| Integrated Impact Assessment Report for Service Specifications | | | |
|---|---|---|---|
| **Service Specification Reference Number** | E01/S/a | | |
| **Service Specification Title** | Clinical Genomics (Adults and Children)<br>Proposal **for routine commission** (source A3.1) | | |
| **Lead Commissioner** | Anita Beer, Genomics Unit, NHS England | **Clinical Lead** | Prof. Bill Newman, Chair: Genomics Clinical Reference Group |
| **Finance Lead** | Hud Manuel, Head of Finance and Information, Genomics Unit | **Analytical Lead** | Claire Strawbridge Contracts Manager, Genomics Unit |

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| About this Impact Assessment: instructions for completion and explanatory notes |
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| • Each section is divided into themes.<br>• Each theme sets out a number of questions.<br>• All questions are answered by selecting a drop down option or including free text.<br>• Free text boxes are provided to enable succinct relevant commentary to be added which explains the rationale for response or assumption. Please limit responses to 3 sentences of explanatory text.<br>• Data in this document is either drawn from one of the relevant service specification documents or a source for the information is provided.<br>• Where assumptions are included where data is not available, this is specified. |

| Section A - Activity Impact | |
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| **A1 Current Patient Population & Demography / Growth** | |
| A1.1 Prevalence of the disease/condition. | Rare diseases are understood to impact on the lives of approximately 6-8% of the population, although it is acknowledged this may be an under representation as we gain in knowledge. It is estimated that at least 80% of rare diseases are genetic, with many of them due to a single genetic or chromosomal change[1]. A total of 305,683 individuals were diagnosed with cancer in 2017, with approximately 5-10% arising in individuals with a genetic predisposition and a significantly increased risk of developing certain tumour types[2].<br>*Source: Service Specification Proposition section 5.1* |
| A1.2 Number of patients currently eligible for the service according to the proposed service specification commissioning criteria. | Up to circa. 128.5k in 2022/23<br>*Source: Local calculation*<br>Please specify<br>**Index Cases:** Percentage incidence of rare diseases and cancer with genetic predisposition included within Service Specification (80% of 6-8% population annual incidence of Rare Disease; 5-10% of cancer annual incidence) compared to Office of National Statistic population forecasts.<br>**Cascade Referrals:** Assumed an average of **one** cascade referral (family member at increased risk of condition with genetic pre-disposition) per Index Case.<br>**Negative Diagnoses:** Assumed 50% of patients that meet the eligibility criteria go on to receive a negative diagnosis.<br><br>Calculations consider the impact of embedding genomics into mainstream clinical pathways and routine care ('mainstreaming'), which is a primary aim of the revised service specification. The revised service specification also aspires to increase equity of access for patients across England and provide the infrastructure required to embrace the impact of the identification of novel conditions |

[1] https://www.raredisease.org.uk/
[2] https://www.ons.gov.uk/peoplepopulationandcommunity/healthandsocialcare/conditionsanddiseases/bulletins/cancerregistrationstatisticsengland/final2016#cancer-incidence-over-the-last-decade

<table>
<tr><td></td><td>and implementation of new testing technology, all of which is expected to increase the eligible population.<br><br>The methodology for calculation does not include weighting of population based on geographical variations in incidence or population demographics. Further work is required to standardise the population based on additional risk factors. In addition, the estimated eligible population includes annual incidence only and does not consider any backlog of undiagnosed patients.</td></tr>
<tr><td>A1.3 Age group for which the service is proposed according to the service specification commissioning criteria.</td><td><u>All ages</u><br>Please specify<br>Eligible population calculations not adjusted for age demographics. There may be disparities in eligible population by age range, as examples 75% of rare diseases affect children; cancer incidence increases significantly in patients over 55 years.</td></tr>
<tr><td>A1.4 Age distribution of the patient population eligible according to the proposed service specification commissioning criteria</td><td>N/A<br>Source: N/A<br>Please specify<br>Further work is required to calculate the age distribution of the eligible population and there are complexities to weighting the demographic profile of England against prevalence and incidence by age for both cancer and rare disease.</td></tr>
<tr><td>A1.5 How is the population currently distributed geographically?</td><td><u>Unevenly</u><br>If unevenly, estimate regional distribution by %:<br><br>NHS England Regional Distribution
<table>
<tr><td>North West</td><td>12%</td><td>South West</td><td>10%</td></tr>
<tr><td>North East & Yorkshire</td><td>12%</td><td>Midlands</td><td>23%</td></tr>
</table></td></tr>
</table>

| | | | | |
|---|---|---|---|---|
| London | 16% | East of England | 8% |
| South East | 16% | | |

**Genomic Laboratory Hub Regional Distribution**

| | | | |
|---|---|---|---|
| North West | 16% | South West | 15% |
| North East & Yorkshire | 19% | Central and South | 14% |
| North Thames | 13% | East of England | 8% |
| South East | 15% | | |

*Source: Service specification proposition section 7.1*

Please specify

The percentages for NHS England Regional Distribution are calculated based on the overall population distribution reported by the Office of National Statistics. However, these may change based on the geographical variations in incidence and risk factors.

We have also included the NHS Genomic Laboratory Hub (NHS GLH) Regional Distribution as the boundaries do not align with those for the NHS England Regions, and the NHS Clinical Genomic Services (NHS CGSs) are expected to align to individual NHS GLHs as part of the overall NHS Genomic Medicine Service (NHS GMS) Structure.

**A2 Future Patient Population & Demography**

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| A2.1 Projected changes in the disease/condition epidemiology, such as incidence or prevalence (prior to | **Increasing** |

<table>
<tr><td>applying the new service specification) in 2, 5, and 10 years?</td><td>It is forecasted that incidence will increase due to the identification of novel genetic conditions and as genomic testing technologies improve and increase. We would also expect the eligible population to increase as genomics and genomic testing becomes further embedded as routine practice across the NHS through implementation of the NHS Long Term Plan, mainstreaming and increased awareness through education.<br>Source: Service specification proposition section 5.1</td></tr>
<tr><td>A2.2 Are there likely to be changes in demography of the patient population and would this impact on activity/outcomes?</td><td><u>Not known</u><br>Please specify<br>Providers of NHS CGSs are required, by way of the revised service specification, to ensure that access to the service is equitable across their entire catchment area. As part of this, providers will work in partnership with their NHS GMS Alliance to reduce barriers to access and improve engagement with communities that have historically not been reached.<br><br>To achieve this, providers will work to understand the needs of their local population and work with their NHS GMS Alliance to address identified unmet need. This will be supported by the development of consistent eligibility criteria during the commissioning implementation of this service specification.<br><br>Whilst the demography of patients is not expected to change, reducing barriers to access, and improving equity of access, is expected to increase demand for some services.<br>Source: Service specification proposition section 6/other</td></tr>
<tr><td>A2.3 Expected net increase or decrease in the number of patients who will be eligible for the service, according to the proposed service specification commissioning criteria, per year in years 2-5 and 10?</td><td><table><tr><td>YR2 +</td><td>3,495</td></tr><tr><td>YR3 +</td><td>4,660</td></tr><tr><td>YR4 +</td><td>5,825</td></tr><tr><td>YR5 +</td><td>6,989</td></tr><tr><td>YR10 +</td><td>11,649</td></tr></table>Source: Service specification proposition section 5.1 and commissioner forecast</td></tr>
</table>

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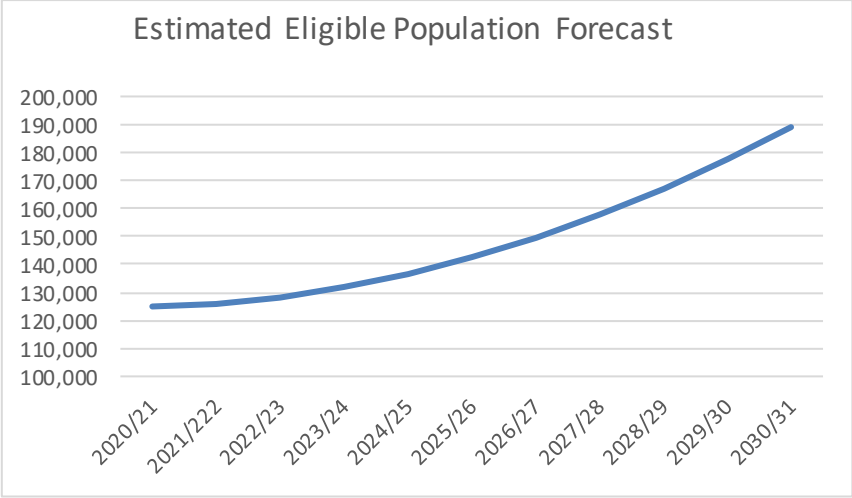
|---|---|
| Are these numbers in line with ONS growth assumptions for the age specific population? If not please justify the growth assumptions made. | **No**<br>The increase in eligible population considers the impact of mainstreaming clinical genomics, equity of access plus the impact of identification of novel conditions and implementation of new testing technologies. The increase in eligible population is expected to be phased over the coming 10 years in response to the transformation work undertaken through the NHS GMS with access to services for a total estimated eligible population of circa. 189k people by 2030/31.<br>  <br> |
| **A3 Activity** | |
| A3.1 What is the purpose of new service specification? | **Revision to an existing published service specification**<br>*PSSAG (Prescribed Specialised Services Advisory Group)<br>Please specify<br>The existing NHS CGS specification was published in 2013 and included both clinical genetics and genomic testing services. Since its publication, the NHS GMS has been established, leading to the separation of the clinical genomic and laboratory functions through the formation of NHS GLHs. It is |

<table>
<tr>
<td></td>
<td>important that the revised specification reflects the infrastructure changes brought about by the formation of the NHS GMS. It also needs to reflect developments within the NHS GMS, including:<br>1) The delivery of NHS CGSs as a network, working in tandem with the NHS GLHs supported by the NHS GMS Alliances.<br>2) Highlighting new ways of working through virtual (online and telephone) and face to face clinics, multi-disciplinary teams (MDTs), genomic test advisory boards (GTABs), and multidisciplinary clinics (MDCs).<br>3) Recognition of the roles of all the health care professional groups within clinical genomics to ensure optimal use of skills.<br>4) Adaptation of the workforce to reflect the rapid advances in genomic testing technology, increased provision of testing including rapid exome sequencing, specialist genetic testing, increased range of cancer genomic testing, and whole genome sequencing.<br>5) To reinforce the need to ensure equity of access and provision of genomic testing to all groups based on clinical need; and<br>6) The increased role of mainstream medicine in the delivery of genomics and how NHS CGSs will support colleagues to facilitate this provision for patient benefit.</td>
</tr>
<tr>
<td>A3.2 What is the annual activity associated with the existing pathway for the eligible population?</td>
<td>140,482 (including provider reported new and follow-up activity)<br>Source: NHS CGS Data Collection - June 2021<br>Please specify<br>Given the reduced demand for services experienced during the COVID-19 pandemic during 2020/21, we anticipate that the activity undertaken in 2019/20 is the most reliable indication of annual activity associated with the existing pathways. However, this activity may be understated due to the variation in completeness of activity reported during the data collection, reportedly caused by the differing methods of recording activity between providers.
<table>
<tr><td></td><td>2018/19</td><td>2019/20</td><td>2020/21</td></tr>
<tr><td>Total New Appointments</td><td>97,540</td><td>96,925</td><td>83,972</td></tr>
<tr><td>Total Follow-Up Appointments</td><td>38,918</td><td>43,557</td><td>39,295</td></tr>
<tr><td>Total Activity</td><td>136,458</td><td>140,482</td><td>123,266</td></tr>
</table>
</td>
</tr>
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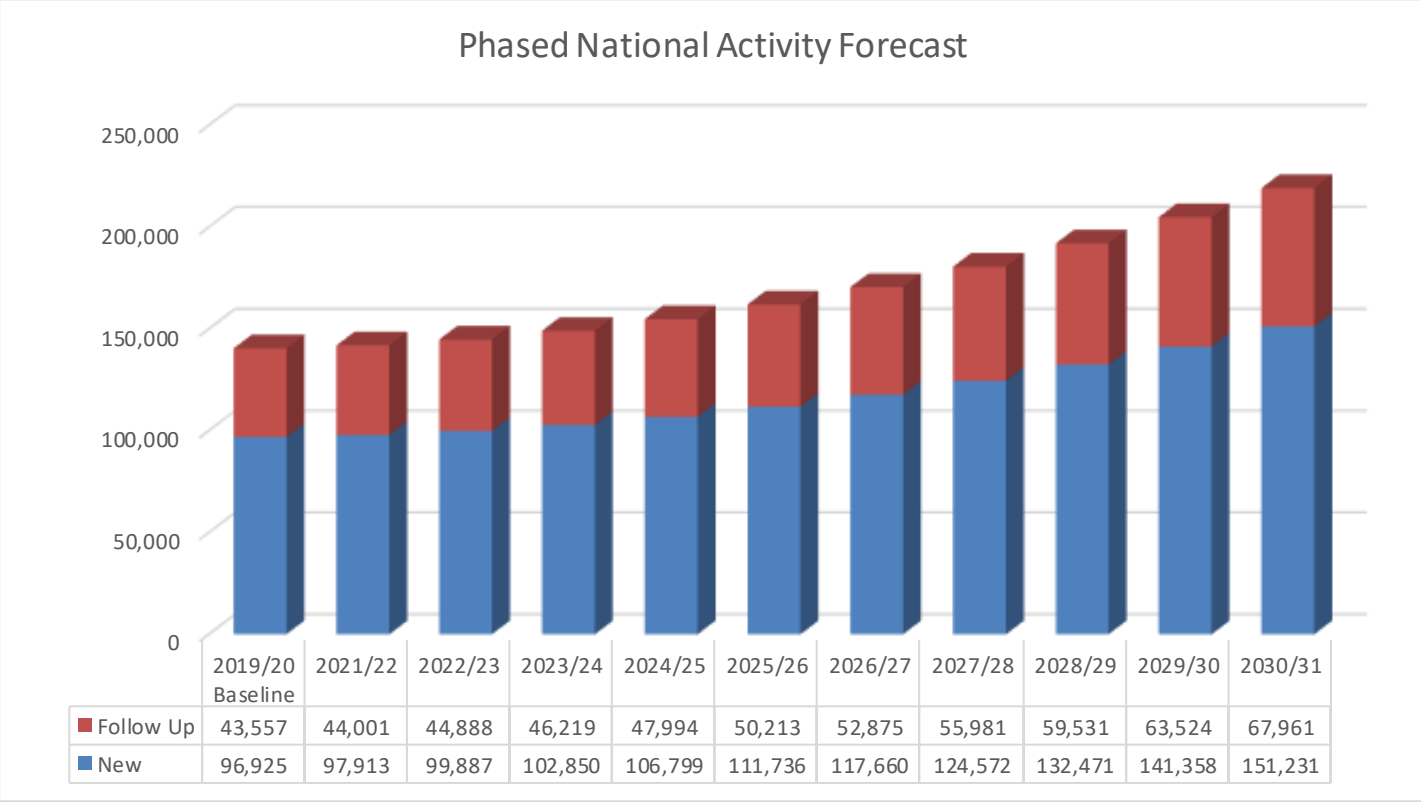
| A3.3 What is the estimated annual activity associated with the proposed service specification proposition pathway for the eligible population? | Phased increase to circa. 219k by 2030/31<br>*Source: NHS CGS Data Collection – June 21 and Local calculations*<br>Please specify<br>The annual activity forecast assumes that the new patient activity reflects 80% of the total eligible population forecast detailed in Section A2.3 by 2020/31. We would not expect 100% of the eligible population to access the service. It also assumes that the new to follow up ratio remains as per the 2019/20 baseline year at circa. 2:1, which is in line with national guidance (Royal College of Physicians Clinical Genetics: Workforce and Job Planning (2017).<br>The Phased National Activity Forecast assumes that demand and activity will increase from the 2019/20 baseline outturn activity to a level that provides access for 80% of the estimated national eligible population by 2030/31.<br>  <br> |

| | 2019/20 Baseline | 2021/22 | 2022/23 | 2023/24 | 2024/25 | 2025/26 | 2026/27 | 2027/28 | 2028/29 | 2029/30 | 2030/31 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| Follow Up | 43,557 | 44,001 | 44,888 | 46,219 | 47,994 | 50,213 | 52,875 | 55,981 | 59,531 | 63,524 | 67,961 |
| New | 96,925 | 97,913 | 99,887 | 102,850 | 106,799 | 111,736 | 117,660 | 124,572 | 132,471 | 141,358 | 151,231 |

| **A4 Patient Pathway** | |
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| A4.1 **Patient pathway**<br>Describe the current patient pathway and service. | Referrals for patients and their family where there is a confirmed or suspected genetic condition are currently received from GPs or clinicians in secondary care. Services are provided through a Hub and Spoke Model, with a single Provider responsible for delivering the NHS CGS from outpatient facilities within the host trust and via outreach within district general hospitals or other suitable settings across the geographical catchment area of the service. Within the clinic, the patient and/or their family will be assessed by the most appropriate clinician(s) to identify the risk of inheriting or developing a genetic condition. Where required, the NHS CGS clinicians will facilitate the collection of tissue/samples for clinical diagnostic testing. Genomic diagnostic tests are performed by the NHS GLHs in line with the National Genomic Test Directory. Follow-up appointments are provided to discuss the results of diagnostic tests. There are also occasions when patients and/or family members are re-referred to the service if new clinical information or risk factors are identified.<br>The NHS CGSs are not restricted to the facilitation of diagnosis, rather they also provide long term management of individuals, particularly those with multi-system disease that require surveillance co-ordinating by the service and counselling services for patients and families. Longer term support may also be required for those patients with a diagnosed mutation that are considering risk reducing surgery later in life. There are also patients that require annual screening programmes, which are again facilitated by the NHS CGSs. Therefore, whilst some patients may require short term care only to facilitate testing and then receive their diagnosis (either positive or negative), there are some that remain under the care of the service for some years.<br>*Source: Service specification proposition section 7.2* |
| A4.2. What are the current service access and stopping criteria? | Current access and exclusion criteria differ between NHS CGSs. A review of current criteria indicates that access to the service is available across all services for:<br>• Patients that live within the NHS CGS catchment population.<br>• General Genetics, such as those patients with or at risk of having a genetic condition themselves or in their family that is not part of the exclusion criteria.<br>• High risk cancers, according to agreed criteria.<br>Significant variation occurs within the exclusion criteria. Five of the thirteen NHS CGSs that provided detail of their criteria exclude self-referrals despite them being described as a potential |

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| | referral source within the currently published service specification. Another area of significant variation is Alpha-1 antitrypsin deficiency, with seven out of thirteen providers explicitly excluding it but others considering it part of the eligible population. There are other examples of disparities in acceptance and exclusion criteria across NHS CGS subspecialties, such as (but not restricted to):<br>• Autism/developmental delay<br>• Familial Hypercholesterolaemia<br>• Hypermobility syndromes, including Ehlers Danlos Syndrome<br>• Cystic Fibrosis Screening<br><br>The differences at a subspecialty level are likely to be due to the ways in which local services have developed such as availability of relevant clinical expertise locally.<br><br>It is expected that the new service specification will lead to a reduction in the number of patients under the care of clinical genetic services for the facilitation of testing only as these will typically be managed within mainstream services once the necessary education and training has been provided to staff within those services. However, the increase in testing within mainstream services is expected to increase the number of patients diagnosed with a rare disease or cancer/risk of cancer with a genetic predisposition which will lead to a higher number of complex patients referred to clinical genomic services that require longer term management and increased clinical input.<br><br>**NOTE:** There is scope to develop consistent and agreed inclusion and exclusion criteria as part of the Commissioning Implementation Plan for the revised Service Specification to ensure equity of access across the population.<br>*Source: NHS CGS Data Collection – June 21 and Service specification proposition section 5.1* |
| A4.3 What percentage of the total eligible population are:<br>a) Referred<br>b) Meet any existing criteria for care<br>c) Considered to meet any existing exclusion criteria | If not known, please specify N/A<br>a) 60%<br>b) 46%<br>c) 10%<br>*Source: NHS CGS Data Collection – June 21* |

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| A4.4 What percentage of the total eligible population is expected to:<br>a) Be referred to the proposed service<br>b) Be eligible for care according to the proposed criteria for the service<br>c) Take up care according to the proposed criteria for the service<br>d) Continue care according to the proposed criteria for the service? | If not known, please specify<br>We have assumed that the NHS CGS increases access to meet 80% of the estimated eligible population. Consistent eligibility criteria will be developed as part of the Commissioning Implementation Phase of specification roll-out. Mainstreaming is expected to increase the proportion of genomic testing that is requested directly through mainstream services, which will reduce the proportion of diagnostic cases referred to the NHS CGS over time, particularly for those patients that go on to receive a negative result. However, given the expected increase in the number of diagnoses made in mainstream services, there is likely to be an increase in the proportion of cases referred to NHS CGSs that meet the eligibility criteria, an increase in the complexity of those patients and the length of time that they remain on the caseload.<br>a) Not known<br>b) 80%<br>c) Not known<br>d) Not known<br>*Source:* Local calculations |
| A4.5 Specify the nature and duration of the proposed new service or intervention. | **Time limited**<br>For time limited services, specify frequency and/or duration.<br>Patients may receive one off diagnostic care that leads to a negative result and no further intervention. Alternatively, patients may require time limited care to receive required counselling along with lifestyle and condition management advice. Lifelong care may be required, with patients re-referred to the services some years after their initial diagnosis as further intervention is required such as advice in relation to risk management surgery.<br>*Source: Commissioner developed text* |
| **A5 Service Setting** | |

A5.1 How is this service delivered to the patient?

*Select all that apply:*

| | |
|---|---|
| Emergency/Urgent care attendance | ☐ |
| Acute Trust: inpatient | ☒ |
| Acute Trust: day patient | ☐ |
| Acute Trust: outpatient | ☒ |
| Mental Health provider: inpatient | ☐ |
| Mental Health provider: outpatient | ☐ |
| Community setting | ☐ |
| Homecare | ☐ |
| Other | ☐ |

Please specify:

Services have historically been provided to patients through a hub and spoke model, with services provided from outpatient facilities across the geographical catchment area of each NHS CGS However, the COVID-19 pandemic has led to a reduction in the number of sites from which services are provided with the cessation of most satellite services. Typically, pre-COVID in 2018/19, most patients were seen face to face by the NHS CGSs, with an average of 27% of consultations undertaken non-face to face. However, during 2020/21, the average percentage of patients receiving non-face to face care increased to a mean of 55%. NHS CGS clinicians may undertake joint consultations with other specialties to improve clinical pathways and reduce the burden on patients. Clinical care including advice, diagnosis and clinical management may also be provided without direct patient care if clinically appropriate; for example, clinicians may review genomic and phenotype data and advise the managing clinician directly of as art of a multi-disciplinary meeting. Patients may also be managed across geographical boundaries if an NHS CGS has specific expertise in a particular specialist area of genomics that is not available within the local NHS CGS.

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| A5.2 What is the current number of contracted providers for the eligible population by region? | <table><tr><td>North West</td><td>2</td></tr><tr><td>North East & Yorkshire</td><td>3</td></tr><tr><td>Midlands</td><td>3</td></tr><tr><td>East of England</td><td>1</td></tr><tr><td>London</td><td>4</td></tr><tr><td>South East</td><td>2</td></tr><tr><td>South West</td><td>2</td></tr></table> |
| A5.3 Does the proposition require a change of delivery setting or capacity requirements? | **yes**<br>Please specify:<br><br>**Delivery Setting**<br><br>A full analysis of the geographical locations of all satellite locations for NHS CGSs has not been possible as the information was not supplied by all providers as part of the data collection exercise. Ten of the seventeen providers supplied details of the satellite services that they provide. The information provided indicates that the number of sites per 100,000 catchment population varies from 0.1 to 0.44, with a mean average of 0.29 and a median of 0.31.<br><br>This indicates that there may be scope to increase the number of satellite services delivered by some providers. However, further work would be required to establish the geographical area that services cover balanced with the impact of increased non-face to face consultations to patient experience and equity of access. In addition, it will be important to consider the reduced availability of satellite locations due to the impact of the Covid-19 pandemic.<br><br>**Capacity**<br><br>Using national workforce planning guidance for both Clinical Geneticists (Clinical Genetics Society Rationale for a job planning document for Consultants in Clinical/Medical Genetics (2020)) and Genetic Counsellors (Association of Genetic Nurses and Counsellors Career Structure for Genetic |

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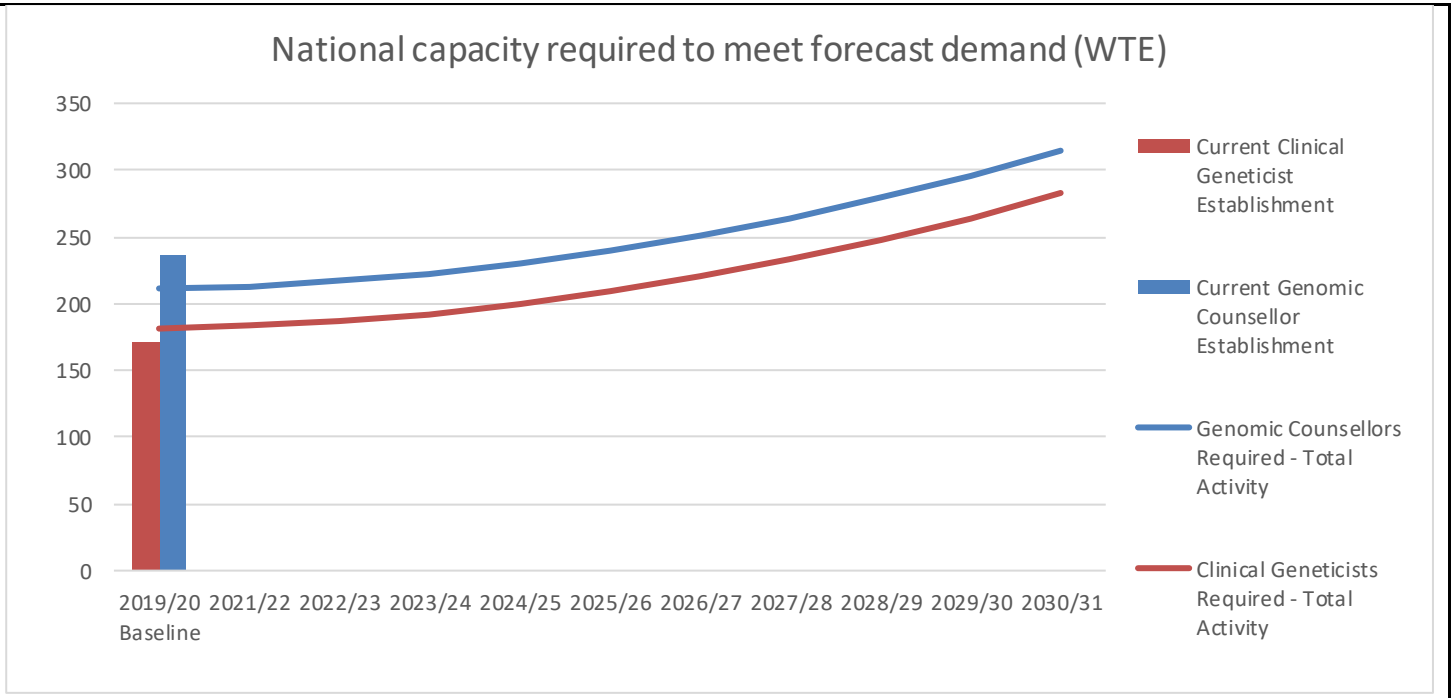
| | Counsellors and Support Roles (January 2021)), the capacity required to meet the 2019/2020 baseline activity has been calculated.<br><br>The capacity required to undertake the 2019/20 baseline activity was compared to the actual establishment across England taken from the NHS CGS data submissions (June 21) and highlights a minimum indicative staffing deficit of approximately 10 clinical geneticists. This problem is compounded by a vacancy rate of over 16% in June 2021 and an expectation that approximately 30 clinical geneticists are likely to retire during the coming 5 years.<br><br>Whilst the overall number of Genetic Counsellors in establishments across England initially appears sufficient to manage baseline activity it's important to note that this is adversely impacted by a vacancy rate of 9.5% at the time of the data submissions. In addition, the Genetic Counsellor establishment at some organisations compares poorly with the number of staff needed to manage demand whilst others compare much better.<br><br>Based on the calculations, the capacity required to meet forecast demand across the coming 10 years is expected to increase assuming there are no changes in first to follow up ratio and the split of activity by clinical geneticist (Consultant Led) and genomic counsellor (Non-Consultant Led) remains the same. |




**NOTE:** Due to the variability in findings across NHS CGSs from the data collection exercise undertaken in June 2021, this analysis is only indicative and requires robust validation and refinement. Rather, it highlights the need for robust activity and finance monitoring frameworks to be developed and introduced to inform detailed activity and demand, workforce and financial planning.

*Source: NHS CGS Data Collection – June 21 and Local Calculations*

**A6 Coding**

A6.1 Specify the datasets used to record the new patient pathway activity.

*Select all that apply:*

| | |
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| Aggregate Contract Monitoring * | ☐ |

*expected to be populated for all commissioned activity

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| Patient level contract monitoring | ☒ |
| Patient level drugs dataset | ☐ |
| Patient level devices dataset | ☐ |
| Devices supply chain reconciliation dataset | ☐ |
| Secondary Usage Service (SUS+) | ☒ |
| Mental Health Services DataSet (MHSDS) | ☐ |
| National Return** | ☒ |
| Clinical Database** | ☐ |
| Other** | ☐ |

**If National Return, Clinical database or other selected, please specify:

Before undertaking a bespoke data collection from each NHS CGS, a review of Secondary Uses Service (SUS) data was undertaken. The historical recording of clinical genomic activity on SUS indicates that this is not currently a robust way to monitor activity.

2019/20 actual outturn activity recorded on SUS indicated that 66,297 new patients were seen, and 37,351 follow up patient appointments were undertaken. SUS activity reflects approximately 68% of the new patient activity reported by providers through the data collection exercise and 85% of follow up activity. In addition, clinical genomic activity (Treatment Function Code 311) was submitted by 68 trusts, significantly more than the 17 commissioned NHS CGSs.

Therefore, it is proposed that a single national dataset for recording the activity and associated costs is developed as part of the Commissioning Implementation Plan for implementation in the first year following publication of the new service specification.

A6.2 Specify how the activity related to the new patient pathway will be identified.

*Select all that apply:*

| | |
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| OPCS v4.8 | ☐ |
| ICD10 | ☐ |

| | |
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| | <table><tr><td>Service function code</td><td>☒</td></tr><tr><td>Main Speciality code</td><td>☒</td></tr><tr><td>HRG</td><td>☐</td></tr><tr><td>SNOMED</td><td>☐</td></tr><tr><td>Clinical coding / terming methodology used by clinical profession</td><td>☒</td></tr></table> |
| A6.3 **Identification Rules for Drugs:**<br>How are any drug costs captured? | **<u>Not applicable</u>**<br>If already specified in the current NHS England Drug / Devices List, please specify drug name and indication for all that apply:<br>N/A<br>If drug(s) NOT already been specified in the current NHS England Drug List please give details of action required and confirm that this has been discussed with the pharmacy lead:<br>N/A<br><br>**NOTE:** There are currently no drug costs in relation to the NHS CGS. However, it is recognised that the introduction of pharmacogenomic prophylactic prescribing of treatments to prevent the occurrence of genomic conditions may be required in the future.<br>A pathway for the management of 'well' patients, at risk of a condition with a genomic predisposition, will need to be considered in response to the implementation of pharmacogenomics as the patients involved are unlikely to require treatment and management by clinical specialities. Therefore, the prescribing of high cost drugs may become the responsibility of the NHS CGSs, the impact of which should be considered as part of the NHS GMS Pharmacogenomic Workstream. |
| A6.4 **Identification Rules for Devices:**<br>How are device costs captured? | **<u>Not applicable</u>**<br>If device(s) covered by an existing category of HCTED please specify the Device Category (as per the National Tariff Payment System Guidance) for all that apply:<br>N/A |

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| | If device(s) not excluded from Tariff **nor** covered within existing National or Local prices please specify details of action required and confirm that this has been discussed with the HCTED team.<br>N/A |
| A6.5 **Identification Rules for Activity:**<br>How are activity costs captured? | **<u>Not captured by an existing specialised service line</u>**<br>If activity costs are already captured please specify the specialised service code and description (e.g. NCBPS01C Chemotherapy).<br>N/A<br>If activity costs are already captured please specify whether this service needs a separate code. **<u>N/A</u>**<br>If the activity is captured but the service line needs amendment please specify whether the proposed amendments have been documented and agreed with the Identification Rules team.<br>N/A<br>If the activity is not captured please specify whether the proposed identification rules have been documented and agreed with the Identification Rules team.<br><br>It is proposed that a single national dataset for the recording and reporting of NHS CGS activity and associated costs is developed as part of the Commissioning Implementation Plan. We anticipate implementation of the revised dataset during Year One of specification roll-out, with development of a financial model during Year Two. Shadow Monitoring against the activity and financial framework is recommended for Year Three, allowing for further refinement as required. Full Implementation of the new financial model is proposed for Year Four. |
| **A7 Monitoring** | |
| A7.1 **Contracts**<br>Specify any new or revised data flow or data collection requirements, needed for inclusion in the NHS Standard Contract Information Schedule. | **<u>Yes - other</u>**<br>Please specify<br>Single national dataset for development during the Commissioning Implementation Phase |

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| Please identify any excluded drugs or devices relevant to the service and their current status with regard to NHS England specialised services commissioning. | |
| A7.2 **Business intelligence**<br>Is there potential for duplicate reporting? | **Yes**<br>If yes, please specify mitigation:<br>The risk of duplicate reporting will be mitigated through the development of a single national reporting framework for implementation in Year One of specification roll-out |
| A7.3 **Contract monitoring**<br>Is this part of routine contract monitoring? | **Yes**<br>If no, please specify contract monitoring requirement:<br>N/A |
| A7.4 **Dashboard reporting**<br>Specify whether a dashboard exists for the proposed service? | **No**<br>If yes, specify how routine performance monitoring data will be used for dashboard reporting.<br>N/A<br>If no, will one be developed?<br>A Quality Dashboard will be developed to reflect the indicators contained within Section 6.2 of the revised specification. The Quality Dashboard will be published on the NHS England website (Specialised services quality dashboards). In addition, development of an activity dashboard in line with the proposed dataset will be considered as part of the Commissioning Implementation Plan. |
| A7.5 **NICE reporting**<br>Are there any directly applicable NICE or equivalent quality standards which need to be monitored in association with the new service specification? | **Yes**<br>If yes, specify how performance monitoring data will be used for this purpose.<br>Adherence to current clinical guidelines, including NICE guidelines for Lynch Syndrome, cancer, familial hypercholesterolaemia will be monitored a part of routine contract monitoring. |

| Section B - Service Impact | |
|---|---|
| **B1 Service Organisation** | |
| B1.1 Describe how the service is currently organised? (i.e. tertiary centres, networked provision etc.) | Services have historically been provided to patients through a hub and spoke model, with services provided from outpatient facilities across the geographical catchment area of the NHS CGS. However, the COVID-19 pandemic has led to a reduction in the number of sites from which services are provided with the cessation of most satellite services.<br><br>The NHS CGSs are an essential core element of the delivery of the NHS GMS, which includes a network of seven NHS GLHs that deliver genomic testing and seven NHS GMS Alliances that lead on service development and transformation across the network.<br><br>There are also links between the NHS CGSs and the Cancer Alliances and Pathology Networks across the country.<br><br>*Source: Commissioner developed text* |
| B1.2 Will the specification change the way the commissioned service is organised? | **Yes**<br>Please specify:<br><br>The service specification aligns the NHS CGSs position in the national NHS GMS network, ensuring that they are a key partner in all transformation and NHS GMS transformations.<br><br>Whilst mainstreaming is expected to lead to increase the proportion of genomic testing that is requested through mainstream services and reduce the proportion of diagnostic cases referred to the NHS CGS that go on to receive a negative result, it is expected to increase demand placed on NHS CGS in other areas such as:<br><br>• Education and training provided to mainstream clinicians<br>• Participation in Multi-disciplinary Team meetings<br>• Expert advice and guidance provided to mainstream services and primary care<br>• Contribution to NHS GMS transformation programmes and NHS GMS Alliances<br>• Eligible patients referred to NHS CGSs<br>• Complexity of patients managed by NHS CGSs |

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| | • Length of time those more complex patients remain on the caseload<br>Therefore, sufficient capacity needs to be built into the workforce to support this additional burden likely to be placed on NHS CGSs.<br>*Source: Commissioner developed text* |
| B1.3 Will the specification require a new approach to the organisation of care? | **Other**<br>Please specify:<br>A full review of the Hub and Spoke model previously delivered by NHS CGSs needs to be reviewed in view of ensuring that services provide equity of access across their geographical area balanced with the impact of increased non-face to face activity, in particular considering any potential impact on patient experience. In addition, it will be important to consider the reduced availability of satellite locations due to the impact of COVID-19.<br>Furthermore, it will also be important to review the scope of professional roles within services to identify areas of shared practice and the distinctions between role profiles for clinical geneticists, genetic counsellors and the new support roles of the genomic associate and genomic practitioner. The appropriate devolution of some areas of practice is likely to free up clinical capacity for direct patient care, supporting services to meet the forecast demand for services in a cost-effective manner. This may also include the increase of joint appointments for NHS CGS staff within other clinical specialties.<br>As part of the commissioning implementation phase, we propose work jointly with Regional Specialised Commissioning colleagues and service providers to develop consistent operating procedures, including intra-specialty collaboration to achieve mainstreaming and consistent eligibility criteria.<br>Consistent pathways to specialist clinical psychology will need to be developed to ensure that individuals affected by certain complex genomic conditions receive sustained psychological support. This may include joint working with Mental Health commissioners to ensure that adequate specialist capacity is available within local mental health services or the recruitment of clinical psychologists directly into the NHS CGS workforce. |
| **B2 Geography & Access** | |

<table>
<tr>
<td>B2.1 Where do current referrals come from?</td>
<td>
Select all that apply:
<table>
<tr><td>GP</td><td>☒</td></tr>
<tr><td>Secondary care</td><td>☒</td></tr>
<tr><td>Tertiary care</td><td>☒</td></tr>
<tr><td>Other</td><td>☒</td></tr>
</table>
Please specify:<br>
Other includes:
<ul>
<li>National Screening Programmes</li>
<li>Emergency departments and hospital wards</li>
<li>Community services, including dental and optometry services</li>
<li>Self-referrals</li>
</ul>
</td>
</tr>
<tr>
<td>B2.2 What impact will the new service specification have on the sources of referral?</td>
<td><u>No impact</u><br>Please specify:<br>N/A</td>
</tr>
<tr>
<td>B2.3 Is the new service specification likely to improve equity of access?</td>
<td><u>Increase</u><br>Please specify:<br>A primary aspiration of the new service specification is to improve equity of access, which is reflected in the forecast demand estimations.<br>Source: Equalities Impact Assessment</td>
</tr>
<tr>
<td>B2.4 Is the new service specification likely to improve equality of access and/or outcomes?</td>
<td><u>Increase</u><br>Please specify:<br>The revised service specification is expected to improve equality of access and patient outcomes. Mainstreaming of genomic services will help to raise awareness of the testing available and increase the diagnosis rate of conditions with a genetic predisposition. This will also lead to an increase in the</td>
</tr>
</table>

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| | number of family members identified at high risk so that their risk of developing conditions with a genomic predisposition can be minimised. In addition, it is hoped to reduce the diagnostic odyssey experienced by many people with rare diseases that often utilise healthcare services heavily and are reliant on 'trial and error' prescribing to manage their condition and associated symptoms.<br><br>*Source: Equalities Impact Assessment* |
| **B3 Implementation** | |
| B3.1 Will commissioning or provider action be required before implementation of the proposition can occur? | **Data monitoring action**<br>**Contract action**<br>**Finance action**<br>**Service organisation action**<br>Please specify:<br><br>To support implementation of the new service specification, it will be essential to develop a robust Commissioning Implementation Plan jointly with commissioners and providers. Specifically, the Commissioning Implementation will need to include a national approach to:<br><br>• Establishing a single, national reporting framework for activity and cost, potentially with shadow monitoring in year one<br>• Consistent operating procedures, including intra-speciality collaboration to achieve mainstreaming and consistent eligibility criteria<br>• A service delivery model that provides equity of access across England, including minimum hub and spoke requirements to meet local geographical demand and achieving equity of service provision<br>• Developing an optimum workforce structure with a skill mix that is capable of meeting forecast demand in view of potential additional roles such as genomic assistants and associates<br>• Work with Health Education England to ensure systems are in place to increase available staff to meet future demand |

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| | • Developing a consistent financial model that supports the required capacity and service delivery model, achieving equity of access and supporting transformational requirements within the NHS GMS. |
| B3.2 **Time to implementation:**<br>Is a lead-in time required prior to implementation? | **<u>Yes - go to B3.3</u>**<br>If yes, specify the likely time to implementation: 2 years |
| B3.3 **Time to implementation:**<br>If lead-in time is required prior to implementation, will an interim plan for implementation be required? | **<u>Yes</u>**<br>If yes, outline the plan:<br>A detailed Commissioning Implementation Plan will be developed jointly with providers and NHSE Regional Specialised Commissioning Teams detailing mobilisation plan and timescales.<br>The plan outline is expected to include four phases:<br>**Phase 1.** Development and implementation of robust activity and financial monitoring frameworks, including development and roll-out of a consistent Patient Level Contract Monitoring across NHS CGSs, with retrospective data submission from October 2022. Development of a nationally consistent methodology to measure the impact the service has on patient outcomes. This phase will also include a full-service review undertaken jointly by NHS England and NHS CGSs, the development of standard operating procedures and consistent eligibility criteria – **Year 1**<br>**Phase 2.** Interpretation of Year 1 reported activity and financial data. Development of a workforce plan in line with actual demand outturn for Year 1 and local population needs (to be established jointly by NHS GCSs and NHS GMS Alliances). Development of a national financial model, including Market Forces Factor structure and weightings for local service specialisms/expertise and population needs. Formal notice of new financial model to be given to providers no later than 6-months prior to expected date of implementation - **Year 2**<br>**Phase 3.** Shadow monitoring of new financial model, activity and workforce to ensure robust, making refinements if required – **Year 3**<br>**Phase 4.** Roll out of new models with contractual arrangements confirmed – **Year 4** |

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| B3.4 Is a change in provider physical infrastructure required? | **No**<br>Please specify:<br>N/A |
| B3.5 Is a change in provider staffing required? | **Yes**<br>Please specify:<br>As a result of the mainstreaming being undertaken, genomics is being integrated into relevant patient pathways across healthcare specialities. The Joint Committee on Genomics in Medicine summarises the impact of mainstreaming in their paper 'Investing in excellence to provide essential core expertise to the NHS Genomic Medicine Services: Role of the Clinical Geneticist' (July 2019) as:<br>• *The specialist genomic workforce within the Genomic Medicine Services (GMSs), the Clinical Geneticists, Genetic Counsellors and Clinical Scientists, will need to* ***support and educate colleagues*** *in the safe application of genomics for patient benefit whilst continuing to be* ***responsible for the diagnosis and management of patients with rare and ultra-rare disease****.*<br>• *Greater genomic awareness of healthcare practitioners (HCPs) in other medical and surgical specialties has resulted in an unprecedented* ***increase in referral rates*** *to GMSs, as well as an increased demand for* ***genomic multi-disciplinary meetings and clinic.***<br>• ***Offering whole genome sequencing to all seriously ill children as part of their care will generate a large workload*** *for Clinical laboratory teams and Clinical Geneticists as these data are complex and difficult to interpret.*<br>• ***Championing the safe implementation of genomic medicine requires investment in the clinical expertise that is core to this mission.***<br>Based on the job planning guidance published by the Clinical Genetics Society (CGS) (Rationale for a job planning document for Consultants in Clinical/Medical Genetics (2020)) and the Association of Genetic Nurses and Counsellors (AGNC) (Career Structure for Genetic Counsellors and Support Roles (Jan 2021)), we have estimated that the current NHS CGS workforce establishment has a deficit of approximately **10 Clinical Geneticists** when compared to the estimated number required to deliver 2019/20 baseline activity. This problem is compounded by a vacancy rate of over 16% in |

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| | June 2021 and an expectation that approximately 30 clinical geneticists are likely to retire during the coming 5 years**.**<br><br>Whilst the overall number of Genetic Counsellors in establishments across England initially appears sufficient to manage baseline activity it's important to note that this is adversely impacted by a vacancy rate of 9.5% (June 2021).<br><br>Based on the apportionment of 2019/20 baseline activity across NHS CGSs, the variance between capacity required and current establishment differs by service. However, this should be considered in view of differing levels of investment across the network per 100k catchment population and reduced levels of activity in some services because there has been a need to limit their activity within available resources rather than develop lengthy waiting lists.<br><br>In addition to the need to increase establishment within the NHS CGSs to enable them to meet the aspirations of the revised service specification for mainstreaming and improving equity of access, it will also be important to ensure a robust skill mix is developed and introduced. This will ensure there is sufficiently skilled capacity to develop new pathways of care required to achieve mainstreaming and provide the education and training required to both build the genomics workforce and develop the competence required in mainstream services. This skill mix will also need to consider the contribution made to the mainstreaming agenda by other parts of the NHS GMS.<br><br>In October 2020, the CGS and AGNC published the 'Scope of professional roles within specialist genomic medicine services', which highlights areas of shared practice and the distinctions between roles for a Consultant Clinical Geneticist, Principal/Consultant Genetic Counsellor and the new support role that is referred to as the 'Genomic Associate'.<br><br>The genomic associate role is introduced through the revised services specification, and provides administrative support for the clinic, the patient and the clinical activities of the clinical geneticist and genomic counsellor. As part of the data collection exercise undertaken in June 2021, only four of the seventeen NHS CGSs indicated that they have genomic associates incorporated into their establishment. There is scope for all NHS CGSs to consider introducing genomic associates to reduce the administrative burden on clinical geneticists and genomic counsellors, enabling them to free up additional time for patient facing activities. |

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| B3.6 Are there new clinical dependency and/or adjacency requirements that would need to be in place? | **Yes**<br>Please specify:<br>The revised service specification emphasises the importance of access to clinical psychology to provide essential management of individuals affected by specific genomic conditions and to provide more complex or sustained psychological support. At the time the workforce data collection was undertaken, just one service employed clinical psychologists within the NHS CGS. NHS CGSs were also asked to confirm access to a psychology pathway if they did not directly employ clinical psychologists within their service. Nine services provided details of access to psychology services, five of which indicated that they had no direct access pathway available. Therefore, there is scope for services to consider employing clinical psychologists within their service or to work with mental health commissioners to secure direct access via robust pathways that are responsive to clinical genomic patients' needs.<br>In addition, mainstreaming may increase the requirement for clinical geneticists and genomic counsellors to be embedded into speciality services. Four of the seventeen NHS CGSs report that they currently have clinical geneticists or genomic counsellors providing embedded sessions within mainstream services.<br>Alongside embedded sessions, there will certainly be an increased need for clinical genomic input into both mainstream and NHS GLH multi-disciplinary team meetings (MDTs). NHS CGSs report that their clinical geneticists currently attend and average of approximately one Programmed Activity (PA) of MDTs per week (Max. 2.3) and genomic counsellors approximately half a session per week (Max. 1.8). The workforce planning and estimation of capacity requirements associated with this Integrated Impact Assessment allow for clinical geneticists to dedicate **two PAs per week** to specialist MDTs, variant interpretation, and additional genomic work; for genomic counsellors it allows for **7.5 hours** (two sessions) for ongoing case management, MDT meetings and variant interpretation. |
| B3.7 Are there changes in the support services that need to be in place? | **No**<br>Please specify:<br>N/A |

<table>
<tr>
<td>B3.8 Is there a change in provider and/or inter-provider governance required? (e.g. ODN arrangements / prime contractor)</td>
<td><u>No</u><br>Please specify:<br>N/A</td>
</tr>
<tr>
<td>B3.9 Is there likely to be either an increase or decrease in the number of commissioned providers? If yes, specify the current and estimated number of providers required in each region</td>
<td><u>No change</u><br>Please complete the table:<br>NHS England Regional Distribution:
<table>
<tr><th>Region</th><th>Current no. of providers</th><th>Future State expected range</th><th>Provisional or confirmed</th></tr>
<tr><td>North West</td><td>2</td><td>2</td><td>C</td></tr>
<tr><td>North East & Yorkshire</td><td>3</td><td>3</td><td>C</td></tr>
<tr><td>London</td><td>4</td><td>4</td><td>C</td></tr>
<tr><td>South East</td><td>2</td><td>2</td><td>C</td></tr>
<tr><td>South West</td><td>2</td><td>2</td><td>C</td></tr>
<tr><td>Midlands</td><td>3</td><td>3</td><td>C</td></tr>
<tr><td>East of England</td><td>1</td><td>1</td><td>C</td></tr>
<tr><td>Total</td><td>17</td><td>17</td><td>C</td></tr>
</table>
NHS Genomic Laboratory Hub Regional Distribution:
<table>
<tr><th>Region</th><th>Current no. of providers</th><th>Future State expected range</th><th>Provisional or confirmed</th></tr>
<tr><td>North West</td><td>2</td><td>2</td><td>C</td></tr>
</table>
</td>
</tr>
</table>

| | | | | |
|---|---|---|---|---|
| North East & Yorkshire | 3 | 3 | C |
| North Thames | 2 | 2 | C |
| South East | 2 | 2 | C |
| South West | 2 | 2 | C |
| Central and South | 3 | 3 | C |
| East of England | 3 | 3 | C |
| Total | 17 | 17 | C |

Please specify:
N/A

B3.10 Specify how revised provision will be secured by NHS England as the responsible commissioner.

*Select all that apply:*

| | |
|---|---|
| Publication and notification of new service specification | ☒ |
| Market intervention required | ☐ |
| Competitive selection process to secure increase or decrease provider configuration | ☐ |
| Price-based selection process to maximise cost effectiveness | ☐ |
| Any qualified provider | ☐ |
| National Commercial Agreements e.g. drugs, devices | ☐ |
| Procurement | ☐ |

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| | Other ☐<br>Please specify:<br>N/A |
| **B4 Place-based Commissioning** | |
| B4.1 Is this service currently subject to, or planned for, place-based commissioning arrangements? (e.g. future CCG lead, devolved commissioning arrangements, STPs) | **No.**<br>Please specify:<br>The commissioning for NHS CGS will remain with NHS England until it reaches a steady state following specification implementation, after which commissioning arrangements will devolve to Integrated Care Systems. |
| **Section C - Finance Impact** | |
| **C1 Tariff/Pricing** | |
| C1.1 How is the service contracted and/or charged?<br>Only specify for the relevant section of the patient pathway | *Select all that apply:* |

| | | |
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| **Drugs** | Not separately charged – part of local or national tariffs | ☐ |
| | Excluded from tariff – pass through | ☐ |
| | Excluded from tariff - other | ☐ |
| **Devices** | Not separately charged – part of local or national tariffs | ☐ |
| | Excluded from tariff (excluding ZCM) – pass through | ☐ |
| | Excluded from tariff (excluding ZCM) – other | ☐ |

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| | | Via Zero Cost Model | ☐ |
| | **Activity** | Paid entirely by National Tariffs | ☐ |
| | | Paid entirely by Local Tariffs | ☐ |
| | | Partially paid by National Tariffs | ☐ |
| | | Partially paid by Local Tariffs | ☒ |
| | | Part/fully paid under a Block arrangement | ☒ |
| | | Part/fully paid under Pass-Through arrangements | ☐ |
| | | Part/fully paid under Other arrangements | ☐ |
| C1.2 **Drug Costs**<br>Where not included in national or local tariffs, list each drug or combination, dosage, quantity, **list** price including VAT if applicable and any other key information e.g. Chemotherapy Regime.<br>NB discounted prices or local prices must not be included as these are subject to commercial confidentiality and must not be disclosed. | N/A | | |
| C1.3 **Device Costs**<br>Where not included in national or local tariff, list each element of the excluded device, quantity, **list or expected** price including VAT if applicable and any other key information. | N/A | | |

| | |
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| NB: Discounted prices or local prices must not be included as these are subject to commercial confidentiality and must not be disclosed. | |
| C1.4 **Activity Costs covered by National Tariff**<br>List all the HRG codes, HRG descriptions, national tariffs (excluding MFF), volume and other key costs (e.g. specialist top up %) | N/A |
| C1.5 **Activity Costs covered by Local Tariff**<br>List all the HRGs (if applicable), HRG or local description, estimated average tariff, volume and any other key costs. Also indicate whether the Local Tariff(s) is/are newly proposed or established and if newly proposed how is has been derived, validated and tested. | Across the 11 providers that receive tariff for all or part of the contracted income there are 44 individual tariffs (See Appendix 1).<br>A comparison of total income received during 2019/2020 with provider declared activity for the same year indicates significant variation of income by unit of activity across services (Min. £210, Max £1,510, Median £370 and Mean £490 per unit of reported activity).<br>Similarly, there is significant variation in the provider declared total income in 2019/20 when calculating the cost per WTE clinical workforce (including both clinical geneticist and genomic counsellor establishment) (2021/22) (Min. £69k per WTE, Max £230k per WTE, Median £139k per WTE and Mean £147k per WTE).<br>Therefore, there is scope to undertake a review of the contractual payment mechanisms cost of service delivery to ensure consistency across the clinical genetic services and ensure that the investment provided enables delivery of the services specification and achieves equity across service provision. This is proposed to be undertaken during Phase 2 of the Commissioning Implementation Plan following the development and implementation of a single, national, dataset for recording the activity and associated costs in Phase 1. |
| C1.6 **Other Activity Costs not covered by National or Local Tariff** | See Section C1.5 |

<table>
<tr><td>Include descriptions and estimates of all key costs.</td><td></td></tr>
<tr><td>C1.7 Are there any prior approval mechanisms required either during implementation or permanently?</td><td><u>No</u><br>Please specify: N/A</td></tr>
<tr><td colspan="2">C2 Average Cost per Patient</td></tr>
<tr><td>C2.1 What is the estimated cost per patient to NHS England, in years 1-5, including follow-up where required?<br><br>Are there any changes expected in year 6-10 which would impact the model?</td><td><table><tr><td>YR1</td><td>£630</td></tr><tr><td>YR2</td><td>£630</td></tr><tr><td>YR3</td><td>Not Known</td></tr><tr><td>YR4</td><td>Not Known</td></tr><tr><td>YR5</td><td>Not Known</td></tr></table>If yes, please specify:<br>Cost per patient for Year 3 and beyond will be established as part of the financial model to be developed during Phase 2 of the Commissioning Implementation Plan.</td></tr>
<tr><td colspan="2">C3 Overall Cost Impact of this Service specification to NHS England</td></tr>
<tr><td>C3.1 Specify the budget impact of the proposal on NHS England in relation to the relevant pathway.</td><td><u>Cost pressure</u><br>Please specify:<br>Work to understand the predicted capacity requirements has commenced and, when compared to the current estimated service budget, there is an indicative cost pressure. However, the estimated budgetary impact is only indicative and requires robust validation and refinement through a full-</td></tr>
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| | service review, workforce modelling, activity monitoring and financial analysis which will be undertaken during Phases 1 and 2 of the Commissioning Implementation Plan. |
| C3.2 If the budget impact on NHS England cannot be identified set out the reasons why this cannot be measured. | N/A |
| C3.3 If the activity is subject to a change of commissioning responsibility, from CCG to NHS England, has a methodology for the transfer of funds been identified, and calculated? | N/A |
| **C4 Overall cost impact of this service specification to the NHS as a whole** | |
| C4.1 Specify the budget impact of the proposal on other parts of the NHS. | Budget impact for CCGs:<br>**No impact on CCGs**<br>Budget impact for providers:<br>**Cost pressure**<br>Please specify:<br>Providers would be unable to meet the forecast demand for services within current resources, leading to an inability to deliver against the proposed revised service specification. This would lead to an inability to achieve the NHS GMS and NHS Long Term Plan for mainstreaming genomics and reduce scope for achieve equity of access for the eligible catchment population. Furthermore, there would be limited scope to address the diagnostic odyssey experienced by many patients, leading to a continuation of many years of costly investigations and 'trial and error' prescribing and the negative impact on patients' quality of life. |

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| C4.2 Taking into account responses to C3.1 and C4.1, specify the budget impact to the NHS as a whole. | **Cost pressure**<br>Please specify:<br>Unknown: to be calculated and specified as part of Phase 1 and 2 of commissioning implementation |
| C4.3 Where the budget impact is unknown set out the reasons why this cannot be measured | N/A |
| C4.4 Are there likely to be any costs or savings for non-NHS commissioners and/or public sector funders? | **No**<br>Please specify:<br>N/A |
| **C5 Funding** | |
| C5.1 Where a cost pressure is indicated, state known source of funds for investment, where identified, e.g. decommissioning less clinically or cost-effective services. | Whilst not quantified, some savings are expected through the reduction in diagnostic odysseys and the associated costs of multiple investigations and 'trial and error' prescribing. Further work to quantify the level of potential savings is required, and to establish a more robust value of the actual cost pressure, through the commissioning implementation phase. |
| **C6 Financial Risks Associated with Implementing this Service specification** | |
| C6.1 What are the material financial risks to implementing this service specification? | The financial risks include the current variation in activity monitoring and financial models across all NHS CGSs, which indicate significant variation in the cost per patient across services. Therefore, without additional work undertaken during commissioning implementation, it is not possible to estimate financial pressure experienced currently across the services. However, it is important that these variations are addressed to ensure that services are resourced in line with their local patient need to ensure equity of access. It is also essential that providers have adequate capacity to |

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| | manage both the direct patient care needs but also the additional workload introduced by the service specification, such as supporting the mainstreaming of genomics, providing expert advice and guidance to clinicians, providing valuable input into MDTs and supporting the transformation work being undertaken by the NHS GMS Alliances. |
| C6.2 How can these risks be mitigated? | Ensuring that robust workforce planning is undertaken in view of a comprehensive understanding of national clinical genomics activity, and development of a financial model that ensures sufficient resource for service delivery whilst delivering best possible value for money. |
| C6.3 What scenarios (differential assumptions) have been explicitly tested to generate best case, worst case and most likely total cost scenarios? | The calculations detailed within this Integrated Impact Assessment are expected to indicate a worst-case scenario. Further modelling will be undertaken when robust and consistent activity monitoring information is available. |
| C6.4 What scenario has been approved and why? | N/A |
| **C7 Value for Money** | |
| C7.1 What published evidence is available that the service is cost effective as evidenced in the evidence review? | **There is no published evidence of cost-effectiveness**<br>Please specify:<br>N/A |
| C7.2 Has other data been identified through the service specification development relevant to the assessment of value for money? | *Select all that apply:*<br>Available pricing data suggests the service specification is equivalent cost compared to current/comparator service specification ☐ |

| | |
|---|---|
| | <table><tr><td>Available pricing data suggests the service is lower cost compared to current/comparator treatment</td><td>☐</td></tr><tr><td>Available clinical practice data suggests the new service specification has the potential to improve value for money</td><td>☐</td></tr><tr><td>Other data has been identified</td><td>☐</td></tr><tr><td>No data has been identified</td><td>☐</td></tr><tr><td>The data supports a high level of certainty about the impact on value</td><td>☐</td></tr><tr><td>The data does not support a high level of certainty about the impact on value</td><td>☒</td></tr></table><br>Please specify:<br>Consistent data collection methodology needs developing and implementing during Phase 1 of the Commissioning Implementation Plan |
| **C8 Non-Recurrent Costs** | |
| C8.1 Are there non-recurrent revenue costs associated with this service specification? | **Yes**<br>If yes, please specify and indicate whether these would be incurred or passed through to NHS England:<br>There are non-recurrent costs expected to facilitate the implementation of a new activity monitoring framework. These costs will be identified as part of developing the Commissioning Implementation Plan.<br>If the costs are to be passed through to NHS England please indicate whether this has been taken into account in the budgetary impact.<br>**No** |

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| C8.2 Are there any non-recurrent provider capital costs associated with the service specification? | **No**<br>If yes, please specify and indicate with there is a separate source of funding identified (commissioners cannot reimburse capital costs).<br>N/A |

**APPENDIX 1**

| | Provider 1 | Provider 2 | Provider 3 | Provider 4 | Provider 5 | Provider 6 | Provider 7 | Provider 8 | Provider 9 | Provider 10 | Provider 11 | Provider 12 | Provider 13 | Provider 14 | Provider 15 | Provider 16 | Provider 17 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| OUTPATIENT - ATTENDANCE - FIRST - MULTI-PROFESSIONAL - CONSULTANT LED | | BLOCK CONTRACT | | | | | BLOCK CONTRACT | | | BLOCK CONTRACT | BLOCK CONTRACT | | | BLOCK CONTRACT | | BLOCK CONTRACT | 363 |
| OUTPATIENT - ATTENDANCE - FIRST - SINGLE PROFESSIONAL - CONSULTANT LED | 562 | | 500 | | 555 | | | | | | | | | | 465 | | 363 |
| OUTPATIENT - ATTENDANCE - FIRST - MULTI PROFESSIONAL - NON-CONSULTANT LED | | | | | | | | | | | | | | | | | 363 |
| OUTPATIENT - ATTENDANCE - FIRST - SINGLE PROFESSIONAL - NON-CONSULTANT LED | | | 500 | 620 | | | | | | | | | | | | | 363 |
| OUTPATIENT - ATTENDANCE - FOLLOW-UP - MULTI-PROFESSIONAL - CONSULTANT LED | | | | 393 | 582 | | | | | | | | | | | | 363 |
| OUTPATIENT - ATTENDANCE - FOLLOW-UP - SINGLE PROFESSIONAL - CONSULTANT LED | 338 | | 500 | 393 | | | | | | | | | | | 465 | | 363 |
| OUTPATIENT - ATTENDANCE - FOLLOW-UP - MULTI PROFESSIONAL - NON-CONSULTANT LED | | | | | | | | | | | | | | | | | 363 |
| OUTPATIENT - ATTENDANCE - FOLLOW-UP - SINGLE PROFESSIONAL - NON-CONSULTANT LED | | | 500 | 393 | | | | | | | | | | | | | 363 |
| OUTPATIENT - NON-FACE TO FACE - FIRST - SINGLE PROFESSIONAL - CONSULTANT LED | | | 500 | 220 | 501 | | | | | | | | | | 67 | | 363 |
| OUTPATIENT - NON-FACE TO FACE - FIRST - SINGLE PROFESSIONAL - NON-CONSULTANT LED | | | 500 | 220 | | | | | | | | | | | | | 363 |
| OUTPATIENT - NON-FACE TO FACE - FOLLOW-UP - SINGLE PROFESSIONAL - CONSULTANT LED | | | 500 | 10 | 537 | | | | | | | | | | 70 | | 363 |

| | Provider 1 | Provider 2 | Provider 3 | Provider 4 | Provider 5 | Provider 6 | Provider 7 | Provider 8 | Provider 9 | Provider 10 | Provider 11 | Provider 12 | Provider 13 | Provider 14 | Provider 15 | Provider 16 | Provider 17 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| OUTPATIENT - NON-FACE TO FACE - FOLLOW-UP - SINGLE PROFESSIONAL - NON-CONSULTANT LED | | | 500 | 10 | | | | | | | | | | | | | 363 |
| OTHER: WARD VISIT | | | | | | | | | | | | | | | | | |
| CLINICAL GENETICS - CLGEN (Band 6) Out Patient First Appointment | | | | | | 612 | | | | | | | | | | | |
| CLINICAL GENETICS - CLGEN (Band 1) Chargeable Letter (Advice and Guidance (A&G)/Results) | | | | | | 104 | | | | | | | | | | | |
| CLINICAL GENETICS - CLGEN (Band 4) Out Patient Follow Up Appointment | | | | | | 414 | | | | | | | | | | | |
| CLINICAL GENETICS - CLGEN (Band 2) Chargeable Phone Call (A&G/Results) | | | | | | 207 | | | | | | | | | | | |
| CLGEN_BAND4b (Telephone clinic initiated pre-COVID) | | | | | | 414 | | | | | | | | | | | |
| OUTPATIENT - NON FACE TO FACE CONTACT | | | | | | | | | | | | 24 | | | | | |
| OUTPATIENT - FIRST ATTENDANCE | | | | | | | | | | | | 644 | | | | | |
| OUTPATIENT - FOLLOW UP ATTENDANCE | | | | | | | | | | | | 599 | | | | | |
| Pre-clinic visit/follow-up - Band A | | | | | | | | | | | | | 171 | | | | |
| New, routine referral - Band B | | | | | | | | | | | | | 338 | | | | |
| Genetics - New, complex referral - Band C | | | | | | | | | | | | | 485 | | | | |
| Genetics - New referral using DNA technology - Band D | | | | | | | | | | | | | 1134 | | | | |
| OUTPATIENT - NON-FACE TO FACE - FIRST - SINGLE PROFESSIONAL - CONSULTANT LED CANCER | | | | | | | | | | | | | | | 117 | | |
| OUTPATIENT - NON-FACE TO FACE - FOLLOW-UP - SINGLE PROFESSIONAL - CONSULTANT LED CANCER | | | | | | | | | | | | | | | 117 | | |

| | Provider 1 | Provider 2 | Provider 3 | Provider 4 | Provider 5 | Provider 6 | Provider 7 | Provider 8 | Provider 9 | Provider 10 | Provider 11 | Provider 12 | Provider 13 | Provider 14 | Provider 15 | Provider 16 | Provider 17 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Clinical Genetics Familiy History Review (virtual) | | | | | | | | 212 | | | | | | | | | |
| Genetics Telephone | | | | | | | | 53 | | | | | | | | | |
| Genetics Consultant | | | | | | | | 630 | | | | | | | | | |
| Genetics Counsellor | | | | | | | | 265 | | | | | | | | | |
| My Medical Record - Message contact | | | | | | | | 26 | | | | | | | | | |
| OUTPATIENT - ATTENDANCE SINGLE PROFESSIONAL | | | | | | | | | | | | | | | | | |
| OUTPATIENT - NON-FACE TO FACE SINGLE PROFESSIONAL | | | | | | | | | | | | | | | | | |
| MDT MULTI-PROFESSIONAL - CONSULTANT LED | | | | | | | | | | | | | | | | | |
| PGD NON FACE TO FACE SINGLE PROFESSIONAL | | | | | | | | | | | | | | | | | |
| OUTPATIENT - ATTENDANCE - SINGLE PROFESSIONAL | | | | | | | | | 332 | | | | | | | | |
| OUTPATIENT - NON-FACE TO FACE - SINGLE PROFESSIONAL | | | | | | | | | 249 | | | | | | | | |
| Preimplantation genetic diagnosis referrals assessment | | | | | | | | | 332 | | | | | | | | |
| Preimplantation genetic diagnosis referrals pre-appt preparation | | | | | | | | | 415 | | | | | | | | |
| Lynch Clinical Genetics led MDT | | | | | | | | | 249 | | | | | | | | |
| WES/WGS Joint GLH/Clinical MDT | | | | | | | | | 249 | | | | | | | | |
| Genomic Tumour Assessment MDT | | | | | | | | | 249 | | | | | | | | |
| Additional Relative - Clinical Advice | | | | | | | | | 83 | | | | | | | | |