Engagement Report for Service Specifications

Unique Reference Number	E01/S/a
Specification Title	**Clinical Genomics (Adults and Children)**
Lead Commissioner	Anita Beer, Genomics Commissioning Lead, Genomics Unit, NHS England
Clinical Reference Group	Genomics Clinical Reference Group
Which stakeholders were contacted to be involved in service specification development?	Clinical Genetics Society Joint Committee for Genomic Medicine Royal College of Pathology Health Education England Neurosciences Clinical Reference Group Cardiac Services Clinical Reference Group Paediatric Neurosciences Clinical Reference Group Chemotherapy Clinical Reference Group Paediatric Cancer Clinical Reference Group Women and Children Care Steering Group Clinical Genetic Services Genomic Laboratory Hubs Cancer Research UK Cancer 52

	Children and Young People with Cancer Coalition British Heart Foundation HEART UK Genetic Alliance Belfast Health and Social Care Trust All Wales Medical Genetics Service University of Aberdeen Royal College of Radiologists Cancer Programme University of Nottingham Cancer Genetics Group Fetal Genomics Group Academy of Medical Royal Colleges Association of Genetic Nurses and Counsellors Cancer Research UK Royal College of General Practitioners NHS Greater Glasgow and Clyde
Identify the relevant Royal College or Professional Society to the specification and indicate how they have been involved	The Royal Colleges are represented on the Genomics Clinical Reference Group and within the Specification Writing Group.
Which stakeholders have actually been involved?	Association of Genetic Nurses and Counsellors and Lead Genetic Counsellor Group British Thoracic Oncology Group Clinical Genetics – Guys and St Thomas's Hospital Clinical Genetics Society Genetic Alliance UK Genomics England Joint Committee on Genomics in Medicine Liverpool Women's Hospital Manchester University Hospitals NHHS Foundation Trust North East Thames Regional Clinical Genetics Service North Thames Genomic Laboratory Hub Oxford University Hospital NHS Trust Regional Clinical Genetics Service Clinical Leads Group

	Roche Products Ltd Peninsula Clinical Genetics Service South West Genomic Medicine Service Alliance Bristol Clinical Genetics Service UK Cancer Genetics Group UK Clinical Expert Group for Lung Cancer and Mesothelioma Wessex Clinical Genetics Service West Midlands Clinical Genetics Unit Yorkshire Regional Genetics Service
Explain reason if there is any difference from previous question	In total, the Draft Service Specification was circulated to 318 individuals. Responses we received from 23 individuals/organisations.
Identify any particular stakeholder organisations that may be key to the specification development that you have approached that have yet to be engaged. Indicate why?	The NHS Genomic Medicine Service (GMS) Alliances were not engaged formally during stakeholder testing as the network was not fully established. However, members of the NHS GMS Alliances primarily emanate from pre-existing services within the NHS GMS so should have had the opportunity to engage through original stakeholder circulation list.
How have stakeholders been involved? What engagement methods have been used?	Three Clinical Genomics Specification workshops took place during July and August 2020 with stakeholders from Clinical Genetics, NHS Genomics Laboratory Hubs (GLHs), primary care, the voluntary sector and devolved nations. Three workshops were undertaken where attendees took part in discussions in relation to four key areas of the service specification, including: • Equity of access • Workforce planning • New models of working • Gaps in the current provision. A Clinical Genomics Service Specification Working Group was formed in July 2020, made up of a sub-section of the Genomics Clinical Reference Group members. The aim of the group was to review the outcomes of the initial workshops and produce the draft revised service specification. An update was provided to the Patient and Communities Forum in March 2021, giving the group an update of the work undertaken to

	revise the service specification to date and to highlight the informal stakeholder testing of the revised specification and associated Equalities and Health Inequalities Impact Assessment (EHIA) due to commence. Informal stakeholder testing took place from the 12th to the 26th March, 2021. An email explaining the stakeholder feedback process was issued to all identified stakeholders from the NHS England Communications and Engagement Team, on behalf of the Genomics Clinical Reference Group, detailing the background and purpose of the stakeholder testing process. Stakeholders were asked to provide feedback on both the revised service specification and the EHIA. All stakeholders were asked to provide their feedback using a standard template, which asked for comments, where applicable, for the following areas: • The service specification • The quality indicators • The Equality and Health Inequalities Assessment
What has happened or changed as a result of their input?	Feedback was received in relation to the content of the service specification, specifically in relation to improving the clarity of network and commissioning governance and consistency of terminology. Changes to the specification in response to feedback were made before sign-off by the Genomics Clinical Reference Group and consideration by the Patient and Public Voice Assurance Group. Feedback also indicated that further consideration was required in relation to: • The affordability of additional roles and changes to patient pathways • Service capacity to manage additional responsibilities e.g. advice and guidance, Multi-disciplinary team meetings and education • The management of increased demand due to increased patient complexity, indirect patient care and virtual consultations for example, particularly as demand is anecdotally already outstripping current capacity • The complexity of data management and reporting for the proposed Quality Framework, which may require investment into Information Technology Systems and benefit from national reporting systems • Clarification of operating procedures, including those in relation to intra-speciality collaboration to achieve mainstreaming, referral criteria and eligibility for the service. This feedback has been considered and informed the Integrated Impact Assessment and will also inform the development of the Commissioning Implementation Plan.
How are stakeholders	Regular updates have been provided to the Clinical Reference Group, Specification Writing Group and Genomics Programme

being kept informed of progress with specification development as a result of their input?	Board. In addition, all previous stakeholders will be further updated through the Public Consultation process, which will include a Stakeholder Consultation Workshop within the first week of the 30-day consultation period.
What level of wider public consultation is recommended by the CRG for the NPOC Board to agree as a result of stakeholder involvement?	A 30-day Public Consultation will be undertaken in June 2022.

Stakeholder/CRG Feedback

Feedback Received	SWG response	Resulting Action
1.1.1 – clarity on the governance of this network needs explanation. 1.1.10 - a number of additional roles above current workforce availability mentioned throughout the document. List below of new/newer roles not currently included in job planning. All require long-term financial commitment rather than short-term project-based goals: • 1.2.3 – GMSA networks • 1.5.2 – mainstreaming and pathway development where not already in place • 1.5.3 – new testing • 1.6.3 – NHSE&I involvement • 1.6.6 – patient and public engagement • 1.6.7 – marginalised community champions Funding – there is (rightly) heavy emphasis on non-face to face clinical activity of various kinds – see below. Demand for this already outstripping supply and majority unfunded in current arrangements. This will need to be recognised in commissioning from the start. • 1.4 – MDT input; limited availability in national job plan and probably insufficient for what is projected in the document • 1.5.2 - increasing complexity requires increased time and expertise input per patient; needs to be reflected in funding streams • 1.5.6 – advice and guidance to non-genetic specialties 1.3.3 – will MDT activity be counted in the clinical interactions under the referral to treatment guidance? Not clear if this is covered by point 1.2.7. 1.4.4 – MDT tools to facilitate MDTs – whose responsibility to develop given that national data sharing important? Some was under development (GEMS) but seems to have disappeared. See also 1.2.1 re IT facilities.	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

1.7.1 & .2 - workforce – whose responsibility will it be to provide the additional staff to ensure the relevant equity of provision/access 2.1.2 – this suggests that there are going to be national referral and triage guidelines – is this the case? 3.3.1 – acknowledges predictable increasing demand. If NHSE&I are the sole commissioner for the NCGS (*cf 1.9.1*) there is no mention of how they will interact with local commissioners – or are they commissioning regional CGS too? This is not clear. 3.2.6 – different geographical needs. In particular those from consanguineous populations (Annexe F) – the additional need here is likely to disproportionately affect some CGS more than others – how will NCGS make sure that the support and funding required for equitable access across regions is available? 101-105: data management will cross Clinical and laboratory Genetics; needs investment in IT systems and would benefit from a national system of reporting. 201-204 and 308: even for units where this is already in place (limited) this is a huge amount of activity and the measurement events are too frequent.		
There would be a need to have an SOP on how relevant clinicians (who are caring for patients with rare diseases) will take part in the CGS activities (MDT attendance, training etc.). The pathway for patient referral from centres with clinical expertise in rare diseases to CGS should be standardized nationally. Similarly, the pathway to refer from CGS to relevant specialties should be standardized and well known. A list of centres with clinical expertise in rare diseases (listed per specialty) should be made available to CGS to help develop appropriate referral pathways. We would recommend that adult respiratory medicine be included in the list of relevant clinical specialties due to sufficient evidence of the role of telomere related gene mutations in the pathogenesis of rare lung diseases, therapeutic response and prognosis (such as in interstitial lung diseases e.g. idiopathic pulmonary fibrosis). Representatives from CGS should also be encouraged to attend clinical MDTs to improve understanding of the rare diseases from the clinical care perspective. It would be helpful to add an indicator referring to the need to have a delegate/representative clinician from specialties involved in the clinical care of rare diseases – i.e. adult respiratory medicine (other than cystic fibrosis, such as interstitial lung disease), transplant, rheumatology, radiologist etc. present during	Acknowledged	Issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

the CGS MDT. It is helpful to promote the creation of networks between clinicians who are treating rare diseases and the CGS by integrating clinical care and genetic MDTs. 205 Self-referral to the service is facilitated It would be helpful to have a process in place for self-referral of asymptomatic cases of relatives of patients with confirmed mutations or high clinical suspicion of rare disease with a genetic component. (In our case – the example of interstitial lung diseases with a familial component or high suspicion of short telomere syndrome).		
The specification does not go very far towards mainstreaming. Genetic counsellors should be based in specialties with links to CGS for education and support – hub and spoke. Will encourage mainstreaming. Inherited cancer services should be embedded in cancer services. At present cancer services only see patients with variants who have cancer. The specification notes the need for interaction but ideally all patients with a cancer pathogenic variant should be seen in the cancer service and cascade testing should be delivered though through GPs with patients seen as required. – 1.4.3 acknowledges the close working needed. Better for patients if based in one service and this needs to be the service that is best placed to give informed advice on management and prognosis. Further, embedding cancer genetic services in cancer services has been shown to encourage to patients to attend appointments and engage with testing, and gives them access to better information and management. Agree that CGS have a significant role in education but there needs to be dedicated funding for consultant PAs and counsellors to deliver education 1.5 1.1.8 Also agree that geneticists have an important role to play in research but again there needs to be dedicated and funded PAs for this. With advances in genomic therapies this will become increasingly important and will generate income from the commercial sector but we need to be research-ready and so advocate funding up front 1.5.5 – cardiac services are an example where genetics could be embedded with clinical genetics only needed for complex syndromic cases maybe. This already works well in some places. Neurology is the other big example here 2.1.5 – most counsellors should be specialty based and experts in these areas working closely with	Acknowledged There are currently no plans to revise the Clinical Genomic Service Structure to reduce the number of services provided, rather ensure that there is clarity in relation to the pathways from existing services within the GLH configuration	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

relevant clinicians who are best placed to discuss prognosis and options. This applies to 2.1.6 and 7. A good example is embedding genetic counsellors in FMUs – at least the tertiary ones – will facilitate delivery of new services such as fetal exome sequencing and NIPD. Good for patients. 2.1.8 – joint appointments are an option for now but the aim should be to mainstream and not need a clinical geneticist at specialty appointments except for complex multisystem cases. Joint appointments waste a lot of time for clinicians and patients 2.1.9 – there is a lot of work ongoing re care coordination for complex genetic disease and if this rests with clinical genetics the burden will be huge. Disagree with this being a role this needs addressing elsewhere with dedicated care coordinators 3.2.5 – if cancer genetic services are embedded in cancer services this will be streamlined A: Please confirm the structure – assume there will be 7 CGS services aligned with the GSA and GLHs so there will be some shift in boundaries served to align CGS with the GLH/GMSA. So some services merged.		
Our organisation welcomes this draft Service Specification for the Clinical Genomics Service, in particular the inclusion of responsibilities related to supporting the whole genome sequencing service, rare disease and cancer genomic implementation, pharmacogenomics, research and polygenic risk score development. Our organisation looks forward to continuing to work with the Clinical Genomics Service on the 100,000 Genomes Project legacy, Genomics England Clinical Interpretation Partnership liaison, the National Genomic Research Library and the National Genomic Research Collaborative.	Acknowledged	Operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase
The service specification states that regional genetics services will provide a mixed model of service delivery with face to face appointments, virtual telephone and video consultations and app based platforms as deemed appropriate by the service. We would request that “assessment and advice” letters are added to this mixed model. A number of patients at increased risk of familial cancer are offered advice by letter only, which includes documentation and recommendation of risk reducing and early detection interventions which we would consider a “clinical interaction”. We note that in point 314 of section 4 a specific indicator is that “there are agreed clinical guidelines as part of the service specification” and in section 1.15, it is stated that the NCGS is committed to rapid adoption of robust new clinical evidence to improve patient care and outcomes. We welcome this commitment, however we would like to take the opportunity to highlight that whilst advances in genetic testing technologies and decreasing testing costs have resulted in the significant advance in the National Genomic Medicine service, for the vast majority of genetic tests currently listed	Acknowledged	Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

in the National test Directory, robust evidence-based clinical guidelines that link accurately predicted genetic risks to the proportionate clinical action are currently lacking. In the absence of systematic, evidenced risk-stratified use of intervention, at the patient-level there is risk of harm and at the population level there is diminution and dilution of the clinical and health-economic benefits that these technologies can offer. Whilst there are recognised bodies in the UK such as the National Institute for Health and Care Excellence (NICE), who are committed to the development of high quality, evidence based clinical management guidelines, their standard approach to evidence testing and guideline development is not always directly transferable or suited to the typically uncommon or rare genetic syndromes affecting modest numbers of individuals in a rapidly evolving field. The development of these guidelines within Clinical Genetics needs to be supported, both in recognition of the time taken to develop such guidance and the final sign-off of guidance by NHSE. In order to address this lack of guidance we would welcome • Recognition by the Genomics Board and NHSE that development and ratification of expert-led evidence-based clinical management guidelines are an essential element of the Genomic Medicine Service • Establishment of expert clinical-academic groups for guideline development • The expert groups should engage the appropriate GLH/GMSA leads, relevant academic experts and broader clinical community to identify and prioritise areas most urgently requiring clinical guidance. In partnership with NHSE, reflecting the principles enshrouded in NICE guideline development, a framework for 'mini-NICE' guideline development should be established resulting in a process of formal endorsement of finalised guidance from NHSE C- We welcome the fact that the service specification excludes “individuals with results from commercial direct to consumer genomic tests which do not meet referral criteria nor referrals from private providers (Annexe c). However, it is hard to relate this to the information in Annexe C and clearer definition and guidance of this would be appreciated.		
This document is predominantly about germline gene testing and not cancer molecular analyses. However, when cancer gene testing we sometimes identify germline variants. This likelihood is increased with use of ctDNA NGS analyses. I therefore have a few comments in addition. 1. The clarification around Cancer genetics services is welcome. However, it is unclear What is the process for Clinical Genetics service review for patients with cancer that undergo somatic cancer NGS and are identified to have “off target” germline risk variants eg germline pathogenic BRCA1 variant in a lung cancer proband without a breast cancer family history, where confirmatory	Acknowledged	Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

germline testing is not currently funded within the NHS test directory? 2. What is the process for Clinical Genetics service review for patients with cancer that undergo somatic cancer NGS and are identified to have “on target” germline risk variants eg germline pathogenic BAP1 variants in mesothelioma, where confirmatory germline testing is not currently funded within the NHS test directory? 3. It is paradoxical that the document talks on p8 about “mainstreaming” genetic testing, but then the funding for tests occurs either via the NHS England Genetics contract if requested by a clinical geneticist or requesting clinician/ the organisation, if another clinician. Unless there is dedicated clear commissioning for genetic tests outside the clinical genetics service this will result in an inequity of care and lack of mainstreaming implementation due to lack of clarity of testing reimbursement and a postcode lottery where some Trusts may pay for testing and some will not.		
By extension this specification creates significant work for GMS Alliances, which may not be explicit in the agreed establishment, priorities and annual business plans of GMS Alliances. Section 1.1.1 States *The network will comprise the Clinical Genomics Services, NHS GMS Alliances and GLHs across each of the seven geographies to optimise access to and provision of Clinical Genomics Services*. But the GMS Alliance ***is*** the network (or Alliance) in which the GLH and clinical genetic services will work. The GMS Alliance has no identify outside of these organisations. The GMS Alliance also includes other partners in their geography, such as acute providers, primary care and ICSs This confusion is repeated in 1.1.6, 1.6.5 and 1.8.1. and 4.2 indicator 303. Therefore 1.8.2 should start *The regional GMS Alliances will underpin the national network.* This confusion is generally repeated throughout by stating “with their GLH and GMS Alliance” – which for these purposes the GLH is a subset of GMS Alliances if not synonymous in the context of many of the statements within the specification. **Annexe D** states that the following are not in scope • Services commissioned as part of another specialised service • Where provision is in mainstream services. I do not think these are defined well, certainly not in the service specifications of the other specialised or mainstream services. Such specifications may not even exist, especially for CCG commissioned services. This leaves considerable room for local disagreement about the amount of clinical work that is expected by this specifications of other service and hence local arguments about funding	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase
The service specification is broadly supported and welcomed by our service as a comprehensive and	Acknowledged	Workforce and funding to be

ambitious document. In order to deliver this fully, additional development, resources and funding will be required, including significant workforce expansion. It would be helpful to have consistency of the terminology used, e.g.'Clinical Genetics' and 'Clinical Genomics' are used interchangeably - this may be confusing if is not clear whether it refers to existing Clinical Genetics services. Will the name of the medical specialty be changing to Clinical Genomics? In addition, 'Genetic diagnosis/Genetic condition' and 'Genomic diagnosis/Genomic condition' are also used variably – there is a difference between these terms. Many patients seen in Clinical Genetics/Genomics services have genetic conditions, and these diagnoses may be made after a genomic test 1.1.6 and 1.5.3 - Polygenic risk scores – not currently within the scope or practice of Clinical Genetics services so additional training and development of this service would be required to deliver it. 1.3.2 – MDT Clinics – services are keen to participate in these, but current funding models limit their number and frequency. 1.6 - Can the relationship between the Clinical Genetics Services and GMSAs be further defined for clarity? How does the network of CGS clinical leads fit in with the existing GLH and GMSA structures, and with NHSE/I? 1.6.8 - Consultant Clinical Geneticists and Genomic Counsellors working within mainstream specialties should be appropriately trained and registered (e.g. with GCRB/HST for Genomic Counsellors) 1.7.2 – how will the discrepancy of workforce staffing and differences in consultants/GCs per head of population between CGSs be addressed? 1.4. and 2.1.11 – MDTs – will patients require referral to MDT, will this form part of the RTT pathway, and how will this clinical activity be recorded, monitored and funded? It would be helpful to distinguish between different types of MDT and ensure consistency of terminology used. 2.1.1 More detailed guidance on guidelines and referral criteria would be welcomed. 2.2.1 – Referral and triage guidance – further clarity regarding how this will be developed and the degree of flexibility to take in to account local and regional variations due to differing populations, workforce and other infrastructure We are broadly supportive of the indicators, but significant development work is likely to be required to ensure they can all be achieved. This may vary across different services, reflecting differences in existing tools, resources and structures. 102 – does this refer to cases from the CGS only, or does it also include those requested by other	Consistency of terminology included in updated specification draft	considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

specialties? 205 – further guidance regarding self-referrals would be helpful, current practice varies across the country. 306 – further guidance (regarding 'the provider') and the funding models for MDT Clinics would be welcomed		
The Clinical Genetics Society broadly supports the proposals outlined in these documents which will provide greater uniformity to the Regional Clinical Genetics Services across England. We would welcome greater clarity on the governance arrangements for the service. For example, the relationship between the NCGS and the regional services as outlined, is confusing: 1.1.1 states that 'The NCGS is delivered by the RCGS'; 1.7.3 'The NCGS will work with the RCGS'. 1.6.3 The clinical leads will represent their service at meetings of NHSE/I as part of their organisational structure. It would be helpful to have an outline of the existing NHSE/I organisational structures for the GLH and GMSA and how the clinical leads for the Regional CGS fit in with those. 1.2.4 'The majority of the NCGS.. *is delivered*.. as part of a managed network of services'. Existing arrangements are largely informal and variable between different services; it would be helpful for arrangements to be formalised and supported by the Genomics Unit in a uniform manner across England to ensure equity of access and provision. 1.7.2 It is well known that there is inequity of provision across different genetics services with huge disparities in the workforce between centres. It is unlikely in the short term that it will be possible to significantly increase the clinical workforce in those services that are relatively understaffed. How might NHSE/I support services to address this disparity? 1.4 and 2.1.11 The terminology used for MDTs is inconsistent and confusing, for example, there is reference to Clinical Genomics MDTs, Genomics MDTs, Genomic Medicine MDTs and Clinical and Specialty MDTs. There are two main subtypes of MDT – those in which clinical management only is discussed, and those that include scientists in which genomic data is incorporated into the discussion. Please could a consistent approach to nomenclature be developed? 1.4.4 'MDT tools…will be required'. The roll out of a national MDT system (GEMS) is awaited. In the short term, are individual clinical services expected to find a solution? 1.3.3 Does discussion at an MDT constitute a patient interaction and affect the referral to treatment pathway? 1.4.5 The availability of access to National expertise is welcomed. Will there be a formal process for	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

setting up National MDTs supported by the Genomics Unit, or will these be developed by the local services in which the expertise exists? 2.1.1 refers to detailed guidelines and referral criteria. These are briefly outlined in Annexe C & D. It would be extremely helpful if the guidelines could be developed in more detail to ensure a national approach. Indicator 306: 'The provider will offer multidisciplinary clinics to ensure that patients with complex needs are referred to clinical genetics…' Please clarify what this means. Does the provider refer to the host Trust or to the clinical genetics service? Does this imply that clinical geneticists/GCs join other specialists in consultations eg neurology, cardiology, or that MDCs should be established for a suite of specific conditions? I'm uncertain how the provision of a clinic can ensure that patients are referred to it. Indicator 314: 'There is adherence to agreed clinical guidelines'. Would these include guidelines produced by expert groups, for example, the Tuberous Sclerosis Complex UK guidelines? Indicator 317: 'The CGS has an operational policy for the storage and usage of genomic data'. As these indicators refer to the clinical service, genomic data would usually only be held as a genomic test result. Is the CGS required to have a policy for the storage of raw data independent of the laboratory?		
The Clinical Leads group broadly welcomes this very comprehensive service specification which defines an ideal and aspirational National Genomic Service. We have concerns that many of the roles that we currently undertake and additional new or expanded roles in the service specification require significant additional funding and workforce expansion to achieve. This includes the greatly increased MDT input, teaching advice and guidance in support of mainstreaming of genetic testing, increased complexity of genetic results , increasing demand, expanded patient and public engagement and work around consanguinity. Proper counselling /consent and organisation of samples for WGS is very time consuming and cannot be absorbed without workforce expansion. There is inconsistent use of genetic/genomic terminology throughout the document. The currently recognised professional title is Genetic Counsellor not Genomic Counsellor. 1.1.6 and 1.5.3 Polygenic risk scores and pharmacogenomics have not been part of the Clinical genetics training curriculum and the workforce will require further development to ensure that they can input into the mainstreaming of these. 1.3.2 joint clinics are often beneficial but previously funding arrangements for these have discouraged their development 1.3.3 Does discussion at an MDT constitute a (funded) patient interaction and affect the referral to treatment pathway?	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

2.1.2 and 2.1.12 How are these being drawn up? Retaining local flexibility in delivery is important 3.2.6 Concerns that there are differing geographic needs eg supporting consanguineous populations is likely to disproportionately affect some CGSs- how will the NCGS make sure support and funding required for equitable access across regions is available? General comment: many centres have concerns about development work needed to be able to collect some of the outcome data and the MDT tools and admin support required. 102-104: will we be expected to report as a trust or only on those tests ordered through the CGS? 105 services not currently set up to be able to retrieve this information without notes review 201: some centres concerned that this is too frequent and need to differentiate between satisfaction/experience/outcomes. Currently centres use different measures, does this need to be standardised? 202: information in braille not readily available to services- in practice most visually impaired patients prefer electronic format. 301: access to psychology services currently limited and mostly not embedded within CGSs though we would welcome the funding to enable this.		
The service specification summarises the range of provision currently offered by Clinical Genetics services. There is nothing missing from the service specification that we currently provide. However we do not currently provide the full range of activities outlined. There are also some plans for future developments although no time scales are offered for these. Terminology in the document changes between Genetics and Genomics - especially with reference to counsellors. It would be helpful to know if our service is being renamed and should professionals have new titles. The geographical area we are responsible for is only defined by a large scale map; it would be helpful to have a confirmation of the boundaries, do these align with the GLH or our historical services? The two are different. We are unable to deliver this service specification in full without additional resources. Our service has very limited access to clinical psychology (mainly through Oncology services). We have no Nurses or Allied Health Professionals and very limited numbers of Genetic/Genomic	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

counsellors. We currently participate in MDTs and work closely with our GLH but have no new resources to do this - it is included in the new service specification and clearly is appropriate. Delivering this alongside our direct patient care in clinic (referrals are rising) is a conflicting demand. To deliver the future suggestions of polygenic risk interpretation and prescribing additional training of staff is needed. Currently our service does not provide any adult cardiac services 1.5.5 - we are close geographically to the Heart Hospital at Barts- they have in house genetic counsellors that are not part of our service. We do not have resources to deliver adult cardiac services. We do have relationships with these staff and with counsellors employed in other local services (ophthalmology, oncology and paediatric cardiology). The document suggests (1.5.8) we should help these staff with CPD and career progression - we aim to do this but without responsibility for line management and financial resources this is challenging. Funding is stipulated but not by whom. We have recently experienced significant challenges with this model with a genomic counsellor employed by another organisation whose performance was poor and the employing organisation did not have experience of the role to manage appropriately. Historically we had a significant network of peripheral clinics in DGHs and Child Development Centres. Prior to the pandemic we were asked for increasing financial sums to access these facilities, or losing them entirely. Post pandemic we have struggled to reopen a significant proportion as local hospitals are retaining space for their in house services with social distancing. Although we have adopted virtual appointments there are still some families who would benefit from face to face assessment that we are unable to provide accessible services for. The service specification while suggesting an outreach model does not help us gain access to these facilities 1.2.4 . This links in to the need for equity of access to service. The quality indicators that are suggested for reporting are significantly more onerous than those we currently submit. We do not currently have resources to identify this data and report it. We do have an EPR that is well embedded (2 years post implementation) and will be in a better position to report the data requested than many services. We have specific fundamental concerns about 201. We have trialled this PROMS previously and abandoned it after sending to a number of families. We had multiple complaints and reports of distress at some of the questions. Specifically about potential “good” coming from genetic problems in the family and similar. We have used a modified version of Berkenstadt’s 1999 questionnaire instead. We have reservations about 204. Organising and maintaining a group of involved patients for a service such as genetics could be challenging, many individuals have only short term contact with our service. To collate, support and manage a group on annual basis would require resources we do not currently		

have. We have previously trialled a visit from the RCP patient group as an alternative. 301 we do not have the staff make up suggested and without financial resource could not develop this. 305 we are not clear if this specifically refers to an in house clinical genetics MDT (which we have weekly) or other MDTs we take part in. Our in house MDT would not include AHPs or Nurses. Several of the reporting requirements are likely to be managed centrally by our host Trust and our knowledge about reporting ability are limited. Some of the quality indicators are unclear - 309 it is not clear exactly what we are asked to report- is it only our local workforce in the CGS or the workforce we interact with? To reiterate our comment there is an increase in reportable data. We are committed to providing the best service we can and acknowledge and want to report back to Commissioners and NHSE, but the volume of data required is significant and collating at the present time would be a considerable challenge.		
On the whole we think this is a good document, but there are a few points that would benefit from clarification or amendment: Clinical **Genomics** Service and Clinical **Genetics** Service are used interchangeably. We presume they refer to the same thing, but this isn't clear. Patients and other clinicians are more likely to know what is meant by genetics and as Clinical Geneticists we would prefer to keep the name Clinical Genetics Service. Genetic Counsellors are registered as Genetic Counsellors by the Genetic Counsellor Registration Board. More involvement in research and education & training is a good thing and should be included. However, it is worth noting that they are currently not supported in job plans by many Trusts. There would be resource implications for these additional roles. 1.1.1, 1.1.6, 1.6.5, 1.8.1 and 4.4.3. The GMSAs are the network and our understanding is that the CGSs are part of the Alliance (together with the GLH and other providers and stakeholders in their geography). This document suggests the GMSA is a separate entity. 1.8.1 Inequity in access – We agree there should be equity of access. Different waiting times are sometimes due to different staffing levels within the CGSs of the GMSA. Is this suggesting clinicians might be expected to work outside their regional CGS geographical area to even up waiting lists across the GMSA? We would not support this without discussion and agreement between services and commissioners 3.3.3 Polygenic risk scores are not currently part of the Clinical Genetics training curriculum (to our knowledge) and not part of what Clinical Geneticists currently do. This would be a big shift in current	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

practice. There is a very large number of indicators – will all be expected? 101-105 – maybe difficult to get accurate data from current patient management systems and patient records – will need data collection systems and admin support 201 – services should be free to use alternative questionnaires. This one has some questions many clinicians and patients are unhappy about e.g. q6 – '*I can see that good things have come from having this condition in my family*'. There is a SWOB patient satisfaction survey that the national Lead Clinicians group agreed to adopt as the national survey, and many services have. 203 and 204 would be new and a considerable amount of work. Are both and 201 required? Whilst good to do they would have significant resource implications. 301 many CGS don't have access to psychology and this is a separately commissioned service. It would be great to have access, but is this service spec the correct place to state it? Annexe D – good to specify exclusions e.g. most direct to consumer testing, hypermobility, recurrent miscarriage and male infertility. Bullet point 3 could be better defined to avoid conflict between CGS and other providers about whose responsibility it is. Whilst CGS doesn't manage CF patients many do (appropriately) see family members and discuss testing and prenatal options		
This is a very comprehensive document outlining an ideal service. It includes a number of roles or expansion of roles above current workforce availability and will require more funding and expansion of the workforce to deliver. Increased MDT input, advice and guidance to non-genetic specialties, increasing complexity and increasing demand need to be reflected in funding streams, as does the increased need for administration support around the quality indicators. There is inconsistent use of genetic/genomic terminology throughout the document. 'Genomic counsellor' is not a recognised professional title and it is unclear what this is (training, skills, scope of practice, regulation). The internationally recognised professional title is 'Genetic Counsellor', which is a regulated professional group with professional standards training and registration. You can however refer to the practice of genomic counselling and genomic testing. 1.9.1. Unclear whether all services will receive the same prorata funding. 1.1.6 and 1.5.3 Polygenic risk scores and pharmacogenomics have not been part of the Clinical genetics training curriculum and the workforce will require further development to ensure that they can input into the mainstreaming of these.	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

1.5.8 For clarification this should only apply to supporting departments within the NHS, not private sector. 3.2.6. There are differing geographic needs eg supporting consanguineous populations is likely to disproportionately affect some CGSs- how will the NCGS make sure support and funding required for equitable access across regions is available? 102 Is the objective of this to look at the number of positive outcomes from WGS or simply the number where outcome is recorded in the notes? 103-104 As above, is this the recording or the number of positive diagnoses? 201 This outcome is unclear. The GCOS score does not measure patient experience, it is a measure of genetic counselling outcomes. There is good research using the GCOS scale and it would be a good measure of patient outcome. However the indicators need to be more clear about whether other measures of patient experience are expected (satisfaction survey, outpatient standards etc). 301- regarding access to clinical psychology services-we would welcome further support but this will depend on commissioning of psychology services providing capacity 307 It is not clear how the delivery of clinics in these areas reflects the indicator of 'training'. 316 This data is already collected in detail by NIHR, is this required as an indicator separate to that? 319 We would need systems in place to be able to link to deprivation scores		
We are extremely pleased to see this draft proposal and support the direction of travel. We request clarification about how the role of the GMSA GC Lead inter-links with the individual GC Leads within each Regional CGS. We are keen to emphasise our support for the following: 1.2.3 The requirement for a Lead GC for each regional service. Lead GCs not only manage the GC team in each local area, but also contribute to management of the whole CGS, including new service/policy development, teaching, training, and research priorities. 1.5.7 GCs being key to mainstreaming agenda. GCs already work across many clinical specialities, and are increasingly employed in embedded roles in non-Genomics departments, providing GC service in situ.	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

1.5.8 The need for funded CPD/career development for GCs in mainstream roles. The AGNC is already offering specific support to GCs working outside of Clinical Genetics centres, and it is clear that ongoing training / development is increasingly important for GCs working in isolation from their professional colleagues. 1.7.1 Increase in specialist workforce of GCs to support mainstreaming of genomics services. As more genomic testing is offered in mainstream settings, the need for GCs will grow, and we wholeheartedly supports the development of the GC workforce. In particular we would like to highlight the importance of supporting of GC trainees through the professional registration process. Areas for clarification/amendment: 1.1.3 Long term management and follow up of patients/families is briefly alluded to. We would like to stress that this is often a significant component of GC workload which should be recognised. 1.4.5 We note the reference to the research initiative CanGene-CanVar. This may be inequitable to other research groups and charities which are not mentioned in the context of a service specification We are keen to emphasise our support for the following: 2.1.2 / 2.1.7 Recognition of the importance of serving the wider family and not just the index patient. Most non-genomics medical specialities are focussed on the immediate patient, whereas GCs consider the whole family and will guide them through the consequent issues. 2.1.5 / 2.1.7 Recognition of importance of psychosocial aspects of Genomics services and the skills of GCs in providing these services. The psychosocial impact of Genomic testing is well documented and it is vital that NHS services do not overlook the importance of supporting patients appropriately through the complex choices and implications of new genomic tests. Areas for clarification/amendment: 2.1.5 States that a clinic appointment with a GC will be offered where a clinical assessment or examination is not required. We would like to highlight that many GCs do conduct some physical examination in clinic, so we suggest that this is changed to 'where a *detailed* clinical assessment /examination is not required' 201 We are in support of the use of the Genomic Counselling Outcome Scale in measuring patient experience, on a regular basis. 301 / 302		

We support the inclusion of GCs at senior levels in the teams described in the service specification. 307 GC led clinics in sub-speciality areas are integral to offering a optimum service in these areas. 309 We are aware that, as with the Clinical Genetics profession, the GC profession is also likely to experience a large number of retirements of GCs at the top of the profession in the next 5-10 years. For this reason we support a significant investment in development of the GC workforce at all levels. As well as taking in new GCs at the lower ranks of the profession, career development and progression is needed for the current workforce to move them into the senior positions that will be vacated through retirements in the coming years. Finally, we note that the document refers to both Genomic Counsellors and Genetic Counsellors. Whilst we accept a change in our title, we suggest adding to Annexe E "Genomic Counsellors – includes both Genetic Counsellors and Genomic Counsellors". Please also note that the links in Annexe E do not work, including the link to the AGNC documents page. They are typed correctly but the hyperlinks have been added incorrectly.		

The wording of the first sentence: "Clinical genetics/genomics is a clinical specialty that advises on the diagnosis and clinical management of patients and families with genomic conditions" should be revised. Consider: "Clinical genetics is a clinical specialty that undertakes diagnostic investigations on patients and families with genetic conditions, including inherited cancer predisposition, advising on clinical management and applying and interpreting genomic and other diagnostic testing for the screening and management of at risk and affected family members." "Clinical genomics" is not currently a recognised clinical specialty; our trainees are Clinical Genetics trainees. "Genomic conditions" is not a well-defined term. "Inherited cancer" should be "inherited cancer predisposition". **1.1.3** - "For both rare diseases and cancer" should read "for both rare diseases and cancer predisposition", unless this means somatic cancer, in which case we would not usually be interpreting those tests. **1.1.6** – The relationship between "Regional CGS" and "NHS GMS Alliance" should be defined. "Ensuring that further elements of genomic medicine are mainstreamed (i.e. primarily delivered in other clinical specialties)". What elements does this refer to? How should clinical genetics be expected to contribute to pharmacogenomics when this is not part of our training or practice? **1.1.7** – See Annexe B. - What is "culturally competent recruitment"? In a small specialty, it is not realistic to expect to "ensure the service reflects the diversity of the population it serves". - What does it mean to ensure "that ongoing genomic research is representative of the population of England"? It is the validity of the scientific question and method that should be judged. **1.1.10** – Not clear how clinical genetics is expected to contribute to the introduction of pharmacogenomics. Tumour specific genomic testing and interpretation is also mentioned and I am unaware of this being part of clinical genetics training or practice. **1.2.1** – Define "Genomic Associates" **1.5.3 and 3.3.3** Clinical genetics services do not include provision or training for giving risk advice based on polygenic risk scores. These will confer at most a moderately increased risk – annexe D excludes from the service women at moderately increased risk of breast cancer, so this is contradictory. **1.5.8 and 1.6.8** – These sections refer to clinical geneticists and counsellors working primarily outside of the regional clinical genetics services but the service spec is for Clinical Genetics; those working in mainstreamed specialties are outside of the spec. **2.1.5** A clinical appointment with a genomic counsellor invariably requires a clinical assessment. The statement "not requiring a clinical assessment" should be removed	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

2.2.2 – Clarify what is expected by "supporting collaboration with industry". There is no requirement to submit data to registries e.g. NCARDRS. There are too many quality indicators and collecting the data will be unduly onerous. Some of the data are not collected systematically e.g. **105**. Outcomes should be subject to review and revised if they are not be discriminatory. Clinical outcome **102** reads as though the proportion of cases undergoing WGS that need to result in definitive diagnosis or change in clinical management has to be >90% - is this what is meant? **103** and **104** suggest the same thing for foetal and rapid exomes respectively. Point **204** about patient involvement groups sounds difficult to set up in anything other than a biased way, since most populations will be much more diverse than can be adequately represented. **313** There are transition pathways in place. Does this mean that all patients under 16 with a diagnosis seen at any point before 16 years, need to be offered a follow up appointment, even if it is many years since their previous appointment? Will resource be put in place to facilitate this major change in practice? **315** stipulates specific audits that should be carried out, including quality of letters and compliance with recording consent/record of discussion. It is unclear what the quality standard is that this should be measured against. **317** says clinical genetics will have a policy for storage and usage of genomic data. Does this mean we will be able to store and access all our patients' genomic data?		
What is the funding mechanism for GMS'? A tariffed service, based on patients seen in outpatient clinics, will fail to deliver on the ambitious proposals on multidisciplinary working & mainstreaming, in the short to medium term. 102, 103, 104. As an example 102; 'proportion of cases undergoing WGS for a rare disease where outcome is recorded resulting in a definitive diagnosis or change in Clinical management'. Numerator; 'No of patients with outcome recorded in patient record'. Denominator; 'Number of cases undergoing WGS for rare disease indication'. Threshold; ' >90% during first year of clinical diagnosis'. Realistically the % of cases resulting in a definitive diagnosis or change in Clinical management is at most 20-30%. The % would be similar for 103 & 104. I can see why they are linked to domain 4, the assumption being that having a diagnosis for instance is	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

a positive experience, but don't think these are useful quality indicators as different GMC's will be using the same testing criteria from the test directory & therefore would expect broadly similar results, so I can't see how they could differentiate between a well & poorly performing service. '317. The CGS has an operational policy for storage & usage of genomic data.' What is meant by genomic data in this case? Does this relate specifically to the data files which will be held by the GLH or to the reports (paper or electronic) relayed to the GMS? If the former, the GMS would not need an SOP		
Overall, this is a good service specification that captures the roles and responsibilities of the current clinical services and I would support it. Clearer definitions around roles and responsibilities of regional CGS/GMSA/NCGS and how they interact would be helpful. Governance – it feels uncomfortable that a clinical service is not accountable to a specialised or higher specialised commissioning group rather than being directly accountable to NHSE/I. Is there a reason for this? Who will administer and fund the networks between regional CGS/GMSA/GLHs Re 1.6.4 – as this is a clinical service should it be inspected by CQC rather than NHSE/I 1.7 Workforce development – there is a statement the workforce is likely to increase. This specification also increases the CGS workload particularly WGS and rapid exomes which require a significantly greater amount of time and more appointments than standard care. Has this been accounted for? The quality indicators are sensible. Collecting the data may prove more of a challenge and help / recommendations of IT systems to capture this would be useful Re 102/103/104 – there is a significant difference between recording an outcome (e.g rapid exome did not turn up a cause) and where one expect 100% and where a diagnosis and or management was changed (< 100%) either these need separated to provide useful data or one chosen to be recorded (or further definition on exact measure) 308 is there a validated survey NHSE/I recommend to capture to determine the value – if not this is not doable. In terms of equity, do other clinical services seek this feedback?	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase
Clinical geneticists are trained in Diagnostic Medicine – please add to role description.	Acknowledged	Workforce and funding to be considered as part of the

There need to be commissioned systems in place that recognise that some patients have a particularly high level of need for genomic analysis and may be ill-served by standard clinical evaluation by non-experts in genomics combined with data analysis by standard filtering pipelines. Systems need to be funded to allow for expert clinical review by clinicians with genomics expertise to undertake clinically driven review of such patients and their genomic data in order to deliver a high quality genomic medicine service to patients with high levels of need eg. patients with ultra-rare disorders or those with hallmarks of a genetic cause for their problems but no diagnosis from routine testing. Whilst the focus here is on Rare Disease it is important to consider input to patients with a cancer diagnosis. Clinical genetics need funded time to attend GTABs to advise on unexpected germline mutations discovered on somatic cancer testing. The amount and type of data return required will place a heavy long-term burden on the submitting services that will require resourcing. We also emphasise that the quality of a result is dependent on sample quality and timeliness of testing and Pathology is key in delivering this but requires resource to achieve this across the service		Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase
This is a tremendous achievement to take the Service Spec so far forward, thank you. 1.1.3 - the definition of rare that is used in the UK is fewer than 1 in 2,000 people affected. There are a handful of genetic conditions that have prevalences very close to this line or over it. An example would be 22q11 deletion which is currently estimated to be rare, but often described as underdiagnosed. What would be the arrangements for conditions that are the rarest of the common? 1.1.9 - the UK Rare Diseases Framework Priority 1 of 4 is for for rare disease patients across the UK to get a final diagnosis faster and for research into previously unrecognised conditions to identify new rare diseases and provide new diagnoses. Priority 3 of 4 commits to a vision for rare disease patients to experience better coordination of care throughout the patient journey. This policy warrants mention here too. 1.5.8 and annex 6 with respect to qualification and registration of genetic counsellors is strongly supported by Genetic Alliance UK While we support all the positive changes in this section, we would like to raise two further issues: - diagnoses of newly identified conditions through genome sequencing - there is unlikely to be a care pathway or non-genomic clinicians who are aware of these conditions in detail - what is the process for identifying appropriate new care pathways in this context - care pathway for non-diagnosed individuals - what is the process for those who do not achieve a diagnosis through the most comprehensive genome sequencing? Is there are pathway to appropriate specialities to make diagnoses of non-genetic rare conditions?	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

These are probably necessary to take account of the above two routes of referral out of CGS?		
Use of Genomic Counsellors with no description until the annexe reads odd. Most of the workforce still practices under Genetic Counsellor - Why would this not be the term with the annexe that Genomic and Genetic counsellors in this group? The service spec should illustrate this clearly at the start or use genetic/genomic counsellors. It is those who practice as genetic counsellors at this stage who will deliver the service. 1.5.3 – Inequality of ascertainment using data and polygenic interpretation is documented. You might need to specify the benefit of 'which' patients. Under what criteria can these tools be used? Most patients? Representative of the patient population? etc. 1.6.7 for the other specifications we use language like co-ordinate, facilitate we talk about the approach etc – suggesting engagement without any direction on how engagement should be approached or delivered seems short sighted and can read tokenistic. 1.7.1 The specialist workforce will need to increase, I don't think this should be a foot note but a specification within itself. The specialist workforce will need to expand, train and develop networks to support the growing need for genomic testing via non-specialist clinicians 1.72. I think you could specify that additional staff will/should be recruited if the services required in a particular region specifically need it in their respective populations 1.7.4. This could be more comprehensive - support how? 2.1 Care pathway referral into specialist service is a key point to address equity to service - there needs to be a point in here about involvement with referrers, collection / monitoring of data to ensure that referral into the service is equitable - development for particular populations may be needed when referral into genetics is not representative of the population it serves 2.1.5 Do we mean 'when a clinical examination is not initially indicated or needed for the management of a patient clinical appointments can usually be offered by the GC.' In addition There are cases where some aspects of clinical examination particularly in cardiac and cancer cases will be undertaken by a genetic counsellor. The need for clinical examination may become apparent after a GC appt or form part of a predictive /diagnostic genomic testing and to ensure patients have full informed consent. 3.3.3 Again with new ways of stratifying healthcare and medicine the bias embedded in that ascertainment may need to be monitored so that measures can be imbedded to ensure equity	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase
We are broadly supportive of the service specification.	Acknowledged	Workforce and funding to be considered as part of the

In addition to the specific concerns, as written, the document includes provision for activity which will likely significantly increase in coming years, such as genomic medicine MDT's, training and education of primary, secondary and tertiary colleagues, working with colleagues in primary and secondary care to establish pathways for end to end care, variant interpretation, patient engagement activities etc. While we recognise this is appropriate and services need to evolve to provide this; currently to deliver this service specification there needs to be significant investment into genetic services. Funding will need to be agreed to meet the specification. For example, activity specified which is not currently funded: **1.4.1**, **1.4.2** Activities currently performed where activity will significantly increase and will need funding: **1.4.3**, **1.1.6**, **1,1,9**, **1,1,10**, **1.5.1**, **1.5.2**, **1.6.1** The NHSE/I genomic unit does not directly commission the clinical service but has asserted authority for accountability. This accountability naturally lies with the host trust and specialist commissioning teams. Peer to peer review by networked regional services and specialist commissioners is appropriate for a clinical service. **1.1.4** We provide specialist MDT clinics offering on-going management for a number genetic disorders. The service specification should be altered to allow for provision of these services within Genetics, eg for clinics such as Marfan or VHL, where the primary diagnostic and management skills are traditionally within clinical genetics or a genetics led MDC/MDT, rather than implying that these will now be delivered by other services such as cardiology or endocrinology. **2.1.11**, we agree that this is appropriate, but given the significant increase in genomic testing anticipated; this has the potential to significantly impact on consultant workload, and will need to be funded. Clinical Indicators 101,102,103,104,104 will have funding/resource implications. Genomic MDT working will need to be properly resourced if to function well. 201 and 203. If to be meaningful they will need to be appropriately funded. 204 – Establishing and maintaining a patient involvement group will take time and resources. Establishing the group to accurately reflect the regional population will require a great deal of input and will need appropriate funding. 308 To be meaningful any user survey will need to be well thought through and if mandated every 2 years will require time to organise and will need to be appropriately funded.		Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

317 'genomic data' needs to be more clearly specified - is this raw sequence data or clinical laboratory reports. If we have clinical records integrated into the EPR system these will contain laboratory reports. This item needs greater clarity. 319 We do not currently have a system in place to capture health inequalities - this would need to be address and funded **Annex A** is an inaccurate map and needs amending to reflect that East Berkshire is served by the OUH Clinical Genomics service (within the C+S GLH and GMSA geography, Section 1.1.1) **Annexe B**- we do not have systems in place to monitor deprivation scores. We do not have the tools or resources to capture this data.		
Most of the content of the documents relates to the breadth of the clinical genetics service and consequently there is very little detail with regard to cancer services. It seems to exclude tumour genomics so not relevant to our group unless dealing with rare germline syndromes that include lung cancer. There are no KPI's relating cancer genomic testing. We have spent a great deal of time discussing turn around times amongst other issues and have heard from numerous colleagues about their concerns about the impact of transition to the GLHs. Is a more detailed service specification relating to cancer is being developed? PPI comment: My only comments are that the patient involvement sections seemed a little lightweight - patient experience to be captured every 2 years and shared with the relevant governance group (P13 201) who would determine who this would be? is this governance group in the genomics service, a Trust or some other body?	Acknowledged	Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase
1.1 The specification states: '*The provision of these opportunities to all individuals on the basis of clinical need, in an equitable manner is a key aim of Clinical Genomics*'. Whilst we welcome this ambition, it is not yet being realised in practice. The GMS is currently only available for cancer patients after they have exhausted all licensed treatment options. Moving forward, Roche would like to see genomic services available to all eligible cancer patients upfront, to direct treatment options. **1.1.8** The specification states: '*Continued delivery of research will ensure the adoption of new advances in genomics where there is evidence of patient benefit*.' We believe that this needs to be beyond just WGS; a comprehensive genomic environment would help to improve identification of patients for clinical trials, which would in turn make the UK more	Acknowledged	Workforce and funding to be considered as part of the Integrated Impact Assessment Governance and operational issues raised to be clarified via Standard Operation Procedures developed in Commissioning Implementation Phase

attractive for investment in research. Currently the Test Directory is limited in its capability and does not cover all the genes required to facilitate adoption in research.

1.1.9
A key part of '*education and training in Genomic Medicine*' must be tailored training for healthcare professionals in interpreting genomic reports.

1.1.10
In relation to the third bullet point – we must get to a place where genomic development is ahead of treatment approvals in a pathway, rather than the other way around.
In relation to the seventh bullet point, the Test Directory is not currently fit for purpose in facilitating participation in research and the WGS turnaround times are too slow.

1.2
Regarding the final sentence, we must ensure that IT infrastructure is future proofed and is set up to harmonize with clinical data infrastructure in hospitals.

1.5.2
Will the genomic pathways mentioned in the first sentence be publicly released?

1.5.4
We would be interested to understand how this ambition will be carried out in primary care, and how the effectiveness/success will be measured. Moreover, we hope that education provision for patients will be considered as part of this.

1.8
We welcome the inclusion of clinical research in the specification, but question the NCGS's ability to deliver on these commitments, given that new genomic alterations are not included in the Test Directory, meaning it isn't possible to gain funding (or to establish routine testing) for new alterations and therefore suitable studies cannot be identified. We are aware of the current negative impact that this is having on clinical research in the UK.

1.8.4
As above, the Test Directory would need to expand in order to fulfil this ambition, particularly for cancer patients.

1.8.5
We would be interested in understanding more about the mechanism for engagement in this.

2.1

Roche had no comment on this specific section. **2.2.2** In relation to the second bullet point – the current system is not fit for purpose for trial recruitment, as per previous comments Whilst we welcome the inclusion of ‘*The CGS will have an operational policy for storage and usage of genomic data*’, we must bear in mind the importance to utilisation of genomic services, development of pathways, assessment of technologies and research. We would welcome more detail on the place of data collection, plans for standardisation and consistency of platforms for data collection and storage in genomics. This data should be harmonised across all GLHs and the data should be collected in a manner that can be federated with future clinical data platforms.		