Genomic Medicine Service Alliance guidance

February 2020

NHS England and NHS Improvement

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Foreword

Across the world, scientific and technological advances in genomics, artificial intelligence and digital technology to for example, are rapidly evolving our understanding of the causes and drivers of disease and the effectiveness of drugs and interventions.

In combination, these advances have the potential to transform the quality of care by improving diagnosis, prediction and prevention of disease.

The NHS, as the single biggest integrated healthcare system in the world, is well placed to harness the potential benefits for NHS patients but also demonstrate the nation's competitive advantage in enhancing our understanding of disease, and ability to facilitate new ways of working with academia and industry to support the discovery of medicines and technologies for earlier detection and stratified treatment.

However, it will only be possible to secure the benefits if everyone, including, patients, the whole multi-professional team across the care system – medics, nurses, midwifes, allied health professionals, healthcare scientists, pharmacists – and academics are able to actively contribute and make this vision a reality.

The NHS Genomic Medicine Service Alliances will have a critical role in facilitating strong collaborations across large geographies, working with patients and the public to build trust in genomics, and providing clinical leadership to enable the multi-professional workforce to use genomics safely, effectively and efficiently. The NHS Genomic Medicine Service Alliances will focus on reducing unwarranted variation and delivering the NHS Long Term Plan commitments to make demonstrable improvements to outcomes for our patients and the populations we serve.

By taking this approach, the NHS will become the first health service in the world to systematically embed genomics into routine care and make significant strides to create a system focused on improving health, not just treating illness, able to predict accurately disease and tailor treatments, with shared decision making at its heart.

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NHS Chief Scientific Officer for England
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Chair NHS Improvement and Interim Chair of Genomics England

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Summary

1. Through the world-leading 100,000 Genomes Project, the NHS was the first health care system to use whole genome sequencing (WGS) as part of clinical care on a large scale. Rather than solely aiming to achieve the sequencing target, the project focused on the transformative potential of genomics. Thirteen NHS Genomic Medicine Centres (GMCs) were contracted by NHS England to consent participants, validate findings, report results, and broaden the use of genomics across clinical specialities.

2. To build on the project infrastructure, seven NHS Genomic Medicine Service (GMS) Alliances are being developed across England. The NHS GMS Alliances will bring together providers to work in partnership across geographies with populations of between five and ten million, aligned to the NHS Genomic Laboratory Hub (GLH) footprints to provide consistency with genomic testing pathways.

3. The NHS GMS Alliances will support the systematic implementation of genomic medicine into the NHS and the delivery of the NHS Long Term Plan ambitions, by creating a learning environment to support the rapid adoption and spread of scientific advances. The NHS GMS Alliances will be accountable for achieving demonstrable improvements across the whole geography in the following areas:

    i) equitable access to standardised end-to-end pathways of care, inclusive of genomic testing and clinical genetics and genomic counselling services;

    ii) access to treatments and medicine optimisation driven by comprehensive genomic and diagnostic characterisation;

    iii) the number of people of accessing clinical trials by ensuring the systematic consideration of eligibility to clinical trials for patients who would potentially benefit; and

    iv) 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.

Developing the NHS GMS Alliance Operating Model

4. This document has been developed to support the introduction of the NHS GMS Alliances and inform the development of local plans.

National coverage across seven geographical areas

5. Thirteen NHS GMCs were established to support the delivery of the 100,000 Genomes Project. Although many Trusts across the country participated in the project, there was not complete engagement all of providers across the country and little or no involvement of primary and community care.

6. Going forward, to enable the NHS Genomic Medicine Service to provide equitable and consistent access to genomic testing services across the whole of England, seven NHS Genomic Medicine Service (GMS) Alliances will be created, aligned to the NHS Genomic Laboratory Hub (GLH) geographies.

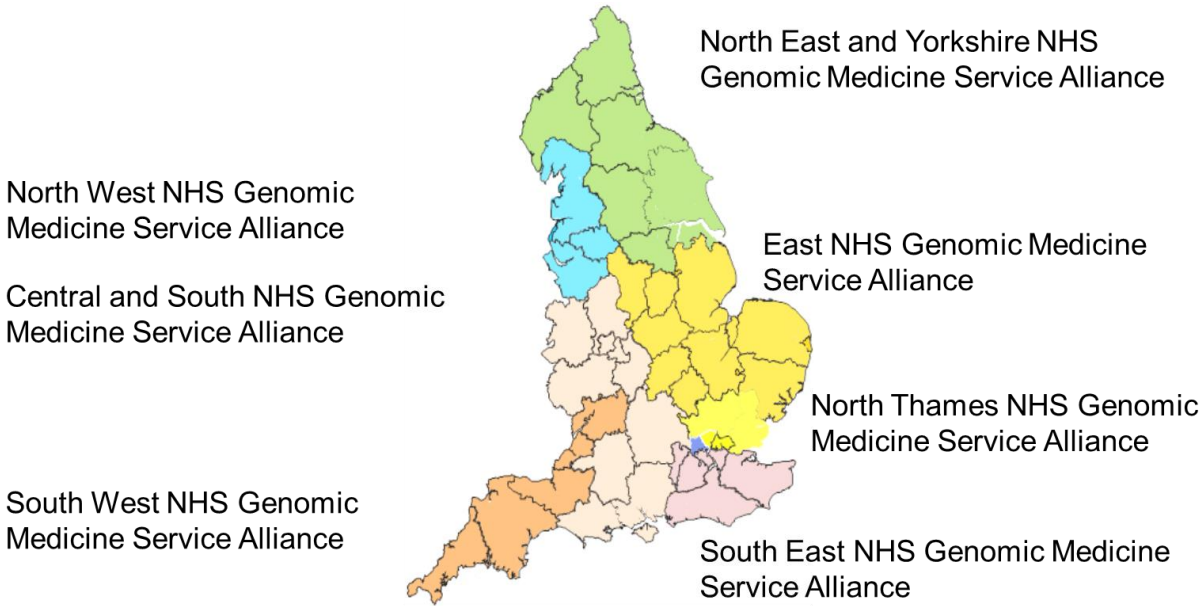

Figure 1: NHS GMS Alliance geographies

Collaboration across providers

7. An NHS GMS Alliance is defined as a collective of providers working in partnership to support the embedding of genomic medicine for a given population. Although all providers within the geography are expected to be

actively engaged with the work of the NHS GMS Alliance, each alliance will be led by a partnership between a small number of partners.

NHS GMS Alliance – a collective of providers working in partnership to support the embedding of genomic medicine for a given population.

NHS GMS Alliance Partner – a provider with direct responsibility for the delivery of specific elements of the NHS GMS Alliance business plan. NHS GMS Alliance Partners will be expected to evidence recognised leadership and experience of bringing partners together, with a track record in the delivery of genomic services (laboratory or clinical) and/or a signification contribution to the 100,000 Genomes Project.

NHS GMS Alliance Network member – all providers and other NHS organisations and networks actively engaged and working together across the geography to support systematic embedding of genomics into care pathways in all relevant clinical applications.

8. A key role of the alliance will be to develop a strong and effective Alliance Network, that facilitates engagement with all other NHS providers and organisations across the geography to achieve the equity and standardisation of genomic medicine, including for example:

    - Primary Care Networks
    - Pathology Networks
    - Cancer Alliances
    - Integrated Care Systems
    - Academic Health Science Networks
    - Academia

9. To support the development of the Alliance Network, the NHS GMS Alliance will need to work with and actively engage with NHS England and NHS Improvement regional teams to develop an appropriate operating model for the geography that the regional team can work with and endorse.

Operating as a national network

10. A single, nominated representative will be identified by the GMS Alliance Partners to work as part of the National Alliance Network. The national representative will be a named senior individual within the NHS GMS Alliance

who has the delegated responsibility to represent the NHS GMS Alliance at a national level and support decision making at a national level.

Integrated governance

11. To facilitate partnership working across the geography, each NHS GMS Alliance will require:

A Partnership Board to drive collaborative working and buy-in across partners to develop and oversee the delivery of the NHS GMS Alliance business plan, with a robust mechanism to track delivery and outcomes. A clear, integrated governance structure with the NHS GLHs and clinical genetic services.

An Alliance Network to facilitate engagement with all other NHS providers and organisations across the geography to achieve the equity and standardisation of genomic medicine, including appropriate and effective engagement with Primary Care Networks, Pathology Networks, Cancer Alliances, Integrated Care Systems, Academic Health Science Networks and Academia.

A clinical leadership team with appropriate experience and seniority setting the strategic direction and driving implementation across the geography. The clinical team will involve individuals from multiple branches of medicine and related multi-professional services, who are able to demonstrate leadership and/or expertise in genomics, with a track record of delivery of change. The clinical leadership structure will need to be integrated with the existing scientific and clinical leadership posts that are already funded as part of NHS services, inclusive of the NHS GLHs and clinical genetic services.

NHS GMS Alliance infrastructure

12. Each NHS GMS Alliance will put in place a core infrastructure to support the operation of the NHS GMS Alliance and the delivery of the business plan. Each NHS GMS Alliance will need to put in place the following:

i) A Clinical Director who will have responsibility for setting the strategic clinical direction and leading all aspects of the clinical activity of the NHS GMS Alliance. The Clinical Director should be able to demonstrate significant leadership and expertise in genomics and be a practicing clinician.

ii) Programme and project management support to coordinate the work of the NHS GMS Alliance.

iii) Communications and PPI expertise that supports communication and engagement across the NHS GMS Alliance geography.

iv) A structure to ensure appropriate and systematic input from the following areas: medical; nursing, midwifery and allied healthcare professionals; pharmacy; research and innovation; workforce development; informatics and data.

13. It is expected that the clinical leadership will be made up of individuals from across multiple branches of medicine, who are able to demonstrate leadership and/or expertise in genomics, with a track record of delivery of change.

14. NHS England and NHS Improvement will review the proposed leadership structure as part of the provider selection process. This could include plans for a shadow leadership structure being put in place from April 2020 with a clear plan and timeline for putting in place the permanent structure.

People and communities engagement

15. Each NHS GMS Alliance will need to develop appropriate and effective mechanisms to ensure that the local and regional communities including patients accessing genomic services, have their voices heard in the set up and delivery of services and in any research projects and initiatives.

16. People and communities should be represented in the governance structures of the NHS GMS Alliance and their views should be considered to inform the operation and development of services; inform changes to models and/or pathways of care; and inform the development of patient information materials.

17. NHS GMS Alliances will be required to recruit a communications and people communities lead to coordinate and implement all communications, engagement and awareness raising activity on behalf of the NHS GMS Alliance.

Provider selection process and timescale

18. A provider selection process will be used to identify the NHS GMS Alliances. The purpose of the selection process is to assess the proposals to ensure support and collaboration across the geography, and to demonstrate there is a clear plan to establish the clincial leadership and governance necessary to creat effective partnerships.

19. The process for prospective NHS GMS Alliances involves two phases.

Phase one – selection phase

20. Each NHS GMS Alliance will need to submit their plans using a national template, which will include:

    i) endorsement from a senior member of the relevant NHS England and NHS Improvement regional team in the NHS GMS Alliance geography;

    ii) the proposed governance;

    iii) leadership and infrastructure, that sets out clear roles and responsibilities within the structure and how they will ensure delivery of the requirements of the NHS GMS Alliance; integration with existing infrastructure; any plans for a shadow leadership structure to be put in place and a timeline for a permanent structure to be established; and requirements for resources to establish the infrastructure; and

    iv) oversight arrangements to ensure delivery of the 100,000 Genomes Project activities.

21. The responses will be scored on the information provided. Submissions will be assessed by a national panel.

22. Following assessment, initial submissions will be given feedback and placed on one of two tracks:

    i) Early adopters: aiming for the NHS GMS Alliance to be operational from April 2020

    ii) In development track: aiming for the NHS GMS Alliance to start contractually from October 2020, allowing for additional time to develop

proposals, prepare and mobilise. Proposals may be subject to a further submission process.

23. Following the provider selection process, NHS GMS Alliances who are part of the early adopter track will receive an infrastructure payment to support leadership and project management support for the GMS Alliance. The funding allocation will be circa £500k to £1 million per NHS GMS Alliance, dependent on the size of the NHS GMS Alliance geography, the complexity of the geography and the delivery requirements of the Alliance.

24. The proxy populations for each GMS Alliance geography is outlined in the table below:

	Proxy Population
Yorkshire & North East	*16.0%*
North West	*12.6%*
East Midland & East of England	*15.1%*
West Midlands, Oxford & Wessex	*19.3%*
South West	*7.7%*
London North	*13.9%*
London South	*15.4%*
Total	***100.0%***

Table 1: Proxy populations for each GMS Alliance geography

25. In addition to the infrastructure payment, up to £10 million of non-recurrent funding will be available in 2020/2021 to support the delivery of the projects agreed in the business case. The funding allocations will be agreed as part of the development of the business case.

26. It expected that project funding will be used to fund both the delivery of the project and any additional infrastructure required to deliver the projects (for example additional clinical leadership or project management).

Phase 2 – development and approval phase

27. Following the selection process, the NHS GMS Alliances will develop their detailed business plans for 2020/21.

28. The business plan should set out the activities the NHS GMS Alliance will undertake to meet all the deliverable and KPIs set out in the priority areas, the timelines, key milestones and funding requirements.

29. The business cases will be reviewed and approved by a panel of representatives from NHS England and NHS Improvement.

Provider selection information sessions

30. To support the provider selection process NHS England and NHS Improvement will hold a series of information sessions as follows:

    i) w/c 24th February – to provide an overview of the process and as an opportunity for submitters to ask any clarifying questions regarding the process the documentation that is required

    ii) weekly virtual surgeries as an opportunity to seek further clarifications prior to the deadline for submissions

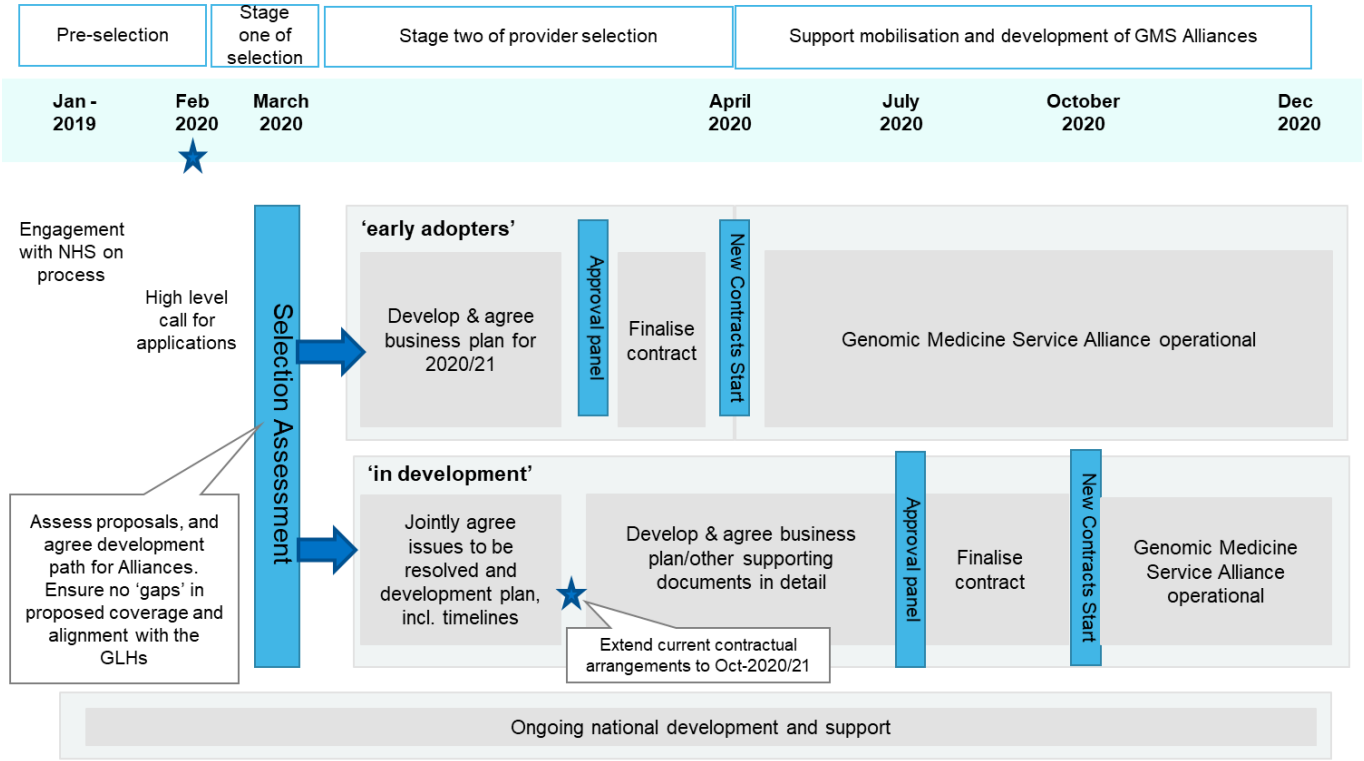

Figure 3: GMS Alliance provider selection process

Timeline

31. The below table provides the timescales for the Provider Selection process:

Activity	Timescale
High level call for applications	Monday 24th February 2020
Information shared to support submission process	w/c 24th February 2020

Weekly surgeries to support submission process	From w/c 2nd March 2020
Submission of application template	13:00 on 23rd March 2020
NHS E/I review panel	w/c 23rd March 2020
Initial feedback from review panel and initiation on partnership working on the business plan	w/c 23rd March 2020
Partnership agreements signed with early adopters	April 2020
Process for 'in development track' to continue	April – October 2020

Developing a 2020/21 business plan

32. Following the provider selection process, each NHS GMS Alliance will need to develop a one-year business plan setting out how the priorities for 2020/21 will be met and how the KPIs (listed in the annex) will be achieved. The priorities, outlined in more detail in the document, are:

    i) Completion of the 100,000 Genomes Project

    ii) Retaining and building patient and public trust in genomics

    iii) Strengthening partnerships across the geography

    iv) Using genomics to improve health and care productivity, quality and efficiency

    v) Supporting the multi-professional workforce to use genomics safely, effectively and efficiently

    vi) Maximising opportunities to facilitate and participate in research and innovation

    vii) Operating as a national network and representing the NHS GMS internationally

33. NHS England and NHS Improvement will work with the successful NHS GMS Alliances to develop the business plan and agree a financial allocation for 2020/21.

Overview of the financial model

34. It is anticipated, the GMS Alliance funding model will comprise the following elements:

    i) Funding allocation for NHS GMS Alliance infrastructure costs as agreed with NHS England and NHS Improvement;

    ii) Activity-based payment for the completion of 100,000 Genomes Project activity (additional findings, pharmacogenomics and reconsenting activity only);

    iii) Non-recurrent funding for the delivery of nationally agreed transformation projects and pilots.

35. In addition, through the NHS GMS Alliances' involvement in the NHS GMS Research Collaborative, it is anticipated that additional funding could be secured for the delivery of agreed research projects.

36. The proposal for the distribution of the funding between the NHS GMS Alliance Partners should be included within the provider selection application.

Contracting model

37. A key principle to the operating model of the NHS GMS Alliance is collaboration. NHS England and NHS Improvement will commission the NHS GMS Alliances through a Network Agreement. To facilitate the NHS GMS Alliances to plan for improving services in the medium term, the agreements will be for up to four years (depending on start dates) in line with the timeline of the NHS Long Term Plan.

38. The Network Agreement will be signed between NHS England and NHS Improvement and all NHS GMS Alliance partners. The Network Agreement will be included in each NHS Standard Contract that a provider holds with NHS England and NHS Improvement regional specialised commissioning team as a document relied upon.

Assurance

39. NHS England and NHS Improvement will be responsible for the overall assurance of the Network Agreement with the NHS GMS Alliances.

40. NHS England and NHS Improvement will undertake quarterly assurance and the NHS GMS Alliance will be expected to be able to demonstrate progress and delivery against the business plan deliverables and KPIs.

Priority 1 – Completion of the 100,000 Genomes Project

41. During Q1 of 2020/21, a small number of NHS GMCs will still have primary diagnostic results to return in line with timetable agreed by NHS England and NHS Improvement. Although there will be no further funding provided for this purpose, the NHS GMS Alliance will need to review, oversee and report on the progress of the return of results to participants.

42. In addition to the completion of the primary diagnosis, there will be a number of other project activities that will need to be completed during 2020/21, these include:

    i) Return of additional findings, including supporting the evaluation of the impact on participants, professionals and the system.

    ii) Return of pharmacogenomics, including supporting both the implementation planning and the evaluation process.

    iii) Re-consenting participants (children when turning 16).

    iv) Patient withdrawal.

43. A key priority for the NHS GMS Alliance will be to ensure that the implementation of the new NHS GMS Alliance model causes no disruption to the completion of the return of results from the 100,000 Genomes Project and that we can continue to learn and gather evidence as the legacy activities complete. NHS GMS Alliances will need to work closely with existing NHS GMCs in their geography through the application phase to agree any requirements for close down and handover activities.

Deliverables

- **By end of Q1, operating clear reporting and oversight arrangements to monitor delivery of all remaining 100,000 Genomes Project activities across the geography.**

- **By end of Q2, agreed plan with NHS England and NHS Improvement requirements for re-consenting patients (children when turning 16) and withdrawal.**

Priority 2 – Retaining and building the trust of people and communities in genomics

Putting patients, people and communities at the centre

44. Ensuring people and communities are at the centre of the work of the NHS to embed genomics into routine care is crucial to ensure the design and delivery of the best possible services.

45. One of the successful elements of the 100,000 Genomes Project was the strength and involvement of the participants within the Project – both through the Genomics England national participation panel and locally through the NHS GMCs.

46. At a national level, NHS England and NHS Improvement is recruiting Patient and Public Voice Partners to participate within the national governance groups and establishing a national genomics people and communities forum. The forum will involve national patient representatives and representatives from the NHS GMS Alliances alongside patient advocacy and research charities.

47. NHS GMS Alliances will need to develop plans to involve representation from people and communities at a local and regional level, including within all governance structures, and monitor effectiveness to assess the positive impact this has on services.

Communications and engagement strategy

48. NHS GMS Alliances will ensure that a communications and engagement strategy is in place to support organisational priorities. The strategy should set out:

    i) how the NHS GMS Alliance will assess local and regional needs in relation to awareness and understanding of the service;

    ii) how people and communities will be involved in the development of the service;

    iii) how they will be kept informed about the service offer and how to access it (for example through local awareness raising activities and surveys);

iv) how the service will ensure it targets the broadest possible diversity of local communities;

Implementing the patient choice framework

49. NHS England and NHS Improvement, in partnership with Genomics England, have developed a patient choice framework, which will initially be implemented for WGS testing only. The framework focuses on two key aspects of choice – clinical care and research. The approach will see the two aspects presented together; however, patients will be assured that choosing not to participate in research will not impact on the standard of their clinical care.

50. The patient choice framework includes the implementation of the national Record of Discussion form, which will be used by clinicians to record that a discussion has taken place between the patient and clinician, that a patient has a full understanding of the implications of having a genomic testing, and the outcome of that conversation.

51. The NHS GMS Alliances will work with NHS England and NHS Improvement to explore whether a national patient choice approach for non-WGS testing, such as WES and large cancer panels, should also be implemented.

52. To support the patient choice framework, NHS England and NHS Improvement has developed a suite of supporting information for clinicians and patients. These materials are being developed by NHS England and NHS Improvement in partnership with Genomics England who will be responsible for the research element, and Health Education England who are supporting the clinical education element.

Deliverables

- **By its first meeting, to have appointed a people and communities representative to the NHS GMS Alliance Partnership Board.**
- **By Q2 have a communications and engagement strategy in place to support the work of the NHS GMS Alliance.**
- **By no later than 3 months after the start of the WGS service established have a mechanism to monitor, identify and agree actions to address any**

issues related to uptake of the research offer in the patient choice discussions.

Priority 3 – Strengthening partnerships across the geography

Securing provider engagement across the whole geography

53. To create strong engagement across the geography the NHS GMS Alliance, with support and buy in from NHS England and NHS Improvement regional teams, will need to actively engage with all providers including primary and community care and seek their participation within the NHS GMS Alliance Network.

54. Throughout 2020/21 it is expected the NHS GMS Alliance Network will be developed to create effective and appropriate mechanisms for:

    i) Collating and sharing information from across the geography with NHS England and NHS Improvement;

    ii) sharing information with providers and clinical teams;

    iii) involving and engaging with patients, the public and clinical teams across the geography; and

    iv) bringing teams together to share best practice, evaluate operating models and improving pathways of care.

Deliverables

- **By end of Q2 have begun operating the NHS GMS Alliance Network to actively engage with all providers and other NHS organisations and networks across the geography.**

Priority 4 – Using genomics to improve health and care productivity, quality and efficiency

Reducing variability and improving equity of access

Access to testing

55. The NHS has been at the forefront of the implementation of genomic medicine, however, variation in access to genomic testing remains a challenge, with too many examples of patients not being able to access appropriate testing leading to unwarranted variation in clinical outcomes.

56. The NHS GMS Alliances, through their multi-professional clinical leadership inclusive of nursing and midwifery, have a role in supporting equity of access to genomic testing, including WGS, through:

    i) Supporting the mapping of end to end genomic testing pathways for all clinical indications for rare and inherited disorders and cancer in the National Genomic Test Directory (the Test Directory).

    ii) Ensuring that all pathways are being delivered systematically across the geography and identifying any barriers to access.

    iii) Improving the education and capability of clinical specialities to request a genomic test and understand appropriate action on receipt of the results through a comprehensive workforce development strategy.

    iv) Introducing new models of care that support early access to genomic testing and understanding the gaps and bottle necks in the system and implementing systematic solutions across the geography.

    v) Understanding areas of unmet clinical need and/or identifying when and where genomic testing may be important to consider as part of a patient's presenting condition.

Lynch syndrome

57. Some cancer types – mainly breast, ovarian, colorectal and prostate – can be strongly influenced by inheriting certain genetic variants which can significantly increase a person's likelihood of developing cancer.

58. Identifying individuals who are at high risk of developing these inherited cancers provides an important opportunity to reduce cancer burden through surveillance and early detection of cancers in this population.

59. Since February 2017 the National Institute for Health and Care Excellence (NICE) has recommended screening of all newly diagnosed colorectal tumours for Lynch Syndrome. Testing for Lynch syndrome by Microsatellite Instability (MSI) testing will simultaneously stratify colorectal cancer patients for treatment with immunotherapy and identify those patients who are unlikely to benefit from chemotherapy. Where MSI testing identifies a deficiency in the DNA mismatch repair pathways, genetic testing of germline DNA should be performed to confirm Lynch Syndrome. In such cases, cascade testing of family members may be performed to identify further at-risk individuals who may benefit from surveillance with a view to early detection of new cancers.

60. Although the Test Directory includes Lynch Syndrome testing, and therefore NHS GLHs are expected to be providing access to this testing within their geography, a wider collaborative approach is required to ensure that individuals are identified and managed appropriately across the end-to-end patient pathway.

61. NHS GMS Alliances are expected to work closely with NHS GLHs, Cancer Alliances and Pathology services within their geography to ensure that pathways are in place to identify individuals with Lynch syndrome and that they (and their families) are managed appropriately.

Familial Hypercholesteremia

62. Expanding access to genetic testing for Familial Hypercholesterolaemia (FH), which causes early heart attacks and affects at least 150,000 people in England, will enable us to diagnose and treat those at genetic risk of sudden cardiac death. It is estimated that currently only 7% of those with FH have been identified, but over the next five years the NHS will aim to improve that to at least 25% in the next five years.

63. During 2020/21 the NHS GLH network will be expected to make demonstratable progress to increase the identification of at-risk individuals. To support an increase in the number of people being identified with the FH, the NHS GMS Alliances will need to work with AHSNs, lipid clinics, primary care and others to identify eligible patients and support the implementation of sustainable models of care.

Sudden Cardiac death

64. During 2020/21 selected NHS GLH representatives will explore with the coronial system, NHS England and NHS Improvement, and British Heart Foundation the possibility of whether a referral pathway between the coronial system and the NHS in England can be developed to enable family members, at risk of sudden cardiac death (SCD), to be identified, and where appropriate genetic testing undertaken. NHS GMS Alliances are expected to support this project to ensure that pathways are set up within their geography where necessary.

Data collection

65. A key enabler to improving the quality of the testing available in the NHS is to collect data and information to monitor whether patients can access the most appropriate testing in an appropriate timeframe.

66. During 2020, the NHS GLHs will establish monthly reporting of genomic testing activity, which will include capturing information on test outcome. The NHS GMS Alliance will need to work with the NHS GLH to understand and evaluate the impact of testing on patient care and subsequent management and identify potential unmet need.

Deliverables

- **Work with the NHS GLH to develop a programme of work to review access to genomic testing included within the National Genomic Test Directory across the geography, including quantifying unmet clinical need and actions to address.**

- **Throughout 2020/21 work in partnership with the NHS GLH and NHS England and NHS Improvement to implement appropriate systems and processes to collect clinical outcomes data and develop appropriate mechanisms to use the data to drive demonstrable improvements for patients.**

- **Throughout 2020/21 oversee the quality of the standardised clinical data being provided to support the introduction of WGS and support the developments of more detailed and standardised clinical and phenotypic data collection for non-WGS testing when agreed.**

- **Throughout 2020/21, work with the NHS GLHs to ensure pathways are in place to identify individuals with Lynch syndrome.**

- **By June 2020, working with the NHS GLHs and other system partners develop a plan to support the identification of individuals at-risk of FH to support the NHS GLH to increase the identification of people with FH, including ensuring end-to-end care pathways are in place to follow up the patient and family members.**

- **If required, actively contribute to the pilot project for the introduction of genetic cascade testing for families affected by sudden cardiac death in collaboration with the coronial system, NHS England and NHS Improvement, and British Heart Foundation and the development of national guidance.**

Access to treatments and interventions based on genomic information

67. The NHS is on a journey towards embedding a personalised medicine approach into mainstream healthcare. For the NHS, we must consider not whether we should go down the route of personalised medicine, but instead how we can best respond and adapt so that those who could benefit have the opportunity – regardless of where they live, the illnesses they have, or where their care is provided.

68. When talking about personalised medicine in the NHS, this includes treatments that fall into the following categories:

    i) Gene therapies and Advanced Therapeutic Medicinal Products e.g. CAR-T therapy

    ii) Targeted treatment where access is based on a genetic or genomic test result e.g. targeted chemotherapy

    iii) Histology-independent, or tumour-agnostic, products; a new class of cancer therapies developed for use for tumours that express a genomic alteration, regardless of where in the body the cancer originated

    iv) Pharmacogenomic test guided therapy e.g. abacavir and HLA-B*5701 or fluoropyrimidines and DPYD

69. Personalised medicine will provide opportunities to improve how we treat disease. Based on comprehensive genomic and diagnostic characterisation, different subtypes of patients within a given condition can be identified, and treatment can be tailored to the underlying cause. Critical to the integral link between the use and optimisation of medicines and the expression of genomic variants will be the involvement of pharmacists and the broader pharmacy workforce.

Improving outcomes and access to clinical trials

70. During the 100,000 Genomes Project, it was identified that different approaches were being adopted across the NHS to validate and interpret results. In some parts of the country, to help improve the pathway for more complex tests, clinical scientists work directly with the referring clinicians to triage the variants and help improve the diagnostic rate of the test. While in other parts of the country, to help clinical teams to determine the actionability of the findings from more complex testing, genomic MDTs have been established.

71. During 2020/21 to standardise the approach to genomic variant triaging, curation and validation across the network, the NHS GLHs will work together to standardise genomic MDT processes to standardise reporting and actionability of genomic results and to engage referring clinicians in a streamlined and efficient way. The NHS GMS Alliances will need to oversee this process to ensure that the standardised approaches are being successfully implemented across the geography.

72. In addition, NHS GMS Alliances will need to identify current testing pathways that are in operation for cancer and ensure that genomic MDT processes are aligned with existing referral and reporting pathways including cancer MDTs.

73. The NHS GMS Alliance should also work to ensure that eligibility to clinical trials is also considered on systematic basis for patients who would potentially benefit, wherever they are in England.

DPYD Testing

74. The dihydropyrimidine dehydrogenase (DPYD) gene encodes an enzyme which plays a role in the rate-limiting catabolism step of 5-Fluorouracil metabolism. Specific sequence variants are known to impact on the activity of the enzyme. The presence of these variants can confer an increased risk of

severe, and even fatal, toxicity when cancer patients with one or more copies of these variants are treated with the fluoropyrimidines; capecitabine or 5-Fluorouracil. If treating clinicians are aware that a patient is heterozygous or homozygous for one of these variants, in many cases they will be able to adjust therapy regimes to reduce the risk of toxicity to the patient.

75. During 2020/21 it is anticipated that DPYD testing will become routinely commissioned through the national genomic testing network. The NHS GMS Alliances will need to work with the NHS GLHs and NHS England and NHS Improvement to develop a plan for implementation.

Pharmacogenomics

76. In November 2018, NHS England and NHS Improvement and Genomics England convened a working group, involving experts from across the UK to explore the science, emerging evidence-base and opportunities for implementing pharmacogenomics in the NHS in England.

77. To help determine future commissioning decisions, it is anticipated that during 2020/21 a pilot will be initiated to evaluate operational and clinical implementation of pharmacogenomic testing

Deliverables

- **Throughout 2020/21 put in place a mechanism to monitor equity of access to clinical trials based on consideration of genomic testing.**

- **Throughout 2020/21 put in place a mechanism to monitor delivery and equity of access to where genomic testing influences personalised treatments and interventions, using existing data sets and working in collaboration with existing oversight committees, such as the Regional Medicines Optimisation Committee.**

- **Support the introduction of DPYD testing in-line with updates to the National Genomic Test Directory.**

- **Where appropriate, actively contribute to the pharmacogenomic pilot, in collaboration with NHS England and NHS Improvement and the NHS GLH.**

Using technology to evaluate and improve services

78. During the NHS GLH procurement process it was expected that the informatics platform referred to as the National Genomics Informatics Service (NGIS) being developed by Genomics England would provide an end-to-end electronic test ordering solution for all genomic tests. However, as the development of the platform has progressed, the scope of the platform for the initial phase has had to be reduced to focus on whole genome sequencing only.

79. Ultimately our aim is to create a national genomic testing network with an underpinning informatics system based on interoperability which enables the NHS to move away from paper-based systems. NHS England and NHS Improvement is currently working with our national partners, such as NHS Digital, and the NHS GLH Informatics Leads to better scope the requirements of the system, define the deliverables and set a clear roadmap that will meet the operational needs of the NHS.

80. Collectively our priority is to work together to define the standards and system requirements so that the informatics solution developed is fit for purpose for the future.

81. Aligned with the work of the NHS GLHs, it is necessary to understand what additional national informatics infrastructure needs to be put in place to drive improvements in health outcomes for patients and provide a pathway that is truly personalised healthcare. This includes linking to the broader digital health integration agenda such as LHCREs, personalised care records and pathology networks to enable the integration of data and linking with integrated diagnostic reporting.

82. To support the development of the national strategy, the NHS GMS Alliance will need to work NHS England and NHS Improvement to develop a genomics informatics and data strategy that will coordinate the informatics approaches across the and facilitate the integration of genomics data within electronic patient records across the geography, including for example pharmacogenomic alerts into EPR systems across the care continuum.

Deliverables

- **Throughout 2020/21 work with NHS England and NHS Improvement to define the national informatics and data requirements to support**

integration of genomics with the broader digital health agenda across the NHS and to support the ongoing development and integration of NGIS.

- **Throughout 2020/21 work with national organisations to integrate genomic data within electronic health records and within integrated diagnostic reporting to improve co-ordination of care, support better clinician decision-making and facilitate seamless clinical pathways.**

Evolving the role of clinical genetics and genetic counselling services

83. Within clinical genetics services there is increasing demand for genetic counsellors and clinical geneticists and demand could outstrip supply. To meet the increased demand, more work is required to integrate genomic testing into certain end-to-end clinical pathways in other clinical specialities.

84. Continued access to the expertise of clinical geneticists and genomic counsellors will be fundamental for the continued delivery of the NHS Genomic Medicine Service. However, further work needs to be undertaken to put in place a sustainable and flexible workforce model that ensures the expertise is being used in the most effective way across the geography working with other health professionals in new and innovative ways.

85. Throughout 2020/2021 the Genomics Clinical Reference Group, with input from the clinical genetic community, will recommend updates to the clinical genetics service specification. The development of the service specification will need to consider new delivery models such as genomic counsellors supporting clinical nurse specialists to embed the skills within defined clinical pathways.

Deliverables

- **Throughout 2020/21 work with existing services to begin to develop coordinated ways of working across the geography to support broader provider access and to widen the involvement of other clinical specialities and services.**

- **Throughout 2020/21 support NHS England and NHS Improvement to define criteria for access to clinical genetics by other specialities and**

support the development of appropriate referral pathways that are sustainable for future service demands.

Priority 5 – Supporting the multi-professional workforce to use genomics safely, effectively and efficiently

Developing the workforce

86. The systematic implementation of genomics within the healthcare system and rising levels of genomic testing presents a challenge for the workforce. There is a growing need for genomic understanding across many clinical specialities as it becomes embedded into clinical pathways. Not all clinicians need to become genomic experts, but the workforce will need sufficient, up-to-date knowledge of genomics in their field; and the skills and confidence to discern when testing may be relevant for their patients, how to request it; and to understand and act appropriately on receipt of the results.

87. To meet growing demand and clinical need for molecular diagnoses to be made to inform treatment decision making, more work is required to integrate genomic testing into end-to-end clinical pathways, embed genomics and its clinical utility across medical specialities and across the whole multi-professional clinical team.

88. More work is needed to understand to the touchpoints where education and training will be required to support and upskill the multi-professional workforce.

89. The NHS GMS Alliances will identify a senior workforce development lead to understand the multi-professional workforce needs to embed genomics into healthcare in a safe and efficient way and establish a workforce development strategy. This should link with the existing work being undertaken by the NHS GLH Education and Training Lead and with the national workforce group lead by NHS England and NHS Improvement and Health Education England.

Nursing collaborative

90. The national nurse and midwifery-led genomics collaborative programme will look at service need and staff development requirements to deliver genomics on the front line for patients. The programme will systematically see nurses and midwives take a lead in testing and redesigning pathways of care in their own clinical work areas, incorporating genomics into daily practices and bringing it into mainstream care. This programme will also include health

visitors and some allied healthcare professions where they are an integral part of the pathway of care.

91. The NHS GMS Alliances will, with the support of the Chief Nurses identified in the NHS GMS Alliance, design a local transformation plan to identify project areas of engagement and transformation.

Medical collaborative

92. The medical collaborative programme involves engaging with the Medical Royal Colleges, the Academy of Medical Royal Colleges, and medical leaders across the geography to agree clinical areas where a transformation and improvement approach to systematically embedding genomics within pathways of care, including primary care, would deliver better outcomes for patients particularly in areas of unmet clinical need and driving personalised and precision medicine.

93. The NHS GMS Alliances are expected to work with NHS England and NHS Improvement and in partnership with the Academy of Medical Royal Colleges and its constituent Colleges, to initiate a Medical Collaborative project.

94. The NHS GMS Alliance will need to develop a plan for transformation projects which meet one or more of the following objectives:

    i) Align and coordinate activity relating to genomics across the clinical community to support systematic implementation across the country of end-to-end genomic pathway design, equity of access to testing, safety and quality

    ii) Gather and assess the emerging evidence on the delivery of genomic medicine across clinical specialities and support the sharing of information, knowledge and understanding of best practice

    iii) Support clinical leadership at a local level to work through the service workflow challenges and obstacles and to identify implementable solutions

    iv) Influence and inform provider / commissioning plans and service specifications to support the embedding of genomic medicine across medical specialities from a practical and logistical perspective

v) Understand the implications on the medical workforce and requirements for job plans and training and education

Pharmacy collaborative

95. The Pharmacy Collaborative will bring together pharmacy system leaders and clinical pharmacy leads with specialist expertise in genomics to achieve the following aims:

i) review workforce development needs and strategically deliver education and training opportunities to support the pharmacy workforce to realise the benefits of genomics for medicines optimisation and personalisation of treatment interventions.

ii) establish the role of pharmacy in clinical pharmacogenomics and in ensuring the safe, effective and equitable use of personalised treatments, including establishing metrics and linking the work of the GMS with the wider medicines optimisation structure in the NHS e.g. Regional Medicine Optimisation Committees

iii) work with NHS England and NHS Improvement to support the strategy for implementation and evaluation of pharmacogenomic testing.

Deliverables

- **In Q1 2020/2021 have worked with NHS England and NHS Improvement to agree a plan for the delivery of the nursing and midwifery transformation projects, including timelines, key milestones and how outcomes will be measured.**

- **By Q2 2020/2021 have worked with NHS England and NHS Improvement to agree a plan for the delivery of the proposed medical programme transformation projects, including timelines, key milestones and how outcomes will be measured.**

- **Throughout 2020/2021 actively support the establishment of the national pharmacy network and the involvement and engagement of chief pharmacists across the geography.**

- **Throughout 2020/2021 work with NHS England and NHS Improvement to develop recommendations on the role of pharmacy in clinical pharmacogenomics and in the personalisation of treatment interventions**

including linked working with regional pharmacy teams and medicines optimisation committees.

Priority 6 - Maximising opportunities to facilitate and participate in research and innovation

NHS GMS Research Collaborative and genomic research projects

96. A key aim of the NHS GMS, in line with the NHS Long Term Plan commitment, is to support research and development including through continuing to develop a single genomic knowledgebase to allow research and industry collaboration on a national scale.

97. Overtime it is anticipated that the number of WGS and non-WGS genomic tests performed in the NHS will increase and more of these non-WGS tests will be at the level of whole exome or large panel testing. To secure the potential benefits from the increased testing we need to secure greater alignment with research to improve the quality of the outputs and improve translation into clinical practice as rapidly as possible.

98. Through working collaboratively, through the NHS GMS Strategic Research Collaborative, there is an opportunity to create an effective approach to embedding research and discovery to advance clinical care for patient and societal benefit.

99. The NHS GMS Research Collaborative will receive and review proposals and creative collaborative bids, support identification of unmet need and respond to emerging technologies. This will include commercial and non-commercial opportunities, including those through the NHS GMS and NHS GLH, and Genomics England structures, that have come from:

    i) Government priority initiatives
    ii) NHS organisations
    iii) Academic institutions
    iv) Voluntary sector / charities / patient groups
    v) Commercial and industry partners

100. The NHS GMS Alliance is expected to play an active role in the NHS GMS Strategic Research Collaborative, working in partnership with the NHS GLHs, and will have a named research and innovation representative to attend NHS

GMS Research Collaborative meetings. This individual will have delegated responsibility to support decision making about research opportunities on behalf of their geography.

101. In addition, the NHS GMS Alliances will provide expertise to help assess the feasibility of a research project that requires genomic testing or use of limited tissue and samples, support identification of required patient populations within their geography to participate in research and provide expert feedback on potential research opportunities from academic or commercial organisations to help determine whether the research could be delivered.

102. As part of the work of the NHS GMS Research Collaborative, metrics will be developed to measure how many projects each NHS GMS Alliance has supported.

Deliverables

- **By Q1 2020 have identified a lead for the NHS GMS Alliance to represent at meetings of the NHS Genomic Research Collaborative.**

- **Throughout 2020/21 create effective genomic research networks across the NHS GMS Alliance geography**

- **Throughout 2020/21 establish metrics to demonstrate the NHS GMS Alliance contribution and commitment to emerging projects and initiatives and to mapping the genomic research and innovation landscape in the NHS.**

Priority 7 –Operating as a national network and representing the NHS GMS internationally

103. To help strengthen the national development of the NHS GMS, the NHS GMS Alliances will be expected to actively participate and contribute to the development of the national network, by supporting the development of national projects, contributing to meetings and actively leading on tasks on behalf of the rest of the network.

104. Many healthcare systems across the world are keen to understand more about the structure and role of the NHS GMS to help inform their own healthcare strategies and approaches to embed personalised medicine. To enable the NHS to benefit from all potential collaboration, investment and development opportunities the NHS GMS Alliances will be expected to share information about potential international collaborations and identify resources as appropriate to support the development of system wide partnerships and international collaborations if deemed appropriate.

Deliverables

- **By Q1 2020/21, identify a single, nominated representative to work as part of the national alliance network and sit on national governance groups.**
- **Actively contribute to the development of the NHS GMS national network.**
- **Actively support the development and promotion of the NHS GMS brand and any nationally agreed international collaborations.**

Annex 1 – draft KPIs

105. Final KPIs will be agreed as part of business planning. Some draft KPIs have been outlined in the table below.

KPI Level	Target Performance Level	Minor KPI Failure	Serious KPI Failure
Remedial Steps	No action required	(1) NHS GMS Alliance to engage in proactive discussion with NHS England and NHS Improvement; (2) NHS GMS Alliance to propose a Corrective Action Plan; and (3) NHS England and NHS Improvement reserves the right to offer the assistance of an improvement team.	(1) NHS GMS Alliance to engage in proactive discussion with NHS England and NHS Improvement; and (2) NHS GMS Alliance to propose a Corrective Action Plan; 3) NHS England and NHS Improvement reserves the right to offer the assistance of an improvement team; and (4) change the configuration of the Partnership (and, in either circumstance, NHS England and NHS Improvement shall have discretion as to whether the NHS GMS Alliance still retains the status designation as a NHS GMS Alliance, taking into consideration the severity of the breach

No.	Key Performance Indicator Definition	Measure of KPI	Measurement Period	KPI Levels
KPI1	99% of all primary diagnostic reports returned to participants by end of Q1 2020.	Number of primary findings results returned to participants.	April – June 2020	Target Performance Level: > = 99% Minor KPI Failure: 95-98.9% Serious KPI: Less than 95%
KPI2	95% of additional findings results returned to participants within 6 weeks of results being made available by Genomics England on the portal, with all results returned by December 2020.	Number of additional findings results returned to participants within 6 weeks from results being made available by Genomics England.	April – December 2020	Target Performance Level: > = 95% Minor KPI Failure: 90-94.9% Serious KPI: Less than 90%
KPI3	80% of patients receiving a WGS test being offered the	Data to be collected from NGIS. *Formula*	Contract period	Target Performance Level: > = 80%

No.	Key Performance Indicator Definition	Measure of KPI	Measurement Period	KPI Levels
	opportunity to take part in research.	Total number of patients receiving a WGS test who have had the opportunity to participate in research *divided by* Total number of patients receiving a WGS test *multiplied by* 100		Minor KPI Failure: 75-79.9% Serious KPI: Less than 75%

KPI4	Development and sustainability of NHS GMS Alliance Network	NHS England and NHS Improvement will create a balanced score from a component balanced score card: each element will be given a 25% weighting and a scoring between 0-5. The following elements will be included, without limitation. For the avoidance of doubt, this is a non-exclusive list of the elements considered: (i) Evidence of comprehensive engagement and involvement with all providers and networks across the geography including: • contribution to the delivery of the NHS GMS Alliance business plan; • that the relationships with NHS GMS Alliance Network are credible; and • that there is explicit involvement of the Academic Health Science Network and active engagement with other networks across the Geographical Area (e.g. strategic clinical networks, Cancer Alliances, professional networks), and clear plans to share knowledge and experience in genomics for patient benefit; (ii) level of clinical engagement across NHS GMS Alliance geography. Evidence of the following required: • Multi-professional clinical leadership across the end-to-end genomic pathway (iv) Patient Public Involvement, including, without limitation: • Communication with patients and communities regarding the genomics and the	Contract period	The Authority will further define these levels in due course.

No.	Key Performance Indicator Definition	Measure of KPI	Measurement Period	KPI Levels
		development of the NHS Genomic Medicine Service • NHS GMS Alliance carrying out PPI activities across the whole geography including:(i) establishing PPI group(s) and demonstrating continuous involvement; and (ii) implementing innovative PPI engagement initiatives across the Geographical Area;		
KPI5	Once established, NHS GMS Alliance geography actively taking part in at least 90% of relevant research projects approved by the NHS Genomic Research Collaborative	Data collated through NHS GMS Research Collaborative. *Formula* Total number of research projects approved by NHS Genomic Research Collaborative which the NHS GMS Alliance actively participates in *divided by* Total number of research projects approved by NHS Genomic Research Collaborative which is appropriate for the NHS GMS Alliance to participate *multiplied by* 100	Contract period	Target Performance Level: > = 90% Minor KPI Failure: 85-89.9% Serious KPI: Less than 85%