Clinical genomic service specification consultation:

NHS England responses to questions asked during webinar events held to support the consultation

Published: 02/11/2022

1 Purpose of this document

1.1 NHS England is seeking views on proposed changes to the Clinical Genomic Service Specification through a public consultation which opened on 1st September 2022 and will close on 6th November 2022. Two webinars were held during the consultation period, on 29th September 2022 and 3rd October 2022, providing opportunities for stakeholders to hear more about the changes and to ask questions. In order to support stakeholders submitting their responses to the public consultation, the questions asked during the two webinars are outlined below, along with NHS England's response.

1.2 Many of the questions asked by stakeholders were related in respect of the subject areas and hence questions have been grouped within this document by theme. Some questions were repeated by a number of webinar attendees, but expressed in different ways and for these, NHS England has combined questions and provided a single response.

1.3 There were some questions asked during the webinars which related to the Genomic Testing Services, which are not the subject of this particular consultation. However, taking into consideration the nature of the questions asked, relevant information about Genomic Testing Services is provided in Section 3.

2 Questions asked and NHS England responses in relation to the Clinical Genomic Services

2.1 Funding the Clinical Genomic Services

Question	NHS England's response
There are inconsistencies in resources made available to Clinical Genomic Services across England. How will this be addressed?	As a result of the work completed to date to revise the Clinical Genomic Service Specification, we recognise this issue of varying levels of resources allocated to each service across England and will seek to address this inconsistency when implementing the revised Service Specification. At the current time, information is anecdotal and there is a need to obtain robust data as part of a service review to evidence inconsistencies in resource allocation and highlight the resulting service provision issues. This service review will allow NHS England to evidence the case for change to provide greater equity of access and provision.
What are the proposals or timelines for any payment methodology changes as a result of the new specification?	There are no immediate changes to be made to the existing payment arrangements following publication of the revised Service Specification. A service review will be completed during the first year after publication. This will inform the development of a national financial model for the Clinical Genomic Service (scheduled for the second-year post publication) and shadow monitoring of the new payment methodology is expected to take place during the third year post publication. These timescales, however, are subject to change depending on how work progresses post publication of the revised Service Specification.

2.2 Geographical boundaries of the NHS Genomic Medicine Service Alliances (GMS Alliances), Genomic Laboratory Hubs (GLHs) and Clinical Genomic Services

Question	NHS England's response

Is it proposed boundaries of the current Clinical Genomic Services change to map those of the GLHs and GMS Alliances?	Currently, there are no plans to change existing geographical boundaries for the Clinical Genomic Services. During the first year following publication of the new Service Specification work will be undertaken to identify how clinical pathways align to ensure they are not disrupted but ensure there is clarity in relation to the most appropriate networking arrangements. The NHS Genomics strategy 'Accelerating genomic medicine in the NHS', published by NHS England in October 2022 (available here: NHS England » Accelerating genomic medicine in the NHS), sets out the plans to incorporate Clinical Genomic Services into the governance structures for the seven NHS Genomic Medicine Services.

2.3 **Commissioning arrangements**

Question	**NHS England's response**
What are the reasons for changing the way the Clinical Genomic Service is commissioned?	A national commissioning model will best enable commissioners and providers to work in partnership to address the issues that impact all or a number of Clinical Genomic Services and threaten the provision of high quality services which meet the needs of patients. For example, several Clinical Genomic Services have reported concerns regarding current gaps in the workforce, due to a combination of lack of adequate funding in some regions and lack of suitably qualified and experienced applicants. A national commissioning model will best enable the gathering of evidence about these and other issues impacting service delivery, building the case for change so that the service meets the requirements of the service specification in a sustainable manner and as part of an integrated system with the wider NHS Genomic Medicine Service. While the commissioning of Clinical Genomic Services will be a central NHS England responsibility in the first instance following publication of the Service Specification, the suitability of devolving commissioning to Integrated Care Boards (ICBs) will be assessed in the future.
How will stakeholders' views be represented when commissioning decisions are made?	NHS England's Genomics Unit recognise the importance of involving stakeholders in the commissioning of the Clinical Genomic Service and in having a Clinical Reference Group operating in accordance with the commissioning of other services falling under specialised commissioning. We will review the existing list of stakeholders and the approach to be taken with regards communication and engagement. Specifically, to ensure clinical views are considered in the commissioning of the service, a Genomics Clinical Leads sub-group of the Genomics Clinical Reference Group will be established including a single representative of each of the services. NHS England's Genomics Unit, alongside NHS England's regional colleagues, will ensure working relationships are developed with ICBs, delivering accountabilities and responsibilities set out in the NHS England Operating Framework (published on 12 October 2022, available here: NHS England » NHS England operating framework). Additionally, as set out in the NHS Genomics strategy, from 2023/24 the NHS Genomic Laboratory Hubs, NHS GMS Alliances and Clinical Genomic Services will establish integrated NHS GMS governance boards with appropriate partnerships and leadership to

	drive the embedding of genomics into the wider NHS. This will support the NHS Genomic Laboratory Hubs, NHS GMS Alliances and Clinical Genomic Services to work together with key partners including ICBs/ICSs, Cancer Alliances and other clinical networks to deliver genomics in their geography.
Does NHS England's Genomics Unit have appropriate experience and leadership to undertake this commissioning role?	The Commissioning team in the Genomics Unit has several years' experience in commissioning the Genomic Testing Services, NHS GMS Alliances and have worked alongside the Genomics Clinical Reference Group since 2018, supporting the revision to the Clinical Genomic Service Specification. Through the completion of this work, the Genomics Unit hold a good understanding of the risks to future service provision. The Commissioning team within the Genomics Unit have previously held roles in commissioning, contracting and planning in Specialised Commissioning, Primary Care Trusts (PCTs) and subsequently Commissioning Care Groups (CCGs) as well as previously holding roles in the NHS provider sector. This experience will allow the Genomics Unit to work effectively with providers and with commissioning colleagues at the national, regional and local levels to ensure the Service Specification is consistently implemented across England to optimise benefit delivered to patients. The Genomics Programme coordinates and oversees genomics across the NHS in England and is led by Professor Dame Sue Hill, the Chief Scientific Officer for England and Senior Responsible Officer for Genomics, who has a broad range of experience in commissioning services at all levels of the NHS. The governance structure is designed to ensure clinical and scientific colleagues, as well as patient and public representatives, provide advice and leadership to inform the ongoing development of the NHS Genomic Medicine Service. The Genomics CRG has worked collaboratively alongside the Genomics Unit in the codesign of the revision to the Service Specification. The Genomics CRG reports to the Genomics Programme Board, which in turn reports to the NHS England Board. Given the NHS Genomic Medicine Service is a specialised commissioned service, changes in commissioning arrangements, including revisions to Service Specifications are also reviewed by NHS England's Specialised Commissioning Oversight Group. This existing governance structure ensures clinicians are involved in commissioning decisions related to the NHS Genomic Medicine Service and this joint working will be strengthened under the new governance structure outlined in the NHS Genomics strategy.

2.4 **Data and information systems**

Question	NHS England's response
Will there be support for trusts to improve their Electronic Patient Record and other IT systems to	We understand this is a challenge and that the Electronic Patient Record Systems and informatic provision vary across trusts. However, securing robust data which reflects service provision is essential to inform the case for change and implementation of a commissioning plan so the service meets the requirements of the

enable accurate data capture (and audit) to support the implementation of the Service Specification?	service specification in a sustainable manner. Discussion will be held with each Clinical Genomic Service to identify what is available and the resources required to support consistent capture of data and information. The data and informatics governance arrangements outlined in the NHS Genomics strategy will enable information technology requirements and solutions to be discussed in the context of the NHS Genomic Medicine Service as a whole.
Will you involve the National Disease Registration Service in capturing equity of access?	Work is underway with the National Disease Registration Service and NHS Digital/NHS Transformation to consider how we can align different data sets and intelligence sources to enable the measurement and monitoring of equity of access across the NHS Genomic Medicine Service.
How will patients' genomics data be handled?	The NHS Genomics strategy highlights progress so far and priority actions to be taken which will enable genomics to be at the forefront of the data and digital revolution to improve patient care, drive health improvements for individuals and populations and maximise diagnosis, access to precision medicine and efficiency.
In terms of increasing equality in access to genomic services, how are the different parts (GLHs, Alliances, GPs) going to procure infrastructure for local/regional genomic data management?	The NHS Genomics strategy highlights work completed to date and further actions planned in the coming years to develop an interoperable informatics and data infrastructure that enables the NHS to use and share genomic data appropriately to improve patient care.

2.5 **Capacity and demand**

Question	**NHS England's response**
How will clinical genomic services be supported to manage pressures resulting from increasing demands and limited capacity, both now and in the future?	An essential aspect of the commissioning process is the assessment of population needs and a forecast of how these may change in the future. This informs capacity and workforce planning to ensure the services are sustainable and can deliver improved health outcomes. As detailed in the Integrated Impact Assessment, initial modelling of eligible population and potential future demand has been completed but will be revisited with input from each service during the first year following publication of the Service Specification. If there is evidence of pressing demand issues impacting some services which need to be addressed in the immediate future, this will be considered by the Genomics Unit and the Genomics Clinical Leads sub-group of the Genomics Clinical Reference Group.

2.6 **Referral criteria**

Question	**NHS England's response**
Will referral criteria be developed, including any criteria for accepting self-	NHS England's Genomics Unit will work with the Genomics Clinical Reference Group and clinical representatives to develop nationally consistent eligibility criteria for Clinical Genomic Services to adopt. This work will be completed in partnership with the Clinical Genomic Service providers.

referrals, and be applied nationally?	
Are you able to tell us more about the proposed pathway of patients being able to self-refer to genomics services in the future?	The previous service specification for Clinical Genomic Services included a provision for patients to self-refer but there may be inconsistencies in how this is applied across the country. There are times when patients may be eligible for these services but are unable to access them through traditional referral routes such as their General Practitioner. We acknowledge that self-referral is not a preferred route to Clinical Genomic Services and would encourage all individuals to be referred to the service through healthcare professionals, where possible. Self-referral is intended to act as a safety net for those that have been unable to gain access.
Will there be scope for other healthcare professionals such as pharmacists to be able to refer within a defined pathway?	Yes, referrals from other healthcare professionals, such as specialist nurses and pharmacists, are entirely appropriate. There is scope for varied health professionals to refer patients and their families into Clinical Genomic Services, in line with nationally consistent eligibility criteria that will be developed for Clinical Genomic Services to adopt and recognising there will need to be a mechanism to ensure these referrals are appropriate.
How can patient advocacy groups signpost extended family members of people with rare genetic diseases to NHS genomic testing if they are planning to start a family?	Details of the option to self-refer will be provided in the relevant section of NHS Trusts' websites and the Genomics Unit would welcome working with patient advocacy groups to ensure there is awareness of this self-referral route. Work is being undertaken to ensure that General Practitioners, who will be the first source of advice for the majority of patients, have awareness of genomics, the services available to patients in the NHS and how to make referrals.

2.7 **Service scope**

Question	**NHS England's response**
During the webinar, an increase in genomics being used in healthcare outside of Clinical Genomic Services was indicated. Please expand on this; where else would genomics be used outside of the Clinical Genomic Service?	Genomics can be and is used effectively in many areas of healthcare. It can be used to identify patients affected by or at risk of developing certain conditions and contribute to the planning of better treatment. It can also be used to support precision medicine, especially in cancer care, to define the use of targeted treatments resulting in improved outcomes. It is already effectively used within several hospital specialties such as haematology, paediatrics, neurology, pre-and post-natal care, cardiology and cancer care. There is also work underway to identify scope for genomics to inform the identification of patients with a risk of high cholesterol in primary care. As technology and knowledge advances, there is scope for genomics to be used more widely to inform the diagnosis and care of many more patients.

2.8 **Workforce planning**

Question	**NHS England's response**

The Service Specification states that a workforce plan will be developed during 2023-24. Where there is clear evidence of the immediate need to take action to address workforce issues (for example in relation to training programmes), will urgent action be taken?	NHS England's Genomics Unit will prioritise the development of workforce planning to ensure Clinical Genomic Services have the appropriate staff available to deliver care in accordance with the Service Specification. Working planning must be based on robust data and evidence, and hence, the proposal for this to follow the completion of a service review to gather activity and finance data and understand population needs. Where there is clear evidence of current workforce issues, NHS England's Genomics Unit will work with stakeholders, for example with our NHS England colleagues and with Health Education England, to agree the appropriate actions.
Will there be work on differentiating the roles of the clinical geneticists and genetic counsellors in the Clinical Genomic Service? Given the different backgrounds it would be helpful to have clarification regarding roles and responsibilities.	We understand there is overlap but also significant differences in the roles of a Clinical Geneticist and a Genetic Counsellor, particularly in terms of training, background and responsibilities. We intend to develop additional documents to support implementation of the Service Specification where it is agreed this would be beneficial, to include best practice and operational guidance, while allowing local flexibility where appropriate.
What do you consider are the reasons for the gaps in recruitment and what are the options for addressing these gaps?	We understand gaps in recruitment exist because of historical variations in resource allocation across Clinical Genomic Services and a lack of detailed workforce planning which is compounded by the availability of suitably trained and experienced applicants for available roles. During the commissioning implementation phase, NHS England will work with services to understand the workforce required to deliver the service specification and the cost pressures associated with their workforce plan. We will also work with Health Education England to identify requirements to develop the available workforce to meet future need.
There has been a significant focus on the development of new roles in providing genomic services and less focus on the staff groups that are providing support to train these new groups whilst managing significant workloads. How will this be addressed?	It is recognised that training and supervision of staff members, while essential, is a significant commitment for more experienced and senior team members. The time required to support training will be taken into consideration to help inform capacity and workforce planning. The funding for Practice Educators and the establishment of the Genomics National Training Academy will contribute to this as well as a systematic approach to genomics workforce planning and development.
Will there be workforce per unit of population recommendations?	It is not intended that NHS England will dictate the workforce to be employed within each Clinical Genomic Service. It is anticipated that quality metrics will relate to outcomes delivered by Clinical Genomic Services, rather than inputs such as workforce employed. That said, where considered helpful, NHS England will coordinate the production and sharing of best practice guidance which may be applied by individual service providers when considering workforce establishment and completing job planning.

The Specification includes a footnote to recommend a change in the Genetic Counsellor professional title to "Genomic Counsellor." While recognising that our profession has evolved with the technology, to go so far as changing the professional title seems wholly unnecessary and may negatively impact relations with international Genetic Counsellor organisations.	We acknowledge the concerns in relation to the proposed change in title to Genomic Counsellor, and the point that it could be argued a similar approach could be taken to medical colleagues. The feedback will be given full consideration as part of the consultation process.
Will the review of service provision and resources to ensure equitability, include looking at administrative support?	The service review and subsequent development of a workforce plan will consider all aspects of workforce required to deliver an effective, efficient and sustainable service including administrative functions.
One fundamental component of genomic counselling is clear communication to patients. There is variability of access to professional interpretation services by Clinical Genomic Services across the country - is this question under the remit of the Clinical Genomic Service spec?	The NHS Standard Contract states (Service Condition 13.2) *'The Provider must provide appropriate assistance and make reasonable adjustments for Service Users, Carers and Legal Guardians who do not speak, read or write English or who have communication difficulties (including hearing, oral or learning impairments).'* However, we recognise that, in practice, delivery in line with this requirement may not be consistently achieved and the Genomics Unit will ensure this service requirement is given consideration as part of the service review.
There is an implication in the draft Service Specification that Clinical Genomic Services will be much more involved in coordinating pathways for patient care. Is this correct, and will this be resourced? (Points 6.1.3 and 6.1.7)	It is considered, at least for some patient pathways, there is the need for improved coordination of care. How this improvement will be delivered is yet to be determined but NHS England expects that Clinical Genomic Services will make a significant contribution in this work. As part of the commissioning implementation phase, it will be important to define the roles and responsibilities that sit within Clinical Genomic Services to support this work. If this requires additional resources, it will be considered as part of the work to establish a financial model.
With regard to upskilling other healthcare professionals to deliver mainstreaming, will there be consideration of changes to traditional portfolios to provide 'specialist' accreditation to professional genomic boards?	The service specification is not the appropriate vehicle to consider the specific training and accreditation of staff that will deliver the service. It will be the responsibility of each Clinical Genomic Service to ensure they have the appropriate professional and skill mix to meet the needs of their population and fulfil the requirements of the commissioned service. It will be appropriate for the Genomics Unit, Health Education England, the professional organisations, registration bodies and the Clinical Genomic Services to devise a workforce plan which will address these needs and the appropriateness of training and accreditation in genomics for a range of health care professionals.

How can Clinical Genomic Services support genomics clinical nurse specialists (CNS)? Genomics CNS are involved in mainstreaming genomics in various specialties and the common link between clinical genomics, medical/surgical specialties, genomic counsellors, laboratory & research.	Genomics clinical nurse specialists form an increasingly important part of the genomics workforce, both embedded within the clinical genetics service and in other clinical services. Where they work in other clinical services their line management would be within these services, but we would expect close liaison and opportunity for continuing professional development in genetics and the joint delivery and co-ordination of care pathways. We would expect and consider within workforce planning whether there is an evidence-based need for an increase in the numbers of genomics clinical nurse specialists to deliver this important role.
The genomic training academy recently established which will play a critical role in the development of the genomics workforce. Can this be signposted in the document?	We agree, it would be helpful to refer to the Genomic Training Academy within the Service Specification. The Genomics Unit will draft an amendment to the Service Specification.
Can you provide further information about the clear referral pathways to Clinical Psychologists, including whether the referral will come from mainstream medicine who undertake testing or via Clinical Genomics?	We have no preconception of the optimal model for access to clinical psychology for patients receiving care from the Clinical Genomic Service. NHS England's Genomics Unit will work with the Clinical Genomic Services and commissioners of Mental Health Services to ensure there is access for patients and their families where required.

2.9 **Impact on pharmacy / consideration of pharmacogenomics**

Question	**NHS England's response**
Where do pharmacogenomic testing services sit within this structure? Or will any developing services run alongside Clinical Genetics, rather than within it?	Currently, genomic testing for defined pharmacogenomics is undertaken by the GLHs. The role of Clinical Genomic Services in pharmacogenomics is yet to be determined but will likely be a supportive role, rather than pharmacogenomics forming an integral part of the Clinical Genomic Service. It will be important for pharmacogenomic services to be aligned with the clinicians and healthcare professionals providing the service to patients and must utilise the expertise of pharmacists.
How do you see the decisions of pharmacists being implemented and reported back to physicians?	As more therapies are identified for individuals with (ultra) rare conditions and for pre-symptomatic treatment of individuals at risk of late onset disorders, it is expected that the Clinical Genomic Service will have a larger direct role in treatment of patients and, will, therefore, develop much closer relationship with the pharmacy profession. Feedback on the current wording of the Service Specification suggests there is scope to include greater acknowledgment of the pharmacist's role in clinical genomics. We will work alongside pharmacy representatives to ensure the Service Specification clearly outlines this aspect.

2.10 **Multi-disciplinary Team provision**

Question	**NHS England's response**

Is a 'Genomics-led MDT' one that is led by the GLH or the Clinical Genomics/Genetics service? There are currently a variety of different MDTs that CG services contribute to and lead, will all of these be reported?	As part of the Commissioning Implementation plan, we will ask Clinical Genomic Services to outline their entire portfolio of MDTs, identifying those they attend and those they lead. The information in relation to their total portfolio will be important from a workforce planning perspective. Outcome measures are expected to be reported for the MDTs that the Clinical Genomic Services are leading. It is recognised that GLH-driven MDTs are supported by Clinical Genomic Services over and above the clinical infrastructure posts that form part of the GLH. We will ensure this contribution is captured and reported.

2.11 **Patient pathway coordination**

Question	**NHS England's response**
How should patients connect different specialties with genetics when seeing numerous different departments in the hospital?	We recognise the difficulties which can occur for patients and their families when there are multiple clinical specialties involved in providing care and will continue work to ensure care provision is as seamless as possible, for example through the provision of multi-disciplinary clinics, multi-disciplinary team meetings and the increased use of video clinic appointments to facilitate the coordination of care across specialties.
How will the service spec align with the role of Community Diagnostic Centres (CDCs) and Cancer Alliances? What are the plans for these pathways?	As outlined in the NHS Genomics strategy, over the next three years, the NHS will expand and maximise the use of the NHS diagnostic infrastructure for genomic testing. For example, exploring the role that Community Diagnostics Centres could play alongside the NHS GMS across several areas, including the collection of samples from family members for inherited disease genomic testing. As part of this, the NHS will also continue to expand the genomic offer into other clinical specialities across the testing spectrum from prevention to treatment.

2.12 **Mainstreaming services**

Question	**NHS England's response**
How is the roll out /adoption of mainstreaming going to be measured? Will the rollout be prioritised for patients/specialisms where treatment choices are reliant on testing taking place?	The mainstreaming of genomic testing has been taking place for years. In fact, most genetic tests are not requested by Clinical Geneticists, but by professionals in other specialities such as cancer care, paediatrics and maternity services. However, there are recent changes: (1) in the range of genomic tests available for those clinical specialties to order and (2) scientific and technological developments mean some clinical specialties who may not have traditionally used genetic testing may now do so as evidence makes it clear that genetic factors play a significant role in certain types of health problems. There are therefore patient pathways for which clinical genomics is mainstreamed and works well. In the past, nearly all individuals who were concerned about a family history of cancer would have been referred to a Clinical Genomic Service. Whereas now, if an individual is at lower risk, they may not need a referral to the

	Clinical Genomic Service and their oncologist or specialist nurse working in cancer care is able to provide advice and order the appropriate genetic testing. Hence, it is important to recognise the Clinical Genomic Service is often best able to provide their expertise by supporting colleagues across a range of specialties, for example by attending MDTs and providing advice via phone and email, rather than every patient requiring a referral to the Clinical Genomic Service to access genomics input. It is recognised that complexities for some patients will mean a referral to the Clinical Genomic Service is the best way to deliver care. Services will continue to ensure this referral option remains open.
With the emphasis on the "mainstreaming agenda", have the necessary links to the Clinical Genetic service been considered? Is there a plan to monitor or feedback serious incidents which occur outside of Clinical Genetics in the realm of mainstreaming (such as missed results, or incorrect interpretation of results)?	NHS England's Genomics Unit will, as commissioner of the service, ensure there are clear protocols in place to support mainstreaming and ensure safe and high quality service provision. We will ensure incident reporting processes are in place. These will support the recognition of quality issues which may arise outside of the Clinical Genomic Service itself, but as part of the mainstreamed service.

2.13 **Delivering the revised Service Specification**

Question	NHS England's response
With the number of vacant posts in admin, GC and consultant posts across CGS, and unacceptably long waiting lists, how realistic do you feel the delivery of the service specification will be?	The Genomics Unit recognise there are significant workforce challenges facing the Clinical Genomic Service, which are impacting current service capacity. There is also an expectation these could increase in the future. It is important these issues and risks are evidenced and quantified to develop a case for change. The work planned, including completion of a service review, development of a workforce plan and a robust activity and service cost monitoring framework will lay the foundations to enable commissioners and providers to address challenges facing the Clinical Genomic Services including workforce issues.

2.14 **Service outcomes**

Question	NHS England's response

Please confirm the quality measures intended to be applied to the Clinical Genomic Service	The five quality measures outlined in the Service Specification reflect the clinical outcomes linked to key priority areas of new development. There is scope to review these annually. Like other NHS services, Clinical Genomic Services will be required to achieve core contractual requirements as relevant and stated within the NHS Standard Contract such as the 18-week maximum waiting time and other mandatory quality requirements. These requirements are not included in the Service Specification as they are already implicit within the Contract and apply to all clinical services. We will build this into the metrics and reporting arrangements.
Will there be clear guidance on how to collect the outcome measures? What would the denominator be for quantifying the clinical outcomes (i.e. proportion of which cases that were discussed at a Genomics-Lead MDT - Will this be all the patients referred to the CGS in the year under consideration?)	For the purposes of reporting, this will relate only to patients under the care of the Clinical Genomic Service, not those referred directly for testing into the GLH by other mainstream services. While some Clinical Genomic Services may already have processes in place to capture the required information, others don't. Therefore, we will work with each Clinical Genomic Service to ensure they are able to collect the required data and understand what information is required. The precise numerators and denominators will be shared with all Clinical Genomic Services.
Why does one of the clinical outcomes relate to discussion of risk-reducing strategies with women at high risk of breast cancer, rather than discussion of all risk management options?	Risk reduction in this context does not solely pertain to surgical options but all options available to modify/reduce risk - chemoprevention, increased screening protocols, avoiding risk increasing factors. It is expected that the full range of options is considered and shared with the at-risk individual, and we will ensure that there is clarity for all centres in measuring this outcome.
Is there an opportunity to establish robust fail-safes to ensure that the patients identified as requiring screening are referred in a timely manner and that these referrals are received? Similarly, is there is a mechanism within genetics centres to do reconciliation audits with receiving centres to ensure that all referrals have been received and actioned? Perhaps this could be a potential patient outcome to be considered.	NHS England's Genomics Unit is aware that there is a need to develop additional quality measures in relation to patient pathways and the quality of services provided more widely. This work will be included as part of the commissioning implementation phase and arise from the work undertaken to map and understand patient pathways. We will engage and collaborate with individuals with the requisite knowledge and expertise, both within NHS England with other commissioning teams responsible for elements of those patient pathways and the providers of Clinical Genomic Services, to identify the required quality measures.
Looking forwards - if we effectively mainstream testing and most of the tests and MDTs are led by other teams the numbers of clinical outcomes we can report reduces - how then can we measure effectiveness?	There will be opportunities to review the clinical outcomes included within the service specification on an annual basis. NHS England has streamlined and simplified the process so the commissioners and providers can ensure that clinical outcomes remain directly relevant to the service provided by Clinical

	Genomic and can be reviewed/refined as required as the service changes.

2.15 **Scope of the Service Specification**

Question	NHS England's response
How will the changes be applied / work in the private healthcare sector?	This Service Specification is to be applied by NHS England in the commissioning of Clinical Genomic Services provided in the NHS in England. It is not designed to be applied in the private healthcare sector.

2.16 **Communication and engagement**

Question	NHS England's response
Is there a plan to have a central information service that provides regular updates to patients on genetic research (per gene or cancer) and risk management updates (like FORCE in the US)?	NHS England's Genomics Unit will consider the approach to be taken with regards making data and information available to patients and their families regarding the development of genomic services in the NHS.
If we are a patient and want to help 'share our story' to try and improve the genetic services for others, who should we contact?	If you have a particular story you would like to share, please contact Andrew Stewart, Senior Communications Manager for Genomics at NHS England via email (andrew.stewart19@nhs.net)

3 Information in relation to the NHS Genomic Medicine Service

3.1 The National Genomic Test Directory specifies which genomic tests are commissioned by the NHS in England, the technology by which they are available, and the patients who will be eligible to access to a test. The National genomic test directory for rare and inherited disorders and cancer can be accessed via the NHS England website: https://www.england.nhs.uk/publication/national-genomic-test-directories/

3.2 Any individual or organisation is able to submit an application to update the National Genomic Test Directory and the review process and supporting documentation is published on the NHS England website: https://www.england.nhs.uk/genomics/the-national-genomic-test-directory/

3.3 All NHS providers of genomic testing services in England are required to submit to NHS England's Genomics Unit activity data on a monthly basis, in accordance with the Genomics Testing Reporting Specification: https://www.england.nhs.uk/publication/genomics-testing-reporting-specification/ This data allows the Genomics Unit to identify the volume and type of tests provided, the

test turnaround times and analyse patient demographic data with the aim of ensuring equity of access.

3.4 The capacity issues currently experienced by the genomic laboratories are recognised. NHS England's Genomics Unit are working with all GLHs to address backlogs in test activity and increase capacity to prevent backlogs developing.

4 Submitting your response to the consultation

4.1 Stakeholders are invited to provide their views on the proposed changes to the Clinical Genomic Service Specification by completing the online survey available here: Clinical genomics service specification consultation - NHS England - Citizen Space

4.2 If you have any questions about the consultation, the documentation or the webinars then please email england.genomics@nhs.net