinicians.

Jointly developed referral pathways will be in place to check that patients with complex needs are referred to NHS Clinical Genomic Services or other services.

There will be support for mainstream care of patients who may not need a referral to Genomics through advice by phone, video or email.

Mainstreaming genomics will mean that fewer service users are referred to NHS Clinical Genomic Services for testing as more speciality clinicians do this.

Greater genomic awareness in mainstream specialties should mean more patients are diagnosed with a rare disease or cancer with a genetic predisposition.

This should lead to more patients with complex disorders referred to Clinical Genomic Services.

2. Co-ordinated Care

Many people with rare diseases need care from lots of services because their condition is complex.

This can mean them going to appointments on different days, with different professionals in different clinics.

They may not have a named care co-ordinator so need to organise appointments, chase test results and repeat their story to different clinicians.

NHS Clinical Genomic Services will co-ordinate care with other parts of the system so care is provided smoothly.

This will link to the work developing pathways with mainstream clinical specialties, but also make sure there are pathways to specialised care, treatment, drugs, mental health and special educational support.

NHS Clinical Genomic Services will also take part in the Rare Disease Collaborative Networks. Each is made up of providers with an interest in a particular disease.

They work together to do research and make patient experience and outcomes better.

3. Equity of Access

Our aim is that all patients who would benefit from genomic testing, and care provided by clinicians confident with genomics, can access it.

We will work with services to make sure *all* eligible people can use services, *regardless* of where they live.

The number of eligible people depends on the type of population in their area. Rare diseases are more often diagnosed in childhood and some genetic conditions are more common in people from certain ethnicities.

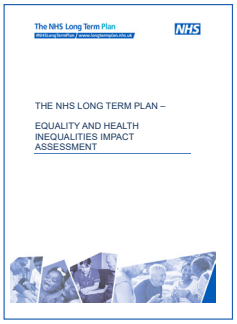
The Service Specification means that each NHS Clinical Genomic Service must prioritise genomic care for all eligible patients.

This means services will engage with seldom heard communities and recruit into patient groups.

NHS Clinical Genomic Services will run activities and act on feedback from the public to develop services.

They will reduce barriers to accessing NHS Clinical Genomic Services, testing and research and better engage with communities not reached in the past.

Services should provide for any person within a protected characteristic group that faces health inequalities.

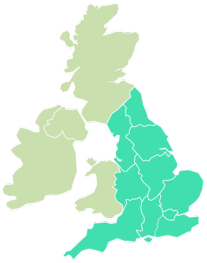


Find out about how NHS England has considered these groups in the Equality and Health Inequalities Impact Assessment.

Where patients cannot access the NHS Clinical Genomic Service through mainstream services or their GP, national guidelines for self-referrals will give more patients access.

COVID-19 has led to fewer sites where clinics are available. However, more patients have consultations with video or over the phone. Services will need to make sure that this does not reduce the quality of the service that patients receive.

NHS Clinical Genomic Services will check on services to make sure the service is available to eligible people across its whole area.



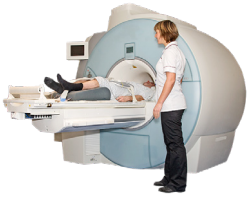
Some clinical geneticists and genetic counsellors have areas of expertise that are not available across all services. We will make this available at national or regional level to patients in *all* geographical locations.

The new Service Specification aims to provide care to those who need longer psychological support.

NHS England will work with services to make sure *all* patients can get clinical psychology if needed.

This may mean bringing clinical psychologists into Clinical Genomic Services or making sure there are referral pathways to psychologists in local mental health services.

4. Making Outcomes Better



With the mainstreaming of genomics and more equal access, we will use the latest scientific advances and deliver better health outcomes at a lower cost.

At the moment, patient can face many referrals, tests and diagnoses before getting their final diagnosis.

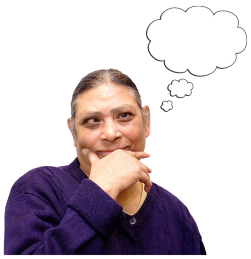
This can last many years and affect their condition and mental health.

The delay in getting a diagnosis can mean patients do not benefit from treatment.

More genomics in patient care will help diagnose rare diseases more quickly.

Using genomics can help target treatments and improve outcomes. NHS Clinical Genomic Services will work with other specialties to offer new care pathways as new genomic tests and therapies become available.

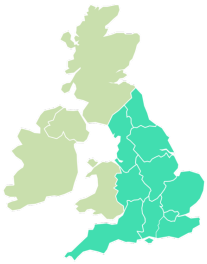
Sometimes genomic testing will identify a disease for which there are no treatments.



In this case, the aim is to help the patient get the best clinical care and support and make decisions about their future.

Genomics can also help through targeted screening. This may mean testing family members of cancer patients, to check if they are at increased risk of cancer.

5. Networked Care

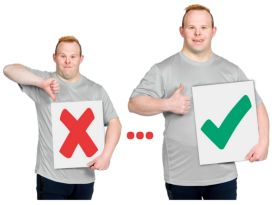


The seventeen NHS Clinical Genomic Services will form networks across the seven NHS Genomic Laboratory Hub and NHS Genomics Medicine Service Alliance areas to make services better.

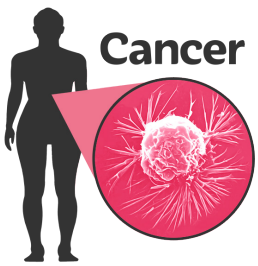
These networks will work as part of the NHS Genomics Medicine Service network to:

- share best clinical practice
- support workforce development
- research and develop guidance
- set direction for the NHS Genomics Medicine Service
- make sure there is equal access for patients and their families.

Services will recruit patients into clinical trials and translate research into practice, bringing new technologies into health care.



Each NHS Clinical Genomic Service will help make clinical pathways better, shorten diagnosis time and reduce the burden on patients.



They will provide clinical expertise to support NHS Genomic Laboratory Hub and clinical specialty colleagues.

Cancer genetics teams will work with cancer specialties to create pathways into inherited cancer teams.

6. Workforce planning and development

The NHS Clinical Genomic Services needs enough staff with the right skills to provide a quality service, without limiting their activity or creating long waiting lists.

NHS Clinical Genomic Services need enough staff to develop new pathways of care, mainstream genomics and, with the NHS Genomics Medicine Service Alliances and Health Education England, provide education, training and develop skills needed in mainstream services.

As well as direct patient care, demand for NHS Clinical Genomic Services will increase in other ways as genomics is embedded into the mainstream, such as:

- Education and training to mainstream services
- Time spent in multi-disciplinary Team meetings
- Time spent providing advice and guidance to mainstream services.

So we need to plan for the workforce to support the extra burden on NHS Clinical Genomic Services.

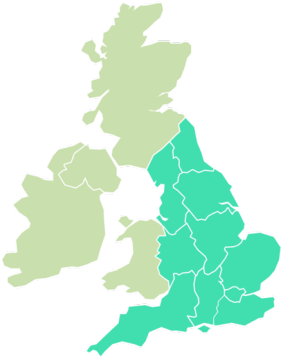
Patients need to receive care from the most appropriate clinician, which may be a Clinical Geneticist, Genetic Counsellor or mainstream clinician.

We will work with each provider to plan the workforce needed for the Service Specification. This will help bring down waiting lists for patients.

We will support NHS Clinical Genomic Services to provide workforce development plans that meet local demand, population needs and aspirations.

How will the changes take place?

NHS England will work with each service to develop a Commissioning Implementation Plan that makes sure there is a national approach to the Specification, but also takes into account *regional* needs.



We expect that the Commissioning Implementation Plan will mean:

- Setting up a national reporting framework, cost of service delivery and for monitoring Quality Standards.
- Developing a tool to measure the impact of service changes on patient outcomes.

- Consistent operating service procedures, including guidance for collaboration between specialties for mainstreaming and consistent eligibility criteria.
- A service delivery model that provides equity of access including minimum requirements for providing services across each area.
- Developing cost effective workforce structures with enough staff to manage care for the eligible population, a skill mix that can develop new care pathways, providing support for mainstream clinicians and makes use of extra roles such as genomic assistants and family history co-ordinators.

- Working with Health Education England to check there are systems to increase staff to meet demand.

- Developing a financial system that supports the capacity needed and achieves equal access.

What do you think of our suggested changes?

NHS England would like to hear what *you* think about the suggested changes to NHS Clinical Genomic Services in England.

Feedback from patients, families and the public

We are asking you about the things that were important if you have used genomic services in that past, and what would be important to you in the future.

1. For patients 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 your experience of services have been better?
2. Everyone, how do you think suggested changes to the Clinical Genomic Services might affect the experience people will have of services in future.
3. If there are any changes that we have not thought of.

Feedback on all Consultation Documents

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

1. How well do you think the Service Specification meets its key aims to:
    a. Embed clinical genomics into mainstream clinical pathways?
    b. Provide co-ordinated care for patients and their families?
    c. Make sure those that would benefit from the Service or genomic testing can access it?
    d. Improve clinical outcomes for patients and their families?
    e. Improve access to and the provision of services by aligning NHS Clinical Genomic Services to the NHS Genomics Medicine Service national network?
    f. Make sure the workforce can implement the key priorities from the Service Specification and meet more demand for services?

2. Do you think the new Service Specification will have a positive effect on patient care, outcomes and experience?
3. How well do you think the Integrated Impact Assessment identifies the potential impact and risks of:
    a. Patients and patient pathways?
    b. The delivery of Clinical Genomic Services?

c. Capacity and workforce planning?

d. Financial risk and cost pressure?

4. How well do you think this Consultation Guide and the Integrated Impact Assessment has identified the actions that need consideration as part of the Commissioning Implementation Plan?

5. How well 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. How well do you think Stakeholder Engagement has:

   a. involved the right stakeholders?

   b. helped stakeholders to be involved?

   c. considered feedback to develop the Service Specification, Integrated Impact Assessment and the actions that will be in the Commissioning Implementation Plan?

7. Are there any changes that need to be made to the Service Specification?

8. Are there any changes that need to be made to the Integrated Impact Assessment?

9. Are there more actions needed for the Commissioning Implementation Plan?

The consultation will run from:
Thursday 1 September - Friday 30 September.

You can provide your views with NHS England by completing the online survey.

You can attend one of our Webinars and Engagement Events.
Webinar 2 – aimed at patients, patient groups, charities, and other stakeholders

Wednesday 14 September, 10am – 11am
Register to attend

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

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