Classification: Official

DRAFT NHS Genomic Medicine Service: Service Specification

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To note

This NHS Genomic Medicine Service (GMS): Service Specification is a draft document and the Commissioner reserves its right to amend this document.

Any guidance or associated document referenced in the Service Specification that is not publicly available can be made available by request. Please submit a request via Atamis as soon as possible.

Contents

Scope

Introduction

1. This Service Specification (the Specification) covers the provision of all the functions of the NHS Genomic Medicine Service (GMS) in England. The purpose of this Specification is to define the requirements, responsibilities, expertise and interactions of each of the functions, all of which will be required to be delivered by the Lead Provider to deliver the NHS GMS in defined geographies.

2. The NHS GMS was established in 2018 by NHS England to support standardised, high quality and equitable access to Genomic Medicine across the NHS in England and will be pivotal to ensuring that the NHS is the best-placed system in the world to harness advances in genomic science coupled with similar advances in data, AI and predictive analytics.

3. The NHS has continued to invest in the infrastructure of the NHS GMS, which has evolved since its inception, including in 2020 the launch of the NHS whole genome sequencing (WGS) service and NHS GMS Alliances. In 2024/25 the genomic testing element of the NHS GMS enabled patients to access over 870,000 cutting edge genomic tests for over 7,000 rare diseases with a genetic cause and 200 cancer indications. Through the Clinical Genetics Service and active involvement of or linkage with multiple other clinical specialities, the NHS GMS has supported rapid diagnosis and access to treatments and interventions for multiple different patient groups, diseases and conditions.

4. In addition to ensuring the widespread integration of genomics into the NHS, the NHS GMS also enables significant investment, including from industry partners, into genomic research and innovation which is translated - at pace - into the NHS. The NHS GMS alignment and linkage with research through but not limited to Genomics England, has enabled hundreds of scientific publications and research projects and initiatives not just in the UK but globally. It is this innovative and world-leading approach that puts the NHS GMS at the front of the global genomics revolution.

5. In 2022, the NHS launched the first ever NHS genomics strategy, Accelerating Genomic Medicine in the NHS, which set out four key pillars of delivery:
   - embedding genomics in the NHS, through a world leading, innovative service model;
   - delivering equitable genomic testing for improved prediction, prevention, diagnosis and precision medicine;

- enabling genomics to be at the forefront of the data and digital revolution; and
- evolving the service through cutting-edge science, research and innovation.

6. This Specification will build on the commitments in the NHS Genomics Strategy and looks towards the future to support the scale of change and ambition set out in the NHS 10 Year Health Plan and the Life Sciences Sector Plan and will be responsive to the anticipated National Cancer Plan. It recognises that transformational change is needed to deliver a Genomic Medicine Service that is fit for purpose and can support the three strategic shifts in healthcare such that by 2035, the healthcare of half of all individuals interacting with the NHS will be informed by genomic insights and other predictive analytics, and Neighbourhood Health Services will have further evolved to fully incorporate genomic data, digital tools and technology.

7. From sickness to prevention, analogue to digital and hospital to community, the NHS GMS will play a key role in reshaping the way in which the NHS delivers care. Moving forward the NHS GMS will be at the heart of bringing together genomics, artificial intelligence and predictive analytics as outlined in the NHS 10 Year Health Plan. Key to this future is a new genomic population health service – a new paradigm - which will move the NHS GMS from a service that has been developed around cancer and rare disease, to one where a third pillar - population health - is of equal focus.

8. To respond to this new paradigm, each NHS GMS Lead Provider will:
   - strategically work with commissioners, providers and other system stakeholders and partners to maximise the benefits that genomics can deliver for patients, their families and carers, and communities more broadly;
   - drive new models of care informed by the use of genomics and changes in care pathways across rare diseases, cancer, more common disease and in approaches to population health, underpinned by operational and clinical guidance and new services, including a new Genomics Population Health Service;
   - provide a comprehensive and equitable genomic testing offer with adoption of new advances and technologies linked to the access to precision and advanced therapeutic medicinal products (ATMP) medicines and to clinical trials;
   - accelerate the use of automation and industrialisation in laboratory services, including through exploring Public Private Partnerships (PPP), to create efficiencies, drive productivity, enable consolidation, deliver high throughput testing and reduce turnaround times;
   - further develop the genomic data and digital infrastructure, enabling genomic and clinical data to be connected in near real time, and ensuring data can be used for multiple purposes;

- develop the multispecialty workforce to continue to mainstream genomics and to make genomics commonplace in the NHS through normalising its use and application across its geography; and
- enable innovation, research and excellence in genomics, through working in partnership with Genomics England and others, in order to generate and build evidence to adopt innovation into commissioning plans and to support broader scientific endeavours and global developments.

Description

9. The NHS GMS is designated as a prescribed specialist service by the Commissioner.

10. The services and requirements that each NHS GMS Lead Provider will be required to deliver or adhere to are outlined at a high level in **Figure 1** and in more detail through this Specification. They are:
    - overarching governance including a leadership and governance model, and close working with the whole NHS system;
    - genomic laboratory services inclusive of delivery of the National Genomic Test Directory ("**Test Directory**"), delivery of genomic testing from the pre-analytical through to the post-analytical stage; delivery of the full repertoire of genomic technologies; and delivery of the genomic testing strategy over the next 10 years;
    - clinical functions including:
        - cancer genomics;
        - rare disease; and
        - population health;
    - promotion of and supporting and leading where relevant science, research and/or innovation, including through partnerships with Genomics England and others;
    - transformation and service improvement, including the delivery of equitable genomic services;
    - people - workforce, education and training; and
    - data and digital development and implementation both across the Geography and on national projects.

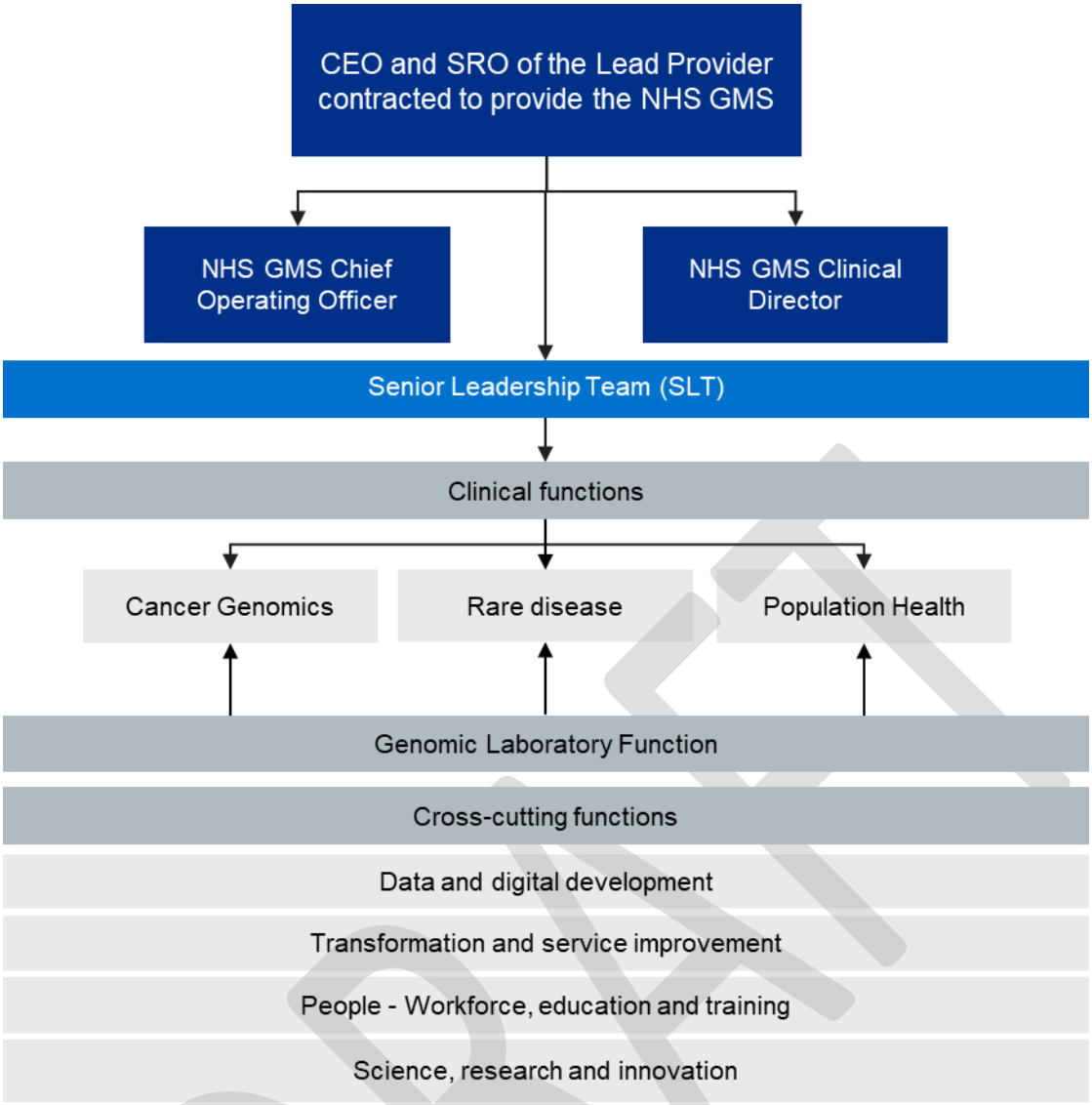


Figure 1 - NHS GMS model

11. This Specification outlines the services to be provided by each NHS GMS Lead Provider for a maximum term of ten years, anticipated to be from 1 April 2026 to 31 March 2036. For the services outlined in this Specification there are three key time frames detailed:
    - 2026 / 2027: activities to be delivered within the first year of each contract;
    - 2027 / 2028 – 2028 / 2029: activities to be delivered by the end of the current comprehensive 'Spending Review' period;
    - 2029 / 2030 onwards: activities to be delivered within the next; and
    - comprehensive 'Spending Review' periods.

12. This Specification may continue to be updated throughout the term and further annexes may be added, enabling the Commissioner to confirm the timings and detailed requirements as plans for delivery evolves. The maximum term of the contract (that underpins the Specification) align to the NHS 10 Year Health Plan.

13. Recognising the need to retain flexibility for any changes in priority or funding during the term, and the need for the Commissioner to effectively manage performance during the full term, the NHS GMS contracts will include mechanisms to address potential changes in the scope of the services over the maximum 10 year term, and performance measures and prescribed routes of escalation in the event that performance is unsatisfactory, with termination potentially being the final outcome.

Population covered and population needs

14. This Specification covers access to Genomic Medicine for both children and young people and adults across cancer, rare diseases inclusive of prenatal care and population health in the NHS in England.

15. A total of 346,000 individuals (inclusive of children, young people and adults) are diagnosed with **cancer** every year in England, a number that has grown each year since 1995, except for 2020. Around half of these diagnoses will be of the most common cancers (namely breast, lung, prostate, and colorectal) and the other half will be of rare or less common types as well as haematological cancers. This rise in incidence rate is due in part to the ageing and growth of the population, which is a result of the overall success of the healthcare system and means that people are less likely to die early from other conditions, such as cardiovascular disease. Cancer recurrence rates in England vary significantly depending on the type of cancer, stage at diagnosis, and treatment received. There is a rising demand for genomic testing in cancer services across a number of areas, including prevention, early diagnosis, precision treatment, monitoring, and access to clinical trials.

16. It is estimated that there are over 7,000 rare diseases and that at least 80% are genetic in nature, with many of them being monogenic. Although rare diseases are individually rare, they are collectively common – with 1 in 17 people being affected by a rare disease at some point in their lifetime. Seventy-five per cent of rare diseases affect children and more than 30% of children with a rare disease die before their fifth birthday. Many patients experience a diagnostic odyssey that involves receiving a large number of diagnostic tests before a diagnosis is made or receiving no diagnosis at all. Prenatal genomic testing can be used to detect genetic disorders and chromosomal abnormalities in a fetus, providing valuable information for expecting parents, for example to inform healthcare decisions about pregnancy management and potential interventions and enabling parents to make future reproductive choices.

17. Genomics can be used in **population health** to either look for rare and other conditions as part of screening programmes or to predict when individuals are at a high relative

risk of developing certain common diseases, such as cancer, diabetes, cardiovascular, and renal disease through for example the use of polygenic risk scores (PRS). It can identify individuals at increased risk of developing a disease that runs in their family, such as inherited forms of cancer and cardiac conditions. With the appropriate support in place, Genomic Medicine can empower individuals and family members to access interventions to either prevent the development of conditions, treat the condition, or manage their individual risk to prevent more serious health impacts. Genomic Medicine in the form of pharmacogenomics can be used to improve the safety of medicines and prevent side effects that some individuals experience when prescribed certain medicines and can optimise medicines use. This use can be either through pre-emptive testing to prepare for any future medicines use or responsive when certain medicines are going to be prescribed.

Excluded from scope

18. Access to genomic tests and the clinical function to embed genomics in end-to-end pathways is covered by this Specification and is the sole commissioning responsibility of the Commissioner. From 1 April 2026, any genomic testing currently undertaken in Highly Specialised Services, particular examples include the genomic tests included in the '6+1 molecular diagnostic tests' and tests used for preimplantation genetic diagnosis (PGD) will be provided by the NHS GMS.

19. The NHS GMS Lead Provider will be required to work with commissioners outside of the scope of the Specification to enable the delivery of services. For example commissioners of Highly Specialised Services for which the NHS GMS Lead Provider delivers the genomic element of the end-to-end pathway.

20. Any genomic test that is not included in the Test Directory is out of the scope of this Specification although they may feature in the work of an NHS Genomic Network of Excellence, or be added over the duration of each contract. This includes:
    - Testing for infectious diseases, such as molecular sequencing of pathogens;
    - Tissue typing testing;
    - Cellular Pathology (including histopathology) that does not use genomic technologies, for example immunochemistry, or provides limited routine molecular pathology testing, such as in situ hybridisation for HER2;
    - Newborn screening undertaken by biochemical testing; and
    - Biochemical antenatal screening and other screening programmes (except for Non-Invasive Prenatal Testing (NIPT), which is in the scope of the Specification).

Overarching governance

Principles

21. Each NHS GMS Lead Provider will be required to apply, and adhere to, the following key principles to:

- identify and work in partnership with the whole NHS system, commissioners and providers, including ICBs, Regions, Primary and Community care, Neighbourhood Health Services and other services and clinical networks;
- work collaboratively as a national network of NHS GMS Lead Providers to drive high quality and universal standards and outcomes for patients;
- adopt a clinical multi-disciplinary leadership approach to give healthcare professionals and clinical teams the skills they need to embed genomics;
- embed genomics through best practice service improvement and transformation methodology to deliver a sustainable and productive business as usual service and end to end clinical pathway model;
- ensure Patient and Public Voice (PPV) involvement is at the heart of the service, including any changes to service design and delivery;
- use robust and up-to-date data to underpin service delivery, drive service improvement and maximise financial outcomes and efficiencies;
- support the delivery of national and regional strategy including the NHS 10 Year Health Plan, Life Sciences Sector Plan and National Cancer Plan;
- reflect patient and population need, identifying, monitoring and reducing health inequalities and tackling inequalities in outcomes, experience and access in line with the Core20Plus5 approach to reducing health inequalities; and
- work collaboratively with academic and industry partners in the life sciences sector, as well research partners such as the National Institute of Healthcare Research, and others in the genomics ecosystem such as Genomics England and Our Future Health, to establish partnerships that support the generation of scientific evidence and models of adoption for new technologies, testing and clinical laboratory practice.

22. Each NHS GMS Lead Provider must ensure that the delivery of the NHS GMS is patient-centred, with PPV embedded at every stage, from strategy to delivery. Each NHS GMS Lead Provider is required to appoint to key PPV roles. PPV is also required to form a central role in the governance of each NHS GMS geography, with a geographical NHS GMS People and Communities Group contributing and informing a national PPV group. Each NHS GMS Lead Provider will regularly carry out and respond to survey outcomes from patients and service users of the NHS GMS.

Leadership and governance model

23. To fulfil the requirements of the Specification, each NHS GMS Lead Provider will need to establish a leadership and governance model to support the relationship with the Commissioner and others in the system including regional teams, Integrated Care Boards (ICBs), NHS Trusts and users including healthcare professionals, and patients and the public.

Leadership model

24. The Chief Executive Officer (CEO) of each NHS GMS Lead Provider - or a designated Board Executive that is approved by the Commissioner - is required to be appointed as the Senior Responsible Officer (SRO) responsible for the strategic direction, delivery and infrastructure of the NHS GMS in its geography. The SRO must secure board level support for the NHS GMS and what this means to be a lead provider for a larger population than their own service footprint and the implications of that. The SRO must also actively engage senior leaders across the geography, including key delivery partners, all providers across the care continuum, emerging Neighbourhood Health Services, ICBs and NHS Regions and other system partners and networks where relevant, to ensure its effective delivery of the functions outlined in this Specification. The SRO must also report regularly to the Board of the Lead Provider and provide evidence of that to the Commissioner.

25. The leadership model of each geographical NHS GMS must include the mandated posts of NHS GMS Chief Operating Officer (COO) and NHS GMS Clinical Director (CD). Job Descriptions set by the Commissioner for these posts are included at **Annex 4** and appointment to these posts are subject to the approval of the Commissioner. The COO and the CD will report into the SRO and will also lead the geographical NHS GMS Senior Leadership Team (SLT).

26. To support the SLT, each NHS GMS Lead Provider must appoint a Service Delivery Team (SDT), including a number of posts mandated by the Commissioner, to ensure delivery of the elements set out in the role descriptions set out in **Annex 4** which must directly support each of the functions set out in the Specification.

27. To support the SLT, each NHS GMS Lead Provider must appoint a Service Delivery Team (SDT), including a number of posts mandated by the Commissioner, that directly support each of the functions set out in the Specification. Role descriptions are set out in **Annex 4**.

28. Each member of the SLT and SDT must be employed by or otherwise contracted by the NHS GMS Lead Provider, or by a NHS GMS third party contractors. Each NHS GMS Lead Provider must be able to provide evidence of the competence of each personnel appointed at the request of the Commissioner.

National governance model

29. Each NHS GMS Lead Provider will work with all other NHS GMS Lead Providers as part of a NHS GMS National Network convened by the Commissioner, providing a comprehensive, coordinated and resilient NHS GMS.

30. The NHS GMS National Network will be overseen by a national NHS GMS Delivery Board, membership of which will be specified by the Commissioner, which will operate with the purpose of:

- providing national operational oversight of the NHS GMS National Network, including identifying areas of variation across and within the NHS GMS geographies;
- working with the commissioner to deliver the genomics commitments set out in the NHS 10 Year Health Plan and Life Sciences Sector Plan and other anticipated plans for workforce and cancer
- developing strategies to address variation in the maturity of each NHS GMS geography as defined in the NHS GMS Maturity Matrix;
- driving implementation of Genomic Medicine across the care continuum within the geography using education and training and other resources, identifying potential barriers and solutions to address them;
- overseeing the implementation and delivery of new services, including the Genomic Population Health Service, and national transformation programmes and priorities;
- ensuring the involvement of patients and the public in the design and delivery of genomic services;
- sharing best practice to support the systematic and equitable implementation of Genomic Medicine into the NHS, including equitable implementation of the Test Directory and promoting equitable access to clinical trials and to precision medicines that are enabled by genomic testing;
- supporting activities to raise awareness and embedding of genomics across all clinical specialties;
- supporting, promoting and where relevant leading research, development and innovation activities and ensuring geographical engagement and involvement working with a range of genomics ecosystem partners;
- ensuring alignment with ICBs, NHS regional teams and other services and networks, to inform planning for population health as well as facilitating delivery; and
- considering people and workforce implications and requirements including education and training and approaches to address them.

31. Each NHS GMS Lead Provider is required to actively participate in regular meetings convened by the Commissioner and shall ensure the attendance of its NHS GMS third-party contractors as required.

32. National oversight groups will be established by the Commissioner to oversee the delivery of the NHS GMS in key areas including the clinical functions for cancer genomics, rare disease and population health, the Genomic Laboratory Services and the cross-cutting enablers, including data and digital, transformation and service improvement and people. Transformation and improvement strategies will form a fundamental part each of the oversight groups. When necessary, time-limited task and finish groups will be established and will report directly to the corresponding national oversight group.

33. The governance will include advisory groups and forums, including the Genomics Clinical Reference Group; NHS Genomics Ethics, Equity and Legal Advisory Group; NHS GMS People and Communities Forum and NHS GMS Science, Research and Innovation Group.

34. The governance will continue to evolve to ensure alignment with the implementation of the NHS 10 Year Health Plan and its requirements.

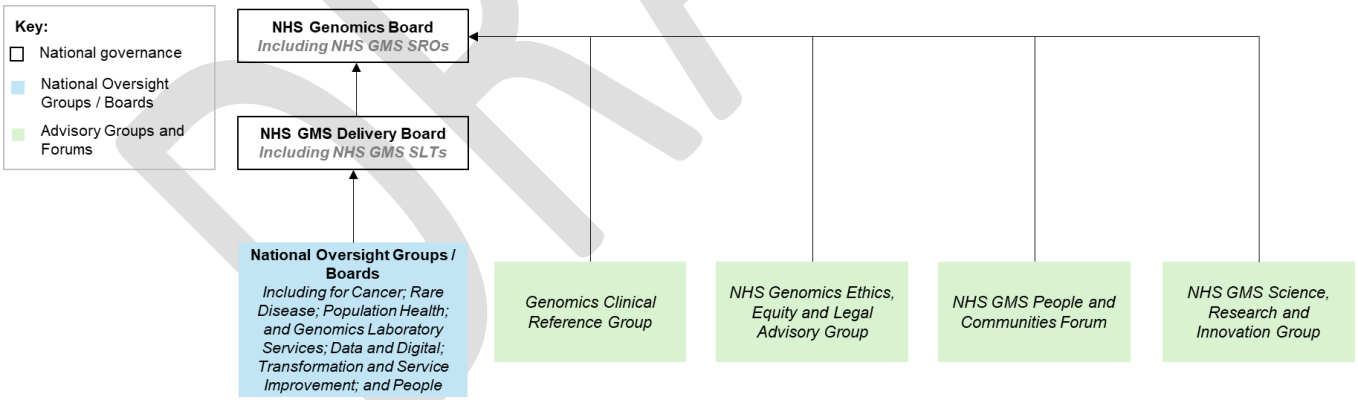


Figure 2 – high level national governance structure

Geographical governance model

35. Each NHS GMS Lead Provider will be responsible for the performance, contract management and quality oversight of each of its NHS GMS delivery partners and third-party contractors.

36. Each NHS GMS Lead Provider is required to establish an NHS GMS Partnership Board including representation from each function of the NHS GMS, as well seeking

involvement from ICBs, NHS regional teams, Pathology Networks, Health Innovation Networks, Cancer Networks, Primary Care Networks and Neighbourhood Health Services once established. It will also be important to ensure senior executive level multiprofessional leadership at this partnership board level to enable the transformative clinical change that needs to be delivered.

37. The purpose of this NHS GMS Partnership Board will be to drive geographical engagement and the strategic development of the NHS GMS within its geography, for the benefit of the patients and population that it serves. Given the level of clinical and patient and public involvement required to deliver all of the functions outlined in this outlined in the Specification it is recommended that a patient and public forum is established as well as a Clinical Senate. Figure 3 shows a high level overview of the NHS GMS geography governance structure.

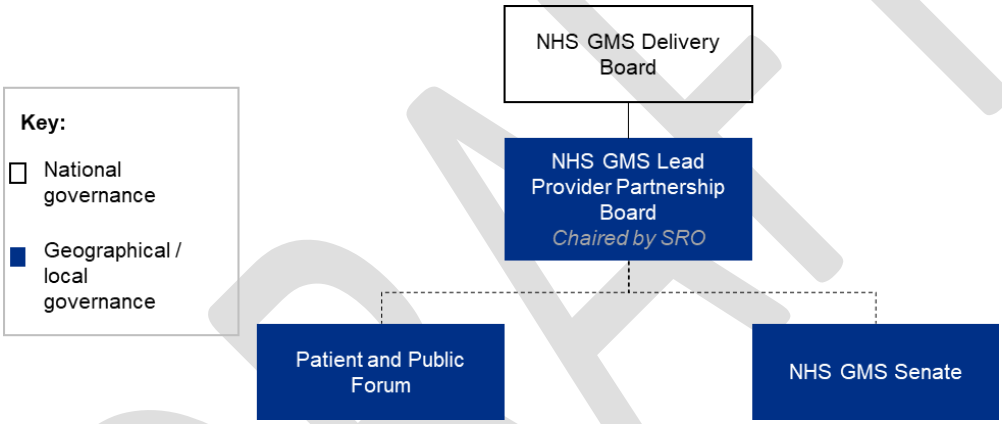


Figure 3 – High level NHS GMS geography governance structure

Assurance and contract monitoring

38. Each NHS GMS Lead Provider will be contractually managed and overseen by the Commissioner for the services that it is commissioned to provide to the NHS GMS. All NHS GMS Lead Providers will operate in accordance with the performance and assurance frameworks and maturity matrices set by the Commissioner.

39. Given the maximum duration of the contract is 10-years, and given the evolving scientific, technological and analytical advances, the requirements of the services set out this Specification may change during the term. Any changes to the requirements of the services set out in the Specification will be dealt with in accordance with the change provisions set out in the contract.

40. Each NHS GMS Lead Provider must develop and maintain a register for the Genomic Laboratory Testin Service function that includes:

- the specific testing inventory that is provided by the NHS GMS Lead Provider;
- the technical platforms and methods that the NHS GMS Lead Provider deploys in respect of the laboratory testing services that it is commissioned to deliver, including the utilisation and throughput; and
- the interpretation services utilised to analyse, interpret and report test results the NHS GMS Lead Provider is commissioned to deliver.

41. Each NHS GMS Lead Provider will use the maturity matrices set by the Commissioner to self-assess its performance against the key functions outlined in the Specification, including governance, leadership, operational management, quality, data and digital, people and research and innovation. This process will support the Commissioner in identifying and reducing any variation of delivery between the NHS GMS Lead Providers, ensuring consistent and high-quality service delivery.

42. Commissioning Intentions and delivery priorities will be set by the Commissioner through the publication of annual NHS GMS Planning Guidance. Each NHS GMS Lead Provider will be accountable to the Commissioner under the Governance and Assurance Framework set out in the Lead Provider Contract. This framework will govern and oversee all functions of the NHS GMS in their entirety.

43. Each NHS GMS Lead Provider will be required to operate under the specific governance arrangements set out by the Commissioner (including under each contract) to ensure that a suitable lead or senior representative attends Board meetings and national oversight groups, which may also include representatives from other key organisations where appropriate as well as any national learning events. If the NHS GMS Lead Provider does not meet this requirement, the Commissioner will escalate this issue through the appropriate contractual route, such as at a quarterly assurance meeting, where the infrastructure funding provided to the NHS GMS Lead Provider may be revised in response by the Commissioner.

44. Each NHS GMS Lead Provider shall be accountable to the Commissioner in accordance with this Specification, and without prejudice to the Commissioner's audit rights set out in the NHS GMS Lead Provider contract, each NHS GMS Lead Provider shall allow the Commissioner (or a third party acting on its behalf) to carry out inspections or assessments of the Services (or any part(s) thereof), including for example the NHS genomic laboratories, including those of its third-party contractors, premises, equipment, and Standard Operating Procedures (SOPs) and processes and their compliance thereto, as the Commissioner deems necessary on an ad-hoc basis.

45. Each NHS GMS Lead Provider will also be expected to report to the Commissioner any activities involving third parties (including those from overseas and research initiatives) that make use of the NHS GMS infrastructure and workforce or are associated with the delivery of the functions set out in this Specification. This is to ensure that the impact on the operational delivery of the NHS GMS is fully understood by the Commissioner. Such activities may include testing, science, research and innovation and data and digital initiatives.

Working with the NHS system

Interdependencies with other Services and organisations

46. It is anticipated that each NHS GMS Lead Provider will have a number of interdependencies with other services and organisations, including without limitation, by way of example:

- clinical networks, for example Cancer Alliances, Pathology Networks and Primary Care networks;
- other NHS GMS Lead Providers;
- a broad range of clinical specialties across the care continuum that use or are dependent on the NHS GMS, from Primary Care (inclusive of Neighbourhood Health Services), through to Secondary and Tertiary Care;
- Clinical Genetics Services; and
- science, research and innovation partners, including for example Genomics England, Our Future Health, Health Innovation Networks and others.

Sub-contracting arrangements

47. Each NHS GMS Lead Provider shall enter into sub-contracts with third-parties (as appropriate) to provide the services set out in this Specification to the NHS GMS. Prior to entering into such sub-contract, each third-party sub-contractor proposed by a NHS GMS Lead Provider must receive prior written approval from the Commissioner. Third-party sub-contracts must be based on the NHS template Sub-Contract (without material amendments) and include a defined scope of work, and provisions in relation to governance, oversight, risk and liability, payment terms, duration, termination clauses and performance standards (including service levels/ key performance indicators and quality indicators).

48. Each NHS GMS Lead Provider must develop and maintain a register that lists each third-party commissioned by the it to provide the services set out in this Specification to the Commissioner. The register must include for any NHS Genomic Laboratory Services delivered by third-parties:

- the specific testing inventory that each third party is commissioned to deliver for which they must have the capability and expertise to perform;
- the technical platforms and methods that each third party must deploy in respect of the laboratory testing services that it is commissioned to deliver, including the utilisation and throughput; and
- the interpretation services and clinical reports that each third party is commissioned to deliver.

49. The Commissioner will perform an annual audit of the register with the NHS GMS Lead Provider.

50. Each NHS GMS Lead Provider shall comply with and be able to demonstrate its compliance and that of its third-party contractors, to all standard operational procedures and service operational plans referred to in this Specification.

NHS Genomic Laboratory Services

1. This Specification sets out the requirements for genomic laboratory services which must be delivered by the NHS GMS Lead Provider in accordance with the following principles:
   - deliver the genomic testing repertoire outlined in the Test Directory to the published turnaround times and in cancer work with Cellular Pathology to deliver clinically relevant end to end cancer diagnostic turnaround times;
   - standardise the approach to genomic testing, interpretation and analysis within and across the NHS GMS network;
   - optimise the strategic approach to genomic testing and remove duplication;
   - streamline end-to-end workflows to improve efficiencies and ensure a cost-effective approach to delivery, benchmarking against other laboratories in the NHS GMS network;
   - operate demand and capacity management and best practice waiting list management and an ongoing and auditable review of diagnostic outcomes
   - provide PLCM data on time and in the structured format required
   - expand the use of genomic data to support access to clinical trials, and data for research and innovation;
   - maintain and deliver a high-quality service including the funded participation in EQA schemes covering the whole testing repertoire; and
   - be agile and rapidly respond to evidence, innovation and the adoption of new technologies.

National Genomic Test Directory

2. Each NHS GMS will continue to deliver the genomic testing offer as set out in the Test Directory. Insights from service delivery should be used to inform proposals for Test Directory amendments through the Test Directory Test Evaluation Process, including the review of gene panel content via PanelApp.

3. The scope of the Test Directory includes:
   - **Cancer**: genomic testing for over 200 cancers, with Clinical Indications across both solid tumour and haematological malignancy where the cancer genomic testing offer will continue to expand over time. This includes somatic (and where relevant paired germline) genomic testing in cancer settings for diagnostic purposes. The scope of diagnostics where genomic findings may be actionable in clinical care includes supporting clinical diagnosis; as part of treatment or surgical decisions; for access to precision medicines and assessing response and acquired resistance mechanisms or as part of conferring eligibility to clinical trials; and for pharmacogenomic use and dose modification of chemotherapy.

- **Rare diseases**: genomic testing for over 7,000 conditions, with a known genetic association, which will expand over time. This includes pre and postnatal diagnostic genomic testing across the range of monogenic and chromosomal disorders, family member testing for known causative variants to predict disease or to support reproductive decision making, and prenatal diagnosis including non-invasive prenatal diagnosis (NIPD) where there is a known genetic cause in the family. Achieving a genomically confirmed diagnosis will support access to treatments and interventions including ATMPs and/or clinical trials.
- **Population Health, inclusive of inherited common diseases and pharmacogenomics:** this will utilise constitutional genomic information in more common disorders to direct clinical care through inherited risk stratification, for example to enable pre-symptomatic screening for inherited breast cancer predisposition or identify risk of cardiovascular disease such as Familial Hypercholesteremia (FH) and as evidence evolves the use of PRS coupled with other diagnostic information to inform predictive and population health approaches. Genomics will be used as part of screening pathways, for example through non-invasive prenatal testing (NIPT) for common aneuploidies. Finally, pre-emptive and / or reactive pharmacogenomic testing that analyses an individual's genome to understand how it influences how they respond to medicines and whether dosing needs to be adjusted or a different medicine identified to reduce adverse drug reactions (estimated to cost the NHS £2.21 billion annually).

Laboratory operating model

4. The genomic testing provision delivered by the NHS Genomic Laboratory Service is categorised as:
   - tests delivered by NHS Genomic Laboratory Services in all NHS GMS geographies; and
   - tests delivered by a National Designated Laboratory (fewer than seven laboratories) including specialist tests. Specialist tests being those within specific clinical groupings (e.g. cardiology, ophthalmology or for certain cancer applications) that benefit, due to rarity and/or specialist knowledge, from delivery by a limited number of providers with expertise in that discipline, technology or test or to drive efficiency and productivity in testing strategy. The provision of whole genome sequencing (WGS) technology, circulating tumour DNA (ctDNA) testing and high volume testing for pharmacogenomics is included in this category to ensure an efficient and cost-effective service appropriate for the predicted activity volume.

5. The NHS GMS Lead Provider shall ensure compliance with the nationally agreed turnaround times as detailed in the Test Directory and align with thresholds set out in the contract. In addition for cancer the NHS GMS must work towards the

implementation of end-to-end cancer diagnostic turnaround times, working closely with Cellular Pathology in the NHS GMS geography. More information is set out in the cancer genomics clinical function and it is a requirement that the NHS Genomic Laboratory Service works in conjunction with this and all clinical functions.

6. The NHS Genomic Laboratory Service infrastructure in each NHS GMS must deliver the following operational requirements:
   - consolidation of wet laboratory functions for rare disease and genomic cancer testing to a maximum of two laboratory sites in total in each NHS GMS;
   - operate at least 6 days per week to meet required turnaround times as well as to deliver an affordable and efficient service;
   - be adaptable, flexible and scalable to be able to rapidly respond to the implementation of new or changing technologies or changing clinical requirements including demand for genomic testing;
   - incorporate clinical trial targets into routine testing and report in line with the nationally agreed protocols;
   - maximise the use of high throughput and cutting-edge testing technologies to deliver clinically meaningful nationally agreed turnaround times and drive efficiency, productivity and affordability;
   - streamline laboratory workflows, including end to end automation of processes from sample receipt (barcoding) to return of result to a clinician, to enable more efficient and effective working;
   - rapidly adopt new approaches to interpretation and analysis including enabling sharing of data across the NHS GMS to improve reference and knowledge derived databases to support genomic data analysis;
   - automated data analysis and reporting for tests where Clinical Scientist interpretation is not required, and to feed into EPRs where appropriate;
   - fully integrated LIMS and data flows across the NHS GMS geography to aid more efficient working;
   - support genomic multidisciplinary meetings;
   - operate a biobank function to support access to DNA, RNA and other samples from NHS GMS patients for research and service evaluation purposes. Further detail to be developed inclusive of operational and consent considerations; and
   - lead or contribute to research and innovation initiatives and to the ongoing development of the Genomic Laboratory service.

7. Each NHS GMS geography Genomic Laboratory Testing Service will consist of the following:
   - a single NHS Genomic Laboratory Hub (NHS GLH) where most of the wet laboratory work is taking place at one site and located at the NHS GMS Lead Provider;

- the NHS GLH will cover rare disease and cancer testing, however where these functions need to be separated due to expertise a single rare disease lead laboratory and a single cancer lead laboratory for the wet laboratory elements, including DNA extraction is acceptable. One of these lead laboratories will be defined as the NHS GLH. Additional wet laboratory sites will only be considered where it provides an efficient and cost-effective service underpinned by the appropriate expertise to be agreed by the Commissioner;
- Cellular Pathology Genomic Centres (CPGC) to prepare samples for genomic testing, enable digital image capture to a defined provider and deliver a small number of designated cancer genomic tests required to support rapid turnaround times or when samples are limited;
- NHS National Designated Genomic Laboratory (NDGL) will provide national services including WGS, rapid WGS for acutely unwell babies and children (R14), rapid sequencing for fetal anomalies (R21), ctDNA testing and minimal residual disease (MRD) testing. Additional laboratories will be designated, within a timeframe to be agreed, for high throughput pharmacogenomic testing and multi-omic testing. These designated laboratories may also be a rare disease lead laboratory, or a cancer lead laboratory; and
- Local Reporting and Coordinating Laboratory (LRCL) to provide distributed reporting expertise and oversee provision and quality management of local point of care testing, near patient testing and genomic tests delivered by CPGCs.

8. The NHS GLH and LRCL will be required to provide an oversight and coordination function to support the use of Point of Care or Near Patient Care Testing across the geography and oversee any designated genomic testing that is undertaken by CPGCs in the NHS GMS geography. This will include being accountable for quality management, providing oversight to ensure that there is appropriate expertise to undertake and analyse the genomic test and working with providers inclusive of Neighbourhood Health Services and others to support implementation.
9. The NHS GMS Lead Provider must drive consolidation to deliver the Genomic Laboratory Service model in full within the first two years of the contract. This includes stopping wet laboratory testing in laboratories that do not fit with the model and ensuring that the operating requirements are met. Any exceptions to the model will need to be approved in writing by the Commissioner. Exceptions will be considered where evidence can be provided on specific expertise being required for the wet laboratory work that cannot be consolidated, negative impact on patient pathway or turnaround times and negative impact on the efficiency and productivity of the Genomic Laboratory Service.

Genomic Testing delivery

Pre-analytical: Sample Collection, Handling and Processing

10. The NHS GMS Lead Provider must ensure that robust standard operating procedures are in place to support the end-to-end delivery of genomic testing, from sample receipt through to the reporting of results. This includes ensuring that:
    - NHS Genomic Laboratory Services operate in accordance with established Standard Operating Procedures and best practice guidance to ensure that all samples are accurately identified, of appropriate quality, and handled and stored correctly to meet the requirements of subsequent genomic testing and for biobanking purposes.
    - The NHS Genomic Laboratory Service is responsible for the management and tracking of all genomic test samples originating within the NHS GMS Geography, including primary care and Neighbourhood Services. The NHS Genomic Laboratory Service across an NHS GMS geography will integrate their Laboratory Information Management Systems (LIMS), including with the National Genomic Informatics Service (NGIS) Sample Tracking for WGS, and LIMS Integration Service via documented APIs. If the ordering entity has also incorporated the sample tracking and LIMS integration service, the NHS GLH will maintain the process of tracking initiated at the ordering entity.
    - Once a genomic test has been requested, the appropriate sample will be handled and prepared by the NHS Genomic Laboratory Service in accordance with standard operating procedures and best practice guidance to ensure it is suitable for generating a reportable result.
    - DNA and RNA extraction should be consolidated to the NHS GLH (plus the lead cancer or rare disease laboratory if different) and only performed elsewhere if defined as part of a National Designated Service or will support rapid turnaround time tests performed in CPGCs.
    - If onward referral of the sample is required for genomic testing at another laboratory, the NHS GLH will handle, prepare, and package the sample in accordance with standard operating procedures, the receiving laboratory's requirements, and relevant best practice guidance.

11. The NHS GMS Lead Provider must ensure that data is recorded on quality of received samples, as outlined below, and is available to be shared with the Commissioner if requested. The report shall include information relating to:
    - the perceived problem(s) with the unacceptable sample;
    - the names and locations of the requestor who provided the unacceptable samples, including identifying if such clinicians/requestors have a high error rate in relation to the samples they provide; and

- if appointed to act as a designated laboratory for a national service, include details of any unacceptable samples it has received from other NHS GMS geographies, so that the referring NHS GMS can identify the names and locations of the clinicians/requestors who have provided the unacceptable sample.

Analytical: Testing, technology and analytical platforms and capability, including bioinformatics

12. Each NHS Genomic Laboratory Service is required to adopt and implement the most efficient and cost-effective technology to deliver genomic testing as outlined in the Test Directory. This includes but is not limited to; automation, reducing the number of sequential tests in favour of broad testing techniques that cover a number of genomic variants and variant types in a single assay, consolidation of tests in line with laboratory operating model outlined in this Specification, utilising cutting edge and high throughput technology with maximum efficiency, implementing assays that support rapid adoption of changes to the Test Directory for futureproofing (e.g. WES versus targeted capture).

13. The NHS GMS is required to work as a national network to share knowledge and reduce duplication of software validation/verification (e.g. decision support tools), bioinformatic developments, variant classification (e.g. through submission to national/internation variant databases including NDRS (under the National Disease Registries Directions 2021), ClinVar (both germline and somatic variants with classifications), and other disease-specific registries. In line with Bioinformatics deliverables the NHS GMS will be required to develop nationally standardised approaches to recording and aggregating variant classification data as part of a national bioinformatics strategy that will be developed and must develop a local strategy for its implementation.

14. The NHS GMS bioinformatics strategy must prioritise data, such as variants detected and their clinical significance, being easily accessible to the whole NHS GMS. Bioinformatics databases and data repositories shall be considered essential NHS GMS infrastructure.

15. The NHS GMS is required to support bioinformatics service delivery, maintenance of bioinformatics resources, continual improvement of bioinformatics clinical utility, and ensuring that methods are standardised across the NHS, in line with priority three of the Accelerating Genomic Medicine strategy and in accordance to ACGS best practice and other appropriate guidelines and be focused on improving clinical utility and patient impact.

16. NHS GMS providers must ensure their workforce are competent to perform the tests they are responsible for and that high equitable standards are achieved across the whole NHS GMS network.

Post-analytical: Clinical interpretation, reporting and returning of results

17. Genomic test results should be interpreted and reported in accordance with relevant national best practice guidance as developed by professional bodies or Medical Royal Colleges unless otherwise stated or covered by guidance from the Commissioner.

18. NHS GMS providers must ensure their workforce are competent to analyse, interpret and report the tests they are responsible for and that high equitable standards are achieved across the whole NHS GMS network.

19. Each NHS GMS must collect and submit outcome data, in line with the PLCM specification and report this to the Commissioner on a regular basis as part of the NHS GMS Contract Management Framework.

20. In 2026, the Commissioner will work with the NHS GMS to develop standardised reporting requirement that then must be implemented by each NHS GMS.

21. NHS GMS Lead Providers should ensure the NHS Genomic Laboratories participate in relevant external quality assessment schemes as noted above including for reporting.

22. The requirements for bioinformatic capability is set out in the Data and Digital section of the Specification.

Referral pathways

23. Each NHS Genomic Laboratory Service shall create, implement, and maintain genomic test referral pathways that cover all providers in the whole NHS GMS geography and ensure that referrals comply with the; end to end clinical pathways, the Test Directory, genomic testing strategy (as outlined in this Specification) and national consent guidance.

24. Prior to accepting a request for testing the NHS GMS should ensure:
    - the Clinical Indication for the test requested is included within the Test Directory;
    - sufficient clinical information has been provided to ensure the patient meets the eligibility criteria, as defined in the Test Directory;
    - the most appropriate test has been requested and there is clinical utility to the test either for clinical management (determining therapeutic decisions and/or clinical

investigations and/or surveillance programmes; patient, parent or adult relative reproductive decision making; unaffected relatives seeking predictive testing;

- the referral is from an appropriate source and within the relevant NHS GMS geography;
- the minimum data has been supplied and submitted in accordance with data models associated with NGIS (currently for WGS centralised service), a function which will transfer to the Order Management System as it goes live and which may be for other genomic tests;
- where required, consent for sample acquisition, clinical testing and DNA and RNA storage should be covered via the genomic test request form and stored within the laboratory information management system, as well as recorded in the patients record;
- ensure that appropriate consent is in place including for sample biobank storage and/or research participation. It is anticipated that, over the course of this Specification, consent models for both clinical and research use will evolve toward digital solutions, encompassing research choices beyond WGS. The NHS GMS must therefore remain responsive and adapt to emerging consent processes in line with national developments.

25. For tests delivered by a National Designated Laboratory, each NHS GMS is responsible for ensuring that relevant information and samples are sent on to the designated providers for that test in the relevant geography.
26. The NHS GMS Lead Provider must audit the performance of all providers in the NHS GMS geography in relation to meeting the above obligations, and react through pathway changes and/or education and training where these audits demonstrate sub-optimal referral pathways with poor diagnostic yield/outcomes or that don't meet quality requirements.

Cellular Pathology Genomic Centres

27. CPGCs create 'one stop' testing laboratories to deliver sample preparation for genomic testing plus Cellular Pathology based tests eg immunohistochemistry for diagnosis, prognosis and treatment. CPGCs optimise workflows and decrease turnaround times through collaborative working with Cellular Pathology (including histopathology). They identify areas of unmet need and, where appropriate support the expansion and repatriation of genomic testing activity to the NHS GMS. CPGCs will also support the ability to monitor performance of the end to end cancer genomic testing pathway including audit and quality improvement.

28. The NHS GMS Lead Provider must ensure CPGCs are commissioned and operating in line with the Specification set out in **Annex 2**. Once defined by the Commissioner,

CPGCs will be commissioned to undertake a defined list of cancer genomic tests requiring rapid turnaround times under the oversight of the NHS GLH or LRCL.

Quality and oversight requirements

29. The NHS GMS Lead Provider will be responsible for ensuring quality, efficiency and standardisation in testing, analysis, interpretation and reporting across the genomic testing function of the NHS GMS geography. This includes:

- defining and implementing requirements for collection, receipt, handling, and processing of all sample types for genomic tests listed in the Test Directory, ensuring compliance with specified standards for labelling, transport, storage, and test-specific and locally developed protocols;
- delivering the Test Directory by ISO 15189:2022 accreditation and ensure continued UKAS accreditation, and promptly notifying the Commissioner and all service users of any change in accreditation status;
- defining the operating model and quality requirements according to ISO 15189:2022 standards for genomic point of care and near patient testing being delivered across NHS GMS geography and for any CPGCs undertaking cancer genomic testing;
- compliance with relevant standard operating procedures, current and future national guidance from the Commissioner and best practice guidance from professional bodies where relevant;
- responsibility for the ongoing monitoring, review, and audit of processes related to the standard operating procedures and service operational plans, ensuring their maintenance and compliance throughout the duration of the Term;
- participation in audits when required by the Commissioner;
- regularly reviewing diagnostic outcomes and benchmarking in accordance with the requirements of the commissioner to ensure clinical utility of genomic testing services;
- ensuring that all laboratories in the NHS Genomic Laboratory Service within the geography participate and consistently demonstrate satisfactory performance in all relevant External Quality Assessment (EQA) schemes (including pilots) funded by the commissioner and appropriate to the testing performed. This includes, but is not limited to, UK NEQAS and GenQA schemes covering DNA extraction from blood, tissue, and saliva; cfDNA extraction in both somatic and prenatal contexts; DNA quantification; histopathological tumour assessment; and variant analysis and classification;
- agreeing to the disclosure of EQA outcomes to the commissioner by the EQA provider, including performance monitoring;
- supporting service evaluation and service improvement linked with the Transformation and Service Improvement enabler; and
- ensure compliance with national output specifications as defined under the Patient Level Contract Monitoring (PLCM) standard (DCB3003) including outcome data, in

accordance with the NHS Standard Contract (as amended) and associated technical and user guidance.

30. Each NHS GMS Lead Provider will be required to collaborate and share information with the commissioner and NHS GMS National Network in order to:
    - ensure sharing of best practice and lessons learned;
    - achieve service improvements;
    - achieve optimisation of processes;
    - provide a conduit for discussions on aspects of the expansion of the NHS GMS; and
    - agree testing priorities across the NHS GMS National Network.

Genomic technologies

31. To deliver the test repertoire requirements outlined in the Test Directory, each NHS Genomic Laboratory Service must either possess or have access to a broad range of genomic technologies.

Sample Types

32. Genomic laboratory testing repertoires should include UKAS accredited processes for; samples handling and DNA extraction from EDTA blood (including from capillary home collection kits), fresh tissue (including frozen or an approved tissue stabiliser), FFPE tissue, bone marrow, saliva, buccal swabs, amniotic fluid, chorionic villus, cultured cells, urine (where relevant), cytology samples including fluids; RNA extraction from blood, fresh tissue (including frozen or in a tissue stabiliser), FFPE tissue, bone marrow or cultured cells, cytology samples including fluids; and cell culture capability to support cytogenetic preparations and DNA/RNA extraction.

DNA and RNA extraction

33. Each NHS Genomic Laboratory Testing Service must be equipped to perform DNA and RNA extraction from the full range of clinically relevant sample types, as outlined in this Specification. These processes must be conducted in accordance with UKAS ISO 15189:2022 accredited protocols to ensure clinical validity and regulatory compliance.

34. Where feasible, laboratories should implement high-throughput and automated extraction technologies to optimise workflow efficiency and support timely service delivery. Automation should be leveraged to reduce manual handling, improve consistency, and enhance scalability.

35. The quantity and quality of extracted nucleic acids must meet the specific requirements for downstream genomic analyses, as defined in the local SOPs for UKAS-accredited tests.

Whole genome sequencing (short read)

36. Provides a comprehensive view of the whole genome and has utility in complex or undiagnosed conditions.

37. In Rare disease WGS will be used in cases with broad and overlapping clinical features caused by genetic variation in multiple genes, affecting coding and non-coding regions and/or multiple variant types (e.g. small variants, copy number variants, short tandem repeat expansions). A limited number of virtual gene panels that are broad in gene to phenotype association, will be used for analysis in the majority of cases A phenotype driven analysis strategy approach may be adopted in the future for some cases, that is agnostic of gene panels. This testing and analysis approach minimizes the requirement for multiple genetic tests (in parallel or sequentially) and reduces the case-by-case reanalysis request burden to provide comprehensive and timely diagnostic testing. Standardisation of data and comprehensive primarily analysis enables automated systematic reanalysis on an ongoing basis to make use of evolving knowledge.

38. WGS offers a transformative approach to cancer (HaemOnc and Solid Tumour) diagnostics by analysing both the tumour genome (from a validated sample type) and the matched germline sample. This dual analysis enables the identification of tumour-specific genomic variants, providing a comprehensive view of the tumour's molecular landscape, and allows analysis of specific germline genes associated with cancer predisposition. WGS supports tumour characterisation, aids in diagnosis and classification, and identifies actionable variants that can inform access to precision therapies or direct patient management. Additionally, it can help predict prognosis and treatment response. In certain cancer types, WGS has the potential to replace multiple targeted assays, thereby streamlining testing pathways, optimising the use of limited patient samples and ensures wise use of NHS funding.

39. Further analysis of the whole genome can also support the return of pharmacogenomic and additional findings for both patients and their families. The utility of this application will be explored with the NHS GMS and guidance developed.

Long Read Sequencing (LRS)

40. LRS offers an alternative to short-read sequencing for whole genome analysis. Unlike short-read approaches, LRS provides several key advantages:
    - Enhanced detection of structural variants, repeat expansions, and complex genomic rearrangements.
    - Direct detection of DNA methylation without the need for chemical modification or bisulfite conversion.

- Longer read lengths, enabling improved phasing and haplotype resolution, which is critical for accurate variant interpretation.
- Rapid turnaround time
- No requirement to batch samples

41. LRS may provide the opportunity to consolidate multiple genomic tests into a single, comprehensive assay—streamlining diagnostics and optimising resource use.

42. In the cancer setting, NHS GMS Pathfinder sites, in collaboration with Genomics England (GEL), have piloted the use of LRS in both solid tumour and haematological malignancy samples.

43. The next phase of development will focus on validating LRS workflows against UKAS ISO 15189:2022 standards, with the goal of implementing LRS-based whole genome sequencing for clinically relevant cancers across the NHS GMS.

44. Additionally, adaptive LRS is being evaluated in the intraoperative setting for central nervous system (CNS) cancers. This approach enables rapid sequencing and analysis during surgery, supporting real-time diagnostic decision-making and guiding surgical interventions.

45. In germline testing for rare and inherited disease LRS will be utilised initially as an additional test where required to phase recessive variants and resolve complex structural rearrangements. But alternative applications for targeted and whole genome LRS will be explored, where LRS offers significant benefit over short read sequencing.

Whole exome sequencing (WES)

46. In Rare disease, whilst it remains most cost effective over WGS, WES with virtual gene panel analysis will be applied to Clinical Indications that are clinically more specific and associated with a smaller number of genes or defined group of diseases. The use of WES will replace targeted NGS capture assays to enable rapid and cost-effective adoption of new gene associations into diagnostic testing. NHS GMS wide adoption of WES assays will support automated reanalysis, future diagnostic discovery and research through cohort wide analysis, as well as provide relevant data for proactive pharmacogenomic testing, additional (looked for) findings, and evidence towards any future adoption of PRS.

47. For cancer, WES offers a more comprehensive approach than targeted NGS panels, though it is not as extensive as WGS. Compared to WGS, WES is more cost-effective while still capturing the most clinically relevant variants. It is particularly effective in

detecting somatic alterations such as single nucleotide variants (SNVs), small insertions and deletions (indels), and copy number variations (CNVs), all of which are critical for cancer diagnosis, classification, and treatment planning. A key advantage of WES is its compatibility with formalin-fixed, paraffin-embedded (FFPE) tissue samples, making it a practical and accessible option for analysing solid tumours in routine clinical settings.

Targeted NGS Panels

48. Targeted next generation sequencing panels are to be used where the number of test targets is small and/or relatively stable but enables cost effective testing at scale. This approach could be used in high throughput population health testing, for example in pharmacogenomics or BRCA1/2 testing.

Circulating tumour DNA

49. The science of detecting circulating tumour DNA (ctDNA) has progressed rapidly over the past decade. Increasingly it is becoming possible to use blood samples to test the circulating free DNA (cfDNA) for the presence of disease-causing variants also known as liquid biopsy testing and ctDNA based testing has now been validated, pan-cancer, in several different clinical trials. ctDNA testing for precision medicine targets in non-small cell lung cancer and breast cancer are already included on the Test Directory.

50. There are four areas where ctDNA could be of significant benefit to the delivery of the cancer commitments in the NHS 10 Year Health Plan and support ambitions expected in the National Cancer Plan:
    - screening asymptomatic populations;
    - routes to diagnosis when there are difficulties in obtaining solid tumour biopsies;
    - rapid selection of therapy to gain a comprehensive view and fully characterise the profile of the tumour to inform precision treatment.
    - Disease monitoring

51. The use of ctDNA testing in non-small cell lung cancer has demonstrated reduced requirements for other diagnostic interventions for removing sample preparation from histopathology and requirements for biopsy or other diagnostic interventions, with the potential to deliver £11m net impact to patients across the healthcare system.

52. The NHS GMS will work with the Commissioner and to continue to develop and review the evidence for the roll out ctDNA testing in the NHS in line with a national implementation plan.

Copy number variants

53. The detection of copy number variants should be included in the design and analysis of all next generation sequencing approaches with additional assays (e.g. MLPA, microarray) performed in parallel only when the genomic architecture of the region/gene to be tested prevents routine copy number variant analysis from sequencing data. Specific assays for the detection of copy number variants (at exon level resolution) should be limited to confirmatory testing or following case specific poor quality or failed CNV analysis result from NGS, unless otherwise stated in the Test Directory. However, in cancer, certain clinical scenarios—such as when a rapid result is required or when there is insufficient material for an NGS panel—fluorescence in situ hybridisation (FISH)-based tests for CNVs may still be preferred due to their speed and minimal sample requirements however the use will be closely monitored and should be audited regularly.

Microarray (including SNParray and methylation microarray).

54. Rare disease microarray testing as a first line test for the genetic diagnosis of rare diseases will be limited to syndromic presentations likely to be of a chromosomal nature. Microarray testing should not be used as a substitute for comprehensive WGS testing where smaller variants are equally or more likely causative than chromosomal variants.

55. In the Cancer the primary application of microarray technology is to detect genome-wide CNVs, such as gains, amplifications and deletions. These findings can inform diagnosis, prognosis, patient management and treatment decisions. In the context of ALL, the SNParray results are mandatory for the ALLTogether trial. When DNA methylation status is also a consideration, methylation microarrays can be employed. These platforms provide both CNV and methylation data from a single assay, offering a more comprehensive molecular profile. This has become particularly important in the classification and diagnosis of central nervous system (CNS) tumours, where methylation signatures can significantly enhance diagnostic accuracy.

Karyotype

56. Karyotype or equivalent assays will primarily be used to detect chromosomal structural variants, genome-wide but at low level resolution and in clinical situations defined in the Test Directory.

57. In cancer, particularly in HaemOnc, karyotyping plays a critical role in identifying chromosomal abnormalities such as translocations, inversions, deletions, duplications and aneuploidies, many of which are characteristic of specific cancer subtypes. This

cytogenetic information is essential for accurate diagnosis and risk stratification, as well as for informing prognosis and guiding clinical management and treatment decisions.

Targeted Genomic Testing

58. A range of technologies are available to support targeted or region-specific genomic assays in cancer, and rare disease and population health contexts. These assays are designed to detect a variety of genomic alterations, including single nucleotide variants (SNVs), structural variants, targeted chromosomal abnormalities, specific exon deletions or duplications, short tandem repeat (STR) expansions, imprinted regions, and methylation status.

59. The Test Directory outlines multiple methodologies that fall under the umbrella of targeted testing. These include PCR and RT-PCR-based assays, as well as fluorescence in situ hybridisation (FISH), among others. The selection of the appropriate technology should be guided by the nature of the genomic target, ensuring that the approach is accurate, cost-effective, and capable of delivering results within defined turnaround times.

Multi-Omics

60. A multi-omic approach integrates data from various ‘omics’, e.g. genomics, transcriptomics, proteomics and epigenomics, from a single patient, enabling a more comprehensive understanding of the link between molecular profiles and clinical presentation. This has significant clinical utility in both cancer and rare disease.

61. In cancer, multi-omic profiling enhances understanding of clinical subtypes, supports better treatment stratification, reveals resistance mechanisms, and helps optimise therapeutic responses. To enable a multi-omic approach there will be a requirement to expand the testing repertoire to include whole transcriptome sequencing and to link this with WGS/WES data, developing bioinformatic pipelines to support this. This will be established in the first instance through a Nationally designated provider (to be designated). Wider data sources which may sit outside of the NHS GMS including proteomic and metabolomic data may also need to be built into to any multi-omic approach in the future.

62. As testing for rare disease expands to include more genes and regions, the number of variants of uncertain significance increases. Improving diagnostic yield requires additional evidence to assess variant causality, often necessitating multi-omic data such as transcriptomic analysis and insights into protein function and biological pathways. In some instances, transcriptomics may be an appropriate alternative to DNA sequencing, so different testing approaches will be explored. The commissioning

of Nationally designated provider(s) to provide alternative testing approaches tailored to meet the needs of individual cases, will be explored.

63. In 2026, a multi-omic service specification will be developed by the commissioner in partnership with the NHS GMS network to meet the needs of the NHS GMS. To inform this each NHS GMS Lead Provider are requested to set out a proposed strategy to effectively use a multi-omic approach, that applies different testing methodologies, to generate information required to support diagnosis or identification of treatment options inclusive of access to clinical trials.

Genomic Point-of-Care and Near-Patient Testing

64. There is a growing requirement to identify genomic point-of-care tests (POCTs) and near-patient testing solutions that can rapidly and accurately analyse genetic variation and determine their clinical appropriateness across healthcare settings. These technologies should be deployable without the need for specialist genomic expertise, enabling timely diagnostic, prognostic, and therapeutic decision-making in a wide range of clinical contexts.

65. A variety of technologies are either available or emerging for use closer to the patient. Potential examples where this maybe applicable include:
    - Pharmacogenomic POCT to assess the risk of aminoglycoside-induced deafness in neonates in an emergency setting.
    - Intraoperative adaptive long-read sequencing (LRS) to support real-time tumour classification and guide surgical decision-making.

66. All such technologies must be CE/UKCA marked and approved for clinical use within the relevant timeframe. Furthermore, all POCT and near-patient tests must undergo rigorous validation and verification for the specific clinical use case in which they are deployed.

67. The commissioner will work collaboratively to develop a strategic approach for the implementation, governance, and evaluation of POCTs, ensuring they are clinically validated, cost-effective, and aligned with national genomic testing standards. If approved for use, these will fall under the quality management service of the respective NHS GMS geography.

Whole genome sequencing delivery model

68. The sequencing function of the current centralised WGS service is delivered through a third-party provider. This will continue between April 2026 and October 2026, but over that time period will transition to a de-centralised sequencing model, with the

centralised model ceasing at the end of October 2026. From April 2026 short-read WGS will begin to be delivered by Nationally Designated NHS GMS WGS laboratories. There will be a mixed operating model of the existing centralised WGS provider and Nationally designated NHS GMS laboratories providing sequencing between April 2026 and October 2026. Over the 7 months transition period, and in line with a nationally defined plan, each NHS GMS geography will switch from sending extracted DNA to the Plater to sending samples to the Nationally designated NHS GMS WGS laboratories. All WGS will be delivered from within the NHS by 31st October 2026.

Centralised Whole Genome Sequencing (April – October 2026)

69. This model involves submission of patient clinical and demographic data into the National Genomic Information System (NGIS) provided by Genomics England by each requesting NHS GMS and transportation of DNA samples by those laboratories to a Single Plating Laboratory at Birmingham Women's and Children's Hospital as part of the Central and South NHS GLH. Samples are transferred to plates and dispatched to the centralised sequencing provider. Genomics England provide bioinformatics analysis and processed data is returned to the NHS GMS for clinical interpretation and reporting via the NGIS portal.

Decentralised Whole Genome Sequencing (from April 2026)

70. This model utilises NovaSeq X Plus technology, distributed sequencing infrastructure, and centralised bioinformatics to enable efficient, cost effective WGS at scale.

71. This model will balance the existing specialist expertise in the system to ensure high quality service delivery whilst creating an affordable and efficient service that can deliver the required turnaround times. The number of NovaSeqX Plus sequencing machines required will be based on expected volumes of WGS tests for rare disease, inclusive of the existing WGS service for acutely unwell children (R14) and the rapid prenatal sequencing service (R21) which will transition to WGSI (in line with the approach developed by the NHS Genomic Prenatal Network of Excellence), and for cancer, both solid tumours and haematological malignancies, including rapid WGS approach for haematological malignancies developed through the NHS Genomic HaemOnc Network of Excellence.

72. The capacity of each NovaSeqX Plus machine will need to be used to maximum efficiency to enable the service to be cost effective. This needs to be considered as part of each NHS GMS broader testing strategy and consolidation arrangements.

73. The will perform DNA extraction services for WGS samples prior to sequencing being undertaken by the Nationally designated NHS GMS WGS laboratory, unless otherwise instructed for specific services (e.g. Rapid HeamOnc WGS or R14).

74. With the exception of specialist national WGS testing services (e.g. R14 and R21), the nationally designated NHS WGS Laboratories will transfer WGS data to Genomics England for bioinformatic analysis who in turn will return processed WGS data to the NHS GMS for analysis, interpretation and reporting, Where specialist analysis is designated the data will be routed by Genomics England accordingly.

75. For specialist nationally designated services delivered by WGS technology (e.g. R14 and R21) bioinformatic analysis, interpretation and reporting will be delivered by the Nationally Designated Provider(s) until such time that a centralised national data storage and bioinformatic pipeline is in place.

76. The nationally designated NHS WGS Laboratories shall provide data regarding, activity and referrals, sample quality issues, library preparation and sequencing failure rates and pathway issues at a frequency defined by the Commissioner though the Contract Management Framework and this will be shared with the NHS GMS National Network.

77. From April 2026 analysis and interpretation of short-read WGS analysis will start to be developed to evaluate incorporating the identification and reporting of pharmacogenomic targets and additional findings. A policy will be developed by the Commissioner, supported by the NHS GMS and Genomics England.

78. The NHS GMS will perform reanalysis in line with the NHS GMS Reanalysis Policy for Rare Disease.

79. The NHS GMS Lead Provider shall provide the WGS Testing services in line with the whole genome sequencing specification as set out in **Annex 3**.

Long Read Sequencing in NHS GMS

80. Long-read sequencing (LRS) offers a transformative alternative to short-read sequencing for whole genome analysis.

81. The distinct operational and clinical characteristics of long-read sequencing (LRS)—including its potential use in intraoperative settings—may favour a distributed delivery model over the current centralised approach used for short-read whole genome sequencing (WGS).

82. As such, the implementation of LRS within the NHS Genomic Medicine Service may require a re-evaluation of infrastructure, logistics, and workforce planning to support a more distributed model. This would ensure that the benefits of LRS—such as real-time diagnostics and streamlined testing pathways—can be fully realised across diverse clinical settings.

83. The commissioner will work collaboratively to develop a strategic approach for the evaluation, implementation and governance of LRS, with consideration given to its use in an intraoperative setting.

Decision Support Services

84. From April 2026 NHS Genomic Laboratories should put in place appropriate Decision Support tools to support the relevant analytical services they are commissioned to deliver.

85. For centralised WGS, current arrangements for Decision Support through Genomics England will remain.

86. In 2026, the commissioner will work with the NHS GMS to set out a national procurement strategy for Decision Support services with the vision being the provision of a number of tools available through cloud infrastructure to support the ability to process both WGS and WES data in the same way but flexible to different service requirements and in an affordable nationally coordinated approach.

Scientific expertise for genomic MDTs

87. The NHS GMS Lead Provider will ensure that a robust model for genomic multidisciplinary teams (MDT), including Molecular Tumour Advisory Boards, is put in place as required for all clinical areas covered by the National Genomic Test Directory.

88. The NHS Genomic Laboratory Testing Service will be responsible for ensuring appropriate scientific expertise is in place to support genomic MDTs either within the applicable parts of their NHS GMS geography or nationally where relevant to support genomic testing requests and interpretation of clinical actionability. Additional requirements on MDTs are set out under the rare disease, cancer and population health clinical functions of this Specification.

Testing being performed outside the NHS GMS scope

89. Each NHS GMS Lead Provider must develop and maintain a list outlining the genomic testing that is being undertaken within the NHS GMS geography that is out of scope of this Specification. This list must be provided on request by the Commissioner and as a

minimum annually. This includes but is not limited to genomic testing being provided for commercial endeavours, for private patients, the Devolved Nations or for other countries. The Commissioner will have no influence on this activity unless it is deemed to be having a negative impact on the capacity, timeliness or quality of the services being provided to support the NHS GMS.

Genomic testing strategy

90. Each NHS GMS Lead Provider will continue to be required to deliver testing as set out in the National Genomic Test Directory. In addition, this Specification sets out areas for development to deliver the most efficient and clinically appropriate testing for patients and the population.

91. The areas for delivery and development are set over three-time horizons. Requirements set out in 2026/2027 horizon will be required to be delivered by each NHS GMS. There is less certainty over the 2027/2028 – 2028/2029 and 2029/2023 onwards horizons and therefore these may be subject to change and continued development. Each NHS GMS will be required to work with the Commissioner to continue to evolve the genomic testing strategy.

Cancer genomic testing strategy

Children and young people cancer genomic testing

2026/27

92. From 1 April 2026 the NHS GMS Lead Provider will:

- Deliver the repertoire of tests outlined in the Test Directory, and where it is possible to use large next generation sequencing panels for solid cancers and haematological malignancies to maximise genomic output for a given patient.
- Deliver access to and WGS provision from the designated national provider and/or the centralised WGS service, comprehensive short-read WGS analysis, including piloting reporting of additional findings and pharmacogenomics findings, to be defined by guidance published by the Commissioner.
- Support evidence generation and planning for the introduction of rapid WGS in haematological malignancies and solid tumours.
- Provide analysis, interpretation and reporting of diagnostic discovery findings from research carried out through the National Genomic Research Library.
- Introduce new testing technologies for relevant cancers e.g. Long Read Sequencing (LRS) for Central Nervous System (CNS) tumours, intraoperative testing to inform surgical management.
- Deliver genomic testing to support access to precision medicines and clinical trials.

- Introduce new testing technologies in relevant cancer types e.g. Long Read Sequencing for Central Nervous System (CNS) tumours and RNA sequencing.
- Deliver pharmacogenomic testing where indicated in the Test Directory.

2027/28 – 2028/29 in addition to above:

- Where the research evidence indicates it is clinically impactful introduce ctDNA testing for patients to advance treatment options where their cancer has recurred or treatment has stopped working.
- Pending evidence generated from HaemOnc NHS Genomic Network of Excellence and transition to a distributed WGS service in 2026/27, deliver rapid WGS for haematological malignancies and solid tumour.
- Provide a comprehensive genomic analysis and molecular profiling including through the use of omic technologies to every CYP cancer patient to guide precision treatment decisions.

2029/30 – onwards in addition to above:

- Introduce genomic technologies that have been evaluated to improve clinical outcomes

Adult cancer genomic testing

2026/27

93. From 1 April 2026 the NHS GMS Lead Provider will:

- Deliver targeted testing and next generation sequencing panels for solid cancers and haematological malignancies as outlined in the Test Directory in line with clinically relevant end to end cancer diagnostic turnaround times.
- Deliver comprehensive short-read WGS analysis for defined solid tumours and develop and implement rapid WGS for haematological malignancies, including piloting the reporting of additional findings and pharmacogenomics findings, to be defined by guidance published by the Commissioner.
- Deliver comprehensive genomic profiling to improve cancer diagnosis and classify tumours and predisposing conditions.
- Provide analysis, interpretation and reporting of diagnostic discovery findings from research carried out through the National Genomic Research Library.
- Deliver genomic testing to support access to precision medicines and clinical trials as set out in the Test Directory or in guidance published by the Commissioner.
- Extend the use of ctDNA for other cancer indications (Cancer of Unknown Primary and others to be notified by the Commissioner.
- Introduce new testing technologies for relevant cancers e.g. Long Read Sequencing (LRS) for Central Nervous System (CNS) tumours, intraoperative testing to inform surgical management.

- Generate evidence on the use of Whole Exome Sequencing to inform decisions on the NHS GMS future testing strategy.
- For haematological malignancies, use cutting edge genomic technology to enable early signs of relapse prior to clinical deterioration or assessing depth of response for treatment de-escalation.
- Deliver pharmacogenomic testing to inform cancer medicines use as outlined in the Test Directory
- Work with Genomics England to deliver diagnostic discovery findings from WGS service

2027/28 – 2028/29

- Phased implementation of WES to replace large panels for cancers providing a more comprehensive genomic profile to inform patient management and access to clinical trials.
- Generate evidence on the use of Whole Transcriptome / RNA Sequencing to inform decisions on the NHS GMS future testing strategy.
- Expand and implement new uses of circulating biomarker testing, for example ctDNA, DNA methylation patterns, microRNA, circulating tumour cells, as well as other biomarker testing on a range of sample types (e.g. urine/saliva) and machine learning to support interpretation;
- Utilise integrated genomic (including MRD, ctDNA and multi-omic data), clinical and survivorship real-world data to inform personalised follow-up intensity, care planning and secondary prevention strategies.
- Provide a comprehensive genomic analysis and molecular profiling for every cancer patient to guide precision treatment decisions and access to clinical trials.
- For haematological malignancies, implement testing for pre-cancerous genetic alterations (CHIP / CCUS) in high-risk individuals with full blood count (FBC) monitoring and where appropriate / available early intervention trials. Use genomics to risk-stratify ‘pre-malignant’ lymphoid conditions.
- For haematological malignancies, implement enhanced MRD monitoring to escalate / de-escalate treatment as appropriate (optimising outcomes, reducing morbidity and saving money) and detect early relapse allowing earlier intervention.

2029/30 – onwards in addition to above

- Introduce genomic technologies that have been evaluated to improve clinical outcomes and expand use of WGS and WTS, other omics and ctDNA.

Rare disease testing and analysis strategy

94. The rare disease genomic testing offer covers three patient populations:

- Prenatal genomic testing

- Children and young people genomic testing; and
- Adult population genomic testing.

Prenatal genomic testing

95. Prenatal diagnosis includes testing for an unknown genetic cause initiated following the detection of fetal anomalies on an ultrasound scan, as well as testing for a known genetic condition, previously detected in a family member, through invasive or non-invasive prenatal sampling.

2026 – 2027

From 1 April 2026 the NHS GMS Lead Provider will:

- Continue to provide prenatal diagnosis as outlined in the Test Directory.
- Support the introduction of WGS for prenatal testing through transitioning the current R21 whole exome sequencing service to whole genome sequencing. The analysis will apply a fetal anomalies gene panel to detect genomic variants associated with structural fetal anomalies.
- Deliver non-invasive prenatal testing (NIPT) for common aneuploidies (for trisomies 13,18 and 21) in line with the NIPT Specification set out in **Annex 1**.

2027/28 – 2028/29 in addition to the above

- Deliver an expansion in the number of diseases tested by non-invasive prenatal diagnosis (NIPD), for example hemophilia.
- Reanalysis of appropriate prenatal data, where it is available, to inform post-natal diagnosis without the need for re-testing.
- Work with the Commissioner to explore future commissioning arrangements for preimplantation genetic testing for monogenic disorders (PGT-M).

2029/30 – onwards

- Deliver PGT for aneuploidy (PGT-A)
- Deliver testing to support in-utero intervention.
- Deliver multimodal testing to increase diagnostic yield, for example through optical genome mapping (OGM), long read sequencing, comparative RNA sequencing and cell-based non-invasive prenatal testing (cbNIPT).

Children and young people rare disease genomic testing

2026 – 2027

From 1 April 2026 the NHS GMS Lead Provider will:

- Continue to provide appropriate and relevant testing as outlined in the Test Directory.
- Provide analysis, interpretation and reporting of diagnostic discovery findings from research carried out through the National Genomic Research Library.

- Deliver testing to provide a rapid diagnosis of genetic conditions for acutely unwell babies and children including those on Neonatal Intensive Care Units (NICU) / Paediatric Intensive Care Unit (PICU), through rapid WGS.
- Support initiatives as they are developed and defined in the Test Directory to streamline WGS analysis approaches and reduce follow up analysis burden, e.g. single large and broad gene panel for all children and young people eligible for rare disease testing by WGS.
- Encourage and support the collection of parent/child Trio samples for WGS testing to improve diagnostic yield e.g. through use of capillary-based home blood collection kits.
- Adhere to national best practice guidance on the reporting of incidental findings including carrier status in children.
- Transition to deliver all non-WGS virtual gene panel analysis from whole exome sequencing data.
- Develop a strategy for the use of different technologies and multi-omics to increase diagnostic yield where a definitive diagnosis has not been achieved through short read WGS. This may include the use of long read sequencing, RNA sequencing, proteomics and DNA methylation.
- Support evidence generation and operational requirements for the reporting of pharmacogenomic targets and additional findings from WGS and WES data generated through rare disease testing.

2027/28 – 2028/29 in addition to the above

- Enable more timely and expanded genomic testing to support access to clinical trials and ATMPs.
- Support the ambitions outlined in the data and digital enabler to develop a lifetime source of genomic data, ensuring genomic data derived through newborn screening or childhood diagnostic testing is re-used rather than retesting the individual.
- Implement a multi-omics strategy to be defined by the Commissioner to increase diagnostic yield.
- Perform systemic automated ongoing reanalysis of genomic results, reflecting new research and discovery in rare disease and utilising new and emerging methods of analysis.
- Expand the genomic analysis offer to include additional findings (in parents of Trios) and pharmacogenomics, from WGS and WES data generated through rare disease testing, if evidence generated supports this.

2029/30 – onwards

- Introduce new genomic technologies and analysis tools that have been evaluated to improve clinical outcomes.

- Further expansion of testing offer via WGS and use of other omics.

Adult rare disease genomic testing

2026 – 2027

96. From 1 April 2026 the NHS GMS Lead Provider will:

- Continue to provide appropriate and relevant testing as outlined in the Test Directory.
- Provide analysis, interpretation and reporting of diagnostic discovery findings from research carried out through the National Genomic Research Library.
- Support initiatives as they are developed and defined in the Test Directory to streamline WGS analysis approaches and reduce follow up analysis burden, e.g. single broad gene panel for adult-onset neurological disorders tested by WGS.
- Adhere to national best practice guidance on the reporting of incidental findings in adult patients and parents.
- Transition to deliver all non-WGS virtual gene panel analysis from whole exome sequencing data.
- Support evidence generation and operational requirements for the reporting of additional findings and pharmacogenomic targets from WGS and WES data generated through rare disease testing.

2027/28 – 2028/29 in addition to above

- Enable more timely and expanded genomic testing to support access to clinical trials and ATMPs.
- Implement a multi-omics strategy to be defined by the Commissioner to increase diagnostic yield.
- Perform systemic automated ongoing reanalysis of genomic results, reflecting new research and discovery in rare disease and utilising new and emerging methods of analysis.
- Expand the genomic analysis offer to include additional findings and pharmacogenomics, from WGS and WES data generated through rare disease testing, if evidence generated supports this.

2029/30 – onwards in addition to above

- Introduce new genomic technologies and analysis tools that have been evaluated to improve clinical outcomes.
- Further expansion of testing offer via WGS and use of other omics.

Population health testing strategy

2026 – 2027

97. From 1 April 2026 the NHS GMS Lead Provider will:

- Identify individuals at high risk of adverse drug reactions and stratify treatments based on metabolic efficiency for selected gene / drug pairs.

- Prepare for pre-emptive pharmacogenomic testing, delivered through panel testing, in severe mental health conditions and other agreed conditions.
- Support evidence generation and develop the supporting infrastructure to pilot the return of pharmacogenomic and additional finding results for patients and their families receiving WGS and WES.
- Support evidence generation towards any future adoption of PRS by providing data obtained through WGS or WES.
- Work with clinical experts in the NHS GMS to explore how genomic testing for pharmacogenomic profiles can be integrated into the NHS over-40s Health Check - and over time, how this can become a universal offer.
- Deliver presymptomatic testing of high-risk individuals e.g. BRCA testing of family members and targeted populations.
- Expand genomic testing for inherited causes of common diseases to allow earlier detection and intervention, including cancer predisposition (e.g. BRCA1/2 genes, Lynch syndrome), and cardiovascular disease predisposition (e.g. Familial Hypercholesterolemia).
- Deliver cascade testing of high-risk family members/individuals to provide the choice to embark on preventative actions, for example surgery or screening.
- Begin integrating genomic insights into cardiovascular disease prevention and other agreed conditions through a trial with Our Future Health on implementing Integrated Risk Scores in Neighbourhood Health Services.
- Support the interpretation and reporting of condition suspected results in the Generation study including undertaking confirmatory testing of genomic variants, as required.

2027/28 – 2028/29 in addition to above

- Deliver a pharmacogenomics panel that covers existing testing, target groups where drug stratification is effective e.g. severe mental health treatment and cardiovascular disease.
- Provide testing for a range of genetic targets relevant to pharmacogenomics and the optimisation of medicines and reduction in adverse events, including HLA testing.
- Return pharmacogenomic and additional finding results for patients and their families receiving WGS and WES.
- Deliver multi-cancer early detection (MCED) tests if evidence supports this.
- Evaluate operational requirements to deliver testing, interpretation and reporting of polygenic risk scores following delivery of integrated risk scores by the Our Future Health programme and ongoing return of PRS results from the programme.
- Support the validation of results in the Adult Population Study undertaking confirmatory testing as required.

- Subject to the availability of capital funding, work towards centralised, automated, high throughput pharmacogenomic testing, requiring minimum scientific input.

2029/30 – onwards in addition to above

- Expand circulating biomarker testing as part of screening for a defined number of conditions.
- Subject to service evaluation, implement population-based polygenic risk scoring alongside other emerging diagnostic tools, enabling identification of and intervention for individuals at high risk of developing common diseases.
- Informed by the Generation Study, implement universal newborn genomic testing and adult population genomic testing.

Clinical Function

1. The clinical functions of the NHS GMS will deliver greater clinical and professional leadership to drive embedding of genomics in end-to-end clinical pathways equitably across the NHS GMS geography and develop the capacity and capability of the multiprofessional workforce.

2. The clinical function will be instrumental in building and maintaining relationships and engaging with key stakeholders across the NHS GMS geography including, for example, NHS regional teams, clinical leaders in all Trusts in the NHS GMS Lead Provider's Geography, including nursing and midwifery and pharmacy, Pathology Networks and other networks such as cardiovascular and primary care, Cancer Alliances, primary and community care and Neighbourhood Health Teams and patient and public representatives.

3. The Specification sets out the requirements that need to be delivered by all the clinical functions and specific requirements for each function.

Governance

4. For each clinical function appropriate governance should be put in place to support delivery and to regularly bring together the clinical leadership and expertise and ensure there is delivery and alignment across the NHS GMS geography. This will ensure that the clinical directors of these functions are able to appropriately represent the NHS GMS at the relevant national groups for these functions as outlined below. These national groups will build upon the example of the current cancer genomics clinical advisory group.

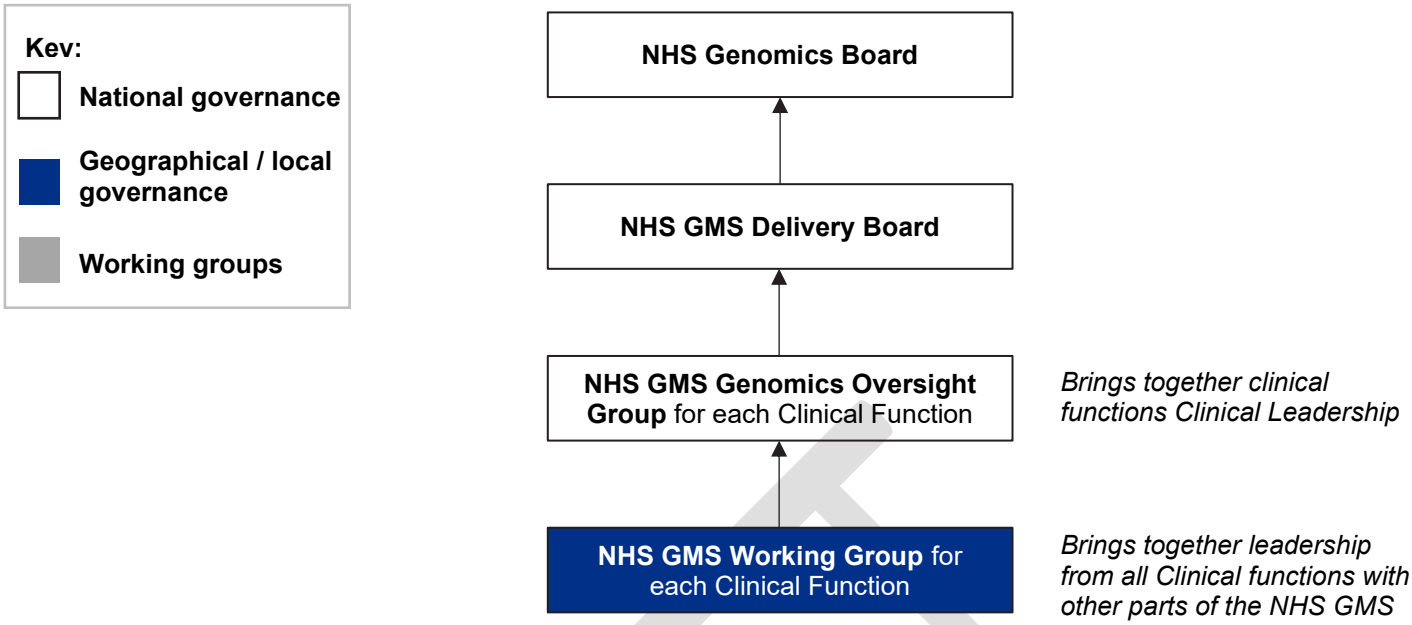


Figure 5 – Clinical functions governance overview

People

5. To deliver each of the clinical functions it is expected that the NHS GMS will appoint an overarching clinical director/s and include within the team multiprofessional clinical leaders with relevant genomic experience and expertise in a range of roles. A full list of these roles is included in **Annex 4**. A number of posts are mandated and must be put in place in each NHS GMS. For others there will be more flexibility to meet local arrangements and requirements, however the national delivery expectations remain the same of all NHS GMS geographies.

Requirements for all clinical functions

6. The following requirements will need to be delivered for all clinical functions and will build upon nationally agreed priority areas for the clinical function which would be reflected in annual planning guidance. This will be set out in an annual delivery plan to outline on how each of the core functions and roles will support delivery. This will be aligned with the following core principles for the clinical functions:
   - provide multiprofessional leadership for clinical change and drive service improvement in areas such as turnaround times;
   - raise awareness of genomics across the NHS workforce and support embedding of genomics into end-to-end clinical pathways inclusive of enabling access to precision medicines and ATMPs and to clinical trials;
   - develop new service models where appropriate;
   - support the education and training of the multidisciplinary workforce to embed a genomics first approach;

- reduce health inequalities and unwarranted variation;
- measure and monitor quality outcomes;
- enable genomic testing interpretation in the context of the clinical presentation;
- support population risk reduction and preventative healthcare; and
- work collaboratively with all functions of the NHS GMS Lead Provider through a matrix approach.

7. To support the delivery of the clinical functions each NHS GMS will map the provision of genomic services in each geography by provider of relevant clinical services, to understand where referrals are being made across the care continuum and for what clinical use. It is expected that this will include primary and community care and over time neighbourhood teams. Each NHS GMS will develop a dashboard – aligned to the national blueprint – to both monitor progress and highlight to providers within the geography, the role that the NHS GMS can play in supporting local clinical priorities, as well as where work is already underway.

8. The NHS GMS will ensure equitable access to and provision of genomic services across clinical pathways including testing services and access to research projects and initiatives and clinical trials. The NHS GMS will use all available national and local data sources, including those relevant to cancer, rare disease and population health and detailed intelligence on the key axes of health inequalities to identify barriers for access, drive strategic decision making and prioritise areas for action, either within clinical areas or for specific communities. The NHS GMS will monitor variation with a focus on outcome data and implement service improvement strategies. The NHS GMS will identify and advise on any gaps in existing data sources, including variation in the quality of data collection and data linkages, to provide an equitable and data-driven service. This will be reported regularly to the Commissioner as part of the Contract Management Framework and to the national clinical oversight groups

9. The NHS GMS will implement a multi-disciplinary approach. To embed a focus on clinical need and outcomes, the clinical function will work closely with the other cross cutting functions including education, service transformation, research and innovation and data and digital to ensure strategies are aligned to clinical service and patient requirements.

10. To support the delivery of a sustainable service, the NHS GMS will develop health economic modelling for the introduction and expansion of genomic services across clinical pathways, including where relevant the re-design of clinical pathways and service models, to build evidence for future investment in genomic services and drive strategic commissioning.

Cancer genomics

Background to the cancer genomics clinical function

11. The ambitions in the NHS 10 Year Health Plan and National Cancer Plan support a move to grow cancer genomic testing exponentially, as well as a drive to deliver a broader testing offer than now across the cancer patient journey as well as to approaches to earlier identification in populations with or without symptoms suggestive of cancer. The cancer genomics clinical function will support this, to bring together multi-professional leadership in cancer genomics to drive pathway improvements, inclusive of improving end-to-end cancer diagnostic turnaround times from tissue acquisition to genomic result, and the uptake of precision medicines and importantly the alignment of routine cancer genomic testing with eligibility for clinical trials. There will be a strong relationship and engagement with NHS pathology providers inclusive of pathology networks for solid tumours and with Specialist integrated haematological malignancy diagnostic services (SIHMDS) for haematological malignancies.

Clinical leadership

12. The clinical leadership team is responsible for ensuring that there is a comprehensive and timely cancer genomic testing offer in place for children and young people and adults in haemato-oncology and solid tumours and in the systematic application of cancer genomic testing across the end-to-end pathway from diagnosis to treatment stratification, disease monitoring and identification of eligibility for clinical trials.

13. Clinical leadership within the NHS GMS will be required to:

- raise awareness of genomic services across the whole cancer multidisciplinary team working with Cancer Alliances and all providers within the NHS GMS geography;
- understand unmet need and variation in equity of access to cancer genomic testing across the NHS GMS geography and recommend actions to address;
- inform the development of national standards and guidelines for cancer genomic pathways inclusive of their interpretation;
- identify transformation opportunities and deliver activities together with pathology and heamatological malignancy services to improve end to end cancer diagnostic turnaround times;
- enable the engagement and development of the multiprofessional workforce in cancer to drive equitable uptake of cancer genomics and the link with and equitable access to precision oncology medicines, as well as the use of pharmacogenomics;
- support the introduction of new technological advances across the care continuum for example circulating tumour DNA testing and research and innovation initiatives such as CVLP;

- inform the national decision making on clinical trial targets to be included as part of routine testing; and
- inform and work in a cross-cutting way with other functions of the NHS GMS in the geography.

Cancer Genomic Tumour Advisory Boards

14. Comprehensive analysis and interpretation of genomic information together with other diagnostic and clinical information through Genomic Tumour Advisory Boards is integral to the diagnosis and management of cancer patients, and supporting and enhancing working relationships with referring clinicians is crucial to maximising the use of genomics data in clinical decision making alongside other diagnostic disciplines.

15. This function will provide leadership and oversight to the establishment and delivery of Genomic Molecular Tumour Boards, and to the submission of data for multiple purposes including the collection of outcome data to demonstrate benefits and ensure the submission of data to registries and to for example variant databases to improve interpretation.

16. This function is required to establish a coordinated approach to the provision of Genomic Tumour Advisory Boards, working both at a geographical level and with other NHS GMS geographies when a national approach is agreed as appropriate. This will include local guidance being required to reflect national and international guidance and best practice.

17. This function will ensure that Genomics Tumour Advisory Boards are aligned with broader cancer MDTs to ensure clinical decisions are informed by genomic data and improve the timeliness of precision diagnosis and subsequent treatment decisions including early access to clinical trials to improve patient outcomes, including survival.

Alignment with Pathology

18. Over recent years there has been a significant increase in the requirement for both cellular pathology (including histopathology) inclusive of IHC and genomic analysis to support the diagnosis of cancer and through the identification of potential actionable targets to determine the appropriate approved treatment, often precision medicines, or access to clinical trials for cancer patients. This increase in demand has put pressure on services with demand outstripping capacity, most notably impacting workforce.

19. The National Cancer Board approved new end-to-end turnaround times for cancer patients in 2023 where the combined histopathology and genomic results are required by day 7 (urgent), day 14 (routine) or day 28 (for future clinical decision-making), from

the time of sample acquisition. Joint working is required across national pathology, genomics and cancer programmes to rapidly drive improvements.

20. This function will support the implementation of innovative service models and collaboration between Pathology Networks, Cellular Pathology Genomic Centres and NHS Genomic Laboratory Testing Service to streamline end to end cancer diagnostics pathways.

21. The NHS GMS will support pathway improvements and modifications, across the entire sample pathway, from the sample biopsy (or blood sample), transportation, histopathological or haematological diagnosis, request for genomic analysis, to the issue of a report to the treating clinical team. This will include the introduction of new technologies such as ctDNA and working with pathology to identify pathways where genomic tissue testing is no longer required.

22. The function will be required to provide a coherent and coordinated approach, and strategic clinical leadership, to the expansion and embedding of cancer genomics into clinical pathways, to the drive to improve turnaround times.

23. To gain a comprehensive view of an individual's diagnosis to inform their care the NHS GMS will coordinate the integration of genomic data with other diagnostic data, including seeking opportunities to align to the advances in digital pathology for example working with National Pathology Imaging Co-operative (NPIC) to evaluate the use of AI, to standardise images for research purposes and to support national genomic MDTs.

Specialist integrated haematological malignancy diagnostic services

24. In line with NICE Guidance (NG47) it is expected that all Haematological Malignancy tests (molecular/genomic and non-molecular) will be ordered via a SIHMDS, with all analysis and interpretation and reporting delivered by the NHS GMS, and then all results returned to the SIHMDS for integration into a final report. The genomic testing associated with haemato-oncology services will be consolidated into a single laboratory in each NHS Genomic Laboratory Testing Service with the exception of those tests where a rapid turnaround time is required (as outlined in the Test Directory) where an agreement with the Commissioner will need to be made.

25. Agreed working arrangements between the NHS GMS and SIHMDS is integral to the diagnosis and management of haematological oncology patients, and supporting and enhancing these close working relationships is crucial to maximising the use of genomics data alongside other diagnostic information.

26. This function will provide the clinical leadership and expertise to work with the NHS GMS Genomic Laboratory function and SIHMDS within their NHS GMS geography to establish referral pathways, reporting pathways and links with genomic haematological oncology MDTs to ensure a streamlined and efficient end to end pathway.

Clinical trials and access to precision medicine

27. This function will include the requirement to:
    - enhance recruitment to clinical trials based on genomic stratification and support strategic developments with industry and with other partners; and
    - drive the equitable uptake of precision medicines in strategic working with other services, such as oncology and pharmacy.

Developing the consent model for cancer

28. This function will support the ambitions outlined in the 10 Year Health Plan and Life Sciences Sector Plan to make it easier for patients to volunteer to participate in clinical trials and support population of the National Genomic Research Library held by Genomics England and the subsequent developments to introduce the Health Data Research Service.

29. Building on the patient choice framework for WGS, working with patient representatives the NHS GMS will develop a streamlined consent model that enables data from cancer genomic testing from large next generation sequencing panels and whole exome sequencing to be included in the National Genomic Research Library. Implementation of the consent model will be consistent with the NHS GMS principles to ensure equity of access for all patients and should be designed to minimise impact on workforce and resource and maximise the use of digital tools including the NHS app. To maximise the utility of genomic data, the consent model for cancer should also provide patients undergoing comprehensive cancer genomic testing in the NHS GMS the opportunity to receive additional findings, including reporting of clinical trial targets and pharmacogenomic analysis, that could influence other areas of their health management.

Cancer Genomic Improvement Programme

30. Each NHS GMS will be required to set out annually a Cancer Genomic Improvement Programme that outlines the objective, milestones that will be delivered and the associated impact to on each of the areas included in the Cancer Genomic clinical function.

Rare disease

Background to the rare disease clinical function

1. Genomic testing for rare disease strives to provide a genomically confirmed diagnosis to patients affected by rare disease with a likely monogenic or chromosomal cause. This diagnosis, or in certain circumstances absence of, may support clinical management, treatment, prognosis as well as impact on reproductive choice and predicted risk of disease in their family members. The testing offered for rare disease therefore covers diagnostic testing, carrier testing, predictive/pre-symptomatic testing as well as prenatal diagnosis.

2. The rare disease clinical function will work to support the commitment in the NHS 10 Year Health Plan to eliminate the diagnostic odyssey experienced by some patients with rare diseases, reducing the time it takes to get a definitive diagnosis from four years to three months (in some instances). It will also raise awareness, identify and address the unmet need in the population for genomic testing in rare disease. It will also support the priorities and commitments in the UK Rare Disease Framework, and the subsequent annual action plans that are published.

3. An important focus of the rare disease clinical function will be working with the transformation and service improvement function to use data to understand equity of access to rare disease, and put in place actions, including with all providers, in order to address any inequitable access to testing across the geography.

Clinical leadership

4. Clinical leadership within the NHS GMS will be required to:
   - raise awareness of genomic services across the whole rare disease multidisciplinary team, and work with all providers to drive patterns of referral where there is a diagnostic odyssey and unmet need in the population;
   - work with specialties where symptoms of monogenic disease most commonly present and get clinicians to ‘think genomics’ establishing pathways of referral and access to education and training resources;
   - enable the engagement and development of the multiprofessional workforce in rare disease, to drive equitable uptake of rare disease testing;
   - understand unmet need and variation in equity of access to rare disease genomic testing across the NHS GMS geography and recommend actions to address, including access to the R14 and R21 service, and raise awareness of these services;
   - support multimodal genomic testing and interpretation for patients who have not yet received a diagnosis;

- streamline pathways to improve diagnostic turnaround times and inform the development of national standards and guidelines for rare disease pathways;
- support the introduction of new technological advances, including omics, across the care continuum;
- support access to clinical trials inclusive of new and emerging ATMPs and the establishment of the Rare Diseases Launchpad;
- work collaboratively with Clinical Genomics Services and with other specialties where there are a high proportion of referrals for genomic testing to both streamline pathways and access results;
- continually review diagnostic outcomes to inform demand management approaches in access to genomic testing across the NHS GMS geography; and
- inform and work in a cross-cutting way with other functions of the NHS GMS in the geography.

5. The clinical leadership within the NHS GMS is responsible for understanding what unmet need exists across NHS GMS geography in areas where genomic testing is already commissioned in the Test Directory, for example monogenic IBD and primary immune deficiency but where there is evidence that communities are not accessing this testing.

6. The clinical leadership will identify patients who have not yet received a diagnosis despite having had WGS and are under the management of Clinical Genetics Services to develop a personalised genomic testing strategy, including reanalysis of existing genomic data for that individual.

Genomics MDTs

7. Genomics MDTs are required to support the interpretation of genomic data through multi-professional discussion of clinical cases and genomic results for patients with rare disease. Genomics MDTs can be particularly helpful for discussing variants of uncertain significance (VUS), incidental findings, and cases where genetic testing has not achieved a diagnosis but clinical suspicion of a genomic diagnosis remains high. In rare disease, this will also include Genomic Rare Disease Advisory Boards (GRDABs) for complex cases.

8. Each NHS GMS Lead Provider will continue to form, coordinate and support Genomics MDTs as set out in the Rare Disease Interpretation and Reporting Guidance and as required and work with other NHS GMS Lead Providers and the Commissioner to share information and learning in order to avoid duplication and enhance the knowledge of the NHS GMS National Network.

Fetal medicine pathway

9. There is considerable variation in care pathways across the country and often poor access of ethnic and other underserved populations to maternity care and pre- and perinatal genomic testing. In 2023 the Commissioner funded the Prenatal NHS Genomic Network of Excellence to ensure equity of access to high quality pre- and perinatal Genomic Medicine for all families across England, capitalising on new technological developments to deliver high quality testing for more conditions.

10. From April 2026 the NHS GMS will be required to increase access to rapid WGS for fetal anomalies and work with fetal medicine to develop optimal care pathways for prenatal genomic testing. Building on the evidence from the Prenatal NHS Genomic Network of Excellence, the NHS GMS should develop innovative approaches to service delivery with a focus on ensuring equitable access for individuals from diverse backgrounds.

Medicines optimisation and clinical trial access

11. Based on national and regional prioritisation, enable equity of access to genomic testing facilitating access to precision medicines and / or ATMPs and support defined medicines optimisation priorities.

12. Linking with research and innovation and the Rare Disease Launchpad to support genomic testing to increase access to clinical trials.

13. Drive uptake of precision medicines and ensure approach to testing that supports reduction in adverse drug reactions.

Rare disease improvement programme

14. Each NHS GMS will be required to set out annually a Rare Disease Improvement Programme that outlines the milestones that will be delivered and the associated impact on each of the areas included in the Rare Disease clinical function and in line with national priorities set out in annual planning guidance.

Clinical genomics service

15. NHS Clinical Genomics Services provide a diagnosis and/or genetic counselling, and, in certain cases, ongoing management for patients of all ages (including prenatally) with a proven or suspected rare genetic condition. NHS Clinical Genomics Services additionally provide genetic counselling and predictive testing to relatives who may have inherited, or are at increased risk, of a rare condition but who are considered asymptomatic.

16. There are seventeen regional NHS Clinical Genomics Services across England. The commissioning of the CGS will remain with NHS Regional Specialised Commissioning and is out of scope of this Specification.

17. A Clinical Genomics Service Transformation Programme is underway and will inform the future commissioning arrangements including developing an updated Service Specification and outcome metrics. The Transformation Programme is looking across the Clinical Genomics Service to agree what action needs to be taken to ensure the Service is sustainable and fit for purpose in the future.

18. Ahead of any decisions being made on the future commissioning arrangements of the Clinical Genomics Service and to support the delivery of Genomic Medicine in the NHS the NHS GMS must ensure that each NHS Clinical Genomics Service within the NHS GMS geography is integrated into the governance structure and part of strategic discussions on the future of genomics across the NHS GMS geography.

19. The NHS GMS should work closely with and ensure that appropriate Clinical Genetics expertise and clinical leadership is part of the delivery of the Rare Disease Clinical Function.

Genomics population health

Background to the genomics population health clinical service

1. Identifying high-risk individuals at a population level, including through cascade testing of family members, is paramount to early detection of disease and preventative medicine, including for example, for inherited cancers and cardiovascular disease. Areas such as Integrated Risk Scores, systematically reanalysing data to generate more diagnoses at a population level; and exploring the clinical utility of genomics in common disease as well as what can be analysed as part of existing genomic testing (for example the additional findings of index and cascade cases), are all areas of genomics population health that require a specific focus.

2. The NHS 10 Year Health Plan included a commitment that the NHS would establish a new genomic population health service, accessible to all, by the end of the decade. This will bring together genomics, new diagnostics and predictive analytics with artificial intelligence, in order to identify genetic predisposition to disease, enable early interventions, support personalised treatments and deliver more effective prevention strategies. This will involve working with new and emerging Neighbourhood health services.

3. A key pillar of the service will be the use of pharmacogenomics – the study of how an individual's genome can influence how they respond to medicines. The current NHS GMS pharmacogenomic testing offer will expand significantly to optimise the use of medicines throughout a patient's lifetime and reduce adverse drug reactions. This will include preparing for pre-emptive pharmacogenomic testing and exploring the development of pharmacogenomic profiles.

4. Relevant systems, tools, standards and resources will be developed nationally, in collaboration with NHS GMS geographies, recognising that local adaptation will be needed to enable partnership working across a broad spectrum of health and care services in specific geographies.

Clinical leadership

5. The clinical leadership within the NHS GMS is responsible for raising awareness of the genomic population health service across the multi-disciplinary workforce and overseeing the development of genomics champions in the neighbourhood, working with the People function outlined in this Specification in conjunction with the Genomics Education Programme and identifying the need for and bringing specialist expertise to

help and guide the neighbourhood teams and in leading approaches to inherited disease testing in high risk populations.

6. Specific clinical leadership will be required in primary care, including working with GPs that have experience in the delivery of population health and understanding of genomics.

7. Clinical leadership within the NHS GMS will be required to:
   - understand the healthcare landscape across the NHS GMS geography and where delivery of genomic population health services will have the greatest impact, including through identifying index cases, family cascade testing and understanding family history;
   - identify high risk populations to target specific inherited genomic testing solutions;
   - collaborate with primary care and Neighbourhood Health Teams to equitably increase the uptake of genomic testing in local communities;
   - support genomic referrals and the interpretation and return of results;
   - provide expertise on understanding and interpreting population based risk including through supporting the proposed evaluative PRS study with OFH and others;
   - develop the workforce in population health in conjunction with the People function;
   - signposting to specialist services e.g. ICC and cancer screening following working with other services in the geography for example coronial services in the case of testing family members from sudden cardiac death cases;
   - support the introduction of new and emerging technologies and approaches in population health across all functions of the NHS GMS, including through generation of evidence; and
   - using service improvement approaches to develop pathways and models of care.

Developing the genomics population health service

8. The Population Health Clinical Function will work with the Commissioner and as part of the NHS GMS National Network, to develop the genomics population health service and strategy for local implementation with primary care and Neighbourhood Health Teams.

9. This will include supporting in the development of a detailed delivery plan for:
   - the implementation of polygenic risk scores in the NHS. This will begin with a Service Evaluation in 2026 inclusive of cardiovascular disease, diabetes and breast cancer and based on the evaluation further scale up and roll out;
   - pre-emptive pharmacogenomics, including from 2026 supporting and working with clinical experts to develop and implement pharmacogenomic panels for severe mental health and CVD working with clinical experts;

- returning additional findings and pharmacogenomic results to all patients and their families who receive WGS and patients who receive WES. This will include agreement on what information is returned and the underpinning data and digital requirements; and
- exploring the future commissioning of newborn genomic testing, following the delivery and evaluation of the Generation Study.
- Working with the Genomics England Adult Population study to help scope and define its approach

Inherited risk

10. The population health function will provide the clinical leadership and expertise to drive the expansion of the NHS GMS to include more comprehensive genomic testing to further an individual's understanding of their inherited risk.

11. This will focus initially on cancer and CVD and expanding to other areas across the duration of the contract. This will include gathering a detailed understanding across the NHS GMS geography of:
    - high risk populations where there is unmet need and undertaking activities to raise awareness of genomic testing
    - the opportunities for improved cascade testing and case finding of index cases across the NHS GMS geography and the actions that can be taken to drive improvements and identification of cases
    - improving the recording and utilisation of family history, supported by the use of digital tools, to improve the uptake of genomic testing and improving patient outcomes.

Genomics Population Health Improvement Programme

12. Each NHS GMS will be required to set out annually a Genomics Population Health Improvement Programme that outlines the milestones that will be delivered and the associated impact to on each of the areas included in the population health clinical function and in line with the priorities set out nationally in the annual NHS GMS planning guidance.

Enabler: Science, research and innovation

1. The NHS 10 Year Plan and the Life Sciences Sector Plan highlights the strength of the UK's life sciences and genomics ecosystem. A fundamental part of this is the NHS GMS infrastructure that has evolved since the 1970's and has continued to develop since the 100,000 Genomes Project and over the last five years since the introduction of the NHS GMS. This continued development has only been possible by embedding a research and innovation pathway into the system from discovery research through to adoption into routine clinical care.

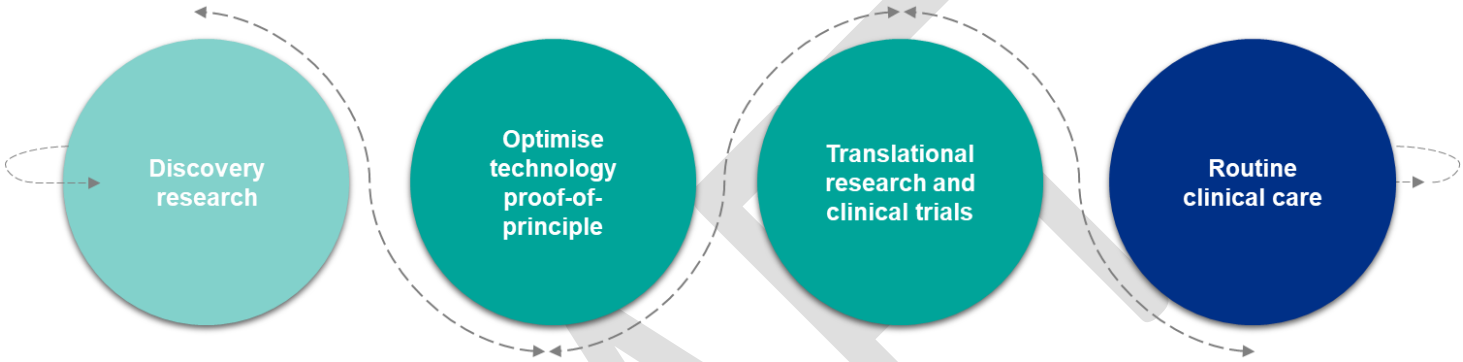


Figure 8 – NHS GMS innovation pathway

2. The research and innovation model that has been developed by the NHS GMS has been recognised as a best practice exemplar by the NHS Innovation Ecosystem Programme. The NHS GMS infrastructure that will be implemented as outlined in this Specification will strengthen the power of the NHS GMS to agree research priorities, leverage funding opportunities, and deliver agreed research priorities across the whole innovation pipeline to benefit NHS patients and the public. Supporting and driving forward research and innovation and genomics excellence will continue to be at the heart of all that the NHS GMS does and supporting a learning health system.

3. It is for this reason that genomics is one of the five transformative technologies ('big bets') that the NHS 10 Year Plan highlights. The NHS GMS Lead Provider will support the ambition to harness the power of genomics, artificial intelligence and predictive analytics to create a new model of care in the NHS and deliver priorities on research and innovation, as well as those in The UK Modern Industrial Strategy, Life Sciences Sector Plan, and other national initiatives and priority programmes, such as the National Cancer Plan.

People and responsibilities

4. The NHS GMS Lead Provider will appoint a Research and Innovation Director, a mandated Consultant level position, where significant expertise and expertise will be

required to lead the science, research and innovation function. The Research and Innovation Director will be responsible for building a team with appropriate expertise and experience, including a Research and Innovation Deputy Director, Senior Programme Manager, further Programme Management support and an NHS Genomic Network of Excellence Lead.

5. The Research and Innovation Team will, on behalf of the NHS GMS Lead Provider, be responsible for being knowledgeable of all genomic research activities being undertaken within each geography that has the potential to impact on the NHS GMS or to influence through evidence generation the commissioning of services nationally and ensure strategic partnerships are in place to influence the delivery of research activity, as well as to align to the strategic ambitions of the NHS GMS.

6. The NHS GMS Lead Provider shall be responsible for developing and maintaining innovative partnerships and collaboration with academia, key research organisations and infrastructure, and industry, to drive research and innovation to deliver technology, scientific, and clinical advances and to facilitate their adoption. This will include working with new initiatives that arise from the NHS 10 Year Health Plan and the Life Sciences Sector Plan in science, research innovation. Coordinating these partnerships, sharing knowledge, promoting best practice and measuring outcomes will be expected at both a regional and national level through governance arrangements outlined in this Specification.

7. This will be overseen by a Research and Innovation Oversight Group at the NHS GMS Lead Provider level, which will report into the NHS Genomics Board, as outlined in **Figure 2**.

Working with partners

8. The NHS GMS Lead Provider will work with the Commissioner and other organisations, including the Health Data Research Service, Health Innovation Networks, the Department of Health and Social Care, and the National Institute for Health and Care Research infrastructure to contribute to scientific and technological advancements; and to support broader research, development, innovation and recruitment to clinical trials.

9. The Commissioner will lead and coordinate collaboration with Our Future Health and Genomics England. As determined by the Commissioner, the NHS GMS Lead Provider will support national priority programmes such as Our Future Health and Genomics England spending review initiatives, including the ongoing Generation Study.

Support for major research studies

10. The NHS GMS Lead Provider will have an important role to play in delivery of a number of existing and future national genomics projects and initiatives (including the supply of additional samples and data for such projects), including:

- a number of projects being delivered by Genomics England:
    i. the Generation Study – which aims to recruit and sequence the genomes of 100,000 newborn babies with parents' consent;
    ii. the Adult Generation Study – which aims to sequence the genomes of 150,000 adults and assess how genomics can be used in routine care; and
    iii. Diverse data initiative – to reduce health inequalities and improve patient outcomes in Genomic Medicine for minoritised communities.
- Our Future Health – service evaluation to explore implementing Integrated Risk Scores in neighbourhood health settings and any broader genomic initiatives that can be aligned with the NHS;
- the Cancer Vaccines Launch Pad; and the Rare Disease Launchpad; and
- other national and international projects approved by the Commissioner.

11. The NHS GMS Lead provider will support academic and local projects, coordinated through the appointed Research and Innovation Director with reporting into the Commissioner as outlined in the governance section of this Specification. This may include developing NHS GMS network research and innovation applications to funding bodies.

12. The Commissioner will lead and coordinate collaboration with industry partners across the NHS GMS. The NHS GMS Lead Provider will deliver activity aligned to national priorities and work with industry partners to support agreed improvement and other activities across the NHS GMS.

13. The NHS GMS Lead Provider will establish a service operating plan to support ongoing research and innovation at both a national and international level and collaboration with industry, including alignment with other current or planned genomics activity, for example, Cancer Alliances, Health Innovation Networks, National Institute for Health and Care Research Biomedical Research Centres, Clinical Research Networks, and others.

14. The NHS GMS Lead Provider shall report to the Commissioner in a timely manner on any collaborative projects it embarks upon in respect of genomics and the use of the NHS GMS infrastructure inclusive of testing services, whether or not it considers the collaboration came about because of its role in the NHS GMS. An appropriate form of

words for any acknowledgements of the NHS GMS infrastructure in any resulting publications will be available from the Commissioner.

15. Subject to compliance with any publication restrictions and existing contractual arrangements, if the NHS GMS Lead Provider undertakes any research or innovation projects, or participates in any genomic projects that are not financed by the Commissioner and covered by this Specification, where possible it must deposit any data collected as a result of such research, innovation or project into National Genomics Research Library and in future linking with the Health Data Research Service.

16. The NHS GMS Lead Provider should enable access to biobanked samples for approved research use, as set out in guidance from the Commissioner.

NHS Genomic Networks of Excellence

17. In line with the commitment in the Life Sciences Sector Plan, the NHS GMS Lead Provider will support the delivery of the NHS Genomic Networks of Excellence, to assess the latest genomic advances in specific priority areas covering genomics, artificial intelligence and predictive analytics. NHS Genomic Networks of Excellence are partnerships between the NHS, academia, the third sector and industry to generate evidence and models of adoption for new technologies and clinical practice in defined topic areas of strategic importance. The ability to leverage funding from partners across the genomics ecosystem will remain a core dependency of establishing any new NHS Genomic Networks of Excellence.

18. Building on the success of the eight NHS Genomic Networks of Excellence that were announced in 2023/24 for a two-year period, the Commissioner will again work with stakeholders to define strategic priorities and establish new NHS Genomic Networks of Excellence to commence from April 2026. This will include continuation of NHS Genomic Networks of Excellence in areas which continue to be of strategic importance such as infectious disease and pharmacogenomics, subject to evaluation and delivery of existing business plans. The NHS GMS Lead Providers will work with partners to put forward expressions of interest and, if successful, business cases for establishing NHS Genomic Networks of Excellence. This documentation will be reviewed by the Commissioner and key experts before successful outcomes are confirmed.

19. Indicative process and timelines for establishing the NHS Genomic Networks of Excellence:

- October 2025: the Commissioner publish guidance, including strategic priorities for the NHS Genomic Networks of Excellence and share expression of interest documentation
- November 2025: NHS GMS Lead Provider submit expression of interest documentation
- November 2025: The Commissioner and key experts review expressions of interest. Successful applications invited to submit formal business case based on a two-year arrangement
- December 2025: NHS GMS Lead Provider submit business case
- January – March 2026: Review of business cases and confirmation of work packages and funding
- April 2026: Delivery of NHS Genomic Networks of Excellence commences.

Support for clinical trials and research

20. The NHS GMS Lead Provider will work with key partners, for example Clinical Research Networks and Clinical Trials Units, to support identification, coordination, and enrolment of eligible participants into clinical trials. This will also involve working with the ABPI and other bodies as directed by the Commissioner

21. The NHS GMS Lead Provider must be able to demonstrate translation of research into clinical practice, working with research and clinical staff to achieve this. The regional NHS GMS will focus on the interface between translational research and service innovation in genomics. In particular, the regional NHS GMS will facilitate clinical trials, the identification of subjects for research and the analysis and / or collection or provision of additional data and biobanked samples where agreed by the Commissioner.

Consent for research

22. The NHS GMS Lead Provider will work with clinicians to support the implementation of guidance on consenting patients for research, in order to support the population of the National Genomic Research Library and diagnostic discovery. Over time it is expected that this will align with the development of the Unified Genomic Record and be more innovative in approach.

23. The NHS GMS Lead Provider, working together with the Commissioner and Genomics England, will agree protocols for the identification and management of existing and potential Intellectual Property Rights and shall comply with all applicable IP standards and IP policies. This will also be considered more broadly for non-Genomics England related activity and in line with relevant national policies and strategies.

Enabler: Transformation and service improvement

People and responsibilities

24. The NHS GMS Lead Provider will appoint a Transformation and Service Improvement Director, a mandated position, to lead a Transformation and Service Improvement Team with relevant transformation and service improvement experience and expertise. This will include experience in appropriate methodologies, for example LEAN. It is expected that specific roles will include a Transformation and Service Improvement Lead for Cancer, Rare Disease and Population Health, an Equality and Diversity Lead, as well as Programme Management Support.

25. As outlined in the independent review of the patient safety landscape across health and care, led by Dr Penny Dash, there are sixteen core functions required to ensure a high-quality health and care system. The Commissioner is responsible for:
    - defining a clear strategy to improve quality of care including defining the purpose and vision of the NHS GMS
    - reviewing the clinical and managerial evidence base, academic research and examples from other industries or other healthcare systems, and defining what 'good' looks like in order to deliver on the NHS GMS purpose and aims
    - identifying national priorities and setting out structures to deliver against these priorities, and enable high-quality and efficient delivery of services, including optimal Commissioner and provider structures and robust governance structures
    - allocating resources in order to maximise delivery against the aims, taking account of the need to balance across competing needs and priorities
    - engaging with users, communities, staff and wider stakeholders on strategy and priority setting.
26. The Commissioner and the NHS GMS Lead Provider will work together to:
    - understand the current performance of the NHS GMS, including what current quality of care (safety, effectiveness, user experience and equity) is, how well managed services are and how well resources are used

27. The NHS GMS Lead Provider is responsible for:
    - **Delivering high-quality care**, including developing, agreeing and implementing highly optimised operating processes and service models to deliver safe, effective, responsive, efficient and equitable services, using standardisation and technology where possible and appropriate. Optimising the resources required at a local level to deliver high-quality care, noting that standardised approaches typically use less resources. Putting in place organisational and governance structures ('from board to ward') to:

    - make clear the standards expected
    - introduce processes for monitoring performance against standards, including continuous review of data and inputs
    - ensure support for improvement where needed
- **Providing leadership** that puts quality at the centre of care, recognises the role of high-quality management - including operational management and people management, embeds, models, recognises and rewards behaviours that enable high-quality care. Training, development and accreditation of staff.
- **Monitoring and assuring quality** including seeking input from users, measuring outputs and outcomes, carrying out audits and investigations, and quantifying the use of resources. Ensuring compliance with planned processes and expected outcomes. Managing the impact of severe harm, taking enforcement action where needed and ensuring redress where appropriate.
- **Continuously improving** - reviewing, learning, listening, changing and adapting - to address sub-optimal adherence to agreed strategies and processes, and expected outputs and outcomes.

28. The NHS GMS Lead Provider will deliver whole scale transformational change and service improvement in line with the five core components of NHS Improving Patient Care Together (IMPACT) that underpin a systematic approach that includes:
    - building a shared purpose and vision;
    - investing in people and culture;
    - developing leadership behaviours;
    - building improvement capability and capacity; and
    - embedding improvement into management systems and processes.

29. It will also be aligned with the five key strands of the Getting it Right First Time (GIRFT) model:
    - a broad data gathering and analysis exercise, performed by health data analysts, which generates a detailed picture of current national practice, outcomes and other related factors;
    - direct clinical engagement via visits or virtual meetings between clinical specialists and individual hospital trusts, which are based on the data – providing an unprecedented opportunity to examine individual trust behaviour and performance in the relevant area of practice, in the context of the national picture. This then enables the trust to understand where it is performing well and what it could do better – drawing on the input of senior clinicians;
    - a national report, that draws on both the data analysis and the discussions with the hospital trusts to identify opportunities for improvement across the relevant services;

- an implementation phase where the service improvement and transformation team supports trusts, commissioners, and integrated care systems to deliver the improvements recommended; and
- best practice guidance and support for standardized / integrated patient pathways and elective recovery work in 'high volume/ low complexity' specialties.

30. The NHS GMS Lead Provider will be accountable for achieving demonstrable improvements in the following areas:
    - delivering genomics services in a timeframe that is aligned with clinical needs to ensure that patients can access a diagnosis and treatments in a clinically relevant timeframe;
    - ensuring that the NHS GMS delivers cost-effective services for the NHS through driving efficiencies and improving processes;
    - delivering high quality genomic services that deliver the highest standards for patients
    - equitable access to standardised end-to-end pathways of care, inclusive of genomic testing and working with clinical genetics and genomic counselling services;
    - access to precision treatments and genomics informed medicine optimisation driven by comprehensive genomic and diagnostic characterisation;
    - enabling access to genomic testing that supports population health, inclusive of inherited disease and pharmacogenomics and over time the use of PRS;
    - increasing the number of people accessing clinical trials by ensuring the systematic consideration of eligibility to clinical trials for patients who would potentially benefit; and
    - active participation and contribution to the nationally coordinated and facilitated approach to genomic research across the country to embed research and discovery to advance clinical care for patient and societal benefit.

Delivering equitable genomic services

Working with partners

31. The NHS GMS Lead Provider will facilitate engagement with all other NHS providers and organisations across each geographical area to achieve the equity and standardisation of Genomic Medicine, including appropriate and effective engagement with NHS Regions, Integrated Care Boards, the National Disease Registration Service (NDRS), Primary Care Networks, Pathology Networks, Cancer Alliances, Health Innovation Networks, academia and industry.

Better data and intelligent use of data

32. The NHS GMS Lead Provider will be responsible for ensuring compliance with the Genomics Testing Reporting Specification for all 'mandatory' and 'mandatory where relevant' data fields, ensuring accuracy of the data provided, and providing this data to

the Commissioner in a timely manner at the agreed frequency. NHS GMS Lead Providers are responsible for coordinating timely collection of the required data fields from providers across their geography.

33. The NHS GMS Lead Provider will work with the Commissioner to demand model population need across NHS GMS geographies; evaluate testing rate in eligible population; routinely report demand and access; and develop priority actions to address inequities highlighted by the data.

Community engagement

34. The NHS GMS Lead Provider will measure and monitor equity of access to testing across the NHS GMS geography, and design and implement initiatives to address unmet need. This will include for example developing strategies and supporting projects to address inequities in access and to use genomics to reduce health inequalities. The NHS GMS Lead Provider will be responsible for identifying and prioritizing cohorts of individuals, for example those with protected characteristics e.g. black, Asian and minority ethnic groups; disabled; LGBTQ+ to actively engage with the NHS GMS and potentially socially excluded cohorts e.g. inclusion health groups such as rough sleepers, the homeless; asylum seekers and Gypsy, Roma and Traveler groups.(Core 20+5).

35. The NHS GMS Lead Provider will be expected to nominate a senior representative to represent the NHS GMS Lead Provider on the NHS Genomics Ethics, Equity and Legal Advisory Group. This representative will be responsible for liaising with the wider NHS GMS Lead Provider Team and the other NHS GMS Lead Providers to tackle issues of equity of access to genomic testing. This will also include working with partners for example the NHS Race and Health Observatory (RHO) to explore current inequities in genomic services, and what actions can be taken.

Enabler: People - Workforce, education and training

People and responsibilities

1. The NHS GMS Lead Provider will appoint a People Director with appropriate experience and seniority, who will bring together an expert multidisciplinary team (and including PPI representation) to drive forward the workforce planning – where relevant - development and upskilling of the clinical and scientific workforce within the NHS GMS geography and to ensure a robust awareness raising package is in place.

2. As well as the People Director, the multidisciplinary team will include a Deputy Director, a Lead for clinical services and laboratory/scientific services, Education and Training Lead, Workforce Lead, Communications and PPI Lead, Workforce Clinical Leads, for example in nursing, pharmacy, midwifery, genomic counselling and other clinical posts, as well as programme management support. The NHS GMS Multidisciplinary People Team will work through a matrix approach with the other functional delivery units of the NHS GMS who are able to demonstrate leadership and/or expertise in education, training and workforce development in genomics, with a track record of delivery of change.

3. The NHS GMS Lead Provider will work with workforce commissioners and NHS workforce leaders in NHS Regions, as well as with the National Genomics Education Programme and any other national education and training function for the healthcare workforce to agree priorities and workforce needs including workforce, education and training requirements, development of established roles and creation of new roles that align to a national genomics workforce, education and training strategic framework, that will be developed.

4. A People Board will be developed and required to oversee the delivery of the function. The People Board will need to link with any national People Boards to ensure that there is strategic alignment.

5. The work will be focused on driving the implementation of Genomic Medicine across the geographical area including in neighbourhood and primary/community health services and in secondary care settings. The work will be in alignment with the 10 Year Health Plan and upcoming 10 year workforce and cancer plans and will take into account any skills developments that could be shared with the life sciences sector and outlined in the Life Sciences Sector Plan. Priorities will be agreed across the NHS GMS geography and shared / coordinated and where needed especially if associated with for example 10 Year Health Plan deliverables agreed at the national level The work will be

overseen by an NHS GMS People Oversight Group, as outlined in **Figure 2**, which will bring together partners as well as representatives from each NHS GMS.

6. A key new area for the NHS GMS will be working with the neighbourhood health service and neighbourhood health teams ensuring genomics is embedded into community care through the upskilling of the workforce to deliver the genomics population health service. Neighbourhood health reinforces a new way of working for the NHS, local government, social care and their partners, where integrated working is the norm and not the exception and where the emphasis is on prevention rather than treatment of disease.

7. Ensuring that the NHS GMS is working closely with neighbourhood health teams, will be critical to the success of the NHS 10 Year Health Plan. The NHS GMS will support the development of neighbourhood teams to harness the benefits of genomics to understand and act on insights from genomics, as part of our approach to ensuring staff have the skills (including some relevant and transferable genomic counselling skills) they need to meet the future.

8. This will include training Genomics Champions in the neighbourhood – local health professionals with the requisite knowledge and ability to increase uptake of local genomic testing in their community, equitably and to drive the uptake of genomic risk-based stratification and other genomic population health-based initiatives.

Workforce

9. Each NHS GMS Lead Provider will work across the professional groups and with other clinical specialties involved in mainstreaming genomics to develop and update an NHS GMS People Plan.

10. Key to this will be working across the geography to gather intelligence on current and future workforce trends that will then inform workforce planning, supply and education and training requirements. Each NHS GMS will provide on an ongoing basis information on demand signaling for the multi-professional workforce to be reflective of genomic needs to feed into the upcoming 10 Year Workforce Plan. This should include working with system partners and relevant clinical networks across the geography to ensure the integration of genomics.

11. The NHS GMS Lead Provider will ensure that all specialist staff working in genomics that require a professional registration, have this in place with the appropriate regulatory body (statutory and non-statutory as relevant) and support continual

professional development (CPD or CME) and regular revalidation or competency assessment as appropriate.

12. The NHS GMS Lead Provider will work with the National Genomics Education Programme to develop neighbourhood and mainstream specialty genomic champions, aligning to where relevant the National Genomics Education Programme genomic champion competency framework.

Education and training

13. The NHS GMS Lead Provider will continue to work in collaboration with the National Genomics Education Programme to drive the implementation of Genomic Medicine through the development of coordinated education and training programmes and resources that are fit for purpose and applicable across the whole of the NHS.

14. The NHS GMS education and training delivery should aim to meet the needs of the multi-professional, multi-specialty workforce with a particular emphasis on the development of neighbourhood teams to spearhead the implementation of Genomic Medicine at the community level and for example the early priority areas of polygenic and inherited risk.

15. Resources should be developed against the priorities that have been identified through the scoping exercise and in communication with the National Genomics Education Programme. Innovative approaches should be used where appropriate with a view to disseminating nationally where applicable in order to share best practice and avoid duplication.

16. The NHS GMS Lead Provider will form part of a national education and training network, led by the National Genomics Education Programme working in conjunction with and as part of the national genomics programme. This collaborative work will ensure delivery of consistent, effective and efficient genomics education and training and workforce development.

17. The NHS GMS Lead Provider will work with partners to support the implementation of changes to the Test Directory, including integration of new, updated and clinically essential tests. This will include working with the multi-specialty and multi-disciplinary workforce to deliver the underpinning education and training.

18. The NHS GMS Lead Provider will develop an awareness and communications strategy, with particular focus on encouraging uptake of genomics within neighbourhood teams as part of preventative population health as well as in cancer care and rare disease.

This strategy should be locally responsive but feed into the national approach, working with the Commissioner and National NHS GMS Network to deliver national initiatives.

19. The NHS GMS Lead Provider will work with patient and public involvement representatives to ensure that any education and training that is delivered also meets the needs of patients, carers and their families by ensuring that it is representative and supports high quality patient care.

Communications

20. Key to delivering all elements of the Specification, but in particular the people function, will be working with partners across the NHS system, as well as the life sciences sector and the genomics ecosystem to communicate effectively on the benefits of genomics.

21. Each NHS GMS Lead Provider will be expected to ensure a communications function is part of the infrastructure, and focused on working with partners to raise awareness of genomics, including new advances being delivered by the NHS GMS; supporting education and training of the workforce; and ensuring that patient and public involvement is at the heart of the design and delivery of the service, in line with the principles outlined in the Specification.

Enabler: Data and digital

22. The requirements in this specification align to each of the digital commitments for genomics in the NHS 10 Year Health Plan and the Life Sciences Sector Plan. NHS GMS clinical and genomic health data will flow seamlessly and securely into every patient's health record, via the Unified Genomic Record, and will be securely accessible by them, and their authorised clinicians, no matter where they are in the NHS. The genomic digital network will ultimately integrate with, and form a key part of, the Genomics Population Health Service, the Health Data Research Service (HDRS) and the Single Patient Record (SPR).

23. The NHS GMS will be responsible for the establishment of sustainable data and digital governance, services and processes across their geographies; and will advance the local adoption of national informatics standards and services amongst NHS GMS partners and across the broader digital landscape within their geography, to achieve the goal of interoperability between NHS GMS LIMS (Laboratory Information Management Systems) and requesting systems (e.g. EPRs and pathology LIMS) via a national broker. Data standardisation, interoperability, and the ability to federate data, within a common cloud environment must be considered a priority across all genomic data and digital services.

24. The Specification details the specific commitments and requirements needed to deliver the national strategy and associated transformations. Specific sections include:
    - governance, people and responsibilities;
    - national systems and standards being delivered by the Commissioner and partners, to support the operation of the NHS GMS;
    - systems and processes expected to be established by each NHS GMS Lead Provider; and
    - specific expectations needed to deliver the service and associated transformations.

Governance

25. Appropriate governance should be put in place to support delivery and to regularly bring together the clinical leadership and expertise and ensure there is delivery and alignment across the NHS GMS geography.

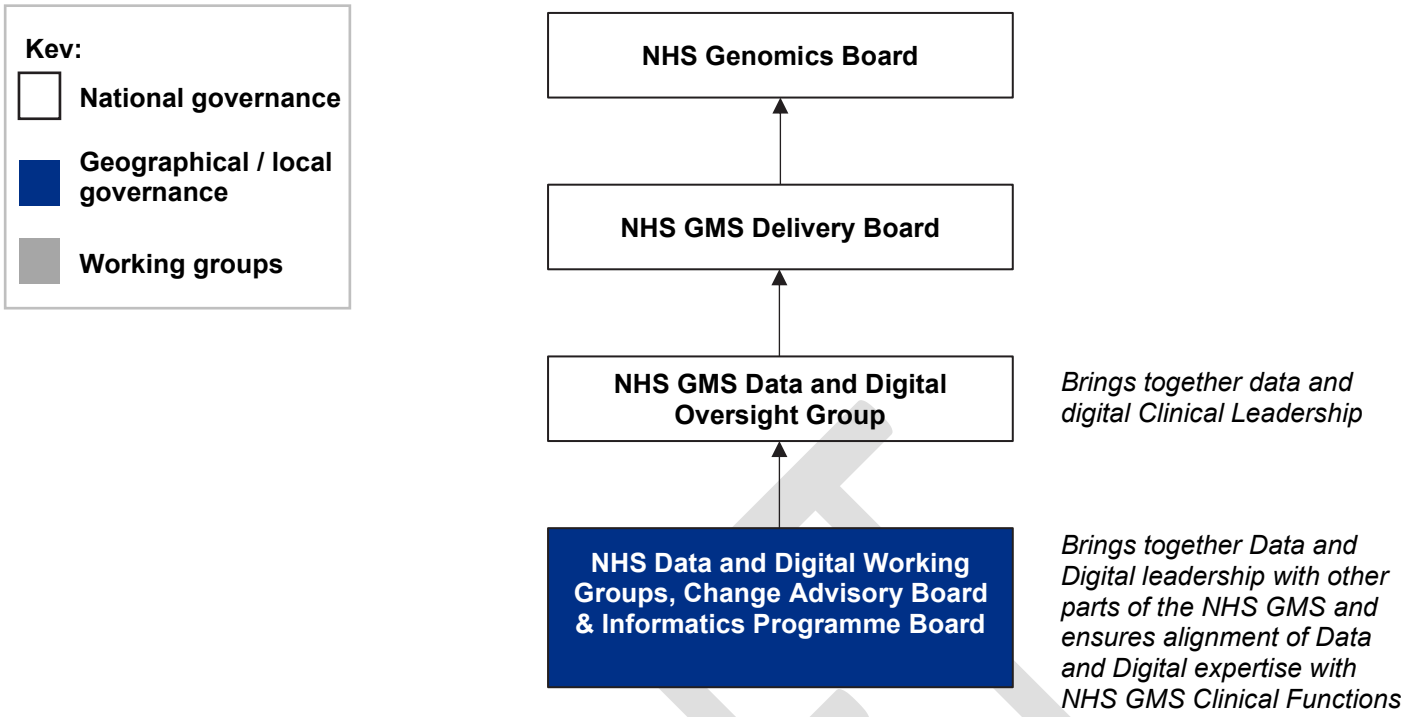


Figure 9 – Data and Digital governance overview

People and responsibilities

26. The NHS GMS is required to have appropriate staffing in place to ensure the fit-for-purpose operation of their data and digital services. It must also maintain strategic alignment with the digital processes and strategies of their associated trusts, Integrated Care Systems and other NHS bodies.

27. The NHS GMS should identify or appoint the following staffing positions:

- A Data and Digital Director, to be responsible for the management and delivery of all NHS GMS data and digital services across the geography.
- A Clinical Informatics Lead, with Chief Clinical Informatics Officer (CCIO) responsibilities, to provide clinical direction and assurance, and to ensure that informatics services are aligned with the wider needs of the NHS GMS; with a supporting team of clinical application and data specialists and clinical / scientific subject matter experts.
- A Bioinformatics Lead, responsible for management and delivery of NHS GMS bioinformatics services, aligned to the national bioinformatics strategy; with a supporting team of bioinformatician clinical scientists and data specialists.
- A Digital Service and Operations Lead, to be responsible for the operation and support of all NHS GMS informatics systems, including their interoperability with other regional (including all providers across the care sectors i.e. secondary care as well as primary community and emerging neighbourhood care) and national systems; with a

supporting team including information governance, systems administrators and service specialists.

- A Data and Analytics Lead, to be responsible for ensuring NHS GMS data is managed appropriately, quality assured and aligned with national data standards; with a supporting team including data engineers, BI developers, population health analysts, data scientists, and governance leads.
- A Data and Digital Head of Programme, to be responsible for the introduction of new digital capabilities across the NHS GMS and establishing connectivity with relevant clinical systems across all providers in their geography; with a supporting team including project managers, business and test analysts, and relevant subject matter experts.

28. The organisation structure must be staffed to meet current demands for each function and should be able to adapt to any changes / growth as the digital programme proceeds.

29. The NHS GMS should develop a data and digital workforce plan, including consideration of training needs and resilience planning. This should include, but is not limited to management training, technical / specialist training, and clinical informatics STP trainees.

30. The NHS GMS should develop a bioinformatics workforce plan, including consideration of training needs, resilience planning on long term resource forecasting. This should include, but is not limited to: management training, ongoing continuous professional development, technical / specialist training, and bioinformatics STP trainees.

Working with partners

31. In support of delivering the digital elements of the national genomic strategy, the NHS GMS in each geography shall work in partnership with the Commissioner and other partners, particularly Genomics England, but also via the NHS Genomic Networks of Excellence with industry and academia, to develop a cutting edge digital infrastructure; to expand and enhance consented datasets for health research; to streamline the testing process and maximise the patient benefit through timely access to data and to accelerate the adoption of health data research insights and evidence into clinical practice.

32. Each NHS GMS Lead Provider will establish and maintain appropriate data and digital governance and management processes. These need to be aligned with, and provide any required assurances to, including but not limited to the following bodies:

- Local trust informatics services.
- Other trusts and regional ICS / ICB / Neighbourhood Health Services.
- Other NHS GMS geographies and Genomic Networks of Excellence.
- The Commissioner, National Data and Analytics and regional commissioners.
- Department of Health and Social Care, and other governmental bodies.
- UK Health Security Agency (UKHSA)
- UKAS, Medicines and Healthcare products Regulatory Agency (MHRA) and other accreditation bodies.
- Academic and research networks, including Health Innovation Networks (HIN), Biomedical Research Centres (BRC), and Local Clinical Research Networks (LCRN).

National Systems and Standards

33. In collaboration with the NHS GMS geographies and with other partners, the Commissioner will continue to build and develop the national digital infrastructure outlined in the national Genomics Data and Digital Framework. The NHS GMS Lead Provider must aim to be interoperable with the national digital infrastructure using the national standards identified within the Specification, and within the timelines laid out below.

34. Detailed timelines for transformation are available in the Data and Digital Programme Tube Map and required NHS GMS contributions are included in the bioinformatics deliverables.

35. National services and commitments include, but are not limited to:
    - **Neighbourhood Health Services:** To support the establishment of the Neighbourhood Health Services, as outlined in the NHS 10 Year Health Plan, genomic data and services will need to be accessible by primary care and other neighbourhood staff and by any supporting secondary care staff. The data and digital services being used and developed by the genomics services need to accommodate the development of new NHS operating models.
    - **Genomic Population Health Service:** To support the goals of the Genomics Population Health service, as outlined in the NHS 10 Year Health Plan, genomic data and systems will be capable of being linked with other clinical services to provide greater insight for our clinicians and patients, and to support the shift towards prevention, predictive testing and personalised medicine.
    - **NGIS / WGS:** To support the continued delivery of Whole Genome Sequencing (WGS) testing beyond the end of the Illumina contract in October 2026 the existing National Genomics Information System (NGIS) will be adapted by Genomics England to accommodate the reconfiguration to distributed sequencing, which will begin to be

delivered by designated NHS laboratories from April 2026. It will also be reconfigured to accommodate the introduction of digital Order Management.

- The NGIS Test Order Management Service (TOMS) will continue to provide a secure web interface for the collection or transcription of request data, until replaced by Order Management.
- The existing 100X series of message data interchanges will be retired and the existing send away process adapted to accommodate the revised sample pathways and dataflows between NHS GMSs and Genomics England.
- The ISO 13485 and ISO 15189 accredited NGIS Bioinformatics pipeline will continue to provide data processing of sequence data.
- In the short term the NGIS Interpretation Browser and Clinical Interpretation Application Programming Interface (CIPAPI) will continue to provide authorised scientists access to patient-level annotated genomic data and the tools to help interpretation. In the medium term the requirements for the integration of third-party tools for interpretation will become less demanding as the scalable architecture evolves in line with the UGR development.
- GEL will continue to transfer appropriately consented and deidentified data to the National Genomics Research Library.
- In the medium-term, elements of NGIS will migrate to use common NHS GMS infrastructure systems and standards. This will enable appropriate, Purpose Based Access (PBAC), to patient data via the UGR including laboratory scientists and, if necessary, clinician access to raw WGS data within each geography.

36. **Digital Genomic Test Service:** To provide access to the digital Test Directory and supporting reference datasets, the existing spreadsheet format test directories will be replaced by the Digital Genomic Test Service (DGTS), in the form of an interactive web portal, which is searchable by users, and supporting Application Programming Interfaces (APIs) to facilitate automated updates by downstream systems.

37. **Order Management:** To establish a consistent use of electronic ordering and results in genomics, the Genomics Order Management (OM) project will deliver several new capabilities:

- A HL7 Genomics FHIR specification conformant to UK Core R4 has been developed and will be maintained.
- A supporting genomic Master Data Set (MDS) has been published and will maintain a standardised data dictionary for genomic data interchange.
- The National Genomics Testing Process (NGTP), a generic process mapping tool used to describe all potential testing pathways, has been developed and will be

maintained, to ensure any specific pathways can be described using consistent terminology.

- The DGTS will be extended to include information on test routing, valid destinations, and other business rules necessary for delivering OM.
- A FHIR order broker is being developed to provide a central point of access for transferring request and result data between clinical requestors and all NHS GMS laboratories providing diagnostic services.
- A FHIR Implementation Guide (IG) has been published and will be maintained, supporting clinical and laboratory system providers to develop and validate native programmatic access to the order broker.
- The standards for order management will continue to be developed to include additional capabilities such as structured reporting.
- A national web ordering portal utilising the FHIR standard via the broker is being developed, to provide Order Management access to users who do not have native programmatic access via a local system, including primary care and neighbourhood teams.

38. **Unified Genomic Record (the UGR):** To provide broader access for clinical users and systems to make use of relevant patient genomic data, the UGR is being developed as the single point of truth for a patient's genomic data as well as limited clinical data /metadata in a FHIR record and will provide links to additional relevant clinical and diagnostic data. Building upon the technology, data standards and data stores used for Order Management, the UGR will provide a single, secure point of access to discover, retrieve and interpret the genomic patient records of NHS patients within England.

- The progress on UGR is being shared with the Single Patient Record, with the intention that the two programmes will be aligned and integrated.
- The initial priority services for UGR are Pharmacogenomics, Inherited Cardiovascular Conditions and Cancer Trials, but other pathways will be identified and developed.
- Patients will be able to view a complete account of their risk of major conditions and manage their personal health risks through the NHS App, drawing data from the UGR.
- The Health Data Research Service (HDRS) will draw pseudonymised genomic data from the UGR to provide research and innovation insights via existing secure data environments in the first instance, whilst HDRS develops.
- Alongside the NHS GMS and Genomics England, Our Future Health and UK Biobank will also develop the cutting edge digital infrastructure needed to deliver a comprehensive genomics ecosystem, enabling insights from multiple datasets to flow with the potential for genomics to contribute to half of all healthcare interventions by 2035. The UGR will be the primary clinical mechanism for access to these services and data.

- The Genomic Population Health Service will enable predictive data analytics utilising the standardised genomic and linked clinical data in the UGR to train AI tools and identify health risks.
- The Neighbourhood Health teams will use the UGR to provide bespoke support to help individuals manage any risks identified.

39. **Bioinformatics Common Cloud:** A standards-based infrastructure will be developed providing a common bioinformatics cloud platform to facilitate alignment of tooling for bioinformatic service delivery across the NHS (and Genomics England). This will enable co-ordinated development and roll-out of NHS-wide tools, reference datasets and platforms to enable translation of new technologies between NHS GMS geographies, prevent duplication of effort, and enable cost-effective scaling.

40. **Bioinformatic Shared Resources:** Bioinformatics assets and resources will be developed and maintained collaboratively by each NHS GMS across all geographies, to allow all patients to benefit from clinical translational efforts. These shared resources will include authoritative datasets defining clinical relevance of variants and genes, validated applications and pipelines. The establishment of national curation teams, national governance for change management, led by NHS GMS bioinformatics staff; will be scoped and will require an NHS GMS contribution, in addition to that of the Commissioner and from Genomics England.

41. **Bioinformatics Operational Standards:** Ways of working and governance processes will be defined by the Commissioner to standardise bioinformatics service delivery and development across NHS GMS. This will lead to creation of reproducible and NHS-portable tooling and pipelines, and adoption of technical data standards.

42. **Contract Monitoring Data**: To support the operational and financial management of the NHS GMS, the mandatory Patient Level Contract Management (PLCM) dataset has been defined. This dataset will be submitted via the Data Landing Portal (DLP), then distributed through the Data Services for Commissioners Regional Offices (DSCROs). In the medium-term development of contracting monitoring may require the development of Aggregate Contract Monitoring (ACM – Standard DCB2050) to further reduce the burden and allow simpler and more consistent reporting. Over the contract term, the PLCM dataset will evolve to include the new services, including potential work associated with Polygenic Risk Scores.

43. **LPP Procurement Framework:** The London Procurement Partnership (LPP) Clinical Digital Solutions (CDS) framework provides a mechanism for NHS and public sector

organizations to procure clinical digital solutions, including those related to genomics. A model genomics LIMS requirements catalogue has been produced and should be utilised to support future procurements.

44. **Registries and NDRS:** Clinical registries are powerful tools in healthcare that provide significant value across clinical care, research, policy, and system improvement by understanding disease patterns, treatment outcomes, and service needs. NHS GMS are required to support data collection for these registries, including but not limited to National Disease Registration Service (NDRS), ClinVar for both germline and somatic variants with classifications, and other external registries.

45. **Single Patient Record:** The Commissioner has committed to the delivery of a Single Patient Record (SPR) by 2028. The UGR will be the route by which genomic data is consolidated and standardised prior to incorporation into the single patient record.

46. **Federated Data Platform:** The Federated Data Platform (FDP) is a powerful platform for data analytics and enabling operational efficiencies. All national projects will be evaluated to ensure that FDP is used to enable more rapid and/or cost-effective implementation of digital services, where appropriate.

47. Other national components and projects may be identified. The scope and requirements for these will be developed in partnership with the NHS GMS geographies, for example the adoption of DICOM standards for digital images (FISH, karyotyping and whole slide digital pathology images) and membership of NPIC (the National Pathology Imaging Co-operative) by CPGCs.

48. The Commissioner will maintain a programme roadmap detailing the key project milestones and dependencies

49. The informatics standards, systems and services developed for the Genomics services will align with other national systems and standards, where relevant. These include:
    - the use of Personal Demographic Service (PDS) / Spine for maintaining a patient index;
    - the use of NHS Care Identity Service 2 (CIS2) / NHS Login / Enhanced Identity and Access Management Patterns for user authentication;
    - The use of API Management (APIM) for access to NHS healthcare programmatic interfaces.

50. Any services and projects should follow an alpha / private beta / public beta / live service release model as described in the UK Gov Agile Delivery Service Manual.
    - Projects in discovery or alpha will be limited to chosen partners and will not involve patient data.
    - Projects in private beta will be limited to chosen partners and will demonstrate capabilities in real clinical practice.
    - Projects in public beta will be available to all regions for clinical use, with specific access subject to agreement with the project team. Systems may be subject to more frequent functionality changes.
    - Projects in live service will be available to all regions for clinical use. Systems should be functionally stable.

51. Although primarily developed for NHS clinical use, options will be explored for the systems above to support collaboration with partners across public, private and charitable sectors.

52. Any record, case, sample or patient identifiers used in the delivery of the services listed above should be considered potentially personally identifying. These should be removed during any data de-identification processes for research or education purposes.

53. Secondary use of the UGR and Order Management is designed to provide pseudo anonymous patient-based data for purposes other than direct clinical care such as healthcare planning, commissioning, public health, clinical audit and governance, benchmarking, performance improvement, medical research and national policy development to users, Commissioners and Providers of NHS funded care.
    - Access to data for secondary uses will be aligned with Health Data Research Service (HDRS) as those standards evolve.

Regional Assurance processes

54. The NHS GMS must develop and maintain fit-for-purpose processes in the following areas. These should meet the legal assurance requirements of their local trusts and should provide sufficient assurances as required by other providers across their geography.
    - Clinical Safety, including the maintenance of all documentation pertaining to DCB0160 and continued active engagement with the national genomics Clinical Safety Group, as part of the local trust processes.
    - Information Governance, including Information Asset Registers.
    - Business Continuity / Disaster Recovery.

- Cybersecurity.
- Data and Digital Risk Management.
- Programme and Project Governance.

55. The NHS GMS must develop and maintain a regional Data and Digital Strategy and Roadmap. These should include:
    - NHS GMS data and digital service development and improvements.
    - The delivery and maintenance of NHS GMS internal systems, including LIMS and bioinformatics systems.
    - The adoption of cloud services and capacity planning for NHS GMS internal systems, particularly bioinformatics systems.
    - The development of interoperability between genomic laboratories and NHS CPGCs.
    - Increased interoperability with provider clinical systems (such as primary and acute EPRs, and pathology and SIHMDS LIMS) across their geography.
    - The regional adoption of national genomic systems and standards.
    - When introduced and the production of polygenic risk scores within genomic reports coupled with other data.
    - Access and connectivity with Artificial Intelligence (AI) systems for research and development, and translation paths for the introduction of AI tools into clinical use.
    - The roadmap should be similar in format, and reconciled with, the national roadmap.

56. The NHS GMS must ensure that the LIMS used in their genomic laboratories meet community standards for functionality and efficiency, are supported by their supplier, and are promptly upgraded to enable interoperability with national systems and standards as these are developed and made generally available.

57. The NHS GMS should ensure that the Electronic Patient Records (EPR), LIMS and other relevant systems used by referring providers in their geography, support the interoperability standards required to connect to genomics systems and services, and they commit to upgrading their systems as new standards are introduced.

58. The NHS GMS Data and Digital Strategy must be aligned with strategies developed by the NHS Trusts within the NHS GMS geography and relevant ICB informatics strategies. The NHS GMS must be able to demonstrate support from the Chief Information Officers (CIOs) / Chief Digital and Information Officers (CDIOs) at both their provider trusts and across their geography.

Data and Digital Service Management

59. The NHS GMS is required to have appropriate processes in place to assure the operation and delivery of all existing and new genomic Data and Digital systems used in their geography. These should be aligned with Information Technology Infrastructure Library (ITIL) or a similar service management framework; and should be able to demonstrate integration with local trusts and/or ICS service management processes, where appropriate.

60. The NHS GMS Lead Provider should establish appropriate processes for incident and request management. This should include consideration of incidents, notifications and requests from:
    - NHS GMS staff.
    - Local and regional clinical users across both primary and secondary care.
    - Trust, ICB and other provider informatics services.
    - Other regional NHS GMSs.
    - The Commissioner and national system delivery partners.

61. The NHS GMS should establish appropriate processes for change and configuration management. This should include consideration and approval of change requests from within the NHS GMS, along with the impact of changes to / from national, local trusts and any other connected systems.

62. The NHS GMS should maintain service documentation in an appropriate format and location.

63. The NHS GMS should maintain a service catalogue, understanding the systems in use, their existing contractual and/or support status, and any other relevant details.

64. The NHS GMS should have appropriate capacity planning and monitoring in place, in particular for: storage, network bandwidth and compute capacity.

65. The NHS GMS should incorporate processes for managing incidents and problems, ensuring timely resolution and minimising service disruptions.

66. The NHS GMS should ensure that adequate data backup and service continuity plans are in place to avoid data loss and maintain service availability during disruptions. Including evidence that plans have been validated.

67. The NHS GMS should have local data retention policies in place, and processes in place to ensure compliance.

68. The NHS GMS should have processes to prioritise and deliver service improvement across data and digital services.

69. The NHS GMS should have process in place to accept the transition of projects, both local and national, into business-as-usual service, including the ongoing maintenance and operations of new services following project closure.

70. The NHS GMS should monitor and ensure compliance of local system with any nationally published information standards notices (ISNs)

Bioinformatics Services

71. The NHS GMS is required to support bioinformatics service delivery, maintenance of bioinformatics resources, continual improvement of bioinformatics clinical utility, and ensuring that methods are standardised across the NHS, in line with priority three of the Accelerating Genomic Medicine strategy.

72. Bioinformatics service delivery in the NHS GMS must be accredited to ISO 15189:2022.

73. Bioinformatics development in the NHS GMS must be performed according to ACGS best practice and other appropriate guidelines and be focused on improving clinical utility and patient impact.

74. The NHS GMS must establish specific processes for the change management of bioinformatics and scientific systems and standards, especially where pathways are delivered across multiple diagnostic providers. The NHS GMS must meet the requirements of UKAS TPS71.

75. The NHS GMS must adopt and adhere to appropriate bioinformatics standards, as identified by the Commissioner, such as GA4GH.

76. Bioinformatics tooling and resources should be developed collaboratively across all NHS GMS geographies, with a requirement to be able to be adopted nationally by all.

77. Bioinformatics algorithms and software should be validated collaboratively across all NHS GMS geographies whenever possible. Collaborative working will be enabled by the establishment of the common cloud infrastructure and shared sandpit for bioinformatics development, with the advent of the UGR.

78. The NHS GMS must submit variants and their classifications to the required repositories, including NDRS (under the National Disease Registries Directions 2021), ClinVar (both germline and somatic variants with classifications), and other disease-specific registries.

79. The NHS GMS must support the development of a national bioinformatics strategy and must develop a local strategy for its implementation.
    - The national and NHS GMS bioinformatics strategies must prioritise data, such as variants detected and their clinical significance, being easily accessible to the whole NHS GMS. Bioinformatics databases and data repositories shall be considered essential NHS GMS infrastructure.
    - The strategy will consider the ability to aggregate sequencing data as well as variant level interpretation data, in accordance with national interpretation guidelines for cancer and rare disease (e.g. SVIG, Canvig-UK, ACGS).
    - These data must be shared securely, using recognised industry formats/protocols and according to approved consent and data governance standards (where these have been finalised).
    - These data will be used to power national genomic knowledgebases to support and expedite interpretation of genomic data within the NHS.

Data and Digital Transformation and Programmes

80. The NHS GMS is required to support the transformation of internal NHS GMS systems such as LIMS, and other provider data and digital systems in their geography, to deliver local improvements as required, and to support the adoption and use of the data and digital systems and services being delivered under the Accelerating Genomic Medicine strategy. In order to deliver these necessary transformations, the NHS GMS should have the following processes in place and integrated with local trust project governance where relevant.
    - The NHS GMS should maintain a process for the development, prioritisation and approval for future transformation requests, including the development of business cases.
    - The NHS GMS should develop and maintain processes for the management of programmes and projects.
    - The NHS GMS should identify a source for project assurance and PMO functions.

- The NHS GMS should identify or establish appropriate bodies to ensure sufficient level of stakeholder engagement across their region.
- The NHS GMS should identify or establish process to ensure regional benefits are identified, mapped and their realisation tracked.

Data and Analytics Services

81. The effective delivery of the NHS GMS relies on a robust and responsive data and analytics infrastructure. The data and analytics service underpins the contractual, operational, clinical, and strategic functions of the NHS GMS by ensuring accurate and timely collection, analysis, and reporting of high-quality data. It supports performance monitoring, financial accountability, equitable access to genomic services, the identification of health inequalities across populations, and service developments review for both genomic testing and supporting clinical pathways.

82. The NHS GMS is required to ensure accurate and timely data to collate robust operational performance and finance reporting, promoting equity of access/Population Health analysis and addressing health inequalities, and service developments review for both genomic testing and supporting clinical pathways.

83. The NHS GMS is required to support and flow contract monitoring data, including but not limited to: the Patient Level Contract Monitoring (PLCM), Backlog reporting, Waiting List MDS, Indicative activity plan (IAP), ClinVar submissions.

84. The NHS GMS must maintain an Information Asset Register to ensure legal and regulatory obligations under the UK General Data Protection Regulation (UK GDPR) and the Data Protection Act 2018, ensuring organisations know what personal data they hold, how it is processed, and who it is shared with.

85. The NHS GMS is required to support the annual planning exercise, aligned with the Commissioner's planning guidance.

86. The NHS GMS is required to identify and support any data development requirements or data quality issues by utilising Data Quality Improvement Plans (DQIP).

87. It is expected that NHS GMS are required to support the uptake of the Federated Data Platform (NHS FDP) aims to enhance patient care and increase efficiency. It securely connects data, breaks down information silos, and provides insights to assist in decision-making, reduce costs, and improve patient outcomes.

Specific deliverables

88. In addition to the general requirements for processes, systems and assurances documented above, there are a number of specific data and digital deliverables required from each NHS GMS geography.

Data Management deliverables

89. The NHS GMS must ensure that all genomic laboratories in their region are submitting Patient Level Contract Monitoring (PLCM) data to the standards and timelines agreed with the Commissioner.

90. The NHS GMS should ensure that any other supplementary datasets are also submitted to the standards and timelines agreed with the Commissioner.

91. The NHS GMS must review any quality metrics available regarding their PLCM and other data submissions and must maintain data quality improvement plans to remedy any known issues.

92. The NHS GMS should engage with any national working groups established to identify and agree relevant coding standards and develop guidelines for their use. These standards include, but are not limited to, SNOMED-CT and HPO. The NHS GMS must adopt these standards once agreed.

93. The NHS GMS must ensure that any processes used to deliver genomic or other data for research or secondary uses are appropriately de-identified.

Informatics Management deliverables

94. The NHS GMS should engage with any national projects requiring support for the development of national model clinical safety cases. The NHS GMS must then support any necessary local adaption and approval processes to permit their use.

95. The NHS GMS should engage with any national projects requiring support for the development of model data protection impact assessment, or other information governance documentation. The NHS GMS must then support any necessary local adaption and approval processes to permit their use.

96. The NHS GMS must develop and maintain appropriate business continuity and disaster recovery processes, to ensure service continuity in the event of a disruption to a

national system, or to notify or update national systems, if necessary, in the event of a local system disruption.

97. The NHS GMS should engage with any national projects requiring support for cybersecurity assessments and must support any local remediation of systems and processes needed due to any vulnerabilities identified.

98. The NHS GMS must identify the impact of any transformation projects on local workforce and service management processes and ensure consideration of these is included in local implementation planning.

99. The NHS GMS must ensure that all NHS GMS systems holding patient data are aligned with NHS Number standards, use NHS number when interacting with genomic systems, and ideally are synchronised with PDS for demographic data.

100. The NHS GMS should ensure that all NHS GMS systems holding patient data are aligned with the use of CIS2 /NHS Login / Enhanced Identity and Access Management Patterns for user authentication.

Bioinformatics deliverables

101. The NHS GMS must contribute to the development of a national bioinformatics strategy, as described above.

102. Once defined, the NHS GMS must demonstrate implementation of the national bioinformatics strategy within their geography that is aligned with implementation across the whole NHS GMS.

103. The NHS GMS must adopt cloud technologies within their regional bioinformatics strategy. This is likely to be assisted by access to a standards-based, national cloud environment, with nationally negotiated pricing.

104. The NHS GMS should engage with the development of community cloud systems and standards for use across all NHS GMS cloud services. Once established, the NHS GMS must migrate to those standards within agreed timescales.

105. The NHS GMS must promote and implement reusable bioinformatics tooling, adopting standards and adhering to best practice.

106. The NHS GMS must adopt a national collaborative approach to bioinformatics development and validation, and work to remove any barriers to working collaboratively between regions.

107. The NHS GMS must collaboratively create shared knowledgebases to ensure that all NHS GMS can use data generated anywhere within NHS GMS for patient benefit.

108. The NHS GMS must collaborate with NHS, academic and industry partners to create and improve bioinformatics systems with clinical utility, ensuring that NHS GMS and NHS interests are protected.

109. The NHS GMS must collaborate on data strategies and critical use cases for development and deployment of AI/ML technologies. Foundational efforts are likely to be necessary to ensure genomic data quality and accessibility.

Transformation and programmes deliverables

110. To prepare for the UGR and the NHS GMS Network Model, developing the national genomics digital infrastructure will be a key enabler as the service rapidly scales and develops.

111. The NHS GMS Network must be:
    - Capable of providing a reliable, predictable and seamless collaboration space service for users (both in terms of data availability and services across the Network);
    - Able to operate as one Network, capable of working as a single clinical scientific and bioinformatics workforce to manage genomic interpretation from a prioritised specialist workflow and/or collaborate on bioinformatics development;
    - Have a commissioning structure which provides stability and some certainty in terms of funding, to enable longer term planning in terms of strategic development and workforce;
    - Separate out service delivery and data controllership and storage; and
    - Be able to respond with agility to support HDRS strategic and tactical imperatives

112. The NHS GMS must engage with the national programme and any national projects. In particular:
    - The NHS GMS should review and respond to national requirement specifications and architecture proposals.
    - The NHS GMS should participate in the testing and evaluation of national deliverables.

- The NHS GMS Data and Digital Director must communicate programme activities within their regional leadership structure and regional informatics community.
- The NHS GMS must identify relevant provider systems for interoperability within their geography, and prioritise their connectivity based on clinical benefit, timetable and upgrade opportunities, and ease of implementation.
- The NHS GMS must maintain and share a regional informatics milestone delivery plan for the adoption of national systems and standards across identified systems, and ensure this remains aligned with any national milestones in the Data and Digital Programme Tube Map, and any local changes in connecting systems.
- The NHS GMS must plan for ongoing adherence to updated standards, including FHIR. It is expected that new versions must be incorporated into any already connected systems within 12 months from the publication of a formal update. NHS GMS and their system suppliers will have access to proposed changes prior to finalisation.
- The NHS GMS must have a strategy for engaging with the use of Artificial Intelligence (AI) for bioinformatics, clinical and operational uses, including partnerships with industry and academia, and secure routes of access to genomic data for training and validation.

113. The NHS GMS must engage with the **distributed WGS** reconfiguration project. In particular:
    - The NHS GMS must support any necessary reconfiguration of systems to support distributed WGS processes according to the timelines set by the project. These include, but are not limited to: order entry processes, the provision of sequence and sample data to Genomics England, and interpretation and reporting via the Interpretation Browser). The technical specification is set out in **Annex 3.**
    - The NHS GMS must develop a detailed regional plan for transition, including business continuity options for connecting to alternate sequencing centres.

114. Initial National Designated NHS GMS Lead Provider(s) sequencing centres must be reconfigured, quality assured and live by 1st April 2026.

115. All NHS GMSs should be capable of sending to a remote sequencing centre via a send-away process, instead of the Plater, by 1st May 2026.

116. Phase 2 sequencing centres, as identified by the project, must be reconfigured, quality assured and live by 1st August 2026.

117. All identified sequencing centres must be running at their allocated capacity by 1st October 2026.

- The NHS GMS must support the delivery of bandwidth upgrades and any other necessary informatics infrastructure upgrades, as agreed with commissioners.
- The NHS GMS must support the decommissioning of any legacy WGS processes, such as audit logs, and plater processes.

118. The NHS GMS must engage with the DGTS project. In particular:
    - The NHS GMS must prepare and execute a transition plan to move to the new system and standards. This includes planning for LIMS import or integration processes and updating LIMS booking processes where needed.
    - The NHS GMS is responsible for defining, validating, and maintaining the routing guidance rules for within their region, to the standards defined by the DGTS.
    - The NHS GMS should participate in any reviews of nationally commissioned changes in the DGTS publication process.
    - The NHS GMS should maintain regional, trial, and non-NHS commissioned test package and test data within the DGTS.
    - The NHS GMS must identify, map and establish processes to be the primary point of contact to support all requesting and laboratory systems who need to accept and integrate changes in the DGTS.

119. The NHS GMS must engage with the Order Management (OM) project and contribute to the development of associated national standards. In particular:
    - The NHS GMS must promote the use of the national OM broker across trusts within its Geography and understand any localised dependencies for adoption.
    - The NHS GMS must advocate for the adoption of FHIR-based integration and should collaborate with regional trusts and their EPR/LIMS vendors to prioritise native interoperability.
    - The NHS GMS must develop a detailed regional plan for adoption, including consideration of the preferred modes of adoption for each requestor (web portal, hybrid integration, or native FHIR integration).
    - When OM enters the private Beta phase:
        - The NHS GMS should review the progress at the evaluation sites, and identify any additional use cases relevant to their region.
        - The NHS GMS should explore options for connectivity with their Genomics LIMS suppliers.
    - When OM enters the public Beta phase:
        - The NHS GMS must prioritise establishing connections with all genomics LIMS in their region, with native or hybrid integration preferred, and the ability to be able to publish detailed status updates, and pass a technical assurance assessment.

    - All genomic LIMS must be capable of receiving and sending send-away requests from other regions via OM within 6 months.
    - The NHS GMS should prioritise connecting with their higher volume requesting systems (pathology LIMS, CPGCs, SIHMDS and EPRs), with native or hybrid integration preferred.
    - The NHS GMS will be able to connect any / all requestors with OM during public beta, with agreement from the project team.
- Once OM enters live service:
    - The NHS GMS must transition any remaining unconnected genomics LIMS from national portal access to native or hybrid integration within 12 months.
    - The NHS GMS must have at least their five highest volume requestors connected to OM within 12 months.
    - The NHS GMS must have all remaining requestors connected to OM within 24 months.

120. The NHS GMS must engage with future phases of the order management project, where the functionality may be extended to cover areas such as structured reporting, and point of care or near patient testing.

121. The NHS GMS must engage with the **UGR** project and the development of associated standards. In particular:
    - The NHS GMS should identify and support the documentation of any emerging clinical and research use cases.
    - The NHS GMS must identify regional priorities, opportunities and blockers that affect adoption of UGR across regional systems. This includes identifying training needs and other digital readiness activities.
    - The NHS GMS should collaborate with ICBs to ensure UGR-related data flows and Decision Support are incorporated into regional digital and clinical strategies.
    - The NHS GMS must support alignment between UGR services and local / neighbourhood health models; and must advocate for and support the integration of UGR services into regional Electronic Health Records (EHRs) and clinical systems.
    - The NHS GMS must support the regional adoption of UGR services that inform and provide access to the genomics population health service. This includes the ability for clinical systems to receive and act upon alerts regarding increased patient risk.
    - The NHS GMS should ensure that UGR data contributions meet quality and semantic interoperability standards.
    - The NHS GMS must contribute to the national evidence base for clinical utility, equity, and outcomes related to UGR-enabled services.
    - The NHS GMS must develop and implement a detailed regional plan for adoption of all UGR services. Currently this includes pharmacogenomics, inherited cardiovascular

conditions and clinical trials, but other services will be introduced over time, for example the population / neighbourhood services by 2028.

- When a UGR service enters alpha or private Beta phase:
    - The NHS GMS may have the opportunity to be chosen as an evaluation implementation site.
    - If not chosen to be an evaluation site, the NHS GMS should monitor the progress, should highlight any additional use cases or factors specific to their region, and should plan for regional implementation opportunities in public Beta.
- When a UGR service enters public Beta phase:
    - The NHS GMS should implement regionally in prioritised areas.
    - The NHS GMS must plan for regional implementation in live service. This includes adoption by regional services in addition to any laboratory functionality.
- When a UGR service enters live service:
    - The NHS GMS must implement regionally according to agreed timescales. This includes adoption by regional services in addition to any laboratory functionality.

122. The NHS GMS must engage with and support the development of **national bioinformatics strategy**, and the development of associated projects or standards that may include:

- innovation and transformation projects to increase NHS GMS productivity and patient outcomes;
- point of care and near patient testing requiring bioinformatics approaches;
- highly scalable, industrialised bioinformatics to support very high throughput genomics applications;
- novel biomarker discovery and delivery for NHS GMS;
- sophisticated Decision Support for clinical scientists that leverages NHS GMS and external data resources;
- bioinformatics to enhance newborn screening tests;
- bioinformatics to enhance adult population screening tests; and
- horizon scanning to identify future technologies applicable to NHS GMS patients such as multiomics approaches.

Annex 1 – NIPT Service Specification

To note this Specification will be updated following UK National Screening Committee recommendations.

Definitions	"**NIPT Service Provider**" means the contract holder for the NIPT service. In this Specification the NHS Genomic Laboratory Hub (GLH). "**NIPT Laboratory Provider**" means the laboratory providing the testing, analysis and reporting of the NIPT sample, where this is different from the NIPT Service Provider.

1 Population Needs

1.1 Context and evidence base

1.1.1 The UK National Screening Committee ("**UK NSC**"), through NHS England has recommended that NIPT be assessed as an additional option to the current screening pathway for Down's syndrome (T21), Edwards' syndrome (T18) and Patau's syndrome (T13) for women with chance results greater than or equal to 1 in 150 at term (1 in 2 to 1 in 150).

1.1.2 The recommendation from the UK NSC is to undertake an evaluative approach to introducing the offer of NIPT, as an additional option, for those women with a higher chance result of 1 in 2, to 1 in 150 at term following an NHS combined or quadruple screening test result.

1.1.3 The UK NSC commissioned a full review of the published scientific and cost evidence (systematic review) relating to NIPT. This was presented to the UK NSC in June 2015. A formal announcement following the UK NSC recommendations was made by the Department of Health on 29 October 2016. Please see the PDF at the following hyperlink for a brief summary on the purpose of non-invasive prenatal testing from the UK NSC; https://legacyscreening.phe.org.uk/policydb_download.php?doc=602.

1.1.4 A further review was undertaken in 2019 to include the offer of NIPT screening in twin pregnancies as part of the evaluation roll-out. In addition, the UK NSC also updated the approved technology for use in NIPT screening as an additional option to the current screening pathway for Down's syndrome, Edwards' syndrome and Patau's syndrome for women with chance results greater than or equal to 1 in 150 at term (1 in 2, to 1 in 150). The NIPT laboratory provider must supply the class of tests which were evaluated in the two Warwick reviews. These were based on sequencing and microarray methodologies. Elements of the test process

may be modified but the overall methodology must remain sequencing and microarray based. This will be effective for the duration of the Contract.

1.1.5 NIPT is a technique that can be used to screen for Down's syndrome, Edwards' syndrome and Patau's syndrome during pregnancy. It involves taking a sample of blood from the pregnant woman. The mother's blood contains a mixture of maternal DNA and the placental DNA. This is known as the total cell free DNA ("**cfDNA**"). In most cases, the placental DNA will be the same as the baby's DNA. The contribution of DNA from the placenta is called cffDNA.

1.1.6 cffDNA can be detected in maternal plasma as early as 5 to 7 weeks gestation. However, test results are more accurate after 10 weeks because the amount of cffDNA increases over time. cffDNA remains in the maternal circulation for only a few hours after each pregnancy, making it suitable for pregnancy-specific testing.

1.1.7 The evaluative roll-out will last for three years after which recommendations will be made by the UK NSC about the future national NHS screening policy for NIPT.

1.1.8 The service will continue for the Contract Term to enable a full evaluation report to be produced and submitted. The report will include full outcome data and will be completed and submitted to the UK NSC before the end of this contract. The UK NSC will review the evidence in the outcome report and make a recommendation advising Ministers and the NHS on implementation of NIPT as formal part of NHS FASP.

1.1.9 Rationale:

The NIPT evaluative roll out formally runs from 1 June 2021 to 31 May 2024. Data about the testing process such as test turnaround times and the timeliness of the pathway is monitored monthly and quarterly, and data is readily available. Complete data will be presented by December 2025 to allow for outcomes to be recorded for testing undertaken at the end of the evaluation.

Full data relating to the performance of NIPT in the higher chance population requires following up the outcomes of all pregnancies and postnatal diagnoses. Full reporting, analysis and reporting will only be made available from December 2025 due to the inherent delays due to birth outcomes and registrations.

This is an annual population of around 15,000 and therefore numbers are small for single years. This makes it important to fully evaluate the pooled data at the end of the three years.

1.1.10 For a final report capturing the full 3 years of data, reporting will continue in line with specifications outlined in section 8 data reporting. The NHS England report in March 2026 will capture data from 1 June 2021 to May 2024 (it includes data from April and May 2024).

2 Outcomes

2.1 Commissioner Responsibilities

2.1.1 The Vaccination and Screening directorate (V&S) forms part of NHS England and exists to protect and improve the nation's health and wellbeing and reduce health inequalities. This is achieved through world-leading science, knowledge and intelligence, advocacy, partnerships, and the delivery of specialist public health services.

2.1.2 The NHS Fetal Anomaly Screening Programme (NHS FASP) is one of the 11 National screening programmes in England and offers screening for Down's syndrome, Edwards' syndrome, Patau's syndrome and 11 physical conditions to all eligible pregnant women as part of their routine antenatal care.

2.1.3 NHS England V&S leads the NHS FASP antenatal screening programme.

2.1.4 The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) was set up in 2015 and collects data on congenital anomalies across England. NCARDRS supports the evaluative roll out of NIPT by collecting data from genomic laboratories on NIPT and cytogenetic tests and the biochemistry screening laboratories on combined and quadruple tests.

Data shows that between 1 July 2021 and 30 September 2022:

- 15,000 NIPT samples tested over the 3 laboratories
- Four fifths followed a higher chance combined test result
- One fifth of NIPT tests followed a higher chance quadruple test
- A result was reported for 96% of these 15,000 samples
- 86% of NIPT results were reported within 5 days
- 7% of NIPT samples tested were reported as higher chance results for one or more of the 3 conditions

Data from maternity services so far shows:

- around three quarters of the women with a higher chance combined or quadruple test chose to have a NIPT screening test and
- 97% of women with a higher chance or a no result NIPT result attended an appointment within 3 days to discuss their results

2.2 Specific defined outcomes

2.2.1 For clarity the service will be an evaluative roll-out. The NIPT Service Provider will be expected to provide feedback and information reporting as

set out in paragraph 8 (Data Reporting) of this Service Specification, as well as adapting the Services as required by NHS England throughout the Term of the Contract. This will enable the programme leads to evaluate the roll-out at the stage identified in paragraph 8 (Data Reporting) of this Service Specification; ensuring any required changes to the pathway and/or screening process can be made efficiently and effectively. If it becomes necessary, the UK NSC would also be able to make a recommendation to cease use of NIPT as part of the NHS screening pathway.

3 Scope

3.1 Aims and Objectives of Services

3.1.1 In delivering the aims and objectives of the Services the NIPT Laboratory Provider will meet all the key indicators (e.g. sample turnaround times) as set out in paragraph 3.7.1 of this Service Specification.

3.1.2 Based upon the UK NSC recommendation in 2019, the overall aims and objectives of the Services are described below.

3.1.3 Pregnant women are already offered a screening test for Down's syndrome, Edwards' syndrome and Patau's syndrome from 10-14 weeks of pregnancy (the NHS combined test, involving an ultrasound scan and blood test), or a screening test for Down's syndrome only (the NHS quadruple test, involving a blood test alone) if booking between 14-20 weeks.

3.1.4 If the NHS screening test results shows that the chance of having a baby with Down's syndrome, Edwards' syndrome and Patau's syndrome is higher than 1 in 150 at term, this is called a higher- chance result. Currently, women who have an NHS higher chance result have the option of having an invasive diagnostic test (amniocentesis or CVS).

3.1.5 The proposed change is for Non-Invasive Prenatal Testing to be offered to women who are deemed at higher chance following the current primary screen. NIPT is not diagnostic, and an invasive diagnostic test is still required to receive a definitive diagnosis.

3.1.6 Key findings supporting the UK NSC recommendation:

i. an invasive diagnostic test carries a small risk of miscarriage. The evidence suggests that NIPT will reduce the number of women being offered an invasive test.

ii. however, while we know that the accuracy of NIPT is good, we don't yet know how it will perform in an NHS screening programme pathway (hence the evaluative roll-out service commissioned here).

iii. for women who choose to have NIPT, this will add in an extra step in the screening programme. The impact of this, and the choices women

make at different points in the pathway, is something that we hope to gain a better understanding of through further evaluation.

3.1.7 A recommendation has therefore been made to evaluate the introduction of NIPT for Down's syndrome, Edwards' syndrome and Patau's syndrome screening. This will include scientific, ethical and user input to better understand the impact on women, their partners and the screening programme around the offer of NIPT or invasive testing following a screening test result where:

i. the NHS screening test chance result for Down's syndrome is greater than or equal to 1 in 150 at term;
ii. the combined test chance result for Edwards' syndrome and Patau's syndrome is greater than or equal to 1 in 150.

3.1.8 The main aims and objectives of this laboratory service are:

I. To allow women to access the NIPT Services with rapid turnaround times to enable appropriate clinical decisions to be taken, at an appropriate time during pregnancy. For clarity, access will be via maternity services if the patient is confirmed as meeting the eligibility criteria which is a screening chance result from NHS combined or quadruple test greater than or equal to 1 in 150 at term.
II. Support the model for the NHS England, as recommended by UK NSC for evaluation as an additional option to the current screening pathway for Down's syndrome, Edwards' syndrome and Patau's syndrome for women with chance results greater than or equal to 1 in 150 at term (1 in 2 to 1 in 150).
III. The NIPT Service Provider will submit data as specified in Table 2: Minimum demographic and result data table to NCARDRS. In addition to this data collected in relation to NIPT samples analysed in line with the NHS England Genomics Unit Test Directory code R445 (NIPT for previous trisomies) will be separated and also submitted.
IV. The extension of the contract period enables eligible pregnant women in England to continue to be offered NIPT as part of the NHS FASP pathway whilst NHS England collects, analyses and prepares a final report for the UK NSC including data and information on the pregnancy outcomes of the cohort of women from the evaluation period. Further details regarding what evidence is required and the frequency of submission is detailed in clause 8 (Data Reporting) of this Service Specification.
V. Set out the roles and responsibilities of the NIPT Service Provider in delivering the Services and meeting the key performance indicators regarding data requirements specified in paragraph 8 (Data Reporting) of this Service Specification. The NIPT Service Provider agrees, as further detailed in the description of the NIPT Services below, to be responsive to the findings of the evaluative roll-out as it proceeds.
VI. The NIPT Laboratory Provider commits to working with the NHS England to develop, adapt and modify the laboratory pathways, ordering

systems, sample management processes and data requirements as the evaluative roll-out progresses. Developments of the pathways and processes would happen as and when required during the evaluative roll-out period.

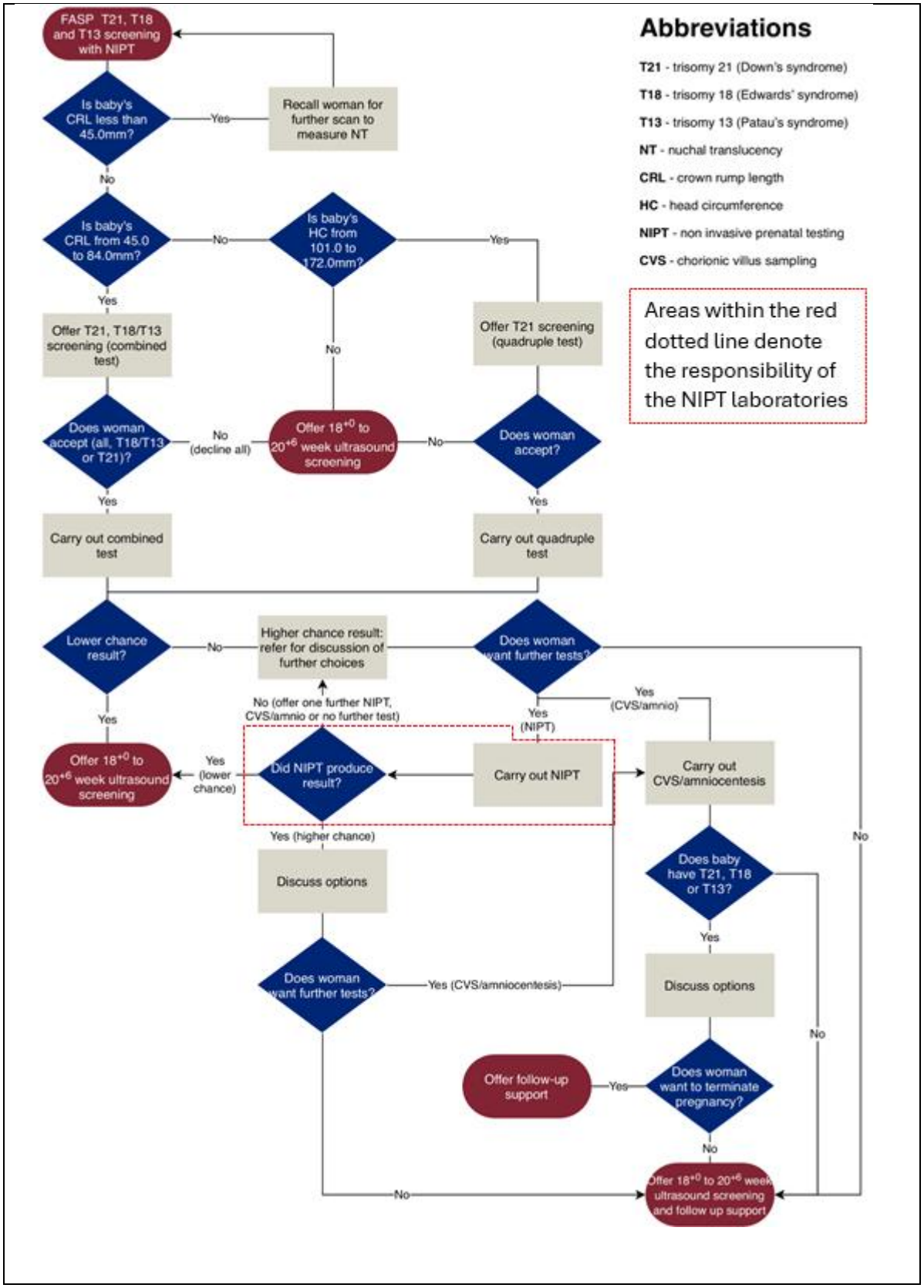
3.1.9 The NIPT Service Provider must comply with all data reporting requirements to fulfil both the on-going quality assurance of laboratory services for NHS England and shorter-term reporting to inform and support the evaluative roll-out of NIPT as part of the screening pathway.

3.2 Laboratory Service description

3.2.1 Genomic testing for the NHS in England is commissioned by NHS England and undertaken through the National Genomic Testing Service and outlined in the National Genomic Test Directory. The National Genomic Testing Service consists of seven Genomic Laboratory Hubs ("GLH") and subcontracted Local Genomic Laboratories ("LGL") and Designated Provider(s). The GLH National Network, working collaboratively, operates a world class resource for the NHS in England to underpin an NHS Genomic Medicine Service and works to a nationally agreed set of standards, quality management and assurance of all processes and data. In order to ensure that the genomic testing element of the NHS FASP NIPT service is an integrated part of the National Genomic Testing Service the NIPT Laboratory Provider shall be part of a GLH either as the lead GLH provider or under a chain of formal sub-contracting agreements to the NIPT Service Provider). Evidence should be provided as to how its provision, governance and quality management of NIPT sits in the context of the overall services provided by the NIPT Service Provider. The NIPT Laboratory Provider must be accredited to ISO 15189:2012 (and in transition to ISO 15189:2022) by United Kingdom Accreditation Service (UKAS) and must participate in an ISO 17043 accredited external quality assessment scheme.

3.2.2 Where NIPT Services are sub-contracted by a NIPT Service Provider the sub-contractor responsible for delivering the services should be defined as an NIPT Laboratory Provider and will comply with the requirements set out in this specification. For clarity, it is the responsibility of the NIPT Service Provider as the Contract holder to ensure Service Specification compliance of any sub-contractor (NIPT Laboratory Provider) and the NIPT Service Provider shall be fully responsible for the delivery of the Services and this Service Specification, irrespective of its contractual relationship with the NIPT Laboratory Provider where the Services are in whole or in part sub-contracted. Where such NIPT Services have been sub-contracted out the NIPT Service Provider lead shall ensure the NIPT Laboratory Provider complies with the provisions and requirements as set out in this Service Specification, including KPIs and performance quality requirements and the terms and conditions of this contract in their entirety.

3.2.3 The following diagram (Table One) is the applicable NHS FASP NIPT Care Pathway which clearly lays out the process that must be followed. The NIPT Laboratory Provider is responsible for carrying out the activities illustrated within the red dotted line, but not the other activities illustrated in Table One.


FASP T21, T18 and T13 screening with NIPT
Is baby's CRL less than 45.0mm?
Yes
Recall woman for further scan to measure NT
No
Is baby's CRL from 45.0 to 84.0mm?
No
Is baby's HC from 101.0 to 172.0mm?
Yes
Yes
Offer T21, T18/T13 screening (combined test)
No
Offer T21 screening (quadruple test)
Does woman accept (all, T18/T13 or T21)?
No (decline all)
Offer 18+0 to 20+6 week ultrasound screening
No
Does woman accept?
Yes
Yes
Carry out combined test
Carry out quadruple test
Lower chance result?
No
Higher chance result: refer for discussion of further choices
Does woman want further tests?
Yes (CVS/amnio)
No (offer one further NIPT, CVS/amnio or no further test)
Yes (NIPT)
Yes
Offer 18+0 to 20+6 week ultrasound screening
Yes (lower chance)
Did NIPT produce result?
Carry out NIPT
Carry out CVS/amniocentesis
Yes (higher chance)
Discuss options
Does baby have T21, T18 or T13?
No
Yes
Does woman want further tests?
Yes (CVS/amniocentesis)
Discuss options
No
Offer follow-up support
Yes
Does woman want to terminate pregnancy?
No
No
Offer 18+0 to 20+6 week ultrasound screening and follow up support
Abbreviations
T21 - trisomy 21 (Down's syndrome)
T18 - trisomy 18 (Edwards' syndrome)
T13 - trisomy 13 (Patau's syndrome)
NT - nuchal translucency
CRL - crown rump length
HC - head circumference
NIPT - non invasive prenatal testing
CVS - chorionic villus sampling
Areas within the red dotted line denote the responsibility of the NIPT laboratories

3.3 Testing Framework

3.3.1 The NIPT Laboratory Provider shall perform NIPT and NHS FASP reporting on Down's syndrome, Edwards' syndrome and Patau's syndrome only in singleton and twin pregnancies with no inclusion of fetal sex or other chromosomal findings.

3.3.2 The NIPT Laboratory Provider shall only undertake NIPT as part of the NHS screening programme:

3.3.2.1 for those women with a higher chance result of 1 in 2 to 1 in 150 at term from combined or quadruple testing offered, as evidenced by the referring maternity services; and

3.3.2.2 where there is evidence provided by the referring maternity services to the NIPT Laboratory Provider that the woman was offered the clinical options as described in the NHS FASP pathway (Table One at paragraph 3.2.4 above) prior to their sample being taken (the "NIPT Eligibility Criteria" – paragraph 3.3.3 below).

3.3.3 Women who have received a higher chance result (1 in 2 to 1 in 150 at term) from either the NHS combined or quadruple tests who choose to have NIPT may accept the offer of screening for:

i. Down's syndrome only,
ii. Edwards' syndrome and Patau's syndrome only
iii. All 3 conditions

3.3.4 The initial NHS FASP NIPT Eligibility Criteria check will be performed by the maternity services that refer the service user to the NIPT Laboratory Provider. The NIPT Laboratory Provider will be required to check that the information provided by the maternity service in the pathology test request indicates the sample meets the Eligibility Criteria prior to processing the sample.

3.4 Sample collection, handling and processing

3.4.1 The NIPT Laboratory Provider must create, maintain and comply with ISO 15189 and compliant Standard Operating Procedures ("**SOPs**") setting out processes in respect of sample receipt, storage, extraction, preparation, sequencing, analysis (including bioinformatics), transportation, and a reporting protocol for NIPT (including specific sample requirements) and the further details required as set out in in this Service Specification.

3.4.2 The NIPT Laboratory Provider will follow the flow chart as defined in Table One (area within red dotted line only) at paragraph 3.2.4 of this Service Specification and meet all the key indicators as specified in paragraph 8 (Data Reporting) of this Service Specification.

3.4.3 The NIPT Laboratory Provider will purchase sample kits and distribute these to the Ordering Entities (i.e the referring services); these sample kits will not be chargeable to the Ordering Entities.

3.4.4 All timings referred use Day 0 definition as the day the NIPT Laboratory Provider receives the sample as evidenced by the Provider logging the sample in its laboratory information management system upon receipt; Day

0 is NOT the day the NIPT Laboratory Provider receipts the sample if samples are not receipted immediately upon receiving.

3.4.5 The Ordering Entity will notify the NIPT Laboratory Provider that a sample is being sent; the NIPT Laboratory Provider should develop (during mobilisation) and provide (during the Contract term) a means of communication for this to occur and a record to be maintained.

3.4.6 If samples are delayed, exception reporting will be undertaken to determine the cause and an appropriate action plan will be agreed with the NIPT Laboratory Provider.

3.4.7 The NIPT Laboratory Provider must provide evidence of and report in the Monthly Performance Report on its compliance with such SOPs.

3.4.8 The SOPs for NIPT shall detail:

a. the provision of specialist cell stabilising blood tubes by the NIPT Laboratory Provider as applicable to Ordering Entities;
b. the provision of appropriate standardised request forms (paper and/or electronic) by the NIPT Laboratory Provider as applicable to Ordering Entities, which meet NHS FASP minimum data requirements;
c. the provision of transportation and storage of samples from Ordering Entities to the NIPT Laboratory Provider as applicable;
d. the competency/grade level of staff which will be required at each step of the NIPT process (which must meet requirements of UKAS);
e. processing times for each stage of the workflow which are consistent with the NIPT Turnaround Times (see paragraph 3.7.1 of this Service Specification);
f. all manual processing of NIPT within the NIPT pathway of the NIPT Laboratory Provider including Quality Control ("**QC**") laboratory protocol for each step of the NIPT process;
g. the processes designed should minimise cross contamination:
    i. processes to ensure that if there is a single test failure in a particular sample run that: only the sample which failed will need to be re-tested or a repeat sample obtained; and none of the other samples tested in that run are affected. These processes should include stop points and re-start points which allow for the repeat of certain steps in the process should a technical failure in the laboratory process occur.
    ii. the number of re-tests that can be run on a single patient sample (minimum of 10 ml sample) without requiring a new sample to be taken.
h. the policy for informing NHS England, Ordering Entities of any downtime in test availability (for example extended holiday periods, instrumentation failure or equipment maintenance). This policy should include a requirement to notify Ordering Entities in advance (where technically possible) of any of the foregoing and advise NHS England of any impact on NIPT Turnaround Times. The NIPT

Laboratory Provider will be required to have contingency plans in place to cover such eventualities.

i. procedures for monitoring, labelling and tracking samples from sample receipt to reporting results back to Ordering Entities, including details of expected timeframes which shall be in line with the NIPT Turnaround Times. The procedure should include a process for cross-referencing samples received against results reported.
j. all consumables required to run NIPT from receipt of patient sample to reporting the result to Ordering Entities and include information as to whether a "kit" is used or separate elements.
k. the quality assurance processes which build in robust QC fail safes into the laboratory pathway for NIPT and set out how samples that fail to meet the QC thresholds are identified;
l. all instruments and equipment used in the screening process;
m. the technology method used for NIPT.
n. the process to collect and follow up outcomes for all NIPT performed as part of the NHS FASP including to confirm screening results by either following up interpupillary distance ("**IPD**") results or assessment of baby at birth for confirmation (this will be collected in collaboration with NCARDRS);
o. the number of DNA samples it can process in a single batch and processes demonstrating that:
    i. DNA samples will be run regularly in batches of scalable numbers; and
    ii. the analysis protocol is able to adjust to scaling up in numbers in order to meet the NIPT Turnaround Times; and
p. a mechanism for the NIPT Service Provider to obtain ordering information and other data where NIPT Services have been subcontracted to an NIPT Laboratory Provider to provide NIPT so that the NIPT Service Provider and the NIPT Laboratory Provider (where applicable) can fulfil their reporting obligations in accordance with this Service Specification.

3.4.9 The NIPT Laboratory Provider shall:

a. receive and acknowledge receipt of the NIPT order in accordance with the SOPs;
b. only process orders which meet the NIPT Eligibility Criteria stated within clause 3.3.3 of this Service Specification;
c. check that all the NIPT Minimum Data has been supplied and submitted correctly alongside the complete sample;
d. check that the sample and accompanying patient referring card/specimen labels provided is complete; if the information is not complete, the NIPT Laboratory Provider must seek this correct information whilst storing and holding the sample(s) provided;
e. check that the choices made regarding conditions that the screening offer is accepted for are clearly documented on the request form. If

they are not clear or the relevant parts of the form have been left blank it is the responsibility of the laboratory to clarify the screening required with the requestor before the sample is analysed;

f. perform the relevant QC on the sample at all steps of the pipeline to determine if the sample is of sufficient quality to proceed to NIPT and at all steps of the procedure;
g. perform NIPT in accordance with the applicable standards for NIPT as set out in this Service Specification; and
h. return the complete results to Ordering Entities in accordance with the NIPT Turnaround Times (see paragraph 3.7.1 of this Service Specification),

3.5 Technical and Analytical platforms and Capability

3.5.1 The Business Continuity Plan and procedures which both the NIPT Service & Laboratory Provider is to have in place, maintain and comply with in accordance with Clause 5 of Schedule 2 (General Terms and Conditions) of the NHS England Contract must include a contingency procedure to ensure continuity of NIPT in the event of service failure.

3.5.2 The NIPT Service Provider should report to NHS England if it proposes to make any upgrades to current technology methodology in development of laboratory equipment, consumables and software for analysis or other similar items used in the performance of NIPT, what the upgrade is for, and an estimated launch date to NHS England.

3.6 Bioinformatics, Annotation and Validation

3.6.1 The NIPT Laboratory Provider shall set out in the SOPs required under paragraph 3.4.9:

a. full details of the process for validation/verification of NIPT results using the ISO 15189:2012 guidance (and working towards ISO 15189:2022). to ISO 15189:2012 (working towards ISO 15189:2022). This must include as a minimum the "ISO 15189:2012 Standard 5.5.1 Selection, Validation and Verification of Examination Procedures" document for NIPT;

b. the specific parameters used to generate final NIPT results. This must include parameters (for example, a prior risk) used to determine the lower and higher chance result;

c. the key quality parameters used in their analysis of NIPT results to pass or fail each sample (for example, minimum/maximum library yield QC, minimum samples for analysis, specific sequencing data quality control, minimum foetal fraction); and

d. the auditing processes and procedures on reporting of NIPT Turnaround Times (see paragraph 3.7.1 of this Service Specification).

3.7 Clinical interpretation, reporting, returning of results and measuring outcomes

3.7.1 The NIPT Laboratory Provider shall ensure that they perform the NIPT and return the result to the Ordering Entity (the maternity service that will provide the service user(s) with the test result(s)) within five (5) calendar days of receipt of the sample at the NIPT Laboratory Provider, as evidenced by the NIPT Laboratory Provider logging the sample in its laboratory information management system ("NIPT Turnaround Times").

3.7.2 If there is a sample testing failure, the NIPT Laboratory Provider shall, report this immediately to the Ordering Entity, advise if a second sample needs to be obtained from a patient, and advise of any impact of such failure on NIPT Turnaround Times.

3.7.3 The NIPT Service Provider must provide, as part of its regular report to NHS England via National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) such information as necessary to fulfil the NIPT Service Provider's obligations) and shall report:

i. examples of NIPT reports (include 'higher chance', 'lower chance' and 'no result');
ii. details of current NIPT 'no result' numbers and reasons for 'no result' and provide evidence for both first sample and re-sample requests;
iii. The NIPT Laboratory Provider will provide data to NCARDRS as specified in Table 2: Minimum demographic and result data to enable reporting on screen positives and screen negatives
iv. details on the NIPT Laboratory Provider's performance against the NIPT Turnaround Times and any failures to meet the NIPT Turnaround Times and the reason for such failure;
v. instances where samples require to be repeated, where additional samples need to be obtained due to test failure, and the impact on the overall NIPT Turnaround Times; any failures to meet the laboratories' QC thresholds.

3.7.4 The NIPT Laboratory Provider shall regularly communicate with NIPT Service Provider on evaluation, reporting and auditing activities performed as part of the NIPT Laboratory Provider's accreditation requirements audit programme to assess screening safety and performance as defined by NHS England requirements. This will include providing returns to **NCARDRS** and providing to NHS England and Ordering Entities:

i. annual data returns and reports on national standards;
ii. details of such returns to NCARDRS; and
iii. other requirements during the evaluative roll-out as defined by NHS England, all within the timescales specified by NHS England
iv. Data return requirements will be ongoing post evaluative roll out

3.7.5 The Maternity Unit is responsible for providing the necessary privacy notices to the patient. This statement overrides GC21.8. This is not the responsibility of the NIPT Laboratory Provider.

3.7.6

3.8 Population covered

3.8.1 The population covered will be eligible women registered for care with a NHS Maternity provider in England who have a higher chance result from an NHS combined or quadruple screening test. Eligibility will be checked and confirmed by the Ordering Entity. See Eligibility Criteria stated within paragraph 3.3.3 of this Service Specification. The NIPT Laboratory Provider will be required to check that the information supplied by the Ordering Entity with the pathology test request indicates that the sample meets the Eligibility Criteria prior to processing the sample.

3.9 Any acceptance and exclusion criteria and thresholds

3.9.1 As per paragraph 3.8 above.

3.10 Interdependence with other services/providers

3.10.1 The Services will be interdependent with the referring healthcare services, which may include, but not limited to, maternity services where the patient is registered with their GP or where they have chosen to receive treatment.

3.10.2 The interdependencies with NHS FASP and NHS England and are described throughout this document and will not be repeated here.

3.11 Activity & Tariff

3.11.1 Activity is anticipated to be between 10,000 to 12,000 tests per annum based on historical activity however this is not guaranteed. This activity will be split between providers.

3.11.2 Payment will be on a tariff basis. The tariff applicable to this service can be found in Schedule 3C of the Contract. This tariff will be reviewed annually.

4 Applicable Service Standards

4.1 Applicable national standards

4.1.1 The NIPT Laboratory Provider if a sub-contractor, must be ISO 15189:2012 accredited (and in transition to ISO 15189:2022) for NIPT, and must participate in an ISO 17043 accredited External Quality Assessment NIPT scheme.

4.2 Applicable standards set out in Guidance and/or issued by a competent body (e.g. Royal Colleges)

4.2.1 The NIPT Laboratory Provider will adhere to all relevant guidance that is issued during the lifetime of this contract.

4.3 Applicable local standards

4.3.1 The NIPT Laboratory Provider will adhere to all relevant guidance and local standards that are issued during the lifetime of this contract.

5 Applicable quality requirements

5.1 Applicable Quality Requirements

5.1.1 See paragraph 8 (Data Reporting) below.

5.2 Quality Assurance

5.2.1 The NIPT Laboratory Provider will provide evidence of its (and any NIPT Subcontractor's) UKAS accreditation to ISO 15189:2012 (working towards ISO 15189:2022) upon request to NHS England

5.2.2 The NIPT Laboratory Provider must provide auditable evidence to NHS England, when requested by either party, that their quality management system incorporates all the requirements of the screening pathway as set out in both Table One (at paragraph 3.2.4 above) and the NHS FASP Guidance, as may be updated from time to time. The NIPT Laboratory Provider shall manage incidents in accordance with the NHS Screening Safety Incidents Framework and the NHS FASP operational guidance.

5.2.3 The NIPT Laboratory Provider shall participate in the ISO 17043 accredited EQA NIPT scheme for aneuploidies. The NIPT Laboratory Provider shall, on request, share its data on EQA performance and the reports on the outcomes of the screening samples set to them via the NEQAS scheme, with NHS England. If the NIPT Service Provider has subcontracted NIPT to an NIPT Laboratory Provider, it shall obtain the performance data and reports relating to such NIPT Laboratory Provider and provide this to the aforementioned parties.

5.2.4 Both the NIPT Service Provider and the NIPT Laboratory Provider will be responsive to the findings of the evaluative roll-out as it proceeds, usually communicated by NHS England. Therefore, the NIPT Service Provider commits (and shall ensure the commitment of any NIPT Laboratory Provider) to working with NHS England to adapt and modify the laboratory pathways, processes and data requirements and SOPs created by the NIPT Laboratory Provider in accordance the evaluative roll-out, as it progresses in accordance with NHS England instructions. The NIPT Laboratory Provider shall also be required to perform data reporting to fulfil both the on-going quality assurance of laboratory services for NHS England in the long term and in the shorter term to

inform and support the evaluative roll-out of NIPT as part of the screening pathway. If either the NIPT Service Provider or the NIPT Laboratory Provider fails to develop or change the Services as required throughout this evaluative roll-out, upon instruction and with a 3-month notice period from NHS England (or 6 months where it would have a material effect upon staff), NHS England reserves the right to terminate the Contract due to failure to provide the contracted service.

6 Governance and organisational structure

6.1 The NIPT Laboratory Provider shall be fully compliant, with the NHS FASP specific requirements and standards. These requirements and standards will be more fully described in the NHS FASP operational guidance ("**NHS FASP Guidance**").

6.2 The NIPT Service Provider shall provide evidence to NHS England upon request, of its laboratory organisational structure for performing NIPT and how its provision of NIPT sits in the context of the overall Services provided by the NIPT Service Provider. On receipt of such evidence, NHS England shall assess (as per points listed below) whether to approve such organisational structure or require changes prior to engaging the NIPT Service Provider to perform the Services. In the event of any changes to the organisational structure of the NIPT Service Provider and/or the NIPT Laboratory Provider during the Term of the Contract, the NIPT Service Provider shall provide details of such changes and shall determine whether to approve the same or request further changes. This organisational structure shall include details of:

i. the named clinical lead for the NIPT Service Provider (and where NIPT has been subcontracted the clinical lead for the NIPT Laboratory Provider) as further detailed in paragraph 6.3, who shall form part of the Service Delivery Team; and
ii. the management and governance structure.

6.3 The named clinical lead referred to in paragraph i6.2i for NIPT screening for T21, T18 and T13 must be a suitably qualified clinician or clinical scientist who:

i. is either at the level of a laboratory director or is directly responsible there to;
ii. has management oversight and responsibilities; and
iii. is FRCPath qualified,

referred to as the "NIPT Screening Lead" ("**NIPT Screening Leads**").

6.4 The NIPT Service Provider shall participate and shall procure the participation of any NIPT Laboratory Provider, in cross-organisational and multi-disciplinary arrangements for the governance, management, communication and development of the screening pathway. This will include the sharing of data and information, as specified by NHS England

on its and any NIPT Laboratory Provider's performance and quality in respect of NIPT (which shall include the submission of Monthly Performance Reports), with NHS England including NCARDRS. If NIPT has been subcontracted in accordance with paragraph 6.5, the NIPT Service Provider shall require the NIPT Laboratory Provider to provide it with sufficiently detailed information on its performance and quality in order for the NIPT Service Provider to fulfil its obligations under this paragraph 6.4.

6.5 The NIPT Service Provider and/or the NIPT Laboratory Provider can only subcontract NIPT to a NIPT Laboratory Provider (providing NHS services) or other LGL or Designated Provider (providing NHS services), with the prior written approval from NHS England prior to any service commencement or entering into contract(s). Where an NIPT Laboratory Provider is used the NIPT Service Provider shall ensure that the relevant NIPT Laboratory Provider Subcontract contains:

6.5.1 details of service level agreements and risk assessed protocols between the NIPT Service Provider and such NIPT Laboratory Provider;

6.5.2 provisions that set out the responsibilities and working arrangements for screening samples sent by Ordering Entities to the NIPT Laboratory Provider;

6.5.3 arrangements and obligations on the NIPT Laboratory Provider in respect of:

- confirming sample receipt;
- meeting NIPT Turnaround Times;
- ensuring all screening results reach the responsible Ordering Entity and the instructing NIPT Laboratory Provider (as further detailed in the NHS FASP Guidance);
- reporting to the NIPT Service Provider all the information that the NIPT Service Provider is required to provide to NHS England in accordance with this Service Specification;
- performing the NIPT Gateway Services;
- complying with the SOPs created by the NIPT Laboratory Provider in accordance with paragraph 3.4.1 above and the NHS FASP Guidance;

6.5.4 performance monitoring and reporting mechanisms to enable the NIPT Service Provider to monitor the compliance of the NIPT Laboratory Provider; and

6.5.5 a requirement on the NIPT Laboratory Provider to be ISO 15189:2012 accredited (Transitioning to ISO 15189:2022) for NIPT and participate in ISO 17043 accredited EQA NIPT schemes and all such provisions and arrangements must align with the SOPs set by the NIPT Laboratory Provider in accordance with Paragraph 3.4.1.

6.6	The NIPT Service Provider must provide details to NHS England of their information governance processes (including those of all NIPT Laboratory Providers).
6.7	The NIPT Service Provider shall comply, and shall procure the compliance of all NIPT Laboratory Providers, with the screening policy and pathways of NHS FASP, as will be set out in the NHS FASP Guidance to support the evaluative roll-out of NIPT as an additional option within NHS FASP.

7	**Workforce Development and Personnel**
7.1.	The NIPT Laboratory Provider must ensure, and evidence on request, that all its staff participating in NIPT (including, all of the NIPT Laboratory Provider's staff who participate in NIPT) undertake Continuing Professional Development ("**CPD**") and revalidation, including the recommended eLearning modules relevant to laboratory staff for the NHS FASP programme. The NIPT Laboratory Provider must ensure that all staff participating in NIPT shall complete the NHS FASP Down's, Edwards' and Patau's Screening e-Learning Resource by the end of the Mobilisation Period. Such e-Learning Resource should be completed by each member of staff (within the NIPT Laboratory Provider) every 24 months for the duration of the contract. Any other specific courses which NHS England requires staff participating in NIPT to complete during the Term of the Contract shall be advised to the NIPT Service Provider from time to time. The NIPT Service Provider shall thereafter advise any NIPT Laboratory Providers of any such additional courses and ensure that the NIPT Laboratory Providers' staff participate in such courses.

8. Data reporting

Data requirements from genomic laboratories to support NIPT evaluation questions

The following information must be provided in line with the timescales stipulated and will be a requirement of the ongoing contractual agreement.

able 1: NIPT evaluative roll-out

Minimum demographic and result data

Reportable to: National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)

Period: samples meeting one of the following criteria within the month:
• Rejected due to ineligiblity for FASP pathway
• Rejected and repeat bloods requested from requesting hospital
• Tested and result reported to requesting hospital
• Tested with no result and either repeat bloods requested from requesting hospital or "no result after two samples" reported to requesting hospital

Submission via: https://nww.api.encore.nhs.uk/users/sign_in on the third week of the month following the reporting period. Any enquires to nhsdigital.ncardrs@nhs.net

Group	Item	Definition	Format	Code	Description	Mandatory/Required/Optional
INTERNAL IDENTIFIERS	**SAMPLE ID**	The unique sample identifier	free			Mandatory
INTERNAL IDENTIFIERS	**HUB ID**	The Genomic Laboratory Hub responsible for receiving and reporting the sample ODS code	an5			Mandatory
INTERNAL IDENTIFIERS	**LAB ID**	The laboratory responsible for receiving and reporting the sample ODS code	an5			Mandatory
INTERNAL IDENTIFIERS	**REQUESTING HOSPITAL ID**	The requesting hospital ODS code	an5			Required
MOTHER	**NHS NUMBER**	The NHS Number of the mother	n10			At least one mandatory combination: a) NHS NUMBER and BIRTH DATE b) FORENAME and SURNAME and BIRTH DATE and POSTCODE All required if available
MOTHER	**SURNAME**	The surname of the mother	free			
MOTHER	**FORENAME**	The forename of the mother	free			
MOTHER	**BIRTH DATE**	The date of birth of the mother	an10 CCYY-MM-DD			

MOTHER	**POSTCODE**	The postcode of the mother's place of residence	max an8			
MOTHER	**ETHNICITY**	Based on the Maternity Services Data Set the ethnicity of the mother as specified by herself	an1		**White**	Required
				A	British	
				B	Irish	
				C	Any other White background	
					Mixed	
				D	White and Black Caribbean	
				E	White and Black African	
				F	White and Asian	
				G	Any other mixed background	
					Asian or Asian British	
				H	Indian	
				J	Pakistani	
				K	Bangladeshi	
				L	Any other Asian background	
					Black or Black British	
				M	Caribbean	
				N	African	
				P	Any other Black background	
					Other Ethnic Groups	
				R	Chinese	
				S	Any other ethnic group	
				Z	Not stated	
				99	Not known	
MOTHER	**WEIGHT**	The weight of the mother at SAMPLE DATE in kilograms	decimal			Required
PREGNANCY	**EXPECTED DELIVERY DATE**	The expected delivery date for the pregnancy by ultrasound	an10 CCYY-MM-DD			Required
PREGNANCY	**FETUSES**		an1	1	Singleton	Required

		The number of fetuses for the pregnancy		2	Twins	
PREGNANCY	**CHORIONICITY**	Where FETUSES = 2, the chorionicity of the twins	an1	1	Dichorionic	Required
				2	Monochorionic	
				3	Other	
				4	Unknown	
PREGNANCY	**IVF**	Is this an IVF pregnancy	an1	1	IVF pregnancy	Required
				2	Not IVF pregnancy	
SAMPLE	**SAMPLE DATE**	The date of sample draw from the mother	an10 CCYY-MM-DD			Mandatory
SAMPLE	**RECEIVED DATE**	The date the sample was received into the laboratory	an10 CCYY-MM-DD			Mandatory
SAMPLE	**SAMPLE SEQUENCE**	Is this the first sample or a repeat sample	an1	1	First sample	Mandatory
				2	Repeat sample	
SAMPLE	**REPEAT REASON**	Where SAMPLE SEQUENCE = 2, the reason for the repeat sample	an1	1	Previous sample rejected	Required
				2	Previous sample did not produce a result after testing	
SAMPLE	**RUNS**	The number of runs completed on the sample	numeric			Required
SAMPLE	**SAMPLE ACCEPTANCE**	Was the sample accepted for testing	an1	1	Ineligible for FASP pathway	Mandatory
				2	Sample rejected prior to testing	
				3	Sample accepted on a discretionary basis after agreement with requesting hospital	
				4	Sample accepted after clarifying missing or discrepant information with requesting hospital	
				5	Sample accepted	
SAMPLE	**INELIGIBLE REASON**	Where SAMPLE ACCEPTANCE = 1 , the reason for ineligibility	an1	1	Maternal cancer (not in remission)	Required
				2	Maternal blood transfusion < 4 months	
				3	Maternal bone marrow or organ transplant surgery	

				4	Maternal immunotherapy (other than IVIg) in pregnancy	
				5	Maternal stem cell therapy	
				6	Vanished twin	
				7	Maternal Down's syndrome or balanced translocation or mosaicism of trisomy 21, trisomy 13, trisomy 18	
				8	Combined test or quadruple test chance threshold not met	
				9	Gestational age at first sample > 21+6 weeks	
				10	Triplet or higher order multiple pregnancy	
				11	Other	
SAMPLE	**REJECTION REASON**	Where SAMPLE ACCEPTANCE = 2 , the reason the sample was rejected	an1	1	Sample not received	Required
				2	Sample in wrong tube	
				3	Sample lacks identifiers or has incorrect identifiers eg mislabelling	
				4	Sample insufficient blood volume	
				5	Sample haemolysis observed	
				6	Sample received outside of lab-specific stability period	
				7	Incorrect blood collection tube used	
				8	Expired blood collection tube used	
				9	Broken/leaking tube received	
				10	High risk infectious disease	
				11	Other	
				12	Sample too long in transit/delayed arrival	
TEST	**CONDITION TESTED FOR**	The condition for which testing is requested	an1	1	T21	Required
				2	T13/T18	
				3	T21/T13/T18	
TEST	**TEST OUTCOME**		an1	1	Not tested	Mandatory

		The outcome of testing for the sample		2	Result	
				3	No result	
RESULT	**AUTHORISED DATE**	The date of test result authorisation by the laboratory	an10 CCYY-MM-DD			Required
RESULT	**REPORTED DATE**	The date the test result was reported to the maternity service (including the reporting of ineligibility, rejection, or "no result")	an10 CCYY-MM-DD			Mandatory
RESULT	**NO RESULT REASON**	Where TEST OUTCOME = 3, the reason for no result	an1	1	DNA extraction	Required
				2	Library prep (sequencing) / pre hybridisation pre (microarray)	
				3	Sequencing / hybridisation	
				4	Low fetal fraction	
				5	High fetal fraction	
				6	Contamination / non declared IVF pregnancy	
				7	Equipment failure	
				8	Technical failure	
				9	Failure to select the correct testing algorithm for the pregnancy	
				10	Other	
				11	Signal:noise failure	
				12	iFACT error	
RESULT	**RESULT REPORTED - T21**	Where TEST OUTCOME = 2, the T21 result of the test for the sample	an1	1	Higher chance	Required
				2	Lower chance	
				3	No result	
RESULT	**RESULT REPORTED - T18**	Where TEST OUTCOME = 2 , the T18 result of the test for the sample	an1	1	Higher chance	
				2	Lower chance	
				3	No result	

RESULT	**RESULT REPORTED - T13**	Where TEST OUTCOME = 2 , the T13 result of the test for the sample	an1	1 2 3	Higher chance Lower chance No result	
RESULT	**Z SCORE-T21**	The T21 Z-score for the sample, where available	decimal			Required
RESULT	**Z SCORE-T18**	The T18 Z-score for the sample, where available	decimal			Required
RESULT	**Z SCORE-T13**	The T13 Z-score for the sample, where available	decimal			Required
RESULT	**LR -T21**	The T21 likellihood ratio for the sample, where available	decimal			Required
RESULT	**LR -T18**	The T18 likellihood ratio for the sample, where available	decimal			Required
RESULT	**LR -T13**	The T13 likellihood ratio for the sample, where available	decimal			Required
RESULT	**POST - T21**	The T21 posterior risk/odds ratio for the pregnancy, where available	decimal			Required
RESULT	**POST - T18**	The T18 posterior risk/odds ratio for the pregnancy, where available	decimal			Required
RESULT	**POST - T13**	The T13 posterior risk/odds ratio for the pregnancy, where available	decimal			Required
RESULT	**FETAL FRACTION**	The estimated fetal fraction for the sample, where available	decimal			Required

8. Data reporting continued

In addition to this data collected in relation to NIPT samples analysed in line with the NHS England Genomics Unit Test Directory code R445 (NIPT for previous trisomies) will be separated and also submitted.

The following experimental FASP metrics must be met on an ongoing basis by all NIPT Laboratory Providers and will be a contractual requirement. NIPT Laboratory Providers must report on their performance against these metrics in the manner stipulated to comply with Contract terms and conditions.

If a provider does not meet the 'Acceptable level' stated in each of the metrics below NHS England will work with the NIPT Laboratory Provider to agree an action plan and timescales with the provider in which they must adhere to rectify performance levels.

FASP NIPT-S02: test: timely receipt of NIPT sample

Description

The proportion of all NIPT samples received in the genomic laboratory ≤ 2 working days.

Rationale

To enable timely reporting of screening results to women so they can make personal informed choices. Delays in sample receipt increases the chances of deterioration and the need for a repeat sample.

Definition

Numerator: number of NIPT samples received by the laboratory ≤ 2 working days of sample collection/draw

Denominator: number of NIPT samples received by the laboratory in the reporting period

Sample received is when the sample is recorded as received on the laboratory information management system

For the purposes of this standard, day of sampling is day 0

We calculate performance by dividing numerator by denominator and multiplying by 100 to give a percentage.

Performance thresholds

Acceptable level: :≥90.0%Achievable level: ≥95.0%

Caveats

None

Data collection and reporting

Data source: genomic laboratories

Responsible for data quality and completeness: genomic laboratories

Responsible for submission: NCARDRS

Reported by: maternity service

Published by: maternity service

Reporting period

Monthly

FASP NIPT-S03: test: turnaround time NIPT

Description

The proportion of NIPT screening test results reported ≤ 5 calendar days of sample receipt (evidenced by the sample being receipted into the NIPT Laboratory Provider's laboratory information management system).

Rationale

To enable timely reporting of screening results to women so they can make personal informed choices.

Definition

Numerator: number of NIPT screening results reported by the genomic laboratory to maternity service ≤ 5 calendar days of sample receipt

Denominator: number of NIPT screening samples received in the genomic laboratory in the reporting period excluding samples received:

- that are not fit for analysis and a repeat sample is requested
- with missing information required for calculating the result

The denominator and numerator include samples that are analysed where the result is 'no result'

Date of sample receipt in the laboratory is counted as day 0.

We calculate performance by dividing numerator by denominator and multiplying by 100 to give a percentage.

Performance thresholds

Acceptable level: ≥85.0%

Achievable level: ≥95.0%

Caveats

None

Data collection and reporting

Data source: genomic laboratory

Responsible for data quality and completeness: genomic laboratory

Responsible for submission: NCARDRS

Reported by: genomic laboratory

Published by: genomic laboratory

Reporting period

Monthly

FASP NIPT-S06: diagnosis and intervention: test turnaround time quantitative fluorescence-polymerase chain reaction (QF-PCR)

Description

The proportion of QF-PCR test results reported in ≤ 3 calendar days of sample receipt.

Rationale

To enable timely reporting of QF-PCR test results to women so they can make personal informed choices.

Definition

Numerator: number of QF-PCR test results reported in ≤ 3 calendar days of sample receipt.

Denominator: number of prenatal diagnostic samples received in the genomic laboratory in the reporting period where the indication for QF-PCR testing is a:

- higher chance NIPT screening result
- ‘no result’ NIPT screening result

Date of sample receipt in the genomic laboratory is counted as day zero.

We calculate performance by dividing numerator by denominator and multiplying by 100 to give a percentage.

Performance thresholds

Acceptable level: ≥ 90.0%

Achievable level: ≥ 95.0%

Caveats

None

Data collection and reporting

Data source: genomic laboratories

Responsible for data quality and completeness: genomic laboratories

Responsible for submission: NCARDRS

Reported by: genomic laboratories

Published by: genomic laboratories

Reporting period

Quarterly: data to be collated between 2 and 3 months after each quarter end

Deadlines: 30 September (Q1), 31 December (Q2), 31 March (Q3), 30 June (Q4)

Annex 2 – Cellular Pathology Genomic Centres technical requirements

Service aims

123. The aim of the service to be provided by the CPGCs will be to optimise and maximise tissue-based predictive immunohistochemistry assessment, sample preparation for genomic testing and to perform a small, prescribed list of cancer genomic tests that require rapid turnaround times and that have been agreed with the Commissioner.

124. The CPGCs will be commissioned, by an NHS GMS Lead Provider to provide the following services on its behalf:

- Efficient tissue handling to provide diagnostic and predictive immunohistochemical information and maximise tissue for genomic analysis. Tissue will be handled to minimise degradation of nucleic acids (i.e. limited time in formalin) and sectioning protocols applied to reduce tissue waste.
- Tissue pathways will be established and/or optimised from other laboratories and hospitals to expedite the transport of the tumour samples into the Centre.
- Tumour diagnosis, identification of a representative tumour block in every cancer report with statement of percentage neoplastic nuclei content, and adequacy assessment for subsequent genomic analysis when appropriate.
- Provision of tumour tissue including FFPE and fresh (frozen or in an approved tissue stabiliser) for all eligible patients for onward referral to NHS Genomics Laboratory for cancer for genomic testing as stipulated by the Test Directory
- Provision of a small number of prescribed genomic tests where fast turnaround time is required under the direction and quality oversight of the NHS GLH inclusive of accreditation requirements and participation in EQA schemes, with the oversight of Local Reporting and Coordinating Laboratory and by prior agreement with the Commissioner .
- Digital macroscopic / microscopic images as required and according to national specifications being established through for example National Pathology Imaging Cooperative (NPIC) and set out in guidance from the Commissioner.

125. The CPGCs will work closely with and be part of the NHS Genomic Testing Laboratory function in the NHS GMS to achieve the following overall objectives of the service:

- To provide an exemplary and comprehensive cellular pathology service for all patients with cancer, working with all providers within their pathology network;

- Deliver a clinically relevant end to end cancer diagnostics turnaround time;
- To facilitate specialist predictive immunohistochemistry (where appropriate)
- To perform tissue sampling, handling, assessment and processing pathways between genomic testing and cellular pathology laboratories;
- To ensure that all aspects of the services are delivered as safely and effectively as possible, conforming to accreditation requirements, national standards and published clinical guidelines;
- To commit to continual service review and improvement;
- To contribute to increasing genomic testing activity in line with the agreement of the NHS GMS and the Commissioner and and return of results within clinically relevant timelines and to defined quality requirements; and
- To enable all referred patients to receive equitable access to an accurate diagnosis and maximise therapeutic options.

Service model

126. The NHS GMS is a multi-professional, multidisciplinary service which delivers integrated pathways focused on patient need, whilst addressing quality, governance and supporting optimal outcomes for patients.

127. Sample acquisition, handling and assessment is provided by a range of professions including clinical and biomedical scientists, genomic technologists, specialists in Genomic Medicine and genomic co-ordinators, in addition to core medical and nursing staff.

128. Each of the professions work together and contribute to the specific role and responsibilities of the NHS Genomic Testing Laboratory function commissioned by the Commissioner to deliver the genomic testing for the NHS GMS as outlined in the Test Directory.

129. Critical to the delivery of the solid tumour genomic testing listed within the Test Directory is the rapid process of assessing, preparing and providing suitable tissue samples alongside immunohistochemistry assessment. Each NHS GMS will receive additional funding from the Commissioner to commission CPGCs in the NHS GMS geography in line with this specification.

130. There is an expectation that the CPGCs will work across the NHS GMS geography and pathology networks so that all providers will be linked to a CPGC within a defined pathology network.

131. The CPGC will be funded to deliver cellular pathology activity for tissue sample preparation for onward referral to NHS GLHs for genomic testing as stipulated by the Test Directory.

132. The CPGC will be funded for formalin fixed paraffin embedded (FFPE) and fresh (frozen or in approved tissue stabiliser) tissue sample preparation for onward referral to NHS Genomics Laboratory for cancer for genomic testing as stipulated by the Test Directory. This will not disrupt SIHMDS pathways

133. This service relates currently to sample preparation of cancer samples only. Throughout the contract duration, the Commissioner may expand the requirement to include tissue for rare disease testing.

134. The CPGC can be funded to perform a prescribed list of genomic tests for specific pathways, subject to the agreement of the NHS GMS Lead Provider to support rapid turnaround times and increased capacity where necessary and in agreement with the Commissioner. The CPGC must have demonstrated the required genomic expertise and competency and the genomic testing must be delivered with the quality management and oversight of the NHS Genomics Laboratory for cancer inclusive of mandatory participation in relevant EQA schemes. The delivery of the genomic test must be cost effective and be in line with the national tariff prices.

135. There is a requirement for each CPGC and NHS Genomics Laboratory for cancer to form a ‘Quality Review Panel’ to define local of local KPIs and quality metrics in line with the Commissioner’s national specification. This should be built into the governance structure.

136. The CPGCs must:
    - be UKAS accredited to ISO 15189;2022 for all activities within the service specification;
    - submit all samples eligible for genomic testing stipulated by the Test Directory to the NHS Genomics Laboratory for cancer unless an alternative approved pathway is in

place e.g., testing performed by an agreed salvage pathway or as part of the prescribed lists of genomic tests to be performed by the CPGC;

- work with cellular pathology and NHS Genomic Laboratory Service teams to deliver all results within a clinically relevant end to end cancer diagnostic turnaround time;
- support the corresponding NHS GMS and pathology network to establish tissue referral pathways from all providers;
- have dedicated consultant histopathologist leadership with demonstrable genomic testing expertise within each CPGC;
- have trained and competent scientific staff with expertise in immunohistochemistry and tissue applications of molecular techniques in place to deliver the optimal sample handling and processing requirements;
- have appropriately trained and competent staff in place to deliver cellularity and tumour assessment for genomic samples, including participation in GenQA Tissue-i tumour assessment external quality assessment modules relevant to their role;
- have dedicated staff in place to identify samples which should be referred for genomic testing stipulated by the Test Directory and enable subsequent forwarding of samples to the NHS Genomics Laboratory for cancer for testing in a timely manner to facilitate clinical care;
- have a mechanism in place for identification and delivery of suitable tissue (scrolls or slide-mounted tissue sections) and other sample types e.g. fresh tissue (frozen or in an approved tissue stabilizer) and cytology samples for genomic testing to the relevant NHS Genomics Laboratory for cancer or for agreed clinical trials
- actively participate in identification and optimised tissue handling and onward submission to the NHS Genomics Laboratory for cancer for patients identified as potentially eligible for cancer vaccine trials according to the national protocol;
- commit to increasing/ implementing use of automated equipment in the cellular pathology laboratory in order to increase processing efficiency and free up staff time for other skilled roles i.e. handling of tissue for genomic testing;
- maximise education, training and staff development opportunities to optimise upskilling of staff, recruitment and retention.
- commit to supporting and delivering sample preparation and data collection for the Cancer Vaccine Launch Pad (CVLP) as required.
- commit to submit the required minimum datasets to GLH.
- participate in regular NHS Genomics Laboratory for cancer /CPGC joint Quality reviews, data collections, audits, activity supporting corrective and preventative measures and ongoing quality improvements.

Pathways

Overall patient pathway

137. Patient samples for genomic testing will either be derived from the existing CPGC internal workload or be referred to the regional designated CPGC/s by cellular pathology laboratories in other trusts in order to access specialist predictive immunohistochemistry and/or GLH genomic testing according to the Test Directory.

People

138. The CPGCs will be staffed by the following essential staff groups for provision of the services:
    - A lead histopathologist with support from fellow consultant histopathologists within their team to implement and provide operational oversight
    - Histopathologists, and appropriately trained laboratory scientists (including Clinical Scientists and Biomedical Scientists) to provide training and reporting of relevant biomarkers including a limited repertoire of genomic tests where agreed.
    - Biomedical scientists to carry out and optimise end to end laboratory tissue handling processes, including validation and introduction of new equipment and techniques
    - Biomedical support workers to provide input to tissue handling processes
    - Administrators to receive and deal with referred samples, requests and enquiries about genomic testing, to track and dispatch samples to GLHs and to submit information and samples to clinical trials where required.

139. All staff must be competent to deliver the services required and be suitably qualified and registered with relevant accredited professional bodies and in line with regulatory requirements. Governance will be led by the Commissioner with the NHS GMS Lead Provider in line with the Contract Management Framework.

Essential equipment and / or technology

140. All CPGCs must have sufficient IT facilities to capture and record all referrals. This will be essential to capture for the purpose of commissioning, and for the purpose of measuring reporting outcomes. CPGCs will also require digital pathology infrastructure in terms of laboratory IT system for recording specimen information and imaging/ slide scanning, storage and retrieval facilities for macroscopic and microscopic images.

Annex 3 – Whole genome sequencing technical requirements

141. Sample handling and DNA extraction requirements remain unchanged between the current centralised sequencing model and new distributed model. The delivery model will change and be outlined in updated guidance. Detailed information can be found in the following guidance documents:
    - Sample Handling Guidance for Whole Genome Sequencing of Germline Samples v1.0
    - Sample Handling Guidance for Whole Genome Sequencing of HaemOnc
    - Sample Handling Guidance for Whole Genome Sequencing of Solid Tumour Samples v1.0

142. The volume and concentration of DNA samples required is described in the following guidance document however this is likely to be amended for the distributed WGS model so that less DNA is required. Relevant guidance documents will be issued or amended to reflect any change.
    - DNA Extraction and Quality Control Guidance for WGS v2.0

143. Test request approval and required data entry into the National Genomic Informatics System (NGIS) also remain unchanged between the two models and is described in the following document, with the exception of Rapid HaemOnc which is described in more detail in the centralised WGS model section.
    - Genomic Laboratory Hub Approval for Whole Genome Sequencing Referrals Pathway

Centralised WGS model

144. Process map 1 outlined below describes the centralised WGS model.

145. Guidance on the process and requirements are provided in the following documents;
    - Packaging and Transportation of Samples for Whole Genome Sequencing in the NHS Genomic Medicine Service v1.0
    - Operational guidance for the transportation of samples for WGS in the NHS GMS
    - Guidance Document for the Plating of DNA Samples for Whole Genome Sequencing (WGS) by the Single Plating Genomic Laboratory Hub (GLH) and submission to the WGS Provider v1.0 - NHS Genomic Medicine Service - Futures

Distributed WGS model

146. Process map 2 describes the indicative activity and data flows in the distributed WGS model for DNA samples sent to the designated NHS GMS WGS laboratories.

147. Process map 3 describes the indicative activity and data flows in the distributed WGS model for fresh samples sent direct from requester (SIHMDS) to the designated NHS GMS WGS laboratories for Rapid HaemOnc.

Sequencing NHS Genomic Laboratory Specification

148. This section outlines the service requirements that a designated WGS NHS GMS Sequencing Laboratory must meet to provide an NHS GMS commissioned WGS service. This specification is designed to minimise variability and promote consistency and standardisation of sequencing data.

149. Note - At this stage, the proposed requirements are based on the current understanding of the future operating model, which remains subject to refinement through pathfinder work underway before the commencement of a live service further guidance will be issued by the Commissioner.

Requirement
NovaSeq X Plus installed, validated, and its use in UKAS ISO15189:2022 scope. If currently not in scope the laboratory must have either submitted an extension to scope or can demonstrate a clear plan to obtain accreditation within 12 months.
Illumina PCR-Free WGS library preparation to be used by standard protocol specified with the assay. This process is to be validated and in UKAS ISO15189:2022 scope. If currently not in scope the laboratory must have either submitted an extension to scope or can demonstrate a clear plan to obtain accreditation within 12 months.
TPS71 is a UKAS (ISO15189:2022) requirement when multiple entities are responsible for different parts of a diagnostic testing pathway. Since distributed WGS represents an end-to-end process, accreditation requires that each part of the process is individually accredited.

TPS71 documentation should be in place as part of work to gain UKAS 15189:2022 accreditation and should support change management of any components or dependencies of the testing and analysis pathways.
The 25B flow cells will need to be used by all rare disease WGS Nationally Designated WGS Providers to maximize cost effectiveness, enable standardised data and support robust copy number variant (CNV) calling across the whole NHS GMS. Non-Rare Disease samples (including germline) will need to be run on 10B flow cells until 25B flow cell are considered operationally and economically viable.
Standardised indexing using Illumina DNA/RNA Unique Dual Indexes (UDI) (96 Indexes, 96 Samples) to support business continuity.
Illumina Dragen v4.0.5b, installed, validated and in UKAS ISO15189:2022 scope. If currently not in ISO15189:2022 scope the laboratory must have either submitted an extension to scope or can demonstrate a clear plan to obtain accreditation within 12 months. The accreditation must be paired between Genomics England and the NHS GMS Sequencing Lab and change managed through TPS71 framework. Note, it is recommended to use the server or cloud version, as the on-board version prevents full utilisation of the sequencers. Data transfer to Genomics England must be automated and streamlined to avoid delays. Illumina Dragen v4.0.5b will be used for demultiplexing by the NHS GMS sequencing laboratory. The resulting FASTQ files, gz/ora compressed, will then be sent to Genomics England. To maintain uniformity across outputs, it is important that all Sequencing providers apply the same parameters and follow a consistent process. The parameters for the demultiplexing command are still to be defined.
The NHS GMS must establish specific processes for the change management of bioinformatics and scientific systems and standards, through a national Genomics Change Advisory Board (CAB), which has yet to be established, especially where pathways are delivered across multiple diagnostic providers. These processes must meet the requirements of UKAS TPS71.
An appropriate Business Continuity and Disaster Recovery (BCDR) plan must be in place and maintained, taking into account volume and turnaround time requirements.

Sign off of National Information Governance (IG) contract to ensure that anyone transferring patient data to another organisation has the appropriate IG documentation in place. The national dWGS DPIA has been updated and any Sequencing NHS GMS Laboratory operating under it must also update their IG documentation accordingly.
Demonstration of capability to run at required scale. The required scale per designated NHS GMS sequencing provider is yet to be confirmed, as it depends on the number of providers.
10Gb upload capability from NovaSeq X Plus to GEL as well as: • Intranet from NovaSeq X Plus to enable local data processing • Internet from local data processing source to GEL Note - A guaranteed 10Gb intranet connection is required for NovaSeq X Plus installation. The 10Gb upload capability must be end-to-end and with guaranteed dedicated bandwidth (either via physical lines or virtually by implementation of virtual networks, Trust-level firewall configuration etc).
A sufficient network infrastructure is in place to support BCDR and comply with TPS71 requirements.
Sequencing NHS GMS Laboratories must have created a Developer Account in the NHS API Platform (also known as the NHS API Management Gateway) and completed the onboarding. The proposed mechanism by which NHS GMS Sequencing Laboratories will transfer the FASTQ files to GEL will involve using NHS API Management.
GEL API (to transfer sequencing data to GEL) standards (yet to be defined) must be adopted. Python packages will be provided to support data transfer from the NHS GMS provider to GEL and demonstrate that the data meets the formatting standards. Note, development and testing are still ongoing to define this process and where such packages will be published.
Sequencing NHS GMS Laboratories must have a unique 6 digit lab ODS code.
The Commissioner in partnership with Genomics England will define the required Quality Control standards for sequencing data to be passed to GEL for processing and for data to be passed from GEL back to the NHS GMS for analysis. Work is still on going to determine

appropriate metrics and thresholds, given that the previous NovaSeq 6000 thresholds may be different to those required for NovaSeq X Plus. Tools will be identified to facilitate the calculation of pre-alignment metrics. The target coverage for germline WGS is 30X and for tumour WGS is 60X.
Once Genomics England confirm that FASTQ data pass QC, NHS GMS sequencing laboratories will delete local copies. This process will be superseded when the laboratory common cloud infrastructure has been developed in line with the UGR. From that point the NHS GMS will store and have access to their own data, in a multi-tenancy single cloud environment.
Standardised quality control definitions, methodologies and metrics are being developed by Global Alliance for Genomics and Health (GA4GH). The NHS GMS will contribute and align to these standards as they are developed and implemented.
NHS GMS Sequencing Laboratories must be able to provide meta data to GEL: See "**Meta Data**" table below.

Meta Data

150. The required metadata outlined represents the minimum data elements that NHS GMS Sequencing Laboratories are expected to send with the FASTQ files. This metadata is necessary to ensure that genomic data can be accurately linked to the clinical data entered into TOMS by the requesting NHS GMS laboratory, or in the case of Rapid HaemOnc testing by the sequencing centre themselves.

151. The validity of the 'Required Meta Data' will be tested and confirmed through the pathfinder work that is underway. Additional data elements may be required and the following list may be subject to change prior to April 2026.

Required Meta Data	Example	Comments
NGIS Referral ID	"r30000000001"	Generated by requesting NHS GMS laboratory prior to sending DNA to NHS GMS Sequencing laboratory.

		For HaemOnc the NGIS Referral ID will be generated by the NHS GMS Sequencing laboratory
Participant ID	"p03751485488"	Populated by requesting NHS GMS laboratory prior to sending DNA to NHS GMS Sequencing laboratory. For HaemOnc the Participant ID will be generated from the NHS GMS Sequencing laboratory
SIHMDS ID	H123/25	For HaemOnc the SIHMDS ID will be generated by the SIHMDS.
Requestor or Sample ID	M1234	Local ID from the clinical Test Ordering Entity
Lab Sample ID (Fluid-X tube number)	"1700009505"	Generated by the requesting NHS GMS laboratory and corresponds to the Fluid-X tube ID the DNA sample sent to the NHS GMS sequencing provider is in. For HaemOnc the Lab Sample Number will be generated by the NHS GMS Sequencing Lab. Fluid-X tubes are not appropriate for HaemOnc fresh samples.

152. For acute leukaemia the result for the three day turn around FISH test as specified in Test Directory data will be submitted (in order to inform ploidy in the bioinformatics pipeline).

Requesting NHS GMS laboratory

153. This section outlines the pre-sequencing activities that must be undertaken by the requesting NHS GMS laboratory prior to sample transfer to the designated NHS GMS Sequencing Laboratory.

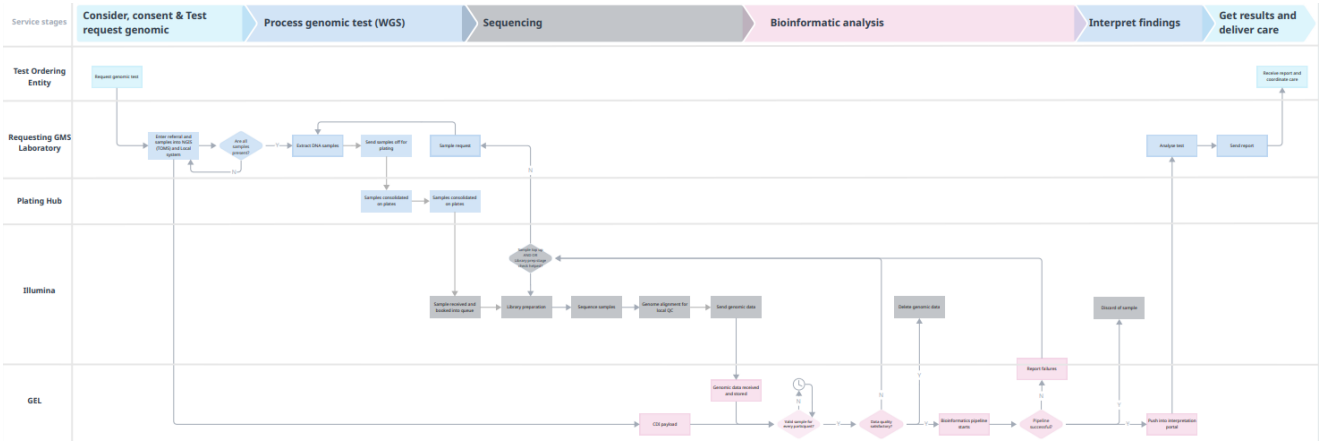
154. For rapid WGS in HaemOnc, the NHS GMS Sequencing Laboratory will perform both DNA extraction and sequencing in order to expedite turnaround times. The Requesting NHS GMS Laboratory should send the sample directly to the NHS GMS Sequencing Laboratory along with the completed test order form and record of discussion form. There is no need for the Requesting NHS GMS Laboratory to perform DNA extraction or enter data into TOMS. The Requesting NHS GMS laboratory in each NHS GMS geography is responsible for identifying and optimising the most efficient pathways to facilitate the rapid turnaround time for HaemOnc. Please refer to Process Map 3 for potential routes. The table below describes the sample requirement. In advance of the introduction of the Order Management (OM), the Requesting NHS GMS Lab are responsible for designing and implementing a process to track the submission of HaemOnc referral (whether direct from Requesting NHS GMS Lab or SIHMDS).

Measurement Name	**Measurement**	**Notes**
Sample volume	Minimum 1ml of EDTA peripheral blood/ bone marrow	Any surplus fresh sample (bone marrow or peripheral blood) sent will not be stored beyond confirmation of successful sequencing. Extracted DNA will be stored in line with standard NHS storage recommendations and in line with biobanking requirements to be set out by the Commissioner.
White cell count	Minimum requirement 2.0 x 10^9/L	If peripheral blood white count is below the minimal requirement, then bone marrow should be sent. Any sample sent should meet the minimum blast/ malignant cell content as specified above.

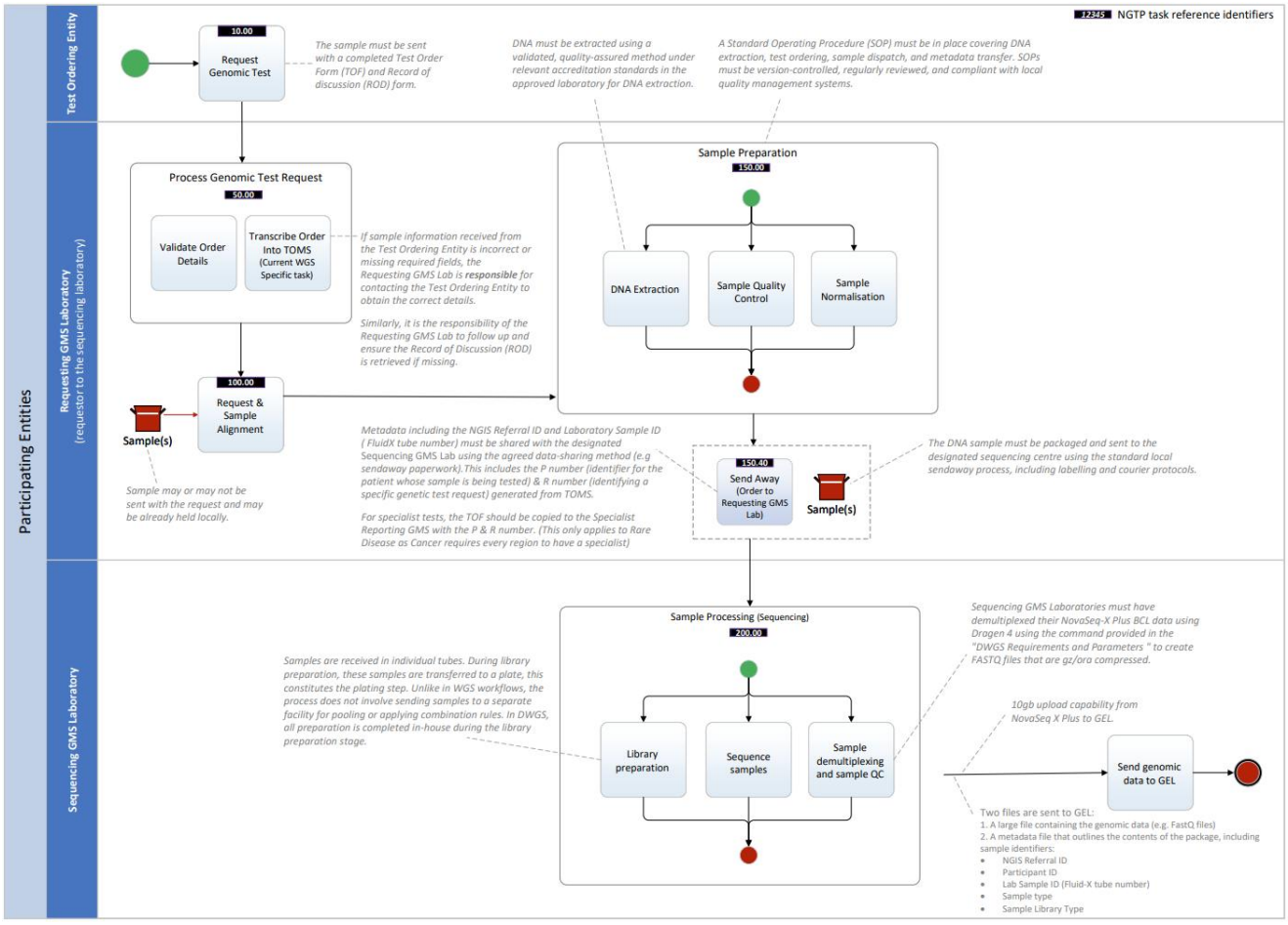
Requirement

The test order must be accurately transcribed and entered into the Test Ordering and Management System (TOMS). For HaemOnc WGS, TOMS requests will be completed by the designated NHS GMS sequencing laboratory.
The Requesting NHS GMS Laboratory will arrange the transportation of DNA samples in FluidX tubes [68-0701-12 FluidX 96-Format, 0.5ml External Thread, Next-Gen Dual-Coded Tube], to their designated NHS GMS Sequencing Laboratory There is no requirement to send samples on specific days, in batches, or to group family members within the same consignment. HaemOnc samples and requests can be sent directly from any requesting laboratory or SIHMDS providing the record of that test request is approved by and follows the process defined by the requesting NHS GMS laboratory for that geography.
The DNA sample or fresh sample (HaemOnc only) must be packaged and sent to the designated NHS GMS Sequencing Laboratory using the standard local sendaway process, including labelling and courier protocols.
The following metadata should accompany the DNA sample (e.g. on a sendaway letter) to enable linking the physical DNA sample to its digital record in TOMS: • NGIS Referral ID • FluidX tube ID • Participant ID *Note this may be subject to change based on the outcomes of the Pathfinder work, a move to electronic messaging and any Order Management developments in the future.* HaemOnc samples sent directly to the NHS GMS Sequencing Laboratory must be accompanied by a competed test order form and record of discussion.
A Standard Operating Procedure (SOP) must be in place covering DNA extraction, test ordering, sample dispatch, and metadata transfer. SOPs must be version-controlled, regularly reviewed, and compliant with local quality management systems.

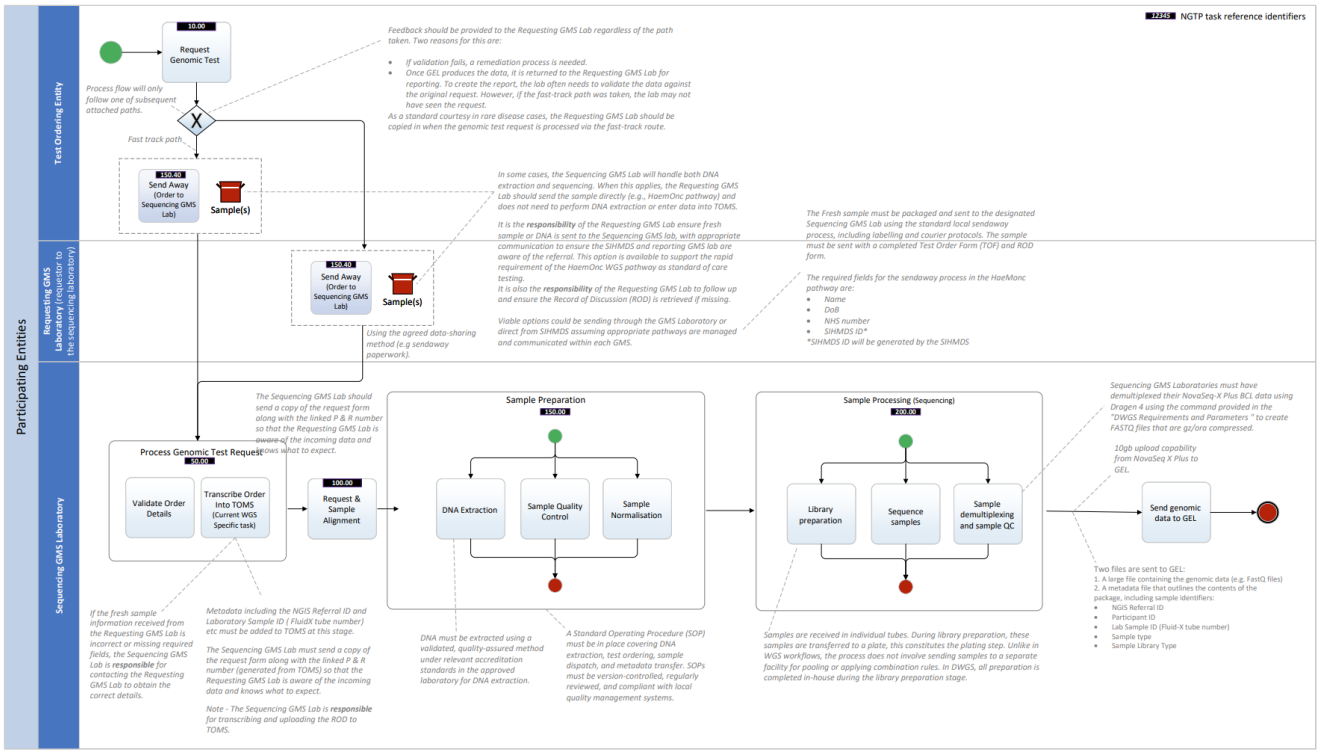
Process Map 1: centralised WGS model



Process Map 2: Distributed WGS (may be subject to amendment)



Process Map 3: Distributed WGS (Rapid HeamOnc) – may be subject to amendment



Annex 4 – People roles

Mandated job roles – Executive leadership

The Commissioner would like to oversee the appointment of NHS GMS Chief Operating Officer and Clinical Director.

The job descriptions below outline the generic duties, responsibilities and person specification for the roles, however they should be tailored to the values and responsibilities of the hiring organisation.

NHS GMS Clinical Director

Position			
Job title	NHS GMS Clinical Director	**Directorate/region**	NHS GMS to add
Pay band	Consultant /Medical	**Responsible to**	NHS GMS add
Salary	NHS GMS to add	**Accountable to**	NHS GMS to add
Tenure	Permanent	**Responsible for**	NHS GMS to add
WTE	1	**Base**	NHS GMS to add

Key experience
A successful candidate should have: • A recognised national/international clinical profile in leading genomics and in contributing to national and international initiatives in Genomic Medicine • Considerable experience of building and developing strong and effective teams and providing robust and visible clinical leadership in a range of public facing and challenging environments across a large geography • A track record of strategic, board level leadership in a large complex public sector, private or third sector organisation • Sound knowledge of clinical governance and effective systems of clinical management such as job planning, appraisal and clinical assessment across the multi-disciplinary, multiprofessional and multi-speciality geography

Key responsibilities
The post holder will have responsibility for managing the multidisciplinary clinical workforce within the NHS GMS geography, to help deliver organisational priorities and compliance. Key responsibilities of the NHS GMS Clinical director include:

- Providing strategic clinical advice leadership and expertise in the design and delivery of the NHS GMS, ensuring services are safe, effective, and aligned with best practices. This includes overseeing workstreams, ensuring visibility of progress and risk among senior leaders.
- Providing strategic direction to the development and improvement of Genomic Medicine pathways, including the mainstreaming of genomic testing across different clinical specialties
- Ensuring equity of access to genomic testing for patients to reduce health inequalities, and overseeing work with local teams and stakeholders to improve this
- Building strong collaborations across the NHS GMS geography, enabling the multi-professional workforce to deliver the ambitions of the NHS GMS and 10 Year Health Plan commitments within genomics
- Leading a multi-specialty and multi-professional team covering the widespread and evolving application of genomics within the NHS GMS geography
- Responsibility for the delivery of clinical governance across the NHS GMS
- Working with the leadership in the cross-cutting functions including People to oversee training and education for the developing clinical genomics workforce and Research and Innovation to ensuring the NHS GMS is at the forefront of innovation and working to translate this into business as usual

Person specification				
Criteria		**Essential**	**Desirable**	**Evidence***
Qualifications	MBBS or equivalent, MRCP or equivalent	✓		**A/I**
	A medical consultant with proven experience of leading, developing and managing a large-scale genomic service	✓		**A/I**
	Able to demonstrate commitment to continuous professional development	✓		**A/I**
Knowledge and experience	Senior clinician or scientist, with a broad understanding of genomics with national leadership experience and credibility	✓		**A/I**
	National leadership experience with international recognition within their own specialist area of genomics	✓		**A/I**
	Experience of developing, applying and reviewing an evidence-based approach to decision making in genomic services	✓		**A/I**

	Experience of chairing multidisciplinary groups	✓		**A/I**
	Knowledge of the genomics clinical and scientific landscape in the NHS and plans for the continued development of the NHS Genomic Medicine Service	✓		**A/I**
	A good understanding of the English healthcare system, and some understanding of international systems	✓		**A/I**
Skills capabilities and attributes	A leading medical consultant with an understanding of advising on the clinical actionability of complex genomic data up to the level of whole genome sequencing	✓		**A/I**
	Previous senior leadership experience in a clinical genomic service or a major academic genomic research group	✓		**A/I**
	The ability to work collaboratively and to deal with ambiguity and complexity	✓		**A/I**
	Highly developed interpersonal skills, negotiation, conflict management, feedback and partnership working	✓		**A/I**
Values and behaviour	*Outline NHS GMS Lead Provider values and behaviours here.*			
Other	Able to travel across multiple sites where required	✓		**I**
	Ability to work flexibly to meet the needs of the service	✓		**A/I**
	Proven proactive approach to work	✓		**A/I**

***evidence will take place with reference to the following information:**

A	Application form
I	Interview
T	Test or assessment
C	Certification

NHS GMS Chief Operating Officer

Position			
Job title	NHS GMS Chief Operating Officer	**Directorate/region**	NHS GMS to add
Pay band	ESM 1/2	**Responsible to**	NHS GMS add
Salary	NHS GMS to add	**Accountable to**	NHS GMS to add
Tenure	Permanent	**Responsible for**	NHS GMS to add
WTE	1	**Base**	NHS GMS to add

Main purpose of the Job

The Chief Operating Officer for the NHS GMS will be expected to oversee and provide the strategic direction for the development of the NHS GMS and all its functions. They will need to have experience in all aspects of change management and operational delivery. They will need to continually improve service delivery, maximising technological advances and capitalise on the opportunities to translate research into clinical practice to ensure the NHS GMS remains a leader in science, research and innovation and delivery of genomic testing.

The Chief Operating Officer will work across the NHS GMS geographical area to:

- Work closely with the Senior Leadership Team to deliver a cutting-edge genomic testing service that is underpinned by all appropriate structures (scientific, educational, quality, managerial and research) to maintain that position.
- Build and develop strong and effective teams and providing robust and visible leadership in a range of public facing and challenging environments across a large geography
- Provide oversight and operational and strategic direction to the delivery of all the functions of the NHS GMS and ensure that outcomes and impact can be measured

Work with partners and all providers within the geography to embed Genomic Medicine in mainstream medicine

Engender a culture that sees science, research and innovation as a key and integral function of the NHS GMS with an emphasis on translation of new technologies to clinical practice to improve genomic services, efficiency, and the care we offer patients.

Key duties and responsibilities
The Chief Operating Officer will be responsible for the overall operational direction and management of the NHS GMS and its staff to include the following areas. **Operational:** • Working with NHS partners to ensure alignment with strategic priorities, including in the NHS 10 Year Health Plan and the Life Sciences Sector Plan • Working with NHS partners to develop new services, for example the Genomics Population Health Service in conjunction with neighbourhood teams • Working with NHS partners across the NHS GMS geography and more broadly if required to support the embedding of genomics in the end to end pathway • Ensuring the NHS GMS has, or has access to appropriate technologies with sufficient capacity to meet service needs • Ensuring the quality and safety of the service in line with ISO 15189 standards and Care • Quality Commission regulatory arrangements for diagnostic services • Overseeing the performance of the NHS GMS against service contracts and signing off of the submissions of KPIs and other monitoring data • Management of risk and reporting of risk for the NHS GMS • Responsibility for line management and personal development of the NHS GMS Senior Leadership Team • Responsibility for all aspects of the NHS operational performance including finance, activity, access (health and safety and risk management in relevant areas) and other operational performance targets. • Responsibility for establishing and overseeing emergency planning and resilience and management of internal/external incidents. • Responsibility for driving standards, productivity, and value for money. • Ensure that the NHS GMS has in place the systems and processes to support the delivery of operational services, has the appropriate leadership and management capability within sites to ensure the delivery of a high-quality service. **Safety, Quality, Governance & Risk** • Establish and manage the quality, governance, and risk processes for the NHS GMS Geography • Develop a culture of continuous quality improvement in which excellence services can flourish • Ensure that laboratories participate in their local safety and quality programmes. • Continually seek to improve the safety, quality, and efficiency of services to support the provision of excellent patient care. • Ensure all services are patient focused and of high quality, optimising utilisation of human, financial and other resources. • Set up mechanisms to ensure high quality service delivery. • To cooperate with the Audit and Governance requirements of the NHS GMS Lead Provider, partners, and the Commissioner

- To facilitate auditors, regulators, and external quality assessors' requests.

Finance & Resource Management

- Oversee the systems to ensure activity can be accurately counted
- Oversee the development of any tariffs, ensuring they cover the systems costs
- Responsibility for ensuring that all management information is collated, and funding appropriately secured and distributed
- Ensure designated cost savings are identified and delivered

Research and Development

- Ensure facilities and protected time for science, research and innovation
- Prospectively identify areas for research and development, identify suitable partners for the research and work with them and the Science, Research and Innovation Director and other academics as appropriate across the geography to identify funding streams and bid for funds to deliver the work

Culture and Behaviour

- Demonstrate participative leadership
- Ensure that all staff are appropriately trained and updated.
- Ensure that all staff managed by the postholder undertake individual performance appraisal, operate within the appropriate Human Resource policies, take up appropriate and required training plans, and contribute to workforce planning activities.
- Promote a culture of diversity
- Ensure that managers and staff are treated consistently and fairly, feeling valued and supported to enable them to achieve their fullest potential in a working environment that is safe and stimulating to work in

Corporate Responsibilities

- Contribute to the achievement of the NHS GMS Lead Provider's corporate objectives.
- Strive to constantly improve the safety, effectiveness and efficiency of care provided to patients
- Share a corporate responsibility for policy and decision making, ensuring high standards of clinical and corporate governance and personal conduct at all times.
- Contribute to the development of best practice in recognising and meeting the needs of patients and staff.

Person specification				
Criteria		**Essential**	**Desirable**	**Evidence***
Qualifications	RCPath / PhD or Equivalent experience in NHS operational delivery	✓		**A/I**

	Proven experience of leading, developing and managing a large-scale genomic laboratory service across multiple providers	✓		**A/I**
	Able to demonstrate commitment to continuous professional development	✓		**A/I**
Knowledge and experience	Significant experience in a senior leadership role and developing partnerships to deliver services	✓		**A/I**
	Experience in service redesign, reconfiguration and consolidation	✓		**A/I**
	Experience in developing and establishing new services, including a clear strategic vision and direction and translation into successful outcomes. Experience in successfully supporting staff through large scale service change and service reconfiguration with knowledge of HR requirements and processes	✓		**A/I**
	Service and process improvement tools and techniques	✓		**A/I**
	Knowledge of appropriate legislation and its application to the running of a large and complex partnership	✓		**A/I**
	Up to date awareness of national policies and legislation affecting the NHS and genomic testing		✓	**A/I**
	Experience of negotiating contracts with NHS, private and public sector partnerships	✓		**A/I**
	A good knowledge of the requirements and delivery of rare		✓	**A/I**

	disease genomic testing and somatic genomic testing			
Skills capabilities and attributes	Demonstrate participative leadership	✓		**A/I**
	Innovative with strong quality focused approach, exceptional communication, interpersonal, negotiating and influencing skills	✓		**A/I**
	Ability to build effective relationships with a range of internal and external stakeholders, including clinicians and partners	✓		**A/I**
	Commitment and passion for patient-focused service with the ability to embed such an ethos at all levels of the organisation	✓		**A/I**
	Ability to develop and maintain effective multi-disciplinary working relationships particularly during times of change	✓		**A/I**
	Ensure that all staff are appropriately trained and updated	✓		**A/I**
	Ensure that all staff managed by the postholder undertake individual performance appraisal, operate within the appropriate Human Resource policies, take up appropriate and required training plans, and contribute to workforce planning activities	✓		**A/I**
	Ensure that managers and staff are treated consistently and fairly, feeling valued and supported to enable them to achieve their fullest potential in a working environment that is safe and stimulating to work in	✓		**A/I**

Values and behaviour	*Outline NHS GMS Lead Provider values and behaviours here.*			
Other	Able to travel across multiple sites where required	✓		**I**
	Ability to work flexibly to meet the needs of the service	✓		**A/I**
	Proven proactive approach to work	✓		**A/I**

***Evidence will take place with reference to the following information:**

A	Application form
I	Interview
T	Test or assessment
C	Certification

Mandated job roles

The job roles below outline roles mandated by the Commissioner to be appointed within each NHS GMS. The job descriptions should be written by the hiring organisation. A suggested WTE and banding is included in the table below but will depend on the size and complexity of the NHS GMS geography.

Post title	Average WTE	Suggested banding	Experience	Key roles
NHS GMS Leadership				
NHS GMS Deputy Chief Operating Officer (*to note may not be required in smaller geographies*)	1	Band 9	• Significant experience in a senior leadership role, developing partnerships to deliver services • Experience in service redesign, reconfiguration and consolidation • Experience in developing and establishing new services, including a clear strategic vision and translation into successful outcomes • Experience in successfully supporting staff through large scale service change and service reconfiguration	• Support the NHS GMS Chief Operating Officer in all work areas and able to deputise for them where needed • Responsible for overseeing the operational aspects of the NHS GMS, including laboratory services, clinical functions and data management systems • Lead initiatives to improve key performance indicators related to genomic testing turnaround times, diagnostic accuracy, and patient outcomes • Collaborate with clinicians, researchers, and other stakeholders to foster a culture of

				innovation and collaboration in Genomic Medicine • Work with education and training programmes to ensure that staff have the necessary skills and knowledge to utilise genomic technologies effectively • Responsible for identifying and mitigating operational risks associated with Genomic Medicine, including data security, ethical considerations, and regulatory compliance.
Finance Lead	0.5	Band 8C	• Significant experience in a senior specialist financial management role • An in-depth knowledge of NHS accounting rules and reporting requirements, and experience in applying these • Experience of interpreting relevant financial legislation and guidance • Experience in formulating annual financial plans with senior leaders, providing effective challenge to ensure these meet financial targets and deliver value for money for the organisation	• Responsible for the financial oversight and management with the NHS GMS geography • Develop and implement financial strategies aligned with the overall goals of the NHS GMS, including long-term financial planning and modelling service costs • Set, monitor, and manage budgets for the NHS GMS, ensuring resources are allocated effectively and efficiently • Responsible for ensuring all financial returns are timely and accurate

				• Oversee the financial billing and invoicing processes for the NHS GMS, ensuring accurate and timely payments • Ensure designated cost savings are identified and delivered, whilst implementing measures to control costs and improve financial efficiency within the NHS GMS
Contracts Lead	0.5	Band 8c	• Extensive relevant experience operating in a senior contract management role • Relevant senior manager experience of negotiating and working with commissioners in a healthcare environment • Experience of working within a complex and challenging environment, and managing demanding contract relationships with multiple stakeholders • Experience of commissioning, procurement, contracting and performance management either within the NHS or a sector of comparable complexity	• Responsible for overseeing all contractual aspects within the NHS GMS geography, ensuring agreements are properly managed, obligations are met, and risks are mitigated • Manage the drafting and negotiation of contracts related to the NHS GMS geography, ensuring they align with organisational goals and legal requirements, including sub-contracts • Responsible for identifying and mitigating potential risks associated with contracts, such as non-compliance, financial exposure, or reputational damage • Track contract performance, ensuring compliance with

				contractual obligations, and identifying potential risks or issues • Work closely with the finance lead to manage contract budgets and payments
NHS Genomic Laboratory Services				
Director of Genomic Laboratory Services	1	Band 9 / ESM1	• Registered with HCPC (essential), FRCPath / PhD or equivalent (essential) • Consultant Clinical Scientist with proven experience of leading and managing large scale clinical and diagnostic NHS genomic laboratory services • An expert within their field at national level as evidenced by involvement in professional (for example scientific, technical, educational or quality) and/or research activities	• Provide oversight and leadership across all laboratories in NHS GMS geography • Work closely with colleagues across the NHS GMS to ensure the laboratories deliver a cutting edge genomic testing service underpinned by appropriate expertise in cancer genomics, rare disease and population health to maintain that position • Responsible for the overall operational direction and management of the laboratory and its staff to include: • workforce planning of the laboratory to ensure sufficient staffing at the right skill level to deliver the service; • ensure the laboratory has, or has access to appropriate; technologies with sufficient capacity to meet service needs

				and deliver an efficient service, for example through the use of automation; • the education and training strategy for the laboratory; • performance of the laboratory against service contracts and sign off of the submissions of KPI and other monitoring data; • management or risk and reporting of risk for the laboratory; • ensure systems are in place for the management of all aspects of the staff of the laboratory; • manage the activity of the laboratory to ensure Genomic Tests are efficiently interpreted and reported in accordance with national guidance and the Turnaround Times; and • act in a consultant capacity on validation and interpretation of genomic data and clinical reporting within the laboratory
Deputy Director of Bioinformatics Services	1	Band 8d	• A principal/ consultant clinical scientist bioinformatician (or equivalent) with significant experience of developing, advising on and implementing bioinformatics solutions in the	• To be responsible for directing and managing all aspects of the bioinformatics activity of the NHS GMS Lead Provider

			genomics setting in an accredited/regulated environment for both rare disease and cancer • Has extensive experience at handling and interpreting genomic data including and up to the level of WGS • Has experience of reviewing and evaluating the latest tools and techniques • Has experience of leading, managing and developing teams • Registered with the HCPC as clinical scientist or equivalent experience	• To work closely with the Data and Digital function to support strategy development and delivery • To ensure that the bioinformatics pipelines and activity are compliant with ISO 15189:2012 standards • To ensure the NHS GMS Lead Provider complies with the implementation of data standards in respect to bioinformatics analysis
Rare Disease Scientific Lead	1	Band 8c	• Significant previous experience of diagnostic service leadership and management and development across a range of specialised genomics services • A recognised expert within own field of practice with a national professional or research profile • Registered with HCPC (essential) • FRCPath / PhD or equivalent (essential)	• Day to day delivery of the full range of Rare Diseases services delivered by the NHS GMS Lead Providers, representing the whole NHS GMS geography • Manages and leads teams of principal, senior and registered clinical scientists and genetic technologists • Ensures that the service is at the forefront of clinical science • Ensures standardisation in relation to referral triage, testing efficiency, interpretation and reporting across the geography

				• Provides senior scientific advice on technical, scientific and clinical aspects of the rare disease service including, appropriateness of referrals and samples, testing strategy, clinical interpretation and actionability of findings • Liaises with referring clinicians on the above • Provides senior clinical scientific input at relevant MDT's • Leads, initiates or collaborates locally or nationally in research and innovation within the service area • Provides advice and leadership on training and education of healthcare scientists within the NHS GMS Lead Provider • Plays a key role at national and local level in professional activities, working and advisory groups
Cancer Scientific Lead	1	Band 8c	• Significant previous experience of diagnostic service leadership and management and development across a range of specialised genomics services	• Day to day delivery of the full range of genomic cancer services of the NHS GMS Lead Provider. This includes liaising with departments including histopathology.

			• A recognised expert within own field of practise with a national professional or research profile • Registered with HCPC (essential) • FRCPath/PhD or equivalent (essential)	• Manages and leads teams of principal, senior and registered clinical scientists and genetic technologists • Ensures that the service is at the forefront of clinical science • Provides senior scientific advice on technical, scientific and clinical aspects of the service including, appropriateness of referrals and samples, testing strategy, clinical interpretation and actionability of findings • Provides senior clinical scientific input at relevant Genomic MDTs • Leads, initiates or collaborates locally or nationally in research and innovation within Your Geographical Area • Provides advice and leadership on training and education of healthcare scientists within the NHS GMS Lead Provider • Plays a key role at national and local level in professional activities, working and advisory groups
Population Health Scientific Lead	1	Band 8c	• Significant previous experience of diagnostic service leadership and management and development	• Day to day delivery of the full range of population health

			across a range of specialised genomics services • A recognised expert within own field of practice with a national professional or research profile • Registered with HCPC (essential) • FRCPath / PhD or equivalent (essential)	services delivered by the NHS GMS Lead Provider • Manages and leads teams of principal, senior and registered clinical scientists and genetic technologists • Ensures that the service is at the forefront of clinical science • Provides senior scientific advice on technical, scientific and clinical aspects of the rare disease service including, appropriateness of referrals and samples, testing strategy, clinical interpretation and actionability of findings • Liaises with referring clinicians on the above • Provides senior clinical scientific input at relevant MDT's • Leads, initiates or collaborates locally or nationally in research and innovation within the service area • Provides advice and leadership on training and education of healthcare scientists within the NHS GMS Lead Provider • Plays a key role at national and local level in professional

				activities, working and advisory groups
Genomic Laboratory Quality Manager	1	Band 8b	• Degree in a relevant scientific field, such as biology, genetics, or molecular biology, is often required • Extensive experience within the clinical laboratory setting • Experience of leading Quality Management and governance activities • A strong understanding of quality management principles, quality control processes, and relevant quality standards (e.g. ISO 15189) • Experience in leading audits, including identifying non-conformances and developing corrective action plans	• Responsible for implementation and coordination of the quality management system in all laboratories within the NHS GMS geography • Ensures the quality and safety of the service in line with ISO 15189 standards and Care Quality Commission regulatory arrangements for diagnostic services • Implementing document control processes, including the development, review and update of standard operating procedures • Monitor key quality indicators and performance against national and international standards, reporting on quality metrics, and identifying areas for improvement • Develop strategies to ensure quality standards are met and improved across the NHS GMS • Lead on the coordination of external assessment visits from regulatory and accreditation bodies at department level (e.g. UKAS, MHRA, and others)

Rare Disease clinical function				
Rare Disease Clinical Lead	0.8	Consultant	• Medical Consultant in rare disease genomics • Proven experience of managing team and leadership at departmental level (lead Consultant) or equivalent • A recognised national/international expert in own field as evidenced by publication record • Recognised expertise in clinically interpreting complex genomic information up to the level of whole genome sequencing	• Provide clinical leadership, direction and oversight across the Rare Disease clinical service • Work with other clinical leads across the NHS GMS as part of a multi-disciplinary team approach. • Work with other clinical leads across other NHS GMS Lead Providers. • Work with other clinical leads outside of the NHS GMS. • Act as the point of contact for clinicians seeking advice on appropriateness of referrals for genomic testing, whom to test within the family, use of HPO terms, understanding the Genomic Test report and actionability of findings and downstream management of the patient and their relatives and when to refer to a clinical genetic clinical or counselling service • Liaise with and advise the staff in the laboratory on referrals, testing strategy, phenotypes, actionability etc.

				• Play a key role in the Genomics MDT and the production of the final clinical interpretation in the context of the presenting phenotype • Play a substantial role in education and training across the clinical specialisms within the NHS GMS geography
Rare Disease Head of Programme	1	Band 8d	• Evidence of leading complex improvement initiatives in a multiple stakeholder environment; including business case preparation, service initiation / commissioning, and the development and monitoring of output and outcome measures • Experience in complex health or care delivery environments, particularly within rare diseases • Experience of establishing effective working relationships and delivering improvement across organisational boundaries. • Significant knowledge of applying budgeting, benefits tracking and demand management principles large scale and complex programmes and projects	• Provide strategic leadership, direction and oversight across the Rare Disease clinical service • Work with various system partners, including clinicians, researchers, patient groups, and industry partners, to advance rare disease research and care • Provide leadership across the rare disease services across the NHS GMS geography, including specialist services, ensuring equitable access across the geography • Maintain oversight of rare disease workstreams, ensuring visibility of progress and risk among senior leaders, and deploy creative solutions to deliver the benefits of genomics across mainstream care

				• Responsible for developing evaluation of the clinical service, benefits realisation, and demonstrating value, with sufficient evidence generation to support the transition of service changes into business as usual • Lead business cases for varied audiences, including new project funding and for service commissioning
Cancer Genomics clinical function				
Cancer Genomics Clinical Lead	0.8	Consultant	• Medical Consultant in cancer with experience in genomics • Proven experience of managing teams and leadership at departmental level (lead Consultant) or equivalent • A recognised national/international expert in own field as evidenced by publication record • Expertise in clinically interpreting complex genomic information up to the level of WGS	• Provide clinical leadership, direction and oversight across the cancer clinical service • Work with other clinical leads across the NHS GMS as part of a multi-disciplinary team approach. • Work with other clinical leads across other NHS GMS Lead Providers. • Work with other clinical leads outside of the NHS GMS. • Act as the point of contact for clinicians seeking advice on appropriateness of referrals for genomic testing up to the level of WGS, data requirements, understanding the Genomic Test

				report and actionability of findings and downstream management of the patient and when to refer to a clinical genetic clinical or counselling service • Liaises with and advises the staff in the laboratories on referrals, testing strategy, phenotypes, actionability etc. • Liaise closely with the histopathology departments to ensure appropriateness of fresh frozen tissue samples for DNA extraction with sufficient DNA and neoplastic cell content • Play a key role in the Genomics MDT and the production of the final clinical interpretation in the context of the presenting diagnosis and other investigations (integrated report) • Play a key role in ensuring a comprehensive plan across the NHS GMS for clinical trials access • Play a key and substantial role in education and training across the clinical specialisms within the NHS GMS geography

Cancer Genomics Head of Programme	1	Band 8d	• Evidence of leading complex improvement initiatives in a multiple stakeholder environment; including business case preparation, service initiation / commissioning, and the development and monitoring of output and outcome measures • Experience in complex health or care delivery environments, particularly within cancer genomics • Experience of establishing effective working relationships and delivering improvement across organisational boundaries • Significant knowledge of applying budgeting, benefits tracking and demand management principles large scale and complex programmes and projects	• Provide strategic leadership, direction and oversight across the Cancer Genomics clinical service • Work with various system partners, including clinicians, researchers, patient groups, and industry partners, to advance cancer research and care • Provide leadership across the cancer services across the NHS GMS geography, including specialist services, ensuring equitable access across the geography • Maintain oversight of cancer genomics workstreams, ensuring visibility of progress and risk among senior leaders, and deploy creative solutions to deliver the benefits of genomics across mainstream care • Responsible for developing evaluation of the clinical service, benefits realisation, and demonstrating value, with sufficient evidence generation to support the transition of service changes into business as usual • Lead business cases for varied audiences, including new project

				funding and for service commissioning
Pathology Clinical Lead	1	Consultant	• Medical Consultant in Cellular Pathology • Proven genomic knowledge and expertise • Proven leadership of managing teams and leadership experience at departmental level • Proven expertise in assessing and processing tissue for genomic testing • A recognised national / international expert in own field as evidenced by publication record and in contributing to national and international initiatives and professional guidelines in pathology and genomic science / medicine	• Understand the requirements and direct the processing of FFPE and fresh tissue for genomic testing and precision medicine IHC • Is responsible for ensuring that tissue is processed in a timely fashion so that genomic testing turnaround times can be met • Ensures that the tissue used for DNA / RNA extraction is of sufficient quality and tumour content to obtain high quality genomic test results and request alternative genomic testing approaches where required. • Liaise with clinical colleagues across the geography to engage, educate, and discuss genomic testing requirements to optimise access to genomic testing and where required onboard activity to the NHS GMS • Has knowledge & experience of digital pathology • Plays a key role in cancer genomic MDTs and ensures that molecular and pathology results are integrated

				• Is responsible for ensuring that medical and scientific staff in Cellular Pathology are fully trained in the processing of tissue for genomic testing • Plays a key role in setting up pathways for tissue transport across the geographical area of the NHS GMS Lead Provider
Population Health clinical function				
Population Health Clinical Lead	0.8	Consultant	• Medical Consultant in population health • Proven experience of managing teams and leadership at departmental level (lead Consultant) or equivalent • A recognised national/international expert in own field as evidenced by publication record • Recognised expertise in clinically interpreting complex genomic information up to the level of WGS	• Provide clinical leadership, direction and oversight across the population health • Work with other clinical leads across the NHS GMS as part of a multi-disciplinary team approach. • Work with other clinical leads across other NHS GMS Lead Providers. • Work with other clinical leads outside of the NHS GMS. • Act as the point of contact for clinicians seeking advice on appropriateness of referrals for genomic testing up to the level of WGS, data requirements, understanding the Genomic Test report and actionability of findings and downstream management of

				the patient and when to refer to a clinical genetic clinical or counselling service • Liaises with and advises the staff in the laboratories on referrals, testing strategy, phenotypes, actionability etc. • Play a key and substantial role in education and training across the clinical specialisms within the NHS GMS geography
Population Health Head of Programme	1	Band 8d	• Evidence of leading complex improvement initiatives in a multiple stakeholder environment; including business case preparation, service initiation / commissioning, and the development and monitoring of output and outcome measures • Experience in complex health or care delivery environments, particularly within population health • Experience of establishing effective working relationships and delivering improvement across organisational boundaries. • Significant knowledge of applying budgeting, benefits tracking and demand management principles	• Provide strategic leadership, direction and oversight across the population health programme • Work with various system partners, including clinicians, researchers, patient groups, and industry partners, to advance population health research and care • Provide leadership across the population health services across the NHS GMS geography, including specialist services, ensuring equitable access across the geography • Maintain oversight of population health workstreams, ensuring visibility of progress and risk among senior leaders, and deploy

			large scale and complex programmes and projects	creative solutions to deliver the benefits of genomics across mainstream care • Responsible for developing evaluation of the clinical service, benefits realisation, and demonstrating value, with sufficient evidence generation to support the transition of service changes into business as usual • Lead business cases for varied audiences, including new project funding and for service commissioning
Population Health Primary Care Lead	0.6	Consultant	• Hold a substantive appointment or honorary contract as a practising clinician in an NHS primary care practice with expertise / specialist interest in genomic medicine • Experience of implementing service improvement programmes. • Experience of participation in education and training • Leadership and/or management experience • Significant subject matter expertise across a number of key areas relating to primary care leadership, transformation and service improvement, large scale	• Working with other NHS GMS Lead Provider Primary Care Leads to develop the genomics primary care strategy. • Develop and lead a primary care strategy to ensure standardisation and embedding the primary care element of clinical pathways. • Develop opportunities to identify priority pathways incorporating genomics. • Facilitate mainstreaming opportunities within the NHS GMS Lead Provider geography.

			change and developing networks and collaborations	• Identify education & training requirements for the region, utlilising existing resources.
Pharmacy Lead	1	Band 8c	• Proven leadership experience on genomics projects relating to the use of medicines. • Recognised expert in genomics relating to the use of medicines and pharmacy. • Significant influencing skills within the wider pharmacy and medicines governance infrastructure to enable the safe and effective implementation of genomics to inform the use of medicines. • Demonstrated understanding of all sectors of the healthcare service and where and how medicines are delivered to patients, as well as a solid understanding of decision and funding routes for intervention and service commissioning. • Clear understanding of the drivers to enable genomics informed medicines optimisation and demonstrable experience of informing their development and deployment in practice	• Responsible for enabling the implementation of genomics informed medicines optimisation across their region in key areas, including cancer, rare and inherited diseases and population health. • Operates as part of the expert genomics MDT locally and nationally guiding the implementation of genomics into mainstream services. • Provides pharmacy leadership in genomics to pharmacy workforce in the locality and their teams, champions and affiliates to support local systems in delivering genomics. • Operates at regional level to drive genomics priorities within existing medicines governance structures and enables the mainstreaming of genomics informed medicines optimisation. • Contributes to local and national tools and resources to support the

				implementation of genomics informed medicines optimisation. • Directly inputs into national priorities and advances in for genomics informed medicines optimisation as key member of relevant governance groups. • Leads the delivery of the pharmacy genomics workforce, training and education strategic framework within their region. • Works with multidisciplinary clinical leadership to enable implementation of genomics informed medicines optimisation. • Informs the development of informatics solutions supporting genomics informed medicines optimisation. • Collaborates with specialised commissioning colleagues to monitor and enable the uptake of genomically informed precision treatments. • Collaborates with cancer alliance colleagues to support the use of genomically informed treatments. • Works with and advises clinical specialist colleagues to ensure the safe and equitable implementation

				of genomic testing into mainstream medicines pathways. • Supports clinical scientist colleagues with implementing testing pathways and reporting for genomics informed medicines optimisation, in line with national testing offer.
Nursing Lead	1	Band 8c/d	• NMC Registered Nurse with extensive clinical experience. • Educated to Masters level or equivalent level of experience of working at a senior level in a specialist area. • Evidence of ongoing continuing professional development. • Leadership and/or management experience. • Significant subject matter expertise across a number of key areas relating to nursing leadership, transformation and service improvement, large scale change and developing networks and collaborations	• Provide clinical leadership, experience and expertise in genomics by working in cohesion with Nursing Leads from all NHS GMS Lead Providers to identify, test and implement improvements in clinical practice to support the embedding of genomic medicine across the geography. • Bring together nursing and Neighbourhood Health Teams to identify and develop champions in nursing through education and training. • Work as part of a multidisciplinary clinical function to deliver priorities including equitable access to genomic testing and supporting the nursing workforce to use genomics safely, effectively and efficiently.

				• Participate in the development and delivery of the NHS GMS Lead Provider business plan, ensuring that nursing forms an integral part of the plan. • Develop innovative strategies to guide and coach nursing teams in developing and strengthening leadership skills in genomics. • Help identify and understand the touchpoints where education and training is needed across the geography, improving the education and capability of the nursing workforce in genomics.
Data and Digital				

Data and Digital Director	1	ESM1	• Senior individual and system leader with proven experience in working in informatics and demonstrable understanding of:
			  • genomic data and systems
			  • establishing and implementing complex informatics solutions across significant geographies
			  • the relationships between clinical and research activities in relation to genomics.
				• Responsible for ensuring the development of genomics informatics solution within the NHS GMS geography.
				• Responsible for ensuring genomics digital service, in alignment with Trust policies, is maintained and managed to ITIL standards.
				• Responsible for ensuring interoperability with other regional (including all providers across the care sectors i.e. secondary care

			• how to influence national and local stakeholders to adopt new systems and services. • Understand the role of informatics in system transformation and is recognised as a leader in the field. • Understands current and emerging NHS best practice for the safe and secure operation of digital and technology platforms.	as well as primary, community and emerging neighbourhood care) and national systems. • Responsible for ensuring the Minimum Data requirements are completed to the quality required and timescales agreed, influencing the Ordering Entities to comply with these requirements. • Responsible for ensuring that the laboratory is compliant with all legal requirements for data security and confidentiality. • Work collaboratively with Genomics England, the Commissioner and others to find innovative solutions for informatics in the NHS GMS.
Digital and Digital Head of Programme	1	Band 8d	• Evidence of leading complex improvement initiatives in a multiple stakeholder environment; including business case preparation, service initiation / commissioning, and the development and monitoring of output and outcome measures • Experience in complex health or care delivery environments, particularly within population health	• Responsible for developing evaluation of the digital service, benefits realisation, and demonstrating value, with sufficient evidence generation to support the transition of service changes into business as usual • Lead business cases for varied audiences, including new project funding and for service commissioning

			• Experience of establishing effective working relationships and delivering improvement across organisational boundaries. • Significant knowledge of applying budgeting, benefits tracking and demand management principles large scale and complex programmes and projects • Significant knowledge of digital and data standards, technical and architectural solutions,	• to be responsible for the introduction of new digital capabilities across the NHS GMS and establishing connectivity with relevant clinical systems across all providers in their geography • Provide strategic leadership, direction and oversight across the data and digital programme • Maintain oversight of all data and digital workstreams, ensuring visibility of progress and risk among senior leaders
Senior Data Analyst	1	Band 8c	• Substantial experience of complex data analysis, including advanced formulas and multiple data sources • Significant experience creating data and analytical reports, including presenting to different audiences • Experience of interpreting national data strategy and implementing this through complex programmes of work	• Responsible for ensuring NHS GMS data is managed appropriately, quality assured and aligned with national data standards • Responsible for the accurate and timely provision of information, including PLCM data encompassing outcome data and genomic end to end turnaround times, for the NHS GMS geography • Provide in depth analysis and reporting, including working with stakeholders to articulate data and reporting requirements

				• The subject matter expert for all data and system related queries, working with other staff to ensure data and system requirements are considered as part of other initiatives • Proactive performance management of services using data to identify areas for improvement, and working with responsible officers and managers to enact change
Data Analyst	1	Band 8a	• Demonstrated experience of data analysis, including advanced formulas and multiple data sources • Experience creating data and analytical reports, including presenting to different audiences	• Support senior analyst on all data workstreams
People – Workforce, education and training				
Workforce, Education and Training Director	1	Band 9 / Medical	• Significant and proven education, leadership and management experience in large and complex public sector organisation • Experience of working across boundaries and influencing senior colleagues at local and governmental level	• Provide strategic and operational leadership for a multi-professional approach, through workforce, education and training programmes within the NHS GMS geography • Develop and implement a comprehensive strategy for workforce development in

			• Knowledge of funding routes and experience of writing successful bids	genomics, aligned with the overall goals of the NHS • Identify current and future workforce gaps in genomics expertise across different professional groups and levels • Collaborate with various stakeholders, across organisational and professional boundaries and the multidisciplinary team • Designing, delivering, and evaluating a range of educational and training initiatives, including formal courses, workshops, and online resources
Workforce, Education and Training Head of Programme	1	Band 8d	• Evidence of leading complex improvement initiatives in a multiple stakeholder environment; including business case preparation, service initiation / commissioning, and the development and monitoring of output and outcome measures • Experience in complex health or care delivery environments, particularly within workforce, education and training • Experience of establishing effective working relationships and	• Provide strategic leadership, direction and oversight across the workforce, education and training • Work with various system partners, including clinicians, researchers, patient groups, and industry partners, to deliver upskilling and workforce development • Provide leadership across the workforce, education and training across the NHS GMS geography, including specialist services,

			delivering improvement across organisational boundaries. • Significant knowledge of applying budgeting, benefits tracking and demand management principles across large scale and complex programmes and projects	ensuring equitable access across the geography • Maintain oversight of workforce, education and training workstreams, ensuring visibility of progress and risk among senior leaders, and deploy creative solutions to deliver the benefits of genomics across mainstream care • Lead business cases for varied audiences, including new project funding and for service commissioning
Communications and PPI Lead	1	Band 8a	• Knowledge and experience of communications, media relations and digital media management techniques. • Experience of writing, editing and proof reading for different channels, styles and audiences, including digital media, press releases and newsletters. • Knowledge and understanding of best practice in communication evaluation techniques. • Experience of leading the development and implementation of communications strategies across digital and traditional media.	• Promote the reputation of the NHS GMS and the NHS GMS Lead Provider, through the creation and maintenance of positive relationships with internal and external stakeholders • Work with the wider NHS GMS Lead Provider Team support implementation of a communications and engagement strategy and action. • Communicate with staff at all levels, including key partners, the media, patients and the public via appropriate communication methods.

			• Ability to provide and receive highly complex and sensitive information, using influencing and negotiating skills to ensure cooperation and reach agreement. • Excellent organisational, problem solving and project management skills. • Experience of using Content Management Systems to organise and publish content. • Experience of assessing web traffic metrics and preparing reports, ideally using Google Analytics.	• Have responsibility for specific presentations to new and potential stakeholders, community presentations, taking the lead in managing associated events and campaigns and planning complex communications activities. • Develop and implement a joined up approach to promoting core activity internally, externally and with other key partners. • Shape the work and provide support to the NHS GMS Lead Provider PPI groups.
Science, Research and Innovation				
Research and Innovation Director	0.8	Consultant / ESM1	• Recognised national/international profile in leading and coordinating research and innovation projects and in contributing to national and international initiatives in genomic science/medicine • Understanding of the UK research funding landscape including experience of working with partners such as the National Institute of Healthcare Research • Knowledge and experience of the NHS innovation landscape and	• Lead the research and innovation function within the NHS GMS geography • Lead the NHS Genomic Networks of Excellence and other projects and initiatives supported within the NHS GMS • Work with the Genomics Research and Innovation Directors across each NHS GMS, the Commissioner, Genomics England and other key stakeholders, to

			partners, as well as what evidence is needed to influence adoption across the NHS GMS	align research and innovation activity • Drive the NHS GMS support for and recruitment to clinical trials • Drive research and innovation across the NHS GMS region working with a range of stakeholders and partners, including industry partners
Research and Innovation Head of Programme	1	Band 8d	• Evidence of leading complex improvement initiatives in a multiple stakeholder environment; including business case preparation, service initiation / commissioning, and the development and monitoring of output and outcome measures • Experience in complex health or care or research delivery environments, particularly within research and innovation • Experience of establishing effective working relationships and delivering improvement across organisational boundaries.	• Provide strategic leadership, direction and oversight across Research and Innovation • Work with various system partners, including clinicians, researchers, key stakeholders including Genomics England and Our Future Health, research funders, patient groups, and industry partners, to advance genomic research and care • Play a key role in ensuring there is a comprehensive plan across the NHS GMS for access to clinical trials • Provide leadership across the NHS GMS geography, ensuring equitable access to research opportunities across the geography

				• Maintain oversight of research and innovation workstreams, ensuring visibility of progress and risk among senior leaders, and deploy creative solutions to deliver genomics research aligned with clinical services • Lead business cases for varied audiences, including new project funding and for service commissioning
Transformation and Service Improvement				

Transformation and Service Improvement Director	1	Band 9	• Proven and significant leadership experience and/or formal management qualification
			• Significant experience in whole scale transformational change, with expert knowledge acquired over a significant period of time in the following areas:
			  • Service Improvement Design and Delivery
			  • Change Management
			  • Quality Management/Assurance
			• Subject matter expertise across a number of key areas relating to patient/treatment pathways
				• Evaluate the overall work programme to ensure improved patient recruitment and pathways
				• Drive the strategy for, supporting and ensuring alignment across the system
				• Drive reform and support organisational change and uptake of initiatives that support excellence
				• Engage with key strategic regional and national policy makers to inform development of strategy and policies
				• Provide expertise of best practice methodologies regulatory

			mapping in acute care settings, clinical leadership, stakeholder management and engagement for diagnostics or relevant clinical specialty	requirements, policy imperatives, innovation and technological developments and stakeholders knowledge

In addition to the mandated roles, the table below outlines additional roles that may be required subject to the requirements of the NHS GMS. Grades and WTE will be dependent on the size and needs of the geography.			
Role	**Grade**	**Mandated**	**Average WTE**
Admin support	Band 5	N	2
Genomic Laboratory Services			
Deputy Director of Genomic Laboratory Services	Band 8d	Depending on size of geography	
Rare disease			
Clinical Genomics Lead	Consultant	N	0.6
Genomic Counsellor Lead	Band 8c	N	0.6
Rare and Inherited Disease Senior Programme Manager	Band 8a	N	1
Rare and Inherited Disease Programme Manager	Band 7	N	1
Cancer Genomics			
Cancer Genomics Deputy Clinical Lead Solid Tumour	Consultant	N	0.6
Cancer Genomics Deputy Clinical Lead Haem-onc	Consultant	N	0.6
Pathology Senior Programme Manager	Band 8a	N	1
Cancer Genomics Senior Programme Manager	Band 8a	N	1
Cancer Genomics Programme Manager	Band 7	N	1
Population Health			
Population Health Deputy Clinical Lead	Consultant	N	0.6
Population Health Neighbourhood Service Lead	Band 8c	N	1
Population Health Senior Programme Manager	Band 8a	N	1
Population Health Programme Manager	Band 7	N	1
Data and Digital			
Digital Service and Operations Lead	Band 9	N	1
Clinical Safety Officer	Consultant	N	0.2
Information Governance Lead	Band 8b	N	0.4
Digital Programme Manager	Band 8a	N	2
Data Analyst	Band 8a	N	1
People - Workforce, education and training			
WTE Programme Manager	Band 8a	N	1
Workforce Clinical Lead	Band 8c	N	2
Research and Innovation			
Research and Innovation Senior Programme Manager	Band 8c	N	1
Research and Innovation Programme Manager	Band 8a	N	2
Transformation and Service Improvement			

Transformation and Service Improvement Lead Cancer	Band 8c	N	1
Transformation and Service Improvement Lead Rare and Inherited Disease	Band 8c	N	0.5
Transformation and Service Improvement Lead Population Health	Band 8c	N	0.5
Transformation and Service Improvement Programme Managers	Band 8a	N	2

Annex 5 - Definitions

Item	Definition
Additional Findings	Additional health information available from a genome sequence.
Annual NHS GMS Planning Guidance	Annual priorities and deliverables set out by the Commissioner for delivery by the NHS GMS.
Bioinformatics	The application of computer science and information technology to analyse and interpret biological data.
Genomic Molecular Tumour Board	A group of medical experts who collaborate to evaluate and provide personalised treatment recommendations for cancer patients based on the genomic characteristics of their tumours.
Cancer Genomics Improvement Programme	Engages representatives from across the entire sample pathway to support the equitable delivery of services for cancer patients.
Cancer Alliances	Bring together clinical and managerial leaders from different hospital trusts and other health and social care organisations, to transform the diagnosis, treatment and care for cancer patients in their local area.
Cellular Pathology Genomic Centre	Aims to optimise and maximise tissue-based predictive immunohistochemistry assessment and sample preparation for genomic testing.
Clinical Genomics Services (CGS)	Deliver a comprehensive clinical genomic and counselling service that directs the diagnosis, risk assessment and lifelong clinical management of service users of all ages and their families, who have, or are at risk of having, a rare genetic and genomic condition, including inherited cancer.

Clinical Indication	A specific medical condition or scenario listed on the National Genomic Test Directory for which a genomic test is available.
Clinical Senate	Provide impartial, strategic evidence-based advice and expert analysis on transformational change, through multi-disciplinary clinical and patient panels.
Commissioner	Means NHS England and any replacement or successor organisation, which for the avoidance of doubt shall include the Department of Health and Social Care.
Commissioning intentions	Provide the context for constructive engagement with providers, with a view to achieving the shared goal of improved patient outcomes and service transformation within the fixed resources available.
Decision Support	The use of genomic data and technologies to aid healthcare professionals in making informed clinical decisions.
Electronic Health Record (EHR)	A patient-centered record that makes information available instantly and securely to authorized users
Genomic Medicine	The use of genomic information and technologies to determine disease risk and predisposition, diagnosis and prognosis, and the selection and prioritisation of therapeutic options.
Genomic Multidisciplinary Team (MDT)	A group of professionals with different expertise to discuss clinical cases and results from genomic testing.
Highly Specialised Services	Highly Specialised Services refers to a subset of specialised services that cater to patients with rare or complex health conditions.

Order Management Service	Part of the core digital service in enabling to accelerate services into clinical settings. The scope of this service is the digitisation of genomics test ordering and reporting within the whole of NHSE, focusing solely on test order management (Whole Genome Sequencing (WGS) and Non-WGS Test Requests) and the management of test results and reports.
Genomic Population Health Service	A service which combines the power of genomics, new diagnostics and predictive analytics with AI, to proactively maintain wellness, identify and mitigating disease risks years – even decades – before symptoms arise.
Genomic Test Turnaround Time (TATs)	Measures the NHS' performance against Genomic turnaround time standards. These measures are used by local and national organisations to monitor the timely delivery of services to patients.
Geography	The area covered by the NHS GMS Lead Provider.
Health Innovation Networks	Established in 2013 to spread innovation at pace and scale – improving health and generating economic growth.
Integrated Care Boards (ICBs)	NHS organisations responsible for planning health services for their local population.
Laboratory Information Management System (LIMS)	Software used in laboratories and hospitals for the effective management of requests/orders, samples and reports.
Multi-omics	An approach to bringing together different omics, including genomics, proteomics, transcriptomics, epigenomics and microbiomics for analysis.
National Genomic Research Library (NGRL)	A comprehensive national database involving a single consent process that stores pseudonymised patient samples and

	health and genomic data for access by approved researchers.
NHS GMS Contract Management Framework	Means the contract management requirements put in place to monitor delivery and performance of all NHS GMS functions. The Framework will be set out as part of the contract documentation.
NHS Genomic Laboratory Hub (NHS GLH)	Responsible for coordinating genomic laboratory services for a particular part of the country.
NHS GMS Lead Provider	The provider which enters into a contract with the Commissioner for the provision of the services set out in this Specification for the relevant Lot.
NHS Genomic Medicine Service National Network (NHS GMS National Network)	Refers to the collective of the NHS GMS Lead Providers.
Neighbourhood Health Service	Deliver more care at home or closer to home, improve people's access, experience and outcomes, and ensure the sustainability of health and social care delivery.
NHS Genomic Network of Excellence	Partnerships between the NHS, academia, the third sector and industry to leverage expertise and resources from the broader genomics ecosystem, and to ensure there is a route towards rapid informing commissioning decisions.
Non-Invasive Prenatal Testing (NIPT)	Non-invasive prenatal testing is a way of screening for chromosomal anomalies in a fetus using a sample of blood from a pregnant woman.
Patient and Public Voice (PPV)	Patient and public voice partners' includes patients, service users, carers and families and the general public.
Patient Level Contract Monitoring (PLCM)	Provides patient level detail as one of a set of standards introduced to enable national consistency of the flow of cost and activity

	information from providers to commissioners.
Pharmacogenomics	The use of genetic and genomic information to tailor pharmaceutical treatment to an individual.
Population Health	Aimed at improving the health outcomes and wellbeing of an entire population while reducing health inequalities.
Precision Medicine	The application of emergent technologies to better manage patients' health and to target therapies to achieve the best outcomes in the management of a patient's disease or predisposition to disease.
Unified Genomic Record (UGR)	Individual patient records and single point of truth for a patient's genomic data, linked to existing NHS digital systems and clinical data, and available nationally.